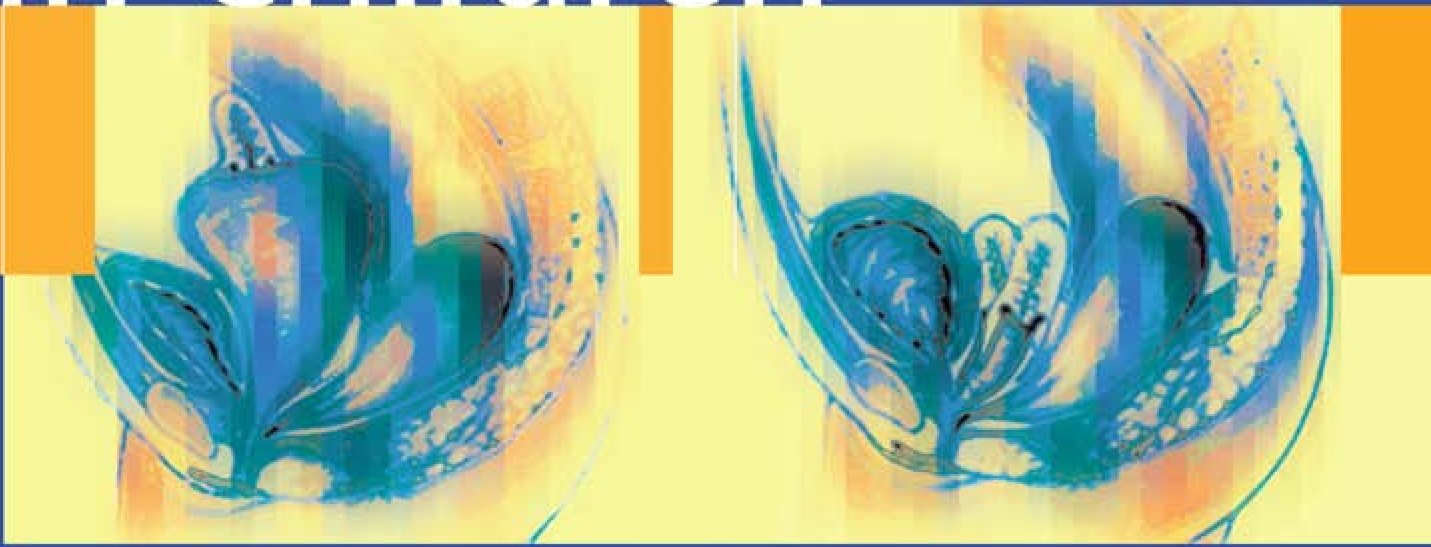
A. M. Holschneider
J.M. Hutson *Editors*

Anorectal Malformations in Children



Embryology
Diagnosis
Surgical Treatment
Follow-up

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Anorectal Malformations in Children

Alexander M. Holschneider · John M. Hutson
Editors

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Embryology, Diagnosis, Surgical Treatment, Follow-up

With 387 Figures

Springer

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Foreword

It is 43 years since we (F. Douglas Stephens, Robert Fowler, and others) produced the first volume of a careful analysis of the anatomical pathology of the many lesions of anorectal anomalies, describing the relationship between the controlling sphincters and the incompletely developed bowel as a logical basis for operative correction [1]. Eight years later under our joint authorship we published the first comprehensive text of the entire subject as known at that time [2], which incorporated the "international classification" developed at a workshop in an international conference held at the Royal Children's Hospital, Melbourne in 1970. The subject was again reviewed 13 years later at another workshop under our chairmanship at the Wingspread Conference Center in Racine, Wisconsin, USA, at which time the classification was simplified. By then, the monumental contribution of Peter De Vries and Alberto Peña regarding the posterior sagittal approach had been published, which revolutionized the operative management of high lesions, and a new edition with multiple authors was called for [3].

Over the next 17 years there were significant studies by younger colleagues, and with our subsequent retirement from clinical surgery, a new work was clearly required. Professor Alexander Holschneider of the Kinderchirurgische Klinik, Lehrkrankenhaus der Universität zu Köln, Germany, is to be congratulated in taking the initiative, and no one is more eminently qualified to do so, having made many important contributions regarding this lesion in his own right. The result is the present volume, superbly edited by our colleague, Professor John Hutson of the Royal Children's Hospital, Melbourne, and Professor Holschneider. It, too, follows an international workshop, organized by Alex Holschneider, who assembled a team of 25 international experts meeting in the picturesque Krickenbeck Castle north of Cologne, Germany, in May 2005.

Although the clinical features and recognition of the various anatomical subtypes has not greatly changed, they are, of course, described in the present work, including a useful summary as an insert. Also included is an atlas of sections of fetal specimens from the original studies of one of us (FDS). This basic anatomical knowledge and clinical recognition is required reading for any pediatric surgeon caring for affected children, and there can be no excuse from not acquiring a thorough grasp of the many complexities of the pathological anatomy of the bowel, fistulas, and surrounding sphincters, including familiarity with the assessment of the muscle integrity and the varieties of sacral nerve outflow. Only on this basis can decisions on management be logically made. Much of the evidence can be acquired from clinical observation with eye and probe, but nevertheless, newer modalities of investigations assisting diagnosis are herein well described – a reevaluation of the technique, posture, and interpretation of the traditional "invertogram", magnetic resonance imaging, electromyography, and endoscopic ultrasound.

New work also includes important new concepts of the early embryological processes of abnormal growth in cloacal membrane development, derived from animal models, and an update on the genetics of anorectal anomalies, including the identification of the genetic basis of Currarino syndrome.

No subject has been more controversial than the classification of anorectal anomalies. The distinction must be made between "classification" based on anatomical pathology and/or embryology, and a "diagnostic plan". To be complete, the former must necessarily be large and complex, describing many subtypes, because that is the nature of this lesion, and was the basis of the international classification of 1970. A diagnostic plan is a recognition of related anatomical features of subtypes in order to make a clinical decision regarding treatment; it is not a classification. In order to reduce the complexity of the international classification, which is not accepted in some centers, the simpler Wingspread classification was introduced in 1984. These classifications are rightly included in the current text and are still required knowledge, but the Krickenbeck workshop took a different approach. One important aspect of an agreed classification is that it facilitates a comparison of operative results from different surgeons operating on the same lesion. However, the number of common operative procedures is much smaller than the number of anatomical subtypes, so it seemed useful to list all those subtypes together for which there was a generally agreed single operative procedure. In this way, the results of a particular procedure could be compared irrespective of the particular subtype. The workshop therefore proposed only a small number (7) of "major clinical groups", each group with its own operative procedure. Reference to this list indicates that the new concept should work well in such "high" groups as rectourethral fistulas, rectovesical fistulas, and cloacal lesions, but perhaps less satisfactorily for lesions traditionally labeled "low" or "intermediate". It is therefore not surprising that some groupings are controversial. Are there different levels of rectovestibular fistula requiring two different operative approaches depending on length of fistula? Are all perineal fistulas treated the same way or do they vary from simple to complex? The category of "no fistula" is its own heading, implying a common method of treatment, yet its subtypes vary from a simple "covered anus" by skin folds, an equally simple "imperforate anal membrane", both of which require very minor surgery, to more complex imperforate anus and rectum ending blindly in levels varying from the area of the bulb of the urethra to high in the pelvis, which require major reconstruction. Experience will establish whether the new scheme will prove satisfactory.

An interesting feature of classification is the major input from colleagues from the Indian and Asian subcontinent, who report considerable differences in the incidence of various lesions; they contribute extensive experience regarding the operative management of several anomalies uncommonly seen elsewhere, and their contribution is essential to this text.

Operative management continues to be dominated, and rightly so, by the enormous contribution and vast experience of Alberto Peña by the introduction of posterior sagittal anorectoplasty (PSARP), and this experience is updated in the current text, especially with respect to cloacal anomalies and total urogenital sinus mobilization; nevertheless, there remain many varieties of detail of PSARP executed by others, while retaining the principal features of this approach. This is especially so in the management of "low" lesions, and alternative approaches are described. In addition, two significant operative procedures are now included – a technique of vaginal reconstruction by Arnold Coran, and the growing experience of the endoscopic repair of several anomalies, which may become the standard approach for high lesions.

One of the major discussions at the Krickenbeck workshop concerned the postoperative assessment of results. Many schemes have been tried in the past, each varying in the parameters to be assessed, and consequently comparison of results has been almost impossible. A simple clinical scheme has now been suggested; it does not result in a numerical "score," but may permit at least a degree of subjective analysis of results. It recognizes the importance of constipation in affecting fecal control, and the value of behavioral training in treatment. Considerable new work is now recorded in the assessment of muscle and nerve integrity and of bowel motility by electromyography, endosonography, and electromanometry, the latter particularly by Alex Holschneider. Adult sexual function is also addressed.

The final chapter is unique and extremely valuable. It is the first time a significant study of results assessed by the direct experience of parents and care support groups has been included in a standard surgical text. Not only is there much detailed factual information of the children's long-term symptoms after surgery, but also some penetrating comments as to how we, as surgeons, have often failed our patients and parents in communication and empathy. No matter how enthused we may be by the practice of surgery, and no matter how dedicated we might be in our endeavor to care for our patients to the best of our ability, none of us can feel to the same extent the depth of the burden suffered by some parents and some children struggling with the practicalities of daily living when results are suboptimal. The input of these writers is beautifully and sensitively written, and it is a salutary reminder that we are always and only the servants, never the masters, of our patients.

We warmly recommend this new book. We congratulate Alexander Holschneider on his enthusiasm and professional expertise in bringing to fruition this new edition after 18 years since the last update, and John Hutson for the masterful editing of a very complex subject. We wish it well.

F. Douglas Stephens
E. Durham Smith

References

1. Stephens DF (1963) Congenital Malformations of the Rectum, Anus and Genito-Urinary Tract. E. and S. Livingstone, Edinburgh and London
2. Stephens FD, Smith ED (1971) Anorectal Malformations in Children. Yearbook Medical Publishers, Chicago
3. Stephens FD, Smith ED (1988) Anorectal Malformations in Children: Update 1988. Alan R. Liss, New York, and March of Dimes Birth Defects Foundation

Preface

This multiauthor book is an update on the science and surgery of malformations of the rectum and anus. It carries on Douglas Stephens' book "Congenital Malformations of the Rectum, Anus, and Genito-urinary Tracts" published in 1963. This first book, which deals exclusively with malformations of the lower end of the digestive and urogenital tracts, was based on fundamental studies on paediatric pathology, surgery and surgical anatomy performed at the Department of Surgical Research of the Royal Children's Hospital, Melbourne, Victoria, Australia. Until today these studies have represented the embryological and pathoanatomical basis of our knowledge in the diagnosis and treatment of anorectal malformations (ARM). In 1971 Douglas Stephens and Durham Smith published the first update of their book, called "Ano-Rectal Malformations in Children". It became the standard work for ARM for the following 17 years. In 1984 an international workshop took place at the Wingspread Convention Center, Wisconsin, USA, hosted by the Department of Surgery, Chicago Children's Memorial Hospital, where Douglas Stephens worked at that time. The chief objects of that meeting were an update of the approximately 170 years of experience with modern treatment of ARM and to set standards for the classification and treatment of this malformation. At the end of the conference the so-called Wingspread classification was settled, technical details for abdominal, sacral, and perineal approaches were proposed and the great variety of ARM listed again. The results of the Wingspread meeting were finally published by Stephens and Smith in 1988 with support of the March of Dimes Birth Defects Foundation at Alan R. Liss, New York [1]. The Wingspread classification of ARM divided ARM into high, intermediate, and low types and correlated the individual underlying pathoanatomy with the appropriate surgical procedures. This meant, roughly speaking, that a perineal approach should be performed for low-type, a sacral approach for intermediate-type, and an abdomino-sacro-perineal pullthrough for high-type malformations. These Wingspread considerations continue to have great influence on the diagnosis and therapy of ARM.

As time went on, however, new aspects were developed, particularly concerning the surgical therapy of children with imperforate anus. Special merit should be given to Alberto Peña, Cincinnati, USA, who described the sacral approach as the method of choice for almost all types of imperforate anus. Peña and de Vries described in 1982 the important details of the posterior sagittal anorectoplasty, which became the classic approach for the treatment of ARM in the subsequent years [2, 3]. This more simplified concept was based on the observation that the anatomical structures described by anatomists could hardly be identified during the operation. The different structures of the levator muscle, the puborectalis sling and the three slings of the external anal sphincter muscle could frequently only be realised as a muscle complex. According to the large experience of Alberto Peña with thousands of patients operated by himself in his former centre for ARM, the Jewish Hospital in Long Island, New York, USA, and throughout the whole world, a therapeutic concept based on anatomical observations seemed to be less important to him than a classification based on clinical experience.

Therefore, in 1990 Peña published an "Atlas of Surgical Management of Anorectal Malformations", describing in detail his new procedure, and in 1995 a clinical classification of ARM according to the type of the associated fistula. By closely comparing both proposals, the Wingspread classification and Peña's suggestions, it became clear that there was no real contradiction between them. Perineal and vestibular fistulas could be regarded as low malformations, bulbar fistulas, imperforate anus without a fistula and some of the vestibular fistulas may be regarded as intermediate-type anomalies, and prostatic and bladderneck fistulas are considered as high-type imperforate anus. However, it became evident that a new conference, 21 years after the Wingspread meeting, would help to clarify these problems. Therefore, an International Conference for the Development of Standards for the Treatment of Anorectal Malformations was organized at Krickenbeck Castle near Cologne, Germany (17–20 May 2005). This workshop brought together 26 international authorities on congenital malformations of the organs of the pelvis and perineum. Recent advances in aetiology and genetics, diagnosis, early and late management and methods of improvement of urorectal continence were reviewed. In addition, the participants developed a new international classification for ARM and a new grouping for follow-up assessment and standard surgical procedures. The principle idea of the Krickenbeck workshop and the subsequent international conference on 21 May 2005 in Cologne was to enhance the current fundamental concepts in the diagnosis and treatment of ARM, to update the recent knowledge on this not infrequent congenital malformation and to prepare this new update of Stephens and Smith's book from 1988 (Fig. 1).

The editors would like to thank Mrs. Gabriele Schröder and Mrs. Stephanie Benko, Springer International Publishers, for their interest and agreement to publish this book. We would also like to thank Mr. Janis Biermann, The March of Dimes Birth Defects Foundation and Alan R Liss, New York for giving us back all rights for publishing, tables, figures and chapters of the previous edition. We are especially pleased and honoured, that the former editors F. Douglas Stephens and E. Durham Smith attended the Krickenbeck Conference and helped with their advice and contributions to continue with their work. Special thanks go to Alberto Peña, who contributed tremendously to this book with many chapters written together with his associate Dr. Marc Levitt. Professor Peña's influence has changed fundamentally the concept of the former edition as he has changed the concept for the diagnosis and treatment of ARM. The Krickenbeck conference and this book are now building up a bridge between the important and still valid pathoanatomical considerations published by Stephens and the large clinical experience described by Peña. The anatomical aspects are supported by an unpublished series of autopsies performed by F. Douglas Stephens in children with imperforate anus who died from other reasons. His findings are presented in this book on a CD with a special index (Chap. 6). They confirm the clinical observations of Peña in a magnificent way. However, they also point out the necessity for an accurate anatomical knowledge of the individual deformity. The new classifications proposed at the Krickenbeck Conference are part of Chaps. 8 and 25. They have also been published by Holschneider et al. as a preliminary report [4]. The authors would like to thank Professor Jay Grosfield for his help for the quick and uncomplicated acceptance of this report.

We would also like to thank all of the co-authors who have contributed their time and effort to the research with or without the support of their parent universities, institutions, or hospitals; none will receive royalties on the sale of this book. Thanks are due to their supporting institutions, the names of which appear in the list of contributors.

Members of many disciplines in hospitals and universities have played important roles in the elucidation of the occult structural anomalies and the overall management of afflicted babies. In this context we are especially grateful to Professor J. Koepke, Head of the Anatomical Institute of the University of Cologne, Professor W. Lierse, former Head of the Institute for Neuroanatomy and Anatomy of the University of Hamburg, and Professor W. Meier-Ruge, Basel, Switzerland, for their support and advice in solving anatomical and pathological questions dealing with the pathophysiology of ARM.

Many other co-workers like physicians, nurses, radiologists, ancillary artists, photographers and hardworking secretaries in many countries have contributed their knowledge and expertise generously to the research, diagnosis, and management of ARM and the manuscript of this book. Mrs. Elisabeth Herschel at the Children's Hospital of the City of Cologne, Germany, and Mrs Shirley D'Cruz at the Royal Children's Hospital in Melbourne, Victoria, Australia, had exceptionally onerous work keeping track of correspondence and manuscripts and retyping, and we thank them for work well done.

Last but not least we would like to thank Dr. Winfried and Danielle Hartwick, Meerbusch, Germany and the Foerderverein Blankenheimer Dorf, Blankenheim, Germany for supported the idea of the Krickenbeck Conference financially. We are grateful to Mr. Thomas Gemein for good cooperation with the Verein der Freunde and Förderer des Kinderkrankenhauses Amsterdamer Strasse, Köln and the WestLB Akademie Schloss Krickenbeck, and Mrs. Svitlana Görden, Düsseldorf/Germany for the organisation of the Krickenbeck Conference.

All of the authors would like to thank all the parents' associations for children with ARM for their confidence and support of our daily work. We are especially grateful for the contribution of their experience and data to this book.

Alexander M. Holschneider, Köln
John M. Hutson, Parkville
April 2006

Fig. 1 Group photograph of the participants at the International Conference for the Development of Standards for the Classification and Treatment of Anorectal Malformations, Krickenbeck, Germany, 17–20 May 2005. *Upper row* (left to right): Naomi Iwai, Kyoto, Japan; Guiseppe Martuciello, Pavia, Italy; Dieter Kluth, Hamburg, Germany; Thomas Boemers, Cologne, Germany; Keith Georgeson, Birmingham, Alabama; Alberto Peña, Cincinnati, USA; Alexander Holschneider, Cologne Germany; Devendra Gupta, New Delhi, India. *Middle row*(left to right): Sudipta Sen, Vellore, India; Middle row right: V.Sripathi, Chennai, India; Sabine Grasshoff, Germany; Feilim Murphy, Dublin, Ireland. *Lower row* (left to right): Reinhold Engelskirchen, Düsseldorf, Germany; Risto Rintala, Helsinki, Finnland; Benno Ure, Hannover, Germany; Samuael Moore, Stellenbosch, South Africa; Michael Davies, Cape Town, South Africa; Arnold Coran, AnnArbor Michigan, USA; Durham Smith, Victoria, Australia; Douglas Stephens, Toorak, Australia; John Hutson, Melbourne, Australia; Subir Chatterchee, Calcutta, India; Jay Grosfeld, Indianapolis, USA; Yunus Sölet, Istanbul, Turkey; Elhamy Bekhit, Parkville, Australia. Photograph taken with permission from Holschneider et al. [4]

References

1. Stephens FD, Smith ED (1988) Anorectal Malformations in Children: Update 1988. Alan R. Liss, New York, and March of Dimes Birth Defects Foundation
2. DeVries P, Peña A (1982) Posterior Sagittal Anorectoplasty. J Pediatr Surg 17:638–643
3. Peña A, DeVries PA (1982) Posterior Sagittal Anorectoplasty: Important Technical Considerations and New Applications. J Pediatr Surg 17:796–811
4. Holschneider A, Hutson J, Peña A, Bekhit E, et al (2005) Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. J Pediatr Surg 40:1521–1526

Krickenbeck Consensus for the Classification, Grouping of Surgical Techniques and Scoring for Follow Up of Anorectal Malformations

Standards for diagnostic procedures: International Classification (Krickenbeck)

Major clinical groups	Perineal (cutaneous) fistula
	Rectourethral fistula
	Bulbar
	Prostatic
	Rectovesical fistula
	Vestibular fistula
	Cloaca
	No fistula
	Anal stenosis
Rare/regional variants	Pouch Colon
	Rectal atresia/stenosis
	Rectovaginal fistula
	H type fistula
	Others

International grouping (Krickenbeck) of surgical procedures for follow up

Operative procedures	Perineal operation
	Anterior sagittal approach
	Sacroperinal procedure
	PSARP
	Abdominosacroperineal pull-through
	Abdominoperineal pull-through
	Laparoscopic-assisted pull-through
Associated conditions	Sacral anomalies
	Tethered cord

Method for assessment of outcome established in Krickenbeck 2005 (patient age > 3 years, no therapy)

1. Voluntary bowel movements	**yes/no**
Feeling of urge	
Capacity to verbalize	
Hold the bowel movement	
2. Soiling	**yes/no**
Grade 1 Occasionally (once or twice per week)	
Grade 2 Every day, no social problem	
Grade 3 Constant, social problem	
3. Constipation	**yes/no**
Grade 1 Manageable by changes in diet	
Grade 2 Requires laxatives	
Grade 3 Resistant to diet and laxatives	

For further details see acknowledgement and chapters 8 and 25.
Preliminaray report in JPS 2005, 40:1521–1526.

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General Aspects

1 ARM – a Historical Overview

Jay L. Grosfeld

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Anorectal malformations (ARM) are relatively frequently encountered anomalies that represent an important component of pediatric surgical practice. Many in our profession have a significant interest in the management of the numerous variants of ARM that affect both boys and girls. This chapter will attempt to bring the reader up to date through a historical overview of these fascinating anomalies from the earliest of days until the current era.

1.1 The Early Era: AD 2–1900

ARM have been a source of concern for centuries and have been recognized in animals since the time of Aristotle in the third century BC [6]. Soranus, who is considered the first pediatrician of Rome, changed the prevailing public attitude in the second century AD by not allowing neonates with anomalies to die and described dividing a thin anal membrane and dilating the opening [70, 144]. Paul of Aegineta pierced an anal membrane and used a wedge-shaped tent dilator in the seventh century [119]. In 1576, Galen described the anal sphincters, levator muscles, and coccyx [46]. There were few recorded references regarding these conditions until 1676, when Cooke treated a child by making a small incision over a blind anal membrane and dilated the aperture with an elder pith. He emphasized care of the sphincter muscles to others who sought to duplicate his success [26, 28]. In 1693, Saviard was the first to attempt treatment of a high termination of the bowel by plunging a trocar through the perineum [143]. In 1787, 94 years later, Benjamin Bell performed the first perineal dissection in two newborns, finding the blind-ending rectum at variable lengths from just above the anal area to the level of the coccyx [10]. A trocar was inserted and fecal content evacuated. Prolonged bouginage was required to preserve the open passage using a sponge tent, gentian root, or other substances that swell with moisture [10, 34]. Bell also described instances of rectovaginal and bladder fistulas [10]. In 1792, Mantell published a report concerning a girl with a rectovaginal fistula [101]. In 1786, he had performed an incision in the perineum and carried it up to a probe placed through the vagina into the fistula, creating an anal communication. Reoperation was required 2 years later for “anal” stricture [101].

Colostomy was popularized in the eighteenth century in France. Following an autopsy in an infant with rectal atresia in France in 1710, Littre proposed that the bowel be brought to the surface of the abdomen to function as an anus [94]. The first successful sigmoid colostomy (termed an “inguinal colostomy” or “procedure of Littre”; Fig. 1.1) was performed by Duret in 1793 on a female infant who survived into adult life [39]. The results described by others were not as successful [31]. In 1798, Martin of Lyon suggested insertion of a sound in the colostomy and pushing distally to identify the blind-ending rectum during a later perineal dissection [102]. In 1856, Chassignac reported successful use of this technique in two infants with a colostomy [22]. However, colostomy in the newborn was neither a popular procedure nor was it widely accepted at the time [34].

In 1834, Roux of Brignoles attempted to preserve external sphincter function and used a midline longitudinal incision extended toward the coccyx [141]. The incision continued through the elliptical sphincter ani muscle and levators and when the rectal atresia was palpated, a bistoury (trocar) was inserted into the

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Fig. 1.1 Illustration of the inguinal colostomy (procedure of Littre) popularized in the eighteenth century in France

Fig. 1.2 JZ Amussat performed the first proctoplasty for a patient with imperforate anus

bowel, releasing meconium. Dressings and bouginage were required to prevent occlusion of the opening [1]. In 1835, Amussat performed the first proctoplasty by suturing the opened rectal atresia to the skin in the midline (Fig. 1.2) [4]. This was a landmark procedure at the time and gained wide acceptance, and was used frequently for the rest of the nineteenth century. Amussat used an extensive T-shaped incision that basically destroyed the sphincter mechanism. In some instances he described removing the coccyx to aid exposure and mobilization of the rectum [4]. Techniques to repair rectovaginal and rectovulvar fistulae were described by Dieffenbach in 1845 [36] and Rizzoli in 1854 and again in 1869 [136, 170]. In 1852, Dunglison described the relationship between the longitudinal smooth muscle fibers of the rectum and the external sphincter muscle and mucous membrane [38]. By 1860, Bodenhamer noted that in some, but not all instances of high rectal atresia, the sphincter muscles were detected [12, 13]. Despite this observation he shunned colostomy and recommended that an artificial anus should always be established in the perineum. He championed the midsagittal incision first described by Roux 27 years earlier. In 1873, Verneuil reaffirmed Amussat's observation that coccygectomy facilitated the dissection of a high blind-ending rectum [4, 167]. In 1879, McLoed described an abdominoperineal (AP) procedure for instances in which the blind rectal atretic end was not found below [107]. By 1882, Amussat's procedure had gained favor in the USA [4, 18]. Before that time only sporadic anecdotal reports concerning ARM were noted in America [53]. Prior to the introduction of the aseptic technique by Lister, the operative mortality for both proctoplasty and colostomy was greater than 60% [34]. In 1886, McCormac was one of the few to suggest a two-stage procedure-preliminary colostomy and subsequent proctoplasty [108]. In 1897, Matas combined a sacral approach to rectal atresia with sacrotomy to aid exposure in instances of high-lying anomalies and predicted that this would be the route of choice for these procedures in the future [106]. Matas was not a proponent of colostomy and favored a one-stage procedure in the neonatal period [106]. His bias influenced the care of babies with ARM for the next four decades.

During the pre 1900 era, appreciation of the pelvic and perineal anatomy was influenced by the observations of Vesalius (1543) [168], Galen (1576) [46], and Santorini (1724) [142] who described the anal sphincters, the levators, and the coccyx. In 1874, Robin and Cadiat reexamined these observations and defined the sphincter ani externus [138]. Gowers described the automatic action of the sphincter ani in 1877 [49], and Holl was the first to describe the puborectalis muscle as a separate entity in 1897 [65].

1.2 Anorectal Anomalies in the Twentieth Century

1.2.1 The Barren Era: 1900–1945

Textbooks of Surgery in 1908 recommended colostomy as a life-saving measure only, otherwise the perineal approach was employed for all other cases [75]. Extension of the long median posterior incision was suggested for those instances associated with a rectovesical fistula [105]. Controversy concerning surgical management continued. In 1908, Mastin noted that the AP procedure had little to attract surgeons and could scarcely be viewed as a rational procedure [105]. In 1915, Brenner on the other hand thought that the AP procedure was a novel technique that promised good results in selected cases [14]. By 1922 it was recognized that that the perineal approach was often futile in instances of high rectal atresia [130]. Despite these sporadic observations, by 1926 laparotomy with or without a colostomy was considered a procedure of last resort and was associated with a high mortality [42].

In 1930, Wangensteen and Rice described the radiographic invertogram as a method for determining the level of termination of the rectal atresia and deciding whether a perineal approach was rational [169]. In 1934, Drs Ladd and Gross at the Boston Children's Hospital proposed a classification for ARM (types 1–4) that closely resembled Bodenhamer's work published 74 years earlier [12, 13, 88]. Only one of their 162 cases was managed by a one-stage AP approach and proctoplasty. At the time the success rate for cases of rectovaginal fistula was 50%, that for rectourethral fistula was 20%, and the mortality was in excess of 50%. Dissection was kept close to the hollow of the sacrum, and the external sphincter was divided in lateral halves and resutured in front and behind the proctoplasty [88]. The role of the levators and puborectalis was unknown. In 1936, Stone described transplantation of a low rectovaginal fistula to the anal site without splitting the rectum sagittally. He termed the condition imperforate anus with rectovaginal cloaca [161]. In 1938, an early perineal operation was still considered the method of choice by many surgeons [11].

1.2.2 Post World War II Era: 1945–1980

1.2.2.1 A Time of Enlightenment and Continued Controversy

Following World War II, things began to change. The availability of antibiotics and improvements in anesthesia had a positive influence on reducing the septic complications associated with bowel surgery. In 1948, Rhoads and colleagues in Philadelphia rekindled interest in a combined AP approach for cases of imperforate anus and high rectal atresia [134]. In 1950, Denis Browne of Great Ormond Street, London, UK, reclassified the defects associated with rectal agenesis using a thesis originally described by Wood-Jones in 1904 and 1915 [16, 173, 174]. The term "covered anus" became popular, and initial colostomy and subsequent AP pull-through through a hole stretched (not cut) in the pelvic floor was advocated for high lesions. Browne also popularized the "cutback" anoplasty for instances of perineal fistula [16]. In 1953, Douglas Stephens, while working with Denis Browne in London, described the sacroperineal rectoplasty and emphasized the role of the levator ani and downplayed the importance of the internal and external anal sphincters [153, 154]. Two of the four patients undergoing this procedure achieved continence. In 1959, Fritz Rehbein of Bremen reintroduced the endorectal pull-through combined with an AP approach for boys with rectourethral fistula [133]. The endorectal concept was described by Hochenneg in Austria in 1889 [63]. Rehbein divided the bowel at laparotomy, stripped the mucosa from the distal rectal atretic end and brought the proximal bowel through the resultant muscular sleeve to the anal dimple to perform an anoplasty [133]. He missed the puborectalis sling in performing this procedure. In 1960, Roumaldi of Italy used the same approach for girls with a rectovestibular fistula [140]. In 1961, after extensive dissections, Stephens proposed the importance of the puborectalis muscle as the main muscle of continence, but recognized that soiling accidents continued to occur because of mucus leakage from the anal canal due to total or functional absence of the external sphincter [155]. He suggested that this was a problem that needed to be solved by the mother and child. He also advocated the use of the pubococcygeal line on the diagnostic radiographic invertogram to improve identification of the level of rectal atresia [156].

Because of the high incidence of incontinence with the AP approach, in 1963 Kiesewetter of Pittsburgh modified Stephens' operation by performing

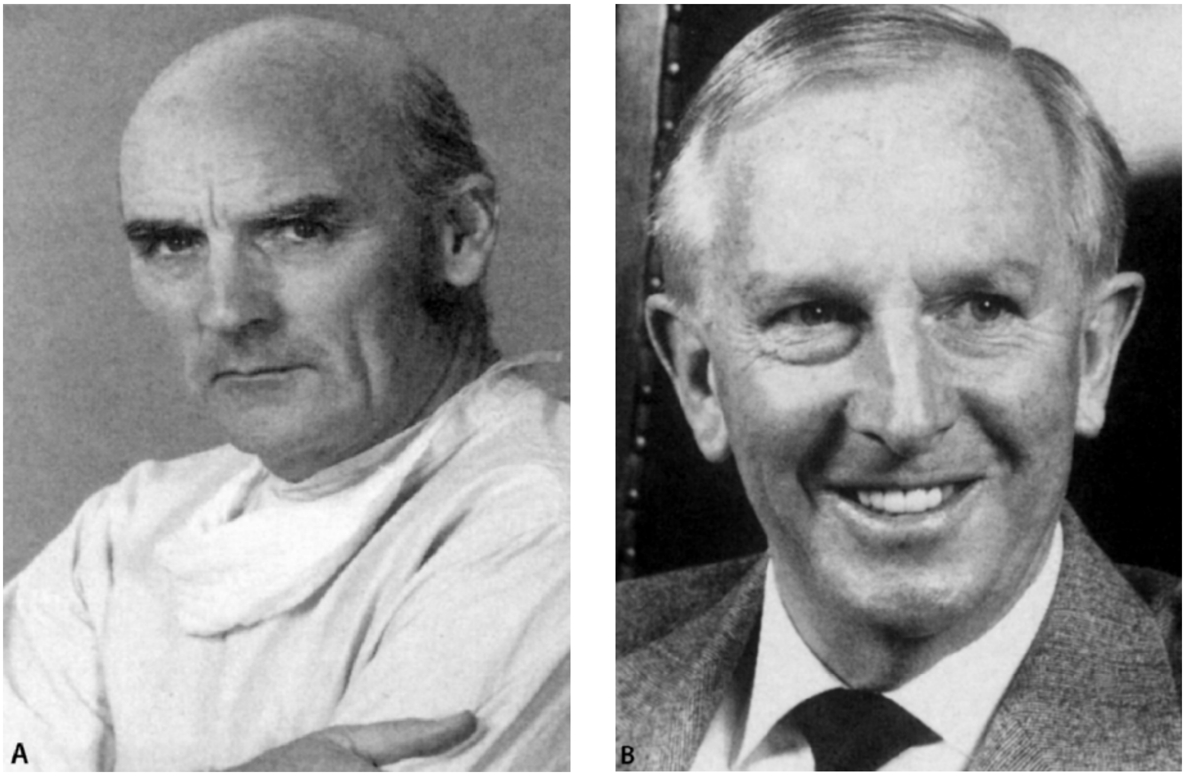

Fig. 1.3 **A** Douglas F. Stephens performed the sacroperineal anoplasty. **B** E. Durham Smith – working with Dr. Stephens edited the first major modern textbook on ARM

an abdominosacroperineal procedure [79]. He used the abdominal approach to isolate the rectourethral fistula and the sacral route to enter the supralevator space by splitting the pubococcygeus and ileococcygeus in the midline. Unlike Stephens, Kiesewetter believed the external sphincter muscle was present and worth saving. Two years earlier (1961), E. Ide Smith and Robert E. Gross had identified a normal-sized external anal sphincter in 15 out of 16 autopsied cases of imperforate anus [150]. Kiesewetter later met with Rehbein and adopted Rehbein's mucosal stripping and endorectal concept, while Rehbein added Stephens' inclusion of the puborectalis sling to his procedure [34]. In 1963, Stephens published the first modern major textbook on the subject [156]. In 1967, Swenson and Donnellan of Chicago described their experience with AP procedures and preservation of the puborectalis sling in the newborn without a colostomy [163]. They thought it was advantageous to locate the anoplasty more anteriorly to avoid tension on the bowel as it passed in front of the puborectalis sling. This completely ignored the external sphincter muscle that Swenson (like Stephens) did not think was important in controlling continence [163]. That same year (1967), Kiesewetter and Nixon found the external sphincter muscle present in all nine cases studied at autopsy, but found a gap between the sphincter and the puborectalis muscle [80]. In 1969, Justin Kelly of Australia reported the innovative use of cineradiographic defecography in an attempt to evaluate the function of the puborecatalis following procedures for ARM [76].

In 1970, Cremin et al. reported that the pubococcygeal line recommended by Stephens on the radiographic invertogram was an unreliable landmark and suggested using an "M" line, especially in cases with missing sacral segments [27]. In 1973, a prone cross-table radiograph was recommended as an alternate to the classic invertogram for diagnosing the level of rectal atresia [112].

In 1970 a workshop was held at the Royal Children's Hospital, Melbourne, Australia to reclassify ARM. A rather complex, male/female low, intermediate, and high classification was brought into effect. The contents of the workshop were published by Stephens and Smith in 1971 [157]. Stephens and Smith (Fig. 1.3a, b) noted that the functional results of the AP procedure and sacroperineal operation were similar. However, they cited problems with the Kiesewetter/Rehbein operation including mucosal prolapse at the anoplasty site, incomplete use of striated muscle, and loss of rectal storage capacity by using the atretic rectal segment for the muscle sleeve. Yet, they agreed that this procedure was more acceptable than an AP approach alone. The controversy continued, and in 1971, Stephens and Smith recommended the sacroperineal approach for redo procedures after an initially inadequate rectoplasty [157]. In 1972, Fowler indicated that since the importance of the puborectalis muscle was recognized, "further finesse in the performance of rectoplasty will center on creating a more functionally efficient sphincter ani" [44]. In 1975, Shafik reported a new concept regarding the anatomy of the anal sphincters and the functional physiology of defecation that he termed the triple loop system [145].

Dissatisfaction with the outcomes of other procedures in 1978 led Mollard to recommend an anterior perineal approach bringing the atretic bowel down in front of the puborectalis sling [110]. Sixty percent of the patients had mucosal prolapse, and soiling continued to be a problem.

Reports recognizing associated anomalies in other midline structures such as esophageal atresia and tracheoesophageal fistula, duodenal atresia, neural tube defects (tethered cord), vertebral anomalies, sacral anomalies, genitourinary anomalies, and instances of congenital heart disease that accompanied imperforate anus became prominent [19, 30, 41, 51, 103, 117, 118, 147, 171]. Acronyms like VATER and VACTERL describe instances of ARM with many of these associated conditions [41, 51, 74, 78, 131]. Certain genetic abnormalities were noted to coexist with ARM, including instances of trisomy 13–15, 16–18, and 21 (Down syndrome) [41, 43, 165]. The absence of associated rectal fistulae in the latter group also became apparent [43, 165]. ARM were also noted to be associated with the cat-eye syndrome (otic atresia and coloboma), Currarino syndrome (rectal stenosis, sacral anomalies, presacral teratoma, or anterior meningocele) and occasionally Hirschsprung disease [24, 29, 41, 86, 89, 98, 104]. It should be noted that although credit has been given to Currarino and associates for recognizing the constellation of defects known as Currarino syndrome, it was Ashcraft and Holder who first described these familial occurrences in 1965 [7, 29]. In India, an unusual subset of patients were identified with imperforate anus and short colon pouch syndrome [8, 17].

The 1970's and early 1980's was a period when children's surgeons reported the results of some of the procedures advocated by Stephens and Smith, Rehbein, Kiesewetter, and others. It became quite clear that different subjective criteria for grading and definitions used by various authors to assess function would make it difficult to compare results. The various studies often focused on the area specifically of interest to the surgeon who advocated the procedure. Comments like excellent, good, fair, and poor characterized the studies, and continence was assessed according to whether one was evaluating puborectalis function or other sphincter activity. Sphincter tone was frequently not mentioned. How motility was evaluated was often in question. Kelly used a scoring system with 6 as a maximum score [77]. Terms like colonic inertia appeared to address the failures and severe constipation that followed the procedure [15, 163]. In 1977, Kiesewetter noted an improvement over time from poor to good in low anomalies and from poor to fair in high anomalies, and that switching from an AP approach to an abdominosacroperineal or sacroperineal approach did not improve function [34]. The same year, Nixon and Puri evaluated 47 children with high anomalies; only seven had a good outcome, 28 were fair, 11 poor, and 7 required a permanent colostomy [114]. The seven good outcome patients soiled for 6–17 years of follow-up. Kiesewetter, Turner and Nixon, and Puri suggested that continence was an "evolutionary process" that improved each year during puberty [79, 114]. In 1979, Hecker and Holschneider presented a manometric and functional classification that evaluated external sphincter contraction, propulsive wave frequency and "critical volume," and noted that "an intelligent patient can effect social continence" [54]. The need for a standardized objective testing method to evaluate outcomes was apparent. Mucosal prolapse and stricture of the anoplasty site remained a problem and a variety of secondary plastic procedures were suggested to revise the anoplasty by Mollard and Rowe [111], Nixon [113], Becmeur et al. [9], and Anderson et al. [5]. Incontinence also continued to be a problem, and free muscle transfer, reverse smooth muscle plasty, gracilis muscle flaps, and artificial sphincters were used in an attempt to correct this problem, with marginal results [34, 64, 66, 67, 149]. Reoperation for a missed puborectalis sling and failed previous procedures was also attempted [81, 87, 127, 149].

1.2.3 The Modern Era: 1980–2005

The 1980s ushered in another procedure, the posterior sagittal anorectoplasty (PSARP), resurrecting the concept initially proposed by Roux [141] in 1834 and subsequently used by Bodenhamer in 1860

[12]. The PSARP operation described by Peter deVries and Alberto Peña and published in 1982 was a new landmark event in the history of ARM, just as influential as the contributions by Amussat and Stephens and was rapidly adopted by many pediatric surgeons throughout the world [4, 33, 153, 154]. They redefined the arrangement of the pelvic muscles and sphincters as a fused sphincter muscle complex. The higher rectal defects were noted to have a wider separation between the subcutaneous and superficial external anal sphincter and the latter muscle from the levators. DeVries and Peña completely divided all the muscles posteriorly in the midline from the anal dimple to the coccyx. They divided the rectourethral fistula from within the atretic segment by separating the mucosa and smooth muscle to avoid urethral and neural damage. The distal atretic segment was tapered to fit within the puborectalis and the divided muscles were sutured posteriorly around and to the neorectum prior to performing the anoplasty. Slight tension was placed on the anoplasty site to draw the skin in and avoid prolapse [120]. The popularity of the PSARP procedure was a reflection of the discontent expressed by many pediatric surgeons with the results obtained after other procedures in the management of intermediate and high ARM. While the procedure allowed a precise anatomic reconstruction, it was not a panacea [55, 148]. In inexperienced hands, urologic injuries were observed during attempted repair of anorectal malformations [68]. Although the rate of continence improved (70% in Peña's hands; Fig. 1.4), it became apparent that many of the children had significant motility disorders, and fecal retention was a major problem [121, 122, 126]. Heightened awareness of this problem led to the establishment of close follow-up programs to assure patient and parent compliance with postoperative anal dilatation and appropriate rectal washouts using enemas [125, 126]. With the advent of the new procedure and continued controversy, an additional conference was held in 1984 at the Wingspread Conference Center in Wisconsin to reevaluate ARM with regard to classification, embryology, anatomy, and treatment. The Wingspread classification simplified the male/female low, intermediate, and high variants decided upon in 1970, transferred many female cloacal variants to separate subgroups and characterized the more uncommon anomalies as rare conditions [34]. Bladder and cloacal exstrophy and the various neuropathies were excluded. The presentations at the Wingspread Conference were edited by Douglas Stephens and E. Durham Smith and were published as a Birth Defects monograph by the March of Dimes in 1988 [158, 159]. The 1980s and early 1990s were periods that were also characterized by children's surgeons paying much closer attention to recognition and repair of cloacal anomalies. Dr. Raffensperger [132] of Chicago, and especially Dr. Hendren of Boston were instrumental in developing methods of repair (Fig. 1.5) [55–62, 97]. Later, Peña (and his associates) employed the posterior sagittal approach to repair cloacal anomalies and developed considerable experience in the management of these patients [92, 93, 124, 128, 151]. Peña also championed the concept of urogenital advancement in the repair of high cloacal defects [124]. Updated information concerning the anatomy and embryology of cloaca was published in a 1996 textbook edited by Stephens, Smith, and Hutson [160].

In 1992, Malone and associates described the antegrade colonic enema concept using an appendicostomy on the anterior abdominal wall or hidden within the umbilicus to flush the colon (MACE procedure) as an alternative to traditional retrograde enema washouts from below in children with incontinence or significant fecal retention [99]. Similarly, subsequent reports by Ellsworth et al. [40], Wilcox and Kiely [172], Squire et al. [152], and Levitt et al. [91] found the procedure useful. The Chait percutaneous cecostomy tube [21, 90] and Gauderer sigmoid button [47] were suggested as alternatives. Brent and Stephens [15] had identified the problem of rectal ectasia in 1976 and this was reaffirmed by Cloutier et al. [25], Cheu and Grosfeld [23] and, more recently, Peña and associates [126]. Of interest is that rectal ectasia occurs more commonly in children with low anomalies and may result from longstanding distension of the rectal segment in utero. Some of these patients (including those with megasigmoid) responded favorably to resection of the abnormal bowel [23, 126]. Despite the apparent popularity of the PSARP operation, in the 1990s several articles appeared in the literature recommending anterior sagittal anorectoplasty, anterior perineal anorectoplasty, and other modifications of the Mollard approach [2, 20, 37, 115].

In the 1990s, advances in technology resulted in new methods of assessment of patients with ARM and their associated anomalies. Transperineal ultrasonography was used to locate the infracoccygeal level of atresia and identify fistulae [52, 82]. Magnetic resonance imaging proved useful in evaluating the pelvic and perineal muscle status and identifying instances of tethered cord, vertebral anomalies, and spinal dysraphic syndromes [96, 137, 166]. Gross and Peña emphasized the value of a pressure-augmented distal colostogram to define the presence and location

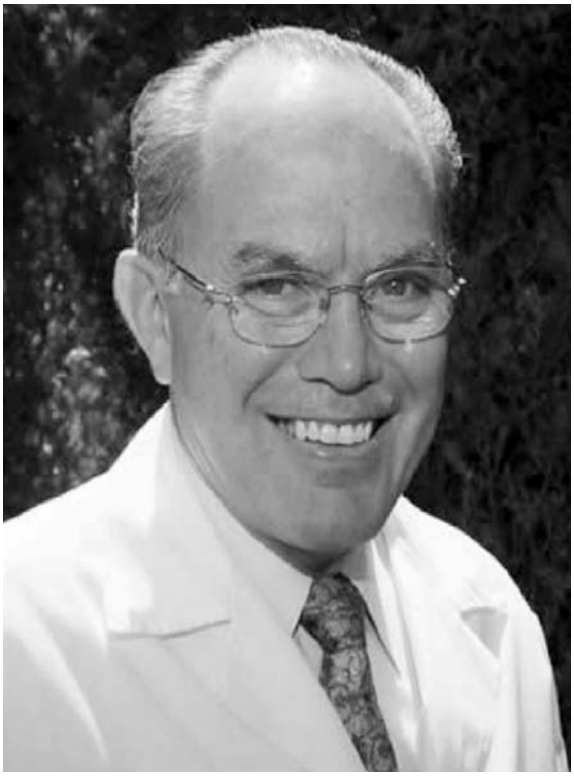

Fig. 4 Alberto Peña – advocated the posterior sagittal anorectoplasty (PSARP) procedure for ARM

Fig. 5 W. Hardy Hendren – was a leader in the management of cloacal anomalies

of the fistula in patients who required an initial colostomy in the neonatal period prior to definitive repair [50]. Postoperative assessment of sphincter muscle function and the position of the pulled-through segment within the sphincter complex is aided by anal endosonography [73, 162]. The advent of minimally invasive surgical techniques also influenced surgical treatment. Georgeson [48] and others [164] employed a laparoscope-assisted one-stage AP pull-through procedure that provided excellent exposure deep in the pelvis, divided the fistula using a harmonic scalpel or endoscopic clips, identified the sphincter complex and puborectalis muscle, and passed the colon through an intact sphincter to the anal dimple where an anoplasty was constructed in the neonatal period. Laparotomy and transsacral incisions were avoided and postoperative pain was diminished. Iwanaka et al. [71] and Yamataka et al. [175] used a laparoscopic muscle stimulator to accurately locate the sphincter during laparoscope-assisted repair. Adeniran supported a one-stage repair of imperforate anus and rectovestibular fistula in girls [2]. Albanese and colleagues advocated a one-stage correction of high ARM in the male neonate as a method of preserving the neural framework for normal bladder and bowel function that exist at the time of birth [3]. The learning period when neuronal circuitry development takes place resulting in normal or near-normal anorectal function occurs early and delayed repair may lose critical time when neural networks and impulses form [3, 45]. The one-stage operation avoids a colostomy and its potential complications, including urinary tract colonization and infection. Results are not yet available to determine whether these modifications will remain enduring and result in favorable long-term outcomes.

Animal models became available to study the embryology and anatomic features of ARM, including cloacal anomalies in pigs [69], mice [84, 85, 95], rats [95, 129], and chick embryos [100]. In mice, defects in sonic hedgehog signaling and mutations in the Gli 2 and 3 genes have been identified that hinder hindgut development [72, 83, 84, 109]. Careful three-dimensional reconstructive studies of embryos from the Carnegie collection by Paidas [116] and Rogers et al. [139] have led to further observations concerning the human embryology of ARM and cloaca, updating de Vries and Friedland's previous contributions [32].

1.3 ARM in the Twenty-First Century

Over the years, a great deal of information has been acquired concerning the many variants of ARM. The current mortality for these anomalies has for the most part been very low and is related to associated anomalies and unfavorable chromosomal and genetic syndromes rather than the result of treatment of the ARM per se. In general, agreement has been reached regarding some embryologic and anatomic considerations, diagnostic evaluation, and preoperative assessment [123, 146, 150]. Most investigators would agree that results are better following repair of low defects when compared to intermediate and high anomalies and those with cloaca. There remain several areas of controversy regarding the choice and timing of the procedure and methodology used to assess results. Certain poor prognostic factors have been identified, including abnormal sacrum, deficient pelvic innervation, poor perineal musculature, and disorders of colonic motility. It may be difficult to establish a prospective randomized study to evaluate some of the differences that exist among surgeons, although a study comparing open and minimally invasive techniques is appealing. Developing an agreeable classification that is stratified for adverse factors (e.g., sacral anomalies, muscle deficiency, pelvic innervation, associated anomalies, motility disorders) may permit surgeons to then assess outcomes more accurately using measures that are more objective than subjective. Designing a protocol that will define the precise location of the pulled-through rectal segment, and pelvic and sphincter muscle assessment using magnetic resonance imaging and/or anal endosonography (neither subject the child to radiation exposure), anal manometrics, functional defecography, and colonic motility studies in addition to traditional evaluation of soiling, sensation, and other more subjective analyses used in the past seems reasonable. For example, recent data suggest that after a properly performed operation, continence is achievable in a subset of patients with high defects that have an internal sphincter and high anorectal resting pressure [135]. I am optimistic that the 2005 conference on ARM in Cologne will address these issues.

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References

1. Academie de Medicine (1984) De l'imperforation de l'anus chez les neuveau-nes: rapport et discussion sur l'operation a tender dans ces cas. Gaz Med Paris 1834:11–112
2. Adeniran JO (2002) One-stage correction of imperforate anus and rectovestibular fistula in girls: preliminary results. J Pediatr Surg 37:16–19
3. Albanese CT, et al (1999) One-stage correction of high imperforate anus in the male neonate. J Pediatr Surg 34:834–836
4. Amussat JZ (1835) Histoire d'une operation d'anus practique avec success par un nouveau procede. Gaz Med Paris 3:753–758
5. Anderson KD, et al (1994) Diamond flap anoplasty in infants and children with an intractable anal stricture. J Pediatr Surg 29:1253–1257
6. Aristotle (1953) Generation of Animals, Peck AL (trans.) Harvard University Press, Cambridge, MA book IV, Chapter IV, pp 444–447
7. Ashcraft KW, Holder TM (1965) Congenital anal stenosis with presacral teratoma: case reports. Ann Surg 162:1091–1095
8. Ashraf KM, et al (1989) Pouch colon syndrome. Indian Pediatr 26:81–2
9. Becmeur F, et al (2001) Three-flap anoplasty for imperforate anus: results for primary procedure or for redoes. Eur J Pediatr Surg 11:311–314
10. Bell B (1787) A System of Surgery (3rd edn). Bell and Bradfute, Edinburgh, pp 275–282
11. Berman JK (1938) Congenital anomalies of the rectum and anus. Surg Gynecol Obstet 66:11
12. Bodenhamer W (1860) Aetiology, Pathology and Treatment of the Congenital Malformations of the Rectum and Anus. Samuel and William Wood, New York
13. Bodenhamer W (1889) Some facts and observations relative to the congenital malformations of the rectum and anus, and to the operation of colostomy in some such cases. NY Med J 49:562
14. Brenner EC (1915) Congenital defects of the anus and rectum. Surg Gynecol Obstet 20:59

15. Brent L, Stephens FD (1976) Primary rectal ectasia: a quantitative study of smooth muscle cells in normal and hypertrophied human bowel. Prog Pediatr Surg 9:41–62
16. Browne D (1951) Some congenital deformities of the rectum, anus, vagina and urethra. Ann R Coll Surg Engl 8:173–192
17. Budhiraja S, et al (1997) A report of 27 cases of congenital short colon with an imperforate anus: so-called 'pouch colon syndrome'. Trop Doct 27:217–220
18. Byrd WA (1882) Verneuil's modification of Amussat's operation for the relief of imperforate anus. Med Surg Reporter X:649
19. Carson JA, et al (1984) Imperforate anus the neurologic implications of sacral abnormalities. J Pediatr Surg 19:838–842
20. Chainani M (1998) The anterior sagittal approach for high imperforate anus: a simplification of the Mollard approach. J Pediatr Surg 33:670–671
21. Chait PG, et al (1997) Fecal incontinence in children: treatment with percutaneous cecostomy tube placement – a prospective study. Radiology 203:621–624
22. Chassignac (1856) Proceedings of the Société de Chirurgie de Paris. Bull Mem Soc Chir February 20
23. Cheu H, Grosfeld JL (1992)The atonic baggy rectum: a cause of intractable obstipation after imperforate anus repair. J Pediatr Surg 27:1071–1073
24. Clark SA, et al (1999) Imperforate anus, Hirschsprung's disease, and trisomy 21: a rare combination. J Pediatr Surg 34:1874
25. Cloutier R, et al (1987) Focal ectasia of the terminal bowel accompanying low anal deformities. J Pediatr Surg 22:758–760
26. Cooke J (1685) Mellificium Chirurgiae: or the Marrow of Chirurgery Much Enlarged. Marshall, London p 669 (and 4th edn p 158)
27. Cremin BJ, et al (1970) The radiological assessment of anorectal malformation. In: Proceedings of the Pediatric Surgical Congress, Melbourne, Australia, March 20
28. Cule JH (1965) John Pugh 1814–1874. A scholar surgeon's operation on the imperforate anus in 1854. Ann R Coll Surg Engl 37:247
29. Currarino G, et al (1981) Triad of anorectal, sacral, and presacral anomalies. AJR Am J Roentgenol 137:395–398
30. Davidoff AM, et al (1991) Occult spinal dysraphism in patients with anal agenesis. J Pediatr Surg 26:1001–1005
31. Delens M (1875) Observations: II. Imperforation anale – enterotomie perineale avec resection du coccyx – Guerison avec prolapses rectal. Bull M Soc Chir 1:217
32. de Vries PA, Friedland GW (1974) The staged sequential development of the anus and rectum in human embryos and fetuses. J Pediatr Surg 9:755–769
33. de Vries PA, Peña A (1982) Posterior sagittal anorectoplasty. J Pediatr Surg 17:638–643
34. de Vries PA (1984) The surgery of anorectal anomalies: its evolution, with evaluation of procedures. Curr Probl Surg 21:1–75
35. de Vries PA (1988) Historical update. In: Stephens FD, Smith ED (eds) ARM in Children; Update 1988. Birth Defects Orig Artic Ser 24:11–15
36. Dieffenbach J (1845) Operative Chirurgie. Leipzig vol 1. p 670
37. Doria do Amaral F (1999) Treatment of anorectal anomalies by anterior perineal anorectoplasty. J Pediatr Surg 34:1315–1319
38. Dunglison R (1846) Human physiology. Lea and Blanchard, Philadelphia, p 497
39. Duret C (1798) Observation sur enfant un ne sans anus, et auguel il a ete fait une ouverture pour y suppléer. Receuil de la Societé de Médicine de Paris, 4:45–50
40. Ellsworth PI, et al (1996) The Malone antegrade colonic enema enhances the quality of life in children undergoing urological incontinence procedures. J Urol 155:1416–1418
41. Engum SA, Grosfeld JL (2005) Anorectal anomalies. In: Yao CD (ed) Shackelford's Surgery of the Alimentary Tract, 6th edn. Elsevier, Philadelphia
42. Fitchet SM (1926) Imperforate anus. Boston Med Surg J 195:25
43. Flageole H (1996) Hirschsprung's disease, imperforate anus, and Down's syndrome: a case report. J Pediatr Surg 31:759–760
44. Fowler R (1972) A reappraisal of surgical approaches for the definitive correction of imperforate anus. Aust NZ J Surg 42:56–61
45. Freeman NV, Burge DM (1980) Anal evoked potentials. Z Kinderchir 31:22
46. Galen C (1576) De musculis sedis. In: Isogogici Libra, 5th edn. Venice Giunta, Chapter 50, p 50
47. Gauderer MW, et al (2002) Sigmoid irrigation tube for the management of chronic evacuation disorders. J Pediatr Surg 37:348–351
48. Georgeson KE, et al (2000) Laparoscopic assisted anorectal pull-through for high imperforate anus – a new technique. J Pediatr Surg 35:927–931
49. Gowers WR (1877) The automatic action of the sphincter ani. Proc R Soc Lond 26:77
50. Gross GW, et al (1991) Augmented-pressure colostogram in imperforate anus with fistula. Pediatr Radiol 21:560–562
51. Haller JO, et al (2004) Tracheoesophageal fistula (H-type) in neonates with imperforate anus and the VATER association. Pediatr Radiol 34:83–85
52. Han TI, et al (2003) Imperforate anus: US determination of the type with infracoccygeal approach. Radiology 228:226–229

53. Hayward G (1853) History of a case of anal malformation successfully treated by an operation. Va Med Surg J 1:268–270
54. Hecker WCh, Holschneider AM (1979) Complications, lethality and long term results after surgery of anorectal defects. In: Kiesewetter WB (ed) Long Term Followup in Congenital Anomalies. Pediatric Surgical Symposium, Pittsburgh
55. Hedlund H, et al (1992) Long-term anorectal function in imperforate anus treated by a posterior sagittal anorectoplasty: manometric investigation. J Pediatr Surg 27:906–909
56. Hendren WH (1980) Urogenital sinus and ARM: experience with 22 cases. J Pediatr Surg 15:628–641
57. Hendren WH (1982) Further experience in reconstructive surgery for cloacal anomalies. J Pediatr Surg 17:695–717
58. Hendren WH (1986) Repair of cloacal anomalies :current techniques. J Pediatr Surg 21:1159–1176
59. Hendren WH, et al (1987) Repair of cloacal malformation using combined posterior sagittal and abdominoperineal approaches. Z Kinderchir 42:115–119
60. Hendren WH (1992) Cloacal malformations: experience with 105 cases. J Pediatr Surg 27:890–901
61. Hendren WH (1996) Urogenital sinus and cloacal malformations. Semin Pediatr Surg 5:72–79
62. Hendren WH (1998) Cloaca, the most severe degree of imperforate anus: experience with 195 cases. Ann Surg 228:331–346
63. Hochenegg J (1889) Beitrage zur chirurgie des rectum und der Beckenorgane. Wien Klin Wschr 2:55
64. Hoffman V Kapp-her S, Koltai I (1981) New methods in the treatment of anorectal incontinence. Z Kinderchir 32:258
65. Holl M (1897) Harn-und Geschlechtsorgane: Die Muskeln und Fascien des Beckenausganges, in Bardeleben: Handbuch der Anatomie des Menschen. Gustav Fischer, Jena, pp 161–300
66. Holschneider AM, Walemar WC (1981) Reverse smooth muscle plasty: a new method of treating anorectal incontinence in infants with high anal and rectal atresia. J Pediatr Surg 16:917–920
67. Holschneider AM, Hecker WC (1981) Flapped and free muscle transplantation in the treatment of anal incontinence. Z Kinderchir. 32:244–58
68. Hong AR, et al (2002) Urologic injuries associated with repair of ARM in male patients. J Pediatr Surg 37:339–344
69. Hori T, et al (2001) Mapping loci causing susceptibility to anal atresia in pigs, using a resource pedigree. J Pediatr Surg 36:1370–1374
70. Ilberg J (1927) Sorani gyneciorium. In: Ilberg J (ed) Corpus Medicorum Graecorum. Teubner, Lipsial et Bertolini, 1927, libri IV ff
71. Iwanaka T, et al (2003) Findings of pelvic musculature and efficacy of laparoscopic muscle stimulator in laparoscopy-assisted anorectal pull-through for high imperforate Surg Endosc 17:278–281
72. Jo Mauch T, Albertine KH (2002) Urorectal septum malformations sequence: insights into pathogenesis. Anat Rec 268:405–410
73. Jones NM, et al (2003) The value of anal endosonography compared with magnetic resonance imaging following the repair of ARM. Pediatr Radiol 33:183–185
74. Kallen KE, et al (2001) VATER non-random association of congenital malformations: study based on data from four malformation registers. Am J Med Genet 101:26–32
75. Keen WW, DaCosta JC (1908) Surgery: Its principles and Practice. WB Saunders, Philadelphia, pp 120–122
76. Kelly JH (1969) Cineradiography in ARM. J Pediatr Surg 4:538–546
77. Kelly JH (1972) The clinical and radiological assessment of anal continence in childhood. Aust NZ J Surg 42:62–63
78. Khoury MJ, et al (1983) A population study of the VACTERL association. Pediatrics 71:815–820
79. Kiesewetter WB, Turner CR (1963) Continence after surgery for imperforate anus: a critical analysis and preliminary experience with the sacro-perineal pull-through. Ann Surg 158:498–512
80. Kiesewetter WB, Nixon HH (1967) Imperforate anus. I. Its surgical anatomy. J Pediatr Surg 2:60
81. Kiesewetter WB, Jefferies MR (1981) Secondary anorectal surgery for the missed puborectalis muscle. J Pediatr Surg 16:921–927
82. Kim IO, et al (2000) Transperineal ultrasonography in imperforate anus: identification of the internal fistula. J Ultrasound Med 19:211–216
83. Kim J, et al (2001) The VACTERL association: lessons from the Sonic Hedgehog pathway. Clin Genet 59:306–315
84. Kimmel SG, et al (2000) New mouse models of congenital ARM. J Pediatr Surg 35:227–230
85. Kluth D, Lambrecht W (1997) Current concepts in the embryology of ARM. Semin Pediatr Surg 6:180–186
86. Kochling J, et al (1996) The Currarino syndrome – hereditary transmitted syndrome of anorectal, sacral and presacral anomalies. Case report and review of the literature. Eur J Pediatr Surg 6:114–119
87. Kottmeier PK (1966) A physiological approach to the problem of anal incontinence through the use of the levator ani as a sling. Surgery 60:1262–1266
88. Ladd WE, Gross RE (1934) Congenital malformations of the anus and rectum. Am J Surg 23:167
89. Lee SC, et al (1997) Currarino triad: anorectal malformation, sacral bony abnormality and presacral mass – a review of 11 cases. J Pediatr Surg 32:58–61
90. Lee SL, et al (2002) Therapeutic cecostomy tubes in infants with imperforate anus and caudal agenesis. J Pediatr Surg 37:345–347

91. Levitt MA, et al (1997) Continent appedicostomy in the bowel management of fecally incontinent children. J Pediatr Surg 32:1630–1633
92. Levitt MA, et al (1998) Gynecologic concerns in the treatment of teenagers with cloaca. J Pediatr Surg 33:194–197
93. Levitt MA, Peña A (2005) Pitfalls in the management of newborn cloacas. Pediatr Surg Int 21:264–269
94. Littre A (1710) Diverses observations anatomatiques. Paris Histoire de l'Academie Royale de Science, pp 36–37
95. Liu MI, Hutson JH (2000) Cloacal and urogenital malformations in adriamycin-exposed rat fetuses. BJU Int 86:107–112
96. Long FL, et al (1996) Tethered cord and associated vertebral anomalies in children and infants with imperforate anus: evaluation with MR imaging and plain radiography. Radiology 200:377–382
97. Lund DP, Hendren WH (2001) Cloacal exstrophy: a 25-year experience with 50 cases. J Pediatr Surg 36:68–75
98. Mahboubi S, Templeton JH Jr (1984) Association of Hirschsprung's disease and imperforate anus in a patient with „cat-eye" syndrome. A report of one case and review of the literature. Pediatr Radiol 14:441–442
99. Malone PS, et al (1990) Preliminary report: the antegrade continence enema. Lancet 336:1217–1218
100. Manner J, Kluth D (2003) A chicken model to study the embryology of cloacal exstrophy. J Pediatr Surg 38:678–681
101. Mantell T (1792) Case of imperforate anus successfully treated. Med Soc London Mem 3:389
102. Martin of Lyon (1798) Recueil des Actes, Societe de Sante de Lyon, p 180
103. Martinez-Frias ML, et al (2000) Anal atresia, vertebral, genital, and urinary tract anomalies: a primary polytopic developmental field defect identified through an epidemiological analysis of associations. Am J Med Genet 95:169–173
104. Martucciello G, et al (2004) Currarino syndrome: proposal of a diagnostic and therapeutic protocol. J Pediatr Surg 39:1305–1311
105. Mastin WM (1908) A resume of the surgical treatment of anorectal imperfection in the newborn. Surg Gynecol Obstet 7:316
106. Matas R (1897) The surgical treatment of anorectal imperforation considered in the light of modern operative procedures. Trans Am Surg Assoc 15:453
107. McLeod N (1880) Case of imperforate rectum with a suggestion for a new method of treatment. Br Med J 2:653
108. McCormac W (1886) On a case of impeforate anus. Lancet 2:12
109. Mo R, et al (2001) ARM caused by defects in sonic hedgehog signaling. Am J Pathol 159:765–774
110. Mollard P, et al (1978) Surgical treatment of high imperforate anus with definition of the puborectalis sling by an anterior perineal approach. J Pediatr Surg 13:499–504
111. Mollard DR, Rowe MI (1982) Plastic principles in high imperforate anus. Plast Reconstr Surg 69:399
112. Narasimharao KI, et al (1983) Prone cross-table view. An alternative to the invertogram in imperforate anus. AJR 140:227–229
113. Nixon HH (1967) A modification of the proctoplasty for rectal agenesis. Pamietnik I-Go Zjazdu 10:5
114. Nixon HH, Puri P (1977) The results of treatment of anorectal anomalies: a thirteen to twenty year follow-up J Pediatr Surg 12:27–37
115. Okada A, et al (1992) Anterior sagittal anorectoplasty for rectovestibular and anovestitibualr fistula. J Pediatr Surg 27:85–88
116. Paidas CN, et al (1999) Septation and differentiation of the embryonic human cloaca. J Pediatr Surg 34:877–884
117. Parrott TS, Woodard JR (1979) Importance of cystourethrography in neonates with imperforate anus Urology 13:607–609
118. Parrott TS (1985) Urologic implications of ARM. Urol Clin North Am 12:13–21
119. Paualus Aegineta (1844) On the imperforate anus. In: Adams F (ed) The Seven Books. The Syndenham Society book VI Section LXXXI, London, pp 405–406
120. Peña A, de Vries PA (1982) Posterior sagittal anorectoplasty: important technical considerations and new applications. J Pediatr Surg 17:796–811
121. Peña A (1983) Posterior sagittal anorectoplasty: as a secondary operation for the treatment of fecal incontinence. J Pediatr Surg 18:762–73
122. Peña A, el Behery M (1993) Megasigmoid: a source of pseudoincontinence in children with repaired ARM. J Pediatr Surg 28:199–203
123. Peña A (1995) ARM. Semin Pediatr Surg 4:35–47
124. Peña A (1997) Total urogenital mobilization – an easier way to repair cloacas. J Pediatr Surg 32:263–267
125. Peña A, et al (1998) Bowel management for fecal incontinence in patients with ARM. J Pediatr Surg 33:133–137
126. Peña A, Levitt MA (2002) Colonic inertia disorders in pediatrics. Curr Probl Surg 39:666–730
127. Peña A, et al (2003) Reoperative surgery for anorectal anomalies. Semin Pediatr Surg 12:118–123
128. Peña A, et al (2004) Surgical management of cloacal malformations: a review of 339 patients. J Pediatr Surg 39:470–479
129. Qi BQ, et al (2002) Clarification of the processes that lead to ARM in the ETU – induced rat model of imperforate anus. J Pediatr Surg 37:1305–1312
130. Quinland WS (1922) Congenital malformation of the intestine – atresia and imperforate anus. Boston Med Surg J 187:870
131. Quan L, Smith DW (1978) The VATER association. Vertebral defects, anal atresia, T–E fistula with esophageal atresia, radial and renal dysplasia: a spectrum of associated defects. J Pediatr 82:104–107

132. Raffensperger JG, Ramenofsky ML (1980) The management of a cloaca. J Pediatr Surg 8:647
133. Rehbein F (1959) Operations der anal – und Rectumatresie mit Recto-Urethralfistal. Chirurgie 30:417–418
134. Rhoads JE, et al (1948) A simultaneous abdominal and perineal approach in operations for imperforate anus with atresia of the rectum and rectosigmoid. Ann Surg 127:552
135. Rintala RJ, Lindahl H (1995) Is normal bowel function possible after repair of intermediate and high ARM. J Pediatr Surg 30:491–494
136. Rizzoli F (1869) A new operative method in four cases of congenital atresia ani, with opening of the rectum into the vulva. Surg Mem 2:321
137. Rivossechi M, et al (1995) Spinal dysrahism detected by magnetic resonance imaging in patients with anorectal anomalies: incidence and clinical significance. J Pediatr Surg 32:462–468
138. Robin C, Cadiat (1874) Sur la structure et les rapports des teguments au niveau de leur junction dans le région anale, vulvaire et du col utérin. J Anat Physiol pp 589–605
139. Rogers DS, et al (2002) Septation of the anorectal and genitourinary tracts in the human embryo: crucial role of the catenoidal shape of the urorectal sulcus. Teratology 66:144–152
140. Romualdi P (1960) A new technique for surgical treatment of some rectal malformations. Langenbecks Arch Klin Chir Ver Dtsch Z Chir 296:371–377
141. Roux J–N (1835) Observation d'imperforation de l'anus et de l'uretre. Memoires de l'Academie Royale de Medicine 4:183–190
142. Santorini JD (1794) Observationes Anatomicae. Baptistam Recurti
143. Saviard (1740) Observations in Surgery. Surgeon FS. London, J. Hodges
144. Scharli AF (1978) Malformations of the anus and rectum and their treatment in medical history. Prog Pediatr Surg 11:141–172
145. Shafik A (1975) A new concept of the anal sphincter mechanism and the physiology of defecation: I The external and anal sphincter – a triple loop system. Invest Urol 12:412–419
146. Shaul DB, Harrison EA (1997) Classification of ARM: initial approach, diagnostic tests and colostomy. Semin Pediatr Surg 6:187–195
147. Smith ED (1968) Urinary anomalies and complications in imperforate anus and rectum. J Pediatr Surg 3:337–349
148. Smith ED (1988) The bath water needs changing, but don't throw away the baby: an overview of anorectal anomalies. J Pediatr Surg 22:335–348
149. Smith ED, et al (1988) Operations to improve continence after previous surgery. Birth Defects Orig Artic Ser 24:447–479
150. Smith EI, Gross RE (1961) The external anal sphincter in cases of imperforate anus: a pathologic study. Surgery 49:807
151. Soffer SZ, et al (2000) Cloacal exstrophy: a unified management plan. J Pediatr Surg 35:932–937
152. Squire R, et al (1993) The clinical application of the Malone antegrade colonic enema. J Pediatr Surg 28:1012–1015
153. Stephens FD (1953) Imperforate rectum; a new surgical technique. Med J Aust 7:202–203
154. Stephens FD (1953) Congenital imperforate rectum, recto-urethral and recto-vaginal fistulae. Aust NZ J Surg 22:161–172
155. Stephens FD (1961) Congenital malformations of the rectum and anus in female children. Aust NZ J Surg 31:90–104
156. Stephens FD (1963) Congenital Malformations of the Rectum, Anus and Genitourinary Tracts. E and S Livingston, Edinburgh and London
157. Stephens FD, Smith ED (1971) ARM in Children. Year Book Medical Publishers, Chicago
158. Stephens FD, Smith ED (1986) Classification and assessment of surgical treatment of anorectal anomalies. Pediatr Surg Int 1:200
159. Stephens FD, Smith ED (1988): ARM in children Update 1988. March of Dimes Birth Defects Foundation Series 24:(4); Alan R Liss, New York
160. Stephens FD, Smith ED, Hutson JM (1996) Congenital Anomalies of the Urinary and Genital Tracts. Isis Medical Media, Oxford
161. Stone HB (1936) Imperforate anus with rectovaginal cloaca. Ann Surg 104:651
162. Stuhldreier G, et al (1997) Endosonographic determination of the volume of anal sphincter muscles as a parameter for continence outcome in childhood. Langenbecks Arch Chir Suppl Kongressbd 114:1330–1332
163. Swenson O, Donnellan WL (1967) Preservation of the puborectalis sling in imperforate anus repair. Surg Clin North Am 47:173–193
164. Sydorak RM, Albanese CT (2002) Laparoscopic repair of high imperforate anus. Semin Pediatr Surg 11:217–225
165. Torres R, et al (1998) ARM and Down syndrome. J Pediatr Surg 33:194–197
166. Tsakayannis DE, Shamberger RC (1995) Association of imperforate anus with occult spinal dysraphism. J Pediatr Surg 30:1010–1012
167. Verneuil M (1873) De la resection du coccyx pour faciliter la formation d'un anus perieale dans l'imperforation du rectum. Bull Soc Chir Paris 2:288
168. Vesalius A (1543) De Humani Corporis Fabrica. Johanes Operinus, Basle
169. Wangensteen OH, Rice CO (1930) Imperforate anus. A method of determining the surgical approach. Ann Surg 92:77

170. Webster R (1971) Historical review. In: Stephens FD, Smith ED (eds) ARM in children. Year Book Publishers, Chicago, pp 11–13
171. Wiener ES, Kiesewetter WB (1973) Urologic abnormalities associated with imperforate anus. J Pediatr Surg 8:151–157
172. Wilcox DT, Kiely EM (1998) The Malone (antegrade colonic enema) procedure: early experience. J Pediatr Surg 33:204–206
173. Wood-Jones F (1904) The nature of malformations of the rectum and urogenital passages. Br Med J 2:1630
174. Wood-Jones F (1915) The explanation of a rectourethral anomaly and some points in normal anatomy. Lancet 2:860
175. Yamataka A, et al (2001) Laparoscopic muscle electrostimulation during laparoscopy-assisted anorectal pull-through for high imperforate anus. J Pediatr Surg 36:1659–1661

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Giuseppe Martucciello

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2.1 Introduction

Anorectal malformations (ARM) present an incidence rate ranging from 1:1,500 to 1:5,000 live births [18, 19, 41, 49, 52, 53, 61, 109, 110, 112], and have variable clinical presentations ranging from mild forms that might require only minor surgical interventions to more complicated cases that need to be managed with multi-staged operations [51, 87, 88, 108].

The cause of ARM is unknown, although arrest of the descent of the urorectal septum towards the cloacal membrane between the 4th and 8th weeks of gestation was previously considered the basic event leading to ARM [31, 32, 55, 71]. Since the molecular determinants during blastogenesis are overlapping for many body systems, and these elements are closely related in timing and spacing, so defects in blastogenesis often involve two or more progenitor fields. This fact may explain the cause of subgroups of ARM that form part of complex phenotypes due to developmental field defects. These complex phenotypes are considered as end results of pleiotropic effects of single causal events that might be chromosomal, monogenic or even teratogenic [72].

Classifying ARM from the genetic point of view is not easy since they present different forms that are believed to be influenced by different factors such as sex and associated anomalies. ARM can be the only pathological finding (non-syndromic) or as part of a more complex phenotype (syndromic), and may occur in a single affected individual (sporadic) or in more than one individual in the same family (familial) with different modes of inheritance. There are gender differences, with remarkably higher preponderance in boys for more complex ARM forms, while the less severe types, with perineal or vestibular fistulae, reported more frequently in girls (Fig. 2.1) [61]. These topics will be discussed in detail below.

2.2 Non-syndromic ARM

Sporadic ARM is a frequent clinical finding; however, different modes of inheritance have been reported. Eleven families have been reported with inherited variants, 7 of these families showing autosomal dominant mode of inheritance. In one family the condition was suggestive of autosomal recessive inheritance, with two affected females to healthy-looking parents. In the remaining three families, it was not possible to differentiate whether the inheritance was autosomal or X-linked recessive (Table 2.1).

Moreover, excluding the isolated autosomal dominant low type, ARM can escape diagnosis easily due to variable expression, such as presenting with a very mild manifestation such as an anteriorly located anus (perineal fistula) [60]. Careful examination of the first-degree relatives, particularly the parents, is highly recommended before calculating the recurrence risk in isolated low ARM due to variable expression [96].

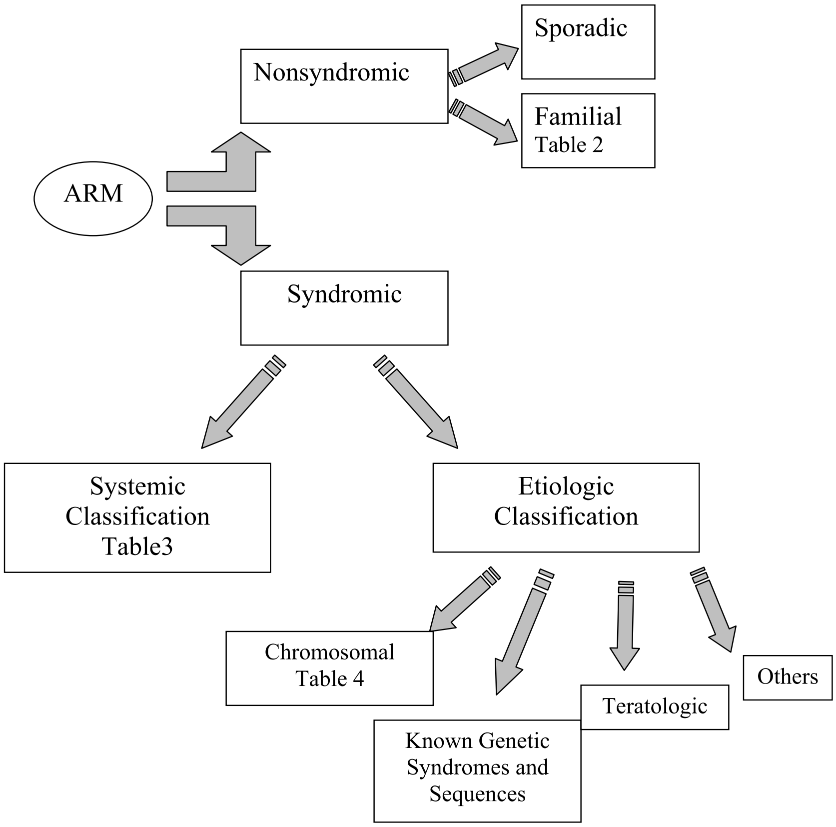

Fig. 2.1 Classification of anorectal malformations (ARM)

Table 2.1 Suggested mode of inheritance for isolated anorectal malformations (ARM)

Reports	Affected members	Suggested inheritance
Manny et al. (1973) [68]	Father-son	Autosomal-dominant
Schwoebel et al. (1984) [106]	Mother, son and daughter	Autosomal-dominant
Boocock and Donnai (1987) [12]	Three father-son pairs	Autosomal-dominant
Weinstein (1965) [124]	Three families with affected sons and healthy parents	Autosomal- or X-linked recessive
Winkler and Weinstein (1970) [126]	Two daughters, healthy parents	Autosomal-recessive
Landau et al. (1997) [60]	Four members in three generations	Autosomal-dominant
Robb and Teebi (1998) [100]	Father-son	Autosomal-dominant

2.3 ARM Associated with Other Systemic Malformations

There is a wide spectrum of possible associations of ARM with various systemic malformations (Table 2.2). These associations may be well-described syndromes with various aetiologies or may be just a random association with other malformations. In a recent review, Ratan et al. [94] studied 416 patients affected with ARM of whom 58% had additional malformations: 68% were male and 32% were female. High-type ARM was found in 58%, but occurred five times more frequently in males than in females, while low-type ARM occurred twice as often in females than in males. High-type ARM were associated 13 times more frequently with other congenital malformations than were low-type ARM. ARM with other anomalies were four times more common among males than females. These findings are in contrast with isolated ARM, for which there is no sex difference and, more surprisingly, the isolated low-type ARM is twice as common as the high type. Ratan et al. [94] also found that boys with high ARM had more vertebral and gastrointestinal tract anomalies, especially for congenital short megacolon, while the boys with low ARM had a higher incidence of genital malformations. On the other hand, girls with high ARM have more skeletal anomalies (other than vertebral) and urinary tract malformations. Girls affected with low ARM frequently suffered from vesicoureteric reflux. The percentages of patients affected by ARM and 2,3, or 4 other body system anomalies were 50%, 29%, and 16% respectively.

The analysis of associated malformations plays an important role in the genetic study of ARM, because diagnosing the associated anomalies can identify specific genes that may represent a starting point for more detailed investigations. Studies have thus focussed on the most frequently associated anomalies, namely the vertebral, genitourinary and gastrointestinal malformations [61].

A condition that is commonly associated with ARM is hypospadias (found in 6.5% of cases) [40]. Moreover, one out of five cases of congenital urethral fistula – a rare condition – is associated with ARM. In a study on rectal innervation in patients with ARM, enteric nervous system anomalies ranged from 79 to 100% of cases depending on the ARM type [61].

The combination of both ARM and intestinal dysganglionoses, particularly Hirschsprung's disease (HD), is another interesting field, since several causative genes for HD are known. The incidence of this combination varies widely from one study to another, ranging from 3.4% [54] to 60% [86]. Lerone et al. suggested that this variability depends on whether suitable techniques for diagnosing intestinal innervation were used or not [61]. Enzymohistochemical techniques studying cryostat sections from biopsy specimens of the fistula and rectal pouch demonstrated an abnormal innervation in 96% of ARM patients [61, 44]. Classical aganglionosis was found in 31% of rectal pouch specimens, hypoganglionosis in 38%, and intestinal neuronal dysplasia type B in 14%; unclassified dysganglionosis was found in 10%. These data demonstrate that ARM and dysganglionoses are not uncommon, and suggest the importance of mutation analysis of HD genes in ARM-affected individuals [61].

2.4 Aetiological Classification

The frequent association of ARM with common chromosomal anomalies is well known (e.g. Down syndrome, trisomy 18, 13q-, cat-eye syndrome (CEM), or genetic syndromes such as Currarino syndrome, FG syndrome, VATER association and others [14, 17, 22, 35, 39, 42, 48, 75, 80, 82, 85, 95, 98, 99, 111, 115, 123]. Teratogenic effects of certain agents such as thalidomide, oestrogen and ethanol intake has also been well described [8, 61, 89].

Table 2.2 Systemic malformations associated with ARM. *US* Ultrasound, *IVU* intravenous urethrogram, *MCU* micturating cystourethrogram, *MRI* magnetic resonance imaging, *ECG* electrocardiogram

Anomalies	Incidence in literature (%)	Investigations	Notes
Urologic	25–55%	US, IVU	Hoekstra, Ratan, Mittal
Vesicoureteric reflux	20–47%	US, MCU	Hassink, Narasimharao, Rickwood, Ratan
Genital	3–18% (boys) 26–39% (girls)		Hoekstra, Metts, Cortes
Vertebral	25–38%	X-ray, MRI	Hassink, Carson, Mittal, Ratan
Other skeletal	13–16%	X-ray	Hassink, Ratan, Mittal
Spinal-cord-related	8–67%	X-ray, MRI	Rivosecchi, Walton, Ratan, Mittal
Cardiac	10–17%	ECG, echocardiography	Mittal, Greenwood, Ratan
Gastrointestinal	7–10%	X-ray, biopsy	Ratan, Mittal, Hassink
Tracheoesophageal	6–8%		Hassink, Ratan
Others	4–5%		Ratan

2.4.1 Chromosomal Anomalies Associated with ARM

In a study of 1,846 babies with ARM [24], chromosomal anomalies were found in 11%, the most frequent form being perineal fistula. The frequencies of trisomy 21, 13, and 18 among babies with ARM were 15, 30, and 90 times higher, respectively, in comparison to neonates in the general population (Table 2.3).

2.4.2 Down Syndrome

The ARM occurs more frequently with Down syndrome than in the general population and there is a higher rate of deformities without fistula [11, 116]. In a study of 1,992 patients with ARM in Japan [30], Down syndrome was seen in 101 patients (5.1%), with no gender variation. The incidence of high, intermediate, and low types were 2.7%, 18.7% and 4.1%, respectively, showing clearly the statistical difference between the intermediate type and the others. Ninety-five percent (96 out of 101) of patients with Down syndrome had deformities without fistula, while only 3 patients had rectourethral fistula; the remaining 2 had a perineal fistula.

Table 2.3 Some forms of chromosomal anomalies associated with ARM (from Cuscheri et al. 2002) [24]

Chromosomal anomalies	Number of cases out of 1,846 ARM patients
Trisomy 8 mosaic	1
Trisomy 13	12
Trisomy18	20
Trisomy 21	39
Trisomy 22	3
Sex chromosome aneuploidy	4
Triploidy	1
Tetrasomy 12 p (Pallister-Killian)	2
Ring (13)	3
Deletion 5p	3
Extra fragment	3
Partial tri/monosomy	21
Other chromosome anomalies	17

2.4.3 Cat-Eye Syndrome

CES is characterised by ARM, coloboma of the iris (total or rarely partial, unilateral or bilateral), coloboma of the choroid and/or optic nerve, microphthalmia (usually unilateral) and variable external ear deformities ranging from unilateral auricular reduction defects to several tags, mostly with atresia of the external auditory canal. These anomalies can be associated with mental retardation in half of the cases; dysmorphic features are the leading signs for the diagnosis, including hypertelorism, downward-slanting palpebral fissures and low root of the nose. Congenital heart disease can also be associated with this syndrome, especially septal defects and anomalous pulmonary venous return, as can different forms of urinary tract malformations such as renal agenesis or hypoplasia, vesicoureteral reflux and bladder anomalies [7, 61, 105].

CES is usually associated with the cytogenetic finding of a supernumerary marker chromosome consisting of duplicated material of chromosome 22. Bisatellite and dicentric markers are usually found (idic(22)(pter→q11.2::q11.2→pter)) and thus results in tetrasomy of the p arm and a part of 22q11.2 [74, 105]. This chromosomal anomaly generally follows a de novo mutation, and the recurrence risk does not increase with subsequent pregnancies. However, in a few cases the anomaly may segregate from an affected parent. Mosaicism can be a frequent finding in blood samples of the affected index case and of his family members. This may draw attention to considerable intrafamilial variability of the phenotypic expression, and the potential recurrence risk for patients with a normal karyotype [105].

2.4.4 Genetic Syndromes Associated with ARM

Seven major syndromes have been considered in this chapter: Townes-Brocks syndrome (TBS), FG syndrome, Pallister-Hall syndrome (PHS), VACTERL (vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies) Association (VATER), sirenomelia, caudal regression syndrome (CRS) and Currarino syndrome.

2.4.4.1 Townes-Brocks Syndrome

Otherwise known as renal-ear-anal-radial syndrome or Townes-Brocks-branchio-oto-renal-like syndrome, this autosomal dominant syndrome was first described in 1972 [116]. It is estimated to have an incidence rate of 1:250,000 live births [70]. TBS has great variability among affected families [89], but in general the main characteristic features of TBS are: (1) ARM (imperforate anus and anal stenosis), (2) hand malformations in the form of preaxial polydactyly with diverse thumb anomalies ranging from vestigial, broad, to triphalangeal thumb, and even distal ulnar deviation of the thumb and (3) external ear malformation (microtia, external auditory atresia, satyr ear with overfolding of the superior helix, preauricular pits and sensorineural deafness). Other associated anomalies are congenital heart malformations, mostly tetralogy of Fallot, ventricular septal defect, truncus arteriosus and genitourinary anomalies (which can include dysplastic kidneys, vesicoureteric reflux and hypospadias); mental retardation has been noted in variable degrees [89]. The major criteria for the diagnosis of TBS are the hands, ears and anus. In the presence of only two major criteria the diagnosis becomes less secure and may be taken in consideration only if: (1) they are accompanied by minor malformations such as cardiac and renal anomalies and deafness, (2) absence of atypical features (i.e. tracheoesophageal or vertebral anomalies), (3) presence of another affected individual in the family and (4) other affected persons in the family who have the missing major feature in the index case. In that case the clinical diagnosis can be confirmed by a mutational analysis of the causative gene *SALL1*. Failure to detect the mutation does not rule out the diagnosis since the detection rate is 64.3–83.3% of patients with "classical" TBS with hands, ears and anal malformations. *SALL1* gene is the only known gene causing TBS [56, 58, 69]. TBS exhibits similar features to other syndromes, namely Goldenhaar, VACTERL or oculo-auriculo-vertebral [89]. SALL1 was mapped first to 16q12.1 by fluorescence in situ hybridisation [57]. Up to 29 mutations were reported in affected individuals with scattered mutations all over the gene [56]. However, it is still unknown whether certain forms of mutations in other parts of *SALL1* would result in different phenotypes. Mutation analysis of *SALL1* has confirmed that penetrance is complete in TBS [56]. Prenatal diagnosis can be performed by searching for mutations of the *SALL1* gene in amniotic fluid and chorionic villus samples. In the case of absence of known mutations of *SALL1* in affected families, prenatal diagnosis can be performed by high-resolution ultrasound between 18 and 22 weeks of gestation, by finding the thumb malformations since ARM can not be seen easily, while renal anomalies can be identified [56].

2.4.4.2 FG Syndrome

FG syndrome, which takes its name from the initials of the first described case [83], is characterised by mental retardation, and multiple congenital anomalies including large head, imperforate anus, congenital hypotonia and partial agenesis of corpus callosum). It can involve many body systems, including: (1) the central nervous system (most markedly mental retardation, congenital hypotonia, convulsions, and sensorineural deafness) – the malformations that can be seen through imaging techniques are partial or total agenesis of the corpus callosum, hydrocephalus, megaloencephaly and neuronal migration defect; (2) dysmorphic features: frontal bossing, macrocephaly, hypertelorism, telecanthus, epicanthal folds and downward-slanting palpebral fissures; (3) severe contipation that can be associated with ARM; (4) genital malformations: cryptorchidism, hypospadias, hernia; (5) patients usually have fine, silky, and soft hair with an anterior upswept hairline and excessive number of hairwhorls; (6) ocular abnormalites in the form of squint and ptosis; (7) broad thumbs and halluces. This syndrome is X-linked recessive with a gene map locus on Xq12-q21.31.

2.4.4.3 Pallister-Hall Syndrome

This is a rare, life-threatening disorder that is characterised by hypothalamic hamartoma (commonly leading to precocious puberty or panhypopituitarism), polydactyly (central), imperforate anus, and respiratory tract anomalies (bifid epiglottis and/or other laryngeal anomalies) [37]. The mode of inheritance is autosomal dominant, with remarkably variable expression [10, 102]. PHS (together with Greig cephalopolysyndactyly), as a distinct and pleiotropic developmental anomaly, is caused by mutations in the gene *GLI3*, which is inherited in an autosomal dominant pattern [50, 121].

PHS disease tends to be expressed mildly in familial cases, while it takes a more dramatic course in sporadic cases. The estimate of recurrence risk and genetic counselling should be based on whether the index case is part of a familial condition. In this case the disease usually tends to repeat itself in successive generations of affected individuals, with a 50% recurrence risk. Incomplete penetrance is not reported and those who harbour the mutation usually manifest the disease, but with highly variable expression. However it is important to inform these families about the possibility of having a baby affected with PHS in a more severe or even milder form than that of the other individuals in the family. Regarding sporadic cases, in whom the disease tends to occur in a severe form and frequently with a higher mortality rate and reduced reproductive fitness, the recurrence risk can be that of the general population. However, the absence of cases with gonadal or germinal mosaicism does not mean that it can not exist, so the family should be informed about the substantial risk of recurrence [9].

2.4.4.4 VACTERL Association (VATER)

VACTERL is an acronym for vertebral anomalies (fusion, hypoplasia), ARM, cardiac malformations, tracheoesophageal fistula with or without atresia, renal anomalies (renal agenesis, hypoplasia or even cystic dysplasia) and limb anomalies (usually involving the radial ray such as radial or thumb hypoplasia, either uni- or bilateral). This term represents the expanded previous acronym of VATER, which stands for vertebral defects, anal atresia, tracheoesophageal fistula, and radial dysplasia, first reported by Quan and Smith [91]. VACTERL is believed to result from an early embryonic insult, more specifically of blastogenic origin occurring during the first 4 weeks of embryogenesis, so the expected effects are primary, polytopic, developmental field defects [70]. This early embryonic event can lead to different defects in various body systems. Of the 416 patients with ARM described by Ratan et al. [93], the additional anomalies occurred in 58%, of whom 2, 3 and 4 additional malformations were observed in 50%, 29%, and 16% of cases, respectively. Only three patients showed the full picture of VACTERL. In another study of 140 patients with ARM, only 2 patients exhibited all of the characteristics of the association, but 44 patients had 3 or more of the components of VACTERL association besides the ARM [76].

This association is frequent and it is estimated to be 1:7,000–10,000 live births [21]. Almost all cases are sporadic and the recurrence risk is minimal. However, VATER with hydrocephalus represents a distinct entity, since an autosomal recessive mode of inheritance has been reported in several families [47, 119]. A novel germline mutation of the *PTEN* gene in a patient with macrocephaly, ventricular dilatation and features of VATER association was recently reported [94]. Another form of VACTERL and hydrocephalus is thought to be X-linked recessive [32, 46, 65]. Chromosomal anomalies have been reported frequently in the literature in association with VACTERL, these can be trisomy 18 or long arm deletion of chromosome 13 [1, 6, 43, 110]. Recently, a case of VATER with 9q+ was reported [2], similarly a case of interstitial deletion of the long arm of chromosome 6: del (6) (q13q15) in association with VACTERL association [122], and another male case with VACTERL association and a karyotype showing mosaicism for a supernumerary ring chromosome in 63% of all the metaphases of both the lymphocytes and fibroblasts. This ring chromosome belongs to chromosome 12 [20]. A patient affected with VACTERL has also been found to harbour a somatic point mutation in mitochondrial DNA obtained from kidney tissue [25].

2.4.4.5 Sirenomelia

Sirenomelia may represent one of the oldest diagnosed congenital deformities, since it was mentioned by the ancient Greeks. Sirenomelia manifests with fusion of the lower limbs at a varying level and degree, with inability to perform normal movements and rotation (apodia, monopodia and dipodia). Not only are limb anomalies found in this lethal phenomenon, but also ARM (variable forms, including cloacal anomalies), genital (absent or arrested development), renal (cystic kidneys, or agenesis as a common cause of death when bilateral), gastrointestinal tract malformation, skeletal (vertebral and rib anomalies), various upper-limb defects, congenital heart disease and more [61]. It has an estimated incidence rate of 1:60,000, and a male:female ratio of 2:7. Almost all cases are sporadic with recurrence risk similar to that of the general population [49]. It was originally believed that sirenomelia is a severe form of CRS; however it has since been suggested that it is rather the result of early embryonic vascular insult leading to ischaemia in the caudal portion of the foetus [5, 15, 117]. It is thought that in sirenomelia an aberrant vessel originating from the vitelline artery shunts the blood supply coming to the high abdominal aorta directly through the umbilical cord to the placenta. The result would be severe hypoperfusion of structures distal to the origin of that aberrant vessel, since this vessel steals the blood supply from the caudal region of the foetus; this vascular stealing phenomenon is thought to be responsible for the pathogenesis of sirenomelia [112].

2.4.4.6 Caudal Regression Syndrome

As the name implies, this syndrome is characterised by a heterogeneous group of caudal anomalies. It may include variable degrees of spinal column agenesis, ARM and genitourinary anomalies. Its effects are not restricted to the caudal part of the body; CRS can also be associated with pulmonary hypoplasia and congenital heart malformations [29]. The estimated incidence rate of CRS is 1:7,500 births, but some authors report a higher incidence of 1:200–1:1,000 [125], while Diel et al. [26] reports an incidence rate of 1:10,000–1:20,000. We believe that the difference in previously reported incidence rates is due to the various presentations of CRS and to the possibility of its association with other multisystemic malformations such as omphalocele, cloacal exstrophy, imperforate anus, and spinal deformities, and VACTERL [104].

Cama et al. [16] outlined some of the typical features of this syndrome: (1) cutaneous signs (such flattening of the buttock and shortening of the intergluteal cleft secondary to lumbosacral agenesis), (2) sacrococcygeal agenesis (partial or total), (3) skeletal deformities (vertebral, rib, or even lower-limb deformities, scoliosis with or without kyphosis, hip dislocation), (4) congenital heart defects (tetralogy of Fallot), (5) ARM, (6) genitourinary disorders (renal aplasia or dysplasia, whether unilateral or bilateral, vesicoureteral reflux, ureterocele, hypospadias and malformed external genitalia) and (7) pulmonary hypoplasia. ARM is considered a frequent finding with CRS: 27–48% of published CRS series of cases were associated with ARM [16, 96]. ARM can be present in CRS in different forms, whether mild or severe.

Nearly all the cases of CRS are sporadic, and the genetic background to the development of CRS is partially known [15, 113, 127]. CRS occurs in up to 1% of pregnancies of diabetic women, and up to 22% of CRS occurs in the offspring of mothers affected with diabetes mellitus type I or type II [118]. The risk seems to be greater for women who are insulin-dependent since it is estimated that they are 200–400 times more likely to have a child with CRS than non-diabetic women, reflecting the fact that CRS is one of the most characteristic abnormalities occurring in foetuses of diabetic women. A reasonable explanation of this combination is that the teratogenic cause underlying CRS in diabetes is hyperglycaemia [33]. To the best of our knowledge, the exact mechanism leading to CRS is not yet known; however, it has been proposed that before the 7th week of gestation, one or more processes of primitive streak migration, primary or secondary neurulation, or differentiation are compromised in the embryonic caudal parts [15, 113, 118, 127].

2.4.4.7 Currarino Syndrome

CS was first described as the triad of ARM, hemisacrum and presacral mass [23]. The radiological aspect of the sacrum described by Currarino is the so-called "sickle-shaped sacrum", which is caused by the presence of a hemisacrum with preservation of the first sacral vertebra. This finding is pathognomonic for the diagnosis CS [103]. According to Cama [16], classification of sacral anomalies presents: (1) total sacral agenesis with normal or short transverse pelvic diameter, and the defect extending to include some lumbar vertebrae, (2) total sacral agenesis with intact lumbar vertebrae, (3) partial agenesis or hypodeveloped sacrum (preserved S1), (4) hemisacrum and (5) coccygeal agenesis. Following this classification, CS has been characterised as a type 4 sacral anomaly. Different forms of ARM can be present in CS, such as rectourethral fistula, rectovestibular fistula and rectocloacal fistula [73]. It is estimated that 29% of ARM are associated with sacral anomalies [97]. The most frequent ARM in CS is ARM with perineal fistula [86]. The same anomaly was reported by some investigators as anorectal stenosis [84]. The presence of perineal fistula leads to difficulty in defecation, and constipation, which can be the leading symptom in the diagnosis of the disease [73]. The presacral mass can be an anterior meningocoele, teratoma, dermoid cyst, rectal duplication, a combination of these or another uncommon tumour with reported malignant forms [81, 114].

This disorder can be associated with other defects, such as urologic abnormalities including horseshoe, duplex, or dysplastic kidney, vesicoureteric reflux, duplex ureter or hypospadias. Gynaecological malformations may include bicornuate uterus, septate vagina and bifid clitoris. Thus, the term syndrome was applied correctly rather than triad [4, 67]. CS can be sporadic or familial, with an autosomal dominant mode of inheritance. It has variable expression (i.e. it can present with all or some of the previously mentioned anomalies), and incomplete penetrance, which means that some individuals have the affected genotype but with normal phenotype. In the biggest series in the literature, the female:male ratio is 1.7:1, with the possibility that the greater number of females is due to the coexistence of the gynaecologic or urologic problems, which are noticed more frequently in females. Moreover, 33% of cases maybe asymptomatic [67].

Previous studies suggest the importance of a locus on chromosome 7q39. This region was identified by linkage analysis to search for the causative gene for CS [3, 66]. The region 7q39 includes three genes; one of the best known is Sonic Hedgehog (*SHH*), a very important molecular factor in early embryogenesis in different body systems; its mutations are responsible for holoprosencephaly in humans [3]. *SHH* was excluded as a causative gene for CS by two thorough investigations performed by Seri et al. [107] and Vargas et al. [120]. The remaining two homeobox genes are also important for early development, namely *EN2* and *HLXB9*. Further linkage analysis on affected families with sacral agenesis showed that the position of the causative gene was more towards the terminal end of the long arm of chromosome 7. This excluded more obviously the involvement of *SHH* and *EN2*, which are located more centromerically. *EN2* was excluded since it is located upstream of *SHH* [101, 107]. The third candidate gene is *HLXB9* (homeobox gene), which was found to be expressed in early development in human embryos and expressed specifically in the anterior horn region of the spinal cord [101]. *HLXB9* is composed of three exons. A combination of DGGE, single-strand conformation polymorphism and direct DNA sequencing experiments were carried out. These studies demonstrated several mutations in the gene coding sequence and in intron-exon boundaries in both sporadic and familial cases, confirming the causative role of *HLXB9* in this syndrome (Fig. 2.2) [4, 36, 101]. To the best of our knowledge, the known

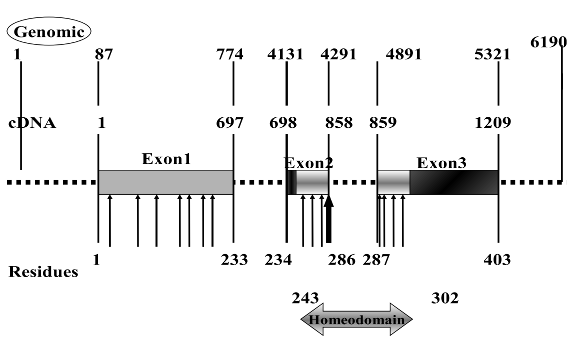

Fig. 2.2 Schematic representation of the structure of HLXB9 gene, showing the three exons, the homeodomain, the numbering according to genomic sequence, and nucleotide numeration according to cDNA sequence, and residues numeration. The arrows indicate the sites of discovered mutations along the gene

mutations causing CS are in total 9 missense, 2 nonsense, 2 splicing, 7 frameshifts, and 6 hemizygous microdeletions. Since the loss of one copy of *HLXB9* gene (haploinsufficiency) can lead to CS, as well as cases involving deletions of the region 7q35-tel, it is quite probable that CS is caused by loss-of-function mutations. The presence of polyalanine expansion with various triplet repeats in homeobox genes can lead to certain pathologic conditions, for example synpolydactyly, oculopharyngeal muscular dystrophy, and cleidocranial dysplasia [13, 34, 79]. This abnormal expansion was not found to determine CS, since they were found in both affected and control cases. In the study performed by Belloni et al. [4], the commonest allele in the heterozygous form for the general population was CGC11, accounting for 90.23% of the 100 total control chromosomes. Other cases observed were CGC12, CGC9 and CGC8, accounting for 1.7, 7.47 and 0.6%, respectively. Only one case was found with homozygous change in the polyalanine tract giving rise to the CGC9/CGC9 allelic combination. The sample was a "control" case, but a closer look and detailed investigations showed that the person lacked posterior arch fusion of the vertebrae and had left-sided scoliosis. The possible relationship between the length of the alanine fragment and the presence of sacral anomalies in either affected or control cases was also excluded in that study [4].

The same studies showed in some familial cases, that the mutation was found as well in asymptomatic patients with normal sacral X-ray giving the disease the characteristic of incomplete penetrance. *HLXB9* mutations are not found in other forms of sacral agenesis and are found only in individuals with CS. However, *HLXB9* mutations are not found in all patients who are diagnosed clinically; this may suggest the possibility of genetic heterogeneity or the presence of some non-genetic components influencing the occurrence of this anomaly, at least for sporadic cases. To the best of our knowledge, no further studies have proved the involvement of any other genes in sacral and anorectal development [104]. However Horn et al. [45] have reported four cases of minimal clinical expression of holoprosencephaly and CS due to different cytogenetic rearrangements affecting both *SHH* and *HLXB9* at 7q36.3

Taking into consideration all of the previous data, we believe that genetic counselling is highly appreciated in any diagnosed case of CS. There is always the need for detailed physical examination and sacral X-ray to the parents to exclude minimal signs of the disease. Finding *HLXB9* mutations in the index case necessitates the molecular genetic study of the parents,

Table 2.4 Examples of genetic syndromes with ARM as a feature. *XR* X-linked recessive, *AD* autosomal-dominant, *AR* autosomal-recessive

Syndrome	Prominent Features	Mode of inheritance	Locus or gene if known
Opitz G	Hypertelorism, hypospadias, swallowing difficulties.	XR,	MID 1 gene
Opitz Frias	The same features of Opitz	AD	22q11.2
Fraser	cryptophthalmos with other malformations, cryptophthalmos-syndactyly syndrome.	AR	FRAS1
Johanson-Blizzard	Hypoplastic alae nasi, deafness, pancreatic insufficiency, hypothyroidism.	AR	-
CHARGE	Coloboma, heart anomaly, choanal atresia, retardation, genital and ear anomalies	Sporadic or AD	CHD7
EEC	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate	AD	P63
Goldenhaar	Hemifacial microsomia, cardiac, vertebral, and central nervous system defects	Sporadic or AD	
Velocardiofacial	Cardiac, thymic, hypocalcaemia, vertebral, others	Sporadic or AD	Chromosome 22 microdeletion
McKusick-Kaufman	Hydrometrocolpus, Hirschsprung, hydronephrosis	AR	MKKS

even if they were asymptomatic, because of incomplete penetrance with possible subsequent extension of the study for further family members in the case of positive results. Since CS is autosomal dominant and the recurrence risk is 50%, the disease has both incomplete penetrance and variable expression. Prenatal diagnosis through ultrasound can only detect the presacral mass, which is not present in all cases, making the molecular diagnosis the most favourable method for diagnosing CS prenatally [67]. ARM can also be associated with many other syndromes, although in some cases it maybe not be a main characteristic feature (Table 2.4) (see Chap. 3).

2.5 Mouse Models with ARM

ARM can be induced by using certain teratogens such as ethylenethiourea (ETU) [41], retinoic acid [28], and adriamycin [64] in mice, rats [59] and piglets [90]. The ARM produced by adriamycin in rat foetuses seem to be more severe and include variations of cloacal agenesis, which is not generally considered the best example with which to study comparable human ARM [27]. ETU treatment of timed-pregnant rats showed ARM in 80% of offspring, with variable phenotypes ranging from simple anal membrane to rectourethral fistula, resembling the ARM seen in the human spectrum [41]. Recently, an explanation of the developmental defects leading to ARM in ETU-induced rats showed the presence of maldevelopment of the cloaca, delayed tailgut regression, increased posterior cloacal wall apoptosis, and underdevelopment of the dorsal aspect of the cloaca itself and its membrane [90].

Although *SHH* mutations were excluded as a cause of ARM in humans [107, 120], *Shh* and the *Shh*-responsive transcription factors *Gli2* and *Gli3* have proved to be essential for mammalian foregut development. Mutant mice for these factors showed a series of multisystemic defects including ARM and other components of VACTERL association in man [54, 63, 77, 78, 92].

The ablation of *Hlxb9* expression was studied in two murine knockout models. Heterozygous mice had no apparent abnormalities, but the homozygous mutants *Hlxb9* -/- were incompatible with life, although they exhibited no obvious morphologic abnormality. Differential models were made for the cartilage and bones, with normal skeletal development and absent sacral defects. However, defects were described in the development of the pancreas, which is not seen in humans affected with CS [38, 62] (Table 2.5).

2.6 Conclusions

Current knowledge of the clinical genetics, cytogenetics and molecular genetics of ARM is still progressing. The genetic basis of these anomalies is very complex

Table 2.5 Human and mouse genotypic/phenotypic correlations. *GCPS* Greig cephalopolysyndactyly syndrome, *PHS* Pallister-Hall syndrome

Gene mutation	Effects in man	Effects in animal model
HLXB9	Currarino syndrome (Heterozygous mutation)	*Hlxb9*: Heterozygous mice (normal skeleton except for curled body and small size) Homozygous mutation (incompatible with life, defects in motor neurons).
SALL1	Townes-Brocks syndrome (heterozygous mutation)	*Sall1:* Heterozygous mice (no detectable phenotype) Homozygous mutation (all died in neonatal period, only renal anomalies)
GLI3	PHS and GCPS (heterozygous mutation)	*Gli3* mutant mice shows similar phenotype to PHS and GCPS
SHH	Holoproscencephaly (heterozygous mutations)	*Shh* and/or its transcription factor mutations: different malformations including gastrointestinal tract malformations (ARM)

because of their phenotypic variability and their multigenic origin. ARM phenotypes may result from alterations of different but functionally related genes that are part of pathways involved in the development of the most caudal region of the hindgut. Moreover, some mutated genes are known to produce an ARM phenotype in mice. ARM in Currarino Syndrome represents the only association for which a single gene has been identified in humans. Loss of function mutations of the HLXB9 gene can lead to the association with anorectal malformation, hemisacrum and presacral mass.

References

1. Allderdice PW, Davis JG, Miller OJ, Klinger HP, Warburton D, Miller DA, Allen FH Jr, Abrams CA, McGilvray E (1969) The 13q-deletion syndrome. Am J Hum Genet 21(5):499–512
2. Aynaci FM, Celep F, Karaguzel A, Baki A, Yildiran A (1996) A case of VATER association associated with 9qh+. Genet Couns 7:321–322
3. Belloni E, Muenke M, Roessler E, Traverso G, Siegel-Bartelt J, Frumkin A, Mitchell HF, Donis-Keller H, Helms C, Hing AV, Heng HH, Koop B, Martindale D, Rommens JM, Tsui LC, Scherer SW (1996) Identification of Sonic hedgehog as a candidate gene responsible for holoprosencephaly. Nat Genet 14:353–356
4. Belloni E, Martucciello G, Verderio D, Ponti E, Seri M, Jasonni V, Torre M, Ferrari M, Tsui LC, Scherer SW (2000) Involvement of the *HLXB9* homeobox gene in Currarino syndrome. Am J Hum Genet 66:312–319
5. Benacerraf BR (1998) Ultrasound of Fetal Syndromes. Churchill Livingstone, Philadelphia, pp 250–254
6. Benady SG, Harris RJ (1969) Trisomy 17–18. A study of five cases, three of whom were associated with oesophageal atresia. Acta Paediatr Scand 58:445–448
7. Berends MJ, Tan-Sindhunata G, Leegte B, van Essen AJ (2001) Phenotypic variability of cat-eye syndrome. Genet Couns 12:23–34
8. Bianchi DW, Crombleholme TM, D'Alton ME (2000) Fetology: Diagnosis and Management of the Fetal Patient, McGraw-Hill, Garamond
9. Biesecker LG (2004) *GLI3* and the Pallister-Hall and Greig cephalopolysyndactyly syndromes. In: Epstein CJ, Erickson RP, Wynshaw-Boris A (eds) Inborn Errors of Development. Oxford University Press, Oxford, pp 257–264
10. Biesecker LG, Abbott M, Allen J, Clericuzio C, Feuillan P, Graham JM Jr, Hall J, Kang S, Olney AH, Lefton D, Neri G, Peters K, Verloes A (1996) Report from the workshop on Pallister-Hall syndrome and related phenotypes. Am J Med Genet 65:76–81
11. Black CT, Sherman JO (1989) The association of low imperforate anus and Down's syndrome. J Pediatr Surg 24:92–94; Discussion p 94
12. Boocock GR, Donnai D (1987) Anorectal malformation: familial aspects and associated anomalies. Arch Dis Child 62:576–579
13. Brais B, Bouchard JP, Xie YG, Rochefort DL, Chretien N, Tome FM, Lafreniere RG, Rommens JM, Uyama E, Nohira O, Blumen S, Korczyn AD, Heutink P, Mathieu J, Duranceau A, Codere F, Fardeau M, Rouleau GA (1998) Short GCG expansions in the PABP2 gene cause oculopharyngeal muscular dystrophy. Nat Genet 18:164–167
14. Briault S, Hill R, Shrimpton A, Zhu D, Till M, Ronce N, Margaritte-Jeannin P, Baraitser M, Middleton-Price H, Malcolm S, Thompson E, Hoo J, Wilson G, Romano C, Guichet A, Pembrey M, Fontes M, Poustka A, Moraine C (1997) A gene for FG syndrome maps in the Xq12-q21.31 region. Am J Med Genet 73:87–90
15. Callen PW (2000) Ultrasonography in Obstetrics and Gynecology, 4th edn. Saunders, Philadelphia, pp 364–367

16. Cama A, Palmieri A, Capra V, Piatelli GL, Ravegnani M, Fondelli P (1996) Multidisciplinary management of caudal regression syndrome (26 cases). Eur J Pediatr Surg 6 (Suppl 1):44–45
17. Carson JA, Barnes PD, Tunell WP, Smith EI, Jolley SG (1984) Imperforate anus: the neurologic implication of sacral abnormalities. J Pediatr Surg 19:838–842
18. Cho S, Moore SP, Fangman T (2001) One hundred three consecutive patients with anorectal malformations and their associated anomalies. Arch Pediatr Adolesc Med 155:587–591
19. Christensen K, Madsen CM, Hauge M, Kock K(1990) An epidemiological study of congenital anorectal malformations: 15 Danish birth cohorts followed for 7 years. Paediatr Perinat Epidemiol 4:269–275
20. Cinti R, Priolo M, Lerone M, Gimelli G, Seri M, Silengo M, Ravazzolo R (2001) Molecular characterisation of a supernumerary ring chromosome in a patient with VATER association. J Med Genet 38:E6
21. Corsello G, Maresi E, Corrao AM, Dimita U, Lo Cascio M, Cammarata M, Giuffre L (1993) VATER/VACTERL association: clinical variability and expanding phenotype including laryngeal stenosis. Am J Med Genet 47:118
22. Cortes D, Thorup JM, Nielsen OH, Beck BL (1995) Cryptorchidism in boys with imperforate anus. J Pediatr Surg 30(4):631–635
23. Currarino G, Coln D, Votteler T (1981) Triad of anorectal, sacral, and presacral anomalies. AJR Am J Roentgenol 137:395–398
24. Cuschieri A; EUROCAT Working Group (2002) Anorectal anomalies associated with or as part of other anomalies. Am J Med Genet 110:122–130
25. Damian MS, Seibel P, Schachenmayr W, Reichmann H, Dorndorf W (1996) VACTERL with the mitochondrial np 3243 point mutation. Am J Med Genet 62:398–403
26. Diel J, Ortiz O, Losada RA, Price DB, Hayt MW, Katz DS (2001) The sacrum: pathologic spectrum, multimodality imaging, and subspecialty approach. Radiographics 21:83–104
27. Diez-Pardo JA, Baoquan Q, Navarro C, Tovar JA (1996) A new rodent experimental model of esophageal atresia and tracheoesophageal fistula: preliminary report. J Pediatr Surg 31:498–502
28. Diez-Pardo JA, Marino JM, Baoquan Q, Delgado-Baeza E, Fernaneez A, Morales MC, Tovar JA (1995) Neural tube defects: an experimental model in the foetal rat. Eur J Pediatr Surg 5:198–202
29. Duhamel B (1961) From the mermaid to anal imperforation: the syndrome of caudal regression. Arch Dis Child 36:152–155
30. Endo M, Hayashi A, Ishihara M, Maie M, Nagasaki A, Nishi T, Saeki M (1999) Analysis of 1,992 patients with anorectal malformations over the past two decades in Japan. Steering Committee of Japanese Study Group of Anorectal Anomalies. J Pediatr Surg 34:435–41
31. Fitzgerald MJT, Fitzgerald M (1994) Human Embryology. Bailliere Tindall, Philadelphia, pp 1–251
32. Froster UG, Wallner SJ, Reusche E, Schwinger E, Rehder H (1996) VACTERL with hydrocephalus and branchial arch defects: prenatal, clinical, and autopsy findings in two brothers. Am J Med Genet 62:169–172
33. Gabbe SG, Niebyl JR, Simpson JL (2002) Obstetrics: Normal and Problem Pregnancies, 4th edn. Churchill Livingstone, New York, pp 1090–1091
34. Goodman FR, Mundlos S, Muragaki Y, Donnai D, Giovannucci-Uzielli ML, Lapi E, Majewski F, McGaughran J, McKeown C, Reardon W, Upton J, Winter RM, Olsen BR, Scambler PJ (1997) Synpolydactyly phenotypes correlate with size of expansions in HOXD13 polyalanine tract. Proc Natl Acad Sci U S A 94:7458–7463.
35. Greenwood RD, Rosenthal A, Nadas AS (1975) Cardiovascular malformations associated with imperforate anus. J Pediatr 86:576–579
36. Hagan DM, Ross AJ, Strachan T, Lynch SA, Ruiz-Perez V, Wang YM, Scambler P, Custard E, Reardon W, Hassan S, Nixon P, Papapetrou C, Winter RM, Edwards Y, Morrison K, Barrow M, Cordier-Alex MP, Correia P, Galvin-Parton PA, Gaskill S, Gaskin KJ, Garcia-Minaur S, Gereige R, Hayward R, Homfray T (2000) Mutation analysis and embryonic expression of the HLXB9 Currarino syndrome gene. Am J Hum Genet 66:1504–1515
37. Hall JG, Pallister PD, Clarren SK, Beckwith JB, Wiglesworth FW, Fraser FC, Cho S, Benke PJ, Reed SD (1980) Congenital hypothalamic hamartoblastoma, hypopituitarism, imperforate anus and postaxial polydactyly – a new syndrome? Part I: clinical, causal, and pathogenetic considerations. Am J Med Genet 7:47–74
38. Harrison KA, Thaler J, Pfaff SL, Gu H, Kehrl JH (1999) Pancreas dorsal lobe agenesis and abnormal islets of Langerhans in Hlxb9-deficient mice. Nat Genet 23:71–75
39. Hasse W (1976) Associated malformations with anal and rectal atresia. Prog Pediatr Surg 9:99–103
40. Hassink EA, Rieu PN, Hamel BC, Severijnen RS, vd Staak FH, Festen C (1996) Additional congenital defects in anorectal malformations. Eur J Pediatr 155:477–482
41. Hirai Y, Kuwabara N (1990) Transplacentally induced anorectal malformations in rats. J Pediatr Surg 25:812–816
42. Hoekstra WJ, Scholtmeijer RJ, Molenaar JC, Schreeve RH, Schroeder FH (1983) Urogenital tract abnormalities associated with congenital anorectal anomalies. J Urol 130:962–963
43. Holman GH, Erkman B, Zacharias DL, Kock HF (1963) The 18-trisomy syndrome – two new clinical variants. One with associated tracheoesophageal fistula and the other with probable familial occurrence. N Engl J Med 268:982–988
44. Holschneider AM, Ure BM, Pfrommer W, Meier-Ruge W (1996) Innervation patterns of the rectal pouch and fistula in anorectal malformations: a preliminary report. J Pediatr Surg 31:357–362

45. Horn D, Tonnies H, Neitzel H, Wahl D, Hinkel GK, von Moers A, Bartsch O (2004) Minimal clinical expression of the holoprosencephaly spectrum and of Currarino syndrome due to different cytogenetic rearrangements deleting the Sonic Hedgehog gene and the HLXB9 gene at 7q36.3. Am J Med Genet 128:85–92
46. Hunter AGW, MacMurray B (1987) Malformations of the VATER association plus hydrocephalus in a male infant and his maternal uncle. Proc Greenwood Genet Center 6:146–147
47. Iafolla AK, McConkie-Rosell A, Chen YT (1991) VATER and hydrocephalus: distinct syndrome? Am J Med Genet 38:46–51
48. Joseph VT, Chan KY, Siew HF (1985) Anorectal malformations and their associated anomalies. Ann Acad Med Singapore 14:622–625
49. Kallen B, Winberg J (1974) Caudal mesoderm pattern of anomalies: from renal agenesis to sirenomelia. Teratology 9:99–111
50. Kang S, Allen J, Graham JM Jr, Grebe T, Clericuzio C, Patronas N, Ondrey F, Green E, Schaffer A, Abbott M, Biesecker LG (1997) Linkage mapping and phenotypic analysis of autosomal dominant Pallister-Hall syndrome. J Med Genet 34:441–446
51. Kiely EM, Peña A (1998) Anorectal Malformations. In: O'Niell JA, Rowe MI, Grosfeld JL, Fonkalsrud EW, Coran AG (eds) Pediatric Surgery. Mosby Year Book, St. Louis, pp 1425–1448
52. Kiesewetter WB, Chang JH (1977) Imperforate anus: a five to thirty year follow-up perspective. Prog Pediatr Surg 10:111–120
53. Kiesewetter WB, Sukarochana K, Sieber WK (1965) The frequency of aganglionosis associated with imperforate anus. Surgery 58:877–880
54. Kim PC, Mo R, Hui Cc C (2001) Murine models of VACTERL syndrome: Role of sonic hedgehog signaling pathway. J Pediatr Surg 36:381–384
55. Kluth D, Hillen M, Lambrecht W (1995) The principles of normal and abnormal hindgut development J Pediatr Surg 30:1143–1147
56. Kohlhase J, Engel W (2004) SALL1 and the Townes-Brocks Syndrome. In: Epstein CJ, Erickson RP, Wynshaw-Boris A (eds) Inborn Errors of Development. Oxford University Press, Oxford, pp 265–271
57. Kohlhase J, Schuh R, Dowe G, Kuhnlein RP, Jackle H, Schroeder B, Schulz-Schaeffer W, Kretzschmar HA, Kohler A, Muller U, Raab-Vetter M, Burkhardt E, Engel W, Stick R (1996) Isolation, characterisation, and organ-specific expression of two novel human zinc finger genes related to the Drosophila gene spalt. Genomics 38:291–298
58. Kohlhase J, Taschner PE, Burfeind P, Pasche B, Newman B, Blanck C, Breuning MH, ten Kate LP, Maaswinkel-Mooy P, Mitulla B, Seidel J, Kirkpatrick SJ, Pauli RM, Wargowski DS, Devriendt K, Proesmans W, Gabrielli O, Coppa GV, Wesby-van Swaay E, Trembath RC, Schinzel AA, Reardon W, Seemanova E, Engel W (1999) Molecular analysis of SALL1 mutations in Townes-Brocks syndrome. Am J Hum Genet 64:435–445
59. Lambrecht W, Lierse W (1987) The internal sphincter in anorectal malformations: morphologic investigations in neonatal pigs. J Pediatr Surg 22:1160–1168
60. Landau D, Mordechai J, Karplus M, Carmi R (1997) Inheritance of familial congenital isolated anorectal malformations: case report and review. Am J Med Genet 71:280–282
61. Lerone M, Bolino A, Martucciello G (1997) The genetics of anorectal malformations: a complex matter. Semin Pediatr Surg 6:170–179
62. Li H, Arber S, Jessell TM, Edlund H (1999) Selective agenesis of the dorsal pancreas in mice lacking homeobox gene Hlxb9. Nat Genet 23:67–70
63. Litingtung Y, Lei L, Westphal H, Chiang C (1998) Sonic hedgehog is essential to foregut development. Nat Genet 20:58–61
64. Liu MI, Hutson JM, Zhou B (1999) Critical timing of bladder embryogenesis in an adriamycin-exposed rat fetal model: a clue to the origin of the bladder J Pediatr Surg 34:1647–1651
65. Lomas FE, Dahlstrom JE, Ford JH (1998) VACTERL with hydrocephalus: family with X-linked VACTERL-H. Am J Med Genet 76:74–78
66. Lynch SA, Bond PM, Copp AJ, Kirwan WO, Nour S, Balling R, Mariman E, Burn J, Strachan T (1995) A gene for autosomal dominant sacral agenesis maps to the holoprosencephaly region at 7q36. Nat Genet 11:93–95
67. Lynch SA, Wang Y, Strachan T, Burn J, Lindsay S (2000) Autosomal dominant sacral agenesis: Currarino syndrome. J Med Genet 37:561–566
68. Manny J, Shiller M, Horner R, Stein M, Luttwak M (1973) Congenital familial anorectal anomaly. Am J Surg 125:639–640
69. Marlin S, Blanchard S, Slim R, Lacombe D, Denoyelle F, Alessandri JL, Calzolari E, Drouin-Garraud V, Ferraz FG, Fourmaintraux A, Philip N, Toublanc JE, Petit C (1999) Townes-Brocks syndrome: detection of a SALL1 mutation hot spot and evidence for a position effect in one patient. Hum Mutat 14:377–386
70. Martinez-Frias ML, Frias JL (1999) VACTERL as primary, polytopic developmental field defects. Am J Med Genet 83:13–16
71. Martinez-Frias ML, Frias JL, Opitz JM (1998) Errors of morphogenesis and developmental field theory. Am J Med Genet 76:291–296

72. Martinez-Frias ML, Bermejo Sanchez E, Arroyo Carrera I, Perez Fernandez JL, Pardo Romero M, Buron Martinez E, Hernandez Ramon F (1999) [The Townes-Brocks syndrome in Spain: the epidemiological aspects in a consecutive series of cases] An Esp Pediatr 50:57–60
73. Martucciello G, Torre M, Belloni E, Lerone M, Pini Prato A, Cama A, Jasonni V (2004) Currarino syndrome: proposal of a diagnostic and therapeutic protocol. J Pediatr Surg 39:1305–1311
74. McDermid HE, Duncan AM, Brasch KR, Holden JJ, Magenis E, Sheehy R, Burn J, Kardon N, Noel B, Schinzel A (1986) Characterization of the supernumerary chromosome in cat eye syndrome. Science 232:646–648
75. Metts JC 3rd, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW 3rd (1997) Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. J Urol 158:1298–1300
76. Mittal A, Airon RK, Magu S, Rattan KN, Ratan SK (2004) Associated anomalies with anorectal malformation (ARM). Indian J Pediatr 71:509–514
77. Mo R, Kim JH, Zhang J, Chiang C, Hui CC, Kim PC (2001) Anorectal malformations caused by defects in sonic hedgehog signaling. Am J Pathol 159:765–774
78. Motoyama J, Liu J, Mo R, Ding Q, Post M, Hui CC (1998) Essential function of Gli2 and Gli3 in the formation of lung, trachea and oesophagus. Nat Genet 20:54–57
79. Mundlos S, Otto F, Mundlos C, Mulliken JB, Aylsworth AS, Albright S, Lindhout D, Cole WG, Henn W, Knoll JH, Owen MJ, Mertelsmann R, Zabel BU, Olsen BR (1997) Mutations involving the transcription factor CBFA1 cause cleidocranial dysplasia. Cell. 89:773–779
80. Narasimharao KL, Prasad GR, Mukhopadhyay B, Katariya S, Mitra SK, Pathak IC (1983) Vesicoureteric reflux in neonates with anorectal anomalies. Br J Urol 55:268–270
81. Norum J, Wist E, Bostad L (1991) Incomplete Currarino syndrome with a presacral leiomyosarcoma. Acta Oncol 30:987–988
82. O'Callaghan M, Young ID (1990) The Townes-Brocks syndrome. J Med Genet 27:457–461
83. Opitz JM, Kaveggia EG (1974) Studies of malformation syndromes of man 33: the FG syndrome. An X-linked recessive syndrome of multiple congenital anomalies and mental retardation. Z Kinderheilkd 117:1–18
84. O'Riordain DS, O'Connell PR, Kirwan WO (1991) Hereditary sacral agenesis with presacral mass and anorectal stenosis: the Currarino triad. Br J Surg 78:536–538
85. Parkkulainen KV, Hjelt L, Sulamaa M (1960) Anal atresia combined with aganglionic megacolon. Acta Chir Scand 118:252–256
86. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
87. Peña A (1996) Anorectal anomalies. In: Puri P (ed) Newborn Surgery. Butterworth-Heinemann, Oxford, UK, pp 379–394
88. Pinsky L (1978) Syndromology of anorectal malformation (atresia, stenosis, ectopia). Am J Med Genet 1:461–474
89. Powell CM, Michaelis RC (1999) Townes-Brocks syndrome. J Med Genet 36:89–93
90. Qi BQ, Beasley SW, Frizelle FA (2002) Clarification of the processes that lead to anorectal malformations in the ETU-induced rat model of imperforate anus. J Pediatr Surg 37:1305–1312
91. Quan L, Smith DW (1972) The VATER association: vertebral defects, anal atresia, tracheoesophageal fistula with esophageal atresia, radial dysplasia. Birth Defects Orig Art Ser VIII:75–78
92. Ramalho-Santos M, Melton DA, McMahon AP (2000) Hedgehog signals regulate multiple aspects of gastrointestinal development. Development 127:2763–2772
93. Ratan SK, Rattan KN, Pandey RM, Mittal A, Magu S, Sodhi PK (2004) Associated congenital anomalies in patients with anorectal malformations – a need for developing a uniform practical approach. J Pediatr Surg 39:1706–1711
94. Reardon W, Zhou XP, Eng C (2001) A novel germline mutation of the PTEN gene in a patient with macrocephaly, ventricular dilatation, and features of VATER association. J Med Genet 38:820–823
95. Reid IS, Turner G (1976) Familial anal abnormality. Pediatrics 88:992–994
96. Renshaw TS (1978) Sacral agenesis. J Bone Joint Surg Am 60:373–383
97. Rich AM, Brock WA, Peña A (1988) Spectrum of genitourinary malformations in patients with imperforate anus. Pediatr Surg Int 3:110–113
98. Rickwood AM, Spitz L (1980) Primary vesicoureteric reflux in neonates with imperforate anus. Arch Dis Child 55:149–150
99. Rivosecchi M, Lucchetti MC, Zaccara A, De Gennaro M, Fariello G (1995) Spinal dysraphism detected by magnetic resonance imaging in patients with anorectal anomalies: incidence and clinical significance J Pediatr Surg 30:488–490
100. Robb L, Teebi AS (1998) Further evidence of autosomal dominant inheritance of the nonsyndromal anorectal malformations. Am J Med Genet 79:400
101. Ross AJ, Ruiz-Perez V, Wang Y, Hagan DM, Scherer S, Lynch SA, Lindsay S, Custard E, Belloni E, Wilson DI, Wadey R, Goodman F, Orstavik KH, Monclair T, Robson S, Reardon W, Burn J, Scambler P, Strachan T (1998) A homeobox gene, HLXB9, is the major locus for dominantly inherited sacral agenesis. Nat Genet 20:358–361
102. Sama A, Mason JD, Gibbin KP, Young ID, Hewitt M (1994) The Pallister-Hall syndrome. J Med Genet 31:740
103. Scherer SW, Cheung J, MacDonald JR (2003)Human chromosome 7: DNA sequence and biology. Science 2:767–772

104. Scherer S, Martucciello G, Belloni E, Torre M, (2004) HLXB9 and Sacral Agenesis and the Currarino Syndrome. In: Epstein CJ, Erickson RP, Wynshaw-Boris A (eds) Inborn Errors of Development. Oxford University Press, Oxford, pp 578–587
105. Schinzel A, Schmid W, Fraccaro M, Tiepolo L, Zuffardi O, Opitz JM, Lindsten J, Zetterqvist P, Enell H, Baccichetti C, Tenconi R, Pagon RA (1981) The "cat eye syndrome": dicentric small marker chromosome probably derived from a no 22 (tetrasomy 22pter to q11) associated with a characteristic phenotype. Report of 11 patients and delineation of the clinical picture. Hum Genet 57:148–158
106. Schwoebel MG, Hirsig J, Schinzel J, Stauffer UG (1984) Familial incidence of congenital anorectal anomalies. J Pediatr Surg 19:179–182
107. Seri M, Martucciello G, Paleari L, Bolino A, Priolo M, Salemi G, Forabosco P, Caroli F, Cusano R, Tocco T, Lerone M, Cama A, Torre M, Guys JM, Romeo G, Jasonni V (1999) Exclusion of the Sonic Hedgehog gene as responsible for Currarino syndrome and anorectal malformations with sacral hypodevelopment. Hum Genet 104:108–110
108. Shaul DB, Harrison EA (1997) Classification of anorectal malformations – initial approach, diagnostic tests, and colostomy. Semin Pediatr Surg 6:187–195
109. Smith ED (1988) Incidence, frequency of types, and etiology of anorectal malformations. Birth Defects Orig Artic Ser 24:231–246
110. Sommer A, Grosfeld JL (1970) Trisomy E and T-E fistula. J Med Genet 7:70–74
111. Spouge D, Baird PA (1986) Imperforate anus in 700,000 consecutive liveborn infants. Am J Med Genet Suppl 2:151–161
112. Stroustrup Smith A, Grable I, Levine D (2004) Case 66: caudal regression syndrome in the fetus of a diabetic mother. Radiology 230:229–233
113. Subtil D, Cosson M, Houfflin V, Vaast P, Valat A, Puech F (1998) Early detection of caudal regression syndrome: specific interest and findings in three cases. Eur J Obstet Gynecol Reprod Biol 80:109–112
114. Tander B, Baskin D, Bulut M (1999) A case of incomplete Currarino triad with malignant transformation. Pediatr Surg Int 15:409–410
115. Torres R, Levitt MA, Tovilla JM, Rodriguez G, Peña A (1998) Anorectal malformations and Down's syndrome. J Pediatr Surg 33:194–197
116 Townes PL, Brocks ER (1972) Hereditary syndrome of imperforate anus with hand, foot, and ear anomalies. J Pediatr 81:321–326
117. Twickler D, Budorick N, Pretorius D, Grafe M, Currarino G (1993) Caudal regression versus sirenomelia: sonographic clues. J Ultrasound Med 12:323–330
118. Twining P, McHugo J, Pilling D (2000) Textbook of Fetal Abnormalities. Saunders, Philadelphia, pp 158–160
119. Vandenborre K, Beemer F, Fryns JP, Vandenborne K [corrected to Vandenborre K] (1993) VACTERL with hydrocephalus. A distinct entity with a variable spectrum of multiple congenital anomalies. Genet Couns 4:199–201
120. Vargas FR, Roessler E, Gaudenz K, Belloni E, Whitehead AS, Kirke PN, Mills JL, Hooper G, Stevenson RE, Cordeiro I, Correia P, Felix T, Gereige R, Cunningham ML, Canun S, Antonarakis SE, Strachan T, Tsui LC, Scherer SW, Muenke M (1998) Analysis of the human Sonic Hedgehog coding and promoter regions in sacral agenesis, triphalangeal thumb, and mirror polydactyly. Hum Genet 102:387–392
121. Vortkamp A, Gessler M, Grzeschik KH (1991) GLI3 zinc-finger gene interrupted by translocations in Greig syndrome families. Nature 352:539–540
122. Walsh LE, Vance GH, Weaver DD (2001) Distal 13q Deletion Syndrome and the VACTERL association: case report, literature review, and possible implications Am J Med Genet 98:137–144
123. Walton M, Bass J, Soucy P (1995) Tethered cord with anorectal malformation, sacral anomalies and presacral masses: an under-recognized association Eur J Pediatr Surg 5:59–62
124. Weinstein ED (1965) Sex-linked imperforate anus. Pediatrics 35:715–717
125. Wilmshurst JM, Kelly R, Borzyskowski M (1999) Presentation and outcome of sacral agenesis: 20 years' experience. Dev Med Child Neurol 41:806–812
126. Winkler JM, Weinstein ED (1970) Imperforate anus and heredity. J Pediatr Surg 5:555–557
127. Zaw W, Stone DG (2002) Caudal Regression Syndrome in twin pregnancy with type II diabetes. J Perinatol 22:171–174

3 Genetics, Pathogenesis and Epidemiology of Anorectal Malformations and Caudal Regression Syndrome

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3.1 Introduction

Anorectal malformations (ARM) represent a complex group of congenital anomalies resulting from abnormal development of the hindgut, allantois and Mullerian duct, leading to incomplete or partial urorectal septal malformations. ARM are a relatively uncommon congenital cause of intestinal obstruction in the newborn, occurring in approximately 1 out of every 4,000–5,000 births (4.05 per 10,000 births) [1]. Approximately 36.4% are isolated lesions and 63.6% are associated with other anomalies [1]. Chromosomal defects are associated in 8% [1] and a family history may be present [2]. There are epidemiological differences in the level and extent of the abnormality. In the landmark study of 2,376 patients by Stephens, 45% were "rectal" and 53% "anal" [3].Although the current understanding of normal development and pathologic variations of ARM is incomplete, the critical period of organogenesis is understood to be at or before the 6–7th week of gestation. As a result, any aetiological defect would have to occur very early on in development [4]. The spectrum of lesions varies from fairly minor lesions (e.g. anal stenosis) to some of the most complicated urogenital lesions including anal agenesis, rectal agenesis and rectal atresia, as well as complex abnormalities, the level being determined by the relationship to the pelvic floor [5].

ARM form a significant load on the surgical services, particularly in developing countries, not only in the emergency situation but also in terms of long-term corrective procedures. Although there have been major advances in the management of these children during the last 15 years, these patients still represent a continuing challenge as a result of the significant reconstructive problems involved, as well as the fact that a significant number suffer from faecal and urinary incontinence, as well as the possibility of sexual inadequacy in later life.

3.2 ARM – Patterns of Occurrence

3.2.1 Incidence

The reported incidence of ARM is 1 in every 2,500–5,000 live births [6,7], but may be even more frequent in certain developing countries (Table 3.1) [1,6,8–25]. Although ARM comprise approximately 0.2–0.3% births [26], they have been reported to comprise up to 1.2% of reported birth defects [16]. From an epidemiological point of view, there is little clarity as to the

Table 3.1 Incidence of anorectal malformations (ARM)

Author	Year	Country	Incidence	No of patients	Population	
Keith	1908	London	1:5,000			
Ladd	1934	USA-Boston	1:7,500			
Malpas	1937	UK - Liverpool	1:3,575			
Crowell and Dulin	1940	USA - Iowa	1:5,300	28	150,354	
Moore et al.	1952	USA - Indianapolis	1:4,500	120		
Kiesewetter	1956	USA - Pittsburgh	1:1,000			
Ivy et al.	1957	USA - Pennsylvania	1:9,630			
Bradham	1958	Michigan	1:5,000	130		
Louw	1965	South Africa	1:1,800	200		
Nixon	1972	London	1:3,000			
Thomas	1977	Australia-Adelaide	1:3,160	35		
Tong	1981	Singapore	1:11,500			
Spouge and Baird	1986	Canada - Vancouver	1:2,524	273	689,118	
Boocock	1987	UK - Manchester	1:5,080	169		
Smith	1988	Australia	1:5,000	5,454		
Christensen	1990	Denmark	1: 3,333	29	96,073	
Schuler	1994	Brazil	1:8,264	121	1,000,000	Approx
Stoll	1997	France	1:2,090	108	225,752	
Naser	2000	Chile	1:1,298	54	70,242	
Niedzielski	2000	Poland-Lodz	1:2,295	30		
Cho	2001	USA Kansas	1:2,500			
Cuschieri	2001	Europe	1:2,469	1,846	4,618,840	
Sipek et al.	2004	Czech Republic	1:3,341	279	932,1253	

real prevalence, epidemiology of various anatomical types and other congenital associations, as reported series vary from unit to unit. The EUROCAT study of 1,846 recorded cases from 33 registries in Europe reported an ARM rate of 405 per 10^6 live births or 4.05 per 10,000 (range 1.14–5.96 per 10,000) [1]. In a small but well-defined area in France, Stoll et al. [23] reported 1 case of anal atresia in every 2,090 live births, or 478 per 10^6.

There appear to be distinct gender differences with respect to ARM, although at least 26% have high lesions, 10.7% intermediate and 57.2% low abnormalities [27] with variation between boys and girls. The Surgical Section of the American Academy of Pediatrics study of 1,142 cases (1965–1969) [28] reported a higher incidence of low lesions (19%) in females, whereas >50% of affected males had "high" lesions. In the EUROCAT studies, high lesions also appeared to be more common among boys (34.5 vs 13.3%) and low lesions more common among girls (70.9% vs 47.6%) [1,29].

The majority of epidemiological data include both isolated lesions and those associated with other anomalies. This collective grouping fails to highlight the characteristics associated with ARM alone without interference from other congenital anomalies. When studied separately, the differences between isolated lesions and those associated with other anomalies may be marked [1].

3.2.2 Geographic Variations

International variation occurs not only in the incidence of ARM but also in individual types, and certain geographic subtypes may occur. There appeared to be inter-registry variation even in Europe from 1.14 to 5.96 per 10,000 [1]. In addition, there appear to be fluctuations between individual years in certain geographical areas [17]. Although these variations may be on the basis of genetic, environmental and socio-economic differences, it is well recognised that genetics may have a significant role to play.

Geographical differences have been observed previously and ethnic differences appear to exist [21,30,31]. It is possible that there is a higher incidence of isolated lesions in developing countries as opposed to those associated with congenital malformations and syndromes. There may also be a higher incidence of low lesions in these populations.

There a body of evidence to show that ARM form a significant clinical burden in Africa [13,32]. Louw [33] reported an overall incidence of 1:1,800 live births in Cape Town in 1965, which was considerably higher than the 1:3,000–5,000 reported from Europe and the North Americas during the same period. He reported an incidence of 1:1,740 in Caucasians, 1:1,770 in coloureds and 1:2,260 in black African live births [33]. The slightly lower incidence in black African patients in his study may be arguably somewhat skewed by demographic factors as well as the governmental policies pertaining to that particular chapter of South African history. Nevertheless, the reported incidence was higher than in other reports from the same period, indicating that ARM may possibly be more common in Africa. In support of this, Shija reported 46 patients seen in a 2-year "sabbatical" in Zimbabwe [32].

At first glance, there is little objective evidence that ARM are more common in black ethnic groups. ARM have been identified as the third most common cause of neonatal intestinal obstruction in one series in Nigeria, representing 13.4% of congenital malformations [34]. In another study it represented 20% of congenital malformations [35]. ARM represented 67% of emergency surgery in neonates in a further study [36]. This suggests that ARM represent a significant burden of disease in Africa, which could represent an expression of either the high birth rate or a higher incidence. There is, however, a great paucity of other objective data from the African continent as to the types, frequency and incidence of ARM encountered. Demographic and clinical factors appear to be associated with incidence, and comparisons between population groups and differences in associated anomalies may exist between population groups. By way of example, the 44.4% of low (anal) lesions reported by Archibong and Idika [37] appears lower than the reported world experience [38]. It is also lower than that reported by Louw in a multiethnic community in Cape Town [33], where 58% were anal by definition. Although some of these differences may be explained on the basis of a lack of a standardized nomenclature in various periods of history, it illustrates the need for more objective data from developing countries to assess geographical differences. A multicentre study is currently underway to examine the epidemiological prevalence of ARM in Africa.

3.2.3 Ethnic Groups

ARM appear to have a worldwide incidence and have been reported from most countries, but there is a great paucity of information as to the relative incidence of ARM between ethnic groups. Smith [7] reviewed the reported incidence and suggested that factors such as definition and inclusion may account for many of the variations observed. In addition to variations in overall incidence, ethnicity and genetic influences may result in individual variations and different patterns of disease may become evident on further study. Anal atresia has been said to have a low incidence in black patients [7,39], but other studies have shown no clear-cut distinction in incidence in black patients and a measure of under reporting could be present. By way of example, the study by Harris et al. in California showed that the observed increase in gastrointestinal atresias was not reflected in anal atresia in black patients [40]. Despite early suggestions to the contrary, there is an unsubstantiated view that ARM are in fact more common in the African population [7,13,33], a view that requires further investigation.

3.2.4 Sex Distribution

In general, the male:female ratio associated with ARM is almost equal, with a 56:44 male:female ratio previously reported in large collective series [7,27] as well as in the EUROCAT study of isolated ARM (male:female ratio = 1.06) [1]. Exceptions to this are anal atresia (male preponderance) and ectopic anus and congenital anal fistula (female preponderance) [29]. In addition, the presence of a fistula appeared to make a difference; a much higher male preponderance was reported in anal atresia without fistula than in those with a fistula ($p < 0.0001$) [1]. This was also shown to be true of both supra- and infralevator lesions. As far as infralevator lesions are concerned, associated anomalies are mostly to be found in male patients, females being largely spared. A male preponderance was also demonstrated in isolated ARM and cloaca, and a marked female preponderance was found in patients with ectopic anus and perineal fistula [1].

Additional anomalies have been reported to occur 13 times more frequently in patients with a high as opposed to a low ARM [41]. In the same study from India, males were said to have a 4:1 incidence of associated anomalies than girls. An equal sex distribution has been reported in patients with ARM with a fistula, but certain other differences have reported between the sexes, which will be discussed below.

The obvious explanation for gender variations, prior to structural or hormonal differences, must lie in the chromosomes themselves – particularly the Y chromosome [42]. On the other hand, geographical differences may exist in the sex incidence, suggesting that other factors also play a part. By way of example, it has been suggested that in Africa there is a preponderance of females with low lesions [37]. These differences should be verified and their aetiology explored.

3.2.5 Familial Associations of ARM

ARM have been reported to recur in families, suggesting a genetic component in their aetiology [43,44]. No real association between birth order, maternal age and relationship to parity has been established, although a slight preponderance among first-born infants has been suggested [7]. Despite the presence of several early reports of familial recurrence [2,44–49], Smith [7] suggested that heredity plays a minor or insignificant role in ARM due to the low familial incidence [50]. There appears to be a low rate of association in families (ranging somewhere between 2.4% [43] and 8% [2]) , but some appear to have an autosomal dominant inheritance pattern. The form of inheritance appears to be autosomal recessive, although X-linked inheritance has been described [50,51].

Consanguinity has been identified as having a higher incidence of ARM [2,16,48], particularly in countries in the Gulf and Middle East regions. ARM have been reported in three generations within a family from this region [52] and syndromic features may be more common [53]. In addition, familial associations of the Currarino association are well established [54] and family members have been shown to have sacral anomalies without the full syndrome [55].

Twin studies also seem to suggest that inheritance plays a fairly minor role. On the other hand, Scandinavian studies have reported an incidence of approximately 3 per 10,000 twin pairs [56] and at least 5 cases have been reported in monozygotic twins [57]. ARM are also common in ischiopagus conjoined twins.

3.2.6 Chromosomal and Genetic Associations of ARM

In an additional 8% of patients, genetic factors are clearly associated with ARM. These include several chromosomal and multiple congenital anomaly syndromes, which include Towne-Brock syndrome [58], FG syndrome [59], Kaufman-McKusick syndrome [60] and Lowe syndrome [61]. In addition, ARM have been described in association with trisomy 8 mosaicism, Down and fragile X syndromes [2,38] and the OEIS (omphalocele, exstrophy, imperforate anus, spinal defects) complex [62]. An extra copy of 12p was identified in an infant with Pallister-mosaic syndrome with ARM [63].

One of the most interesting associations is with Down syndrome [of the order of 2.2% [64] to 5.1% [27]. In the latter large Japanese cohort, Down syndrome had the highest incidence of females with a covered anus (73%) . Similarly, girls with associated anomalies but no fistula had a significantly higher incidence of Down syndrome than boys without a fistula ($p < 0.001$) [27].

Other congenital anomalies such as cardiac, Hirschsprungs disease (HSCR) [65] and other gastrointestinal atresias [66], raise interesting questions with regards to potential genetic associations.

3.3 Descriptive Epidemiology of ARM

3.3.1 Supralevator Versus Infralevator ARM

There appear to be distinct differences in the epidemiology of isolated cloaca, supralevator, infralevator anal atresia (with or without fistula) , ectopic anus and congenital anal fistula [29]. It is possible that differences in pathogenetic pathways can be identified for supralevator as opposed to infralevator lesions. For instance, isolated lesions only had a 10% incidence of supralevator lesions as opposed to a much higher incidence within the syndromic group [1] and those with additional anomalies [29]. This is in contrast to the 26% incidence of supralevator lesions reported in a large Japanese study [27]. Supralevator lesions were more common in multiple congenital abnormality syndromes associated with ARM [25]. In a large study from India, additional anomalies were 13 times more frequently associated with high lesions than with low lesions [41].

3.3.2 The incidence of Fistulas

There also appears to be a difference in the incidence of fistulas occurring in supra- and infralevator lesions, with fistulas being identified in 53% of isolated supralevator lesions and in only 37% of infralevator lesions [29]. Early studies showed a higher rate of fistula than more recent reports [29].

The fistula in ARM is now broadly accepted as an ectopic anal canal and remains the one defining feature between groups of ARM. In animal models, the fistula appears to represent failure of the urorectal septum to fuse to the cloacal membrane [67]. It is therefore interesting that sex differences are also seen in relation to the presence of fistulas. The Surgical Section of the American Academy for Pediatrics study reported a 72% incidence of fistula in boys and a 90% incidence in girls [28]. The anatomical and embryological differences between the genders is an obvious potentially significant factor in the pathogenesis of these fistulas.

In a large Japanese cohort, males with fistulas were divided into rectourethral (adjacent to the ejaculatory duct 40%) and rectobulbar entering the bulb of the urethra below the urogenital diaphragm [27]. Rectourethral fistula was identified in 81.4% of patients with high lesions; these authors suggested that recto-urethral fistulas should be classified into subgroups depending on the level of the blind-ending rectum.

The presence of a fistula also appears to affect outcome; a higher incidence of foetal death was encountered in patients without a fistula [1].

3.3.2 Associations with Other Anomalies or Syndromes

3.3.2.1 Isolated ARM

The epidemiology of these isolated anomalies appears to differ somewhat from those associated with other abnormalities and syndromes [1]. The study of isolated anorectal lesions identifies those characteristics that can be associated with ARM alone without interference from other congenital anomalies [1]. In particular, the incidence of supralevator and infralevator lesions and of fistulas, as well as ectopic anus, congenital anal fistula and cloacal persistence varied between the two groups. In the EUROCAT study, isolated lesions only had a 10% incidence of supralevator lesions as opposed to a much higher incidence within the syndromic group, suggesting that associated anomalies occur more frequently with high lesions. This is in keeping with that reported by Louw [33], where a 60% incidence of associated anomalies was associated with anorectal agenesis, as opposed to a 30% incidence in anal malformations. Also in keeping with this, a 13 times higher incidence of high lesions has recently been reported with associated abnormalities [41].

A male preponderance was demonstrated in isolated ARM and cloaca, an equal sex distribution in patients with ARM with a fistula and a marked female preponderance in patients with ectopic anus and perineal fistula [1]. In isolated ARM, the most common associations are genitourinary, spinal and extremity anomalies, congenital heart disease and gastrointestinal and esophageal atresia [28,68]. Hypospadias seems to be one of the most common genital anomalies [68].

3.3.2.2 ARM Associated with Other Anomalies and Syndromes

Incidence

Early studies stressed the fairly high risk of associated congenital anomalies in ARM, the incidence ranging between 28 and 72% (Table 3.2) [13,15,25,32,68–72]. Although post-mortem studies have demonstrated an extremely high (97 and 94%, respectively) incidence of associated anomalies [15,73] this can probably be partly attributed to patient selection, as those with multiple anomalies are most likely to succumb to the condition.

Since 1990 there have been several reports that have included careful identification of associated anomalies. One UK report suggested a 53% incidence and a Japanese study of 1,992 cases a 45.2% association [27]. In the more recent Eurocat study, a 64% incidence was identified [1]. Several more reports suggest a significant association (Table 3.2) [6,16,41,74–76]. The reported occurrence of associated anomalies probably depends on how extensive the investigation of the patient has been. It may therefore be higher in areas of high resources and much lower in developing countries.

Epidemiology of Associated Lesions

The epidemiology of associated anomalies may be further affected by the level of the lesion as well as the sex of the patient. Associated anomalies are important as these are not uncommon causes of death and may determine quality of life in survivors. Abnormalities can be broadly classified into minor and major anomalies as well as chromosomal and malformation

Table 3.2 Anomalies associated with ARM

Author	Year	Country	ARM (*n*)	No associated abnormality	Incidence (%)	Notes
Ladd and Gross	1934	USA	214	60	28	
Lee	1944	USA - Madison	16	29	69	
Bacon and Hering	1948	USA	98	16	30	
Norris et al.	1949	USA Los Angeles	52	24	46	
Mayo and Rice	1950	USA- Rochester	165	63	44	
Moore and Lawrence	1952	USA - Indianapolis	120	86	97	Postmortem
Louw	1965	South Africa	200	105	52	
Taneja	1970	India - New Delhi	74	38	49	
Hasse	1976	Germany - Berlin	1,420	592	42	
Tong	1981	Singapore	49	20	31	
Shija	1986	Zimbabwe	46	12	26	
Boocock and Donnai	1987	UK - Manchester	169	84	53	
Endo	1999	Japan	1,992	896	45	
Chen	1999	Taiwan	108	63	58	
Naser	2000	Chile	54	31	59	
Cho	2001	USA - Kansas	103	73	71	
Cuschieri	2002	Europe	1,846	1,181	64	
Mittal	2004	India	140	83	59	
Chalapathi	2004	India - Chandigarh	125	35	28	
Ratan	2004	India - Haryana/Delhi	416	241	58	
		Total	6,876	3,430	Mean 50.45%	

syndromes, associations or sequences. In the study reported by Louw [33] the associated anomalies were not only found more frequently in association with high lesions, but were more severe. Clinical groups evident in the EUROCAT study of associated anomalies [29] included the following:

1. Syndromes of known cause (including monogenic, teratogenic and chromosomal causes)
2. Recognised syndromes and sequences
3. VACTERL (mnemonic: Vertebral anomalies, Anal atresia (no hole at the bottom end of the intestine), Cardiac defect, most often ventricular septal defect, TracheoEsophageal fistula (communication between the oesophagus and trachea) with esophageal atresia (part of the esophagus is not hollow) , Renal (kidney) abnormalities and Limb abnormalities, most often radial dysplasia (abnormal formation of the thumb or the radius bone in the forearm) associations
4. Multiple congenital anomalies (MCA) with two or more anomalies without a recognisable pattern

Associated anomalies occur frequently and range from 36.4% (672 cases) in the EUROCAT study [29] to 45.2% in another large series (Table 3.2) [27]. Although these would appear to be higher in supralevator lesions as opposed to low lesions [25–41], it is of considerable interest that low vaginal anomalies in females appear to have as high an incidence of associated anomalies as high rectal lesions [13,73].

ARM may be seen to represent not only localised lesions, but may also be part of a broader spectrum of field defects. As a result, the epidemiological characteristics of those occurring with other associated anomalies or as part of other syndromic phenotyopes are of considerable interest. The EUROCAT study has shown at least 15% of cases to be associated with chromosomal variations (monogenic or teratogenic syndromes) [29]. Twins (including conjoined twins [77]) were more frequent in those with chromosomal sequences, VACTERL or multiple anomalies than in isolated or monogenetic associations [29]. Those patients with syndromes or MCA were associated with a significantly lower birth weight and significantly higher incidences of foetal death or termination of pregnancy. Syndromes were mostly associated with high or supralevator lesions, whereas in low lesions, associated anomalies were mostly identified in male patients, females being largely spared. Cloacas only represented 0.9% of the total [29]. Of the remainder, 60.2% had multiple anomalies, 15.4% were associated with VACTERL syndromes and 9.3% with sequences. A considerable overlap was reported between those with VACTERL associations and those with MCA.

One of the problems in ARM epidemiology is that the lesion may be hidden among other multiple anomalies in syndromes, some of which are life threatening. The ARM may then not be properly identified and classified. By way of example, a South American study of more than 1 million births in 11 countries [78] identified 121 (8.5%) of 1,428 babies with multiple anomalies that included anal, renal and genital anomalies. Three or more VACTERL associations were seen in 21 of these cases (17.4%) . A second group are encountered as part of a complex group of multiple anomalies, which include exomphalos and the OEIS complex, bladder exstrophy and colonic anomalies among others. Cuschieri [29] reported a high incidence of exomphalos, half of which were associated with cloacal exstrophy. In one study urinary tract anomalies were identified in 42.5%, skeletal in 26% and the cardiovascular system in 18.5% [16]. ARM have also been reported in association with malrotation [79], and with Pallister-Hall [80], Currarino [81] and Down syndromes [65,82].

Congenital anomalies may be less common in developing countries, occurring in only 27.9% of cases in one African study [83]. The question of under-reporting is a real issue in many developing countries, with the result that direct comparisons are difficult.

3.3.2.3 Individual Systems with Associated Anomalies

Urogenital Anomalies

Urogenital associations are one of the more common associations seen in ARM and occur in 20–54% of cases [84–87]. In the tertiary referral series by Peña, a 48% overall incidence was reported [88], being 14% in low lesions. In other series the incidence was as high as 54% in supralevator lesions [68] and 90% in cloacas [89]. Anomalies may affect the upper and lower urinary tracts. In a study of 162 female patients, [90] the urinary tract was involved in 40% overall, with 25% of these associated with the upper urinary tract. Unilateral renal agenesis and hydronephrosis were the most common lesions. Renal tract anomalies were mostly associated with communicating as opposed to non-communicating lesions (29.6% vs 10.7%) . In 51%, the anomalies were located outside of the genitourinary tract. Genital defects are most common where anorectal and renal anomalies co-exist [91], but are generally excluded from the VACTERL association and have previously been included in certain series [22,91,92].

Urinary tract anomalies occurred in 25.6% in one study from a developing country, occurring more frequently in high lesions [93]. These included genital anomalies (14%) and vesicoureteric reflux in more than half of them.

The VACTERL Association

The association of ARM and the VATER syndromes by Quan and Smith in 1973 [94] was expanded to include limb deformities in the VACTERL association soon afterwards [95,96]. VACTERL has been reported in 1 out of every 5,000 live births and appears to represent a developmental "field defect" [97]. Most cases are sporadic with a recurrence risk of 1%, familial cases being rarely reported [98,99].

The aetiology of VACTERL is largely uncertain, although a considerable body of evidence suggests that it is genetic in origin. Anomalies of the ribs and lumbosacral vertebrae are not uncommon associations of ARM and can be reproduced in animal models, suggesting a common notocordal pathogenesis [100]. Studies of the adriamycin animal model have linked it to defective sonic hedgehog (Shh) signalling [101,102]. Kim et al. [101] found that although there was significant association with the sonic hedgehog pathway, it did not explain all cases and speculated that environmental factors such as teratogens possibly trigger the event. This is supported by animal models utilizing a variety of toxic chemicals [103].

The associations differed markedly with different types of ARM lesions [29]. Although VACTERL only contributed 15.4% of the total associated anomalies, more than 50% of patients with MCA displayed two or more VACTERL associations, in more or less the same frequency as in the full syndrome itself. Three or more VACTERL associations were observed in 37.43% [76], which suggests a common pathogenetic pathway.

In a report from the International Clearing House for Birth Defects Monitoring Systems (representing 17 major registries worldwide, and reflecting more than 10 million newborn infants) , 286 out of 2,295 cases with multiple anomalies had 5 or more VACTERL associations [91]. In 74.8% of these, additional defects were identified, significantly genital and small bowel atresias ($p < 0.001$) . It is clear that VACTERL associations are partly the result of genetic factors, and animal models exist [104]. The association of VACTERL with craniofacial anomalies and sirenomelia suggest overlapping developmental pathways [105]. Mutation of the Fanconi anaemia complementation group C gene (FAC) has been reported in twins in association with VACTERL and hydrocephalus [106].

Apart from VACTERL associations, other associations may occur and the OEIS complex associates bladder exstrophy with exomphalos, ARM and spinal anomalies [107], which include the VACTERL associations (Shh signalling) [104,108] and the Currarino Triad [108–111]. Both of these have been extensively researched via animal models, and defects of the 22q11.2 site as well as exon 1 deletions (1q41-q42) in the homeobox gene HLXB9 have been described [109]. It is thus clear that genetic mutations in the sonic hedgehog and homeobox genes may result in caudal mesodermal maldevelopment, particularly in association with other abnormalities in syndromes.

Cardiovascular System Anomalies

Although part of the VACTERL association, cardiovascular associations occur in approximately 9% of patients [73]. Greenwood et al. [112] found a 14.9% association in his series of 222 patients. A further literature review revealed cardiovascular anomalies in 17 out of 1,898 patients. The reported spectrum of lesions includes atrial and ventricular septal defects, tetralogy of Fallot, truncus arteriosis, transposition of the great vessels, infundibular stenosis and aortic coarctation [73].

Gastrointestinal Associations

Gastrointestinal anomalies are relatively uncommon but have been reported in as many as 10% (25/246) of patients [73]. Apart from the VACTERL association with oesophageal atresia, they include malrotation [113], HSCR [80,81,114,115], gastrointestinal duplications [116] and duodenal obstructions [73]. Malrotation is of particular interest because of its reported association with ARM [79] and the hedgehog signalling systems, which are known to be affected.

One of the most significant of the gastrointestinal associations occurs with HSCR. It is generally accepted that the association between HSCR and ARM is uncommon [39,82] but may be under-reported (being confused with anal stenosis if there is a very short aganglionic segment) . In a study of five North American centres, Kiesewetter [39] reported a 3.4% incidence. In a further large collective series of more than 1,200 HSCR cases, a 2.5% association with ARM was reported [117]. It was also reported in nine cases from a single centre over a 10-year period in another series [118]. ARM and HSCR has been recorded in two siblings of consanguineous parents [119] and has also been associated with trisomy 21 [65], suggesting some genetic association. The association remains uncommon, however, and in our series we encountered only 1 out of 408 cases [120,121], which is in keeping with several other reports [69,73]. If HSCR is present with ARM, it may lead to diagnostic delay because of the initial diagnosis of the ARM and the fact that the dysfunctioning colostomy is proximal to the affected bowel.

Vertebral and Spinal Cord Anomalies

Animal studies suggest that the notocord controls the development of the spinal cord, vertebral column and anorectum and appears pivotal in the development of ARM [104]. As a result, ARM are also commonly associated with underlying vertebral and spinal cord anomalies, which include the hemisacrum of Currarino triad. In one study of murine ARM embryos, the neural tube was observed to form an anomalous, irregularly branched mass in the sacral region [122] in the presence of normally developed pelvic musculature. A further association is with a "tethered" spinal cord in cases of ARM. In one study of 55 patients with tethered cord in Japan, 10 (18%) had a high ARM [123].

There is considerable variation in abnormalities in sacral development, but a sacral ratio of less than 0.52 can be considered pathological [124]. Currarino triad links sacral agenesis with ARM and includes a presacral mass (teratoma) , partial sacral agenesis (hemisacrum) and anorectal defects. Currarino syndrome has been associated with haploinsufficiency of the HLXB9 gene [111,125,126], but there is considerable variation in penetrance [127] Other tumours such as sacrococcygeal teratoma have been described in association with a low lesion [128] and malignant degeneration of the presacral teratoma has been described in a familial case [129]. Seri et al excluded the sonic hedgehog pathway as being responsible for the Currarino syndrome [130]. Subsequent linkage of Currarino syndrome to chromosome 7q36 [131,132] and the HLXB9 homeobox gene [111] shed further light on its pathogenesis (see Chap. 2).

Other Associations

Other skeletal anomalies occur and limb defects tend to be pre-axial in nature [133]. Syndactyly has been reported in association with other craniofacial anomalies [134]. The morphogenesis of the internal anal sphincter has been related to the Hoxd-12 and Hoxd-13 genes in animal models [135], which overlap with digit development in the foetus, thus providing a potential link to digital anomalies.

Association with craniofacial syndromes such as Apert and Pfeiffer syndromes may be related to fibroblast growth factor receptor gene variations [134]. This is of particular interest due to recent experiments where Fgf10 invalidation produced ARM [136].

Further associations with nasal and renal anomalies in four consanguinous siblings may represent some autosomal recessive syndrome [53]. Choanal atresia has also been associated [137]. The cat-eye or Schmid-Fraccaro syndrome has been described with ARM and provides a further link to chromosome 22 [138]. Other associations include Stratton-Parker syndrome in association with growth hormone deficiency [139].

Associations with Gastrointestinal Innervation

In addition to spinal cord associations, aberrations of the enteric nervous system (ENS) have been demonstrated in both animal models [140] and humans, which may be partly responsible for postoperative dysfunction. Mostly nerves staining for vasointestinal peptide, SP-100 and neurone-specific enolase showed marked reduction in the rectum and fistulous tract of high lesions in ethylenethiourea-treated rats, thus giving possible explanations for postoperative colonic dysfunction [140]. In humans, several reports have indicated disturbed innervation in the affected segment of bowel, being as high as 81.82% in one study [141].

The association with HSCR [39,60,69,81,114,115,119] suggests a connection with the major susceptibility genes for that condition (*RET* and *EDNRB*) . Although no known associations with *RET* mutations are known, there is a reported association between ARM and chromosome 13 (the "13q syndrome" [115], suggesting that the long arm of chromosome 13 (and thus *EDNRB*) is associated with anogenital abnormalities. *EDNRB* mutations have also been identified in 60% of patients with penoscrotal transposition [142].

Early studies suggested association between vascular events and rectal stenosis [109]. More recently, an association with thalidomide and ARM [110] has been reported, and in the light of its known anti-angiogenic properties, it reopens the possibility of a vascular association. The vascular hypothesis for ARM has been explored since the 1960s. Correlation between ARM and the origin of intestinal atresia [104] has yielded conflicting results, suggesting that the origin of ARM, with the exception of isolated to atresia, is not associated with vascular malperfusion during embryogenesis. On the other hand, other vascular malformations are not uncommon in anorectal malformations [143,144]. In addition, retinoic acid, a well known modulator of the endothelin system, has been shown to induce caudal regression syndrome and ARM in a mouse model [145], thus adding support to the angiogenesis theory. The overlap in spatiotemporal expression in the developing gut mesoderm between retinoic acid receptor beta, cellular retinal binding protein 1, CRBP1 and Hox b5 and c-ret supports the hypothesis that retinoic acid is involved in the neuromuscular development of the gastrointestinal tract [146]. As such it can affect both innervation and embryologic development. The pertinent question is therefore whether genetic factors influence the vascularisation, development and differentiation of smooth muscle and nerves, which may be among the most significant events in the aetiology of ARM.

3.4 Associations with an Increased Genetic Risk

The application of epidemiologic techniques to study genetic risk factors is growing rapidly due to major advances in our understanding of the human genome. This opens the door to the study of genetic risk factors and environmental factors as part of genetic epidemiology. ARM are generally accepted as being a largely genetically based disorder at a cellular level. From an epidemiological point of view, it could be argued that either the genetic influences pertain to only a few cases (particularly those with recognized clinical syndromes) , or alternatively they could be more subtle and broad-based defects in genetic influences on signalling pathways and therefore implicated in the pathogenesis of ARM in general.

Many of the associated syndromes appear to have a complex aetiology and it may be that several different signalling pathways are involved in producing field defects. Developmental field defects would then include the coming together of associated pathways and sequences during blastogenesis [147].

The causes of ARM are unknown, but it is generally understood that development of the anorectal region appears to depend upon the normal development of the terminal portion of the hindgut, the critical period of organogenesis being at or before the 6th or 7th weeks of gestation. Abnormal development would have to commence in early embryogenesis, possibly due to the limitations in the dorsal portion of the cloacal membrane resulting in persistent attachment of the hindgut to the urogenital sinus, resulting in the associated fistula [148]. As such, the aetiology of ARM is probably multifactorial and may include both genetic and environmental factors.

Despite the paucity of information on the genetic associations of ARM in humans and a fairly low familial incidence, the probability of a genetic association has increased due to the association with other chromosomal abnormalities and syndromes [58,60,61,80]. In addition, Scandinavian studies suggest an increased incidence of chromosomal anomalies [56]. There is also a considerable body of evidence from animal experiments to substantiate the genetic associations [104,108–111]. Among others, the VACTERL association has been linked to defective Shh signalling (*22q11.2q)* [104] and the related *Gli2,Gli3, ATRA* pathways [102,122].

Several other cytogenetic deletions have been reported between the 7p36 site and the hemisacrum of Currarino syndrome [109–111]. In the female, it has been shown that the p63 signalling pathway is important in the development of epithelial stromal signalling, urorectal septation and modelling of the external genitalia [149], thus opening a further area for investigation. The significance of the ephrin pathways and complete lack of cloacal septation in the animal model [150] needs to be further investigated. Furthermore, Fgf10 invalidation in experimental animals results in a genetically reproducible ARM [136]. One further significant candidate in this regard is endothelin-1 and its action via the B receptor gene, which will be discussed later.

Previous work has shown that the sonic hedgehog system (which induces mesodermal gene expression) is required for the normal development of mid-axial organs including the developing gut [103,104]. It is thus involved in the pathogenesis of the VACTERL phenotype. Shh null mutant mice have persistent cloaca [102], whereas the Shh-responsive transcription factors Gli2- or Gli3-deficient mice demonstrate ARM with fistula [151]. Although it therefore appears that mutations in Shh signalling are involved in an animal model phenotype that mimics human ARM, this does not necessarily provide an explanation for the full clinical spectrum.

In addition to Shh, B-subclass ephrin and ephrin molecules have been linked to midline cell–cell adhesion and fusion events [150]. Ephrins are a family of membrane-bound proteins involved in neural guidance during ENS development [152] that are possibly mediated by Jak/Stat proteins. A complete lack of ephrin B2 reverse signalling has been reported to result in the complete lack of cloacal septation in an animal model [150].

Martinez-Frias [153] explains the differences between isolated ARM and those associated with other anomalies by suggesting that the entire field is a pathogenetic unit and responds in the same manner to the effect of different aetiological factors, which include genetic predisposition and environmental trigger factors. The timing of the effect could explain an isolated or syndromic phenotype.

In addition, it is possible that some common genetic background may be shared with HSCR susceptibility genes [80,81,115]. There are further associations with pig chromosome 15 [154]. As a result, a more modern understanding of genetic influences on signalling pathways reopens the debate. Initially, ARM were thought to result from a vascular accident or insufficiency during development. This idea is supported by some early studies that suggested the existence of a vascular component, particularly in low malformations [155]. Early studies in experimental animal models produced only rectal stenosis and atresia [156]. Although further supported by certain anatomical [157] and post-mortem studies in animals [158], an ischaemic hypothesis appears insufficient to explain the full spectrum of observed related conditions, particularly those associated with other abnormalities and known clinical syndromes. Nevertheless, animal models not uncommonly demonstrate a single umbilical artery and/or radial artery hypoplasia [143,144]. In addition, studies noting an abnormal blood supply in lesions such as isolated rectal atresia or the Indian H-type lesions [159,160], further suggest some measure of vascular compromise.

Reported associations with thalidomide, a well-known anti-angiogenesis drug [161] as well as an association between retinoic acid and caudal regression [145] lend further support for a vascular/angiogenesis hypothesis. In addition, the adriamycin/ethylenethiourea (ETU) animal model [101,104,153] suggests that toxic effects may be a trigger for disrupted genetic pathways. The way seems open to explore a potential combination of genetic and vascular influences as well as a genetic-based vascular hypothesis for ARM.

There appears to be some additional evidence that endothelins may be influential in the pathophysiology and possible development of ARM. A potential link exists to chromosome 13 (and thus endothelin receptor type B, EDNRB) exists via associations with the “13q syndrome” and penoscrotal transposition, with a 60% ARM association [115]. *EDNRB* mutations have also been identified in 60% of patients with penoscrotal transposition [142]. Other potential links to EDNRB include those syndromes where ARM are associated with HSCR (Kaufman-McKusick) [60], Pallister-Hall [80] or sensorineural deafness (Towne-Brock [58] and Lowe [61] syndromes) where a common genetic background may be postulated.

Endothelins have an established role in the proliferation, differentiation and migration of neural crest cells [162–164]; enteric neuroblasts require an intact endothelin-3/EDNRB system to develop normally [165]. EDNRB relates to all three endothelins (ET-1, ET-2 and ET-3) [166–168], and its role in neural cell differentiation appears to be an anti-apoptotic one [169], whereby it may prevent early neuroblast differentiation. It appears that EDNRB inhibition results in impairment of DNA repair and a decreased resistance to pro-apoptotic signals [169].

Endothelins are also involved in angiogenesis [170], and it is clear from recent animal studies that the exceptional circulatory physiology of the newborn is dependent upon the endothelin-controlled behaviour, which is not the case 1 month after birth [171]. In the study by Nankervis, the mesenteric arteries of newborn pigs demonstrated significant diameter changes in response to the blockade of endogenous nitric oxide production or blockage of endothelin receptors (EDNRB) compared to 1 month later. In addition, hypoxia-ischaemia experiments on 14-day-old sl/sl rats have shown that ETB-deficient animals have an increased susceptibility to in vitro hypoxia, with a significant decrease in surviving neuronal cells [172]. In further experiments with endogenous "rescued" ETB receptor-knockout mice, Murakoshi et al. [173] showed that chronic inhibition of the ETB by the receptor antagonists A-192621 was harmful to vascular remodelling following injury. This is still further supported by the reported inhibition of EDNRB, resulting in induction of VEGF expression in melanoma cells [169]. These actions of endothelin suggest that it plays a major role in development, possibly as a regional morphogen (perhaps via co-coordinated control of genes and signalling pathways) and possibly in conjunction with *GDNF* as well as laminin-1 downregulation.

In our own study of 14 children (6 males and 8 females) with ARM [174], we showed mobility shift aberrations and variations in the *EDNRB* gene of all patients with ARM, which included one previously described polymorphism in exon 4 (831G/A) previously reported in association with HSCR. Six novel polymorphisms were identified in exons 1 (178G/A), 2 (552C/T and 561C/T) and 3 (702C/T) in patients with ARM. Analysis of the total patient group with non-syndromic ARM compared to controls revealed statistically significant differences for the polymorphism 178G/A ($p < 0.01$, χ^2 with Yates correction = 8.24) , which was identified in 3 out of 4 affected individuals (75%) compared to 1 out of 84 (1%) control samples. The genetic variations encountered in this study also appeared to correlate with the level of the lesion. By way of example, the polymorphism identified in exon 1 (178 G/A) was present in 3 out of the 4 (75%) low lesions, but not the high or intermediate lesions. A different exon 3 (702C/T) single nucleotide polymorphism was present in 3 out of 5 (60%) of the supralevator lesions. Further research is required to establish the validity of the hypothesis that EDNRB plays a major role in the pathogenesis of ARM and to understand its mode of action.

3.5 Environmental Factors

3.5.1 The Effect of Drugs in Pregnancy

Although there is very little objective evidence of drugs in the pathogenesis of ARM, Stoll et al. [23] suggested from their relatively small sample that mothers of children with congenital anal atresia took drugs more often during pregnancy than did controls. Previous reports have implicated thalidomide [161] and tridione as possible aetiological agents [2]. More recently, Bonnot et al. [175] have reported 6 cases of anal atresia out of 262 congenital malformations identified in 13,703 patients exposed to benzodiazepines during pregnancy. They then demonstrated a significant association ($p = 0.01$) between lorazepam and anal atresia (five out of six patients)

3.5.2 Associations with Toxins

The known associations with toxins have been used to develop animal models as well as indicate possible involved pathways. The adriamycin-exposed animal model is well established [103] and ETU toxicity is the basis of a current animal model of ARM [104].

Retinoic acid is thought to be involved in the development of patterns in the developing embryo. Teratogenic doses of retinoic acid results in truncation of the embryonic body axis in the mouse, which corresponds to caudal regression syndrome as described in humans [146]. All-transretinoic acid has been used as an animal model of high ARM with fistula, suggesting that it interferes with the normal caudal migration along the urogenital system posterior wall [176].

In a further study on the effects of retinoic acid in the developing foetus, 100% of offspring had craniofacial anomalies, 94% anorectal, 90% limb and 55% neural tube defects [177]. Etretinate (which targets the tail bud) produces a mouse model of Currarino syndrome [178]. Although it is not clear at this stage whether this is a toxic effect or has to do with blocking of receptors during normal development, this association appears to indicate that (retinoic acid and other drug) teratogenesis affects the neural crest developmental pathway. Retinoic acid is of particular interest as it appears to inhibit Shh signalling and downstream bone morphogenetic protein 4 synthesis [179].

An association with thalidomide [161] has been reported, which reopens the possibility of a vascular component to the aetiology, because of its recently discovered anti-angiogenic properties. This raises the possibility of a genetically based vascular hypothesis for ARM, which may combine the two hypotheses.

3.5.3 Association with Infectious Agents

Associations with infectious agents remain uncommon, but foetal exposure to cytomegalovirus and toxoplasmosis have been reported as possible aetiological factors [29].

3.5.4 Environmental Exposure and ARM

The results of epidemiological studies are inconsistent as far as environmental exposure is concerned, but only weak potential associations with risk factors have been identified. The adverse effects of smoking, previous abortions and cycle disorders before pregnancy could not be established [180].

Stoll et al. [23] suggested that in their relatively small sample, fathers of ARM babies were more exposed to hazardous substances than fathers of controls. Other environmental factors such as exposure to electromagnetic radiation have proved to be difficult to determine from an epidemiological point of view.

3.5.5 Environmental Versus Genetic Factors – Increased Susceptibility to Environmental Factors

Epidemiologic techniques are not infrequently used to study risk factors and interactions between gene susceptibility and environmental factors. The role of environmental factors in the development of ARM is probably small. Events during pregnancy or parental exposure (maternal or paternal) could theoretically be of significance and further research is clearly warranted. A clear distinction between environmental and genetic factors is not necessarily justified, as demonstrated in the concept of ecogenetics [181].

Although a considerable spectrum of anorectal maldevelopment has been shown to result from the toxic effects of ETU administration to timed-pregnant rats [104], another study demonstrated that discordance among twin animals suggested that teratogens could be eliminated as a major aetiological factor [182].

References

1. Cuschieri A (2001) Descriptive epidemiology of isolated anal anomalies: a survey of 4.6 million births in Europe. Am J Med Genet 103:207–215
2. Boocock GR, Donnai D (1987) Anorectal malformations: familial aspects and associated anomalies. Arch Dis Child 62:576–579
3. Stephens FD (1970) Embryologic and functional aspects of "imperforate anus". Surg Clin North Am 50:919–927
4. Kluth D, Hillen M, Lambrecht W (1995) The principles of normal and abnormal hindgut development. J Pediatr Surg 30:1143–1147
5. N'Guessan G, Stephens FD (1986) Covered anus with anocutaneous fistula: the muscular sphincters. J Pediatr Surg 21:33–35
6. Cho S, Moore SP, Fangman T (2001) One hundred three consecutive patients with anorectal malformations and their associated anomalies. Arch Pediatr Adolesc Med 155:587–591
7. Smith ED (1988) Incidence, frequency of types, and etiology of anorectal malformations. Birth Defects Orig Artic Ser 24:231–246
8. Bradham RR (1958) Imperforate anus; report of 130 cases. Surgery 44:578–584
9. Crowell EA, Dulin JW (1940) Congenital anomalies of the anus and rectum. Surgery 529–539
10. Ivy RH (1957) Congenital anomalies; as recorded on birth certificates in the Division of Vital Statistics of the Pennsylvania Department of Health, for the period 1951–1955, inclusive. Plast Reconstr Surg 20:400–411
11. Keith A (1908) Three demonstrations on malformations of the hind end of the body. Br Med J , 1736–1741
12. Ladd WE, Gross RE (1934) Congenital malformations of the anus and two: report of 162 cases. Am J Surg 23:167–183
13. Louw JH (1959) Malformations of the anus and rectum: a report on 85 consecutive cases. S Afr Med J 33:874–881

14. Malpas P (1937) The incidence of human malformations and the significance of changes in the maternal environment in the causation. J Obstet Gynaecol Br Commw 44:434–454
15. Moore TC, Lawrence EA (1952) Congenital malformations of the rectum and anus. II. Associated anomalies encountered in a series of 120 cases. Surg Gynecol Obstet 95:281–288
16. Nazer J, Hubner ME, Valenzuela P, Cifuentes L (2000) Anorectal congenital malformations and their preferential associations. Experience of the Clinical Hospital of the University of Chile. Period 1979–1999. Rev Med Chil 128:519–525
17. Niedzielski J (2000) Incidence of anorectal malformations in Lodz province. Med Sci Monit 6:133–136
18. Nixon HH (1972) Anorectal anomalies: with an international proposed classification. Postgrad Med J 48:465–470
19. Schuler L, Salzano FM (1994) Patterns in multimalformed babies and the question of the relationship between sirenomelia and VACTERL. Am J Med Genet 49:29–35
20. Sipek A, Gregor V, Horacek J, Masatova D (2004) Survival of children born with selected types of birth defects in Czech Republic. Ceska Gynekol 69(Suppl 1) :47–52
21. Smith ED (1988) Incidence frequency of types and etiology of anorectal malformations. In Smith ED, Stephens FD (eds) Anorectal Malformations in Children, Update 1988 edn. March of Dimes Birth Defects Foundation and Alan R. Liss, New York, pp 238–240
22. Spouge D, Baird PA (1986) Imperforate anus in 700,000 consecutive liveborn infants. Am J Med Genet Suppl 2:151–161
23. Stoll C, Alembik Y, Roth MP, Dott B (1997) Risk factors in congenital anal atresias. Ann Genet 40:197–204
24. Thomas MP (1977) Incidence of some surgically correctable congenital abnormalities in South Australia. J Pediatr Surg 12:693–701
25. Tong MC (1981) Anorectal anomalies: a review of 49 cases. Ann Acad Med Singapore 10:479–484
26. Skandalakis JE, Gray SW, Ricketts R (1994) The colon and rectum. In: Skandalakis JE, Gray SW (eds) Embryology for Surgeons, 2nd edn. Williams and Wilkins, Baltimore, pp. 242–281
27. Endo M, Hayashi A, Ishihara M, Maie M, Nagasaki A, Nishi T, Saeki M (1999) Analysis of 1,992 patients with anorectal malformations over the past two decades in Japan. Steering Committee of Japanese Study Group of Anorectal Anomalies. J Pediatr Surg 34:435–441
28. Santulli TV, Schullinger JN, Kiesewetter WB, Bill AH Jr (1971) Imperforate anus: a survey from the members of the Surgical Section of the American Academy of Pediatrics. J Pediatr Surg 6:484.–487
29. Cuschieri A (2002) Anorectal anomalies associated with or as part of other anomalies. Am J Med Genet 110:122–130
30. Chadha R, Bagga D, Malhotra CJ, Mohta A, Dhar A, Kumar A (1994) The embryology and management of congenital pouch colon associated with anorectal agenesis. J Pediatr Surg 29:439–446
31. Chadha R (2004) Congenital pouch colon associated with anorectal agenesis. Pediatr Surg Int 20:393–401
32. Shija JK (1986) Some observations on anorectal malformations in Zimbabwe. Cent Afr J Med 32:208–213
33. Louw JH (1965) Congenital abnormalities of rectum and anus. Curr Probl Surg 31:1–64
34. Adeyemo AA, Gbadegesin RA, Omotade OO (1997) Major congenital malformations among neonatal referrals to a Nigerian university hospital. East Afr Med J 74:699–701
35. Akamaguna AI, Odita JC (1985) Intestinal obstruction of infancy and childhood in Benin City, Nigeria. Trop Geogr Med 37:160–164
36. Ameh EA, Dogo PM, Nmadu PT (2001) Emergency neonatal surgery in a developing country. Pediatr Surg Int 17[5–6:448–451
37. Archibong AE, Idika IM (2004) Results of treatment in children with anorectal malformations in Calabar, Nigeria. S Afr J Surg 42:88–90
38. Smith ED, Stephens FD (1988) High, intermediate, and low anomalies in the male. Birth Defects Orig Artic Ser 24:17–72
39. Kiesewetter WB, Turner CR, Sieber WK (1964) Imperforate anus. Review of a sixteen year experience with 146 patients. Am J Surg 107:412–421
40. Harris J, Kallen B, Robert E (1995) Descriptive epidemiology of alimentary tract atresia. Teratology 52:15–29
41. Ratan SK, Rattan KN, Pandey RM, Mittal A, Magu S, Sodhi PK (2004) Associated congenital anomalies in patients with anorectal malformations – a need for developing a uniform practical approach. J Pediatr Surg 39:1706–1711
42. Lubinsky MS (1997) Classifying sex biased congenital anomalies. Am J Med Genet 69:225–228
43. Forrester MB, Merz RD (2002) Descriptive epidemiology of anal atresia in Hawaii, 1986–1999. Teratology 66 (Suppl 1):S12–16
44. Van Gelder DW, Kloepfer HW (1961) Familial anorectal anomalies. Pediatrics 27:334–336
45. Briard ML, Frezal J, Kaplan J, Nihoul-Fekete C, Valayer J (1977) Ano-rectal abnormalities and esophageal atresia; familial and epidemiological studies. Arch Fr Pediatr 34 (7 Suppl):CLXXII
46. Goldschmidt H, Globl H (2005) Familial occurrence of anorectal abnormalities associated with vertebral abnormalities. Helv Paediatr Acta 27:361–370
47. Manny J, Schiller M, Horner R, Stein H, Luttwak EM (1973) Congenital familial anorectal anomaly. Am J Surg 125:639–640
48. Naveh Y, Friedman A (1976) Familial imperforate anus. Am J Dis Child 130:441–442

49. Schwoebel MG, Hirsig J, Schinzel A, Stauffer UG (1984) Familial incidence of congenital anorectal anomalies. J Pediatr Surg 19:179–182
50. Murken JD, Albert A (1976) Genetic counselling in cases of anal and rectal atresia. Prog Pediatr Surg 9:115–118
51. Weinstein ED (1965) Sex-linked imperforate anus. Pediatrics 35:715–718
52. Landau D, Mordechai J, Karplus M, Carmi R (1997) Inheritance of familial congenital isolated anorectal malformations: case report and review. Am J Med Genet 71:280–282
53. Al-Gazali LI, Bakir M, Hamud OA, Gerami S (2002) An autosomal recessive syndrome of nasal anomalies associated with renal and anorectal malformations. Clin Dysmorphol 11:33–38
54. Kurosaki M, Kamitani H, Anno Y, Watanabe T, Hori T, Yamasaki T (2001) Complete familial Currarino triad. Report of three cases in one family. J Neurosurg Spine 94:158–161
55. Iinuma Y, Iwafuchi M, Uchiyama M, Yagi M, Kondoh K, Ohtani S, Kanada S, Mishina T, Saitoh H, Suzuki N (2000) A case of Currarino triad with familial sacral bony deformities. Pediatr Surg Int 16:134–135
56. Christensen K, Madsen CM, Hauge M, Kock K (1990) An epidemiological study of congenital anorectal malformations: 15 Danish birth cohorts followed for 7 years. Paediatr Perinat Epidemiol 4:269–275
57. Kubiak R, Upadhyay V (2005) Isolated imperforate anus in monozygotic twins: case report and implications. J Pediatr Surg 40:E1–4
58. Towne, PL, Brock (1972) Hereditary syndrome of imperforate anus with hand foot and ear anomalies. J Pediatr 81:321–326
59. Thompson EM, Baraitser M, Lindenbaum RH, Zaidi ZH, Kroll JS (1985) The FG syndrome: 7 new cases. Clin Genet 27:582–594
60. Kaufman RL, Hartman A, McAlister WH, et al (1972) Family studies of congenital heart disease: a syndrome of Hydrometrocolpus, proximal polydactyly and congenital heart disease. Birth Defects Original Series 8:85–87
61. Lowe J, Kohn G, Cohen O, et al (1972) Dominant anorectal malformations, nephritis and nerve deafness: a possible new entity? Clin Genet 24:191–193
62. Carey JC, Greenbaum B, Hall BD (2005) The OEIS complex (omphalocele, exstrophy, imperforate anus, spinal defects). Birth Defects Orig Artic Ser 14:253–263
63. Adachi M, Urata R, Takashima R, Miyamoto H, Tsuneishi S, Nakamura H (2003) Pallister-Mosaic syndrome and neuronal migration disorder. Brain Dev 25:357–361
64. Zlotogora J, Abu-Dalu K, Lernau O, Sagi M, Voss R, Cohen T (1989) Anorectal malformations and Down syndrome. Am J Med Genet 34:330–331
65. Clarke SA, Van der Avoirt A (1999) Imperforate anus, Hirschsprung's disease, and trisomy 21: a rare combination. J Pediatr Surg 34:1874
66. Agarwala S, Goswami JK, Mitra DK (1999) Pyloric atresia associated with epidermolysis bullosa, malrotation, and high anorectal malformation with recto-urethral fistula: a report of successful management. Pediatr Surg Int 15:264–265
67. Bai Y, Chen H, Yuan ZW, Wang W (2004) Normal and abnormal embryonic development of the anorectum in rats. J Pediatr Surg 39:587–590
68. Hasse W (1976) Associated malformation with anal and rectal atresiae. Prog Pediatr Surg 9:99–103
69. Bacon HE, Hering AC (1948) Surgical management of congenital malformations of the anus, rectum and colon. J Atheroscler Thromb 4:41
70. Lee MJ Jr (1944) Congenital anomalies of the lower part of the rectum. Am J Dis Child 68:182–189
71. Norris WJ, Brophy TW, Brayton D (1949) Imperforate anus: a case series and preliminary report on the one stage abdomino-perineal operation. Surg Gynecol Obstet 88:623–625
72. Taneja OP, Sharma MM, Mukerji AC, Taneja S (1970) Management of congenital anorectal malformations in India. Arch Surg 100:47–54
73. Smith ED, Saeki M (1988) Associated anomalies. Birth Defects Orig Artic Ser 24:501–549
74. Chalapathi G, Chowdhary SK, Rao KL, Samujh R, Narasimhan KL, Mahajan JK, Menon P (2004) Risk factors in the primary management of anorectal malformations in Northern India. Pediatr Surg Int 20:408–411
75. Chen CJ (1999) The treatment of imperforate anus: experience with 108 patients. J Pediatr Surg 34:1728–1732
76. Mittal A, Airon RK, Magu S, Rattan KN, Ratan SK (2004) Associated anomalies with anorectal malformation (ARM). Indian J Pediatr 71:509–514
77. Janik JS, Hendrickson RJ, Janik JP, Bensard DD, Partrick DA, Karrer FM (2003) Spectrum of anorectal anomalies in pygopagus twins. J Pediatr Surg 38:608–612
78. Schuler L, Salzano FM (1994) Patterns in multimalformed babies and the question of the relationship between sirenomelia and VACTERL. Am J Med Genet 49[1:29–35
79. Poenaru D, Uroz-Tristan J, Leclerc S, Murphy S, Bensoussan AL (1995) Imperforate anus, malrotation and Hirschsprung's disease: a rare association. Eur J Pediatr Surg 5:187–189
80. Haynes JH, Bagwell CE (2003a) Hirschprung's disease and imperforate anus in Pallister-Hall syndrome: a new association. J Pediatr Surg 38:1411–1412
81. Baltogiannis N, Mavridis G, Soutis M, Keramidas D (2003) Currarino triad associated with Hirschsprung's disease. J Pediatr Surg 38:1086–1089
82. Flageole H, Fecteau A, Laberge JM, Guttman FM (1996) Hirschsprung's disease, imperforate anus, and Down's syndrome: a case report. J Pediatr Surg 31:759–760
83. Adejuyigbe O, Abubakar AM, Sowande OA, Olayinka OS, Uba AF (2004) Experience with anorectal malformations in Ile-Ife, Nigeria. Pediatr Surg Int 20:855–858

84. Belman AB, King LR (1972) Urinary tract abnormalities associated with imperforate anus. J Urol 108:823–824
85. Hoekstra WJ, Scholtmeijer RJ, Molenaar JC, Schreeve RH, Schroeder FH (1983) Urogenital tract abnormalities associated with congenital anorectal anomalies. J Urol 130:962–963
86. Munn R, Schillinger JF (1983) Urologic abnormalities found with imperforate anus. Urology 21:260–264
87. Parrott TS (1985) Urologic implications of anorectal malformations. Urol Clin North Am 12:13–21
88. Rich MA, Brock WA, Peña A (1988) Spectrum of genitourinary malformations in patients with imperforate anus. Pediatr Surg Int 3:110–113
89. Peña A (1990) Surgical Management of Anorectal Malformations, 1st edn. Springer Verlag, New York
90. Fleming SE, Hall R, Gysler M, McLorie GA (1986) Imperforate anus in females: frequency of genital tract involvement, incidence of associated anomalies, and functional outcome. J Pediatr Surg 21:146–150
91. Botto LD, Khoury MJ, Mastroiacovo P, Castilla EE, Moore CA, Skjaerven R, Mutchinick OM, et al (1997) The spectrum of congenital anomalies of the VATER association: an international study. Am J Med Genet 71:8–15
92. Rittler M, Paz JE, Castilla EE (1996) VACTERL association, epidemiologic definition and delineation. Am J Med Genet 63:529–536
93. Sangkhathat S, Patrapinyokul S, Tadtayathikom K (2002) Associated genitourinary tract anomalies in anorectal malformations: a thirteen year review. J Med Assoc Thai 85:289–296
94. Quan L, Smith DW (1973) The VATER association. Vertebral defects, anal atresia, T–E fistula with esophageal atresia, radial and renal dysplasia: a spectrum of associated defects. J Pediatr 82:104–107
95. Kaufman RL (1973) Birth defects and oral contraceptives. Lancet 1:1396
96. Nora AH, Nora JJ (1975) A syndrome of multiple congenital anomalies associated with teratogenic exposure. Arch Environ Health 30:17–21
97. Martinez-Frias ML, Bermejo E, Frias JL (2001) The VACTERL association: lessons from the Sonic hedgehog pathway. Clin Genet 60:397–398
98. McMullen KP, Karnes PS, Moir CR, Michels VV (1996) Familial recurrence of tracheoesophageal fistula and associated malformations. Am J Med Genet 63:525–528
99. Nezarati MM, McLeod DR (1999) VACTERL manifestations in two generations of a family. Am J Med Genet 82:40–42
100. Qi BQ, Beasley SW, Arsic D (2004) Abnormalities of the vertebral column and ribs associated with anorectal malformations. Pediatr Surg Int 20:529–533
101. Kim J, Kim PCW, Hui C-C (2001) The VACTERL association: lessons from the Sonic hedgehog pathway. Clin Genet 59:306–315
102. Mo R, Kim JH, Zhang J, Chiang C, Hui CC, Kim PC (2002) Anorectal malformations caused by defects in sonic hedgehog signaling. Am J Pathol 159:765–774
103. Beasley SW, Diez Pardo J, Qi BQ, Tovar JA, Xia HM (2000) The contribution of the adriamycin-induced rat model of the VATER association to our understanding of congenital abnormalities and their embryogenesis. Pediatr Surg Int 16:465–472
104. Qi BQ, Beasley SW, Frizelle FA (2003) Evidence that the notochord may be pivotal in the development of sacral and anorectal malformations. J Pediatr Surg 38:1310–1316
105. Duncan PA, Shapiro LR (1993) Interrelationships of the hemifacial microsomia – VATER, VATER, and sirenomelia phenotypes. Am J Med Genet 47:75–84
106. Cox PM, Gibson RA, Morgan N, Brueton LA (1997) VACTERL with hydrocephalus in twins due to Fanconi anemia (FA): mutation in the FAC gene. Am J Med Genet 68:86–90
107. Kallen K, Castilla EE, Robert E, Mastroiacovo P, Kallen B (2000) OEIS complex – a population study. Am J Med Genet 92:62–68
108. Bohring A (2002) OEIS complex, VATER, and the ongoing difficulties in terminology and delineation. Am J Med Genet 107:72–76
109. Kochling, Karbasiyan M, Reis A (2001) Spectrum of mutations and genotype-phenotype analysis in Currarino syndrome. Eur J Hum Genet 9:599–605
110. Nagai T, Katoh R, Hasegawa T, Ohashi H, Fukushima Y(1994) Currarino triad (anorectal malformation, sacral bony abnormality and presacral mass) with partial trisomy of chromosomes 13q and 20p. Clin Genet 45:272–273
111. Ross AJ, Ruiz-Perez V, Wang Y, Hagan DM, Scherer S, Lynch SA, Lindsay S, Custard E, Belloni E, Wilson DI, Wadey R, Goodman F, Orstavik KH, Monclair T, Robson S, Reardon W, Burn J, Scambler P (1998) A homeobox gene HLXB9 is the major locus for dominantly inherited sacral agenesis. Nat Genet 20:358–361
112. Greenwood RD, Rosenthal A, Nadas AS (1975) Cardiovascular malformations associated with imperforate anus. J Pediatr 86:576–579
113. Mahajan JK, Kumar D, Chowdhary SK, Rao KL (2003) Anorectal malformation with malrotation of gut. Eur J Pediatr Surg 13:63–65
114. Hartman EE, Oort FJ, Aronson DC, Hanneman MJ, van der Zee DC, Rieu PN, Madern GC, van Heurn LW, van Silfhout-Bezemer M, Looyaard N, Sprangers MA (2004) Critical factors affecting quality of life of adult patients with anorectal malformations or Hirschsprung's disease. Am J Gastroenterol 99:907–913
115. Shanske A, Ferreira JC, Leonard JC, Fuller P, Marion RW (2001) Hirschsprung disease in an infant with a contiguous gene syndrome of chromosome 13. Am J Med Genet 102:231–236

116. Wang J, Shi C, Yu S, Wu Y, Xu C (2003) A rare association of rectal and genitourinary duplication and anorectal malformation. Chin Med J (Engl) 116:1955–1957
117. Meijers C, Mulder M (1995) Anteroposterior differences within caudal hindbrain neural crest cell populations and the development of the enteric nervous system. Presented at the Second International Meeting: Hirschsprung Disease and Related Neurocristopathies. Cleveland, Ohio, October 1995
118. Watanatittan S, Suwatanaviroj A, Limprutithum T, Rattanasuwan T (1991) Association of Hirschsprung's disease and anorectal malformation. J Pediatr Surg 26:192–195
119. Takada Y, Aoyama K, Goto T, Mori S (1985a) The association of imperforate anus and Hirschsprung's disease in siblings. J Pediatr Surg 20:271–273
120. Moore SW, Rode H, Millar AJ, Albertyn R, Cywes S (1991) Familial aspects of Hirschsprungs disease. Eur J Pediatr Surg 1:97–107
121. Moore SW (1993) A study of the etiology of post-surgical obstruction in patients with Hirschsprungs disease. Doctoral Thesis, University of Cape Town
122. Bitoh Y, Shimotake T, Sasaki Y, Iwai N (2002) Development of the pelvic floor muscles of murine embryos with anorectal malformations. J Pediatr Surg 37:224–227
123. Morimoto K, Takemoto O, Wakayama A (2003) Tethered cord associated with anorectal malformation. Pediatr Neurosurg 38:79–82
124. Torre M, Martucciello G, Jasonni V (2001) Sacral development in anorectal malformations and in normal population. Pediatr Radiol 31:858–862
125. Horn D, Tonnies H, Neitzel H, Wahl D, Hinkel GK, von Moers A, Bartsch O (2004) Minimal clinical expression of the holoprosencephaly spectrum and of Currarino syndrome due to different cytogenetic rearrangements deleting the Sonic Hedgehog gene and the HLXB9 gene at 7q36.3. Am J Med Genet 128A:85–92
126. Le Caignec C, Winer N, Boceno M, Delnatte C, Podevin G, Liet JM, Quere MP, Joubert M, Rival JM (2003) Prenatal diagnosis of sacrococcygeal teratoma with constitutional partial monosomy 7q/trisomy 2p. Prenat Diagn 23:981–984
127. Riebel T, Kochling J, Scheer I, Oellinger J, Reis A (2004) Currarino syndrome: variability of imaging findings in 22 molecular-genetically identified (HLXB9 mutation) patients from five families. Rofo Fortschr Geb Rontgenstr Neuen Bildgeb Verfahr 176:564–569
128. Kumar A, Gupta AK, Bhatnagar V (2005) Low anorectal malformation associated with sarococcygeal teratoma. Trop Gastroenterol 25:101–102
129. Urioste M, Garcia-Andrade Mdel C, Valle L, Robledo M, Gonzalez-Palacios F, Mendez R, Ferreiros J, Nuno J, Benitez J (2005) Malignant degeneration of presacral teratoma in the Currarino anomaly. Am J Med Genet A 128:299–304
130. Seri M, Martucciello G, Paleari L, Bolino A, Priolo M, Salemi G, Forabosco P, Caroli F, Cusano R, Tocco T, Lerone M, Cama A, Torre M, Guys JM, Romeo G, Jasonni V (1999) Exclusion of the Sonic Hedgehog gene as responsible for Currarino syndrome and anorectal malformations with sacral hypodevelopment. Hum Genet 104:108–110
131. Lynch SA, Bond PM, Copp AJ, Kirwan WO, Nour S, Balling R, Mariman E, Burn J, Strachan T (1995) A gene for autosomal dominant sacral agenesis maps to the holoprosencephaly region at 7q36. Nat Genet 11:93–95
132. Belloni E, Martucciello G, Verderio D, Ponti E, Seri M, Jasonni V, Torre M, Ferrari M, Tsui LC, Scherer SW (2000) Involvement of the HLXB9 homeobox gene in Currarino syndrome. Am J Hum Genet 66:312–319
133. Rosano A, Botto LD, Olney RS, Khoury MJ, Ritvanen A, et al (2000) Limb defects associated with major congenital anomalies: clinical and epidemiological study from the International Clearinghouse for Birth Defects Monitoring Systems. Am J Med Genet 93:110–116
134. Kodaka T, Kanamori Y, Sugiyama M, Hashizume K (2004) A case of acrocephalosyndactyly with low imperforate anus. J Pediatr Surg 39:E32–34
135. Kondo T, Dolle P, Zakany J, Duboule D (1996) Function of posterior HoxD genes in the morphogenesis of the anal sphincter. Development 122:2651–2659
136. Fairbanks TJ, De Langhe S, Sala FG, Warburton D, Anderson KD, Bellusci S, Burns RC (2004) Fibroblast growth factor 10 (Fgf10) invalidation results in anorectal malformation in mice. J Pediatr Surg 39:360–365
137. Saxena AK, Morcate JJ, Schleef J, Reich A, Willital GH (2004) Rectal atresia, choanal atresia and congenital heart disease: a rare association. Technol Health Care 12:343–345
138. Kollarova A, Misovicova N, Malis V (1999) The Schmid-Fraccaro syndrome. Cesk Slov Oftalmol 55:362–366
139. Spadoni E, Castelnovi C, Maraschio P, Stacul E, Beluffi G, Bozzola M, Danesino C (2004) Growth hormone deficiency, anorectal agenesis, and brachycamptodactyly: a phenotype overlapping Stratton-Parker syndrome. Am J Med Genet A 128:57–59
140. Mandhan P, Qi BQ, Beasley SW (2005) Aberrations of the intrinsic innervation of the anorectum in fetal rats with anorectal malformations. J Pediatr Surg 40:397–402
141. Martucciello G, Mazzola C, Favre A, Negri F, Bertagnon M, Morando A, Torre M, Gambini C, Jasonni V (1999) Preoperative enzymo-histochemical diagnosis of dysganglionoses associated with anorectal malformations (ARM) with recto-vestibular and recto-perineal fistula. Eur J Pediatr Surg 9:96–100
142. Parida SK, Hall BD, Barton L, Fujimoto A (1995) Penoscrotal transposition and associated anomalies: report of five new cases and review of the literature. Am J Med Genet 59:68–75
143. Bass J (1991) Radial artery hypoplasia: a further association with the VATER syndrome? J Urol 146:824–825

144. Merei JM (2003) Single umbilical artery and the VATER-animal model. J Pediatr Surg 38:1756–1759
145. Padmanabhan R (1998) Retinoic acid-induced caudal regression syndrome in the mouse fetus. Reprod Toxicol 12:139–151
146. Pitera JE, Smith VV, Woolf AS, Milla PJ (2001) Embryonic gut anomalies in a mouse model of retinoic acid-induced caudal regression syndrome: delayed gut looping, rudimentary cecum, and anorectal anomalies. Am J Pathol 159:2321–2329
147. Hersh JH, Angle B, Fox TL, Barth RF, Bendon RW, Gowans G (2002) Developmental field defects: coming together of associations and sequences during blastogenesis. Am J Med Genet 110:320–323
148. Kluth D, Lambrecht W (1997) Current concepts in the embryology of anorectal malformations. Semin Pediatr Surg 6:180–186
149. Ince TA, Cviko AP, Quade BJ, Yang A, McKeon FD, Mutter GL, Crum CP (2002) p63 Coordinates anogenital modeling and epithelial cell differentiation in the developing female urogenital tract. Am J Pathol 161:1111–1117
150. Dravis C, Yokoyama N, Chumley MJ, Cowan CA, Silvany RE, Shay J, Baker LA, Henkemeyer M (2004) Bidirectional signaling mediated by ephrin-B2 and EphB2 controls urorectal development. Dev Biol 271:272–290
151. Kimmel SG, Mo R, Hui CC, Kim PC (2000) New mouse models of congenital anorectal malformations. J Pediatr Surg 35:227–230
152. Hess R, Scarpelli DG, Pearse AG (1958) The cytochemical localization of oxidative enzymes II Pyridine nucleotide-linked dehydrogenases. J Biophys Biochem Cytol 4:753–760
153. Martinez-Frias ML, Bermejo E, Frias JL (2001) The VACTERL association: lessons from the Sonic hedgehog pathway. Clin Genet 60:397–398
154. Hori T, Giuffra E, Andersson L, Ohkawa H (2001) Mapping loci causing susceptibility to anal atresia in pigs, using a resource pedigree. J Pediatr Surg 36:1370–1374
155. Trusler GA, Mestel AL, Stephens CA (1959) Colon malformation with imperforate anus. Surgery 45:328–334
156 Stone HH, Wilkinson AW (1983) Experimental production of rectal stenosis and atresia in the rabbit. J Pediatr Surg 18:89–90
157. Bourdelat D, Labbe F, Pillet J, Delmas P, Hidden G, Hureau J (1988) A study in organogenesis: the arterial supply of the anorectal region in the human embryo and fetus. Anatomic and embryologic bases of anorectal malformations. Surg Radiol Anat 10:37–51
158. Lambrecht W, Riebel T, Weinland G (1986) Vascular supply of the rectum in anal atresia. Angiography studies in newborn swine. Z Kinderchir 41:340–343
159. Stephens FD, Donnellan WL (1977) "H-type" urethroanal fistula. J Pediatr Surg 12:95–102
160. Willems M, Kluth D, Lambrecht W (1996) Anorectal malformation: a new anatomic variant resembling an H-type fistula. J Pediatr Surg 31:1682–1684
161. Ives EJ (1962) Thalidomide and anal anomalies. Can Med Assoc J 87:670–672
162. Baynash AG, Hosoda K, Giaid A, et al (1994) Interaction of endothelin-3 with endothelin-B receptor is essential for development of epidermal melanocytes and enteric neurons. Cell 79:1277–1285
163. Hosoda K, Hammer RE, Richardson JA, et al (1994) Targeted and natural (piebald-lethal) mutations of endothelin-B receptor gene produce megacolon associated with spotted coat color in mice. Cell 79:1267–1276
164. Dembowski C, Hofmann P, Koch T, Kamrowski-Kruck H, Riedesel H, Krammer HJ, Kaup FJ, Ehrenreich H (2000) Phenotype, intestinal morphology, and survival of homozygous and heterozygous endothelin B receptor-deficient (spotting lethal) rats. J Pediatr Surg 35:480–488
165. Pla P, Larue L (2004) Involvement of endothelin receptors in normal and pathological development of neural crest cells. Int J Dev Biol 47:315–325
166. Arai H, Nakao K, Takaya K, et al (1993) The human endothelin-B receptor gene: structural organisation and chromosomal assignment. J Biol Chem 268:3463–3470
167. Sakurai T, Yanagisawa M, Takuwa Y, Miyazaki H, Kimura S, Goto K, Masaki T (1990) Cloning of a cDNA encoding a non-isopeptide-selective subtype of the endothelin receptor. Nature 348:732–735
168. Shichiri M, Kato H, Marumo F, Hirata Y (1997) Endothelin-1 as an autocrine/paracrine apoptosis survival factor for endothelial cells. Hypertension 30:1198–1203
169. Lahav R, Suva ML, Rimoldi D, Patterson PH, Stamenkovic I (2004) Endothelin receptor B inhibition triggers apoptosis and enhances angiogenesis in melanomas. Cancer Res 64:8945–8953
170. Yanagisawa M, Kurihara H, Kimura S, Goto K, Masaki T (1988) A novel peptide vasoconstrictor, endothelin, is produced by vascular endothelium and modulates smooth muscle Ca2+ channels. J Hypertens Suppl 6:S188–191
171. Nankervis CA, Reber KM, Nowicki PT (2001) Age-dependent changes in the postnatal intestinal microcirculation. Microcirculation 8:377–387
172. Siren AL, Lewczuk P, Hasselblatt M, Dembowski C, Schilling L, Ehrenreich H (2002) Endothelin B receptor deficiency augments neuronal damage upon exposure to hypoxia-ischemia in vivo. Brain Res 945:144–149
173. Murakoshi N, Miyauchi T, Kakinuma Y, Ohuchi T, Goto K, Yanagisawa M, Yamaguchi I (2002) Vascular endothelin-B receptor system in vivo plays a favorable inhibitory role in vascular remodeling after injury revealed by endothelin-B receptor-knockout mice. Circulation 106:1991–1998
174. Moore SW, Zaahl MG (2006) Association of Endothelin-ß receptor (EDNRB) gene variants in Anorectal malformations. J Pediatr Surg (in press)

175. Bonnot O, Vollset SE, Godet PF, d'Amato T, Dalery J, Robert E (2003) In utero exposure to benzodiazepine. Is there a risk for anal atresia with lorazepam? Encephale 29:553–559
176. Hashimoto R, Nagaya M, Ishiguro Y, Inouye M, Aoyama H, Futaki S, Murata Y (2002) Relationship of the fistulas to the rectum and genitourinary tract in mouse fetuses with high anorectal malformations induced by all-trans retinoic acid. Pediatr Surg Int 18:723–727
177. Yu J, Gonzalez S, Martinez L, Diez-Pardo JA, Tovar JA (2003) Effects of retinoic acid on the neural crest-controlled organs of fetal rats. Pediatr Surg Int 19:355–358
178. Liu Y, Sugiyama F, Yagami K, Ohkawa H (2003) Sharing of the same embryogenic pathway in anorectal malformations and anterior sacral myelomeningocele formation. Pediatr Surg Int 19:152–156
179. Sasaki Y, Iwai N, Tsuda T, Kimura O (2005) Sonic hedgehog and bone morphogenetic protein 4 expressions in the hindgut region of murine embryos with anorectal malformations. J Pediatr Surg 39:170–173
180. Angerpointner T, Radtke W, Murken JD (1981) Catamnestic investigations in children with malformations of the gastrointestinal tract and the abdominal wall. Z Kinderchir 32:129–144
181. Mulvihill JJ (2005) Clinical ecogenetics – cancer in families. N Engl J Med 312:1569–1570
182. Jo Mauch T, Albertine KH (2002) Urorectal septum malformation sequence: Insights into pathogenesis. Anat Rec 268:405–410

4 The Embryology of Anorectal Malformations

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4.1 Introduction

Despite many efforts, the embryology of numerous congenital anomalies in humans remains a matter of speculation. This is due to a number of reasons, such as a shortage of study material (both normal and abnormal embryos), various technical problems (difficulties in the interpretation of serial sections, shortage of explanatory three-dimensional reconstructions), and misconceptions and/or outdated theories concerning normal and abnormal embryology. Fortunately, there are now several animal models that allow advanced embryological studies in various embryological fields, including that of anorectal malformations (ARM).

Appropriate and illustrative findings in various fields of embryology are still lacking. This explains why today many typical malformations are still not explained satisfactorily. Pediatric surgeons are still confused when they are confronted with the embryological background of normal and abnormal development.

For misconceptions and/or outdated theories, Haeckel's "biogenetic law" [1] is one example. According to this theory, a human embryo recapitulates in its individual development (ontogeny) the morphology observed in all life-forms (phylogeny). This means that during its development an advanced species is seen to pass through stages represented by adult organisms of more primitive species [2]. This theory still has an impact on the nomenclature of embryonic organs. This explains why human embryos have "cloacas" like adult birds and "branchial" clefts like adult fish.

Another very popular misconception is the theory that malformations actually represent "frozen" stages of normal embryology ("Hemmungsmißbildung") [3]. As a result, our understanding of normal embryology stems more from pathological-anatomic interpretations of observed malformations than from proper embryological observations. The theory of the "rotation of the gut" as a step in normal development is a perfect example for this misconception (see below for detailed discussion).

The most widely recognized descriptive embryology of the anorectal region dates mostly from the late nineteenth century. Anorectal separation from the urogenital structures was thought for a long time to occur either as a result of a cranially orientated septum growing down to reach the cloacal membrane and fuse with it [4] or from lateral folds encroaching on the lumen of the cloaca from either side and fusing in the middle [5], or from a combination of the two processes [6]. These theories have been seriously questioned in the literature from time to time during the last century [7–10], but the surgical community largely ignored the doubts raised. Recent developments in our understanding of the mechanisms by which embryos grow and develop, the roles of local cytokines, and molecular messengers from adjacent epithelia, mesenchyme, and the regional nerve supply, have sparked renewed interest in reviewing both normal and abnormal development of the perineum. The clarification of normal embryological development and insights into abnormal development through studies of animal embryos with either genetic or chemically induced ARM now allows us to speculate much more accurately as to the mechanisms that may be involved in the pathogenesis of human anorectal and urogenital malformations.

As molecular biology delivers more insights into the molecular keys that trigger the various stages in development we will learn more about the pathogenesis of ARM. However, the key is to first understand the normal embryological development of the human perineum and the subtle differences that occur across species so as to avoid drawing false deductions from the abnormal development seen in experimental animals.

The value of detailed histological examination of abnormal human specimens cannot be underestimated in adding pieces of information to the jigsaw puzzle of ARM. The detailed work of Douglas Stephens [6] was invaluable and more recently has been complemented by further studies (see below).

By putting together the pieces of the puzzle, the pathogenesis of ARM in humans may be hypothesized and a classification system proposed. This chapter illustrates what we have learned in the field of normal and abnormal hindgut development, using both standard histological sections and scanning electron microscopy (SEM) to illustrate the findings, as SEM allows documentation of three-dimensional structures in superior detail [11,12].

4.2 Early Controversies About Development of the Hindgut

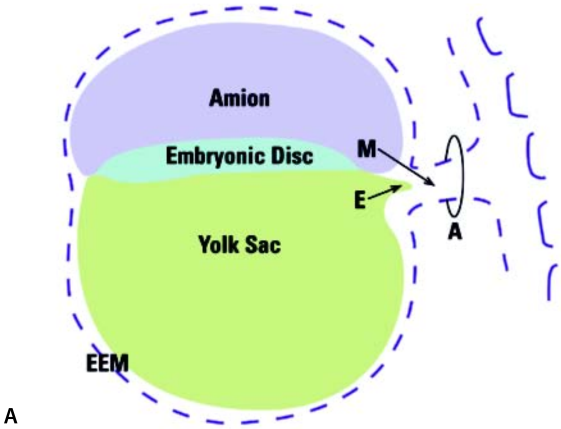
4.2.1 The "Anorectal Septum" of the Hindgut

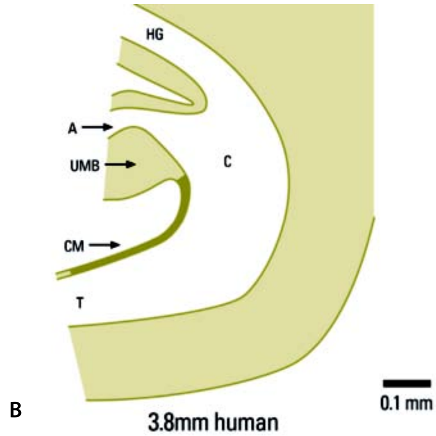
In very young embryos, the hindgut is a simple structure. Cranially, it is in continuity with the midgut; caudally, it is in direct contact with the ectoderm, thus forming the "cloacal membrane." When development progresses, the caudal part of the hindgut, the "cloaca," differentiates into two separate organ systems – the urogenital tract and the anorectal tract. As mentioned above, since the work of Tourneux [4] and Retterer [5] at the end of the nineteenth century, it has been generally accepted that the normal development of these tracts depends upon the proper subdivision of the cloaca by a septum, the so-called urorectal septum. According to this theory, abnormal septal development should always result in abnormal cloacal development. However, there is no agreement among investigators about the nature of this septum and the way it develops. Whereas Tourneux [4] thought that the septum moves down from cranial to caudal "like a French curtain," Retterer [5] speculated that lateral folds or ridges appear in the lumen of the cloaca. These ridges should fuse and thus form the septum, beginning cranially and ending caudally at the level of the cloacal membrane. In the past, numerous investigators supported one or another of these theories. Stephens [6] combined both theories, believing that this could best explain the various forms of ARM. He claimed that the cranial part of the septum should grow downward, as explained by Tourneux, while in the caudal part lateral ridges should fuse to form the septum in this area. In 1986, van der Putte [13] first showed that the role of the urorectal septum in the process of "cloacal" differentiation was actually very minor.

4.2.2 The "Migration" of the Rectum

Studying the morphology of ARM in human newborns, Bill and Johnson [14], and later Gans and Friedman [15] stated that in most forms of ARM the fistula may represent an ectopic anal opening. They concluded from these observations that the rectum actually migrates during normal development, from a rather high position to the normal area of the anal opening. If this process of migration is stopped before the anus has reached its definitive position in the area of the perineum, an ectopic anal canal would result. Although this speculation is rather attractive, neither these investigators nor other researchers were able to show any embryological evidence of this migration.

In 1986, van der Putte [13] modified the theory of a rectal or anal migration. Studying normal and abnormal pig embryos, he proposed that a shift or rotation of the dorsal cloaca takes place. This shift should bring the dorsal cloaca down to the area of the tail





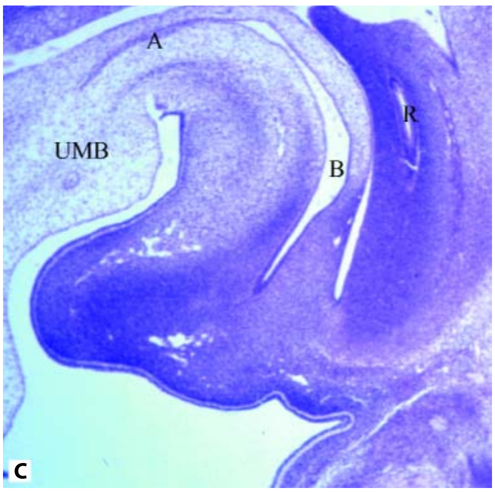


Fig. 4.1 Allantois: The allantois develops as an endodermal and mesenchymal outpouching of the yolk sac within the body stalk (**A**). As the caudal part of the embryo develops and the gut tube forms, the allantois and umbilicus become displaced to the ventral aspect of the embryo where the allantois is in continuity with the cranial aspect of the primitive cloaca. The endodermal allantois therefore extends a short distance into the proximal umbilical cord and is in continuity with the cloaca (**B**, diagram 3.8-mm human embryo; **C**, photomicrograph of 18-mm human embryo). *HG* Hindgut, *T* tailgut, *C* cloaca, *CM* cloacal membrane, *UMB* umbilical stalk, *E* endodermal allantois, *M* mesodermal allantois, *A* allantois within body stalk, *EEM* extraembryonic mesoderm, *B* developing bladder, *R* developing rectum

groove, thus establishing there the future anal opening (see below).

4.3 Normal Anourogenital Development

4.3.1 The Primitive Streak and Allantois

The earliest evidence of craniocaudal orientation in the developing embryo occurs with the formation of the primitive streak at the caudal end of the dorsal aspect of the embryo adjacent to the connecting stalk. From the primitive streak the mesenchymal cells first develop and migrate between the layers of the bilaminar disc and extend throughout the embryo, except where the endoderm and ectoderm are fused at the cloacal and oropharyngeal membranes. The cloacal membrane may be found immediately caudal to the primitive streak. The notochord develops from the notochordal process that also grows between the ectoderm and endoderm in the midline cranially from the primitive pit at the cranial end of the primitive streak. Dorsal to the notochord is the neural plate, the forerunner of the neural tube and future spinal cord.

As the embryo rapidly elongates with the production of the mesoderm and the development of the notochord and the neural tube, the primitive streak itself becomes relatively smaller and is finally succeeded by the caudal eminence and tail bud, which is then the source of most of the caudal mesoderm, the tailgut and, by secondary neurulation, the distal spinal cord [16].

The term "allantois" is often used to describe the endodermal outpouching of the dorsal yolk sac that appears close to the cloacal membrane and extends into the connecting stalk at around day 16 in human embryos (Fig. 4.1). However, it is the combination of the endodermal pouch with its surrounding specialized mesenchyme that is the true allantois that is common to reptiles, birds, and mammals. The specialized mesenchyme of the allantois is also derived from the ectoderm of the primitive streak [16]. In birds and reptiles the allantois is a relatively well-developed structure that is a significant organ of respiration and is involved in the storage of waste products. In mammals it is fundamental to the development of the umbilical cord as well as being a site of angiogenesis and the first site of formation of blood cells. The structure is highly variable. In humans the endodermal component of the allantois is small, while in rodents the allantois is an entirely mesenchymal structure [16].

The allantois develops at what is initially the most caudal end of the cloacal membrane, but with the disproportionate growth of the dorsal aspect of the embryo and the development of the tail structures, the cloacal membrane is displaced to the ventral aspect of the embryo, together with the developing umbilical cord, which therefore lies at the cranial end of the cloacal membrane on the ventral surface of the embryo. The yolk sac is restricted and starts to develop a narrower connection to the mid-part of the developing gut tube.

4.3.2 The Cloaca and Cloacal Membrane

As the embryonic mesenchyme grows and spreads from the dorsal aspect of the embryonic disc the allantois comes to connect the developing cloaca to the developing umbilical cord on the ventral aspect of the embryo. In humans the endodermal component of the allantois becomes an outpouching at the cranioventral aspect of the newly formed cloaca and extends into the proximal umbilical cord. The cloacal membrane limits the cloaca ventrally and the tail gut extends as a narrow projection from the cloaca into the ventral aspect of the embryonic tail. The hindgut is that part of the endoderm-lined gut tube between the yolk sac and the allantois that has developed as a result of mesenchymal growth in that area. The mesonephric ducts open into either side of the hindgut soon after it first appears and at around the same time the endodermal allantois is first seen in humans. The presence of the mesonephric ducts and allantoic diverticulum marks the beginning of the cloaca proper (Fig. 4.2).

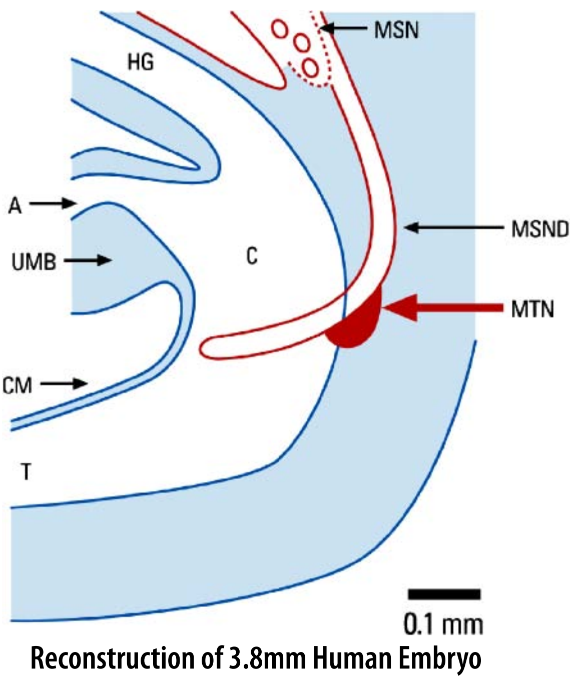


Fig. 4.2 The cloaca: The cloaca is bounded ventrally by the cloacal membrane, is continuous with the tailgut caudally, with the hindgut dorsocranially, and the allantois ventrocranially. This is seen in the human embryo at 3.8 mm length in a midsagittal reconstruction with the mesonephric attachment drawn in the lateral plane. *MSND* Mesonephric duct, *MTN* metanephric blastema, *MSN* mesonephros

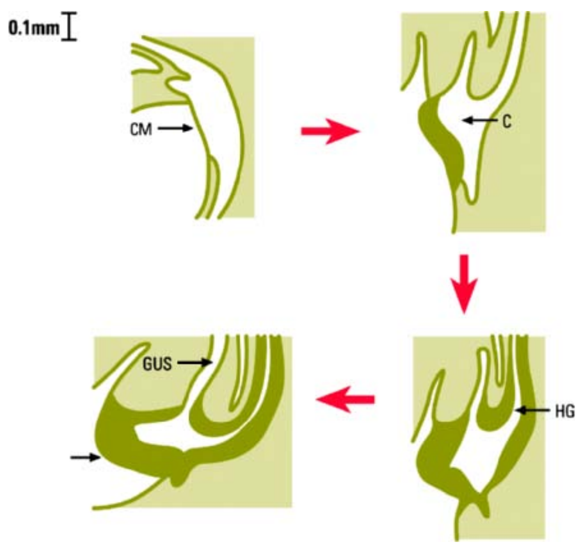
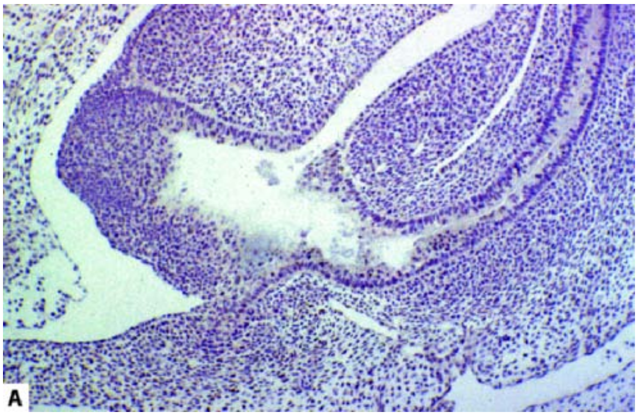


Fig. 4.3 Showing the development of the genital tubercle (cloacal eminence). Growth of the mesenchyme on either side of the cloaca as well as in the midline in the infraumbilical region results in the formation of the genital tubercle (cloacal eminence) as well as a change in the shape of the cloacal cavity and a displacement of the cloacal membrane such that a nearly solid plate of epithelial cells fills the caudal part of the developing tubercle, extending from the tail fold to the tip of the developing tubercle. *GUS* Genitourinary sinus (developing bladder and urethra)



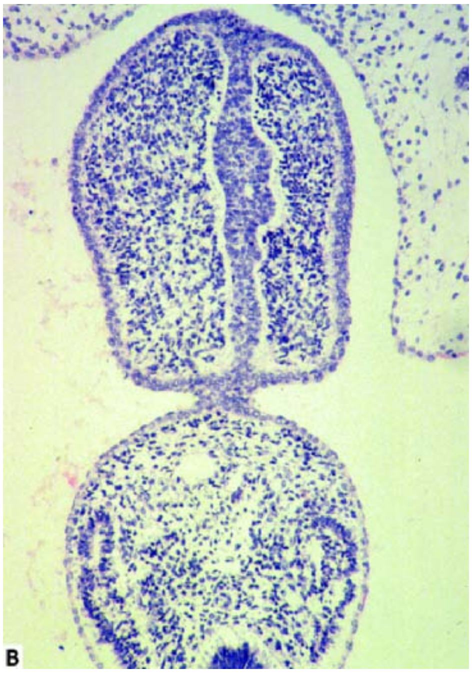


Fig. 4.4 Photomicrographs illustrate the relationship between the cloacal plate and the genital tubercle (caudal eminence) in A 13.5-day developing mouse embryo (magnification ×100). *GT* Genital tubercle, *T* tail; **A** Sagittal rectum; **B** transverse rectum

The cloacal membrane extends to the umbilical cord at this early stage and there is not yet any ventral abdominal wall caudal to the umbilical cord. The first mesenchyme to develop between the umbilical cord and the cloacal membrane is that of the genital tubercle (also known as the cloacal eminence), which normally develops as a single infraumbilical bulge of densely cellular distinctive mesenchyme that displaces the cloacal membrane caudally in the midline and surrounds the ventral portion of the cloacal cavity on either side laterally (Fig. 4.3). The membrane itself assumes the characteristics of a multilayered epithelial plate orientated in the sagittal plane [17]. (Fig. 4.4). This occurs chiefly as a result of lateral compression of the walls of the cloaca consequent to the proliferation of the mesenchyme surrounding the cloaca, but also to a smaller extent near the tip of the genital tubercle by the ingress of ectodermal cells from the surface into the urogenital portion of the plate in the region of the developing glans [18].

4.3.3 The Urogenital Sinus and Anal Canal

As the genital tubercle grows, so the cloacal cavity is displaced and its shape altered. At the same time growth of the mesenchyme surrounding the hindgut results in elongation of the hindgut. Growth in the ventral part of the cloaca and its surrounding mesenchyme adjacent to the endodermal allantois signals the beginning of bladder and urethra development (Fig. 4.5). This part is commonly referred to as the urogenital sinus. The tail gut is resorbed by a process of apoptosis or programmed cell death and the dorsal wall of the cloaca shortens by the same process [19–21].

As the mesenchyme surrounding the various structures associated with the cloaca grows, the cloaca becomes relatively smaller. The mesenchyme between the hindgut and the developing bladder appears to approach the cloacal plate; however, the apparent "descent of the urorectal septum" or "fusion of the lateral walls of the cloaca" described in the past is an illusion created by the changes in the relative size and position of the mesenchymal structures surrounding the cloaca as they grow, and their examination in two-dimensional sections.

As the cloaca develops, the cloacal plate becomes rapidly thinner in the dorsal part until it once again resumes the appearance of a membrane. The appearance of the cloacal plate and the reformed cloacal membrane and their relationship to the mesenchyme between the urogenital sinus and the rectum varies widely across species leading to the mistaken belief that the "uroroectal septum" fuses with the cloacal membrane during normal development (as it appears to do in some species). In humans, however, it is clear that the cloacal membrane breaks down without ever fusing with the urorectal septum, thus exposing both the anal and urogenital compartments and the

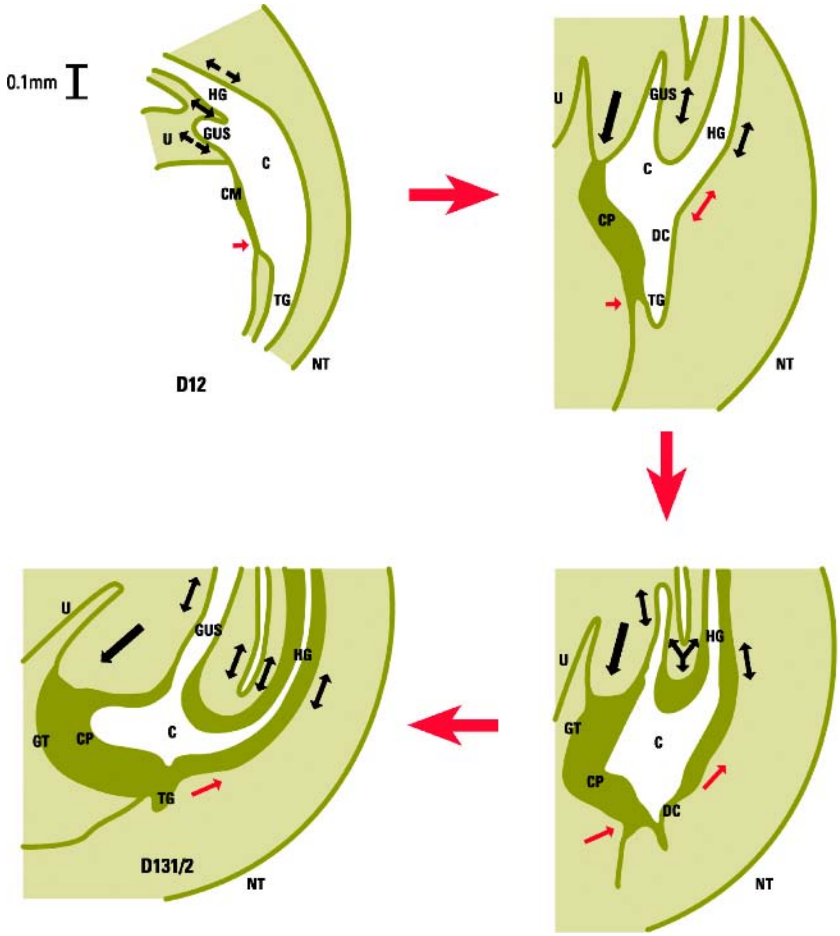
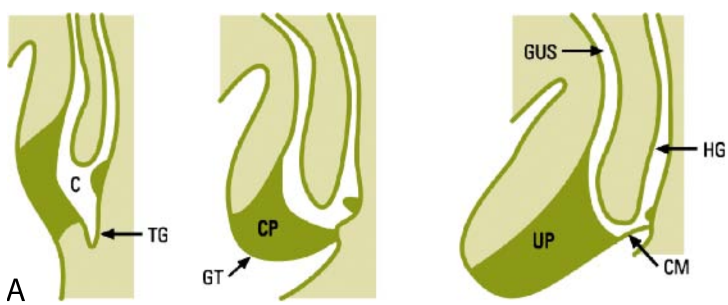


Fig. 4.5 Diagram showing the development of the ventral infraumbilical part of the embryo. Ventral growth in the mesenchyme surrounding the cloaca combined with shortening of the dorsal cloacal wall and regression of the tailgut by apoptosis results in a fundamental change in the shape of the cloaca and the relationship of the associated structures during normal development. *Red arrows* mark areas of regression and programmed cell loss (apoptosis). *Black arrows* mark areas of strong mesenchymal (and associated endodermal) growth. Embryos are aged from day 12 (D12) to day 13½. *NT* Neural tube, *U* umbilicus, *DC* dorsal cloaca



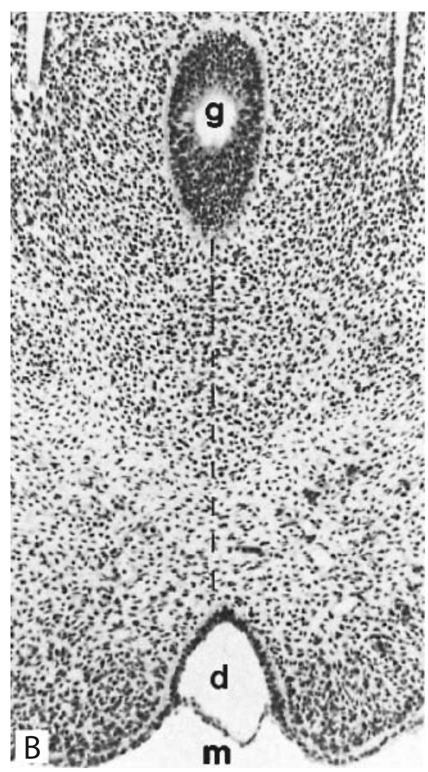


Fig. 4.6 **A** Diagrams redrawn from van der Putte [39]. showing the changes seen in the cloacal plate in the pig embryo during the formation of the genital tubercle and the cloacal plate. Note how the cloacal plate gradually loses height dorsally and reverts to a membrane. **B** Photomicrograph reproduced from van der Putte's paper describing anorectal development [13]. The section passes through the distal hindgut, through the free lower border of the urorectal septum and the dorsal reformed cloacal membrane shortly before it ruptures. *g* Hindgut, *d* remnant of cloacal cavity just prior to rupture of the cloacal membrane, *m* cloacal membrane

intermediate communication, which thus becomes the median cloacal groove (Fig. 4.6), the future midperineum, and the source of the future raphe [22].

The metanephric buds form as an outpouching from the mesonephric duct close to the urogenital sinus. They are induced by the metanephric blastema, which lies at the caudal end of the nephrogenic ridge adjacent to the dorsolateral aspect of the distal mesonephric ducts, and grow directly into the metanephric blastema, thus commencing nephrogenesis. The mesonephric duct distal to the developing ureter (the common excretory duct) is incorporated into the urogenital sinus by a process that combines both apoptosis of the lining of the distal mesonephric duct and replacement by urogenital sinus epithelium, thus forming separate openings for the mesonephric and metanephric (ureteric) ducts into the urogenital sinus [22].

4.3.4 Sexual Differentiation in the Perineum

The median cloacal groove becomes the surface of the midperineal region between the anal and urogenital openings and is enhanced by growth in the contiguous lateral mesenchyme [23]. The mesenchyme on either side of the cloaca initially grows more rapidly than that in the midline leading to labioscrotal swellings on either side of the developing perineum. In the male, growth of the mesenchyme in the midline "fills in" the medial perineal groove and the median aspect of the scrotal swelling, allowing it to develop as a single mass. Strong midline growth increases the distance between the anal canal and the urogenital opening as well as the distance between the anus and the midline scrotal swelling and is associated with the development of the strong perineal body in the male and the muscles of the perineum and the superficial part of the corpus spongiosum of the phallus. Failure of midline development in the male for whatever reason is associated with a shortened distance between the anus and phallus, bifid scrotum, and/or chordee or hypospadias.

In the female the strong growth in the midline perineal structures does not occur, leaving a shorter anophallic distance, a rudimentary perineal body, and separate labial folds with a midline cleft surrounded anteriorly and cranially by the erectile tissue of the labia minora and the corpora cavernosa of the clitoris [24].

Uterine and vaginal development commences with fusion of the paramesonephric or Mullerian ducts in the midline between the hindgut and the developing bladder and extending into the mesenchymal mantel

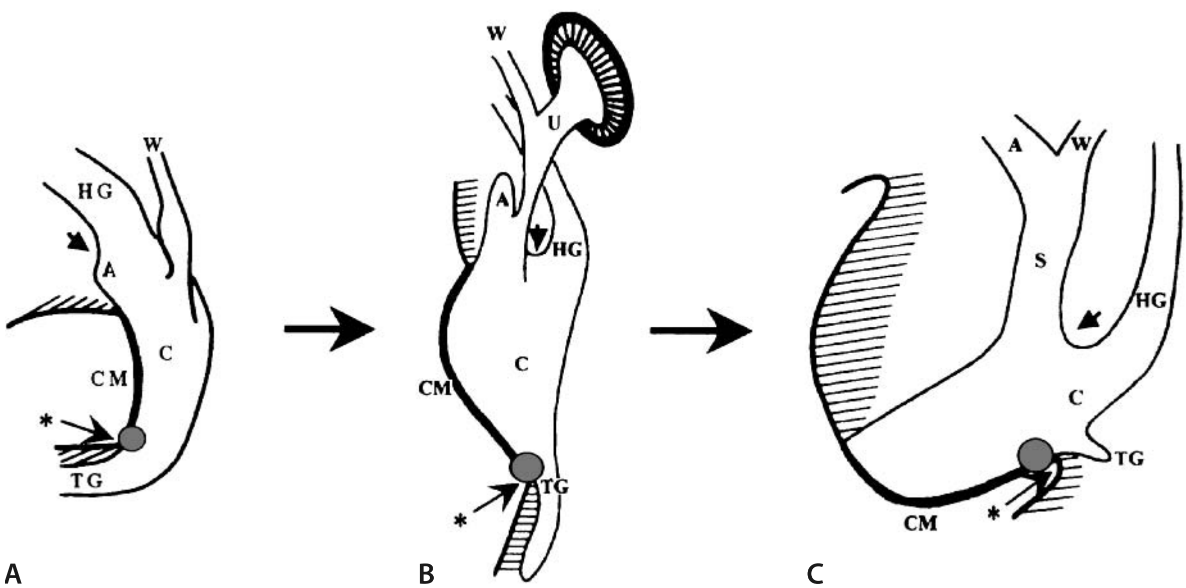


Fig. 4.7 Schematic drawing of normal cloacal development in rats (drawn after SEM photographs). **A** A 12.5-day embryo. **B** A 14-day embryo. **C** A 15-day embryo. Note the movement of the cloacal membrane (*CM*) from a vertical to a horizontal position. This movement is caused by the ventral outgrowth of the genital tubercle and the cloaca. Note the descent of the urorectal fold (*short arrows*). The dorsal part of the cloacal membrane (*gray dots*) is the area of the future anal opening. Arrows with an *asterix* point to the tail groove. This area is the fixed point in development of the cloaca. *S* Sinus urogenitalis, *W* Wolffian (mesonephric) duct, *U* ureter

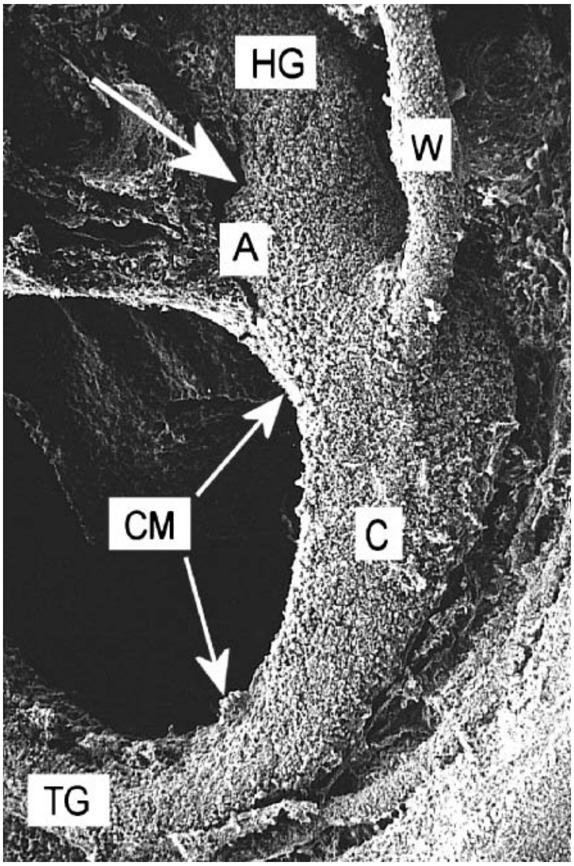


Fig. 4.8 Scanning electron micrograph (SEM) of the cloaca of a 12.5-day-old rat embryo. Lateral of view the cloaca after microdissection. The mesenchyme has been removed. See text for details. *Arrows* point to the cranial and caudal borders of the cloacal membrane. The *large arrow* points to the shallow urorectal fold

of the developing proximal urethra. The vaginal anlage derived from the paramesonephric ducts grows as a practically solid epithelial plate that gains access to the urogenital sinus through the openings of the regressing mesonephric ducts. Epithelial proliferation on the urogenital sinus adjacent to the developing vaginal plate is responsible for development of the hymen and introitus.

Disproportionate growth of the proximal urethra and development of the trigone of the bladder leads to elongation of the urethra and relative movement of the opening of the vagina caudally. As the vagina grows it gradually matches and then exceeds the size of the adjacent urethra such that the vagina that once developed in the mesenchymal wall of the urethra now houses the urethra in its anterior wall. The close relationship between vagina and urethra and their shared mesenchyme is borne out in the complications that beset those that have tried to separate them in surgical repairs of ARM.

4.4 SEM Studies in the Normal Embryonic Rat

In 1995, Kluth et al. [16] studied hindgut development in 245 staged rat embryos between the 10th and the 15th gestational day (comparable to human embryos between the 3rd and 7th week of gestation) using SEM [16]. The essential findings of the study are summarized in Fig. 4.7. In contrast to earlier reports, it was found that: (1) septation of the cloaca by means of fusion of the lateral folds does not take

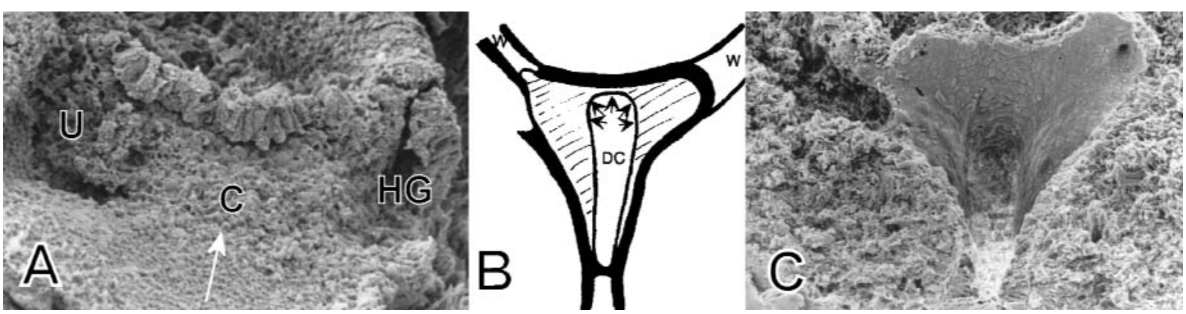


Fig. 4.9 SEM of the cloaca of a 14-day-old rat embryo. **A** Lateral view of the cloaca. A lateral ridge that would divide the dorsal and ventral cloacas is not seen (*arrow*). **B** Ventral view of the cloaca (schematic drawing of SEM shown in **C**). Signs of fusion of the lateral wall components are missing. The shape of the lower tip of the urorectal fold (*small arrows*) is evidence against fusion from cranial to caudal. *W* Left and right orifices of Wolffian ducts, *DC* dorsal (anorectal) part of the cloaca

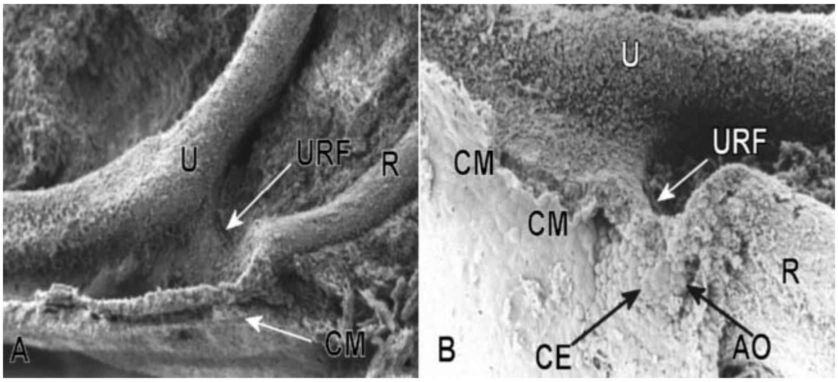


Fig. 4.10 SEMs of the cloacas of 16-day-old rat embryos. **A** Lateral view of the cloaca after microdissection. The mesenchyme has been removed. The urorectal fold (*URF*) has nearly reached the cloacal membrane. **B** In this slightly older embryo, the tip of the urorectal fold has reached the level of the cloacal membrane. Local disintegration of the cloacal membrane is obvious. *U* Urethra, *R* rectum, *AO* anal opening, *CE* cloacal epithelium

place, and (2) migration of the anal opening cannot be observed.

4.4.1 Early Cloacal Development

The starting point of this series was the cloaca in an 12.5-day-old rat embryo (Fig. 4.8). At this stage, all features of a typical cloaca were present: the hindgut (HG) enters the cloaca (C) from dorsocranial, while the allantois (A; the forerunner of the bladder) can be identified as a cranioventral diverticulum. Between this diverticulum and the hindgut, the urorectal fold (arrow) can be seen. This fold marks the cranial border of the undifferentiated hindgut, the so-called cloaca. The mesonephric duct (Wolffian duct; W) enters the cloaca in its cranial part but in a relatively dorsal position. Caudally, the cloaca continues directly into the tailgut (TG). The cloacal membrane (CM) extends in a slight concave curve from the caudal border of the body stalk to the tail, where the tailgut enters the cloaca. At this stage, the cloaca has the shape of a triangle standing on its top. A genital tubercle is missing. In the subsequent stages, the cloacal shape starts to change. This is caused by the ventral growth of the genital tubercle, a process that can be traced easily in a 14-day-old rat embryo (Fig 4.9B). This growth results in two processes: (1) a remarkable outgrowth of the cloaca in a ventral direction, and (2) a rectangular displacement or rotation of the cloacal membrane (Fig. 4.9A–C), which swings down from a vertical to a horizontal position.

4.4.2 The Septum in Normal Cloacal Development

In a 12.5-day-old rat embryo (Fig. 4.8), a tiny depression can be noted between the diverticulum of the urachus and the rectum. This fold is the first indication or the so-called urogenital septum. Using the junction between the mesonephric duct and the cloaca as a marker, the relative movement of this fold can be discerned with ease (Fig. 4.7). To see directly what happens during this so-called process of septation, we sagitally opened cloacas of 13-day-old rat embryos to inspect them from inside. However, there was no sign of the lateral cloacal ridges or of fusion of the lateral cloacal wall components (Fig. 4.9).

4.4.3 The Fusion of the Urorectal Fold with the Cloacal Membrane in Normal Cloacal Development

In our studies we saw a disintegration of the cloacal membrane in the area where the tip of the urorectal fold meets the cloacal membrane (Fig. 4.10).

4.4.4 The Region of the Future Anal Orifice

It is interesting to note that in the period of ventral cloacal rotation (between day 11 and day 15), the dorsal part of the cloacal membrane and the dorsal cloaca always remains in close contact with the tail region. This region, which carries the anlage of the future anal orifice, is the "fixed point" in cloacal development (Fig. 4.7).

4.4.5 Nomenclature

It must be kept in mind that the term "cloaca" is used to describe not only a transitional organ system in human embryos, but also a congenital anomaly and a normal organ in birds. This can lead to the false conclusion that the morphology of these three entities is similar, which is not the case. Despite the same name, embryonic cloacas are completely different morphologically from cloacas in females with ARM and in birds. The main difference is the presence/absence of the area of the future anal opening. In embryonic cloacas, the future anal region is always present, whereas the future anus is always missing in the human malformation that we call "cloacas". This confusion in the terminology is, as mentioned previously, the result of two outdated theories: Haeckel's [1] "biogenetic law" and the theory of the malformation as a "frozen" stage of normal embryology ("Hemmungsmißbildung") [3].

4.4.6 Conclusions From SEM Studies

The SEM studies described here indicate clearly that the subdivision of the cloaca is not the result of a process of fusion of lateral cloacal wall components [2]. In our opinion, the importance of the process of septation has been overestimated in the past. According to these results, the normal development of the hindgut depends primarily on the normal formation of the cloacal membrane. In all normal embryos, we could identify the region of the future anal orifice in the dorsal part of the cloacal membrane close to the tail groove. This observation makes the theory of a migration of the rectal opening to the perineum obsolete. Furthermore it is obvious from our SEM observations that the embryonic cloaca never passes through a stage that is similar to any form of ARM in neonates, including the so-called cloacas in females.

The most impressive feature in most cases of ARM is the missing anus, which seems to enter the urogenital tract as an "ectopic" rectal opening or is simply misplaced ventrally into the perineum. Obviously, this misplacement cannot be explained by a faulty septation alone because this would result in a persistent embryonic cloaca, with the area of the future anal orifice still in its place. According to these findings, the relative downgrowth of the urorectal septum is the result of normal cloacal development, not its cause. A fusion of the urorectal fold with the cloacal membrane could not be observed. When the fold comes into contact with the cloacal membrane, it disintegrates locally.

4.5 Abnormal Anourogenital Development

Thus far there is only one study of human embryos at an early stage of maldevelopment. Padmanabhan et al. [25] examined embryos of 7.5 and 8 postovulatory weeks that had a blind-ending rectum, abnormal genital tubercle development, and sacral vertebral column defects. One also exhibited agenesis of the ureters. Our understanding of abnormal development of the anogenital region depends principally on observations made in other mammalian embryos combined with detailed histological examination of

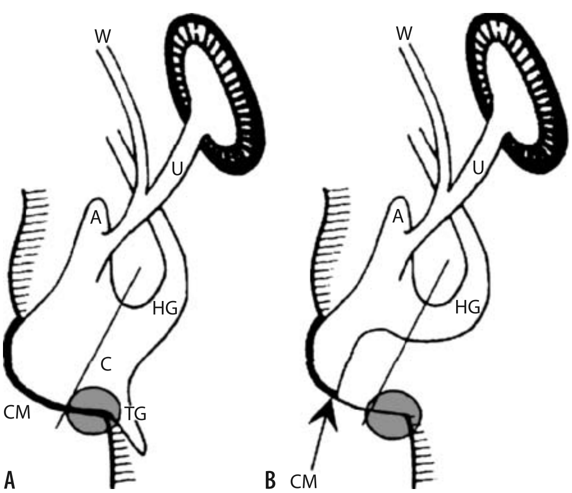


Fig. 4.11 Schematic drawings of a normal (**A**) and an abnormal (**B**) cloaca. In the abnormal embryo, the cloacal membrane is too short (*arrow*); it does not extend to the region of the tail groove (*gray area*). The dorsal cloaca is missing. In the normal embryo (A), the cloacal membrane is of normal length and extends to the region of the tail groove (*gray area*)

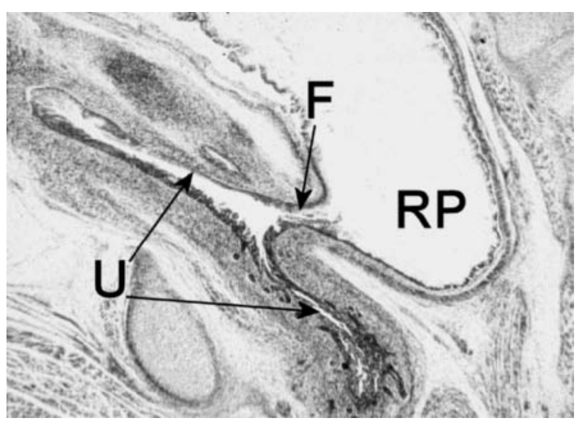


Fig. 4.12 Histological section of the pelvic organs of an SD-mouse (newborn). This newborn presents the features of anorectal malformations (ARM) with a rectourethral fistula (*F*) and a blind ending rectal pouch (*RP*)

human fetuses and infants who succumbed to multiple malformations, including ARM. The observations in animal embryos fall into three broad categories: embryos from strains of animals with a high incidence of hereditary ARM, embryos exposed to teratogens in early development, and genetically modified strains of mice where a single gene product has been "knocked out," resulting in ARM. Each group of data add to our overall understanding of the likely pathogenesis of ARM in humans.

4.5.1 Hereditary Congenital Malformations in Pigs and Mice

Detailed reports are available regarding the embryonic development of timed pregnant Dutch minipigs [22,26] and short-tailed Danforth mice [27]. In all of these animals there was a high incidence of congenital ARM. In the pig population there were also many animals with normal anorectal development, allowing a direct comparison of normal and abnormal embryos. The common finding in all of these studies was that the all of the abnormal embryos had defects in the dorsal part of the cloacal membrane and the adjacent dorsal cloaca from the earliest stages of development studied (Fig. 4.11). The size of the defect in the cloacal membrane and dorsal cloaca determines the severity of the ARM and the level of communication between the hindgut and the urogenital sinus. The reason why the defect is present in the dorsal cloaca and cloacal membrane is speculative, but the possibilities include abnormal infiltration of mesenchyme from the primitive streak into the caudal part of the early cloacal membrane, abnormal apoptosis of the dorsal cloacal membrane and adjacent cloaca, or disturbance of growth in the region of the dorsal cloaca and cloacal membrane.

4.5.2 Abnormal Cloacal Development

In 1940, a mutant of the normal house mouse, the SD-mutant, was described by Dunn et al. [28]. These mice, first bred by Danforth [29], prominently feature a short tail and therefore also are known as "Dan-

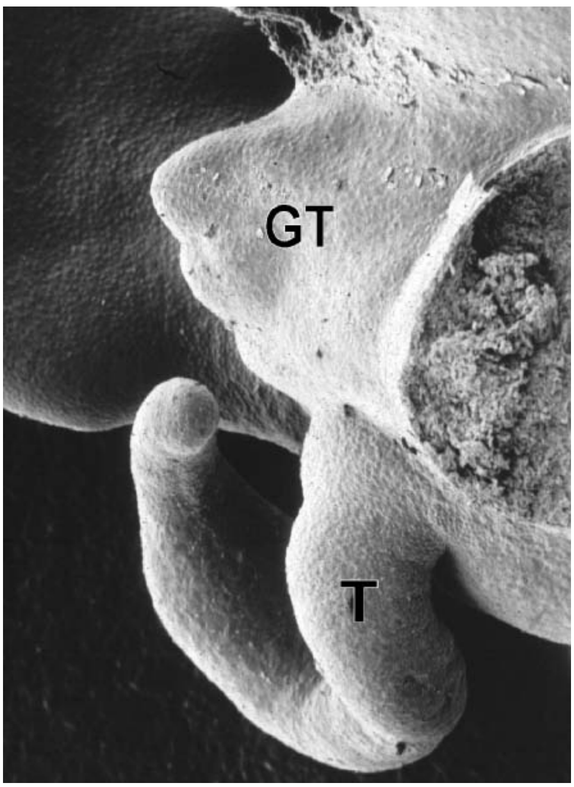


Fig. 4.13 SEM. An abnormal SD-mouse embryo. Note the crippled tail and the hypoplastic genital tubercle

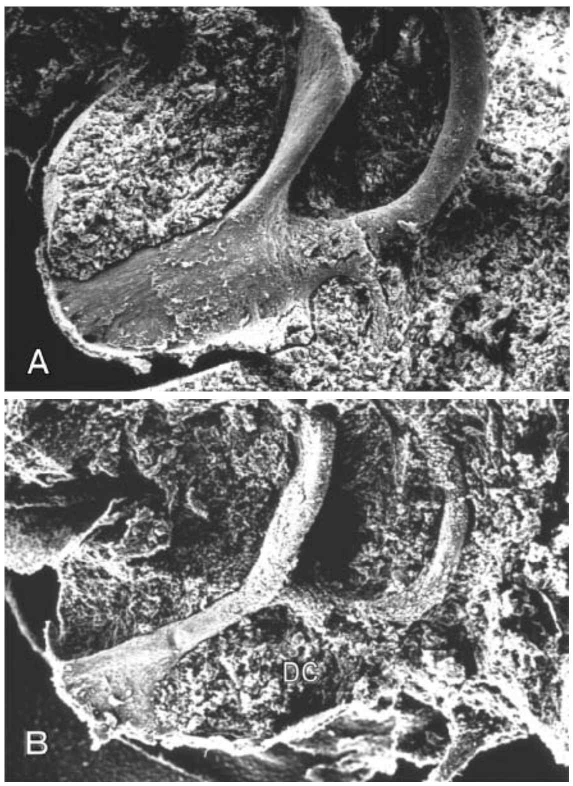


Fig. 14 SEM of a normal (A) and an abnormal (B) 14-day-old SD-mouse embryo. The findings are identical to those shown in Fig. 4.11. Note that the dorsal cloaca (*DC*) is missing

forth's short-tail mice" [30]. However, the SD gene influences not only the axial skeleton, but also the rectum and the urogenital system, causing a spectrum of ARM and urogenital anomalies [30]. Recently, we analyzed the spectrum of ARM in this model [27]. The pathologic-anatomic findings in the heterozygous (SD/+) SD-mouse group (Fig. 4.12) were identical to those described earlier in pigs [13] and humans [6,31]. Since the percentage of abnormal animals per litter is high and breeding of SD-mice is simple and inexpensive, we believe that the SD-mouse model is ideal for studying the embryological background of disturbed cloacal development.

4.5.2.1 Recent Studies in SD-Mouse Embryos

The SD-mice used for this study originally were received from Philip Harris Biological (UK), in 1985, and were subsequently bred continuously in our facility in accordance with German federal and local regulations. A total of 80 abnormal SD-mouse embryos were easily identified by their shortened or crooked tails. In several of these, the genitals were also abnormal (Fig. 4.13). After microdissection, typical morphological changes could be observed when abnormal and normal cloacas were compared (Figs. 14A, B). In all abnormal cloacas, we found: (1) the cloaca had an unusual shape – the dorsal cloaca was always missing, (2) the cloacal membrane was too short – in all cases the dorsal part of the cloacal membrane was absent, and (3) an abnormal junction between the proximal hindgut and the cloaca.

4.5.2.2 Drug-Induced Deformities in Rodents

Several agents have been identified that, when given to timed-pregnant rodents at the appropriate time in development, result in a high incidence of ARM. Etretinate, a long-acting synthetic retinoid, has been the most widely studied in mice and produces ARM together with several other deformities in that region and elsewhere. Ethylenethiourea has also been used with similar results. Adriamycin, in rats a potent inducer of abnormalities similar to those seen in the VACTERL (vertebral anomalies, anal atresia, cardiac defect, tracheoesophageal fistula, renal (kidney) abnormalities, and limb abnormalities) anomaly in humans, also produces variable ARM. Unfortunately it also uniformly causes bladder agenesis in rats, thus muddying the interpretation of changes in the cloacal region that might be related to anorectal development.

Again the findings in the embryos treated with teratogens at embryo day 9 in mice is that of a deficiency of the dorsal cloacal membrane and the adjacent dorsal cloaca [32]. In addition, etretinate also has an effect on the proliferation of neuroepithelial cells in the presacral region, resulting in anterior sacral myelomeningocele. These animals also developed significant abnormalities in the sacral vertebrae and short or absent tails. Most of the ARM that developed were high lesions with rectourethral fistulas in males or "cloacal" malformations in the females. Embryos exposed to etretinate at an earlier stage of development have more severe deformities and many do not survive, but of those that are not resorbed, many display syringomyelia, with the total absence of lower midline trunk and genital structures and fusion of the lower limb anlage (unpublished data). This would support the suggestion by Liu et al. [32] that the effects of etretinate are initiated by its effects on the tail bud/caudal eminence.

Given that the primitive streak is responsible late in its development for the production of mesenchyme in the region of the distal cloaca as well as the development of the caudal segments of the embryo (i.e., sacral somites and neural tissue and tail gut), it seems likely that this is the site of action of etretinate that ultimately results in the development of ARM. In other words, ARM may develop not, as was once thought as a result of excessive regression of the embryonic tail (caudal regression syndrome) but as a result of a primary abnormality of "tail" development.

4.5.2.3 Genetically Modified Rodents

Abnormalities in SHH, Gli2, Gli3, Hoxa-13/Hoxd-13 and bone morphogenic protein 4 (BMP4) expression have all been linked to abnormal anorectal development, although there also appears to be a link to the effects produced by retinoids. Retinoids play an essential part in normal growth and development and are thought to act at least in part by regulating the expression of *Hox* genes as well as the distribution of retinoic acid receptors. *Hox* genes in the mesoderm are also regulated by the protein product of the sonic hedgehog (Shh) gene. Mutant mice that do not express Shh also exhibit a range of ARM similar to those found in humans [33].

Shh is produced in epithelial cells and also induces BMP4 in the mesoderm (an example of epithelial-mesenchyme interaction). Animals with defective *Shh* genes have isolated anal malformations rather than the regional deformities noted in Hoxa-13/Hoxd-13 mice and those treated with retinoids [34,35]. Animals treated with Retinoids show defects in Shh and BMP4 expression in both rectal and urogenital tissues compared to controls [36]; however, information demonstrating its effects at the stage when the cloacal membrane is first being formed is yet to be presented.

Other gene products known to relate to epithelial adhesion (or de-adhesion) [37] have also been linked to ARM in gene-knockout models. The morphological appearance of the developing anorectum in all of these models is similar to those seen in the teratogenic and hereditary models, that is a shortening of the cloacal membrane and deficiency of the dorsal cloaca. Fgf10 has also been reported as being important in anorectal development; however, the null mutant mouse strain exhibits rectal agenesis and is therefore not included for discussion here.

4.5.2.4 Detailed Histology of Affected Human Infants

The histological analysis of specimens from human fetuses and newborns with nonviable malformations including imperforate anus [18] has revealed the following main findings:

1. The malformation typically affects the anal canal rather than the rectum, which is generally only secondarily affected.
2. The anal canal is displaced ventrally and ends either on the perineum or forms an anourogenital connection. The communications demonstrate a lining of pseudostratified columnar epithelium from which extends anal glands. Around this is a lamina propria of very dense, longitudinally oriented stroma with longitudinal grooves and ridges and fibromuscular differentiation, surrounded by a smooth muscle coat that is an extension of the muscularis propria of the rectum. Connections to the perineum have features of both the superficial and deep parts of the anal canal, while connections to the urogenital sinus have features of the deep part of the anal canal only and were therefore not truly ectopic anal canals.
3. In those specimens where the rectum is blind-ending there are commonly found signs of a partly regressed preexisting connection. Such a connection is more likely to be lost in a region where there is rapid growth (e.g., midline perineum in the male). Some have no anal elements at all and are a truly blind-ending rectum, while others have a transition from rectum to anal canal but then have no further connection to the urogenital structures.
4. Where there is a communication from the rectum to the urogenital sinus structures (rather than the perineum) there is a gradual transition from the anal mucosa normally found in the proximal anal canal, into the mucosa of the urogenital system. In rectovestibular fistulas in females, the transition is from proximal anal canal mucosa to the mixed pseudostratified columnar and metaplastic noncornifying stratified squamous epithelium of the vestibulum. This would concur with the clinical impression reported by Peña that these lesions should be characterized as proximal rather than distal lesions [38].
5. The most proximal connections to the urogenital sinus are found in the region of attachment of the mesonephric ducts to the urogenital sinus. In the so-called rectovesical fistulas, the rudimentary prostate gland is found consistently in the region where the abnormal anal canal meets the urogenital structures [6]. In these specimens the development of the trigone of the bladder, the upper urethra, and the urethral sphincter is also abnormal. In the female, vaginal development is grossly disturbed leading to an abnormal persistence of the urogenital sinus caudal to the mesonephric ducts (referred to clinically as a persistent cloaca).
6. The striated muscles of the perineum often have a very abnormal configuration, with the fibers of the external anal sphincter forming a median concentration of longitudinal bundles in the absence of an anal canal. In high lesions the bulbospongiosus muscle is also displaced medially. The external urethral sphincter and ischiocavernosus muscle are also variably affected, as is the puborectalis sling, depending on the severity of the lesion.
7. The more proximal the connection between rectum and structures of the urogenital sinus, the greater the likelihood of associated abnormalities in the development of the pelvis and perineum as well as bladder, ureters, and phallus.

4.6 Proposed Pathogenesis of ARM in the Human

Many of the previous theories regarding the pathogenesis of ARM in humans were based on theories of normal development that have not stood up to scrutiny. It is now clear that lateral fusion plays no part in the division of the cloaca into anal and urogenital parts, nor does fusion of an anorectal septum with the cloacal membrane. The distribution of common sites of anogenital communication (or lack thereof) reflects the sites of rapid localized growth within the embryo rather than a series of layers of lateral fusion, as has been previously hypothesized. The evidence from animal models and from the detailed study of human fetuses with major anomalies suggests strongly that the earliest morphological defect leading to ARM is a deficiency in the dorsal component of the cloacal membrane and the adjacent dorsal cloaca. This is likely to be consequent on a malfunction, as yet ill-defined, of the primitive streak and tail bud, very early in the development of the caudal part of the embryo.

The extent of the defect in the dorsal part of the cloacal membrane determines the severity of the defect that develops. The smaller defects lead to distal defects and anocutaneous fistulas, covered anus, and so on. The larger defects are associated with major malformations in the region as well as urogenital fistulas and occasionally even abnormalities in the development of the genitourinary sinus also with urethral hypoplasia and genital and scrotal malformations. The larger defects are also associated with abnormal development of the striated muscle that would normally develop in the mesenchyme associated with the missing cloacal part, namely the anal and urethral sphincters, the bulbo and ischiocavernosus muscles, and the pelvic floor.

4.7 Conclusions

The embryology of the anal and urogenital region is quite different from that which has been understood by the pediatric surgical community until now. Previous classifications of ARM based on surgically apt theories of development must now be revised in the light of this new (and sometimes really quite old) evidence that has to date been ignored. A better understanding of the normal stages of anorectal and urogenital development will enhance future investigation, particularly into the molecular and genetic basis of ARM. This may in the future identify factors responsible for promoting the ARM, and the relationship between the adjacent structures may temper surgical expectations from repair.

The evidence that is currently available would support a classification of ARM that describes merely high and low (proximal and distal, severe, and mild) lesions with or without communication (see Chap. 8). In the female, communication between the rectum and vestibulum should be considered "high" lesions. Abnormalities affecting the development of the vagina where there is a persistence of the urogenital sinus below the openings of the vaginae might be more accurately referred to as persistent urogenital sinus with high ARM rather than a persistent cloaca, as has been the case to date. Finally, the term "caudal dysgenesis sequence" should replace "caudal regression sequence" to describe the association between ARM and other abnormalities of caudal embryonic development.

References

1. Haeckel E, cited in Starck D: (1975) Embryologie (3rd edn). Thieme, Stuttgart, Germany
2. Gilbert SF (2003) Developmental Biology (7th edn), Chapter 23. Sinauer, Sunderland, MA
3. Schwalbe E (1906) Die Morphologie der Missbildungen des Menschen und der Tiere. 1. Teil Allgemeine MiBbildungslehre (Teratologie). Gustav Fischer, Jena, Germany, pp 143–144
4. Tourneux F (1888) Sur les premiers développements du cloaque du tubercule génital et de l'anus chez l'embryon de mouton. Anat Physiol 24:503–517
5. Retterer E (1890) Sur l'origin et de l'evolution de la region ano-gentiale des mammiferes. J Anat Physiol 26:126–210
6. Stephens FD (1963) Congenital Malformations of the Rectum, Anus, and Genitourinary Tract. Livingstone, Edinburgh, UK
7. Politzer G (1931) Uber die entwicklung des dames beim menschen. Z Anat Entwicklungsgesch 95:734
8. Politzer G (1932) Uber die entwicklung des dammes beim menschen. Z Anat. Entwicklungsgesch 97:622
9. Wijnen HP (1964) Hypothesen over enkele congenitale vitia van het caudale gedeelte van het menselijk lichaam aan een morphologisch embryologisch onderzoek getoetst. Doctoral Thesis, University of Amsterdam
10. Ludwig KS (1965) Ueber die beziehungen der kloakenmembran zum septum urorectale bei menschlichen embryonen von 9 bis 33 SSL. Z Anat Entw Gesch 124:401–403
11. Penington EC, Hutson JM (2003) The absence of lateral fusion in cloacal partition. J Pediatr Surg 38:1287–1295
12. Kluth D, Lambrecht W (1997) Current concepts in the embryology of anorectal malformations. Semin Ped Surg 6:180–186
13. van der Putte SCJ (1986) Normal and abnormal development of the anorectum. J Pediatr Surg 21:434–440

14. Bill AH, Johnson RJ (1958) Failure of migration of the rectal opening as the cause for most cases of imperforate anus. Surg Gynecol Obstet 106:643–651
15. Gans SL, Friedman NB (1961) Some new concepts in the embryology, anatomy, physiology and surgical correction of imperforate anus. West J Surg Obstet Gynecol 69:34–37
16. Kluth D, Hillen M, Lambrecht W (1995) The principles of normal and abnormal hindgut development. J Ped Surg 30:1143–1147
17. Penington EC, Hutson JM (2002) The cloacal plate: the missing link in anorectal and urogenital development. BJU Int 89:726–732
18. van der Putte SC (2005) The development of the perineum in the human. A comprehensive histological study with a special reference to the role of the stromal components. Adv Anat Embryol Cell Biol 177:1–131
19. Airhart MJ, Robbins CM, Knudsen TB, Church JK, Skalko RG (1996) Developing allantois is a primary site of 2'-deoxycoformycin toxicity. Teratology 53:361–373
20. Smits-van Prooije AE, Vermeij-Keers C, Poelmann RE, Mentink MMT, Dubbeldam JA (1988) The formation of mesoderm and mesectoderm in 5- to 41-somite rat embryos cultured in vitro, using WGA-Au as a marker. Anat Embryol 177:245–256
21. Kluth D, Lambrecht W, Reich P (1988) Pathogenesis of hypospadias – more questions than answers. J Pediatr Surg 23:1095–1101
22. van der Putte SCJ, Neeteson FA (1983) The normal development of the anorectum in the pig. Acta Morphol Neerl Scand 21:107–132
23. Paidas CN, Morreale RF, Holoski KM, et al (2000) Septation and differentiation of the embryonic human cloaca. J Ped Surg 34:877–884
24. Forsberg JG (1961) On the development of the cloaca and the perineum and the formation of the urethral plate in female rat embryos. J Anat 95:423–435
25. Padmanabhan R, Naruse I, Shiota K (1999) Caudal dysgenesis in staged human embryos: Carnegie stages 16–23. Am J Med Genet 87:115–127
26. Hori T, Giuffra E, Andersson L, Ohkawa H (2001) Mapping loci causing susceptibility to anal atresia in pigs, using a resource pedigree. J Pediatr Surg 36:1370–1374
27. Kluth D, Lambrecht W, Reich P, et al (1991) Sd-mice – an animal model for complex anorectal malformations. Eur J Pediatr Surg 1:183–188
28. Dunn LC, Gluecksohn-Schoenheimer S, Bryson V (1940) A new mutation in the mouse affecting spinal column and urogenital system. J Hered 31:343–348
29. Danforth CH (1930) Developmental anomalies in a special strain of mice. Am J Anat 45:275–287
30. Kluth D, Kaestner M, Tibboel D, et al (1995) Rotation of the gut: fact or fantasy? J Pediatr Surg 30:448–453
31. Gray SW, Skandalakis JE: (1972) Embryology for Surgeons. Saunders, Philadelphia, PA, pp 87–216
32. Liu Y, Sugiyama F, Yagami K, Ohkawa H (2003) Sharing of the same embryogenic pathway in anorectal malformations and anterior sacral myelomeningocele formation. Pediatr Surg Int 19:152–156
33. Sukegawa A, Narita T, Kameda T, Saitoh K, Nohno T, Iba H, Yasugi S, Fukuda K (2000) The concentric structure of the developing gut is regulated by Sonic hedgehog derived from endodermal epithelium. Development 127:1971–1980
34. Kimmel SG, Mo R, Hui CC, Kim PC (2000) New mouse models of congenital anorectal malformations. J Pediatr Surg 35:227–230
35. Mo R, Kim JH, Zhang J, Chiang C, Hui CC, Kim PC (2001) Anorectal malformations caused by defects in sonic hedgehog signalling. Am J Pathol 159:765–774
36. Sasaki Y, Iwai N, Tsuda T, Kimura O (2004) Sonic hedgehog and bone morphogenetic protein 4 expression in the hindgut region of murine embryos with anorectal malformations. J Pediatr Surg 39:170–173
37. Dravis C, Yokoyama N, Chumley MJ, et al (2004) Bidirectional signalling mediated by ephrin-B2 and EphB2 controls urorectal development. Devel Biol 271:272–290
38. Peña A (1995) Atlas of Surgical Management of Anorectal Malformations. Springer-Verlag, New York
39. van der Putte SCJ, Neeteson FA (1984) The pathogenesis of hereditary congenital malformations of the anorectum in the pig. Acta Morphol Neerl Scand 22:17–40

5 Recent Advances Concerning the Normal and Abnormal Anatomy of the Anus and Rectum

Michael R.Q. Davies and Heinz Rode

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5.1 Introduction

Knowledge about a problem brings with it understanding, allowing assessment of the situation and its ultimate solution. This flow of activity is interrupted where knowledge remains conjectural and unclear. This state, which is characterized by uncertainty, clouds what is written and spoken about by experts on the subject of congenital anorectal malformations (ARM). Major significant recent advances have been made, but ignorance due to a lack of clarification on the subject still pervades much of what we think is the truth concerning these malformations. Because of this sorting out of "fact from fiction," finding the true pathway to follow through the literature on the subject is extremely difficult and subject to observer bias.

5.2 The Pelvic Floor

The sheet-like muscle of the pelvic floor has been arbitrarily divided into seemingly different muscles based on their site of origin and perhaps insertion. This is of no practical importance to the pediatric surgeon. Furthermore, conventional morphologic descriptions, those of the anatomist, are of limited value to the clinical practitioner [12]. They do, however, form the basis, the core, around which the pelvic structures can be discussed. In practice what can be visualized in the living provides more guidance and insight to the nature of the problem addressed [91]. It is for this reason that imaged appearances (sonar, magnetic resonance – MR imaging, and computed tomography – CT scan) of the pelvic structures discussed are used in preference to those seen by the anatomist [120].

5.2.1 Levator Ani

This is the major muscle of the pelvic diaphragm and is attached anteriorly to the pubic bone. The ventromedial segment, is termed the pubovisceralis muscle as it holds the urethra, vagina, and anorectum within its sling-like fibers. It is drawn caudally by the viscera passing through it to which it is attached. A further subdivision of this muscle, a segment composed of fibers passing, but intimately in contact with, the anorectum in the shape of a U-loop from pubis to pubis is named the puborectalis. It forms the anorectal junction, defining the rectum from the surgical anal canal, the anorectal ring. This muscle participates in the formation of the external anal sphincter, from which it can be distinguished, forming the most cranial significant component of this structure. The pubovisceralis is part of the levator, separated from it by its function, closing the urogenital and anorectal hiatuses by contraction.

5.2.2 The Nerve Supply

Gross dissection studies carried out in the female suggest that the levator muscle is not innervated by the pudendal nerve, but by a levator ani nerve (S3–S4), which travels on the superior surface of the pelvic floor and is made up of nerves that are direct branches that split from nerve roots proximal to the sacral plexus [4]. It should be noted that this supply is anatomically distinct from that of the external anal sphincter, which is supplied by nerve fibers traveling with the pudendal nerve [73]. This dual motor supply indicates that the pelvic floor muscles do not necessarily have to behave as a unitary mass. This has been demonstrated in the cat, where bilateral section of the pudendal nerves entirely abolishes both the tonic activity and phasic responsiveness of the external anal sphincter without affecting the activity of the levator ani [27]. In spite of proposals that the external anal and urethral sphincters and bulbocavernosus muscle originate from the puborectalis and can be demonstrated to act as one muscle (they contract or relax en masse), a voluntary selective individual muscle function exists in man, such that each named component can act independently of the other [108–110]. Yet to be clarified in the human is the suggestion that there is a clear segregation of the segmental motor neuron pools innervating these muscular sections of the pelvic floor. Quantitative analysis of these motorneurons in the rat model with ARM of various forms has shown that there are significantly fewer motor nerve cells in these cases [135].

Evolutionary considerations regarding the pelvic floor muscles indicate that they develop with specific attachments and function; they do not derive from preexisting muscles [26]. For instance, the external anal sphincter has strong connectivity with its surrounding skin and has muscle spindles, but it is devoid of the phasic monosynaptic component of the stretch reflex. Similarly, in quadrupedal mammals it has fast-twitch muscle characteristics, but in the human it is a slow-twitch muscle. Onuf's nucleus, which innervates the pelvic floor muscles and is suggested to occupy an intermediate position between the visceral and somatic nuclei, receives an important group of suprasegmental afferents, probably direct corticospinal fibers.

Sensory perception originating from the levator muscles plays a fundamental role in signaling the arrival of rectal content at the pelvic floor. Its role in anal continence mechanisms is undisputed. Sphincter-saving operations in humans have shown clearly that the rectum is essential for neither the appreciation of impending evacuation, nor for the sphincter inhibitory reflexes (rectoanal reflex) [62]. Because of this, sensory nerve endings in the puborectalis and anal region have been evaluated in humans with ARM and equated with normal material [65,82]. The reported findings must be interpreted in light of the fact that these sensory receptors appear to present themselves at different ages (i.e., there are no sensory nerve endings in the anal canal of the newly born, but they are abundant in later life). In normal pelvic muscle, muscle spindles are not present in every part, being well represented in only the front two-thirds of the puborectalis and the middle segment of the external anal sphincter.

In ARM, not only are these monitoring structures decreased in number, but their location is limited to the middle segments of the puborectalis; the greater the degree of developmental regression, the greater the impact on this alteration (i.e., the decrease in density corresponds to the degree of anorectal agenesis).

Structural differences have been shown in the human with regard to the ultrastructure of the periurethral levator ani muscle. The external urethral sphincter consists of a single population of type I (slow-twitch) fibers in the absence of muscle spindles; in contrast, the periurethral levator possesses muscle spindles and a heterogeneous population of type I and type II (fast-twitch) fibers, indicating that it could be of considerable importance in producing active urethral closure during continence [43, 137].

5.3 The Anal Canal

This section of the terminal part of the hindgut remains difficult to define; it means different things to different people. To anatomists, the canal has clearly defined borders: from the anal valve line (pectinate line) to the anus. To the surgeon, however, its upper margin is determined by puborectalis contraction – the upper border of the anorectal ring. Seen from a logical point of view, the intrinsic structures of the rectal wall stretch nearly to the skin, the anal canal being that part of it held within the external anal sphincter. This implies that the anal canal is a modified part of the rectum [107]. New tissue introduced from surface structures is only encountered within its wall at and distal to the pectinate (valve) line.

In ARM the anal canal may be either agenetic, where the terminal hindgut is not surrounded by a well-formed external anal sphincter, or dysgenetic, where the terminal hindgut is incompletely formed but surrounded by an external sphincter [30,113].

5.3.1 The Anal Canal in ARM

The following analysis is based on the assumption that the following anatomical considerations are correct:

1. The anal canal stretches from the anorectal ring to the anus; it is defined and its length is determined by the presence of the striated muscle sphincter that surrounds it.
2. That the canal is divided into two component parts: an upper canal formed intrinsically from structures of rectal origin (embryologically of endodermal origin) and a lower canal composed of tissue of somatic (surface) origin (embryologically of ectodermal origin). This is not absolute as endodermally derived structures of rectal origin are contained within its wall. The site of anatomical division between the parts of the canal is the pectinate or anal valve line.
3. Congenital malformations involving the canal are classified as: (1) rectal malformations, where the upper canal and rectum are primarily involved, and anal malformations, where the lower canal, external genital, and cutaneous structures are primarily involved.
4. In rectal malformations, the malformed endodermally derived tissues retain the anatomical features of the parent structures to variable degrees [60]. In anal malformations, the ectodermally derived anatomical components do not retain these features.

Given these assumptions, certain conclusions can be drawn. These are now discussed.

5.3.1.1 Agenesis of the Anal Canal

In agenesis of the anal canal, the hindgut terminates above the anorectal ring or at the pelvic floor with a noncommunicative (blind) pouch or a pouch that communicates by fistula with an epithelial surface. The malformation involves the rectum and the upper anal canal. It is therefore a rectal anomaly. Where a fistula is present, it retains, as predicted, anatomical features of its parent structure the rectum. Because of this the fistula is correctly called a terminal channel: a rectal terminal channel in this instance. Indeed, any fistula with its origin in the rectum or upper anal canal should be given this name. This is of importance anatomically as the fistulae that arise in the lower anal canal differ. This terminology is used as follows: (1) the Fistula's site of origin is the rectum/upper anal canal, (2) Its endpoint is the bulbar urethra, (3) the name used is anal canal agenesis with terminal rectobulbar urethral channel. When this terminal channel traverses, or moves through, the muscular structures of the pelvic floor, it picks up (gains) a coating of striated muscle fibers, which are likened to a forme fruste of an external anal sphincter. Because of this, from a functional point of view, under physiological circumstances, it can act as an anal canal. It is either: (1) a pressure-determined incontinent (refluxing) channel, or (2) a stress-controlled continent channel. The factor that determines this difference is the length of the translevator channal, which is an obvious variable.

5.3.1.2 *Dysgenesis of the Anal Canal*

In dysgenesis of the anal canal, the hindgut terminates beneath the anorectal ring, at or below the pectinate or anal valve line, as a noncommunicative (blind) pouch, or the pouch communicates by fistula with an epithelial surface. Where the fistula is present it retains none of the features of its parent structure, the lower anal canal, as this structure is anatomically malformed. Because of this the fistula is correctly called a terminal tract. This terminology is used as follows: (1) the Fistula's site of origin is the lower anal canal, (2) Its endpoint is the genital or perineal skin, (3) the name used is anal canal dysgenesis with terminal anocutaneous tract. The anatomical distinctions to be emphasized here are that the tract does not have the mural features of the rectal or normal anal wall and that it loses (sheds) surrounding striated muscle fibers. Functionally, it is not a sphincter-controlled communication.

5.4 The Anal Canal – Relevant Anatomy

For descriptive purposes the anal canal can be divided endosonographically into three sections [37]:

1. The high anal canal: A level midway between the inferior border of the puborectalis and complete formation of the external sphincter ring anteriorly.
2. The middle anal canal: Completion of the external ring anteriorly in combination with maximal internal sphincter thickness.
3. The low anal canal: Immediately caudal to the termination of the internal sphincter, it comprises the superficial external sphincter.

The length of the normal anal canal (external sphincter length) varies with age. There are no accurate data for the term baby, but measurements are available for children (Table 5.1). In adults there are no statistically significant gender-determined differences in overall canal length (Table 5.2). Females have a significantly shorter external anal sphincter anteriorly [9]. In ARM, measurements of the length of potential sphincter muscles have been reported upon; predictably influenced by the severity of the malformation.

5.4.1 Potential Sphincteric Structures

The striated muscle complex (SMC) and parasagittal fibers have been visualized using three-dimensional

Table 5.1 The length of the normal anal canal as determined sonographically in healthy children [6]

	Sphincter length (mm)	Range (mm)
At rest	20.9 ± 3.1	17–28
At squeeze	22.4 ± 4.4	17–31

Table 5.2 The length of the normal anal canal as determined by magnetic resonance (MR) imaging in the adult [102]

	Sphincter length (mm)
Females	
• Lateral wall	27.1 ± 5.4
• Anterior wall	14.0 ± 3.0
Males	
• Lateral wall	28.6 ± 4.3
• Anterior wall	27.0 ± 5.3

image reconstruction in patients before anorectoplasty and in controls [88,117,118]. The data, which have not been confirmed, have been reported to show: extreme variation, with differing fiber configurations between cases and controls [128]. At a level where the parasagittal and vertical fiber components of the potential striated sphincter muscles meet there is scanty representation, in comparison to controls, of vertical components in major malformations. In major malformations there is an increased length of parasagittal fibers in comparison to controls. Variations due to age were eliminated.

5.4.1.1 *Endoanal Anatomy*

The mucosal type and pattern in terminal rectal channels emulates that found in the normal upper anal canal [39]. Rectal mucosa gives way to transitional epithelium and then the valve line (forme fruste). Minor abnormalities show zonal changes, although individual variations are common. Major abnormalities are associated with both underrepresentation and absence of the described features.

In the newborn piglet model, minor, lower anal canal anomalies have all three mucosal linings (rectal, transitional, and squamous) represented, whereas in major anal canal agenesis, the fistula is completely lined by the transitional form, with a notable absence of squamous mucosa. A comment is made that anal glands were demonstrated in all studied animals with or without ARM [61].

Anal Cushions

Anal Cushions (corpus/cavernosum recti) [26] have grown in importance as a factor that determines anal continence. No study has looked at this in the human with an anal anomaly (see also Chaps. 7 and 8).

5.4.1.2 *Mucosal Nerve Supply*

Sensory nerve endings present themselves during fetal life and go through the stages of immaturity through to morphologically mature structures [65]. Abundantly organized nerve endings are encountered at term in the wall of the anal canal. Free nerve endings are found in the epithelium of the canal and perianal skin. The densities of nerve bundles in the subepidermis and dermis of the perineal region associated with ARM are decreased; this correlates inversely with the degree of anorectal agenesis. This is also reflected by the density of free nerve endings in the region of the valve line (crypts and valves), where they are usually most numerous.

5.5 The Internal Anal Sphincter

This is the terminal portion of the inner circular smooth muscle layer of the rectum. The functional importance of this muscle is emphasized, as incontinence after anal surgery is characterized by the virtually universal presence of an internal sphincter injury resulting in a reversal of the normal resting pressure gradient in the anal canal [67,115]. The internal anal sphincter in the adult human has recently been described as being composed of 26 rings, flat ring-like slats, of smooth muscle bundles stacked like the slats of a Venetian blind, one on top of the other, and arranged to form 3 equally sized columns around the anal canal [124]. This is a morphological study and has not yet been confirmed by imaging studies.

At the anorectal junction, the muscularis propria of the rectum changes [102]. The inner circular layer thickens to become the internal sphincter. Imaging studies divide the length of the anal canal into three zones – high, middle, and low. The internal sphincter surrounds the upper two, with two-thirds of its length above the anal valve line. Its lower border is identified by the intersphincteric groove on the skin, thus defining the upper limit of the low canal zone.

5.5.1 Internal anal sphincter thickness

The thickness of the internal anal sphincter has been determined in neonates using infracoccygeal

Table 5.3 Internal anal sphincter thickness as determined by infracoccygeal transperineal ultrasound (ITU) in neonates [46] and by endosonography in healthy children (average age = 12 years) [6]

Subjects	Measurement technique	Internal anal sphincter thickness	
		Thickness (mm)	Range (mm)
Neonates	ITU	1.3 ± 0.3	0.8–1.9
Children (average age 12 years)	Endosonography	2.5 ± 0.66	2–4

transperineal ultrasound, and by endosonography in healthy children (average age 12 years; Table 5.3) [6,46]. Using this technique it is seen as an echo-poor structure, an extension of the muscular wall of the rectum, which is homogeneous in appearance, varying only slightly in thickness. Normal values for sphincter dimensions differ between the imaging techniques employed. Therefore, muscle thicknesses obtained are not interchangeable and true values of thickness in normal individuals have not yet been determined [5]. The purpose of measurement, however, is to distinguish the normal from the abnormal muscle thickness with reproducible reliability.

Thicknesses can be determined by different methods, such as endosonography, endoanal MR imaging, or phased-array MR imaging. The effect of an endoanal coil – causing sphincter muscle stretch – is reflected by the difference in internal anal sphincteric thickness obtained between the two above MR techniques. Also of note, there is an inherent inaccuracy of endoanal ultrasound in defining and measuring the external sphincteric muscle.

Evaluating the internal sphincter in the adult using data obtained manometrically, by ultrasound and MR techniques reveal:

1. No gender differences.
2. No difference in length (as a percentage of the overall anal canal sphincter length).
3. The proximal sphincter is nearly equal in thickness – anteriorly/posteriorly.
4. Measured in the middle/high canal, the thickness tends to increase with increasing age (using endoanal MR, the thickness is reported to increase significantly with age).
5. Homogeneous except in the elderly, where the appearance altered (not uniformly noted).
6. In the adult, using endosonography, the internal sphincter thickness has been assessed at the mid-canal level (Table 5.4) [5].

Table 5.4 Internal anal sphincter thickness as determined at mid-anal canal level, endosonographically in the adult [5]. These figures are thicker than those obtained with phased-array MR imaging and significantly thicker than that determined by endoanal MR

	Internal anal sphincter thickness (mm)
Females	3.8 ± 1.2
Males	3.4 ± 1.4

5.5.2 Nerve Supply

The internal anal sphincter is innervated by the intrinsic enteric nervous system: autonomic (sympathetic, parasympathetic, and nitrinergic). It receives this supply uninterrupted from the distal rectum, which is innervated in a craniocaudal (isoperistaltic) direction [85]. The enteric nervous system is ganglionated in the rectum, where it is arranged in the form of named plexuses [mucosal (Henle); submucosal (Meissner); intermyenteric (Auerbach)], which contain ganglia and interconnecting nerve fibers. In the wall of the anal canal its nature alters [1], with the upper canal (high) hypoganglionated, the mid canal aganglionic with nerve fibers present, and the low canal aganglionic with nerve fibers prominent. The ganglia, ganglion cells, and microglia, contain immature cells, even at term [11]. This, in the preterm must be combined with the fact that the plexuses develop within the wall of the bowel in an "out to in" fashion. The intermyenteric plexus is always the most prominent. The rectoanal inhibitory reflex tests the integrity of this supply.

5.5.3 The Internal Anal Sphincter in ARM

Depending inversely on the degree of dysgenesis present, it has been well recognized for many years that a functional internal anal sphincter is present where the anal canal is partially formed [7,94]. Its presence is associated with a normal rectoanal inhibitory response [100]. With rectal anomalies it has been established that a translevator fistula (terminal channel), when present in instances of anal agenesis/dysgenesis, represents and is the terminal portion of the hindgut, with its opening an ectopically sited anus [64,106]. These channels have the mural features of the upper anal canal, identified classically by the presence of a terminally thickened portion of circular smooth muscle – a forme fruste of an internal anal sphincter [54]. This emulates the features shown to be present in the pig model with anal agenesis:

1. A terminal hindgut end surrounded by an internal sphincter.
2. The enteric nervous system of the pouch in the region of the fistulous connection, as well as the channel itself, show features of intestinal hypo- or aganglionosis with neuromatosis.
3. The proximal fistula is lined by transitional epithelium (see also Chap. 7).

Endoanal imaging in humans managed surgically using fistula-preserving techniques substantiate the idea that a mural sphincter with features determined by the severity of the anomaly present can be detected and functionally assessed [24]. Its appearance, however, is often structurally abnormal and it may be scarred by the surgery done.

5.6 The Longitudinal Muscle of the Anal Canal

This is a structure that can be imaged clearly in the high anal canal [120]. It is the continuation of the longitudinal outer smooth muscle of the rectum, which is situated between the internal and external anal sphincters. Terminal fibers of this muscle insert into the submucosa of the canal, musculus canalis ani; others traverse the superficial external anal sphincter to reach the skin, the musculus corrugator cutis ani. It connects and tethers the visceral and somatic parts of the anal sphincteric complex together. It is seen on MR imaging within the fat tissue of the intersphincteric space, and is best evaluated by a phased-array MR technique where the sphincter complex is unstretched.

5.6.1 Dimensions of the Longitudinal Muscle

Length measurements (adult studies) indicate that in the female the longitudinal muscle ends cranial to the superficial external sphincter, while in the male it extends to the caudal end of the sphincter. In both males and females its thickness decreases with age (Table 5.5) [5]. Histologically, Pacinian corpuscles, pacemaker cells of Cajal, and ganglia can be identified within its muscular tissue [41,45].

Table 5.5 The longitudinal muscle of the anal canal: thickness measurements in adults [5]

Imaging technique	Longitudinal muscle thickness (mm)	
	Female	Male
Endoanal ultrasound	2.9 ± 1.0	2.3 ± 1.0
Endoanal MR	0.8 ± 0.2	0.8 ± 0.2
Phased-array MR	1.0 ± 0.2	0.9 ± 0.2

5.7 External Anal Sphincter

5.7.1 Localization and Nature

There are perianal sphincter fibers of striated muscle that stretch from the lower border of the puborectalis cranially to the distal caudal termination of its fibers. No constant plains of cleavage within it are seen but it has a changing pattern at different levels conforming to a trilaminar arrangement [2]. The sphincter has three open U-loop like sections, which give it an ellipsoidal shape morphologically [8]. It is situated between the fat-filled ischiorectal fossae laterally, it attaches anteriorly to the perineal muscles, and posteriorly to the anococcygeal raphe and coccyx [55]. It seems not to form complete circles in certain planes in neither the male nor the female [35]. Sexual differences of the ventral part of this sphincter have been noted even in the fetus [79].

The classically described tripartite subdivision of this sphincter (see Chaps. 6 and 7] can not be confirmed using imaging techniques, but it can be divided into sections that are continuous with one another, subdivisions of a single muscular structure [102]. From caudal to cranial, these are:

1. The superficial external sphincter (subcutaneous). This surrounds the lower anal canal and is traversed by the coat tails of the longitudinal anal canal muscle (in the male). Histologically, it can be seen as two parallel muscle strips in the axial plane; confirmed operatively as the so-called parasagittal fibers and by MR imaging.
2. The deep external sphincter, surrounds the middle anal canal anteriorly in both sexes. It is continuous through the high canal with the puborectalis fibers of the levator ani cranially; with which seen in the sagittal plane, it gives a posteriorly positioned teardrop-like appearance to the anal sphincters perpendicular to the axis of the canal.
3. The puborectalis fibers of the levator ani. Contraction of this section of the levator muscle forms the anorectal ring. During contraction it cannot be separated from the external anal sphincter caudally, but from the rest of the levator-formed pelvic diaphragm cranially. Its lower border defines the upper extent of the high anal canal, which is deficient in external sphincteric muscles anteriorly in the female – the major anatomical difference between the male and female anal sphincter (see Chaps. 6 and 7). The puborectalis is continuous with, but not described as part of the anatomic external anal sphincter; it has a separate motor nerve supply.

Normal variants of the external sphincter may have a closed circular configuration or may have an open configuration anteriorly and posteriorly.

5.8 The Perineum and Perineal Body

The urogenital hiatus is bridged by a diaphragm, the fibrous perineal membrane. This is triangular in shape as it spans the inferior ischiopubic rami from the pubis to the ischial tuberosities. Posteriorly, the superficial and deep transverse perinei run along its free edge [102]. The genitalia are attached to its inferior surface. The perineal body is a point of fusion at the posterior free edge of the urogenital diaphragm. It is a site of insertion of:

1. Muscle: the superficial and deep perinei, bulbocavernosus, levator ani fibers, rectourethralus, external anal and striated urethral sphincters, (the perineum acts as an insertion site for the striated muscles that anchor the anal sphincter to the surrounding pelvis), the striated urethral sphinter.
2. Fascia: the perineal membrane, Denonvilliers, Colle's, and the outer pelvic stratum.

The deep transverse muscle is the primary muscle of the diaphragm, with the puborectalis muscle superior to it. It is penetrated by the urethra and the vagina.

The format of the perineum differs between the sexes [102]. The superficial transverse perineal muscle is directly superior, and overlaps the external sphincter in females, whereas it is directly anterior to the external sphincter in males. In addition, the female perineum is imaged as a pronounced intermingling of muscle fibers with the formation of a definable perineal body. In the male, in contrast, it seen as a point of aggregation of fibers, directly superior to the root of the penis; inferior to the prostate gland.

It is proposed that anatomically, the external anal sphincter, urethral sphincter, and bulbocavernosus muscle originate from the puborectalis muscle and, furthermore, that the bulbocavernosus muscle is an integral part of the external anal sphincter. For this reason this muscle complex in its entirety has been named the “anogenital muscle”.

5.8.1 Gender Differences – External Anal Sphincter

5.8.1.1 The Female Sphincter

This is significantly shorter anteriorly, but there is no statistically significant difference between sexes in the length of the canal plus puborectalis muscle, for the same age (see Chaps. 6 and 7) [102]. There is a trend for the thickness of the external anal sphincter (plus puborectalis) to decrease with increasing age (not a significant finding). Also, the thickness of the external anal sphincter in young adult women is less than in young adult men (age-related atrophy has been observed and is thought to be associated with late-onset fecal incontinenced (Table 5.6). The major variation in anatomy lies in the anterior part of the anal sphincter [102], where in the male the central perineal body acts as a central insertion point, while in females there is not a point, rather an area of woven muscular fibers.

5.8.2 Nerve Supply of the External Anal Sphincter

The external anal sphincter is under voluntary control and is innervated by the pudendal nerves (S2, 3, and 4), which supply the inferior rectal nerve, which in turn traverses the lower half of the ischiorectal fossa from out of the pudendal canal, and terminates in the external anal sphincter at the 3 and 9 o'clock positions [111]. The puborectalis has a separate nerve supply on its superior surface (S3 and 4).

Table 5.6 The thickness of the external anal sphincter as determined in the neonate and child [6], and in the adult male and female [5]

Subject	Technique	Thickness (range)
Neonate child	Endoanal ultrasound	1.6 mm (1.2–2.3 mm)
	Endoanal ultrasound	(6.2 ± 1.64 mm)
Adult male	Endonal ultrasound	(6.1 ± 1.7 mm)
	Endoanal MR	(1.3 ± 0.4 mm)
	Phased-array MR	(1.4 ± 0.5 mm)
Adult female	Endonal ultrasound	(7.2 ± 2.3 mm)
	Endoanal MR	(1.2 ± 0.3 mm)
	Phased-array MR	(1.3 ± 0.2 mm)

5.8.2.1 Overlapping Innervation (Monkey)

Unilateral pudendal neurectomy causes histological changes consistent with partial denervation on both sides of the midline, and is most marked ipsilaterally [132]. This indicates overlap in supply or the interdigitation of muscle fascicles across the midline.

5.8.2.2 Resting Tone

An inherent permanent spontaneous activity characterizes the resting state of the external sphincter. Combined positron emission and CT have confirmed this an extremely sensitive reflex state [116]. Resting tone is controlled by sensory (stretch) nerve endings, muscle spindles, within the sphincter. Pressure receptors (Pacinian corpuscles) are found between the external anal/internal anal sphincters and in the presacral space. This is part of an abundant supply of sensory organized nerve endings (Meissners corpuscles, Golgi Mazzoni bodies, Krause bulbes, and globular nerve endings) encountered within the wall of the anal canal, with numerous free nerve endings [28,65].

5.8.3 Spinal Control of the Pelvic Floor Muscles

There is a prevalent notion in the literature that the pelvic floor muscles behave as a unitary mass. Uncontrolled stimulation of the pelvic floor substantiates this position. Personal experience could, however, question it. Experiments carried out in the spinal cat model show [27]:

1. Electrical stimulation of different sacral ventral nerve roots elicited twitch responses from different muscles.
2. Dependent upon the intensity of the stimulation, various muscles of the pelvic floor could be reflexly reactivated either individually or as a mass unit.
3. The external anal sphincter responded to tactile or electrical stimulation of the pudendal regions elicited on either side of the body; this is in contrast to other muscles that could be lateralized.
4. Unilateral section of one pudendal nerve did not alter the level of tonic activity of the sphincter, and bilateral section abolished it, without affecting the activity of the levator muscles.

Based upon this evidence, it was concluded that the neural apparatus supplying the pelvic floor is capable of activating the different muscles that make up its structure individually, and that the external anal sphincter and the levator ani are subserved by different neuronal circuits. The extrapolation of these findings to the human could require modification, but support the concept that a segregation of the segmented neuronal pools innervating the different pelvic floor muscles exists.

5.8.4 The Voluntary Anorectal Inhibition Reflex

This is a postulated viscerosomatic reflex: external anal sphincter contraction preventing emptying causes reflex rectal wall relaxation [108]. This illustrates the close functional relationship between pelvic visceral and somatic structures that may be traced back to Onuf's nucleus, which innervates pelvic floor muscles and occupies an intermediate position between visceral and somatic nuclei.

5.8.5 Neurophysiological Dysfunction

There is a well-documented association between abnormal neurology and constipation in adults [57]. The innervation of the external sphincter has been shown to be abnormal in ARM, concentrating on ventral spinal cord motor neuron damage [134]. Nerve arc studies determining latencies, spinoanal responses (pudendoanal reflex; dorsal nerve penis→cord→to external sphincter) were recorded. In ARM, conduction latencies and evoked potentials were abnormal and grossly prolonged, documenting the presence of serious motor neuron lesions involving the sacral spinal centers [133]. Of great interest, loss of one sacral vertebra (S5) significantly affected the assessment results.

5.8.6 Endosonography of the Anal Sphincters

Endosonography is the tool of choice for assembling the component parts of the sphincter in a practical and available fashion. The individual and gender-related variability is emphasized, as shown by a study carried out in adult volunteers [121]. There was no plane of cleavage between components of the external sphincter. A trilaminar arrangement conforming to the changing patterns encountered in the external sphincter at different levels was noted, with a deep external sphincter annular in 72–76% of cases. The superficial external sphincter was elliptical in 76% of females and 86% of males. The subcutaneous component of superficial external sphincter was conical in 56–57% of cases. The external sphincter was shorter anteriorly in females. Abnormal insertions from the external sphincter were encountered anteriorly in 14% of cases. The longitudinal muscle layer was identified in all men, and in 60% of women it was indistinguishable from the external sphincter sonographically. Subepithelial tissues and the internal sphincter were easily identified in all subjects. Manometrically in healthy adults there is no relationship between resting and squeeze pressures and internal or external sphincter thickness [40].

5.8.7 Individual Versus Common Sphincter Control of Pelvic Organ Continence

The external anal and urethral sphincters are examples of individual sphincters. The puborectalis in contradistinction gives origin to these individual sphincters, but at the same time embraces them as their common sphincter. This arrangement allows for common, individual or double sphincter control [108].

5.9 The Pelvic Floor and External Anal Sphincter in ARM

The work of Peña has had a major influence on our understanding of the anatomy in these congenital anomalies (see also Chaps. 6 and 7). We have read, digested, and found the structures he has described. Imaging techniques have supported his interpretations and recent human studies have consolidated rather than refuted his concepts of the presence of "potential" striated muscle tissue – designated, but not used for the formation of an external anal sphincter [88, 119]. A spectrum of deformities as has been stressed by Peña [104]. The normal anal canal and its opening, and the rectum with their surrounding anatomical structures are either fully represented with only very minor modification, or else nonrepresentation or absent development is encountered. The visceral and parietal anatomical defects usually parallel one another in severity or degree. This facilitates their clinical detection and predicts the functional prognosis on completion of current surgical treatment.

The size of the anal orifice in the neonate has been expressed as a function of the birth weight (BW) [29]: 1.3 + (BW in kg × 3), expressed in millimeters (Hegar size). The minimal functional (adequate) size at term is estimated to be at least 12 mm (12 Hegar).

5.9.1 Position of the Anus

The ectopic position of the opening is of importance when it is obvious and symptoms attributed to it are present. The orifice in these instances is usually stenotic. Minor variants of dystopia appear to be common. The perineum increases in length as the child matures, an important variable that makes accurate determination of the position of the orifice on clinical grounds imperfect. The proposition that in females the anus lies on the midpoint between the fourchette and the tip of the coccyx led to the conclusion that anterior ectopia was a common cause of constipation, and gave rise to an unusual incidence of this anomaly [63]. Ectopia was related in a causal way with chronic constipation, as it was necessarily associated with a prominent levator (puborectal) shelf. Surgical treatment was directed at this feature. Building on this theme, gender-based ratios were quoted: the ratio of midanus to fourchette length to midanus to coccyx length being < 0.34 in females (< 0.46 in males).
In practical terms, anterior displacements can be excluded where the anus lies less than 2/3 of the distance from coccyx to fourchette [98]. The issue of posterior displacements remains controversial. Imaging techniques may clarify these situations in the future by positioning the canal centrally within or partially eccentrically outside the sphincter complex.

5.9.2 The Anal (Canal Opening) Target Site

The normal anus has a cutaneous areola, a surrounding halo of pigmentation and puckering due to fibrous sphinteric attachments. Where this opening is absent, these changes or forme fruste thereof can be present [117, 118]. The low anal canal, below the intersphincteric groove, develops from the proctodeum (anal pit), and partially from this structure as it stretches up to the valve line. It had been suggested, (now refuted) that these cutaneous developments were induced by the approaching terminal hindgut as it nears the skin. In ARM, the skin over the expected anal site may appear unaltered. This suggests to the clinician that the anomaly could be of a major nature, although this rule is not absolute. A spectrum of anal areolar maldevelopment can be encountered; in the presence of an absent perineal anus, changes from near normality to total absence are seen; its epicenter may be depressed (anal fossette) or frequently everted as if it had not been sucked up to meet the approaching hindgut.

In the male, the perineum is divided by a midline (sagittal) raphe. In anal dysgenesis this may appear hypertrophied; inappropriately cross over the target site and /or have the appearance of a bucket handle. It can contain an epidermal tract that connects the lumen of the malformed anal canal to the raphe. Prenatally this is unfilled, although epithelial detritus accumulates within it and can give it an appearance of a string of pearls. Postnatally, meconium finds its way into it, turning it into a black track that may rupture anywhere along the length of the raphe.

In the female the normally featureless perineum can have a sagittally placed midline groove – the perineal groove – lined by unkeratinized epithelium.

5.9.3 The Perineum

Where the anus is obviously absent, so too the perineal body is not formed. Where the perineal muscles form part of the striated muscle sphincters of the anal canal or urethra, they are only represented where these structures traverse the levator ani and perineal diaphragm.

5.9.4 The Common Wall Phenomenon

Most obvious where the abnormal hindgut opening is found within the vestibule or posterior urethra, the anterior wall of its accompanying channel above the opening is unified cranially for a variable distance (2–4 cm) with the muscular wall of the urethra or vagina. The length of this union is determined by the length of the intralevator segment of the hindgut channel, which is longest with rectobulbar connections and absent with a T-shaped termination where the rectum opens above the bladder base or higher. In cloacal variants, the vagina may in a similar fashion be united with the urethra along its anterior wall. The implications of this anatomy are of great importance during the surgical correction of these anomalies.

5.9.5 Striated Muscle Tissue, "Potential" Sphincteric Fibers in ARM

This tissue is found in the infralevator space, the levator muscle, perineal diaphragm, and the perineal body/male phallus [88, 119].

5.9.5.1 The Levator Component – The Pubovisceralis

Dependent upon the anomaly present the following may be observed:

1. A Puborectalis; in anal dysgenesis where the hindgut has descended through the levator floor (noncommunicating: communicating anomalies in males and females).
2. A Pubourethralis; a fistula opens into the urethra above its membranous section, the prostatic urethra, or the bladder neck.
3. A Pubovaginalis in females. A urethral and genital opening is present within the external genitalia.
4. A Pubourogenitalis in females. The cloacal variants.

There is a spectrum of severity, with an open pelvic osseous ring at one extreme.

Here, diastasis of the symphysis pubis is present. The pubovisceral section of the levator muscle is absent. The pelvic floor lies high and is flat in shape (imaging has shown that this diaphragm has a domed shape when normal). The pelvic viscera, where an ARM is present, lie outside the pelvic ring. There is foreshortening in the sagittal length of the true pelvic cavity. With a closed pelvic osseous ring, the pelvic viscera can abut the symphysis pubis. The regional effects of caudal regression are apparent and may include hypoplasia of the gluteae (flat-bottomed appearance; absent, shortened, or shallow natal cleft) with associated absent sacrum or underdevelopment and sagittally foreshortened external genitalia (this can facilitate phallic prominence). In addition, the cavity of the vulva may appear to be open or partially closed. The "potential" striated sphincteric muscle tissues of the levator floor are severely underdeveloped, absent and/or compromised.

5.9.5.2 The Striated Muscle Complex (Peña)

This terminology is used to name the striated muscle tissue destined for, but never used to form the external anal sphincter. The tissue is found between the pubovisceralis cranially, and the predestined anal target site at skin level caudially. Peña describes it according to the main direction of its component fibers, with deep, vertical fibers in a column of muscle that stretches between the pubovisceralis and the skin where it interdigitates with superficial horizontal fibers, the parasagittal fibers, at right angles [88]. The original description of these fibers was based upon the morphological appearance of the structures encountered during posterior sagittal anorectoplasty. Subsequent imaging studies have confirmed this anatomical configuration.

The Deep Component

This is a strictly midline column that is held laterally between the fat-filled fascial pockets of the ischiorectal fossae (Gonzales hernia). It is a poorly represented structure in major malformations. It has a defined anterior and posterior boundary.

The Superficial Component

This has parasagittal fibers that run horizontal to the vertical column and that interdigitate posteriorly (raphe) and anteriorly (perineal body) in the midline, and superiorly with the vertical fibers that tend to fan out into it. It is an easily seen subcutaneous structure that can be better represented in major malformations.

5.10 The Rectum

The rectum imperceptibly begins with the disappearance of the sigmoid mesentery opposite the third sacral vertebra, a site where the taenia coli fuse to form a continuous longitudinal layer as its outer coat. At the anorectal ring it is angulated posteriorly as it descends as part of the anal canal, its mural structures continuing in modified form to beneath the pectinate line. It is covered anteriorly at its upper two-thirds by parietal peritoneum and is separated in the male from the bladder by the rectovesical pouch and in the female from the uterus by the rectouterine pouch of Douglas. The lower third is "bare" and lies within the retroperitoneal space where its anterior wall and adventitial sheath, beneath the seminal vesicles or posterior fornix of the vagina, is in contact with Denonvilliers' or rectovaginal fascia. It terminates at its most inferior part as an ampulla, which is attached to the posterior urethral sphincter and perineal body. It is innervated by the laterally placed pelvic plexus, and its blood supply reaches it via the superior (from the inferior mesenteric), middle (from the internal iliac), and inferior (from the internal pudendal) rectal arteries [87, 112]

5.10.1 Rectal Ectasia

Dilatation of an unexpected magnitude of the most distal hindgut is seen in association with ARM (see Chap. 12) [19]. In its most extreme form it is encountered in pouch colon cases (see Chap. 11) [14]. This is suspected when on a plain abdominal X-ray study the erect picture shows the distended colon to be greater than 50% of the diameter of the abdomen. Detailed histopathology of the reservoir's wall [15] has not been extensively researched. It would appear that the wall is reported to be normal in most instances, with no enteric nervous system anomaly.

5.10.1.1 Pouch Anatomy

Usually a thin-walled, sac-like structure; taenia coli are absent or poorly developed with no appendices epiploicae [13]. Rarely it is observed as thick walled and has an associated wide genitourinary fistula. There is a characteristically abrupt transition between the sac and normal-appearing proximal intestine. In summary, this anomaly of the colon has four major characteristics: a reservoir-like pouch, a foreshortened colon, the presence of a genitourinary fistula (which is rarely absent or represented by a fibrous cord), and an ARM.

5.10.1.2 Ectasia

Significantly less dramatic and more localized dilatation of the terminal intestine immediately proximal to the fistula in ARM or to the blind-ending termination are reported [136]. These bulbous terminations not involving the fistula are labeled rectal ectasias. They are reported to be congenital or, when acquired due to anatomical defects in the wall of the bowel, deemed to be part of the ARM. In description they are minor variants of the pouch anomalies, although outlet obstruction and the effect of urinary retention in the segment play etiological roles. The significant nature of the dilatation draws attention to their presence.

The normal rectal pouch (termination) with secondary dilatation due to outlet ARM may be attributable to poor or incomplete pouch emptying (see Chap. 12).

5.11 Fetal Defecation

Dilatation of the rectosigmoid in newborn infants with ARM supports the hypothesis that fetal colonic peristalsis and defecation is a normal physiological process [56]. What is contentious is whether colonic emptying occurs during the third trimester. The determination of fetal intestinal disaccharidases in the amniotic fluid suggests that they are released by defecation [92]. Their disappearance after the 22nd week suggests that rectal emptying is absent during the third trimester.

This evidence must be correlated with morphological data regarding the anatomical development of the anal sphincteric mechanism [10]. Digestive enzyme assays indicate that anal membrane perforation occurs at about 12 weeks of gestation; their absence in amniotic fluid subsequent to 22 weeks of gestation apparently cannot be explained by development of anal continence, as morphological evidence shows that the functional components of the sphincteric mechanism are not adequate by the 30th week of gestation. The use of ultrasound to observe anal activity revealed that the greatest frequency occurs between weeks 28 and 34 of gestation; however, defining fetal defecation as the expulsion of contents through the anus showed that emptying of rectal content occurs until term. The discrepancy between these studies remains speculative.

Hypoxic defecation in the mature fetus is unchallenged when meconium staining of liquor occurs. If, as it is suggested, defecation in utero is a physiological fetal function, what happens to the evacuated material? Filtration mechanisms clearing unwanted content have been demonstrated in animal experiments, which are rendered inoperative during hypoxic stress [17,18]. These findings point to the presence of a similar physiological cycle in the human. It is obvious that in the fetus with an ARM and fistula, emptying of the rectum is possible and determined by the size and site of this channel/tract. Meconium reaches the colon by the 18th week of gestation and builds up thereafter in the colon. How much of it is evacuated before term remains a matter of conjecture [97].

5.12 The Pelvic Connective Tissue

See also Chaps. 6 and 7. In the mind of the surgeon, the fibrous tissue within the pelvis is thought of as fibrous sheets; membranous fasciae that line the pelvic wall covering muscle and bone or fibrous ligaments (aggregations), which hold or tether the pelvic viscera to prevent visceral prolapse, especially in the female [122]. Little mention is made of the loose areola tissue (retroperitoneal fascia) that surrounds the pelvic viscera and encloses the vessels and nerves that supply these structures. Intraoperative mobilization of the pelvic viscera is possible as this tissue can be divided by blunt and sharp dissection, developing surgical plains of dissection freeing up the structure to be removed [47]. During this dissection, vessels and nerves are divided [36,51].

What is termed fascia in the pelvis does not imply that the structure is merely collagenous; pelvic fascia is rich in elastic tissue and contains smooth muscle. This indicates that it plays a major role in the support and possible function of the pelvic viscera. This connective tissue has been arbitrarily divided into outer, intermediate and inner strata.

5.12.1 The Outer Stratum, the Parietal Layer (Leaf)

This lines the inner surface of the pelvic wall and is continuous with the transversalis layer of the abdomen. Where it covers the sacrum it is known as Waldeyer's fascia, and it is through this that the surgeon must enter before a retrorectal dissection plane can be developed.

5.12.2 The Intermediate Stratum

This is the tissue in which the pelvic viscera are embedded. Its loose areolar nature is compressible, allowing it to accommodate their emptying and filling. Because of this potential, four spaces can be developed in it: retropubic, paravesical, rectogenital, and retrorectal. This stratum coalesces around vessels and nerves and thickens to form well- and ill-defined named ligaments that suspend the viscera.

5.12.3 The Inner Stratum: The Visceral Layer (Leaf)

This layer clothes the viscera and is well defined where these structures are covered by peritoneum; it flows imperceptibly into the intermediate stratum when it is not.

Other named areas of importance are the aventitia surrounding the rectum, the rectogenital septum (Denonvilliers' fascia), the rectovaginal fascia, and the perianal connective tissue.

5.12.4 The Aventitia Surrounding the Rectum

This extends down from the sigmoid colon and continues to the pelvic floor. It is not well seen in the cadaver. It is of extreme importance because within it are hidden the autonomic nerve supply to the rectum and bladder. The sympathetic supply originates from the lateral column of grey matter in the thoracolumbar spinal cord (T10–L2), and forms the superior hypogastric plexus, which contains sympathetic fibers from the celiac plexus and first four lumbar splanchnic nerves. It divides into two hypogastric nerves to enter the pelvis beneath the outer stratum medial to the internal iliac vessels and anterior to the sacrum. Paravertebral sympathetic trunks descend into the pelvis deep to major vessels and medial to the sacral foramina to fuse in front of the coccyx as the ganglion impar. The parasympathetic nerves originate in the intermediolateral cell column of the sacral cord (S2–4), and reach the pelvic plexus as the nervi erigenti.

5.12.5 The Pelvic Plexus

The pelvic plexus, or inferior hypogastric plexus, is a flat plexus of nerves and ganglia that is oriented in a semisagittal plane anterolateral to the surface of the rectum, with its midpoint sited at the seminal vesicles (in adults ± 4–5 cm long) [3,32,50]. It is tranversed by numerous vessels going to and from the rectum, bladder, and internal genitalia. The right and left plexus communicate behind the rectum and anterior and posterior to the bladder neck in the male. Branches of the plexus follow blood vessels to reach the viscera they supply.

The nervi erigenti leave their sacral nerve roots of origin well lateral to the midline behind Waldeyer's fascia, through which they penetrate, then travel within the intermediate stratum of fascia to join the pelvic plexus.

5.12.6 The Neurovascular Bundle of Walsh

Since the advent of nerve-sparing radical prostatectomies, this structure has increased in importance as it contains the innervation of the prostate and corpora cavernosa [126,127]. Inferior to the seminal vesicles, these nerves lie within the leaves of the intermediate stratum near its junction with but outside Denonvillers' fascia. They travel at the posterolateral border of the prostate on the surface of the rectum, lateral to the prostatic capsular arteries and veins. The nerves are composed of multiple fibers and are not visible on gross inspection. Blood vessels serve as surgical landmarks for their course. At the apex of the prostate they approach the capsule of the gland at the 5 and 7 o'clock positions. On reaching the membranous urethra, its superficial branches travel on the surface of the striated urethral sphincter at 3–9 o'clock positions. At the base of the penis, they join to form 1–3 discrete bundles related to the urethra at 1–11 o'clock positions. They enter the corpora cavernosa with the arteries to supply the erectile tissue. In the female these nerves travel between the anterior vaginal wall and bladder in association with lateral venous plexuses.

5.12.7 The Rectogenital Septum (Denonvilliers' Fascia)

This is a thin layer of connective tissue between rectum and the bladder, seminal vesicle, and prostate [25,66]. It is thought to represent an obliterated part of the rectovesical pouch, but has no macroscopically discernible layers once it has been separated from the adventitia (fascia propria) of the rectum. The pelvic plexus of nerves lie posterior to it [16].

5.12.8 The Rectovaginal Fascia

This is situated between the rectum and vagina and ends superficially in the perineal body supporting this structure [69]. It serves to subdivide the subperitoneal space into dorsal rectal and ventral urogenital compartments. It connects the rectum firmly to the vagina and serves as a good guiding structure during nerve-sparing surgery as nerves, part of the pelvic plexus, lie within the dorsal rectal compartment.

5.12.9 The Perianal Connective Tissue

The coattail intramural termination of the longitudinal layer of the rectal muscular wall has been noted. This is a fibroelastic network (web) that continues through the surrounding perineal fat to the pelvic wall, connecting the lower levator fascia to the perianal skin, firmly anchoring the anus [44]. The development and organization of pelvic connective tissue has been studied in the human fetus [32,34]. The early general arrangement of this tissue in fetal life is the same in the male and female. No ligaments, save for the pubovesical and puboprostatic ligaments, can be identified; while easily identifiable compartments exist in the young fetus, the later development of adipose tissue within them clouds this appearance, as is encountered in the neonate [33]. This situation is carried through into adult life, where interperson, age, and ethnic differences have a major influence on the morphology of this tissue.

5.13 Intestinal Neuronal Dysplasia

Also see Chap. 25

5.13.1 General

If we accept that the terminal part of the fistula not only represents, but is also structurally similar to the proximal anal canal, then it follows that its intrinsic nerve supply and neuronal plexuses will conform anatomically with that present in the normal anal canal [99]. The nearer the opening of the fistula is to the normal anatomical position of the anus, the greater the similarity would be expected to be. The appearance of the internal anal canal mucosal surface in many instances does indeed follow this rule. The columns of Morgagni, and an improperly formed anal valve line, the pectinate line, are often discernible even in instances where the fistula opening is in the upper reaches of the urethra in the male. This expectation is confirmed in the more minor anal malformations.

5.13.2 Specific

The enteric nervous system has been looked at and aberrations in its anatomical appearance reported [74,76]. As our knowledge and ability to demonstrate this supply has increased, so too have these structural appearances become defined and found to have causative roles in the pathophysiology encountered in cases with anorectal dysfunctional states [75]. Quantitative and qualitative alterations in this system have been demonstrated to involve the fistula in ARM [52,53,71,77].

Prior to our current state of knowledge concerning the fistula and the enteric nervous system, aganglionosis with features of congenital intestinal aganglionosis were detected and reported [52,53,86.]. These findings were classified as abnormal and related to visceral malfunction. This analysis, however, was never accepted universally by pediatric surgeons, so that rectal biopsy sampling has never been used as a routine screening technique that would alter the primary management of this group of malformations.

5.13.3 Normal Anatomy

At birth, only one-third of the nerve cells within the ganglia of the submucosal plexus are mature [11]. Features consistent with those encountered with intestinal neuronal dysplasia, such as giant ganglia and neuromatosis, may be found, although where the definition of this aberration is strictly applied and related to the patient's age, it is accepted that these findings in most instances are normal [20,129,130]. The nearer to the anal opening from where the tissue is sampled, the more likely are these changes to be encountered.

5.13.4 Abnormal Anatomy

Hypoganglionosis has been detected and reported in patients with ARM [21]. Hyperganglionosis with associated submucosal and mucosal nerve plexus proliferation consistent with a definition of an intestinal neuronal dysplasia has also been identified [59]. The significance of this altered anatomy involving the enteric nervous system is difficult to assess [57,72,103]. What role it may play, how it affects anorectal function in ARM, can only be postulated. Whether it is a true malformation of the nervous system, an alteration acquired during fetal life, or the consequence of an obstructed gut is a further unclarified feature because intestinal neuronal dysplasia has been reported to occur in such instances [38,52,53,58,78,101].

If the appropriate animal model can be used to aid us in evaluating the effects and significance of these changes, then the altered microanatomy is abnormal and must play a role in the pathophysiology present in ARM [93]. This is even more important today where the fistulous termination of the intestine is used surgically to establish an anal canal [31,95,114].

5.14 Caudal Regression

This was introduced by Bernard Duhamel as a concept that features a continuum of anorectal, urogenital, and skeletal congenital anomalies, from sirenomelia to ARM at its two extremes [22,70]. It characterizes what is a spectrum of mesodermal dysplasia axial in position, which has been extensively studied in several different animal models, the most important of which are the mouse, rat, and pig [89]. Information such as the absence of expression of Sonic Hedgehog and bone morphogenic protein 4 in murine embryos with ARM has been obtained, advancing our knowledge regarding causative factors in mammals [105]. So too has the role played by the notochord been exposed, indicating a common etiology for ARM and vertebral anomalies in these models [96]. Clinicians have used this association in the assessment of the degree of regression present in the human, by looking at the osseous skeleton radiographically. This has prognostic implications. Soft-tissue MR imaging has shown up this link to even greater degree, revealing associated anomalies of neurilation not known about and not assessed or taken into consideration during case management [49,83].

Maldevelopment of the dorsal cloaca has shown similarities and differences in these models to that in the human [84]. In human and pig embryos, the urorectal septum formed by the process of embryonic caudal folding and the incorporation of surrounding extraembryonic mesoderm does not grow in the direction of the cloacal membrane, and consequently has never been observed to fuse with it [125]. On rupture of this structure as the result of apoptotic cell death, the cloaca becomes part of the amniotic cavity, thus exposing both the anorectal and urogenital tracts. In ARM the maldevelopment focuses on the cloacal plate, which is a vertically oriented midline plate of epithelial cells in the caudal half of the genital tubercle [90]. The anterior portion of this plate persists as the urethral plate and participates in the formation of the urethra in the male and the vestibule in the female. Events limiting the movement of the dorsal part of the plate and adjacent hindgut onto the body surface and the normal development of the surrounding structures are operative in ARM and have been demonstrated in the pig [23]. In contrast to previous hypotheses, this rearrangement of dorsal cloacal structures takes place as the consequence of differential tissue growth, apoptosis, and changes in the curvature of the developing spine. Cloacal partition, as previously described by lateral fusion of its side walls, appears not to play any role; the emphasis now is placed upon remodeling of structures [123]

5.15 Spinal Dysraphism

Axial, caudal dysgenesis is always present where cloacalplate-related clinical malformations are present. What was not realized in the past is their incidence in minor anomalies [42,48]. The apparent central role of the notochord in the closure of the caudal neuropore, as part of the process of neurilation of the neural plate, has been emphasized.

5.15.1 Defects in the Development of the Axial Vertebral Structures

These defects will be discussed in terms of the sacrum, the vertebral canal, and the spinal cord [68,81].

5.15.1.1 The Sacrum

This is clinically the most obvious, with a wide spectrum in severity of the osseous maldevelopment encountered.

Sacral Hypoplasia

Its degree relates pari passu to the severity of the visceral maldevelopment. A sacral index determined radiologically measures this and is used to predict functional outcome. The pathology represents tissue underdevelopment, an underrepresentation not an absence.

Segmental Sacral Dysgenesis and Agenesis

The structural complement of the sacrum may be intact but disorganized or scrambled; an absence or maldevelopment is encountered within this context (dysraphism). Complete absence of a part or the whole of a vertebral segment represents a degree of sacral agenesis. The most caudal are the most affected and are the most commonly absent. Hemisacral agenesis is encountered.

Sacral Agenesis

This has significant functional implications if more than two of the five sacral vertebrae are absent.

5.15.1.2 The Vertebral Canal

Defective development may be reflected by an abnormally shaped canal (J-shaped sacrum – Currarino syndrome) or incomplete axial closure (spina bifida occulta or spina bifida cystica). This may occur anteriorly or posteriorly. The complete vertebral canal may be stenotic. Conventional radiology has focused attention on the associated osseous anomalies in hindgut malformations. Modern imaging techniques have uncovered the regional nature of this maldevelopment.

5.15.1.3 The Spinal Cord, Cauda Equina and their Coverings and Attachments

Abnormality in cord shape (caudal regression syndrome), cord fixation (tethered cord), cord morphology (syrinx, diastematomyelia), and conal level occur. Cauda equina involvement concerns absent nerve roots, while cord-covering abnormalities are associated with spina bifida.

Within this group, clinical attention has been focused on the tethered cord in association with ARM (see Chap. 18). Abnormalities of the filum terminale and cord pulsation are documented by MR imaging and sonographically when possible. The interpretation of these studies where minor variants of the normal anatomy are present is difficult and their clinical relevance uncertain [80].

It is clear that neurological deficit can be congenital or acquired. Where it is congenital, evaluation of the motor neuron innervation to the anal sphincter mechanism in the rat model is reported to indicate the presence of a quantitative deficiency in this supply. Whether this deficiency is inadequate in relation to the mass of muscular sphincter fibers present, whether the sphincter is present but partially denervated, is not certain. The implications are that this scenario could be possible. If we can extrapolate urodynamic findings in patients with ARM not previously operated upon to the rectal continence-controlling mechanisms, then intrinsic defective motor neuron innervation to the potential external sphincteric muscle fibers could be part of the primary anomaly. This could contribute to the poor postoperative anorectal function seen in surgically corrected cases.

A further factor to consider is abnormal cord anatomy that restricts or causes neuronal dysfunction, which could occur during fetal life. This would compound any intrinsic innervation defect present. Acquired neurology would be based upon impaired cord development due to growth restriction.

It can be concluded that spinal dysraphism occurs in the absence of any osseous vertebral anomaly, in both anal and rectal malformations in both sexes, and that if abnormal anatomy is detected, its functional role in the physiological derangement of anorectal function has yet to be quantified accurately in these cases.

There may be early and late defects, with agenesis of the spinal cord being a disturbance of primary neurilation (early embryonic period). Tethered cord may be a disturbance of degeneration or differentiation (late embryonic period). The conus medullaris [131] normally ascends between the 30th and 40th week postmenstrual age to reach the L1/L2 interspace by the 40th week. This suggests that the conus does not ascend during normal childhood.

Neuroradiological evaluation of patients with ARM has shown [64]: (1) abnormalities detected in proportion to the severity of the ARM (all cases of cloacal exstrophy and caudal regression, 60% of cases with anal agenesis, and 20% of cases with anal dysgenesis), (2) the detected presence of sacral dysraphism/hypoplasia indicates the predicted presence of abnormal cord imaging in 60% of cases, and (3) there exists a spectrum of neuroanatomic anomalies (see Chap. 18).

References

1. Aldridge RT, Campbell PE (1968) Ganglion cell distribution in the normal rectum and anal canal. A basis for the diagnosis of Hirschsprung's disease by anorectal biopsy. J Pediatr Surg 4:475–490
2. Ayoub SF (1979) Anatomy of the external anal sphincter in man. Acta Anat (Basel) 105:25–36
3. Baader B, Herrmann M (2003) Topography of the pelvic autonomic nervous system and its potential impact on surgical intervention in the pelvis. Clin Anat 16:119–130
4. Barber MD, Bremer RE, Thor KB, Dolber PC, Kuehl TJ, Coates KW (2002) Innervation of the female levator ani muscles. Am J Obstet Gynecol 187:64–71
5. Beets-Tan RGH, Morren GL, Beets GL, Kessels AGH, el Naggar K, Lemaire E, Baeten CGMI, von Engelshoven JMA (2001) Measurement of anal sphincter muscles: endoanal US, endoanal MR imaging, or phased-array MR imaging? A study with health volunteers. Radiology 220:81–89
6. Benninga, MA, Wijers OB, Van Der Hoeven CW, Taminiau JA, Klopper PJ, Tytgat GN, Akkermans LM (1994) Manometry, profilometry and endosonography: normal physiology and anatomy of the anal canal in healthy children. J Pediatr Gastroenterol Nutr 18:68–77
7. Bill AH, Johnson RJ (1958) Failure of migration of the rectal opening as a cause for most cases of imperforate anus. Surg Gynecol Obstet 106:643–648
8. Bogduk N (1996) Issues in anatomy: the external anal sphincter revisited. Aust NZ J Surg 66:626–629
9. Bollard RC, Gardiner A, Lindow S, Phillips K, Duthie GS (2002) Normal female anal sphincter: difficulties in interpretation explained. Dis Colon Rectum 45:171–175
10. Bourdelat D, Muller F, Droulle P, Barbet JP (2001) Anatomical and sonographical studies on the development of continence and sphincter development in human fetuses. Eur J Pediatr Surg 11:124–130

11. Bughaighis AG, Emery JL (1971) Functional obstruction of the intestine due to neurological immaturity. Prog Pediatr Surg 3:37–52
12. Bustami FM (1988–89) A reappraisal of the anatomy of the levator ani muscle in man. Acta Morphol Neerl Scand 26:255–268
13. Chadha R (2004) Congenital pouch colon associated with anorectal agenesis. Pediatr Surg Int 20:393–401
14. Chadha R, Bagga D, Malhotra CJ, Mohta A, Dhar A, Kumar A (1994) The embryology and management of congenital pouch colon associated with anorectal agenesis. J Pediatr Surg 29:439–446
15. Chadha R, Gupta S, Tanwar US, Mahajan JK (2001) Congenital pouch colon associated with segmental dilatation of the colon. J Pediatr Surg 36:1593–1959
16. Church JM, Raudkivi PJ, Hill GL (1987) The surgical anatomy of the rectum – a review with particular relevance to the hazards of rectal mobilisation. Int J Colorectal Dis 2:158–166
17. Ciftci AO, Tanyel FC (1998) In utero defecation: a new concept. Turk J Pediatr 40:45–53
18. Ciftci AO, Tanyel FC, Bingol-Kologlu M, Sahin S, Buyukpamukcu N (1999) Fetal distress does not affect in utero defecation but does impair the clearing of amniotic fluid. J Pediatr Surg 34:246–250
19. Cloutier R, Archambault H, D'Amours C, Levasseur L, Ouellet D (1987) Focal ectasia of the terminal bowel accompanying low anal deformities. J Pediatr Surg 22:758–760
20. Coerdt W, Michel JS, Rippin G, Kletzki S, Gerein V, Muntefering H, Arnemann J (2004) Quantitative morphometric analysis of the submucous plexus in age-related control groups. Virchows Arch 444:239–246
21. Coerdt W, Muntefering H, Rastorguev E, Gerein V (2004) Congenital disorders of the colonic innervation. A diagnostic guide. Pathologe 25:292–298
22. Cohen AR (1991) The mermaid malformation: cloacal exstrophy and occult spinal dysraphism. Neurosurgery 28:834–843
23. Davidoff AM, Thompson CV, Grimm JM, Shorter NA, Filston (1991) Occult spinal dysraphism in patients with anal agenesis. J Pediatr Surg 26:1001–1005
24. De Souza NM, Ward HC, Williams AD, Battin M, Harris DN (1999) Transanal MR imaging after repair of anorectal anomalies in children: appearances in pullthrough versus posterior sagittal reconstructions. Am J Roentgenol 173:723–728
25. Denonvilliers C (1836) Anatomie du perinée. Bull Soc Anat de Paris 2:105
26. Dubrovsky B, Filipini D (1990) Neurobiological aspects of the pelvic floor muscles involved in defecation. Neurosci Biobehav Rev 14:157–168
27. Dubrovsky B, Martinez-Gomez M, Pacheco P (1985) Spinal control of pelvic floor muscles. Exp Neurol 88:277–287
28. Duthie HL, Gairns FW (1960) Sensory nerve endings and sensation in the anal region of man. Br J Surg 47:585–595
29. El-Haddad M, Corkery JJ (1985) The anus in the newborn. Pediatrics 76:927–928
30. Endo M, Hayashi A, Ishihara M,Maie M, Nagasaki A, Nishi T (1999) Analysis of 1,992 patients with anorectal malformations over the past two decades in Japan. J Pediatr Surg 34:435–441
31. Favre A, Briano S, Mazzola C, Brizzolara A, Torre M, Cilli M, Martucciello G (1999) Anorectal malformations associated with enteric dysganglionosis in Danforth's short tail (Sd) mice. J Pediatr Surg 34:1818–1821
32. Fritsch H (1989) Topography of the pelvic autonomic nerves in human fetuses between 21–29 weeks of gestation. Anat Embryol (Berl) 180:57–64
33. Fritsch H (1993) Development and organization of the pelvic connective tissue in the human fetus. Anat Anz 175:531–539
34. Fritsch H (1994) Topography and subdivision of the pelvic connective tissue in human fetuses and in the adult. Surg Radiol Anat 16:259–265
35. Fritsch H, Brenner E, Lienemann A, Ludwikowski B (2002) Anal sphincter complex: reinterpreted morphology and its clinical relevance. Dis Colon Rectum 45:188–194
36. Fritsch H, Lienemann A, Brenner E, Ludwikowski B (2004). Clinical anatomy of the pelvic floor. Adv Anat Embryol Cell Biol 175:1–64
37. Frudinger A, Halligan S, Bartram CI, Price AB, Kamm MA (2002) Female anal sphincter: age-related differences in asymptomatic volunteers with high-frequency endoanal US. Radiology 224:417–423
38. Galvez Y, Skaba R, Vajtrova R, Frantlova A, Herget J (2004) Evidence of secondary neuronal intestinal dysplasia in a rat model of chronic intestinal obstruction. J Invest Surg 17:31–39
39. Gans SL, Friedman N (1961). Some new concepts in the embryology, anatomy and surgical correction of imperforate anus. West J Surg Obstet Gynecol 69:34–37
40. Gantke B, Schafer A, Enck P, Lubke HJ (1993) Sonographic manometric and myographic evaluation of the sphincters morphology and function. Dis Colon Rectum 36:1037–1041
41. Gerdes B, Kohler HH, Stinner B, Barth PJ, Celik I, Rothmund M (1997) The longitudinal muscle of the anal canal. Chirurg 68:1281–1285
42. Golonka NR, Haga LJ, Keating RP, Eichelberger MR, Gilbert JC, Hartman GE, Powell DM, Vezina G, Newman KD (2002) Routine MRI evaluation of low imperforate anus reveals unexpected high incidence of tethered spinal cord. J Pediatr Surg 37:966–969

43. Gosling JA, Dixon JS, Critchley HO, Thompson SA (1981) A comparative study of the human external sphincter and periurethral levator ani muscles. Br J Urol 53:35–41
44. Haas PA, Fox TA (1977) The importance of the perianal connective tissue in the surgical anatomy and function of the anus. Dis Colon Rectum 20:303–313
45. Hagger R, Gharaie S, Finlayson C, Kumar D (1998) Distribution of the interstitial cells of Cajal in the human anorectum. J Auton Nerv Syst 73:75–79
46. Han TI, Kim IO, Kim WS, Chung JY, Choeh K, Song MK, Yoon YG (2000) US identification of the anal sphincter complex and levator ani muscle in neonates: infracoccygeal approach. Radiology 217:392–394
47. Havenga K, De Ruiter MC, Enker WE, Welvaart K (1996) Anatomical basis of autonomic nerve preserving total mesorectal excision for rectal cancer. Br J Surg 83:348–388
48. Heij HA, Nievelstein RA, de Zwart I, Verbeeten BW, Valk J, Vos A (1996) Abnormal anatomy of the lumbrosacral region imaged by magnetic resonance in children with anorectal malformations. Arch Dis Child 74:441–444
49. Hirano H, Tomura N, Wataral J, Kato T (1998) Caudal regression syndrome: MR appearance. Comput Med Imaging Graph 22:73–76
50. Hoer J, Roegels A, Prescher A, Klosterhalfen B, Tons C (2000) Preserving autonomic nerves in rectal surgery. Results of surgical preparation on human cadavers with fixed pelvic sections. Chirurg 71:1222–1229
51. Hollabaugh RS, Steiner MS, Sellers KD, Samm BJ, Dmochowski R (2000) Neuroanatomy of the pelvis: implications for colonic and rectal resection. Dis Colon Rectum 43:1390–1397
52. Holschneider AM, Koebke J, Meier-Ruge W, Land N, Jesch NK, Pfrommer W (2001) Pathophysiology of chronic constipation in anorectal malformations. Eur J Pediatr Surg 11:305–310
53. Holschneider AM, Pfrommer W, Gerresheim B (1994) Results in the treatment of anorectal malformations with special regard to the histology of the rectal pouch. Eur J Pediatr Surg 4:303–309
54. Husberg B, Rosenborg M, Frenckner B (1997) Magnetic resonance imaging of anal sphincter after reconstruction of high and intermediate anorectal anomalies with post sagittal anorectoplasty and fistula preserving technique. J Pediatr Surg 32:1436–1442
55. Hussain SM, Stoker J, Lameris JS (1995) Anal sphincter complex: endoanal MR imaging of normal anatomy. Radiology 197:671–677
56. Kimble RM, Trudenger B, Cass D (1999) Fetal defecation: is it a normal physiological process? J Paediatr Child Health 35:116–119
57. Kobayashi A, Yokota H, Kobayashi H, Yamataka A, Miyano T, Haya Y (2004) Mucosal neuroendocrine cell abnormalities in patients with chronic constipation. Asian J Surg 27:197–201
58. Kobayashi H, Yamataka A, Lane GJ, Miyano T (2004) Inflammatory changes secondary to postoperative complications of Hirschsprung's disease as a cause of histopathologic changes typical of intestinal neuronal dysplasia. J Pediatr Surg 39:152–156
59. Koletzko S, Jesch I, Faus-Kebetaler T, Briner J, Meier-Ruge WA, Muntefering H, Coerdt W, Wessel L, Keller KM, Nutzenadel W, Schmittenbaecher P, Holschneider AM, Sache P (1999) Rectal biopsy for diagnosis of intestinal neuronal dysplasia in children: a prospective multicentre study on interobserver variation and clinical outcome. Gut 44:853–861
60. Lambrecht W, Kluth D, Lierse W (1989) Epithelium and anal glands in rectal pouches and fistula. Histologic studies of swine with congenital anal atresia. Z Kinderchir 44:41–46
61. Lambrecht W, Lierse W (1987) The internal sphincter in anorectal malformations: morphologic investigations in neonatal pigs. J Pediatr Surg 22:1160–1168
62. Lane RH, Parks AG (1977) Function of the anal sphincter following colo-anal anastomosis. Br J Surg 64:596–599
63. Leape LL, Ramenosfsky ML (1978) Anterior ectopic anus: a common cause of constipation in children. J Pediatr Surg 13:627–630
64. Levitt MA, Patel M, Rodriguez G, Gaylin DS, Peña A (1997) The tethered spinal cord in patients with anorectal malformations. J Pediatr Surg 32:462–468
65. Li L, Li Z, Hou HS, Wang HZ, Wang LY (1990) Sensory nerve endings in puborectalis and anal region: normal findings in the newborn and changes in anorectal anomalies. J Pediatr Surg 25:658–664
66. Lindsey I, Guy RJ, Warren BF, Mortensen NJ (2000) Anatomy of Denonvilliers' fascia and pelvic nerves, impotence and implications for the colorectal surgeon. Br J Surg 87:1288–1299
67. Lindsey I, Jones OM, Smilgin-Humphreys MM, Cunningham C, Mortenson NJ (2004) Patterns of fecal incontinence after anal surgery. Dis Colon Rectum 47:1643–1649
68. Long FR, Hunter JV, Mahboubi S, Kalmus A, Templeton JM (1996) Tethered cord and associated vertebral anomalies in children and infants with imperforate anus: evaluation with MR imaging and plain radiography. Radiology 200:377–382
69. Ludwikowski B, Oesch Hayward I, Fritsch A (2002) Rectovaginal fascia: an important structure in pelvic visceral surgery. About its development structure and function. J Pediatr Surg 37:634–638.
70. Martins JL, Martins EC (2003) Anorectal anomaly associated with caudal regression: late evaluation after posterior sagittal anorectoplasty. Pediatr Surg Int 19:106–108

71. Martucciello G, Mazzola C, Favre A, Negri F, Bertagnon M, Torre M, Gambini C, Jasonni V (1999) Preoperative enzymo-histochemical diagnosis of dysgangionosis associated with anorectal malformations (ARM) with recto-vestibular and recto-perineal fistula. Eur Pediatr Surg 9:96–100
72. Martucciello G, Torre M, Pini Prato A, Lerone M, Campus R, Jasonni V (2002). Associated anomalies in intestinal neuronal dysplasia. J Pediatr Surg 37:219–223
73. Matzel KE, Schmidt RA, Tanagho EA (1990) Neuroanatomy of the striated muscular and continence mechanism. Dis Colon rectum 33:666–673
74. Meier-Ruge WA, Longo-Bauer CH (1997) Morphometric determination of the methodological criteria for the diagnosis of intestinal neuronal dysplasia (IND B). Pathol Res Pract 193:465–469
75. Meier-Ruge WA, Ammann K, Bruder E, Holschneider AM, Scharli AF, Schmittenbecher PP, Stoss F (2004) Updated results on intestinal neuronal dysplasia (IND B). Eur J Pediatr Surg 14:384–391
76. Meier-Ruge WA, Bronnimann PB, Gambazzi F, Schmid PC, Stoss F (1995) Histopathological criteria for intestinal neuronal dysplasia of the submucosal plexus (type B). Virchows Arch 426:549–556
77. Meier-Ruge WA, Holschneider AM (2000) Histopathologic observations of anorectal abnormalities in anal atresia. Pediatri Surg Int 16:2–7
78. Meier-Ruge WA, Holschneider AM, Scharli AF (2001) New pathogenic aspects of gut dysmotility in aplastic and hypoplastic desmosis of early childhood. Pediatr Surg Int 17:140–143
79. Morren GL, Beets-Tan RG, Von Engelshoven JM (2001) Anatomy of the anal canal and perianal structures as defined in phased-array magnetic resonance imaging. Br J Surg 88:1506–1512
80. Mosiello G, Capitanucci ML, Gatti C, Adorisio O, Lucchetti MC (2003) How to investigate neurovesical dysfunction in children with anorectal malformations. J Urol 170:1610–1613
81. Muthukumar N, Subramaniam B, Gnanaseelan T, Rathinam R, Thiruthavadoss A (2000) Tethered cord syndrome in children with anorectal malformations. J Neurosurg 92:626–630
82. Nagashima M, Iwai N, Yanagihara J, Iwata G (1995) Sensation in the anal region and rectum after surgery for anorectal malformations. Eur Pediatr Surg 5:167–169
83. Nievelstein RA, Valk J, Smit LM, Vermeij-Keers C (1994) MR of the caudal regression syndrome: embryologic implications. AJNR Am J Neuroradiol 15:1021–1029
84. Nievelstein RA, van der Werff JF, Verbeek FJ, Valk J, Vermeij-Keers FJ (1998) Normal and abnormal embryonic development of the anorectum in human embryos. Teratology 57:70–78
85. Okamoto E, Satani M, Kuwata K (1982) Histologic and embryologic studies on the innervation of the viscera in patients with Hirschsprung's disease. Surg Gynecol Obstet 155:823–828
86. Parkulainen KV, Hjelt L, Sulamaa M (1950) Anal atresia combined with aganglionic megacolon. Acta Chir Scand 118:252
87. Patricio J, Bernades A, Nuno D, Falcao F, Silveira L (1988) Surgical anatomy of the arterial blood supply of the human rectum. Surg Radiol Anat 10:71–75
88. Peña A (1988) Potential Anatomic Sphincters of Anorectal Malformations in Females. March of Dimes Birth Defects Foundation. Birth Defects: Original Article Series 24:163–175
89. Penington EC, Hutson JM (2002) The cloacal plate: the missing link in anorectal and urogenital development. BJU Int 89:726–732
90. Penington EC, Hutson JM (2003) The absence of lateral fusion in cloacal partition. J Pediatr Surg 38:1287–1295
91. Peschers UM, De Lancey JO, Fritsch H, Quint LE, Prince MR (1997) Cross-sectional imaging anatomy of the anal sphincters. Obstet Gynecol 90:839–844
92. Potier M, Melancon SB, Dallaire L (1976) Fetal intestinal disaccharidases in human amniotic fluid. Biomedicine 25:167–69
93. Puri P (2003) Intestinal neuronal dysplasia. Semin Pediatr Surg 12:259–264
94. Puri P, Nixon HH (1976) The internal anal sphincter in translevator (low) anal anomalies. J Pediatr Surg 11:553–556
95. Puri P, Shinkai T (2004) Pathogenesis of Hirschsprung's disease and its variants: recent progress. Semin Pediatr Surg 13:18–24
96. Qi BQ, Beasley SW, Arsic D (2004) Abnormalities of the vertebral column and ribs associated with anorectal malformations. Pediatr Surg Int 20:529–533
97. Ramony Cajal CL, Martinez RO (2003) Defecation in utero: a physiologic fetal function. Am J Obstet Gynecol 1888:153–156
98. Reisner SK, Sivan Y, Nitzen M, Merlob P (1984) Determination of anterior displacement of the anus in newborn infants and children. Pediatrics 73:216–217
99. Ricciardi R, Counihan TC, Banner BF, Sweeney WB (1999) What is the normal aganglionic segment of anorectum in adults. Dis Colon Rectum 42:380–382
100. Rintala R, Lindahl H, Sariola H, et al (1990) The rectourogenital connection in anorectal malformations is an ectopic anal canal. J Pediatr Surg 25:665–668
101. Rintala R, Rapola J, Louhimo I (1989) Neuronal intestinal dysplasia. Prog Pediatr Surg 24:186–192
102. Rociu E, Stoker J, Eijkemans MJ, Lameris JS (2000) Normal anal sphincter anatomy and age - and sex-related variations at high-spatial-resolution endoanal MR imaging. Radiology 217:395–401

103. Ryan DP (1995) Neuronal intestinal dysplasia. Semin Pediatr Surg 4:22–25
104. Sachs TM, Applebaum H, Touran T, Taber P, Darakjian A, Colleti P (1990) Use of MRI in evaluation of anorectal anomalies. J Pediatr Surg 25:817–821
105. Sasaki Y, Iwai N, Tsuda T, Kimura O (2004) Sonic hedgehog and bone morphogenetic protein 4 expression in the hindgut region of murine embryos with anorectal malformations. J Pediatr Surg 39:170–173
106. Scott JES (1966) The microscopic anatomy of the terminal intestinal canal in ectopic vulval anus. J Pediatr Surg 1:441
107. Shafik A (1975) New concept of the anatomy of the anal sphincter mechanism and physiology of defecation. Invest Urol 12:412
108. Shafik A (1984) Pelvic double-sphincter control complex. Theory of pelvic organ continence with clinical application. Urology 23:611–618
109. Shafik A (1998) A new concept of the anatomy of the anal sphincter mechanism and the physiology of defecation: mass contraction of the pelvic floor muscles. Int Urogynecol J Pelvic Floor Dysfunct 9:28–32
110. Shafik A (1999) Physioanatomic entirety of external anal sphincter with bulbocavernosus muscle. Arch Androl 42:45–54
111. Shafik A, Doss S (1999) Surgical anatomy of the somatic terminal innervation to the anal and urethral sphincters: role in anal and urethral surgery. J Urol 161:85–89 and J Urol (2001) 164:784
112. Shafik A, Mostafa H (1996) Study of the arterial pattern of the rectum and its clinical application. Acta Anat (Basel) 157:80–86
113. Shaul DB, Harrison EA (1997) Classification of anorectal malformations – initial approach, diagnostic tests and colostomy. Semin Pediatr Surg 6:187–195
114. Shono T, Nagasaki A, Hirose R, Ohgami H, Yakabe S (1997) Acetylcholinesterase (ACE) staining shows the abnormal innvervation of a pulled-through rectum in a case of repaired anorectal malformation. Eur J Pediatr Surg 7:248–251
115. Speakman CT, Burnett SJ, Kamm MA, Bartram CI (1991) Sphincter injury after anal dilatation demonstrated by anal endosonography. Br. J Surg 78:1429–1430
116. Stelzner F, Rodel R, Biersack HJ, Jaeger UE, von Mallek D (2003) Proof of natural spontaneous activity of pelvic and anal musculature by combined position emission therapy and computed tomography. Importance for diagnosis and therapy. Chirurg 74:834–838
117. Stephens FD, Smith ED (1971) Individual deformities in the male. In: Ano-Rectal Malformations in Children. Year Book Medical Publishers, Chicago, pp 33–80
118. Stephens FD, Smith ED (1971) Individual deformities in the female. In: Ano-Rectal Malformations in Children. Year Book Medical Publishers, Chicago, pp 81–117
119. Stephens FD (1988) Potential anatomic sphincters of anorectal malformations in males. March of Dimes Birth Defects Foundation. Birth Defects: Original Article Series 24:155–161
120. Stoker J, Halligan S, Bartram CI (2001) Pelvic floor imaging. Radiology 218:621–641
121. Sultan AH, Kamm MA, Hudson CN, Nicholls JR, Bartram CI (1994) Endosonography of the anal sphincters: normal anatomy and comparison with manometry. Clin Radiol 49:368–374
122. Tamakawa M, Murakami G, Takashima K, Kato T, Hareyama M (2003) Fascial structures and autonomic nerves in the female pelvis: study using macroscopic slices and their corresponding histology. Anat Sci Int 78:228–242
123. Tsakayannis DE, Shamberger RC (1995) Association of imperforate anus with occult spinal dysraphism. J Pediatr Surg 30:1010–1012
124. Uz A, Elhan A, Ersoy M, Tekdemir I (2004) Internal anal sphincter: an anatomic study. Clin Anat 17:17–20
125. Van der Putte SC (1986) Normal and abnormal development of the ano rectum. J Pediatr Surg 21:434–440
126. Walsh PC, Donker PJ (1982) Impotence following radical prostatectomy: insight into etiology and prevention. J Urol 128:492–497
127. Walsh PC, Lepor H, Eggleston JC (1983) Radical prostatectomy with preservation of sexual function: anatomical and pathological considerations. Prostate 4:473–485
128. Watanabe Y, Ando H, Seo T, Kaneko K, Katsuno S, Shinohara T, Toriwaki J (2003). Three-dimensional image reconstruction of an anorectal malformation with mulitidetector-row helical computed tomography technology. Pediatr Surg Int 19:167–171
129. Wester T, O'Briain S, Puri P (1998) Morphometric aspects of the submucous plexus in whole-mount preparations of normal human distal colon. J Pediatr Surg 33:619–622
130. Wilder-Smith CH, Talbot IC, Merki HS, Meier-Ruge WA (2002) Morphometric quantification of normal submucous plexus in distal rectum of adult healthy volunteers. Eur J Gastroenterol Hepatol 14:1339–1342
131. Wolf S, Schneble F, Troger J (1992) The conus medullaris: time of ascendance to normal level. Pediatr Radiol 22:590–592
132. Wunderlich M, Swash (1983) The overlapping innervation of the two sides of the external anal sphincter by the pudendal nerves. J Neurol Sci 59:97–109
133. Yuan Z, Bai Y, Zhang Z, Ji S, Li Z, Wang W (2000) Neural electrophysiological studies on the external anal sphincter in children with anorectal malformations. J Pediatr Surg 35:1052–1057
134. Yuan ZW, Lui VC, Tam PK (2003) Deficient motor innervation of the sphincter mechanism in female rats with anorectal malformation: a quantitative study by fluorogold retrograde tracing. J Pediatr Surg 38:1383–1388

135. Yuan ZW, Wang WL, Pkh T (2003) Quantitative analysis of motorneurons innervating the muscle levator ani in rats with anorectal malformation. Zhonghua Yi Xue Za Zhi 83:1266–1269
136. Zia-ul-Miraj M, Brereton RJ (1997) Rectal ectasia associated with anorectal anomalies. J Pediatr Surg 32:621–623
137. Lierse W, Holschneider AM, Steinfeld J (1993) The relative proportion of type I and type II muscle fibers in the external sphincter ani muscle in different ages and stages of development – observations on the development of continence. Eur J Pediatr Surg 3:28–32

6 Photographic Album of Anorectal Malformations and the Sphincter Muscles

F. Douglas Stephens

The specimens presented in the accompanying compact disc were collected and sectioned at the Royal Children's Hospital, Melbourne, Australia and the Children's Memorial Hospital, Chicago, USA. The colored sections were enlarged directly from the stained histologic sections, which were selected from serial sections of the pelvis of newborn babies who had died of multiple anomalies. The sections show the normal and abnormal anatomy of the viscera and the muscular components of the levator ani and sphincter muscles of the anorectum. Kascot Media Incorporated (Chicago, Illinois, USA) rephotographed the Cibachromes and diagrams and arranged the layout in the original atlas.

The prints have been traced and colored. The schematic diagrams are color-coded: yellow for urinary structures, green for smooth muscle of the bowel, red for the levator complex, brown for external sphincter muscle, purple for genital organs, and blue for cartilage. The red spots on the schematic diagrams serve to orient the sections of each specimen. The blue dots on some diagrams indicate the course of a fistula or bowel lumen.

"P-C" represents the pubococcygeal line, which is drawn on a true lateral radiograph between the center of the boomerang-shaped ossific center of the pubic bone through the junction of the cranial one-quarter and the caudal three-quarters of the ischial ossific center, and a point just distal to the ossific center of the fifth sacral vertebra.

The 1984 classification of ARM is the first illustration (Table 6.1, Fig. 6.1). Figure numbers 6.2–6.28 show female anomalies and 6.29–6.55 show male anomalies. Tables 6.2 and 6.3 list the malformations in females and males with corresponding Figure numbers.

Table 6.1 Index of anomalies

Index of Anomalies	
Anomaly: female	
• Normal female	1F, 2F
• High anomaly	3F
• Intermediate anomalies	4–6F
• Low anomalies	7–10F
Anomaly: male	
• Normal male	1M
• High anomaly	2M
• Intermediate anomalies	3–5M
• Low anomalies	6–10M

Table 6.2 Female malformations with Figure number

Malformation	Figures
Normal female pelvis	6.2–6.3
Normal female pelvis	6.4–6.5
Rectovesical fistula (with phallic urethra)	6.6–6.8
Rectovestibular/low cloaca (absent vagina)	6.9–6.12
Cloaca with short common channel (atrophic vagina)	6.13–6.15
Anorectal atresia	6.16–6.18
Anovestibular fistula	6.19–6.20
Anterior Anus (slightly oblique transverse section)	6.21–6.22
Anocutaneous fistula	6.23–6.25
Perineal groove	6.26–6.28

Table 6.3 Male malformations with Figure number

Malformation	Figures
Normal male pelvis	6.29–6.30
Rectoprostatic urethral fistula	6.31–6.33
Rectobulbar urethral fistula (paremedian)	6.34–6.35
Rectobulbar fistula	6.36–6.38
Rectobulbar fistula (A+B)	6.39–6.40
Anocutaneous fistula	6.41–6.43
Anocutaneous fistula	6.44–6.45
Anocutaneous fistula (bucket handle)	6.46–6.48
Anocutaneous fistula	6.49–6.51
Imperforate anal membrane	6.52–6.55

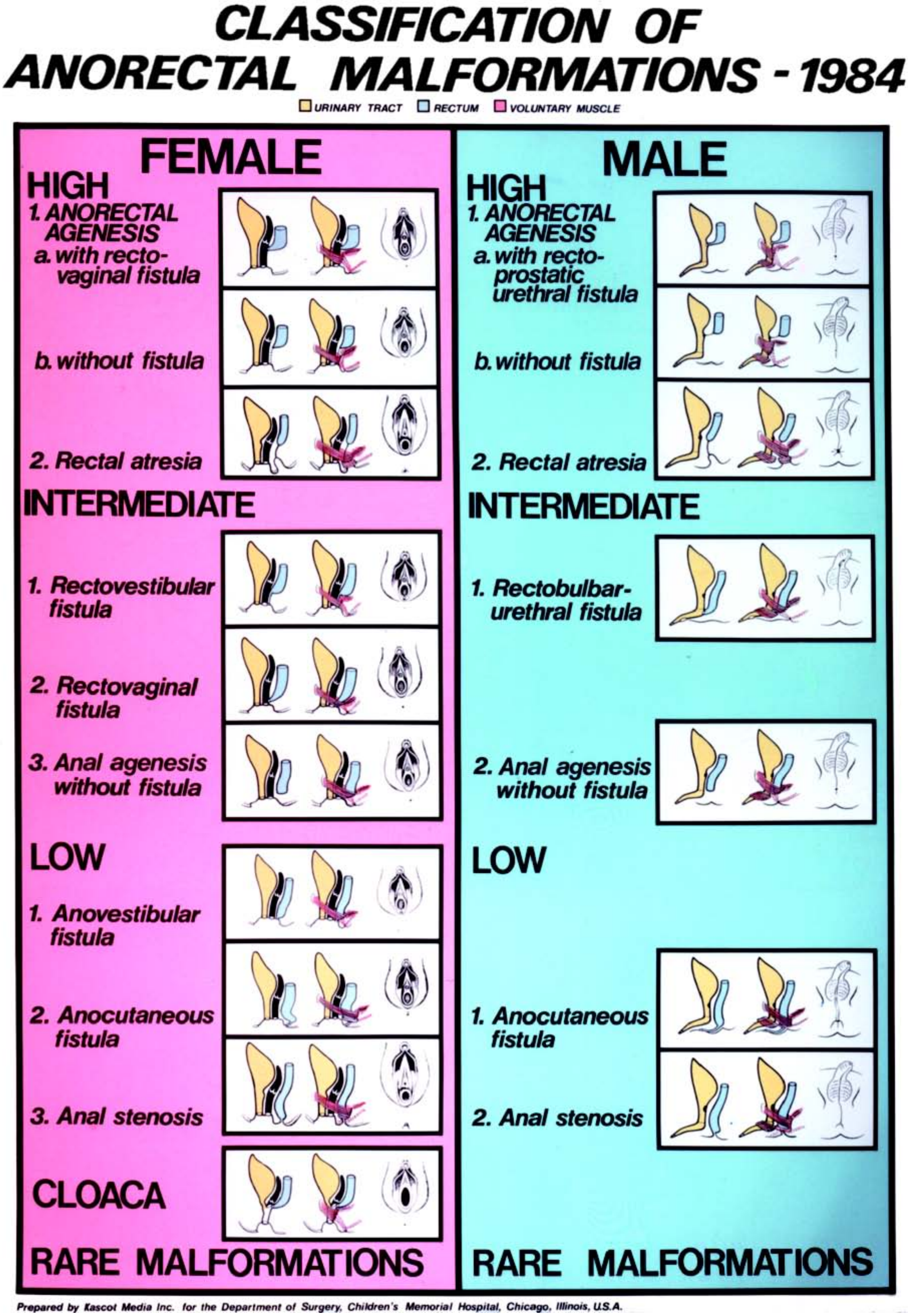

CLASSIFICATION OF
ANORECTAL MALFORMATIONS - 1984
URINARY TRACT
RECTUM
VOLUNTARY MUSCLE
FEMALE
HIGH
1. ANORECTAL AGENESIS
a. with recto-vaginal fistula
b. without fistula
2. Rectal atresia
INTERMEDIATE
1. Rectovestibular fistula
2. Rectovaginal fistula
3. Anal agenesis without fistula
LOW
1. Anovestibular fistula
2. Anocutaneous fistula
3. Anal stenosis
CLOACA
RARE MALFORMATIONS
MALE
HIGH
1. ANORECTAL AGENESIS
a. with recto-prostatic urethral fistula
b. without fistula
2. Rectal atresia
INTERMEDIATE
1. Rectobulbar-urethral fistula
2. Anal agenesis without fistula
LOW
1. Anocutaneous fistula
2. Anal stenosis
RARE MALFORMATIONS
Prepared by Kascot Media Inc. for the Department of Surgery, Children's Memorial Hospital, Chicago, Illinois, U.S.A.

Fig. 6.1

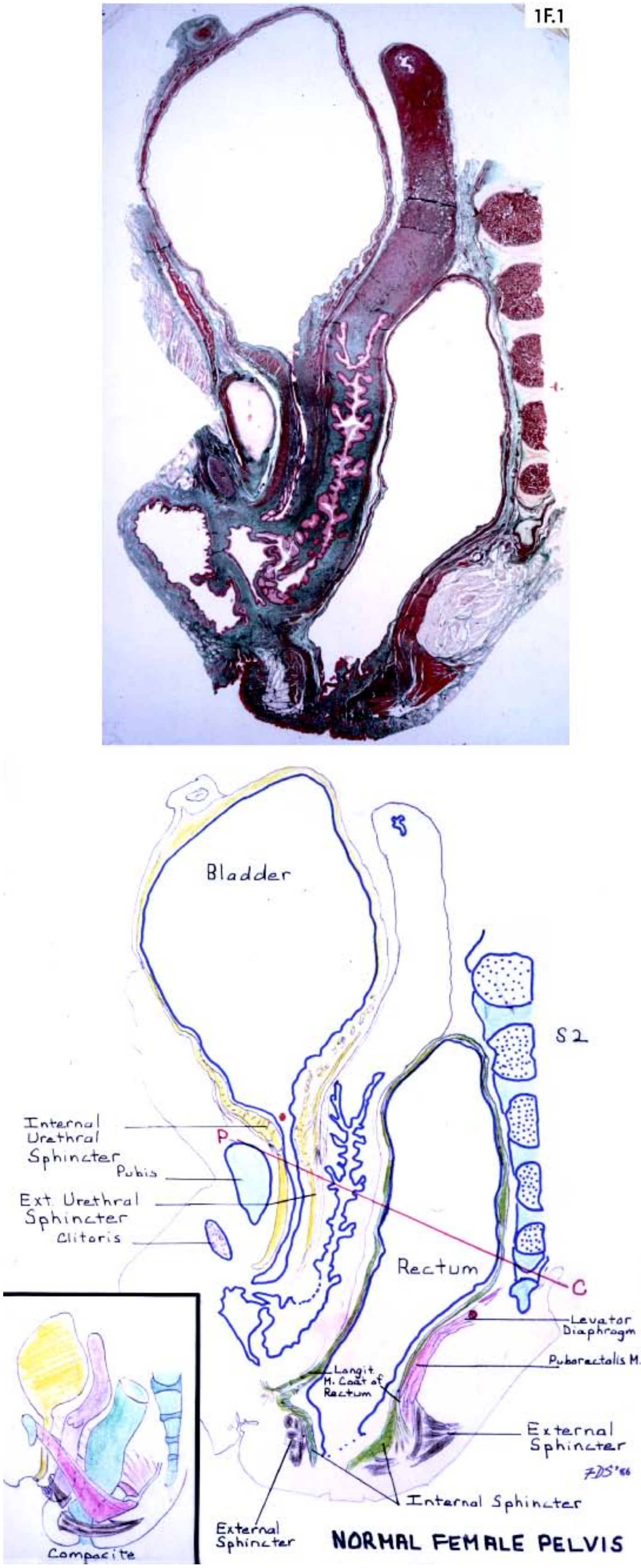


Fig. 6.2 Normal female pelvis (1F.1)

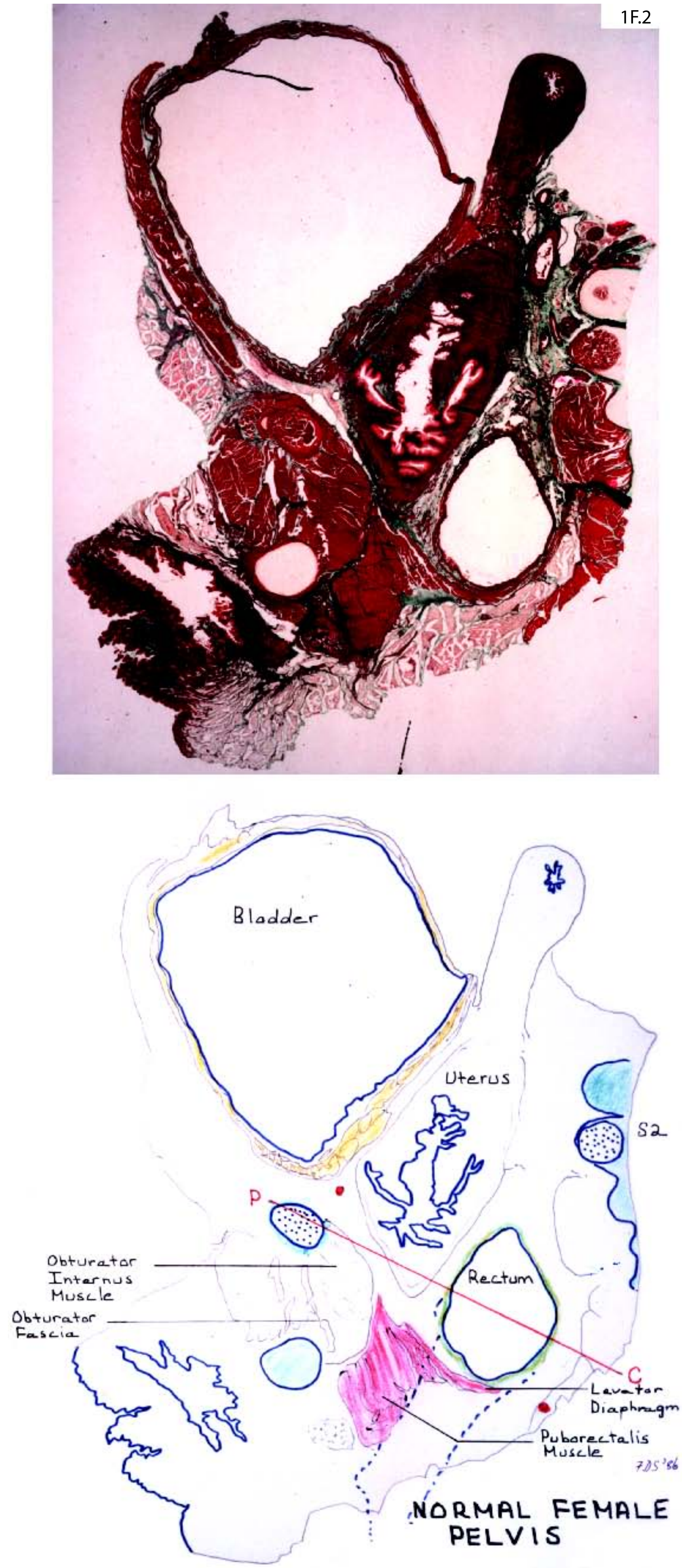
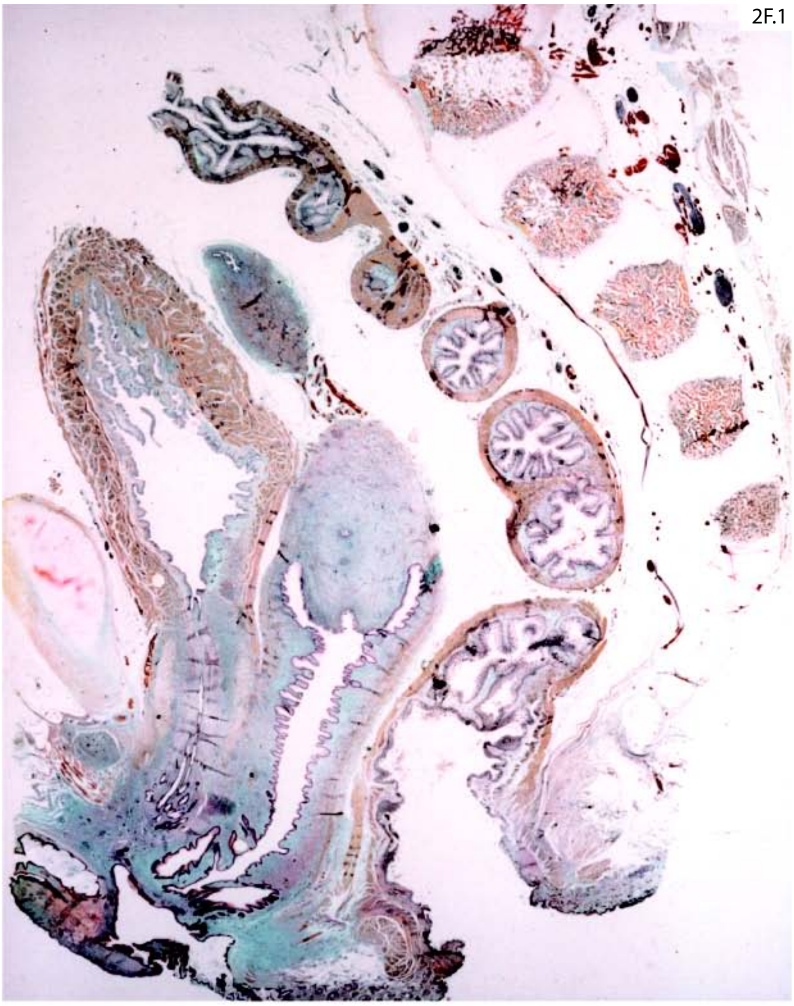


Fig. 6.3 Normal female pelvis (1F.2)



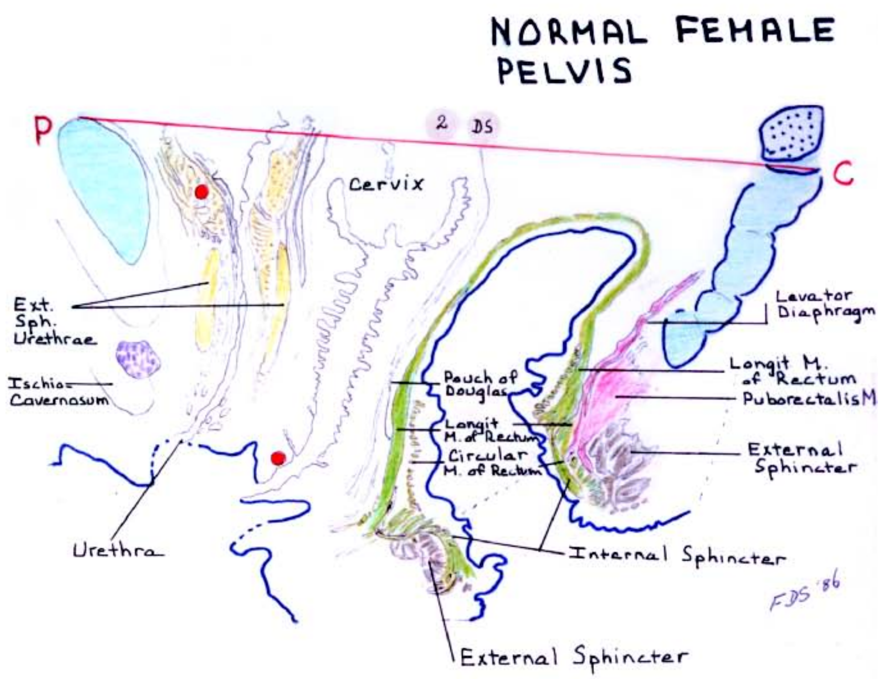


Fig. 6.4 Normal female pelvis (2F.1)

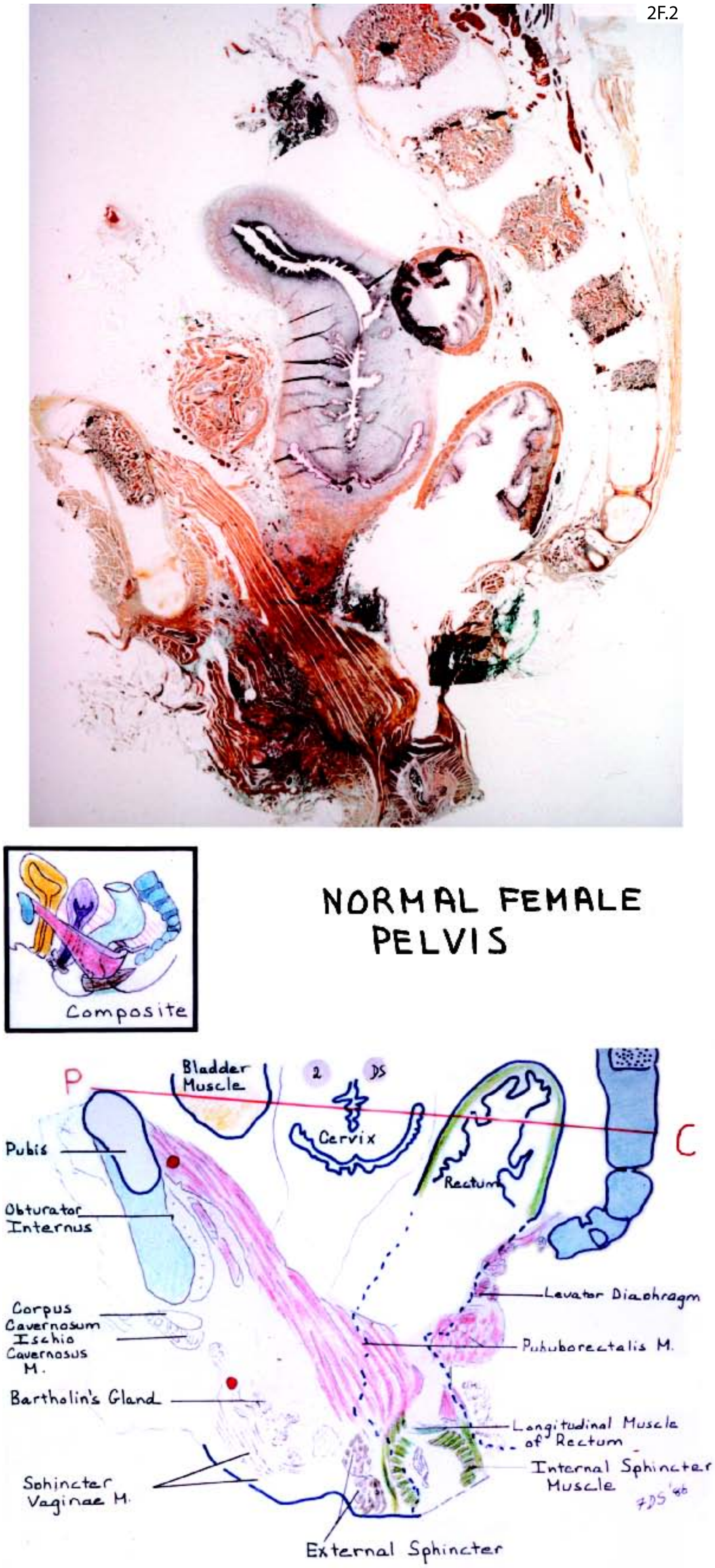


Fig. 6.5 Normal female pelvis (2F.2)

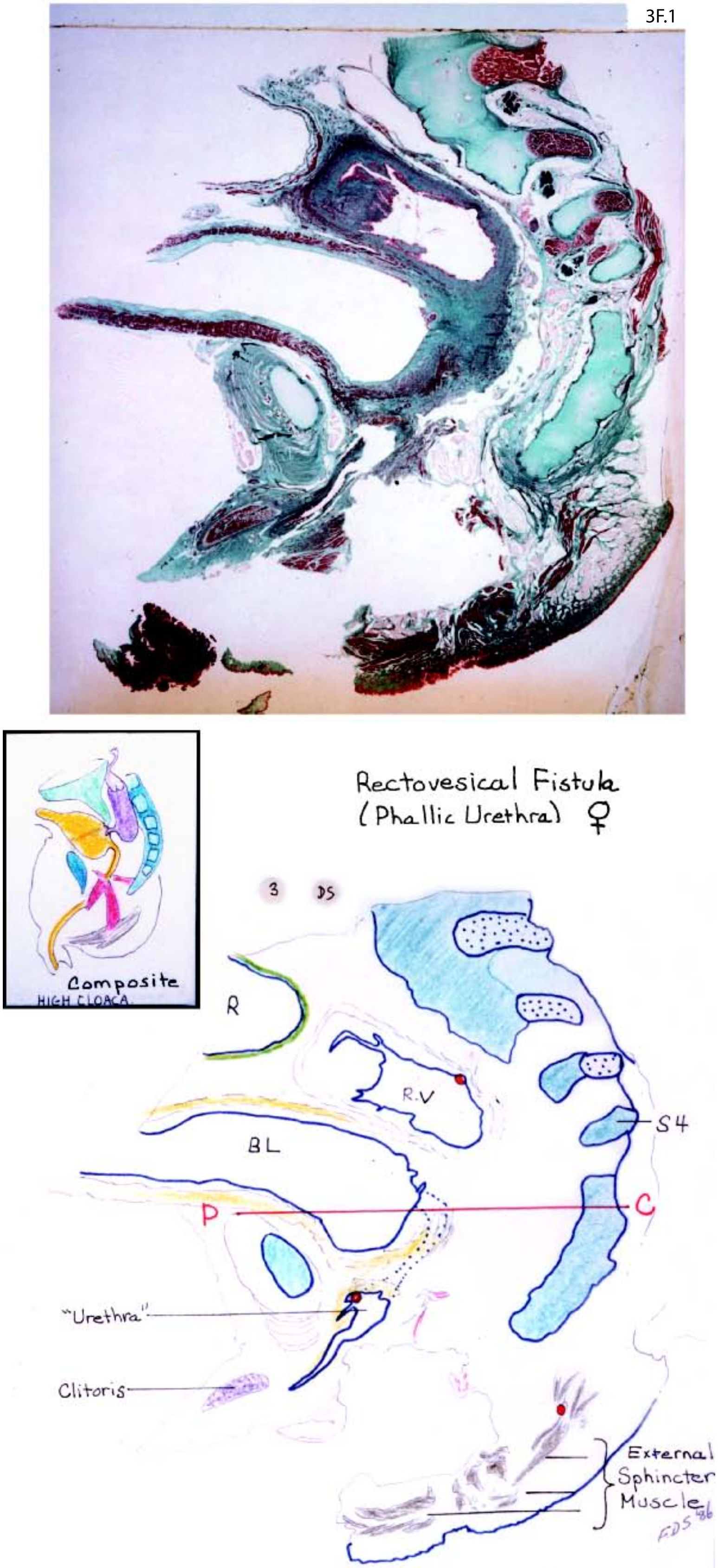


Fig. 6.6 Rectovesical fistula (phallic urethra), female (3F.1)

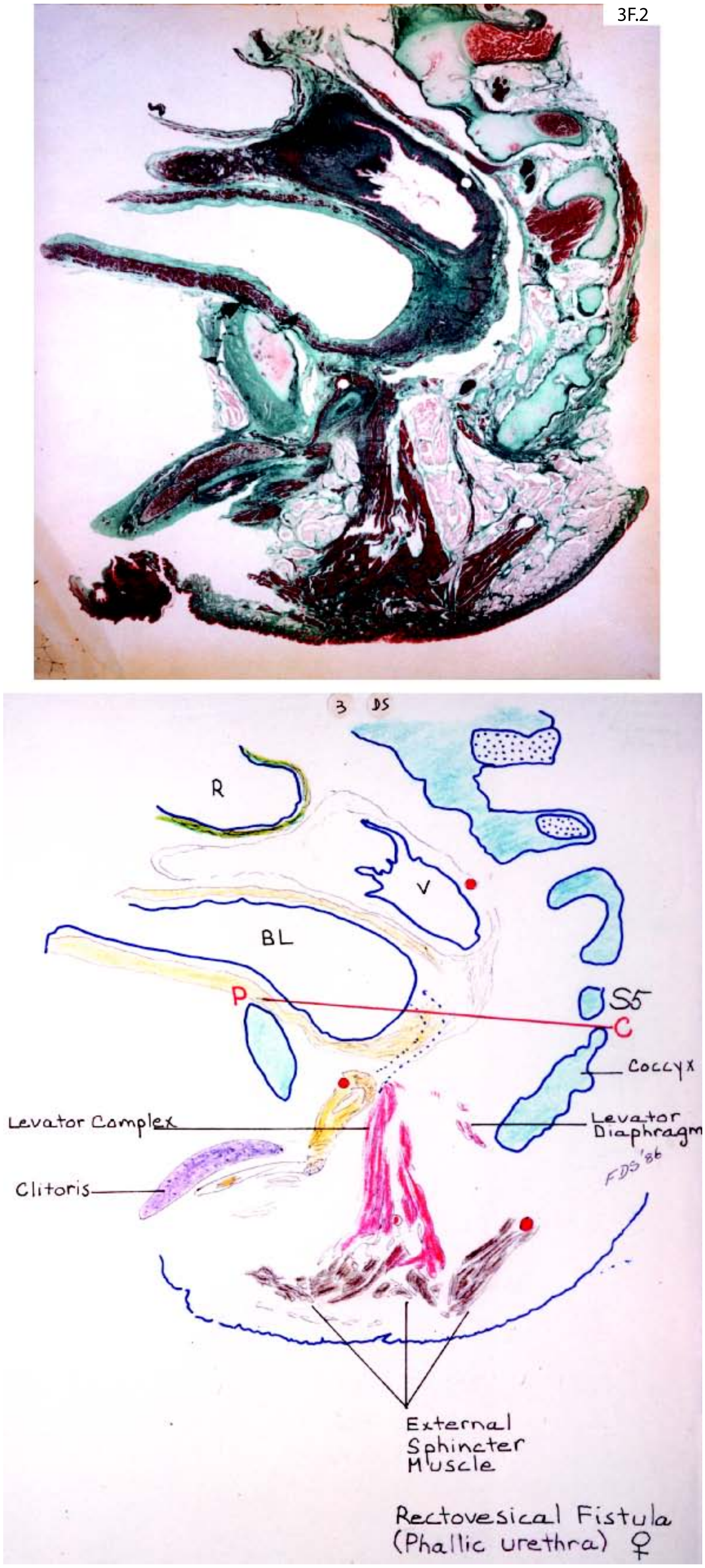


Fig. 6.7 Rectovesical fistula (phallic urethra) (3F.2)

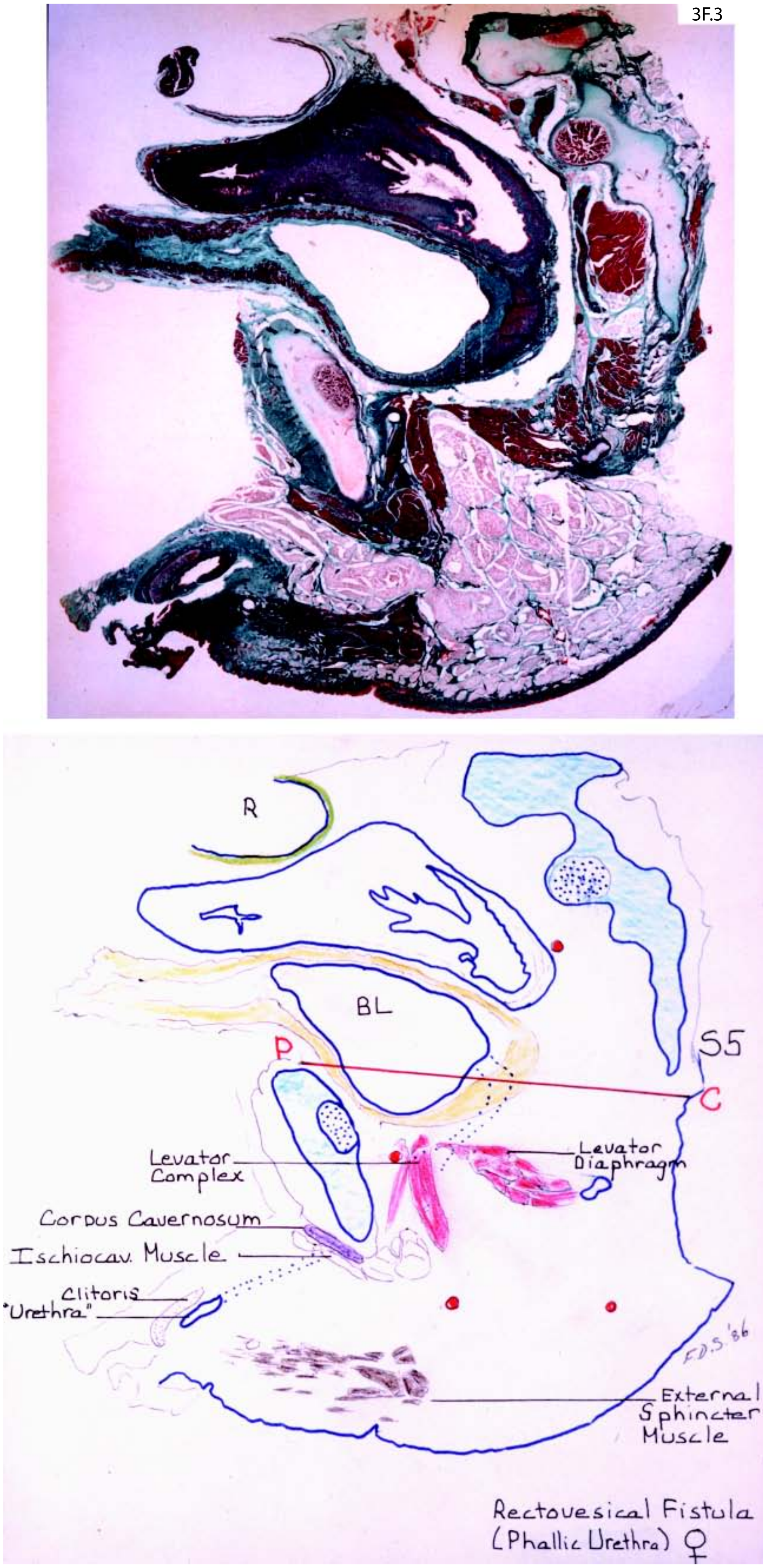


Fig. 6.8 Rectovesical fistula (phallic urethra) (3F.3)

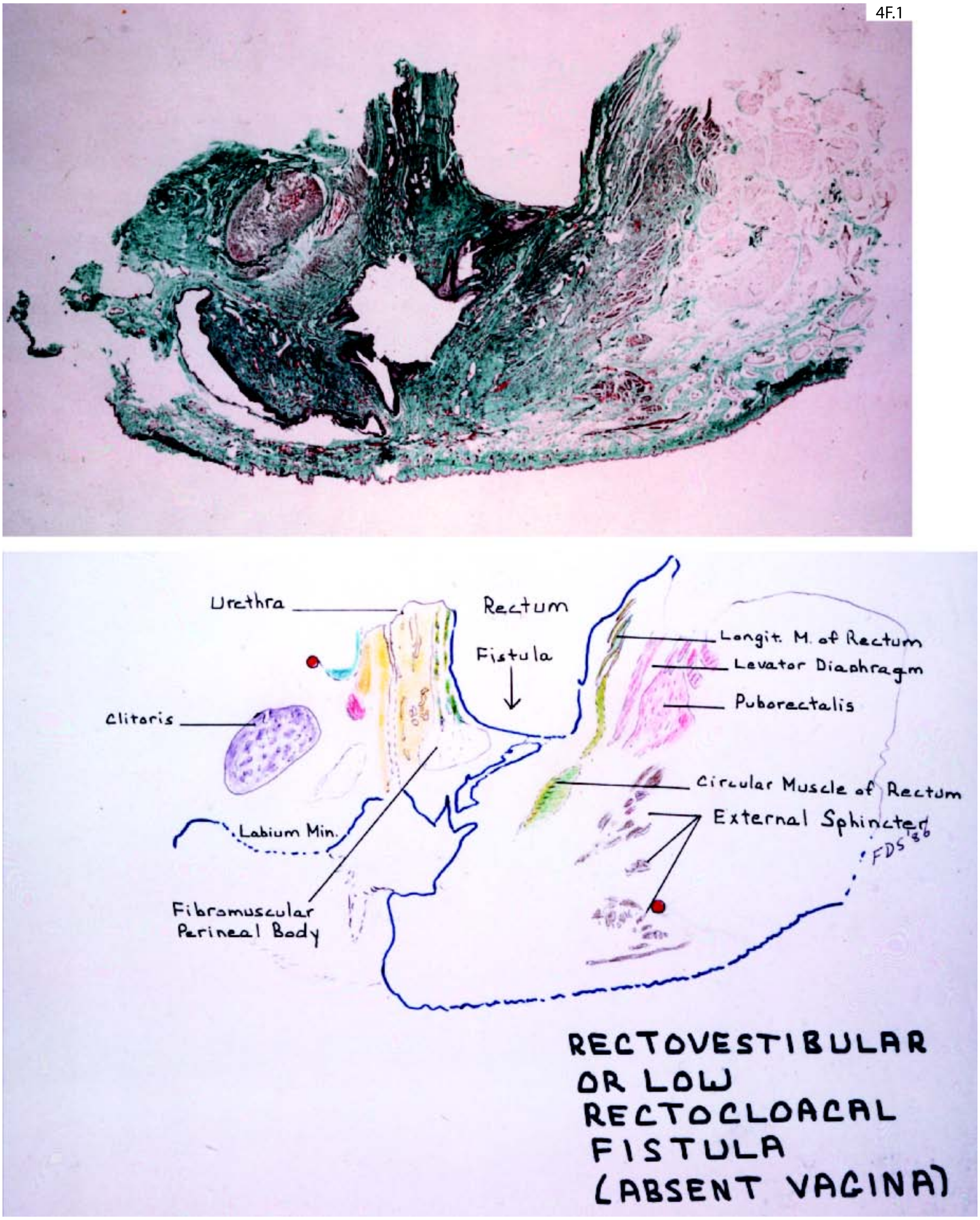
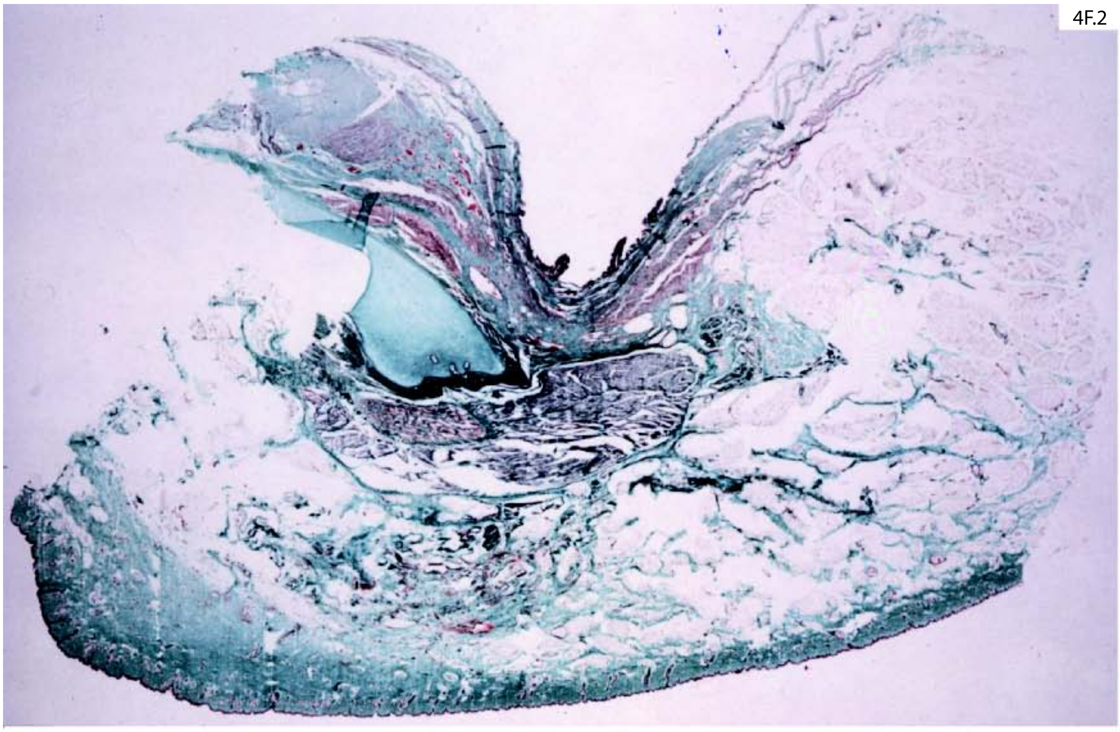


Fig. 6.9 Rectovestibular or low rectocloacal fistula (absent vagina) (4F.1)



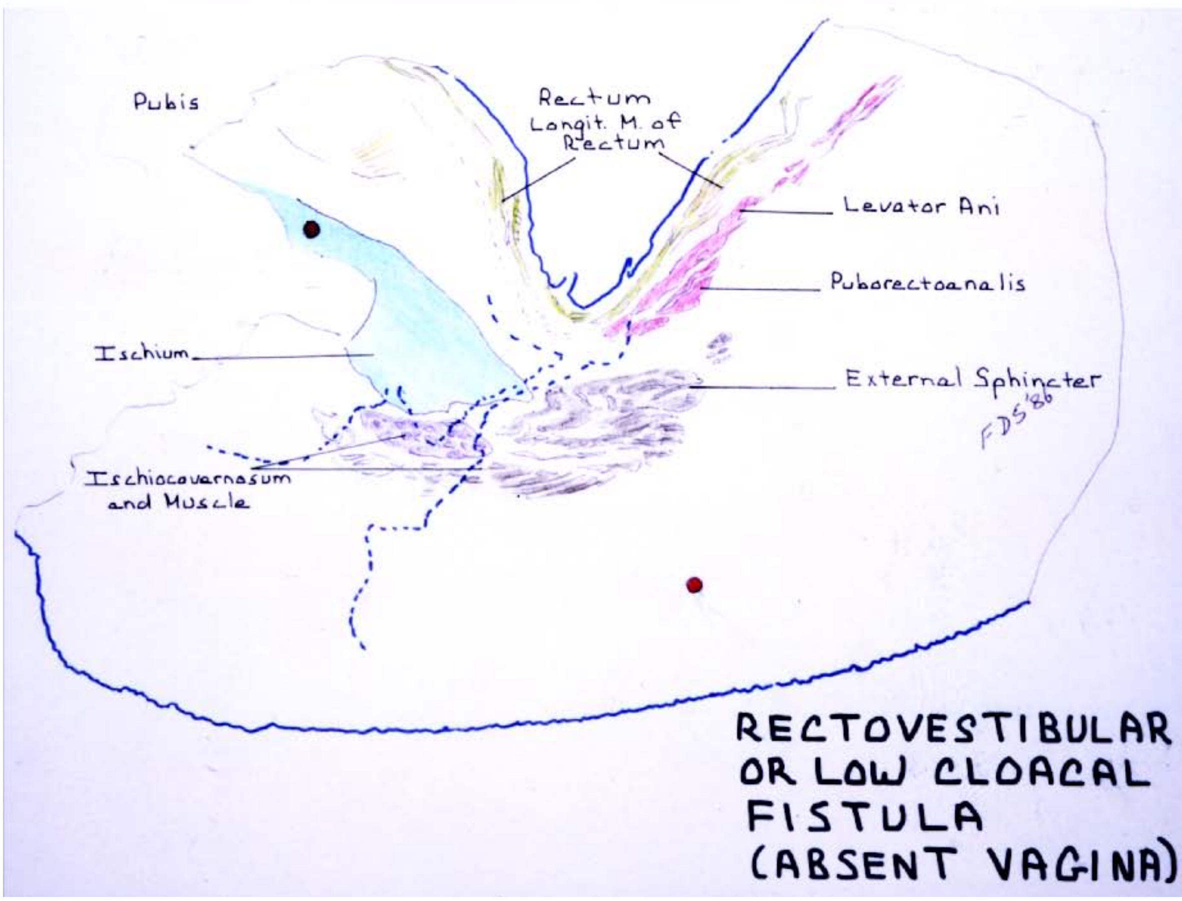


Fig. 6.10 Rectovestibular or low cloacal fistula (absent vagina) (4F.2)

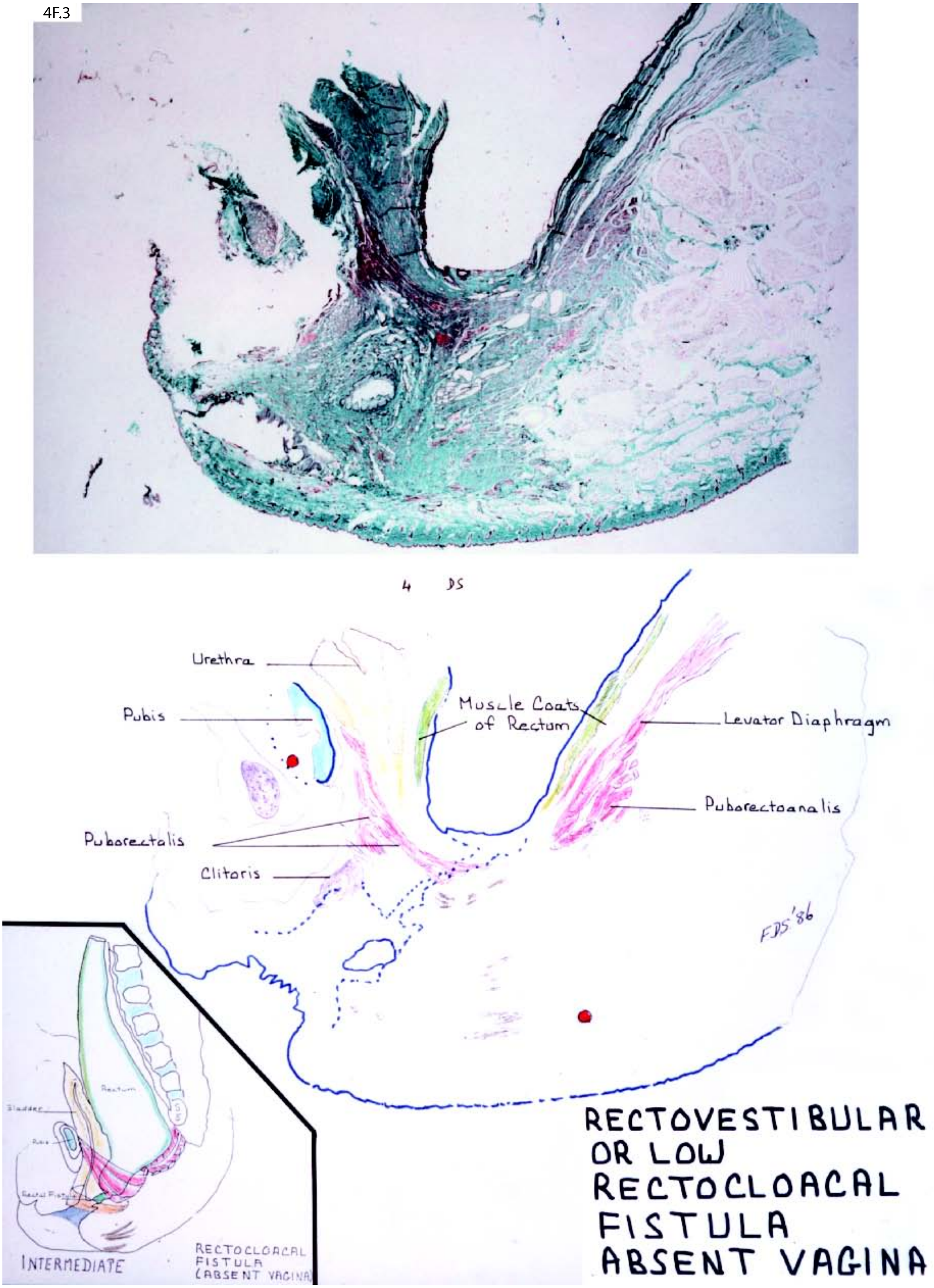


Fig. 6.11 Rectovestibular or low rectocloacal fistula (absent vagina) (4F.3)

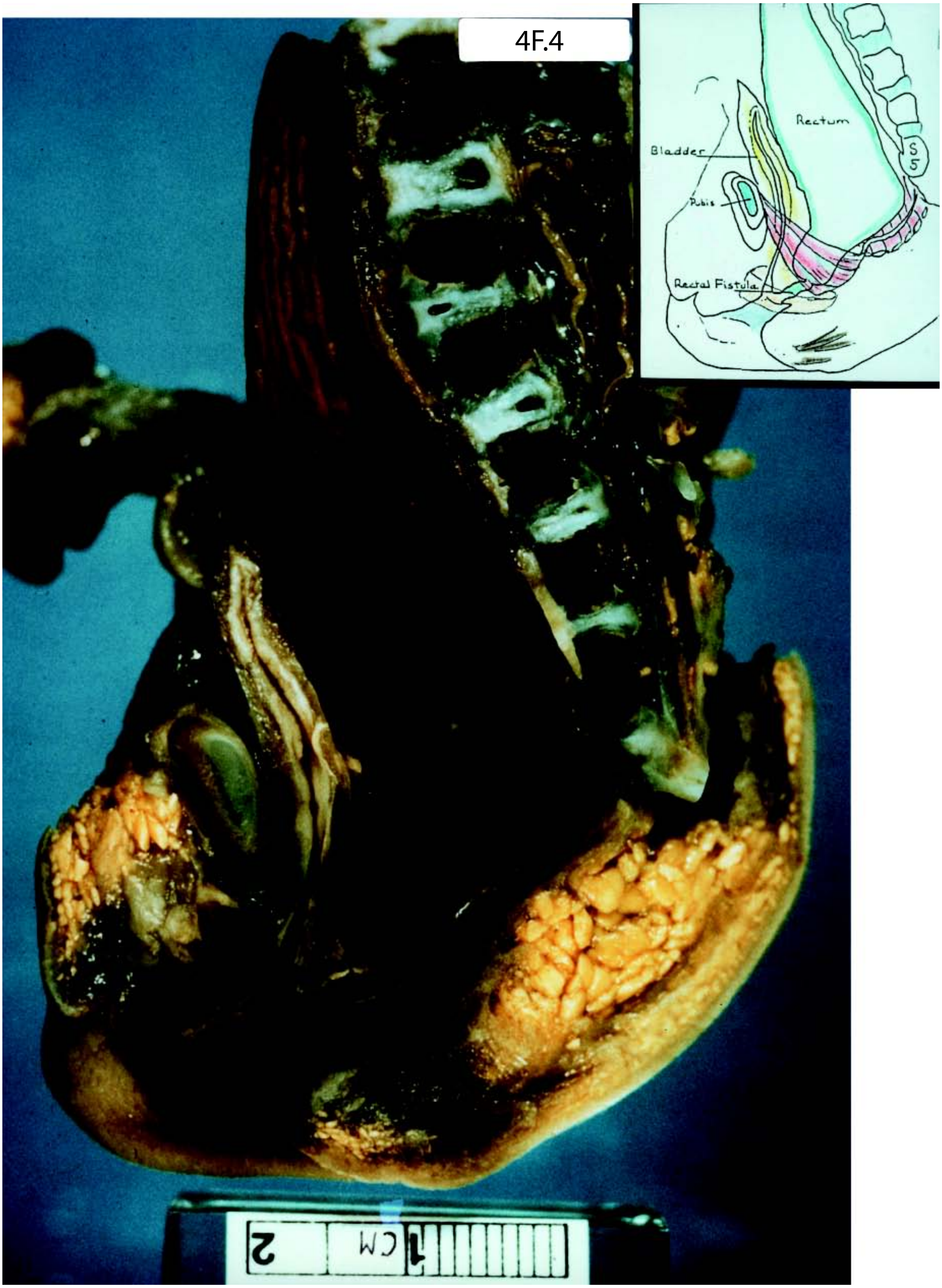
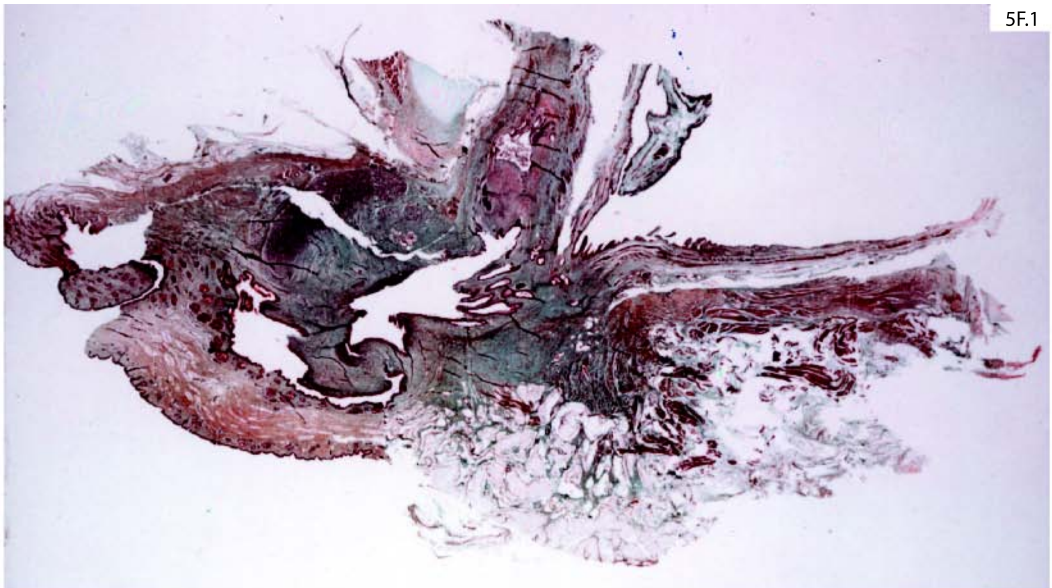


Fig. 6.12 Cloacal anomaly (absent vagina, same as Fig. 6.11) (4F.4)



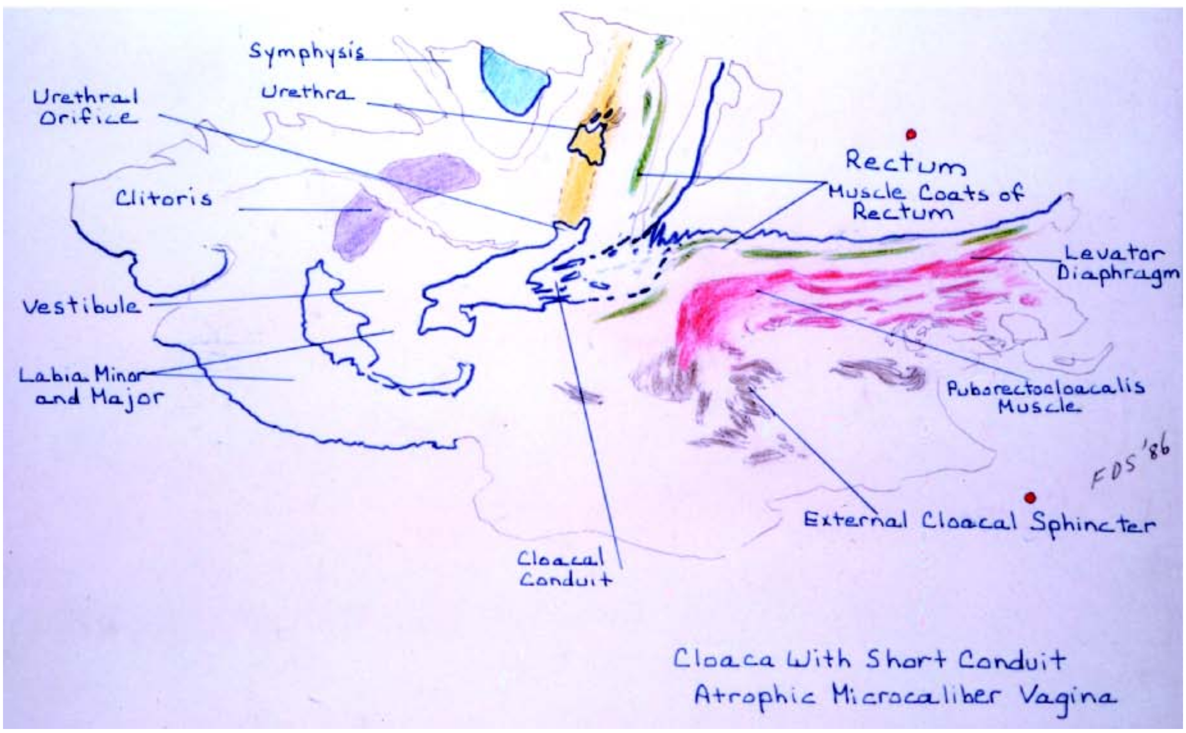


Fig. 6.13 Cloaca with short conduit atrophic microcaliber vagina (5F.1)

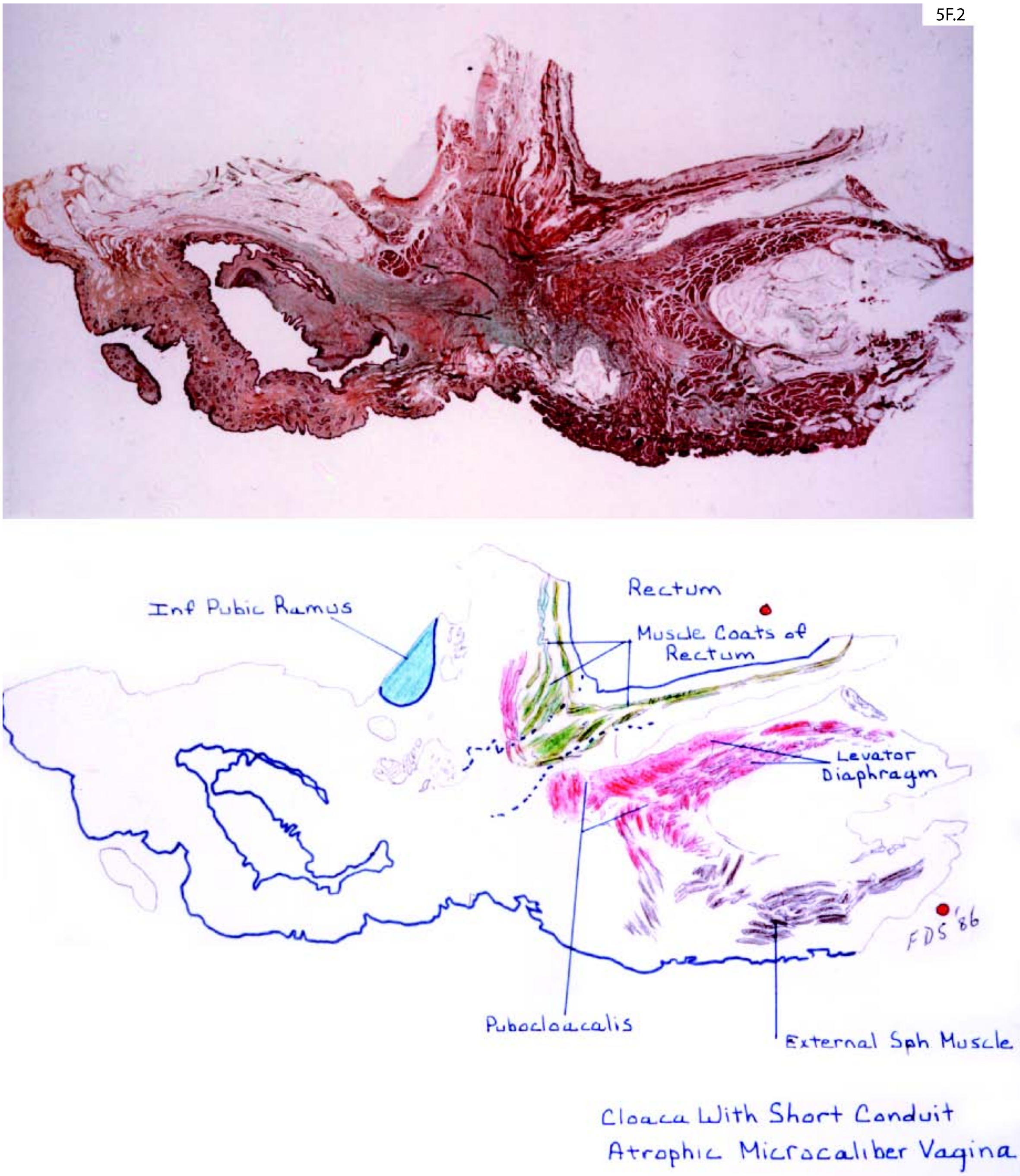


Fig. 6.14 Cloaca with short conduit atrophic microcaliber vagina (5F.2)

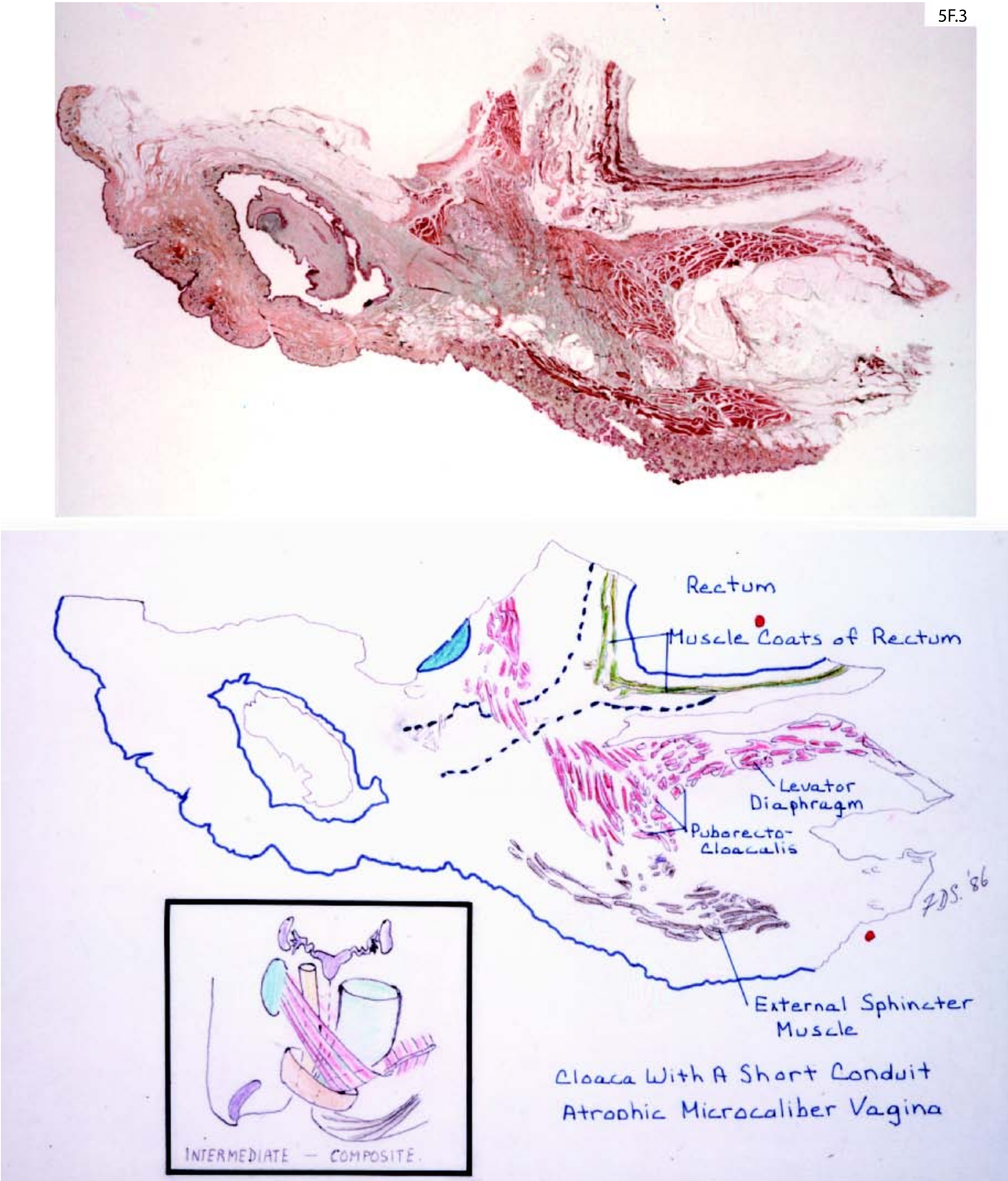


Fig. 6.15 Cloaca with short conduit atrophic microcaliber vagina (5F.3)

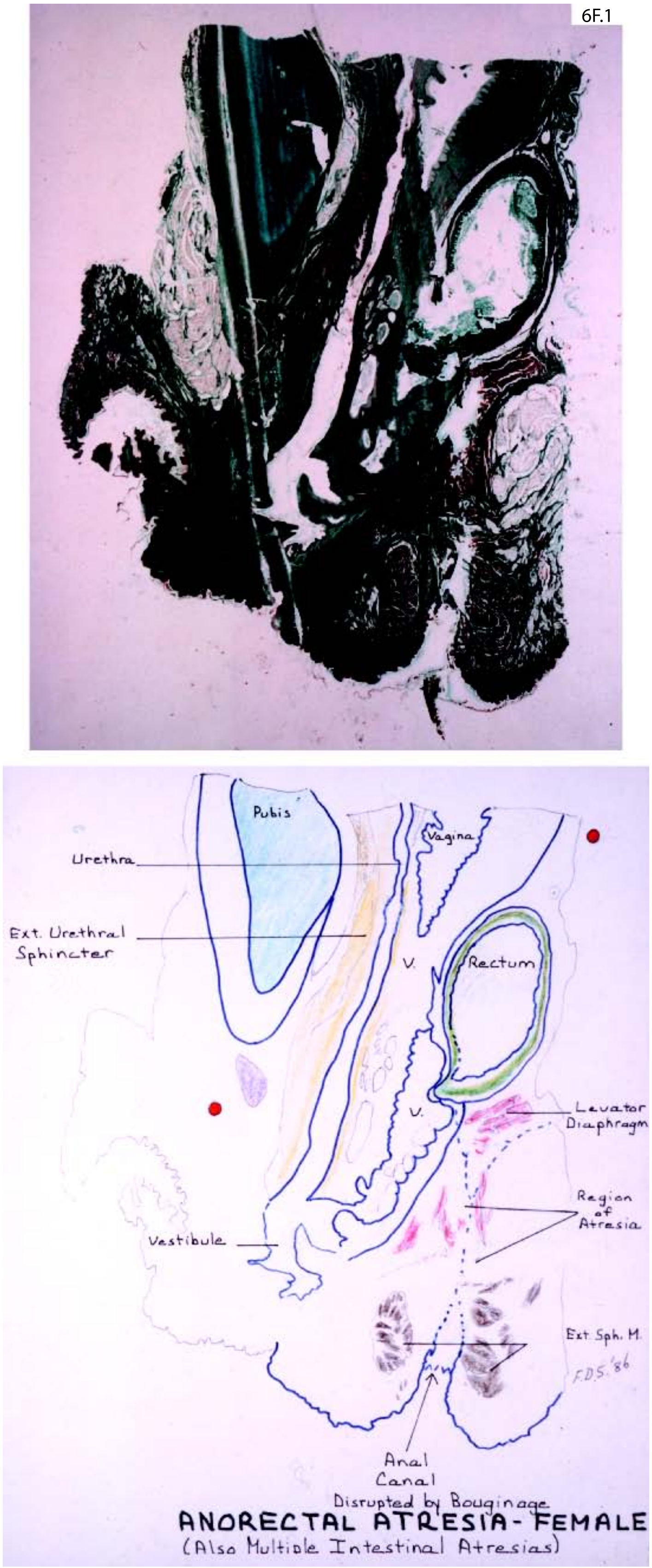


Fig. 6.16 Anorectal atresia, female (also multiple intestinal atresias) (6F.1)

6F.2

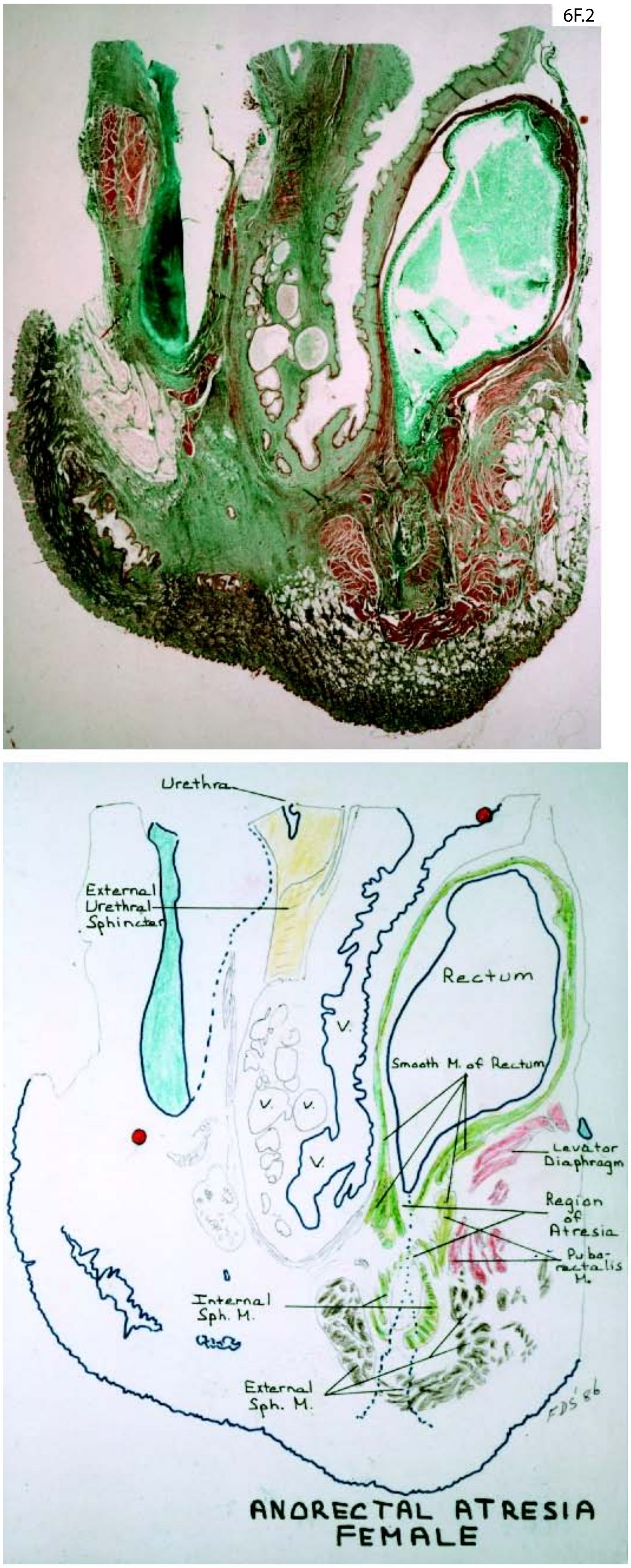
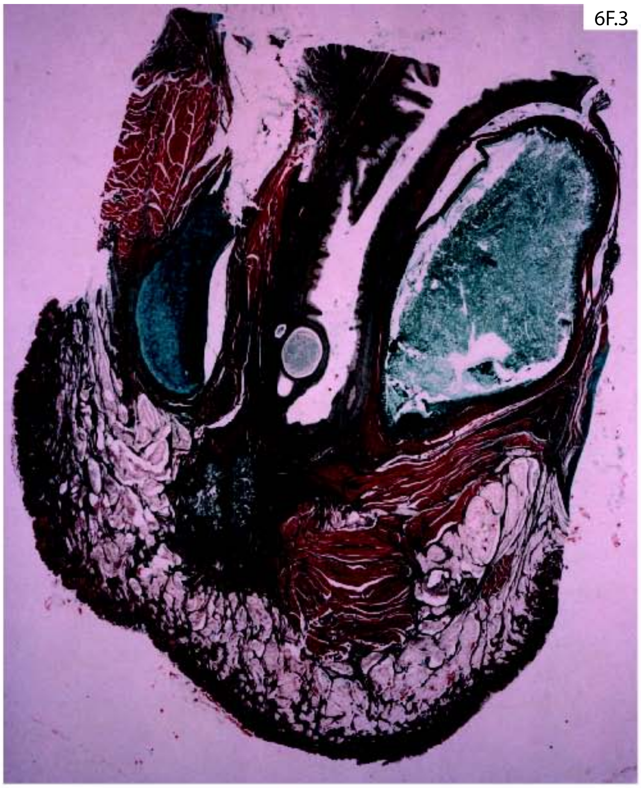


Fig. 6.17 Anorectal atresia, female (6F.2)



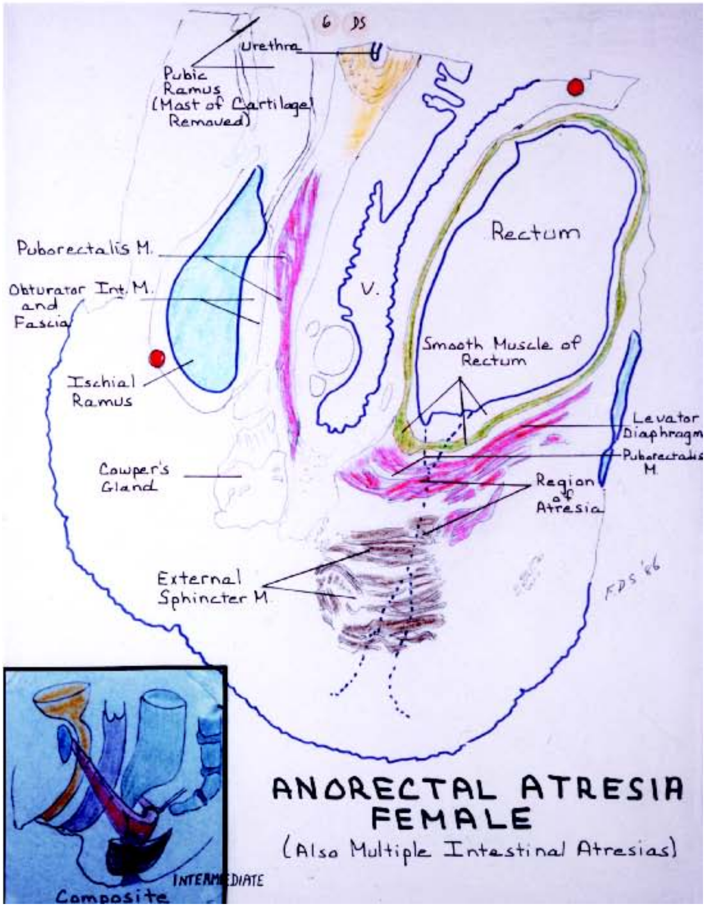


Fig. 6.18 Anorectal atresia, female (also multiple intestinal atresias) (6F.3)

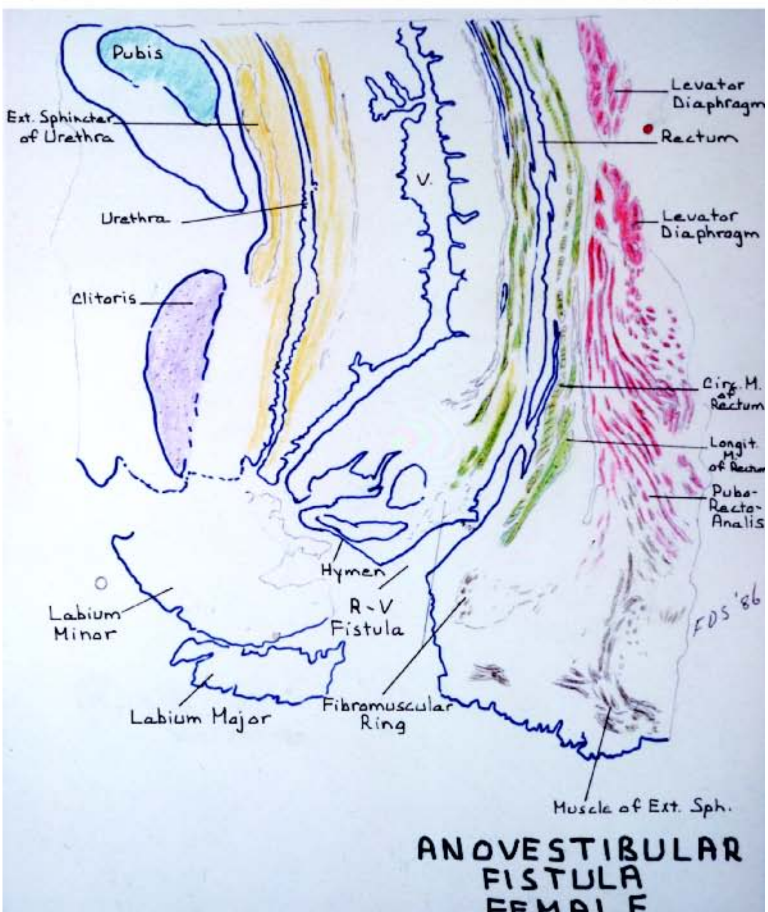


Fig. 6.19 Anovestibular fistula, female (7F.1)

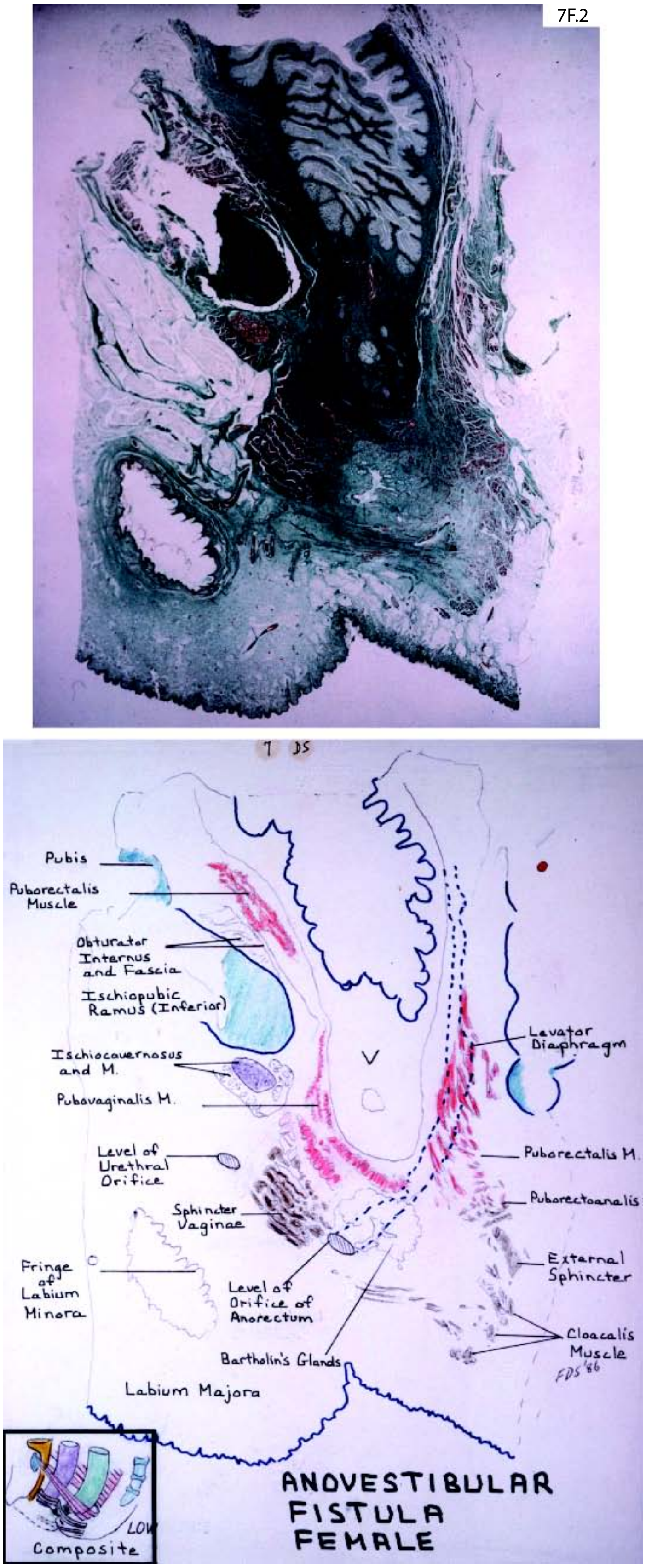


Fig. 6.20 Anovestibular fistula, female (7F.2)

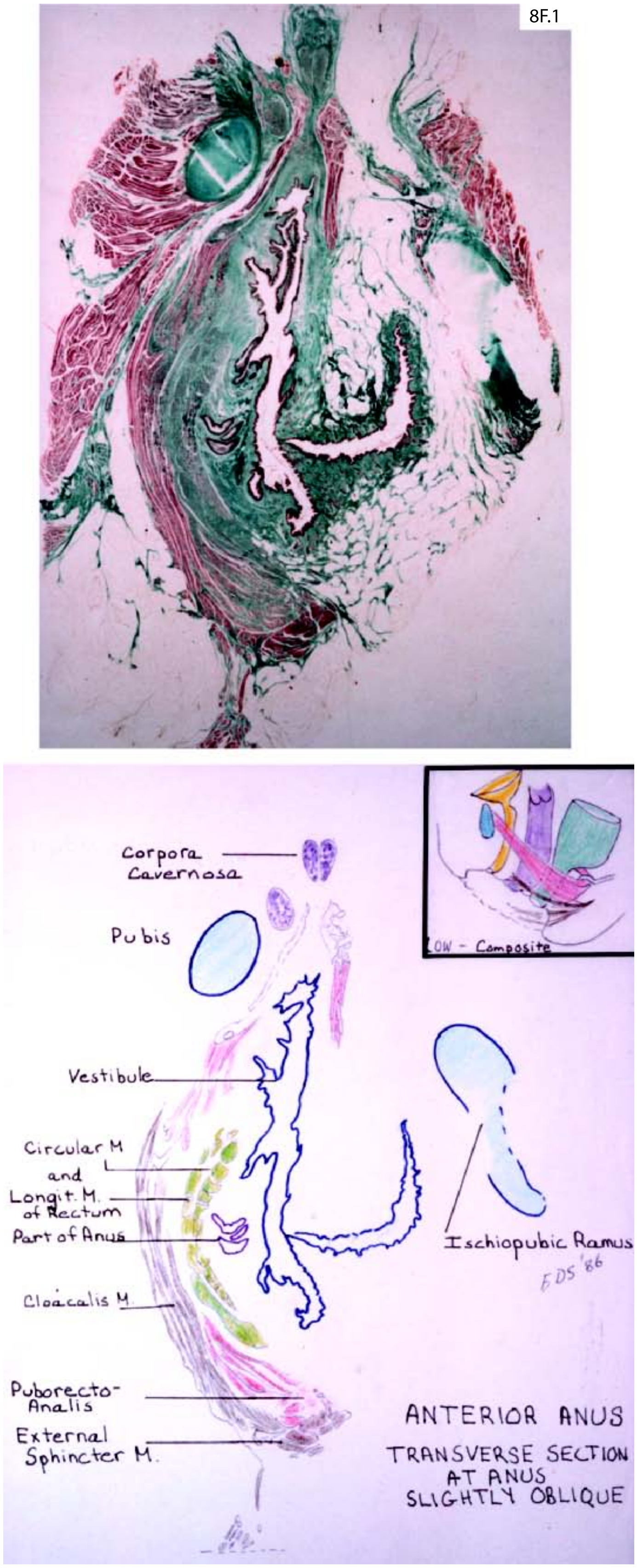


Fig. 6.21 Anterior anus, transverse section at anus slightly oblique (8F.1)

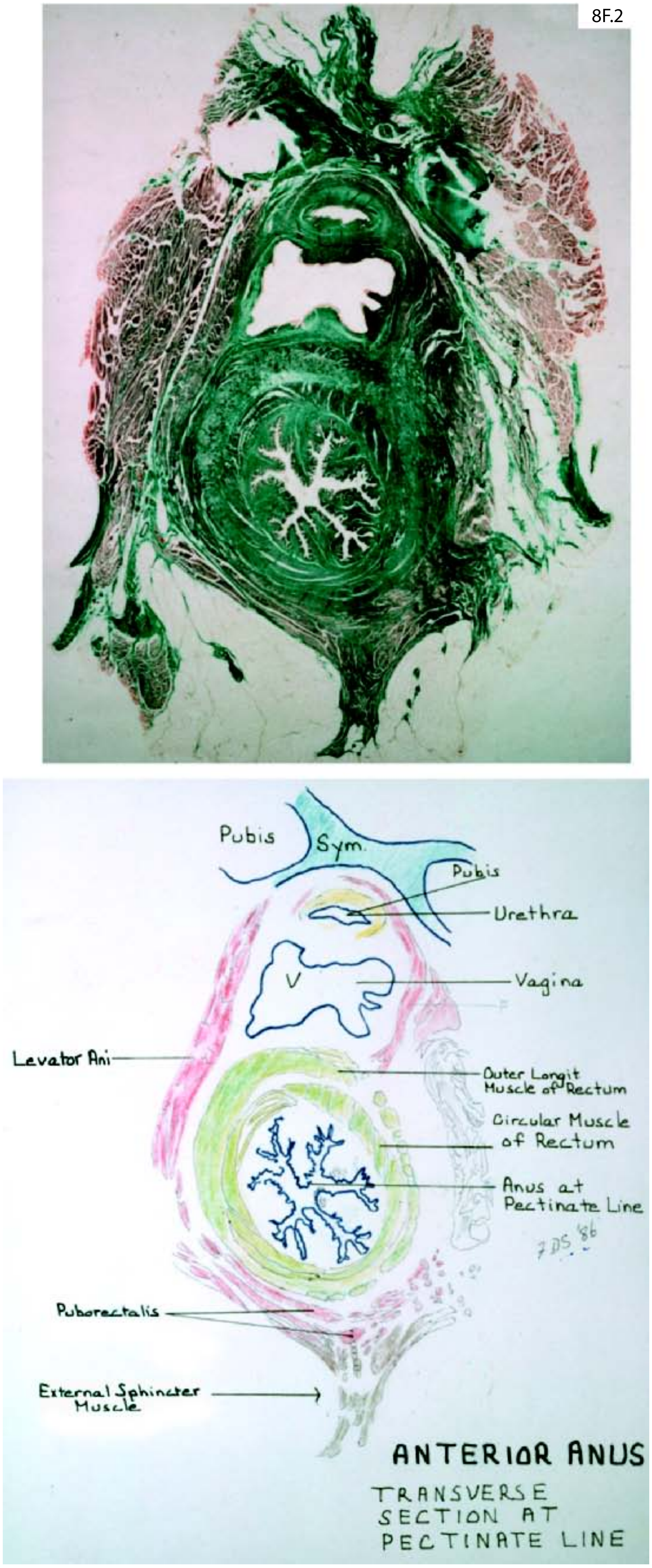


Fig. 6.22 Anterior anus, transverse section at pectinate line (8F.2)

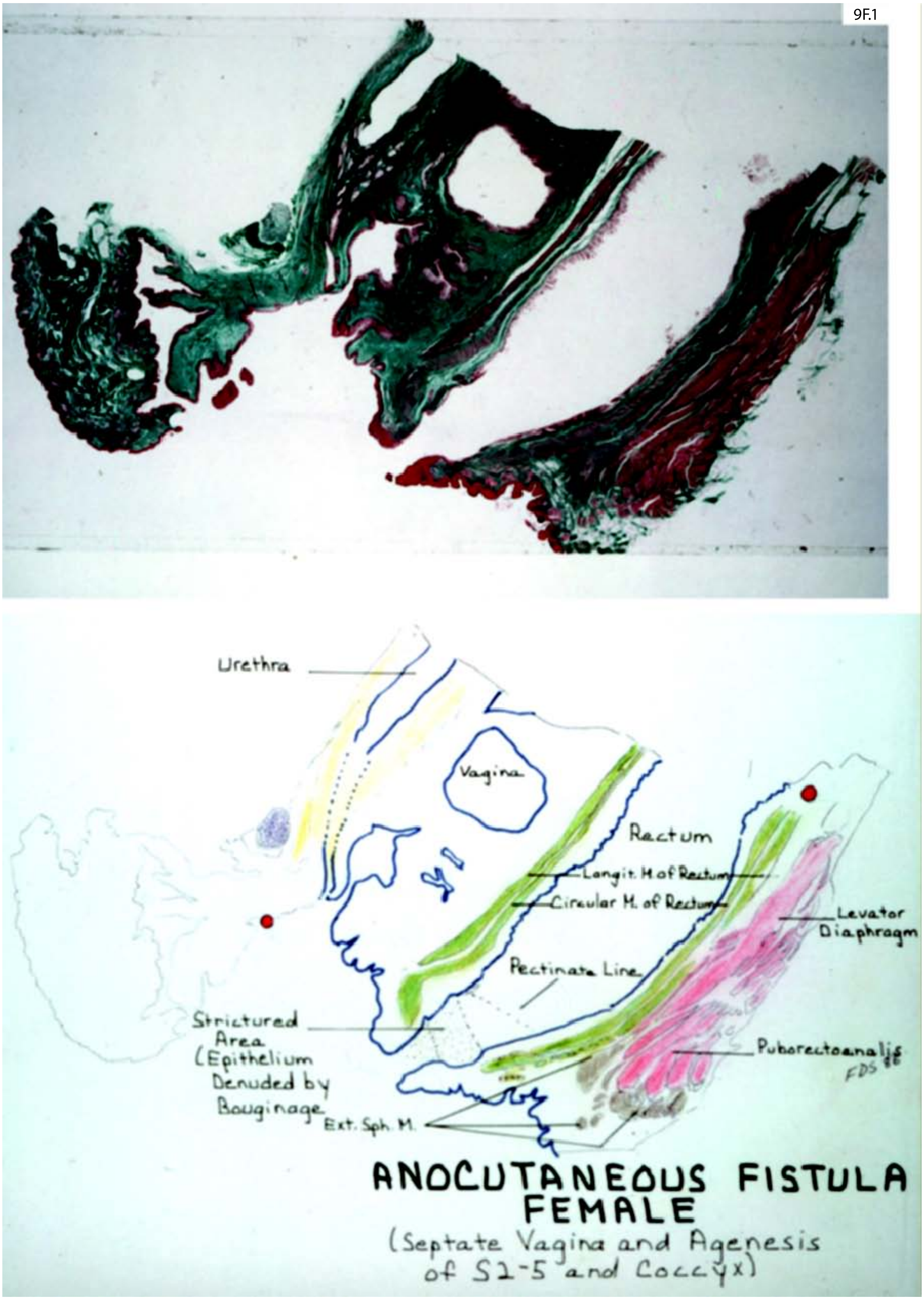


Fig. 6.23 Anocutaneous fistula, female (septate vagina and agenesis of S2–5 and coccyx) (9F.1)

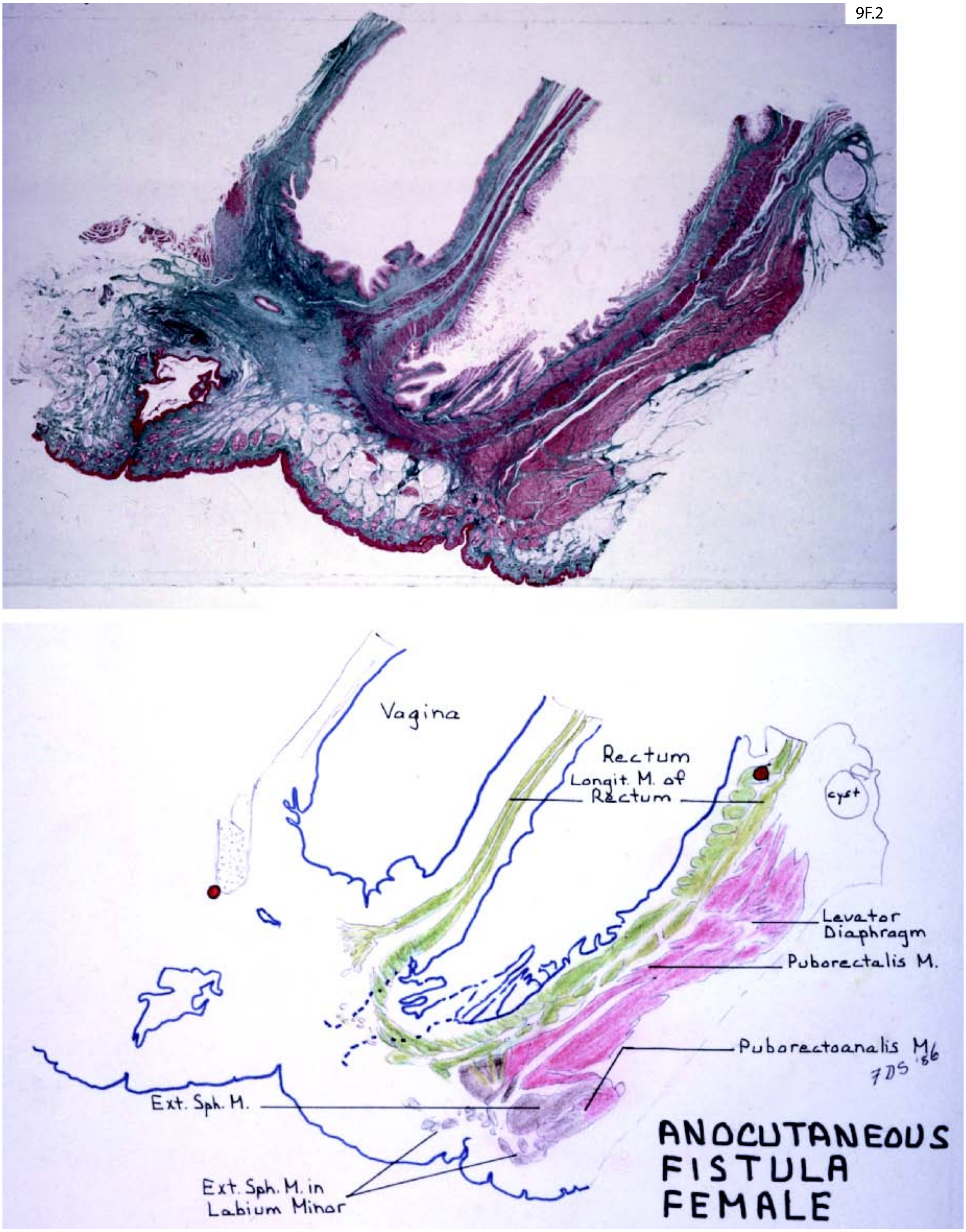


Fig. 6.24 Anocutaneous fistula, female (9F.2)

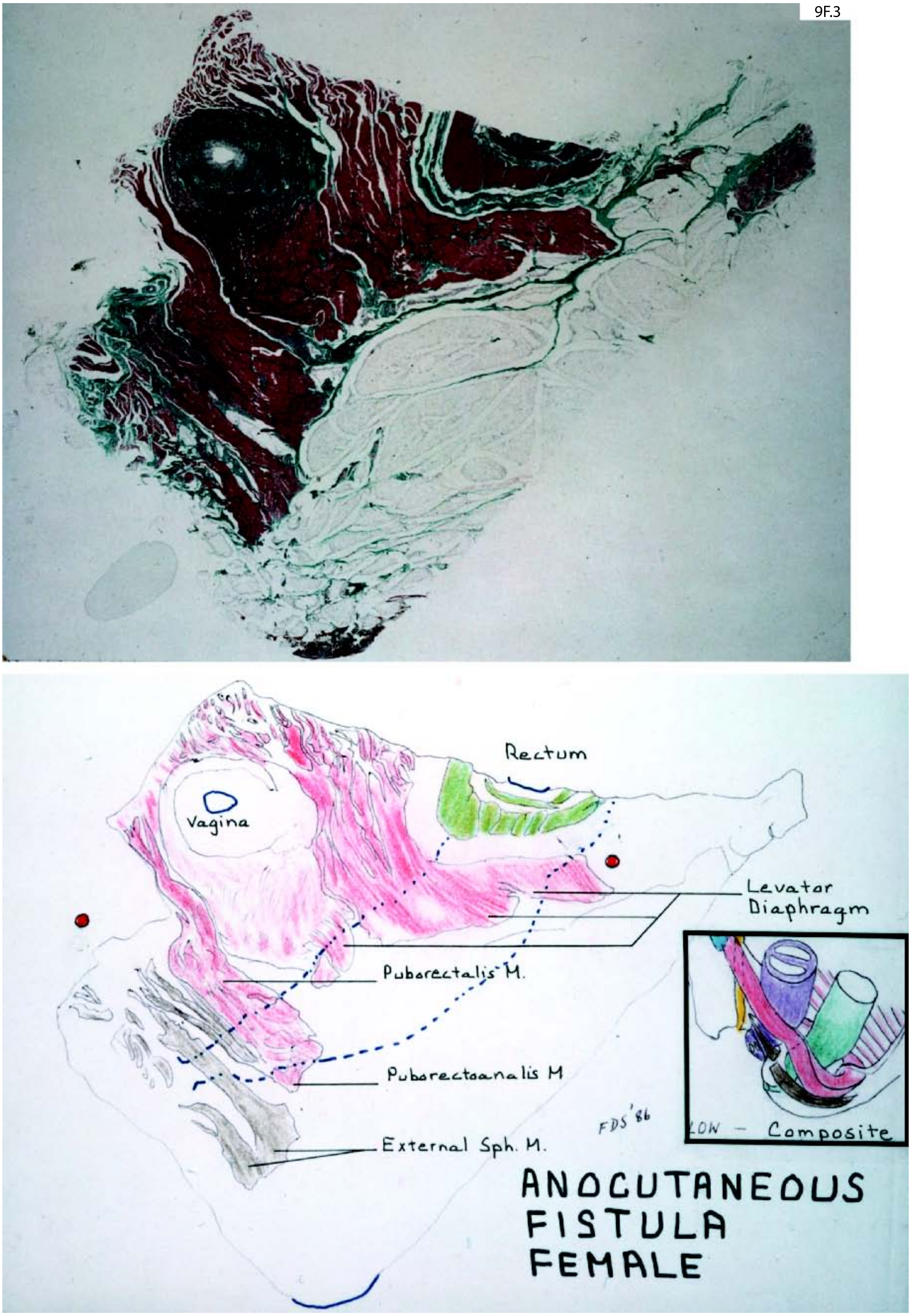


Fig. 6.25 Anocutaneous fistula, female (9F.3)

10F.1

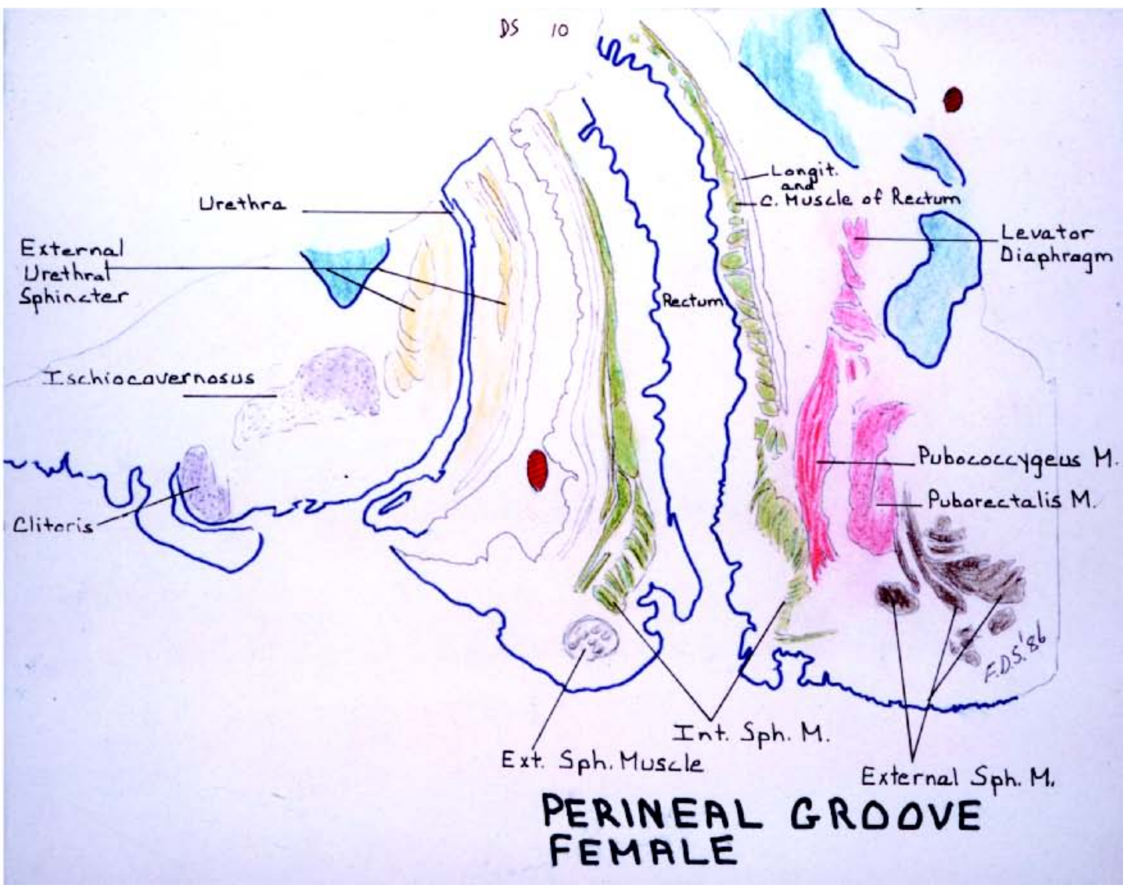


Fig. 6.26 Perineal groove, female (10F.1)

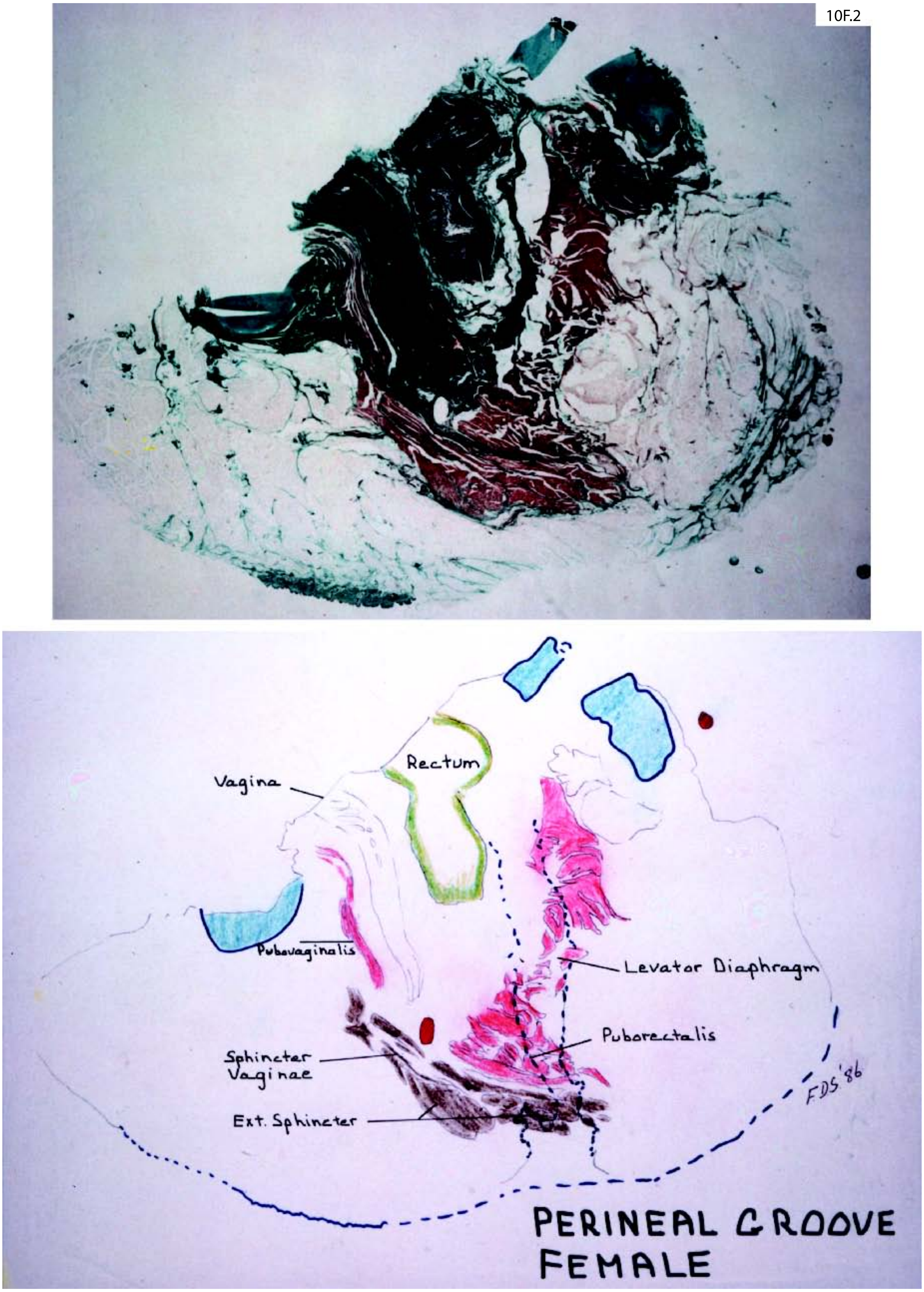


Fig. 6.27 Perineal groove, female (10F.2)

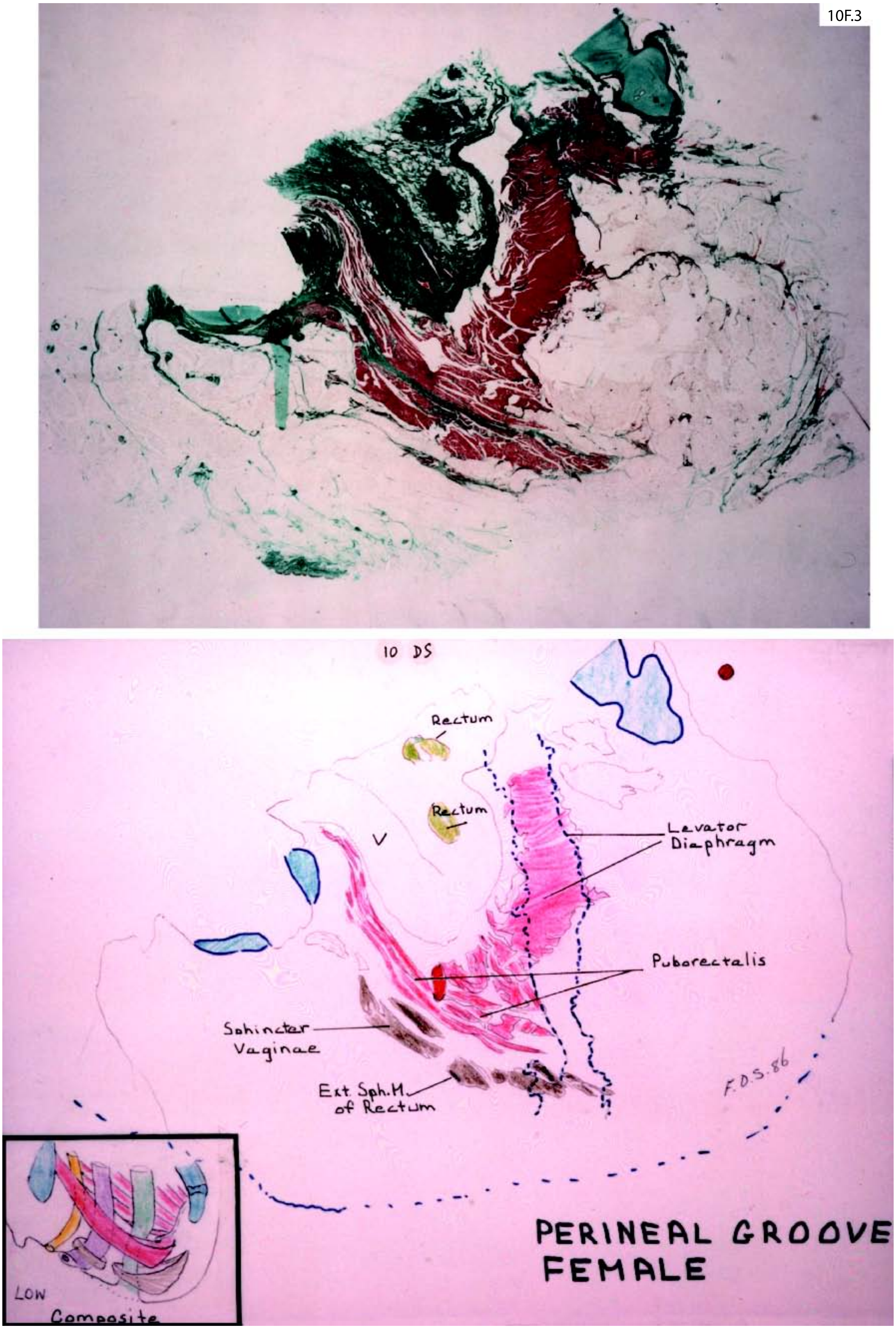


Fig. 6.28 Perineal groove, female (10F.3)

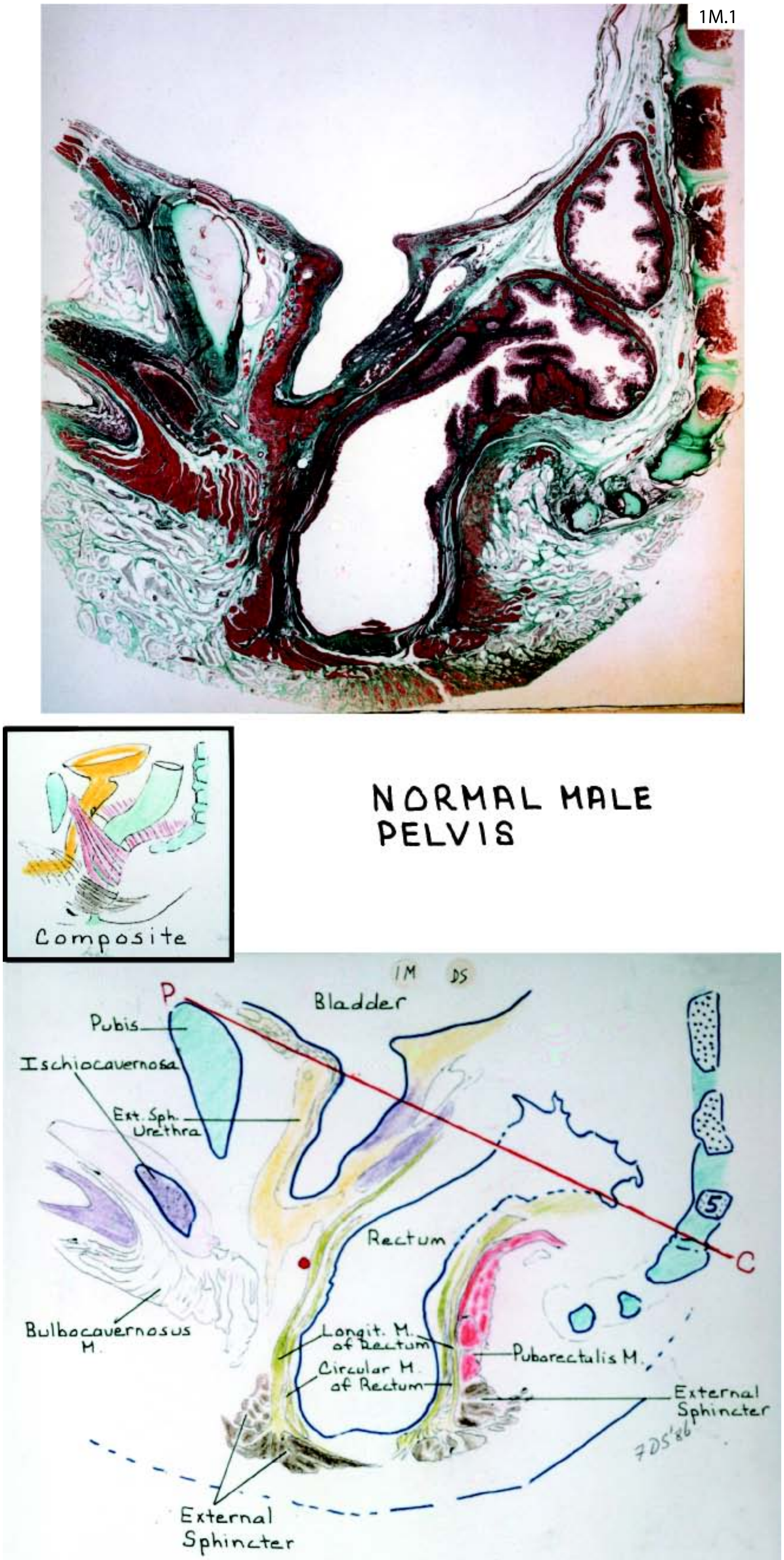


Fig. 6.29 Normal male pelvis (1M.1)

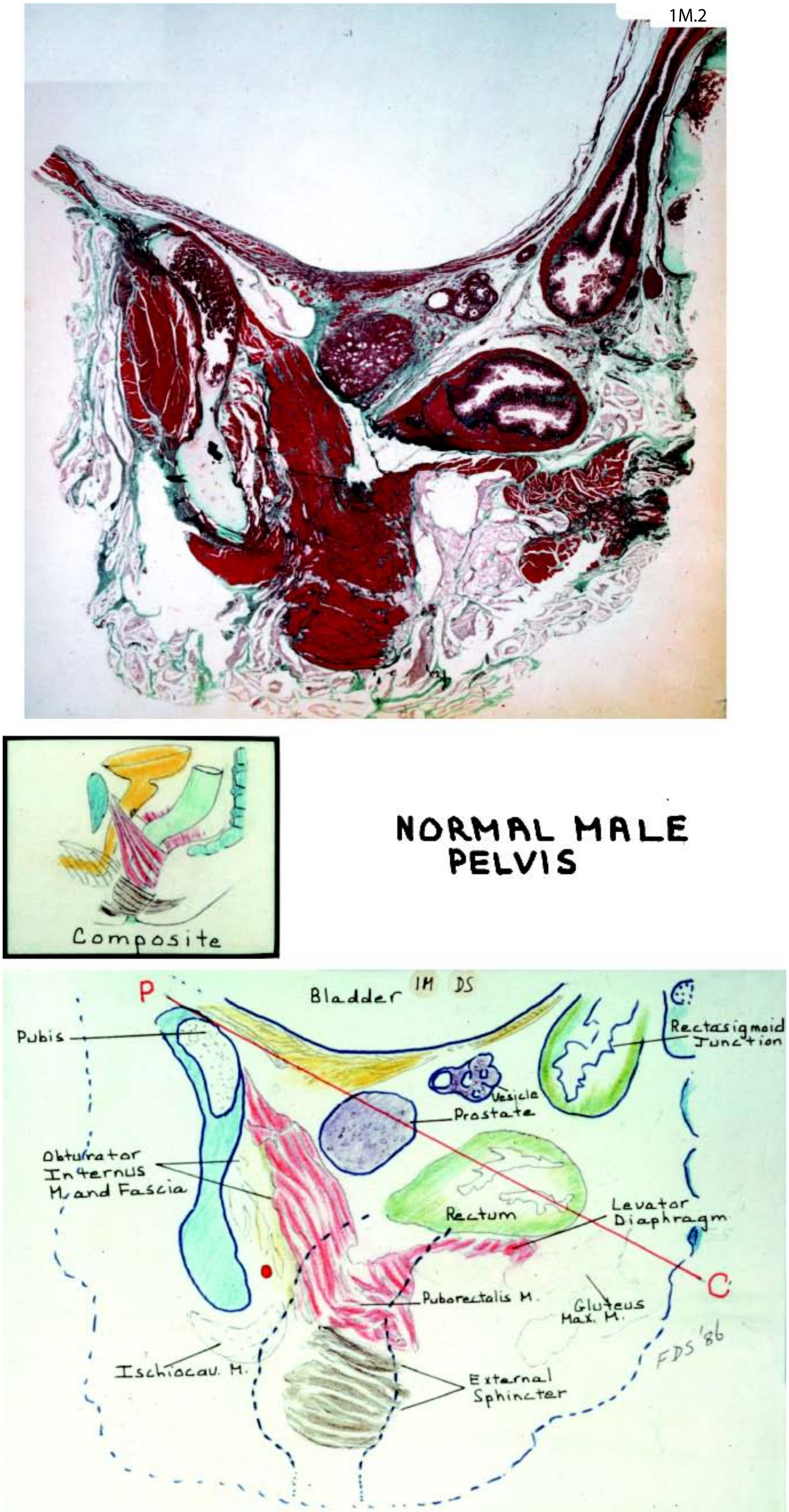


Fig. 6.30 Normal male pelvis (1M.2)

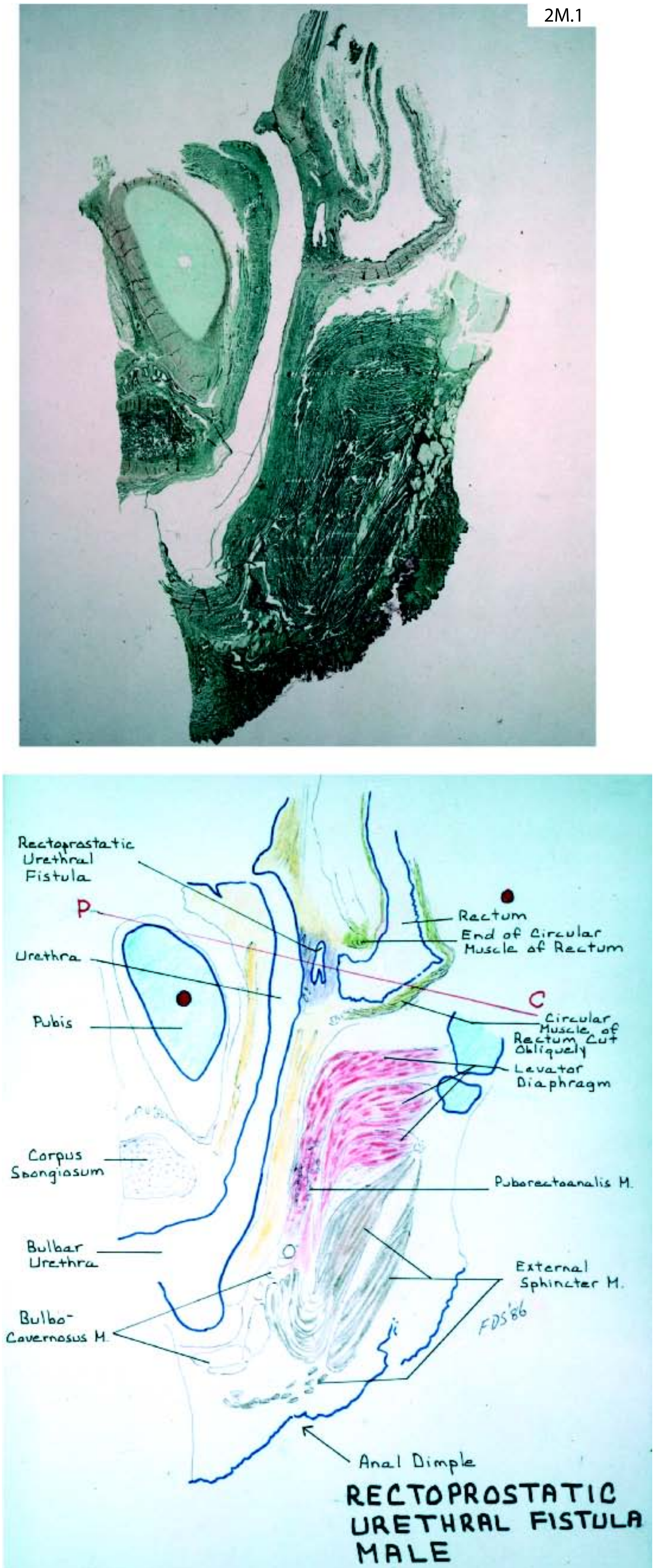


Fig. 6.31 Rectoprostatic urethral fistula, male (2M.1)

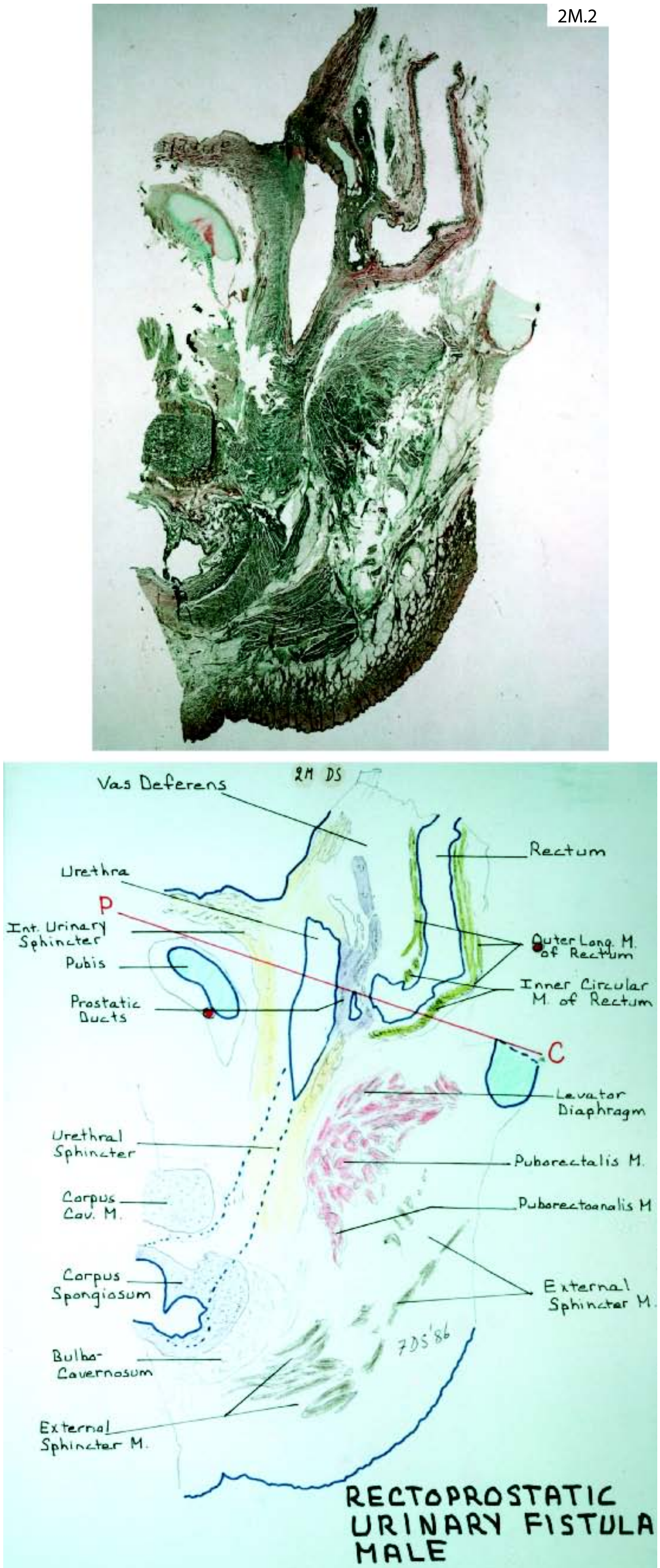


Fig. 6.32 Rectoprostatic urinary fistula, male (2M.2)

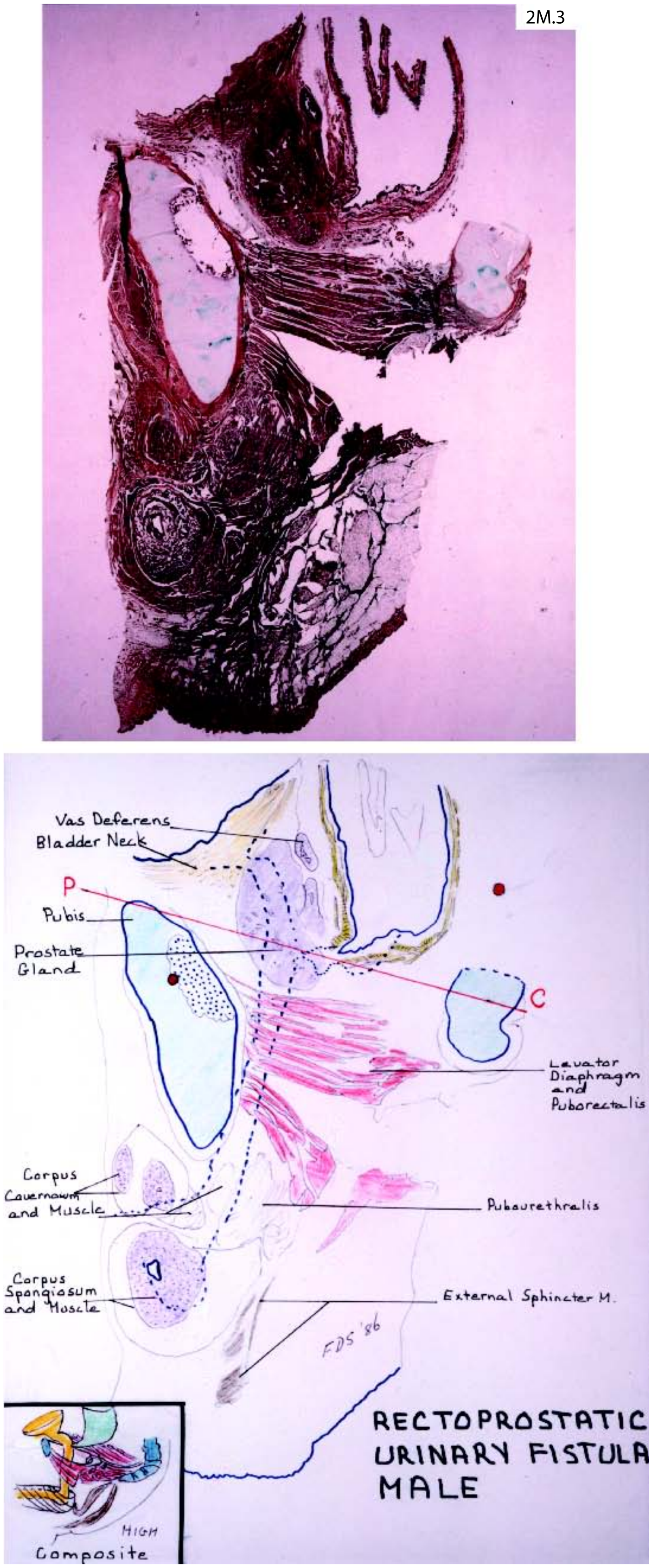
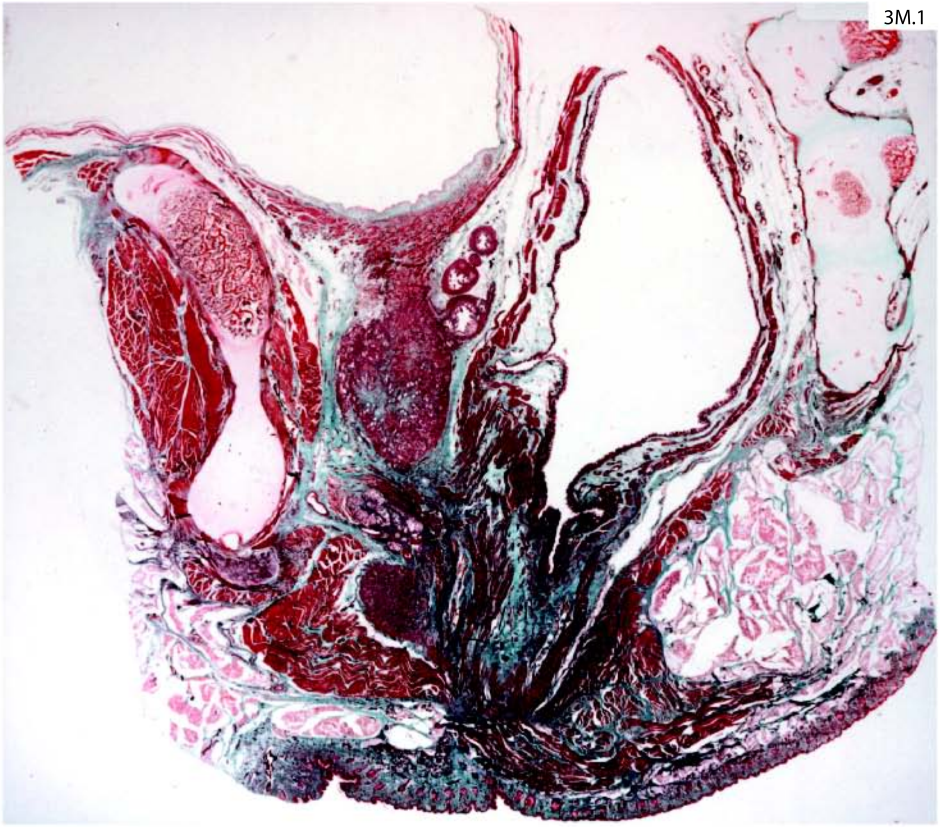


Fig. 6.33 Rectoprostatic urinary fistula, male (2M.3)



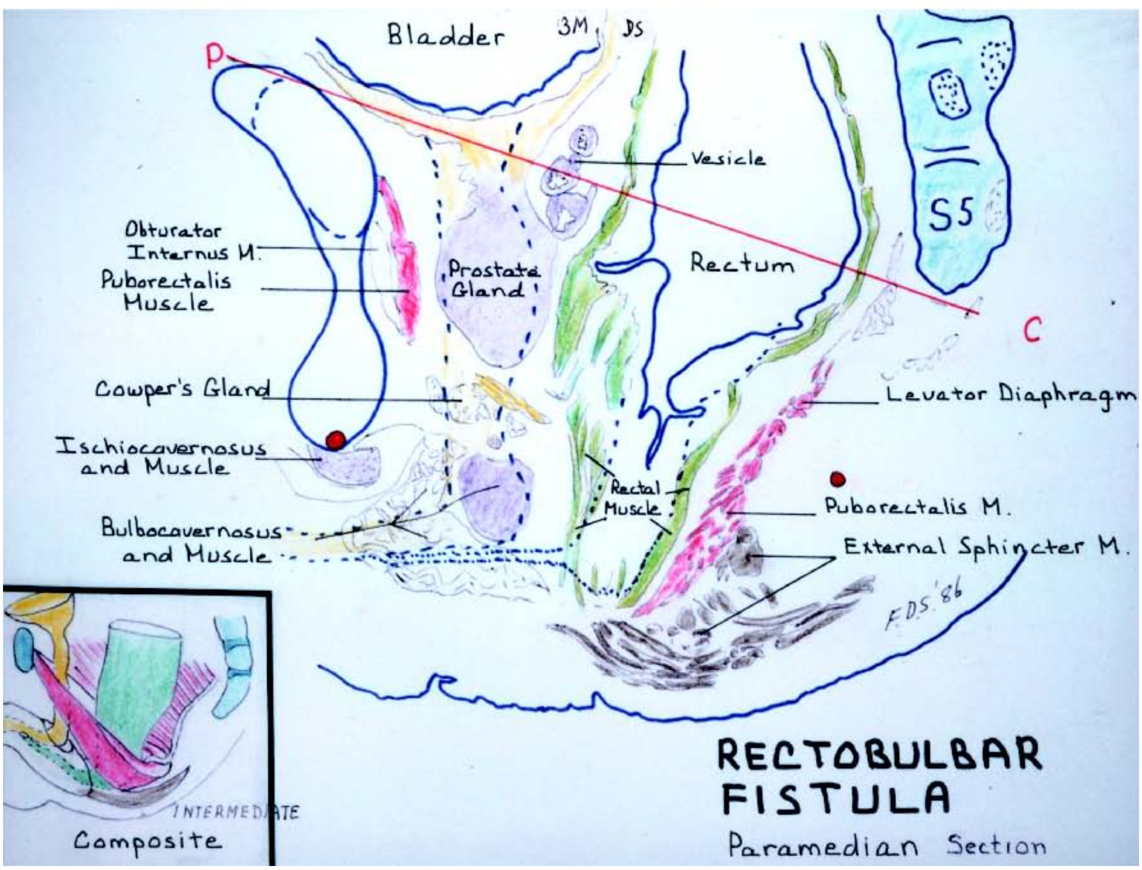


Fig. 6.34 Rectobulbar fistula, paramedian section (3M.1)

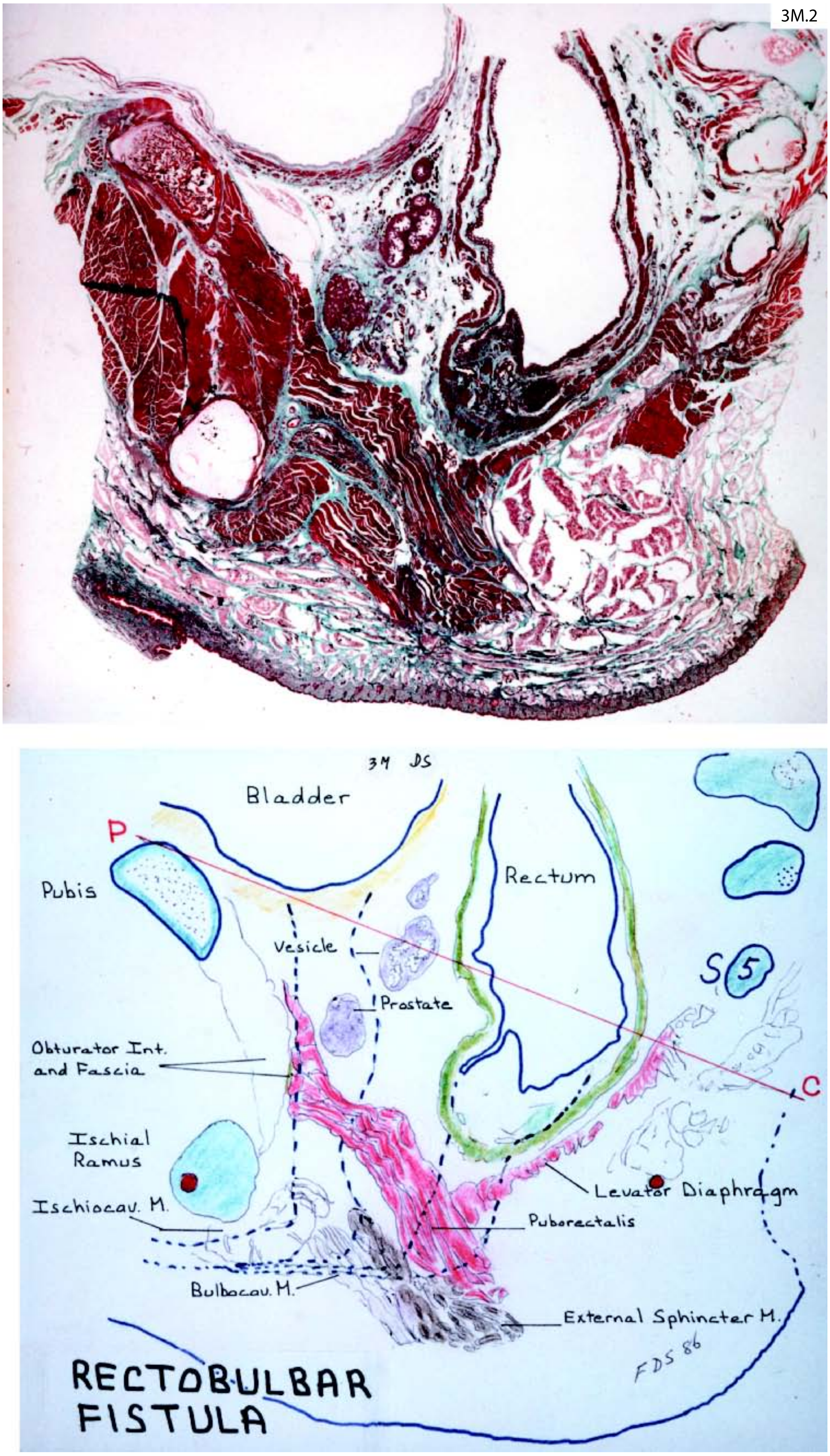


Fig. 6.35 Rectobulbar fistula (3M.2)

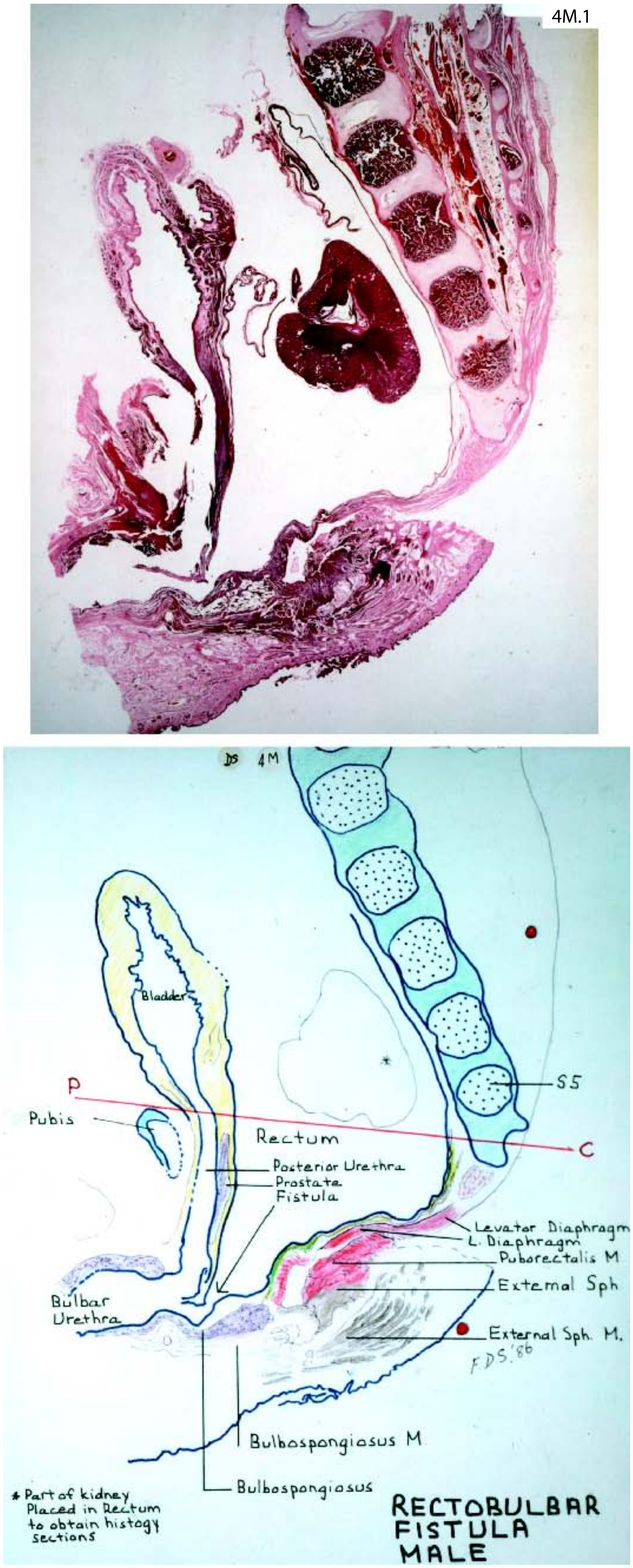


Fig. 6.36 Rectobulbar fistula, male (4M.1)

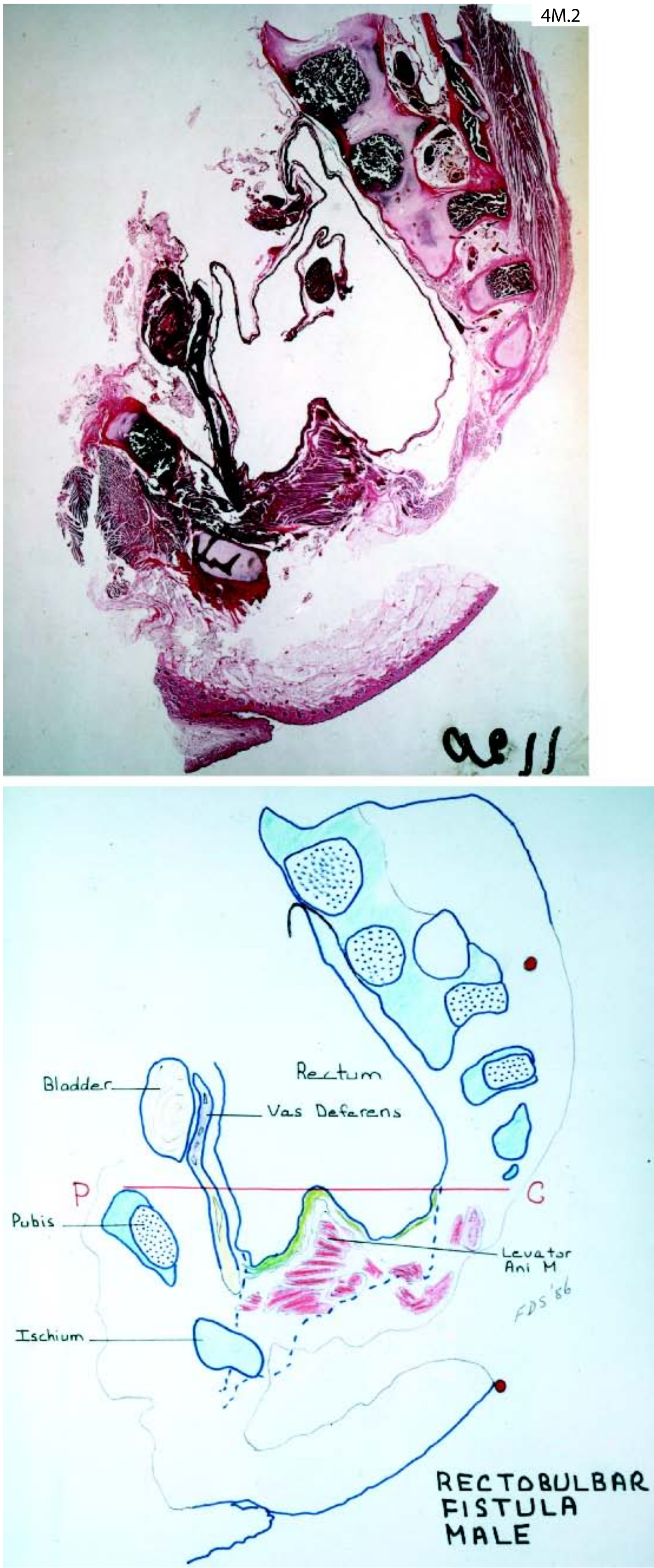


Fig. 6.37 Rectobulbar fistula, male (4M.2)

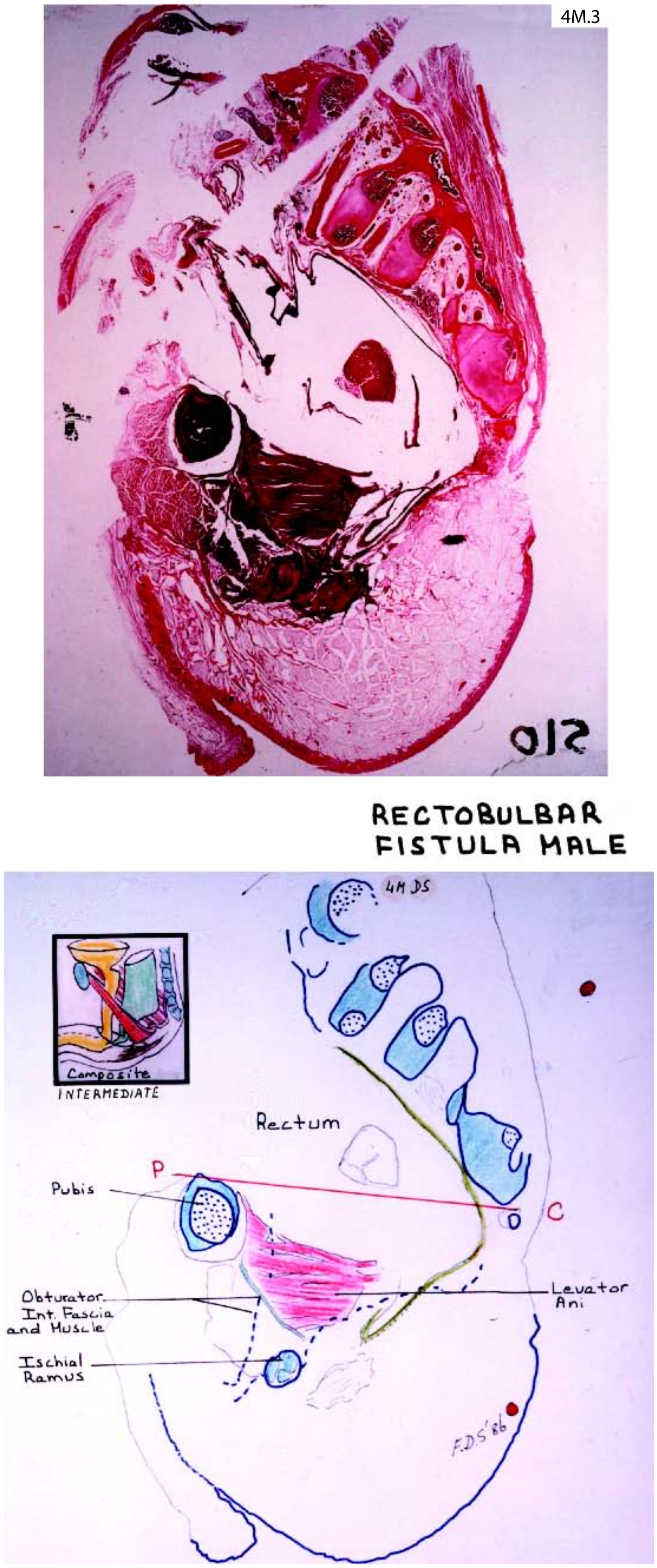


Fig. 6.38 Rectobulbar fistula, male (4M.3)

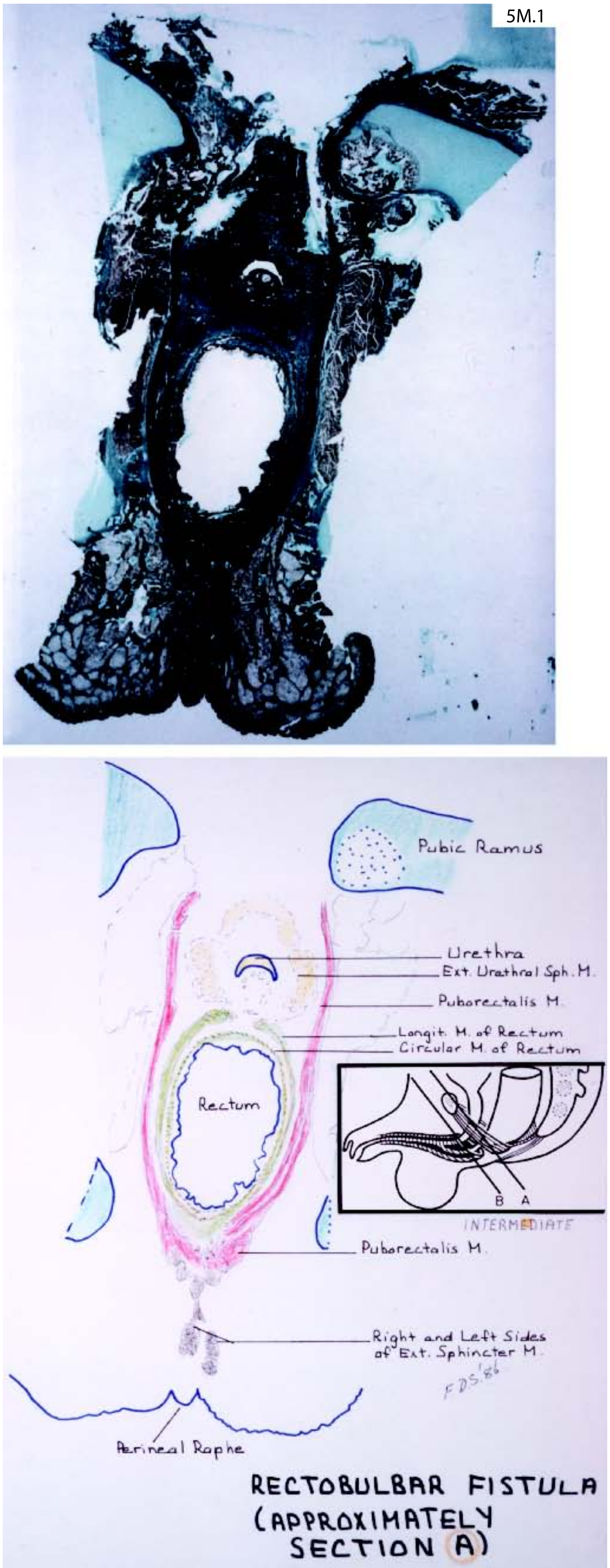


Fig. 6.39 Rectobulbar fistula (approximately section A) (5M.1)

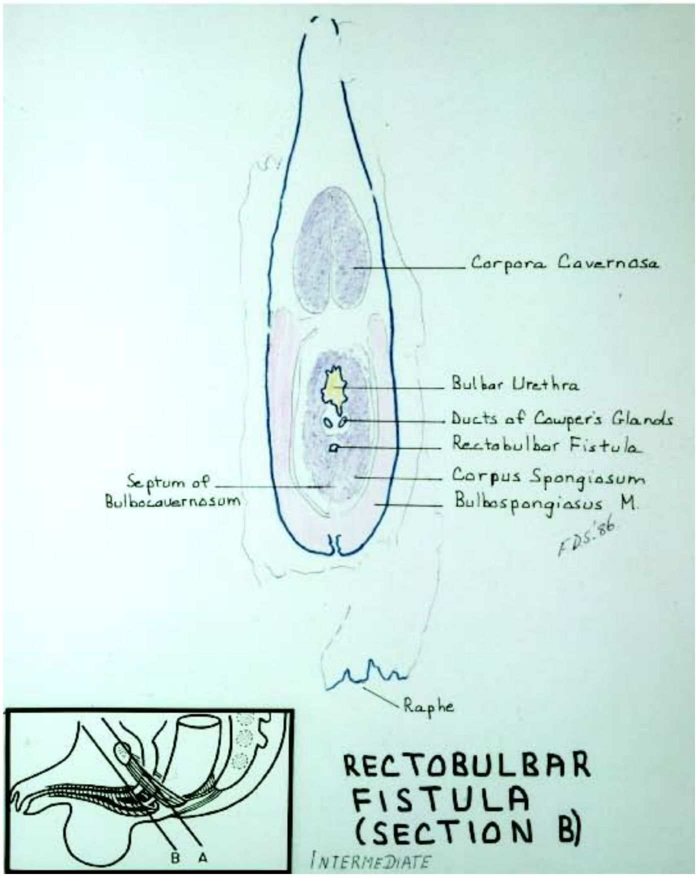
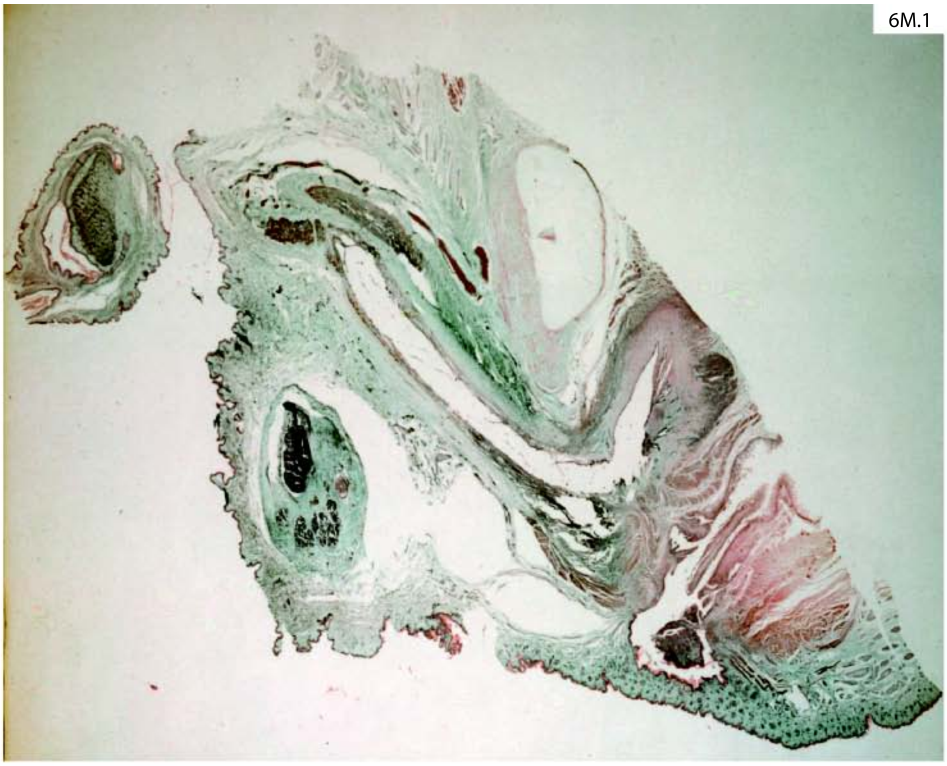


Fig. 6.40 Rectobulbar fistula (approximately section B) (5M.2)



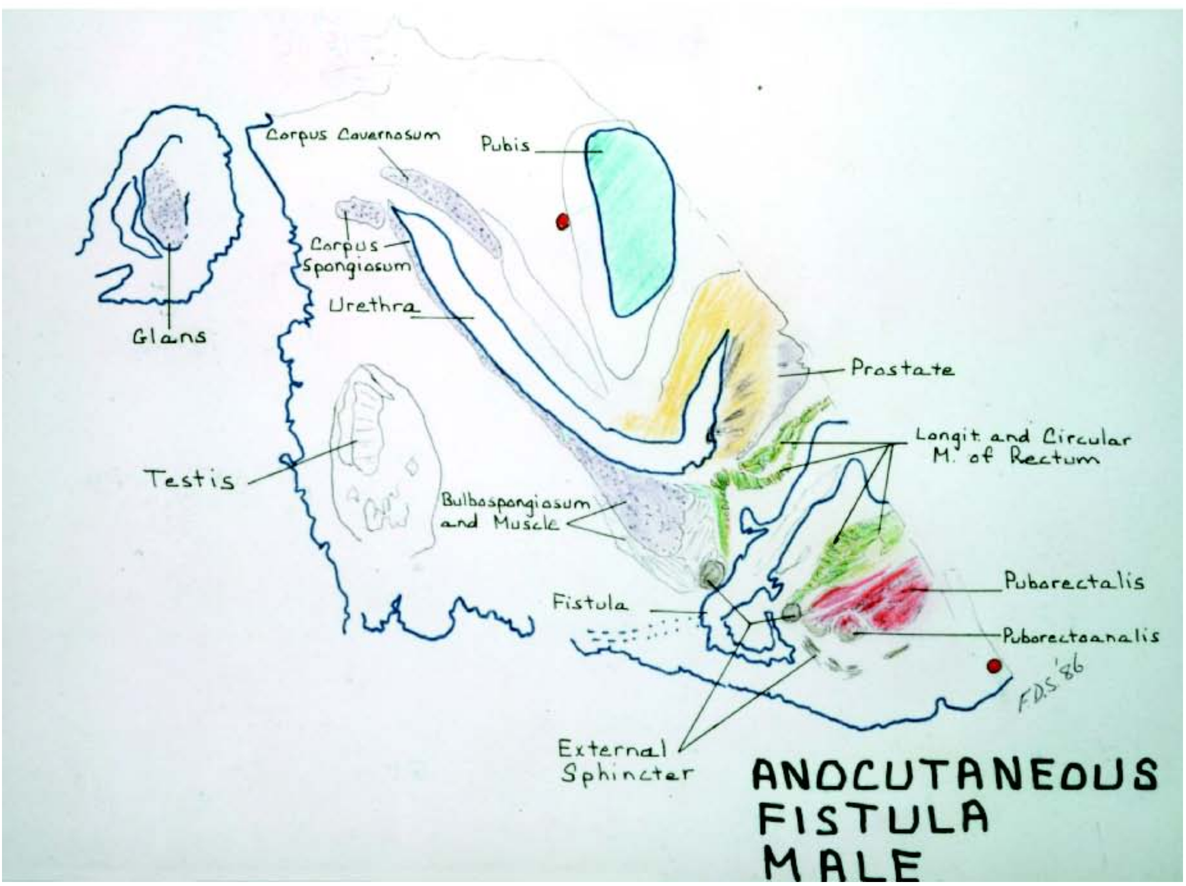
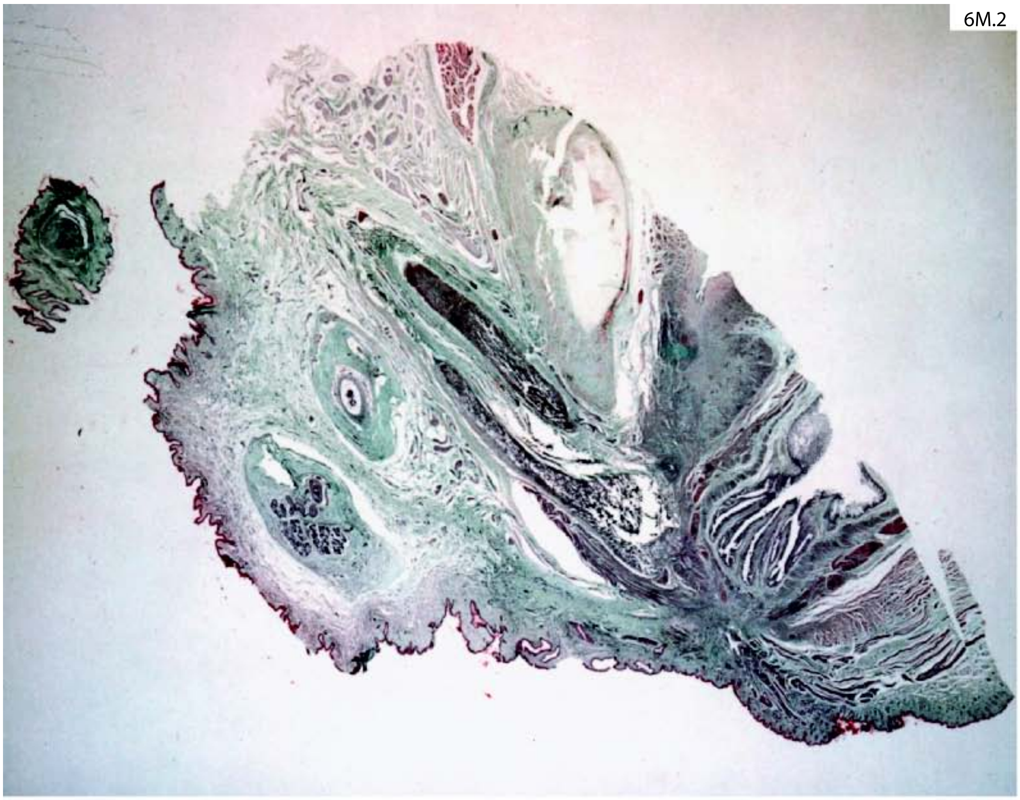


Fig. 6.41 Anocutaneous fistula, male (6M.1)



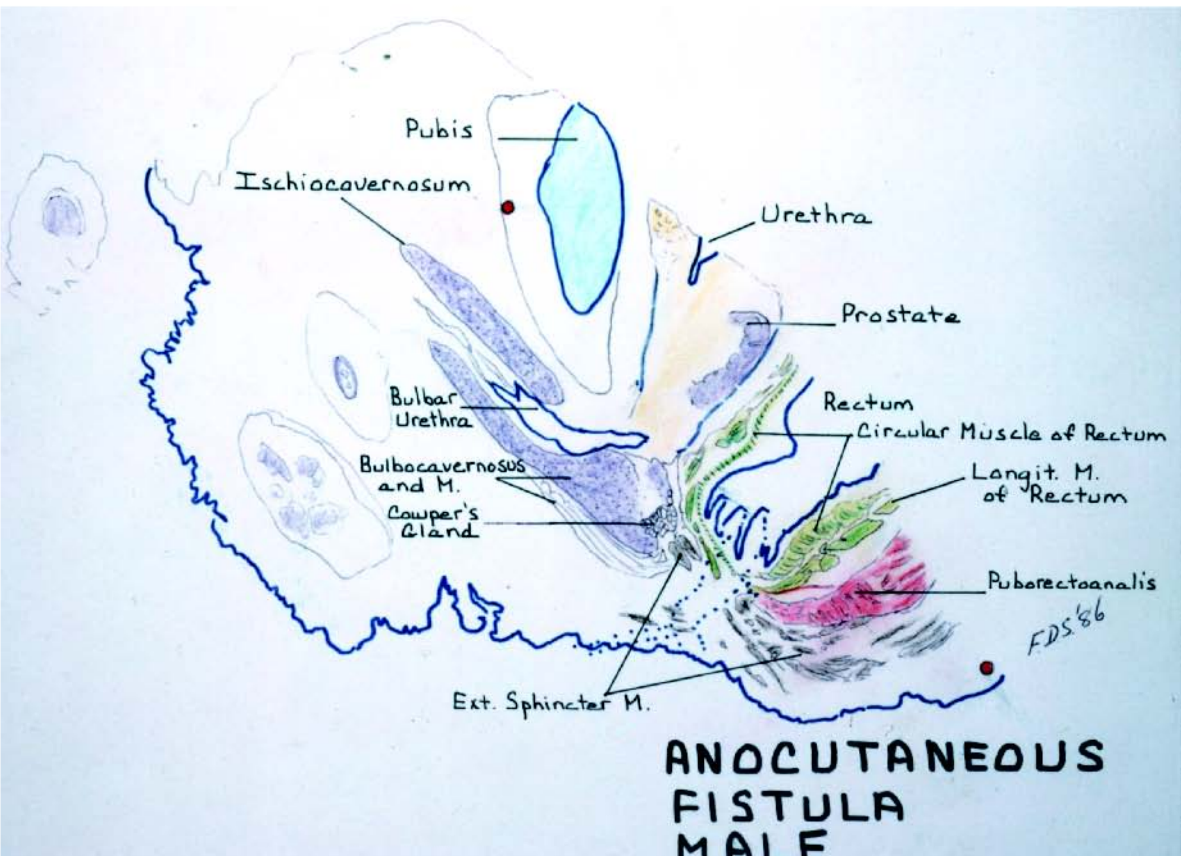


Fig. 6.42 Anocutaneous fistula, male (6M.2)

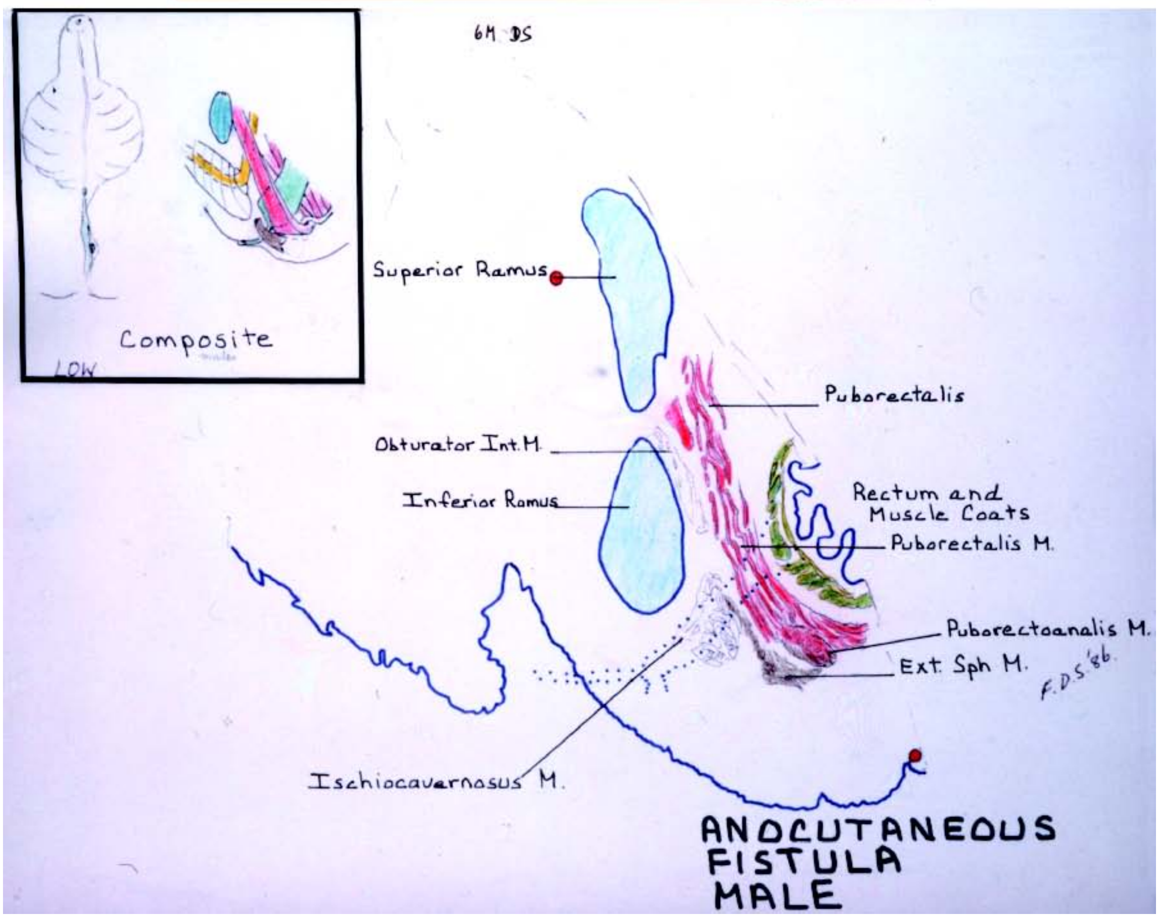


Fig. 6.43 Anocutaneous fistula, male (6M.3)

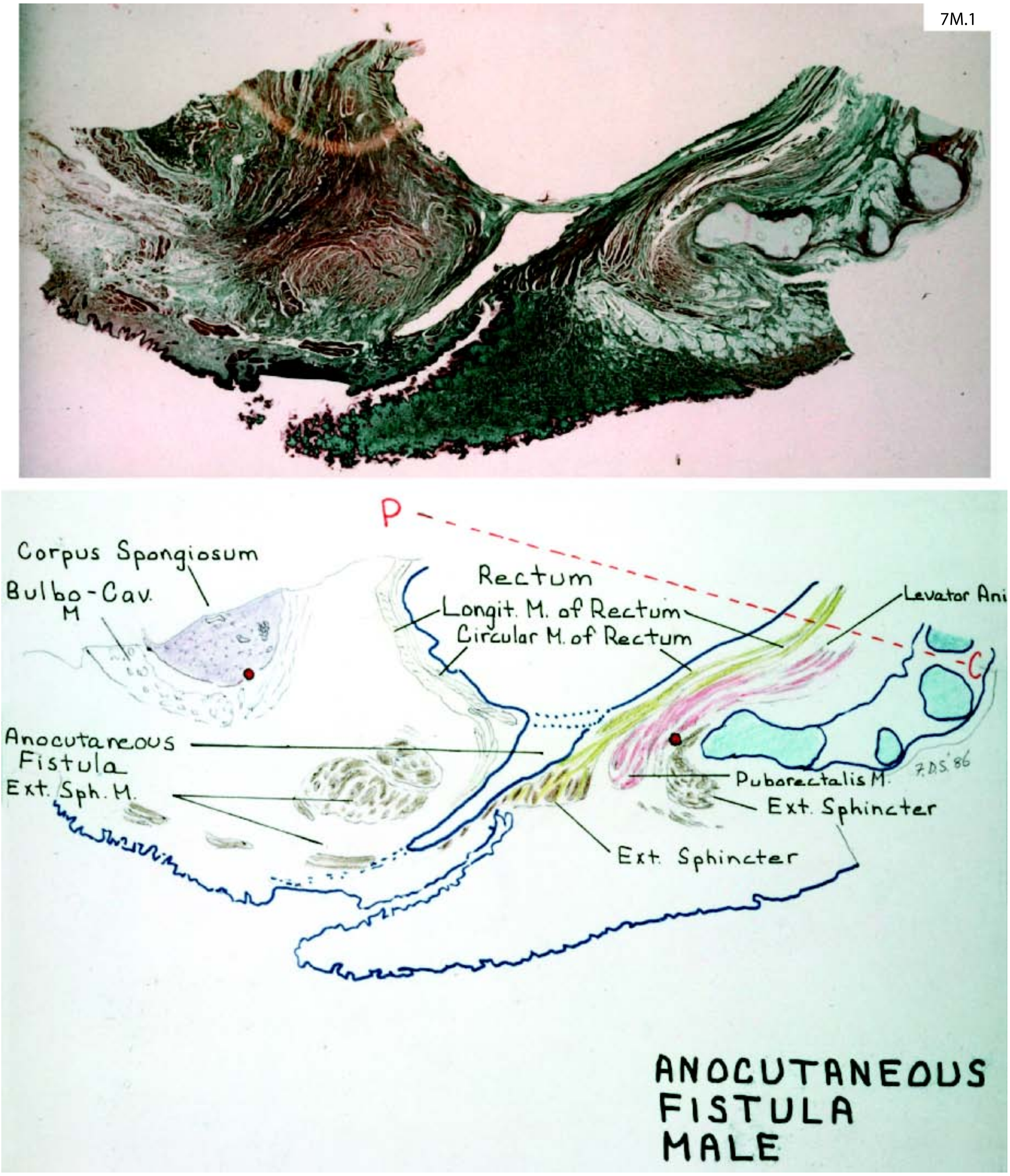


Fig. 6.44 Anocutaneous fistula, male (7M.1)

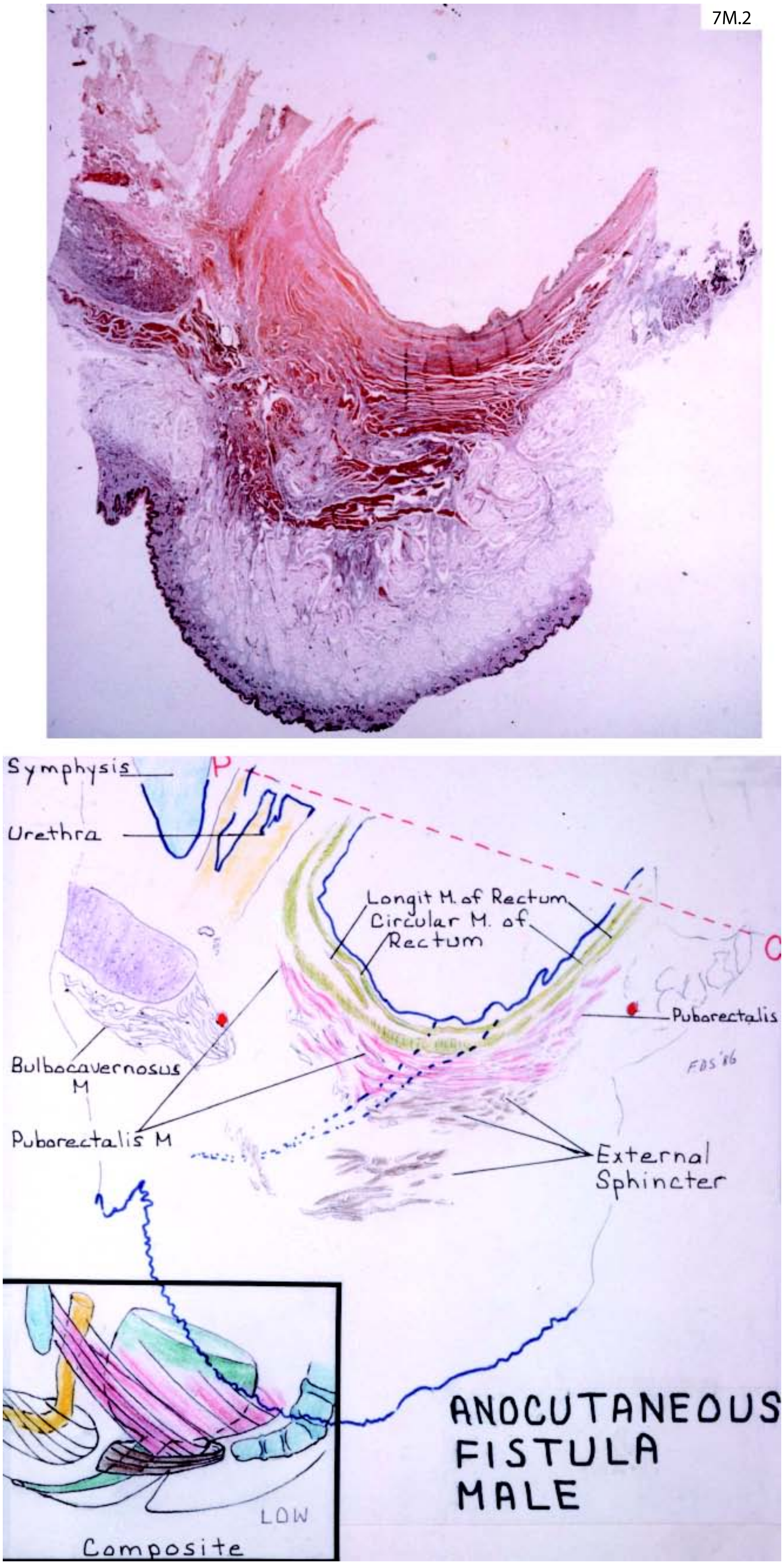


Fig. 6.45 Anocutaneous fistula, male (7M.2)

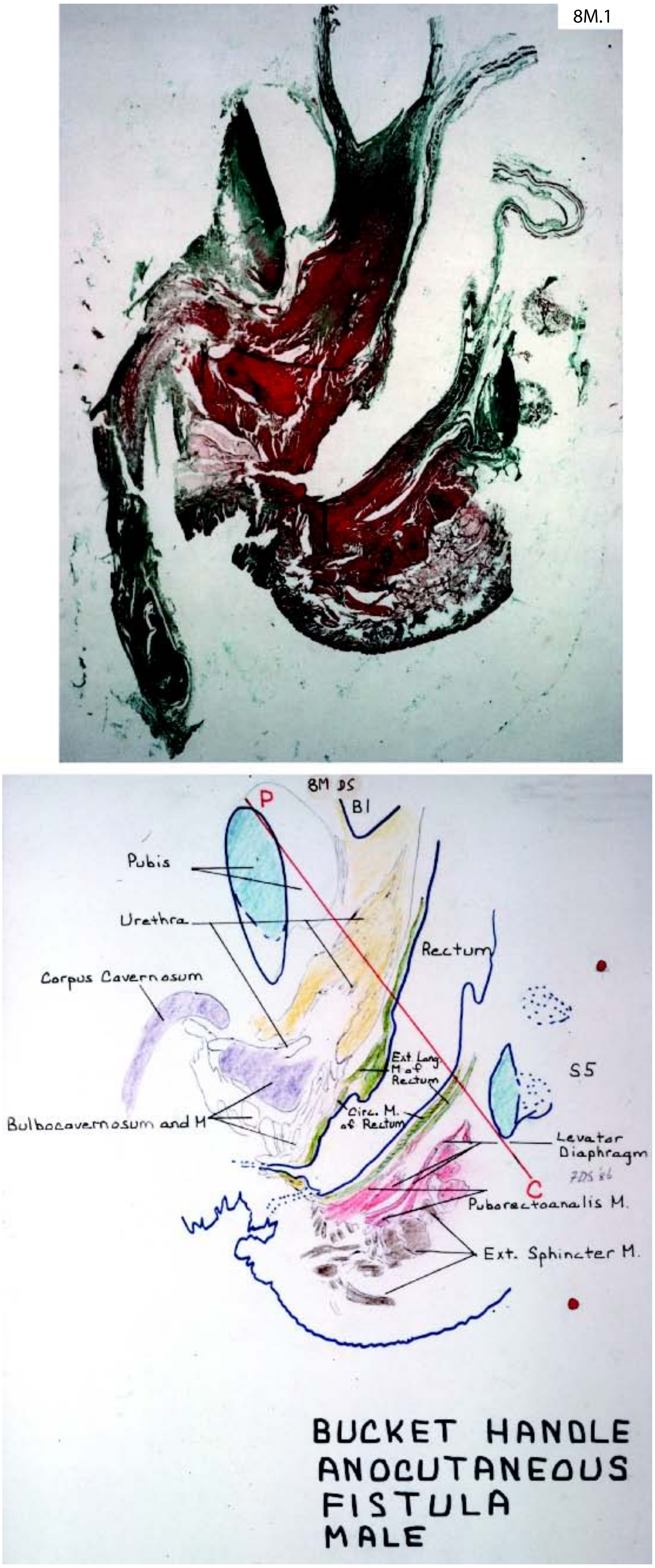


Fig. 6.46 Bucket handle anocutaneous fistula, male (8M.1)

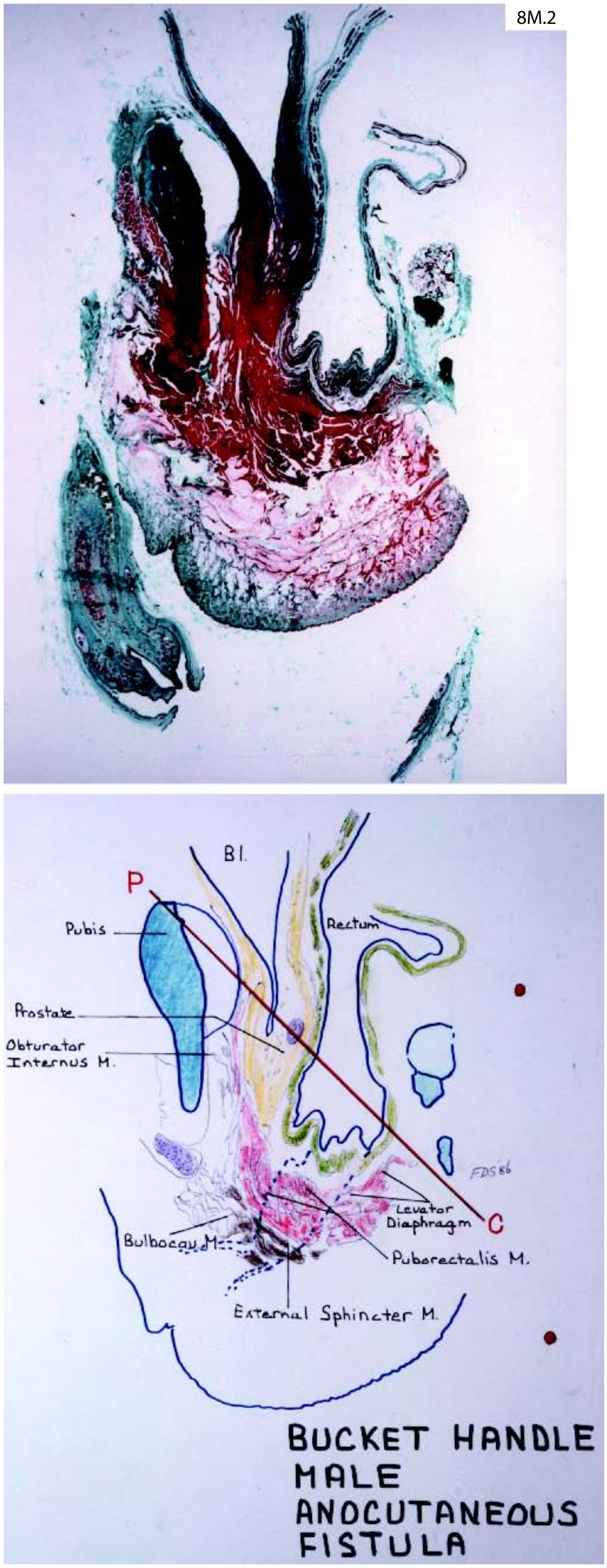


Fig. 6.47 Bucket handle male anocutaneous fistula (8M.2)

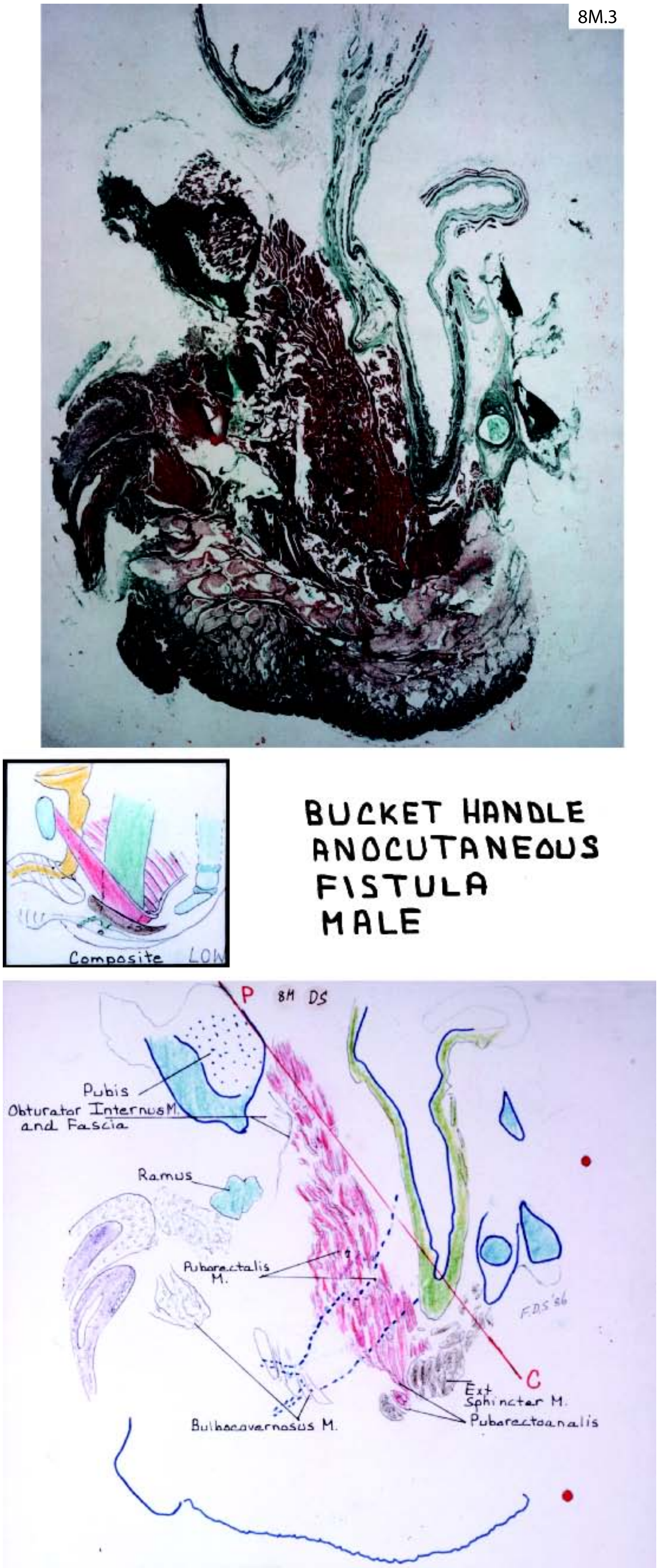


Fig. 6.48 Bucket handle anocutaneous fistula, male (8M.3)

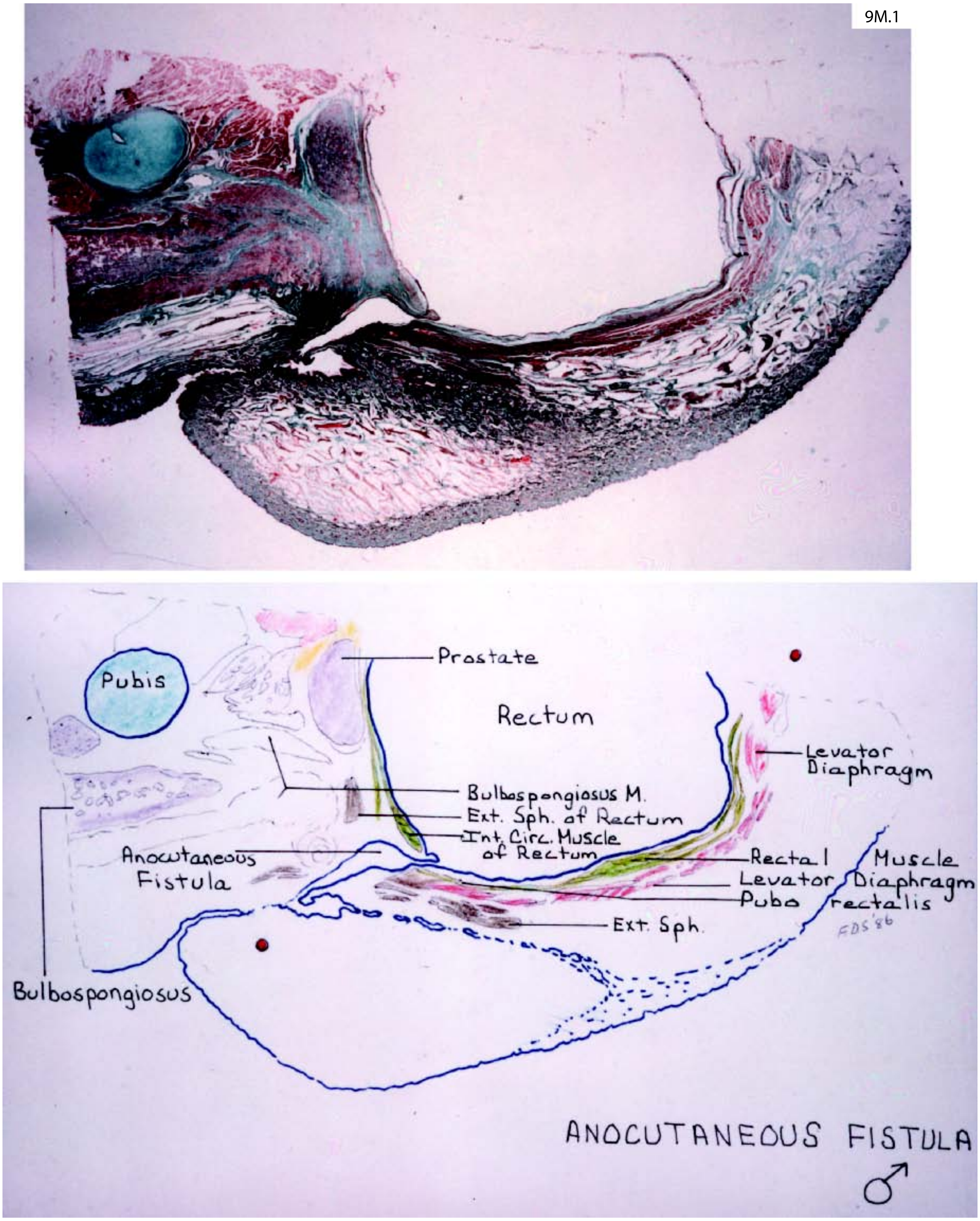


Fig. 6.49 Anocutaneous fistula, male (9M.1)

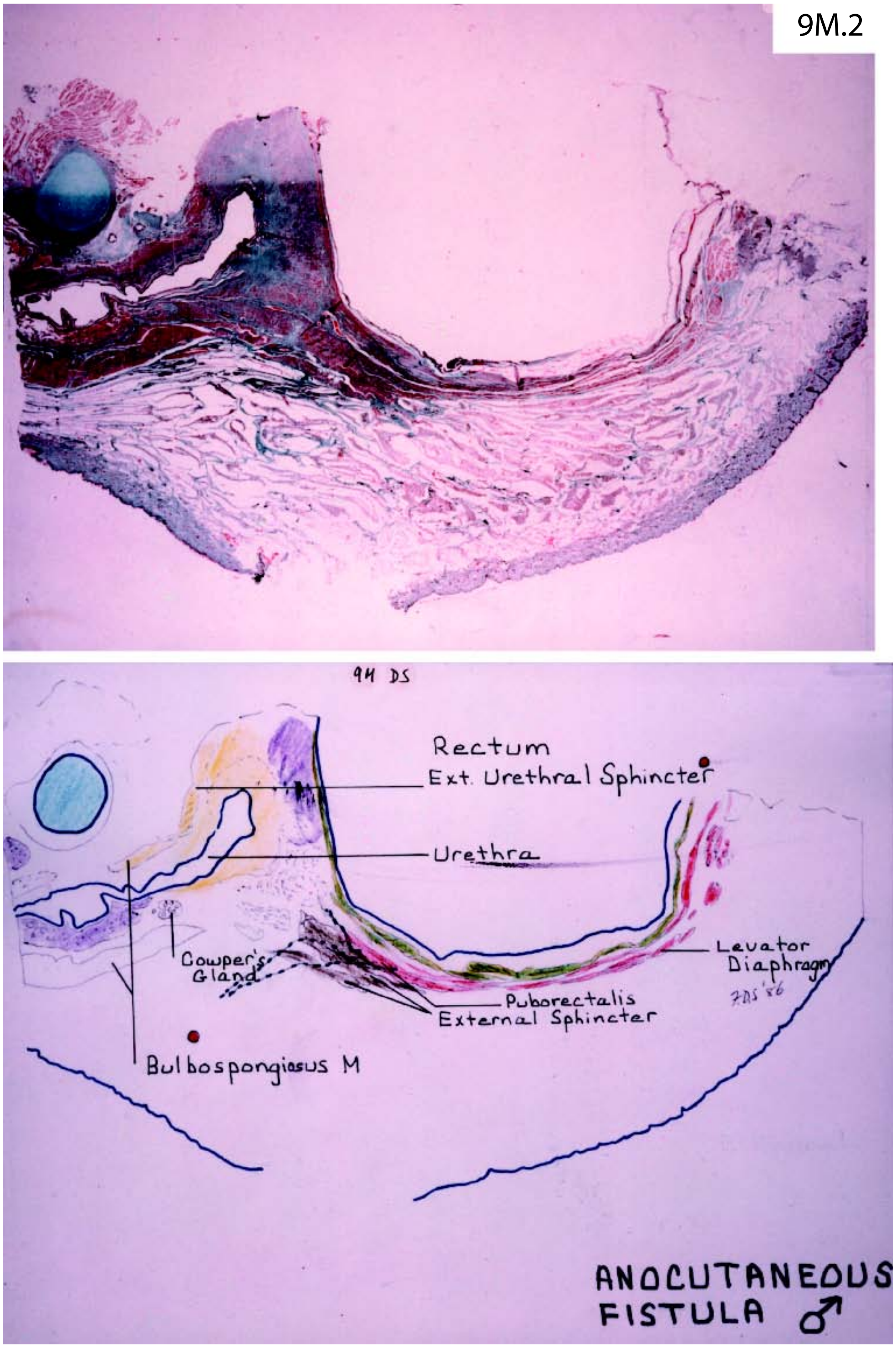


Fig. 6.50 Anocutaneous fistula, male (9M.2)

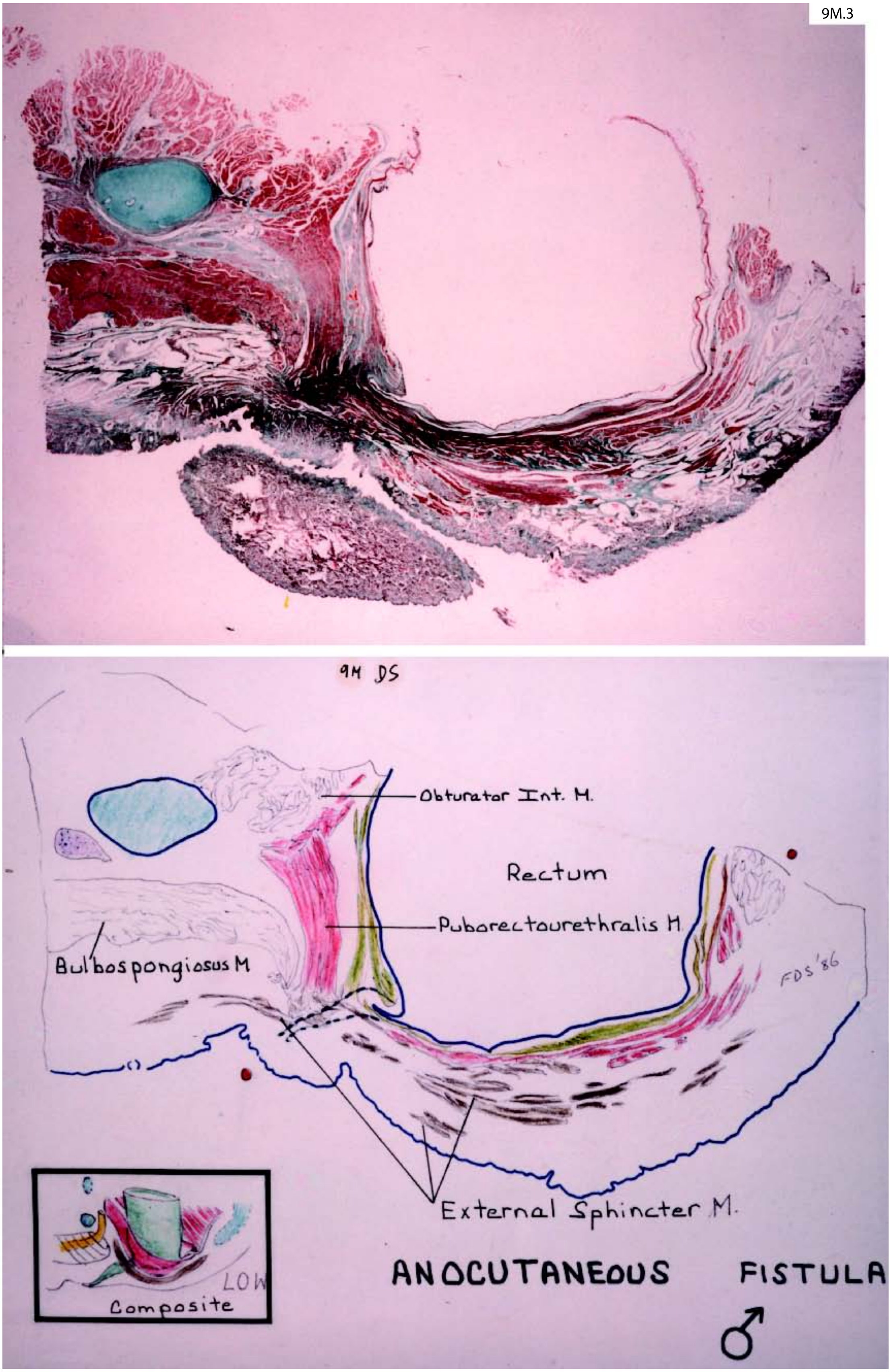
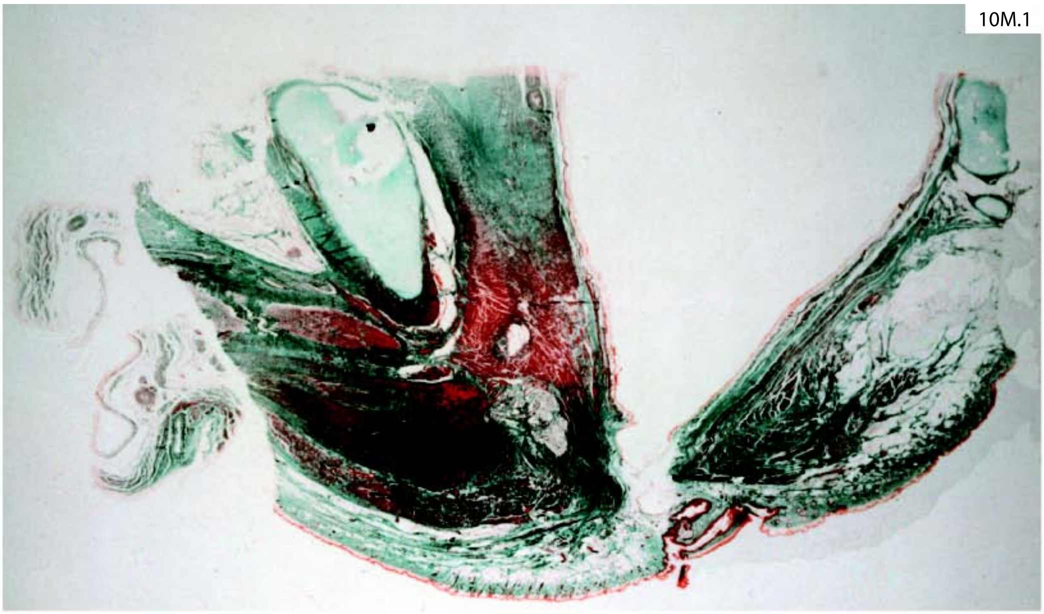


Fig. 6.51 Anocutaneous fistula, male (9M.3)



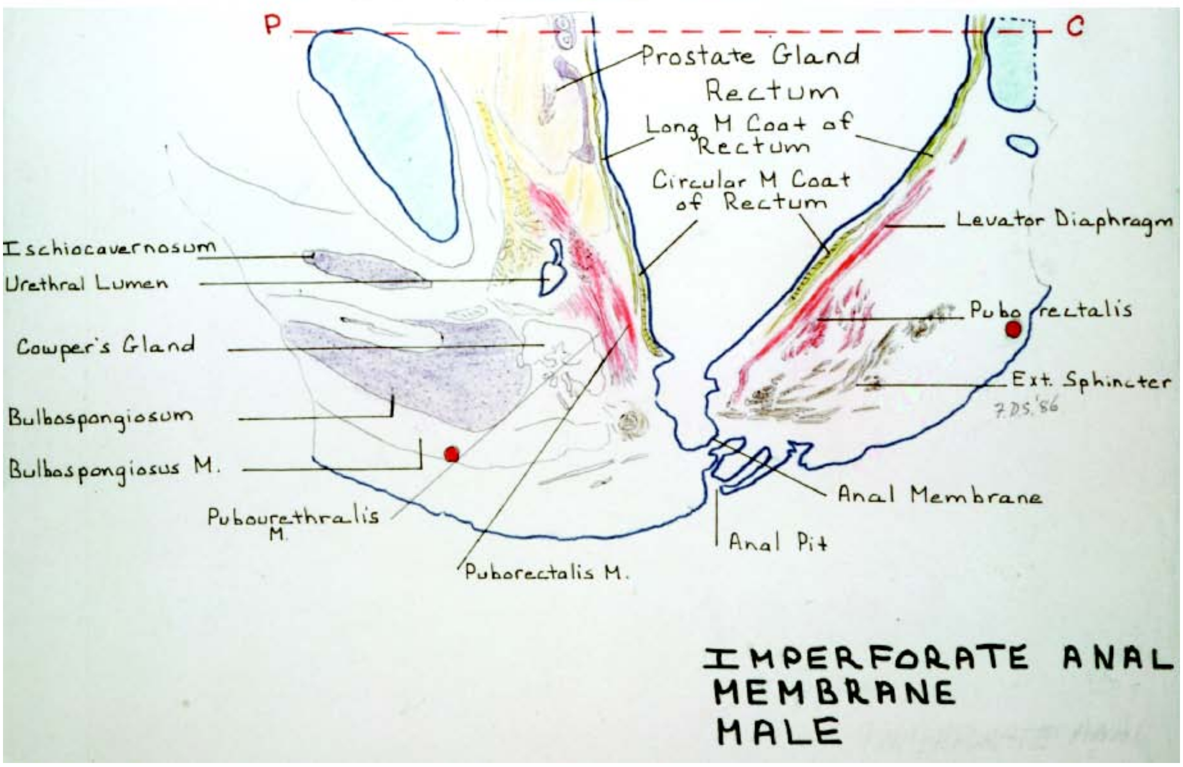


Fig. 6.52 Imperforate anal membrane, male (10M.1)

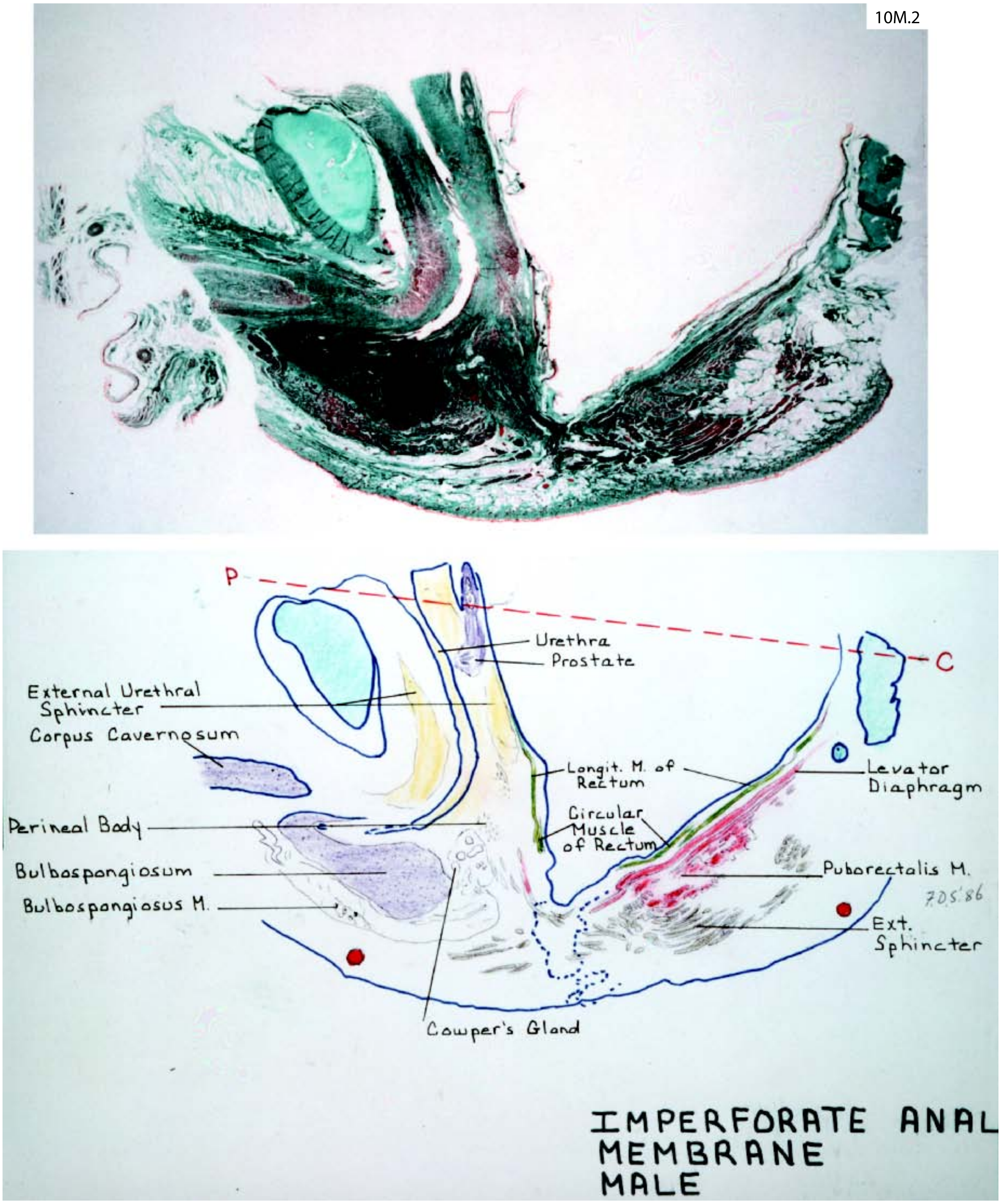
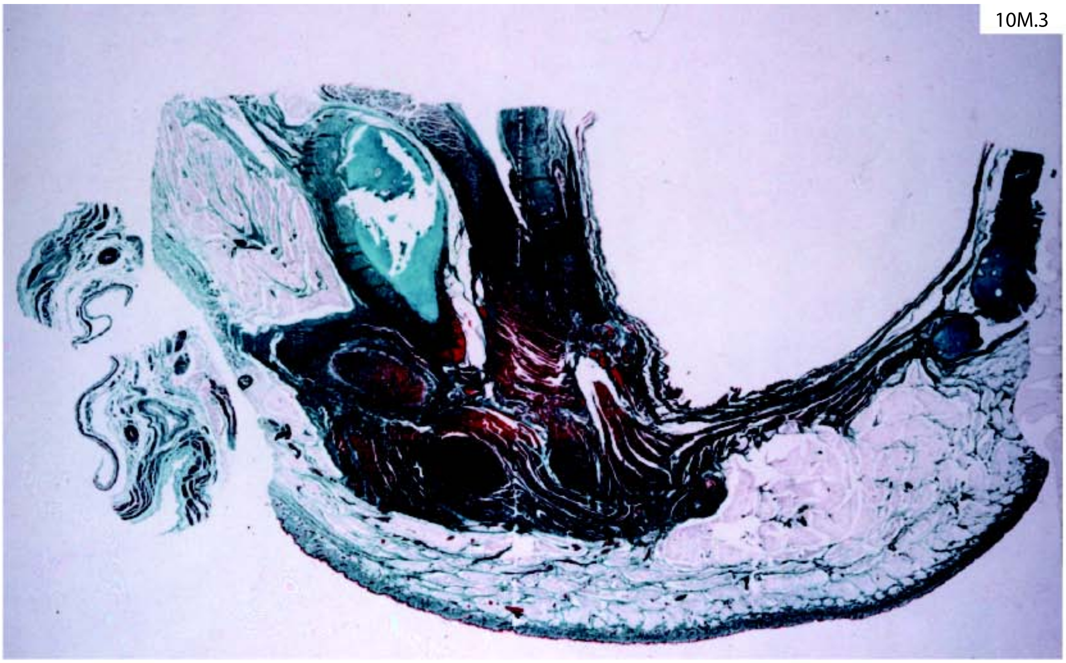


Fig. 6.53 Imperforate anal membrane, male (10M.2)



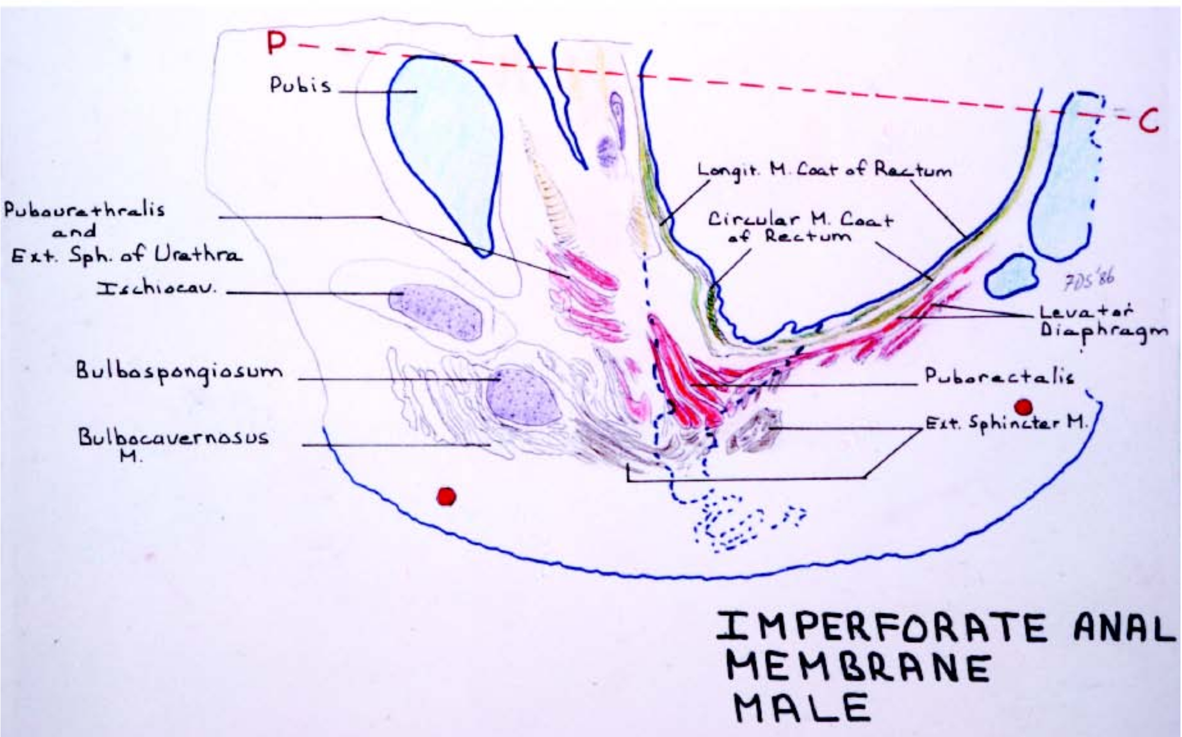


Fig. 6.54 Imperforate anal membrane, male (10M.3)

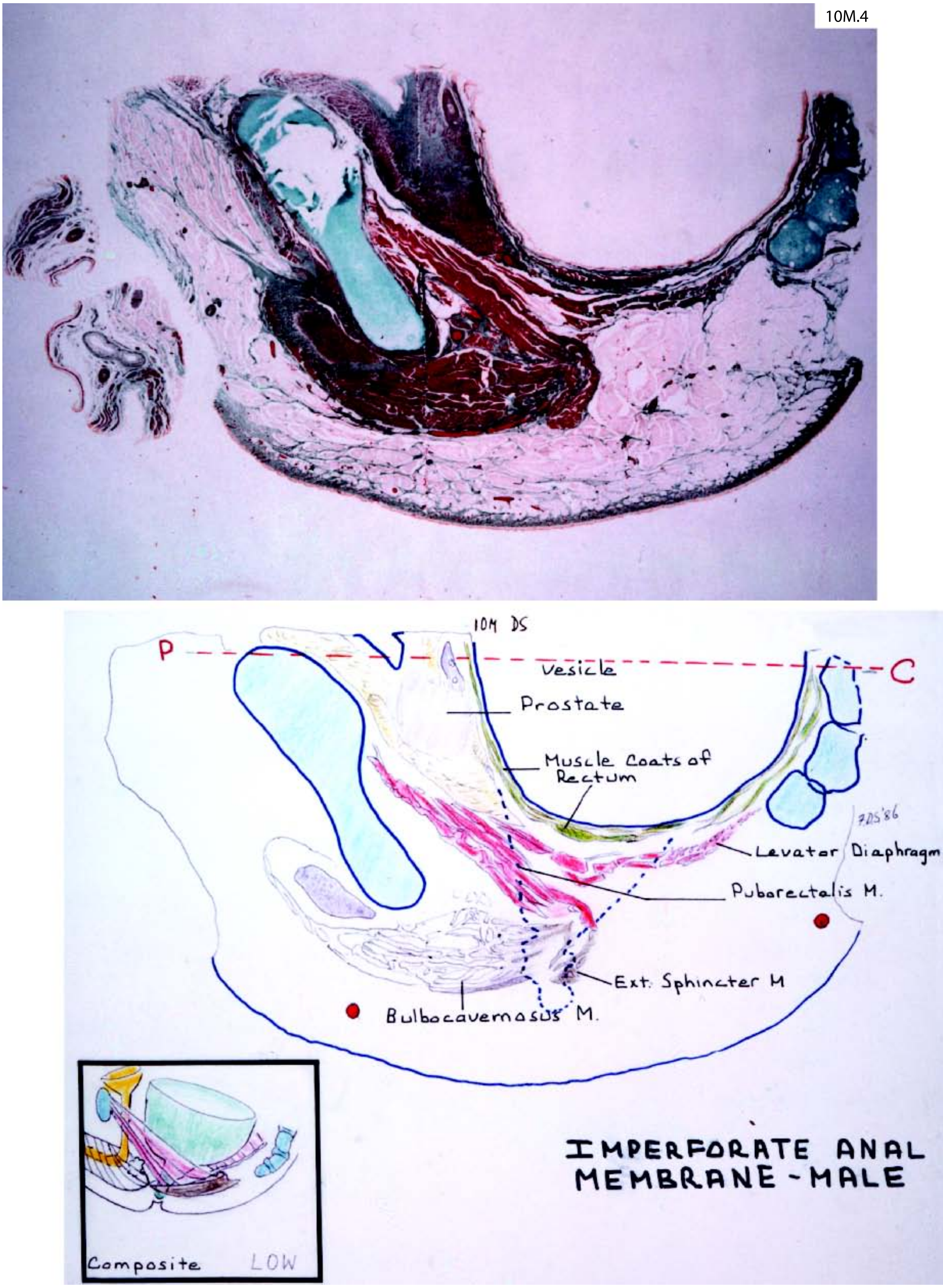


Fig. 6.55 Imperforate anal membrane, male (10M.4)

7 Anatomy and Function of the Normal Rectum and Anus

Alexander M. Holschneider, Helga Fritsch, and Philipp Holschneider

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7.1 Introduction

Sphincteric control, either actual or potential, is the prime consideration in dealing with malformations of the anorectum. The management depends upon the predicted capacity of the sphincter muscles to maintain an adequate measure of cleanliness. It is important to understand not only the function of the muscles governing normal sphincters, but also the potential function of the muscles found in standard visceral malformations.

7.2 Comparative Anatomy

It is believed that the "tail" muscles are adapted in the human to form the pelvic diaphragm and rectal sphincters. Magnus [1] considered that the anococcygeal body [2] of the human is phylogenetically the tail of lower animals and that some of the tail muscles of the lower animals are rearranged around the raphe in humans. In man, these muscles form the pelvic diaphragm and its raphe and are composed of the ilio- and pubococcygeus muscles.

The puborectalis muscle, which is grouped anatomically and embryologically with the levator ani muscle, is not found in lower animals. It appears to be a modification of the external or cloacal sphincter [3]. According to morphological data, the anal canal and the anal sphincter complex are situated beneath the level of the caudal tips of the vertebral column [4].

7.3 The Anal Canal

The anal canal can be defined embryologically as that part of the proctodeum lying between the anal valves (or pectinate line) and the anal orifice [5]. It is surrounded by the anal sphincter complex [6], which is composed of the internal sphincter, longitudinal muscle layer, and external sphincter. The surgeon's definition is based on the "functional anal canal" as determined by digital examination of the subject in the conscious state, extending from the anorectal ring or cranial margin of the puborectalis muscle in the contractile state to the orifice [7]. In normal individuals, the anal orifice is located in the middle of a line drawn between the ischial tuberosities. In mature neonates, the anus will normally admit a 12-Fr (4 mm) dilator.

7.3.1 Epithelial Lining

The epithelium of the anal canal changes abruptly at the pectinate line from the stratified squamous skin of the anus to the stratified columnar mucosa of the rectum. This line also demarcates the level of the deep part of the external sphincter, the lowermost limit of the puborectalis sling, and the junction of the upper one-third with the lower two-thirds of the internal sphincter. It is firmly tethered to the internal sphincter by the submucosa ani.

7.3.1.1 Epithelium Caudal to the Pectinate Line

Adjoining the valves is the pecten, a smooth zone of pink shiny skin that lacks hair and sebaceous glands, and which extends distally to the caudal margin of the internal sphincter. At the orifice and in the surrounding skin, hair follicles and sebaceous glands appear. The skin is puckered by the pull of the coattails of the longitudinal muscle of the bowel wall and, in this puckered perianal area, the skin is brownish in color.

7.3.1.2 Epithelium Cranial to the Valves

Stratified columnar epithelium lines the zone of the anal columns to near the level of the anorectal ring. Aldridge and Campbell, who examined the zone in premature and full-term babies and in children up to age 12 years, have estimated that the length varies in prepared specimens from 0.1 to 1.0 cm [8]. It lacks specialized structures. At the level of the anorectal ring, the epithelium thickens and exhibits crypts, goblet cells, and mucus-secreting glands typical of rectal mucosa.

7.3.2 Sphincter Anatomy

The anal canal is well endowed with involuntary and voluntary muscles, the sphincters, which together with the longitudinal muscle constitute the sphincter complex. The smooth muscle of the internal sphincter is intrinsic to the bowel wall and spans the distal two-thirds of the anal canal. Morphologically there are two components of the external sphincter embracing the distal half of the canal, outside the internal sphincter. The levator ani complex, including the puborectalis, operates in sling-and-sleeve fashion upon the cranial half of the canal.

7.3.2.1 The Internal Sphincter

The internal sphincter is a thickening of the inner circular muscle coat of the bowel wall supplemented and permeated by the dividing coattails of the longitudinal muscle coat. The caudal rim is palpable digitally as a prominent cushion at the mucocutaneous junction of the anal orifice. This cushion is separated from the lowermost fibers of the external sphincter muscle by a palpable circumferential groove called the anal intermuscular groove. At this level the external sphincter turns in and forms a muscular continuum with the internal sphincter and the longitudinal muscle layer [6]. The coattails also penetrate the muscle bundles of the external sphincter, terminating in the perineal body [9] and in the perianal skin [10].

This complex network of coattails knits the mass of the sphincters to the perineum, holds the canal firmly in its grasp, tethers the mucosa and skin above and below the pectinate line to the circular muscle of the internal sphincter, corrugates the perianal skin, and exerts an opening action on the internal and external sphincters during defecation. According to the detailed morphometric studies by Taffaszoli, there is no uniform distribution pattern of ganglia or nerve cells in the internal anal sphincter, but a continuous decrease towards the anus [11].

7.3.2.2 The External Sphincter

This voluntary striated muscle is an irregular collar around the anal canal from the region of the anal valves to the anal orifice, suspended between the perineal body and the anococcygeal body (Fig. 7.1). It is the subcutaneous or superficial portion that is cut into fasciculi and moored to the skin by the coattails of the longitudinal muscle coat of the rectum. Its cranial extremity is contiguous with, and cradles the caudal cuff of the sling of the puborectalis muscle. Above the perineum the external sphincter seems deficient in the midline ventrally [6, 12], thus ensuring a continuity of voluntary muscle throughout the length of the anal canal. Note the differences between male and female sphincters (Fig. 7.2) [6].

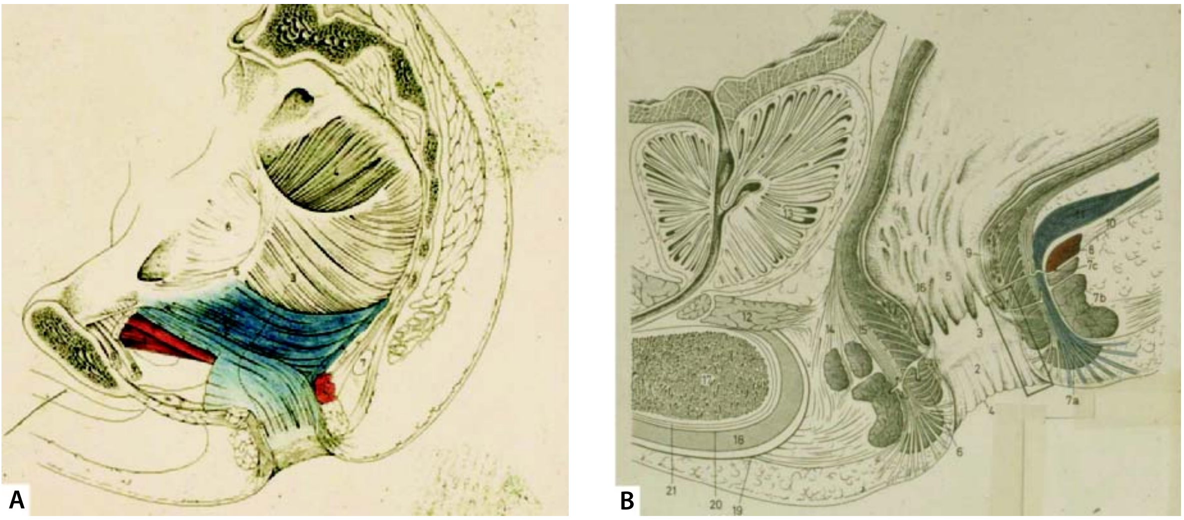


Fig. 7.1 **A** Sagittal view of the pelvis in man, especially levator ani musculature (reproduced from Stelzner [3]). *1* Puborectalis muscle, *2* pubococcygeus, *3* iliococcygeus, *4* coccygeus, *5* linea alba, *6* fascia of the levator muscle above the obturator fascia, *7* ischiorectal groove, *8* pubourethralis muscle, *9* puboperinealis muscle. **B** Sagittal view of the anorectal continence organ in males (reproduced from Stelzner [3] with permission of the publishers). *1* rectum, *2* anal canal, *3* dentate line, *4* anocutaneous line, *5* anorectal line, *6* internal anal sphincter, *7a* external subcutaneous sphincter, *7b* superficial external sphincter, *7c* deep external sphincter, *8* puborectalis muscle, *9* corpus cavernosum of rectum, *10* anococcygeal ligament, *11* levator ani muscle, *12* deep transverse perinei muscle, *13* prostate gland, *14* prerectal muscle, *15* corrugator muscle, *16* muscle of anal canal, *17* corpus cavernosum penis, *18* bulbourethralis muscle, *19* Colles fascia (superficial layer), *20* Colles fascia (deep layer), *21* Buck fascia

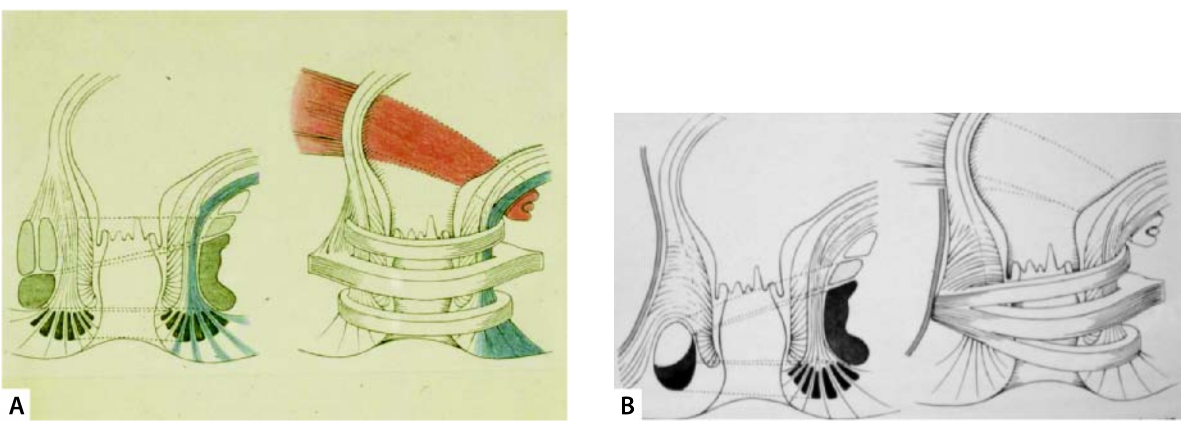


Fig. 7.2 **A** Diagram illustrating the triple loop system of the external anal sphincter in men. The external subcutaneous part has the form of a ring, whereas the superficial part is stronger coccygeally than perineally. The deep external sphincter ani externus profundus muscle, however, is stronger perineally than coccygeally. Nevertheless, the muscle cuff is of the same strength on both sides (reproduced from Stelzner [3]). **B** Diagram illustrating the triple loop system of the external anal sphincter in women. Perineally the muscle cuff is only half as strong as the coccygeal muscle (reproduced from Stelzner [3])

7.3.2.3 The Pelvic Diaphragm

The outlet of the bony pelvis is a wide, diamond-shaped area bounded in front by the inferior pubic and ischial rami and posteriorly by the sacrotuberous and spinous ligaments of the coccyx. The floor of the front half is a triangular ligament, the ligamentous diaphragm, is thick in the male and thin in the female. In the posterior half, the floor is resilient and muscular, the muscular diaphragm.

7.3.2.4 The Muscular Diaphragm

The levatore ani muscles arise directly or indirectly from the inside walls of the true pelvis and converge on the midline to form a bipennate, staggered, muscular hammock posteriorly and a funnel-shaped portal of exit for the anal canal. The muscle spans most of the pelvic outlet, except for two small gaps on the posterolateral aspects, which are filled by thin, fibromuscular structures called the ischiococcygei muscles.

The levator musculature comprises the iliococcygeus, pubococcygeus, and puborectalis subdivisions (Fig. 7.1A), The anlagen of the levator ani muscle can already be subdivided into the three portions during early fetal development [13]. The iliococcygeus muscle arises from the white line of the obturator fascia posterior to the obturator nerve and unites with the muscle of the opposite side and with the sides of the coccyx to form the caudal lamina of the posterior half of the pelvic diaphragm. The pubococcygeus has a linear attachment to the back of the body of the pubis and the anterior part of the white line as far back as the obturator canal. The fibers take a posteromedial and medial course to attach to the coccyx and the muscle of the opposite side to form a lamina of the pelvic diaphragm, which is more extensive than, and cranial to, that of the iliococcygeus. This muscle, depending on its tone, appears to be funnel-shaped. The pubococcygeus and iliococcygeus elevate, straighten, steady, and suspend the rectum.

The puborectalis muscle is the third component that originates from the myotomes S1–S4. It is a sling-like ribbon of muscle that is firmly anchored anteriorly to the inferior ramus of the pubic bone at both sides. The sling is set on an inclined plane from the pubis to the back of the rectal wall. It is approximately 1–2 cm deep, is attached to the rectum several millimeters above the valves, and hugs the back and sides of the terminal rectum. It is delicately adherent to the iliococcygeus. The caudal edge of the puborectalis sling is cradled posteriorly by the upper extremity of the deep external voluntary sphincter at approximately the level of the pectinate line. It has been shown that already in fetal stages the puborectalis and external anal sphincter cannot clearly be separated [13]. By its action, the puborectalis apposes the back and side walls of the rectum against the anterior wall and jams the rectum against the fixed structures of the triangular ligament; the anal canal is thereby tilted anteriorly, shut, and elevated, and the rectum is angulated between the anal canal and the ampulla. Generally, it can be said that if the myotomes of S1/S2 are missing, there is no puborectalis muscle and continence is poor. If the S3 vertebra is missing, the puborectalis sling is very thin and continence is doubtful. If S4 is not developed, the puborectal sling is weakened, although continence is favorable; only if S5 exists will continence be good [14].

That part of the anal canal cranial to the pectinate line is intimately wrapped in the pubococcygeus and puborectalis muscles – the sleeve-and-sling complex, whereas the part distal is clothed by the encircling internal and external sphincters (Fig. 7.1B). The narrow zone of the pectinate line is ringed by the deep part of the external sphincter muscle. Wilson [15] considered that it is more correct and more logical to call the puborectalis muscle the puborectoanalis muscle because of its intimate relationship to that part of the rectum that forms part of the anal canal. That is the reason why in anorectal malformations (ARM) we see only a so-called muscle complex instead of different pelvic floor and sphincter muscles.

7.3.2.5 The Suspending Mechanism

The rectum is mainly held in place by muscles that counterbalance the abdominal pressures exerted on it as by coughing or by the erect posture. Wilson [15] believed that there is a direct suspender effect of the fascia of Waldeyer where its favial "claws" gain attachment to the rectum. Recent studies show that there is a plane of cleavage between the striated muscles of the pelvic diaphragm and the muscular coat of the rectum laterally [16]. Indirectly, the parietal fascia and its septal components serve to stabilize the anorectum. The pubococcygeus, in which are found numerous membranous fibers, and the iliococcygeus are the chief pelvic muscular suspenders.

The external sphincter is attached directly or indirectly to the perineal body and to the coccyx. Other so-called ligaments are condensations of the pelvic connective tissue and indirectly serve to take the strain and create a firm anchorage. Stelzner [3] and

El Shafik [17] suggest a triple loop concept of the attachment and functioning of the external sphincters of the anal canal (Fig. 7.2). However in radiologic defecography it is difficult to demonstrate this triple loop concept (Fig. 7.3).

We consider, however, that the fascial suspension, although important, is not the essential mechanism and that the perineal membrane, from which the perineal body gains fixation, and the muscular diaphragm are the chief supporters of the viscera. In addition, the rectogenital septum, which is found in both males and females, may play an important role in stabilizing the anorectum during defecation [9, 18].

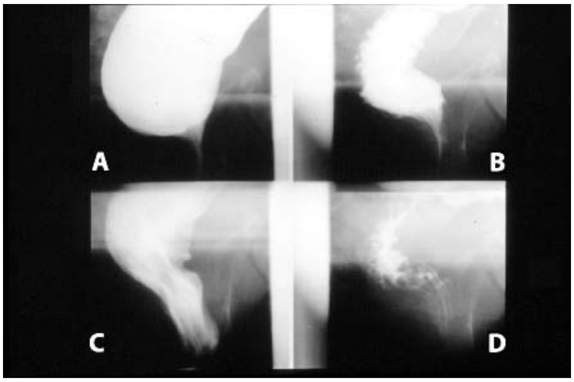


Fig. 7.3 Defecography in a healthy child in sagittal position. **A** Normal anorectal angle formed by the puborectalis sling and the deep part of the external anal sphincter; **B** internal sphincter relaxation starting with opening of the proximal one-third of the anal canal. The middle and superficial parts of the external anal sphincter are still closed; **C** complete opening of the internal anal sphincter with simultaneous reflex inhibition of puborectalis/levator ani and external sphincter muscles leading to defecation; **D** almost complete emptying of the rectum after defecation and restoring of the anorectal angle (reproduced from Holschneider and Puri [23])

7.4 The Striated Muscle Complex

Electrostimulation of the perineum and study of the striated muscle bundles using the midsagittal approach [19] revealed the presence of a "striated muscle complex," which represents the external sphincter in the cases of low, intermediate, and high ARM. Recent studies [20, 21] show that it is possible to define accurately the normal pelvic musculature, and also that of patients with ARM, using computed tomography scans (Fig. 7.4).

7.5 The Nerve Supply of the Normal Rectum and Sphincters

The second, third, and fourth sacral segments of the spinal cord are the nerve centers of the arcs that subserve the receptors and effectors of the rectum, anus, bladder, and urethra, and, together with higher centers in the brain, are responsible for continence. These centers in the spinal cord also subserve cutaneous sensation in the anal canal to the level of the valves and in the perianal region. The sympathetic supply, however, arises in the second, third, and fourth lumbar segments. Malformations of the spinal cord pertaining to the sacral segments involve all systems, but damage of nerves within the pelvis or perineum may have more localized effects.

7.5.1 Parasympathetic Nerves

The parasympathetic nerves to the bowel arise on either side of the pelvis from the anterior divisions of the third and fourth sacral nerves, with twigs sometimes from the second. These preganglionic nerve fibers usually join to form two nervi erigentes, which give short branches directly to the rectum at the level of the ischial spine (Fig. 7.5 A–C) and continue as longer trunks to the inferior hypogastric or pelvic plexus, where they are redistributed to pelvic organs, directly or via blood vessels. In the wall of the rectum, these fibers relay in the ganglia of Auerbach's plexus. Other small parasympathetic nerves from the anterior divisions of the third and fourth sacral nerves join and ascend in the presacral sympathetic nerve and then follow the ramifications of the inferior mesenteric artery.

These delicate, tenuous nervi erigentes run lateral to the rectum, directly attached to the rectal fascia [16] close to the ischial spine or, in the newborn baby, at the level of the pubococcygeal (PC) line [56]. The main trunks can be separated safely from the rectum because a natural plane of cleavage can be found between the perirectal connective tissue, rectal fascia, and nerves. Hence, bladder and urethral function is spared in excision of the rectum for nonmalignant conditions.

The nervi erigentes in rectal deformities are separated throughout their course by the rectum if it descends to the level of the PC line (see chapter 25).

When the rectum is located higher in the pelvis than the PC line, these nerves run a more medial course with the perirectal connective tissue (perirectal

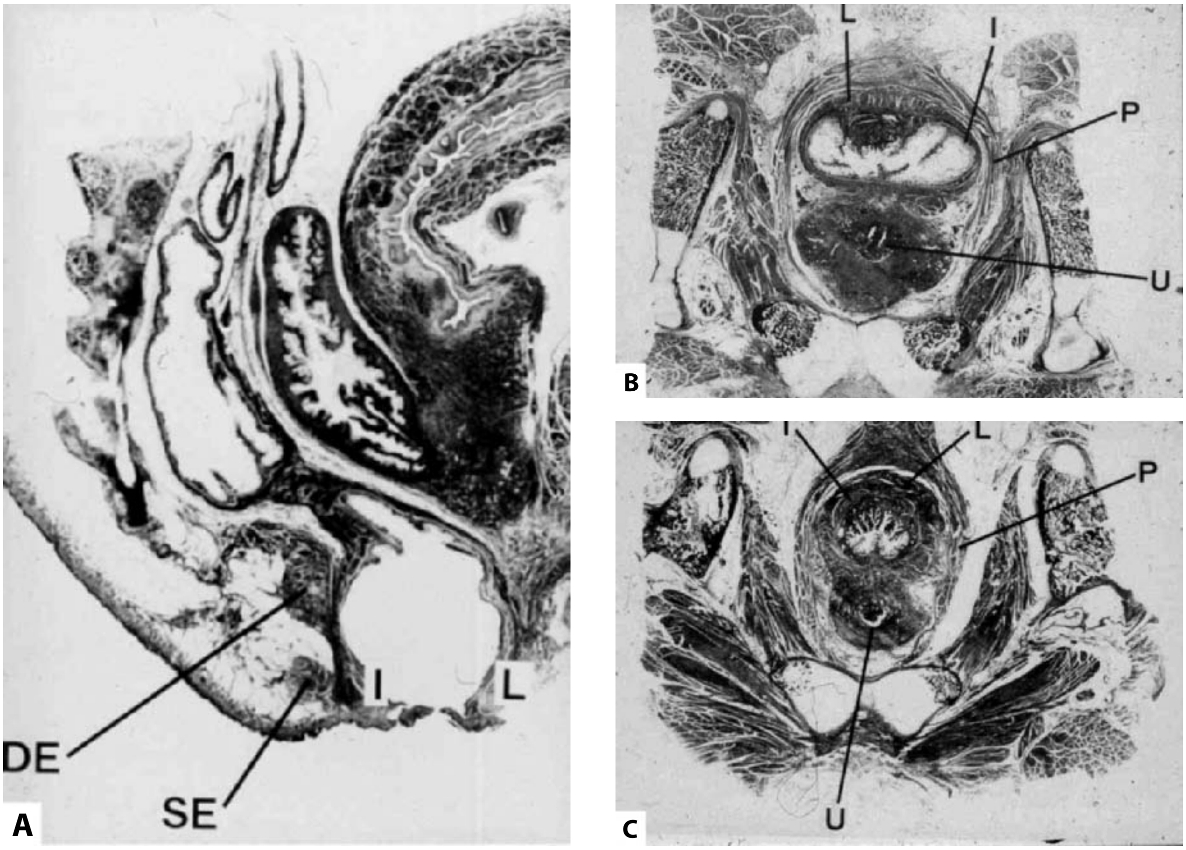


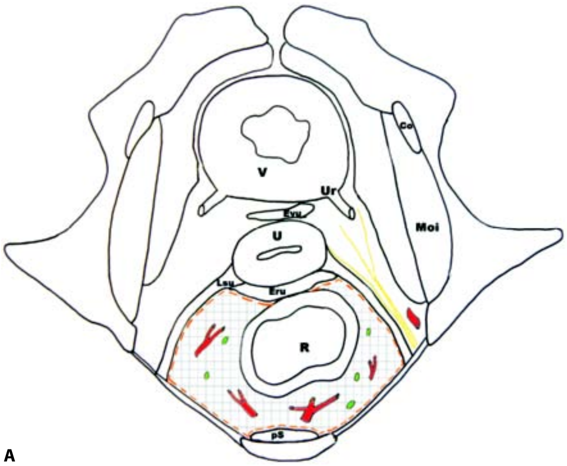
Fig. 7.4 **A** Sagittal section of a normal pelvis at the level of the pubic arch (*P*). The longitudinal muscle (*L*) is thickened and blended with the external anal sphincter. *DE* Deep external anal sphincter, *SE* superficial anal sphincter, *I* internal anal sphincter. **B** Transverse section of a normal male pelvis at the level of the pubic arch. Inner circular muscle (*I*) and longitudinal muscle (*L*) are thickened at this level. *P* Puborectalis muscle, *U* urethra. **C** Rectourethral fistula in a boy with a high anorectal malformation. At this level, 2 cm above the connection with a fistula, the thickening of the inner circular muscle can be seen. The puborectalis muscle is just adjacent to the rectal wall. **A**–**C** reproduced from Yokoyama et al. [21] with the permission of the publisher

fascia) beneath the blind ending rectum to reach the region of the bladder base and neck. In this situation, they are more vulnerable, especially if mobilization of the rectum is attempted from the sacrococcygeal approach [55]. Furthermore, in some patients the nervi erigentes and nerves to levator ani have a common stem or origin before dividing and diverging in their different fascial investments, in which event, if the common trunk is damaged, the function of both the bladder and the levator ani would be affected [3].

7.5.2 Sympathetic Nerves

The sympathetic nerves arise in the second, third, and fourth lumbar ganglia and the preaortic plexus. They unite on either side and form the hypogastric plexus in front of the fifth lumbar vertebra and then continue down the posterolateral pelvic walls as the presacral nerves, which join the pelvic ganglion on either side of the pelvis. Several fine sympathetic nerves from the second and third ganglia of the sacral sympathetic chain also join the pelvic ganglion in close company with the parasympathetic nervi erigentes.

The pelvic ganglion is a flat pannus that lies closely applied to the base of the bladder and prostate, the



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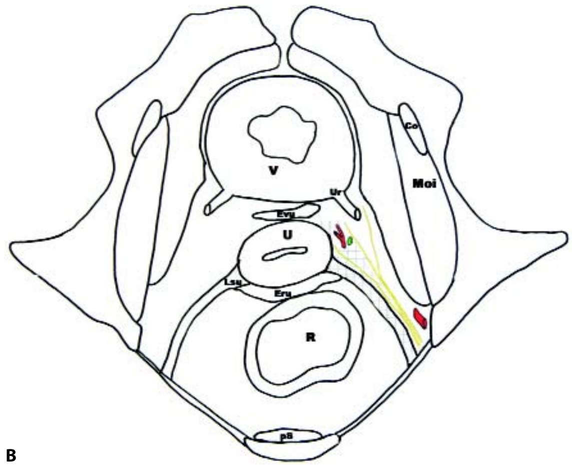

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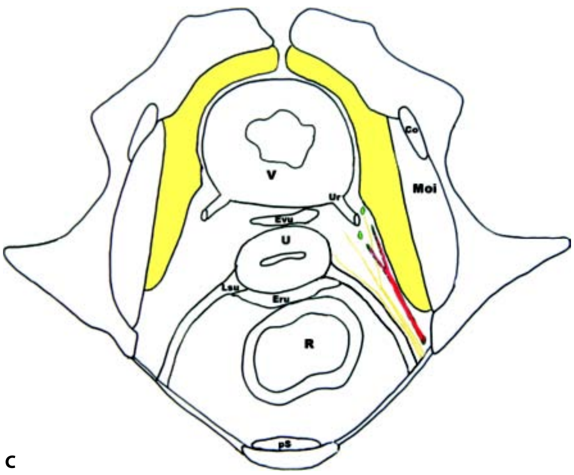

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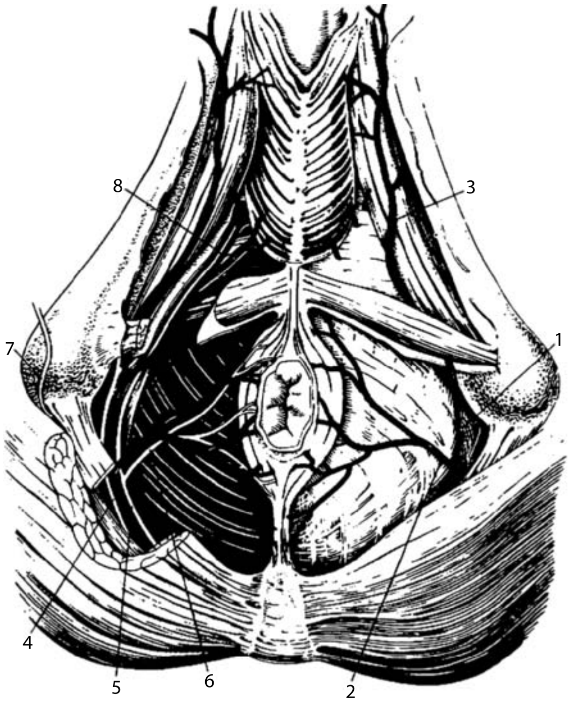
Fig. 7.5 **A**–**C** Schematic view of the compartments of the female pelvis. **A** Dorsal compartment with *hatched* perirectal subcompartment [57]; **B** ventral compartment with marked (*yellow/hatched*) paravisceral fat body [57]; **C** middle compartment with *hatched* paracervical, adventitial connective tissue and sacrouterine ligament [57]. *V* Bladder, *U* uterus, *R* rectum, *PS* os coccyx, *Co* canalis obturatorius, *Moi* Musculus obturatorius internus, *Lsu* sacrouterine ligament (see also chapter 25, Fig. 25.6)

region of the uterine cervix, and the adjoining anterolateral wall of the rectum. The ureter passes through it to get to the bladder. The ganglion has two posterior dog ears, one in the line of the presacral nerves and one adjacent to the third and fourth sacral segments, reaching backwards toward the contributions from the nervi erigentes. The pelvic ganglion is composed of multiple convoluted nerves and large clusters of ganglion cells packed into the tessellated pannus. It lies in the parietal layer of pelvic fascia and can be separated from the rectum, which can be freed and resected without interference with function of the urinary or genital tracts (see Fig. 7.5A–C).

The sympathetic and parasympathetic nerves to the rectum and anal canal are responsible through the ganglion plexuses of Auerbach and Meissner for organized peristalsis and tone in the internal sphincter. The sympathetic fibers are said to be inhibitors of the bowel wall and motor to the involuntary internal sphincter, whereas the parasympathetic nerves are motor to the bowel and inhibitors of the sphincters [22, 23]. The parasympathetic nerves carry, in addition, sensory fibers conveying knowledge of distention of the rectum [24], which are supposed to be located at the ventral rectal wall [9].



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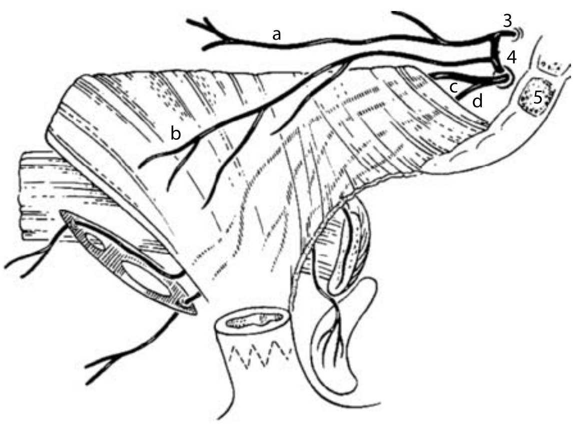

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Fig. 7.6 **A** Pudendal nerves and arteries and perineal branches of S4 (reproduced from Stelzner [3] with the permission of the publishers). Course of the pudendal nerve with radial branches to the pubococcygeus and puborectalis muscles. Perineal branch of S4 to the puborectalis and external sphincter muscles. Note that the midline zone around and in front of the coccyx is free from nerves and safe for dissection. *1* Pudendal artery, *2* anal branch, *3* perineal branch, *4* perineal nerve, *5* dorsal nerve of the penis (*4–6* branches of the pudendal nerve). **B** Nervi erigentes and nerves to the levator ani (reproduced from Stephens and Smith [55] with the permission of the publishers). Right half of the pelvis from within. *a* Right nervi erigentes arising from the roots of S3 and S4, *b* branch of S3 and S4 to cranial aspect of levator ani, *c* pudendal nerve giving branch to the caudal aspect of levator ani and to the external anal sphincter, *d* perineal branch of S4 to puborectalis and external sphincter

7.5.3 Nerves to the Levator Ani Muscles and the External Sphincter

Branches from the anterior roots of the third and fourth sacral nerves unite to form the main nerve pathway to the ilio- and pubococcygeus muscles. The trunk runs a lateral course on the cranial or pelvic surface of the levator ani muscle, not far from and parallel to the white line of origin. Its branches run obliquely, anteriorly, and medially on these muscles. This nerve may be single with peripheral oblique branchings, a single stem with two main branches, or may be represented by two separate nerves running parallel to each other, arising independently from the nerve roots of third and fourth sacral nerves.

The pudendal nerve, which arises from the anterior divisions of the second, third, and fourth sacral nerves, clings to the lateral wall of the pelvis in the pudendal, or Alcock's canal. It supplies branches to both the ilio- and pubococcygeus muscles and to the puborectalis [25] through its inferior hemorrhoidal and perineal branches, which cross the ischioanal space to enter the muscles (Fig. 7.6).

The perineal branch of the fourth sacral nerve, a nerve that must be distinguished from the perineal branches of the pudendal nerve, enters the ischiorectal fossa medial to the ischial spine on the caudal and lateral aspect of the coccygeus muscle, and its branches are directed medially to the posterior fibers of the puborectalis sling and external sphincter [15]. This nerve is at surgical risk only when deep lateral cuts are directed from the vicinity of the coccyx and anococcygeal body.

The coccyx and the distal sacral vertebrae are absent in many patients exhibiting ARM, and the coccygeal nerves and corresponding sacral nerves in some such patients are also defective. Generally, it can be observed that bilateral loss of all sacral nerve fibers S2–S4 leads to complete incontinence. There are no longer anorectal reflex mechanisms or sensitivity.

If only the sacral nerve supply of S1 and S2 is developed bilaterally, the feeling of fullness and the ability to discriminate solid, liquid, or gaseous stools is disturbed, as well as the rectosphincteric reflex mechanism to the external anal sphincter and the puborectalis muscles. The complete unilateral loss of the sacral nerves has almost no consequences [26].

7.6 Rectal and Anal Sensation and Control

Efficient control of the rectum occurs only if the sensory afferent messages from the bowel and pelvis are correctly interpreted by the control mechanism of the brain. There is still much to be learned concerning the location and nature of the afferent receptors, of the muscles that guard continence by day and by night, and of the differential function of the sphincter muscles.

7.6.1 Intrinsic Sensory Receptors of the Anal Canal

Duthie and Gairns [27] carefully plotted the sensory nerve ends in the anal canal. They found an abundance of conventional nerve endings, such as those presumed to denote pain (free intraepithelial), touch (Meissner's corpuscles), cold (Krause end-bulbs), pressure or tension (corpuscles of Pacini and Golgi-Mazzoni), and friction (genital corpuscles), together with unnamed, unconventional receptors in the anal canal of adults, lying distal to the valves and to a point 0.5–1.5 cm cranial to these valves. These receptors were responsible for acute and fine sensory discrimination, which in the skin beyond the pecten was mediated through receptors around the hair follicles. There was a crescendo of free nerve endings and genital corpuscles on the valve line, waning in the stratified columnar zone cranial to the valves. No receptors were found in the rectal mucosa, although myelinated and nonmyelinated nerve trunks were present under the epithelium, and Meissner's plexus of ganglion cells was readily identified. The rectal mucosa of the anal canal did not appreciate any of the above stimuli when tested by the techniques used and appeared to lack the appropriate receptors. They considered that receptors may be present in the rectum to receive distension stimuli, but that they were unable to demonstrate them by present staining methods.

In two other papers, Duthie and Bennett [28] and Duthie and Watts [29] suggested that the effect of rectal distention (as assessed using balloons in these experiments) was to relax the internal sphincter and contract the external sphincter. They claimed that the relaxed internal sphincter allowed feces to contact the very sensitive and effective anal canal receptors that induced external sphincter contraction, which is thus important in the fine control of continence. We suggest in the following section that the initiating signal of distention of the rectum may not be only from the rectal mucosa. In the rectal deformities discussed, both the internal and external sphincters may be rudimentary, yet a high degree of continence can be achieved.

7.6.2 Extrinsic Sensory Receptors

Work and observations on malformation of the anus and rectum led us to evaluate the absence of receptors in the rectal mucosa in a different way. We consider that coarse perception of distention of the rectum is in part a function of the parasympathetic nerves conveying impulses from the muscle spindles in the walls of the rectum and colon, but that fine appreciation of distention, even of minor changes, is the function of the muscles surrounding the anal canal. Furthermore, the pubococcygeus and puborectalis, with their intimate sleeve-and-sling relationship to the anal canal on the cranial aspect of the valves, provide the warning of impending peristaltic progress towards the anus. With the bowel empty and at rest, no sensation is registered, but gas, solid, or liquid content moving into the sleeve-and-sling zone provides a stretch that is immediately and keenly appreciated.

Goligher and Hughes [30], in studies in adults using balloon distention of the bowel brought down in pullthrough operations, also concluded that the response to distention probably arose in structures surrounding the bowel. Similarly, Parks et al. [31] and Porter [32], in studies on the pelvic floor muscles in rectal prolapse, suggested that the receptors lie in the rectal wall and the surrounding pelvic floor muscles. Kiesewetter and Nixon [33], in their anatomic and physiologic studies of rectal sensation in patients following surgical correction of ARM, considered that the sensory receptors responsible for a measure of rectal sensation were probably present in the puborectalis muscle.

The investigations of Freeman et al. [34] showed that anal sensation as detected by evoked cortical responses was not present at birth, but showed maturation in the first 3–4 months of life. If the eye of a newborn kitten is kept closed for 4–5 weeks after birth and then opened, the eye is permanently blind; appropriate repetitive somatosensory stimuli during the critical interval of brain development have not occurred. On this basis, they argued that the definitive pullthrough operation should be completed by 3–4 months of age to achieve the best functional results [34]. The results in neonatal pullthrough operations lend support to the above hypothesis [35].

7.7 Continence

7.7.1 Electric Properties of the Mechanism of Defecation

The internal anal sphincter has two functions: (1) it is persistently tonically contracted, and (2) it initiates the act of defecation by reflex dilation in response to rectal distention. This apparently contradictory behavior can be explained by the electric property of the smooth musculature of the sphincter. In the internal sphincter, a basic electric activity can be demonstrated similar to that found in the colon or rectum.

Electromyographic investigations of the smooth intestinal musculature carried out by Bulbring et al. [36], Bortoff [37], Bolzer [38], and Christensen [39] have shown that the changes in intraluminal intestinal pressure depend upon changes in the electric potential of these smooth muscle cells. These are slow, rhythmic potential changes of the membranes, the so-called basal electric rhythm (BER), and, in addition, super-added, fast, spike-like action potentials, which are triggered by a pacemaker cell causing segmental musculature contractions. The development of a propulsive wave of contraction is coordinated by various pacemakers in the longitudinal and circular musculature. These pacemakers are synchronized in an oral-aboral direction (Fig. 7.7). The frequency of the BER and the mechanical activity diminishes in the same direction, but increases again in the region of the rectosigmoid in the direction towards the anus. The frequency of the pressure waves in the lower rectum is greater than in the sigmoid and especially in the anal canal [40]. Here too, therefore, is an area where the pressure runs in the oral direction; thus, it is possible that the intestinal contents can be transported back into the more proximal segments of the colon, so that normally the rectum is empty. In ARM, this rectal property is acquired by the pulled-down colon several years after the pullthrough procedure [22, 23].

7.7.2 Pharmacologic Properties of the Mechanism of Defecation

Anorectal motility is frequently disturbed in ARM. It is therefore important to consider briefly the physiology of normal bowel movements. Both the origin and the propagation of the propulsive waves, and in all probability the segmental contractions, are regulated via the intramural bowel-wall plexus. Distension of the bowel wall by a stool bolus produces an excitatory impulse, which, after traversing the submucous plexus and being transmuted by the myenteric plexus, leads to a cholinergic contraction oral to the bolus and to a nonadrenergic, noncholinergic (NANC) relaxation that is mediated by nitric oxide (NO)-containing inhibitory neurons, aboral to the bolus. Adrenalin modulates the acetylcholine release at cholinergic synapses. Nitric oxide has recently been recognized as a neurotransmitter that mediates relaxation of the smooth muscles of the gastrointestinal tract. Besides NO-containing inhibitory neurons, many other peptidergic neurons that store, for example, vasoactive intestinal peptide (VIP), substance P, neurokinin A, and many others, are involved in the peristaltic reflex. In addition, the interstitial cells of Cajal have important regulatory functions in the human gut musculature and on bowel motility. If they are disturbed or even lacking, severe chronic constipation may result (Fig. 7.8A) [23].

Two different pharmacologic regions can be demonstrated in the internal sphincter. In the proximal part, acetylcholine will cause a contraction exactly as in the rectum and in the rest of the alimentary tract, and nitric oxide will cause relaxation (Fig. 7.8B). Ganglion cells are present here. In the more distal parts of the internal anal sphincter, alpha-stimulating, beta-relaxing receptors and especially NANC relaxing nerve fibers are present. The number of ganglia and ganglion cells diminishes in an anal direction. Relaxation is also mediated by VIP fibers; Cajal cells also play an important role in the function of the internal anal sphincter.

It is thought that relaxation is stimulated by tension receptors in the puborectal perception field and is transmitted via the ganglion cells of the mesenteric plexus to NANC neurons running to the sphincter. In the distal two-thirds of the internal sphincter, the impulses travel more electronically via a nexus in an anal direction [23].

Electromanometrically, a direct proportionality of the distending volume to the duration and amplitude of the relaxation can be shown. Electromyographically, the BER of the smooth muscle cells is desynchronized during the first part of the relaxation reflex (Fig. 7.9). This action can be demonstrated by cineradiography (see Fig. 7.3). In the course of defecation, the anorectal angle becomes less acute, and the proximal third of the anal canal becomes dilated while the sphincter is still markedly contracted. Because this sympathetic nerve causes contractions via alpha receptors, the opening up of the anal canal lasts only for a short period and defecation occurs in the form of a number of propulsion waves. In patients with transverse section of the spinal cord secondary to lumbar myelomeningocele, the sympathetic action is absent and marked and delayed relaxation occurs [22]. The reflex arc, however, remains intact; it passes not via the spinal cord, but via the rectal ganglion cells of the myenteric plexus, which can be confirmed by the absence of the reflex in Hirschsprung's disease [22, 23].

In high ARM, although the sphincter is rudimentary, there might be some circular smooth muscle fibers persisting at a higher level so that some kind of rudimentary internal sphincter relaxation is demonstrable in a few patients. In low deformities, the sphincter can be fully developed, as in patients with imperforate anal membrane or orifices at the perineal site, but can also be rudimentary, as in girls with anovestibular fistula or in patients with anal agenesis (Figs. 7.6–7.9C).

According to Schweiger [41], the internal sphincter ani muscle contributes to 75% of the anorectal pressure profile and, according to Frenckner and von Euler [42], under resting conditions to 85% and under rectal distension and relaxation to 40–65%. After spinal or pudendal anesthesia [43] or in patients with paraplegia [31, 44], there is almost no diminution of the anorectal sphincter profile under resting conditions. Therefore, the internal anal sphincter has to be regarded, together with the puborectalis muscle, as one of the most important factors in anorectal continence. The complete strength of the anorectal sphincter mechanism, internal and external anal sphincters, and puborectalis muscle can be demonstrated by the anorectal resting-and-squeezing pressure profile (ARRP or ARSP; Fig. 7.10).

7.7.3 Puborectalis Muscle: Reflex Contractions

The voluntary contraction of the puborectalis muscle will interrupt the start of defecation. The puborectalis muscle and the external anal sphincter function act here as a unit [6]. The persistent tonic contraction of these muscles is based on a proprioceptive reflex mechanism where the receptors are situated in the striated muscles of the pelvic floor and the ganglia in the lumbosacral spinal cord. This has been proved by the investigations of Parks et al. [31] in patients with tabes. Here the dorsal roots are destroyed, and the proprioceptive afferent nerve paths are therefore eliminated. No motor activity can be demonstrated in the muscles of the pelvic floor at rest. On the other hand, voluntary contractions of the pelvic floor and the external anal sphincter remain because the

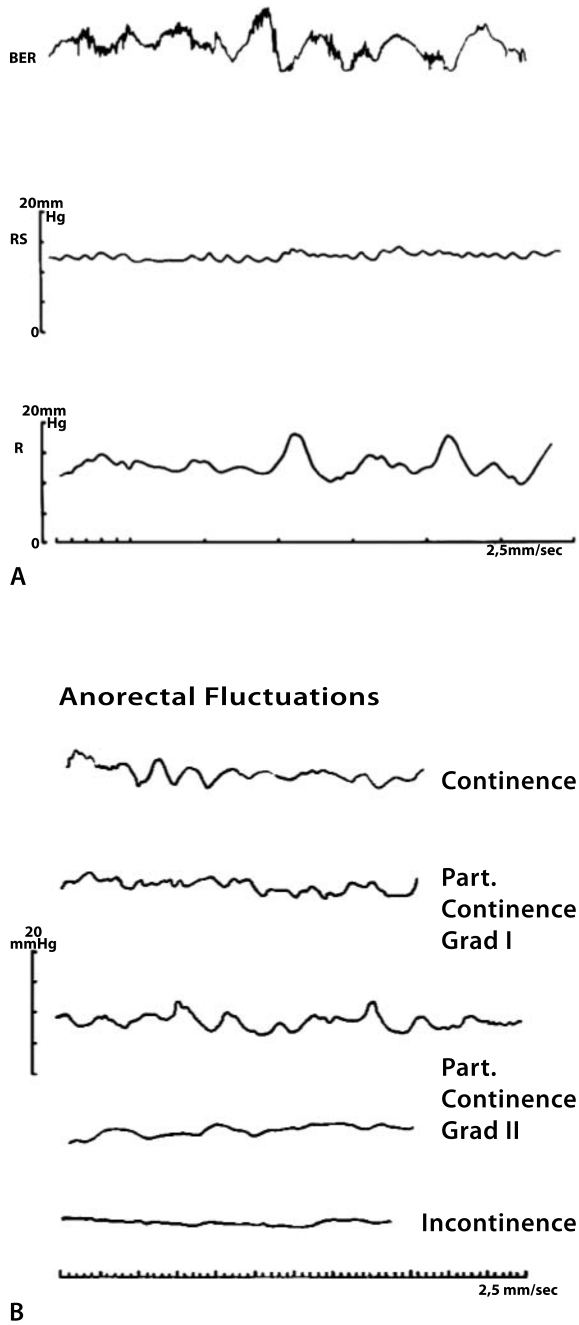
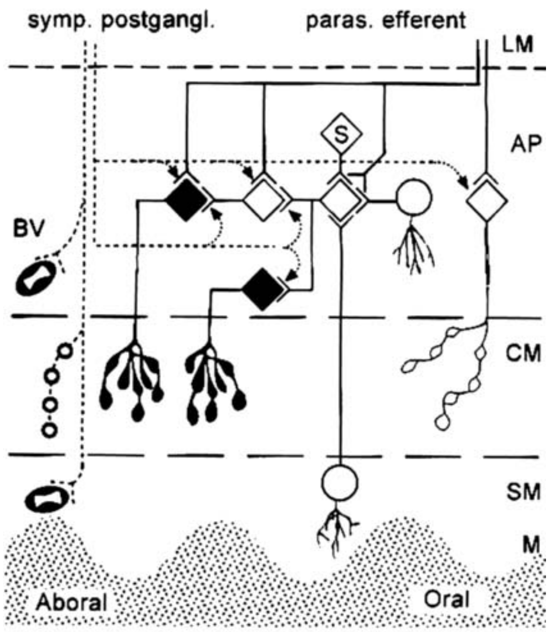


Fig. 7.7 **A** Basal electrical rhythm (*BER*): anorectal fluctuation of waves. Note the slow waves and the bursts of spike activity on the top of the waves. Simultaneous electromechanical contractions occur in the rectum. *R* Rectum, *RS* rectosigmoid. **B** Different pathologic patterns of anorectal fluctuations. The different morphology corresponds with different degrees of fecal incontinence. *Part.* Partial, *Grad* grade. **A** and **B** are reproduced from Holschneider [22, 23] with permission of the publishers)



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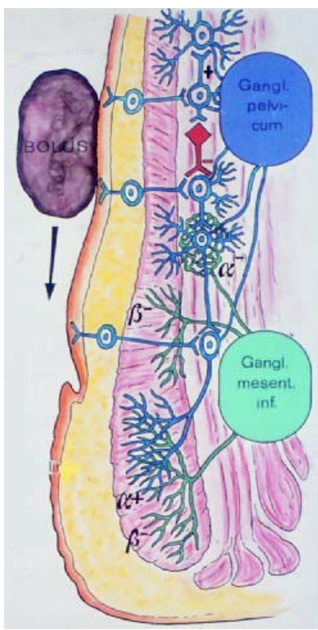

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Fig. 7.8 **A** Schematic portrayal of the peristaltic reflex, showing the intramural plexus and the efferent postganglionic adrenergic and preganglionic cholinergic axons entering the bowel. *SM* submucosa, *CM* circular muscle, *AP* Auerbach's plexus, *symp.* sympathetic, *paras.* parasympathetic, *postgangl.* postganglionic, *M* mucosa, *LM* longitudinal muscle layer; *peptidergic transmitters. The sensory neurons are indicated by *circles*. The impulses from the mechanoreceptor cells are transmitted via interneurons (*white squares*) over cholinergic synapses to the nonadrenergic-non-cholinergic (*NANC*) inhibitory neurons (*dark squares*). The finely drawn neurons with *white circles* in their terminal axons represent postganglionic, adrenergic axons. The circles marked *BV* indicate blood vessels. The neurons labeled *S* symbolize pacemaker neurons with spontaneous activity, situated in the interstitial Cajal cells of Stach's plexus (plexus submucosus extremus). Stimulation of the NANC neurons leads to a neurogenically induced and peptidergic-transmitted relaxation aboral to the bolus. Oral to the bolus, a myogenically produced contraction of the circular muscle occurs (rebound excitation). The sympathetic system acts as a modulator of acetylcholine release at the cholinergic synapses (reproduced from Holschneider and Puri [23]). **B** Schematic drawing of the innervation of the internal anal sphincter. Note: alpha-stimulating, beta-relaxing receptors and especially NANC relaxing fibers. Relaxation is also mediated by vasoactive intestinal peptide (*VIP*) fibers. In contrast, in the proximal bowel there are beta- and alpha-relaxing influences [23].

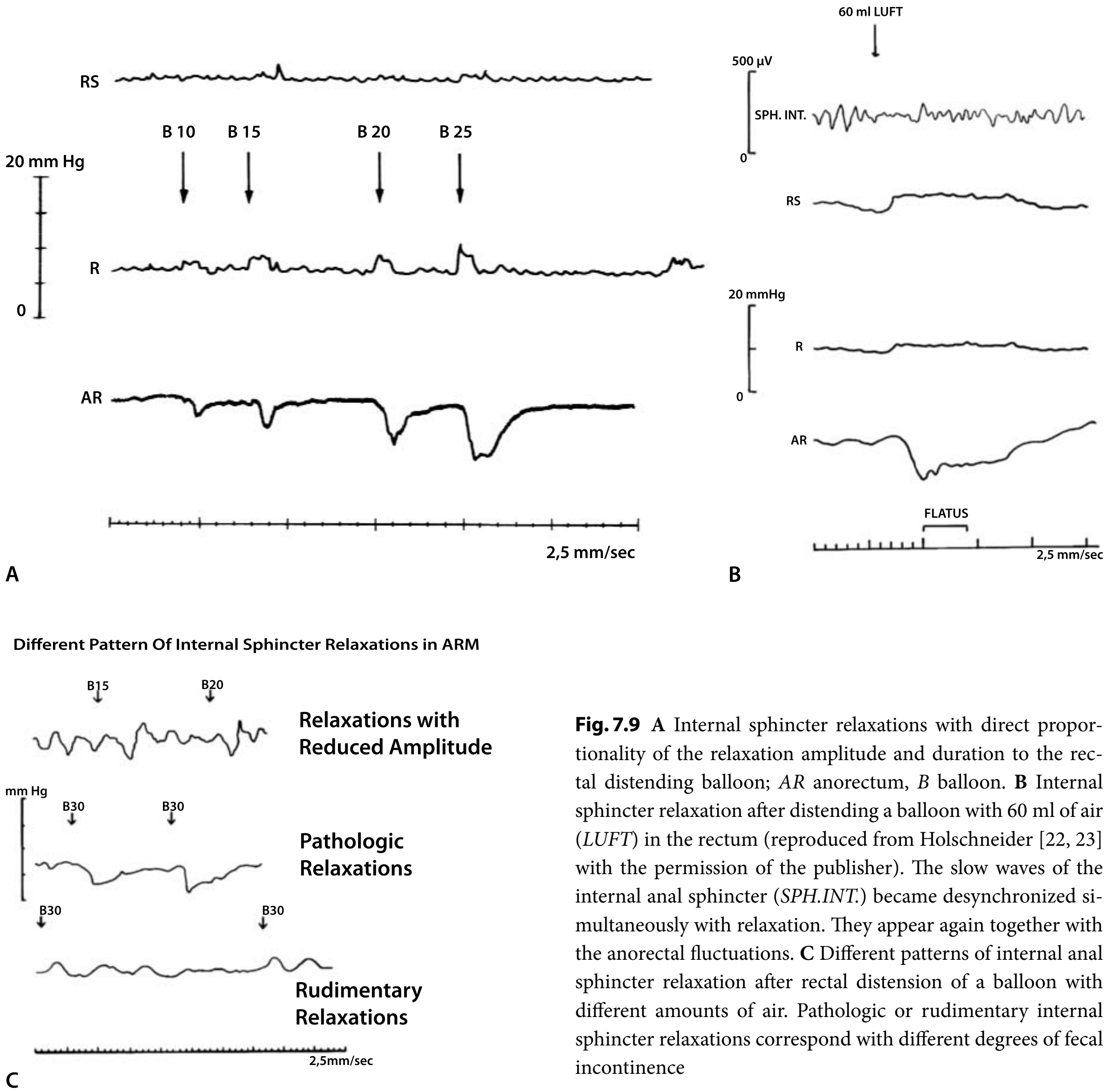

Fig. 7.9 **A** Internal sphincter relaxations with direct proportionality of the relaxation amplitude and duration to the rectal distending balloon; *AR* anorectum, *B* balloon. **B** Internal sphincter relaxation after distending a balloon with 60 ml of air (*LUFT*) in the rectum (reproduced from Holschneider [22, 23] with the permission of the publisher). The slow waves of the internal anal sphincter (*SPH.INT.*) became desynchronized simultaneously with relaxation. They appear again together with the anorectal fluctuations. **C** Different patterns of internal anal sphincter relaxation after rectal distension of a balloon with different amounts of air. Pathologic or rudimentary internal sphincter relaxations correspond with different degrees of fecal incontinence

function of the anterior horn cells is left intact. The increase in activity in these striated muscles of the pelvic floor when speaking or coughing, when taking a deep breath, or when touching the rectum, shows that the puborectalis muscle is a much more sensitive receptor for alterations in pressure than the rectum (Fig. 7.11). The reflex contraction of the striated anal sphincter muscles during internal anal sphincter relaxation (called continence reaction or continence reflex) is a further important factor supporting anal continence. The receptors for this proprioceptive reflex mechanism lie eventually in the parapuborectalis tissues, and so this reflex remains intact even after amputation of the rectum or low anastomosis.

7.7.4 Adaptation Reaction

The adaptation reaction causes a delay in the passage of the intestinal contents. It thus prevents the changes in volume that will produce a rise in pressure, which would endanger continence (Fig. 7.12). This reaction shows itself electromanometrically as a steep rise in pressure followed by a slow pressure decrease at rest. The change in volume per unit of change of pressure, the so-called compliance, can be calculated for the rectum and is a measure of the elasticity of this organ. In patients with a rigid anorectum, or when the colon is surrounded by fibrous tissue, as can be found after abdominoperineal pullthrough operations, an injection of even a small volume of air will lead to a steep rise of the intraluminal pressure, as there is a markedly lower compliance. On the other hand, in patients with secondary megacolon with a widely dilated rectosigmoid, the injection of even large volumes of air will cause only a very small rise in pressure, so that there the compliance is abnormally high. It can therefore be said that the function of the rectum is not so much as a storage or reservoir organ but as an organ that delays the passage and promotes the return of the stool into the more proximal intestinal segments in an attempt to prevent irritation of the puborectal perception field. Both these functions depend on the length of the rectum. Continence is not impaired after the resection of the colon, provided the distal 4–5 cm of rectum is left intact. If only 3cm of rectum is left behind, continence occurs in only 80%. If only 2 cm is left, 50% of the patients will not be continent.

7.7.5 Feeling of Fullness

If the rectum is filled with intestinal contents, tension receptors in the rectal wall and in the neighboring

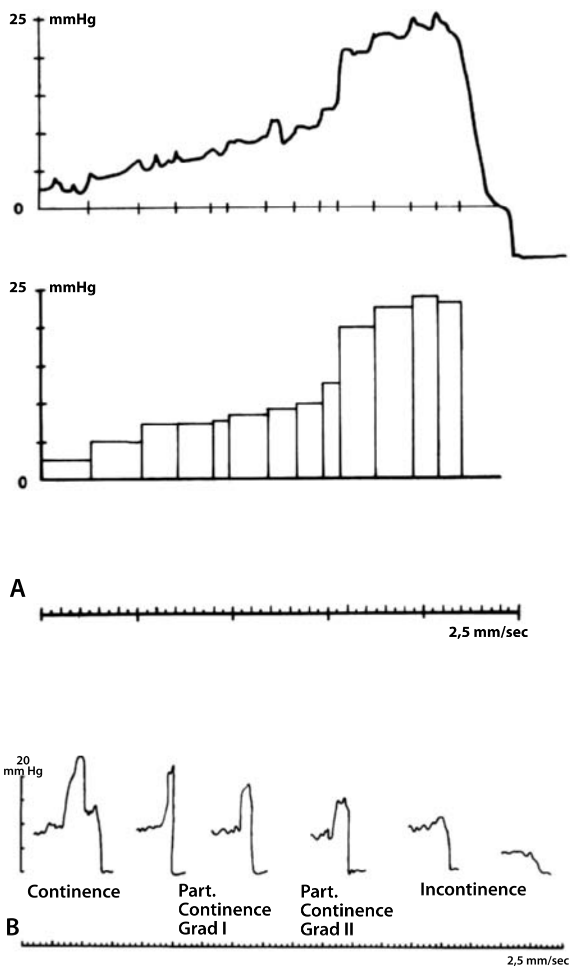

Fig. 7.10 **A** Anorectal resting pressure profile. The horizontal lines symbolize 1 cm of anorectal length each (reproduced from Holschneider [22, 23] with the permission of the publisher). The catheter is pulled out from the rectal area (pressure 3 mmHg) to the anus (pressure in the internal sphincter area 20–25 mmHg). **B** Different patterns of anorectal pressure profile in relation to different degrees of fecal continence

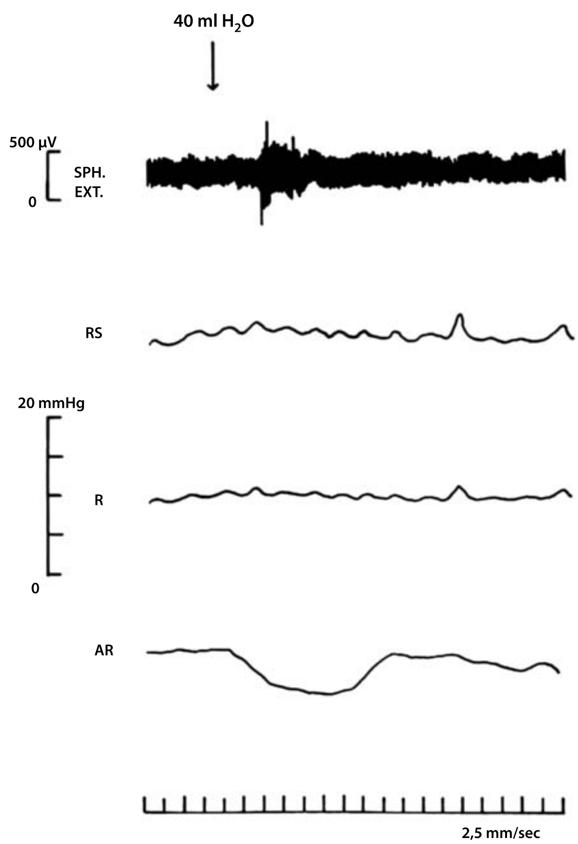

Fig. 7.11 Increasing activity of the external anal sphincter (*SPH.EXT.*) during injection of 40 ml of physiological saline in the rectosigmoid. Note the increasing reflex activity in the external anal sphincter (continence reaction, external sphincter reflex; reproduced from Holschneider [22, 23] with the permission of the publishers)

puborectal muscles are stimulated, and the nerve impulses passing via the spinal cord up to the cortical centers will transmit a feeling of fullness and a desire to pass stool. At the same time, the reflex mechanism of the sphincters is triggered off, causing the relaxation of the internal anal sphincter and the simultaneous contraction of the external anal sphincter and the puborectalis [45, 46].

7.7.6 The Anorectal Angle

In this connection, investigations carried out by Denny Brown and Robertson [47] are of interest. These authors were able to show that in kittens, in which during the 1st week of life the musculature loses its red color and as well as some lipids, there is an increased ability for rapid contractions. This seems to indicate that the pale, rapidly contracting muscle fibers of animals develop only after birth. Holschneider and Lierse [23] were able to show that in man, the small-diameter red fibers are used for tone and the pale, large-diameter fibers are used for phasic contractions, and that in young infants the appearance of continuous electrical activity in the striated muscles of the pelvic floor is associated with the child's ability to attain an erect posture.

Apart from the reflex contractions, the striated muscles of the pelvic floor are governed by cortical centers and can be contracted voluntarily. The voluntary and involuntary contractions of these muscles depend upon two different types of muscle fibers, those with a high content of myoglobin, which are responsible for slow tonic contractions, and those (pale fibers) with a low content of myoglobin, which are responsible for rapid phasic contractions. Investigations by Dubowitz and Pearse [48, 49] showed that as well as anatomic differences, there are also biochemical and histochemical differences in the fat, glycogen, and enzyme contents of these muscle fibers. The difference between these two types of fibers is, therefore, based on different metabolism. The red fibers contain several oxidizing enzymes and obtain their energy from the Krebs cycle, whereas the pale fibers have a high content of phosphorylase and need glycogen for their energy. Because the Krebs cycle is a better source of energy than glycolysis, the red fibers are better able to maintain tonic activity, whereas the pale fibers containing phosphorylase are designed for a sudden rapid increase of activity.

7.7.7 Corpus Cavernosum Recti

The corpus cavernosum recti is a network of arterial vessels situated at the area of the anorectal line, working as a second plugging mechanism of the anal canal together with the corrugator and pubococcygeus. Its venous flow drains transsphincterically to the portal vein. These vessels fill during internal contraction, thereby obstructing the upper part of the canal.

7.7.8 The Rectum

The rectum and a colon of adequate length for the resorption of water are also important factors in anorectal continence. The most important function of the rectum is delaying the passage of intestinal contents. This is achieved by a pressure gradient that runs against the direction of normal peristalsis (i.e., in a cranial direction), and also by the adaptation reaction.

7.7.9 Mechanism of Defecation

Various reflexes, such as the gastrocolic reflex and the ileocolic reflex contraction of the colon, caused by filling the stomach and the ileum, respectively, as well as voluntary contraction of the abdominal musculature, may initiate defecation by suddenly filling the rectum with colonic contents. The increasing intrarectal pressure stimulates the distention receptors in the puborectalis muscle and the parapuborectal tissues, and the desire to pass a stool is consciously felt. At the same time, a reflex relaxation of the internal anal sphincter occurs. This allows even the smallest amounts of stool to reach the anal canal. The hypersensitive mucosa of the anal canal in the region of the anal valves is able to discriminate between flatus and liquid or solid stool. The reflex contraction of the external anal sphincter and the puborectalis will prevent the expulsion of stool from the anal canal and thus inhibit fecal soiling. This effect is increased by the compression of the lower anal canal by the corpus cavernosum of the rectum, and by the corrugator muscle of the anus. This allows the rectum time to adapt itself to the increased intraluminal pressure. The aboral–oral pressure gradient of the rectum will propel the stools upward into a more proximal rectal segment. This, however, will stimulate further propulsive waves via a feedback mechanism. An intrarectal pressure of between 25 and 30 mmHg will stimulate a reflex inhibition of the anorectal sphincters and the

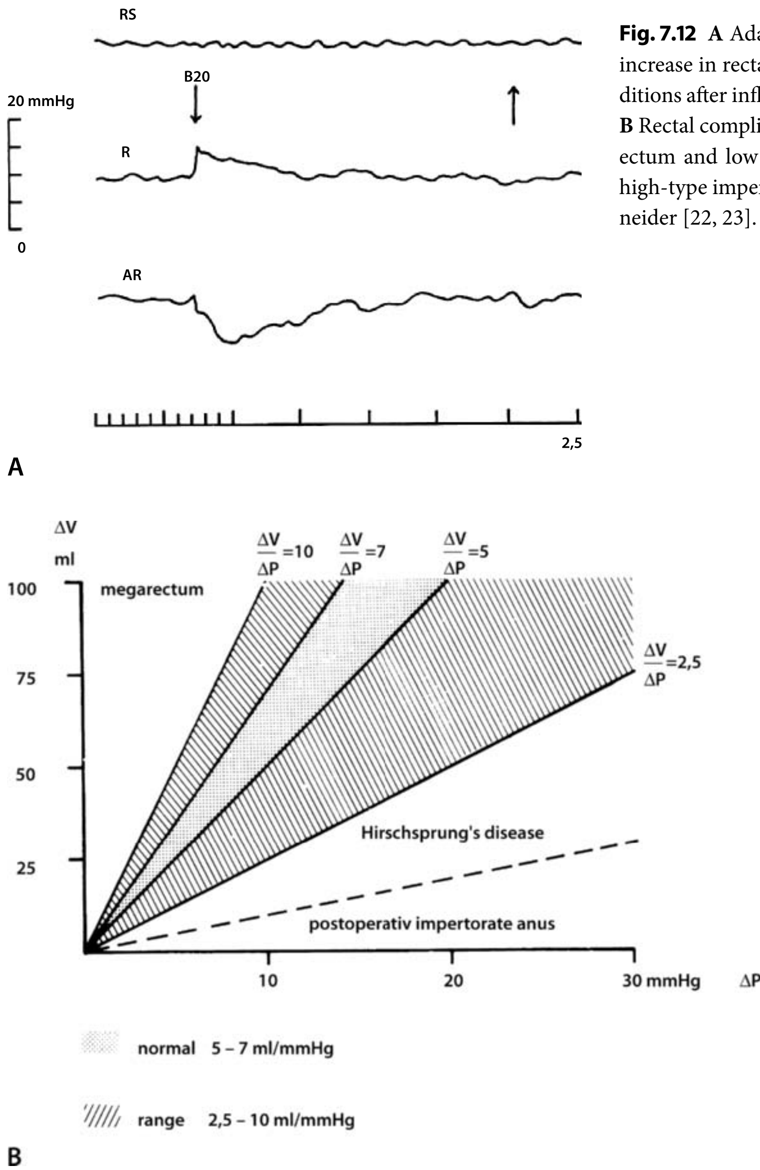

Fig. 7.12 **A** Adaptation reaction in the rectum. Note the sharp increase in rectal pressure and its slow decrease to resting conditions after inflation of a rectal balloon with 20 ml of air (B20). **B** Rectal compliance. The compliance is high in cases of megarectum and low after pullthrough procedures in patients with high-type imperforate anus. **A** and **B** reproduced from Holschneider [22, 23]. *V* volume, *P* pressure

puborectalis muscles. The voluntary contractions of the abdominal muscle will also cause a reciprocal inhibition of the striated muscles of the pelvic floor. This, in turn, will decrease the acuteness of the anorectal angle formed by the puborectalis muscle, and defecation commences. When the rectoanal reflex operates following sudden distention of the rectum, sampling of whether the waste is solid, liquid, or gas occurs at approximately the level of the anal valves. If defecation is not intended, voluntary contraction of the puborectalis muscle will return the contents back into the rectum off the sensitive zone and the desire to defecate will diminish.

The external sphincter is a powerful muscle that is brought into action in moments of stress to supplement the sling action in arresting defecation or deflation. It too has a resting tone that mildly occludes the anus, and when forced open by flatus under high pressure it exhibits a flutter valve action with the accompanying characteristic noise. The tone of the internal and external sphincters that surround the skin-lined anal canal is probably responsible for prevention of wetting of this part of the canal with mucus secreted from the adjoining rectal mucosa in the long intervals between acts of defecation. Neither of these sphincters accounts for the minute-to-minute day and night fecal continence, which appears to be the function of the sleeve-and-sling.

Further observations of children who become chronically constipated indicate that the sleeve-and-sling become easily tired by impacting feces, become relaxed, and permit shortening of the anal canal to the length only of the skin-lined anus. It is found that then the short passage, although encircled by external and internal sphincters, is barely sphincteric, permitting constant leakage, which is momentarily arrested only at the time of conscious muscular contractions of the external sphincter surrounding the skin-lined anus.

Finally, the anal canal constructed in patients exhibiting a congenital rectourethral fistula is endowed with a high degree of sensation, content discrimination, and muscular sphincter function if the new canal is lodged within the striated muscle complex, which is then its only sphincter. If the canal is directed to the perineum through the muscular diaphragm posterior to the sling, the bowel lacks appreciation of its content and all power to control defecation [50–52].

The observations of Duthie and Gairns [27], in our view, support this extrinsic theory of rectal sensitivity as far caudally as the anal valves, and we take cognizance of their findings of keen intrinsic receptiveness of the short, skin-lined anus to, for example, distention, temperature, and friction. A skin-lined anal canal is vital to continence. Kiesewetter and Nixon [33] showed an ingrowth of sensory fibers from the perianal skin following pullthrough operations. Anoplasty, to create a skin-lined canal if prolapse occurs after pullthrough operations, improves continence [53]. Stephens and Smith [54], however, consider that proper function of the puborectalis muscle is adequate for near complete continence, including content discrimination, as is found in patients after rectoplasty operations for ARM.

References

1. Magnus RV(1968) Rectal atresias as distinguished from rectal agenesis. J Pediatr Surg 3:595–598
2. Federative committee on anatomical terminology (1998) Terminologia Anatomica, International Anatomical Terminology. Georg Thieme Verlag, Stuttgart
3. Stelzner F (1981) Die anorektalen Fisteln, 3 Aufl.. Springer, Berlin, pp 1–30
4. Fritsch H (1988) Developmental changes in the retrorectal region of the human fetus. Anat Embryol 177:513–522
5. Jones FW (1904) The nature of malformations of the rectum and the urogenital passages. Br Med J 2:1630–1634
6. Fritsch H, Brenner E, Lienemann A, Ludwikowski B (2002) Anal sphincter complex. Reinterpreted morphology and its clinical relevance. Dis Colon Rectum 45:188–194
7. Milligan ETC, Morgan CN (1934) Surgical anatomy of anal canal with special reference to anorectal fistula. Lancet 2:1150–1155
8. Aldridge RT, Campbell PE (1968) Ganglion cell distribution in the normal rectum and anal canal. A basis for the diagnosis of Hirschsprung's disease by anorectal biopsy. J Pediatr Surg 3:475–490
9. Aigner F, Zbar AP, Ludwikowski B, Kreczy A, Kovacs P, Fritsch H (2004) The rectogenital septum: morphology, function, and clinical relevance. Dis Colon Rectum 47:131–140
10. Taffozoli K, Soost K, Wessel L, Wedel T (2005) Topographic peculiarities of the submucous plexus in the human anorectum – consequences of histopathologic evaluation of rectal biopsies. Eur J Pediatr Surg 15:159–163
11. Fowler R (1957) Landmarks and legends of the anal canal. Aust NZ J Surg 27:1–18
12. Peschers UM, DeLancey JO, Fritsch H, Quint LE, Prince MR (1997) Cross-sectional anatomy of the anal sphincters. Obstet Gynecol 90:839–844
13. Fritsch H, Fröhlich B (1994). Development of the levator ani muscle in human fetuses. Early Hum Develop 37:15–25
14. Schärli AF (1971) Die angeborenen Missbildungen des Rektums und Anus. Aktuelle Probleme in der Chirurgie. Huber Verlag, Bern Stuttgart Wien
15. Wilson PM (1967) Anchoring mechanism of the anorectal region. S Afr Med J 41:1127–1132
16. Fritsch H, Lienemann A, Brenner E, Ludwikowski B (2004) Clinical anatomy of the pelvic floor. Adv Anat Embryol Cell Biol 175:1–64
17. El Shafik A (1975) The new concept of the anatomy of the anal sphincter mechanism and the physiology of defecation. The external anal sphincter: a triple loop system. Invest Urol 12:412–419
18. Ludwikowski B, Oesch-Hayward I, Fritsch H (2002) Rectovaginal fascia: an important structure in pelvic visceral surgery? About its development, structure, and function. J Pediatr Surg 37:634–638
19. DeVries P, Peña A (1982) Posterior sagittal anorectoplasty. J Pediatr Surg 17:638–643
20. Ikawa H, Yokoyama J, Sanbonmatsu T, Hagane K, Endo M, Katsumata K, Kohda E (1985) The use of computerized tomography to evaluate anorectal anomalies. J Pediatr Surg 20:640–644
21. Yokoyama, J, Hayashi A, Ikawa H, Hagane K, Sanbonmatsu T, Endo M, Katsumuta K (1985) Abdomino-extended sacroperineal approach in high-type anorectal malformation – A new operative method. Z Kinderchir 40:150–157
22. Holschneider AM (1983) Elektromanometrie des Enddarmes. Diagnostik und Therapie der Inkontinenz und der chronischen Obstipation, 2 Aufl. Urban u. Schwarzenberg, München, pp 52–86

23. Lierse W, Holschneider AM, Steinfeld (1993) The relative Proportions of Type I and II Muscle fibers in the external anal Sphincter ani muscle at different ages and stages of development – Observations on the development of continuence for J Pediatr Surg 3:28–32
24. Nixon HH, Callaghan RP (1964) Anorectal anomalies: physiological considerations. Arch Dis Child 39:158–160
25. Roberts WH, Harrison CW, Mitchel DA, Fischer AF (2005) The levator ani muscle and the nerve supply of its puborectalis component. Clin Anat 1:267–283
26. Gunterberg B, Kewenter J, Petersen I (1976) Anorectal function after major resections of the sacrum with bilateral or unilateral sacrifice of sacral nerves. Br J Surg 63:546–554
27. Duthie HL, Gairns FW (1960) Sensory nerve endings and sensation in the anal region of man. Br J Surg 47:585–595
28. Duthie HL, Bennett RC (1964) Anal sphincteric pressure in fissure in ano. Surg Gynecol Obstet 119:19–21
29. Duthie HL, Watts JM (1965) Contribution of the external anal sphincter to the pressure zone in the anal canal. Gut 6:64–68
30. Goligher JC, Hughes ESR (1951) Sensibility of the rectum and colon. Its role in the mechanism of anal continence. Lancet 1:543–548
31. Parks AG, Porter NH, Melzak J (1962) Experimental study of the reflex mechanisms controlling the muscles of the pelvic floor. Dis Colon Rectum 5:407–414
32. Porter NH (1961) Megacolon: a physiological study. Proc R Soc Med 54:1043–1047
33. Kiesewetter WB, Nixon HH (1967) Imperforate anus I. Its surgical anatomy. J Pediatr Surg 2:60–68
34. Freeman NV, Burge DM, Soar JS, Sedgwick EM (1980) Anal evoked potentials. Z Kinderchir 31:22–30
35. Freeman NV, Bulut M (1986) High anorectal anomalies treated by early (neonatal) operation. J Pediatr Surg 21:218
36. Bulbring E, Brading A, Jones A, et al (1970) Smooth Muscle. Edward Arnold, London
37. Bortoff A (1972) Digestion: motility. Ann Physiol 34:261–290
38. Bozler E (1948) Conduction, automaticity and tonus of visceral muscles. Experientia 4:213–218
39. Christensen J (1971) The controls of gastro-intestinal movements: some old and new views. N Engl J Med 285:85–98
40. Holschneider AM (1974) Elektromyographische Untersuchungen der Musculi sphincter ani externus und internus in Bezug auf die anorektale Manometrie. Langenbecks Arch Chir 333:303–316
41. Schweiger M (1979) Eine Methode zur Differenzierung zwischen dem Anteil der glatten und quergestreiften Analsphinktermuskulatur am Ruhetonus. Chir Forum für experimentelle und klinische Forschung. Langenbecks Arch Chir Suppl, pp 151–155
42. Frenckner B, von Euler C (1975) Influence of pudendal block of the function of the anal sphincters. Gut 16:482–489
43. Frenckner B, Ihre T (1976) Influence of autonomic nerves on the internal anal sphincter in man. Gut 17:306–312
44. Howard ER (1970) Anorectal Pressure Studies and their Clinical Applications. Recent Advances in Pediatric Surgery. Grune and Stratton, New York
45. Holschneider AM, Metzler E (1975) Manometrische Studien zur anorektalen Kontinenz im Kindesalter. Brun Beitr Klin Chir 221:14
46. Holschneider AM (1976) The problem of anorectal continence. Prog Pediatr Surg 9:85–97
47. Denny Brown D, Robertson EG (1935) An investigation on nervous control of defecation. Brain 58:256–307
48. Dubowitz V, Pearse AGE (1960) A comparative histochemical study of oxidative enzyme and phosphorylase activity in skeletal muscle. Histochemie 2:105–117
49. Dubowitz W, Pearse AGE (1960) Reciprocal relationship of phosphorylase and oxidative enzymes in skeletal muscle. Nature 185:701–702
50. Stephens FD (1963) Congenital Malformations of the Rectum, Anus, and Genitourinary Tracts. ES Livingstone, Edinburgh
51. Stephens FD (1965) Congenital rectal fistulae and their sphincters. Aust Paediatr J 1:107
52. Kiesewetter WB, Turner CR (1963) Continence after surgery for imperforate anus: a critical analysis and preliminary experience with the sacroperineal pullthrough. Ann Surg 158:498–512
53. Freeman NV (1984) The foreskin anoplasty. Dis Colon Rectum 27:309–313
54. Stephens FD, Smith ED (1971) Anatomy and function of the normal rectum and anus. In: Stephens FD, Smith ED, Ano-Rectal Malformations in Children. Year Book Medical Publishers, Chicago p 14
55. Stephens FD, Smith ED (1988) Anorectal malformations in children: update 1988. March of Dimes Birth Defects Foundation. Birth Defects Original Series 24 (4) Alan R Liss, New York
56. Huber AV, Hochstetter AHC, Allgöwer M (1983) Transsphinktere Rektumchirurgie. Topographische Anatomie und Operationstechnik. Springer, Berlin Heidelberg New York Tokyo
57. Fritsch H (2005) Gliederung des Bindegewebes im weiblichen Becken. Pathologe 26:273–275

Clinical Aspects

8 Incidence and Frequency of Different Types, and Classification of Anorectal Malformations

Feilim Murphy, Prem Puri, John M. Hutson and Alexander M. Holschneider

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8.1 Incidence

The incidence of anorectal malformations (ARM) is reported as 2.0–2.5 per 10,000 live births. However, there are significant variations in the prevalence between regions throughout the world. Confusion regarding the incidence is further increased with the addition and/or exclusion of anal stenosis in the studies in the literature. While no significant association between ethnicity and ARM has been identified, there are several studies that identify variations between ethnic groups [1,2].

In North America the Texas Birth Defects Registry reported 542 cases during the period of 1999–2001, thus a prevalence of 5.03 per10,000 live births for all cases of "stenosis or atresia of large intestine, rectum, or anal canal" [3]. In California, similar to Texas, a lower rate of incidence has been identified within the African-American community than within the Hispanic [3,4]. Between 1981 and 1986, the Centre for Disease Control monitored 4,617,613 births. Information on specific race and ethnicity was available for 92.6% of these births. These data revealed a higher incidence of ARM amongst Native American Indians and a lower incidence amongst African Americans than the rest of the population [2].

ARM are a common cause of bowel obstruction amongst neonates in Nigeria, where in one centre ARM accounted for 13.4% of all neonatal malformations and 39% of emergency neonatal surgery [5–7]. Louw reported an incidence of 5.5 per 10,000 live births in South Africa, with the highest rates noted among the Caucasians (5.7 per 10,000) and lower rates amongst the Bantu (4.4 per 10,000) [8]. However, further detailed prevalence rates are not available for sub-Saharan Africa.

The incidence rate in British Columbia was 4.0 per 10,000 live births (273 cases out of 689,118 consecutive liveborn infants) between 1964 and 1982 [9]. According to the Latin American Collaborative Study for Congenital Malformations (ECLAMC, Estudio Colaborativo Latino Americano de Malformaciones Congenitas), its frequency is 4.1 per 10,000 live births. The highest recorded incidence is 7.7 per 10,000 in Chile [10].

In Europe, several countries have reported prevalence rates: for example in Denmark an incidence of 3.8 per 10,000 live births was reported in Denmark over a 13-year period from 1980 to 1993; in Budapest, Hungary, 1.8 per 10,000 live births between 1970 and 1977 [11,12]. A study of different ethnic groups from Birmingham, UK, revealed that rectal atresia occurs more frequently among Europeans and South Asians than amongst those from the Caribbean [13]. A more detailed analysis was performed by EUROCAT (European network of population-based registries for the epidemiologic surveillance of congenital anomalies). Data from 1980 to 1994 from 33 registries revealed a prevalence of 4.05 per 10,000 births from a total of 4,618,840 births throughout Europe. It also revealed significant differences between registries; the lowest being in Luxembourg, with a prevalence of 1.14 per 10,000, to the highest in Finland with 6.13 per 10,000. There were also significant differences between registries within the same country, such as between Galway (1.2 per 10,000) and Dublin (4.1 per10,000) in Ireland [14]. This may reflect different ethnic groups within nations or Europe, or different embryological factors.

In the Indian subcontinent, numerous studies have revealed a high prevalence, with ARM accounting for 15% of admissions to one unit; however, overall incidence figures are not presently available [15,16]. It is important to note the concept of the pouch colon with regard to the Indian subcontinent. Pouch colon

Table 8.1 Summary of incidences of anorectal malformations (ARM) from registries

ARM Registry	Region	Incidence (per) 10,000 live births
Texas Registry [3]	USA	5.0
Kansas [24]	USA	2.5
Louw [8]	South Africa	5.7
ECLAMC [10]	South America	4.1
EUROCAT [14]	Europe	4.1
Haeusler [25]	Europe	1.4
Finley [26]	Sweden	2.5
Stoll [27]	France	4.8

syndrome, or congenital short colon, is defined as a sac-like dilation of the shortened colon, which can lead to a massively dilated distal "pouch", and which fistulates into the genitourinary tract. This unusual condition, which is associated with ARM, is much more common in North India than in the rest of the world. Indian patients account for 92% of all reported cases and pouch colon syndrome accounts for 6–13% of all ARM in Northern India [17–19].

In the Far East, the prevalence is reported as 3.5, 5.04 and 3.38 per 10,000 for South Korea, Japan and China [20,21]. The incidence in the Singapore population is reported as 0.86 per 10,00 live births [22]. In Victoria, Australia they have reported an increase in the overall prevalence between 1983 and 1995 to 5.6 per 10,000, followed by a decline to 2.9 per 10,000 between 1999 and 2000. Similar to the increase in the incidence of gastroschisis born to teenage mothers within the state, they noted a significantly increased prevalence of births to mothers less than 20 years old (11.2 per 10,000) as opposed to those 25–29 years old (4.5 per 10,000) [23].

Some argue that the differences in the prevalence results are due to problems with case inclusion; however, ARM patients who are captured by a registry or a study are generally assessed by paediatric surgeons who are quite specific about the type of abnormality [14]. Thus, despite the variable prevalence rates ranging from 0.86 to 7.7 per 10,000, the overall incidence is 2.0–2.5 per 10,000 live births (Table 8.1).

8.1.1 Associated Anomalies

ARM occur in 2.2% of children with trisomy 21, with a range of 2–8% [28–31]. This incidence is 15 times more common than the rate within the general population [32]. In racial groups such as African Americans, the incidence of ARM is much higher in children with Down's syndrome than its incidence in the general population [33]. Black and Sherman were the first to describe the particular association between Down's syndrome and ARM without fistula [34].

A small number of studies have demonstrated an increased incidence among first-degree relatives of affected patients. Data from the 1950s placed the risk of a second child in a family being born with ARM at 1%; however, this has not been confirmed in later studies [35]. There are case reports of siblings and parents and children both having the anomalies, especially anal stenosis [36]. In 1957, Kaijser et al. reported the case of a mother and two daughters in whom each had an imperforate anus with a rectovaginal fistula [37]. In 1961, VanGelder commented on a family with three known and one possible ARM extending over four generations. The pattern suggested a dominant inheritance with reduced penetrance [38]. Seitz et al. reported anterior ectopic anus in a mother and her two daughters [39].

In India, Mittal et al. reported that a detailed family history revealed 21 out of 140 patients (15%) had a sibling with an ARM [40]. In a report from Manchester, 15 out of 186 patients (8%) had a strong family history of ARM [41]. Schwoebel et al. describe two families with nine members affected with ARM over two to three generations, with nine members affected with ARM, with a recurrence risk of 10–20% for first degree relatives [42]. Some studies reveal evidence of autosomal recessive inheritance, while others demonstrate an autosomal dominant syndrome [38]. Christensen et al. found an increased frequency of chromosome anomalies among children with ARM [43].

Genetic factors are believed to be more important in anorectal stenosis than other anal anomalies. However, there is little evidence to imply that genetic predisposition plays a significant role in the aetiology of this condition [44].

In 1981, Currarino et al. described the triad of anorectal stenosis (low ARM), an anterior sacral defect and a presacral mass [45]. The complete Currarino triad is rare and is familial in 50% of cases [46]. A screening program with lumbosacral ultrasound and plain radiography for siblings of those with the Currarino triad should be obligatory [47].

Cuschieri and the EUROCAT group demonstrated that of the 1,846 patients in the registry with ARM, 1,174 children had other defects [31]. Chromosomal abnormalities occurred in 11%, most frequently those with anal stenosis. VACTERL association was present in 181 patients (10%) and multiple congenital anomalies in unrecognised patterns were present in 711 (38%) [31]. VACTERL stands for Vertebral, Anorectal, Cardiac, Tracheo-oEsophageal, Renal and Limbs particularly radial, anomalies. When three or more anomalies are present together then VACTERL is considered to exist.

Associated anomalies are more common in boys (52–63%) and the higher the ARM, the higher the risk of associated anomalies, commonly ranging from 44% to 67% [48]. Associated anomalies can be twice as prevalent in patients with higher anomalies than in those with lower lesions [40]. However, Javid et al. reported a 61% incidence of associated anomalies in girls with low ARM [49].

Vertebral and spinal anomalies are commonly associated, especially with supralevator lesions, with an incidence of 4.6–40%. Abnormalities such as vertebral defects, tethered spinal cord or diastematomyelia can occur, but by far the most frequent is a sacral anomaly consisting of the absence of one or more sacral vertebrae [14].

Ratan et al. from Haryana in India presented data from 416 patients demonstrating that males had a significantly higher incidence of genital anomalies in association with low ARM, and gastrointestinal tract anomalies in association with high ARM. Unlike other reports, girls with high ARM had more urological anomalies compared with the boys with high ARM [50].

Genitourinary anomalies occur in 21–61% of patients [14]. Up to 26% of boys had genitourinary problems, as opposed to 5% of girls, with upper urinary tract anomalies present in 50% of boys and 30% of girls, respectively. The risk for both sets of problems increased with the level of the anorectal lesion [51]. Low lesions such as perineal cutaneous fistula have less than a 10% chance of having a urinary anomaly, while rectovesical fistulas have a 90% risk. The incidence of genital malformations increased in the presence of renal or spinal lesions. Conversely, the incidence of urinary and spinal anomalies increased in patients with genital malformations [51]. Undescended testes are commonly associated with ARM and are reported to occur in up to 19% [52].

Gastrointestinal anomalies are less common, with reported incidence of 10–25%, the commonest deformity being tracheoesophageal fistula (13%) followed by duodenal atresia [31,44]. Cardiac malformations occur in 9–20% of defects equally in patients with high and low lesions, with tetralogy of Fallot being the commonest diagnosis [44]. Several hypotheses exist for this association of abnormalities. Abnormal notochord development may be pivotal in producing neural tube defects and ARM, possibly by altering sonic hedgehog signalling [53].

8.2 Frequency

ARM occur more frequently in boys than girls. The sex ratio varies from 55% to 70% in favour of boys [22,54]. Smith and Stephens initially reviewed 2,376 cases in the literature from 36 published reports in 1970, and then 5,454 cases in 42 reports in 1988, which was in keeping with a male preponderance (Table 8.2).

Studies show a higher incidence of supralevator lesions in boys as compared to girls, ranging from 44% to 65%; however, there is significant variation between studies. In Nigeria, female neonates were affected more than males in a ratio of 1.5:1, with high lesions present in 30 (55.6%) and low lesions in 24 (44.4%) [57] (Table 8.3).

The frequency of each type of anomaly is difficult to define clearly. The reason for this is the failure of a single classification to be adopted. The medical literature is full of references and papers on ARM. In fact there are 1,211 references available on PubMed at the time of writing. However, the confused terminology and classification systems make comparisons near impossible. In order to define the frequency of different forms we will focus on the larger cohorts.

Table 8.2 Gender ratio of anorectal anomalies

	Stephens [44]	Endo et al. [55]
Male	57%	57%
Female	43%	43%
Total No. of patients	3,645	1,992

Table 8.3 Frequency of types of ARM

Authors	High	Intermediate	Low
Mittal et al. [40]	52%		48%
Cook [41]	28%	13%	51%
Rich et al. [56]	28%	54%	18%
Melbourne series [58]	37%		62%
Stephens series [44]	46%		54%
Chen [59]	20%	47%	33%
Endo et al. [55]	26%	11%	57%

Table 8.4 Incidence of subtypes (Stephens and Smith [44])

Type of anomaly	Incidence (%)	
	Male	Female
Rectourethral fistula	36	
Rectocloacal fistula		5
Rectovesical fistula	6	5
Rectovaginal fistula		19
Anorectal agenesis (no fistula)	8	4
Anterior anus	4	17
Anovestibular fistula		18
Covered anus with fistula	25	18
Covered anal stenosis	10	4
Total	1,429	951

The seminal work of Stephens and Smith reported the relative incidence of the subtypes in a series of 11 authors in 1988 using the "International" classification (Table 8.4).

Stephens and Smith's paper reported that in boys, 52% of lesions were high and 48% were low, while in girls 65% were high and 35% were low. They demonstrated that the rectourethral fistula is the commonest lesion in males, while lower anal lesions are twice as common as rectal anomalies in girls.

Kiesewetter's data demonstrated anal stenosis in 8% of cases, with rectourethral fistula in 29% of boys and rectovaginal fistula in 46% of girls; pure rectal atresia occurs only rarely [60,61].

EUROCAT data show that in patients with isolated ARM there is a male predominance for ARM without fistula, a female predominance for ectopic anus and an equal gender representation for ARM with fistula [14]. The same patterns of preponderance are present in those ARM that occur in association with VACTERL [31].

In 1999 the Japanese Study Group of Anorectal Anomalies (JSGA) published their results of a total of 1,992 patients (1,183 boys and 809 girls) registered from 1976 to 1995 [55]. They used the international 1970 classification and demonstrated low lesions in 57% of cases, intermediate in 11% and high in 26%. In males, 48% of lesions were low, 13% were intermediate and 36% were high. Among the girls, had 71% low lesions, 7% intermediate lesions and 13% high lesions. The most frequent deformity reported in Japan was male anocutaneous fistula (30%; $n = 364$), followed by male rectourethral fistula (28%; $n = 333$) and female anovestibular fistula (30%; $n = 241$). The incidence of rectovesical fistulas among the boys was 4% ($n = 42$), and that of rectocloacal fistulas among the girls was 11% ($n = 93$). Differences with Stephens and Smith consisted of a higher incidence of covered anus complete occurring at the same frequency (10.1% of low deformities) as covered anal stenosis. The JSGA series revealed a significantly lower rate of rectovaginal and rectovestibular fistula.

Peña states from his case load that the commonest defect in males is the rectourethral fistula followed by the cutaneous perineal fistula. In females the commonest defect is the vestibular fistula followed by the cutaneous perineal fistula. Rectovaginal fistulas are quite rare. Rectovesical fistulas occur in only 10% of all cases in both boys and girls. ARM without fistula occur in 5% [55,62].

8.3 Classification

ARM represent a wide spectrum of defects and conditions. A clear understanding of normal anorectal anatomy and the different types of ARM is necessary for both the planning of surgery and the procedure itself. An appreciation of the classification systems is useful in practice to the surgeon. This is, however, much easier said than done. The classification systems are notoriously difficult and unwieldy. There are multiple classifications in use in different centres throughout the world, making comparisons difficult. A brief examination of the literature demonstrates the multitude of classification systems available and in daily use. In order to explain the classifications that have been proposed over the years and that still exist we will discuss the history and development of the classification systems

Amussat, the father of the proctoplasty in 1835, was the first to attempt a classification system of ARM [63]. He described five groups: (1) a narrowed anus, (2) a closed anal membrane, (3) rectum interrupted by a septum at some distance from the opening, (4) imperforate anus and (5) the presence of a rectal fistula. Numerous other authors such as Stieda in 1903, Jones in 1904, Breener in 1915 and Frazer in 1926 created clinical classification systems [58]. However, it was the classification system of Ladd and Gross (1934) that prevailed and became the standard (Table 8.5) [64].

In 1963 a Melbourne team lead by Stephens classified the lesions into two categories, either high or low (Table 8.6). This classification recognises the importance of the puborectalis muscle and its effects in continence. Lesions above the pubococcygeal (PC) line were described as high and below as low. The PC line is drawn on a lateral pelvic radiograph “invertogram” between the midpoint of the pubis and the inferior aspect of the sacrum [65]. It represents the level of the levator ani attachment to the pelvic wall.

The Melbourne classification allowed Stephens to pioneer the sacrococcygeal approach in order to preserve the puborectalis. Although this classification was deficient in clinical information and overly

Table 8.5 Ladd and Gross classification 1934 [64]

Type	Anomaly
I	Anal and anorectal stenosis
II	Imperforate anus
III	Imperforate anus with blind ending pouch with fistula
IV	Rectal Atresia

Table 8.6 Stephens and Smith 1963 classification based on embryological concepts

Male	Female
Anorectal deformities	
A: Defect of the partition of the internal cloaca	
1. Anorectal agenesis (no fistula)	1. Anorectal agenesis (no fistula)
2. Rectovesical fistula	2. Rectovesical fistula
3. Rectourethral fistula	3. Rectourethral fistula
	4. Rectovaginal fistula
	5. Rectovestibular fistula
B: Secondary defects after Partition	
Rectal atresia	Rectal atresia
Rectal deformities	
A: Defects of the perineum	
1. Anterior perineal anus	1. Anterior perineal anus Perineal Vulvar
	2. Anovestibular fistula
	3. Perineal groove
	4. Perineal canal
B: Defects of the genital fold	
1. Covered anus complete	1. Covered anus complete
2. Anocutaneous fistula	2. Anocutaneous fistula
3. Anobulbar fistula	3. Anovulvar fistula
C: Defects of the proctodeal pit	
1. Anorectal agenesis	1. Anorectal agenesis
2. Imperforate anal membrane	2. Imperforate anal membrane
3. Anal stenosis (i) Covered anal stenosis (ii) Anal membrane stenosis (iii) Anorectal stenosis	3. Anal stenosis (i) Covered anal stenosis (ii) Anal membrane stenosis (iii) Anorectal stenosis
Unclassified	
1. Vesicointestinal fissure	
2. Duplication of the rectum and anus	
3. Combination of usual deformities	

Type of anomaly	Female	Male
Low, infralevator	I. Anal stenosis	I. Anal stenosis
	II. Anal membrane	II. Anal membrane
	III. Anal agenesis	III. Anal agenesis
	A. Without fistula	A. Without fistula
	B. With fistula	B. With fistula
High, supralevator	I. Rectal agenesis	I. Rectal agenesis
	A. Without fistula	A. Without fistula
	B. With fistula	B. With fistula
	II. Rectal atresia	II. Rectal atresia

Table 8.7 Anorectal anomalies based on a simplified Santulli classification [66]

Type of anomaly	Female	Male
High	1. Anorectal agenesis	1. Anorectal agenesis
	A: Rectal atresia	A: Rectal atresia
	B: With fistula Rectocloacal fistula Rectovaginal/high	 Rectovesical fistula Rectourethral fistula
	2. Rectal atresia	2. Rectal atresia
Intermediate	1. Anal agenesis A. Without fistula B. With fistula Rectovaginal fistula low Rectovestibular fistula	1. Anal agenesis A. Without fistula B. With fistula Rectobulbar fistula
	2. Anorectal stenosis	2. Anorectal stenosis
Low	1. At normal anal site Covered anus – complete Covered anal stenosis	1. At normal anal site Covered anus – complete Covered anal stenosis
	2. At perineal site Anocutaneous fistula Anterior perineal anus	2. At perineal site Anocutaneous fistula Anterior perineal anus
	3. At vulvar site Vulvar anus Anovulvar fistula Anovestibular fistula	
Miscellaneous	Anal membrane stenosis	Anal membrane stenosis
	Imperforated anal membrane	Imperforated anal membrane
	Perineal groove	Perineal groove
	Perineal canal	Perineal canal

Table 8.8 A simplified version of the 'International' classification

HIGH DEFORMITIES (SUPRALEVATOR)

1. Anorectal Agenesis—Male

(a) Without Fistula

1. ANORECTAL AGENESIS (Without Fistula)

The rectum ends blindly at any level in the pelvis above the levator with or without a fibrous cord to urethra. No internal sphincter; rudimentary external sphincter.

Diagnosis:

(i) No anus or distinguishing diagnostic sign. (ii) No gas or meconium in urine. (iii) Gas at or above P/C line on invertography. (iv) Urethrogram shows no fistula, nor any angulation to shape of urethra. (v) No fistula seen on colonogram later.

(b) With Fistula

(i)

2. RECTOVESICAL FISTULA

The rectum enters the bladder, usually towards the base. Usually associated with other serious anomalies, and all sphincters may be very rudimentary.

Diagnosis:

(i) No anus or distinguishing diagnostic sign. (ii) Gas or meconium in urine. (iii) Gas above P/C line on invertography. (iv) Gas in bladder. (v) Cystogram may show fistula.

(ii)

3. RECTOURETHRAL FISTULA

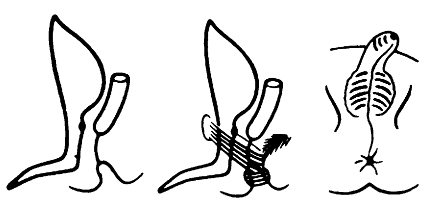
The most common high anomaly in males. Fistula is fine, and most commonly enters posterior urethra adjacent to the ejaculatory duct, but may be slightly higher or lower. Puborectalis foreshortened; intimately around urethra and below or at the fistula. Absent internal sphincter; variable external sphincter.

Diagnosis:

(i) No anus or distinguishing diagnostic sign. (ii) Gas or meconium in urine. (iii) Gas at P/C line, or just below, on invertography. (iv) May have gas in bladder. (v) Urethrogram may show fistula or characteristic angulation of urethra. (vi) Colonogram may show fistula later.

2. Rectal Atresia—Male

4. RECTAL ATRESIA



Rectum terminates at any level; thence a short or long fibrous cord to the sacrum or to the distal bowel. The anus and anal canal are normal. All sphincters present, and in normal relationship to anal canal. The lesion is probably an acquired vascular defect.

Diagnosis:

(i) Normal anus present. (ii) Obstruction on digital examination. (iii) Gas at or above P/C line on invertography. (iv) Distal canal shown by dye, and length of atresia shown by colonogram later.

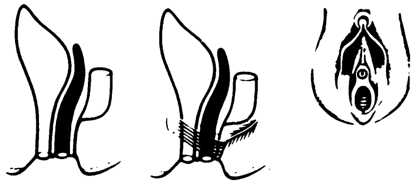
Fig. 8.1 Pictorial account of the 'International' classification

HIGH DEFORMITIES (SUPRALEVATOR)

1. Anorectal Agenesis—Female

(a) Without Fistula

14. ANORECTAL AGENESIS (Without Fistula)



As in the male.
The blind strand, if present, connects with vagina or perineum.

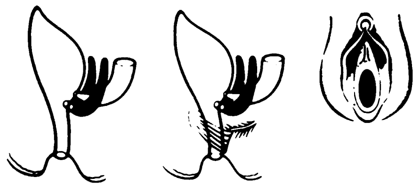
Diagnosis:

(i) Two perineal orifices (urethra and vagina) and no anus. (ii) No gas or meconium in urine. (iii) Gas at or above P C line on invertography. (iv) No fistula seen on colonogram later.

(b) With Fistula

(i)

15. RECTOVESICAL FISTULA



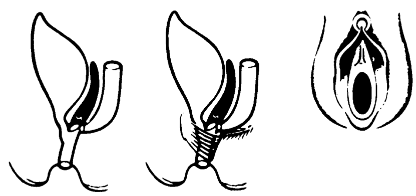
Rectum enters bladder between two separate vaginae which form a common cloaca at the bladder outlet. Other serious anomalies.

Diagnosis:

(i) A single cloacal orifice. (ii), (iii), (iv), (v) as in male.

(ii)

16. RECTOCLOACAL FISTULA



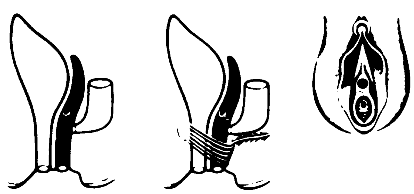
Rectum, vagina and urethra enter a common cloaca. Vagina may be short, single, or septate.
Sphincters as in male, with the puborectalis intimately surrounding the cloacal canal below the fistula.

Diagnosis:

(i) A single cloacal orifice in vestibule. (ii) Gas or meconium in urine. (iii) Invertography unreliable re gas levels. (iv) Vaginoscopy and vaginogram may show fistula. (v) Colonogram may show fistula.

(iii)

17. RECTOVAGINAL FISTULA High



Vagina usually normal, rectal fistula opens in midline posteriorly, orifice largish.
Sphincters as in rectocloacal fistula.

Diagnosis:

(i) Normal two orifices in vestibule. (ii) Meconium from vagina. (iii), (iv), (v) As in rectocloacal fistula.

2. Rectal Atresia—Female

18. RECTAL ATRESIA

As in male, very rare.

Diagnosis:

As in male.

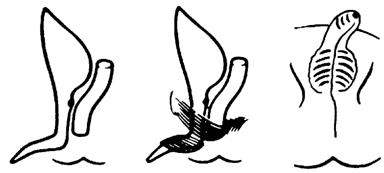
Fig. 8.1 (*continued*)

INTERMEDIATE DEFORMITIES

1. Anal Agenesis—Male

(a) Without Fistula

5. ANAL AGENESIS (Without Fistula)



The rectum terminates blindly at the upper border of the bulbocavernosus muscle close to the urethra. The puborectalis muscle is foreshortened around the caudal end of the gut; internal sphincter absent, and external sphincter rudimentary.

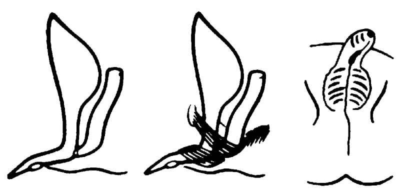
Diagnosis:

(i) No distinctive perineal apperance. (ii) No gas or meconium in urine. (iii) Gas near I point,* **i.e.** below P/C line, on invertography, with or without an anterior "beak." (iv) Normal urethrogram, without fistula. (v) Colonogram to confirm later.

*I point = tip of comma-shaped ossific centre of ischial bone.

(b) With Fistula

6. RECTOBULBAR FISTULA



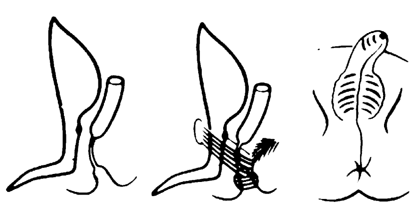
The rectum terminates above the bulbocavernosus muscle by a wide fistula into the bulb (rectobulbar), or enters the urethra more distally as a fine fistula. The rectum and fistula lie in a hammock of puborectalis, and the most caudal part of the fistula penetrates the sling; internal sphincter absent; external sphincter variable.

Diagnosis:

(i) Often no distinguishing perineal sign, but there may be a thin perineum, hypospadias, or cleft scrotum. (ii) Gas and meconium in urine. (iii) Probe passes along urethra into rectum. (iv) Gas may be seen near I point, below P/C line on invertography. (v) Urethrogram may show fistula. (vi) Colonogram to confirm later.

2. Anorectal Stenosis—Male

7. ANORECTAL STENOSIS



Anus present and anal canal may or may not be involved in the stenosis. Stenosis extends into lower rectum. Normal sphincters.

Diagnosis:

(i) Normal anus. (ii) Obstruction on digital examination, but not to probe. (iii) Fistulogram to show length.

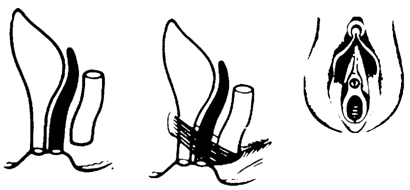
Fig. 8.1 (*continued*)

INTERMEDIATE DEFORMITIES

1. Anal Agenesis—Female

(a) Without Fistula

19. ANAL AGENESIS (Without Fistula)



As in male, with caudal end of blind gut near lower end of vagina.

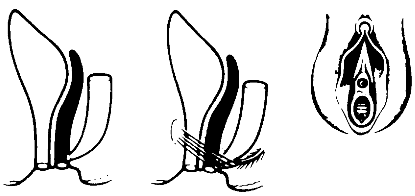
Diagnosis:

(i) Normal vestibule, with urethral and vaginal orifices; no distinguishing mark at the anal site. (ii), (iii) as in male, gas at or below I point.

(b) With Fistula

(i)

20. RECTOVAGINAL FISTULA (Low)



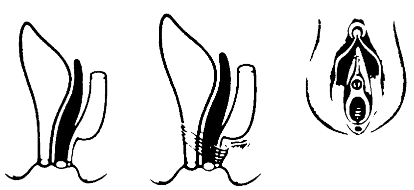
Fistula usually just above hymen. The puborectalis muscle surrounds the caudal bowel, although the fistula may penetrate the sling anteriorly.

Diagnosis:

As in high rectovaginal fistula.

(ii)

21. RECTOVESTIBULAR FISTULA



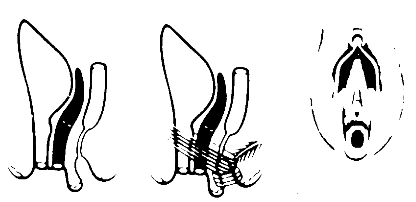
The rectum terminates at or just below P C line, with an anterior fine fistula 1-2 cms. long to the vestibule, coursing immediately adjacent to the posterior wall of the vagina. The terminal gut is supralevator, the fistula translevator. Vagina often double.

Diagnosis:

(i) Three orifices in vestibule, urethra, vagina, and a fine rectal fistula in fossa navicularis. Orifice totally surrounded by red vestibular mucosa. (ii) Probe into fistula passes cranially only. (iii) Measure length of fistula by marked Foley catheter. (iv) Fistulogram may define length.

2. Anorectal Stenosis—Female

22. ANORECTAL STENOSIS



Description and Diagnosis as in male.

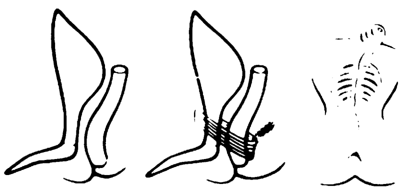
Fig. 8.1 (*continued*)

LOW DEFORMITIES (TRANSLEVATOR)

1. At Normal Anal Site—Male

(i) "COVERED ANUS—COMPLETE"
*includes No. 12

8. COVERED ANUS—COMPLETE



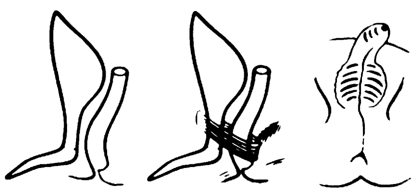
It was agreed that an occluded anus at the normal site may be due to either hypertrophy of genital folds or to imperforation of the anal membrane. The latter is so rare that it is placed in the "miscellaneous" group; most occlusions are of the former type.

The anus is "covered" at the normal site by excessive posterior development of the genital folds, shown as a thick raphe over the site, by anterior or posterior folds, or by a fused central band ("congenital median band").
Puborectalis surrounds normal anorectal canal, external sphincter variable.

Diagnosis:
(i) Perineal appearance of hypertrophied folds. (ii) Gas below I point on invertography. (iii) Percutaneous needling and X-ray dye study shows level.

(ii) "ANAL STENOSIS"
**includes No. 13

9. COVERED ANAL STENOSIS



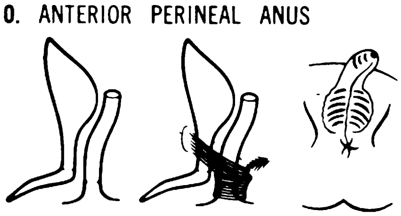
A stenotic anus at the normal site may be due to partial covering by genital folds, or to partial perforation of the anal membrane. A common term "anal stenosis" was chosen to include both types, although most will be covered deformities. The membrane deformity is classified in the "miscellaneous" group.

Identical to "covered anus—complete," except that the covering is incomplete resulting in a stenotic canal, or a fistula either side of a median band.

Diagnosis:
(i) Perineal appearance of hypertrophied folds. (ii) Fistulogram may show length.

2. At Perineal Site—Male

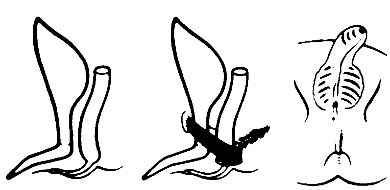
(i)
10. ANTERIOR PERINEAL ANUS



A normal anus situated further forward in the perineum. All sphincter components are normal.

Diagnosis:
(i) Normal anus, but anteriorly placed. (ii) No genital fold defects.

(ii)
11. ANOCUTANEOUS FISTULA
(Covered Anus—Incomplete)



The most common male deformity. Excessive posterior fusion of genital fold covers the anus and projects the orifice anteriorly as a fistula. The fistulous orifice may open anywhere from the normal anal site to the tip of the penis, but is usually just behind the scrotum. The anal canal is complete to the valves; the fistula is covered by thin skin of the raphe.
Puborectalis is normal, but internal sphincter absent, and external sphincter variable.

Diagnosis:
(i) Distinctive appearance of perineum—fistula orifice visible, or obscured by speck of meconium; fistulous track filled with meconium or by white epithelial pearls; the anal site a dimple, raphe or hypertrophied fold. (ii) Probe passes directly back along fistula into anal canal. (iii) Invertogram unnecessary, but gas below I point, provided levator not contracted.

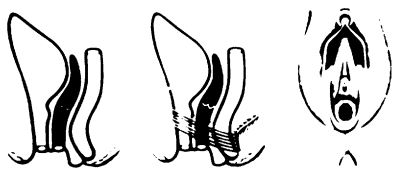
Fig. 8.1 (*continued*)

LOW DEFORMITIES (TRANSLEVATOR)

1. At Normal Anal Site—Female

(i) "COVERED ANUS COMPLETE"
*includes No. 30

23. COVERED ANUS—COMPLETE

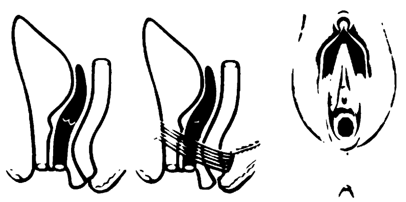


As in male.

Very rare in female.

(ii) "ANAL STENOSIS"
**includes No. 31

24. COVERED ANAL STENOSIS



Description of Anus: as in male.

Diagnosis

Three orifices in perineum, **i.e.** urethra, vagina and stenotic anus.

2. At Perineal Site—Female

(i)

25. ANTERIOR PERINEAL ANUS

A normal anus situated between the normal site and the fourchette. The common site is in the dry skin contiguous with the posterior limit of the vulva.

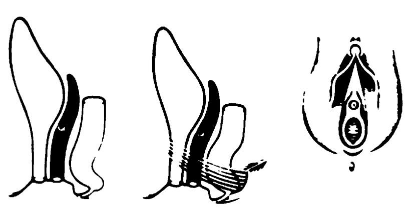
All sphincter components probably present.

Diagnosis:

As in male.

(ii)

26. ANOCUTANEOUS FISTULA
(Covered Anus—Incomplete)



As in male. The fistulous orifice is stenotic, bevelled anteriorly, and lies between anal site and the dry skin posterior to the vulva.

Diagnosis:

(i) Three orifices in perineum, **i.e.** urethra, vagina, and fistulous orifice in the dry skin of the perineum. (ii), (iii) & (iv) as for male.

Fig. 8.1 (*continued*)

LOW DEFORMITIES (TRANSLEVATOR) Contd.

3. At Vulvar Site—Female

(i)

27. VULVAR ANUS

A variety of anterior anus, but with so little development of the perineum that the anus lies in the vestibule. Anal orifice is normal, not stenotic, usually surrounded by wet vestibular epithelium; or it may be on the rim of the fourchette, wet in front, dry skin behind. The minoral tails pass in front and behind the anus.
Sphincters are all represented.

Diagnosis:

(i) Three orifices in the vestibular area—urethra, vagina, and normal anus.

(ii)

28. ANOVULVAR FISTULA

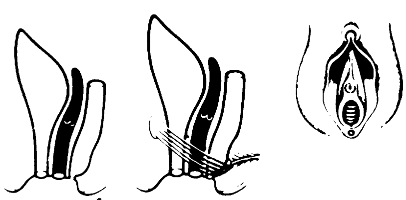
Another variety of covered anus (see ano-cutaneous fistula above) with the fistula projected into the posterior vulva.

Diagnosis:

(i) Three orifices in vulva—urethra, vagina, and fistula. (ii) Fistula orifice on rim of vestibule, with wet epithelium in front and dry skin behind. (iii) Minoral tails may project posteriorly as a raphe. (iv) Probe into fistula passes preferentially backwards close to skin, but also cranially. (v) Fistulogram may confirm.

(iii)

29. ANOVESTIBULAR FISTULA



A lesion probably embryologically identical with rectovestibular fistula but antomically dissimilar in that the fistula is very short. Rectum lies adjacent to the vagina and extends to the vestibule as a normal lumen except for a very short terminal fistula. Puborectalis normal, rudimentary internal sphincter, and variable external sphincter.

Diagnosis:

(i) Three orifices in vestibule—urethra, vagina and fistula. (ii) Fistula is tucked up in the fossa navicularis, completely within the vestibule, and totally surrounded by wet vestibular mucosa. No minoral tails.

(iii) Probe into fistula passes preferentially cranially, but also posteriorly. (iv) Fistulogram may confirm.

Fig. 8.1 (*continued*)

MISCELLANEOUS DEFORMITIES

FEMALE

(i)

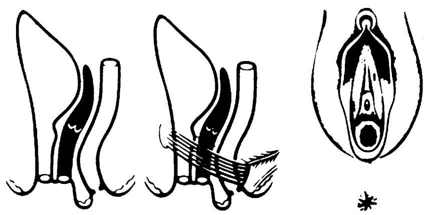
30. IMPERFORATE ANAL MEMBRANE*

As in male.

*See 23.

(ii)

31. ANAL MEMBRANE STENOSIS**



As in male.

**See 24.

(iii)

32. PERINEAL GROOVE

A normal vestibule anteriorly but a wet groove extends posteriorly to the anus, bordered laterally by hypertrophied minoral tails and labia majora. Anus is of normal calibre but with anterior ectropion into the groove. The groove later epithelialises into normal skin.

(iv)

33. PERINEAL CANAL

A normal anus and vestibule, but there is a fistula from the anal canal (near the valves) to the fossa navicularis. Sometimes the anal canal is anteriorly placed and stenotic.

(v)

VESICOINTESTINAL FISSURE
As in male.

(vi)

DUPLICATIONS OF ANUS, RECTUM & GENITO-URINARY TRACTS
As in male.

(vii)

COMBINATIONS AND VARIANTS OF THE USUAL DEFORMITIES
e.g. — complicated varieties of rectocloacal fistulae.
— rectovesical plus rectocloacal fistulae.
— rectovaginal fistula plus normal anus.
— rectovaginal fistula plus anal stenosis.
— anovulvar or rectovaginal fistula plus exstrophy of bladder.
— etc.

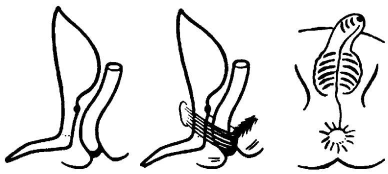
Fig. 8.1 (*continued*)

MISCELLANEOUS DEFORMITIES

MALE

(i)

12. IMPERFORATE ANAL MEMBRANE*



Anus at normal site, but proctodeal membrane intact. Puborectalis normal; no internal sphincter below the anal valves; deep external sphincter intact, but rudimentary superficial sphincter.

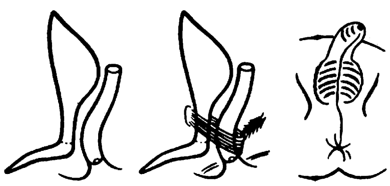
Diagnosis:

(i) Superficially normal anus, but membrane may bulge with meconium, or be found on digital examination. (ii) No genital fold defects. (iii) Gas well below I point on invertography.

*See 8.

(ii)

13. ANAL MEMBRANE STENOSIS**



As in imperforate anal membrane, but membrane partially perforate.

**See 9.

(iii)

VESICOINTESTINAL FISSURE

A complicated anomaly of exstrophy of the ileo-caecal region, short terminal blind bowel, exstrophy of paired bladders, and other associated anomalies.

(iv)

DUPLICATIONS OF ANUS, RECTUM & GENITO-URINARY TRACTS

About 40 recorded cases in various combinations of blind and fistulous openings.

(v)

COMBINATIONS AND VARIANTS OF THE USUAL DEFORMITIES

e.g. — rectovesical plus rectourethral fistulae.
— rectourethral fistula plus normal anus.
— colourethral plus coloneurenteric canal fistulae.
— rectobulbar fistula plus normal anus.
— covered anus with track deep to scrotum.
—etc.

Fig. 8.1 (*continued*)

Table 8.9 Wingspread Conference classification

Level of anomaly	Male	Female
High	1. Anorectal agenesis A. Rectovesical fistula B. Without fistula 2. Rectal Atresia	1. Anorectal agenesis A. Rectovaginal fistula B. Without fistula 2. Rectal Atresia
Intermediate	1. Rectourethral fistula 2. Anal agenesis without fistula	1. Rectovestibular Fistula, 2. Rectovaginal fistula 3. Anal agenesis without fistula
Low	1. Anocutaneous (perineal) fistula 2. Anal stenosis	1. Anovestibular (perineal) fistula, 2. Anocutaneous (perineal) fistula 3. Anal stenosis
Miscellaneous	Rare malformations	Persistent cloacal anomaly Rare malformations

complex, it allowed ARM to be viewed in a structured fashion. "Intermediate" was later added to this classification to describe those lesions where the rectum ends below the PC line.

In 1964 Santulli proposed his classification system, which was based on the work of Ladd and Gross. This also divided lesions into low, infralevator, and high, supralevator (Table 8.7) [66].

In 1970 the "International" classification was proposed at a symposium on Anorectal Malformations at the paediatric surgical congress in Melbourne in order to further decrease confusion (Table 8.8). Based on the early work of Smith and Stephens, the 1970 International classification was based on the principles of normal and abnormal anatomy and divided the lesions into three groups high (supralevator), intermediate and low (translevator). Although it proved much too complex for most surgical groups due to the fact it contained nearly 40 subtypes, it is still in use in the literature and is the primary reporting mechanism for the JSGA (Fig. 8.1) [55].

There are several terms that are used quite commonly in the classifications that can cause confusion for the modern reader. The term "covered anus" has been used to describe both the presence of the anal membrane and the cutaneous fistula. It was defined by Smith et al. in 1970 as a normally placed anus that is covered by excessive development of the genital folds or a fused congenital median band [58]. "Covered anus incomplete" is described as the commonest male anomaly, which is best described as a low perineal cutaneous fistula [58]. The "perineal groove" describes a normal vestibule but with a groove extending from the vestibule to the anus, which is both normal sized and positioned. The "perineal canal" is defined as a normal anus and vestibule with the presence of a fistula from the anal canal to the fossa navicularis [58].

The "International" classification also describes the anovulvar fistula as a variation of the covered anus with a fistula into the posterior fourchette. The vulvar anus was viewed as a variation of the anterior anus with so little development of the perineum that the anus, which is normal, is in the vestibule. The term rectocloacal fistula is used to describe all situations where the rectum, bladder and urethra enter a single channel as a common cloaca. The 1970 classification proposed that "rectovesical fistula" be used to describe the rectum entering the bladder between two separate vaginas to form a "common cloaca" at the bladder outlet [58].

The term "ectopic" anus still excites debate. It is used to describe a stenosed anus that has migrated to the vulva or, more commonly, the vestibule in the female, or any abnormally positioned anal opening in the male. It is also described as an anterior displacement of the anus and "anterior ectopic anus". It was on occasion also used to describe rectovaginal fistulae [67]. An ectopic anus is described as an anal index of less than 0.34 in girls and less than 0.46 in boys. The anal index is defined as the ratio of the scrotal–anal distance to the scrotal–coccygeal distance in males, and as the ratio of the fourchette–anal distance to the fourchette–coccygeal distance in females [68]. Kluth's embryological and histological evidence of hindgut development implies that any abnormal anal opening could be viewed as an anal fistula. Peña argues strongly from his experience of 1,460 patients that a normal anus surrounded by a normal voluntary

Fig. 8.2 Perineal fistula or an anterior "ectopic" anus

Fig. 8.3 Anterior "ectopic" anus or perineal fistula

sphincter in an abnormal position does not occur and that the term is overused (Figs. 8.2 and 8.3) [69].

New research and variations in surgical technique in the late 1970s and early 1980s altered previously fixed concepts. This led to the "Wingspread classification", which evolved from a conference held in the Wingspread Convention Center, Racine, Winconsin (USA) in 1984 [70]. It was created in order to update the "International" Melbourne classification that was described at the time as "unwieldy". It is based on high, intermediate and low anomalies and is presented in Table 8.9. Rarer subtypes that had cluttered the 1970 classification were removed. The cloacal lesion was also placed in its own separate class as the other class divisions depended on the length of the cloacal canal. The members of the workshop accepted that the classification would not be the final word on the issue, and indeed it is not.

It is well accepted that the Wingspread classification has not been fully endorsed and used within the surgical practice as it is based on anatomical principles [71]. Its important concept of a spectrum of disease are frequently alluded to and it is used to classify newly identified rarer anomalies such as translevator anal anomalies with cutaneous fistulae passing deep to the scrotum [72]. Yet even a perfunctory review of the literature reveals the ongoing use of the original Stephens, International and Wingspread classifications. This confusion has led to heated discussion on the need for even more classifications.

Peña argues that the terms "low", "intermediate" and "high" are arbitrary and not based on outcome, therapeutic management and prognosis. He further argues that other classification systems overcomplicate the issue leading to misdiagnosis and unnecessary surgery. Peña's classification is built on the concept of high and low lesions and their requirement for colostomy or not, and hence the primary principle is the management algorithm in the neonate. He argues that his classification is based on the anatomical defects and how they correlate with surgical management (Table 8.10).

A brief discussion on the subtypes on Peña's classification is necessary so that they can be compared to the other classification types. Male defects will be discussed initially. Cutaneous perineal fistula is the simplest ARM and has the lowest part of the rectum opening anterior to the sphincter. This can present in several manifestations in the male, which can cause confusion. In general, a midline fistula can appear

Table 8.10 Peña's classification

Males	Females
Perineal (cutaneous) fistula	Perineal (Cutaneous) fistula
Rectourethral fistula Bulbar Prostatic	Vestibular fistula
Rectovesical fistula	Persistent Cloaca
Imperforate anus without fistula	Imperforate anus without fistula
Rectal atresia	Rectal atresia

anywhere from the base of the penis to the midline raphe to just anterior to the centre of the sphincter. The anal membrane is also included in this group (Figs. 8.4 and 8.5).

Rectourethral fistula describes the rectum connecting to the urethra. The fistula most commonly enters the bulbar urethra, but can also enter the prostatic urethra. An important anatomical feature in the rectourethral fistula is the common wall between the fistula and the urethra. Rectovesical fistula involves the rectum opening into the bladder neck above the sphincter and bladder neck. On examination, the perineum is flat. The sacrum and pelvis can appear dysmorphic or underdeveloped (Fig. 8.6).

High imperforate anus without fistula implies that the rectum ends blindly at the level of the bulbar urethra without a fistula. ARM without fistula occurs in 5% equally in both males and females and is an uncommon presentation in the normal population. However, 50–95% of patients have trisomy 21 and the others tend to suffer from syndromes such as Apert [73,74].

Rectal atresia is a rare condition that occurs in 1% of all cases, although it is common in regions of

Fig. 8.4 Low anorectal lesion with fistula in the scrotum

Fig. 8.6 High lesion in the male with flat perineum

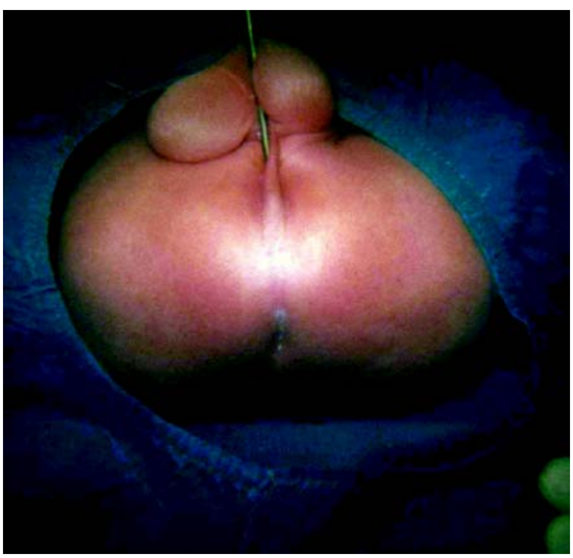


Fig. 8.5 Perineal fistula in a boy with probe in situ

Fig. 8.7 Imperforate anus with anterior fistula

southern India. It is frequently misdiagnosed due to the normal external appearance of the anus. The atresia lies approximately 1–2 cm above the perineum. There can be a thick fibrous band separating the rectum from the anus or a thin membrane with a small pinhole opening.

Female defects consist of several subtypes such as the cutaneous perineal fistula, which has an opening anterior to the sphincter and posterior to the introitus (Fig. 8.7). The vestibular fistula enters the vestibule superficial to the hymen. The vagina and the rectum contain a common wall and the fistula is of variable length. The opening can be difficult to see and requires careful inspection (Fig. 8.8).

Cloacal anomalies are a separate, complex, wide variety of malformations. Cloaca is derived from the Latin term for sewer or latrine and is defined as a common channel for the opening of the rectum, vagina and urinary systems. It is extremely important that it is correctly identified prior to definitive surgery. On examination of the perineum a single orifice is identified. The size of the introitus is smaller than in the normal female in most cases. Of note, the longer the common channel, the higher the defect and the more complex the lesions. Multiple subtypes of cloacal anomalies have been described and a full description of the cloacal anomaly is available in Chap. 10.

Peña argues that unusual and uncommon presentations should be viewed as complex malformations that require an individualised approach to each patient; hence, no generalised guidelines can be created [74].

In reality, whether one adheres strictly to the Wingspread or Peña's classification was a matter of personal choice. From a practical point of view, Peña's classification allowed a concise and appropriate clinical management structure. The Wingspread classification allowed the anatomical detail to be further elucidated. All descriptions of ARM require a detailed anatomical description of the lesion in order to illustrate the defect and allow comparisons between centres.

In May 2005 an international congress for the development of standards for the classification, treatment and follow up of ARM took place in Krickenbeck Castle in Westphalia, Germany. At this meeting the need for a new, unifying, international classification system that enabled everyone to talk the same language was quite clear. Thus was born the new standards for diagnostic procedures international classification system "Krickenbeck" (Table 8.11) [75]. This new classification system was reached by consensus within the symposium. It does not focus on anatomical or embryological features or on imaging. It is divided into two main groups "major clinical groups" and "rare/regional variants" and is based on frequency of occurrence and allows management outcomes to be measured. The addition of the "rare/regional variants" allows lesions that are less common in the Western world yet quite common in India and

Fig. 8.8 Vestibular fistula

Table 8.11 Standards for diagnostic procedures: International Classification (Krickenbeck) [69]

Major clinical groups	Perineal (cutaneous) fistula
	Rectourethral fistula
	Bulbar
	Prostatic
	Rectovesical fistula
	Vestibular fistula
	Cloaca
	No fistula
	Anal stenosis
Rare/regional variants	Pouch Colon
	Rectal atresia/stenosis
	Rectovaginal fistula
	H type fistula
	Others

the Far East to be included. For example, the H-type fistula, where together with a normally placed anal canal there is a fistulous communication between the anorectum and the genital tract, has an incidence of 3% in Finland but is found more commonly in India [76]. We accept that cloacae are uncommon, but due to the significant impact of the potential errors from an incorrect initial diagnosis it has been placed in the major group. Also, despite the controversy over anal stenosis, the symposium agreed to include it in the major group.

Beside the new international "Krickenbeck" standards for diagnostic procedures, an international grouping of surgical procedures for follow was developed at the Krickenbeck meeting (Table 8.12). This second standardisation seemed to be necessary to make the different surgical procedures comparable with each other. Perineal (cutaneous) or ano-vestibular fistulas could be operated either by a perineal operation or by an anterior sagittal approach (former Pott's procedure). The PSARP technique is used for prostatic and bulbar or recto-vestibular fistulas. To be able to compare the results of the different operations with the results of other authors, not only the type of the fistula or malformation has to be compared but also the type of the operation used. One should always keep in mind, that the postoperative results after the repair of anorectal malformations are strongly correlated to the extension of intraoperative mobilisation of the fistula and the blind pouch. This can be determined by the new international classifications for the diagnosis, the procedures and the new follow up scoring for postoperative results (see Chaps. 25 and 27).

It is therefore, envisaged that the new three Krickenbeck classifications will enable comparable follow up of patients with anorectal malformations.

Table 8.12 International grouping (Krickenbeck) of surgical procedures for follow up [75]

Operative procedures
• Perineal operation
• Anterior sagittal approach
• Sacroperineal procedure
• PSARP
• Abdominosacroperineal pull-through
• Abdominoperineal pull-through
• Laparoscopic-assisted pull-through
Associated conditions
• Sacral anomalies
• Tetherd cord

References

1. Stoll C, et al (1997) Risk factors in congenital anal atresias. Ann Genet 40:197–204
2. Chavez GF, et al (1988) Leading major congenital malformations among minority groups in the United States, 1981–1986. MMWR CDC Surveill Summ 37:17–24
3. Texas Birth Defects Registry Report of Birth Defects Among Deliveries (2000) Texas Department of State Health Services, 1100 West 49th Street, Austin, Texas
4. Harris J, et al (1995) Descriptive epidemiology of alimentary tract atresia. Teratology 52:15–29
5. Ameh EA, Chirdan LB (2000) Neonatal intestinal obstruction in Zaria, Nigeria. East Afr Med J 77:510–513
6. Ameh EA, et al (2001) Emergency neonatal surgery in a developing country. Pediatr Surg Int 17:448–451
7. Adeyemo AA, et al (1997) Major congenital malformations among neonatal referrals to a Nigerian university hospital. East Afr Med J 74:699–701
8. Louw JH (1965) Congenital abnormalities of the rectum and the anus. Curr Probl Surg 31:1–64
9. Spouge D, Baird PA (1986) Imperforate anus in 700,000 consecutive liveborn infants. Am J Med Genet Suppl 2:151–161
10. Nazer J, et al (2000) Anorectal congenital malformations and their preferential associations. Experience of the Clinical Hospital of the University of Chile. Period 1979–1999. Rev Med Chil 128:519–525
11. Garne E, et al (2002) Gastrointestinal malformations in Funen county, Denmark – epidemiology, associated malformations, surgery and mortality. Eur J Pediatr Surg 12:101–106
12. Czeizel A (1981) Birth prevalence of five congenital abnormalities of medium frequency in Budapest. Acta Paediatr Acad Sci Hung 22:299–308
13. Leck I, Lancashire RJ (1995) Birth prevalence of malformations in members of different ethnic groups and in the offspring of matings between them, in Birmingham, England. J Epidemiol Community Health 49:171–179
14. Cuschieri A; EUROCAT Working Group (2001) Descriptive epidemiology of isolated anal anomalies: a survey of 4.6 million births in Europe. Am J Med Genet 103:207–215
15. Hashmi MA, Hashmi S (2000) Anorectal malformations in female children – 10 years experience. J R Coll Surg Edinburgh 45:153–158
16. Chowdhary SK, et al (2004) An audit of neonatal colostomy for high anorectal malformation: the developing world perspective Pediatr Surg Int 20:111–113
17. Chadha R (2004) Congenital pouch colon associated with anorectal agenesis. Pediatr Surg Int 20:393–401
18. Rao KL, Menon P (2005) Congenital pouch colon associated with anorectal agenesis (pouch colon syndrome). Pediatr Surg Int 21:125–126

19. Sharma AK, Harjai MM (1997) Simplified colorrhaphy and posterior sagittal anorectoplasty for the management of congenital short colon with imperforate anus. Br J Surg 84:389
20. Yang JH, et al (2004) A multi-center study for birth defect monitoring systems in Korea. Korean Med Sci 19:509–513
21. Borman B (2000) Annual report with data for 2000 of the International Clearinghouse for birth defects monitoring systems. Published by The international centre for birth defects Roma, Italy
22. Tong MC (1981) Anorectal anomalies: a review of 49 cases. Ann Acad Med Singapore 10:479–484
23. Riley MM, et al (1998) Congenital malformations in Victoria, Australia, 1983–95: an overview of infant characteristics. J Paediatr Child Health 34:233–240
24. Cho S, et al (2001) One hundred three consecutive patients with anorectal malformations and their associated anomalies. Arch Pediatr Adolesc Med 155:587–591
25. Haeusler MC, et al (1994) Prenatal ultrasonographic detection of gastrointestinal obstruction: results from 18 European congenital anomaly registries. Prenat Diagn 22:616–623
26. Finley WH, et al (1994) Birth defects surveillance: Jefferson County, Alabama, and Uppsala County, Sweden. South Med J 87:440–445
27. Stoll C, et al (1996). Evaluation of prenatal diagnosis of congenital gastro-intestinal atresias. Eur J Epidemiol 12:611–616
28. Zlotogora J, et al (1989) Comment on Anorectal malformations and Down syndrome. Am J Med Genet 34:330–331
29. Buchin PJ, et al (1986) Down's syndrome and the gastrointestinal tract. J Clin Gastroenterol 8:111–114
30. Joseph VT, et al (1985) Anorectal malformations and their associated anomalies. Ann Acad Med Singapore 14:622–625
31. Cuschieri A, EUROCAT Working Group (2002) Anorectal anomalies associated with or as part of other anomalies. Am J Med Genet 110:122–130
32. Hassink EA, et al (1996) Additional congenital defects in anorectal malformations. Eur J Pediatr 155:477–482
33. Shapiro BL (2003) Down's syndrome and associated congenital malformations. J Neural Transm Suppl (67):207–14
34. Black CT, Sherman JO (1989) The association of low imperforate anus and Down's syndrome. J Pediatr Surg 24(1):92–94
35. Anderson RC, Reed SC, (1954) The likelihood of recurrence of congenital malformations. Lancet 74:175–176
36. Manny J, et al (1973) Congenital familial anorectal anomaly. Am J Surg 125:639–640
37. Kaijser K, et al (1957) Ano-rectal abnormalities as a congenital familial incidence. Acta Paediatr 46:199–200
38. VanGelder DW, Kloepfer HW (1961) Familial anorectal anomalies. Pediatrics 27:334–336
39. Seitz A, Bautze HJ (1951) Familiar anus vestibularis. Zentralbl Gynakol 73:1543–1545
40. Mittal A, et al (2004) Associated anomalies with anorectal malformation. Indian J Pediatr 71:509–514
41. Cook RCM (1990) Anorectal malformations. In: Lister J, Irving I (eds) Neonatal Surgery, 3rd edn, Butterworth, London, pp 547–570
42. Schwoebel MG, et al (1984) Familial incidence of congenital anorectal anomalies. J Pediatr Surg 19:179–182
43. Christensen K, et al (1990) An epidemiological study of congenital anorectal malformations: 15 Danish birth cohorts followed for 7 years. Paediatr Perinat Epidemiol 4:269–275
44. Stephens FD (1988) Anorectal Malformation in Children Update 1988, Birth Defects Original Article Series vol 24 (4). John Wiley and Sons
45. Currarino G, et al (1981) Triad of anorectal, sacral, and presacral anomalies. Am J Roentgenol 137:395–398
46. Gegg CA, et al (1999) An unusual case of the complete Currarino triad: case report, discussion of the literature and the embryogenic implications. Neurosurgery 44:658–662
47. Riebel T, et al (1999) The spectrum of imaging in Currarino triad. Eur Radiol 9:1348–1353
48. Boocock GR, Donnai D (1987) Anorectal malformation: familial aspects and associated anomalies. Arch Dis Child 62:576–579
49. Javid PJ (1998) Immediate and long-term results of surgical management of low imperforate anus in girls. J Pediatr Surg 33:198–203
50. Ratan SK, et al (2004) Associated congenital anomalies in patients with anorectal malformations – a need for developing a uniform practical approach. J Pediatr Surg 39:1706–1711
51. Metts JC, et al (1997) Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. J Urol 158:1298–1300
52. Cortes D, et al Cryptorchidism in boys with anal atresia. Ugeskr Laeger 158:2845–2848
53. Qi BQ, et al (2003) Evidence that the notochord may be pivotal in the development of sacral and anorectal malformations. J Pediatr Surg 38:1310–1316
54. Otte JB (1983) Imperforate anus. Various Belgian epidemiologic data. Acta Chir Belg 82:158–162
55. Endo M, et al (1999) Analysis of 1,992 patients with anorectal malformations over the past two decades in Japan. Steering Committee of Japanese Study Group of Anorectal Anomalies. J Pediatr Surg 34:435–441
56. Rich MA, et al (1988) Spectrum of genitourinary malformations in patients with imperforate anus. Pediatr Surg Int 3:110–113
57. Archibong AE, Idika IM (2004) Results of treatment in children with anorectal malformations in Calabar, Nigeria. S Afr J Surg 42:88–90
58. Stephens FD, Smith ED (1971) Anorectal Malformation in Children, Year Book Medical Publications, Chicago

59. Chen CJ (1999) The treatment of imperforate anus: experience with 108 patients. J Pediatr Surg 34:1728–1732
60. Kiesewetter WB, Chang JH (1977) Imperforate anus: a five to thirty year follow–up perspective. Prog Pediatr Surg 10:111–120
61. Kiesewetter WB, et al (1964) Imperforate anus. A review of sixteen years of experience. Am J Surg 107:412–421
62. Peña A (2003) Anorectal malformation. In: Ziegler MM, Azizkhan RG, Weber TR (eds) Operative Pediatric Surgery. McGraw-Hill Professional, pp 739–762
63. Amussat JJ (1835) Observation sur une operation d'anus artifical pratiquee avec success par un nouveau procede, Gaz Med, Paris
64. Ladd WE, Gross RE (1934) Congenital malformations of rectum and anus: report of 162 cases. Am J Surg 23:167–183
65. Wangensteen OH, Rice CO (1930) Imperforate anus. A method of determining the surgical approach. Ann Surg 92:77–81
66. Santulli TV, et al (1964) Malformations of the anus and rectum. Surg Clin North Am 45:1253–1271
67. Nixon H, O'Donnell B (1976) The Essentials of Paediatric Surgery, 3rd edn. William Heinemann Medical Books, pp 28–36
68. Herek O, Polat A (2004) Incidence of anterior displacement of the anus and its relationship to constipation in children. Surg Today 34:190–192
69. Holschneider, et al (2005) Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. J Pediatr Surg 40:1521–1526
70. Peña A (2004) Comments on anterior ectopic anus. Pediatr Surg Int 20:902
71. Stephens F.D, Durham-Smith E (1986) Classification, identification, and assessment of surgical treatment of anorectal anomalies. Pediatr Surg Int 1:200–205
72. Holschneider AM, et al (2002) Surgical methods for anorectal malformations from Rehbein to Peña – critical assessment of score systems and proposal for a new classification. Eur J Pediatr Surg 12:73–82
73. Fitzgerald RJ, et al (2002) Translevator anal anomalies with cutaneous fistulae passing deep to the scrotum. J Pediatr Surg 37:1326–1329
74. Torres R, et al (1998) Anorectal malformations and Down's syndrome. J Pediatr Surg 33:194–197
75. Peña A (2000) Imperforate anus and cloacal anomalies. In: Ashcraft MD (ed) Pediatric Surgery, 3rd edn. WB Saunders, London, pp 473–492
76. Rintala RJ, et al (1996) H-type anorectal malformations: incidence and clinical characteristics. J Pediatr Surg 31:559–562

9 The Clinical Features and Diagnostic Guidelines for Identification of Anorectal Malformations

Elhamy Bekhit, Feilim Murphy, Prem Puri, and John M. Hutson

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9.1 Clinical Features

The diagnosis of anorectal malformations (ARM), with the exception of anal stenosis, should be made shortly after birth during the routine neonatal examination. The vast majority of lesions will be detected even after the most cursory examination of the perineum. However, rectal atresia in isolation may not be detected until later. In regions of the world where early neonatal review is not practiced, such as sub-Saharan Africa, ARM presents commonly with gross abdominal distension, bowel obstruction, and sepsis on the 4th day of life [1, 3].

The importance of a complete medical history should not be neglected in order to focus on the examination of the perineum. There is always time to do a detailed evaluation and examination. The family, antenatal, and birth history should be obtained. Stephens and Smith found no relationship between the age of the mother and the birth of the affected child [44]. Cushieri reports that odds ratios for mothers above 35 years old were increased for supralevator atresia without fistula, supralevator atresia with fistula, and congenital anal fistula [12]. Maternal recreational drug use and parental consanguinity have been blamed as risk factors for ARM [45]. An association with maternal residence at high altitude and paternal occupation in vehicle manufacture has also been described [27].

9.1.1 Prenatal Diagnosis

Prenatal ultrasonography has a low sensitivity and specificity for the detection of ARM. A normal anus is visualized as a circular rim of hypoechogenicity in the perineum together with a central linear echogenic stripe. The absence of this circular rim is described as imperforate anus on the prenatal scan. Enterolithiasis has been described on prenatal scans of rectovesical or rectourethral fistula in patients with ARM [2]. Harris retrospectively viewed prenatal scans of children with ARM and demonstrated dilated colon on prenatal ultrasonograms [16]. Oligohydramnios and a highly distended vagina are signs of imperforate anus on ultrasound. However, due to the frequency of concurrent anomalies associated with ARM it is these other lesions that are usually diagnosed on the prenatal scan. Polyhydramnios associated with an upper gastrointestinal obstruction, sacral anomalies, and vertebral and renal defects are much more readily seen on ultrasonography than an imperforate anus. At birth, a history of excess oral secretions and central cyanosis implies the potential presence of VACTERL (Vertebral, Anorectal, Cardiac, Tracheo-Esophageal, Renal and Limbs particularly radial) anomalies.

9.1.2 Neonatal Examination

There are several important clinical features that must be elicited on perineal examination, such as the presence or absence of the anus, the presence of vaginal and urethral openings, the presence and exact position of the fistula, the size of the anal dimple and midline groove, and the presence of a presacral mass. Due to the VACTERL condition, a full and detailed examination of the entire child is mandatory after the discovery of an ARM. Particular stress must be placed on the examination of the spine, the pelvis, the esophagus, and the cardiovascular system to ensure a life-threatening abnormality has not been overlooked because of an absent rectum.

In order to identify anomalies correctly, a detailed understanding of the normal anatomy of the perineum is required. On inspection of the perineum the position of the anus or its absence should be noted. The normal position of the anus in a girl is one third of the distance from the coccyx to the fourchette. If the anus is present and in the correct position the passage of a soft catheter greater than 2 cm into the rectum and the presence of meconium passage rules out atresia. The parasagittal muscle fibers are located on either side of the midline of the normal anal position. These fibers are responsible for the midline buttock groove. The higher the fistula in the urethra, the fewer the parasagittal fibers present, hence the flatter the perineum appears. The midline groove between the buttocks and the anal dimple also becomes less prominent the higher the fistula (Fig. 9.1).

Anal stenosis is a subjective diagnosis that is frequently made following a "tight" digital examination. The normal size of the anus is described as $1.3 + (3 \times \text{birth weight in kg})$ in millimeters.

In the presence of an anal membrane, the anus is usually in the normal position. However, a thin epithelial lining is present overlying the opening. Meconium can be seen bulging behind the membrane.

In boys, the midline raphae and the scrotum need to be examined for a fistulous opening. The urethral meatus is assessed for the presence of meconium staining, which occurs with rectourethral fistula (Fig. 9.2). A damp clean swab can be left at the meatus to assess for meconium staining and microscopy should be performed on the urine. The presence of meconium or squamous epithelium in the urine indicates a fistula into the urinary system. Other lesions that can occur include a midline subepithelial tract along which meconium can be milked or a midline skin tag described as a "bucket handle". On occasion a thin membrane (called the anal membrane) obstructs the meconium, which can be seen behind it.

In females a key step is determining the number of orifices present. The presence of the hymen and the small size of the newborn perineum can make this quite challenging and leads to significant confusion. Three frequently made errors are failure to diagnose the presence of a single orifice only (cloaca) and failure to visualize the vestibular fistula or to label ves-

Fig. 9.1 Flat perineum associated with a high anomaly

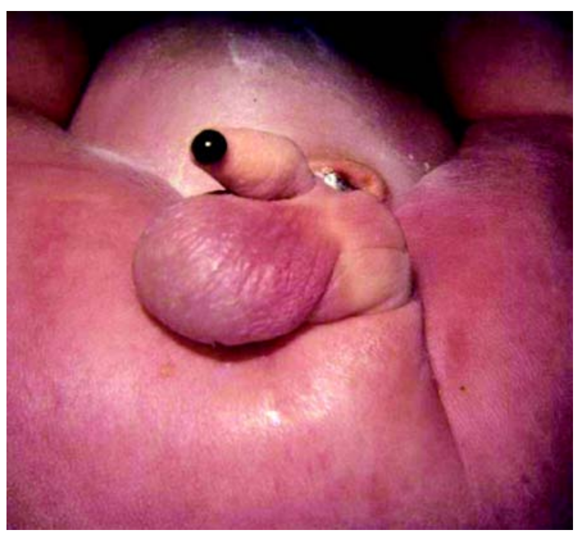

Fig. 9.2 Meconium at the urethral meatus, demonstrating presence of a fistula

tibular fistulas as vaginal. Careful inspection with a good light source is necessary to avoid these pitfalls. If the size of the introitus appears smaller than in a normal female, the possibility of a cloacal anomaly is very high. On probing a rectovestibular fistula with a fine forceps, the probe will pass posteriorly and backwards. The probe in a rectovaginal fistula will pass only upwards and this has long been stated as a method of differentiating between the two.

Rosen et al. in 2002 assessed the incidence of rectovaginal fistula and demonstrated the importance of the correct initial diagnosis. A review of 617 patients over a 20-year period revealed that only 6 (1%) had a true rectovaginal fistula. Of a total of 139 patients who were referred after a previous repair, 42 had a diagnosis of rectovaginal fistula. The diagnosis was incorrect in all cases. Twelve patients had a rectovestibular fistula, and 30 had a cloaca with a persistent urogenital sinus requiring a second complete repair [34].

In the rare female with a perineal groove anomaly, the anus is not stenotic but there is a mucosal strip extending through a slightly bifid perineal body to the introitus. In the “perineal canal” variant of an H-fistula in girls the anus is patent (± stenosis). Deep in the fourchette is a fistulous orifice and mucosal-lined canal running through the perineal body to the anterior wall of the anal canal.

9.2 Diagnostic Guidelines

The initial management of a newborn baby with ARM should be made only after accurate determination of the exact type and level of the anomaly. A decision can then be made as to whether to do a primary perineal operation, or to perform a colostomy, deferring definitive repair. In addition to determining the exact level and anatomical type, further information is required, such as the integrity of the neuromuscular components of the pelvis and the presence of any associated anomalies, particularly in the urinary tract. Most of the information required can be determined from the clinical observations of the baby and a radiological study of the pelvis, along with renal, spinal, and cardiac ultrasound. The aims of the initial assessment are threefold:

1. To determine the level of the malformation in relation to the muscular sphincters and the site of any fistulous communications.
2. To determine the integrity of sphincters and their nerve supply.
3. To document any associated anomalies that may affect survival.

9.3 Assessment of the Level of Anomaly and Presence of a Fistula

The internal anatomy is predicted by the clinical examination, “invertogram” radiology, the presence of gas on radiology in other viscera, radiopaque contrast studies of the fistula, urinary tract or the bowel, ultrasound examination of the abdomen, pelvis and spine, computed tomography (CT) and magnetic resonance imaging (MRI), and finally the occasional use of endoscopy. Under anesthesia, the use of a muscle stimulator can give a good assessment of the sphincter muscles, but this will be discussed in other chapters. The emphasis in physical diagnosis varies between the sexes and is therefore described separately.

9.4 Male Prediction from Clinical Appearance

The perineal appearance should be documented with photography and recorded on a standard assessment form. An orifice visible externally in the perineum predicts a translevator anomaly, except in a few anomalies such as rectal atresia (see Chap. 12). There may be a normal anus, although displaced anteriorly in the perineum, as in anterior perineal anus. In the case of a perineal fistula, the opening may be difficult to locate, especially if it is associated with genital fold anomalies. A careful search should be made for a minute orifice along or beside the perineal raphae from behind the normal site to the tip of the penis. A speck of meconium or a bubble of gas may direct attention to the orifice. The fistulous track may be suggested by a fine bluish line, a bulge of meconium, or white epithelial pearls along its course. In some forms of cutaneous perineal fistula, the opening is in the folds at the normal anal site. In the rare anomalies of rectal atresia and imperforate anal membrane or anorectal stenosis, the superficial appearance of the anus may be normal, but the patients may have ribbon stools, and a stricture on rectal examination assists in the diagnosis.

Where there is no opening in the perineum it is not possible to predict the level of the anomaly with certainty, although most will have rectal lesions. There is nothing distinctive about the perineal shape, which may possess a raphe, hypertrophied genital folds, a dimple, or a pigmented patch. The diagnosis of a very low ARM without fistula can be suspected from the presence of hypertrophied genital folds or a median band over the normal anal site, especially if there is a bluish tinge of meconium through the skin and bulging of the anal area on abdominal compression.

In many patients with rectobulbar urethral fistula, the diagnosis may be suspected because of an associated genital anomaly in the anterior part of the perineum, such as atypical hypospadias, cleft scrotum, a slender perineum, or a flimsy floor to the urethra.

Palpation of the urethra may produce meconium at the urethral meatus, and if urine is collected meconium is likely to be present. If urine is not passed, a catheter should be inserted and left in place for subsequent radiological studies. Where no meconium is present in the urine, either a fistula is blocked with meconium or there is a noncommunicating abnormality such as ARM without fistula. Further imaging is required to identify the specific abnormality in this case.

Where meconium is present in the urine, the rectum is confirmed to communicate with the urinary tract. This occurs in rectoprostatic urethral fistula, rectobulbar urethral fistula, and rectovesical fistula. All three versions usually require initial colostomy prior to definitive treatment. Prone cross-table lateral x-rays and delayed contrast studies including pressure-augmented colostogram and micturating cystourethrography (MCUG) are useful.

9.4.1 Inversion Radiography

Wangensteen and Rice [50] first described the use of inversion radiography in 1930 to indicate the distance between the gas bubble within the terminal colon and the perineal skin. Many authors [6, 8, 11, 13, 19, 22, 29, 39, 48] then subsequently related the distance from the skin to the underlying abnormality, although the measurement of the distance alone does not provide the essential information required, which is the relationship of the blind-ending rectum to the levator ani and sphincter muscle complex.

An upside-down inversion x-ray is no longer performed, having been replaced by a prone, cross-table lateral examination of the pelvis, with the hips elevated over a bolster [26]. This examination allows for easier positioning of the patient and better delineation of the anatomy. Use of bony landmarks on the pelvic x-ray allows the clinician to relate the gas bubble to the origin of the levator ani and the apex of the levator sphincter muscle complex near the lower point of the ischium.

The pubococcygeal line (PC line) and the ischial (I) point were determined by Stephens [40–43] from dissection of 25 stillborn pelves in documenting the attachment of the levator ani to the pelvic wall both macroscopically and radiologically, with opaque wires marking the attachment of the levator ani. Using these techniques the PC line stretches from the upper border of the symphysis pubis to the sacrococcygeal junction. At the symphyseal end this line is taken as the center of the "boomerang" shape of the os pubis, which corresponds with the upper border of the symphysis. The top of the "boomerang" corresponds with the superior pubic ramus. Ossification of the pubis begins in the second fetal month and extends along the superior pubic ramus medially to the body.

At the coccygeal end, the C point is just caudal to the last or fifth ossific center of the sacrum. The ossification center of this fifth piece normally appears at the 4th month of fetal development. The coccyx, by contrast, does not ossify until 2–5 years after birth. If the caudal segments of the sacrum are deficient, the PC line can be developed by projection from the pubis through the same site on the ischium, which is approximately the junction between the upper quarter and the lower three-quarters. The soft tissues lying at the level of the PC line are the bladder neck, the verumontanum, the pelvic reflection from the rectum to the prostate, and the external os of the cervix.

In assessing the gas bubble in an ARM, its relation to the PC line defines the essential factor of whether the blind pouch is above or below the attachment of levator ani to the pelvic wall (Fig. 9.3). The ischial line (I line) and I point are related to the ossification center of the ischium, which is a comma shape in the neonate. The I point is demarcated on the x-rays at the inferior end of the ischial comma. The I line is drawn through the I point parallel to the PC line and corresponds to the upper surface of the bulb of the urethra in the male and the upper limit of perineal body and the level of the triangular ligament in the female. The anal pit is normally 1–2 cm caudal to the ossified ischium.

Kelly [18] has demonstrated the extent of the attachments of the levator ani in dissections of neonatal pelvis. Using wires from the specimens and then taking x-rays, he was able to identify the attachments of the levator ani and the apex of the funnel-shaped muscle complex in the normal neonate. Kelly also dissected the pelves in neonates with rectal and anal anomalies and found that the origin of the levator muscles was constant and followed closely the PC line in both normal patients and those with malformations of the rectum. The triangle bounded by the PC line and the I point denotes the radiographic markings of the levator complex in babies with a significant ARM, with the bowel terminating above the sphincter muscle complex. In Kelly's study group, in those with rectoprostatic urethral fistulae, the wire marking the

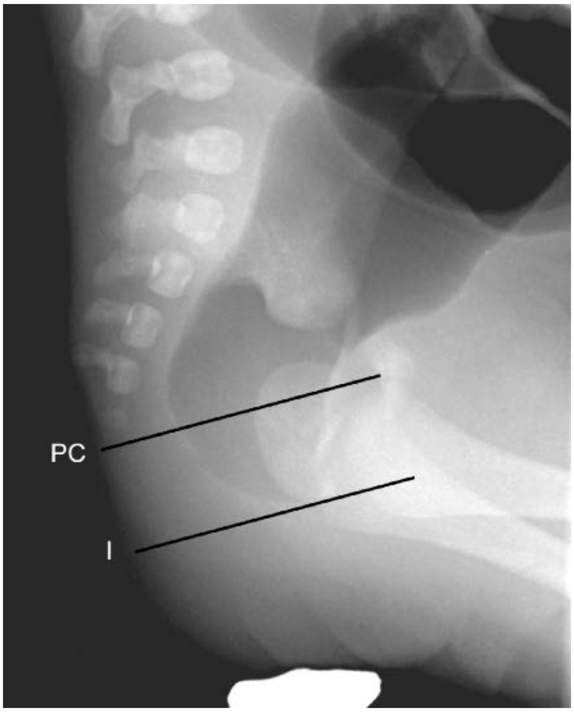

Fig. 9.3 Prone, cross-table lateral radiograph of a pelvis demonstrating the pubococcygeal (*PC*) line between the pubic symphysis and coccyx, and the ischial (*I*) line running parallel to the PC line at the inferior aspect of the ischium. In the this example the terminal bowel gas extends to the I point in a child with a rectoprostatic urethral fistula

levator ani demonstrates the extent of the puborectalis and the bowel opening into the back of the urethra well above the I point. The fistula is at or very close to the PC line. In perineal anomalies the gas bubble on radiology extends well below the I point to within a very short distance from the skin.

A true lateral view of the pelvis with accurate centering on the greater trochanter is essential. It ensures that the ossification centers of the pubic bones are superimposed and readily recognized and that the two ischial bones are accurately superimposed, appearing as one, with the anal dimple and natal cleft outlined with barium or a radiopaque marker correctly aligned. In these circumstances the PC line can be drawn accurately and the visceral anatomy interpreted. The common error, centering on the middle of abdomen, results in an angled projection of the right and left ischial bones. This causes distortion of the picture and difficult interpretation. The hips should be slightly extended so that the femoral shadows are clear of the pubic ossification centers.

The prone, cross-table lateral x-ray should be delayed for 12–24 h after birth to allow gas to reach the distal rectum. The baby should be placed in the prone position for 3 min before taking the film to allow gas to displace meconium and rise to the termination of the pouch. Barium paste or contrast-soaked gauze placed in the natal cleft is more accurate than a metal marker. A catheter may be placed in the urethra to make delineation of the urethra more obvious, although this is not essential. The greater trochanter should be marked with a marker pen on the upper thigh and the x-ray beam centered on this spot.

Common causes for erroneous interpretation of prone, cross-table lateral x-ray include:

1. Insufficient time for gas to reach the terminal bowel.
2. Meconium plug in the terminal gut may produce an erroneously high shadow if the gas does not displace the meconium.
3. Active contraction of the levator ani/sphincter muscle complex can push the gas shadow higher.
4. Gas escape through a fistula may confuse the x-ray, but clinical diagnosis should then be obvious.
5. Distortion by x-ray magnification resulting in the appearance of a longer gap between the gas within the terminal colon and skin.
6. Inappropriate placement of an anal marker may cause an error of assessment of the exact site of the anus on the skin.
7. Erroneous estimation of level of the lesion inside the sphincter muscle complex may occur if the pelvic floor muscles are relaxed, or if there is a sacral anomaly.
8. Finally, gas in the vagina may be mistaken for gas in the distal bowel.

9.4.2 Contrast Studies

9.4.2.1 Pressure-Augmented Colostogram

Beyond the neonatal period, in those who have undergone a diverting colostomy, the anatomy of the terminal colon and fistula can be accurately depicted fluoroscopically by performing a pressure-augmented colostogram. This method was first described by Cremin et al. [10] in 1972 and has subsequently been studied by Lernau et al. [21] and others [14, 46, 49], showing that it is a safe technique that can successfully demonstrate the presence or absence of a fistula in all published cases. The investigation involves the injection of water-soluble contrast medium into the distal limb of the colostomy using a balloon catheter. The balloon is inflated and traction applied to the catheter to occlude the stoma. Water-soluble contrast medium is then injected under constant gentle pres-

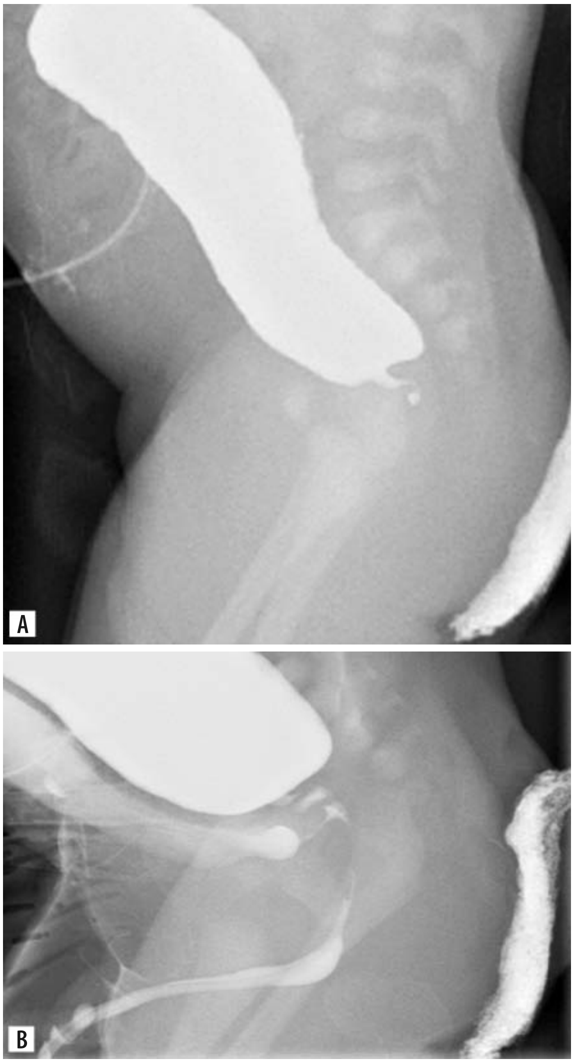

Fig. 9.4 Pressure-augmented colostogram. The initial fluoroscopic image (**A**) demonstrates the nipple arising from the terminal colon. Following further injection of contrast medium (**B**), a rectoprostatic urethral fistula is demonstrated. A second catheter has been placed in the urethra to help demonstrate the bladder and urethra proximal to the fistula

Fig. 9.5 **A** Micturating cystourethrogram demonstrating a rectoprostatic urethral fistula. **B** Retrograde urethrogram performed following a micturating cystourethrogram on withdrawal of the urinary catheter, demonstrating a rectoprostatic urethral fistula

sure until a fistula is filled, and visualized with the aid of fluoroscopy. The presence of a fistula is initially indicated by the appearance of a nipple of contrast arising from the terminal colon (Fig 9.4).

9.4.2.2 Micturating Cystourethrography

MCUG should be performed in all neonates with an ARM due to the high incidence of renal tract anomalies in these patients. This can be used to assess for the presence of associated vesicoureteric reflux. The antegrade urethrogram component of the study, together with a retrograde urethrogram performed whilst removing the catheter, may demonstrate the site of a rectourinary fistula. This is not as reliable at demonstrating a fistula as a pressure-augmented colostogram, however [21]. The MCUG can be combined with the pressure-augmented colostogram by filling the bladder via the colostogram until an episode of voiding is achieved (Fig. 9.5). Even if a fistula is not identified on MCUG, there may be the impression

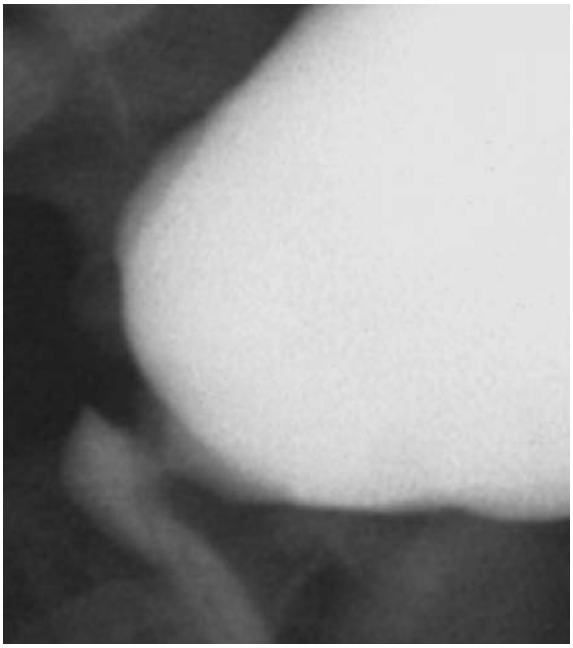

Fig. 9.6 Voiding phase of a micturating cystourethrogram demonstrating no urinary fistula; however, there is an area of acute angulation and tenting of the posterior urethra, suggestive for the presence of a fistula

Fig. 9.7 Axial plane, infracoccygeal ultrasonogram demonstrating the levator ani (*straight arrows*) with the colon (*curved arrow*) passing through the levator sling in a normal child

of an abnormal communication by the presence of a region of acute angulation or tenting of the urethra (Fig. 9.6).

An alternative to this is a "flushing technique" first described by Shopfner [38] in 1965, where the urethra is flushed with water-soluble contrast medium in a retrograde manner whilst a Foley catheter is in situ. The balloon of the Foley catheter is used to occlude the bladder neck by applying gentle traction, to stop contrast from entering the bladder. This may prove difficult, however, due to the size of these patients.

9.4.2.3 *Other Contrast Studies*

Contrast studies by direct puncture of the distal bowel via the perineum may be performed in some cases to identify the level of the terminal colon, although pressure-augmented colostograms, MCUG, and retrograde urethrography, as well as cross-sectional imaging have mostly obviated the need for this.

9.4.3 Ultrasound

Several authors [28, 37, 52] have postulated the use of ultrasound to determine the position of the terminal colon. Initial reports involved scanning from the anal dimple and measuring the distance between the terminal colon and the skin at the anal dimple site. This provides similar information to the prone, cross-table lateral x-ray; however, like that study it also has limitations, including the application of too much pressure at the anal dimple distorting the distance to the terminal colon and the colon being difficult to visualize if decompressed by surgery or a large fistula.

Recently, a new infracoccygeal scanning technique has been described [15], looking at the pelvic floor in an axial plane whilst directly visualizing the terminal colon and its relationship to the pelvic floor (Fig. 9.7). This technique may prove to be useful in determining the position of the terminal colon with respect to the levator sling. It cannot demonstrate the presence or site of any associated fistula. Whilst this technique may prove itself useful in the future, it will require time before pediatric radiologists and surgeons alike are comfortable with the procedure, and its efficacy is established.

9.4.4 Computed Tomography

Current multidetector CT allows for multiplanar imaging; this, however, comes at a radiation cost to the patient. CT provides increased detail, particularly of the osseous structures, as well as being able to demonstrate the pelvic floor musculature. There is difficulty, however, in distinguishing meconium from the rectal wall and adjacent musculature, due to limitations in contrast resolution. This limits its usefulness in detecting the site of any fistula, although it has been used to demonstrate the level of the terminal colon [17, 20].

9.4.5 Magnetic Resonance Imaging

MRI provides the same multiplanar capability as CT but without the radiation burden. Its improved contrast resolution also allows easy differentiation of meconium from the rectal wall and levator musculature [24]. In the neonatal period, meconium is easily visualized on MRI as hyperintense material on T1-weighted imaging, due to its lipid content (Fig. 9.8) [9].

The role of MRI in the assessment of the level of anomaly and presence of a fistula in neonates, however, is unclear as their small body size impedes spatial resolution. MRI can accurately detect the level of the terminal bowel, and the state of the pelvic floor musculature. In the newborn period prior to any surgery the fistula may be visualized in approximately 20% of cases [24]. In older children, due to their larger size and therefore improved resolution, this improves slightly, approximately one-third of fistulae being identified on MRI (Fig. 9.9) [4]. This can be improved in children who have undergone a colostomy by the use of an oil-based contrast agent such as Vaseline oil within the distal colon [46]. Pressure-augmented colostograms, however, are still more sensitive at detecting any associated fistula [4, 24, 46].

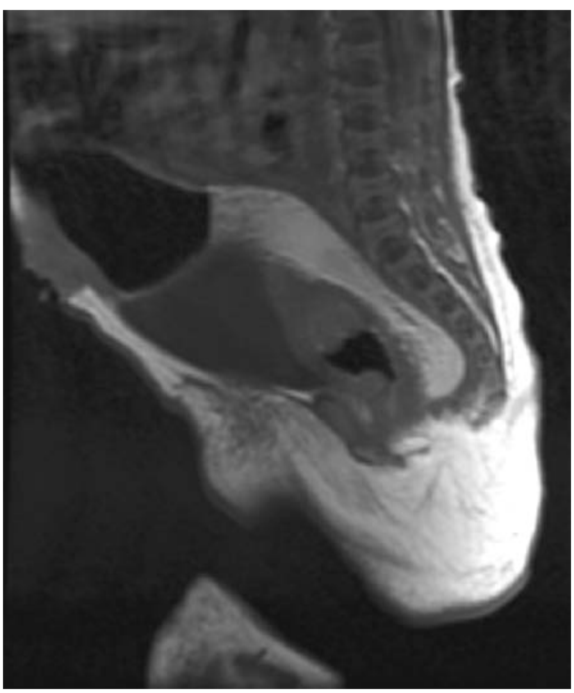

Fig. 9.8 Sagittal T1-weighted magnetic resonance image of the pelvis in a 2-day-old neonate, demonstrating the hyperintense meconium within the colon

9.5 Female Prediction from Clinical Appearance

Even more than the male, the internal anatomy of the female can nearly always be predicted from a careful study of the visible orifices in the perineum. Non-communicating anomalies are rare in the female, so that some clinical evidence for a fistulous communication can nearly always be determined by physical examination for the emission of meconium or gas. A photograph of the perineal anatomy is useful for the record as a basis for future comparison. There may be some fusion of the labial folds, preventing adequate inspection of the vulva. The possibility of a cloacal abnormality is very high particularly if the size of the introitus appears smaller than in the normal female.

The site, number and configuration of all of the orifices should be carefully documented by inspection and with a probe. Special attention should be given to the vestibule, where a small orifice may be hidden under the overhanging fourchette. When only one orifice is present in the vulva, a cloacal abnormality is predicted. Meconium and urine issue from the common orifice and colostomy is necessary. The precise anatomy can then be determined later by endoscopy or contrast studies through the common channel or the colostomy.

Where two orifices are present in the vulva, one of these can be predicted to be the urethra. Meconium issuing from the second orifice indicates the presence of a rare rectovaginal fistula. Inspection alone cannot distinguish between high or low communication of the rectal fistula into the vagina. Care should be taken to ensure that this is not actually a cloacal anomaly. Two orifices in the vulva but a bulging of the anal skin, particularly on abdominal compression suggests a rare covered anus.

Where there are three orifices in the vulva and perineum and the urethral and vagina orifices are normal, the third orifice is likely to be a fistulous opening or abnormal anus. If there is an opening at the normal anal site there can be seven different anomalies: perineal groove, perineal canal, imperforate anal mem-

Fig. 9.9 A series of axial T2-weighted magnetic resonance images in a 5-week-old male. The images were taken at a level just above (**A**), at (**B**), and just below (**C**) the pubococcygeal plane. Initially the rectum (*curved arrow*) lies posterior to the urethra (*straight arrow*). On the image below the level of the pubococcygeal plane (**C**) a rectoprostatic urethral fistula (*arrowhead*) is demonstrated

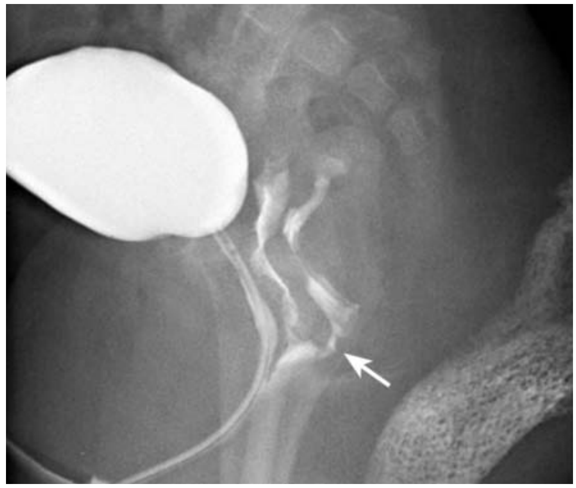

Fig. 9.10 A 3-month-old female with a normal-appearing anus, but also passing fecal material per vagina. Contrast study (sinugram) demonstrating passage of contrast from the vagina posteriorly into the rectum via a perineal canal (*arrow*)

brane, anal membrane stenosis, covered anal stenosis, anorectal stenosis, and rectal atresia.

In the case of a perineal groove, the anus is not stenotic but there is a mucosal strip extending through a slightly bifid perineal body to the introitus. With a perineal canal the anus may or may not be stenotic, but is patent and normal if at the normal anal site. Deep in the fourchette are a fistulous orifice and a canal running through the perineal body to the anterior wall of the anal canal (Fig. 9.10).

If there is an opening anterior to the anal dimple in the perineum, two deformities may be present, either anterior perineal anus or a cutaneous fistula. If there is an orifice outside the vagina but inside the vestibule, then the abnormality may be vulvar anus, anovular fistula, or a vestibular fistula. A rectovestibular fistula sometimes requires a different management approach. Anovestibular fistula can be distinguished from rectovestibular fistula by passing a probe through the orifice and seeing if this can be directed toward the coccyx. By contrast, in the case of a rectovestibular fistula, the probe only passes cranially along the posterior wall of the vagina.

9.5.1 Inversion Radiography

Prone, cross-table x-rays are needed only occasionally in the female, as noncommunicating abnormalities are rare; however, the technique and interpretation is otherwise the same as for males.

9.5.2 Contrast Studies

9.5.2.1 *Micturating Cystourethrography*

As in the male, this examination is useful in documenting associated urinary abnormalities. It is rare, however, for a female to have a rectourinary communication (See Chap. 10 and 14).

9.5.2.2 *Cloacagram*

In a female child with a clinical cloacal malformation, water-soluble contrast medium can be instilled into the cloaca to determine the anatomy and length of the common channel (Fig. 9.11). Multiple catheters may be required to outline the genital and urinary components with contrast medium. Alternatively, a balloon catheter can be used, with the balloon inflated outside the patient and applied to the cloacal opening to occlude it. If contrast does not reflux into the colonic component and a colostomy is present, then a pressure-augmented colostogram can be performed, as in a male.

Barium paste or contrast-soaked gauze can be applied to the perineum to help determine the length of the common channel. An alternative method is to place a radiopaque marker of known dimensions at the cloacal orifice, which can then be used as a scale to directly measure the length of the common channel with minimal error from distortion by x-ray magnification.

9.5.2.3 *Other Contrast Studies*

As for males, radiological imagining with contrast medium injected into the fistula or abnormal anus might be required, as well as direct percutaneous needle injection of the rectal pouch (only rarely used). The need for such imaging however is rare, due to the availability of other imaging techniques.

9.5.3 Endoscopy

Endoscopy of the orifices within the urogenital sinus or common cloacal channel in particular, may be helpful.

9.5.4 Cross-Sectional Imaging

As for the male, ultrasound, CT, and MRI may also be useful in determining the anatomy of the malformation in indeterminate cases for surgical planning. The procedures are the same as for male infants.

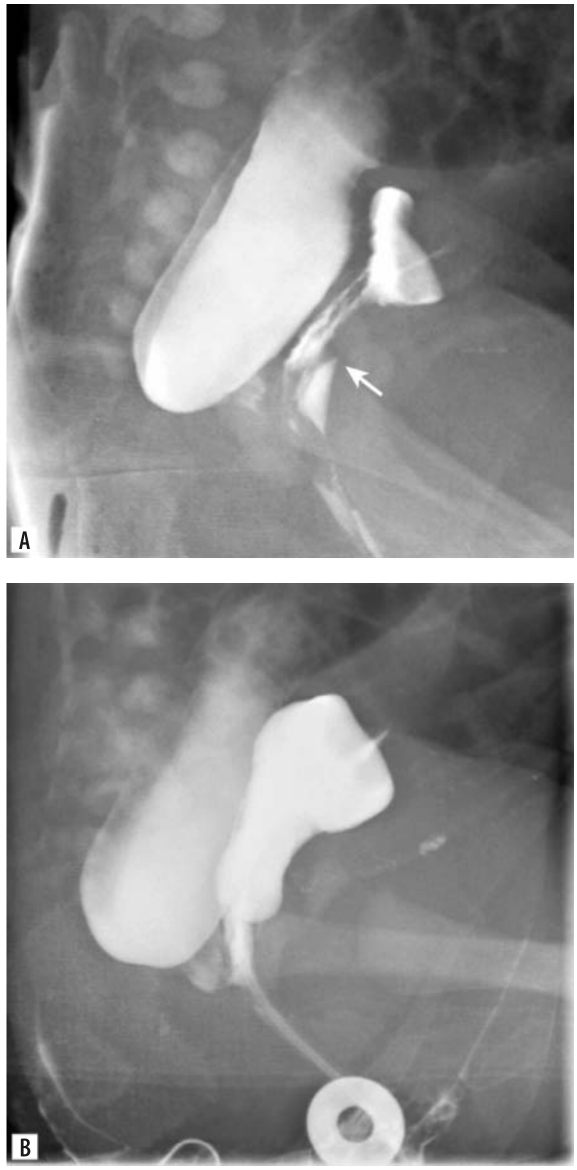

Fig. 9.11 Cloacagram in a 2-day-old female, demonstrating a catheter in the common channel with contrast in the vagina and rectum. On the early filling image (**A**) a small jet of contrast was also identified in the urethra (*arrow*). Subsequently (**B**), an 11-mm-diameter washer was placed on the perineum to indicate scale. This demonstrated the common channel to be approximately 20 mm in length. This was confirmed at surgery

9.6 Assessment of the Sphincter Complex and Nerve Supply

There is a close relationship between development of the sacrum and the extent of formation of the levator ani and sphincter muscle complex. Normal bony development is correlated closely with normal neurological development, and absence of the bony segments very commonly predicts absence of the nerve roots. When the nerve roots and the bony segments are missing, the sphincter muscle and levator ani muscle is deficient. This is now recognized by ultrasound of the spinal cord, plain x-rays of the sacrum, and MRI or CT scan of the pelvis to identify the amount and level of the muscle and presence or absence of the nerve roots.

A detailed study of specimens of ARM and sacral abnormalities predict the following general conclusions:

1. Deficiency of the fourth and fifth sacral vertebrae usually allows normal innervation of the bladder and levator ani and adequate development of the levator.
2. Deficiency of the third, fourth, and fifth sacral vertebrae is usually accompanied by variable abnormal nerve and muscle development, and most patients are incontinent.
3. Deficiencies involving the first or second sacral segments are always associated with incontinence and poorly developed and innervated levator ani and pelvic floor musculature.
4. The innervation and muscle development accompanying hemisacral defects is unpredictable, but if only one or two vertebrae show the hemisacral defect, then innervation and muscle development may be adequate, and can be determined on cross-sectional imaging.
5. Finally, the presence of a lumbosacral myelomeningocele always involves a serious defect in innervation. An anterior sacral myelomeningocele (Currarino syndrome) is also associated with significant abnormalities of the pelvic floor (see Chap. 2).

A problem arises in children in whom the sacral segments are present but may be dysmorphic, fused, or hypoplastic. There, innervation may be unpredictable and due to the distortion it may be difficult to accurately identify the number of sacral segments present, so as to use this to help predict the likelihood of adequate innervation and function. This is illustrated by the above prediction for hemisacral defects, based on specimen observations. In such cases Peña [30] has suggested the use of a sacral ratio, where the size of the sacrum is compared to that of the pelvis. This can be performed on either an anterior–posterior (AP) or lateral radiograph of the pelvis. The lateral ratio is thought to be more reliable, as the sacrum on the AP view may be foreshortened if the pelvis is tilted (Fig. 9.12).

The sacral ratio is calculated by dividing the distance from the lowest point of the sacrum to the lowest point of the sacroiliac joints, by the distance from the iliac crests to the lowest point of the sacroiliac joints. There is good inter- and intraobserver repeat-

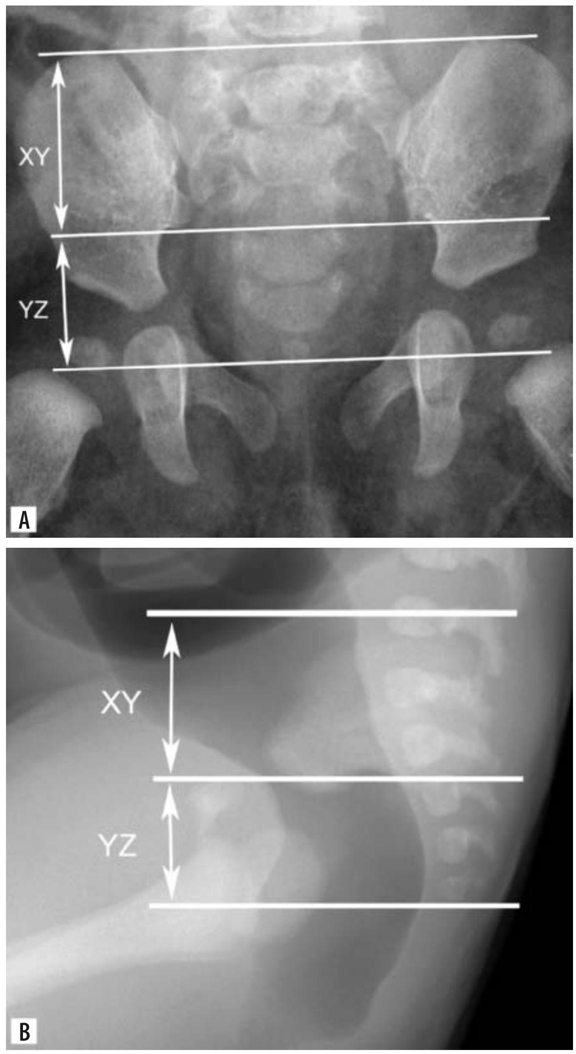

Fig. 9.12 Anteroposterior (**A**) and lateral (**B**) radiographs of the pelvis demonstrating the measurement of a sacral ratio. The ratio is calculated by dividing the distance from the inferior most point of sacrum to the inferior point of the sacroiliac joint (*YZ*) by the distance from the iliac crests to the inferior point of the sacroiliac joint (*XY*). Sacral ratio = *YZ/XY*

ability for this measurement [51]. The mean values in normal children for the AP sacral and lateral sacral ratios are 0.74 and 0.77, respectively [30, 47, 51]. There is a wide range of normal values, however, with some overlap between children with a normal sacrum and those with an abnormal sacrum. Those with a sacral ratio below 0.50, however, have a significantly decreased chance of normal function [30, 47].

A clinical assessment of sacral outflow should also be performed. As the same sacral segments (second, third, and fourth) innervate the levator ani and sphincter muscles as well as the bladder sphincter, abnormalities of bladder sphincter function can be used to predict anal sphincteric muscle complex function, and vice versa. Careful observation of urinary control in the first few days will give a good indication of not only urinary continence, but also neurological function of the sphincter muscles. Neurological abnormalities affecting the bladder will present with either continuous dribbling of small volumes through a patulous urethra, or overflow leakage with a firm full bladder that empties irregularly in episodes up to 1–2 h. The most constant and useful physical sign of a neurogenic bladder is the ability to express it by manual pressure. Even in the neonate, urine normally cannot be expressed from a bladder with normal innervation.

In babies with abnormality of the innervation of the bladder it will be either freely expressible with little pressure or only expressible with considerable force when the abdominal muscle is relaxed. Neurological assessment of the sphincter muscles can also be determined by response to pinprick stimulation of the perineal skin, absence of which predicts abnormal sacral outflow.

If imaging is required to demonstrate the pelvic floor and sphincter muscle, this can be achieved using both CT (Fig. 9.13) and MRI (Fig. 9.14) [4, 17, 20, 33, 36]. Both modalities can now demonstrate the bulk and development of the musculature in multiple planes. MRI, however, offers the advantage of being able to image the spinal cord as well as the musculature and does not involve the use of ionizing radiation.

9.7 Assessment for Associated Malformations

Associated malformations are identified in approximately 50% of children with ARM (see Chaps. 16–18) [23, 25, 31]. Some of these defects may be life threatening or may have a greater impact than the ARM

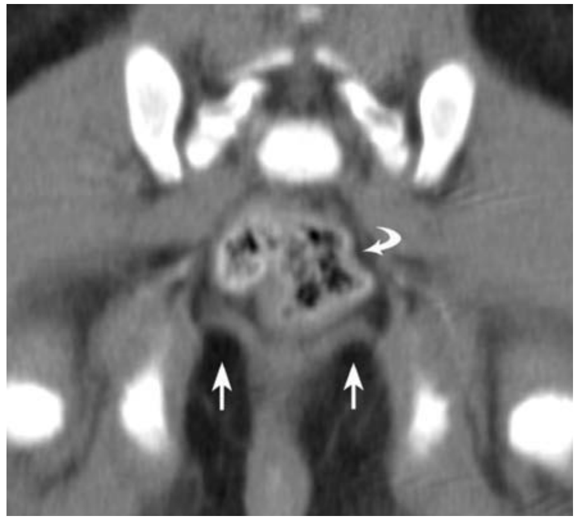

Fig. 9.13 Coronal reconstruction of a computed tomography examination of the pelvis demonstrating the pelvic floor musculature (*arrow*) and the rectal ampulla (*curved arrow*) sitting superior to the levator sling

itself on the quality of survival of the child. It is imperative, therefore, that these are identified prior to undertaking surgery to treat the ARM. Some of these will be suggested or obvious on clinical examination, such as limb anomalies in the VACTERL complex, myelomeningocele, exomphalos, and cardiac lesions. The clinical examination should include the passage of an orogastric tube to exclude the presence of an esophageal atresia. Other defects may not be apparent clinically, but can be detected on screening investigations.

Screening ultrasound of the genitourinary tract should be performed before any initial surgery [7, 31]. This is a quick, noninvasive test with high sensitivity and specificity that can be performed at the patient's bedside if required (Fig. 9.15). Depending on the clinical situation, this should be delayed for between 24 and 72 h after birth, as during this time the urinary output may be low, resulting in a false negative scan for hydronephrosis. The examination should include ultrasound of the pelvis in females to look for hydrocolpos, which may compress the bladder base producing ureteric obstruction and hydronephrosis. Pelvic ultrasound in female will also provide information about the presence of uterine or vaginal anomalies such as uterus didelphys. MCUG should also be performed to assess for vesicoureteric reflux, as well as to help in assessing the bladder in those with spinal anomalies.

Screening of the spine and spinal cord can be obtained by the combination of radiographs of the spine

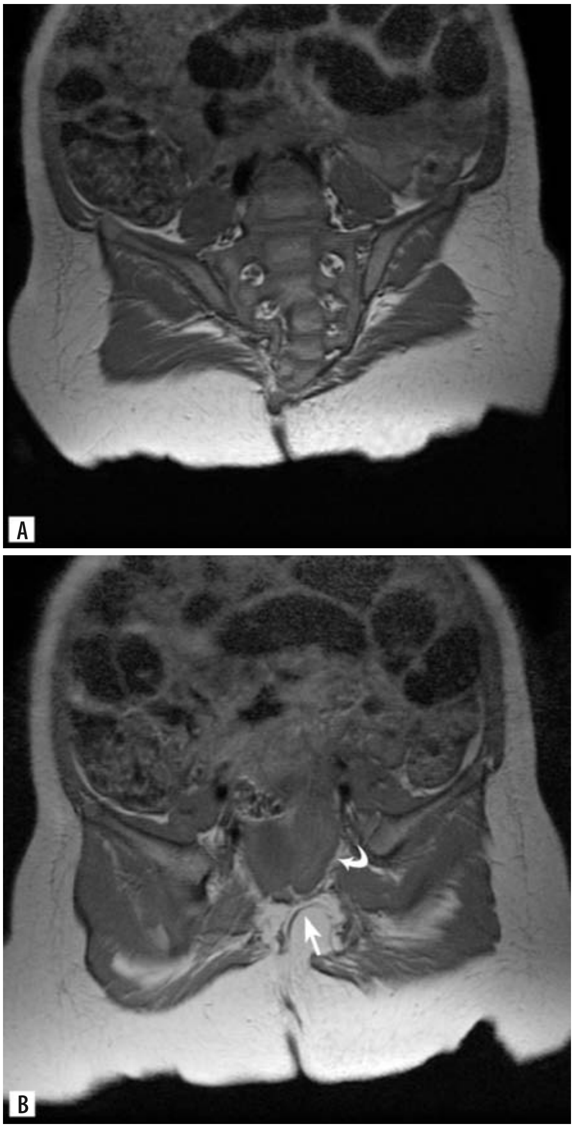

Fig. 9.14 Coronal T1-weighted magnetic resonance images in a 1-year-old male demonstrating a right hemisacral defect (**A**) and associated poor development of the right leaf of the levator sling (**B**). The normal left half of the levator is easily identified (*arrow*), with the rectal ampulla (*curved arrow*) immediately superior to it

in conjunction with an ultrasound examination of the spine and spinal cord. In the hands of an experienced pediatric radiologist, this combination is as sensitive as MRI in detecting vertebral and sacral anomalies as well as abnormalities of the cord, including tethering (Fig. 9.16) [5, 7, 23]. If abnormalities are detected ultrasonographically and further delineation of the anomaly is required, then this can readily be obtained with MRI.

Fig. 9.15 Coronal ultrasonogram of a kidney, demonstrating dilatation of the pelvicaliceal system

If experience with spinal ultrasound is not available, or MRI is being performed to evaluate the anatomy or state of the pelvic floor, then MRI may be the modality of choice to screen for associated anomalies as well as evaluating the anatomy. This has been postulated by some authors as a “one-stop shop” in the investigation of ARM [4, 35]. In such cases the field of view can be expanded to encompass the renal fossae and spine, so as to evaluate both the spinal cord and kidneys at the same time. The trade-off, however, is the decrease in spatial resolution due to the increase in the field of view. If dedicated imaging of the spine, renal tracts, and pelvis is performed so as to improve resolution, the trade-off then is the greatly increased scan times.

Cardiac anomalies are found in approximately 12% of children with ARM [25], and can produce significant morbidity. Screening for these can be obtained with echocardiography, which is a noninvasive, high-resolution technique that is currently the dominant imaging test for congenital heart disease [32].

Other investigations may be required depending on clinical findings, such as limb radiographs in those with associated limb anomalies.

9.8 Conclusion

The initial assessment of a newborn with ARM is aimed at identifying the anatomy of the malformation, the presence of any associated anomalies that may impact on immediate survival or future quality of life, and the state of the pelvic floor musculature and its innervation, which will impact on future function. The assessment must include a thorough clinical examination and appropriate imaging. An initial

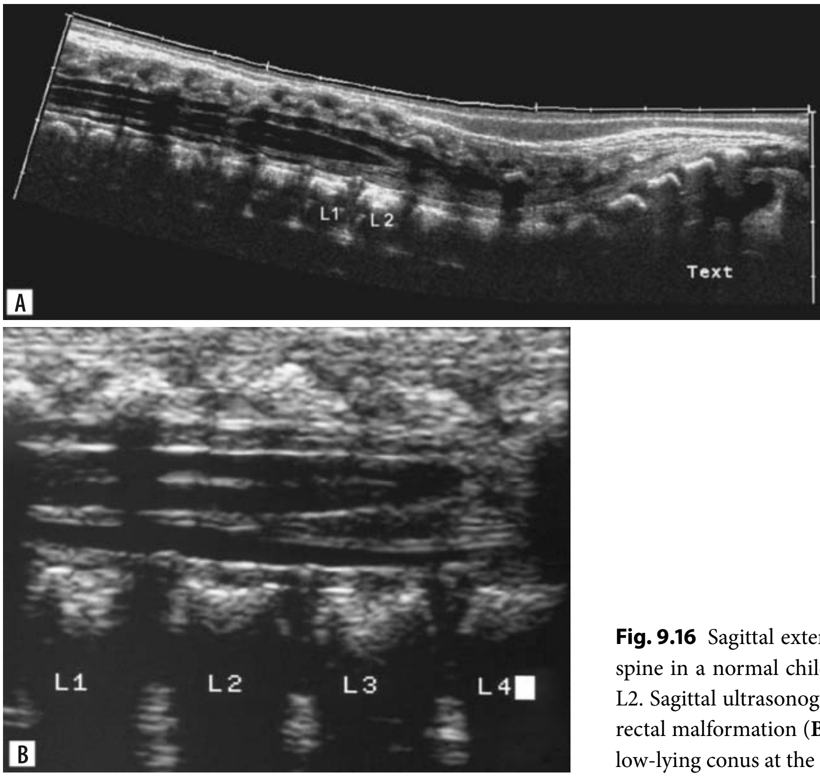

Fig. 9.16 Sagittal extended field of view ultrasonogram of the spine in a normal child (**A**) demonstrating a normal conus at L2. Sagittal ultrasonogram of the spine in a child with an anorectal malformation (**B**) demonstrating an abnormally shaped, low-lying conus at the L3/L4 level

period of observation for 12–24 h is required to allow adequate gaseous distention of the colon to develop. In clinically indeterminate cases, a prone, cross-table lateral x-ray can then be performed to help determine whether the terminal colon is likely supralevator or translevator in location. Screening ultrasound of the genitourinary tract, spine, and heart, as well as radiographs of the spine should be performed in the immediate newborn period on all children.

Following either definitive repair for translevator anomalies or diverting colostomy for supralevator lesions, delayed imaging should include MCUG and, when indicated, pressure-augmented colostogram, cloacagram, or other contrast studies as required.

References

1. Ameh EA, Chirdan LB (2000) Neonatal intestinal obstruction in Zaria, Nigeria. East Afr Med J 77:510–513
2. Anderson S, Savader B, Barnes J, Savader S (1988) Enterolithiasis with imperforate anus. Report of two cases with sonographic demonstration and occurrence in a female. Pediatr Radiol 18:130–133
3. Archibong AE, Idika IM (2004) Results of treatment in children with anorectal malformations in Calabar, Nigeria. S Afr J Surg 42:88–90
4. Aslam A, Grier DJ, Duncan AW, Spicer RD (1998) The role of magnetic resonance imaging in the preoperative assessment of anorectal anomalies. Pediatr Surg Int 14:71–73
5. Beek FJ, Boemers TM, Witkamp TD, van Leeuwen MS, Mali WP, Bax NM (1995) Spine evaluation in children with anorectal malformations. Pediatr Radiol 25 Suppl 1: S28–32
6. Berman JK (1938) Congenital anomalies of rectum and anus. Surg Gynecol Obstet 66:11
7. Boemers TM, Beek FJ, Bax NM (1999) Review. Guidelines for the urological screening and initial management of lower urinary tract dysfunction in children with anorectal malformations – the ARGUS protocol. BJU Int 83:662–671
8. Bradham RR (1958) Imperforate anus; report of 130 cases. Surgery 44:578–584
9. Cohen MD (1990) Gastrointestinal System. In: Cohen MD, Edwards MK (eds) Magnetic Resonance Imaging of Children, Vol. 1, 1st edn. BC Decker, Philadelphia, Pennsylvania, pp 611–678
10. Cremin BJ, Cywes S, Louw JH (1972) A rational radiological approach to the surgical correction of anorectal anomalies. Surgery 71:801–806
11. Crowell EA, Dulin JW (1940) Congenital anomalies of the anus and rectum. Surgery 7:529
12. Cuschieri A (2001) Descriptive epidemiology of isolated anal anomalies: a survey of 4.6 million births in Europe. Am J Med Genet 103:207–215

13. Gans SL, Friedmam NB, David JS (1963) Congenital anorectal anomalies: changing concepts in management. Clin Pediatr 2:605
14. Gross GW, Wolfson PJ, Peña A (1991) Augmented-pressure colostogram in imperforate anus with fistula. Pediatr Radiol 21:560–562
15. Han TI, Kim IO, Kim WS (2003) Imperforate anus: US determination of the type with infracoccygeal approach. Radiology 228:226–229
16. Harris RD, Nyberg DA, Mack LA, Weinberger E (1987) Anorectal atresia: prenatal sonographic diagnosis. AJR Am J Roentgenol 149:395–400
17. Ikawa H, Yokoyama J, Sanbonmatsu T, Hagane K, Endo M, Katsumata K, Kohda E (1985) The use of computerized tomography to evaluate anorectal anomalies. J Pediatr Surg 20:640–644
18. Kelly JH (1969) The radiographic anatomy of the normal and abnormal neonatal pelvis. J Pediatr Surg 4:432–444
19. Kiesewetter WB, Turner CR (1963) Continence after surgery for imperforate anus: a critical analysis and preliminary experience with the sacroperineal pull-through. Ann Surg 158:498–512
20. Kohda E, Fujioka M, Ikawa H, Yokoyama J (1985) Congenital anorectal anomaly: CT evaluation. Radiology 157:349–352
21. Lernau OZ, Jancu J, Nissan S (1978) Demonstration of rectourinary fistulas by pressure gastrografin enema. J Pediatr Surg 13:497–498
22. Mayo CW, Rice RG (1950) Ano-rectal anomalies: a statistical study of 165 cases with special reference to "distal loop trouble". Surgery 27:485
23. McHugh K (1998) The role of radiology in children with anorectal anomalies; with particular emphasis on MRI. Eur J Radiol 26:194–199
24. McHugh K, Dudley NE, Tam P (1995) Pre-operative MRI of anorectal anomalies in the newborn period. Pediatr Radiol 25 Suppl 1:S33–36
25. Mittal A, Airon RK, Magu S, Rattan KN, Ratan SK (2004) Associated anomalies with anorectal malformation (ARM). Indian J Pediatr 71:509–514
26. Narasimharao KL, Prasad GR, Katariya S, Yadav K, Mitra SK, Pathak IC (1983) Prone cross-table lateral view: an alternative to the invertogram in imperforate anus. AJR Am J Roentgenol 140:227–229
27. Nazer J, Hubner ME, Valenzuela P, Cifuentes L (2000) [Anorectal congenital malformations and their preferential associations. Experience of the Clinical Hospital of the University of Chile. Period 1979–1999]. Rev Med Chil 128:519–525
28. Oppenheimer DA, Carroll BA, Shochat SJ (1983) Sonography of imperforate anus. Radiology 148:127–128
29. Palmer JA (1956) Congenital malformations of the anus. Can Med Assoc J 74:882
30. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
31. Peña A, Hong A (2000) Advances in the management of anorectal malformations. Am J Surg 180:370–376
32. Pignatelli RH, McMahon CJ, Chung T, Vick GW 3rd (2003) Role of echocardiography versus MRI for the diagnosis of congenital heart disease. Curr Opin Cardiol 18:357–365
33. Pringle KC, Sato Y, Soper RT (1987) Magnetic resonance imaging as an adjunct to planning an anorectal pull-through. J Pediatr Surg 22:571–574
34. Rosen NG, Hong AR, Soffer SZ, Rodriguez G, Peña A (2002) Rectovaginal fistula: a common diagnostic error with significant consequences in girls with anorectal malformations. J Pediatr Surg 37:961–965; discussion 961–965
35. Sachs TM, Applebaum H, Touran T, Taber P, Darakjian A, Colleti P (1990) Use of MRI in evaluation of anorectal anomalies. J Pediatr Surg 25:817–821
36. Sato Y, Pringle KC, Bergman RA, Yuh WT, Smith WL, Soper RT, Franken EA Jr (1988) Congenital anorectal anomalies: MR imaging. Radiology 168:157–162
37. Schuster SR, Teele RL (1979) An analysis of ultrasound scanning as a guide in determination of "high" or "low" imperforate anus. J Pediatr Surg 14:798–800
38. Shopfner CE (1965) Roentgenologic evaluation of imperforate anus. South Med J 58:712–719
39. Smith CC (1953) Seven cases of imperforate anus treated by the abdominoperineal placement of the rectum. Acta Chir Scand 105:305–310
40. Smith ED (1988) Anorectal malformations. Classifications. Birth Defects Orig Artic Ser 24:211–222
41. Stephens FD (1953) Congenital imperforate rectum, recto-urethral and rectovaginal fistulae. Aust NZ J Surg 22:161
42. Stephens FD (1953) Malformations of the anus. Aust NZ J Surg 23:9
43. Stephens FD, Smith ED (1971) Ano-Rectal Malformations in Children. Year Book Medical Publications, Chicago
44. Stephens FD, Smith ED (1988) Anorectal malformations in children: update 1988. Birth Defects Orig Artic Ser 24:1–604
45. Stoll C, Alembik Y, Roth MP, Dott B (1997) Risk factors in congenital anal atresias. Ann Genet 40:197–204
46. Taccone A, Martucciello G, Dodero P, Delliacqua A, Marzoli A, Salomone G, Jasonni V (1992) New concepts in preoperative imaging of anorectal malformation. New concepts in imaging of ARM. Pediatr Radiol 22:196–199
47. Torre M, Martucciello G, Jasonni V (2001) Sacral development in anorectal malformations and in normal population. Pediatr Radiol 31:858–862
48. Trusler GA, Wilkinson RH (1962) Imperforate anus: a review of 147 cases. Can J Surg 5:269–277
49. Wang C, Lin J, Lim K (1997) The use of augmented-pressure colostography in imperforate anus. Pediatr Surg Int 12:383–385

50. Wangensteen OH, Rice CO (1930) Imperforate anus. A method of determining the surgical approach. Ann Surg 92:77
51. Warne SA, Godley ML, Owens CM, Wilcox DT (2003) The validity of sacral ratios to identify sacral abnormalities. BJU Int 91:540–544
52. Willital GH (1971) Advances in the diagnosis of anal and rectal atresia by ultrasonic-echo examination. J Pediatr Surg 6:454–457

10 Persistent Cloaca – Clinical Aspects

Alexander M. Holschneider and Horst Scharbatke

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10.1 Introduction

Persistent cloaca represents the most complex deformity in female anorectal, vaginal, and urogenital malformations. It is defined as a defect in which the rectum, one or two vaginas and the urinary tract converge into one common channel. It is very rare and occurs in 1:250,000 newborns [1]. It is physiological in some reptiles, birds, and a few mammals. In humans, however, it represents a malformation that occurs at a very early stage of development.

According to Qi et al. [2] and Nievenstein et al. [3], the rat tail gut immediately distal to the hindgut starts to regress by apoptosis on day 12 of gestation in a craniocaudal direction and has regressed completely by day 13.5. This tail-gut regression and the urorectal septum play an important role in the process of cloacal separation by cellular proliferation and differentiation. Rupture of the anal membrane plays an additional role. Cloacal malformations, according to these authors, are early defects, while anorectal malformations (ARM) with the anus in the normal position are late embryonic defects. However, in the opinion of Kluth and Lambrecht [4], the embryonic cloaca never passes through a stage that is similar to any form of ARM in neonates, including the cloaca. In contrast, the cloacal membrane is always too short in abnormal rodent embryos and the region of the future anal opening is missing, in contrast to normal mouse embryos (see Chap. 4 for a detailed description) [5].

Several experimental models of ARM including persistent cloaca exist. They are based on the teratogenic effect of ethylenethiourea in the rat [6] and exposure of rat fetuses to adriamycin [7] or etretinate, a long-acting vitamin A analog, in mice [8].

A molecular basis for ARM was first shown by Kimmel et al. [9]. In the murine model of ARM; Gli3-/-mutants exhibited anal stenosis and ectopic anus, Gli2-/-mutants exhibited imperforate anus and rectourethral fistula, and Gli2-/-Gli3+/- mutants developed a cloacal abnormality. In addition, isochromosome 18q has been shown to cause megacystis, intrauterine growth retardation, and cloacal dysgenesis sequence in a fetus [10]. Keppler-Noreuil [11] suggests a possible etiologic role for homeobox genes, such as HLXB9, with mutations resulting in ARM and spinal abnormalities.

10.2 General Clinical Aspects

Prenatal diagnosis of persistent cloaca has been reported, but is not always accurate [12,13]. In contrast, clinical diagnosis is simple. In girls, a single opening on the perineum is always suspicious of a cloacal malformation. The length of the introitus is characteristically shorter than in a normal girl. Cloacas have only rarely been reported in boys in whom the urethra and rectum has coalesced into a common channel that is connected to the external surface in the perineal or anal area [14]. In girls, an abdominal mass and severe abdominal distension resulting from hydrometrocolpos and/or rectal obstruction can frequently be observed. Additional malformations of the lower limbs, genitalia, skin (hemangioma), urogenital tract, vertebral, cardiac, and gastrointestinal deformities, among others may occur. A rarity is the posterior cloaca, described first by Peña and Kessler [15]. In these patients the vagina and urethra fuse together but the urogenital sinus (UGS) opens into the anterior rectal wall; the rectum is normal or minimally mislocated anteriorly. The most severe type of deformity is cloacal exstrophy, a combination of cloaca with bladder exstrophy that was first reported by Rickham in 1960 [16]. It is now called vesicointestinal fissure (see Chap. 14 for details).

10.3 Classification of Persistent Cloacas

A detailed description of the varying anatomy of cloacal malformations has been published by Hendren [1, 17–22] and Peña [23–25]. Hendren distinguishes between anomalies of the perineum, UGS, vagina, and rectum (Table 10.1).

Table 10.1 Classification of cloacal malformations. *UGS* urogenital sinus, *PC* pubococcygeal

Type	Anomaly
Type I: Forme fruste	Anteposition of anus with ultra short UGS and normal female genitalia
Type II: Low cloacal malformation	Short UGS < 3 cm (confluence below PC line)
Type III: High cloacal malformation	long UGS > 3 cm (confluence at or above PC line)
Type IV:	Vagina and/or rectum into bladder cavity
Rare cloacal malformations	Posterior cloaca in boys Cloacal exstrophy

10.3.1 The Perineum

According to Hendren [1] there is a wide range of deformities of each pelvic structure. At the mild end of the spectrum of perineal malformations there is an almost normal-looking vaginal opening with an anal orifice that is situated very close, but not incorporated into, the UGS. In the next degree the vaginal introitus might be incompletely formed and the anus displaced forward with a dysplastic perineum between both openings. There might be a large sinus urogenitalis opening and the anal opening just behind it, or a single perineal opening covered by the clitoris, which could be either hyper- or hypoplastic. In a very few cases an accessory hypoplastic urethra can be observed at the tip of the phallus in addition to the proximal urethra fusing with the UGS. The perineal skin can be covered by a hemangioma and abnormal pigmentation (Fig. 10.1 A–F).

10.3.2 UGS Variations

The UGS may exhibit an ending on the tip of the clitoris, a subclitoral meatus, a wide opening to a short common channel, or a long sinus with the junction of urethra, vagina(s), and rectum above the pubococcygeal (PC) line. By using this anatomical projection of the upper border of the prostate and the ischial (I) point representing the plane of the upper border of the internal anal sphincter, high, intermediate, and low types of cloaca can be distinguished according to the Wingspread classification of ARM [26]. However, there are so many variations of anomalies of the vagina, urethra, and rectum in cloacae that this classification, which is very useful in ARM, is not very helpful for the classification of cloacae. Peña's sacral ratio [25], which should be in 0.77 in normal children, gives an idea of sacral hypodevelopment and, therefore, the probable degree of neurogenic bladder dysfunction and disturbed bowel control, but is no classification. Nevertheless, it is important to distinguish between high and low cloacal deformities because, for example, a UGS less than 3 cm long can be treated by total UGS advancement, whereas in longer channels an abdominal approach is necessary (Fig. 10.2) [27].

10.3.3 Vaginal Variations

The two most common variations are a single vaginal opening in the upper urethra, with the rectal opening below this orifice, and a double side-by-side vagina with the rectum ending on the septum between both vaginas. In cases of hemivaginas and an incomplete septum, the rectal fistula can be situated high on the septum; in cases with two separated vaginas it can usually be found between the vaginal openings. There might be two vaginas but atresia, or even absence of one, double diverging vaginas, two separate vaginas, or both, or two vaginas entering the bladder (Fig. 10.3 A–G).

10.3.4 Rectal Variations

The rectum can enter the UGS separate from and below the vagina, may enter the posterior wall of the

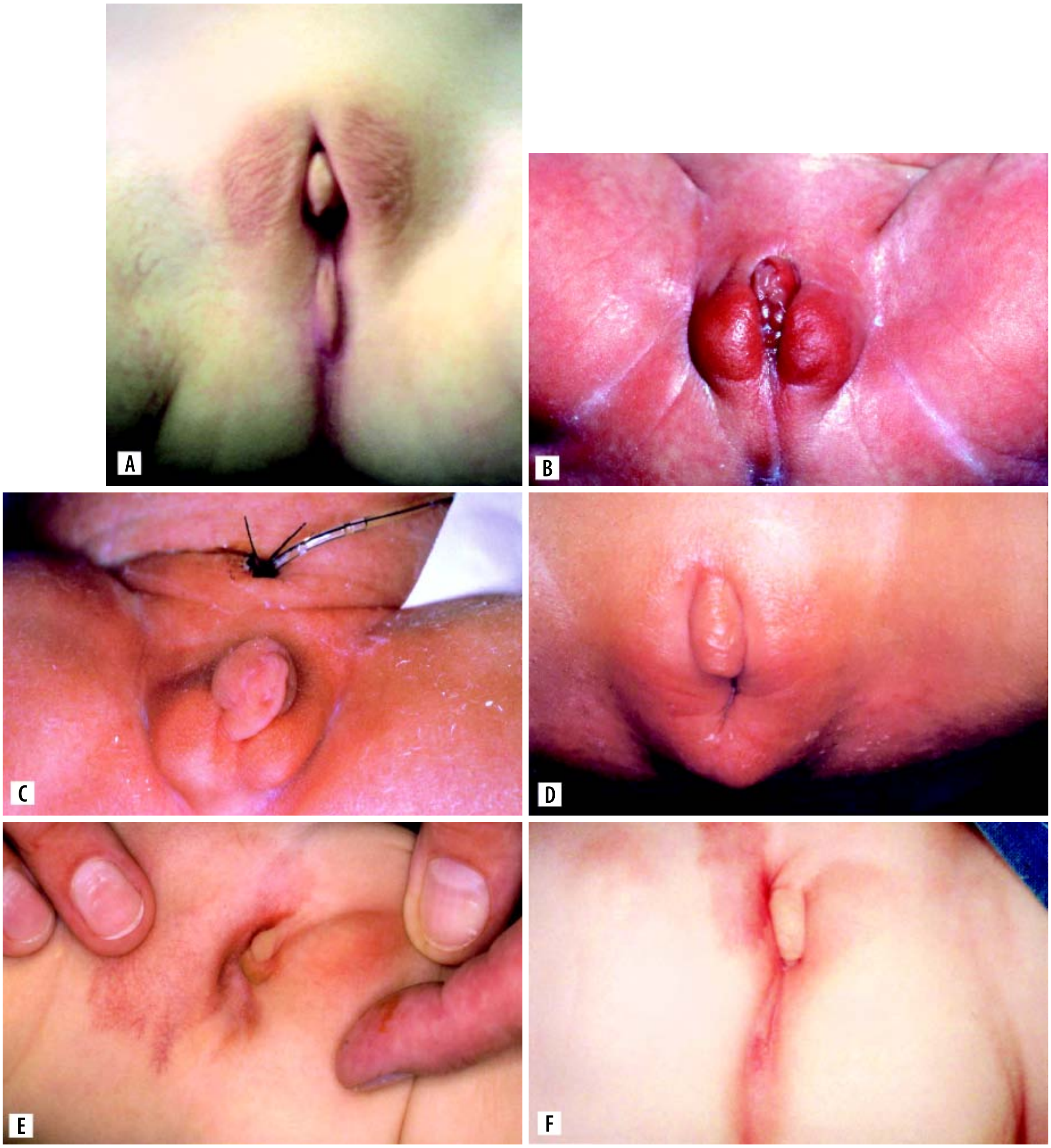

Fig. 10.1 Clinical aspects of cloacal malformations. **A** Almost normal-looking female genitalia. **B** Enlarged labia majora. **C** Male aspect of female genitalia (after drainage of hydrocolpos). **D** Malformation of the labia majora. **E** Total malformation of the introitus; the labia majora are visible. **F** Labia majora not developed

vagina or the bladder, it can be positioned between and anterior to two vaginal openings, or run as a long fistula all along the posterior wall of the vagina down to the UGS. The rectal pouch at the upper end of the fistula is very high up in these cases, sometimes even above the peritoneal reflection (Fig. 10.4).

10.4 Associated Malformations

Being a very complex deformity of a very early stage of human development, many associated anomalies can be observed in patients with cloacal malformations (Table 10.2).

Table 10.2 Some of the associated anomalies found in children with cloacal anomalies

Vesicoureteral reflux	Absence of uterus
Malformations of the kidneys	Bladder exstrophy
Tethered cord	Gastrointestinal duplications
Neurogenic Bladder	Pouch colon
Diastematomyelia	Cardiac malformations
Myelomeningocele	Vertebral deformities
Lower-limb deformities	Esophageal atresia
Occluded tubes	Cerebral anomalies
Absence of one ovary	Abnormalities of the enteric nervous
Hypoplastic labia	

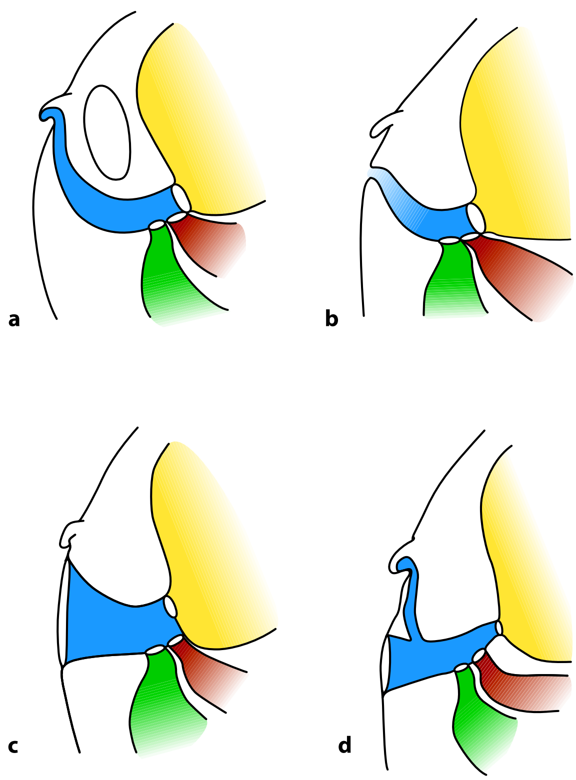

Fig. 10.2 Urogenital sinus (*UGS*) variations [modified according to Hendren (1992) J Pediatr Surg 27:890–901, Fig. 3]. **a** Long UGS ending in the tip of the clitoris. **b** Subclitoral meatus. **c** Wide opening of the UGS (like a vagina). **d** With accessory tract

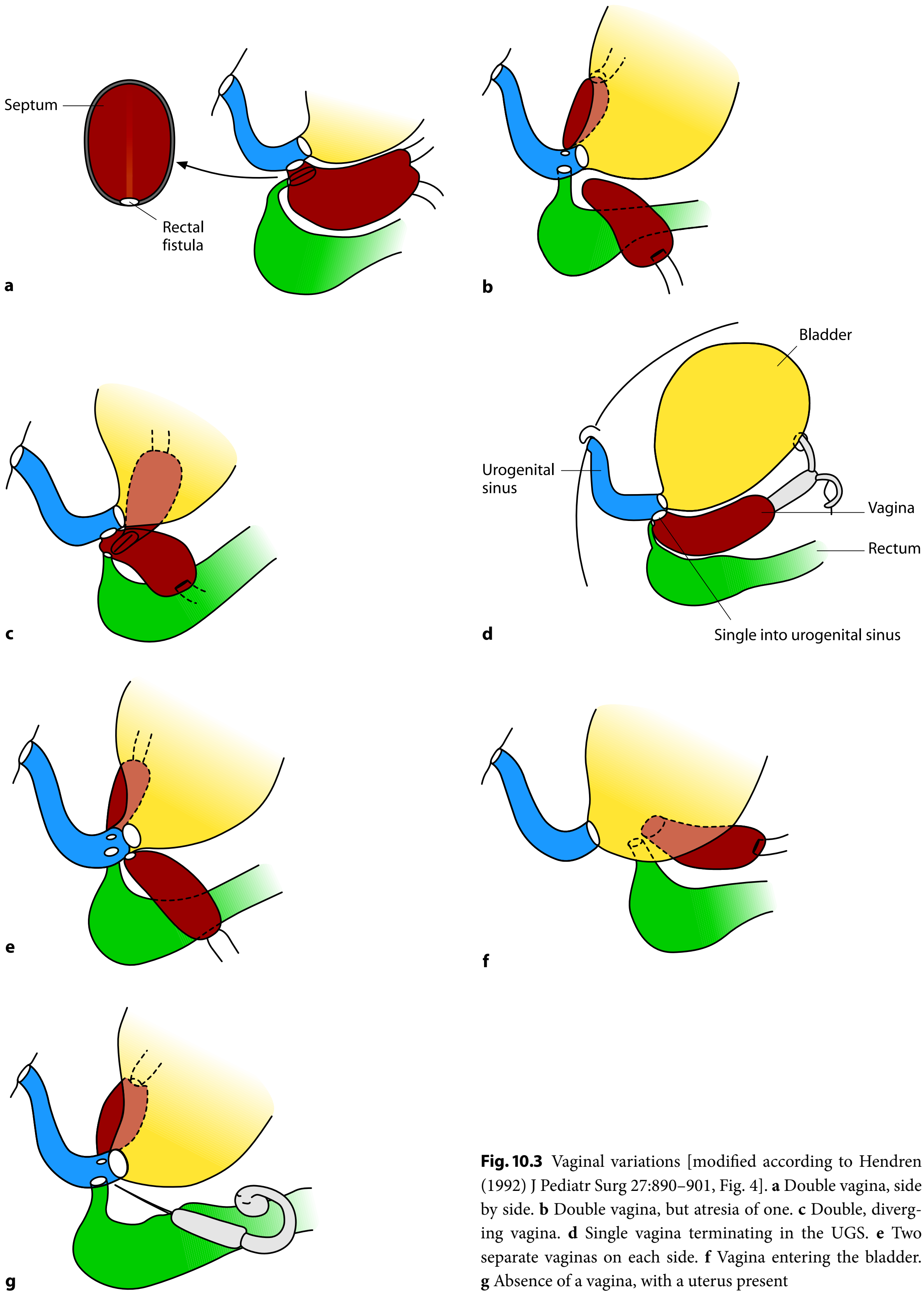

Fig. 10.3 Vaginal variations [modified according to Hendren (1992) J Pediatr Surg 27:890–901, Fig. 4]. **a** Double vagina, side by side. **b** Double vagina, but atresia of one. **c** Double, diverging vagina. **d** Single vagina terminating in the UGS. **e** Two separate vaginas on each side. **f** Vagina entering the bladder. **g** Absence of a vagina, with a uterus present

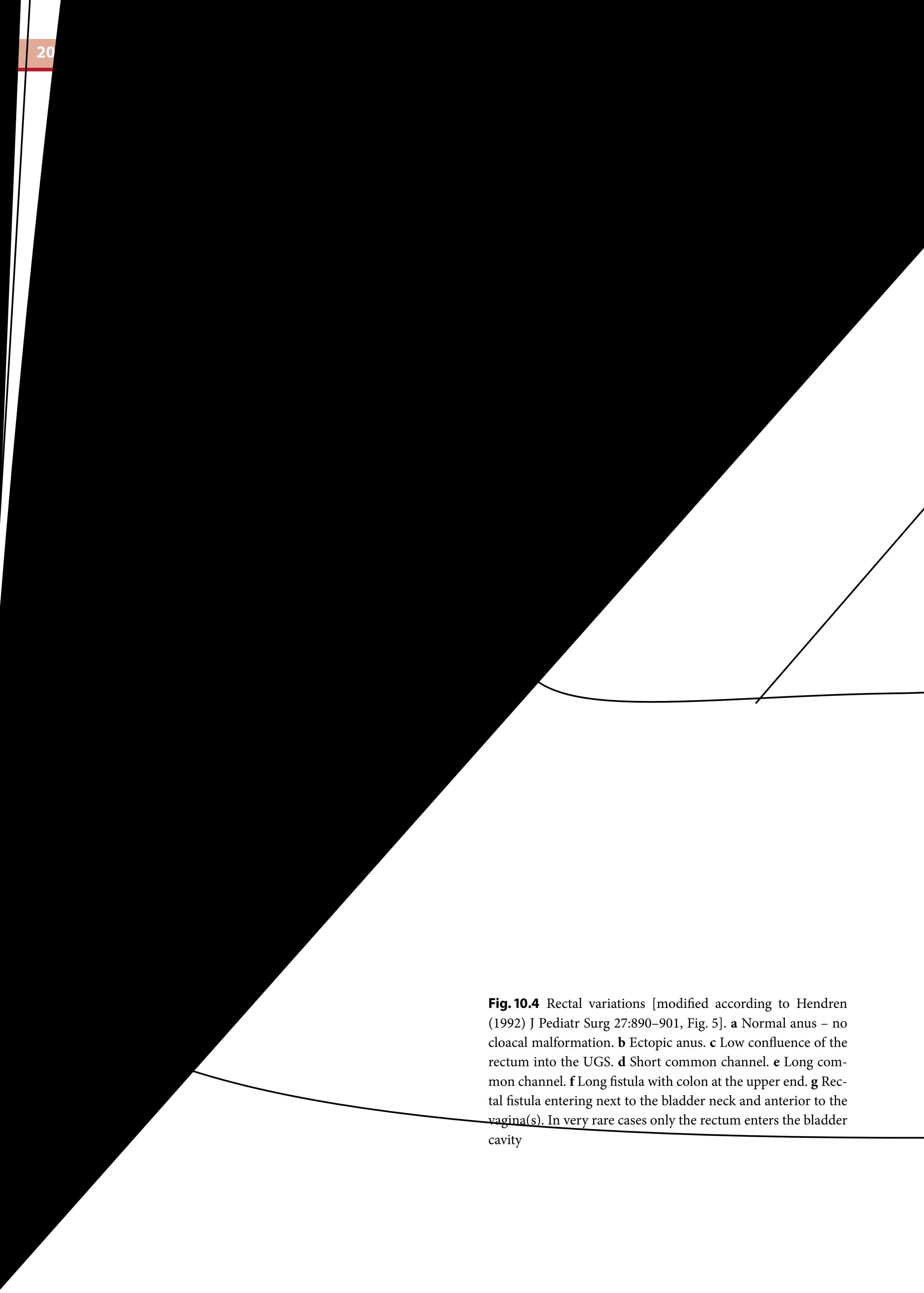

Fig. 10.4 Rectal variations [modified according to Hendren (1992) J Pediatr Surg 27:890–901, Fig. 5]. **a** Normal anus – no cloacal malformation. **b** Ectopic anus. **c** Low confluence of the rectum into the UGS. **d** Short common channel. **e** Long common channel. **f** Long fistula with colon at the upper end. **g** Rectal fistula entering next to the bladder neck and anterior to the vagina(s). In very rare cases only the rectum enters the bladder cavity

16.3 Other Intestinal Malformations

Other intestinal malformations that may accompany ARM include jejunal-ileal atresia and colonic malformations. Given the association between notochordal malformations and intestinal malformations, some of the same processes that produce EA/TEF and ARM may be applicable to other malformations. Many years ago we reported a congenital malformation of the colon associated with a teratoma arising from L2 that was associated with a low ARM and in whom the aorta was noted to be bifid below the level of T12 [12]. Other cases, although rare, of dilated, dysfunctional colon found in patients with ARM may be of similar etiology (see Chap. 11 on congenital pouch colon).

16.4 Hirschsprung's Disease

The coincidence of HD and ARM is not easily determined. In the aforementioned extensive series of lesions associated with ARM, the incidence of classically proven HD was thought to be very low [9]. Hypoganglionosis of the terminal gut has been observed in cases of ARM where it was felt to be a result of the etiologic factors producing the ARM rather than due to a de novo failure of ganglion cell migration [13,14]. It has been suggested that the terminal bowel in cases of high ARM be biopsied and that the aganglionic or hypoganglionic segments be resected during the pull-through [15], but most surgeons now feel that the association between HD and ARM is sufficiently rare that biopsy sampling is unnecessary. Constipation following repair of an ARM is unlikely to be due to HD and can be managed by ensuring patency of the anus and attention to dietary factors.

More recently, the subject of NID and the occurrence of classical HD in the presence of low ARM have entered the complex picture of innervation disorders of the anorectum and terminal colon. Using the more modern techniques of histochemical analysis of the fistula or terminal rectum in patients with ARM, the criteria for both HD and NID have been seen with somewhat alarming frequency [16]. All fistulas were "abnormally" innervated. The four patients with low ARM and recognizable innervation disorders of the distal centimeter of the rectal pouch portion of the resection ultimately had severe constipation. Only 5 of 19 patients with intermediate or high ARM and innervation disorders suffered from constipation or soiling. The authors' recommendation was that since the clinical course could not be correlated with the type or severity of the distal innervation disorder, the fistula and the distal segment of rectal pouch should not be used in the repair of the ARM. Perhaps the safest course in the absence of the histochemical analysis is to follow the authors' recommendation and discard the distal 1 cm of the rectal pouch.

16.5 Cardiac Malformations

Because the association between ARM and cardiac malformations exceeds that of any other "correctable" associated malformation, it is necessary to obtain a cardiology consultation and an echocardiogram as part of the initial evaluation (Fig. 16.3). The most common associated cardiac malformation is a ventricular septal defect (VSD). Some are associated with tetralogy of Fallot (TOF) and others are isolated defects. It is unusual for a VSD of any type to be associated with cyanosis or cardiac failure in the newborn except for those with complex cardiac malformations such as pulmonary atresia or transposition of the great vessels. In the very rare instance of these latter lesions being associated with ARM patency of the ductus arteriosus may be maintained by the use of prostaglandin while the colostomy is established. Additionally, the patent foramen ovale may need to be enlarged by the balloon technique in the case of in-

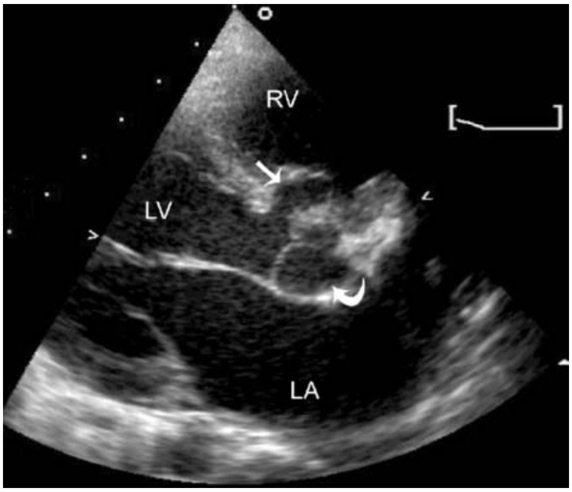

Fig. 16.3 Transthoracic echocardiogram demonstrating a gap between the left ventricle (*LV*) and right ventricle (*RV*) that does not involve the left ventricular outflow tract and aorta (*curved arrow*), in keeping with a perimembranous ventricular septal defect (*arrow*). Image courtesy of Dr. G. Lane, Department of Cardiology, Royal Children's Hospital, Melbourne, Australia

adequate atrial mixing. This is probably the only time the cardiac lesion will take precedence over the repair of the ARM. The usual associated cardiac malformations of VSD and TOF will be addressed following the repair of the ARM.

When EA/TEF is present to further complicate the clinical and therapeutic course, many factors enter into the treatment algorithm. Among these is the possibility of ligation or division of the distal TEF along with a colostomy to temporize. If the baby has TOF the descending aorta may be on the right side, making the definitive repair of the EA/TEF easier from the left thoracic approach.

Only in the presence of a cyanotic cardiac lesion without ductal patency will a systemic-to-pulmonary shunt be required. Although most of these are currently done by the interposition of a Gore-Tex graft from the subclavian artery to the ipsilateral pulmonary vessel, the classic Blalock-Taussig shunt is created on the side of the innominate artery and may cause the later repair of the EA/TEF to be performed through a previously violated pleural cavity.

The mortality rate of the commonly associated cardiac malformations has dropped significantly over the last decade or two, but the constellation of a more severe, cyanotic cardiac defect with EA or ARM will obviously increase the complexity of the surgical approach as well as the mortality and morbidity.

16.6 Spinal Deformities

These are the most commonly associated malformations with ARM. The most common of the spinal deformities are aberrations in the numbers and symmetrical development of the pelvic vertebrae. There are usually hemivertebrae present that will tilt the pelvis to a varying degree and, if extensive, may result in severe deformity of the lower body. The absence of several pelvic vertebrae is more often associated with urinary incontinence than with bowel incontinence.

At times the spinal malformation may be very severe with lack of development of the sacrum and sacral nerve plexus, a condition know as caudal regression or caudal dysgenesis sequence. In these instances the baby's buttocks are flat and the gluteal crease is barely discernable. The clinical import of this condition is that the pelvic musculature and innervation are impossible to utilize. The establishment of an anal opening on the perineum will result in a perineal colostomy with little or no bowel control and no possibility of maintaining a colostomy bag seal. In these cases, establishment of a permanent colostomy is required following separation of the distal bowel from the urogenital tract.

Some of the associated spinal malformations are accompanied by a tethered spinal cord. The filum terminale is fixed to the abnormal sacral vertebrae and the spinal cord is stretched, resulting in dysfunction of the most distal spinal nerves first – those innervating the levator/sphincter mechanism, which results in a flaccid anus. There is usually is some skin abnormality such as a hairy nevus located over the sacrum or distal lumbar vertebrae. Ultrasonography will usually detect the tethered cord or will indicate the presence of an intraspinal lipoma or meningeal cyst that might also eventually prevent the patient from being satisfactorily continent following repair of an ARM. There continues to be no hard evidence that the presence of a tethered cord affects continence or that release of the tethered cord alone will improve continence when it is suboptimal. The reader is directed to Chap. 18 for a more complete discussion of this topic.

References

1. Kimmel SG, Mo R, Hui C, et al (2000) New mouse models of congenital anorectal malformations. J Pediatr Surg 35:227–231
2. Ioannides AS, Henderson DJ, Spitz L, et al (2003) Role of sonic hedgehog in the development of the trachea and esophagus. J Pediatr Surg 38:29–36
3. Spilde T, Bhatia A, Ostlie D, et al (2003) A role for sonic hedgehog signaling in the pathogenesis of human tracheoesophageal fistula. J Pediatr Surg 38:465–468
4. Gillick J, Mooney E, Giles S, et al (2003) Notochord anomalies in the adriamycin rat model: a morphologic and molecular basis for the VACTERL association. J Pediatr Surg 38:469–473
5. Qi BQ, Beasley SW, Frizelle FA (2003) Evidence that the notochord may be pivotal in the development of sacral and anorectal malformations. J Pediatr Surg 38:1310–1316
6. Mortell A, Gillick J, Giles S, et al (2005) Notable sequential alterations in notochord volume during development in the adriamycin rat model. J Pediatr Surg 40:403–406
7. Gross RE (1953) Malformations of the anus and rectum. In: Gross RE (ed) The Surgery of Infants and Children. WB Saunders, Philadelphia, pp 348–368
8. Kiesewetter WB (1981) Imperforate anus. In: Holder TM, Ashcraft KW (eds) Pediatric Surgery. WB Saunders, Philadelphia, pp 401–417
9. Smith ED, Saeki M (1988) Associated anomalies. In: Stephens D, Smith ED (eds) Anorectal Malformations. Birth Defects: Original Article Series. 24, Number 4:501–549

10. Ratan SK, Rattan KN, Pandey RM, et al (2004) Associated congenital anomalies in patients with anorectal malformations – a need for developing a uniform practical approach. J Pediatr Surg 39:1706–1711
11. Holder TM, Cloud DT, Lewis JE Jr, et al. (1964) Esophageal atresia and tracheoesophageal fistula: a survey of its members by the Surgical Section of the American Academy of Pediatrics. Pediatrics 34:542–549
12. Ashcraft KW, Holder TM (1966) Congenital megaileocolon (basketball bowel) with teratoma. J Pediatr Surg 1:178–183
13. Parkkulainen KV, Hjelt L, Sulamaa M (1959) Anal atresia combined with aganglionic megacolon. Acta Chir Scand 118:252–254
14. Okamoto E, Ueda T (1967) Embryogenesis of intramural ganglia of the gut and its relation to Hirschsprung's disease. J Pediatr Surg 2:437–443
15. Kiesewetter WB, Sukaraochana K, Sieber WK (1965) Frequency of aganglionosis associated with imperforate anus. Surgery 58:887–880
16. Holschneider AM, Ure BM, Pfrommer W, et al (2005) Innervation patterns of the rectal pouch and fistula in anorectal malformations: a preliminary report. J Pediatr Surg 31:357–362

17 Urological Problems in Children with Anorectal Malformations

Duncan T. Wilcox and Stephanie A. Warne

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17.1 Introduction

The importance of investigating and treating the urinary tract in patients born with anorectal malformations (ARM), has until recently been underemphasized [5]. This is surprising as the association between urological anomalies and ARM is well described and is a known feature of both the VATER (acronym of Vertebral and vascular anomalies, Anal atresia, Tracheoesophageal fistula, Esophageal atresia, and Renal anomalies, Radial dysplasia) and VACTERL (acronym of Vertebral abnormalities, Anal atresia, Cardiac defects, Tracheoesophageal fistula with Esophageal atresia, Radial and renal defects, and Lower-limb abnormalities) associations [40,56]. These children can have both structural and functional abnormalities of the upper and lower urinary tract as well as significant genital anomalies [37]. Anomalies of the genitourinary tract can have a dramatic impact on the length and quality of these children's lives [27].

Genitourinary anomalies occur frequently in patients with ARM and previous retrospective reviews report incidences from 20 to 50% [27,28,36,41,54,55]. In one large series from Japan consisting of 1,992 patients, 425 had genitourinary problems [16]. This association is easily understood when one considers that the embryological development of the rectum and genitourinary tract develop simultaneously and in close proximity [10]. A common embryological insult affecting the caudal portion of the embryos development at a critical time during early gestation can cause a spectrum of defects in the anorectal, genital, urinary, and spinal tract [26,32,47].

Overall, approximately 40% of patients have a urinary tract anomaly and 10% have a genital anomaly (Table 17.1) [27,28,30,41]. Historical studies have used different classification systems; in this chapter it has only been possible to compare reports using the "high, intermediate and low" definitions. In addition, female patients with cloacal anomalies will be considered separately. The incidence of urinary anomalies increases according to the severity of the anorectal lesion [28,37,42] and is shown in Table 17.2. The incidence of an associated genitourinary anomaly also increases when a lumbosacral defect is present [28,37,42].

Genital maldevelopment is less frequent, but still a significant problem. Interestingly, those patients with a urinary anomaly are more likely to have a genital tract problem (26%) compared with those without a

Table 17.1 Incidence of genitourinary anomalies related to type of *ARM* Anorectal malformation

Fistula level	Associated genitourinary malformation %
Cloaca	88
Bladder neck	92
Prostatic urethra	66
Vestibular	30
Bulbar urethra	25
Perineal	0
No fistula	25

urinary defect (14%) [28]. However, a genital anomaly is much better at predicting a urinary problem, as 55% of these patients have both [28].

The incidence of genitourinary problems depending on the sex of the patient is confusing. In their early series, Metts et al. showed clearly that boys had more problems than girls (50% versus 30%), and this difference continued to be significant even when the severity of the abnormality was considered [28]. Ratan, however, showed the opposite, with girls more frequently affected [40]. McLorie and Warne identified no significant difference between the sexes for renal abnormalities [27,41,51 52].

This chapter will outline the structural and functional anomalies seen in the genitourinary tract and describe the abnormalities seen according to the severity of the underlying ARM. In addition, the potentially adverse effects of surgical reconstruction on the urological outcome will be discussed. Finally, a suggested outline for the evaluation of these patients will be proposed.

17.2 Renal Anomalies

The majority of anomalies associated with the kidney and ureter have been described in patients with ARM. This review will outline those seen and the management of these problems.

17.2.1 Structural

17.2.1.1 Position

Kidneys placed ectopically have been well described [27,28,30,41]. Kidneys can either be single and ectopically positioned (i.e., pelvic) or can be joined. Joining of the kidneys can either be midline (horseshoe kidney; Fig. 17.1) or joined both on the same side (crossed fused ectopia; Fig. 17.2). Renal ectopia occurs in approximately 5% of patients and renal fusion in 5% [27,28]. These patients do not need specific management of these renal problems; however, they do predispose to urinary tract infections and to vesicoureteric reflux, which needs to be considered.

17.2.1.2 Duplication

Renal duplication, either partial or complete, is seen in between 2 and 5% of patients [28,30]. The management of these patients is the same as those without ARM. An ectopic ureter associated with an upper pole moiety must be considered as a cause of incontinence in these children.

17.1.2.3 Hydronephrosis

The incidence of hydronephrosis varies greatly between the series reported in the literature. Hydrone-

Table 17.2 Genitourinary anomalies according to severity of the ARM [28]

Abnormality	ARM Boys $n = 21$	ARM Girls $n = 12$	Cloaca $n = 12$	Total $n = 45$
Abnormal sacrum	10 (48%)	5 (42%)	6 (50%)	21 (47%)
Abnormal spinal cord	2 (9%)	4 (33%)	5 (42%)	11 (24%)
Abnormal kidneys	11 (52%)	3 (25%)	3 (25%)	17 (38%)
Vesicoureteric reflux	6 (29%)	3 (25%)	1 (8%)	10 (22%)
Abnormal genitalia	11 (52%)	1 (8%)	12 (100%)	24 (53%)
Abnormal urodynamics	13 (62%)	6 (50%)	10 (75%)	29 (64%)

Fig. 17.1 Intravenous urogram showing a horseshoe kidney

Fig. 17.2 Intravenous urogram showing a right-crossed, fused, ectopic kidney

phrosis was the most common problem identified in the renal tract by Ratan and colleagues, but in other series the rate is between 2 and 10% [41]. In most series hydronephrosis is used to refer to a uretero-pelvic junction impairment to urine flow. The management of these patients has not been specifically discussed; the general approach is prophylactic antibiotics and observation, with approximately 25% requiring surgical reconstruction [14]. Surgical reconstruction is needed in those patients with an increasing hydronephrosis, deteriorating renal function, or symptoms [14].

17.2.1.4 *Renal Dysplasia*

Renal dysplasia is seen in patients with both low and high ARM [27,30]; the incidence varies from 2 to 8%. The diagnosis of dysplasia has been made in a variety of ways including pathological specimens and by nuclear renography. The latter technique is not as accurate. In addition to dysplasia, patients with ARM may also have a multicystic dysplastic kidney, which is nonfunctioning. This abnormality appears less commonly, in about 1–3% of patients [27,30]. The importance of dysplasia and renal agenesis cannot be overlooked, as chronic renal failure is one of the major causes of mortality in these patients [27].

17.2.1.5 *Renal Agenesis*

Unilateral renal agenesis is a common problem, it is reported in 3–5% of children with low anomalies and up to 20% of patients with high anomalies [27,28,30,42].

17.2.2 Functional

Studies looking at renal function in prospective cohorts of patients with ARM have not been reported. Patients with chronic renal failure (defined as a glomerular filtration rate, GFR - corrected for surface area- of less than 80 ml/min) have been described in both low and high anomalies. Misra described 4 of 95 patients who had chronic renal failure; the underlying diagnosis was bilateral renal dysplasia, bilateral vesicoureteric reflux, and neuropathic bladder. One of these patients required renal transplantation before 18 years of age [30]. The incidence of chronic renal failure in patients with high lesions is not well documented, but between 2 and 6% of these patients die from renal insufficiency, compared with 1.1% with low lesions [27]. The management of chronic renal failure and end-stage disease in children with ARM raises specific challenges. Peritoneal dialysis and subsequent renal transplantation can be technically de-

Table 17.3 The incidence of urinary tract anomalies in 64 patients with a persistent cloaca

Urinary tract anomalies	Number	%
Bilateral renal dysplasia	15	23
Unilateral renal dysplasia	2	3
Solitary kidney (dysplastic)	8	13
Pelviureteric junction obstruction	4	6
Bilateral duplex	6	9
Horseshoe kidney	1	3
Pelvic kidney	4	6
Crossed fused ectopia	4	6
Hydronephrosis	10	16
Ureterocele	1	2
Patent urachus	1	2
Bladder diverticulum	2	3
Bladder atresia	3	5

manding due to the previous abdominal operations. In addition, those patients who go on to renal transplantation need specific attention to exclude neuropathic bladder, which could continue to damage the new kidney.

In a recent long-term outcome review of 64 cloaca patients, an abnormality of the kidneys, ureters, and bladder was identified in 83% at presentation (Table 17.3) [52]. Other authors also describe a similarly high incidence of genitourinary abnormalities in girls with persistent cloaca [20,21,42]. Bilateral renal dysplasia and dysplasia of a solitary or cross-fused ectopic kidney was diagnosed in the early neonatal period, with abnormal renal function on presentation in 15 (23%). At an average age of 11 years, 50% of the group had developed chronic renal failure (with GFR < 80 ml/min/1.73 m²). This was severe or end-stage renal failure in 17% (with GFR < 25 ml/min/1.73 m²). Six patients (9%) required renal transplantation. One died posttransplantation from renal vein thrombosis in the grafted kidney. A further three patients died as a consequence of renal failure, giving an overall mortality rate of 6% from renal failure [52].

17.3 Ureteric Anomalies

17.3.1 Vesicoureteric Reflux

The reported incidence of vesicoureteric reflux varies greatly from 2 to nearly 50% [27,41]. The variation appears to depend entirely upon the number of patients who undergo a diagnostic micturating cystogram to detect reflux. In a cohort who all had cystograms, 33% presented with reflux; however, this represented a selected group [27]. In a prospectively studied group, 10 out of 45 (22%) had vesicoureteric reflux; 4 of these (9%) had structurally normal upper tracts at presentation. All grades of reflux have been described: in 1996, Boemers et al. reported that 27% of their cohort had reflux; of the 24 patients (37 kidneys) with reflux, 6 were grade I, 4 were grade II, 5 were grade III, 9 were grade IV, and 3 were grade V [3,4]. This suggests that higher-grade reflux is seen than in patients with primary vesicoureteric reflux; however, large series have not been well reported and an association between reflux with and that without a neuropathic bladder has not been documented.

The management of reflux follows the same principles as all patients with primary reflux. In 18 patients with reflux, 10 spontaneously improved, 3 are still being observed, and 5 had the reflux corrected [30]. Neither the initial grade of reflux nor the indication for surgical treatment, however, was mentioned. Division of the rectourinary fistula and subsequent decrease in bacterial contamination frequently leads to resolution of lower grades of reflux [9,43]. This report and others, suggest that a policy of observation and expectant management is appropriate for these patients.

17.3.2 Megaureters

Megaureters are rarely observed (approximately 1–3%) [28,30]. As with hydronephrosis, a policy of prophylactic antibiotics and observation is appropriate. The necessity for surgical reconstruction is not well reported.

17.3.3 Ureteric Ectopia

Ureteric ectopia is a rare clinical entity. It is associated with complete ureteric duplication in 80% of cases. Single system ureteric ectopia usually presents with persistent urinary incontinence and hydroureteronephrosis and is more frequently seen in patients with cloacal anomalies (A. Trainer, personal communication) [51]. It is associated with a renal abnormality such as horseshoe kidney, crossed-fused renal ectopia, malrotated kidney, renal dysplasia, and pelvic kidney. Surgical options to correct this condition usually include ureteric reimplantation and procedures to increase bladder outlet resistance.

Table 17.4 Urodynamic findings in a prospective cohort of patients with an ARM according to fistula position. *NFU Abn* Abnormality found on natural filling urodynamics *DO* detrusor overactivity, *M* male, *F* female

	Cloaca	Vesical fistula	High, no fistula	Urethral fistula	Vestibular fistula
*n**	12	5	4	12	10
Gender	F	M	2M, 2F	M	F
Normal	2 (17%)	0	0	6 (50%)	6 (60%)
NFU Abn	10 (83%)	5 (100%)	4 (100%)	6 (50%)	4 (40%)
Idiopathic DO	3	0	1	2	1
Neurogenic DO	7	2	1	4	2
Asynchronous	0	0	2	0	1
Inadequate	0	3	0	0	0

17.4 Bladder Anomalies

17.4.1 Structural

Structural abnormalities of the bladder occur infrequently in patients with ARM. However, fistulas between the rectum and bladder or bladder neck occur in around 10% of all ARM and represent the most complex malformations in males [37,42]. The bladder is absent in up to 8% of female infants with a persistent cloaca; in these patients it is associated with bilateral, single-system ectopic ureters [51].

17.4.2 Functional

The etiology of lower urinary tract dysfunction in ARM and cloacal anomalies is poorly understood [2–4,50,51]. However, bladder dysfunction causes significant urological morbidity in the pediatric population, resulting in renal damage from recurrent urinary tract infections and urinary incontinence, both of which can cause profound morbidity and disability [7]. Recurrent urinary tract infection may be overlooked or attributed to coexisting vesicoureteric reflux or renal anomalies, which are prevalent in a high proportion of these children [7,27]. Consequently, detecting bladder dysfunction at an early age is essential in avoiding deterioration in renal function [5,18,24,45,46].

17.4.2.1 Preoperative

In newly diagnosed ARM and cloacal malformations, surgical and medical treatment, for the first few years of life, focuses primarily on construction of a new anus and to restore continuity to the gastrointestinal tract [37,38]. Bladder dysfunction can easily be overlooked and is difficult to evaluate since surgical reconstruction is performed at an age where the majority of patients have not reached the age to achieve urinary continence.

Recently, De Gennaro and colleagues described a 4-h voiding observation in infants and observed that 50 out of 89 patients (55%) had abnormal bladder function. In the neonates, five out of nine had incomplete voiding [32]. This compares well with the overall incidence of bladder dysfunction as assessed by urodynamics, which is between 7 and 30% [4,49]. The majority of children with bladder dysfunction had abnormal spines, 88% in a recent series [32]. However, there are multiple reports of ARM patients with abnormal bladder function with a normal bony sacrum [32,49]. Therefore it is not possible to use spinal radiography alone as the investigation to exclude spinal dysraphism or spinal cord lesion and potential neurogenic bladder in ARM patients. Ultrasound and magnetic resonance imaging (MRI) are comparable in depicting the presence or absence of intraspinal pathology [44,48]. Spinal ultrasound provides rapid information on the bony sacrum, the spinal cord, filum terminale, and overlying soft tissue [15,49] and is a useful screening investigation during the first 3 months of life. MRI delineates the specific nature of the spinal abnormalities more clearly [1] and is the modality of choice in older children, or where ultrasound has failed.

The incidence of neurogenic bladder increases with the severity of the underlying ARM. Mosiello and colleagues observed a neurogenic bladder in 8 out of 39 patients with low lesions, 18 out of 45 patients with

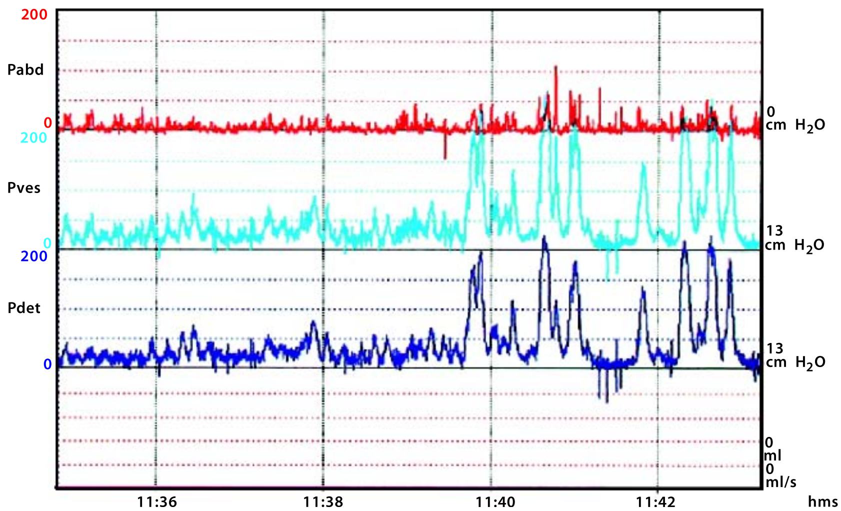

Fig. 17.3 Urodynamic tracing in a patient with an anorectal malformation. The bottom line represents the detrusor pressure. Marked instability, indicative of a neuropathic bladder, can be clearly seen. *Pabd* Abdominal pressure, *Pves* vesicular pressure, *Pdet* detrusor pressure

high lesions, and all 5 cloaca patients [32]. This was confirmed by Warne, who prospectively studied a group of 45 ARM patients by natural filling urodynamics and found that 58% of ARM patients and 83% of cloaca patients had bladder dysfunction on presentation (Table 17.4, Fig. 17.3) [49]. The high rate of bladder dysfunction in this series may be explained by the predominance of high lesions, and the incidence of bladder dysfunction increases with the severity of the ARM. These data suggest that a neurogenic bladder can be seen in all varieties of patients with ARM, either with or without an abnormal spine [32].

17.4.2.2 Postoperative Urinary Continence

The rate of urinary incontinence varies greatly in the literature depending on the severity of the original ARM and on the definitions used to describe continence. Overall incontinence rates in the literature vary from 10% up to 25% [3,4,37]. Rintala reviewed continence in adult high and intermediate ARM patients with mean age of 35 years and reported urinary incontinence in 33%, which greatly affected their quality of life [12]. The rate of incontinence in girls born with a cloacal anomaly can approach 60–70% [37,50].

17.4.2.3 Bladder Physiology

Denervation of the lower urinary tract has been observed following posterior sagittal anorectoplasty (PSARP) and with other techniques used to reconstruct ARM [2,4,8,11,55]. In prospective series, the incidence of new neurological damage following PSARP for anorectal malformations varies between 5 and 10% [2,49]. However, the rate of change is much greater in patients with a cloacal anomaly who undergo reconstruction using PSARP and total urogenital mobilization, with 50% showing deterioration in bladder function [50]. The bladder tended to change from an unstable bladder to a denervated, low-pressure, high-capacity bladder [49]. This change was more commonly noted when the length of the common channel was greater than 3 cm. This change is supported by the high incidence of patients requiring postoperative catheterization observed by Peña [37]. The deterioration in bladder function following surgery may be partially due to the pelvic autonomic plexus lying more midline and in close proximity to the fistula in these children than in those without ARM [13].
Constipation may also contribute to abnormal bladder function. More recently, Warne and colleagues observed that in 12 of 45 patients a wave of peristalsis could initiate an abnormal bladder contraction. This work suggests that there is a direct link between bowel motility and urinary incontinence.

17.4.2.4 Management of Urinary Incontinence

Unlike fecal incontinence, urinary incontinence impacts on other organ systems. Incontinence caused by a neurogenic bladder may be associated with recurrent urinary tract infections and vesicoureteric reflux. This combination can result in ongoing damage to the kidneys, resulting in renal failure [27]. Consequently, the early aggressive management of these patients is important to prevent renal damage. The important factors are to ensure that the bladder is emptied regularly and that the intravesical pressure remains low. Bladder emptying, when necessary, is best performed by clean intermittent catheterization (CIC). CIC can be performed either urethrally or via a Mitrofanoff stoma [6,24]. In the majority of patients with an ARM the urethra is sensate, consequently urethral CIC can be uncomfortable, especially in the older patient. By starting in the first 3 months of life, Boemers found a higher compliance rate [6]. Reducing bladder pressure should first be attempted medically using anticholinergic agents; when this fails, bladder augmentation may be necessary. These techniques can be used to achieve social continence in many of the patients [6].

17.5 Urethral Problems

Posterior urethral valves, megaurethra, and urethral duplication have been reported in association with ARM [28,41]. The most commonly reported urethral problems are iatrogenic and include urethral strictures, large diverticula, or remnants of the rectal pouch from incomplete dissection of the rectal fistula at the time of pull-through [8,37]. These complications are now seen less frequently with the development of PSARP, which allows good visualization of the urethral fistula. Stones may form if a urethral diverticulum is left, precipitating recurrent infections, and it can be technically difficult to catheterize the urethra in those patients who require CIC for neurogenic bladder [36,39].

17.6 Genital Anomalies

17.6.1 Male

Among 21 male ARM patients, abnormalities of the genitalia were identified in 11 (52%) [28]. An associated renal abnormality was detected in nine of these boys and in all male patients with hypospadias, bifid scrotum, bilateral undescended testes, or penoscrotal transposition. There were two boys with a unilateral undescended testis who had normal kidneys. The presence of a genital abnormality in boys with ARM appears to be associated with a renal abnormality [28].

17.6.1.1 Penis and Urethra

A penile abnormality has been described in 14–25 % of male ARM patients [28,42]. Hypospadias is the most common penile abnormality detected, but chordee, rotational anomalies of the penis, and epispadias are also reported. Megaurethra and ectopic urethra are uncommon, and penile duplication has occurred in a few patients [28].

17.6.1.2 Testicles and Scrotum

Cryptorchidism is a common finding and the incidence varies between 10 and 40% of male ARM patients [27,28,42]. McLorie reports that this is more common in high versus low malformations (27% vs 7%). A bifid scrotum commonly occurs and is often with the more severe variants of hypospadias. Penoscrotal transposition has also been described (Fig. 17.4) [27].

Fig. 17.4 A patient with penoscrotal transposition and hypospadias

17.6.1.3 Epididymis, Vas, and Ejaculatory Ducts

There is an increased incidence of epididymitis in ARM boys and this is seen prior to and after surgical closure of rectourinary fistula [8,35]. The exact mechanism for this is unknown, but abnormalities such as urethral strictures, ectopic ureters, diverticulum at the previous fistula site, and a neurogenic bladder may all be contributory factors. Ectopia of the vas has been reported [28], and Wolffian duct abnormalities are more common on the same side as renal anomalies [23]. The vas and seminal vesicles are prone to iatrogenic injury in patients with prostatic and bladder neck fistulas [11]. Holt et al. reported a high incidence of male infertility in adult males treated for ARM. Half of the groups studied were azospermic and there was a high incidence of ejaculatory problems [23]. This highlights the importance of treating recurrent epididymitis, performing early orchidopexy, and referring these patients to adolescent urology for assessment in early adult life.

17.6.2 Females

Mullerian abnormalities occur in 30–45 % of girls with ARM [17,19,30,31]. Cloacal anomalies are characterized by a confluence of the urethra, vagina, and rectum into a single channel, or persistent cloaca with a solitary opening on the perineum. This anomaly occurs as a result of a complex defect of perineal development, and is associated with maldevelopment of the Mullerian tubercle, sinovaginal bulbs, vaginal plate, and urogenital sinus. The resulting abnormalities show great variation depending on whether the confluence is high or low. Some degree of septation of the uterus and vagina, ranging from a partial septum in a large vagina with single cervix and uterus to a completely separated double vagina with double cervix and uteri, is seen in 60% of cloaca patients [25,33,37,53].

Abnormalities of the external genitalia are rare, but hamartomas and hemangiomas of the labia majora have both been observed.

17.7 Management Suggestions

17.7.1 Initial Evaluation

Since around 40% of ARM patients have a structural abnormality of the urinary tract, it is important to document these early in life and initiate treatment where appropriate. Ultrasound scan, cystogram, and isotope renogram should therefore be performed in all ARM patients at presentation [5]. In all patients with bilateral renal disease, GFR measurement should be assessed and joint management with nephrology specialists may be required. As 50% of cloaca patients develop renal impairment we would recommend that all new cloaca patients have a baseline GFR measurement at around 1 year of age [52].

Spinal status should be documented before assessing bladder function in all new ARM patients. All should have full vertebral radiographs in the anterior–posterior and lateral views, and either spinal ultrasound or MRI. Spinal ultrasound is useful as a screening tool; however, MRI is the most sensitive tool with which to detect spinal cord abnormalities [1]. If the investigation is performed early in infancy then sedation may not be necessary.

There should be a high index of suspicion for bladder dysfunction in all new ARM patients. This can be investigated by noninvasive bladder function assessment, as described by Holmdahl and Mosiello, within the first few months of life [22,32]. This will probably suffice for patients with low malformations, who have a low incidence of bladder dysfunction. However, all other ARM patients, particularly those with high malformations, persistent cloaca, and those with documented spinal pathology, should have formal urodynamics assessment by conventional cystometrogram or natural filling techniques [4,49,57].

17.7.2 Postreconstruction Evaluation

As bladder function does not change following PSARP, the urodynamic study can be performed at any time during the 1st year of life providing the initial bladder function assessment showed good bladder emptying and the child does not suffer from urinary tract infections. In cloaca patients there is a frequent association with bladder dysfunction and as there may be delay before cloaca reconstruction, it is recommended that all cloaca patients should be screened using urodynamics within the first few months of life. Since a high proportion of the cloaca group showed deterioration after surgical reconstruction, bladder function should be reassessed again postoperatively by urodynamics [49].

17.7.3 Long-Term Evaluation

All ARM patients who are diagnosed with a genitourinary malformation, spinal cord abnormality, or bladder dysfunction on initial assessment, and all cloaca patients require regular review. Patients with renal abnormalities and vesicoureteric reflux should have serial ultrasound scans to monitor their renal status. In patients suffering from urinary tract infection, whatever the etiology, a Tc-99m dimercaptosuccinic acid renogram is recommended to diagnose renal scarring.

Gynecological abnormalities are common, especially in cloaca patients [25,30,53], but may remain asymptomatic until puberty or adult life (Fig. 17.5). All cloaca patients and those with documented Mullerian and vaginal anomalies should be reassessed and monitored in the peripubertal period. Ultrasound scan of the pelvis is a useful investigation for screening [34], whereas an MRI scan is the investigation of choice to document the complex anatomy in these patients, particularly when reconstructive surgery is necessary [29]. Examination under anesthesia and vaginoscopy are also recommended to assess vaginal patency, adequacy, and the presence of a cervix (cervices). Findings previously documented in infancy may be misleading. An apparently vestigial uterus may develop enough to produce menstrual flow and hence obstruction if the genital tract is not patent [53]. The size and adequacy of the vagina in proportion to the size of the child may also appear to have changed at puberty, so it is of utmost importance to reassess the patient at this stage.

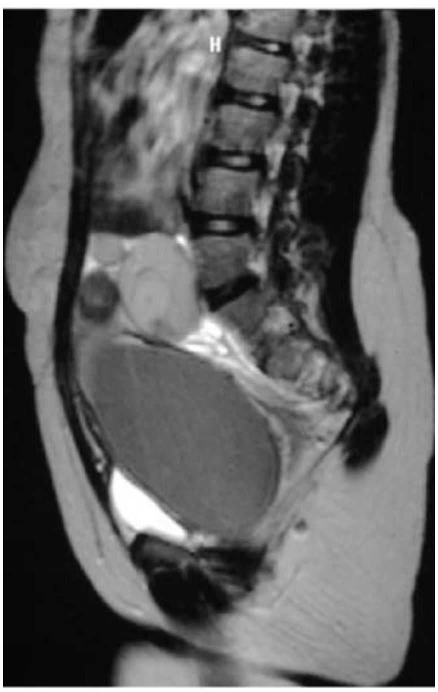

Fig. 17.5 Magnetic resonance imaging scan showing an obstructed uterus presenting as hematocolpos. The bladder, with contrast in it, can be seen anteriorly to the distended uterus

References

1. Beek FJ, Boemers TM, Witkamp TD, et al (1995) Spine evaluation in children with anorectal malformations. *Pediatr Radiol* **25**:S28–32
2. Boemers TM, Bax KM, Rovekamp MH, van Gool JD (1995) The effect of posterior sagittal anorectoplasty and its variants on lower urinary tract function in children with anorectal malformations. J Urol 153:191–193
3. Boemers TM, Beek FJ, van Gool JD, et al (1996) Urologic problems in anorectal malformations. Part 2: Functional urologic sequelae. J Pediatr Surg 31:634–637
4. Boemers TM, Beek FJ, van Gool JD, et al (1996) Urologic problems in anorectal malformations. Part 1: Urodynamic findings and significance of sacral anomalies. J Pediatr Surg 31:407–410
5. Boemers TM, Beek FJ, Bax KM (1999) Review. Guidelines for the urological screening and initial management of lower urinary tract dysfunction in children with anorectal malformations – the ARGUS protocol. BJU Int 83:662–671
6. Boemers TM (1999) Neurogenic bladder in infants born with anorectal malformations: comparison with spinal and urologic status. J Pediatr Surg 34:1889–1890
7. Borzyskowskyi M, Mundy A (1988) The management of neuropathic bladder in childhood Pediatr Nephrol 2:56–66
8. Brock WA, Peña A (1992) Cloacal abnormalities and imperforate anus. In: Kelais PP, King LR, Belman AB (eds) Clinical Pediatric Urology, 3rd edn. WB Saunders, Philadelphia, Vol 2, Chap 19, pp 920–942
9. Carson J Barnes P, Tunnell W, et al (1984) Imperforate anus: the neurologic implication of sacral abnormalities. J *Pediatr* Surg 19:838–842
10. Churchill BM, Hardy BE, Stephens CA (1978) Urologic aspects of malformations and common abnormalities of the anus and rectum. Urol Clin North Am 5:141–154
11. Davies M, Kiss A (1994) Intraoperative damage to the male urethra and pelvic visceral nerves during posterior sagittal anorectoplasty. Pediatr Surg Int 9:8–11
12. Davies MC, Creighton SM, Wilcox DT (2004) Long-term outcomes of anorectal malformations. Pediatr Surg Int 20:567–572

13. Davies MR (1997) Anatomy of the nerve supply of the rectum, bladder, and internal genitalia in anorectal dysgenesis in the male. J Pediatr Surg 32:536–541
14. Dhillon HK (1998) Prenatally diagnosed hydronephrosis: the Great Ormond Street experience. Br J Urol 81 Suppl 2:39–44
15. Dick EA, Patel K, Owens CM, de Bruyn R (2002) Spinal ultrasound in infants. Br J Radiol 75:384–892
16. Endo M, Hayashi A, Ishihara M, Maie M, Nagasaki A, Nishi T, Saeki M (1999) Analysis of 1,992 patients with anorectal malformations over the past two decades in Japan. Steering Committee of Japanese Study Group of Anorectal Anomalies. J Pediatr Surg 34:435–441
17. Fleming S, Hall R, Gysler M, McLorie G (1986) Imperforate anus in females: frequency of genital tract involvement. J Pediatr Surg 21:146–150
18. Geraniotis E, Koff SA, Enrile B (1988) The prophylactic use of clean intermittent catheterization in the treatment of infants and young children with myelomeningocele and neurogenic bladder dysfunction. *J Urol* **139**:85–86
19. Hall JW, Fleming S, Gysler M, McLorie G (1985) The genital tract in female children with imperforate anus. Surg Gynecol Obstet 151:169–171
20. Hendren WH (1998) Urological aspects of cloacal malformations. J Urol 140:1207–1213
21. Hendren WH (1998) Cloaca, the most severe degree of imperforate anus. Ann Surg 228:331–346
22. Holmdahl G, Hansson E, Hansson M, et al (1996) Four-hour voiding observation. J Urol 156:1809–1812
23. Holt B, Pryor JP, Hendry W (1995) Male infertility after surgery for imperforate anus. J Pediatr Surg 30:1677–1679
24. Kaefer M, Pabby A, Kelly M, et al (1999) Improved bladder function after prophylactic treatment of the high risk neurogenic bladder in newborns with myelomeningocele. J Urol 162:1068–1071
25. Levitt MA, Stein DM, Peña A 1998 Gynecologic concerns in the treatment of teenagers with cloaca. J Pediatr Surg 33:188–193
26. Maizels M, Simpson S (1983) Primitive ducts of renal dysplasia induced by culturing ureteral buds denuded of condensed renal mesenchyme. Science 219:509–510
27. McLorie G, Sheldon M, Fleisher M, et al (1987) The genitourinary system in patients with imperforate anus. J Pediatr Surg 22:1100–1104
28. Metts J, Kotkin K, Kasper S, et al (1997) Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. J Urol 158:1298–1300
29. Minto CL, Hallings N, Hall-Craggs M, Creighton SM (2001) Magnetic resonance imaging in the assessment of complex Mullerian anomalies. BJOG 108:791–797
30. Mollitt DL, Schullinger JN, Santulli TV (1981) Complications at menarche of urogenital sinus with associated anorectal malformations. J Pediatr Surg 16:349–352
31. Moore K (1983) The Developing Human: Clinically Orientated Embryology, 3rd edn. WB Saunders, Philadelphia, p262
32. Mosiello G, Capitanucci ML, Gatti C, et al (2003) How to investigate neurovesical dysfunction in children with anorectal malformations. J Urol 170:1610–1613
33. Nussbaum-Blask A, Sanders R, Rock J (1991) Obstructed uterovaginal anomalies: demonstration with sonography (Part II. Teenagers). Radiology 179:84–88
34. Parrott TS, Woodward JR (1979) Importance of cystourethrography in neonates with imperforate anus. Urology 8:607–609
35. Parrott TS (1985) Urologic implications of anorectal malformations. Urol Clin N Am 12:13–25
36. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
37. Peña A, Hong A (2000) Advances in the management of anorectal malformations. Am J Surg 180:370–376
38. Persky L, Tucker A, Izant RJ (1974) Urological complications of correction of imperforate anus. J Urol 111:415–418
39. Quan L, Smith DW (1973) The VATER association. J Pediatr 82 104–108
40. Ratan SK, Rattan KN, Pandey RM, Mittal A, Magu S, Sodhi PK (2004) Associated congenital anomalies in patients with anorectal malformations – a need for developing a uniform practical approach. J Pediatr Surg 39:1706–1711
41. Rich M, Brock W, Peña A (1988) Spectrum of genitourinary malformations in patients with cloacal malformations. Pediatr Surg Int 3:110–113
42. Rickwood AMK, Spitz L (1980) Primary vesicoureteric reflux in neonates with imperforate anus. Arch Dis Child 55:149–150
43. Rohrschneider WK, Forsting M, Darge K, Troger J (1996) Diagnostic value of spinal US: comparative study with MR imaging in pediatric patients. Radiology 200:383–388
44. Santulli T Schullinger Keisewetter, et al (1971) Imperforate anus: a survey from members of the surgical section of the American Academy of Pediatrics. J Pediatr Surg 6:484–486
45. Sidi AA, Dykstra DD, Gonzalez R (1986) The value of urodynamic testing in the management of neonates with myelodysplasia. J Urol 135:90–93
46. Stephens FD, Smith ED (1971) Ano-rectal Malformations in Children. Year Book Medical, Chicago
47. Stephens FD, Smith ED (1986) Classification, identification, and assessment of surgical treatment of anorectal anomalies. Pediatr Surg Int 1:200
48. Unsinn K, Geley T, Freund M, Gassner I (2000) Ultrasound of the spinal cord in newborns. Radiographics 20:923–938
49. Warne SA, Godley ML, Wilcox DT (2004) Surgical reconstruction of cloacal malformation can alter bladder function: a comparative study with anorectal anomalies. J Urol 172:2377–2381

50. Warne SA, Wilcox DT, Ransley PG (2002) Long-term urological outcome in patients presenting with persistent cloaca. J Urol 168:1859–1862
51. Warne SA, Godley ML, Wilcox DT (2003)Urological status in children with anorectal malformations – are there gender differences? Abstracts of the XIVth ESPU, Madrid, Spain, 12–15 March, 2003. BJU Int 91:12
52. Warne SA, Wilcox DT, Ledermann SE, Ransley PG (2002) Renal outcome in patients with cloaca. J Urol 167:2548–2551
53. Warne SA, Wilcox DT, Creighton S, Ransley PG (2003) Long-term gynecological outcome of patients with persistent cloaca. J Urol 170:1493–1496
54. Wiener ES, Kieswetter WB (1973) Urologic abnormalities associated with imperforate anus. *J Pediatr Surg* **8**:151–157
55. Williams DI, Grant J (1969) Urological complications of imperforate anus. *Br J Urol* **41**:660–665
56. Winter RM, Baraitser M (2000) London Dysmorphology Database. Oxford University Press, Oxford, UK
59. Yeung CK, Godley M, Duffy PG, Ransley PG (1995) Natural filling cystometry in infants and children. BJU Int 75:531–537

18 Tethered Spinal Cord in Patients with Anorectal Malformations

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18.1 Prevalence

Anorectal malformations (ARM) are commonly associated with maldevelopment of neighbouring structures derived from the caudal cell mass. Bony spinal anomalies and malformations of the spinal cord are among the more frequent findings. The association of ARM and tethered cord has been repeatedly documented [1–5]. The reported prevalence of spinal cord abnormalities in patients with ARM varies between around 10% (4/44 [5], 15/106 [6], 22/223 [7]), and 50% and more (40/76 [8], 25/50 [9], 54/89 [10]). A prevalence of between 20 and 35% appears most plausible (27/111 [2], 22/63 [11]). The majority of tethered cord lesions will be missed unless routine screening with ultrasound and magnetic resonance imaging (MRI) is applied, as was shown by Tuuha et al. in a retrospective 10-year analysis. With systematic screening, the percentage of spinal cord abnormalities found jumped from 4% to 20% [7]. In surgical series of tethered cord, associated ARM are comparatively rare (7/435 [7], 35/480 own experience). Historically, a risk stratification subdividing between low, intermediate and high level of ARM has been proposed, with supposedly higher incidence in high versus low lesions [1,6,12]. However, similar incidences of tethered cord were found in those subgroups in more recent surveys [7,9,10]; one study even recorded a higher incidence in low lesions [11]. The incidence of intraspinal anomalies is significantly higher in patients with anomalies of the sacrum on plain x-rays, typically hypoplasia of lower sacral segments [2,13]. Classification of ARM is a complex topic that is covered extensively in this book. While the frequent association of cloacal exstrophy and spinal cord malformation has long been established [1,8,14], there are no comprehensive assessments so far. There are strong indications in line with embryological considerations, however, that the prevalence of tethered cord is correlated with the severity of the ARM, as was proposed and demonstrated by Peña's workgroup [2]. Tethered cord prevalence was found in 11% of vestibular versus 18% of bulbar and prostatic fistulas, 27% of cloacal common channels, 40% of cloacal exstrophies, and 43% of complex malformations [2]. The risk of tethered cord increased with the number of associated anomalies. Presacral mass, sacral hemivertebrae, and single or ectopic kidney were each associated with tethered cord in 50–60% of cases, reflux only in 10% [2].

18.2 Clinical Presentation, Diagnosis

Closed spinal dysraphism reveals itself through skin abnormalities in 50% to more than 80% of cases [15–18]. In the context of ARM, vascular nevus, lumbosacral subcutaneous mass, skin dimple and deviation of the natal cleft should be searched for. The manifestations of tethered cord have rightfully been called protean by Hoffman and coworkers in 1976 [19]. Presenting symptoms may include motor and sensory deficits in the lower extremities, gluteal and genital region, leg or back pain (especially in older children and adults), foot deformities, leg length discrepancies and scoliosis (neuro-orthopaedic syndrome) or bladder and bowel dysfunction (Fig. 18.1). The majority of our patients had abnormal findings in one or several of these aspects (27/35), but only a minority (6/35) presented with clear-cut progressive symptoms. Most authors agree, that faecal incontinence and urinary abnormalities are more common in patients with tethered cord [2,5,20], whereas constipation is more common in ARM without tethered cord [2]. When the type of ARM is accounted for, faecal incontinence

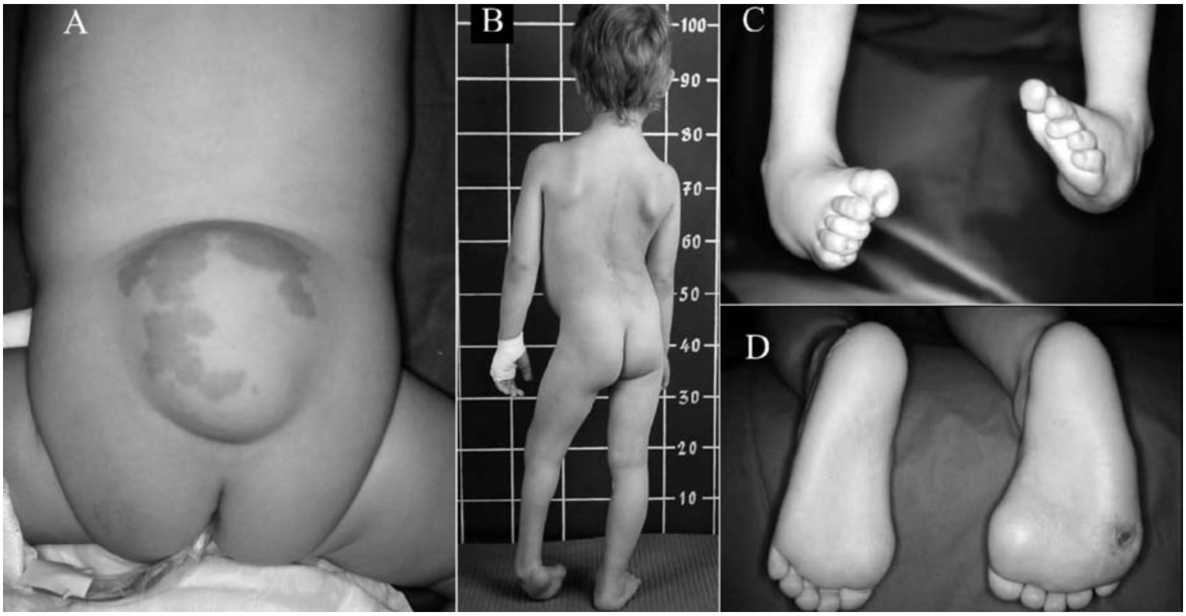

Fig. 18.1 Signs and symptoms of tethered cord. **A** Subcutaneous mass and reddish vascular naevi point to a lumbosacral lipoma of the spinal cord. **B** Neuro-orthopaedic syndrome. **C** Club feet. **D** Pressure sore due to sensory loss and trophic problems on deformed foot

and constipation are similar; only urinary dysfunction remains slightly more common [2]. This relationship and the commonly found dysplasia of the sacral bone point to maldevelopment of the respective innervation rather than secondary factors.

Abnormalities of the sacral bone and lumbar spine point to the presence of intraspinal pathology, but plain x-rays are less sensitive than ultrasound and MRI and are therefore no longer advocated as a screening procedure for intraspinal pathology. All neonates with ARM should be subjected to an ultrasound examination of the spine and spinal canal, with emphasis on conus shape and position as well as exclusion of irregular intraspinal tissue and abnormal fluid collections (syrinx, arachnoid cyst, meningocele, myelocystocele). An MRI is indicated at 3 months of age if either ultrasound or clinical signs (skin lesion, neurological or neuro-orthopaedic abnormalities of the lower extremities) are positive. In the neonatal period, the image quality of MRI is significantly reduced. From a neurosurgical standpoint, early MRI is indicated only in cases requiring early intervention, such as large myelocystoceles precluding supine positioning of the infant. Whether all infants with negative ultrasound and without clinical signs and symptoms should still be screened with MRI at about 1 year of age is not entirely clear. The sensitivity of MRI is higher than that of ultrasound, estimated at 95.6% and 86.5%, respectively. Specificity is similar, at 90.9% and 92.9%, respectively [21,22]. It appears prudent, therefore, to screen all patients using MRI, at least those patients with higher risk (e.g. cloacal and complex malformations).

18.3 Pathophysiology, Malformation Entities

Tethered cord is not synonymous with malformation of the spinal cord. In caudal regression syndrome, absence of the lower sacral spinal cord segments and according neurological deficits often occur without associated tethering lesions. Tethered cord describes a potentially harmful pathological fixation of the spinal cord, either directly or via inelastic tissue (bone, fat, fibrous or fatty filum terminale). It is typically located in the lumbosacral region and most commonly caused by spinal cord malformations. The negative impact on the spinal cord is believed to result from a stretching effect brought about by everyday activities, especially flexion of the spine and pelvis. Elongation has been demonstrated to be most severe at the point of fixation and extends upward for up to five segments, but not beyond the lowest insertion of the dentate ligament at the thoracolumbar junction. Such traction on the spinal cord causes impairment of capillary blood flow and results in reduced oxygenation of the spinal cord. Experimental studies in animals have shown a

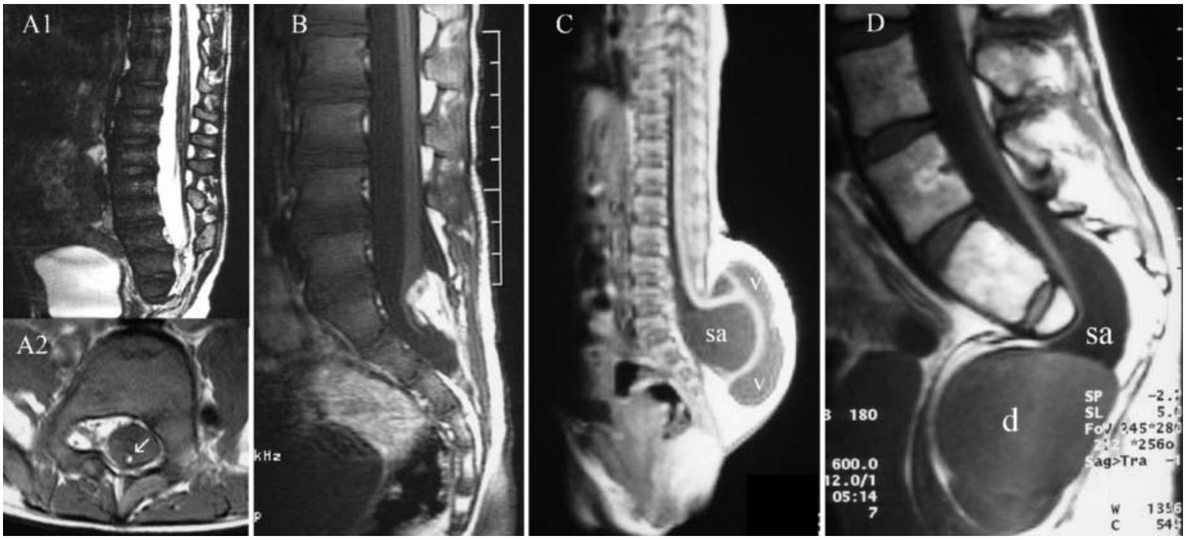

Fig. 18.2 Magnetic resonance imaging findings typical for tethered cord in patients with anorectal malformations. **A** Thickened filum terminale (see *arrow* in **A2**), absence of the lower segments of the sacral bone. **B** Lipoma of the spinal cord (*arrow*). **C** Terminal myelocystocele. Note the terminal ventricle (*v*) dorsal to the malformed spinal cord, which is pushed out far above the fascial plane by an enlarged subarachnoid space (*sa*). **D** Ventral sacral meningocele with large dermoid (*d*) adjacent to the subarachnoid space

close correlation between the severity and duration of traction, and reduction of blood flow, deterioration of interneuron potentials and neurological deficits [23]. These results have been corroborated by intraoperative measurements of spinal cord blood flow [24] and cytochrome-C redox state [23]. Intraoperative improvement of those parameters was correlated with the neurological outcome.

Spinal cord malformations causing tethered cord are a heterogeneous group of lesions resulting from maldevelopment of mesoderm invagination and preneurulation midline formation, errors of primary and secondary neurulation and post-neurulation events. As would be expected, the spectrum of entities associated with ARM consists of the subgroup related to secondary neurulation and subsequent processes (see Fig. 18.2 for typical MRI findings). Most common is a thickened, shortened or fatty filum terminale, followed by lumbosacral lipoma or lipomyelomeningocele. Other lesions are terminal myelocystocele and ventral sacral meningocele. Caudal regression syndrome, also labelled caudal suppression syndrome or, better, caudal dysplasia syndrome, occurs without tethering lesion or in combination with the lesions mentioned above. The complexity of these lesions and their impact on neurological function, natural history, difficulty of surgical repair, outcome and related long-term problems varies enormously, similar to the spectrum of ARM.

Pathological filum is the prototype of pure spinal cord tethering. Dysplasia of the spinal cord and cauda is minor or absent, unless caused by associated malformations. The dura is intact, the arachnoid membranes only slightly abnormal if at all. Patients are less likely to have neonatal neurological deficits, deteriorate later than patients with more severe malformations, but continue to be at risk for deterioration throughout their life [25,26].

Spinal cord lipomas and lipomyelomeningoceles occur in a wide spectrum of mild to severe cases in terms of malformation size, degree of spinal cord involvement, extent of dysplasia of the spinal cord and cauda and intensity of spinal cord tethering and/or compression. At birth, about 20% have functional deficits attributable to the malformation, the majority will deteriorate during infancy and childhood [27–30].

Terminal myelocystocele is a malformation that is not rarely associated with cloacal malformations [31,32]. It has all the features of a lipomyelomeningocele (i.e. extension of the spinal cord into a meningocele, where it is infiltrated by and tethered to fat tissue). In addition, the central canal of the spinal cord is distended into a terminal ventricle, which forms another component of the cele. The spinal cord tends to be both dysplastic and hypertrophic at the level of the malformation, a syrinx may extend upwards into functionally normal spinal cord segments. Patients tend to be more severely affected at birth and throughout life than average spinal cord lipoma patients.

Ventral sacral meningocele together with ARM and dysplasia of the sacral bone (scimitar sacrum) often presents as a component of the Currarino syndrome. A high proportion of cases are hereditary with autosomal dominant inheritance [33]. Tethered cord is a common finding in ventral sacral meningoceles. The spinal cord is attached to the caudal wall of the meningocele, either directly or by way of a pathological filum terminale. The spinal cord can be elongated to an extent that it may be addressed as a mere pathological filum on imaging. Dermoid tumours are commonly associated, located in or adjacent to the caudal wall of the meningocele. Comorbidities other than faecal abnormalities are uncommon. Early progression of tethered cord syndrome is uncommon, but we have seen some cases with deterioration even in adult age.

18.4 Tethered Cord Surgery

The decision for surgical treatment of tethered cord in patients with ARM is controversial. Patients suffering from progressive motor or sensory deficits are unanimously considered good candidates for surgery [7–9,11]. The relationship, however, between faecal and urinary abnormalities and spinal cord tethering is highly questionable in the context of caudal dysplasia with ARM [2,13]. The natural history of patients with ARM appears to harbour only a low risk of neurological deterioration [2,7]. Tuuha found that only 2% of 435 patients with ARM had tethered cord symptoms and only 18% of the subgroup were diagnosed with tethered cord [7]. It is concluded by some authors, therefore, that patients with ARM may form a subgroup of tethered cord patients less prone to deterioration who may not profit enough from untethering to outweigh the risks of this procedure [2,7], while other authors and most of the paediatric neurosurgical community keep recommending prophylactic surgery for all tethered cord patients [9,34]. When tethered cord is diagnosed late and the patient has been stable so far, further observation is preferred. Warf et al. did so in patients beyond 10 years of age [8]; non-progressive patients should certainly not be operated on in adult age [35]. The controversy regarding prophylactic surgery is less applicable for terminal myelocystocele, where large lumps on the back and the threat of direct transcutaneous pressure transmission through the terminal ventricle and syrinx into the normal lumbar spinal cord warrant early intervention. Expanding intrapelvic meningocele or detection of an inclusion tumour are additional reasons for prophylactic surgery in ventral sacral meningoceles. Constipation is not an indication for surgery, at least in our experience, since we never saw improvement after untethering plus closure or removal of the meningocele.

18.5 Results/Outcome

Data on the outcome of tethered cord surgery specific for patients with ARM remain scarce. There is no reported mortality, and permanent morbidity such as injury to neurological or urological function, while certainly possible, appears to be low. Specifically, there is no reported iatrogenic deficit in 6 series with an accumulated number of 87 patients [1,2,7,8,11,34], which parallels our own results in 35 patients. Motor and sensory deficits usually stop deteriorating. Postoperative motor improvement is reported in a cumulative 8/15 patients [1,2,7,8]. Unanimously, however, very little influence of surgery on manifest urinary or faecal incontinence has been found, as is our experience. A surprising exception is the paper by Muthukumar, who reported improvement of urinary, but not faecal incontinence in all eight patients operated on, without giving details or plausible explanations [34]. A 9% rate (2/22) of late deterioration ascribed to retethering has been reported in one series [11].

In the general tethered cord population, the complication rate and long-term results are significantly different in patients with pathological filum compared to lipomas of the conus. In the former, surgery is straightforward and the rate of neurological injury approaches zero in large series. Secondary deterioration during follow-up is rare [29,30]. Deficits present at the time of surgery will usually stop worsening, but will improve in only 53% of cases [29] and resolve completely in only 19–41% [29,30]. Prophylactic surgery of pathological filum terminale and of lipomas of the filum therefore continues to be unanimously recommended [29,30]. Conus lipoma (and lipomyelomeningocele), on the other hand, often pose significant surgical problems; complete untethering is impossible in up to 20% of the patients and the risk of neurological injury is not negligible, at around 4% [29]. Figure 18.3 illustrates the different situations encountered intraoperatively. There is a disturbingly high rate of secondary deterioration during follow-up, ranging from 28% [30] to 36% after 5 years or more [29]. Some have been moved by these results to offer surgery only when symptoms evolve [29], while most

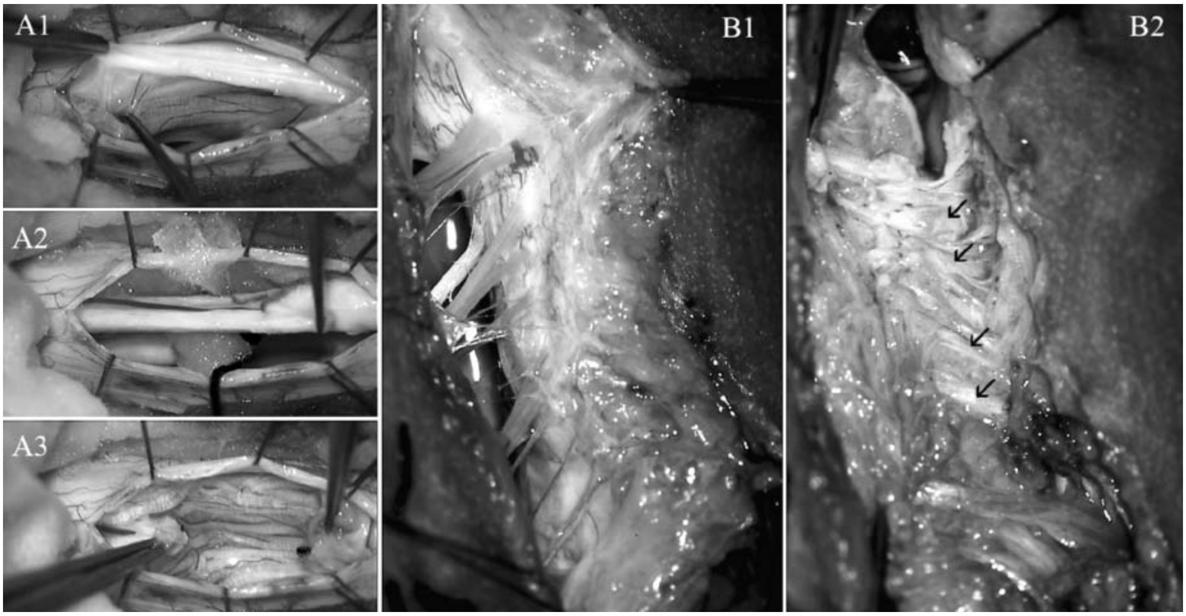

Fig. 18.3 **A1–3** Separation and division of thickened filum terminale. **A3** The divided filum retracted by 10–15 mm, the ends have been pulled back into view by forceps. **B** Lipoma of the spinal cord. Complete untethering is impossible because several segments of nerve roots are inseparably incorporated into the malformation on the right side (**B2**). Free, but shortened subarachnoid course of the nerve roots shown after untethering on the less affected side (**B1**)

of the neurosurgical community continue to advocate prophylactic surgery. The main arguments for that position remain that existing symptoms are only stabilized after surgery in the majority of patients, rather than reversed, and that deterioration over time, while not completely preventable, appears to be mitigated [30]. In our experience (122 patients, unpublished data), deterioration during a 5-year follow-up period was less common after complete untethering (16%) than after incomplete untethering (48%).

18.6 Conclusion

Spinal cord malformations and tethered cord are no less heterogeneous a group as are ARM. While the specific aspects of tethered cord in patients with ARM warrant more extensive investigation, these patients cannot be adequately addressed and treated as a homogeneous subgroup of tethered cord. Existing data are insufficient to either reject or mandate prophylactic tethered cord surgery conclusively. The true natural history remains unknown. The lifelong deterioration risk, sufficiently demonstrated for the general tethered cord population, however, cannot be completely denied, although its incidence and dynamics need further quantification. Prospective studies, while welcome, would need to span decades to properly balance the risks and benefits of operative versus conservative treatment. Since the risk:benefit ratio of tethered cord surgery is so much better and the retethering risk so much lower for pathological filum compared to lipomas of the spinal cord, prophylactic surgery appears to be difficult to reject at least for this subgroup.

References

1. Karrer FM, Flannery AM, Nelson MD Jr, McLone DG, Raffensperger JG (1988) Anorectal malformations: evaluation of associated spinal dysraphic syndromes. J Pediatr Surg 23:45–48
2. Levitt MA, Patel M, Rodriguez G, Gaylin DS, Peña A (1997) The tethered spinal cord in patients with anorectal malformations. J Pediatr Surg 32:462–468
3. Sato S, Shirane R, Yoshimoto T (1993) Evaluation of tethered cord syndrome associated with anorectal malformations. Neurosurgery 32:1025–1027
4. Carson JA, Barnes PD, Tunell WP, Smith EI, Jolley SG (1984) Imperforate anus: the neurologic implication of sacral abnormalities. J Pediatr Surg 19:838–842
5. Davidoff AM, Thompson CV, Grimm JM, Shorter NA, Filston HC, Oakes WJ (1991) Occult spinal dysraphism in patients with anal agenesis. J Pediatr Surg 26:1001–1005

6. Tunell WP, Austin JC, Barnes PD, Reynolds A (1987) Neuroradiologic evaluation of sacral abnormalities in imperforate anus complex. J Pediatr Surg 22:58–61
7. Tuuha SE, Aziz D, Drake J, Wales P, Kim PC (2004) Is surgery necessary for asymptomatic tethered cord in anorectal malformation patients? J Pediatr Surg 39:773–777
8. Warf BC, Scott RM, Barnes PD, Hendren WH III (1993) Tethered spinal cord in patients with anorectal and urogenital malformations. Pediatr Neurosurg 19:25–30
9. Rivosecchi M, Lucchetti MC, Zaccara A, De GM, Fariello G (1995) Spinal dysraphism detected by magnetic resonance imaging in patients with anorectal anomalies: incidence and clinical significance. J Pediatr Surg 30:488–490
10. Mosiello G, Capitanucci ML, Gatti C, et al (2003) How to investigate neurovesical dysfunction in children with anorectal malformations. J Urol 170:1610–1613
11. Golonka NR, Haga LJ, Keating RP, et al (2002) Routine MRI evaluation of low imperforate anus reveals unexpected high incidence of tethered spinal cord. J Pediatr Surg 37:966–969
12. Long FR, Hunter JV, Mahboubi S, Kalmus A, Templeton JM Jr(1996) Tethered cord and associated vertebral anomalies in children and infants with imperforate anus: evaluation with MR imaging and plain radiography. Radiology 200:377–382
13. Taskinen S, Valanne L, Rintala R (2002) Effect of spinal cord abnormalities on the function of the lower urinary tract in patients with anorectal abnormalities. J Urol 168:1147–1149
14. Tank ES (1970) Latent anomalies associated with imperforate anus. Arch Surg 100:512–514
15. Drolet BA (2000) Cutaneous signs of neural tube dysraphism. Pediatr Clin North Am 47:813–823
16. Hall DE, Udvarhelyi GB, Altman J (1981) Lumbosacral skin lesions as markers of occult spinal dysraphism. JAMA 246:2606–2608
17. Humphreys RP (1996) Clinical evaluation of cutaneous lesions of the back: spinal signatures that do not go away. Clin Neurosurg 43:175–187
18. Schropp C, Sorensen N, Collmann H, Krauss J (2006) Cutaneous lesions in occult spinal dysraphism–correlation with intraspinal findings. Childs Nerv Syst 22:125–131
19. Hoffman HJ, Hendrick EB, Humphreys RP (1976) The tethered spinal cord: its protean manifestations, diagnosis and surgical correction. Childs Brain 2:145–155
20. Kaplan WE, McLone DG, Richards I (1988) The urological manifestations of the tethered spinal cord. J Urol 140:1285–1288
21. Santiago ML, al-Orfali M, Zurakowski D, Poussaint TY, DiCanzio J, Barnes PD (1999) Occult lumbosacral dysraphism in children and young adults: diagnostic performance of fast screening and conventional MR imaging. Radiology 211:767–771
22. Rohrschneider WK, Forsting M, Darge K, Troger J (1996) Diagnostic value of spinal US: comparative study with MR imaging in pediatric patients. Radiology 200:383–388
23. Yamada S, Zinke DE, Sanders D (1981) Pathophysiology of "tethered cord syndrome". J Neurosurg 54:494–503
24. Schneider SJ, Rosenthal AD, Greenberg BM, Danto J (1993) A preliminary report on the use of laser-Doppler flowmetry during tethered spinal cord release. Neurosurgery 32:214–217
25. Yamada S, Won DJ, Yamada SM, Hadden A, Siddiqi J (2004) Adult tethered cord syndrome: relative to spinal cord length and filum thickness. Neurol Res 26:732–734
26. Hendrick EB, Hoffman HJ, Humphreys RP(1983) The tethered spinal cord. Clin Neurosurg 30:457–463
27. Hoffman HJ, Taecholarn C, Hendrick EB, Humphreys RP (1985) Lipomyelomeningoceles and their management. Concepts Pediatr Neurosurg 5:107–117
28. Kanev PM, Bierbrauer KS (1995) Reflections on the natural history of lipomyelomeningocele. Pediatr Neurosurg 22:137–140
29. Pierre-Kahn A, Zerah M, Renier D, et al (1997) Congenital lumbosacral lipomas. Childs Nerv Syst 13:298–334
30. LaMarca F, Grant JA, Tomita T, McLone DG (1997) Spinal lipomas in children: outcome of 270 procedures. Pediatr Neurosurg 26:8–16
31. Byrd SE, Harvey C, Darling CF (1995) MR of terminal myelocystoceles. Eur J Radiol 20:215–220
32. Weaver KB, Matthews H, Chegini S, King H, Shurtleff DB, McLaughlin JF (1997) Vertebral column and spinal cord malformation in children with exstrophy of the cloaca, with emphasis on their functional correlates. Teratology 55:241–248
33. Lynch SA, Bond PM, Copp AJ, et al (1995) A gene for autosomal dominant sacral agenesis maps to the holoprosencephaly region at 7q36. Nat Genet 11:93–95
34. Muthukumar N, Subramaniam B, Gnanaseelan T, Rathinam R, Thiruthavadoss A (2000) Tethered cord syndrome in children with anorectal malformations. J Neurosurg 92:626–630
35. Hüttmann S, Krauss J, Collmann H, Sorensen N, Roosen K (2001) Surgical management of tethered spinal cord in adults: report of 54 cases. J Neurosurg 95:173–178

Initial Management

19 Management in the Newborn Period

Marc A. Levitt and Alberto Peña

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19.1 Males

Figure 19.1 shows the decision-making algorithm for the initial management of male patients with anorectal malformations. When one is called to see a newborn male with an anorectal malformation, a thorough perineal inspection must be performed. This usually gives the most important clues about the type of malformation that the patient has. It is important not to make a decision about colostomy or primary operation before 24 h of life because significant intraluminal pressure is required for the meconium to be forced through a fistula orifice, which is the most reliable sign of the location of the fistula. If meconium is seen on the perineum, it is evidence of a perineal fistula. If there is meconium in the urine, a rectourinary fistula is present.

Radiologic evaluations do not show the real anatomy before 24 h because the rectum is collapsed and does not yet have enough intraluminal pressure to overcome the muscle tone of the sphincters that surround it. Therefore, radiologic evaluations done too early (before 24 h) will most likely reveal the false impression of a "very high rectum."

During the first 24 h, the baby should receive intravenous fluids and antibiotics. The presence of associated defects should be investigated. These include cardiac conditions, esophageal atresia, duodenal atresia, and urologic and spinal defects. An echocardiogram of the heart can be taken. A nasogastric tube is passed to detect the presence of esophageal atresia. An x-ray film of the lumbar spine and the sacrum are helpful

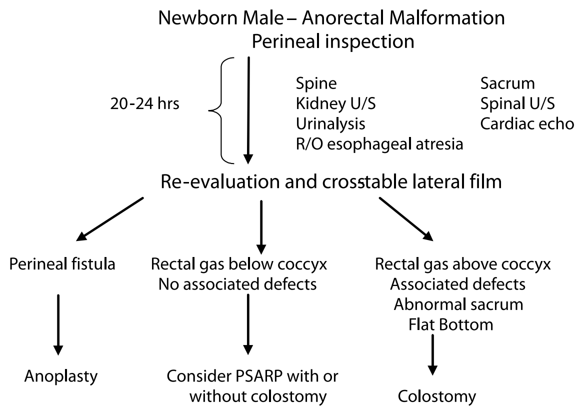

Fig. 19.1 Decision-making algorithm for male newborns with anorectal malformations (ARM). *U/S* Ultrasound, *PSARP* posterior sagittal anorectoplasty, *R/O* rule out (Reprinted from Pediatric Surgery, 4th ed., Ashcraft, Whitfield & Murphy eds. Peña A, Levitt MA. Imperforate Anus and Cloacal Malformations, p 501, Elsevier Saunders, Philadelphia (2005), with permission from Elsevier.)

to determine the presence of associated spinal and sacral anomalies. A spinal ultrasound in the newborn period is a good screen for tethered cord and other spinal anomalies. An ultrasound of the abdomen will rule out the presence of hydronephrosis [1]. If the baby has signs of a perineal fistula, an anoplasty can be performed during the first 48 h of life without a protective colostomy (Fig. 19.2). If the baby's clinical condition warrants waiting to do surgery, such as for a premature baby or one with an associated cardiac defect, dilatations of the fistula with a delayed repair is acceptable. Such a baby must be watched closely to be sure that the colon is adequately emptying through the fistula. After 24 h, if no meconium is seen on the perineum, a cross-table, lateral x-ray film with the baby in a prone position often shows the location of the distal rectum. If the gas in the rectum is located below the coccyx and the baby is in a good condition with no significant associated defects, depending on the surgeon's experience, a posterior sagittal opera-

Fig. 19.2 Newborn anoplasty

Fig. 19.3 Radiograph of cross-table lateral x-ray

tion without a protective colostomy can be considered (Fig. 19.3). If the rectal gas is seen located above the coccyx and the patient has meconium in the urine, or has significant associated defects, an abnormal sacrum, or a flat bottom, a colostomy is the safest approach, with postponement of the main repair for a subsequent operation [2]. The definitive operation can be performed as early as 4–8 weeks later provided the baby is gaining weight normally. Performing the definitive repair early has important advantages for the patient, including less time with an abdominal stoma, less size discrepancy between the proximal and distal bowel at the time of colostomy closure, easier anal dilatation, and avoidance of psychological sequelae from painful perineal maneuvers. In addition, placing the rectum in the right location early in life, and using it, may represent an advantage in terms of the potential for acquired local sensation [3].

A temptation to repair these defects without a protective colostomy always exists [4, 5]. Such a repair without colostomy is performed without adequate anatomic information about the specific type of defect. Catastrophic complications have been seen in patients in whom the surgeon did not have a distal colostogram, approached the patients posterior sagittally looking for the rectum, and during the search for the rectum, injured to important structures (urethra, bladder, ureters, vas deferens, or seminal vesicles) occurred [6].

19.2 Females

Figure 19.4 shows a decision-making algorithm for the initial management of female patients. As in males, the perineal inspection is the most important step in the diagnosis and decision-making. The first 24 h should also be used to rule out important associated defects, as described previously.

The perineal inspection may disclose a single perineal orifice, which establishes the diagnosis of a cloaca. The clinician should know that such patients have a high likelihood (90%) of having an associated urologic defect. The patient needs a urologic evaluation and the presence of hydrocolpos should be ruled out by ultrasound.

Babies with a cloaca should undergo a diverting colostomy. It is important to perform the colostomy proximally enough to allow for the future repair of the malformation without interference from the colostomy. The surgeon must leave enough redundant distal rectosigmoid to allow a pull-through and, if needed, a vaginal reconstruction.

During the opening of the colostomy, it is mandatory to drain a hydrocolpos if present [7]. If the hydrocolpos is not large enough to reach the abdominal wall above the bladder, it can be drained with a rubber tube. Because a significant number of these patients have two hemivaginas, the surgeon must be certain that the tube inserted into the hydrocolpos is drain-

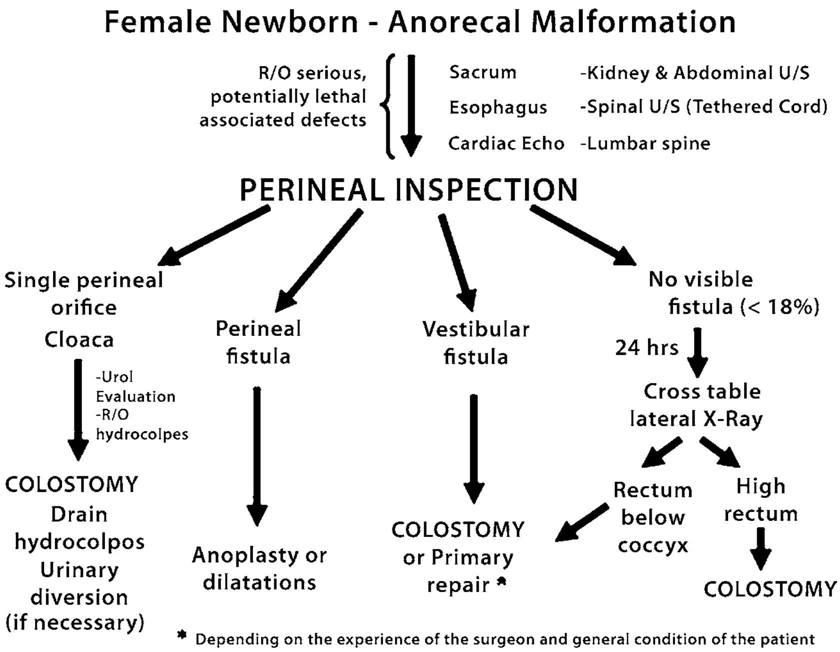

Fig. 19.4 Decision-making algorithm for female newborns with ARM. *Urol.* Urological (Reprinted from Pediatric Surgery, 4th ed., Ashcraft, Whitfield & Murphy eds. Peña A, Levitt MA. Imperforate Anus and Cloacal Malformations, p 502, Elsevier Saunders, Philadelphia (2005), with permission from Elsevier.)

ing both hemivaginas. Occasionally, the surgeon has to open a window in the vaginal septum in order to drain both with a single tube. The hydrocolpos can be so large that it may produce respiratory distress; vaginas that large may be drained by suturing the vaginal wall to the abdominal wall as a stoma.

Drainage of the hydrocolpos will relieve the ureteral obstruction, as the dilated vagina compresses the trigone. In rare cases, the bladder remains distended, and this may be evidence of particularly long, or narrow common channel. In such circumstances, the baby may require a vesicostomy or a suprapubic cystotomy. During the same anesthetic, it is helpful to perform an endoscopy to try to determine the anatomy, particularly the length of the common channel, which will help planning of the definitive operation.

The perineal inspection may show the presence of a perineal fistula, for which a primary anoplasty without a colostomy may be performed. Occasionally, the surgeon may have to care for a baby with severe associated defects or one who is very premature. If that is the case, dilatation of the fistula facilitates emptying of the colon, and a definitive operation can be planned for a future time.

The presence of a rectovestibular fistula is the most common finding in female patients. This malformation can be repaired during the neonatal period without a protective colostomy. A newborn pull-through in such patients is ideal, but unfortunately also represents the most common source of complications in these patients. To decide to repair this malformation primarily or to open a colostomy is a personal decision that should be based on the experience of the surgeon. Colostomy is still the most effective way to protect the pull-through.

Occasionally (less than 10% of the cases) there is no visible fistula and there is no meconium coming out from either the perineum or the urinary tract, even after 24 h of observation. For this small group of patients, a cross-table, lateral film is valuable. If the x-ray shows the gas in the rectum located very near the skin, it is likely that the patient has a perineal fistula. If the patient has a blind rectum located about 1 or 2 cm above the skin, the patient probably suffers from an imperforate anus with no fistula. One can consider, in this case, performing a primary operation without a colostomy, depending on the surgeon's experience (many of these patients with no fistula also have Down syndrome) [8].

Patients with a rectovestibular fistula who are very sick or premature can have dilatations of the fistula to allow decompression of the colon, with a repair to be performed later in life. That repair can be done with a protective colostomy or in a primary fashion, depending again on the surgeon's experience.

When patients with rectovestibular fistulas are repaired primarily in the newborn period, we recommend keeping them hospitalized for 5 days with nothing by mouth, and on parenteral nutrition. On the other hand, when the patients are subjected to a primary repair of a perineal fistula or rectovestibular fistula later in life, strict preoperative bowel irrigation is vital in order to be sure that the intestine is completely clean. We insert a central venous line and keep the patient hospitalized for 7–10 days with nothing by mouth, receiving parenteral nutrition. With this regimen, there have been no cases of perineal infection.

19.3 Colostomy

A divided descending colostomy is ideal for the management of anorectal malformations (Fig. 19.5). The completely diverting colostomy provides bowel decompression as well as protection for the final repair of the malformation. In addition, this type of colostomy facilitates the distal colostogram, which represents the most accurate diagnostic study for determining the anatomy of these defects [9].

A descending colostomy has advantages over a right or transverse colostomy [10, 11]. There is a relatively short segment of defunctionalized distal colon. Atrophy of the bowel distal to a more proximal colostomy and development of a microcolon with megarectosigmoid may result when a higher colostomy is utilized. Mechanical cleansing of the distal colon prior to the definitive repair is much less difficult when the colostomy is located in the descending colon. In the case of a large rectourethral fistula the patient may pass urine into the colon, whereas a more distal colostomy allows urine to escape through the distal stoma without significant absorption. If urine remains in the colon and is absorbed, metabolic acidosis may develop.

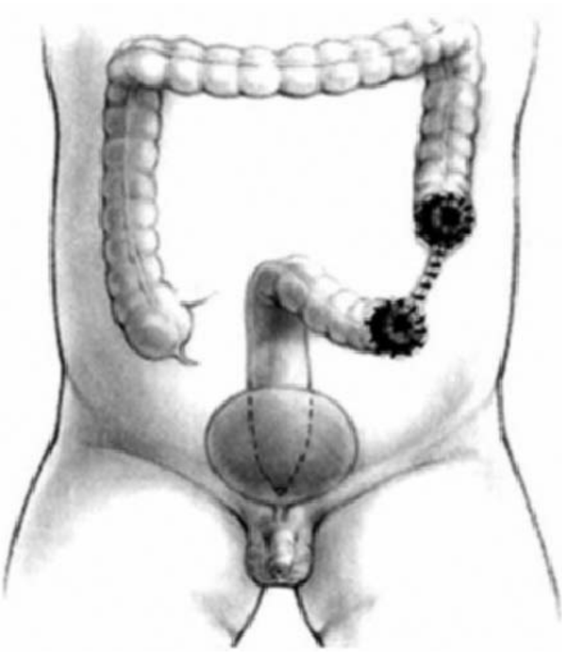

Fig. 19.5 Ideal colostomy (Reprinted from Atlas of Surgical Management of Anorectal Malformations, Peña A. Colostomy, p 19, Springer-Verlag, Inc. (1989), with permission of Springer Science and Business Media.)

Loop colostomies permit the passage of stool from the proximal stoma into the distal bowel, which produces urinary tract infection, distal rectal pouch dilatation, and fecal impaction. Prolonged distention of the rectal pouch may produce irreversible bowel damage, leading to a significant bowel hypomotility disorder and severe constipation later in life. Loop colostomies are also prone to prolapse [11].

A colostomy created too distal in the area of rectosigmoid may interfere with the mobilization of the rectum during the pull-through and is a common error.

References

1. Peña A, Hong AR (1999) Anorectal malformations – the state of the art. Colon Rectal Surg 2:1–19
2. Shaul DB, Harrison EA (1997) Classification of anorectal malformations – initial approach, diagnostic tests and colostomy. Semin Pediatr Surg 6:187–195
3. Freeman NV, Burge DM, Soar JS, et al (1980) Anal evoked potentials. Z Rinderchir 31:22–30
4. Goon HK (1990) Repair of anorectal anomalies in the neonatal period. Pediatr Surg Int 5:246–249
5. Moore TC (1990) Advantages of performing the sagittal anoplasty operation for imperforate anus at birth. J Pediatr Surg 25:276–277
6. Hong AR, Rosen N, Acuña MF, Peña A, Chaves L, Rodriguez G (2002) Urological injuries associated with the repair of anorectal malformations in male patients. J Pediatr Surg 37:339–344
7. Levitt MA, Peña A (2005) Pitfalls in the management of newborn cloacas. Pediatr Surg Int 21:264–269
8. Torres P, Levitt MA, Tovilla JM, Rodriguez G, Peña A (1998) Anorectal malformations and Down's syndrome. J Pediatr Surg 33:1–5
9. Gross GW, Wolfson PJ, Peña A (1991) Augmented-pressure colostogram in imperforate anus with fistula. Pediatr Radiol 21:560–563
10. Wilkins S, Peña A (1988) The role of colostomy in the management of anorectal malformations. Pediatr Surg Int 3:105–109

11. Peña A, Krieger M, Levitt MA (2005) Colostomy in anorectal malformations: a procedure with serious but preventable complications. J Pediatr Surg 41:748–756
12. Peña A, Levitt MA (2005) Imperforate anus and cloacal malformations. In: Ashcraft KW, Holcomb W, Murphy JP (eds) Pediatric Surgery 4th edn. Elsevier Saunders, Philadelphia, pp 501–502
13. Peña A (1989) Colostomy. In: Peña A (ed) Atlas of surgical management of anorectal malformations, Springer-Verlag, New York, pp 26–44

20 Operative Management of Anomalies in Males

Marc A. Levitt and Alberto Peña

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20.1 Introduction

Anorectal malformations (ARM) represent a wide spectrum of defects. The terms low, intermediate, and high are arbitrary and not very useful in therapeutic or prognostic terms. Within the group of ARM traditionally referred to as high, there are defects with different therapeutic and prognostic implications. For instance, retroprostatic fistula and rectobladderneck fistula were both considered high, yet the first can be repaired with a posterior sagittal approach alone, but the second also requires an abdominal approach. The prognosis for each type is completely different. Therefore, anatomic descriptions of the malformations are more useful clinically (Table 20.1).

In the past, many surgical techniques to repair ARM have been described. These included endorectal dissection [1–3], anterior perineal approach to a rectourethral fistula [4], and many different types of anoplasties [5]. However, most pediatric surgeons now use the posterior sagittal approach to repair these malformations with or without laparotomy or laparoscopy. The debate recently has been centered more on the possibility of performing these operations primarily without a protective colostomy and using laparoscopy if an abdominal component is needed [6–8].

20.2 Posterior Sagittal Approach

The patient is placed in the prone position with the pelvis elevated. The use of an electrical stimulator to elicit muscle contraction during the operation is very helpful. This contraction serves as a guide to keep the incision precisely in the midline, leaving equal amounts of muscle on either side. The length of the incision varies with the type of defect and can be extended to achieve the necessary exposure to effect a satisfactory repair. Thus, a perineal fistula requires a minimal posterior sagittal incision (2 cm), whereas higher defects may require a full posterior sagittal incision that runs from the middle portion of the sacrum towards the base of the scrotum in the male. The incision includes the skin, subcutaneous tissue, parasagittal fibers, muscle complex, and levator muscles (Fig. 20.1). In simple defects (perineal), the incision includes the parasagittal fibers and the muscle com-

Table 20.1 Classification of anomalies (according to Peña [14], with permission)

Males
Cutaneous (perineal fistula)
Rectourethral fistula
Bulbar
Prostatic
Rectobladderneck fistula
Imperforate anus without fistula
Rectal atresia
Females
Cutaneous (perineal fistula)
Vestibular fistula
Imperforate anus without fistula
Rectal atresia
Cloaca
Complex malformations

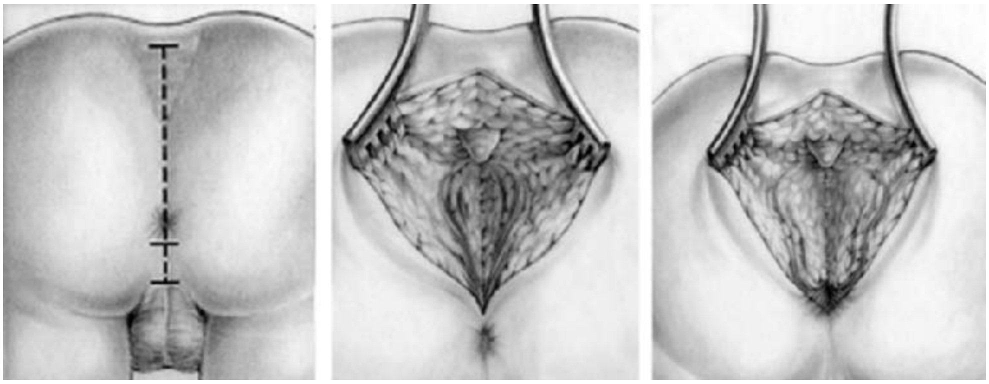


Fig. 20.1 Posterior sagittal incision. Separation of the parasagittal fibers and exposure of the muscle complex ([15], with permission)

plex, and it is not usually necessary to expose the levator ani muscle. Once the sphincter mechanism has been divided, the next most important step of the operation is the separation of the rectum from the urogenital structures, which represents the most delicate part of the procedure. Any kind of blind maneuver exposes the patient to a serious injury during this part of the operation [9].

About 90% of defects in boys can be repaired via the posterior sagittal approach alone without an abdominal component [10]. Each case has individual anatomical characteristics that mandate technical modifications. An example is the size discrepancy frequently seen between an ectatic rectum and the space available for pull-through. If the discrepancy is significant, the surgeon must tailor the rectum to fit. The number of rectums that need tapering is decreasing, probably because patients are receiving better colostomies, and the babies are undergoing the main repair earlier in life so that the distalrectosignoid does not have time to dilate.

A posterior sagittal approach should never be attempted without a technically adequate high-pressure distal colostogram to determine the exact position of the rectum and the fistula [11]. Attempting the repair without this important information significantly increases the risk of nerve damage, damage to the seminal vesicles, prostate, urethra, ureters, bladder neck, and bladder denervation [9].

20.3 Repair of Specific Defects in Boys

20.3.1 Rectoperineal Fistulas

Rectoperineal fistula is what traditionally was known as a low defect. The rectum is located within most of the sphincter mechanism. Only the lowest part of the rectum is anteriorly mislocated (Fig. 20.2). Sometimes the fistula does not open into the perineum, but rather follows a subepithelial midline tract, opening somewhere along the midline perineal raphe, scrotum, or even at the base of the penis. The diagnosis is established by perineal inspection. No further investigations are required. Most of the time, the anal fistula opening is abnormally narrow (stenosis). The terms “covered anus,” “anal membrane,” and “anteriorly mislocated anus,” as well as “bucket-handle malformations” refer to different external manifestations of perineal fistulas. We prefer the term “rectoperineal fistula” as this is most descriptive. The opening is not an anus as it is not a normal anal canal and is not surrounded by sphincter. The term “fistula” therefore is more accurate.

The operation is performed in the prone position with the pelvis elevated. Multiple 6-0 silk stitches are placed in the fistula orifice. An incision, usually about 2 cm, is created dividing the entire sphincter mechanism located posterior to the fistula. The sphincter is

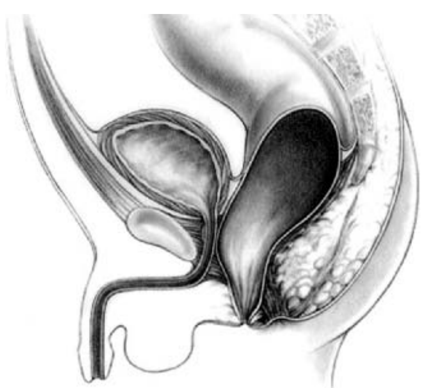


Fig. 20.2 Perineal fistula ([15], with permission)

divided and the posterior rectal wall is identified by its characteristic whitish appearance. Dissection of the rectum begins laterally, which makes dissection of the anterior rectal wall easier to visualize.

Dissection of the anterior rectal wall is the most critical because even when these patients have a low malformation, the rectum is still intimately attached to the urethra. The most common, and feared complication in these operations involves injury to the urethra. The patient must have a Foley catheter in place. To avoid a urethral injury, the surgeon must be meticulous during the dissection of the anterior rectal wall, and must keep in mind that the common wall has no plane of dissection and two walls must be created out of one.

These patients have an excellent functional prognosis in terms of bowel control. However, they suffer from the highest incidence of constipation. When the problem of constipation is not treated properly, chronic fecal impaction and overflow pseudoincontinence can occur.

20.3.2 Rectourethral Fistulas

Imperforate anus with rectourethral fistula is the most frequent defect in male patients [10]. The fistula may be located at the lower part of the urethra (bulbar urethra; Fig. 20.3) or the upper urethra (prostatic urethra) (Fig. 20.4). Immediately above the fistula site, the rectum and urethra share a common wall, an anatomic fact with significant technical and surgical implications. The rectum is usually distended and surrounded laterally and posteriorly by the levator ani muscle. Between the rectum and the perineal skin, a portion of striated voluntary muscle, called the muscle complex, is present. Contraction of these muscle fibers elevates the skin of the anal dimple. At the level of the skin, a group of voluntary muscle fibers, called parasagittal fibers, are located on both sides of the midline.

Lower urethral (bulbar) fistulas are usually associated with good-quality muscles, a well-developed sacrum, a prominent midline groove, and a prominent anal dimple. Higher urethral (prostatic) fistulas are more frequently associated with poor-quality muscles, an abnormally developed sacrum, a flat perineum with a poor midline groove, and a barely visible anal dimple. Of course, exceptions to these rules exist.

A Foley catheter is inserted through the urethra. About 20% of the time, this catheter goes into the rectum rather than the bladder. Under these circumstances, the surgeon can attempt catheterizing again using a catheter guide, or can relocate the catheter into the bladder under direct visualization during the operation. The incision is performed as previously described (Fig. 20.1); the parasagittal fibers, muscle complex, and levator muscle fibers are completely divided. Sometimes, the coccyx can be split in the midline with a cautery, particularly in those cases of rectoprostatic fistula in which the surgeon requires more exposure in the upper part of the incision. The higher the malformation, the deeper the levator muscle is located. When the entire sphincter mechanism has been divided, the surgeon expects to find the rectum.

It is at this point in the operation that the importance of a good high-pressure distal colostogram cannot be overstated. If the radiologic image shows the presence of a rectourethral bulbar fistula, the surgeon can expect that the rectum will be found just below the levator, and there is no way to injure the urinary tract because the rectum extends all the way down to the area of the bulbar urethra. On the other hand, if the preoperative image of the distal colostogram

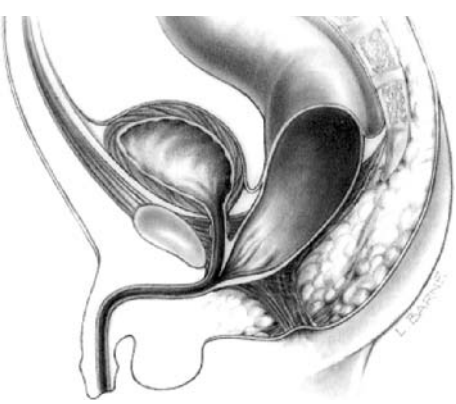


Fig. 20.3 Rectourethrobulbar fistula ([15], with permission)

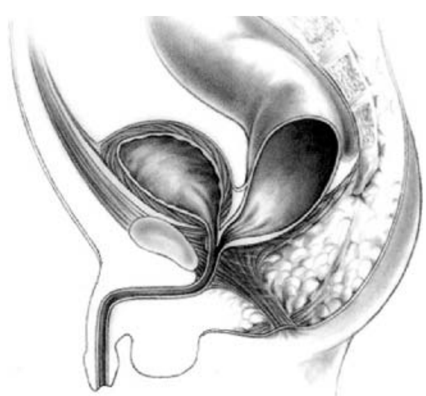


Fig. 20.4 Rectourethroprostatic fistula ([15], with permission)

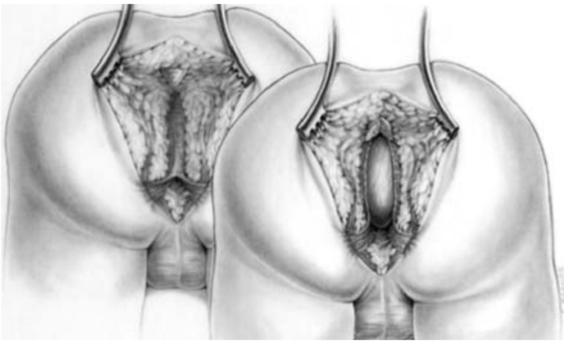


Fig. 20.5 Dividing the muscle complex and levator muscle. The rectum is exposed ([15], with permission)

shows a rectoprostatic fistula, the surgeon must be particularly careful and look for the rectum near the coccyx. Looking for the rectum lower than that exposes the patient to the risk of urinary tract injury. Also, if the high-pressure distal colostogram discloses the presence of a rectobladder-neck fistula, the surgeon should not even look for the rectum posterior sagittally because it is not there, and during a blind search, injury to the genitourinary tract could occur. In patients with rectourethrobulbar fistula, the rectum actually bulges through the incision when one completes division of the entire sphincter mechanism (Fig. 20.5).

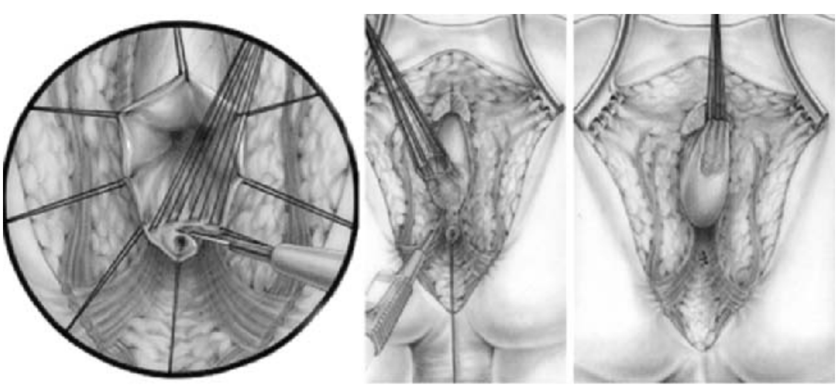
Silk stitches are placed in the posterior rectal wall on both sides of the midline. The rectum is opened in the midline and the incised distally, exactly in the midline, down to the fistula site. Temporary silk stitches are placed on the edges of the opened posterior rectal wall. When the fistula site is visualized, a final silk stitch is placed in the fistula orifice itself.

The anterior rectal wall above the fistula is a thin structure. It is actually a common wall with no plane of separation between the urinary tract and the rectum. Therefore, a plane of separation must be created in that common wall. For this, multiple 6-0 silk traction stitches are placed in the rectal mucosa immediately above the fistula site. The rectum is then separated from the urethra, creating a submucosal plane for approximately 5–10 mm above the fistula site (Fig. 20.6). This dissection is the source of the most serious complications during this repair. Creating a lateral plan first makes the anterior dissection easier.

The rectum is covered by a thin fascia that must be completely removed to be sure that one is working as close as possible to the rectal wall, to avoid denervation and injury to neighboring structures and to insure mobilization. Once the rectum is fully separated, a circumferential perirectal dissection is performed to gain enough rectal length to reach the perineum. In cases of rectourethrobulbar fistula, the dissection is rather minimal because only a short gap exists between the rectum and the perineum. In cases of rectoprostatic fistulas, the perirectal dissection is significant.

During this dissection, uniform traction is applied on the multiple silk stitches that were originally placed on the rectal edges and on the mucosa above the fistula. Uniform traction shows the rectal wall and identifies bands and vessels that hold the rectum in the pelvis. These bands must be carefully separated from the rectal wall and cauterized because they are vessels that tend to retract into the pelvis. The dissection should be performed as close as possible to the rectal wall without injuring the wall itself. Injury to the rectal wall can disrupt the intramural blood supply, upon which the pulled-through rectum depends.

The bands that are divided around the rectum are actually vessels and nerves. One would think that this denervation would provoke dysmotility, which leads to the problem of constipation in these patients. Thus, patients with higher malformations (which require



A B C

Fig. 20.6 A, B Separation of the rectum from the urethra. C The rectum is completely separated ([15], with permission)

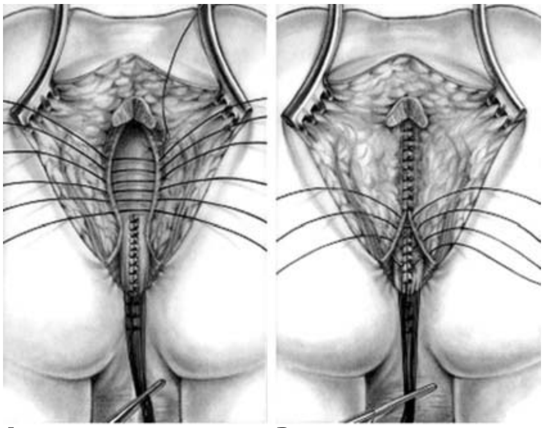


Fig. 20.7 **a** The rectum is passed in front of the levator muscle. **b** Muscle complex sutures anchor the rectum ([15], with permission)

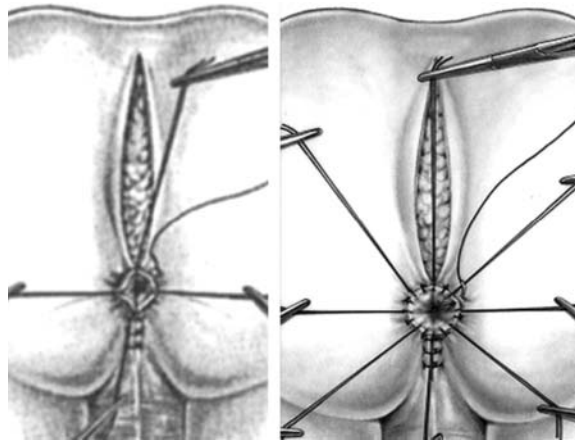


Fig. 20.8 Anoplasty ([15], with permission)

more dissection) would be expected to suffer from more severe constipation. However, the opposite is true in that patients with lower defects suffer more severe constipation than patients with higher defects [10]. The explanation for the observed dysmotility remains elusive [12].

The circumferential dissection of the rectum continues until the surgeon feels that enough length has been gained to allow a tension-free rectoperineal anastomosis. At this point, the size of the rectum can be evaluated and compared with the available space. If necessary, the rectum can be tapered, removing part of the posterior wall. In such cases, the rectal wall is reconstructed with two layers of interrupted long-lasting absorbable stitches.

The anterior rectal wall is frequently damaged to some degree as a consequence of the mucosal separation between rectum and urethra. To reinforce this wall, both smooth muscle layers can be stitched together with interrupted 5-0 long-lasting absorbable stitches. The urethral fistula is sutured with the same material. The rectal tapering should never be performed anteriorly as this would leave a rectal suture line in front of the urethral fistula repair, and may lead to a recurrent fistula.

The limits of the sphincter mechanism are determined electrically and marked with temporary silk stitches at the skin level. Those limits are sometimes easily visible without electrical stimulation in patients with a good sphincter mechanism. The limits of the sphincter are represented by the crossing of the muscle complex (the voluntary muscle structure that runs from the levator all the way down to the skin parallel to the direction of the rectum) with the parasagittal fibers (which run perpendicular and lateral to the muscle complex and parallel to the posterior sagittal incision.).

The perineal body is reconstructed, bringing together the anterior limits of the external sphincter, which was marked previously with the temporary silk stitches. The rectum must be placed in front of the levator and within the limits of the muscle complex (Fig. 20.7 a). Long-lasting 5-0 absorbable stitches are placed on the posterior edge of the levator muscle.

The posterior limit of the muscle complex must also be reapproximated behind the rectum. These stitches must take part of the rectal wall to anchor it to avoid rectal prolapse (Fig. 20.7 b). An anoplasty is performed with 16 interrupted long-lasting absorbable stitches (Fig. 20.8). The ischiorectal fossa and the subcutaneous tissue are reapproximated and the wound is closed with a subcuticular absorbable monofilament.

All of these patients have a Foley catheter inserted prior to starting the operation. The patient receives broad-spectrum antibiotics for 24–48 h and can be fed postoperatively on the same day of surgery.

Repair of rectourethral fistulas has been performed laparoscopically [7] with separation of the fistula through the abdomen, and pull-through of the rectum through a minimized perineal incision. The preliminary experience shows that these procedures are feasible. It is unclear whether a laparoscopic approach for a rectourethral fistula is less invasive than the posterior sagittal approach. Prevention of prolapse is by a pelvic hitch rather than tacking of the posterior rectum via the posterior sagittal incision. Technical challenges with this approach include gaining adequate rectal length, and tapering an ectatic rectum if necessary.

20.3.3 Rectobladderneck Fistula

In this defect, which affects 10% of males, the rectum opens at the bladder neck (Fig. 20.9). The patient has a poor prognosis because the levator ani muscle, muscle complex, and external sphincter are frequently poorly developed. Consistent with the caudal regression, the sacrum and entire pelvis is often deformed and underdeveloped. The perineum is often flat, with evidence of poor muscle development.

For this repair, a total body preparation is performed; the sterile field includes the entire lower part of the patient's body. The initial incision is posterior sagittal. All of the muscle structures are divided in the midline. The channel for the rectum, which lies just under the coccyx, is created bluntly. This posterior sagittal incision can be created with the child in supine position and the legs lifted up. At this point, laparoscopy represents an excellent minimally invasive alternative to a laparotomy.

These patients have the rectum connected to the bladder neck, located approximately 2 cm below the peritoneal reflection. Interestingly, the higher the malformation, the shorter the common wall between the rectum and the urinary tract. This means that the rectum in this group of defects opens in a perpendicular fashion into the bladder neck, which makes its dissection much easier. The laparoscopic approach provides an excellent view of the peritoneal reflection, and one can also see the ureters and the vas deferens, which tend to run in the direction of the bladder neck. During the dissection of the rectum, therefore, these structures must be kept under direct view to prevent damage to them. The serosa that covers the most distal part of the rectum should be divided, creating a plane of dissection around the rectum; it is this plane that is used to continue the dissection distally. The rectum rapidly narrows down, and this is where the fistula should be ligated.

At this point the surgeon must gain adequate length for the rectum to reach the perineum. The vessels that supply the distal rectum must be meticulously divided. Laparoscopically, or through a laparotomy, the space in the retroperitoneum for pull-through of the rectum is visualized (Fig. 20.10). An instrument or trocar from the perineum can be introduced to grab the rectum and pull it down. This allows the surgeon to see from above the tension lines that represent the vessels that it must be divided until the rectum reaches the perineum.

Tapering of the rectum, if that is required, is difficult with a laparoscopic approach, and mobilization of a very high rectum is technically challenging. In addition, passage of the trocar from perineum into the pelvis must be done carefully to avoid injury to the bladder neck and ureters.

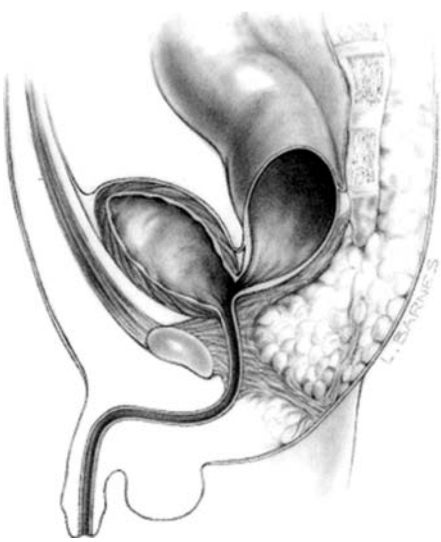


Fig. 20.9 Rectobladderneck fistula ([15], with permission)

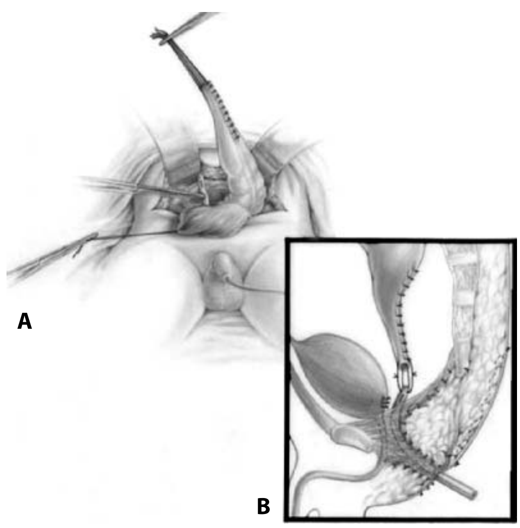


Fig. 20.10 **A** Abdominal approach for high defects (rectobladder-neck fistula). The rectum has been separated from the bladder neck. The presacral rubber tube is identified. **B** The rectum is anchored to the rubber tube to guide the pull-through ([15], with permission)

20.3.4 Imperforate Anus Without Fistula

Most patients with this unusual defect have a well-developed sacrum and good muscles, and thus they have good prognosis for bowel function. The rectum ends approximately 2 cm from the perineal skin. Even though the patient has no communication between rectum and urethra, these two structures are separated only by a thin, common wall, which is an important anatomic detail with technical implications. About half of the patients with no fistula also have Down's syndrome and more than 90% of the patients with Down's syndrome and imperforate anus suffer from this specific defect, hinting at a chromosomal link [13]. The fact that these patients have Down's syndrome does not seem to interfere with the good prognosis in terms of bowel control.

In these cases, the blind end of the rectum is usually located at the level of the bulbar urethra. The rectum must be carefully separated from the urethra, because both structures share a common wall even though no fistula is present. The rest of the repair must be performed as described for the rectourethral fistula.

20.3.5 Rectal Atresia and Stenosis

This is an extremely unusual defect that occurs in only 1% of male patients, occurring in 1% of our cases, in which the lumen of the rectum may be atretic or stenotic. The upper pouch is dilated and the lower portion is a small anal canal in the normal location. The narrow area starts at a depth of approximately 1–2 cm. These two structures may be separated by a thin membrane or by a dense portion of fibrous tissue. Patients with this defect have all the necessary elements to be continent and have an excellent functional prognosis. Because they have a well-developed anal canal, they have normal sensation in the anorectum. They also have almost normal voluntary sphincters.

The approach to these malformations is also via a posterior sagittal incision. The upper rectal pouch is open, as well as the short distal anal canal. An end-to-end anastomosis is performed under direct visualization, followed by a meticulous reconstruction of the sphincter mechanism posterior to the rectum (see Chap. 12)..

20.4 Postoperative Management and Colostomy Closure

In male patients with rectourethral fistulas, the Foley catheter remains in for 7 days. If the catheter comes out accidentally before that, patients usually void without any problem and do not require catheter replacement. Intravenous antibiotics are administered for 48 h. Antibiotic ointment is applied to the perineum for 7 days. Most patients usually go home after 2 days, or after 3–4 days if the abdomen was entered.

Two weeks after the repair anal dilatations are started. A dilator that fits snugly into the anus is passed twice daily by the parents. Every week, the size of the dilator is increased, until the rectum reaches the desired size, which depends on the patient's age (Table 20.2). Once this desired size is reached, the colostomy may be closed. The frequency of dilatation may be reduced once the dilator goes in easily with no resistance according to the following protocol: once a day for 1 month, every 3rd day for 1 month, twice a week for 1 month, once a week for 1 month, and once a month for 3 months. Nonmanageable, severe strictures are seen in cases in which the dilatation program was not carried out as indicated or when the blood supply of the distal rectum was damaged during the pull-through.

After the colostomy is closed, the patient usually has multiple bowel movements and may develop perineal excoriation. A constipating diet may be helpful in the treatment of this problem. After several weeks, the number of bowel movements decreases and most patients then start to have constipation. After 3 months, the patient develops a more regular bowel movement pattern. A patient who has one to three bowel movements per day, remains clean between bowel movements, and shows evidence of a feeling or pushing during bowel movements, has a good bowel movement pattern and is usually able to potty train. A

Table 20.2 Size of dilator according to age ([14], with permission)

Age	Hegar dilator (no.)
1–4 months	12
4–12 months	13
8–12 months	14
1–3 years	15
3–12 years	16
>12 years	17

Table 20.3 Global functional results. *Pts* Patient ([14], with permission)

	Voluntary bowel movement		Soiling		Totally continent		Constipated	
	Pts	%	Pts	%	Pts	%	Pts	%
Perineal fistula	39/39	100	3/43	20.9	35/39	89.7	30/53	56.6
Rectal atresia or stenosis	8/8	100	2/8	25	6/8	75	4/8	50
Vestibular fistula	89/97	92	36/100	36	63/89	70.8	61/100	61
Imperforate anus without fistula	30/35	86	18/37	48.6	18/30	60	22/40	55
Bulbar urethra fistula	68/83	82	48/89	53.9	34/68	50	52/81	64.2
Prostatic fistula	52/71	73	67/87	77.1	16/52	30.8	42/93	45.2
Cloaca: short common channel	50/70	71	50/79	63.3	25/50	50	34/85	40
Cloaca: long common channel	18/41	44	34/39	87.2	5/18	27.8	17/45	34.8
Vaginal fistula	3/4	75	4/5	80	1/3	33.3	1/5	20
Bladder neck fistula	8/29	28	39/43	90.7	1/8	12.5	7/45	15.6

patient with multiple bowel movements or one who passes stool constantly without showing any signs of sensation or pushing usually has a poor functional prognosis.

20.4.1 Evaluation of Results

Each defect described herein has a different prognosis. When evaluating clinical results, the error of oversimplification should be avoided. Categories such as high, intermediate, and low, or even high and low, include specific malformations, each with a different prognosis. The patients with low defects can be expected to have excellent results. Table 20.3 shows the results obtained in our series. A patient with a very abnormal sacrum (a ratio of less than 0.3) and a flat perineum can be expected to suffer from fecal incontinence regardless of the type of malformation.

References

1. Kiesewetter WB (1967) Imperforate anus, II. The rationale and technique of sacroabdominoperineal operation. J Pediatr Surg 2:106–117
2. Louw JH, Cywes S, Cremin BJ (1971) The management of anorectal agenesis. S Afr J Surg 9:21–30
3. Rehbein F (1967) Imperforate anus: experiences with abdominoperineal and abdomino sacroperineal pullthrough procedures. J Pediatr Surg 2:99–105
4. Mollard P, Soucy P, Luis D, Meunier P (1989) Preservation of infralevator structures in imperforate anus repair. J Pediatr Surg 24:1023–1026
5. Nixon JJ (1988) Nixon anoplasty. In: Stephens FD, Smith ED (eds) Anorectal Malformations in Children: Update 1988. Alan R. Liss, New York, pp 378–381
6. Sydorak RM, Albanese CT (2002) Laparoscopic repair for high imperforate anus. Semin Pediatr Surg 11:217–225
7. Georgeson KE, Inge TH, Albanese CT (2000). Laparoscopically assisted anorectal pull-through for high imperforate anus – a new technique. J Pediatr Surg 35:927–931
8. Goon HK (1990) Repair of anorectal anomalies in the neonatal period. Ped Surg Int 5:246–249
9. Hong AR, Rosen N, Acuña MF, Peña A, Chaves L, Rodriguez G (2002) Urological injuries associated with the repair of anorectal malformations in male patients. J Pediatr Surg 37:339–344
10. Peña A, Levitt MA (2005) Imperforate anus and cloacal malformations. In: Ashcraft KW, Holder TM, Holcomb W (eds) Pediatric Surgery, 4th edn. WB Saunders, Philadelphia, pp 496–517
11. Gross GW, Wolfson PJ, Peña A (1991) Augmented-pressure colostogram in imperforate anus with fistula. Pediatr Radiol 21:560–563
12. Peña A, Levitt MA (2002) Colonic inertia in pediatrics. Curr Prob Surg 39:661–730
13. Torres P, Levitt MA, Tovilla JM, Rodriguez G, Peña A (1998) Anorectal malformations and Down's syndrome. J Pediatr Surg 33:1–5
14. Peña A, Levitt MA (2005) Imperforte anus and cloacal malformations. In: Ashcraft KW, Holcomb W, Murphy JP (eds) Pediatric Surgery 4th edn.
15. Peña A (1989) Male defects. In: Peña A (ed) Atlas of surgical management of anorectal malformations, Springer-Verlag, New York, pp 26–44

21 Operative Management of Anomalies in the Female

Marc A. Levitt and Alberto Peña

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Many surgical techniques to repair anorectal malformations (ARM) in females have been described including an endorectal dissection [1–3], an anterior perineal approach [4], and many different types of anoplasties [5]. The majority of pediatric surgeons now use the posterior sagittal approach to repair these malformations. The debate recently has been centered more on the possibility of performing these operations primarily without a protective colostomy.

21.1 The Posterior Sagittal Approach

The posterior sagittal approach is utilized as described in Chap. 20.

21.2 Repair of specific Defects in Girls

21.2.1 Rectoperineal Fistulas

From the therapeutic and prognostic points of view, this common defect is equivalent to the perineal fistula described in the male patients. The rectum is located within the sphincter mechanism, except for its lower portion, which is located anteriorly. The rectum and vagina are well separated (Fig. 21.1). This defect is repaired in the same way as described for male patients with the exception of course that the dissection is off the posterior vaginal wall rather than the urethra. Complete mobilization of the rectum is vital until the surgeon sees the areolar tissue that demonstrates that the rectal and vaginal walls are completely separated, so that the anoplasty is performed with no tension. Failure to complete this separation can lead to retraction of the pull-through and to dehiscence of the perineal body.

21.2.2 Rectovestibular Fistulas

Patients with rectovestibular fistulas are frequently erroneously diagnosed as having a rectovaginal fistula. Rectovestibular fistula is by far the most common defect in females, It has an excellent functional prognosis. The precise diagnosis is a clinical one requiring only a meticulous inspection of the newborn genitalia. The clinician observes a normal urethral meatus, a normal vagina, and a third hole in the vestibule, which is the rectovestibular fistula (Fig. 21.2). About 5% of these patients also have two hemivaginas, and a vaginal septum is visible, which should be removed at the time of the pull-through.

This defect may be repaired without a protective colostomy. This is a well-recognized trend in the management of ARM [6, 7], avoids the potential morbidity of a colostomy, and reduces the number of operations to one rather than three (colostomy, main repair, and colostomy closure). Many patients do very well with a single neonatal primary operation without a protective colostomy. However, a perineal infection followed by dehiscence of the anal anastomoses and recurrence of the fistula provokes severe fibrosis, which may interfere with the sphincteric mechanism. In such a case, the patient may have lost the best opportunity for an optimal functional result, because secondary operations do not render the same good prognosis as successful primary operations [8]. Thus, a protective colostomy is still the safest way to avoid these complications. The decision related to the opening of a colostomy or operating primarily must be taken by individual surgeons, taking into consideration his or her experience and the clinical condition

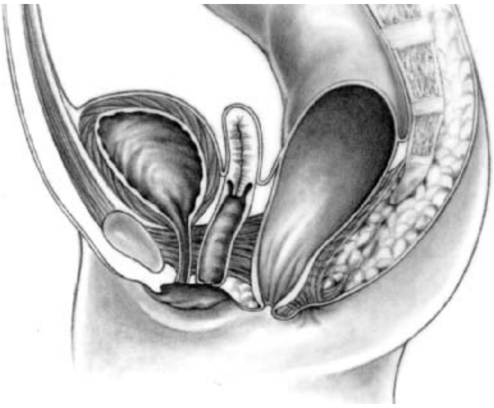

Fig. 21.1 Perineal fistula ([11], with permission)

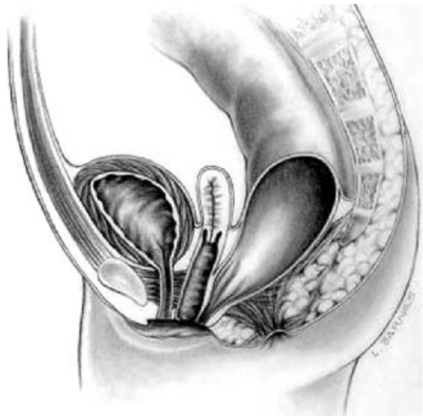

Fig. 21.2 Vestibular fistula ([11], with permission)

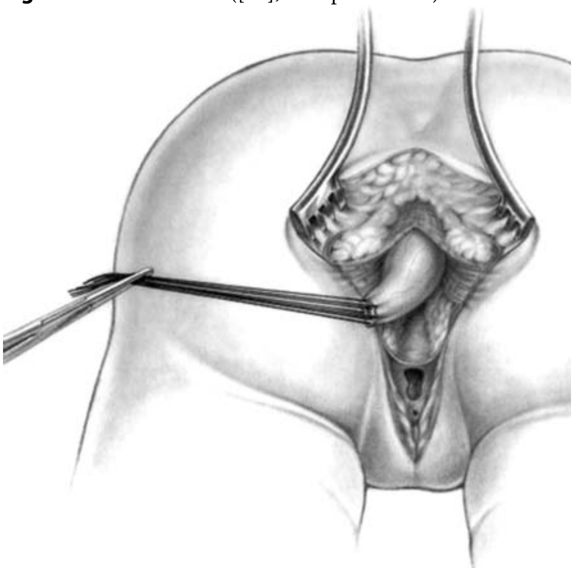

Fig. 21.3 Repair of rectovestibular fistula. The rectum is completely separated from the vagina ([11], with permission)

Fig. 21.4 Repair of the rectovestibular fistula. The perineal body is repaired ([11], with permission)

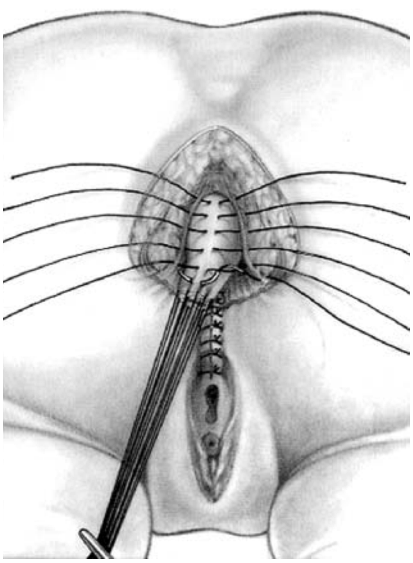

Fig. 21.5 Repair of the vestibular fistula. Muscle complex sutures anchor the rectum ([11], with permission)

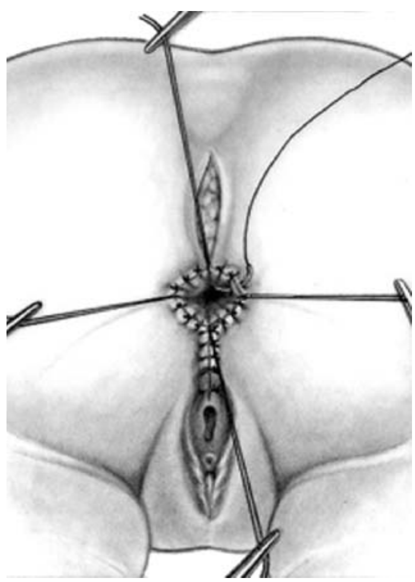

Fig. 21.6 Repair of the vestibular fistula. Anoplasty ([11], with permission)

of the patient. At our institution, patients who are born with this kind of malformation without serious associated defects are operated primarily as newborns without a colostomy.

The term "rectovaginal fistula" is often misused in patients who actually have a rectovestibular fistula or a cloaca. A real rectovaginal fistula occurs in less than 1% of all cases [9, 10].

The complexity of the rectovesticular fistula defect is frequently underestimated. Multiple 6-0 silk stitches are placed at the mucocutaneous junction of the fistula. The incision used to repair this defect is shorter than that used to repair the male rectourethral fistula. The incision continues down to the fistula and around the fistula into the vestibule. Once the entire sphincter mechanism has been divided, the posterior rectal wall is evident by its characteristic whitish appearance. The fascia that surrounds the rectum must be removed to be sure that the dissection is performed as close as possible to the rectal wall. The dissection continues creating the plane of dissection along the lateral walls of the rectum while applying traction on the multiple silk stitches. The last part is the most important part of this dissection, which is the separation of the rectum from the vagina.

There is a long common wall, and two walls must be created out of one using a meticulous technique, trying to keep both walls of the rectum and vagina intact. The dissection continues cephalad until both walls of the rectum and vagina are fully separated (a location identified when the surgeon encounters areolar tissue between rectum and vagina). At this point both walls are full thickness (Fig. 21.3). If the rectum and the vagina are not completely separated, a tense rectal anastomosis would be created, which, as for the rectoperineal fistula, would predispose the patient to dehiscence and retraction.

Once the dissection has been completed, the perineal body is repaired (Fig. 21.4). The anterior edge of the muscle complex is reapproximated as described previously. The muscle complex must be reconstructed posterior to the rectum, with the stitches including the posterior edge of the muscle complex and the posterior rectal wall to avoid rectal prolapse (Fig. 21.5). The anoplasty is then performed (Fig. 21.6).

21.2.3 Imperforate Anus Without Fistula

This defect in female patients carries the same therapeutic and prognostic implications as described for male patients. The surgical technique is similar to that described for males, with the obvious difference being a separation of the rectum from the posterior vaginal wall rather than the posterior urethra.

21.2.4 Postoperative Care

The dilatation protocol is similar to that described for males in Chap. 20. The Foley catheter can be removed in the first 24 h.

References

1. Kiesewetter WB (1967) Imperforate anus, II: the rationale and technique of sacroabdominoperineal operation. J Pediatr Surg 2:106–117
2. Rehbein F (1967) Imperforate anus: experiences with abdominoperineal and abdomino sacroperineal pullthrough procedures. J Pediatr Surg 2:99–105
3. Soave F (1969) Surgery of the rectal anomalies with preservation of the relationship between the colonic muscular sleeve and puborectal muscle. J Pediatr Surg 4:705–712
4. Mollard P, Soucy P, Luis D, Meunier P (1989) Preservation of infralevator structures in imperforate anus repair. J Pediatr Surg 24:1023–1026
5. Nixon HH (1988) Nixon anoplasty. In: Stephens FD, Smith ED (eds) Anorectal Malformations in Children: Update 1988. Alan R. Liss, New York, pp 378–381
6. Goon HK (1990) Repair of anorectal anomalies in the neonatal period. Pediatr Surg Int 5:246–249
7. Moore TC (1990) Advantages of performing the sagittal anoplasty operation for imperforate anus at birth. J Pediatr Surg 25:276–277
8. Peña A (1988) Posterior sagittal anorectoplasty: results in the management of 332 cases of anorectal malformations. Pediatr Surg Int 3:94–104
9. Rosen, NG, Hong AR, Soffer SZ, Rodriguez G, Peña A (2002) Rectovaginal fistula: a common diagnostic error with significant consequences in girls with anorectal malformations. J Pediatr Surg 37:961–965
10. Bill AH, Hall DG, Johnson RJ (1975) Position of rectal fistula in relation to the hymen in 46 girls with imperforate anus. J Pediatr Surg 10:361–365
11. Peña A (1989) Female defects. In: Peña A (ed) Atlas of surgical management of anorectal malformations, Springer-Verlag, New York, pp 50–55

22 Treatment of Cloacas

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22.1 Introduction

Persistent cloaca was, in the past, considered an unusual defect, with a high incidence of "rectovaginal fistula" reported in the literature [1]. In retrospect, it seems that cloaca represents a much more common defect in female patients than previously thought, and rectovaginal fistula is an almost nonexistent defect that is present in fewer than 1% of all cases, except in a few specific geographical areas [2]. Most patients suffering from a persistent cloaca were erroneously thought to have a rectovaginal fistula. Many of those patients underwent surgery with repair of the rectal component only and were left with the urogenital sinus untouched [2], requiring a complete redo operation.

This group of defects represents the extreme in the spectrum of complexity of female malformations. A cloaca is defined as a defect in which the rectum, vagina, and urinary tract meet and form a confluence exiting the perineum via a single common channel (Figs. 22.1–22.3). The diagnosis of persistent cloaca is a clinical one. This defect should be suspected in a female born with imperforate anus and small-looking genitalia. Careful separation of the labia discloses a single perineal orifice.

The length of the common channel varies from 1 to 7 cm. Common channels longer than 3 cm are usually associated with complex defects (Fig. 22.3). The mobilization of the vagina is difficult and often some form of vaginal replacement is needed during the definitive repair. A common channel of less than 3 cm usually means that the defect can be repaired with a posterior sagittal approach only without opening the abdomen (Fig. 22.1).

The rectum sometimes opens high into the dome of the vagina. Therefore, a laparotomy or laparoscopy must be part of the procedure to mobilize the bowel. The vagina is frequently abnormally distended and full of secretions (hydrocolpos; Fig. 22.3). This distended vagina can compress the trigone and interfere with the drainage of the ureters, leading to megaureters. The dilated vagina can also become infected, pyocolpos, which can lead to perforation and peritonitis. On the other hand, such a large vagina may represent a technical advantage for the repair, because having more vaginal tissue will facilitate its reconstruction.

A frequent finding in cloacal malformations is the presence of different degrees of vaginal and uterine septation or duplication (Fig. 22.3). The rectum usually opens in between the two hemivaginas. These patients may also suffer from cervical atresia and during puberty are unable to drain menstrual blood through the vagina; the menstrual blood accumulates in the peritoneal cavity and sometimes requires emergency surgery [3].

Low cloacal malformations (less than 3 cm; Figs. 22.1 and 22.2) are usually associated with a well-developed sacrum, a normal-appearing perineum, and adequate muscles and nerves. Therefore, a good functional prognosis is expected.

22.2 Surgical Treatment of Cloacas

Prior to undertaking the repair of cloacal malformations, the surgeon should perform endoscopy to determine the length of the common channel. There are two well-characterized groups of patients with cloaca

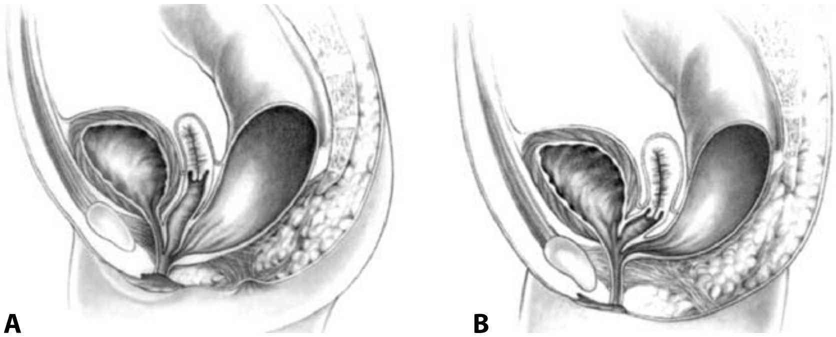

Fig. 22.1 Spectrum of cloacae. **A** Most common channel. **B** Long common channel ([6], with permission)

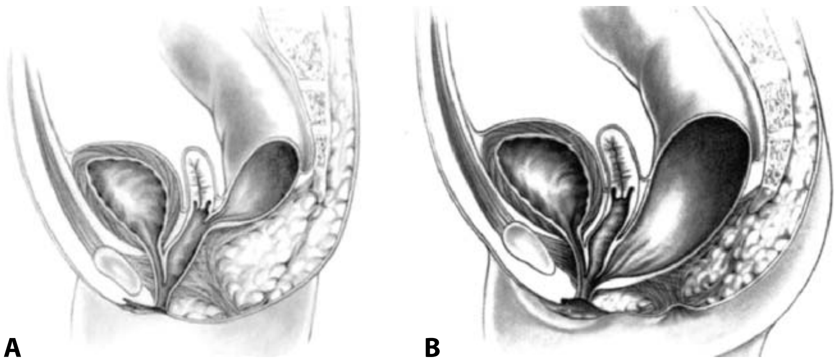

Fig. 22.2 Spectrum of cloacae. **A** High rectal implantation into the vagina. **B** Short common channel ([6], with permission)

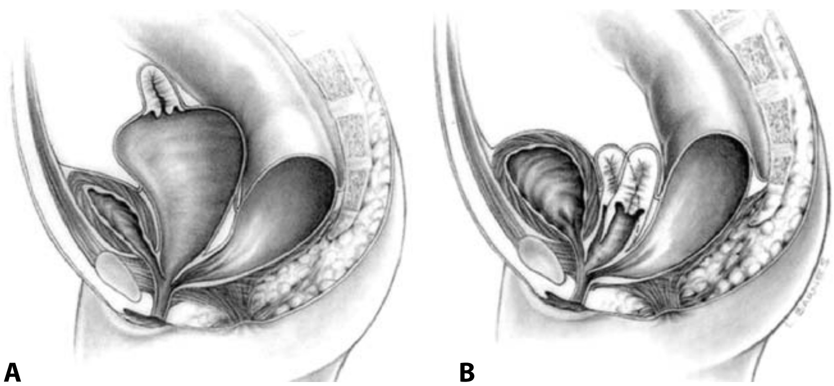

Fig. 22.3 **A** Associated hydrocolpos. **B** Double vagina and double uterus ([6], with permission)

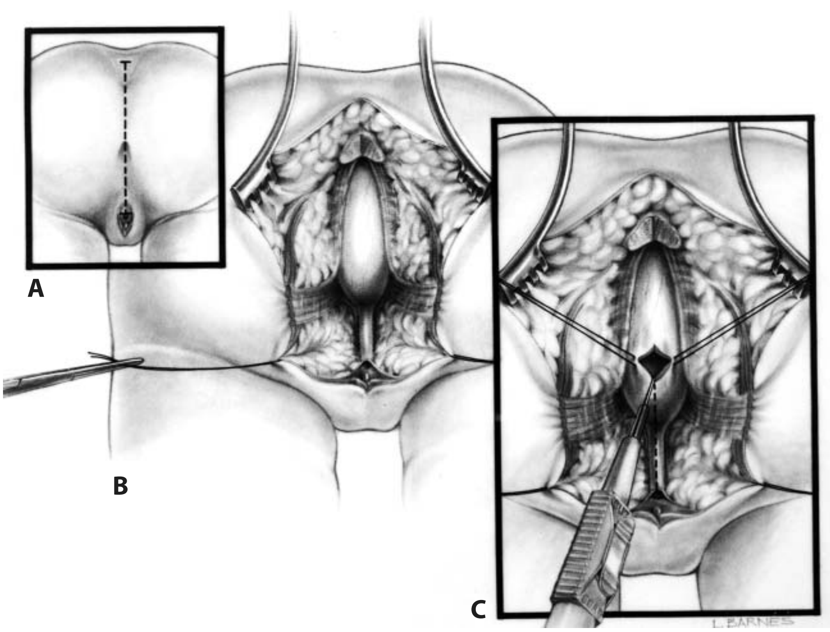

Fig. 22.4 Cloaca repair. **A** Incision. **B** Rectum and common channel are exposed. **C** Rectal opening ([6], with permission)

[4], and each represent different technical challenges that must be recognized preoperatively.

The first is represented by patients who are born with a common channel shorter than 3 cm. Fortunately, these patients represent the majority (over 60%) of all cloacas. The great majority of these patients can be repaired using only a posterior sagittal approach, without a laparotomy, and the operation is a reproducible one that can be performed by most general pediatric surgeons.

The second group is represented by patients with longer common channels. These patients usually need a laparotomy and a decision-making algorithm for the reconstruction that requires a large experience and special training in urology. These patients are therefore best cared for in centers with special expertise in the repair of these defects.

Fig. 22.5 Total urogenital mobilization. The rectum is separated from the vagina ([7], with permission)

22.2.1 Cloacas With a Common Channel Shorter than 3 cm

The incision extends from the middle portion of the sacrum down to the single perineal orifice. The entire sphincter mechanism is divided in the midline. The first structure that the surgeon finds after the division of the sphincter mechanism is the rectum (Fig. 22.4). Because of the complexity of these malformations, the surgeon must be prepared to find bizarre anatomic arrangements of the rectum and vagina.

The rectum is opened precisely in the midline (Fig. 22.4) and silk stitches are placed along the edges of the posterior rectal wall. The incision is extended distally through the posterior wall of the common channel. The entire common channel is exposed, which allows measurement of the common channel under direct vision. The rectum is then separated from the vagina (Fig. 22.5) in the same way as described for the repair of rectovestibular fistula. The rectum and vagina share the same type of common wall that has already been described.

Once the rectum has been completely separated from the vagina, a total urogenital mobilization is performed [5]. In the past, the vagina was separated from the urinary tract, which was a technically challenging maneuver associated with a significant morbidity. Total urogenital mobilization consists of the mobilization of both the vagina and urethra as a unit. After the rectum has been separated, multiple silk stitches are placed, taking the edges of the vagina and the common channel, in order to apply uniform traction on the urogenital sinus for its mobilization. Another series of fine stitches is placed across the urogenital sinus approximately 5 mm proximal to the clitoris (Fig. 22.6). The urogenital sinus is transected between the last row of silk stitches and the clitoris, and is dissected, taking advantage of the fact that there is a natural plane between it and the pubis. Very rapidly, and in a bloodless field, one can reach the upper edge of the pubis. There, a fibrous, avascular structure is identified that gives support to the vagina and bladder and is called the "suspensory ligament of the urethra and bladder." While applying traction to the multiple stitches, these suspensory ligaments are divided, which provides significant mobilization (2–3 cm) of the urogenital sinus. In addition, one can then dissect the lateral and dorsal walls of the vagina to gain a further 5–10 mm (Fig. 22.7).

This dissection is enough to repair about 60% of all cloacas and is a reproducible maneuver. It has the additional advantage of preserving an excellent blood supply to both the urethra and the vagina, and placing the urethral opening in a visible location to facilitate intermittent catheterization if necessary (Fig. 22.8). It also provides a smooth urethra that can be catheterized easily.

What used to be the common channel is divided in the midline creating two lateral flaps that are sutured to the skin, creating the new labia. The vaginal edges are mobilized to reach the skin to create the introitus. The limits of the sphincter are then determined electrically and the perineal body is reconstructed, bringing together the anterior limit of the sphincter. The rectum is placed within the limits of the sphincter as described previously.

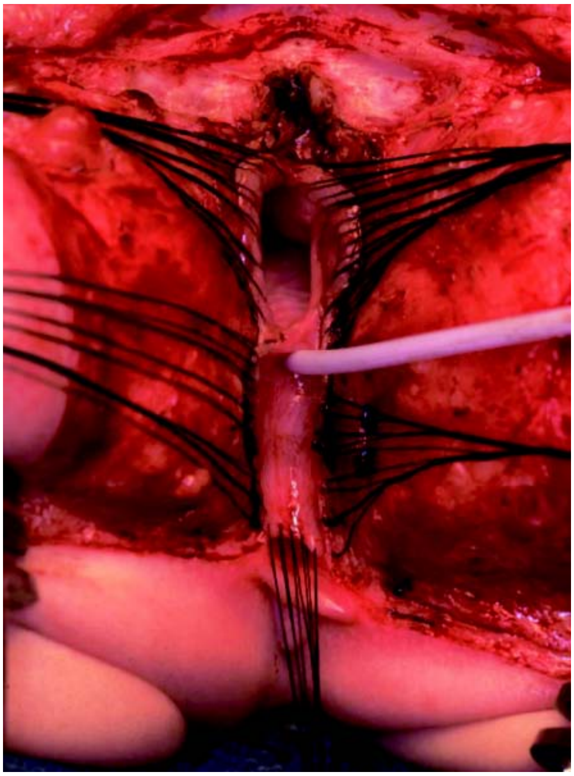

Fig. 22.6 Total urogenital mobilization. Stitches placed on the edges of the sinus and across, near the clitoris ([8], with permission)

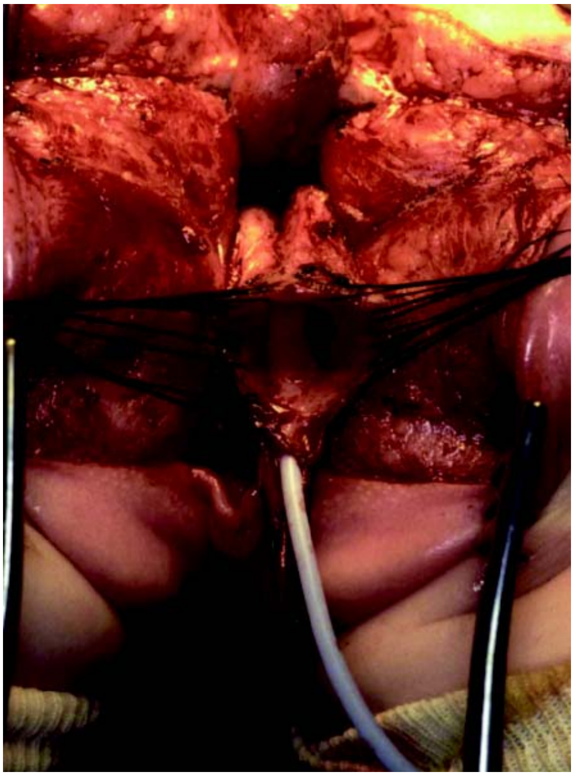

Fig. 22.7 Total urogenital mobilization. Urogenital sinus fully mobilized ([8], with permission)

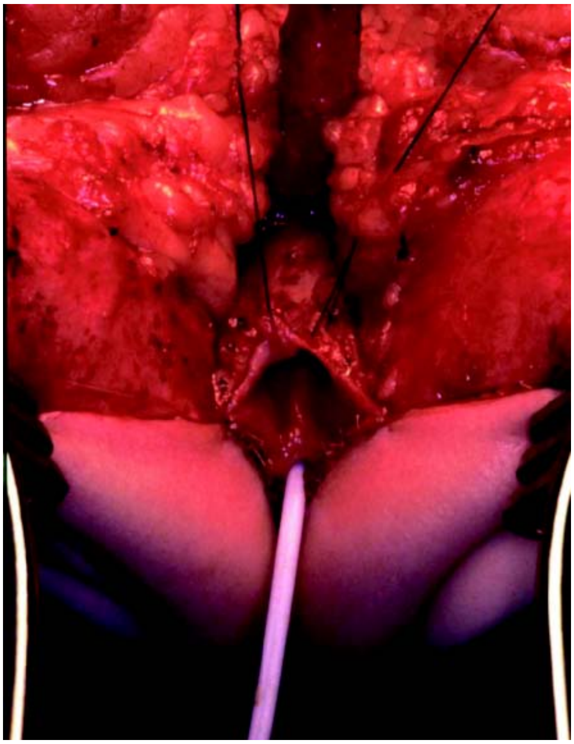

Fig. 22.8 Total urogenital mobilization. Urethra and vagina sutured into their new positions ([8], with permission)

22.2.2 Cloacas with a Common Channel Longer than 3 cm

When the endoscopy shows that the patient has a long common channel, the surgeon must be prepared to face a very significant technical challenge. In the presence of a long common channel, patients should receive a total body preparation, as it is likely that they will require a laparotomy. The rectum is separated from the vagina and urethra. The presence of a very long common channel (more than 5 cm) means that there is no way that the total urogenital mobilization will be enough to repair that malformation, and it is therefore advisable to leave the common channel in place using it for the urethra for intermittent catheterization.

In that situation, it is suggested that the surgeon tries to separate the vagina from the urinary tract from within the abdomen, rescuing it from the back of the common channel, and then closing the posterior aspect of the common channel, which will become the catheterizable urethra. The bladder is opened in the midline and feeding tubes are placed into the ureters to protect them. In these types of malformations, there is a significant common wall between the vagina and the bladder. The ureters run through that common wall and must therefore be consistently identified and palpated during the separation of the vagina from the urinary tract. The surgeon must be familiar with the different techniques of ureteral reimplantations because these patients frequently require that operation during the same procedure.

Once in the abdomen, the patency of the Mullerian structures are investigated by passing a no. 3 feeding tube through the fimbriae of the Fallopian tubes and injecting saline through them to be sure that they are patent. If one of the systems is not patent, excision of the atretic Mullerian structure without damage to the blood supply of the ovary is recommended. When both Mullerian structures are atretic, they should be left in place, and the patient followed closely with further decisions made when she reaches puberty.

With the abdomen open, the surgeon has to a make decisions based on the specific anatomic findings. In the presence of a single mid-sized vagina, the surgeon must separate the vagina from the urinary tract, being sure to preserve the blood supply of this structure, which comes from the uterine vessels. When the vagina is found to be too short, the patient requires some form of vaginal replacement that can be performed using tissue from the rectum, colon, or small bowel.

22.2.3 Vaginal Switch Maneuver

There is one specific group of patients who are born with hydrocolpos and two hemivaginas. The hemivaginas are very large and the two hemiuteri are very separated, the distance between one hemiuterus and the other is longer than the vertical length of both the hemivaginas. In those cases, it is ideal to perform a maneuver called a “vaginal switch” (Figs. 22.9 and 22.10). One of the hemiuteri and the ipsilateral Fallopian tube is resected (Fig. 22.9), with particular care given to preserving the blood supply of the ovary. The blood supply of the hemivagina of that side is sacrificed and the blood supply of the contralateral hemivagina is preserved. Most of the time it provides

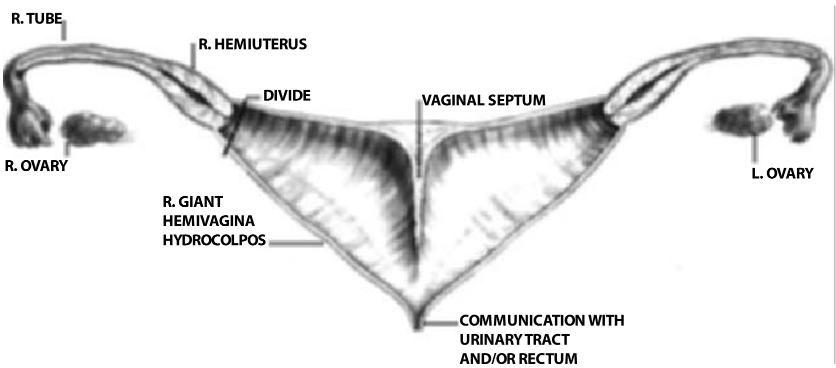

Fig. 22.9 Bilateral hydrocolpos and very high vagina. Ideal anatomy to be repaired with vaginal switch maneuver. *L* Left, *R* right ([9], with permission)

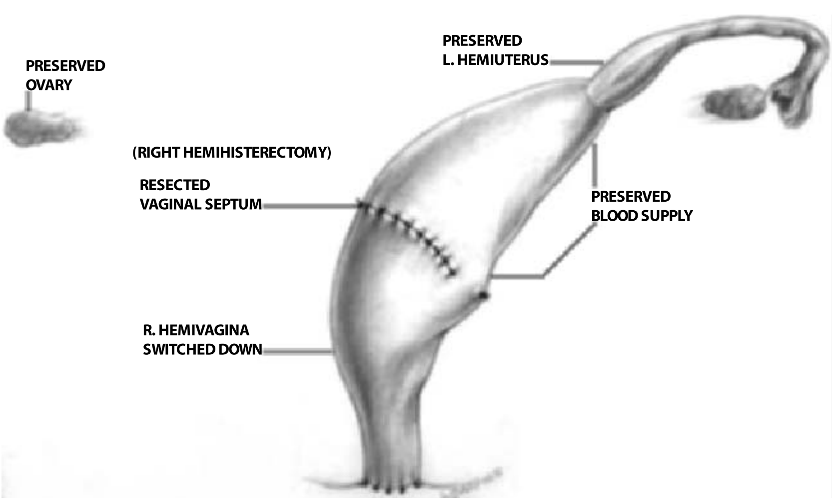

Fig. 22.10 Vaginal switch maneuver, where one vagina is brought to introitus, with excision of ipsilateral hemiuterus and tube and vaginal septum. ([9], with permission)

good blood supply for both hemivaginas. The vaginal septum is resected, and both hemivaginas are tubularized into a single vagina, taking advantage of the long lateral dimension of both hemivaginas together. Then, what used to be dome of the hemivagina, where the hemiuterus was resected, is turned down to the perineum (Fig. 22.10). This is an excellent maneuver that can be performed only when the anatomic characteristics fulfill these unique requirements.

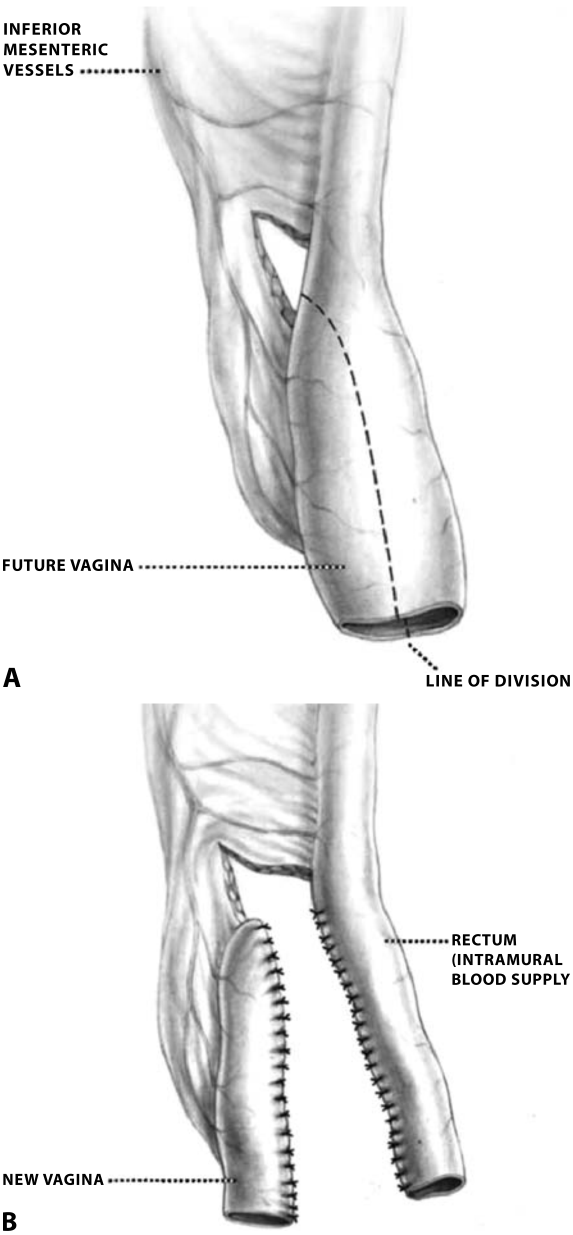

Fig. 22.11 **A** Vaginal replacement with rectum. Planning the neovagina. **B** Neovagina separated from rectum ([10], with permission)

22.2.4 Vaginal Augmentation and/or Replacement

The vagina can be augmented or totally replaced with bowel tissue when it is very small and is located very high, or in cases of absent vagina. The choices are: rectum, colon, or small bowel. Vaginal replacement with rectum is only feasible in patients who have a megarectum that is large enough to be able to divide it longitudinally into a portion with its own blood supply that will represent a new vagina and to leave another half with enough circumference to reconstruct an adequate-sized rectum (Fig. 22.11). The blood supply of the rectum will be provided transmurally from branches of the inferior mesenteric vessels.

The colon is an ideal substitute to replace the vagina (Fig. 22.12). However, sometimes, the location of the colostomy interferes with this type of reconstruction. When available, the sigmoid colon is preferable. One must take the most mobile portion of the colon in order to use a piece that has a long mesentery. When the patient has internal genitalia or a little cuff of vagina or cervix, the upper part of the bowel used for replacement must be sutured to the vaginal cuff. When the patient has no internal genitalia (no vagina and no uterus), the vagina is created and is left with

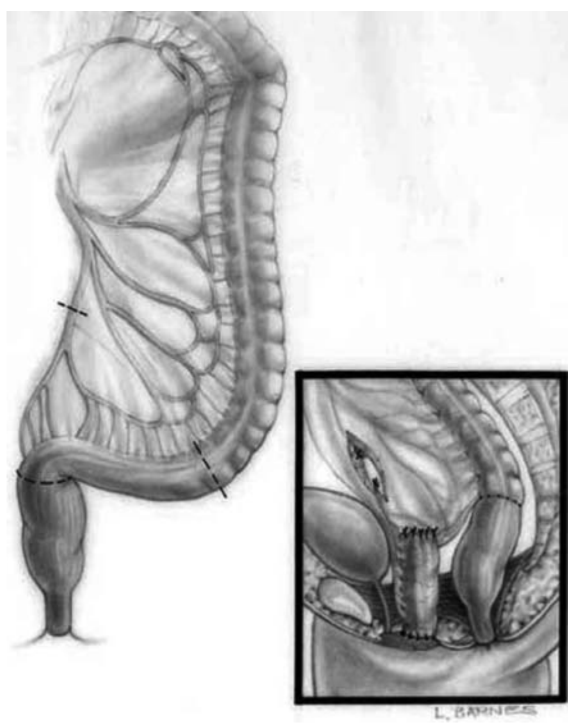

Fig. 22.12 Vaginal replacement with colon ([10], with permission)

its upper portion blind, and is used only for sexual purposes, not for reproduction.

When the colon is not available, then the most mobile portion of the small bowel is utilized for vaginal reconstruction. The mesentery of the small bowel is longer in an area located approximately 15 cm proximal to the ileocecal valve. This is the best portion of small bowel to be used for vaginal replacement. A portion of this ileum is isolated and pulled down, preserving its blood supply (Fig. 22.13).

22.2.5 Two Hemivaginas Attached to the Bladder Neck

In the highest type of cloaca one may find two little hemivaginas attached to the bladder neck or even to the trigone of the bladder. In these cases, the rectum also opens into the trigone. Separation of these structures is performed abdominally. Unfortunately, when that separation is completed, the patient is frequently left with no bladder neck or a severely damaged blad-

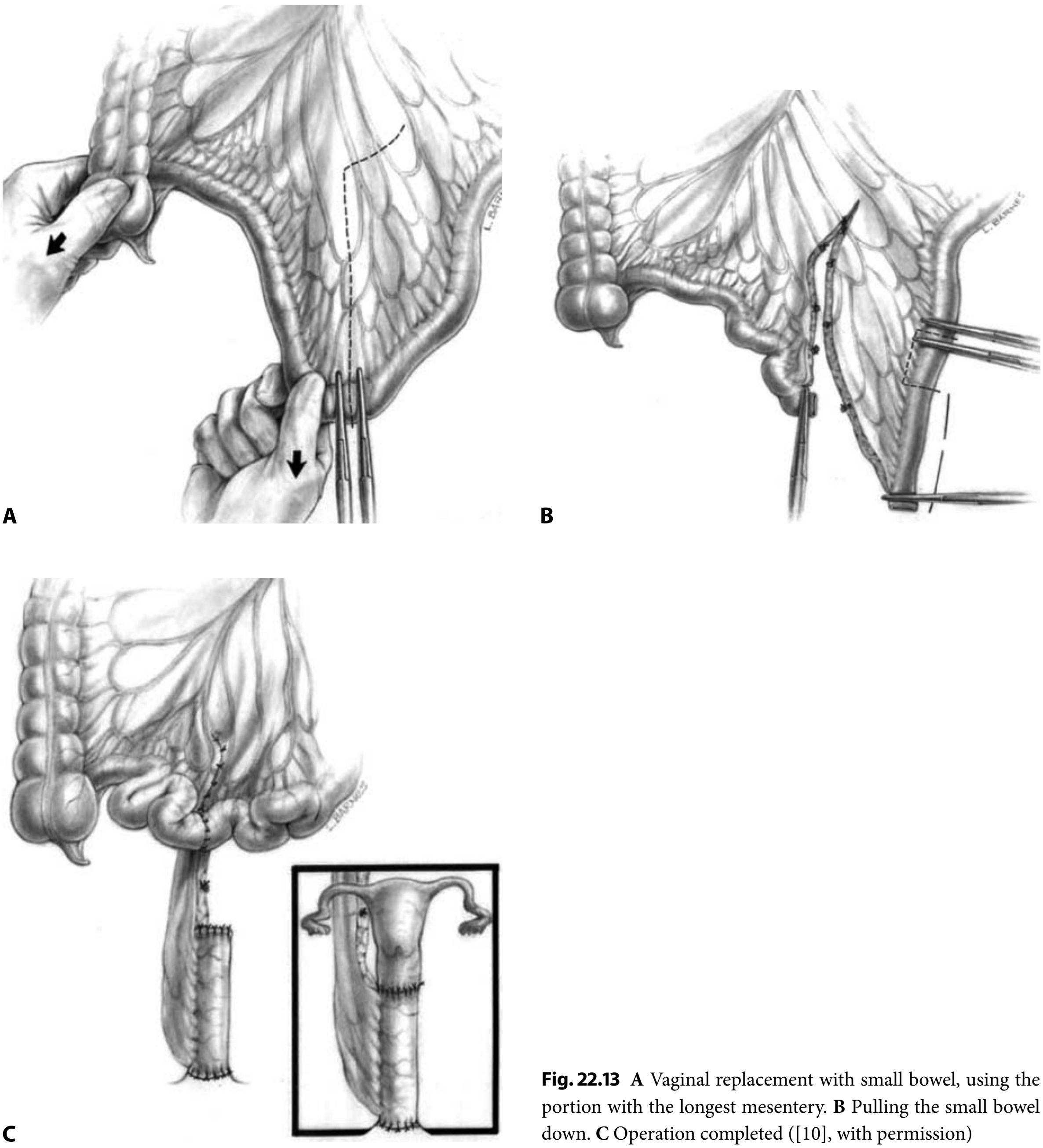

Fig. 22.13 **A** Vaginal replacement with small bowel, using the portion with the longest mesentery. **B** Pulling the small bowel down. **C** Operation completed ([10], with permission)

der neck. At that point, the surgeon must have enough experience to make a decision as to whether to reconstruct the bladder neck or to close it. In the first situation, most patients will need intermittent catheterization to empty the bladder and there is no guarantee that the bladder neck reconstruction will work. In the second situation (permanent closure of the bladder neck), a vesicostomy is created, and the patient will require a continent diversion-type of procedure at the age of urinary continence (3–4 years old). In this particular type of malformation, the patient also needs a vaginal replacement.

22.3 Postoperative Care

The patients with cloacas usually keep the Foley catheter for 10–14 days. In our series, about 20% of patients with a cloaca with common channel shorter than 3 cm require intermittent catheterization to empty the bladder. Patients with common channels longer than 3 cm require intermittent catheterization 70–80% of the time. Therefore, we leave the Foley catheter in place as long as the patient shows signs of swelling in the genitalia and until the urethral meatus is not readily visible. Once we are able to see the urethral orifice, the Foley catheter can be removed in the clinic, and the baby watched to see if she is capable of emptying the bladder. If she cannot pass urine, then we can teach the parent to pass the catheter intermittently.

In cases with very long common channels, we prefer to leave a suprapubic tube. One month after surgery, we can perform a suprapubic cystogram and start clamping the tube and measuring the residual urine, which is an indicator of the efficiency in the function of the bladder. The suprapubic tube remains in place until we have evidence of a good bladder function or the caregiver learns to catheterize the bladder when indicated. The rectal dilatation protocol is the same as described in Chaps 20 and 21. Once the rectum is the appropriate size, the colostomy can be closed. An endoscopy should always be performed prior to colostomy closure to inspect the repair.

References

1. Stephens FD, Smith ED (1971) Incidence, frequency of types, etiology. In: Stephens FD, Smith ED, Paul NW (eds) Anorectal Malformations in Children. Year Book Medical, Chicago, pp 160–171
2. Rosen, NG, Hong AR, Soffer SZ, Rodriguez G, Peña A (2002) Recto-vaginal fistula: a common diagnostic error with significant consequences in girls with anorectal malformations. J Pediatr Surg 37:961–965
3. Levitt MA, Stein DM, Peña A (1998) Gynecological concerns in the treatment of teenagers with cloaca. J Pediatr Surg 33:188–193
4. Peña A, Levitt MA, Hong AR, Midulla PS (2004) Surgical management of cloacal malformations; a review of 339 patients. J Pediatr Surg 39:470–479
5. Peña A (1997) Total urogenital mobilization – an easier way to repair cloacas. J Pediatr Surg 32:263–268
6. Peña A (1989) Female defects. In: Peña A (ed) Atlas of surgical management of anorectal malformations, Springer-Verlag, New York, pp 60–64
7. Peña A (1997) Total urogenital mobilization – an easier way to repair cloacas. J Pediatr Surg 32:263–264
8. Peña A, Levitt MA (2005) Imperforate anus and cloacal malformations. In: Ashcraft KW, Holcomb W, Murphy JP (eds) Pediatric Surgery 4th edn. Elsevier Saunders, Philadelphia, p 509
9. Kiely EM, Peña A (1998) Anorectal malformations. In: O'Neil YA, Rowe MI, Grosfeld JL et al. (eds) Pediatric Surgery. Mosby Yearbook, Elsevier, p 1442
10. Peña A, Levitt MA, Hong AR, Midulla P (2004) Surgical management of cloacal malformations: a review of 339 patients. J Pediatr Surg 39:474–475

23 Laparoscopy-Assisted Anorectal Pull-Through

Keith E. Georgeson and Oliver J. Muensterer

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23.1 Introduction

Posterior sagittal anorectoplasty (PSARP [3]) is the current standard surgical management for patients with anorectal malformations (ARM). Despite the excellent exposure of the anatomy and the exact placement of the distal rectum within the muscle complex with this operation, postoperative fecal continence is less than ideal [4,7]. Tsuji et al. reported a careful analysis of postoperative anorectal function, comparing PSARP with older, conventional operations [10]. They found that patients in both groups had a similar manometry and long-term function. Most of the patients needed bowel management. Other authors have reported similar findings [1,9]. Increased constipation after PSARP compared to a more limited surgical approach has also been reported [2].

The goals of laparoscopic assisted pull-through for ARM include avoiding the dividing and weakening of the external sphincters, diminishing perirectal scarring while allowing for precise placement of the rectum through the external sphincters, and the potential development of a primary procedure in the newborn, which would avoid the morbidity associated with a colostomy.

23.2 Operative Technique

A standard proximal sigmoid colostomy is performed in the newborn. About 2–4 months later, the patient is positioned transversely at the end of the operating table (Fig. 23.1). Circumferential skin preparation is performed from the nipples down to the toes. A catheter is passed into the bladder in all cases, even if cystoscopy is required to do so. A pneumoperitoneum with pressures of 12 cm of water is established. A 4-mm trocar is placed in the anterior-axillary line just below the liver. A 5-mm port is placed through the umbilicus using an open technique. A 3- or 4-mm port is placed in the anterior-axillary line just above the anterior superior iliac spine (Fig. 23.1).

Laparoscopic rectal dissection is initiated at the peritoneal reflection. Using a hook cautery, the distal mesorectum is divided. The dissection is continued circumferentially around the rectum down to the rectourethral or rectovesical fistula. It is important to keep this dissection in the definitive plane between the longitudinal rectal muscles and surrounding tissues. Just proximal to the entrance of the fistula into the urinary tract, a loop ligature is preloaded through the 5-mm trocar in the umbilicus over a Maryland clamp placed through right lower quadrant trocar. This clamp is placed on the fistula several millimeters proximal to the entrance of the fistula into the urinary tract (Fig. 23.2). The fistula is divided proximal to the

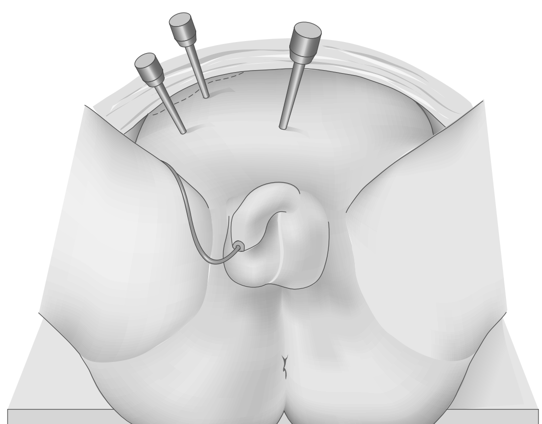

Fig. 23.1 The patient is positioned transversely on the operating table, a bladder catheter is placed, and the trocars are positioned in the illustrated sites (umbilical – 5 mm, right upper and lower quadrant – 4 mm)

placement of the Maryland clamp. The loop ligature is then passed around the Maryland clamp and the fistula and snugged in place, adjacent to the urethra. A second loop can be placed on the rectal fistula proximally in a similar fashion (Fig. 23.3). The rectum is then retracted out of the pelvis. The pubococcygeus muscle can often be visualized when it is present (Fig. 23.4). In some patients with ARM, particularly the higher lesions, the levator ani muscle is poorly developed. However, in many patients with a rectoprostatic fistula, the muscle is quite well developed and can be seen from above with the endoscope.

A transperineal dissection follows division of the rectourethral fistula. The external anal sphincters are mapped using a transcutaneous electrostimulator. The area of maximal contraction is identified and marked appropriately with sutures. A 1-cm vertical midline incision is made at the site of the maximal muscle contraction. The intersphincteric plane is gently dissected from below the level of the levator sling up through the muscle complex bluntly (Fig. 23.5). A radially expanding trocar is then passed over a Veress needle through this intersphincteric plane and advanced between the two bellies of the pubococcygeus muscle in the midline just posterior to the urethra, using laparoscopic guidance. If the needle is inaccurately passed to either side of the midline it is readily apparent due to the laparoscopic surveillance. The Veress needle is

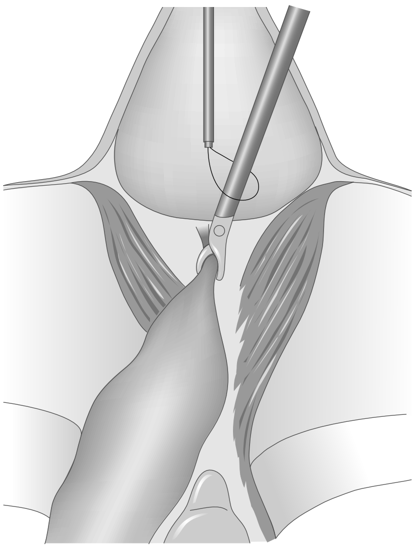

Fig. 23.2 After circumferential dissection of the rectum, the fistula is grasped with a Maryland clamp preloaded with a loop ligature. The fistula is then divided on the rectal side of the clamp, and the ligature is tightened around the urethral side of the clamp

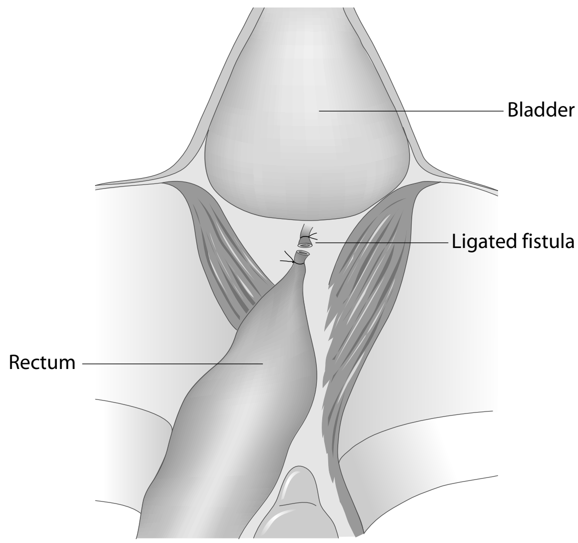

Fig. 23.3 A second loop ligature is used to close the fistula on the rectum

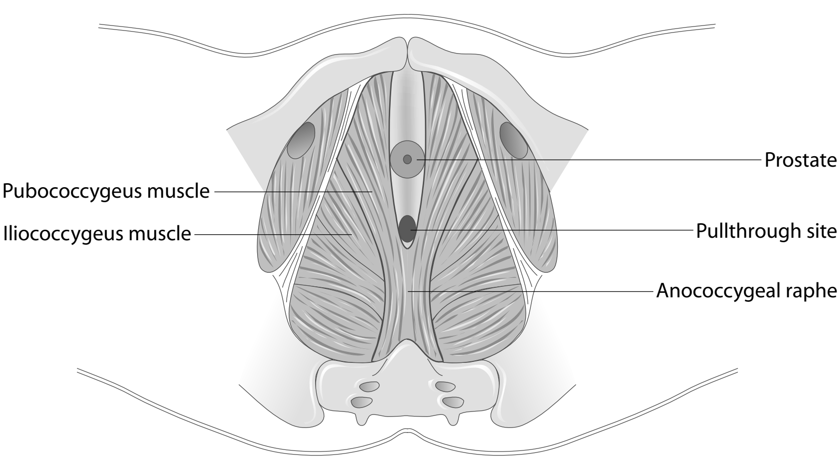

Fig. 23.4 Anatomic diagram of the pull-through site in relation to the pubococcygeus muscle

redirected to the correct position prior to the dilatation of the tract through the expandable trocar sleeve. The tract is dilated radially up to 10–12 mm. The rectal fistula is then grasped through the transperineal trocar and is pulled down onto the perineum trailing the trocar (Fig. 23.6). The anastomosis between the rectum and the anus is completed with a polyglycolic acid suture. The rectum is retracted cephalad laparoscopically and secured in this retracted position with 2-0 silk sutures (Fig. 23.7). It is important to place these hitch stitches to avoid prolapse of the rectal mucosal wall through the anus and also to lengthen the skin-lined anal canal.

Patients are fed on the first or second postoperative day. Graduated anorectal dilatation is started 2–3 weeks after surgery. The colostomy is closed 2–3 months after the pull-through procedure is completed.

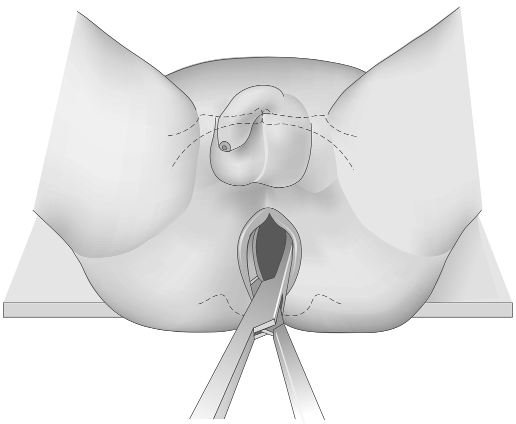

Fig. 23.5 Transperitoneal blunt dissection of the intersphincteric plane is performed through a 1-cm vertical incision using a clamp (the underlying external muscle complex and the pubococcygeus muscle are *dotted*)

23.3 Results

The best comparative study between the laparoscopic-assisted anorectal pull-through and the PSARP has been reported by Lin et al [6]. Nine patients had a laparoscopy-assisted pull-through and 13 had a PSARP. Lin et al. reported equal centrality of the pull-through segment when comparing the PSARP approach and the laparoscopic approach. However, sphincter asymmetry was much greater with the posterior sagittal approach, as was sphincter irregularity. Megarectum and constipation were also greater in the PSARP group. Eight of the 9 laparoscopy-assisted pull-through patients developed an anorectal reflex after laparoscopic pull-through, while only 4 out of 13 PSARP patients developed an anorectal reflex after surgery. As many other authors have noted, eventual continence is re-

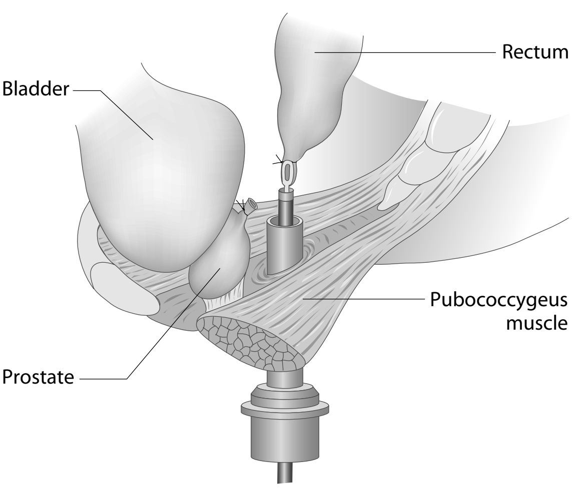

Fig. 23.6 Schematic diagram of the trocar passing from the peritoneum between the two bellies of the pubococcygeus muscle into the abdomen. A grasper is advanced through the port to grasp the distal end of the dissected rectum. The rectum is then pulled down through the perineum trailing the trocar

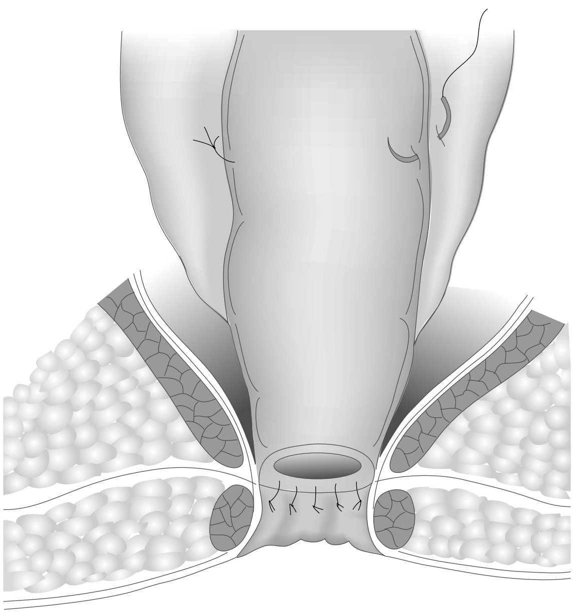

Fig. 23.7 After performing the anastomosis between the rectum and anus, the rectum is retracted cephalad and secured to the presacral fascia using lateral hitch stitches to avoid prolapse of the rectal mucosa through the anus

lated to a positive anorectal reflex [5,8]. Lin et al. also reported similar anal resting pressures after the two operations. However, rectal compliance was much better in the laparoscopy-assisted approach when compared to the PSARP group of patients [6].

23.4 Discussion

Laparoscopy-assisted anorectal pull-through seems to achieve some of its stated goals. There does not seem to be any question that the centrality of the pull-through inside the sphincter complex is achieved successfully. In addition, there appears to be less scarring with the laparoscopic approach when compared to the PSARP approach, as demonstrated by magnetic resonance imaging [11]. Long-term follow-up for fecal continence has yet to be determined. Because the internal sphincter is not well developed in these patients, long-term continence may not be significantly improved. A normally functioning internal sphincter is certainly a great aid to fecal continence.

In summary, laparoscopy-assisted anorectal pull-through is anatomically sound and leaves the external sphincter muscles intact. This technique allows for the centrality of the pull-through inside the sphincter complex. There is a higher incidence of the anorectal reflex in patients after laparoscopy-assisted pull-through than after PSARP. There is also less scarring of the pelvic floor, resulting in better rectal compliance. Long-term follow-up for continence is needed for further evaluation of this technique.

References

1. Bliss DP Jr, Tapper D, Anderson JM, et al (1996) Does posterior sagittal anorectoplasty in patients with high imperforate anus provide superior fecal continence? J Pediatr Surg 31:26–30
2. Chen CC, Lin CL, Lu WT, et al (1998) Anorectal function and endopelvic dissection in patients with repaired imperforate anus. Pediatr Surg Int 13:133–137
3. DeVries PA, Peña A (1982) Posterior sagittal anorectoplasty. J Pediatr Surg 17:638–643
4. Langemeijer RA, Molenaar JC (1991) Continence after posterior sagittal anorectoplasty. J Pediatr Surg 26:587–590
5. Lin CL, Chen CC (1996) The rectoanal relaxation reflex and continence in repaired anorectal malformations with and without an internal sphincter-saving procedure. J Pediatr Surg 31:630–633
6. Lin CL, Wong KK, Lan LC, et al (2003) Earlier appearance and higher incidence of the rectoanal relaxation reflex in patients with imperforate anus repaired with laparoscopically assisted anorectoplasty. Surg Endosc 17:1646–1649
7. Rintala RJ, Lindahl H (1995) Is normal bowel function possible after repair of intermediate and high anorectal malformations? J Pediatr Surg 30:491–494
8. Rintala RJ, Lindahl HG (2001) Fecal continence in patients having undergone posterior sagittal anorectoplasty procedure for a high anorectal malformation improves at adolescence, as constipation disappears. J Pediatr Surg 36:1218–1221
9. Schuster T, Lagler F, Pfluger T, et al (2001) A computerized vector manometry and MRI study in children following posterior sagittal anorectoplasty. Pediatr Surg Int 17:48–53
10. Tsuji H, Okada A, Nakai H, et al (2002) Follow-up studies of anorectal malformations after posterior sagittal anorectoplasty. J Pediatr Surg 37:1529–1533
11. Wong KK, Khong PL, Lin SC, et al (2005) Post-operative magnetic resonance evaluation of children after laparoscopic anorectoplasty for imperforate anus. Int J Colorectal Dis 20:33–37

24 Complications after the Treatment of Anorectal Malformations and Redo Operations

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24.1 Introduction

In spite of the technical advances in the surgical repair of anorectal malformations (ARM) that have occurred over the last 20 years, complications that require a secondary procedure are still common. Surprisingly, this need is not isolated to the group of patients with very complex malformations. Instead, it spans the entire spectrum of malformations seen, and in fact is quite common for the relatively benign malformations.

Reoperative surgery may be considered for several reasons (see Chap. 25). Fecal incontinence may be present after the first operation, and a surgeon may wish to attempt to improve on the results. Other patients may have suffered significant, sometimes catastrophic complications because of technical errors and require revisional surgery to alleviate pain, discomfort, and other sequelae. Finally, mismanagement of constipation can lead to significant sequelae.

It is clear that a patient's best chance for a good functional result is when the proper operation is performed during the first definitive procedure and complications are avoided [1]. This is especially true in those patients born with a defect that has a good prognosis. It is unfortunate when such patients end up with fecal or urinary incontinence as a result of avoidable complications of the surgical repair.

Complications in patients who have undergone surgical repair of an ARM can be grouped into three categories: (A) those with fecal incontinence requiring a reoperation, (B) those who have suffered a perioperative complication, and (C) those suffering from sequelae resulting from constipation (Table 24.1) [2].

Table 24.1 Complications associated with anorectal malformations

Group A	Patients with fecal incontinence requiring reoperation
Group B	Perioperative complications • Wound infection • Femoral nerve palsy • Rectal problems: dehiscence, retraction, infection and/or acquired atresia • Rectourinary fistula • Rectovaginal fistula • Persistent urogenital sinus • Acquired vaginal atresia • Acquired urethral atresia • Posterior urethral diverticulum • Urologic injuries • Neurogenic bladder • Complications involving the laparoscopic approach • Rectal prolapse
Group C	Complications from mismanagement of constipation

24.1.1 Group A, Patients with Fecal Incontinence Requiring Reoperation

Reoperation to improve a patient's functional prognosis is indicated in several circumstances. During the first 5 years of our experience, reoperative surgery was performed on every patient we evaluated who underwent a repair at another institution and suffered from fecal incontinence. During those years, we hoped that the new posterior sagittal approach would give these patients an opportunity to recover bowel control. When the results were evaluated [3,4], only 30% of those patients experienced a significant improvement. Therefore, the indications for surgery were modified.

At the present time, reoperation for fecal incontinence is recommended only for patients with very special criteria. Those born with a malformation associated with a goog prognosis, with a rectum that is completely mislocated, with an intact rectosigmoid, a normal sacrum, and an intact sphincter mechansim are candidates. The rectal location can be evaluated by magnetic resonance imaging if its mislocation is not obvious by inspection.

For these reoperations, the rectum is approached posteriorly. Multiple silk stitches are placed at the mucocutaneous margin in order to apply uniform traction to facilitate the dissection and mobilization of the rectum. A full rectal dissection and mobilization is performed, staying as close as possible to the bowel wall but avoiding injury. The limits of the sphincters, including the parasagittal fibers, muscle complex, and levator muscle, are determined by electrical stimulation and the rectum is repositioned within it.

In many cases, the patient is found to have had the colon, and not the rectum pulled down, which is identified by the presence of mesentery attached to the bowel. In such cases, the mesenteric fat must be trimmed from the last few centimeters of the rectum to allow for direct contact between the sphincter mechanism and the colonic wall. An anoplasty performed within the limits of the sphincter mechanism completes the reconstruction.

The number of patients that require a reoperation for fecal incontinence has decreased significantly over the years. This is probably due to the increased use of the posterior sagittal approach, which provides superior exposure and prevents the complete mislocation of the rectum that was seen with other techniques.

Years ago many patients underwent abdominal perineal pull-through procedures with endorectal dissections of the rectosigmoid [5]. This procedure essentially resulted in loss of the rectosigmoid. These patients do not suffer from constipation. Instead they suffer from increased colonic motility and a tendency to diarrhea. It took several years to recognize this specific group of patients, and today revisional surgery is not offered to them, because it is clear that they never regain bowel control. Fortunately, endorectal pull-throughs are no longer performed for ARM and it is rather unusual to see these patients.

In addition to the aforementioned group, those patients born with poor prognosis defects and fecal incontinence are also considered inappropriate candidates for reoperation. These patients typically have an abnormal sacrum, flat perineum, and poor sphincters. There is usually evidence that they were born with a high rectoprostatic or rectobladderfistula, or a cloaca with a common channel longer than 3 cm. Their sacral ratio is almost always less than 0.4. We do not reoperate on these patients, even if they have a completely mislocated rectum because they do not improve after reoperation. Instead they are offered a bowel management program [6] in order to prevent soiling and to keep them completely clean (see Chaps. 29 and 30).

When revisional surgery for fecal incontinence is offered, the likelihood of the patient regaining bowel control is reviewed with the family. Even with those patients who are expected to improve, the bowel management program is implemented prior to surgery. If it turns out that the patient does not improve enough after reoperation to avoid enemas, the already tested bowel management is reinstituted.

24.1.2 Group B, Perioperative Complications

Group B includes those who sustained a perioperative complication during or shortly after the first operation [2]. The specific types of complication are described below.

24.1.2.1 Wound Infection

Wound infection of the posterior sagittal incision is very uncommon in the immediate postoperative period, but is more prevalent in the presence of a loop colostomy, which might not be completely diverting, or in cases operated without a colostomy. Fortunately, the infections usually affect only the skin and subcutaneous tissue and heal secondarily, without functional sequelae.

24.1.2.2 Femoral Nerve Palsy

Transient femoral nerve palsy can be observed, particularly in adolescent patients, and is a consequence of excessive pressure in the groin during the PSARP operation. This problem can be avoided by adequate cushioning of the patient's groin area.

24.1.2.3 Rectal Problems

Patients can experience dehiscence, retraction, infection and/or acquired atresia of the rectum related to technical problems arising during the pull-through procedure. These are usually the result of excessive tension or inadequate blood supply. In addition, anal strictures may result when families do not follow the prescribed protocol of dilatations.

Reoperation for these patients proceeds posterior sagittally. In cases of retraction, dehiscence, and acquired atresia, the rectum is usually located somewhere high in the pelvis and is surrounded by a significant amount of fibrosis. Multiple 6-0 silk sutures are placed in the rectal wall in order to exert uniform traction and facilitate a circumferential dissection of the rectum, again trying to stay as close as possible to the rectal wall without injuring it. Bands and extrinsic vessels surrounding the rectum are divided and cauterized circumferentially until enough rectal length is gained so as to place the rectum within the limits of the sphincter mechanism.

Short ring-like rectal strictures can be treated with a Heineke-Mikulicz type of plasty. Strictures that are longer than 1 cm must be resected, with the rectum mobilized until the fibrotic portion can be removed, and a fresh nonscarred portion of rectum pulled down, creating a new anus.

Based on our anatomic findings during these reoperations, we speculate that retraction, dehiscence, and acquired rectal atresia were most likely due to a poor technique used to mobilize the rectum. During a primary procedure, the rectum, when seen posterior sagittally, is covered by a very characteristic white fascia that contains vessels to the rectum. The surgeon must dissect this fascia off the rectum, remaining as close as possible to the rectal wall. Uniform traction provided by multiple silk sutures is imperative to facilitate the dissection. Bands and the extrinsic rectal blood supply must be divided to gain rectal length. The intramural blood supply of the rectum is excellent; and the rectum can be dissected to gain significant length provided the rectal wall is not injured. The most likely cause for difficulty in dissection of the rectum is working outside the fascia. Alternatively, dissection too close to the rectum can injure the rectal wall, interfere with the intramural blood supply, and provoke ischemia. The result of all this is an incomplete mobilization, rectal ischemia, and a rectal-to-skin anastomosis under tension, which may explain most of these complications.

We speculate that rectal strictures are also most likely due to ischemia of the distal part of the rectum. When the rectum is correctly mobilized and the blood supply kept intact, it is extremely unlikely to see an anal stricture. A few patients of ours who were operated on primarily failed to follow our protocol of dilatations and returned months after their operation with strictures. These patients had a thin fibrotic ring in the area of the anoplasty, which was easy to treat either with an anoplasty or dilatations. A long narrow stricture is most likely due to rectal ischemia.

Some surgeons do not have their patients follow a protocol of anal dilatations. In order to avoid painful maneuvers to the patient, they follow a specific plan consisting of taking the patient to the operating room every week and under anesthesia performing forceful dilatations. Those dilatations can actually provoke lacerations in the anal verge, which then heal with scarring, only to be reopened during the next forceful dilatation, leading ultimately to an intractable ring of fibrosis.

24.1.2.4 Rectourinary and Rectovaginal Fistulae

Patients may have various types of rectogenitourinary tract fistula complications. Fistulae can be persistent when the original rectourethral fistula remains untouched during the main repair, even when the rectum was repaired. Recurrent fistulae may occur if the surgeon repaired the fistula but it reopened. Acquired rectourethral fistulae are those that are inadvertently created during the repair of a malformation [7].

Acquired rectovaginal fistulae can occur during a failed attempt at repair of a rectovestibular fistula. In the past urethra-vaginal fistula was the most common and feared complication in cases of persistent cloaca prior to the introduction of the total urogenital mobilization maneuver [8]. In certain circumstances, the vagina can be rotated to try to prevent this complication [9], but even with that maneuver, these fistulae can occur.

Persistent rectourethral fistulae can occur in patients who were born with a rectourethral bulbar fistula and underwent a repair that did not address the fistula. We speculate that surgeons following the old diagnostic approach performed an invertogram and found the bubble of rectal air close to the skin. This may have led to an approach through the perineum, with identification of the rectum and subsequent pull-through and anoplasty. Since the surgeon was completely unaware of the low rectourethral bulbar fistula, it was not repaired.

Recurrent rectourethral fistulae may result if the fistula is closed, but the rectum is not mobilized adequately, leaving the anterior wall under tension. A dehiscence of the anterior rectal wall may explain the recurrence of the fistula. Also, leaving sutures in the rectum adjacent to the sutures in the urethra may lead to the formation of a recurrent fistula. Consequently, an injured rectum that requires a repair to the anterior wall may lead to the development of a recurrent fistula. The same explanation may apply to rectovaginal fistulae.

Acquired rectourethral fistulae can occur in male patients who were born with rectoperineal fistulae if they undergo their first operation without a Foley catheter in the urethra. During the mobilization of the anterior wall of the rectum, an unrecognized urethral injury can occur and, if not mobilized to leave normal rectal wall in front of the urethral injury, an acquired rectourethral fistula will form.

Fistulous complications can be approached posterior sagittally. The posterior rectal wall should be opened and the fistulae identified and closed. The rectum then needs to be separated from the urinary tract or the vagina, and mobilized as so as to be sure that a completely normal anterior rectal wall is left in front of the urethral or vaginal suture line.

24.1.2.5 Persistent Urogenital Sinus

This problem occurs in patients born with a cloaca who underwent an operation in which the rectal component of the malformation was repaired, but the urogenital sinus was ignored (Fig. 24.1). Their reoperation can also be approached posterior sagittally. The rectum must be completely dissected and reflected out of the way. This allows exposure of the urogenital sinus, which can be repaired using the same technique that is employed during the treatment of a cloaca [8,9].

All of the patients in one of our reports came to us with an original diagnosis of "rectovaginal fistula" [10]. The word cloaca was never mentioned in the operative report. Only the rectal component of the malformation was repaired and the urogenital sinus was left unattended. In our experience, a true congenital rectovaginal fistula is an extremely unusual defect [1], yet in the literature from previous years, we have found frequent mention of this malformation and very little reference to cloacas [11–17]. In the recent literature, there is an increase in the diagnosis of cloacas and less mention of rectovaginal fistulae, which, we think, reflects the improved understanding of the true anatomy of these lesions [18–21].

24.1.2.6 Acquired Vaginal Atresia

Complete fibrosis of the vagina can occur as a result of excessive dissection in an attempt to mobilize a high vagina, during a failed attempt to repair a cloaca. This comlication requires a reoperation and sometimes a vaginal replacement. It usually occurs when the vagina is separated from the urethra during the repair of cloacas. Such maneuvers are rarely used since the advent of total urogenital mobilization [8]. The separation of the vagina from the urinary tract is not an easy maneuver; the vagina may become devascularized and as a consequence, patients develop ischemic vaginal atresia. Those patients with acquired vaginal atresia are treated using the same surgical approach as for persistent urogenital sinus.

24.1.2.7 Acquired Urethral Atresia

Acquired urethral atresia in cloaca patients can occur as a result of devascularization of the pulled-through

Fig. 24.1 Pull-through only of the rectum in a patient with a cloacal malformation, leaving the urogenital sinus untouched ([27], with permission)

or reconstructed neourethra. In male patients, it has occurred when the urethra was accidentally transected during an attempt to repair an anorectal malformation. This complication mostly occured when the surgeon approached a patient posterior sagitally without a pre-operative high pressure distal colostogram. To repair this problem the rectum must be mobilized in order to expose the urethral area. Both urethral ends need to be identified, dissected, and mobilized enough to perform a tension-free end-to-end anastomosis.

24.1.2.8 Posterior Urethral Diverticulum

This complication can occur when a retained portion of the rectum is left attached to the posterior urethra (Fig. 24.2). The same reoperative approach to fix it can be utilized, with a posterior sagittal incision, mobilization of the rectum to expose the posterior aspect of the urinary tract, identification of the diverticulum, and dissection of it down to the urethra. The diverticulum can be separated from the urethra in the same manner as in a primary repair of ARM, with the urethra closed and the diverticulum resected.

This complication occurs in patients born with a rectourethral bulbar fistula that was repaired transabdominally. It is easy to understand that the surgeon was unable to reach the fistula site through the abdomen. Consequently, the rectum was amputated, leaving a piece of rectum attached to the urethra. This potential complication is possible with a laparoscopic approach, particularly if the rectum reaches well below the peritoneal reflection.

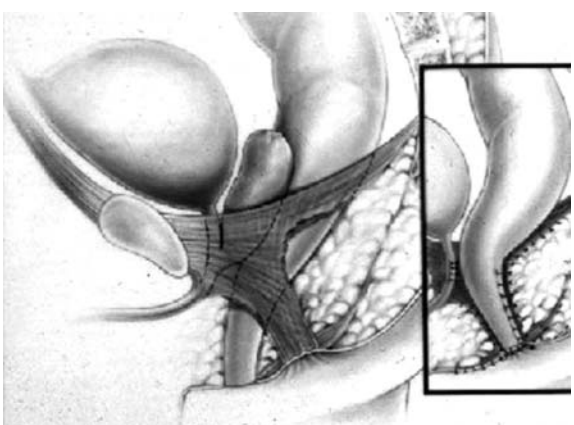

Fig. 24.2 Posterior urethral diverticulum, representing the undissected former distal rectum ([27], with permission)

These patients, are initially asymptomatic, but after years develop symptoms such as passage of mucus through the urethra, dribbling, orchioepididimitis, urinary tract infections, and urinary pseudoincontinence. In addition, we saw one such patient who, after 30 years, developed an adenocarcinoma in the piece of rectum left attached to the urethra.

24.1.2.9 Other Urologic Injuries

Significant urologic injuries such as transection of the bladder neck, transection of the urethra, and injury to the vas deferens, seminal vesicles, prostate, and ectopic ureters have occurred, usually when the posterior sagittal approach was performed without a good previous distal colostogram, and thus the precise anatomy was not known prior to the posterior sagittal dissection [7].

24.1.2.10 Neurogenic Bladder

Neurogenic bladder in male patients with ARM is extremely unusual. In our series, we have only seen it in patients with a very abnormal sacrum or associated spinal anomalies. If it does occur, it mostly likely represents a denervation of the bladder and bladder neck during the repair. It is very important to note that patients with cloacal malformations are different with regard to this subject. Patients with cloaca often have a deficient emptying mechanism of the bladder. They do not have the typical "Christmas tree" type of image of a neurogenic bladder seen in patients with a spina bifida. They have rather a flaccid, smooth, large bladder that does not empty completely. Fortunately, most patients with cloacas have a very good bladder neck. The combination of a good bladder neck with a floppy, flaccid bladder, make these patients ideal candidates for intermittent catheterization, which keeps them completely dry.

Two exceptions to this rule exist. One is represented by patients who have a very long common channel, in which the hemivaginas as well as the rectum are attached to the bladder neck and after these are separated the patients are left with no bladder neck or a very damaged bladder neck. The second group is represented by a small number of cloaca patients who are born with separated pubic bones, who could be described as having a covered exstrophy [22]. These patients have no bladder neck congenitally and they eventually require a continent diversion type of operation.

24.1.2.11 Complications Specific to the Laparoscopic Approach to ARM

Long-term results and complications have not yet been described for the laparoscopic approach to imperforate anus, which is a relatively new approach that was developed to avoid a laparotomy and to minimize the posterior sagittal incision [23]. We have managed several complications with this approach, and can anticipate others.

Avoidance of the exposure required to perform the posterior sagittal approach can lead to inadvertent injuries, such as injury to the bladder neck, urethra, or an ectopic ureter. Precise understanding of the anatomic relationships between the pelvis and the laparoscopic view is vital to avoid these problems. Like the transabdominal approach, there is potential for leaving behind the distal rectal cuff, leading to a posterior urethral diverticulum, particularly for malformations below the peritoneal reflection, such as rectobulbar fistula. Finally, to avoid rectal prolapse, a pelvic hitch is employed; if this step is omitted or done incorrectly, the incidence of prolapse will probably be significant. With the avoidance of the posterior sagittal incision, the described laparoscopic operation omits several key steps of the PSARP that are very important to avoid prolapse [24], particularly tacking of the posterior rectal wall to the muscle complex.

24.1.2.12 Rectal Prolapse

Rectal mucosal prolapse occurs following PSARP, with an incidence of 3% [24]. It is more common in patients with higher malformations and with poor sacral and pelvic musculature. Significant prolapse may lead to ulceration, bleeding, and mucus production, and can interfere with anal canal sensation and thus impact upon a patient's functional prognosis. Correction of the prolapse can be performed transanally, with mobilization of the redundant full-thickness rectum, and redo-anoplasty. This is ideally done prior to colostomy closure. Sometimes, however, prolapse only develops after colostomy closure and in the presence of constipation [24]. The repair of the prolapse in patients without a colostomy requires a strict preoperative bowel preparation, insertion of a central line, nothing by mouth for 7 days, and administration of parenteral nutrition.

24.1.3 Group C, Sequelae of Constipation

Group C includes those patients referred to our institution because of fecal incontinence who actually had untreated severe constipation, chronic impaction, and therefore suffered from overflow pseudoincontinence. All of these patients have several factors in common. All were born with a malformation with good functional prognosis and underwent a technically correct, successful operation. Postoperatively, they all had severe constipation which was not adequately treated and developed megasigmoid and chronic fecal impaction. Adequate treatment of their constipation, with or without a sigmoid resection [25], rendered them fecally continent (see Chaps. 29, 30 and 32].

Constipation is the most common functional disorder observed in patients who undergo posterior sagittal anorectoplasty [1]. Interestingly the incidence of constipation is inversely related to the frequency of voluntary bowel movements. This means that patients with the best prognosis for bowel control have the highest incidence of constipation. Patients with very poor prognosis, such as bladder neck fistula, have a rather low incidence of constipation.

It seems from analysis of our series that constipation is related to the degree of preoperative rectal ectasia. Colostomies that do not allow cleaning and irrigation of the distal colon lead to megarectum. Transverse colostomies lead to a micro left colon with dilatation of the rectosigmoid. Loop colostomies allow for passage of stool and distal fecal impaction. It is clear that keeping the distal rectosigmoid empty and not distended from the time the colostomy is established and proceeding with pull-through and subsequent colostomy closure as early as possible within several months results in better ultimate bowel function [26].

All patients in this pseudoincontinent group underwent a laxative test to determine whether they were fecally continent. First, large-volume enemas were administered until the patient's colon was clean (disimpacted). Daily laxatives were then administered, increasing the amount each day until the amount necessary to produce colonic evacuation was determined. A plain abdominal x-ray was obtained every day to assess the colonic emptying (see Chaps. 29, 30 and 32). If the patient demonstrated the capacity to feel the stool in the rectum, reach the bathroom, have voluntary bowel movements, and remain clean every day, the patient was considered continent. The patient was then offered the option of continuing the treatment with large quantities of laxative for an indefinite period of time or a sigmoid resection [25] in order to make the constipation more manageable and thereby decreasing the laxative requirement.

It is extremely important to recognize this group of patients. Some may be wrongly diagnosed as suffering from true fecal incontinence and some have even undergone reoperations such as gracilis muscle or artificial sphincters, which can actually make the patient worse. This problem should be suspected when one sees a patient who was born with a benign malformation, who underwent a technically correct operation, but who was not treated correctly for constipation.

24.2 Conclusion

Unfortunately, despite great advances in pediatric surgical care, there remain a significant number of patients who undergo attempted anorectal repairs with significant complications, many of which are preventable. One must have a thorough understanding of these malformations, utilize a meticulous technique, and employ rigorous, careful postoperative management if repairs are to be successful. These basic fundamentals need to be emphasized in the training of young pediatric surgeons, so as to improve the outlook for children born with ARM.

References

1. Peña A (1995) Anorectal Malformations. Semin Pediatr Surg 4:35–47
2. Peña A, Hong AR, Midulla P, Levitt M (2003) Reoperative surgery for anorectal malformations. Semin Pediatr Surg 12:118–123
3. Peña A, deVries PA (1982) Posterior sagittal anorectoplasty: important technical considerations and new applications. J Pediatr Surg 17:796–811
4. Peña A (1990) Advances in the management of fecal incontinence secondary to anorectal malformations. In: Nyhus L (ed) Surgery Annual. Appleton and Lange, Connecticut, pp 143–167
5. Kiessewetter WB (1967) Imperforate Anus. II - The rationale and technique of the sacroabdominoperineal operation. J Pediatr Surg 2:106–110
6. Peña A, Guardino K, Tovilla JM, Levitt MA, Rodriguez G, Torres R (1998) Bowel management for fecal incontinence in patients with anorectal malformations. J Pediatr Surg 33:133–137
7. Hong AR, Rosen N, Acuña MF, Peña A, Chaves L, Rodriguez G (2002) Urological injuries associated with the repair of anorectal malformations in male patients. J Pediatr Surg 37:339–344
8. Peña A (1997) Total urogenital mobilization. An easier way to repair cloacas. J Pediatr Surg 32:462–468
9. Peña A, Levitt MA, Hong A, Midulla P (2004) Surgical management of cloacal malformations: a review of 339 patients. J Pediatr Surg 39:470–479
10. Rosen NG, Hong AR, Soffer SZ, Rodriguez G, Peña A (2002) Rectovaginal fistula: a common diagnostic error with significant consequences in girls with anorectal malformations. J Pediatr Surg 37:961–965
11. Wangensteen O, Rice C (1930) Imperforate anus: a method of determining the surgical approach. Ann Surg 92:77–81
12. Brenner EC (1938) The rectum and anus. In: Brenner EC (ed) Pediatric Surgery. Lea and Feibiger, Philadelphia, pp 537–571
13. Ladd WE, Gross RF (1941) Malformations of the anus and rectum In: Ladd WE, Gross RF (eds) Abdominal Surgery in Infancy and Childhood. WB Saunders, Philadelphia, pp 166–187
14. Santulli TV (1952) The treatment of imperforate anus and associated fistulas. Surg Gynecol Obstet 95:601–614
15. Gross RE (1953) Malformations of the anus and rectum. In: Gross RE (ed) The Surgery of Infancy and Childhood. WB Saunders, Philadelphia, pp 348–367
16. Santulli TV (1962) Imperforate anus. In: Benson CD, Mustard WT, Ravitch MM, et al (eds) Pediatric Surgery. Year Book Medical Publishers, Chicago, pp 821–836
17. Swenson O, Donellan WL (1967) Preservation of the puborectalis sling in imperforate anus repair. Surg Clin North Am 47:173–193
18. Santulli TV, Schullinger JN, Kieseweitter WB, et al (1971) Imperforate anus: a survey from the members of the Surgical Section of the American Academy of Pediatrics. J Pediatr Surg 6:484–487
19. Bill AH, Hall DG, Johnson RJ (1975) Position of rectal fistula in relation to the hymen in 46 girls with imperforate anus. J Pediatr Surg 10:361–365
20. The Japan Study Group of Anorectal Anomalies (1970) A group study for the classification of anorectal anomalies in Japan with comments to the International Classification. J Pediatr Surg 17:302–308
21. Stephens FD, Smith ED (eds) (1988) Anorectal Malformations in Children: Update 1988. March of Dimes Birth Defects Foundation, Birth Defects: original article series 24(4), Alan R. Liss, New York
22. Soffer SZ, Rosen NG, Hong AR, Alexianu M, Peña A (2000) Cloacal exstrophy: a unified management plan. J Pediatr Surg 35:932–937
23. Georgeson KE, Inge TH, Albanese CT (2000) Laparoscopically assisted anorectal pull-through for high imperforate anus – a new technique. J Pediatr Surg 35:927–931
24. Belizon A, Levitt M, Shoshany G, Rodriguez G, Peña A (2005) Rectal prolapse following posterior sagittal anorectoplasty for anorectal malformations. J Pediatr Surg 40:192–196

25. Peña A, El-Behery M (1993) Megasigmoid – a source of pseudo-incontinence in children with repaired anorectal malformations. J Pediatr Surg 28:1–5
26. Peña A, Krieger M, Levitt MA (2006) Colostomy in anorectal malformations – a procedure with significant and preventable complications. J Pediatr Surg 41:748–756
27. Peña A (1989) Secondary operations. In: Peña A (ed) Atlas of surgical management of anorectal malformations, Springer-Verlag, New York, pp 80–83

Results and Aftercare

25 Postoperative Pathophysiology of Chronic Constipation and Stool Incontinence

Alexander M. Holschneider, Jürgen Koebke, William A. Meier-Ruge, and Stefanie Schäfer

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25.1 Introduction

The treatment of ARM either by a perineal, sacral, abdominal, or combined approach is technically demanding and requires strict adherence to the finer anatomical and physiological details to achieve good results. The most important problem after imperforate anus repair is chronic constipation. It is astonishing that in the period from 1953, when Stephens presented first his sacral approach, up to 1982, when Peña and de Vries introduced the posterior sagittal anorectoplasty (PSARP) procedure, fecal incontinence represented the main postoperative problem [1,2]. After the introduction of PSARP, primary fecal incontinence dropped considerably down from 31 to 11.8% [3], but chronic constipation with overflow incontinence became the most important postoperative sequelae [4–9].

25.2 Postoperative Outcome after ARM

In 2001, Holschneider et al. [3] compared two groups of patients after ARM treatment: results achieved before the introduction of PSARP in the period 1962–1984 ($n = 381$) and those achieved after the introduction of Peña's technique (period 1985–1997; $n = 197$). Incontinence decreased during these two periods in high types of ARM from 55% to 24%, in intermediate types from 30% to 11%, and in low forms from 8% to zero. The frequency of partial continence increased in the same time interval from 32% to 56% in the high types and decreased from 60% to 47% in the intermediate forms. It remained unchanged at 25% and 21%, respectively, in the low malformations. Continence increased from 13.5% to 20% in high, 10% to 42% in intermediate, and 67% to 79% in low anomalies (Fig. 25.1).

The main postoperative problem in our last series from 1985 to 1997 of 197 children operated according to Peña was chronic constipation with overflow incontinence: 63.2% of the patients showed no constipation and had normal defecation habits, but 28.6% of the children were occasionally, and 8.3% always constipated. In other series too, chronic constipation is a well-known phenomenon. In 1992, Hedlund and Peña reported on 9 out of 30 severely constipated children and 6 with additional overflow soiling [10].

In 1995 Peña described his results in 285 children [4]. He observed chronic constipation to varying degrees in 61.4% with vestibular fistulae, 55.0% with bulbar fistulae, and 41.4% with prostatic fistulae. Soiling occurred in 0–82.6% of his patients depending on the type of the fistula. It is well known that vestibular, bulbar, and prostatic fistulae usually need more preparation to allow sacroperineal pull-through than do perineal and bladder-neck fistulae. Therefore, the possibility of damage to the extramural nerve supply to the rectum is higher in intermediate-type lesions than in low- or high-type malformations. On the other hand, in bladder-neck fistulae the development of the striated muscle complex is hypoplastic, favor-

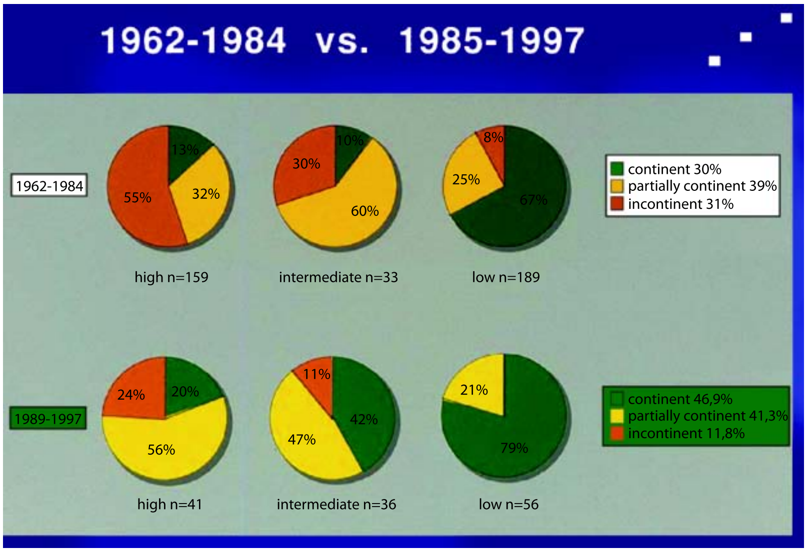


Fig. 25.1 Continence, partial continence, and incontinence in two groups of patients (periods 1962–1984 and 1985–1997; from Holschneider et al. 2001 [3])

ing anal incontinence as a result of a reduced anorectal pressure barrier. Therefore, soiling was observed in 82.6% of patients with bladder-neck fistulae. Bliss et al. [11] and Chau et al. [12] found similar results (Table 25.1).

In addition, chronic constipation is also an ongoing problem after repair of low ARM. In 1997 Rintala et al. [13] found constipation in 17 out of 40 (42%) children after correction of low ARM. In addition, four patients (10%) suffered from daily soiling by overflow incontinence. Only half of the children with low ARM had age-appropriate normal bowel function.

25.3 Continence and the Mechanism of Defecation in Normal Individuals

The most important physiological factors contributing to fecal control in normal individuals are a normal propulsive activity of the rectum, good rectal wall compliance, a high resting tone, contractility of the smooth and striated anal sphincters, and intact sensation. It is important to remember that 70–80% of the anorectal resting pressure profile is established by the internal anal sphincter. The striated muscle fibers of the external anal sphincter and pelvic floor muscles are an additional help, but not the main factors of the anorectal pressure barrier (Table 25.2) [14,15].

The relationship between continence and the mechanism of defecation is described in Chap. 7 and can be summarized as follows. Normally, the rectum is empty. Various reflexes, such as the gastrocolic reflex and the ileocolic reflex (contraction of the colon caused by filling of the stomach or ileum, respectively) as well as voluntary contraction of the abdominal musculature, may initiate defecation by filling the rectum with colonic contents. The increasing intrarectal pressure stimulates the distension receptors in the puborectalis muscle and the parapuborectal tissues, and desire to pass stool is consciously felt. At the same time, a reflex relaxation of the internal anal sphincter occurs. This allows even the smallest amounts of stool to reach the anal canal. The hypersensitive mucosa of the anal canal is able to distinguish the difference between flatus and liquid or solid stool. The reflex contraction of the external anal sphincter and the puborectalis will prevent expulsion of stool from the anal canal and thus inhibit fecal soiling. This effect is increased by the compression of the lower anal canal by the engorged hemorrhoidal vessels of the rectum and the corrugator muscle of the anus. This allows the rectum time to adapt itself toward the increased intraluminal pressure. The aboral–oral pressure gradient of the rectum will propel the stool upward into a more proximal rectal segment. This will, however, stimulate a further propulsive wave via a feed-back-mechanism. An intrarectal pressure of 25–30 mmHg will stimulate a reflex inhibiting the anorectal sphincters and the puborectalis muscles. Voluntary contraction of the abdominal wall muscles will also cause a reciprocal inhibition of the striated muscles of the pelvic floor. This, in turn, will decrease the acuteness of the anorectal angle formed by the puborectalis muscle, and defecation commences. The increasing desynchronization of electrical activities in the smooth muscle fibers will cause dilatation of the anal canal at first in its upper third, and later throughout

Table 25.1 Frequency of continence and soiling according to Peña 1995 [4]

		Soiling (%)			Constipation (%)		
		Grade I	Grade II	Total	Grade I	Grade II	Total
Low	Perineal Fistula (n = 14)	0.0	0.0	0.0	21.5	7.1	28.6
	Vestibular Fistula (n = 44)	20.9	9.3	30.2	34.1	27.3	61.4
Intermediate	Bulbar Fistula (n = 42)	29.1	35.4	64.6	11.1	44.4	55.5
	Imperforate without Fistula (n = 17)	5.5	33.3	38.9	16.7	33.3	50.0
	Vaginal Fistula (n = 4)	50.0	50.0	100	0	25.0	25.0
High	Cloacal Fistula (n = 38)	23.6	44.7	68.4	2.7	25.0	27.7
	Prostatic Fistula (n = 57)	18.9	55.1	74.1	15.5	25.9	41.4
	Bladderneck Fistula (n = 19)	4.3	78.2	82.6	4.6	13.6	18.2
Other	Rectal Atresia or Stenosis (n = 5)	0.0	0.0	0.0	20.0	20.0	40.0
Total	(n = 245)	18.7	38.2	57.0	15.4	27.6	43.1

Table 25.2 Physiological factors contributing to anorectal continence. *ARRP* Anorectal resting pressure profile

Anatomical site	Physiological factor
Rectum	Propulsive bowel motility Compliance (adaptation reaction)
Pelvic floor	Anorectal angle Resting tone and contractility of striated muscle complex Sensibility: feeling of fullness
Internal anal sphincter	Resting tone (70–80% of ARRP) Internal sphincter relaxation reflex
External anal sphincter	Resting tone and voluntary contractility of muscle fibers Fine continence (hemorrhoidal plexus) Rectosphincteric reflex
Anorectum	Discrimination Warning period

its whole length, and thus the intestinal contents of the descending colon, rectosigmoid, and rectum will be expressed by contraction of the abdominal muscles with a closed epiglottis in a few strong propulsive waves [16].

The etiology of defecation problems after PSARP is multifactorial and includes: (1) sacral malformations, (2) altered rectosigmoid motility, (3) sphincteric insufficiency, and (4) secondary psychological problems. In some cases, fecal incontinence is a complication of surgery (e.g., a mislocated rectum); however, in most children fecal incontinence is secondary to the defect. Children with sacral agenesis, and males with a rectal fistula to the bladder neck had the highest rates of fecal incontinence, followed by females with a high confluence cloaca [4].

25.4 Surgical Anatomy of the PSARP Approach

The most important factor for postoperative continence is adequate surgical technique. It is therefore necessary to mention the anatomical sections of a PSARP approach performed by Huber et al. in 1983 [17]. His preparations in a human cadaver with normal pelvic anatomy correspond with the recent PSARP procedure in children with ARM (Fig. 25.2).

In newborn babies, the anatomical situation is similar to that in adults, but is more difficult to demonstrate, especially under surgical conditions. However, the nerve supply inside the ailerons latéraux, the lateral fascial wings, shows the same course and density [3] (Fig. 25.3). These postganglionic extramural cholinergic nerve fibers running from the pelvic plexus are responsible for the propulsive rectal evacuation and descend from segments S2 to S4. They should only be cut inside Waldeyer's fascia and directly on the rectal wall.

Unfortunately, in patients with ARM the anatomical structures of the rectum, anorectal sphincters, and their nerve supply are frequently not well developed. This includes malformations of the intramural plexus, pelvic fascia, and the blood supply to the blind pouch (see CD Chap. 6 and Chap. 7).

In piglets with anal atresia, the ailerons latéraux appear to be less well developed and the nerve sup-

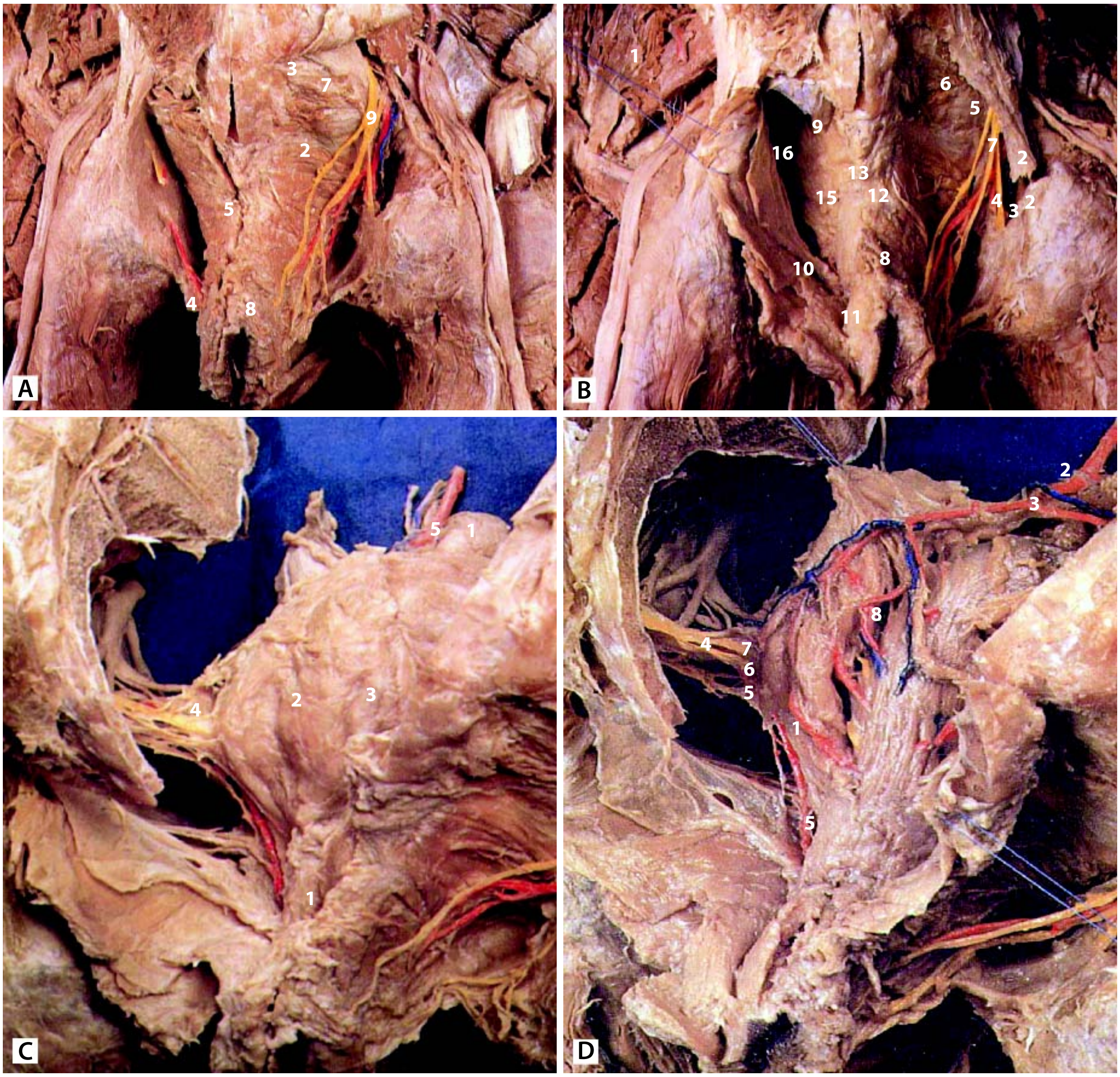


Fig. 25.2 **A** The external anal sphincter (8), the urogenital diaphragm (4), the puborectalis muscle (5) as part of the levator ani muscles (2), and the pudendal nerves (9) running lateral into Alcock's canal, as well as branches of the pudendal artery and the inferior rectal nerves (9). The external anal sphincter is innervated by the pudendal nerves; however, the levator ani muscles and the upper part of the puborectalis muscle are innervated by branches of the sacral plexus laying on the inner surface of the pelvic floor. Sacrospinal ligament (7), coccygeal muscle (3). **B** The pelvic floor is opened and the levator ani (10) and sphincter muscles cut (8). The internal muscle fibers of the levator muscles can be seen inserting into the longitudinal muscle layer of the rectum (11). The sacrotuberal ligament (2) is cut as well and elevated. The inferior rectal nerves and the inferior rectal artery (7) are visible. Here, the pudendal nerves can be seen more closely (4), passing into Alcock's canal (3) after having left the pelvis. Very important are the rectal fascias (15) and next to them, the lateral rectal wings (also called "ailerons latéraux" (16), the lateral fixation of the rectum. Ischial spines (5), sacrospinal ligament (6), anococcygeal ligament (12/13). **C** After having removed the sacrum, the rectal fascias (2) can be demonstrated. Only the dorsal part of the rectal fascia is called Waldeyer's fascia (3). Laterally, the rectal fascia passes over into the internal parietal pelvic fascia. mesosigmoid (5), rectum (1). **D** This is the most important view for PSARP. It shows branches of the medial rectal artery (5), the venous plexus (6), and the parasympathetic splanchnic nerves (the erigentes and pelvic nerves). They run together with the sympathetic nerve fibers of the inferior hypogastric plexus of the pelvic sympathetic trunk inside the ailerons lateraux to the rectum (7). The endarteries and the tiny endings of the nerve fibers are passing through Waldeyer's perirectal fascia into the muscular wall of the rectum. Pouch of Douglas (8) (reproduced from Huber et al 1993) [17]

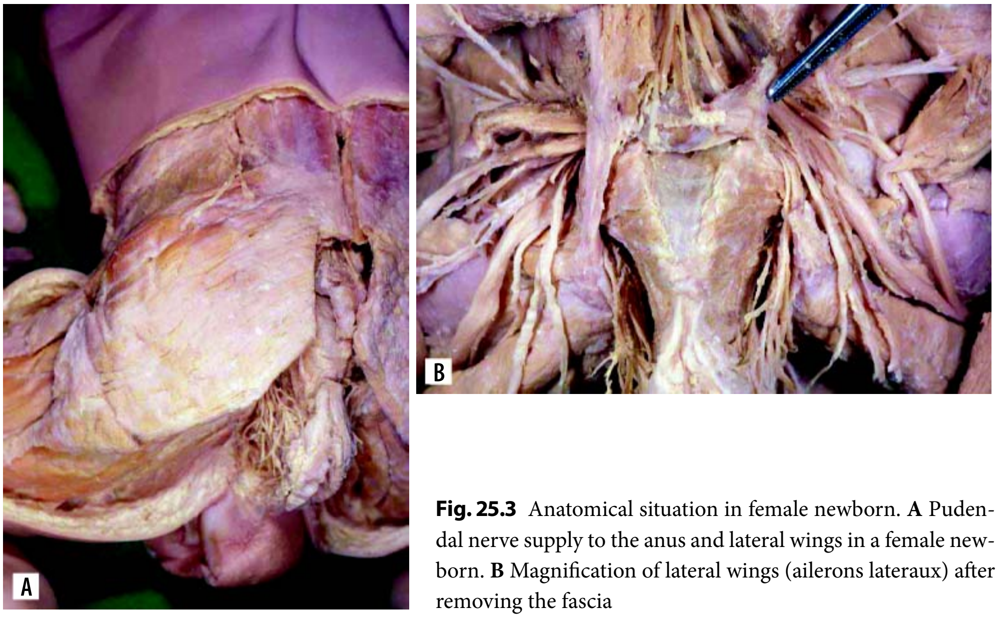


Fig. 25.3 Anatomical situation in female newborn. **A** Pudendal nerve supply to the anus and lateral wings in a female newborn. **B** Magnification of lateral wings (ailerons lateraux) after removing the fascia

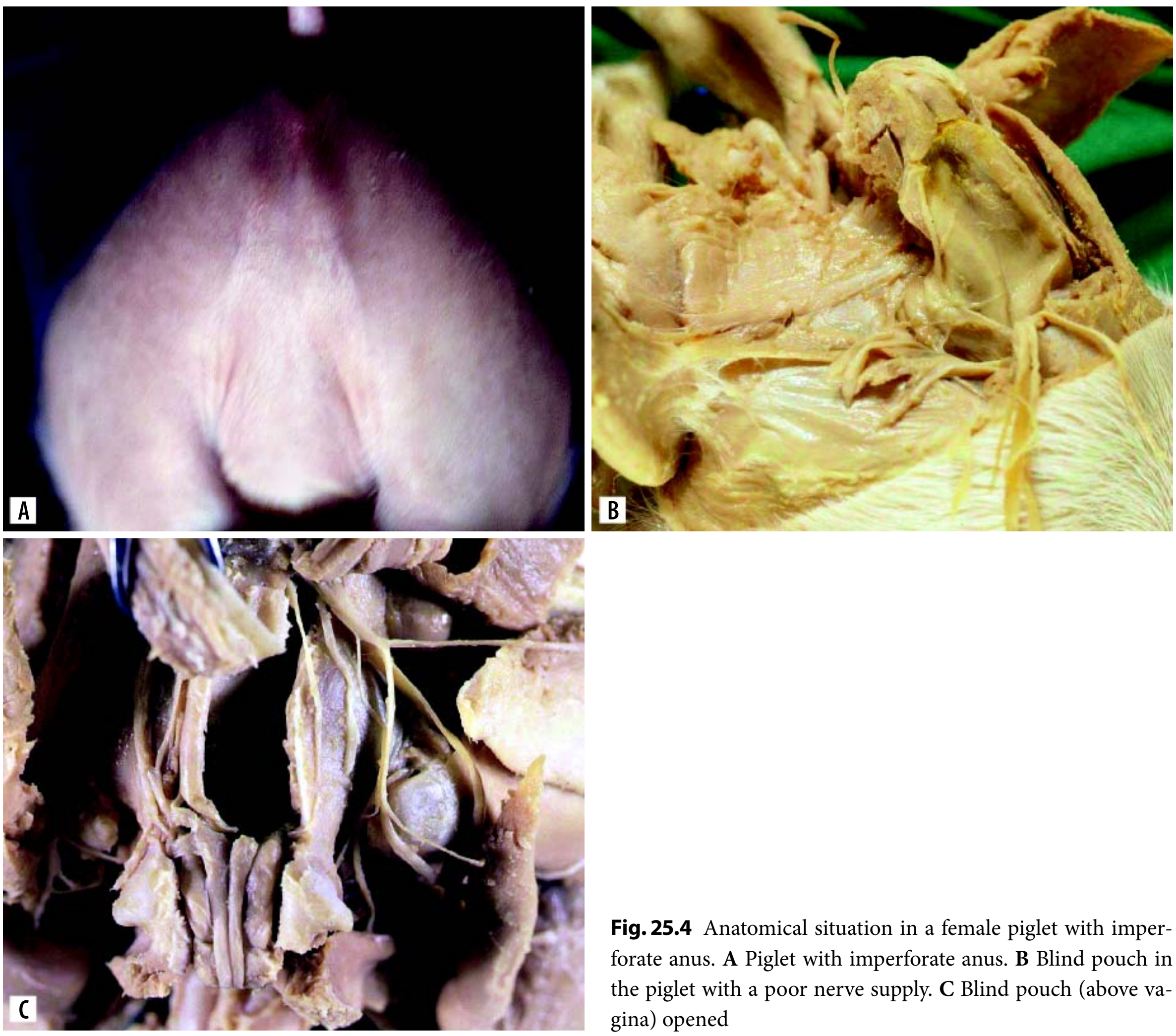


Fig. 25.4 Anatomical situation in a female piglet with imperforate anus. **A** Piglet with imperforate anus. **B** Blind pouch in the piglet with a poor nerve supply. **C** Blind pouch (above vagina) opened

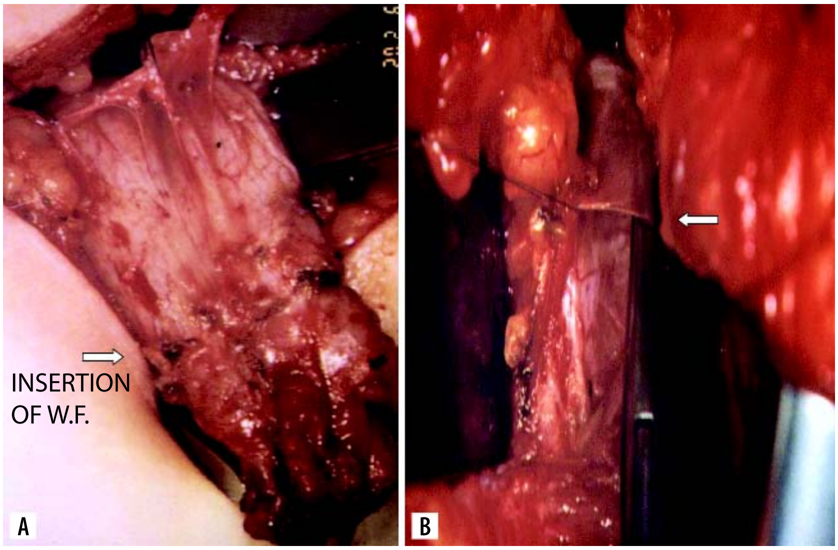
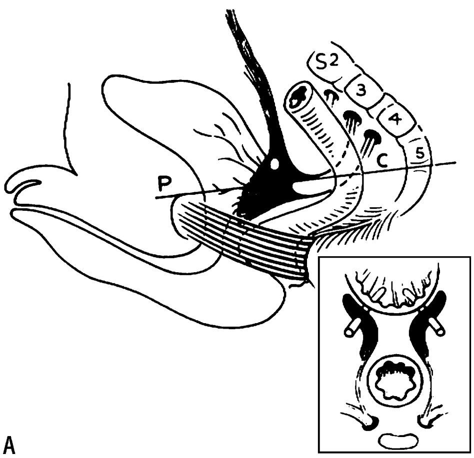


Fig. 25.5 **A** Insertion of Waldeyer's fascia (*W.F.*) in a patient with intermediate anal atresia. **B** Waldeyer's perirectal fascia incised (*green arrow*)



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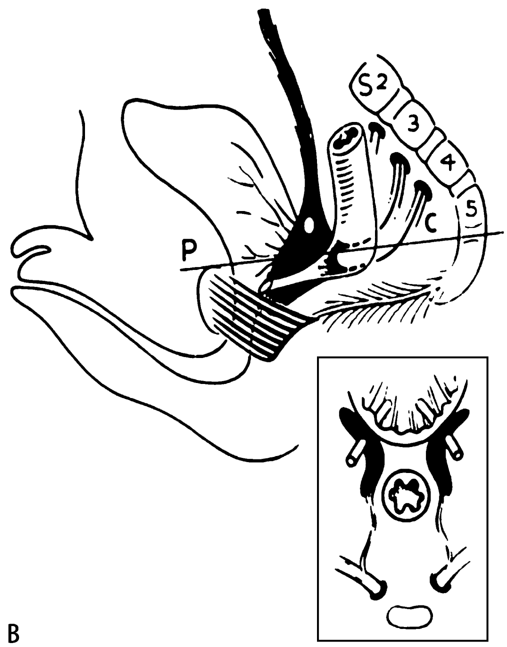

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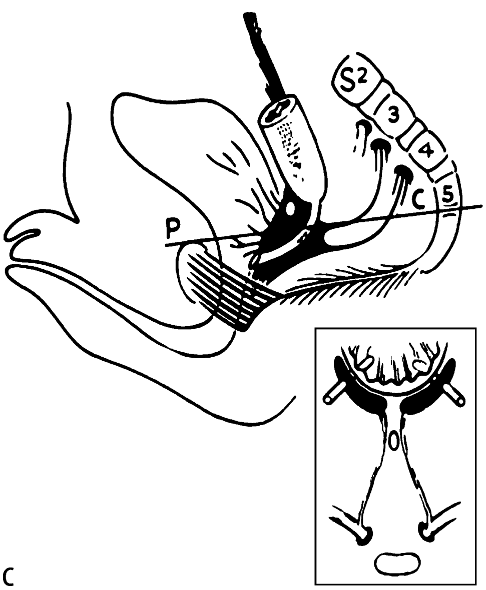

C

Fig. 25.6 Course of the erigentes nerve, according to Stephens 1988 [2]. **A** Normal situation. **B** Intermediate type imperforate anus with rectourethral fistula. **C** High-type anorectal malformation (ARM). *P---C* Pubococcygeal line

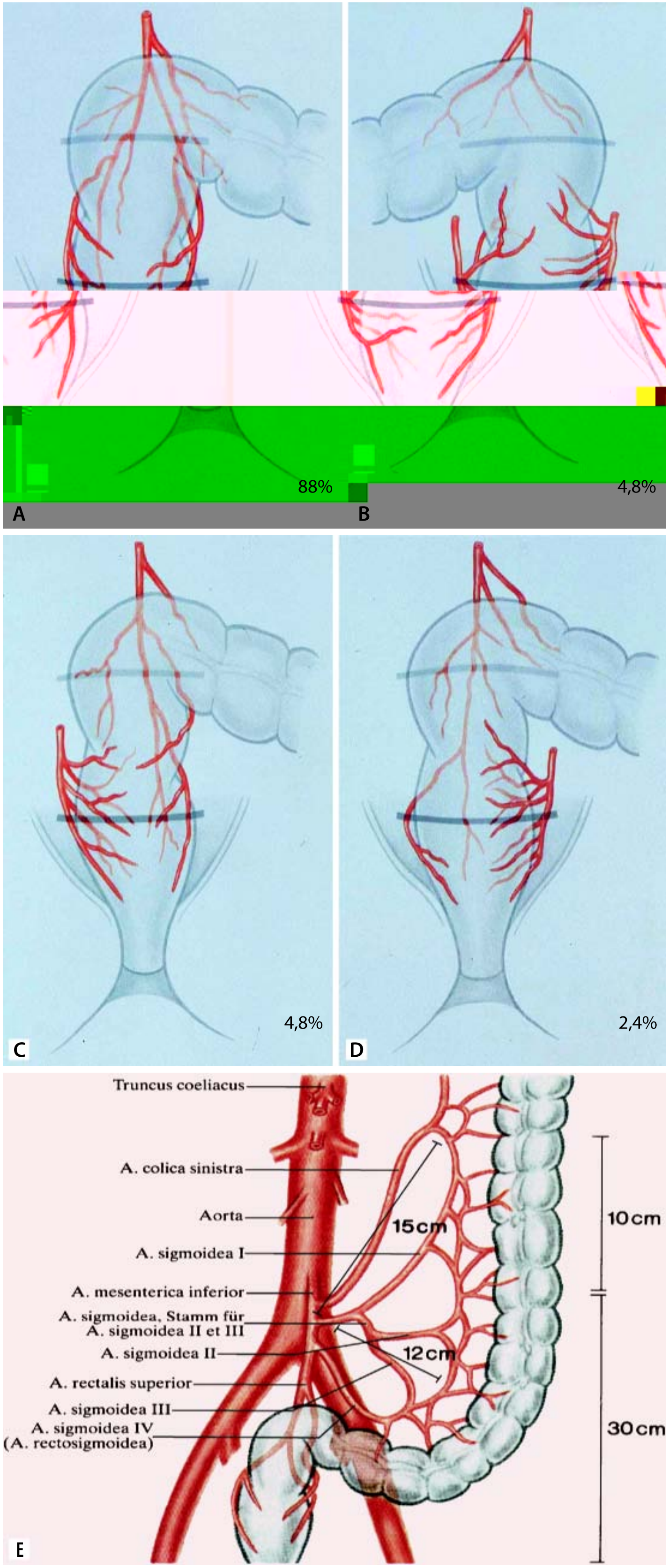

Fig. 25.7 **A**–**D** Variations of arterial supply to the rectum and rectosigmoid according to van Lanz and Wachsmuth 1982 [22]. **A** Normal course of rectal arteries in 88% of patients. **B** Bilateral middle rectal arteries (4.8%). **C** Only the left branch coming from superior rectal artery (4.8%). **D** Only the right branch coming from superior rectal artery (2.4%). **E** Branches of inferior mesenteric artery to the rectosigmoid. *A* Artery

ply seems to be poorer (Fig. 25.4). Likewise, the nerve supply of the external anal sphincter seems to be less dense, which might be explained by the purely reflex-controlled fecal continence of these animals [3]. Contribution of the ailerons latéraux to fixation of the rectum in the small pelvis might account for stronger development in men, particularly in adults.

25.5 Pathologic Anatomy in ARM

25.5.1 Extrinsic Malformations of the Anorectum

In patients with ARM, Waldeyer's fascia is hardly visible and is very difficult to demonstrate (Fig. 25.5). It is therefore difficult to identify the erigentes nerves during preparation of the blind pouch. For that reason it is even more important to dissect directly on the rectal wall and to identify the individual neurovascular bundles at the point where they penetrate the rectal wall, and to dissect them as little as absolutely necessary for the pull-through.

Peña et al. had already pointed in 1993 [18] that after perisacral approach (PSA) and perirectal dissection in dogs, 57.1% of the animals were able to hold an enema; however, after additional intensive perirectal dissection only 44.4% could do so. They concluded that PSA did not hurt the perirectal nerve supply, but extended perirectal dissection does.

In this context it is important to remember the studies of Stephens et al. [1,2] and Kelly [19,20]. These authors pointed out that the stimulating parasympathetic pelvic nerves (erigentes nerves) take an atypical course in patients with ARM. They demonstrated that pelvic nerves normally pass the pubococcygeal line separately on the right and the left of the rectum. In patients with high ARM, nerve fibers pass more medially. They may pass medially and caudally along the lower pole of the rectal pouch only in its inferior part and around the fistula (Fig. 25.6).

These observations are in accordance with findings made by Davies [21]. In specimens from corpses, Davies found that the autonomic nerves pass laterally to the rectal wall and are covered with endopelvic fascia. He stressed the importance of Denonvillier's fascia (fascia prostatoperinealis) on the ventral side of the blind pouch. The nerves of the pelvic plexus, which supply the urogenital intestines, lie on Denonvillier's fascia anterior to the rectum. In ARM, the fascia, too, is frequently not developed, and for that reason nerve fibers are particularly easily damaged. Denonvillier's fascia should not be touched. A neurogenic bladder can result from an excessively extensive mobilization inside this fascia prostatoperinealis.

The vascular supply to the rectum also varies to a great extent [22]. There might exist one well-developed middle rectal artery on each side, or there may be only one on one side, the other half of the rectum receiving its vascular supply as a branch of the superior rectal artery. Or else both of the middle rectal arteries are lacking and the whole vascular supply of the rectum comes from the superior rectal artery. This becomes especially important in pull-through procedures, in particular in cloacal malformations. The branches of the inferior mesenteric and sigmoid vessels, usually 15 and 12 cm long, respectively, might be too short to allow a pull-through for both the vagina and the rectum. An enforced trial to do so may result in vascular problems, leading to stenosis (Fig. 25.7).

25.5.2 Intrinsic Malformations of the Anorectum

Fetal experiments by Pickard et al. [23] revealed that an intestinal obstruction evolving in early fetal life might cause secondary alterations in the plexus of the intestinal wall. According to our own studies in 1994 and 1996, aganglionosis, hypoganglionosis, intestinal neuronal dysplasia (IND), desmosis, and deficiency of interstitial cells of Cajal may occur [24–26]. We also observed a case with duplication of the plexus, and another where the internal anal sphincter consisted of longitudinal muscle fibers of the rectum instead of, as usual, the circular muscle layer (Fig. 25.8).

Martuciello et al. [27] examined 100 suction rectal biopsy samples from the fistulae, and at 6, 4, 3, and 2 cm distance to the opening of the fistula in 22 patients with ARM. They detected innervation disorders of the rectal wall in 81.8% of the biopsy samples. All female patients with vestibular fistulae exhibited dysganglionosis. Aganglionosis was found in 18% of all patients.

Masumoto et al. [26] described anomalies of enteric neurons, intestinal pacemaker cells, and smooth muscle in human intestinal atresia. They observed a hypoplasia of the myenteric ganglia and a marked reduction of intramuscular nerve fibers, including nitrergic neurons, in the dilated proximal segment of intestinal atresia. C-kit-positive cells were localized around the myenteric plexus, but were rarely found within the muscularis propria in the proximal segment. A reduced staining intensity for monoclonal antibodies to α-smooth muscle actin was observed mainly in the hypertrophic circular muscle layer of

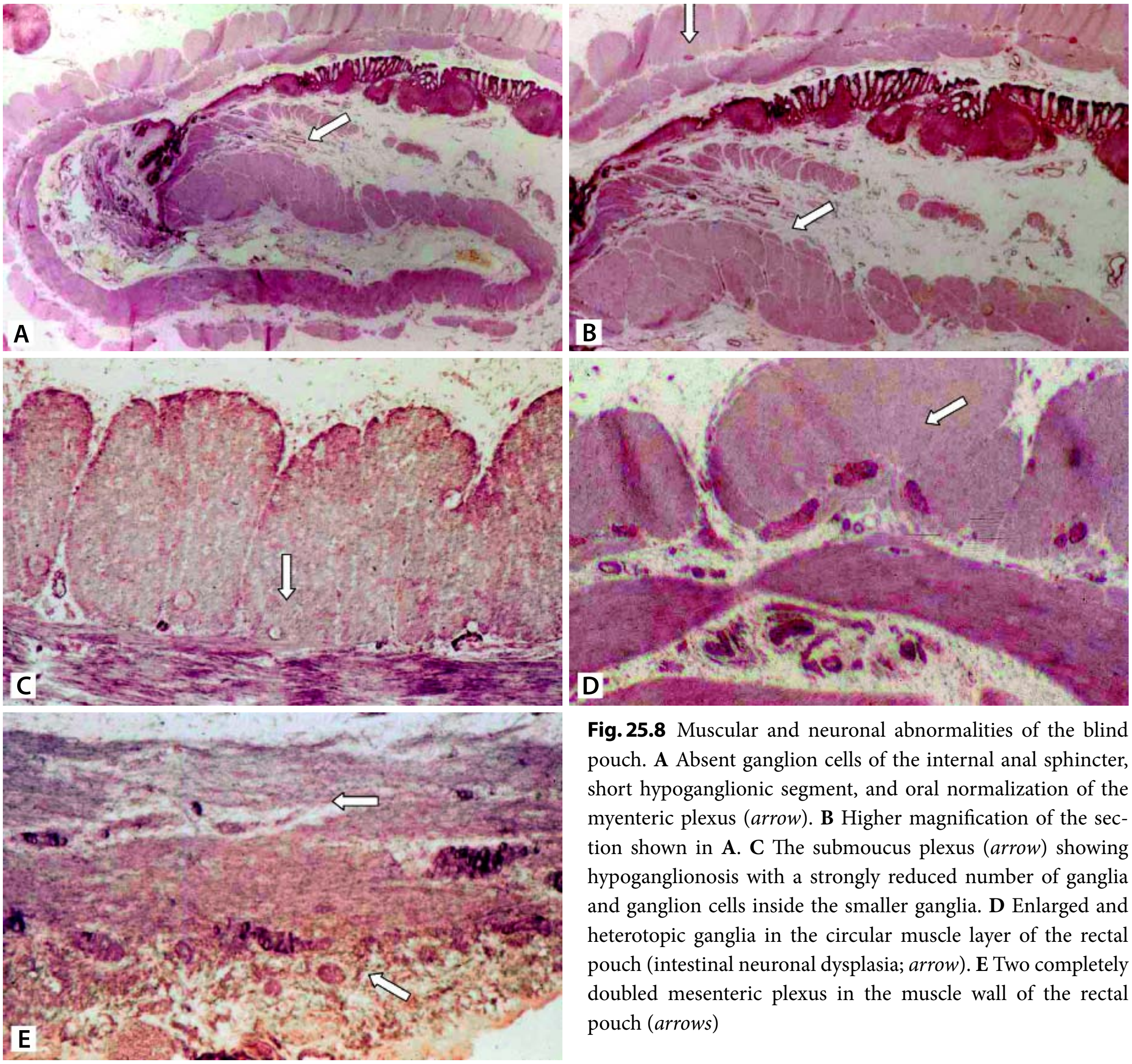

Fig. 25.8 Muscular and neuronal abnormalities of the blind pouch. **A** Absent ganglion cells of the internal anal sphincter, short hypoganglionic segment, and oral normalization of the myenteric plexus (*arrow*). **B** Higher magnification of the section shown in **A**. **C** The submoucus plexus (*arrow*) showing hypoganglionosis with a strongly reduced number of ganglia and ganglion cells inside the smaller ganglia. **D** Enlarged and heterotopic ganglia in the circular muscle layer of the rectal pouch (intestinal neuronal dysplasia; *arrow*). **E** Two completely doubled mesenteric plexus in the muscle wall of the rectal pouch (*arrows*)

the proximal segment. These data suggest that anomalies of the intramural nerve plexus are less important for chronic constipation than is malformation or damage to the extramural cholinergic nerve fibers. In addition, biopsy samples of the rectal wall in 25 patients with severe postoperative constipation investigated by Holschneider et al. revealed no correlation between histomorphological findings and the clinical symptoms. Only four out of nine patients with aganglionosis and two out of eight children with hypoganglionosis suffered from severe chronic constipation. However, all patients without histological abnormalities showed normal bowel evacuations [7,13]. Therefore, aganglionosis, hypoganglionosis, and IND of the most distal part of the rectal pouch and in the fistula may represent a normal situation.

25.6 Pathophysiology of Continence and Defecation in ARM

The pathophysiological conditions arising from the aforementioned pathoanatomical findings are summarized in Table 25.3.

25.6.1 Rectum

The rectum in children with ARM is usually dilated and does not shrink very much after the establishment of a colostomy. Its voiding function is reduced due to the impairment of parasympathetic nerve fibers and, in a few cases, of the intramural nerve plexus. A dilated bowel segment is not usually able to

Table 25.3 Pathologic conditions for stool continence in anorectal malformations. *ARM* Anorectal malformations, *AG* Aganglionosis, *HG* hypoganglionosis, *IND* intestinal neuronal dysplasia

Anatomical site	Pathological condition
Rectum	Blind-ending, dilated rectal pouch Malformation of extramural nerve supply -tethered cord, spinal dysraphism, caudal regression, over-distension of cholinergic nerve fibers, damage, and atypical course of the erigentes nerves Malformations of intramural nerve plexus and bowel wall structures -AG, HG, IND, desmosis, absence of interstitial cells of Cajal Reduced fixation of the rectum in the small pelvis after pull-through: disposition for rectal prolapse
Pelvic floor	Hypoplastic striated muscle complex, frequently not in direct contact with external sphincter muscle fibers and much better developed close to the sacrum than at the side of the anal dimple No clearly visible anorectal angle
Internal anal sphincter	Reduced number and weaker smooth muscle fibers localized around the fistula and not always at the deepest point of the blind pouch, with the exception of ARM without fistula
External anal sphincter	Hypoplastic and sometimes malpositioned muscle fibers Hemorrhoidal plexus on a higher level No longitudinal muscle fibers of the rectal muscle coat running through the external anal sphincter into the anal skin: disposition for mucosal prolapse No anoderm: reduced sensibility

contract. The slow waves are disturbed and the evocations of spike potentials are reduced [15]. This is a well-known phenomenon in other diseases like ileus, segmental dilatation of the small bowel loops, and jejunal atresia. However, rectal dilatation in ARM does not usually need tapering like jejunal obstructions. Partial resection might be necessary only in a very few cases of inert rectum that are resistant to washouts. However, one always has to keep in mind that patients with ARM need a certain degree of constipation to become clean with washouts.

In 1997, Rintala [13] demonstrated that total colonic transit is significantly prolonged in patients with ARM and even more prolonged in high types (median 24% prolonged transit) than in low types and healthy individuals (median 10% prolonged transit). However, Nagashami et al. in 1992 [28] found no difference in either contractile or myoelectric activity (spike bursts and slow waves) of the rectum between the two groups. Heikenen et al. [29] observed high-amplitude propagating contractions in severely constipated children with ARM soiling after repair. The same phenomenon is found in children with myelomeningocele [13,24,30]. Thus, overflow incontinence in ARM seems mainly to be a motility disturbance of the rectum and not a problem of the anorectal sphincters. This can also be concluded by comparing the functional situation in ARM with that of patients with Hirschsprung's disease. In Rehbein's procedure it is permitted to leave 4–5 cm of aganglionic anorectum in situ. However, chronic constipation occurred postoperatively in only 7.9% of these patients [31]. The frequency of postoperative chronic constipation in children after megacolon repair is lower than in children after PSARP for imperforate anus. In addition, hypoganglionosis of the lower most part of the blind pouch represents the normal ganglion cell distribution in the internal anal sphincter in healthy individuals.

Motility disturbance could theoretically also result from malformations of the parasympathetic nerve supply to the rectum. Christensen pointed out that the large intestine receives its extrinsic nerve supply through the vagus nerves, from the pelvic nerves and from the mesenteric nerves [16]. The vagus nerves provide a parasympathetic innervation to the whole gastrointestinal tract and to the rostral end of the large intestine. The pelvic nerves also distribute cholinergic fibers, the sacral component of the craniosacral outflow, to the whole of the large intestine. They form the pelvic plexus from which colonic nerve branches pass to the large intestine. The mesenteric-adrenergic nerves emerge from the three prevertebral ganglia, which send branches along the three arteries to the gut. The vagal branches extend no further than about the middle of the transverse colon. The pelvic nerves distribute nerve fibers to the remainder of the large intestine. Branches of these colonic nerves extend rostrally in the myenteric plexus as far as the transverse colon. These are called ascending nerves of the colon. They may well overlap to some degree with that of the vagus nerve and vary widely.

In children with ARM this descending, ascending, and intramural nerve supply could be disturbed and its stimulating influence diminished leading to the varying degree of constipation observed in these children. This aspect is supported by studies of Mandhan et al. [32] showing that the imunoreactivity of neuron-specific enolase, vasoactive intestinal peptide, NSE, VIP, and nuclear protein SP-100 is markedly reduced in the rectum and fistulous tract of high ARM and slightly reduced in low ARM compared with controls. Intramural nerves immunostained with VIP and SP-100 antisera are decreased in both types of ARM, indicating that both inhibitory and excitatory motor neural elements are affected. This may also explain the distal colonic dysmotility seen postoperatively in both high and low ARM.

However, the degree of rectal dysmotility varies widely. Kayaba et al. [33], performing fecoflowmetric studies in 16 patients after repair of ARM, demonstrated that 7 out of 16 children exhibited periodical contractions of the rectum synchronized with relaxations of the anal canal during saline infusion, as did controls, and had significantly better clinical scores than other patients. Only two children with severe chronic constipation lacked rectal contraction. The fecoflow parameters, such as the maximum flow, average flow, and tolerable volume of saline infused into the rectum, were significantly lower in the patients with poor clinical scores than those of controls. The maximal squeeze pressure and resting anal pressure were not significantly different between the patients and controls. This also indicates that anorectal dysfunction in patients with ARM is more a problem of dysmotility of the rectum than of anal sphincter incompetence. However, a low anorectal pressure barrier might be an additional problem leading to overflow incontinence. This is supported by the studies of Heikenen et al. [29]; they performed motility studies in 13 children with repaired ARM and fecal soiling and found high-amplitude propagating contractions with an average of 80% propagation into the neorectum. Internal sphincter resting pressure was low in six out of ten patients. Internal sphincter relaxation was also present in six out of ten children. Only one out of five patients was able to cooperate and therefore capable of generating a normal squeeze pressure. The authors conclude that fecal soiling in patients with repaired ARM is a multifactorial problem including propagation of excessive numbers of high-amplitude propagating contractions into the neorectum as well as internal sphincter dysfunction. However, high-amplitude contractions are a typical electromanometric sign of chronic constipation [34]. In addition, in 20% of Heikenen's patients the rectum was "lazy", showing no motility. These patients probably needed rectal resection [29].

25.6.2 Pelvic Floor

From the anatomical sections by Stephens (see CD) it becomes obvious that the muscle complex structures vary individually in a wide range that is not only dependent upon the level of the malformation. In one and the same type of ARM in one patient the muscle complex can be well developed, while in an other patient it is hypoplastic. In high lesions there is not always a continuity of well-developed muscle fibers from the upper parts of the pelvic floor down to the external anal sphincter fibers and there might be instead some very tiny, hypoplastic muscle fibers around the urethra simulating a gap filled with fatty ischiorectal tissue. The striated muscle fibers covering the dorsal wall of the urethra are usually much better developed proximal to the coccyx than in the more caudal region below the rectal pouch. The development of the pelvic floor muscles seems to correspond with the growth of the rectal pouch inside the muscle complex structures.

25.6.3 Internal Anal Sphincter

Lambrecht and Lierse [35] demonstrated a normal internal anal sphincter surrounding the proximal part of the fistula in 33 neonatal piglets with anal atresia [30]. The position of the internal sphincter depended on the localization of the fistula orifice into the rectal pouch. This differed greatly. The form and thickness of the internal sphincter also varied to a large degree. Sometimes the muscle had the form of a tube, or an acute-angled funnel as in healthy piglets. However, mostly the internal sphincter was spread out wide and had the form of a disc or a flat dish. In addition, the rectal pouches in the region of the internal anal sphincter as well as the fistula are hypoganglionic.

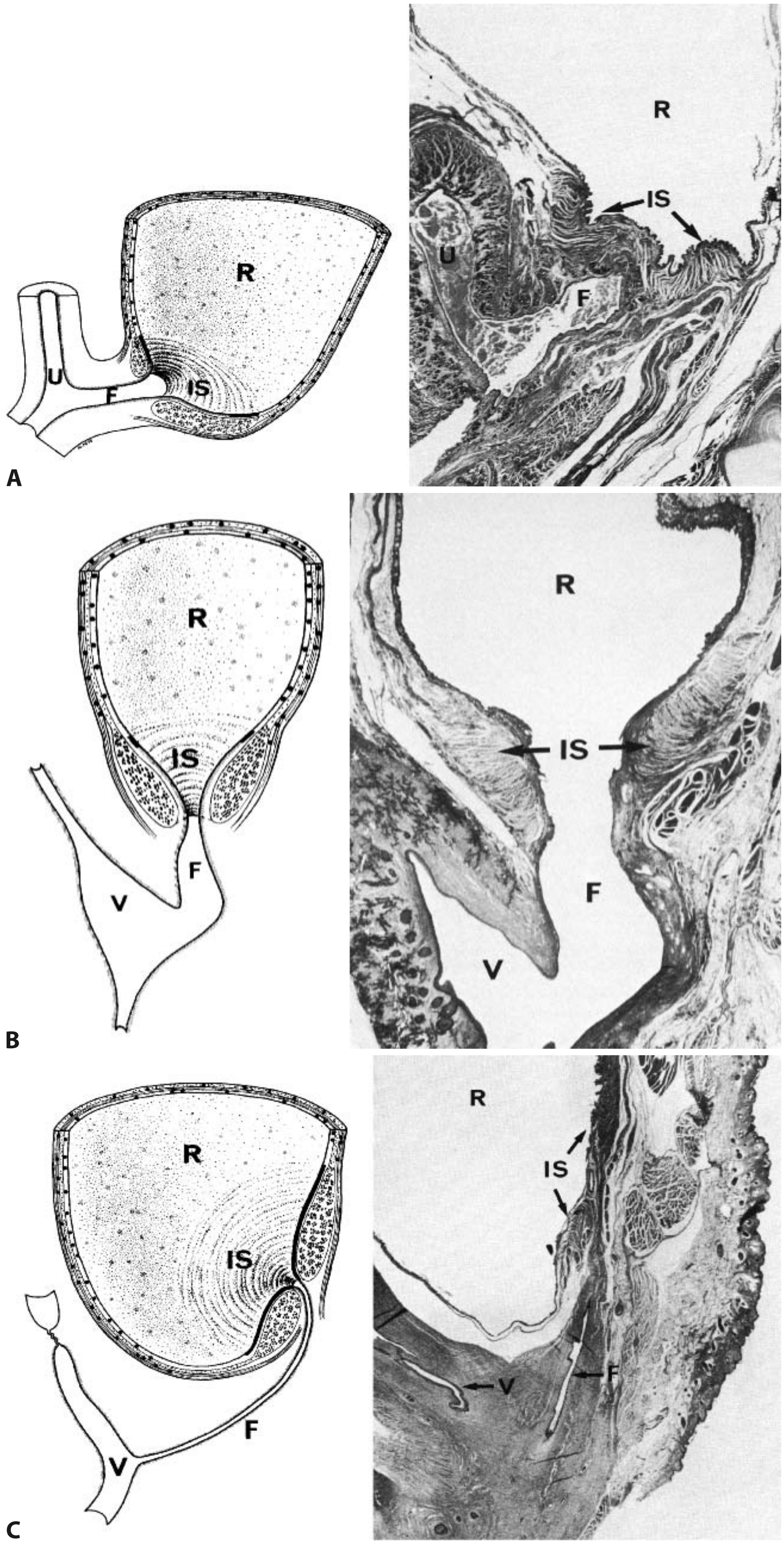

Fig. 25.9 **A–C** Histological sections through the internal anal sphincter fibers in the rectal pouch of piglets with imperforate anus (*right*). Schematic drawings of the histological sections are shown on the *left* (from Lambrecht and Lierse 1987 [35] with permission from the publishers). **A** Female piglet with high ARM and rectovaginal fistula. Note the funnel-shaped internal anal sphincter. **B** Male piglet with high ARM and rectourethral fistula. Note the disc-type development of internal anal sphincter. **C** Female piglet with high ARM and rectovaginal fistula. Note the wide spreading out of the smooth muscle fibers. *R* Rectum, *V* vagina, *F* fistula, *IS* internal anal sphincter, *U* urethra

The proximal fistula region is lined by transitional epithelium, and it contains anal glands. The authors concluded, therefore, that the fistula should be regarded as an ectopic anal canal (Fig. 25.9).

The questions as to whether these smooth muscle fibers should be preserved during the operation and whether they play an important role for postoperative continence remains open. In 1985, Iwai reported that internal sphincter relaxation correlated well to the Kelly score of continence [36]. However, in 1992 Peña and Hedlund reported that the correlation between electromanometric findings and clinical results is incomplete [37]. In contrast, Rintala observed good continence in children with positive internal anal sphincter relaxation and a high anorectal pressure profile [8,9,38]. These findings are in contrast to follow-up studies of Chen-Lung in 1998 [39], who reported that internal sphincter relaxation was not correlated with the surgical procedure. However, if the internal sphincter relaxation was positive and a high anorectal pressure profile was established, the development of constipation was six times more likely to occur. By studying 24 infants less than 3 years who had ARM, Sangkhathat et al. came to the conclusion that the rectoanal inhibitory reflex plays a crucial role in emptying function after anoplasty in such children [40]. Internal sphincter relaxation was present in 93% of cases without constipation and 12% of the patients with constipation. One patient who had clinical conversion from constipation to a good result also showed a positive conversion of the internal sphincter relaxation reflex. This means that these functions should be preserved during reconstruction if possible. This corresponds with Lambrecht's suggestions.

The deepest point of the rectal pouch, however, does not always correspond with the rectal origin of the fistula or the confluence to the urethra. Only in patients without fistula is the lowest point of the rectal pouch thickened by a cluster of smooth muscle cells that could easily be implanted in the perineum inside the external sphincter fibers. In many patients with fistulae, however, the smooth muscle fibers surrounding the fistulae are on a higher level than the deepest point of the pouch and can hardly be brought to the midline to be used as additional sphincter support.

25.6.4 External Anal Sphincter

The external anal sphincter fibers develop from the anal tubercules. The fibers are not circularly arranged, which may impair concentric contraction. They can easily be identified by electrical stimulation and are usually situated in a line between the ischial tuberosities. However, they can also be antepositioned. There are no longitudinal muscle fibers penetrating the smooth muscle fibers of an internal anal sphincter, as in normal individuals, and inserting into the skin. Therefore, there is no corrugator ani muscle for the so-called fine continence helping to avoid staining or smearing, and fixing the mucosa to the surrounding tissue, which avoids mucosal prolapse. Rectal prolapse with a protrusion of all muscle layers of the pulled-down rectum, on the other hand, is unusual if the lateral wings continue to fix the neorectum inside the pelvis or the colon is fixed to the presacral fascia. The hemorrhoidal plexus is developed on a higher level and therefore not supporting fine continence by obstructing the lower anal canal by filling its vessels.

Abnormalities of innervation of the external sphincter are another important factor affecting postoperative anorectal function. Yuan et al. investigated 45 patients with ARM by neural electrophysiologic methods [41]. They found the latencies of the pudendoanal reflex, the spinoanal response, and the conduction times of sacral spinal centers are significantly prolonged in patients with ARM. There was a significant difference between the rectourethral fistula group and the vestibular fistula group, as well as the low-type deformity group on the other side. There was significant negative correlation between the latencies and clinical scores. Although pudendoanal reflex latency was longer in patients who had PSARP than those who had abdominoperineal pull-through procedures, the difference was not significant. The neuronal lesions resemble those of lumbosacral deformity but vary for each type of anal lesion. Nevertheless, they are important for clinical outcome.

Abbaso et al. came to the same result [42]. Five out of eight children showed well-preserved electrical activity of the external sphincter muscle in postoperative electromyographic studies. In one patient there was no activity and in only two patients the activity was normal. Six out of eight children showed a normally localized anus.

This is contradictory to studies in rats performed by Yuan et al. in 2003 showing that the innervation to the sphincter mechanism is defective and should be regarded as a primary anomaly that coexists with the alimentary tract anomaly in ARM during fetal development [43]. However, in the animal model of ethylenethiourea (ETU)-treated rats, the development of the muscle complex is decreased, especially in type II deformities showing only a muscular cord without any intestinal tube [44]. ETU-induced malformations are more complex, showing additional neural tube defects in 24 out of 29 animals studied by Qi et al. [45]. In addition, in Bai's ETU experiments the only types of ARM induced were the rectourethral fistula and common cloaca [46]. According to Mortell et al. adriamycin induces notochord hypertrophy, resulting in a significant increased volume of notochord during the critical phase of development [47,48]. This may interfere with organogenesis, resulting in vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb (VACTERL) association. ETU-induced ARM cannot, therefore, be compared with human alterations.

25.6.5 Anoderm (Skin of the Anal Canal)

In many cases the anoderm is well developed and should be preserved or reconstructed, for example by using Nixon's plasty or retraction at the neoanus at the end of the procedure after having sutured the fistula under tension to the perineal skin. This technique could also help to increase sensibility and to improve the rectosphincteric reflex.

Fecal incontinence in patients having undergone PSARP repair for ARM improves at adolescence, as constipation disappears. Continence was ameliorated in 14 out of 22 children followed up by Rintala et al. after puberty [49]. Constipation improved in 13 out of 15 patients and only 2 needed a regular enema regimen after puberty. This improvement with time is probably related to a reinforced sphincter function and an increasing use of gluteal and pelvic floor muscles, and is a manifestation of the adaptation and adjustments made by the patient himself to achieve a socially acceptable status.

However, we believe that future continence behavior can be predicted within the 1st year of life. Regular bowel movements with evidence of straining and observation of signs suggestive of imminent defecation by the mother without staining of the napkins suggests a good outcome.

25.7 Conclusion

The reconstruction of anal sphincters is much improved by preservation of the blind pouch, but rectal motility remains disturbed. It has become apparent that there should be an equilibrium between the propagating forces of propulsive waves from the normal colon down to the rectum and the stool-retaining forces of the anorectal pressure barrier. However, this state of balance is frequently disturbed by reduced motility of the blind pouch on one side and lowered anorectal resting and squeeze pressures on the other. Therefore, the most important goal of repair of ARM should be to preserve as many anatomical and physiological continence-improving structures as possible and to avoid any damage to the nerve supply of the blind pouch and the anorectal sphincters during surgery. In addition, the dysmotility of the blind-ending rectum should be used as a help to slow down the propulsive waves of the normal descending colon, otherwise these movements could overwhelm the weakened sphincter barrier leading to incontinence. However, storage of fecal material in a dysfunctioning rectum with overflow incontinence should be avoided.

These aforementioned anatomical and pathophysiological considerations yield the following conclusions for clinical purposes:

1. The introduction of PSARP has been of great benefit to children with ARM.
2. Chronic constipation with overflow incontinence is more frequent after PSARP than it is after abdominoperineal pull-through procedures, which more frequently lead to stool incontinence.
3. The rectal pouch cecum should always be preserved.
4. It is of greater importance, however, to mobilize the pouch as little as possible and keep directly on the rectal wall.
5. If possible, the fistula and the surrounding smooth muscle fibers, representing an ectopic anus, should be implanted into the perineum.
6. In constipated children one should always be aware of associated malformations of the intramural plexus. In addition, malformations of the smooth and striated muscle fibers and Cajal's cells could be present. In cases of severe chronic constipation, suction, or even full-thickness biopsy samples should be taken.
7. Postoperative electromanometric studies are of great importance for the differentiation between motility disturbances of the rectum and incompetence of the anorectal sphincter.
8. There is no place for ongoing postoperative sphincter dilatations. After a few weeks the anus should grow elastically by itself.

References

1. Stephens FD, Smith ED (1971) Anorectal Malformations in Children. Year book Medical Publishers, Chicago
2. Stephens FD, Smith ED, Paul NW (1988) Anorectal Malformations in Children: Update 1988. Alan R. Liss, New York, March of Dimes Birth Defect Foundation, Birth Defect Original Series Vol 24:4
3. Holschneider AM, Koebke J, Meier-Ruge W, Land N, Jesch NK (2001) Pathophysiology of chronic constipation on anorectal malformations. Long-term results and preliminary anatomical investigations. Eur J Pediatr Surg 11:305–310
4. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
5. Holschneider AM, Jesch NK, Stragholz E, Pfrommer W (2002) Surgical methods for anorectal malformations from Rehbein to Peña – critical assessment of score systems and proposal for a new classification. Eur J Pediatr Surg 12:73–82
6. Langemeijer RATM, Mo1enaar JG (1991) Continence after posterior sagittal anorectop1asty. J Pediatr Surg 26:57–90
7. Rintala R, Mildh L, Lindahl H (1992) Fecal continence and quality of life in adult patients with an operated low anorectal malformation. J Pediatr Surg 27:902–905
8. Rintala R, Lindahl H (1995) Internal sphincter saving PSARP for high and intermediate anorectal malformations: technical considerations. Pediatr Surg Int 10:345–349
9. Rintala RJ, Lindahl HO (1995) Is normal bowel function possible after repair of intermediate and high anorectal malformations. J Pediatr Surg 30:491–499
10. Hedlund H, Peña A, Rodriguez G, et al (1992) Long term anorectal function in imperforate anus treated by posterior saggita1 anorectoplasty: manometric investigation. J Pediatr Surg 27:906–909
11. Bliss DP, Tapper D, Sanderson JM, Schaller RT, Hatch EJ, Morgan A, Hall DG, Sawin RS:(1996) Does posterior sagittal anorectoplasty in patients with high imperforate anus provide superior fecal continence? J Pediatr Surg 31:26–32
12. Chau-Jing C (1999) The treatment of imperforate anus: experience with 108 patients. J Pediatr Surg 34:1728–1732
13. Rintala RJ, Lindahl HG, Rasanen M (1997) Do children with repaired low anorectal malformations have normal bowel function. J Pediatr Surg 32:823–826
14. Holschneider AM (1976) The problem of anorectal continence. Progress Pediatr Surg 9:85–97
15. Holschneider AM (1983) Elektromanometrie des Enddarms. Diagnostik und Therapie der Inkontinenz und der chronischen Obstipation. 2.Auflage. Urban und Schwarzenberg , München, Wien, Baltimore,
16. Christensen J (2000) Normal colonic motor function and relevant structure. In: Holschneider AM, Puri P (eds) Hirschsprung's Disease and Allied Disorders. Harwood Academic publishers, Australia p 89
17. Huber A, Hochstetter AHC van, Allgöwer M (1993) Transsphinktere Rektumchirurgie. Topographische Anatomie und Operationstechnik. Springer, Berlin, Heidelberg, New York, Tokyo
18. Peña A, Amroch D, Baeza C, et al (1993) The effect of the posterior sagittal approach on rectal function (experimental study). J Pediatr Surg 28:773–778
19. Kelly JH (1969) The radiographic anatomy of the normal and abnormal neonatal pelvis. J Pediatr Surg 4:432–444
20. Kelly JH (1969) Cine radiography in anorectal malformations. J Pediatr Surg 4:538–546
21. Davies MRQ (1997) Anatomy of the nerve supply of the rectum, bladder and internal genitalia in anorectal dysgenesis in the male. J Pediatr Surg 32:536–541
22. Lanz T van, Wachsmuth W (1984) Praktische Anatomie. Zweiter Bad, Teil 8A, Becken Springer Verlag, Berlin, Heidelberg, New York, Tokyo p 245
23. Pickard LR, Santoro S, Wyllie RG, Haller JA (1982) Histochemical studies of experimental fetal intestinal obstruction. J Pediatr Surg 16:256–260
24. Holschneider AM, Pfrommer W, Gerresheim B (1994) Results in the treatment of anorectal malformations with special regard to the histology of the rectal pouch. Eur J Pediatr Surg 4:303–309
25. Holschneider AM, Ure BM, Pfrommer W, Meier-Ruge WM (1996) Innervation patterns of the rectal pouch and fistula in anorectal malformations: a preliminary report. J Pediatr Surg 31:357–362
26. Masumoto K, Suita S, Nada O, Taguchi T, Guo R (1999) Abnormalities of enteric neurons, intestinal pacemaker cells, and smooth muscle in human intestinal atresia. J Pediatr Surg 34 1463–1468
27. Martucciello G, Mazzola C, Favre A, Negri F, Bertagnon M, et al (1999) Preoperative enzymo-histochemical diagnosis of dysganglionosis associated with anorectal malformations (ARM) with rectovestibular and recto-perineal fistula. Eur J Pediatr Surg 9:96–100
28. Nagashami M, Iwai N, Yanagiihra J, Shimotake G (1992) Motility and sensation of the rectosigmoid and the rectum in patients with anorectal malformations. J Pediatr Surg 27:1273–1277
29. Heikenen JB, Werlin SL, Di Lorenzo C, Hyman PE, Cocjin J, Flores AF, Reddy SN (1999) Colonic motility in children with repaired imperforate anus. Dig Dis Sci 44:1288–1292
30. Pigeon N, Leroi AM, Devroede G, Watier A, Denis P, Weber J, Arhan P (1997) Colonic transit time in patients with myelomeningocele. Neurogastroenterol Motil 9:63–70
31. Holschneider AM (1982) Hirschsprung's Disease. Hippokrates Verlag, Stuttgart, Thieme-Stratton, New York

32. Mandhan P, Qi BQ, Beasley SW (2005) Aberrations of the intrinsic innervation of the anorectum in fetal rats with anorectal malformations. J Pediatr Surg 40:397–402
33. Kayaba H, Hebiguchi T, Yoshino H, Mizuno M, Yamada M, Chihara J, Kato Tetsuo (2002) Evaluation of anorectal functions of children with anorectal malformations using fecoflowmetry. J Pediatr Surg 37:623–628
34. Holschneider AM, Puri P (2000) Hirschsprung's Disease and Allied Disorders. Harwood Academic Publishers, Australia
35. Lambrecht W, Lierse W (1987) The internal sphincter in anorectal malformations: morphologic investigations in neonatal pigs. J Pediatr Surg 12:1160–1168
36. Iwai N, Kaneda H, Tamiguchi T, et al (1985) Postoperative continence assessed by electromyography of the external sphincter in anorectal malformations. Z Kinderchir 40:87–90
37. Hedlund H, Peña A, Rodriguez G, et al (1992) Long term anorectal function in imperforate anus treated by posterior sagitta1 anorectoplasty: manometric investigation. J Pediatr Surg 27:906–909
38. Rintala R, Lindahl H (1995) Secondary PSARP for anorectal malformations: a long term follow-up extending beyond childhood. Pediatr Surg Int 10:414–417
39. Chen-Lung L, Chiu-Chiang C (1998) The rectoanal relaxation reflex and continence in repaired anorectal malformations with and without an internal sphincter saving procedure J Pediatr Surg 31:630–633
40. Sangkhathat S, Patrapinyokul S, Osatakul N (2004) Crucial role of rectoanal inhibitory reflex in emptying function after anoplasty in infants with anorectal malformations. Asian J Surg 27:125–129
41. Yuan Z, Bai Y, Zhang Z, Ji S, Li Z, Wang W (2000) Neural electrophysiological studies on the external anal sphincter in children with anorectal malformation. J Pediatr Surg 35:1052–1057
42. Abbaso Iu, Tansu Salman F, Baslo B, Ier S, Gün I (2004) Electromyographic studies on the external anal sphincter in children with operated anorectal malformations. Eur J Pediatr Surg 14:103–107
43. Yuan ZW, Lui VC, Tam PK (2003) Deficient motor innervation of the sphincter mechanism in fetal rats with anorectal malformation: a quantitative study by fluorogold retrograde tracing. J Pediatr Surg 38:1383–1388
44. Arana J, Villanueva A, Guarch R, Aldazabal P, Barriola M (2001) Anorectal atresia. An experimental model in the rat. Eur J Pediatr Surg 11:192–195
45. Qi BQ, Beasley SW, Francis FA (2003) Evidence that the notochord may be pivotal in the development of sacral and anorectal malformations. J Pediatr Surg 9:1310–1316
46. Bai Y, Hui Ch, Zheng WY, WeiLin W (2004) Normal and abnormal embryonic development of the anorectum in rats. J Pediatr Surg 39:587–590
47. Mortell A, O'Donnell AM, Giles S, Bannigan J, Puri P (2004) Adriamycin induces notochord hypertrophy with conservation of sonic hedgehog expression in abnormal ectopic notochord in the adriamycin rat model. J Pediatr Surg 39:859–863
48. Mortell A, Gillick J, Giles S, Bannigan J, Puri P (2005) Notable sequential alterations in notochord volume during development in the adriamycin rat model. J Pediatr Surg 40:403–406
49. Rintala R, Lindahl H (2001) Fecal continence in patients having undergone posterior sagittal anorectoplasty procedure for a high anorectal malformation improves at adolescence, as constipation disappears. J Pediatr Surg 36:1218–1221

26 Postoperative Electromanometric, Myographic, and Anal Endosonographic Evaluations

Naomi Iwai, Eiichi Deguchi, Takashi Shimotake, and Osamu Kimura

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The goal of treatment for anorectal malformations (ARM) is to achieve anal continence, and various attempts have been made to obtain objective assessments for it. These objective measures give us accurate information about postoperative sphincter function or sphincter distribution, and provide us with further strategies for postoperative treatments.

We have two main objective assessments: physiological examinations and imaging studies. In this chapter we describe our experience with postoperative electromanometric, electromyographic, and anal endosonographic evaluations, in addition to the use of biofeedback therapy.

26.1 Electromanometric Evaluation

For the complete evaluation of postoperative continence, anorectal manometric studies have been performed on patients with ARM [1,2]. We also investigated whether there was a correlation between clinical assessment and manometric assessment [3].

26.1.1 Manometry Study

Manometry was done without anesthesia, except in restless children, who required mild sedation at the time of examination. The probe was filled with water before the examination, but was not perfused during the test. The anorectal pressure profile was first recorded in centimeters by withdrawing the probe that was introduced 8 cm above the mucocutaneous line into the rectum. The presence or absence of an anorectal reflex was determined by distending the balloon in the rectum.

26.1.2 Results of Anorectal Manometry

Patients with a good clinical result after staged abdominoperineal rectoplasty or perineoplasty exhibited the same anorectal pressure profile as normal subjects, with a high-pressure zone in the anal canal (Fig. 26.1). They also had an adequate anorectal pressure difference that was not significantly different from that of normal subjects. On the other hand, patients with a poor clinical result showed a slight radial change in the anorectal pressure profile and did not have such a high-pressure zone as was found in normal subjects (Fig. 26.2). The anorectal pressure difference was as low as 3 cmH_2O. Most of the patients with a good clinical result, regardless of the type anomaly, exhibited an anorectal reflex. In high anomalies, however, some patients with a good clinical result did not necessarily show the reflex.

Manometric investigations showed that good clinical results after surgery were associated with a normal function of the anorectum. The anorectal pressure profile, observed in all patients with adequate continence, characteristically had a marked high-pressure zone, as did normal subjects. Thus, the presence of normal anal pressure at rest as well as an adequate anorectal pressure difference was found to correlate well with continence after surgery for ARM. However, the anorectal reflex in high anomalies did not necessarily correlate well with continence. Accordingly, it seems that the reflex is not essential to achieve continence, at least in patients with high anomalies. This might be explained as follows. In a high anomaly, only a mechanical resistance remains without sensi-

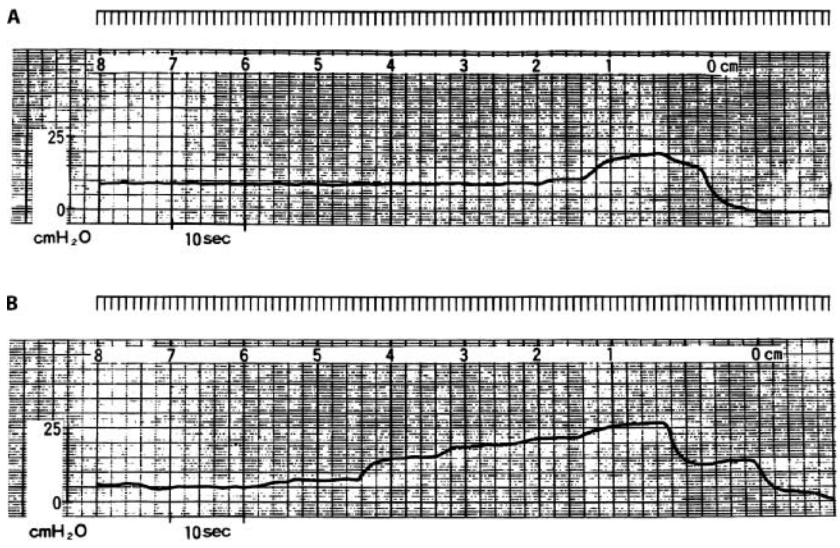

Fig. 26.1 Anorectal pressure profile in patients with good results. **A** Staged abdominoperineal rectoplasty. **B** Perineoplasty. The same anorectal pressure profile with a high-pressure zone was observed

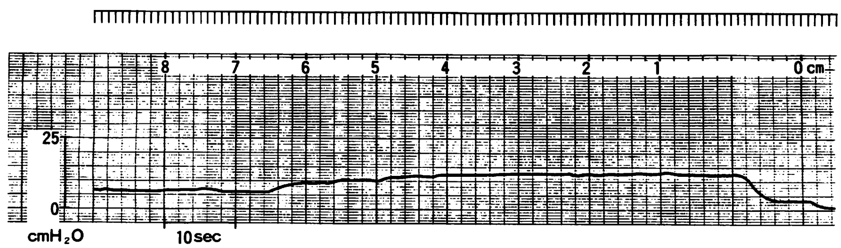

Fig. 26.2 Anorectal pressure profile in a patient with a poor result after staged abdominoperineal rectoplasty. A slight radial pressure change in the anal canal was found without a high-pressure zone

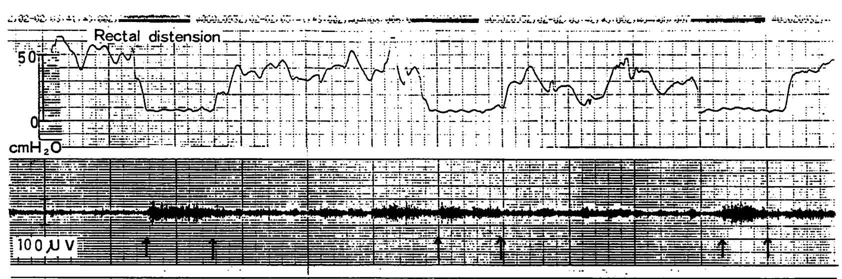

Fig. 26.3 Electromyographic recording of the external sphincter (EAS) during rectal distension. In response to each rectal distension, relaxation of the anal canal pressure and contraction of the external sphincter were observed (*arrows*)

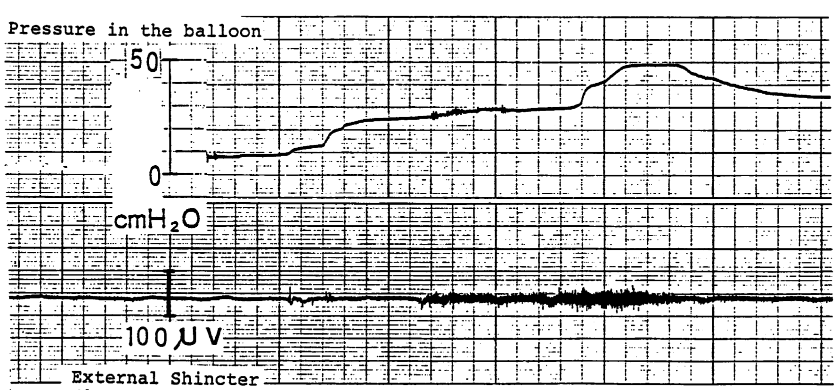

Fig. 26.4 Electromyographic recording of the EAS during further rectal filling. The further rectal filling elicited renewed contraction of the external sphincter

tive receptors in the mucosa, which is concerned with initiating the anorectal reflex. Therefore, normal anal resting pressure and an adequate anorectal pressure difference in a high anomaly are apparently more important factors relating to continence after reconstructive surgery for ARM.

26.2 Electromyography

In the treatment of ARM it seems that the function of the external sphincter has not been emphasized sufficiently. The external sphincter muscle provides fine control, especially at the time of a "sense of urgency". The external sphincter function was assessed by electromyography (EMG) and voluntary contraction pressure.

26.2.1 EMG study

The EMG was recorded by two surface electrodes placed just outside the anal orifice, as reported previously [4]. The patient was grounded with a similar electrode. The time constant was 0.03 s. The patients were examined awake, without sedation, and in a supine position. The external sphincter electrogram at rest was recorded first. When the rectum was transiently distended by a balloon, contraction of the external sphincter was observed. This response was defined as the presence of an inflation reflex (Fig. 26.3). The rectal balloon was further inflated to a maximum tolerable level, and the electrical activity was observed during rectal filling (Fig. 26.4). In cooperative patients, the presence or absence of phasic activity during voluntary anal contraction pressures were measured at 2 cm and at 1 cm from the anal verge.

26.2.2 Results of the EMG Study

In patients with low anomalies, the inflation reflex was observed, and electrical activity during further rectal filling was increased. Phasic activity was present in all of them. On the other hand, in patients with intermediate or high anomalies, most did not show an inflation reflex, and electrical activity was stationary in spite of further rectal filling. Phasic activity was present in all.

Adequate electrical activity of the external sphincter at rest was observed in patients with low and intermediate anomalies. On the other hand, tonic activity was observed less often in patients with high anomalies. These results suggest that patients with high anomalies have a congenital functional problem of the external sphincter muscle. Molander and Frenckner [5] showed that the presence of an inflation reflex correlated well with the development of voluntary anal continence. In our study the inflation reflex was much more common in normal subjects and in patients with low anomalies. From the point of view of the inflation reflex, the function of the external sphincter is more frequently disturbed in patients with high anomalies. Electrical activity during further rectal filling is an index of external sphincter function in patients with high anomalies, as is tonic activity or the inflation reflex. The results of phasic activity indicate that although patients with high anomalies may have congenitally rudimentary external sphincter muscles, they may still be able to improve their external sphincter function. Therefore, patients with high anomalies may achieve compensatory voluntary continence of defecation if the external sphincter muscle is developed by voluntary bowel training such as biofeedback training (Fig. 26.5). These results have led us to devise a new computer-assisted biofeedback therapy in patients who have fecal incontinence after surgery for ARM [6,7].

26.2.3 Anal Endosonography

Information on sphincter morphology cannot be obtained by physiological examination. Accordingly, anal endosonography has come into use for the morphological evaluation of the external anal sphincter (EAS) and the internal anal sphincter (IAS) [8,9], and we have applied this method to assess ultrasonically the structures around the anal canal in ten patients with high-type anomalies and five patients with intermediate anomalies [10].

An ultrasonographic scanner (ASU-61; Aloka, Tokyo, Japan) was used with a 7.5-MHz rotating endoprobe. The endoprobe is protected by a hard sonolucent plastic cone with a diameter of less than 2 cm, designed to fit within the anal canal. The presence or absence of a well-defined uniform hypoechoic band that corresponds to the IAS was examined. Whether the hypoechoic band was well or moderately defined from the surrounding tissues and whether continuity of the band was interrupted were also examined. In addition, the appearance of the layer and its continuity were checked.

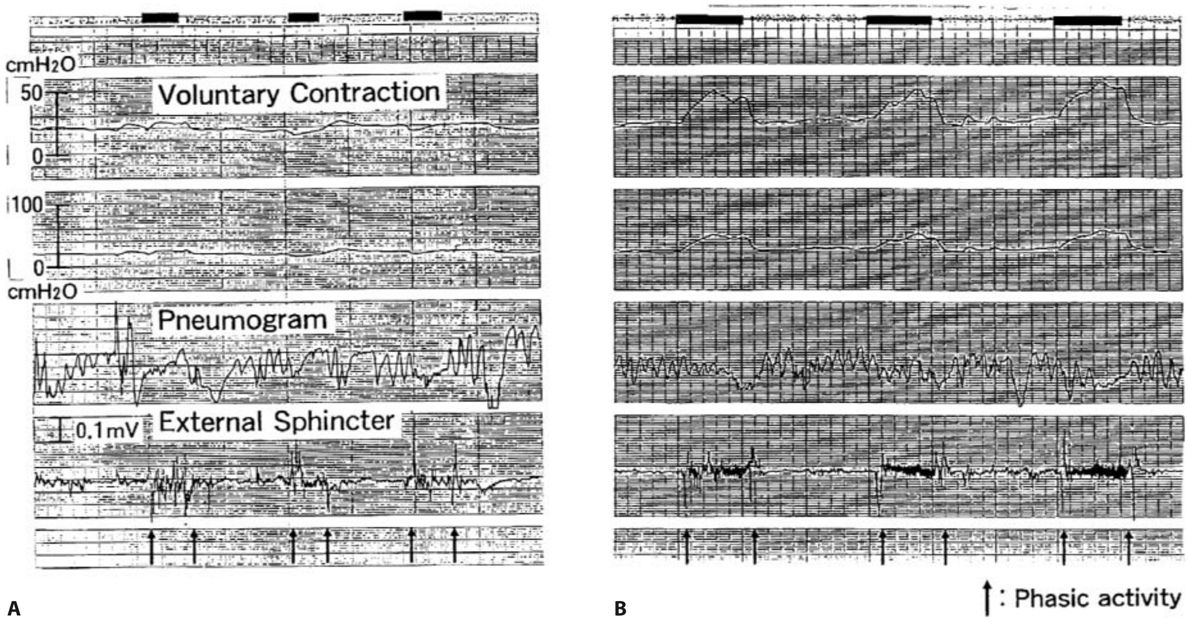

Fig. 26.5 Voluntary contraction pressures and phasic activities of the external sphincter before (**A**) and after (**B**) biofeedback therapy. Voluntary contraction pressures and electrical activities of the phasic activities are increased after therapy

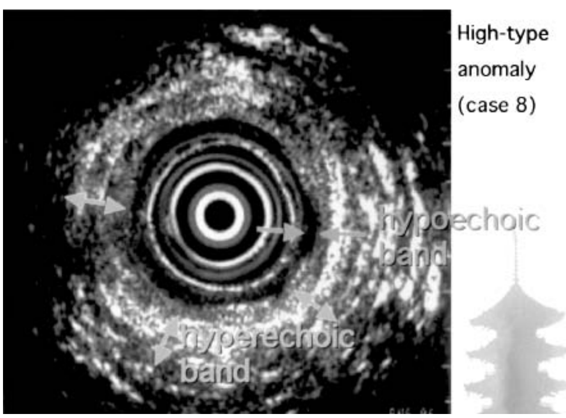

Fig. 26.6 An endosonographic image showing the hyperechoic band and hypoechoic band (high anomaly). The hyperechoic band layer is moderately defined from the surrounding tissues, and the continuity is uninterrupted. The hypoechoic band layer is well defined and the continuity is uninterrupted

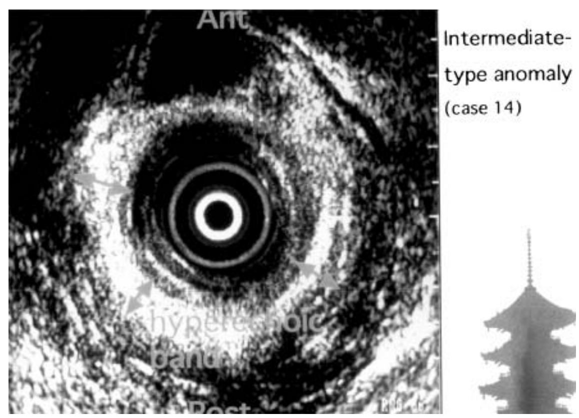

Fig. 26.7 An endosonographic image showing the hyperechoic band corresponding to the EAS (intermediate type). The lower portion of the hyperechoic band is well defined from the surrounding tissues, and the continuity is not interrupted. *Ant* Anterior, *Post* posterior

26.2.4 Results of Anal Endosonography

An image of the EAS was obtained in all ten patients with high anomalies; however, the distribution of the image was inadequate. The hyperechoic layer corresponding to the EAS was not completely defined from the surrounding tissues, although it was moderately defined in all ten patients (Fig. 26.6). The continuity of the hyperechoic layer was partially interrupted in two patients, and was complete in eight. The IAS was seen in five of the ten patients with high anomalies. Four out of the five patients showed a well-defined layer corresponding to the IAS with circular continuity, but the other showed complete interruption of the hypoechoic band.

The EAS was seen in all five patients with intermediate anomalies (Fig. 26.7). In addition, a well-defined layer corresponding to the EAS and uninterrupted continuity of the EAS image were noted in two. A moderately defined layer was observed in the other three patients. The IAS was seen in one patient with an intermediate anomaly, and was not seen in the remaining four patients.

Anal endosonography has demonstrated that patients with high anomalies have less adequate distribution of the EAS compared with those with intermediate anomalies, especially for the hyperechoic layer. These results indicate that patients with high anomalies have a congenitally rudimentary EAS.

The IAS has been regarded as being congenitally absent in patients with high or intermediate anomalies. However, Rintala reported the presence of the IAS even in patients with high anomalies, and stressed the importance of preserving the IAS at the time of surgery [11]. In the present study, five of the ten patients with high-type anomalies and one of the five patients with intermediate-type anomalies showed the hypoechoic band that corresponds to the IAS. Therefore, if the IAS is present in patients with high or intermediate anomalies, its preservation might contribute to the improvement of postoperative anorectal function.

References

1. Scharli AF, Kiesewetter WB (1969) Anorectosigmoid pressure studies as a quantitative evaluation of postoperative continence. J Pediatr Surg 4:694–704
2. Arhan P, et al (1976) Manometric assessment of continence after surgery for imperforate anus. J Pediatr Surg 11:157–166
3. Iwai N, et al (1979) A clinical and manometric correlation for assessment of postoperative continence in imperforate anus. J Pediatr Surg 14:538–543
4. Iwai N, et al (1988) Voluntary anal continence after surgery for anorectal malformations. J Pediatr Surg 23:383–397
5. Molander ML, Frenckner B (1983) Electrical activity of the external anal sphincter at different ages in childhood. Gut 24:218–221
6. Iwai N, et al (1997) Is a new biofeedback therapy effective for fecal incontinence in patients who have anorectal malformations? J Pediatr Surg 32:1626–1629
7. Hibi M, et al (2003) Results of biofeedback therapy for fecal incontinence in children with encopresis and following surgery for anorectal malformations. Dis Colon Rectum 46:S54–S58
8. Law PJ, Bartron CL (1989) Anal endosonography: technique and normal anatomy. Gastrointest Radiol 14:349–353
9. Sultan AH, et al (1993) Anal endosonography and correction with in vitro and in vivo anatomy. Br J Surg 80:508–511
10. Fukata R, et al (1997) A comparison of anal endosonography with electromyography and manometry in high and intermediate anorectal anomalies. J Pediatr Surg 32:839–842
11. Rintala R (1990) Postoperative internal sphincter function in anorectal malformations – a manometric study. Pediatr Surg Int 5:127–130

27 Scoring Postoperative Results

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27.1 Introduction

Standardized assessment of clinical outcome after repair of anorectal malformations (ARM) is essential for appropriate quality control in series of patients treated in single or different institutions, and for comparing different treatment modalities. Clinical assessment is subjective and may be biased by the observer, who is often the surgeon treating the patient. Therefore, pediatric surgeons performing clinical research need scales and scores that provide reliable information on the condition and functional status of their patients. However, appropriate methods and instruments for collecting data on the outcome after repair of ARM have been a matter of debate for decades.

In 1960, Scott [30] introduced a simple score, which was based on his clinical experience and which included exclusively clinical data. Since then, approximately ten scores have been introduced and used with varying frequency in patients after ARM repair. These scores are of different complexity with regard to clinical, functional, and other parameters. However, none of the instruments has undergone a proper validation process. This may be the main reason why no single score has been generally accepted to date. Therefore, the reported differences in results of different series of patients with ARM undergoing different methods of repair remain difficult to interpret [10], and a generally accepted score remains mandatory.

27.2 Principles of Scaling and Scoring

A scale is an instrument that is used to measure clinical phenomena, such as the degree of incontinence or the squeezing pressure of the anal sphincter. A score is a value on a scale in a given patient. The simplest and most complex scales have similar structures. They consist of elements and questions and their answers. Scores in specific patients may be dichotomous (yes/no), or rank-ordered. Thus, qualitative scores can be differentiated from numerical scores.

Principally, a score may serve three functions: prediction, evaluation over time, or description at a certain time point [2]. Scores have to be within a reasonable range of variation, with repeated administrations to the same patient by the same and different observers (reproducibility). In addition, a score has to be a valid measure of what it is supposed to be measuring (validity), and should show changes when the patient changes and no change when the patient is stable (responsiveness). Ideally, the process of ensuring reproducibility, validity, or responsiveness of a specific scale or score should not be based on the observer's clinical knowledge and common sense, but on a structured process.

Patients with ARM have mostly been scored descriptively. None of the scores suggested for use in these patients has undergone a standardized validation process concerning reproducibility, validity, or responsiveness, with the exception of specific quality-of-life scores. The latter were not specifically designed for patients with ARM. In addition, the problem of definition of endpoints has not been sufficiently addressed. Constipation, intermittent soiling, or other symptoms are not uniformly defined.

27.3 Specific Scores used in Patients with ARM

There is consensus that fecal continence represents the most important endpoint in patients with ARM. Therefore, specific scores for assessment of long-term results are focused on differentiating various degrees of fecal incontinence. No consensus has been achieved on including and scoring other symptoms such as constipation, urinary incontinence, electromanometric and endosonographic findings, or quality-of-life measurements.

27.3.1 The Scott Score

In 1960, Scott [30] established a qualitative score that differentiates between "good", "fair," and "poor" continence (Table 27.1). The items used are defecation habits, stool control, perianal soreness, and the function of the puborectalis muscle on digital examination. "Good" continence is defined as spontaneous regular defecation with or without occasional soiling during stress situations. Patients who are scored as "fair" have spontaneous and regular defecations or chronic constipation. They are continent for normal, but not for liquid stool. They suffer from frequent soiling and intermittent perianal soreness. The strength of the puborectalis sphincter is reduced. Patients with "poor" continence suffer from frequent stools, constant soiling, and perianal soreness, and have no puborectalis sphincter tone. This score was not validated, and a clear definition of specific items such as constipation, and puborectalis sphincter pressure was not given. However, the score was used and modified subsequently in early series of children with ARM [17,23].

27.3.2 The Kelly Score

In the Kelly method [15], the criteria are somewhat similar to the Scott score, but continence is scored quantitatively (Table 27.2). The determination is based on leakage phenomena, on the strength of the puborectalis sphincter, and on sensitivity. Factors include the appearance of staining or smearing, accidental defecation or soiling, sensitivity, the strength of the puborectal is muscle action on digital examination, and "feeling of defecation". A total of 5–6 points is considered "good," 3–4 points is "fair," and 2 points is "poor." Although not validated, the score has gained increasing popularity [6,13,19,22,29] and probably represents the most commonly used instrument for assessment of fecal incontinence today. It is not often used as a single instrument, but is compared with other more objective measures, such as manometry, electromyography, and quality-of-life data.

Table 27.1 Assessment criteria for continence according to Scott et al. (1960) [30]

Evaluation	Assessment criteria	Clinical result
Good	Defecation	Spontaneous/regular
	Stool control	Normal/occasional soiling during stress situations
	Soreness perianal	None
	Sphincter tension	Strong
Fair	Defecation	a) Regular b) Chronic constipation
	Stool control	Frequent soiling, continence restricted to normal stool consistency
	Soreness perianal	Only with frequent soiling
	Sphincter tension	Reduced
Poor	Defecation	Frequent
	Stool control	Constant soiling
	Soreness perianal	Constant
	Sphincter tension	None

Table 27.2 Kelly score (Kelly, 1972) [15]

Staining/smearing	none	2
	occasional	1
	constant	0
Accidental defecation/soiling	none	2
	occasional	1
	constant	0
Strength of puborectalis muscle	strong	2
	weak	1
	none	0

27.3.3 The Holschneider Score

Holschneider and Metzer [11] introduced a quantitative clinical score, including the parameters frequency of defecation, fecal consistency, soiling, rectal sensation, ability to hold back, discrimination, and need of therapy. Each of these seven parameters is scored as 0–2 according to the degree of impairment. In contrast to previous scores, items such as “frequency of stools” or “warning period” are clearly defined. A score of 10–14 points is “continent,” 5–9 points “fair,” and 0–4 points “incontinent.” Later, the authors felt that the parameters of rectoanal sensibility were overrepresented. They modified the score, reduced the clinical parameters, and included manometric data without changing the numerical scoring (Table 27.3) [8]. Both the initial and the modified scores have been used by some other authors in recent years [27].

Holschneider et al. [10] recently stated that neither a reference to the course of the anal or rectal fistula, nor a rating as “good,” “satisfactory”, or “sufficient”, nor the current score systems are suitable for comparative postoperative studies. The authors suggested renouncing the assessment of fecal continence, taking chronic constipation into account. With regard to three subgroups, the types of partial continence, but not the degrees of continence, are differentiated. The authors used their score in 78 patients and postulated that with regard to therapeutic conclusions, the results were more evident and more correct as compared to other scores.

Table 27.3 Clinical evaluation of continence according to Holschneider (1983) [8]. For these scores, 14 points means normal bowel habits, 10–13 points means good (social continence, few limitations in social life), 5–9 points means fair (marked limitations in social life), and 0–4 points means poor bowel habits (total incontinence)

Frequency of defecation	Normal (1–2/day)	2
	Often (3–5/day)	1
	Very often	0
Fecal consistency	Normal	2
	Soft	1
	Liquid	0
Soiling	No	2
	Stress/diarrhea	1
	Constant	0
Sensitivity	Normal	2
	Reduced (no discrimination)	1
	Missing	0
Anorectal resting pressure profile	≥ 20–24 mmHg	2
	14–19 mmHg	1
	< 13 mmHg	0
Maximum pressure at maximum squeezing	≥ 30 mmHg	2
	20–29 mmHg	1
	< 20 mmHg	0
Adaptation reaction	Normal	2
	Small amplitude, shortened	1
	Not detectable	0

27.3.4 The Wingspread Score

In the Wingspread score [31,32], the grades of continence are scored qualitatively. They fall into the four main categories of “clean,” “staining,” “intermittent fecal soiling,” and “constant fecal soiling” (Table 27.4). Subcategories include the need of occasional or constant therapy. In an additional category, related complications concerning the anorectum, urinary, genital, or other functions are noted. The grades are scored qualitatively and the instrument has been widely used in recent years [3,18,22,25,27].

27.3.5 The Rintala Score

Rintala and Lindahl [25] established a clinical score for the evaluation of fecal continence. The score is derived from standardized questionnaires and physical examination is not required (Table 27.5). The score consists of seven factors, which are scored from 0 to 3, except the factor of frequency of defecation, which is scored 1–2. The maximum bowel function score is 20. The authors used the score initially in 46 consecutive patients who had undergone surgical repair of high or intermediate ARM, and compared the results with data obtained from 70 healthy children with a similar age and sex distribution. Only 52% of the children of the control population obtained a completely normal bowel function score of 20 points. Functional aberrations in controls were occasional staining in 42% and constipation in 15%. The authors considered a score of 18 or higher to be normal. The score differentiated an excellent outcome with a normal score in 35% of the patients after repair. Another 35% of the patients were scored as “good,” having occasional staining and infrequent accidents; this group of patients scored 9–16 points. Patients with “fair” results had intermittent daily soiling or staining and scored 7–11 points. Patients with “poor” results scored 6–9 points and had to use daily enemas because of severe constipation or had constant soiling.

Table 27.4 Wingspread Score according to Stephens et al. (1988) [32]

1. Clean
1.1 No accumulated feces
1.11 No therapy
1.12 Occasional therapy
1.13 Therapy dependent
1.2 Accumulated feces
1.21 No therapy
1.22 Occasional therapy
1.23 Therapy dependent
2. Staining
2.1 No accumulated feces
2.11 No therapy
2.12 Occasional therapy
2.13 Therapy dependent
3. Intermittent fecal soiling
3.1 No accumulated feces
3.11 No therapy
3.12 Occasional therapy
3.13 Therapy dependent
3.2 Accumulated feces
3.21 No therapy
3.22 Occasional therapy
3.33 Therapy dependent
4. Constant fecal soiling
4.1 No accumulated feces
4.11 No therapy
4.12 Occasional therapy
4.13 Therapy dependent
4.2 Accumulated feces
4.21 No therapy
4.22 Occasional therapy
4.23 Therapy dependent
Related complications (specify)
1. Anorectal
(a) abnormal position
(b) stenosis
(c) prolapse
(d) fistula
(e) lack of contractility
(f) abnormal length
2. Urinary
3. Genital
4. Other

There were some validation steps. The scores derived from the questionnaires and the clinical outcome noted in the hospital records were positively correlated. In addition, pathological findings on plain spinal radiography or magnetic resonance imaging in 11 patients were negatively correlated with the bowel function score. Manometric findings did not differentiate between patients with excellent and good clinical outcome, but showed a significantly reduced anal resting pressure in patients with "fair" or "poor" clinical outcome. The authors used the score subsequently in a series of patients with low ARM [28]. Only half of these children had age-appropriate bowel function as compared to the control group. The main problem was constipation.

Table 27.5 Evaluation of fecal continence according to Rintala and Lindahl (1995) [25]

Ability to hold back defecation	
Always	3
Problems less than 1/week	2
Weekly problems	1
No voluntary control	0
Feels/reports the urge to defecate	
Always	3
Most of the time	2
Uncertain	1
Absent	0
Frequency of defecation	
Every other day to twice a day	2
More often	1
Less often	1
Soiling	
Never	3
Staining less than 1/week, no change of underwear required	2
Frequent staining, change of underwear often required	1
Daily soiling, requires protective aids	0
Accidents	
Never	3
Fewer than 1/week	2
Weekly accidents; often requires protective aids	1
Daily, requires protective aids during day and night	0
Constipation	
No constipation	3
Manageable with diet	2
Manageable with laxatives	1
Manageable with enemas	0
Social problems	
No social problems	3
Sometimes (foul odors)	2
Problems causing restrictions in social life	1
Severe social and/or psychic problems	0

Table 27.6 Evaluation of Bowel Function according to Peña 1995 [24]

1. Voluntary bowel movements	
Feeling of urge	
Capacity to verbalize	
Hold the bowel movement	
2. Soiling	
Grade 1	Occasionally (once or twice per week)
Grade 2	Every day, no social problem
Grade 3	Constant, social problem
3. Constipation	
Grade 1	Manageable by changes in diet
Grade 2	Requires laxatives
Grade 3	Requires enemas
4. Urinary incontinence	
Grade 1	Mild dribbling/wetness day and night
Grade 2	Complete incontinence

The advantages of the score are threefold. First, the questionnaires are completed by patients or parents; the assessment is thus observer-independent. Second, a physical examination is not required. And finally, data from a control group of children with normal bowel habits are available, and a drawback is the overlapping of scores in the different groups of continence (i.e., patients with a score of 9–11 may have either "good" or "fair" continence), which is the result of a lack of clear cut-off points.

27.3.6 Peña 1995

Peña [24] suggested a specific methodology for evaluation of long-term results according to his personal experience (Table 27.6). He evaluated 387 out of 792 patients who had undergone a posterior sagittal anorectoplasty. At the time of evaluation, none of the patients was allowed to be subjected to any type of medical management. Four parameters are evaluated:

1. Voluntary bowel movements, which are defined as feeling the urge to use the toilet to have a bowel movement, the capacity to verbalize it, and to hold the bowel movement.
2. Soiling is defined as involuntary leaking of small amounts of stool, which may be present with or without voluntary bowel movements. Soiling grade 1 occurs occasionally (once or twice per week). Grade 2 refers to soiling that occurs every day, but does not cause social problems. Grade 3 represents constant soiling with social problems.
3. Constipation is defined as the incapacity to empty the rectum spontaneously without help every day (grade 1: when the patient is manageable by diet; grade 2: when he requires laxatives; grade 3: when he requires enemas).
4. Urinary incontinence is considered grade 1 when the patient has mild dribbling and wetness of the underwear day and night, and grade 2 when he is completely incontinent. Patients with voluntary bowel movements and no soiling are considered totally continent. (These were 41% of the total series.)

Peña presented a detailed analysis of the postoperative results related to the types of fistula and malformation. However, to our knowledge, this score has not yet been widely used.

27.3.7 Other Scores

Other authors have suggested their own method of assessing continence and investigated small series of patients. Ditesheim and Templeton [4] introduced a "qualitative fecocontinence score." Points are assigned for the degree of awareness of impending stool, occurrence of accidental defecation, need for extra underwear or liners, social problems related to odor, restriction of physical activity, and presence of rashes. The items are scored from 0 to 1; a total score of 4–5 points is "good continence", 2–3.5 points is "fair continence", and 0–1.5 points is "poor continence."

Kiesewetter and Chang [16] categorized continence as "continent most of the time, suffering only occasionally from soiling when diarrhea or unusual physical stress were encountered," fair as "occasional soiling or staining with a normal consistency stool, but with a socially acceptable degree of continence," and poor as "frank incontinence, with occasional times of control, or permanent colostomy established after a period of definitive therapy."

27.4 Other "Objective" Methods of Scoring

Electromanometry has been used to determine the degree of incontinence since the early 1960s [11–13]. Holschneider [8] electromanometrically defined four grades of continence. These grades were derived from numerous parameters, such as anorectal pressure profile, fluctuations, relaxation of the internal sphincter, external sphincter contractions, puborectalis sphincter contractions, pressure tolerance, defecation reflex, adaptation, compliance, and critical volume. The criteria for grading continence were well established, but there remained some inconsistency due to overlapping of the parameters in the different groups. The author stated that electromanometric scoring reveals more unfavorable results when compared to clinical scores, but data derived from a large series of patients undergoing both clinical and electromanometric scoring are lacking. However, he suggested including selected manometric data in his clinical score for obtaining a more objective result [8].

Diseth and Emblem [3] confirmed that anal canal resting pressure and squeeze pressure correlated negatively with fecal incontinence. In a study of Hedlund et al. [7], abnormal anal resting tone was found in 14 out of 17 patients with soiling 5–10 years after repair. However, the correlation to clinical results was incomplete and some patients without soiling had an abnormal resting tone. Other authors confirmed a lack of correlation of some manometric parameters with clinical continence. In a long-term study of 22 patients with high ARM, Rintala et al. [26] found that the only manometric parameter that correlated with the continence outcome was voluntary squeeze pressure.

Schuster et al. [29] recently used computerized vector manometry in 17 patients with various types of ARM. Besides computerized software supported by data on standard manometric parameters, a score assessing three pressure zones of the anal canal (0–16 points) was established. However, the authors found a poor correlation between quantitative manometric parameters and clinical results, which were assessed by a modified Kelly score.

Fukata et al. [6] compared endosonography and electromyography of the external anal sphincter with electromanometry and clinical data derived from the Kelly score. Endosonographic findings for the external anal sphincter corresponded well with electromyographic findings, but not with manometry. Only 15 patients were investigated. Jones et al. [14] compared endosonography with magnetic resonance imaging after repair of ARM. The findings were comparable in only 9 out of 14 patients. Fukuya et al. [5] compared magnetic resonance imaging with clinical assessment on the basis of the Kelly score. The proportion of "fair" or "poor" developed muscles was not significantly different between the continence groups according to Kelly. Therefore, no conclusion concerning the correlation of endosonographic and magnetic resonance imaging findings with clinical scores can be drawn to date.

27.5 Quality-of-Life Measurements

Quality of life is a multidimensional concept, which includes, but is not limited to, the social, physical, and psychological functioning of the individual. Validated instruments are supposed to objectively measure the domains of quality of life, and to exclude observer bias. The relevance of quality-of-life assessment in children with ARM was confirmed in an early study by Ditesheim and Templeton [4], who used a questionnaire scoring system that included items such as school attendance, social relationships, and physical capacities. Today it is well known that children and adolescents with fecal incontinence may suffer from emotional problems, internalizing behavior problems, and depressive symptoms. Various measures of quality of life have been used for quantitative and qualitative scoring of children and adolescents with fecal incontinence and constipation. However, the results presented below are not conclusive and none of the suggested instruments has been generally accepted.

Diseth and Emblem [3] used semistructured interviews and questionnaires, such as the Child Assessment Schedule, Child Behavior Check List, and self report in 33 adolescents with ARM. Psychosocial functions were impaired in 73% of the adolescents, and 58% met the criteria for psychiatric diagnosis. The authors found a significant correlation of the degree of flatus incontinence with the degree of psychosocial impairment and of continence of flatus with mental health symptom scores.

Ludman and Spitz [19] assessed the quality of life by self-report questionnaires, such as the Depression Self-Rating Scale and Self-Perception Profile, in 157 children and adolescents. In addition, the authors assessed parents and teachers using the Child Behavior Checklist and other instruments. The level of continence, which was defined by the Kelly score, did not influence psychological adjustment, with exception of incontinent young girls. There were no significant differences between continent and incontinent children concerning the global self-worth measure. However, 29% had some psychiatric disorder and these were more likely children with incontinence. Other work confirmed that mental health or psychological problems were found more frequently in 160 children and adolescents with ARM as compared to the normal population, but the incidence of these problems was similar in continent and incontinent patients [20].

Bai et al. [1] used the Achenbach's Child Behavior Checklist in 71 children with ARM and found quality of life to be significantly reduced as compared to a normal control group. The authors established tenta-

Table 27.7 Quality-of-life scoring criteria for children (aged 8–16 years) with fecal incontinence according to Bai et al. (2000) [1]. Note that the higher the scores, the better the quality of life

Item	Criteria	Points
Soiling	Absent	4
	Accidental	3
	Frequent	2
Incontinence	Accidental	1
	Frequent	0
School absence	Never	2
	Accidental	1
	Frequent	0
Unhappy or anxious	Never	2
	Accidental	1
	Frequent	0
Food restriction	No	2
	Somewhat	1
	Much	0
Peer rejection	Never	2
	Accidental	1
	Frequent	0

tive quality-of-life scoring criteria, including somatic assessment, social aspects, and psychological investigation (Table 27.7).

Quality-of-life assessment in patients with ARM is essential. However, no instruments taking the specific symptoms and problems into account have yet been established. The use of currently available instruments and the calculation of scores remains difficult and time consuming. Therefore, a specific instrument for scoring quality of life after ARM repair cannot be recommended for routine use yet.

27.6 Comparison of Scores and Outcomes

Studies comparing different scores in the same study population are scarce. Ong and Beasley [21] compared 4 scoring methods in 37 patients who had undergone sacroperineal rectoplasty. Continence was scored as "good," "fair," and "poor" using two numerical (Kelly [15], Templeton and Ditesheim [33] and two qualitative scores (Kiesewetter and Chang [16], Wingspread – Stephens and Smith [31]). The Wingspread score was adapted to three categories for comparison purposes. The results differed considerably. The authors found "good" continence in approximately 30–70% of patients with high ARM, and "poor" continence in approximately 5–20%. The Templeton score assigned a higher score of fecal continence than the other three, with excellent concordance. In patients requiring revision after Stephens rectoplasty, the Templeton and Kelly scores, in contrast to the others, did not show incontinence in two and three patients, respectively [22].

Table 27.8 Method for assessment of outcome established in Krickenbeck 2005 (patient age > 3 years, no therapy; Holschneider et al. [9])

1. Voluntary bowel movements		**yes/no**
Feeling of urge		
Capacity to verbalize		
Hold the bowel movement		
2. Soiling		**yes/no**
Grade 1	Occasionally (once or twice per week)	
Grade 2	Every day, no social problem	
Grade 3	Constant, social problem	
3. Constipation		**yes/no**
Grade 1	Manageable by changes in diet	
Grade 2	Requires laxatives	
Grade 3	Resistant to diet and laxatives	

27.7 Conclusions and Results of the Krickenbeck Meeting 2005

A clear recommendation concerning specific instruments cannot be derived from clinical experience or from data in the literature. To date, the Kelly score [15] has been most widely used. The Holschneider score [8] is the only one that includes an objective parameter (electromanometry) and gives more detailed information regarding bowel habits. The score introduced by Rintala and Lindahl [25] underwent a validation process and data from a control group of children with normal bowel habits are available. Quality-of-life measurement reveals the most relevant and detailed information. However, the appropriate instruments for quality-of-life measurement of children and adolescents with incontinence remain a matter of discussion.

At the Krickenbeck Meeting in 2005, consensus was achieved concerning the assessment of outcome after ARM repair (Table 27.8). The method comprises three parameters: voluntary bowel movements (yes/no), soiling (yes/no, if yes grade 1–3), and constipation (yes/no, if yes grade 1–3). The assessment should be performed in children more than 3 years of age who are not undergoing therapy. The surgeon, who is involved in the treatment and follow-up of the patients who are to be scored, may bias the results of scoring. Therefore, the assessment and analysis of data should preferably be done by a person who is not involved in the treatment of the patient. This may bring pediatric surgeons closer to psychologists and methodologists, who are essential for establishing validated instruments for assessment of children and adolescents with ARM in the future.

References

1. Bai Y, Yuan Z, Wang W, Zhao Y, Wang H, Wang W (2000) Quality of life for children with fecal incontinence after surgically corrected anorectal malformation. J Pediatr Surg 35:462–464
2. Charlson ME, Johanson NA, Williams PG (1991) Scaling, scoring, and staging. In: Troidl H, Spitzer WO, McPeek B, Mulder DS (eds) Principles and Practice of Research. Springer, New York, pp 192–200
3. Diseth TH, Emblem R (1996) Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. J Pediatr Surg 31:638–643
4. Ditesheim JA, Templeton JM Jr (1987) Short-term v long-term quality of life in children following repair of high imperforate anus. J Pediatr Surg 22:581–587
5. Fukuya T, Honda H, Kubota M, Hayashi T, Kawashima A, Tateshi Y, Shono T, Suita S, Masuda K (1993) Postoperative MRI evaluation of ARM with clinical correlation. Pediatr Radiol 23:583–586
6. Fukata R, Iwai N, Yanagihara J, Iwata G, Kubota Y (1997) A comparison of anal endosonography with electromyography and manometry in high and intermediate anorectal anomalies. J Pediatr Surg 32:839–842
7. Hedlund H, Peña A, Rodriguez G, Maza J (1992) Long-term anorectal function in imperforate anus treated by a posterior sagittal anorectoplasty: manometric investigation. J Pediatr Surg 27:906–909
8. Holschneider AM (1983) Elektromanometrie des Enddarms. 2. neubearbeitete, erweiterete Auflage. Urban and Schwarzenberg, München-Wien-Baltimore
9. Holschneider A, Hutson J, Peña A, Beket E, et al (2005) Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. J Pediatr Surg 40:1521–1526
10. Holschneider AM, Jesch NK, Stragholz E, Pfrommer W (2002) Surgical methods for ARM from Rehbein to Peña – Critical assessment of score systems and proposal for a new classification. Eur J Pediatr Surg 12:73–82
11. Holschneider AM, Metzer EM (1974) Elektromanometrische Untersuchungen der Kontinenzleistung nach rektoanalen Fehlbildungen. Z Kinderchir 14:405–412
12. Iwai N, Ogita S, Kida M, Fujita Y, Majima S (1979) A clinical and manometric correlation for assessment of postoperative continence in imperforate anus. J Pediatr Surg 14:538–543
13. Iwai N, Yanagihara J, Tokiwa K,Takahashi T (1989) Rectoanal pressure studies and postoperative continence in imperforate anus. Prog Pediatr Surg 24:115–120
14. Jones NM, Humphreys MS, Goodman TR, Sullivan PB, Grant HW (2003) The value of anal endosonography compared with magnetic resonance imaging following the repair of ARM. Pediatr Radiol 33:183–185
15. Kelly JH (1972) The clinical and radiological assessment of anal continence in childhood. Aus N Z J Surg 42:62–63
16. Kiesewetter WB, Chang JHT (1977) Imperforate anus: a five to thirty year follow-up perspective. Prog Pediatr Surg 10:111–120
17. Kiesewetter WB, Turner RC (1963) Continence after surgery for imperforate anus: a critical analysis and preliminary experience with the sacroperineal pull-through. Ann Surg 158:498–502
18. Liem NT, Hau BD (2001) Long-term follow-up results of the treatment of high and intermediate ARM using a modified technique of posterior sagittal anorectoplasty. Eur J Pediatr Surg 11:242–245
19. Ludman L, Spitz L (1995) Psychosocial adjustment of children treated for anorectal anomalies. J Pediatr Surg 30:495–499
20. Ludman L, Spitz L, Kiely EM (1994) Social and emotional impact of faecal incontinence after surgery for anorectal abnormalities. Arch Dis Child 71:194–200
21. Ong N-T, Beasley SW (1990) Comparison of clinical methods for the assessment of continence after repair of high anorectal anomalies. Pediatr Surg Int 5:233–237
22. Ong N-T, Beasley SW (1991) Long-term continence in patients with high and intermediate anorectal anomalies treated by sacroperineal (Stephens) rectoplasty. J Pediatr Surg 26:44–48
23. Partridge JP, Gough MH (1961) Congenital abnormalities of the anus and rectum. Br J Surg 49:37–40
24. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
25. Rintala RJ, Lindahl H (1995) Is normal bowel function possible after repair of intermediate and high ARM? J Pediatr Surg 30:491–494
26. Rintala RJ, Lindahl HG, Rasanen M (1997) Do children with repaired low ARM have normal bowel function? J Pediatr Surg 32:823–826
27. Rintala R, Mildh L, Lindahl H (1992) Fecal continence and quality of life in adult patients with an operated low anorectal malformation. J Pediatr Surg 27:902–905

28. Rintala RJ, Lindahl HG (2001) Fecal continence in patients having undergone posterior sagittal anorectoplasty procedure for a high anorectal malformation improves at adolescence, as constipation disappears. J Pediatr Surg 36:1218–1221
29. Schuster T, Lagler F, Pfluger T, Dietz HG, Joppich I (2001) A computerized vector manometry and MRI study in children following posterior sagittal anorectoplasty. Pediatr Surg Int 17:48–53
30. Scott JE, Swenson O, Fisher JH (1960) Some comments on the surgical treatment of ARM. Am J Surg 99:137–143
31. Stephens FD, Smith ED (1986) Classification, identification, and assessment of surgical treatment of anorectal anomalies. Pediatr Surg Int 1:200–205
32. Stephens FD, Smith ED, Paul NW (1988) ARM in children: Update 1988. March of Dimes Birth Defect Foundation. Original Series Vol. 24 (4). Alan R. Liss, New York
33. Templeton JM, Ditesheim JA (1985) High imperforate anus – quantitative results of long-term fecal incontinence. J Pediatr Surg 20:645–652

28 Results Following Treatment of Anorectal Malformations

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28.1 Introduction

The outcomes of patients with anorectal malformations (ARM) have greatly improved during the last decades as a result of modern surgical techniques and improved neonatal care facilities. Early survival is today a rule except in some rare cases with cardiac, urogenital, or chromosomal anomalies that are not compatible with life. The overall long-term functional outcome expectancy in terms of fecal and urinary continence is today relatively optimistic. The majority of patients reaching adolescence and adulthood are able to maintain themselves socially continent.

The present review is based mainly on pertinent literature. In addition, the author's personal experience with 245 posterior sagittal anorectoplasty (PSARP) procedures for high and intermediate malformations, and management of 130 low anomalies between 1984 and 2004 is used as a basic material to address specific previously unpublished issues in the management of ARM.

28.2 Short-Term Outcome

28.2.1 Mortality

ARM are very often a part of a malformation complex. Some associated anomalies, especially cardiovascular malformations, may be uncorrectable and there is therefore always going to be a degree of mortality, albeit low, among these patients. The VACTERL (acronym: Vertebral anomalies, Anal atresia, Cardiac defect, TracheoEsophageal fistula with esophageal atresia, Renal abnormalities, and Limb abnormalities) association carries considerable mortality, especially if there is a combination of esophageal atresia and cardiovascular anomaly as well as an ARM [1,2].

The mortality of patients with ARM during the last decades has been between 10 and 20% of all cases [2,3]. The mortality of patients with high and intermediate anomalies is about three times higher than that of patients with low anomalies, which corresponds to

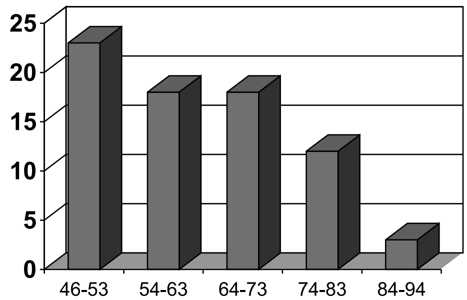

Fig. 28.1 Reduction in mortality (%) in patients with anorectal malformations between 1946 and 1994

the higher incidence of severe associated anomalies. Only a minority of deaths are directly related to the ARM and its treatment [1–3]. At the Children's Hospital, University of Helsinki, the mortality of ARM has decreased from 23% in the late 1940s and early 1950s to 3% in the 1980s and 1990s (Fig. 28.1). This decrease is clearly due to improvements in the care of severe associated anomalies, especially cardiac defects.

28.2.2 Operative Complications – High and Intermediate Anomalies

A neonatally performed colostomy carries a high degree of morbidity. The most common complications are colostomy prolapse and stricture [4,5]. Stoma complications appear to be less common with a completely divided sigmoid colostomy than with transverse or loop colostomy [6,7]. The reported total incidence of complications of infant colostomies ranges between 17 and 68% of cases; the complications also include a few colostomy-related deaths.

Early complications occur following all commonly used reconstructions for high and intermediate ARM. Peritonitis, retraction or dehiscence of the pull-through segment, and refistula between the bowel and the urogenital tract are typical severe early complications. The incidence of these major complications ranges between 10 and 30% following abdominoperineal or sacroabdominoperineal pull-through operations [2–4]. Severe complications seem to be less common following posterior sagittal anorectoplasty. In the large series of Peña, serious complications requiring major reoperative surgery occurred in 2% of the cases, mainly following repair of a cloaca [8]. In the author's series of 245 patients there were 4 major early complications requiring reoperations: 2 patients with a persistent cloaca developed urethrovaginal fistulae, 1 patient with rectovaginal fistula had retraction of the pulled-through anal canal, and 1 with rectovestibular fistula had dehiscence of the reconstructed perineal body.

Postoperative anal complications have been common following traditional pull-through operations. Anal stenosis and mucosal prolapse have been found in up to 15-78% of patients [2–4,9,10]. Stenosis has usually been attributed to inadequate anal dilatations during the follow-up period. Anal stenosis may respond to dilatations; in refractory cases, surgical excision of scar tissue is needed. Mucosal prolapse usually requires operative treatment to reduce mucosal soiling and to improve sensation in the neoanal canal. On the other hand, local anal problems have been rare following posterior sagittal anorectoplasty. Peña reported very few local complications in his series of 792 patients [8]. In the author's series of 245 posterior sagittal anorectoplasties, anal stenosis requiring anoplasty occurred in 3 patients; in addition, 3 patients, early in the series, required local operation for a minor mucosal ectopy.

28.2.3 Operative Complications – Low Anomalies

Early complications after neonatal treatment of low anomalies are uncommon. However, local complications may occur later and are usually caused by inappropriate long-term follow-up and care. A typical problem is postoperative anal stenosis, which can be prevented by gradual postoperative dilatations and careful follow-up of the patient. Untreated anal stenosis may cause secondary megacolon, which may require operative treatment [2,3,11]. In some cases the rectal blind pouch is primarily so ectatic that it is symptomatic without any associated anal stricture or stenosis [11,12].

28.2.4 Urological Complications

Lesions in the urinary tract may complicate surgery for ARM. Before the definitive repair, it is essential to minimize the risk of urinary tract infections caused by the rectourogenital connection, if present. This is best accomplished by establishing a completely diverting colostomy and by careful washout of the rectal pouch [8]. Infection may cause permanent damage to the kidneys, because upper urinary tract anomalies, and especially vesicoureteral reflux, are common [13–15]. Infection may occur after definitive repair and is caused in most cases by a urological anomaly, vesicoureteral reflux being the most common [15]. Urinary infections may be caused by a rectourinary fistula remnant that is too long [16]. Damage to the pelvic innervation and urethra during the dissection of rectal blind pouch may cause urinary incontinence or urethral stricture [17]. The incidence of urological injuries associated with surgery are strongly related to the experience of the surgeon [18]. A detailed imaging of the rectourinary communication by contrast studies may decrease the possibility of injury. Many of the functional urinary abnormalities previously attributed to surgical intervention are congenital [13,19].

28.2.5 Functional Problems During the First Years of Life

Bowel function is not normal in patients with repaired high or intermediate ARM following closure of the protecting colostomy. The most common problem is frequent bowel movements, which may cause perineal skin excoriation. In patients with traditional pull-through operations, this stage of bowel function, which is best characterized as uncontrollable soiling, may continue for a long period of time, often for years. This is particularly the case with operations where the terminal rectum is resected as a part of the procedure and not pulled-through (sacroabdominoperineal pull-through and endorectal abdominoperineal pull-through) [2,20].

Constipation is a major problem with patients who have had PSARP or anal transposition for rectovestibular fistula [2,8,11,21]. Constipation may develop secondary to untreated anal stenosis, but more commonly is a consequence of disordered colonic motility [8,22]. Constipation may begin early after the operation, and its severity is related to the degree of the initial dilatation of the rectal blind pouch [8,21]. Constipation is also the most common early functional problem in patients with low anomalies [2,23]. Severe soiling with low anomalies is rare and is caused by operative sphincter damage or severe sacral defects.

Associated anomalies cause significant morbidity in patients with ARM. Congenital heart defects occur in more than 15% of the patients [2] and often require major cardiac surgery during early childhood. Associated gastrointestinal anomalies are found in almost 20% of the patients [2]. Half of these patients have esophageal atresia, which may require additional surgery such as esophageal dilatations and fundoplication, despite successful neonatal repair.

Genitourinary tract abnormalities are encountered in more than 40% of patients with ARM [2,24]. These anomalies are more common in patients with high malformations. Upper urinary tract dilatation, which is most commonly related to vesicoureteral reflux, requires careful follow-up and sometimes surgery. Congenital neurovesical dysfunction may complicate successful treatment of structural urinary tract abnormalities [13,25].

Vertebral anomalies occur frequently in patients with ARM [2]. Hemivertebral anomalies may cause scoliosis, which may require operative stabilization or bracing early in childhood. Spinal dysraphism, detected by magnetic resonance imaging (MRI), has recently been found to be more common than has previously been appreciated [26]. This may have significant impact on the functional outcome in terms of urinary function and fecal incontinence. Routine spinal and cord MRI studies have been advocated for all patients [27]. Careful neuroradiological follow-up is warranted in patients with spinal dysraphism.

28.3 Long-Term Outcome

28.3.1 Evaluation of Long-Term Functional Outcome

28.3.1.1 Clinical Evaluation

In the literature there is a great variation in the functional results after repair of ARM. There is no generally agreed method by which to assess the bowel function of patients with ARM, and the main problem in comparing different series is the highly variable criteria used in the evaluation of fecal continence. Evaluation of bowel function during childhood may be biased because the information concerning the functional outcome is derived mainly from the parents; they may not want to report unfavorable results to a surgeon who has been responsible for the treatment of their child. The parents may also ignore minor and moderate defects in continence in a child whose bowel function has been deficient from birth or, in the case of smaller children, may consider them to be part of normal maturation of defecation. The final outcome may not be assessed until the patients have reached adulthood and, as independent individuals, can evaluate the social consequences of possibly defective bowel control.

Assessment of long-term results has been made at different ages by different pediatric surgeons. An appropriate age to perform functional assessment is the age when normal children are fully toilet trained. In the Western world children are usually toilet trained when they reach the age of 3 years. Peña [8] evaluated his patients with PSARP when they were older than 3 years of age, Rintala et al. [28] 3 years after the closure of the colostomy, and Langemeijer et al. [29] and Bliss et al. [30] at the age of 4 years.

Clinical assessment based mainly on history has been the most common method to evaluate functional outcome. The outcome has usually been presented as good, fair, or poor. Kiesewetter and Chang [31] used fecal soiling as a basis for clinical evaluation. A patient with a good result was defined as being continent most of the time, suffering only occasional soiling during diarrhea and physical stress. A fair result was one in which there was occasional soiling with normal stool consistency, but acceptable social continence. Frank incontinence or permanent colostomy was considered a poor result. Nixon and Puri [32] used a questionnaire to the parents, hospital records, personal interview, and clinical examination to classify bowel control into three groups: normal control, occasional soiling, and frequent soiling or colostomy. Swenson and Donnellan [33] suggested that motility problems causing constipation were the most frequent long-term complications in patients who had experienced abdominoperineal pull-through. He based his nonscoring clinical evaluation on the occurrence of symptoms of constipation. Scoring methods for the assessment of postoperative functional outcome are discussed in Chap. 27.

28.3.1.2 Objective Methods

Manometric assessment has been the principal method used to obtain objective data of postoperative sphincter function. However, there is no standard for manometric evaluation. Some authors [34] have used single, fluid-filled balloons in the anal canal. Balloon devices allow only stationary measurements; others [35,36] favor, therefore, the open-tip perfused catheter method. The numerical pressure recordings in stationary and dynamic studies are not directly comparable; the values of manometric tracings tend to be higher with the balloon method.

The manometric data are most often combined with clinical information [28,37–39]. Only Hecker and Holschneider have used manometric findings alone as an indicator of functional outcome [3]. Manometric and clinical results have often been found contradictory. Some investigators have found a positive correlation between clinical continence and the anal resting pressure profile [28,34,38], others have found no such correlation, but have observed a correlation between continence and voluntary squeeze force [40]; still others have reported no correlation at all between clinical continence and pressure profile or squeeze force [35,37]. However, a clear correlation between the presence of an inhibitory rectoanal reflex and clinical continence have been stated by several authors [28,34,35,38,39]. Decreased rectal sensitivity at rectal distension has been reported to correlate with poor functional outcome [34,35,40].

Anal sphincter myography has also been used to evaluate the postoperative results. A positive inflation reflex has been associated with good fecal continence [40,41]. Boyd et al. [42] found significant abnormalities in the external sphincter in most of their patients with incontinence following repair of an ARM. The typical findings were asymmetry of the sphincter in relation to the anal orifice, a sphincter that is completely separate from the neoanus, and no sphincter activity at all. They suggested that these findings are consistent with sphincter damage during primary corrective surgery.

Shandling et al. have developed a simple method to measure anal sphincter force in patients with ARM [43]. They used a balloon catheter attached to a hand-held dynamometer. The device records the anal sphincter force in grams when pulled out from the distal rectum. The main findings were that patients with high ARM had significantly lower anal sphincter force than normal subjects, and also lower than patients with spina bifida.

Imaging studies to assess long-term anorectal function in patients with ARM have been reported infrequently. Dynamic radiographic examination of the anorectal function by defecography has been used to objectively analyze anorectal function [44–46], but has not gained widespread popularity, mainly because it is cumbersome and requires the full cooperation of the patients to provide valid information. Modern imaging modalities such as intra-anal ultrasound, computed tomography (CT) and MRI provide excellent information about the postoperative anatomy of an ARM. Anal ultrasound has been found to be a reliable tool in the evaluation of sphincter integrity in patients with repaired ARM [47,48]. Good sphincter anatomy at ultrasound is also correlated with good function [47]. CT provides good anatomical data concerning the relationship between a pulled-through bowel and the sphincter system [49], but gives a high dose of ionizing radiation. Only a few studies have shown any

Table 28.1 Functional outcome during childhood - low malformations

Older series	***N***	**Good**	**Fair**	**Poor**
Stephens and Smith [4]	46	83%	15%	2%
Trusler and Wilkinson [17]	20	80%	20%	–
Kiesewetter and Chang [31]	68	88%	7%	5%
Partridge and Gough [55]	150	86%	11%	3%
Recent series	***N***	**Normal**	**Constipated**	**Soil/smear**
Yeung and Kiely [23]	35	47%	53%	28%
Rintala et al. [57]	40	52%	42%	10%
Laboure et al. [59]	27	52%	48%	
Javid et al. [60]	28	53%	47%	

correlation between clinical outcome and the integrity of the sphincter complex at CT [35]. MRI may have an advantage over CT because of superior soft-tissue characterization, multiplanar imaging, and lack of ionizing radiation. At MRI, a hypoplastic sphincter complex, misplacement of the bowel in relation to the sphincters, and an obtuse anorectal angle have been related to poor outcome [50,51]. However, correlations between clinical results and findings in MRI have not been convincing. It may also be that MRI is not superior to anal ultrasound, which is a much cheaper and easier imaging method [48]. The main value of MRI in patients with ARM is not in detecting abnormalities in the sphincter complex, but that it reveals associated spinal dysraphism, which is common in these patients [52,27].

28.3.2 Long-Term Bowel Function

The variable criteria used to evaluate long-term outcome in patients with ARM makes comparison of the reported outcomes difficult. Moreover, these criteria are mainly designed for high anomalies [4,20,31,53,54]. In high anomalies, a good result usually means socially acceptable continence, which is not equivalent to normal anal function. A patient with a high anomaly and a good functional result rarely has normal bowel function and, although socially continent, often has a minor degree of smearing or soiling associated with physical straining or loose stools. Although many patients with low malformations have normal bowel function at long term, a method designed to assess long-term outcome in high anomalies may underestimate minor defects in bowel function, which may become significant when the patient leads an independent adult life.

28.3.3 Results in Low Anomalies

28.3.3.1 Long-Term Bowel Function During Childhood

Traditionally, the long-term results of low malformations are considered to be good in the great majority of patients (Table 28.1) [4,17,31,32,55]. The poor results have been considered to be related to neurological damage and mental retardation [56] or insufficient long-term care of the patients [3,32]. Recently, more critical and comprehensive analyses have demonstrated clearly a significant number of patients with functional aberrations, especially chronic constipation (Table 28.1). In the series of Yeung and Kiely [23], 15 of 32 children with a follow-up between 1 and 7 years had normal bowel function. Of the remaining 17 patients, all had constipation and 9 occasional or frequent soiling requiring treatment. Rintala et al. [57] used a scoring system to compare the bowel function of 40 children with low anomalies with that of healthy children. Only 52% of their patients had scores within the range of healthy children; constipation was found in 42% and soiling in 10% of the patients. Ong and Beasley [58] reported a follow-up of 70 patients with low ARM; of the 35 children under the age of 15, 9 were clean, 14 had occasional smearing, and 12 soiling. Laboure et al. [59] reported 27 male patients with 3–20 years follow-up; 48% of the patients had long-term problems related to constipation or soiling. Javid et al. [60] found, in their series of 44 girls with low imperforate anus, that 47% experienced at least occasional fecal incontinence. The median follow-up time in this series was 11 years.

28.3.3.2 Long-Term Bowel Function in Adults

There are only a few reports concerning the functional results of low ARM, with a follow-up extending beyond childhood. Karkowski et al. [61] reported good continence in 12 (80%) of his 15 patients with low malformations. Nixon and Puri [32] found normal control under all circumstances in 23 (74%) of their 31 adult or adolescent patients. The remaining patients had occasional or frequent soiling. These results reflects the traditional view of the long-term outcome in these patients. More recently, the large series of Ong and Beasley [58] and Rintala et al. [62] have demonstrated that a significant percentage of patients have abnormal anorectal function at adult age. Ong and Beasley had 35 patients with a follow-up of more than 15 years. Although the majority had good continence according to commonly used clinical scoring methods, only 13 (37%) of the patients were clean at all times. Seventeen patients (49%) had smearing and 5 (14%) intermittent soiling. Rintala et al., using a quantitative scoring method, compared the bowel function of 83 patients with that of healthy individuals with a similar age and sex distribution. All controls, but only 60% of the patients with low anomalies, had good continence. The social problems related to deficient fecal control were reported by 39% of the patients. In addition, 13% of the patients had difficulties in sexual function. Other health problems were reported by 52% of the patients, but only by 6% of the controls.

28.3.4 Results in High and Intermediate Anomalies

28.3.4.1 Prognostic Factors

The level of the rectourogenital connection is an important prognostic factor of bowel function. Males with a bladder-neck fistula and females with a high-confluence cloaca [8] have worse prognosis than patients with a lower connection. It has been shown clearly that the bowel function of patients with intermediate anomalies (rectobulbar fistulae in males, low-confluence cloacae and rectovestibular fistulae in females) is better than of those with higher anomalies [28,38]. The obvious cause of poorer prognosis in high anomalies is the more marked hypoplasia of the voluntary sphincter muscles [8].

Severe sacral abnormalities adversely affect long-term functional outcome. More than two missing sacral vertebrae or other major sacral deformities, such as hemivertebrae and vertebral fusions, worsen the functional outcome when compared with patients with a more normal sacrum [8,28]. The poor outcome in patients with severe sacral anomalies is usually related to sphincter insufficiency. Sacral dysplasia may also cause severe constipation by impairing rectal sensibility [21,28].

Modern ultrasound and MRI imaging has enabled noninvasive evaluation of the spinal cord in patients with ARM. Occult myelodysplasias have been found to be common, occurring in 20-50% of the patients [26,52]. Myelodysplasias are more common in high lesions and usually associated with sacral deformities. At the moment it is unclear if these lesions have any effect on long-term bowel function.

The functional role of the internal sphincter following repair of ARM is controversial. The internal sphincter has been considered to be either missing or insignificant for the development of fecal continence [4]. Embryological, animal and clinical studies have, however, documented the presence of the internal sphincter in the region of the fistulous bowel termination [63–65]. The functioning internal sphincter can be demonstrated by the presence of rectoanal relaxation reflex on anorectal manometry. Most patients with a low anomaly have a positive rectoanal reflex [2,38,56]. In patients with more complex malformations, the rectoanal relaxation reflex has traditionally been present in only a minority of patients [37,38,56]. However, when the rectourogenital connection has been preserved at the anorectal repair, a functional internal sphincter has been demonstrated later in 40-80% of patients [28,66,67]. The presence of internal sphincter has been shown to correlate with favorable functional outcome [28,34,36,39,66,68].

Abnormal colonic motility, usually presenting with constipation, has been reported to be a problem in patients with low ARM and in females with a vestibular fistula [2,8]. Since the advent of PSARP for higher anomalies, however, cumulative evidence has shown that chronic constipation is one of the main functional complications encountered following repair [8,21,69]. The incidence of constipation following the PSARP procedure varies in the literature between less than 10% [29] and 73% [28,70]. Constipation seems to be more common when internal-sphincter-preserving techniques have been used [28,66,70]. The cause of constipation is unclear; extensive mobilization of the anorectum may cause partial sensory denervation of the rectum and impair rectal sensation. Rectosigmoid hypomotility has also been suggested

[8]. Many patients suffering from postoperative constipation have a dilated rectosigmoid. In some, the dilatation of the rectum is present at birth [12], others appear to develop dilatation later in life. The dilatation is only rarely related to stenosis of the bowel outlet [8,28]. Segmental colonic transit time studies in patients with ARM has shown that those with low anomalies have rectosigmoid hypomotility, whereas those with high anomalies have a generalized colonic motility disturbance [22].

It is likely that the surgical method of anorectal reconstruction is a significant prognostic factor. However, this is very difficult to prove since there are no randomized controlled studies. In the literature, comparisons between surgical methods have been performed by comparing the results from different institutions [4,20,71] or by comparison with historical controls within one institution [20,69,72]. Several factors bias both methods. Methods of assessment as well as the age at assessment may vary significantly between institutions. When historical controls are used for comparisons, the age distribution between groups is usually quite different. It is difficult to find reliable criteria to compare bowel function in small children and that of adolescents or adults. Retrospective case note evaluation is unreliable unless strictly defined criteria for long-term bowel function have been used or one single investigator has reviewed all the patients during the entire follow-up period. These conditions are rarely fulfilled. Nevertheless, some authors have made such comparisons. Kiesewetter and Chang [31] found abdominoperineal pull-through to be slightly better than sacroabdominoperineal operation in a series of 78 patients. The age distribution in both groups is not given. Holschneider et al. [69] reported significantly better continence outcome in 21 patients who had PSARP compared with a historical cohort of 16 patients who underwent abdominoperineal pull-through with or without submucosal rectal resection. In a retrospective case note study, Mulder et al. [72] compared 15 patients who had undergone sacroabdominoperineal pull-through with 25 patients who had had PSARP. A good continence outcome was found in 40% of patients in both groups. Templeton and Ditesheim [20] reviewed the outcomes in several series without taking into account the method of continence evaluation or the age at the time of the assessment. They suggested that the use of full-thickness terminal bowel (abdominoperineal pull-through, sacroperineal pull-through) in the anorectal reconstruction might give better long-term outcome than endorectal pull-through procedures (sacroabdominoperineal pull-through, anterior perineal pull-though). In a similar literature review, deVries [71] could not find evidence to support the superiority of any procedure in anorectal reconstruction. However, like Templeton and Ditesheim, he suggested that the preservation of full-thickness terminal bowel gives an individual the best chance for fecal continence.

The age of the patient at the time of the anorectal repair has been suggested to influence the long-term functional outcome. Neonatal abdominoperineal reconstruction of the anorectum was popularized by Rhoads et al. in 1948 [73] and used extensively in the 1950s and 1960s. However, many surgeons were dissatisfied with the functional results following this procedure, therefore a staged procedure became the preferred approach [4,74]. The anorectal reconstruction was usually performed at the age of 6-12 months, following a neonatal colostomy. Recently, early repair of high ARM during the first 3-6 weeks of life has gained popularity [8,75]. Some surgeons advocate neonatal repair [76–80]. Although the operation is technically more demanding, there are some advantages in performing the definitive reconstruction early. The critical anal dilatations are easier to perform in an infant. Moreover, early reconstruction may theoretically allow the early development of neural pathways between the anal canal and the brain, facilitating better anorectal sensation and sphincteric function. However, at present there is no evidence that a neonatal or early repair would provide better functional results than a repair at the age of 6-12 months, although some reports suggest that this might be the case [8,77,81].

28.3.4.2 Long-Term Bowel Function During Childhood

Reported long-term functional outcomes in patients with high ARM are highly variable. Most series grade the results as good, fair, or poor. It has to be remembered that a good outcome does not mean that the patient has a normal bowel function. Patients with a good result have usually been considered socially continent, which implies that the defects in bowel function do not cause significant social disability. Prior to PSARP the reported percentages of patients, evaluated with clinical criteria, with a “good” result varied between 6 and 56%. The percentage of poor results, which means more or less total incontinence, varied between 10 and 70% (Table 28.2). It is unlikely that such a variation in the long-term results would reflect true differences in function. The operative methods used in these series were routine procedures for anorectal reconstruction and the number of patients in each series relatively large, which implies that the reporting authors/centers were experienced in the repair of ARM. The probable explanation for the observed variation is the differences in assessment criteria. The two large series of Templeton and Ditesheim [20] and Rintala et al. [2] both used a quantitative multifactorial evaluation for continence. Both these series identified a lower percentage of poor results than the other series using mainly qualitative criteria. These multifactorial quantitative assessments seemed to grade continence higher than a qualitative clinical assessment, probably because they are a more sensitive index of the patient's social adaptation to abnormal anorectal function [82]. Only Taylor et al. [37] and Rintala et al. [2] have reported the incidence of patients with a completely normal bowel function without soiling or staining in any circumstances, which in both series was only 7.5%. In Rintala's series all such patients had an intermediate anomaly. It is probable that this grim figure reflects the true incidence of unequivocally good long-term outcome in patients treated with abdominoperineal and sacral approaches.

There are still few reports concerning long-term functional outcome following PSARP, and the results have been inconsistent (Table 28.3). Some surgeons report a dismal outcome, with most patients requiring adjunctive measures to maintain social continence [29]. On the other hand, in the series of Peña [8], approximately one-third of the patients with high or intermediate anomalies could be considered as totally continent. In the series of Rintala and Lindahl [28], in which the bowel function of the patients was compared to that of healthy children with a similar age and sex distribution, 35% of the patients had age-appropriate, normal bowel function. A fair outcome, with intermittent soiling requiring frequent change of underwear or protective aids, or poor outcome, with intractable constipation or total incontinence, was found in 30% of the patients.

Many authors report an improvement in fecal continence with increasing age in patients who have undergone abdominoperineal procedure with or without a sacral approach [2,20,31]. In the series of Rintala et al. with similar patients, the frequency of good outcome increased from 35% at 5-10 years of age to 58% at 11 and 15 years of age [2]. The improvement in fecal continence is more clearly shown in series including adolescent or adult patients [31,32]. It is not clear whether this improvement is the result of a true improvement of sphincter function or just adaptation to their handicap. In contrast, reports by Peña, and Rintala and Lindahl [8,28] have shown that in favorable cases, patients who have had PSARP may gain normal or near normal bowel function as early as 3 years of age provided that the inherent functional complications related to the procedure, especially constipation, are treated early and vigorously. In many cases, soiling during the early years after PSARP has been a consequence of severe constipation with overflow incontinence rather than sphincter insufficiency. The treatment of soiling associated with constipation is much more rewarding than treatment of soiling related to sphincter insufficiency. The functional out-

Table 28.2 Functional outcome during childhood – high malformations: before the era of posterior sagittal anorectoplasty (PSARP)

	N	Good	Fair	Poor
Partridge and Gough [55]	63	33%	43%	24%
Trusler and Wilkinson [17]	15	26%	20%	54%
Stephens and Smith [4]	25	56%	32%	12%
Taylor et al. [37]	45	24%	20%	56%
Cywes et al. [45]	38	42%	35%	23%
Smith et al. [88]	18	6%	28%	66%

Table 28.3 Functional outcome during childhood – high malformations: PSARP. *Adolescents

	Total continence	Significant soiling	Constipation
Peña [8]	36%	41%	43%
Rintala and Lindahl [28]	35%	30%	60%
Langemeijer and Molenaar [29]	7%	56%	5%
Rintala and Lindahl [70]*	50%	22%	9%

come following PSARP procedure appears to further improve at adolescence. Rintala and Lindahl [70] reported that 50% of adolescents who had undergone a PSARP procedure were fully continent, and a further 14% had only occasional minor soiling. The improvement was attributed to the disappearance of constipation and overflow soiling at puberty.

28.3.4.3 *Long-Term Bowel Function at Adult Age*

The pediatric surgical literature has only a few reports concerning the functional outcome of high anomalies in adults (Table 28.4). These results, however, illustrate the end-point in the development of bowel function. Moreover, an adult patient as an independent individual may be able to provide a more reliable picture of the overall outcome, including the social consequences of defective bowel function. Nixon and Puri [32] found normal bowel control in 7 (15%) out of 47 adolescent and young adult patients. Twenty-nine patients (62%) had occasional soiling and 11 (23%) frequent soiling or a colostomy. In this series, "normal control" is not defined; moreover, all seven patients who were clean at the time of the study soiled 6-17 years after the reconstruction before achieving continence. More recently, Rintala et al. [83] studied 33 adult patients with a mean age of 35 years, using a questionnaire-based scoring system. Healthy adults with an age and sex distribution similar to the patients were used as controls. None of the patients reached a score indicating normal bowel function, and only 6 (18%) had a good continence score. All controls had good scores and 80% had normal bowel function. Eighteen (54%) patients had fair continence, and 9 (27%) were totally incontinent or had a colostomy. Thirty-one (94%) of the 33 patients reported some degree of fecal soiling. Hassink et al. [84] evaluated 58 patients with a median age of 26 years using a similar scoring method. None of their patients met the criteria for normal bowel habits, 21 (36%) had good continence scores, 25 (43%) scored fair, and 12 (21%) were totally incontinent or had a permanent stoma. About 80% of the patients had soiling. In both these series most patients had undergone an abdominoperineal repair as a primary reconstruction. In the series of Nixon and Puri [32], 68% of the patients had major secondary surgery to improve continence. In the series of Rintala et al. [83] and Hassink et al. [84], 30% and half of the patients had secondary sphincter repairs, respectively.

According to these reports it is obvious that almost all adult patients who have undergone repair of a high ARM using traditional methods (direct perineal, abdominoperineal, or sacroabdominoperineal operation) have some form of fecal incontinence despite secondary sphincter reconstructions. Although many patients report being satisfied with their current level of fecal continence [84], objective evaluation of the data yields a different picture. It is likely that the adult patients have accepted their handicap. The patients have developed measures to cope with unsatisfactory bowel control, such as staying near toilets, wearing liners or diapers, having regular enemas, or having dietary restrictions [83–85].

In adulthood, defective fecal continence has significant social consequences. The main problem is fecal soiling that restricts social activities. In the series of Rintala et al. [83], 85% of the adult patients reported social disability related to soiling. Other problems, especially those that disturb occupational life, were inability to hold back flatus and fecal urgency. Hassink et al. [85] reported that adult patients had a significantly lower educational level than expected. A striking finding in both these series was that after their childhood, most of the adult patients were not followed up by clinicians who were familiar with ARM. Consequently, their medical and social support has not been appropriate and most of the patients had had to attain bowel control by themselves.

28.3.5 Long-Term Problems Related to Associated Malformations

28.3.5.1 *Urinary Tract Problems*

Urinary tract anomalies occur in more than 40% of all patients with ARM. It is not unexpected that urologi-

Table 28.4 Functional outcome in adults – high malformations: operated before the era of PSARP

	N	Normal	Good	Fair	Poor
Hassink et al. [84]	58	0	36%	43%	21%
Rintala et al. [83]	33	0	18%	54%	27%
Nixon and Puri [32]	47	15%	62% (good/fair)		23%

cal long-term morbidity is common. The incidence of long-term urological morbidity is difficult to assess because only a few long -term follow-up reports have specifically addressed urological problems. Late surgery to repair concomitant urogenital anomalies is required in approximately 20% of the patients. There is a significant risk of death from renal failure in patients with ARM. McLorie et al. [14] reviewed a large series of patients and found that death from renal failure occurred in 6.4% of patients with high anomalies and 1.1% of those with low anomalies.

A high incidence of neurovesical dysfunction in patients with ARM has been reported by several authors [13,26,86,87]. Neurovesical dysfunction is usually congenital and often associated with lumbosacral or intraspinal abnormalities [24,87]. This is reflected by the dominance of hyperreflexic findings in cystometry and radiological investigations, suggesting upper motor neuron lesion. Vesicoureteral reflux in patients with ARM is commonly associated with neurovesical dysfunction, and therefore carries a high risk of recurrent urinary infection and subsequent renal damage. Urinary incontinence is related to dysplastic sacrum, urethral and bladder anomalies, and neurovesical dysfunction. Surgical damage to the bladder neck or urethra accounts for a minority of causes of urinary incontinence [24,87].

Table 28.5 Urinary incontinence

	N	Incontinence
Low anomalies		
Peña [8]	14	0%
Rintala and Lindahl [28]	40	0%
Trusler and Wilkinson [17]	20	10%
Smith et al. [88]	29	10%
Rintala et al. [62]	83	11%
High anomalies (before posterior sagittal anorectoplasty)		
Trusler and Wilkinson [17]	15	33%
Wiener and Kiesewetter [24]	90	31%
Smith et al. [88]	18	28%
Rintala et al. [83]	33	33%
Hassink et al. [84]	58	22%
High anomalies (posterior sagittal anorectoplasty)		
Peña [8]	233	10%
Rintala and Lindahl [75]	65	8%

Urinary incontinence is uncommon in patients with low anomalies, which probably reflects the lower incidence of spinal anomalies and neurogenic bladder in these patients. Rintala and Lindahl [28] and Peña [8] reported no cases with urinary incontinence in their recent work; Trusler and Wilkinson [17] and Smith et al. [88] found 10% of their patients to have some degree of urinary incontinence. Rintala et al. [62] reported urinary incontinence in 11% of 83 adult patients with low ARM (Table 28.5).

Urinary incontinence is significantly more common in patients with high anomalies (Table 28.5). Trusler and Wilkinson [17] reported that 5 out of 15 (33%) patients who had undergone abdominoperineal pull-through had urinary incontinence. Smith et al. [88] found urinary incontinence in 5 (28%) out of 18 patients with high anomalies treated by sacroabdominoperineal operation. Wiener and Kiesewetter [24] found neurogenic bladder in 28 (31%) out of 90 children with high malformations, most of whom had urinary incontinence. These figures compare well with the incidence of urinary incontinence among adult patients with high ARM. Rintala et al. [83] found urinary incontinence in 11 (33%) out of 33 adults who were operated on by abdominoperineal or direct perineal procedures during their early childhood, and Hassink et al. [84] reported urinary incontinence in 13 (22%) of their 58 patients operated on by similar methods.

It appears that patients who have had PSARP have lower incidence of neurogenic bladder and urinary incontinence than patients operated on by traditional methods, despite the fact that PSARP requires extensive dissection behind the urethra and bladder neck (Table 28.5). Peña [8] found a 10% incidence of urinary incontinence in 233 children who had PSARP for high or intermediate anomalies including rectovestibular fistulae. The incidence was highest (69%) in patients with high-confluence cloaca. Excluding these patients, urinary incontinence was related to poor sacrum. In the series of Rintala and Lindahl [75], of 65 patients with high or intermediate ARM repaired by internal-sphincter-preserving PSARP, 8% had long-term postoperative urinary incontinence. Urinary incontinence was related to severe sacral anomalies and high cloacal deformities. The incidence of urinary incontinence among patients who had PSARP was lower than in the older series, even if rectovestibular fistulae, which were often classified as low anomalies, are excluded. If vestibular fistulae are excluded, Peña's series reports an incidence of urinary incontinence of 11% and that of Rintala and Lindahl one of 10%.

28.3.5.2 Genital Anomalies, Fertility, and Sexual Problems

Patients with ARM have a high incidence of genital anomalies. Genital anomalies occur in 26% of boys [89], the most common being undescended testes and hypospadias. The most common in girls are vaginal and uterine septation anomalies and vaginal agenesis [90]. Genital tract function has been reported to be impaired in almost half of patients because of vaginal scarring [90]. In sexually active females this often causes coital problems; Matley et al. [91] found dyspareunia in four of nine adult females who had undergone repair of a vestibular anus. Vaginal scarring may interfere with deliveries, and today most females with significant ARM deliver by cesarean section [91,92]. Vaginal deliveries and pregnancies may also worsen fecal continence in patients with a history of vestibular fistula [91,93]. Late gynecological problems are especially common in patients with cloaca. A high percentage of postpubertal females with cloaca develop obstruction of some part of Mullerian structures, with subsequent cystic collections of menstrual blood [94].

There is little information about fertility in patients with ARM. In the series of Rintala et al. of 83 adult patients [62] with low malformations, 47 (57%) had offspring of their own. In the same study, 54% of the healthy controls of similar age and sex distribution had children of their own. On the other hand, in another study from the same institution concerning high malformations, only 39% of the patients had children, which was significantly less than the healthy controls (60%) [83]. In Hendren's [92] large series of cloaca patients, 7 out of 24 adults have had children of their own. Obviously, the low frequency of offspring in patients with high anomalies reflects true infertility in a significant percentage of patients. Ejaculatory duct obstruction has been reported in males [95], some have weak or missing erections, or retrograde ejaculations [83], and some females have Mullerian structure agenesis [90]. On the other hand, some patients may avoid sexual contacts because of defective fecal continence. Rintala et al. [62,83] reported that 20% of patients with high anomalies and 13% of those with low anomalies avoided sexual intercourse because of poor bowel control.

28.3.5.3 Vertebral Anomalies and Myelodysplasias

In the literature there are essentially no reports concerning late problems related to vertebral anomalies in patients with ARM. In the author's consecutive series of 375 ARM during the period 1984-1994, 2 patients have required spinal bracing and 4 required operative spinal stabilization because of progressive scoliosis. A report from the same institution in adults with ARM noted that 18 (16%) out of 116 patients reported symptoms, mainly chronic back pain, related to their vertebral anomalies [62,83].

Recently, much attention has been placed on the occurrence of myelodysplasias in patients with ARM [26,96,97]. The effect of spinal abnormalities, especially tethered cord, on long-term functional outcome in terms of bladder and bowel function or neurological symptoms in the lower extremities is unclear, although some recent reports suggest that worsening of the function due to spinal anomalies is possible. DeGennaro et al. [86] found a higher incidence of spinal dysraphism associated neurovesical dysfunction in older patients than in infants. Sato et al. [97] reported aggravation of neurological symptoms related to tethered cord in a small group of patients with ARM. In the author's series, only 3 out of 375 patients have required detethering because of progressive neurological symptoms, which were completely alleviated only in 1 patient. There appears to be no evidence to support prophylactic detethering of patients who do not have specific symptoms related to tethering [98,99]. There is also no evidence to support the concept that tethered cord affects functional outcome in terms of fecal or urinary continence in patients with ARM [98,100]. Screening for spinal abnormalities for diagnostic purposes is, however, indicated in patients with ARM, including those with low anomalies [52,96]. Normal vertebral anatomy on spinal x-rays does not preclude the presence of spinal cord abnormalities [96]. Screening can be performed by ultrasound during early infancy or by MRI at any age.

28.4 Functional Outcome Following Secondary Surgery for ARM

Secondary reconstructions to improve poor anorectal function have been used extensively in patients with ARM. In most long-term follow-up series extending to adulthood, a significant proportion of patients have undergone redo-surgery [32,83,84]. A clear message arises from these reported series: the long-term outcome is not better in patients who have had secondary surgery [83] and may be worse than in those who had only one reconstruction [84]. It is possible, however, that the patients who have undergone redo-operations had initially worse continence than those with only one operation, but there are no reports that have specifically addressed this question.

Gracilisplasty has been a common method for secondary sphincter reconstruction. Several reports have shown a clear improvement in fecal continence in the short term [101,102]. The improvement in continence is caused by a somewhat increased resting pressure [101] and significantly increased squeeze pressure. In adults who have had gracilisplasty during childhood, the functional results are not very encouraging [32,83,84]. Fecal continence is no better, and may be worse, than in patients with only a primary reconstruction. The main functional limitation of conventional gracilisplasty is that a skeletal muscle like the gracilis muscle can contract only voluntarily. The initially increased tone of a tight muscle wrap around the anus tends to weaken with time [101]. Recently, electrically stimulated gracilisplasty has been used to improve deficient fecal continence in adult patients with ARM. The patients underwent conventional gracilisplasty followed by implantation of a muscle stimulator [103]. After a training period the stimulator was used continuously to maintain constant anal tone. Short-term clinical and manometric results were promising, but after a median follow-up of 4 years only one-third of the patients gained satisfactory fecal continence [104].

Levatorplasty, originally described by Kottmeier and Dzaidiw [105], was popularized as a secondary sphincter reconstruction by Puri and Nixon [106]. Several authors [32,107,108] have published encouraging results. The functional improvement following this procedure has been thought to be related to the creation of an acute anorectal angle, because actual resting or squeeze pressures are not changed at the level of the anal canal. Again, as in patients who had gracilisplasty during childhood, the long-term outcome in adults does not seem to be encouraging. In the author's institution, 15 children with high malformations underwent secondary levatorplasty for poor fecal continence in the late 1970's. At adulthood, only 1 of these 15 patients have gained a satisfactory fecal continence. There does not seem to be any significant differences in fecal continence between those who had secondary levatorplasty and those who have undergone only primary reconstruction [83,84].

Rerouting of the pulled-through bowel has been advocated for patients who have a misplaced anal canal following primary operation [4,109]. In patients with previous abdominoperineal, sacroperineal, or sacroabdominoperineal pull-through, a typical misplacement is anterior to the external sphincter funnel [110–112]. An essentially identical procedure for the rerouting and repair of the muscular anal canal has been suggested by Stephens and Smith, Peña, and Kiesewetter and Jeffries [4,109,113]. The repair is performed through a posterior sagittal sacroperineal incision and includes splitting of the voluntary sphincter muscles in the midline, as in standard PSARP.

Anterior sagittal repair for anterior misplacement has been suggested by Okada et al. [112] and Bass and Yazbeck [114]. The reported outcomes in terms of improved fecal continence have been variable. Following redo PSARP, Peña found a very significant improvement in 52% of his 62 patients, mild improvement in 18%, and no improvement in 12%; the length of follow-up was not stated. The patients who improved had a lesser degree of sacral dysplasia than those who did not improve. Mulder et al. [72] reported that 25% of their 20 patients became continent following this procedure; the mean follow-up period was 3.5 years. Both these series allowed occasional soiling in patients with a good outcome. Brain and Kiely [115] had a success rate of 16% following a relatively short follow-up period. Rintala and Lindahl [111] followed-up 16 patients with redo-PSARP beyond childhood (mean follow-up period 6 years). Although the clinical continence and manometric findings initially improved in 13 of the 16 patients, at adult age only 4 (25%) of the patients could be considered more or less continent, despite a significant increase in both anal resting and squeeze pressures. According to the results of these reports, the role of secondary PSARP in the treatment of fecal incontinence after primary reconstruction of ARM remains to be established.

Late fecal soiling may be related to intractable constipation [8,11,28,116]. This occurs most commonly in patients with a repaired vestibular fistula [2,8,11]. Many of these patients have an adynamic megarectum, which cannot be emptied with medical management or regular enemas. Aganglionosis, although a rare occurrence in patients with ARM, should be ruled out by rectal biopsies. In recalcitrant cases, resection of the dilated distal colon has given favorable results. Peña et al. [117, 118] and Cheu and Grosfeld [116] have used anterior resection with good outcome. Rintala et al. and Powell et al. [2,11] combined resection of the megarectum with endorectal pull-through, which cured the constipation in all of their patients. In the series of Rintala et al., which comprised 13 patients with surgically treated megarectum, 1 had an associated short-segment aganglionosis. The constipation may not be resolved permanently, however; recurrent constipation has been reported following rectosigmoid resection of a megarectum [22].

28.5 Conclusion

Despite significant developments in the understanding of the pathological anatomy and physiology, and innovation of novel surgical techniques, the results of surgical therapy of ARM remain far from perfect. Completely normal bowel function, comparable to that of healthy individuals, is not possible in many patients with high and intermediate malformations. At present it is evident that PSARP and its modifications can improve the dismal outcome found in adult patients who have undergone conventional repairs. It remains to be seen whether novel mini-invasive laparoscopic techniques further improve the promising results that have been achieved with PSARP. Clearly the most important role of a pediatric surgeon in the management of patients with ARM is to perform as optimal repair as possible, taking into account the unique anatomy of each patient. An optimal operation restores normal anatomy and preserves all potential sphincter structures. Secondary surgery for failed or inadequate primary reconstruction is unlikely to provide results that are comparable to those achieved following a successful primary operation. In contrast to more or less commonly accepted earlier assumptions, many patients with low malformations also have functional problems that continue beyond childhood. Therefore, not only the patients with high anomalies, but all patients with ARM need careful follow-up, preferably in a specialized unit, throughout their childhood. Functional complications, especially treatable ones, such as constipation, should be detected and treated early to achieve an optimal outcome. The treatment of defective continence should be commenced well before the child reaches school age to overcome the devastating social consequences of fecal soiling and to integrate the child to the social context of his peers. Because the management of ARM requires years of commitment and special knowledge concerning the anatomical and physiological characteristics of this complex group of congenital malformations, these children should be treated by specialized medical personnel in specialized referral centers.

References

1. Kiesewetter WB, Hoon A (1979) Imperforate anus: an analysis of mortalities during a 25-year period. Prog Pediatr Surg 13:211–220
2. Rintala R, Lindahl H, Louhimo I (1991) Anorectal malformations – results of treatment and long term follow-up of 208 patients. Pediatr Surg Int 6:36–41
3. Hecker WC, Holschneider AM, Kraeft H, et al (1980) Complications, lethality and long-term results after surgery of anorectal atresia. Z Kinderchir 29:238–244
4. Stephens FD, Smith ED (1971) Anorectal Malformations in Children. Year Book Medical Publishers, Chicago
5. Mollitt DL, Malangoni MA, Ballantine TVN (1980) Colostomy complications in children. Arch Surg 115:455–458
6. Wilkins S, Peña A (1988) The role of colostomy in the management of anorectal malformations. Pediatr Surg Int 3:105–109
7. Gardikis S, Antypas S, Mamoulakis C, et al (2004) Colostomy type in anorectal malformations: 10 years experience. Minerva Pediatr 56:425–429
8. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
9. Guttman M, Laberge Y, Yazbeck S (1988) Anterior perineal approach for high imperforate anus using the Mollard technique. In: Stephens FD, Smith ED (eds) Anorectal Malformations in Children: Update 1988. Birth defects: Original article series, vol 24, Number 4, pp 349–355
10. Kiesewetter WB (1979) Rectum and anus: malformations. In: Ravitch MM, Welch KJ, Benson CD, Aberdeen E, Randolph JG (eds) Pediatric Surgery, 3rd edn. Year Book Medical Publishers, Chicago, London, pp 1059–1072
11. Powell RW, Sherman JO, Raffensperger JG (1982) Megarectum: a rare complication of imperforate anus repair and its surgical correction by endorectal pull-through. J Pediatr Surg 17:786–795
12. Brent L, Stephens FD (1976) Primary rectal ectasia: a quantitative study of smooth muscle cells in normal and hypertrophied human bowel. Prog Pediatr Surg 9:41–62
13. Sheldon CA, Gilbert A, Lewis AG, et al (1994) Surgical implications of genitourinary anomalies in patients with imperforate anus. J Urol 152:196–199
14. McLorie GA, Sheldon CA, Fleischer M, et al (1987) The genitourinary system in patients with imperforate anus. J Pediatr Surg 22:1100–1104
15. Rickwood AM, Spitz L (1980) Primary vesicorureteric reflux in neonates with imperforate anus. Arch Dis Child 55:149–150

16. Vinnicombe SJ, Good CD, Hall CM (1996) Posterior urethral diverticula: a complication of surgery for high anorectal malformations. Pediatr Radiol 26:120–126
17. Trusler GA, Wilkinson RH (1962) Imperforate anus: a review of 147 cases. Can J Surg 5:269–277
18. Hong AR, Acuna MF, Peña A, et al (2002) Urologic injuries associated with repair of anorectal malformations in male patients. J Pediatr Surg 37:339–344
19. Boemers TM, Bax KM, Rovekamp MH, van Gool JD (1995) The effect of posterior anorectoplasty and its variants on lower urinary tract function in children with anorectal malformations. J Urol 153:191–193
20. Templeton JM, Ditesheim JA (1985) High imperforate anus – quantitative result of long-term fecal continence. J Pediatr Surg 20:645–652
21. Rintala R, Lindahl H, Marttinen E, Sariola H (1993) Constipation is a major functional complication after internal sphincter-saving posterior sagittal anorectoplasty for high and intermediate anorectal malformations. J Pediatr Surg 28:1054–1058
22. Rintala R, Marttinen E, Virkola K, et al (1997) Segmental colonic motility in patients with anorectal malformations. J Pediatr Surg 32:453–456
23. Yeung CK, Kiely EM (1991) Low anorectal anomalies: a critical appraisal. Pediatr Surg Int 6:333–335
24. Wiener ES, Kiesewetter WB (1973) Urologic abnormalities associated with imperforate anus. J Pediatr Surg 8:151–158
25. Boemers TM, de Jong TP, van Gool JD, Bax KM (1996) Urologic problems in anorectal malformations. Part 2: functional urologic sequelae. J Pediatr Surg 31:634–637
26. Rivosecchi M, Lucchetti MC, De Gennaro M, et al (1995) Spinal dysraphism detected by magnetic resonance imaging in patients with anorectal anomalies: incidence and clinical significance. J Pediatr Surg 30:488–490
27. Mosiello G, Capitanucci ML, Gatti C, et al (2003) How to investigate neurovesical dysfunction in children with anorectal malformations. J Urol 170:1610–1613
28. Rintala R, Lindahl H (1995) Is normal bowel function possible after repair of intermediate and high anorectal malformations. J Pediatr Surg 30:491–494
29. Langemeijer RATM, Molenaar JC (1991) Continence after posterior sagittal anorectoplasty. J Pediatr Surg 26:587–590
30. Bliss DP, Tapper D, Anderson JM, et al (1996) Does posterior sagittal anorectoplasty in patients with high imperforate anus provide superior fecal continence. J Pediatr Surg 31:26–32
31. Kiesewetter WB, Chang JHT (1977) Imperforate anus: a five- to thirty-year follow-up perspective. Prog Pediatr Surg 10:110–120
32. Nixon HH, Puri P (1977) The results of treatment of anorectal anomalies: a thirteen to twenty year follow-up. J Pediatr Surg 12:27–37
33. Swenson O, Donnellan WL (1967) Preservation of the puborectalis sling in imperforate anus repair. Surg Clin North Am 47:173–193
34. Hedlund H, Peña A, Rodriquez G, et al (1992) Long-term anorectal function in imperforate anus treated by a posterior sagittal anorectoplasty: manometric investigation. J Pediatr Surg 27:906–909
35. Doolin EJ, Black CT, Donaldson JS, et al (1991) Rectal manometry, computed tomography and functional results of anal atresia surgery. J Pediatr Surg 28:195–198
36. Rintala R (1990) Postoperative internal sphincter function in anorectal malformations – a manometric study. Pediatr Surg Int 5:127–130
37. Taylor I, Duthie HL, Zachary RB (1973) Anal continence following surgery for imperforate anus. J Pediatr Surg 8:497–503
38. Iwai N, Hashimoto K, Goto Y, et al (1984) Long term results after surgical correction of anorectal malformations. Z Kinderchir 39:35–39
39. Rintala RJ, Lindahl HG (1999) Posterior sagittal anorectoplasty is superior to sacroperineal-sacroabdominoperineal pull-through: a long-term follow-up study in boys with high anorectal anomalies. J Pediatr Surg 34:334–337
40. Molander ML, Frenckner B (1985) Anal Sphincter function after surgery for high imperforate anus – a long term follow-up investigation. Z Kinderchr 40:91–96
41. Iwai N, Kaneda H, Taniguchi T, et al (1985) Postoperative continence assessed by electromyography of the external sphincter in anorectal malformations. Z Kinderchir 40:87–90
42. Boyd SG, Kiely EM, Swash M (1987) Electrophysiological studies of puborectalis and external anal sphincter in incontinent children with corrected high ano-rectal anomalies. Pediatr Surg Int 2:110–112
43. Shandling B, Gilmour R, Ein S (1991) The anal sphincter force in the evaluation of postoperative imperforate anus. J Pediatr Surg 26:1369–1371
44. Kelly JH (1969) Cine radiography in anorectal malformations. J Pediatr Surg 4:538–546
45. Cywes S, Cremin BJ, Louw JH (1972) Assessment of continence after treatment for anorectal agenesis: a clinical and radiologic correlation. J Pediatr Surg 6:132–137
46. Yagi M, Iwafuchi M, Uchiyama M, et al (2001) Postoperative fecoflowmetric analysis in patients with anorectal malformation. Surg Today 31:300–307
47. Emblem R, Diseth T, Morkrid L (1997) Anorectal anomalies: anorectal manometric function and anal endosonography in relation to functional outcome. Pediatr Surg Int 12:516–519
48. Jones NM, Humphreys MS, Goodman TR (2003) The value of anal endosonography compared with magnetic resonance imaging following the repair of anorectal malformations. Pediatr Radiol 33:183–185
49. Ong NT, de Campo M, Fowler R (1990) Computerised tomography in the management of imperforate anus patients following rectoplasty. Pediatr Surg Int 5:241–245

50. Fukuya T, Honda H, Kubota M, et al (1993) Postoperative MRI evaluation of anorectal malformations with clinical correlation. Pediatr Radiol 23:583–586
51. Wong KK, Khong PL, Lin SC (2005) Postoperative magnetic resonance evaluation of children after laparoscopic anorectoplasty for imperforate anus. Int J Colorectal Dis 20:33–37
52. Beek FJA, Boemers TML, Witkamp TD, et al (1995) Spine evaluation in children with anorectal malformations. Pediatr Radiol 25:S28–S32
53. Stephens FD, Smith ED (1986) Classification, identification and assessment of surgical treatment of anorectal anomalies. Pediatr Surg Int 1:200–205
54. Holschneider AM (1983) Elektromanometrie des Enddarms. Urban and Schwarzenberg Munich-Wien-Baltimore, pp 213–218
55. Partridge JP, Gough MH (1961) Congenital abnormalities of the anus and rectum. Br J Surg 49:37–50
56. Scharli AF, Kiesewetter WB (1969) Imperforate anus: anorectosigmoid pressure studies as a quantitative evaluation of postoperative continence. J Pediatr Surg 4:694–704
57. Rintala RJ, Lindahl HG, Rasanen M (1997) Do children with repaired low anorectal malformations have normal bowel function. J Pediatr Surg 32:823–826
58. Ong NT, Beasley SW (1990) Long-term functional results after perineal surgery for low anorectal anomalies. Pediatr Surg Int 5:238–240
59. Laboure S, Besson R, Lamblin MD, et al (2000) Incontinence and constipation after low anorectal malformations in a boy. Eur J Pediatr Surg 10:23–29
60. Javid PJ, Barnhart DC, Hirschl RB, et al (1998) Immediate and long-term results of surgical management of low imperforate anus in girls. J Pediatr Surg 33:198–203
61. Karkowski J, Pollock WF, Landon CW (1973) Imperforate anus. Eighteen to thirty year follow-up study. Am J Surg 126:141–147
62. Rintala R, Mildh L, Lindahl H (1992) Fecal continence and quality of life in adult patients with an operated low anorectal malformation. J Pediatr Surg 27:902–905
63. Lamprecht W, Lierse W (1987) The internal sphincter in anorectal malformations: morphologic investigations in neonatal pigs. J Pediatr Surg 22:1160–1168
64. Kluth D, Hillen M, Lamprecht W (1995) The principles of normal and abnormal hindgut development. J Pediatr Surg 30:1143–1147
65. Rintala R, Lindahl H, Sariola H, et al (1990) The rectourogenital connection in anorectal malformations is an ectopic anal canal. J Pediatr Surg 25:665–668
66. Husberg B, Lindahl H, Rintala R, Frenckner B (1992) High and intermediate imperforate anus: Results after surgical correction with special respect to internal sphincter function. J Pediatr Surg 27:185–189
67. Mollard P, Meunier P, Mouriquand P, et al (1991) High and intermediate imperforate anus: functional results and postoperative manometric assessment. Eur J Pediatr Surg 1:282–286
68. Gil-Vernet JM, Asensio M, Marhuenda C, et al (2001) Nineteen years experience with posterior sagittal anorectoplasty as a treatment of anorectal malformation. Cir Pediatr 14:108–111
69. Holschneider AM, Pfrommer W, Gerresheim B (1994) Results in the treatment of anorectal malformations with special regard to the histology of the rectal pouch. Eur J Pediatr Surg 4:303–309
70. Rintala RJ, Lindahl HG (2001) Fecal continence in patients having undergone PSARP procedure for a high anorectal malformation improves at adolescence as constipation disappears. J Pediatr Surg 36:1218–1221
71. de Vries P (1988) Results of treatment and their assessment. In: Stephens FD, Smith ED (eds) Anorectal Malformations in Children: Update 1988. Birth defects: original article series, Vol 24, Number 4. Alan R. Liss, New York, pp 481–500
72. Mulder W, deJong E, Wauters I, et al (1995) Posterior sagittal anorectoplasty: functional results of primary and secondary operations in comparison to the pull-through method in anorectal malformations. Eur J Pediatr Surg 5:170–173
73. Rhoads JE, Pipes RL, Randall JP (1948) A simultaneous abdominal and perineal approach in operations for imperforate anus with atresia of the rectum and rectosigmoid. Ann Surg 12:552–556
74. Kiesewetter WB (1967) Imperforate anus II. The rationale and technique of the sacro-abdomino-perineal operation. J Pediatr Surg 2:106–110
75. Rintala R, Lindahl H (1995) Internal sphincter saving PSARP for high and intermediate anorectal malformations – technical considerations. Pediatr Surg Int 10:345–349
76. Moore TC (1990) Advantages of performing the sagittal anoplasty operation for imperforate anus at birth. J Pediatr Surg 25:276–277
77. Aluwihare APR (1990) Primary perineal rectovaginoplasty for supralevator imperforate anus in female infants. J Pediatr Surg 24:278–281
78. Goon HK (1990) Repair of anorectal anomalies in the neonatal period. Pediatr Surg Int 5:246–249
79. Liu G, Yuan J, Geng J, et al (2004) The treatment of high and intermediate anorectal malformations: one stage or three procedures? J Pediatr Surg 39:1466–1471
80. Albanese CT, Jennings RW, Lopoo JB, et al (1999) One-stage correction of high imperforate anus in male neonate. J Pediatr Surg 34:834–836
81. Freeman NV, Bulut M (1986) "High" anorectal anomalies treated by early "neonatal" operation. J Pediatr Surg 21:218–220
82. Ong N, Beasley SW (1990) Comparison of clinical methods for the assessment of continence after repair of high anorectal anomalies. Pediatr Surg Int 5:233–237
83. Rintala R, Mildh L, Lindahl H (994) Fecal continence and quality of life in adult patients with an operated high or intermediate anorectal malformation. J Pediatr Surg 29:777–780

84. Hassink EA, Rieu PN, Severijnen RS, et al (1993) Are adults content or continent after repair for high anal atresia? A long-term follow-up study in patients 18 years of age and older. Ann Surg 218:196–200
85. Hassink EA, Rieu PN, Brugman AT, et al (1994) Quality of life after operatively corrected high anorectal malformation. A long-term follow-up study in patients 18 years of age and older. J Pediatr Surg 29:773–776
86. De Gennaro M, Rivosecchi M, Lucchetti MC, et al (1994) The incidence of occult spinal dysraphism and the onset of neurovesical dysfunction in children with anorectal anomalies. Eur J Pediatr Surg 4:12–14
87. Ralph DJ, Woodhouse CRJ, Ransley PG (1992) The management of the neuropathic bladder in adolescents with imperforate anus. J Urol 148:366–368
88. Smith EI, Tunell WP, Williams GR (1978) A clinical evaluation of the surgical treatment of anorectal malformations (imperforate anus). Ann Surg 187:583–591
89. Metts JC, Kotkin L, Kasper S, et al (1997) Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. J Urol 158:1298–300
90. Hall R, Fleming S, Gysler M, et al (1985) The genital tract in female children with imperforate anus. Am J Obstet Gynecol 151:169–171.
91. Matley PJ, Cywes S, Berg A, et al (1990) A 20-year follow-up of children born with vestibular anus. Pediatr Surg Int 5:37–40
92. Hendren WH (1998) Cloaca, the most severe degree of imperforate anus: experience with 195 cases. Ann Surg 228:331–346
93. Rintala R, Luukkonen P, Järvinen HJ (1989) Surgical repair of vulvar anus in adults. Int J Colorect Dis 4:244–246
94. Levitt MA, Stein DM, Peña A (1998) Gynecologic concerns in the treatment of teenagers with cloaca. J Pediatr Surg 33:188–193
95. Pryor JP, Hendry WF (1991) Ejaculatory duct obstruction in subfertile males: analysis of 87 patients. Fertil Steril 56:725–730
96. Davidoff AM, Thompson CV, Grimm JK, et al (1991) Occult spinal dysraphism in patients with anal agenesis. J Pediatr Surg 26:1001–1005
97. Sato S, Shirane R, Yoshimoto T (1993) Evaluation of tethered cord syndrome associated with anorectal malformations. Neurosurgery 32:125–127
98. Levitt MA, Patel M, Rodriguez G, et al (1997) The tethered spinal cord in patients with anorectal malformations. J Pediatr Surg 32:462–468
99. Tuuha SE, Aziz D, Drake J, et al (2004) Is surgery necessary for asymptomatic tethered cord in anorectal malformation patients? J Pediatr Surg 39:773–777
100. Taskinen S, Valanne L, Rintala R (2002) Effect of spinal cord abnormalities on the function of the lower urinary tract in patients with anorectal abnormalities. J Urol 168:1147–1149
101. Holschneider AM, Pöschl U, Hecker WC (1979) Pickrell's gracilis muscle transplantation and its effect on anorectal continence. A five year prospective study. Z Kinderchir 27:135–143
102. Raffensperger J (1979) The gracilis sling for faecal incontinence. J Pediatr Surg 14:794–797
103. Baeten CGMI, Konsten J, Heineman E, et al (1994) Dynamic graciloplasty for anal atresia. J Pediatr Surg 29:922–925
104. Koch SM, Uludag O, Rongen MJ, et al (2004) Dynamic graciloplasty in patients born with an anorectal malformation. Dis Colon Rectum 47:1711–9
105. Kottmeier PK, Dziadiw R (1967) The complete release of the levator ani sling in fecal incontinence. J Pediatr Surg 2:111–117
106. Puri P, Nixon HH (1976) Levatorplasty: A secondary operation for fecal incontinence following primary operation for anorectal agenesis. J Pediatr Surg 11:77–82
107. Ninan GK, Puri P (1994) Levatorplasty using a posterior sagittal approach in secondary faecal incontinence. Pediatr Surg Int 9:17–20
108. Kottmeier PK, Velcek FT, Klotz DH, et al (1986) Results of levatorplasty for anal incontinence. J Pediatr Surg 21:647–650
109. Peña A (1983) Posterior sagittal anorectoplasty as a secondary operation for the treatment of fecal incontinence. J Pediatr Surg 18:762–772
110. Peña A (1988) Surgical management of anorectal malformations: a unified concept. Pediatr Surg Int 3:82–93
111. Rintala R, Lindahl H (1995) Secondary PSARP for anorectal malformations – a long term follow-up extending beyond childhood. Pediatr Surg Int 10:414–417
112. Okada A, Tamada H, Tsuji H, et al (1993) Anterior sagittal anorectoplasty as a redo-operation for imperforate anus. J Pediatr Surg 28:933–938
113. Kiesewetter WB, Jeffries R (1981) Secondary anorectal surgery for the missed puborectalis muscle. J Pediatr Surg 16:921–925
114. Bass J, Yazbeck S (1987) Reoperation by anterior perineal approach for missed puborectalis. J Pediatr Surg 22:761–763
115. Brain AJL, Kiely EM(1989) Posterior sagittal anorectoplasty for reoperation in children with anorectal malformations. Br J Surg 76:57–59
116. Cheu HW, Grosfeld JL (1992) The atonic baggy rectum: a cause of intractable obstipation after imperforate anus repair. J Pediatr Surg 27:1071–1074
117. Peña A, El behery M (1993) Megasigmoid: a source of pseudoincontinence in children with repaired anorectal malformations. J Pediatr Surg 28:199–203
118. Peña A, Hong AR, Midulla P, et al (2003) Reoperative surgery for anorectal anomalies. Semin Pediatr Surg 12:118–123

29 Treatment of Fecal Incontinence

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29.1 Introduction

Most patients who undergo repair of an anorectal malformation (ARM) suffer from a degree of functional defecating disorder, and all suffer from an abnormality in their fecal continence mechanism. Approximately 25% of patients are deficient enough in these mechanisms that they are fecally incontinent and cannot have a voluntary bowel movement. The others are capable of having voluntary bowel movements, but may require treatment of an underlying dysmotility disorder, which manifests as constipation. This management is discussed in Chaps. 29, 30 and 33.

Fecal incontinence represents a devastating problem that often prevents a person from becoming socially accepted, which in turn provokes serious psychological sequelae. These patients require an artificial way to keep them clean and in normal underwear, a regimen termed bowel management.

29.2 Fecal Continence

Fecal continence depends on three main factors: (1) voluntary sphincter muscles, (2) anal canal sensation, and (3) colonic motility [1].

29.2.1 Voluntary Sphincter Muscles

The voluntary muscle structures are represented by the levators, the muscle complex, and external sphincter. They are normally used only for brief periods, when the rectal fecal mass reaches the anorectal area, pushed by the involuntary peristaltic contraction of the rectosigmoid motility. This voluntary contraction occurs only in the minutes prior to defecation, and these muscles are used only occasionally during the rest of the day and night. Patients with ARM have abnormal voluntary striated muscles with different degrees of hypodevelopment. However, voluntary muscles can be used only when the patient feels that it is necessary to use them. For that sensation, the patient needs information that can only be derived from an intact anal sensory mechanism, a mechanism that many patients with ARM lack.

29.2.2 Anal Canal Sensation

Exquisite sensation in normal individuals resides in the anal canal. Except for patients with rectal atresia, most patients with ARM are born without an anal canal; therefore, sensation either does not exist or is rudimentary. It does seem that patients can perceive distention of the rectum, but this requires that the rectum has been properly located within the muscle structures. This sensation seems to be a consequence of stretching of the voluntary muscle (proprioception). The most important clinical implication of this is that liquid stool or soft fecal material may not be felt by the patient with ARM, as it does not distend the rectum. Thus, to achieve some degree of sensation and bowel control, the patient must have the capacity to form solid stool.

29.2.3 Bowel Motility

Perhaps the most important factor in fecal continence is bowel motility; however, the impact of motility has been largely underestimated. In a normal individual, the rectosigmoid remains quiet for variable periods of time (one to several days), depending on specific defecation habits. During that time, sensation and voluntary muscle structures are almost unnecessary because the stool, if it is solid, remains inside the colon. The peristaltic contraction of the rectosigmoid that occurs prior to defecation is normally felt by the patient. The normal individual can voluntarily relax the striated muscles, which allows the rectal contents to migrate down into the highly sensitive area of the anal canal. There, accurate information is provided concerning the consistency and quality of the stool. The voluntary muscles are used to push the rectal contents back up into the rectosigmoid and to hold them, if desired, until the appropriate time for evacuation. At the time of defecation, the voluntary muscle structures relax.

The main factor that provokes the emptying of the rectosigmoid is a massive involuntary peristaltic contraction that is sometimes helped by a Valsalva maneuver. Most patients with ARM suffer from disturbance of this sophisticated bowel motility mechanism. Patients who have undergone a PSARP or any other type of sacroperineal approach, in which the most distal part of the bowel was preserved, show evidence of an overefficient bowel reservoir (megarectum). The main clinical manifestation of this is constipation, which seems to be more severe in patients with lower defects (see Chaps. 29 and 30) [2].

Constipation that is not aggressively treated, in combination with an ectatic distended colon (sometimes associated with a loop colostomy that allows fecal impaction in the blind rectal pouch), eventually leads to severe constipation, and a vicious cycle ensues, with worsening constipation leading to more rectosigmoid dilation, leading to worse constipation. The enormously dilated rectosigmoid, with normal ganglion cells, behaves like a myopathic type of hypomotile colon [1]. For these patients who are fecally incontinent, a daily enema successfully cleans the colon, which then remains clean until the next enema [3].

Those patients treated with techniques in which the most distal part of the bowel was resected behave clinically as individuals without a rectal reservoir. This is a situation equivalent to a perineal colostomy. Depending on the amount of colon resected, the patient may have loose stools. In these cases, medical management consisting of enemas plus a constipating diet, and medications to slow down the colonic motility is indicated.

29.3 Bowel Management Program

The bowel management program consists of teaching the patient or his/her parents how to clean the colon once daily so as to stay completely clean in the underwear for 24 h. This is achieved by keeping the colon quiet in between enemas. The program, although simplistic, is implemented by trial and error over a period of 1 week. The patient is seen each day and an x-ray film of the abdomen is taken so that they can be monitored on a daily basis for the amount and location of any stool left in the colon as well as the presence of stool in the underwear. The decision as to whether the type and/or quality of the enemas should be modified as well as changes in their diet and/or medication can be made [3].

It is important to differentiate real fecal incontinence from overflow pseudoincontinence. In patients with real fecal incontinence, the normal mechanism of bowel control is deficient for the reasons described. Pseudoincontinence occurs when a patient behaves like they are fecally incontinent, but really have severe constipation and overflow soiling. Once the disimpaction is treated and the patient receives enough laxatives so as to avoid constipation, he/she becomes continent. This patient group is described in Chaps. 29 and 30. It is extremely important to distinguish between real incontinence and pseudoincontinence in order to identify the origin of the problem and consequently to plan the best treatment.

Of all children with ARM who have undergone a correct and successful operation, 75% have voluntary bowel movements after the age of 3 years [2]. About half of these patients soil their underwear on occasion. Those episodes of soiling are usually related to constipation. When the constipation is treated properly, the soiling frequently disappears. Thus, approximately 40% of all children have voluntary bowel movements and no soiling. In other words, they behave like normal children. Children with good bowel control may still suffer from temporary episodes of fecal incontinence, especially when they experience severe diarrhea. Some 25% of all children suffer from real fecal incontinence, and these are the patients who must receive bowel management to keep them clean.

The surgeon should be able to predict in advance which children have good functional prognosis and which children have a poor prognosis. Table 29.1 shows the most common indicators of good and poor prognosis. After the main repair and the colostomy closure, it is possible to establish the functional prog-

Table 29.1 Prognostic signs

Good prognosis signs:	Bad prognosis signs:
• Good Bowel movement patterns: 1–2 bowel movement per day – no soiling in between	• Constant soiling and passing stool
• Evidence of sensation when passing stool (pushing, making faces)	• No sensation (no pushing)
• Urinary control	• Urinary incontinence, dribbling of urine

Table 29.2 Predictors of prognosis

Indicators of good prognosis for bowel control	Indicators of poor prognosis for bowel control
• Normal sacrum	• Abnormal sacrum
• Prominent midline groove (good muscles)	• Flat perineum (poor muscles)
• Some types of anorectal malformations: - Rectal atresia - Vestibular fistula - Imperforate anus without a fistula - Cloacas with a common channel < 3 cm - Less complex malformations: perineal fistula	• Some types of anorectal malformations: - Rectobladderneck fistula - Cloacas with a common channel > 3 cm - Complex malformations

nosis (Table 29.2). Parents must be realistically informed as to their child's chances for bowel control, avoiding needless frustration later on. It is imperative to establish the functional prognosis of each child as early as possible, sometimes even in the newborn period, in order to avoid creating false expectations for the parents. Once the diagnosis of the specific defect is established, the functional prognosis can be predicted. If the child's defect is of a type associated with good prognosis, such as a vestibular fistula, perineal fistula, rectal atresia, rectourethral bulbar fistula, or imperforate anus with no fistula, one should expect that the child would have voluntary bowel movements by the age of 3 years. These children will still need supervision to avoid fecal impaction, constipation, and soiling (see Chaps. 29, 30 and 33).

If the child's defect is of the type associated with a poor prognosis, for example, a very high cloaca with a common channel longer than 3 cm, a rectobladderneck fistula, or if they have a very hypodeveloped sacrum or associated spinal anomalies the parents must understand that their child will most likely need a bowel management program to remain clean. This should be implemented when the child is 3 or 4 years old, before he/she begins spending a great deal of time away from home. Children with rectoprostatic fistula have an almost 50:50 chance of having voluntary bowel movements or of being incontinent. In these children, an attempt should be made to achieve toilet training by the age of 3 years. If this proves to be unsuccessful, bowel management should be implemented. Each summer, during school vacation, reattempts can be made to assess the child's ability to potty train.

In patients previously operated on for an ARM with fecal incontinence, a reoperation with the hope of obtaining good bowel control can be considered if the child was born with a good sacrum, good sphincter mechanism, a malformation with good functional prognosis, and the rectum is mislocated (see Chap. 24). A redo posterior sagittal anorectoplasty (PSARP) can be performed and the rectum can be relocated within the limits of the sphincter mechanism. Approximately 50% of the children operated on under these very specific circumstances have a significant improvement in bowel control [4].

Patients with fecal incontinence and a tendency toward constipation cannot be treated with laxatives, but instead need bowel management for fecal incontinence. Such children are usually those born with a defect that carries a bad prognosis and severe associated defects (e.g., defect of the sacrum, poor muscle complex).

Children operated on for ARM that suffer from fecal incontinence can be divided into two well-defined groups that require individualized treatment plans: (1) those with constipation (colonic hypomotility), and (2) children with loose stools and diarrhea.

29.3.1 Children with Constipation (Colonic Hypomotility)

In these children the motility of the colon is significantly reduced. The basis of the bowel management program is thus to teach the parents to clean the child's colon once a day with a suppository, an enema, or colonic irrigation. No special diet or medications are necessary in these cases. The fact that they suffer from constipation (hypomotility) is very helpful, as it guarantees that they will remain clean in between enemas. The real challenge is to find an enema that is capable of cleaning the colon completely. Definitive evidence that the colon is truly empty following the enema requires a plain abdominal radiograph. Soiling episodes or severe "accidents" occur when there is an incomplete cleaning of the bowel, with feces that progressively accumulates.

29.3.2 Children with Loose Stools and Diarrhea

The great majority of children who suffer from this kind of problem were operated on before 1980, prior to the introduction of the PSARP technique. In those years the techniques frequently included resection of the rectosigmoid (reservoir) [5,6]. Therefore, this group of children have overactive colons. Rapid transit of stool results in frequent episodes of diarrhea. This means that even when an enema cleans their colon rather easily, stool keeps passing fairly quickly from the cecum to the descending colon and the anus. To prevent this, a constipating diet and/or medications to slow down the colon are necessary. Eliminating foods that further loosen bowel movements will help the colon to slow down. Some children, however, may have in addition an "irritable" colon, which makes the management more challenging.

29.4 Evaluation and Treatment

The keys to success of the bowel management program are dedication and sensitivity from the medical team. The basis of the program is to clean the colon and keep it quiet, and thus the patient remains clean for the 24 h after the enema, colonic irrigation, or suppository that is given once a day. Sometimes manipulation of diet and medication are utilized for patients with a hypermotile colon.

The program is an ongoing process that is responsive to the individual patient and differs for each child. The program is usually successful within a week, during which family, patient, physician, and nurse undergo a process of trial and error, tailoring the program to the specific patient. More than 90% of the children who follow this program are artificially clean and dry for the whole day and can lead a completely normal life. It is unacceptable to send a child with fecal incontinence to school in diapers when his classmates are already toilet trained, and proper treatment to prevent this is perhaps more important than the surgical procedure itself. Children who require diapers or who have accidents while in school because of fecal incontinence are exposed to ridicule from their peers, which can lead to adverse psychological sequelae.

The first step is to perform a contrast enema study with hydrosoluble material. The study should never be done with barium; it is also important to obtain a picture after the evacuation of the contrast material. This study shows the type of colonic motility, hypomotility – constipated (Fig. 29.1) or hypermotility (Fig. 29.2).

The bowel management program is then implemented according to the patient's type of colon, and the results are evaluated every day. Changes in the volume and content of the enemas are made until the colon is successfully cleaned. For this, an x-ray film of the abdomen, taken every day, is invaluable in determining whether the colon is empty.

29.4.1 Types of Enema

There are different types of solutions to use for enemas: there are some ready-made solutions that can be bought in a pharmacy or drugstore, or solutions that can be prepared at home based on water and salt (0.9% saline can be made by adding 3–4 teaspoons to 1 liter of water). The use of phosphate enemas is most convenient since it is already in a prepared vial. However, saline enemas are often just as effective and some families find it easier and less expensive. Occasionally, children will complain of cramping with the phosphate enema, but have no complaints with the saline one. Children older than 8 years of age or heavier than 30 kg may receive one adult phosphate enema daily (240cc). Children between 3 and 8 years of age or between 15 and 30 kg in body weight may receive one pediatric phosphate enema each day (120cc). Children should never receive more than one phosphate enema a day because of the risk of phosphate intoxication, and others with impaired renal function should use these enemas with caution.

Fig. 29.1 Contrast enema of megarectosigmoid (Reprinted from Current Problems in Surgery, 39, Peña A., Levitt M. Colonic Inertia Disorders in Pediatrics, p 681, Mosby (2002), with permission from Elsevier.)

Fig. 29.2 Contrast enema in a patient who has had their rectosigmoid resected (Reprinted from Current Problems in Surgery, 39, Peña A., Levitt M. Colonic Inertia Disorders in Pediatrics, p 695, Mosby (2002), with permission from Elsevier.)

The enema administered on a regular basis should result in a bowel movement followed by a period of 24 h of complete cleanliness. If one enema is not enough to clean the colon (as demonstrated by an x-ray, or if the child keeps soiling), then the child requires a more aggressive treatment, and a saline enema is added to the phosphate one. If the addition of the saline enema still results in inadequate results, then glycerin can be added, or high colonic washings with a balloon catheter may help. The "right" enema is the one that can empty the child's colon and allow him to stay clean for the following 24 h. This can be achieved only by trial and error and learning from previous attempts.

Children with loose stool have an overactive colon and most of the time they do not have a reservoir. This means that even when an enema cleans their colon rather easily, new stool passes quickly from the cecum to the descending colon and the anus. To prevent this, a constipating diet and/or medications (such as loperamide) to slow down the colon are recommended. Eliminating foods that loosen bowel movements will help the colon to move slowly.

Parents are provided with a list of constipating type of foods to be given and a list of laxative foods to be avoided. The diet is very rigid (e.g., banana, apple, baked bread, white pasta with no sauce, boiled meat). Fried foods and dairy products must be avoided (Table 29.3). Most parents know which meals provoke diarrhea and which constipate their child. To determine the right combination, the treatment starts with enemas, a very strict diet, and loperamide. Most children respond to this aggressive management

Table 29.3 Constipating foods

Constipating diet	
No	**Yes**
Milk or milk products	Apple sauce
Fats	Apple without skin
Fried foods	Rice
Fruits	White bread
Vegetables	Bagels
Spices	Boiled, broiled, baked meat, chicken or fish
Fruit juices	Soft drinks
French fries	Banana
Chocolate	Pasta
	Pretzels
	Tea
	Potato
	Jelly (no jam)

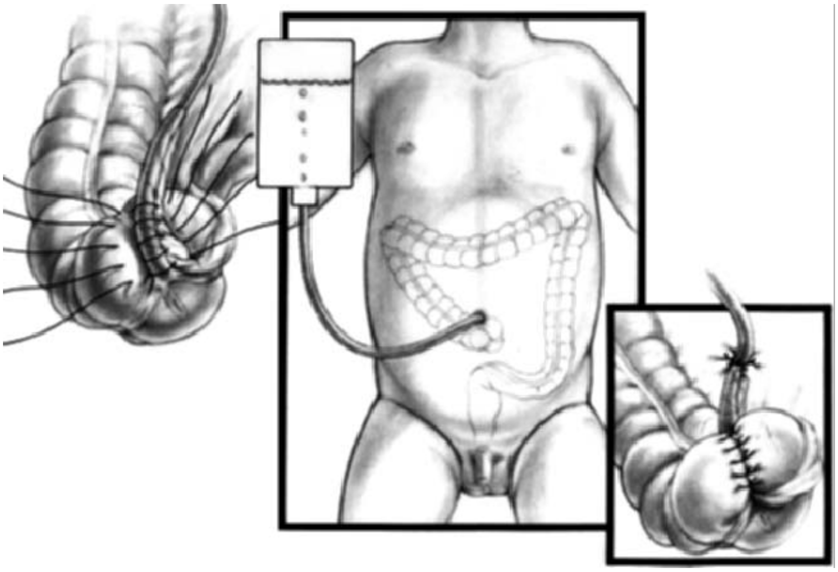

Fig. 29.3 Malone appendicostomy (Reprinted from J Pediatr Surg, 32, Levitt MA, Soffer SZ, Peña A Continent Appendicostonomy in the Bowel Management of Fecal Incontinent Children, p 1631, Elsevier (1997), with permission of Elsevier.)

within a few days. The child should remain on a strict diet until clean for 24 h for 2–3 days in a row. The child can then choose one new food every 2–3 days, observing the effect on his/her colonic activity. If the child soils after eating a newly introduced food, that food must be eliminated from the diet. Over several months, the most liberal diet possible should be sought for the child. If he or she remains clean with a liberal diet, the dose of the medication can gradually be reduced to the lowest effective dose to keep the child clean for 24 h.

In children in whom a successful bowel management program has been implemented, the parents frequently ask if this program will be needed for life. The answer is "yes" for those patients born with a poor prognosis type of defect. However, since we are dealing with a spectrum of defects, there are patients with some degree of bowel control. These patients are subjected to the bowel management program in order not to be exposed to embarrassing accidents of uncontrolled bowel movements. However, as time goes by the child becomes more cooperative and more interested in his/her problem. It is conceivable that later in life, a child may stop using enemas and remain clean, following a specific regimen of a disciplined diet with regular meals (three meals per day and no snacks) to provoke bowel movements at a predictable time.

Every summer, the children with some potential for bowel control can try to find out how well they can control their bowel movements without the help of enemas. This is done during vacations to avoid accidents at school, during a time that they can stay home and try some of the strategies.

Most preschool and school-age children enjoy a good quality of life while undergoing the bowel management program. However, when they reach puberty, many express a high degree of dissatisfaction. They feel that their parents are intruding on their privacy by giving them enemas. It is feasible, but rather difficult for them to administer the enema themselves. An operation called a continent appendicostomy or a Malone procedure has been designed for this specific group of children (Fig. 29.3) [7]. It is important to stress that the Malone procedure is just another way to administer an enema, and therefore, before implementing the Malone procedure, the child has to be perfectly clean with his/her regular bowel management. The operation consists of connecting the appendix to the umbilicus, and creating a valve mechanism that allows catheterization of the appendix for the enema fluid, but avoids leakage of stool through it. If the child has lost his/her appendix, it is possible to create a new one from the colon. This is called a continent neoappendicostomy (see Chap. 34, for further continence imroving operations see Chap. 31).

References

1. Peña A, Levitt M (2002) Colonic inertia disorders in pediatrics. Curr Prob Surg 39:661–732
2. Peña A (1995) Anorectal malformations. Semi Pediatr Surg 4:35–47

3. Peña A, Guardino K, Tovilla JM, Levitt MA, Rodriguez G, Torres R (1998) Bowel management for fecal incontinence in patients with anorectal malformations. J Pediatr Surg 33:133–137
4. Peña A, Hong AR, Midulla P, Levitt M (2003) Reoperative surgery for anorectal anomalies. Semin Pediatr Surg 12:118–123
5. Kiesewetter WB (1967) Imperforate anus II. The rationale and technique of the sacroabdominoperineal operation. J Pediatr Surg 2:106–117
6. Rehbein F (1967) Imperforate anus: experiences with abdomino-perineal and abdomino-sacro-perineal pull-through procedures. J Pediatr Surg 2:99–105
7. Levitt MA, Soffer SZ, Peña A (1997) Continent appendicostomy in the bowel management of fecal incontinent children. J Pediatr Surg 32:1630–1633
8. Peña A, Levitt MA (2002) Colonic inertia disorders in pediatrics. Current problems in surgery 39:681
9. Peña A, Levitt MA (2002) Colonic inertia disorders in pediatrics. Current problems in surgery 39:695
10. Levitt MA, Soffer SZ, Peña A (1997) Appendicostomy in the bowel management of fecal incontinent children. J Pediatr Surg 32:1631

30 Dietary Prevention of Constipation

Petra Stommel and Alexander M. Holschneider

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30.1 Introduction

After the definitive correction of anorectal malformations (ARM), two different new diseases frequently occur: chronic constipation and stool incontinence. Chronic constipation is sometimes associated with smearing, staining, or overflow incontinence, which should not be confused with true incapability to retain stools due to an insufficient sphincter mechanism. Digital and electromanometric investigations under resting and squeezing conditions are very helpful for the differentiation. According to Holschneider et al., only 11.8% of the high-, 22.7% of intermediate-, and 63.3% of low-type malformations became completely continent without need of any additional help [1]. Total continence corresponds to the continence behavior of a healthy person who does not soil, does not have constipation, and can regularly and voluntarily have bowel movements. Another group of patients become continent with some aid, which means they need occasionally a light constipating diet or laxatives for the regulation of their stools. 23.5% of the patients with a high, 13.6% of children with an intermediate, and 22.7% of cases with a low type of imperforate anus behave this way. Taking these two groups of patients together, 35.3% of the high-, 36.3% of the intermediate-, and 86.3% of the low-type anal atresias became acceptably continent. In the literature this behavior is usually called "good continence".

The so-called "satisfactory results" in the literature involve two groups of patients: children who are chronically constipated and patients with a partially incompetent sphincter. However, the problems of the patients in both groups can be managed sufficiently by conservative means. Only the last small group in Holschneider's new classification [1], the "bad results" with complete therapy-resistant fecal incontinence need surgical therapy either due to untreatable chronic constipation or complete incompetence of the anorectal sphincters. The surgical therapy for incurable constipation is described in Chap. 32 and consists of resection of a megarectum. In contrast, complete insufficiency of the anal sphincters needs strengthening, sometimes of the external anal sphincter muscles, by a continence-improving operation, which will be described in the Chap. 31. This very unsatisfactory group of children comprises 20.6% of the high-, 9.1% of the intermediate-, and 4.5% of the low-type malformations.

The largest group of patients is, as mentioned above, the so-called "satisfactory results." The continence behavior of these patients is, per se, not satisfactory at all. They consist of two different, postoperative newly appearing diseases, which have to be treated by different therapeutic means: chronic constipation and stool incontinence.

Fecally incontinent patients suffer from hypoplasia of the muscle complex and an absence of smooth muscle fibers. For this group of patients, the diet must be constipative (e.g., bitter chocolate with 70% or more portion of cocoa, blueberries, bananas, apple pie, and carrot soup). Carrot pie however, may have a laxative effect due to its high amount of cellulose. Administration of activated carbon for medical use or loperamide, or the use of soft anal tampons after sufficient bowel cleaning may help.

Chronic constipation needs a totally different diet. The definition of the term constipation is difficult and imprecise. If bowel movements are only possible when the patient exerts the utmost pressure or after convulsive cramps, if there is a sensation that the bowel has not been completely emptied, if the stools are hard, if no bowel movements occur for a period of 3–4 days, or if an overflow soiling occurs, all of these occurrences are referred to as constipation [2,3]. Overflow soiling is the most common sign of chronic constipation in children after ARM repair. Rectal examination shows an impacted rectum that cannot be evacuated completely. The first therapeutic procedure described in Chap. 29 consists, therefore, of rectal washouts, as described in Chap. 31. After cleaning of as much colon as possible, either retro- or anterogradely, a laxative diet should be administered. This diet should take into account the underlying reasons for the constipation. Constipation can occur in an acute form (e.g., due to a change in diet when traveling, after febrile illnesses, after being bedridden for some time, because of local anal complaints, or after taking medication). They can occur, for example, if too little food is ingested, if insufficient roughage is consumed, if the amount of ingested liquids is insufficient, if the defecation stimulus is repressed, if the patient uses too many laxatives over longer periods, and/or if the patient has too little exercise. A medical examination will be necessary to clarify whether we are dealing with an acute and/or a chronic disorder, or if the constipation is the result of organic disease, (e.g., Hirschsprung's disease, dysganglionoses, or anal stenoses). The most frequent cause for chronic constipation in ARM are inborn motility disorders of the extra- or intramural nerve supply to the rectum, malformations of the rectal smooth muscle structure, or damage to the neuronal or vascular supply of the rectum during surgery. Whatever the reasons for the digestive complications, an optimal diet can have positive effect on constipation [3,4].

The best effect is achieved by a combination of different factors. These may include treatment of the underlying disease, a diet calculated to loosen the stools, drinks, bowel training, and exercise.

30.2 Dietary Fiber

Roughage, or fiber, plays an important role in the passage of chyme. Roughage increases the digestive juices in the gastrointestinal tract. In the intestine the roughage swells due to its absorption of water. It serves as a culture medium for the bacteria in the colon, allowing them to multiply more quickly and contribute to the volume increase. The breakdown of the fibers by bacteria creates gases and acids, which in turn stimulate the peristalsis of the intestinal wall. The consistency of the stools becomes softer, and the distension of the intestinal wall and increased propulsive motility shortens the transit time and reduces water resorption.

For roughage to have the optimal effect, it is important to drink enough liquids. Children between the ages of 1 and 4 years should drink at least 950 ml, children between 4 and 10 years of age should drink at least 1100 ml, between 10 and 13 years at least 1200 ml, and children between 13 and 15 years at least 1300 ml [5,6]. The more roughage the food contains, the more should be drunk. It is also important to increase the fluid intake if there is increased sweating, for example during sports. Insufficient fluid intake may lead to bowel obstruction. Mineral water, still mineral water, unsweetened fruit tea or herbal teas, and sugar-free fruit juices diluted with mineral water are all suitable. The amount drunk can be monitored using a checklist. Certain types of receptacles, drinking bottles, or jugs are useful aids to monitor fluid intake. To begin with, the current fluid intake should be monitored by recording all fluids ingested and then the amounts should be slowly increased until the desired daily amount is reached. Milk is not considered as a drink, but as a liquid meal. Too much milk often results in too little being eaten or drunk.

Roughage is indigestible vegetable material, which can be found in leaves, fruits, or roots. It is also referred to as raw fiber, vegetable fiber, or indigestible carbohydrate. Roughage cannot be broken down by digestive enzymes, but it can be partially broken down by the bacteria in the colon. The most important types of roughage are water-insoluble cellulose, lignin, and the partially water-soluble hemicellulose, together with water-soluble pectins. No single type of roughage is an essential food; however a certain amount of roughage is indispensable, beginning in the second half of the 1st year of life at the latest, to ensure that the bowel functions properly [7,8].

Nonpurified vegetable fibers are the fibers found in cereals, fruits, and vegetables. Purified vegetable fibers are fibrous and polymer substances such as lignin, cellulose and pectins, if they are ingested alone. These must be differentiated from synthetic fibers, such as crystalline cellulose, lignin, and cellulose, which are used in synthetic products. They are referred to as fillers because they have only a limited ability to swell. Hemicellulose and pectin are both bulking agents; however, pectin absorbs more water. Vegetable foodstuffs usually have less than 15% roughage. The declaration of the raw fiber content of foodstuffs in nutritional indices always refers to cellulose, hemicellulose, and lignin [7,8].

To increase the roughage intake, the amount of vegetable foodstuffs should be increased and the amount of animal products ingested should be reduced. This will result in a mechanical stimulation of the bowel. An increase in roughage can be achieved not just by increasing the percentage of vegetable foodstuffs ingested, but also by choosing products with more roughage. As fruits and vegetables largely consist of cellulose, such a substitution is limited, because the roughage in fruits and vegetables, with the exception of pulses, is only around 1–3% [7,8]. Berries and dried fruits have the highest roughage content. The vegetables with the highest amount of roughage are green peas, leeks, cabbages, and pulses; however, they are also more indigestible and can lead to flatulence. One should eat four to five portions of vegetables, uncooked vegetables, salads, fruit, and/or fruit juices every day.

Cereals consist in the main of hemicellulose that has a high capacity to absorb water. If wholegrain, multigrain, or wholemeal breads, pumpernickel, Graham bread, crispbread, or products such as granola, linseed, wholemeal gruel, wholemeal noodles, or brown rice are eaten instead of white bread, the roughage intake can be increased without increasing the size of the portions.

The roughage intake can also be increased by eating wheat bran, oat bran, or products containing wheat or oat bran. The volume of chyme is increased by the coarse bran's capacity to bind water and swell. Products made with coarse meal are more effective than those made of finely ground meal or bran flour, as their water absorption is limited. Wheat and oat bran are both available as supplements that can be added to granola and can be purchased either roasted or crisped.

Due to the hydrophilic nature of bran, ingesting around 5–10 g bran will require an additional 200 ml of liquid to be drunk; in other words, after the ingestion of bran it is important to drink enough. If it is not possible to ensure sufficient liquid intake, then the bran should be soaked prior to consumption, for example in water or juice. If sufficient amounts of liquid are ingested together with the bran, the time required for passage can be reduced. Bran can also be added to milk products, compotes, soups, and stews, and even to dishes with minced meat, to potato dumplings, and potato pancakes [4,9].

Various roughage supplements with different effects are available. These supplements have a high capacity to bind water and can increase the moisture content in the stools, increase the volume of the stools, and/or serve as a nutrient substrate for colon cells. The choice of supplement is an individual decision [4,9,10].

Nuts, almonds and sesame seeds also contain relatively high amounts of roughage and can promote the passing of stools.

Foodstuffs made of very finely ground meal (for example white bread rolls, toast bread, milk bread rolls, cake, and biscuits), are very unsuitable because these foodstuffs contain very little roughage. Other products with little roughage are noodles made of semolina wheat flour, white rice, desserts, sweets, and confectionery [10].

Foodstuffs that consolidate the stools, such as bananas, blueberries, boiled carrots, rice, low-fat curd cheese, hard-boiled eggs, cocoa and black tea, should initially be avoided. If the symptoms improve it may be possible to reintroduce them.

30.3 Dietary Stimulants

In addition to the mechanical stimulation provided by roughage, a chemical stimulation may also improve bowel peristalsis. The laxative effect of lactic acid, for example, is well known. It has an impact on intestinal motility via the bacterial flora and shortens the transit period. Lactic acid is found in yogurt, buttermilk, soured milk, kefir, vegetables that have been pickled, such as sauerkraut and pickles, and vegetable juices such as sauerkraut juice or red beet juice. Other organic acids, such as the tartaric acid found in grape juice, malic acid, which is found in grape juice but also in the juice of pip fruits and stone fruit, especially in apple, prune or fig juice, the citric acid in citrus fruits, and the acetic acid in wine vinegar are also believed to improve intestinal peristalsis.

Lactose or concentrated sugar solutions made of lactose also stimulate intestinal motility because lactose is digested more slowly than sucrose (normal sugar) due to the physiologically reduced activity of lactase in the small intestine. If larger amounts are ingested the lactose will reach the intestine without being digested. The laxative effect is due to the increase in the amount of liquid because of the osmotic activity of the lactose. The increased distension stimulus in the colon leads to an increase in intestinal motility. Lactose is partly broken down in the colon by bacteria in a similar manner to water-soluble roughage. This results in the creation of short-chain fatty acids, such as lactic acid, acetic acid, and formic acid, and carbon dioxide. In addition to the increased motility because of intestinal gas formation, the short-chain fatty acids help regenerate the intestinal mucosa and create an acidic intestinal milieu. Intestinal bacteria, such as lactobacilli and bifidobacteria, which prefer an acidic environment, multiply more rapidly, reducing the numbers of pathogenic organisms.

Sugar substitutes such as fructose, sorbite, sorbitol, mannitol, and xylitol also have a laxative effect. Their resorption is passive and therefore slower than that of glucose, which has an osmotic effect. Flatulence is a frequent side effect. Here again, it is important to gradually increase the dose.

Thermal stimulation through cold drinks, for example one glass of cold mineral water or fruit juice drunk before breakfast on an empty stomach, can have a gastrocolic effect. Cold food or drinks in the small intestine can improve colonic peristalsis.

Carbonated drinks can also accelerate the progress of chyme through the intestine.

In addition to gradual changes in diet so as to include more roughage and a sufficient amount of fluids, it is necessary to consistently train the intestine until it reacts with a normal defecation reflex. Defecation training should be carried out at certain fixed times of the day, preferably 30 min after breakfast, lunch, and dinner. Active pressing, carried out every day at the same time, together with sufficient calm, should accustom the bowel to being voided. This requires practice and does not work on command. If the urge to defecate occurs at a different time, it should on no account be suppressed.

30.4 Lifestyle

Modern life is often characterized by a lack of exercise. This means that muscles are exercised less. The measures outlined should ideally be combined with regular sports activities and other types of exercise. This will stimulate the metabolism, which in turn will also stimulate intestinal activity. A stomach massage using slow, circular, clockwise movements or damp, warm compresses may bring some relief.

At the beginning of the change in diet there may be some abdominal discomfort, which will take the form of flatulence and spasmodic cramps due to the formation of intestinal gases, but this can be expected to disappear after some time. It is possible to make food more digestible by thoroughly chewing it, making sure that meals are unhurried, and ensuring that there are sufficient rest periods between meals. It is best to begin with more easily digestible foodstuffs such as fruit purées, boiled vegetables, salads, oatmeal, and bread made of more finely ground wholemeal. Flatulence or a feeling of fullness can be relieved by drinking fennel tea, caraway tea, fennel-caraway-aniseed tea, or mint tee, or caraway can be added to food during cooking.

From infancy on a child should be given a varied mixed diet which should include products made of wholemeal, potatoes, vegetables, fruit, and curdled milk products. Sweets, cookies, cakes, and ice cream should not be forbidden, but they should make up only a small part of the daily food intake. A list of the appropriate amounts of food depending on the child's age can be obtained from the Research Institute for Child Nutrition or the German Society for Nutrition (Table 30.1).

30.5 Weaning

The possibilities available during the 1st year of life are much smaller. In breastfed babies who receive only breast milk, normal bowel movements may range from several times a day to once every 10 days, because breast milk is very digestible [2]. If the baby is given formula, it is important to ensure that the choice of formula is appropriate for the baby's age, that the formula is properly prepared, and that the baby receives the optimum daily amount at the proper intervals. These points must all be resolved before attempting to loosen the stools by giving the baby lactose, germ oil, or medium-chain triglyceride oil. The digestive tract of a baby is not yet fully developed and cannot fully resorb larger amounts of fat, so fat allows the chyme to slide more easily through the bowel. When beginning with solids it is important to avoid giving the baby foodstuffs that will consolidate the stools. The solids given at lunch can be prepared once or twice a week using wholemeal noodles or brown rice instead of potatoes. Wholemeal flakes and fruit purées should be used to make the evening porridge of milk and cereals. It is very important to ensure that the baby drinks sufficient amounts of liquid in the form of fruit tea or water. The more solid food the baby is fed instead of milk, the higher the liquid intake should be.

30.6 Laxatives

Many different laxatives exist and different groups have different effects. Laxatives should not be given indiscriminately, but only after consultation with a doctor and for short periods of time. They do speed up the passage of food through the bowel and result in defecation. If they are the only form of therapy

Table 30.1 Desirable and undesirable food in the treatment of chronic constipation

	Desirable food	Undesirable food
Fruits	All kinds of fruit (better fresh than cooked), nuts, almonds	Bananas, blueberries, raw apples
Vegetables	Vegetables of all kinds, especially legumes, cabbages and potatoes, if possible raw (except raw carrots)	Cooked carrots
Cereals and breads	Wholemeal bread, rye bread, bread with linseed, sesame bread, crisp bread, wholemeal rolls, muesli, bran, millet, linseed, cake or biscuits with wholemeal, whole-wheat pasta, whole-wheat rice, porridge	White bread, sandwiches, rolls, croissants, cakes, crescents, biscuits, cookies, pudding (nothing with starch flour), pasta, rice, semolina puddings, semolina dumplings, sweets only occasionally, farinaceous products
Fat	Butter, plant margarine, olive oil	Lard
Bread spreads	Berry marmalades (blackberry, raspberry, strawberry, gooseberry), honey	Peanut butter, chocolate cream, marmalade without seeds
Milk and milk products	Buttermilk, yoghurt	Cream cheese, cheese (under 30% fat)
Liquids	Mineral water, water, tea, fruit spritzer, fruit juice with fibre, fruit juice (after meals), malt coffee	Skim milk, curds, tea (with a lot of sugar), cocoa
Sweets	Dried fruit (figs, prunes, apricots, dates) with a lot of water to increase volume	Ice cream, marzipan, nougat, chocolate, sweet paste made from cocoa, sugar and crushed nuts
Other	Milk sugar (lactose), malt sugar (maltose), juice from figs or plums	
Not forbidden, but restricted foods	Meat, ham, cream, eggs, cheese with more than 30% of fat	Strongly forbidden: fast food

used without any corresponding change in diet or lifestyle, they will not be effective in the long term. Inappropriate use may result in complications such as fluid imbalance, loss of electrolytes, and disturbances of the acid-base metabolism. If laxatives are taken to supplement the altered diet and lifestyle, they should not be stopped abruptly, but gradually phased out.

30.6.1 Slow-Transit Constipation

If constipation is caused by intestinal neural dysplasia or slow-transit constipation, a diet with large amounts of roughage may be contraindicated. Due to the lack of distension stimulus the bowel peristalsis will not be sufficient for the passage of chyme. In this case it is important that the child receives a laxative diet with very little roughage.
As mentioned above, a laxative effect can be elicited by:

1. Acids, such as lactic acid, wine vinegar or cider vinegar, and citric and acetic acid.
2. Carbonated drinks.
3. Thermal stimulation.
4. Lactose.

Fig syrup also has a slightly laxative effect as it contains inverted sugar.

Malt extract is a polysaccharide, which consists chemically of various starch breakdown products that have been reduced enzymatically, a lot of maltose, and a small amount of dextrines. Depending on the amount of maltose, it affects the bowel by promoting fermentation and can therefore have a positive effect on constipation. One teaspoon of malt extract can be added 1–3 times per day to tea or in soup. The use of lactulose syrup may also be helpful. The amount given will depend on the child's age.

For children with intestinal neural dysplasia, in addition to a laxative diet with little roughage and the intake of lactulose syrup it is important to avoid the ingestion of foodstuffs that will consolidate the stools; the child must also drink sufficient amounts of liquid.

References

1. Holschneider AM, Esch NK, Tragholz E, Pfrommer W (2001) Surgical methods for anorectal malformations from Rehbein to Peña – critical assessment of score systems and proposal for a new classification. Eur J Pediatr Surg 12: 3–82
2. Wachtel U, Hilgarth R (1994) Ernähung und Diätetik in der Pädiatrie und Jugendmedizin. Band II Diätetik. Thieme Verlag, Stuttgart, pp 76–80
3. Müller S-D (2000) Ernährung bei gastrointestinalen Erkrankungen. VitaMinSpur 15:121–124
4. Küpper C (2003) Obstipation und diätetische Abhilfemöglichkeiten. Ernährung Med 18:44–47
5. Deutsche Gesellschaft für Ernährung und Aid (2004) Vollwertig essen und trinken nach den 10 Regeln der DGE. 21 Auflage
6. Verschiedene Broschüren des Forschungsinstituts für Kinderernähung. Dortmund
7. Wachtel U, Hilgarth R (1994) Ernähung und Diätetik in der Pädiatrie und Jugendmedizin. Band I Ernährung. Thieme Verlag, Stuttgart, pp 20–25
8. Berg G (1978) Ernährung und Stoffwechsel. Schöningh Paderborn UTB, pp 145–147
9. Sailer D (1999) Obstipation und Ernährung. Ernährungsumschau 46:380–382
10. Morlo M (2004) Gut gekaut ist halb verdaut. VFED aktuell 79:14–18

31 Operations to Improve Continence after Previous Surgery

Alexander M. Holschneider and Philipp Holschneider

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31.1 Introduction

Several techniques have been described to restore continence after previous repair of anorectal malformations (ARM). In principle, these attempts have been based on: (1) secondary repair of the levator ani, (2) substitution for the levator by muscle grafts, (3) strengthening of the levator by muscle grafts, (4) construction of a sphincter from the bowel itself, (5) electronic devices to stimulate the muscle, and (6) restoration of dermal sensibility especially after mucosal ectopy or rectal prolapse.

A secondary operation for a patient born with an ARM is generally necessary in two main groups of patients. The first group is represented by those patients who had a failed operation for the original defect. This may include retraction of the descended bowel, severe wound infection, severe stricture, recanalization of a fistula, or a combination of these. The second group is represented by patients who had a previous procedure but who suffer from fecal incontinence.

A good candidate for a reoperation is considered to be a patient who was born with a rather benign type of malformation, namely rectobulbar urethral fistula in the male or rectovestibular or low vaginal fistula in the female, with a good sacrum and a "good-looking perineum." In addition, if we see a very mislocated rectum, patulous and prolapsed, most frequently anterior to the center of the external sphincter, significant improvement by reoperation through a posterior sagittal approach can be expected. On the other hand, we no longer operate on a patient who has a grossly abnormal sacrum and a rectum that was well placed at the center of the external sphincter.

The redo posterior sagittal anorectoplasty (PSARP) is an operation that takes advantage of the available muscles. If the patient does not have good muscles and the rectum is relatively well located by the previous operation, we must assume that reoperation will not benefit him (see Chap. 25)

31.2 Stephens' Secondary Pull-Through [1,2]

In all patients who need a sacroperineal or sacroabdominoperineal reexploration and levator repair, magnetic resonance imaging (MRI) and a cinedefecography should be performed before the secondary repair. Prior to intended repair, defecography indicates whether there is complete or partial loss of anorectal angulation and movement so that the incontinence is indeed due to muscle inaction rather than to some simply correctable factor such as constipation.

MRI is able to show the presence of muscular damage or hypoplasia and the wrong position of the pulled-down rectum.

For reconstruction, we usually use the technique of Stephens and Smith [2], which is basically a sacroperineal exposure of the entire muscle complex, as shown in Figs. 31.1 and 31.2. The purpose of the operation is to place the rectum so that it lies adjacent to the urethra or vagina, with the levator muscles drawn together behind it. The most anterior muscle bundles are approximated in the midline as far anteriorly as possible, preferably indenting the rectum to the point of occlusion.

The age of these patients is usually 3 years or older, and for this reason it is deemed unnecessary to perform a preliminary colostomy. The pelvis and the levator muscles are larger in these patients than in the newborn and are larger compared with the size of the collapsed rectum, which is tucked up anteriorly against the urethra or vagina. Needless to say, the bowel should be empty. The patient is anesthetized, a bladder catheter is passed into the urethra, and the patient is turned into the jackknife position. A gauze with Betadine is inserted into the anal canal. A natal cleft incision is made from sacrum to anus, which is encircled by a circumferential racket extension. The incision is deepened in the midline through the fat using a muscle stimulator to identify muscle as it comes into view. It is possible to detect the activity and direction of muscle pull and thus incise the pelvic diaphragm as close to the raphe as possible. The coccyx should be split or excised and the supralevator space dissected up under the tip of the sacrum. It is then possible to separate the tissues from the rectum from above down, within and outside the supralevator space. In this way, the pelvic diaphragm is laid open, the rectum is isolated from the levator muscles, and the route that the rectum has taken can be identified.

The nerves to the levator muscles pass from the lateral pelvic wall toward the midline on or in the substance of the levator muscle. Midline operations with expansion of the window thus created should not impair these nerves. It may be necessary at this juncture to insert a metal obturator or gloved finger into the rectum to facilitate isolation of the wall from the surrounding very adherent fibrous and muscular tissues.

The rectum and anus are cleared and thoroughly mobilized from the pelvis and perineum, and replaced in a collapsed and empty state, with the gauze removed, against the urethra or vagina, and the levator muscle, ragged and barely recognizable, partly obscured by fat and fascia, is sutured in the midline as far anteriorly as possible to the indent or, if possible, to occlude temporarily the rectal lumen and thus heal in this position. The remainder of the diaphragm and the fat of the perineum are approximated, and the anus, divested of any cutaneous cuff still adhering to the wall of the rectum, is restored in its original site.

A drain tube is brought out of the incision in the vicinity of the coccyx. A rectal tube is then passed into the rectum through the indented zone and is left in place for several days. Regular saline irrigations of the rectum are recommended to deflate the lumen and wash away fecal matter as it accumulates. The tube may be removed on the 4th day. To reduce residue, the patient should be given fluids by the intravenous route and only water by mouth for at least 3–4 days, and longer if tolerated after surgery. A low-residue diet is continued for 1–2 weeks postoperatively.

At 10 days postoperatively, gentle dilatation of the indented zone of the rectum is then performed daily with the surgeon's or parent's finger until the sling is softened and agile. This may take several weeks.

In a paper describing 29 secondary operations, with "acceptable" continence in three-quarters of the patients, Kiesewetter and Jeffries [3] described the advantages of the sacral route in revision repairs. Furthermore, it is interesting to note the similarity of this reconstruction to that of Peña's PSARP [4], even to the use of a muscle stimulator.

31.3 Stephens' Secondary Repair of Damaged or Hypoplastic Muscle Complex [2]

Once the decision has been made to operate on a specific patient, a protective colostomy should be created. We always prefer a right transverse colostomy. The technique corresponds in principle to Peña's procedure for PSARP and redo operations [5].

The patient is placed in the prone position, as described previously. The electrical stimulator allows the surgeon to make a full evaluation of the available muscles in the perineum. Multiple 5-0 stitches are placed at the mucocutaneous junction of the anus. All of these stitches are used for traction as in a primary PSARP (Fig. 31.3). The incision runs from a point immediately below the coccyx midsagittally and around the mucocutaneous junction in a "racket-like" fashion. Since the rectum is usually anteriorly mislocated, the midline incision necessarily cuts through the midline external sphincter and divides it into two halves. The dissection around the rectum must run as close as possible to the serosa of the rectum (colon) to avoid unnecessary damage to the surrounding stri-

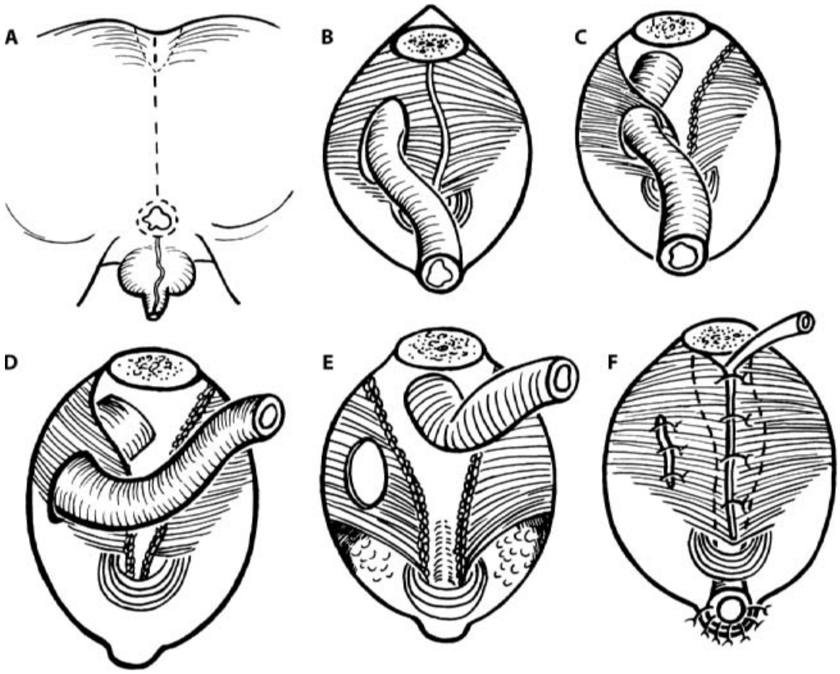

Fig. 31.1 Secondary repair of the levator ani muscle following inadequate rectoplasty. Rerouting the rectum from the lateral diaphragmatic to the sphincteric parts of the levator ani (From Stephens and Smith [2], Fig. 21-1). **A** Perineal incision in the natal cleft and around the anus. **B** Rectum located in the lateral levator diaphragm. **C** Levator incised in the line of the raphe. **D** Rectum then dissected from its tunnel through the levator muscle. **E** Rectum drawn up through the lateral tunnel. **F** Rectum rerouted through the puborectalis funnel; levator ani sutured in the midline, lateral rent closed, and anus resutured

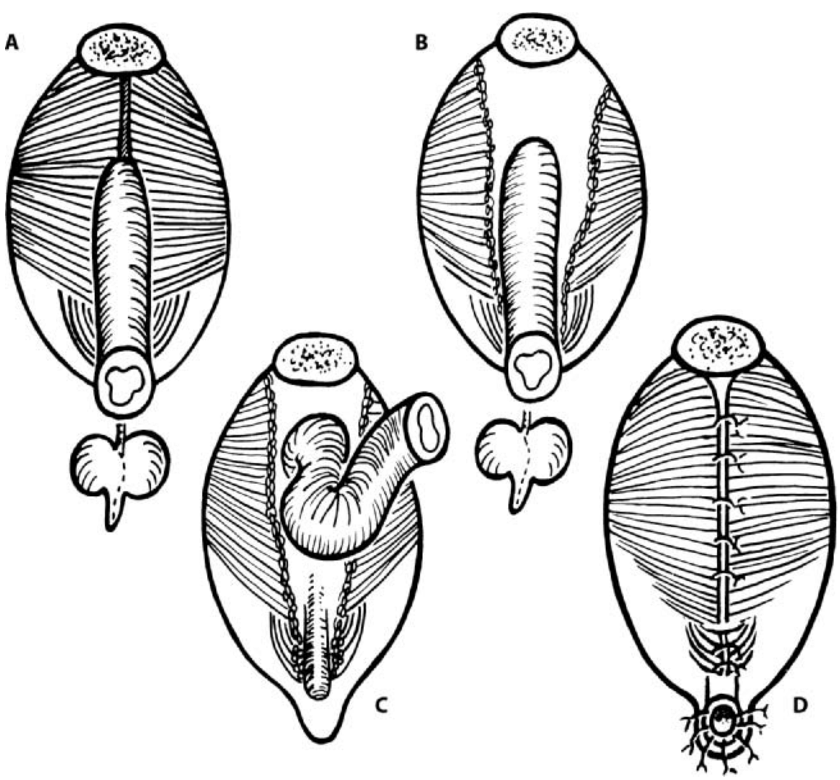

Fig. 31.2 Posterior sagittal anorectoplasty with repair of the levator ani muscle following inadequate rectoplasty. Rerouting the rectum in the sagittally divided levator ani muscle (from Stephens and Smith [2], Fig. 21-2). **A** Rectum emerging from the split diaphragmatic part of the levator ani. **B** Liberation of the rectum from the adherent levator muscles. **C** Levator ani incised in the midline behind and around the rectum. **D** Rectum tucked closely against the urethra and the levator ani muscle, including the remains of the puborectalis, approximated caudal to it. End of the rectum resutured to the skin of the perineum.

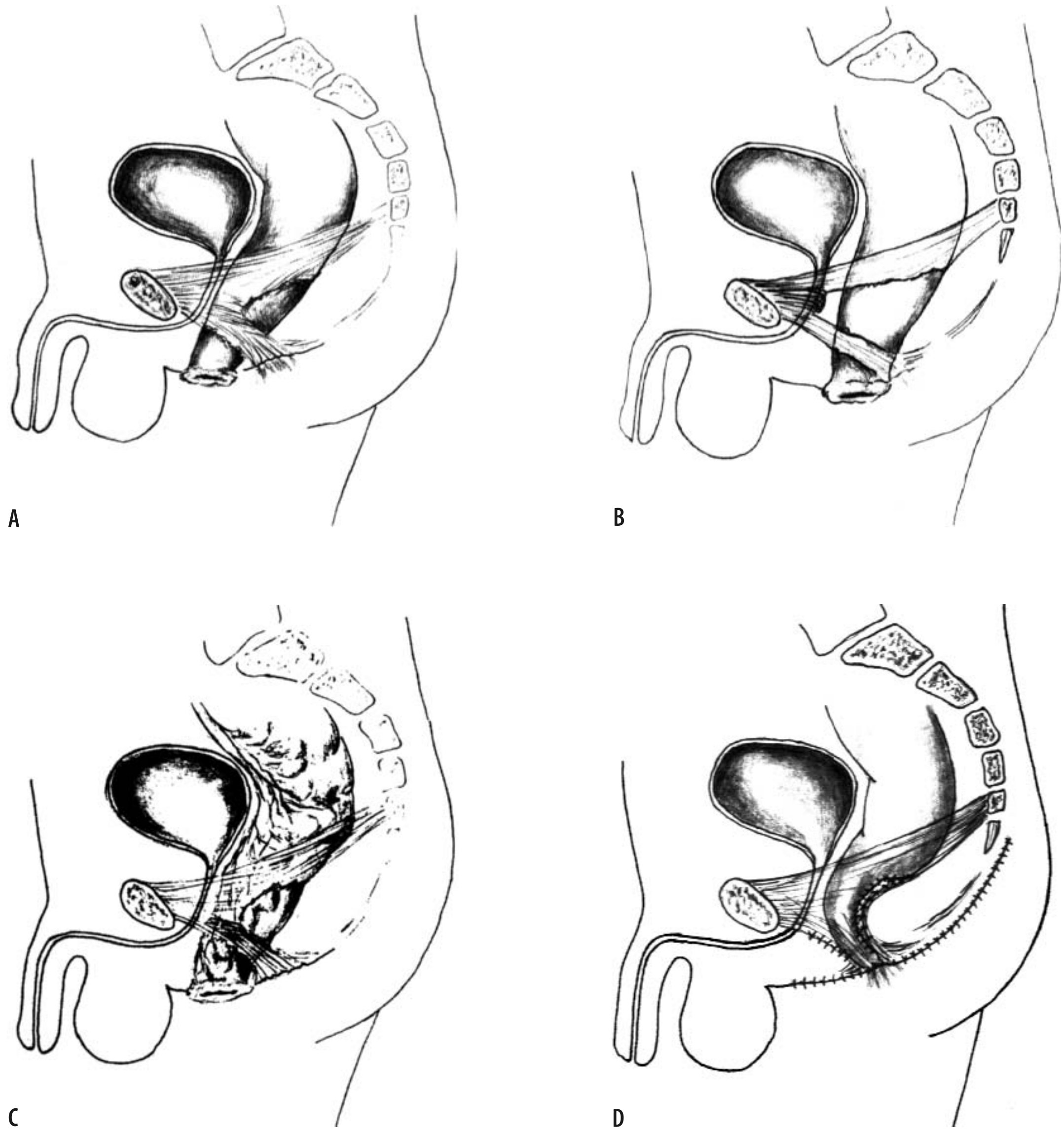

Fig. 31.3 Different types of inadequate rectoplasty (from Stephens and Smith [2], Fig. 21-7–21-9). **A** Muscle complex damaged and levator muscle not completely closed. **B** Parts of the muscle complex anterior the rectum. **C** Mesenteric fat adding to the rectal wall. **D** Finished repair showing tapered rectum, reconstruction of the muscle complex with reestablished anorectal angle including striated muscle of external anal sphincter

ated muscle structures. The midline incision is deepened until the posterior rectal wall is located. The lowest part of the levator muscle, the muscle complex, and the external sphincter usually have to be divided. The rectal dissection is carried out high enough until the posterior urethra is identified. The electrical stimulator allows one to determine whether there is any striated muscle left behind the urethra and in front of the rectum, which could be identified as the "missed puborectalis" [2].

Once the dissection is complete, the operative field is evaluated and the strategy of reconstruction is established. Moreover, each case represents a different problem. There have been several basic findings in most of these cases, including:

1. A prolapsed patulous anus was found located anterior to an intact external sphincter.
2. An intact muscle complex had been left behind the rectum, which was pulled down in a rather straight manner.

3. There was a significant piece of mesenteric fat surrounding the rectum (colon) that represented the colon mesentery, which was pulled down to preserve the blood supply of the colon. The mesenteric fat occupies a significant space and interferes with the muscle function, since it lies in between the bowel wall and the striated muscle.
4. There have been different degrees of lateral mislocation of the rectum.
5. There have been a few patients in whom the rectum was pulled down behind the muscle complex and behind the external sphincter, leaving a significant portion of the striated muscle (which represents the lowest part of the levator), which also could be identified as a "missed puborectalis" [2]. In these patients the rectum was pulled straight behind, leaving a good portion of levator muscle in front of the rectum and behind the urethra.
6. There have also been different degrees of levator muscle scarring and destruction, particularly in its lowest portion, as a consequence of a pull-through of a very large rectum or muscle hypoplasia, or just an inappropriate preparation of the muscle complex. In some patients there is a gap between the upper part of the muscle complex being well developed at its posterior coccygeal attachment and hypoplastic close to the external sphincter fibers.

The goal of the operation is to reconstruct the patient's anatomy to make it resemble, as closely as possible, the normal configuration (Fig. 31.3 D). Thus, the rectum (which is not frequently necessary in secondary operations) might be tapered and should be relocated and placed in front of the levator and in the middle of the muscle complex. Then the new anus is created at the center of the contraction of the external sphincter. The entire mesenteric fat should be resected, leaving the blood supply of the rectum through its transmural circulation. During this type of operation, one can gain a very accurate idea of the final prognosis of the patient based on the appearance of the muscles and on their strength of contractions.

The reconstruction is carried out with 5-0 Vicryl or Dexon interrupted sutures. The same principles described for the primary reconstruction of these patients [6,7] are used for the secondary repair.

31.4 Reinforcement and Substitution for the Levator Ani

Procedures for the reinforcement and substitution of the levator ani are applicable when the levator is destroyed or is congenitally hypoplastic, or when neuropathic paralysis of the sacral outflow occurs.

31.4.1 Kottmeier's Levatorplasty (Levator Release)

Kottmeier [8] and coworkers [9,10] described an operation to gain increased effectiveness of the levator ani in secondary repair. An attempt was made to improve fecal continence through the utilization of salvageable remnants of the levator sling through: (1) enhancement of the motor function by increasing the anterior shutter action of the levator muscle, and (2) increasing the area of sensory perception of the levator by enlarging the levator sling through a posterior release and plication (Fig. 31.4).

With the patient in either a lateral position with knees flexed or a jackknife position, the perineum is opened through a posterior midline incision or through an inverted chevron incision. The coccygeal attachment of the levator muscle is transected, and the dissection is carried into the presacral space. The rectum or the pulled-through intestine is freed from its presacral attachment and pushed anteriorly. This maneuver will reveal the levator sling with its upper components, the ileococcygeal and pubococcygeal muscles. The ileococcygeal muscle is split sharply in a horizontal plane parallel to its fibers. The entire levator sling, consisting of ileococcygeal, pubococcygeal, and if present, puborectalis muscle, is then liberated from its posterior attachment. A finger is placed into the rectum, and the levator sling is then snugly tightened around the rectum with interrupted nonabsorbable sutures to achieve an acute anterior angulation. The presacral space is irrigated with saline and Betadine and closed with multiple interrupted absorbable sutures. The skin is closed with sutures. Drainage of the presacral space is not necessary.

Follow-up examination of 18 patients with a levatorplasty for incontinence following repair of ARM showed that complete restoration of continence occurred in 8 children, 6 improved, and gross incontinence persisted in 4 children [8]. Kottmeier later reported [see 4] 35 children in whom this procedure was performed, with similar results.

It is remarkable that the nerve supply to the levator is not divided in this procedure; nevertheless, the accompanying radiographs show forward and backward movement of the muscle, with a considerable shutter effect on the rectal lumen. Gross [11] also described a similar procedure.

31.4.2 Puri and Nixon's Levatorplasty (Levator Plication)

This procedure [12] is based on the Kottmeier [8] concept, together with the levator plication described by Gross [11]. Of 15 patients, 4 became fully continent, 7 were improved, and 4 did not improve. The technique is illustrated in Fig. 31.5.

31.4.3 Free Autogenous Muscle Transplant for Strengthening of the Levator Ani (Palmaris Longus Transplant)

Hakelius [13] and Hakelius et al. [14–16] originally described the procedure in which a graft of muscle (usually the palmaris longus, less commonly the sartorius, or extensor digitorum brevis) is first denervated 2 weeks before transplantation and is then transplanted as a U-shaped sling around the rectum, in close contact with the puborectalis, and anchored to the pubic bone (Figs. 31.6 and 31.7). Denervation allows muscle survival as it initiates a lower level of energy consumption. The graft becomes reinnervated from the puborectalis over a period of 9 months. Central atrophy of the muscle occurs, but the surviving one-third facilitates continence.

Grotte et al. [16] reported the follow-up of 21 patients. Continence was virtually normal in ten patients, nine were "socially satisfactory", and in two the results were poor. The procedure has undoubted merit. Successful reports were also given by Mollard et al. [17] and by Holschneider and Hecker [18] (Fig. 31.8).

The problem with this operation is similar to that of transposition of denervated gracilis muscle described by Holle et al. [19]. Scarring of the pelvic floor prevents reinnervation, and fibrosis of the regenerating muscle graft remains a critical factor for the ingrowing nerve fibers. A further problem is the degree to which the graft is prestretched. On the one hand, it is necessary to prestretch the muscle sufficiently to obtain a better angulation of the anorectal angle and to achieve a compression of the rectum from both

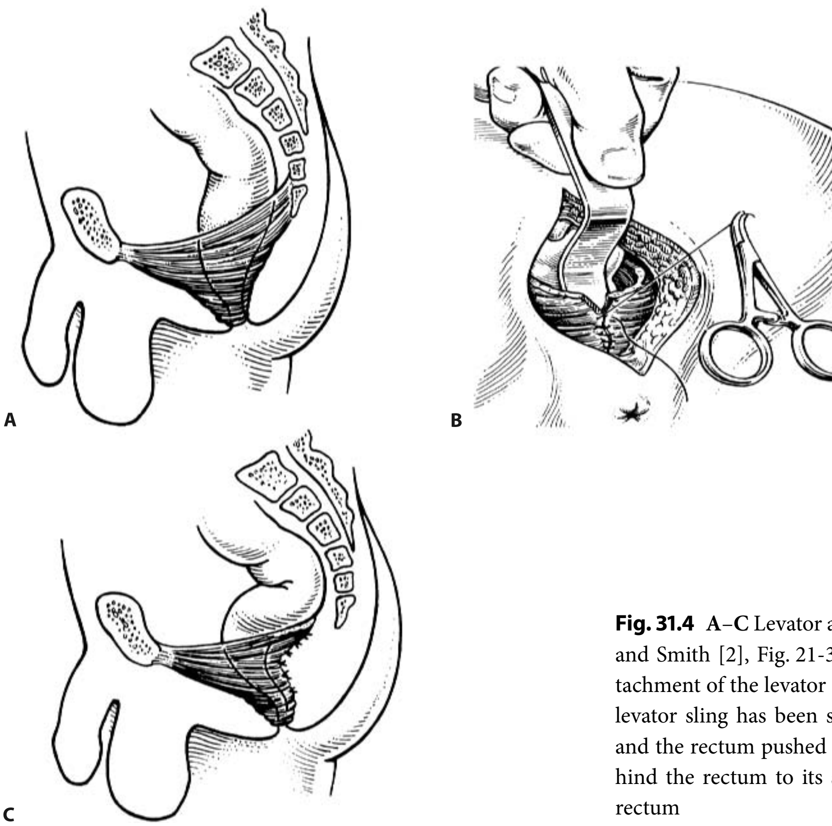

Fig. 31.4 **A**–**C** Levator ani release and plication(from Stephens and Smith [2], Fig. 21-3–21-5). **A** The posterior coccygeal attachment of the levator sling is illustrated schematically. **B** The levator sling has been severed from its posterior attachment, and the rectum pushed anteriorly. **C** The levator tightened behind the rectum to its sheath on the acute angulation of the rectum

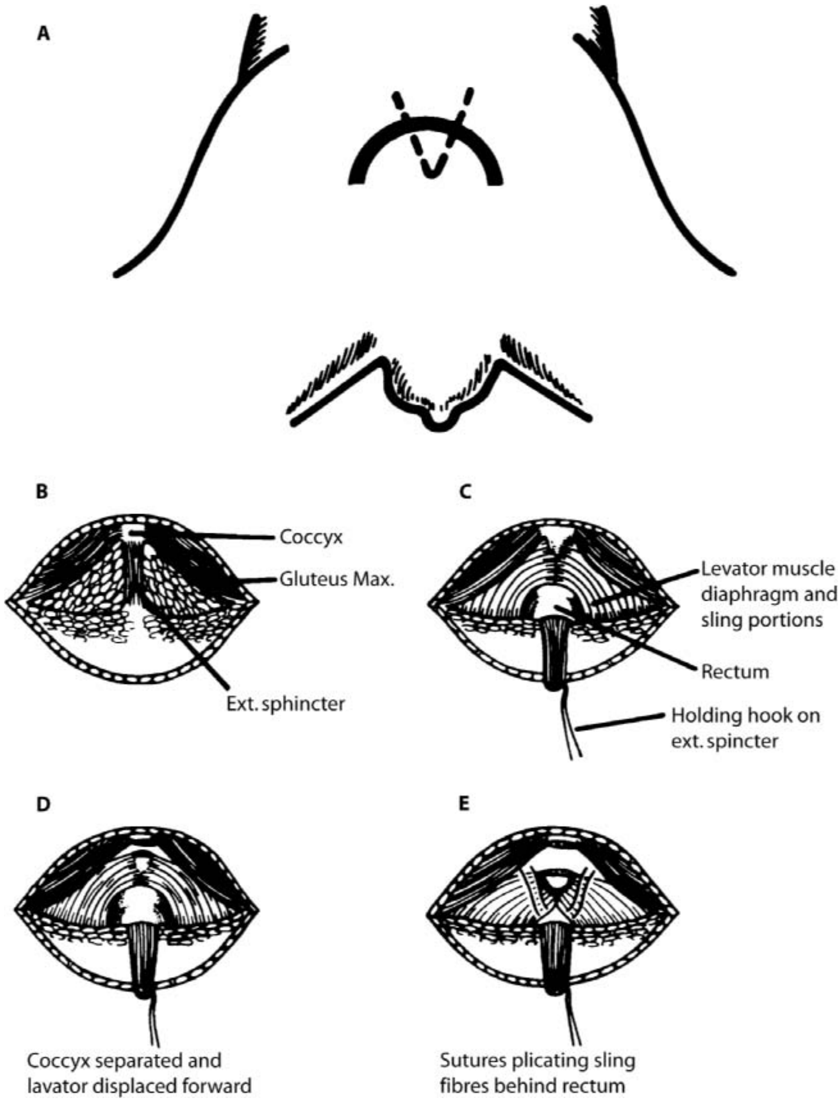

Fig. 31.5 Steps of levatorplasty (reproduced from Puri and Nixon [12] with permission of the publishers). **A** Incision. **B** Beginning of flap reflection. **C** Exposition of the levator ani in the midline. **D** Coccyx separated from the sacrum, and levator freed from adjacent attachments. **E** Plication of the mobilized levator. *Max* Maximus, *Ext* external

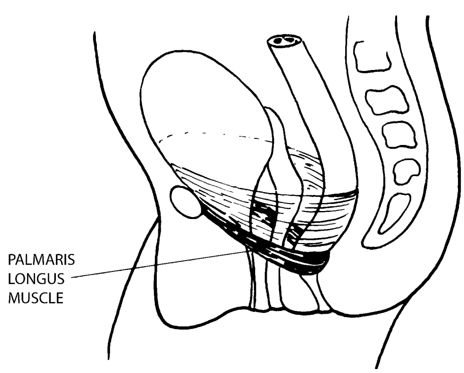

Fig. 31.6 Schematic representation of palmaris longus transplantation [13,14]. Reproduced from Holschneider and Hecker [18] with permission of the publisher)

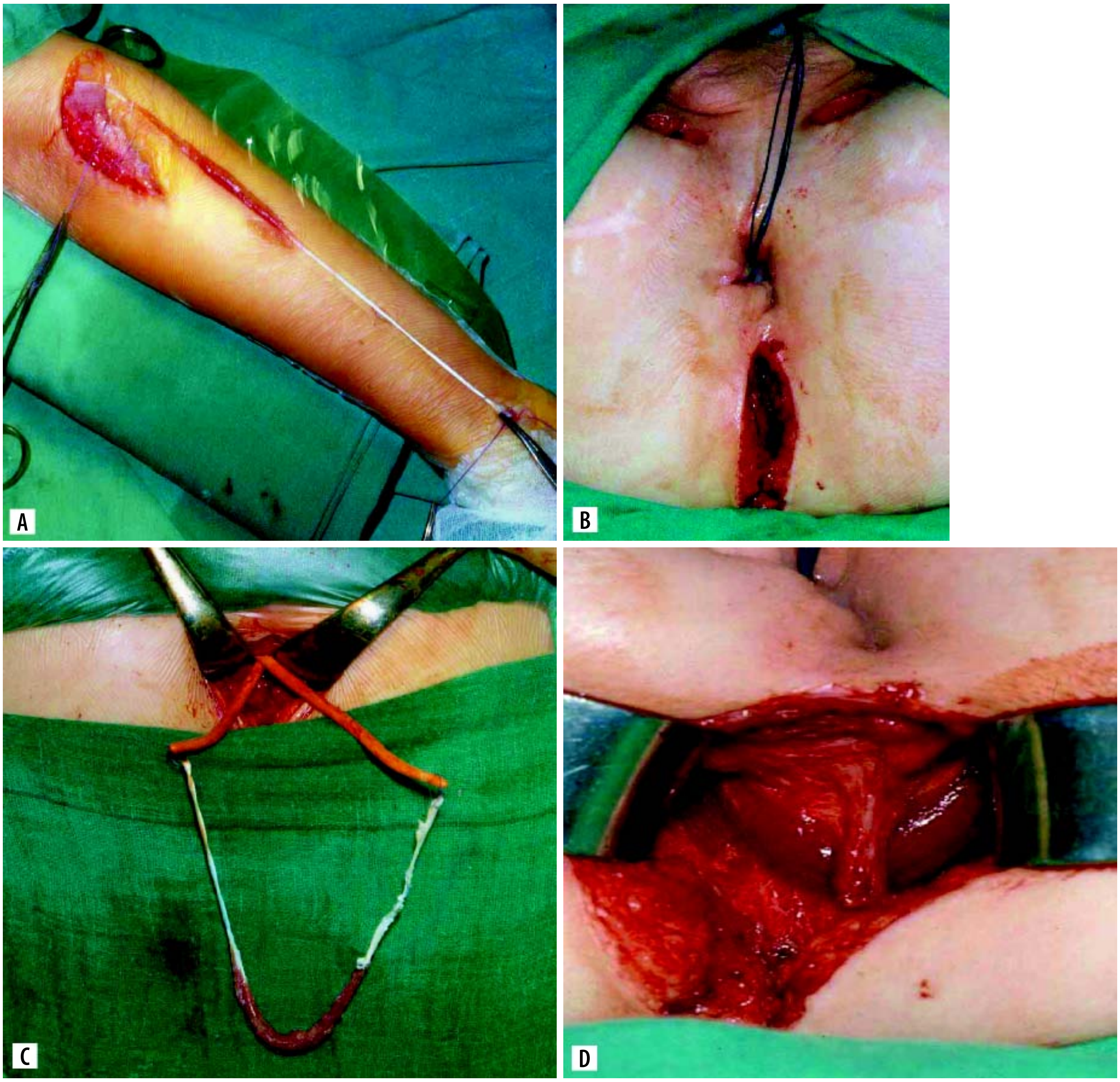

Fig. 31.7 **A**–**D** Palmaris longus transplantation. **A** Preparation of the palmaris longus muscle, which was denervated 14 days previously. **B** Dorsal incision in the natal cleft. **C** Tendon of the palmaris longus muscle divided and resutured to the muscle belly, creating a sling. Sling ready for transposition. **D** Palmaris longus sling in situ close to the muscle complex

sides. On the other hand, striated muscles can only be prestretched about 15% over resting conditions for the fibers to survive.

Studies in the rat [20] have shown that necrosis increases in proportion to the degree of prestretching. In human studies, six out of nine patients treated with Hakelius-Grotte free muscle transplantation became continent for solid bowel contents, and three for liquid content, but none were continent for flatus. These results are less favorable than those reported by Hakelius et al. [13–15] and Grotte et al. [16]. Postoperative electromanometric studies have shown that the anorectal pressure difference is improved by transplantation and that there is some reflex response coincidental with puborectalis sling contractions.

31.5 Substitution of Striated Anal Sphincter

31.5.1 Gracilis Muscle Transplant

Pickrell et al. [21] described the original procedures; further reports and modifications have since been

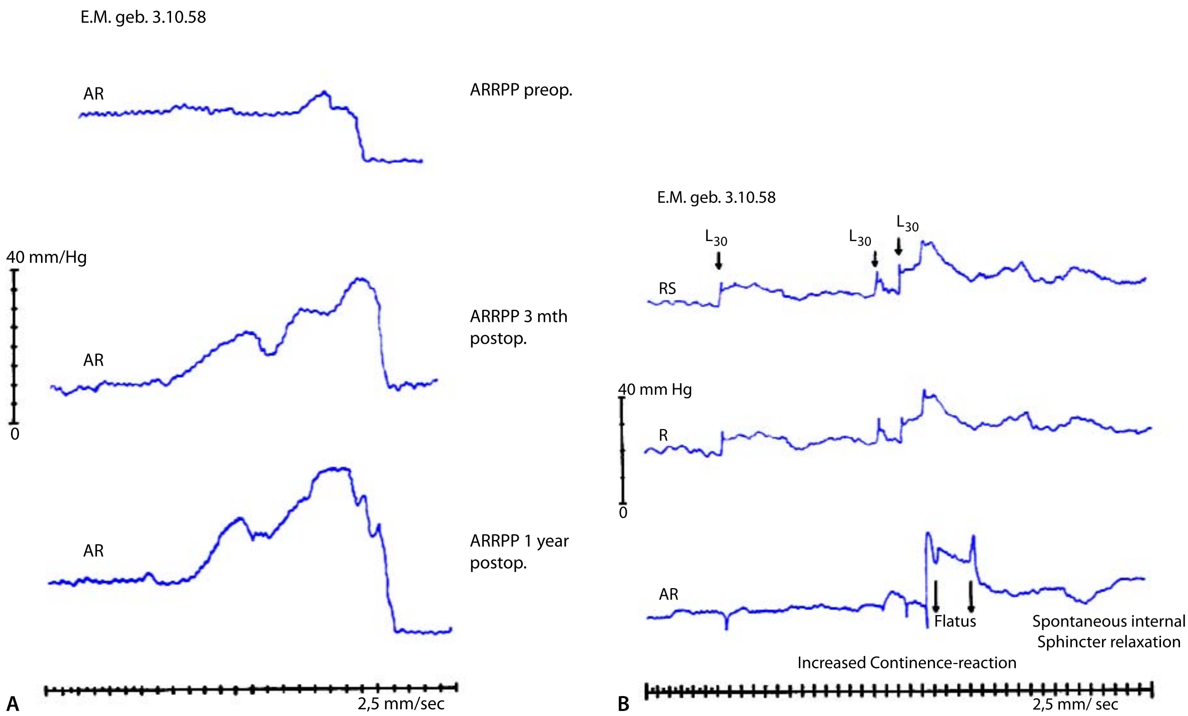

Fig. 31.8 **A** Electromanometric investigation of a child after palmaris longus transplantation: preoperatively (top trace), 3 months after operation (middle trace), and 1 year after the operation (bottom trace). **B** Continence after palmaris longus transposition. Injection of 30 ml of air (*L30*) into the rectosigmoid (*RS*). Contractions of the muscle complex reinforced by the palmaris longus muscle interrupting the evacuation of flatus. Note the spontaneous relaxation of the internal anal sphincter. *AR* Anorectum, *ARP* anorectal resting pressure profile (also *ARRPP*), *R* rectum, *prop.* preoperatively, *postpone.* postoperatively

made [22–26], including those by Brandesky and Holschneider [27] in 24 patients, showing definite improvements in 21; Berger et al. [28], who obtained poor results in 8 patients; and Brandesky et al. [29], who achieved improvement in 8 of 11 patients, but had poor results in myelomeningocele children. In the original method [21], the gracilis muscle was used as a neurovascular pedicle transplant. The muscle was detached distally and freed from the surrounding tissue up to its proximal one-third, where the neural innervation from the femoral nerve joins the muscle medially. From this point the muscle was transposed to the perineum, where it surrounded the anorectum subcutaneously, then dorsally, and again anteriorly. The tendon of the gracilis muscle was then fixed to the contralateral ischial tuberosity (Fig. 31.9).

Holschneider states that to obtain a good functional result, some requirements must be met. First, the gracilis muscle should be long enough so that the muscle belly and not the tendon surrounds the anal canal. Second, the gracilis muscle should have one good neurovascular supply in the most proximal one-third of the thigh, because more distal vessels and nerves must be divided during the operation to allow the transposition of the muscle. Third, the muscle should be fixed with minimal tension to the contralateral ischial tuberosity. Dilatation should be unnecessary if the tension is correct. Fourth, although infection is a high risk in gracilis muscle transplantation, Holschneider never uses a colostomy and states that the infection rate is very low because of 24-h postoperative antibiotic prophylaxis and very careful postoperative care.

Holschneider and Lahoda [30] were able to show that commencing a few weeks to 9 months postoperatively, the patient learns to contract the gracilis muscle sling without a simultaneous contraction of the other adductor muscles of the thigh. On the other hand, the patient learns to contract his adductor muscles without simultaneous contraction of the gracilis muscle sling, A good result is shown in Fig. 31.10. However, variations in the innervation of the gracilis muscle, with a low neurovascular support joining the muscle at its distal end in about 15–20% of patients, may lead

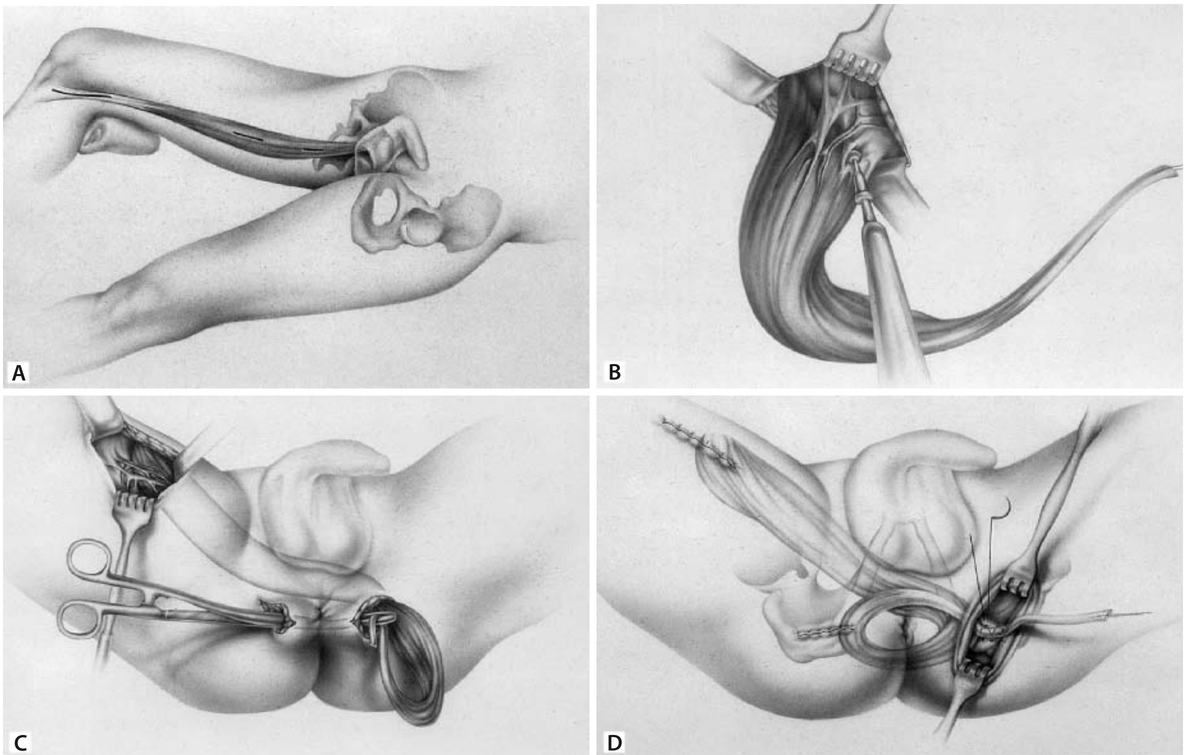

Fig. 31.9 **A–D** Schematic drawing of gracilis muscle transplantation according to Pickrell [21]. **A** Incisions on the thigh. **B** Electrostimulation of the innervating branches of the femoral nerve. **C** Pull-through of the gracilis muscle to the perineum and around the anorectum. **D** Transposition of the muscle finished. The gracilis muscle is fixed at the contralateral ischial tuberosity

to poor results. To avoid these problems Hartl's modification [31] of Pickrell's method used the gracilis muscles of both sides simultaneously. A disadvantage of this technique is that if an infection affects both muscles, there is no possibility for a second gracilis transposition.

In another modification, Holle et al. [19] first denervated and then transposed the gracilis muscle sling as a vascularized pedicle transplant (Fig. 31.11). After the muscle is transposed to the perineum, it is divided into two equal parts and attached closely to the pelvic floor. Both parts of the muscle belly are sutured to the ischiococcygeal ligament so that the anorectum is now lying in between the gracilis muscle belly. According to Holle et al., reinnervation of the denervated muscle will take place from the pudendal nerve within about 9 months [19]. Therefore, the gracilis muscle can be contracted simultaneously with the pelvic floor muscles and is able to exert a passive continence reaction, which was never possible following the original operation procedure [21].

The success of the operation of Holle et al. [19] depends on two points: (1) There should be no scarring of the pelvic floor from previous operations, because this disturbs the ingrowing nerve fibers, and (2) the vascular supply should be uniform in both parts of the gracilis muscle; unfortunately, this is not always the case.

Holschneider et al. [20] have extended these studies in goats by performing a microsurgical anastomosis between the pudendal nerve and the gracilis branch of the femoral nerve. Although the nerve pathways were reestablished, the division of the muscle into two parts (as in the Holle technique) sometimes leads to muscle death. Holschneider et al. are therefore not in favor of the Holle method.

A further modification of Pickrell's method was made by Dittertová and Grim [32], who combined Pickrell's gracilis muscle transplantation with Hartl's modification. They use the proximal two-thirds of both gracilis muscles, but prepare the neurovascular supply of both muscles so that the muscles and their neurovascular flaps can be transposed at the pelvic floor. Both parts of the gracilis muscle are sutured at the os pubis of each side and fixed to each other behind the anorectum, forming a muscle sling. The is-

Fig. 31.10 Gracilis muscle transplant (from Holschneider [55], with permission of publisher). **A** Under resting conditions. **B** When contracting

sue with this modification is avoidance of damage to the innervation of the transplanted muscle segments (Fig. 31.12).

In assessing their own results, Holschneider et al. stated that gracilis transplantation creates a myogenic stenosis that is able to establish a high-pressure barrier and to relax, but is not able to contract reflexly [20]. Electromanometrically they observed a postoperative rise in the anorectal pressure barrier. The patient was able to stop propulsive waves by voluntary contractions of this muscle and to avoid defecation. The anorectal squeezing pressure profile also increased (Figs. 31.13 and 31.14).

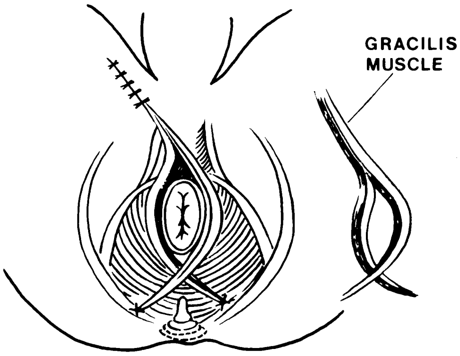

Fig. 31.11 Schematic representation of gracilis transplantation of Holle [19], reproduced from Holschneider [55] with permission of the publisher

31.5.1.1 Follow-Up

Eight out of 56 of Holschneider's patients with Pickrell's gracilis transplantations acquired a postoperative infection. Eight others developed anal stenosis that could be gently dilated. Five children developed an ileus because of adhesions as a late complication of the primary pull-through procedure. In one patient mucosal ectopy persisted, and three other children developed a keloid scarring of the distal thigh [25]. Twenty-eight out of 40 patients in whom the operation was performed at least 2 years previously developed good continence. The children ceased soiling and had regular stools or suffered a little staining under conditions of stress and diarrhea. Three children had a small degree of persistent soiling, but did

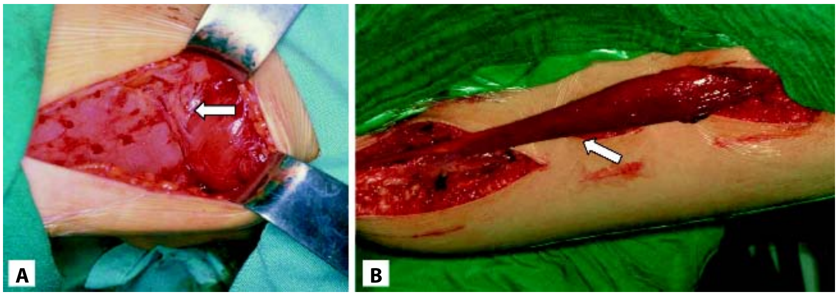

Fig. 31.12 Technical details of gracilis transplantation. **A** Branches of the femoral nerve to the gracilis muscle in situ. **B** Additional vessel in the distal part of the muscle. Note damage to the nerve supply and an additional distal vessel will lead to atrophy and fibrosis of the muscle

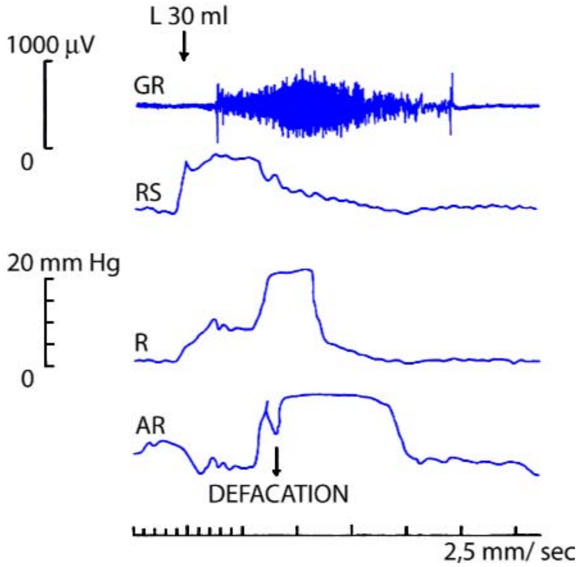

Fig. 31.13 Electromyography and electromanometry after gracilis muscle transplantation. Injection of 30 ml of air (*L*) into the rectosigmoid (*RS*), leading to a propulsisve wave in the rectum (*R*) and anorectum (*AR*). Defecation starts but is immediately interrupted by a vigorous contraction of the gracilis muscle. *GR* Electromyography of the gracilis muscle

not wear napkins. Nine patients remained completely incontinent. Manometrically, the anorectal pressure profile increased to values over 20 mmHg in 31 patients, and remained poor (under 15 mmHg) in 9 patients. The squeezing pressure profile increased to values higher than 20 mmHg in 38 children and remained poor (under 19 mmHg) in 2. The adaptation reaction was normal in 23 patients, remained shortened in 14, and was lacking in 3.

The increased pressure profile led to a maturation of the pulled-through colon, which acquired rectal qualities. During some years, however, the contractile force of the gracilis muscle diminished, probably because of atrophy of the muscle. Nevertheless these patients did not become incontinent, because in the meantime the pelvic floor muscles grew strong enough to balance the diminished anorectal resting pressure barrier. Holschneider's results are similar to those reported in the literature.

Recently, Kotobi et al. published their results with the Pickrell intervention in 23 children for anal incontinence secondary to ARM [33]. After a mean follow up of 6 years the functional result was estimated to be good in 25%, intermediate in 45% and poor in 30%.

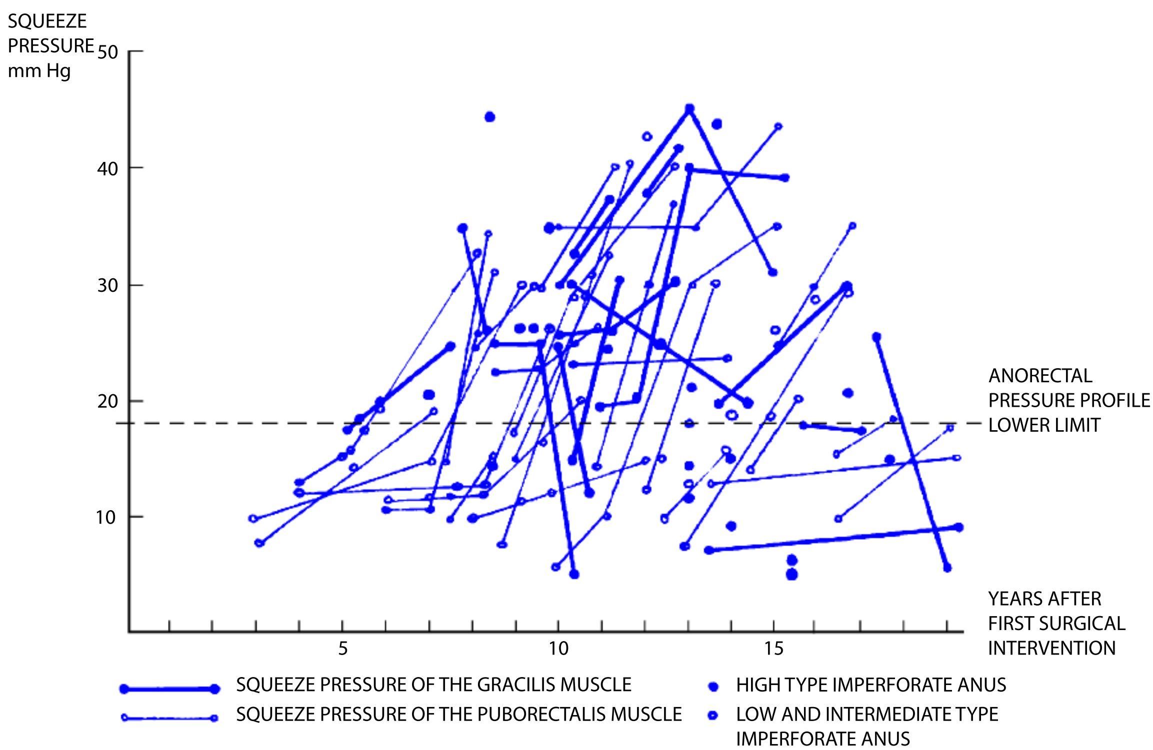

Fig. 31.14 Squeeze pressure of 33 patients, 5 years after gracilis muscle transplantation surgery. In seven patients the maximal squeeze pressure dropped down again in this follow-up period after a primary increase in all transplanted children. From Holschneider [55] with permission of the publishers

Clinical improvement could therefore be obtained in 70% of the cases. This corresponds to our earlier experience.

31.5.2 Dynamic Graciloplasty

In general, two local muscles are used for intensification of the striated muscle complex, especially the external anal sphincter, the gracilis, and the gluteus muscles. With both muscles, long-term muscle contractions are difficult to maintain due to muscle fatigue. The gracilis muscle, however, is technically much easier to transfer, and most activities of daily living and even sports are still possible. The patients are able to contract the gracilis without contraction of the other adductor mucles of the thigh in a neurophysiological adaptation process that takes about 7–9 months and needs intensive cooperation by the child. However, the gracilis muscle is not contracted under resting conditions and can only be used to support voluntary efforts to restain defecation if the patient wishes. Therefore it replaces only the voluntary function of the external anal sphincter, not the resting pressure barrier.

Experimental studies have shown that electrical stimulation of skeletal muscles can transform fatigue-prone muscles into fatigue-restistant muscles [19]. Electrical stimulation leads to morphologic and metabolic transformation of the muscle, with an increase in slow-twitch fibers (type I) [34–36] and a change in mitochondrial metabolism [37–39], thus enhancing the muscle's ability for continuous contraction. Therefore, in 1986 Baeten et al. [40–42] started to perform graciloplasty procedures with intramuscular electrodes connected to an electrical stimulator. More than 200 patients, especially adults, have been treated since then in his institution [42]. The mean age of the patients was 48 years; in only 28 patients the cause of stool incontinence was congenital. Of 200 patients, 76% were considered to have successful outcomes, but patients whose cause of incontinence was trauma or pudendal nerve pathology tended to respond better to this treatment than patients with ARM [43].

Koch et al. reported on 28 patients (median age of 25.5 years) born with ARM and operated on by dynamic graciloplasty [44]. Rectal inhibitory reflex was present in 17% of these patients. Satisfactory continence was achieved in 35%; however, 7.1% of his patients gained this continence score by additional bowel irrigation. Twenty-nine percent of patients were incontinent for loose stools, 36% were incontinent for formed stool. Satisfactory continence was reached in only 18% of patients with high ARM, but in 100% in patients with low ARM. However, in the total group of patients with dynamic graciloplasty, satisfactory continence was obtained in 76%.

Unfortunately many complications with this new technique have also been reported. Da Silva et al. [45] compared 2 groups of patients, 11 had an artificial bowel sphincter and 5 had the gracilis neosphincter. Complications occurred in six patients (50%), including three with fecal impaction (all artificial sphincter), three with device migration (two gracilis neosphincter, one artificial bowel sphincter), and in two patients with concomitant wound infection (one gracilis, one artificial sphincter). None of the patients had the device explanted. The quality of life scales increased with both the artificial and the gracilis sphincter.

Saunders et al. used the electrically stimulated gracilis neoanal sphincter in combination with continent colon conduits [46]. Seven patients (50%) had a successful outcome, defined as continent to solid and liquid stool, and eight (57%) reported some degree of improvement in their bowel function; an end stoma had to be formed in six (43%). Only one out of four boys treated by Rückauer [47] became almost continent. It seems, therefore, that this method could only be the last choice in really desperate cases of stool incontinence. It should only be performed in adults, but it is possible to create a gracilis transplantation in childhood and activate it with success later in adulthood [40,41,48].

31.5.3 Gluteus Maximus Transplant

Chetwood [49] first used the gluteus maximus muscle to support the rectum; further reports were made by Shoemaker [50], Bistrom [51], Prochiantz [52], Hentz [53], Skef et al. [54], and Holschneider [55] (Fig. 31.15). Skef et al. stated that the muscle "normally functions as an accessory muscle of anal continence" [54]. The coccygeal fibers of the gluteus are wrapped around the anal canal with its intact neuromuscular bundle. The pull is direct and sling-like. Shoemaker [50] reported that six out of six patients became continent, but there have been only isolated reports since then (Fig. 31.16). Kücükaydin et al. recently presented 5 patients with gluteus maximus transplant who in the follow-up period showed no continence problems.

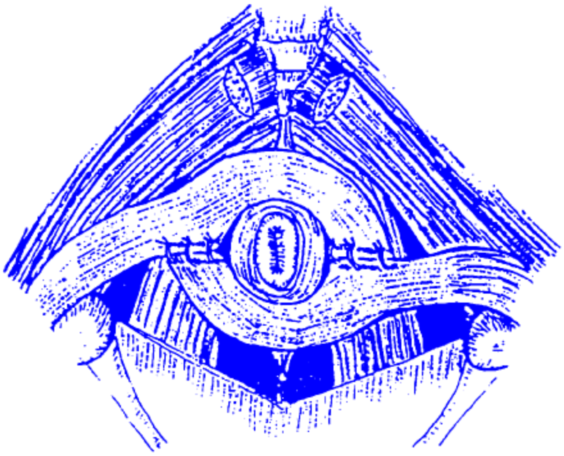

Fig. 31.15 Gluteus muscle transplant after Shoemaker [50]

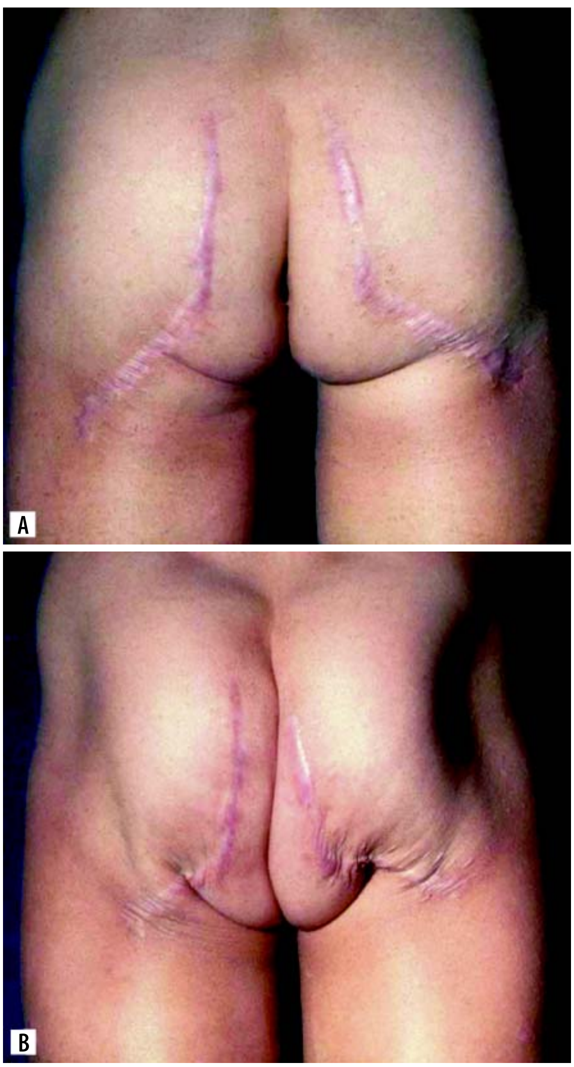

Fig. 31.16 Patient with gluteus maximus transplant (reproduced from Holschneider [55] with permission of the publishers) in the relaxed condition (**A**) and contracted (**B**)

31.6 Construction of Sphincters from Bowel Wall (Free Smooth Muscle Transplantation)

Free smooth muscle transplantation was introduced by Schmidt [56], who used this method to create a continent colostomy in adult patients after rectal resection. After resection of a segment of ascending colon, Schmidt excised the mucosa, prestretched the seromuscular cuff of the bowel by at least 120%, and applied it around the colostomy in a circular manner. Schmidt claimed development of a continent colostomy using this method in more than 300 patients. However, in manometric studies, Holschneider could not show any physiological sphincter mechanism. The smooth muscle graft was not able to relax, and the anorectal pressure barrier was simply the result of an external obstruction of the bowel. Most of his patients had to evacuate by enema. His results in children with imperforate anus were not encouraging; all four children acquired anal stenosis that had to be dilated by bouginage and/or a Heinieke/Mikulicz-plasty, resulting in incontinence.

31.7 Flap Smooth Muscle Transplantation

Because of the poor aforementioned results, Holschneider and Hecker [57] introduced the flap smooth muscle transplant for the treatment of high ARM. The procedure can be performed as a primary operation following the abdominosacroperineal pull-through procedure in high anomalies, or as a secondary procedure in failed cases.

As a primary operation, Holschneider first performs an abdominoperineal or abdominosacroperineal pull-through procedure. After having pulled down the colon through the puborectalis sling and the muscle complex, the mucosa is resected from the colon. The seromuscular cuff is turned back 180°, prestretched to 120–140% of the original length, and then attached to the serosa of the pulled-down colon with Vicryl sutures. The reversed muscle segment is brought back just above the pelvic floor (Figs. 31.17 and 31.18).

Postoperative studies in goats and in children have shown normal internal sphincter relaxations with an amplitude and duration directly proportional to the distending rectal volume, a normal anorectal pressure profile, and normal defecation behavior, with opening and closing of the anal canal during defecation (Figs. 31.19 and 31.20).

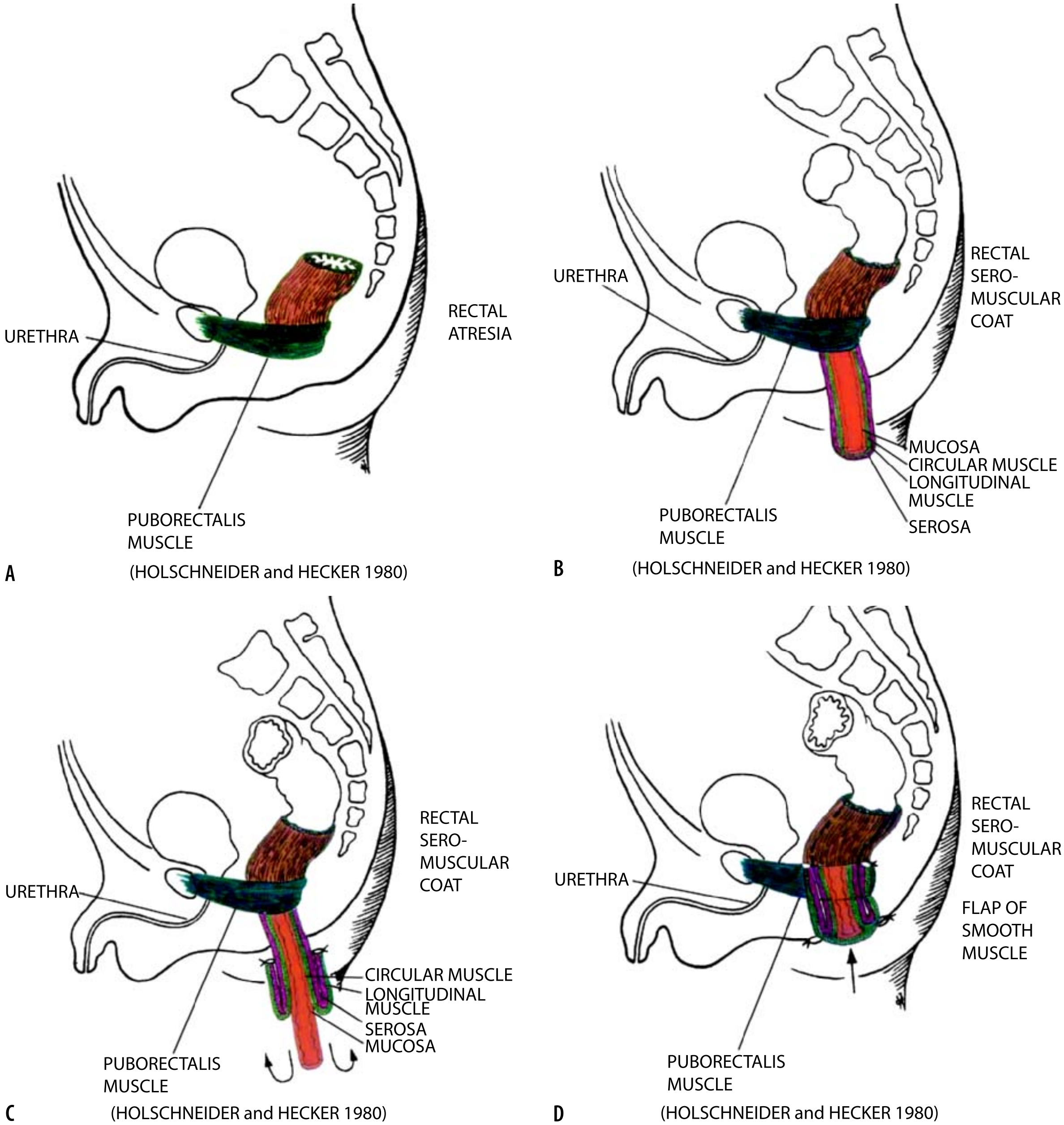

Fig. 31.17 **A**–**D** Reverse smooth muscle plasty according to Holschneider (reproduced from Holschneider and Hecker [58] with permission of the publisher). **A** High anorectal malformation. **B** After abdominoperineal pull-through of the colon to the perineum. **C** The seromuscular cuff is turned up 180 degrees orally and sutured to the serosa of the pulled-through colon. **D** The bowel is pulled back cranially within the puborectalis sling.

It is not necessary to prestretch the muscle cuff more than one-third over the resting condition, as proposed by Hofmann-von Kap-herr and Koltai [58], as this leads to stenosis.

Flap smooth muscle transplantation has been added to the Peña and de Vries PSARP as a secondary procedure. When the muscle complex is too hypoplastic to become a satisfactory sphincter, one can incise the seromuscular cuff of the pulled-down rectum or colon, detach the mucosa from the circular muscle layer, prestretch the seromuscular cuff to about 120–140%, and suture both seromuscular layers to each other. The most important point in this kind of procedure is the amount of prestretching of the muscle

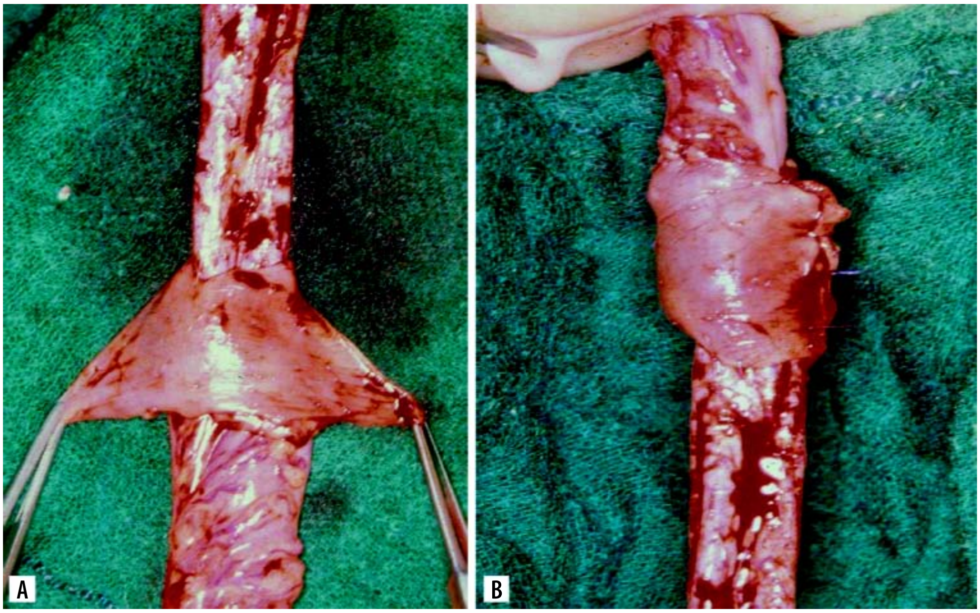

Fig. 31.18 Flapped smooth muscle transplantation in situ (from Holschneider and Hecker [58] with permission of the publisher). **A** Extirpation of the mucosa and turning back the smooth muscle layer. **B** Prestretching of the smooth muscle segment by about one-third over resting conditions and suturing to the serosa of the pulled-down colon

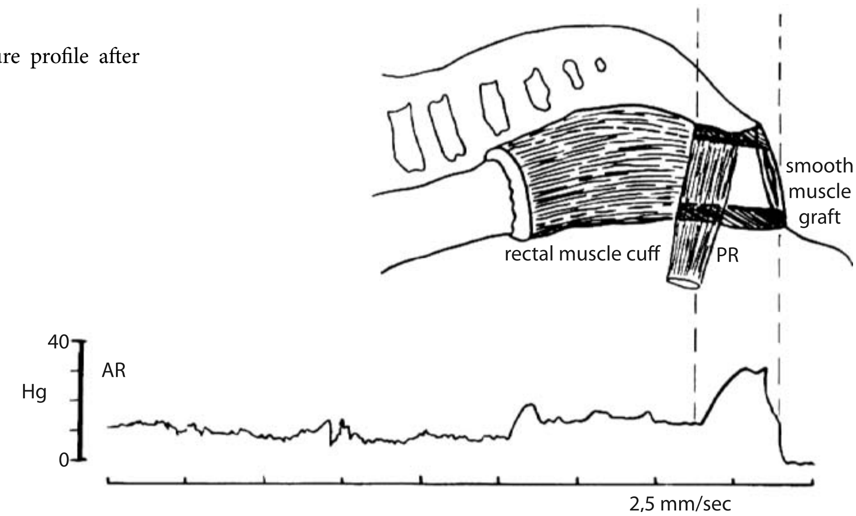

Fig. 31.19 Anorectal resting pressure profile after reverse smooth muscle plasty

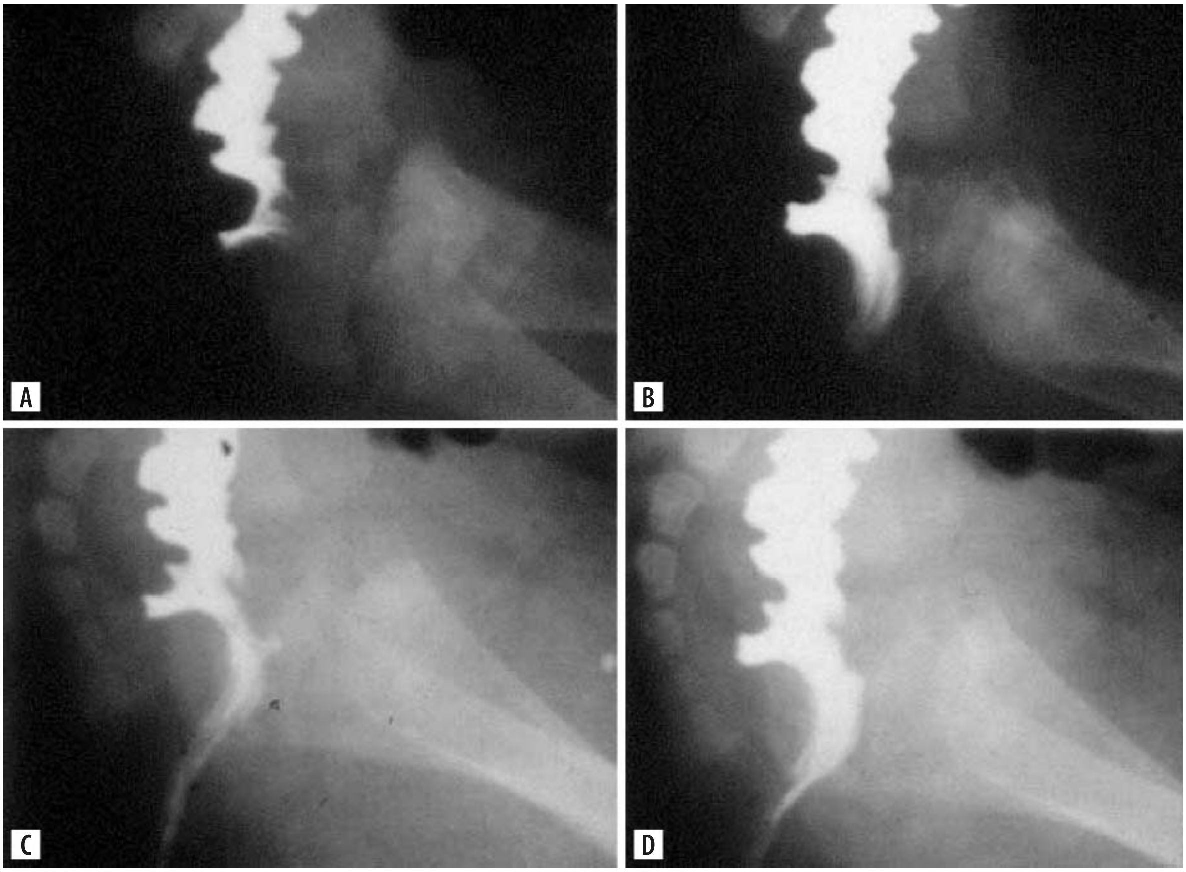

Fig. 31.20 **A–D** Defecography after reverse smooth muscle plasty. Narrowing and opening of the strengthened internal anal sphincter has been effected by the plasty

graft. Hofmann-von Kap-herr et al. [59] modified the Holschneider technique by creating a smooth- muscle fold-over double-plasty (SMSD-plasty), but prestretched the muscle cuff more severely, in some parts not only duplicating, but triplicating the muscle layers. In recent studies in the goat, Holschneider could show that such a high amount of prestretching results in stenosis and fibrosis of the muscle graft.

In Holschneider's series with his own technique, ten patients were operated upon. Eight of them showed normal internal sphincter relaxation and an anorectal resting pressure of over 20 mmHg, six children became completely continent or suffered from soiling only under stress conditions and diarrhea, one remained incontinent, and in the other child an anal stenosis required bouginage.

A secondary smooth muscle flap plasty was also performed in 12 children; 7 became completely continent, 2 remained incontinent because of stenosis and required bouginage, and 3 improved but continued to suffer from soiling under stress conditions and diarrhea. However, after the introduction of PSARP, this procedure has only been used by us in children with rectal prolapse combined with spinal lesions. In these patients with a flat bottom and neurogenic defecation disorders, a sacroperineal redo procedure combined with smooth muscle plasty and fixation of the rectum to the sacral or ischial fascia could be supported. In cases with simple ectopy of parts of the rectal mucosa, a perineal skin flap plasty is recommended. One should carefully distinguish between mucosal and rectal prolapse. In patients with mucosal prolapse the mucosa is not fixed by the longitudinal muscle fibers of the rectum at the perineal skin. Therefore, gliding in the submucosal layer is possible. In contrast, rectal prolapse involves all muscle layers. It occurs because the pulled-down rectum is not fixed to the lateral pelvic fascia after the lateral wings (ailerons latereaux) have been cut during preparation. In these cases a laparoscopic fixation of the rectum to the presacral fascia is the method of choice. However, this is not always possible and resection of the prolapsing bowel may be necessary. This should be performed with great care to the sphincter fibers.

31.8 Electrical Devices

Various attempts have been made to stimulate the sphincters tonically to gain continence [60–65]. In 1987 Christiansen and Lorentzen [66] developed an artificial anal sphincter (AMS 800; American Medical Systems, Minneapolis, MN, USA). This sphincter offers great benefit to patients with intractable fecal incontinence [66–70]. However, only half of the patients who underwent implantation of AMS 800 obtained satisfactory results. Amae et al. [71] and Nishi et al. [72] reported recently the development of an artificial sphincter using a shape memory alloy (AS-SMA) that had a simple structure and good durability. The aim of this device is to sandwich the bowel between two plates covered by silicon rubber. They are designed to close and to generate a pressure of 40 mmHg when electrical current is not supplied. When electrical current is supplied to the heaters, the temperature of the SMA plates immediately starts to rise, and the two plates bend to form an almond-like shape with a maximal gap of 33 mm between the two plates. First results in piglets with colostomy seem to be promising.

31.9 Secondary Procedures for Anal Prolapse or Stricture

31.9.1 Nixon Anoplasty

We recommend the "Nixon anoplasty" as the most satisfactory repair [73,74]. The lateral flap proctoplasty [73] was originally added to the "Kiesewetter-Rehbein" pull-through. It can be added to any form of pull-through and can also be used as a secondary procedure to correct postoperative prolapse.

This simplest form of anoplasty was fist described by Nixon in 1967 [73], but is not widely known. We and others use it regularly in both primary and secondary rectoplasties or in a few cases for revision anoplasty after stricture or mucosal ectopy. It creates a skin-lined anus, which reduces mucus discharge, preserves anal sensation, and is cosmetically satisfactory. A series is also reported by Davies and Cywes [74]. Nixon originally advocated incision of triangular areas of skin, but later suggests "kite"-shaped areas. The resulting anus is not so "tubular" and easier to clean.

Kite-shaped excisions of skin are taken from the perineum in front of and behind the anal site so that lateral advancement flaps are formed. (Fig. 31.21) The incision at the anal site must be long enough to form the circumference of an adequate anus (proximately 1.5–2 cm). The sutures include sphincter muscle as well as skin and bowel wall and are tied loosely to avoid cutting out. When the surgery is carried out secondarily for prolapse it may be necessary to excise a considerable amount of redundant "rectum," but control of prolapse depends mainly on the sutures attaching the bowel to the sphincter muscle. This has not proved to be necessary in the usual case treated by the minimal mobilization inversion anoplasty, but has been used on rare occasions in a slightly modified form to allow for the presence of the "Mollard flap," the large anterior and smaller posterior excision

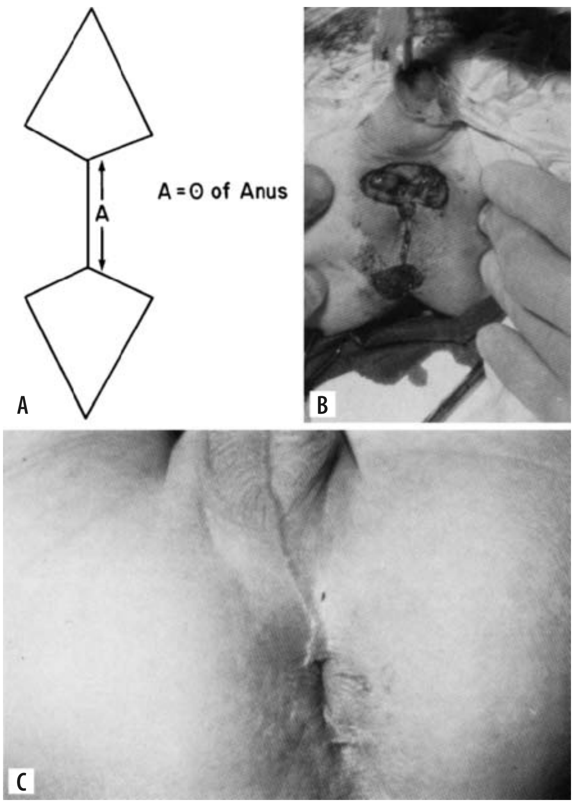

Fig. 31.21 **A**–**C** Nixon anoplasty (from Stephens and Smith [2] with permission of the publishers). **A** Incisions in the perineum, as shown by the *lines*. The kite-shaped areas of skin are excised. Each edge of the incision is sutured to 180° of the circumference of the bowel. By closing the "kites" of the skin flaps, the anastomosis ascends cephalically and creates a skin-lined anus. **B** The operative appearance before the closure of the "kites". **C** Postoperative appearance

forming posterolateral flaps, which retain an adequate base for their blood supply.

Mirsi et al. also described a sliding skin graft technique, and Freeman has utilized the penile foreskin to create a skin-lined anal canal. Millard and Rowe [75] described a double skin flap procedure, one flap being a dorsally based trap-door incision at the anal site, and the defect being covered by a second flap from the scrotum and thigh.

31.9.2 Mollard-Laberge Operation

In 1975, Mollard et al. [76] first reported his experience using an anterior perineal approach for high imperforate anus pull-through operations. For the reconstruction of the anoderm his group preferred a three-flap anoplasty, which has been described in detail by Caouette-Laberge et al. [77,2]. A posterior flap measuring 1.5 x 3 cm and lateral flaps 1.5 x 4 cm are created dorsal and lateral of the anus (Figs. 31.22 and 31.23). The V-shaped perineal flap is first raised to allow dissection parallel and just posterior to the urethra, as in the original Mollard technique (Fig. 31.22 A, B). The external anal sphincter is then identified in its subcutaneous portion, separated bluntly in its center, and gradually divided in the midline anteriorly up to the level of the puborectalis (Fig. 31.22 C). The colon is brought just below the puborectalis. The posterior flap is elevated, rotated by 180° and sutured into a 1-cm slit made on the posterior wall of the colon (Fig. 31.22 D).

The left flap is raised with enough subcutaneous tissue to protect its blood supply. It is then spiraled in the anal canal, its tip going beyond the midline. The interrupted 5-0 resorbable sutures include skin,

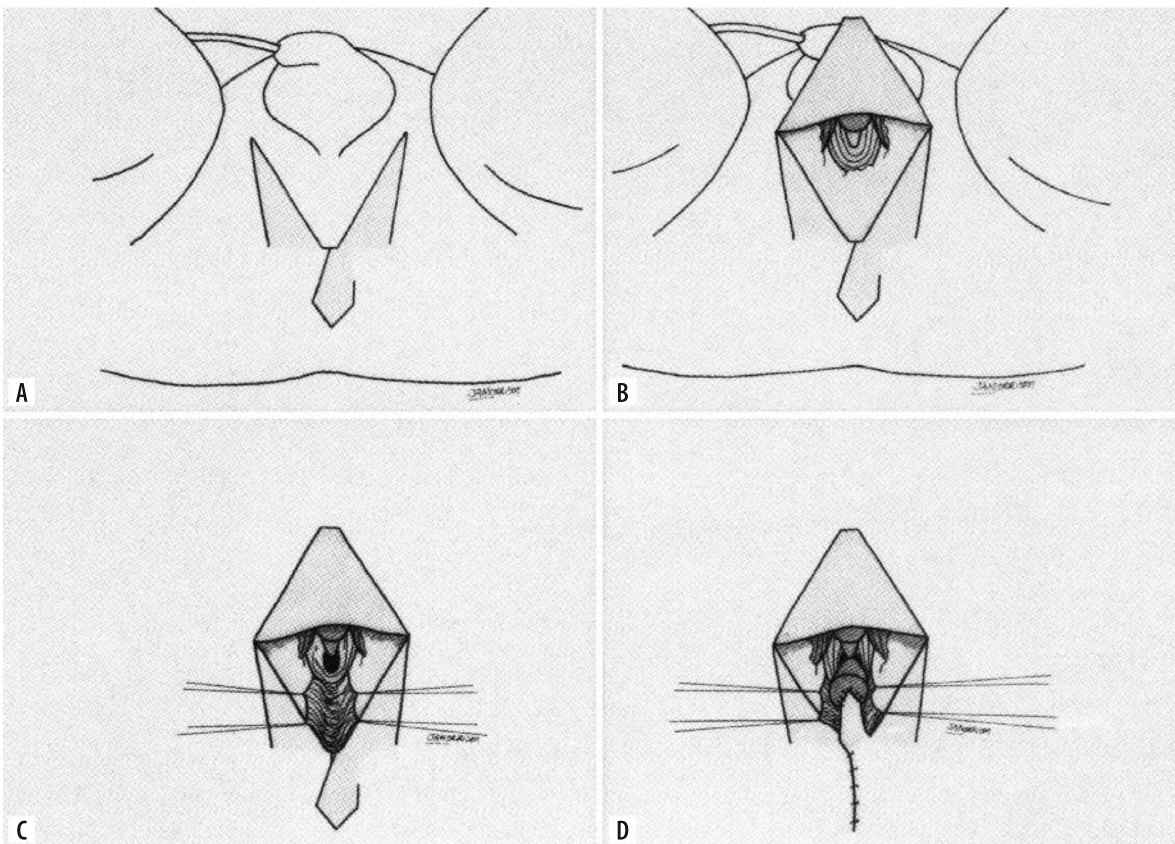

Fig. 31.22 Three-flap operation of Caouette-Laberge [79,80]. **A** Outline of the incision. The posterior flap measures 1.5 x 3 cm and the lateral flaps 1.5 x 4 cm. **B** The V-shaped perineal flap is first raised to allow dissection parallel and just posterior to the urethra. **C** The external sphincter is then identified in its subcutaneous portion, separated bluntly in its center and gradually divided in the midline anteriorly up to the level of the puborectalis. **D** The colon is brought below the puborectalis. The posterior flap is elevated, rotated 180° and sutured into a 1-cm slit made on the posterior wall for the colon

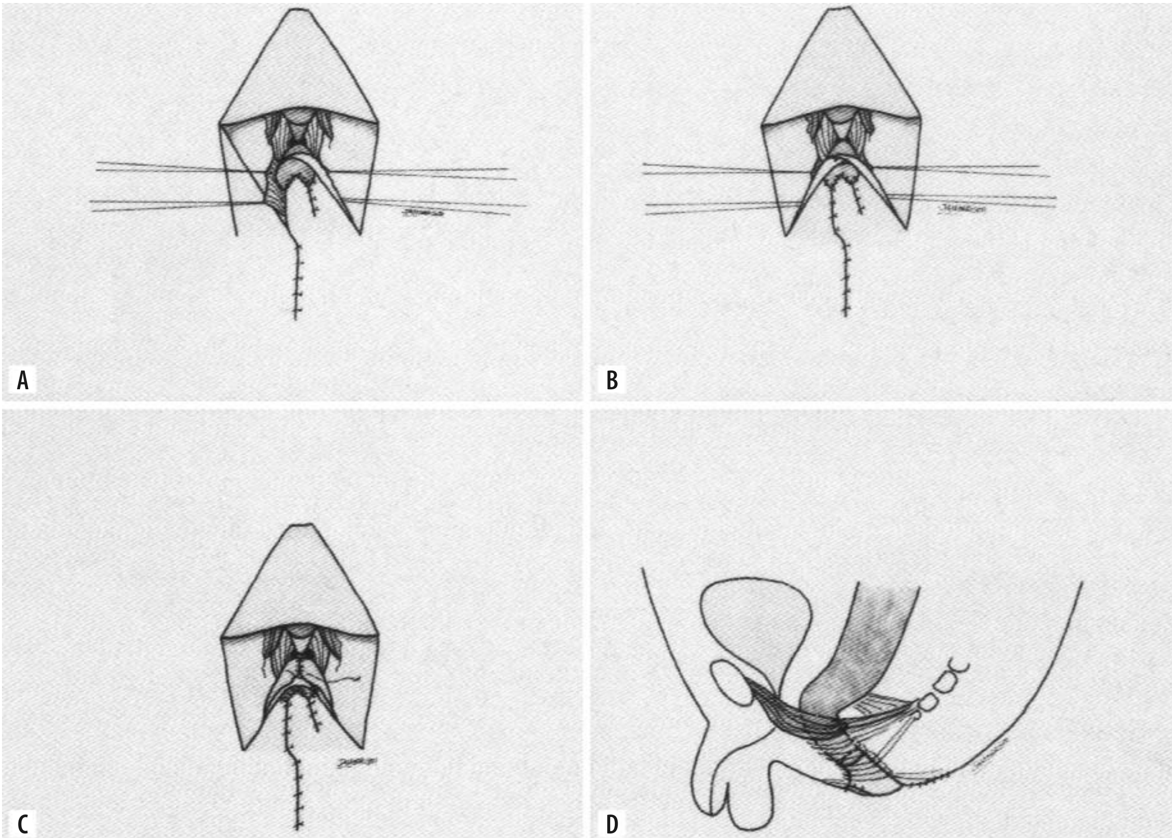

Fig. 31.23 Three-flap operation of Caouette-Laberge [79,80] continued. **A** The left flap is raised with enough subcutaneous tissue to protect its blood supply. It is then spiraled into the anal canal, its tip going beyond the midline. The interrupted 4-0 resorbable sutures included skin, sphincter fibers, and colon. **B** The right perineal flap is now in place overlapping the left one across the midline. **C** The external sphincter fibers are then closed around the distal colon and the newly created anal canal. **D** Sagittal view of the completed operation. the puborectalis is intact and the external sphincter surrounds a skin-lined anal canal

sphincter fibers, and colon (Fig. 31.23 A). The right perineal flap is now in place, overlapping the left one across the midline (Fig. 31.23 B). The external sphincter fibers are then closed around the distal colon and the newly created anal canal (Fig. 31.23 C). After the procedure the external anal sphincter should surround a skin-lined anal canal as in the normal anatomy (Fig. 31.23 D).

We use this technique not for primary reconstruction because the goal of this approach, a skin-lined anal canal for better discrimination, can be achieved easier using the techniques of Peña or Nixon technique. However, the three-flap procedure is very useful and superior to Nixon anoplasty in cases of severe rectal prolapse or circular mucosal ectopia.

Daily dilatation with Hegar bougies are started on the 10th postoperative day. As the caliber of the anal canal increases, dilatations are performed less frequently and are discontinued 6–12 weeks postoperatively. The anus is now growing itself and should remain elastic.

Very good results with this three-flap anoplasty in the repair of ARM have recently been reported by Yazbeck et al. [78], Bass et al. [79] and Becmeur et al. [80]. A cruciate incision creating four flaps was described by Dorio de Amaral in 1999 [81] as an approach for perineal anorectoplasty.

In cases with severe rectal prolapse following PSARP for ARM, the three-flap technique is of course only the last step of the surgical repair. At first, circumferential incision and mobilization of the redundant rectum with preservation of anal skin and muscle fibers has to be performed, as recently shown by Belizon et al. [82].

References

1. Stephens FD, Smith ED (1971) Anorectal Malformations in Children. Year Book Medical Publishers, Chicago
2. Stephens FD, Smith ED (1988) Anorectal Malformations in Children: update. March of Dimes Foundation Birth Defects. Original Article Series Volume 24, Number 4. Alan R. Liss, New York
3. Kiesewetter WB, Jeffries MR (1981) Secondary anorectal surgery for the missed puborectalis muscle. J Pediatr Surg 16:921–926
4. Peña A (1983) Posterior sagittal anorectoplasty as a secondary operation for the treatment of fecal incontinence. J Pediatr Surg 18:762–773
5. Peña A (1990) Atlas of Surgical Management of Anorectal Malformations. Springer-Verlag, New York
6. De Vries PA, Peña A (1982) Posterior sagittal anorectoplasty. J Pediatr Surg 17:638–643
7. Peña A, de Vries PA (1982) Posterior sagittal anorectoplasty. Important technical considerations and new applications. J Pediatr Surg 17:796–811
8. Kottmeier PK (1966) A physiological approach to the problem of anal incontinence through use of the levator ani as a sling. Surgery 60:1262–1266
9. Kottmeier PK, Dziadiw R (1967) The complete release of the levator ani sling in fecal incontinence. J Pediatr Surg 2:111–117
10. Kottmeier PK, Velcek FT, Klotz DH, Coren CV, Hansbrough F, Price AP (1986) Results of levatorplasty for anal incontinence. J Pediatr Surg 24:647–650
11. Gross RE (1970) „An Atlas of Children‘s Surgery. W.B. Saunders, Philadelphia
12. Puri P, Nixon HH (1976) Levatorplasty: a secondary operation for fecal incontinence following primary operation for anorectal agenesis. J Pediatr Surg 11:77–82
13. Hakelius L (1975) Free autogenous muscle, transplantation in two cases of total anal incontinence. Acta Chir Scand 141:69–75
14. Hakelius L, Gierup J, Grotte G, Jorulf H (1978) A new treatment of anal incontinence in children: free autogenous muscle transplantation. J Pediatr Surg 13:77–82
15. Hakelius L, Gierup J, Grotte G (1980) Further experience with free autogenous muscle transplantation in children for anal incontinence. Z Kinderchir 31:141–147
16. Grotte G, Hakelius L, Trykberg T, Rasmundsson T (1984) Nine years of free autogenous muscle transplantation for anal incontinence in children. Z Kinderchir 39:80–82
17. Mollard P, Valla V, de Beaujeu MJ (1979) Incontinence anale apres imperforation: traitment per transplantation musculaire libre. Chir Pediatr 20:205–208
18. Holschneider AM, Hecker WC (1981) Flapped and free muscle transplantation in the treatment of anal incontinence. Z Kinderchir 32:244–258
19. Holle J. Freilinger G, Mamoli B, Spangler HP, Braun F, Krenn R (1975) Neue Wege zur chirurgischen Rekonstruktion der analen Sphinktermuskulatur. Wien Med Wochenschr 125:735–743
20. Holschneider AM, Amano S, Urban A, Lohrs U, Biese K, Donhauser G, Kampf B (1985) Free and reverse smooth muscle plasty in rats and goats. Dis Colon Rectum 28:786–794
21. Pickrell KL, Georziade N, Crawford H, et al (1952) Construction of a rectal sphincter and restoration of anal continence by transplanting the gracilis muscle. Ann Surg 135:853–862
22. Feuchtwanger MM, Ben-Hur N (1968) The surgical correction of anal incontinence by complete perineoplasty: case report. Plast Reconstr Surg 41:268–272
23. Raffensperger J (1979) The gracilis sling for fecal incontinence. J Pediatr Surg 14:794–797
24. Landeen JM, Habal MB (1979) The rejuvenation of the sphincteroplasty. Surg Gynecol Obstet 149:78–80
25. Holschneider AM, Poschl J, Kraeft H, Hecker WC (1979) Pickrell‘s gracilis muscle transplantation and its effect on anorectal continence. A 5-year prospective study. Z Kinderchir 27:135–143
26. Holschneider AM, Hecker WC, Schimmel (1983) Ergebnisse kontinenzverbessernder Operationen nach anorektalen Fehlbildungen. In: Hoffmann-von Kap-Herr S (ed) Anorektale Fehlbildungen. Gustav Fischer Verlag, Stuttgart
27. Brandesky G, Holschneider AM (1976) Operations for the improvement of fecal incontinence. Prog Pediatr Surg 9:105–114
28. Berger D, Genton N, Berger K (1979) Efficacité et limites de I‘ano-myoplastique selon Pickrell dans le traitement de I‘incontinence postoperatoire des malformations ano-rectales. Chir Pediatr 20:57–62
29. Brandesky G, Geley L, Janout D (1984) Results of the modified Hartl gracilis plasty. Prog Pediatr 17:115–122
30. Holschneider AM, Lahoda F (1974) Elektromyographische und elektromanometrische Untersuchungen zur gracilis plastik nach Pickrell. Z Kinderchir 14:288
31. Hartl M (1972) A modified technique of gracilis plastic. Paediatr Paedol 2:99–107
32. Dittertová L, Grim M (1983) Puborektalisersatz durch modifizierte Grazilis-lastik. In: Hoffmann-von Kap-herr S (ed) Anorektale Fehlbildungen. Gustav Fischer Verlag, Stuttgart
33. Kotobi H, Forin V, Larroquet M, Khairouni A, Loch P, Grapin C, Audry G (2000) Pickrell intervention in children for anal incontinence secondary to anorectal malformations. Ann Chir 125:954–960
34. Pette D, Vrova G (1992) Adaptation of mammalian skeletal muscle fibers to chronic electrical stimulation. Rev Physiol Biochem Pharmacol 120:115–202

35. Salmons S, Vroba G (1996) The influence of activity on some contractile characteristics of mammalian fast and slow muscle. J Physiol 201:535–549
36. Chaques JC, Grandjean PA, Carpentier A (1996) Dynamic cardiomyoplasty: experimental cardiac wall replacement with a stimulated skeletal muscle. In: Chin RJC (ed) Cardiomyoplasty and Muscle-powered Devices. Futura, New York, pp 59–84
37. Mayne CN, Sutherland H, Jarvis JC, et al (1996) Induction of a fast oxidative phenotype by chronic muscle stimulation: histochemical and metabolic studies. Am J Physiol 270:313–320
38. Reichmann H, Wasl R, Simeneau JA, et al (1991) Enzyme activities of fatty acid oxidation and the respiratory chain in chronically stimulated fast twitch muscle of the rabbit. Pflügers Arch 428:572–574
39. Rouanet P, Bacou F (1993) Changes in fibre type, metabolic character and acetylcholinesterase forms in rabbit skeletal muscle following stretch and electrical stimulation. Neuromuscul Disord 3:401–405
40. Baeten CG, Bailey HR, Bakka A, et al (2000) Safety and efficacy of dynamic graciloplasty for fetal incontinence: report of a prospective multicenter trial. Dynamic graciloplasty therapy study group. Dis Colon Rectum 43:743–751
41. Baeten C, Spaans F, Fluks A (1988) An implanted neuromuscular stimulator for fecal incontinence following previously implanted gracilis muscle: report of case. Dis Colon Rectum 31:134–137
42. Baeten C, Uludag O, Rongen MJ (2001) Dynamic graciloplasty for fecal incontinence. Microsurgery 21:230–234
43. Baeten CG, Konsten J, Heineman E, Soeters PB (1994) Dynamic graciloplasty for anal atresia. J Pediatr Surg 29:922–925
44. Koch SM, Uludag Ö, Rongen M-J, Baeten CG, Gemert W (2004) Dynamic Graciloplasty in patients born with anorectal malformation. Dis Colon Rectum 47:1711–1719
45. da Silva GM, Jorge MN, Belin B, Nogueras JJ, et al (2004) New surgical options for fecal incontinence in patients with imperforate anus. Dis Colon Rectum 47:204–294
46. Saunders JR, Williams NS, Eccersley AJ (2004) The combination of electrically stimulated neoanal sphincter and continent colonic conduit: a step forward to total anorectal reconstruction? Dis Colon Rectum 47:354–363
47. Rückauer KD (2001) Dynamic graciloplasty in children with fecal incontinence: a preliminary report. J Pediatr Surg 36:1036–1039
48. Seccia M, Banti P, Zocco G, Viacava P (2001) Restoration of fecal continence with chronic electrostimulation of gracilis muscle 17 years after Pickrell's operation. Int J Colorectal Dis 16:391–394
49. Chetwood CH (1902) Plastic operation for restoration of the sphincter ani, with report of a case. Med Rec 61:529–534
50. Shoemaker J (1909) Les nouveaux procedées opértoires pour la reconstruction du sphincter anal. Semin Med Paris 29:160
51. Bistrom O (1944) Plastischer Ersatz des M. sphinkter ani. Acat Chir Scand 90:431–448
52. Prochiantz A (1979) Myoplastic fessière a visée sphinctérienne pour le traitement des incontinences fecales des agenesies sacrées et des malformations anorectales operées. Chir Pediatr 20:63–67
53. Hentz VR (1982) Construction of a rectal sphincter using the origin of the gluteus maximus muscle. Plast Reconstr Surg 70:82–85
54. Skef Z, Radhakrishnan J, Reyes HM (1983) Anorectal continence following sphincter reconstruction utilizing, the gluteus maximus muscle. A case report. J Pediatr Surg 18:779–781
55. Holschneider AM (1983) Elektromanometrie des Enddarms. Diagnostic und Therapie der Inkontinenz und der chronischen Obstipation, 2nd edn. Urban und Schwarzenberg, München-Wien-Baltimore
56. Schmidt E (1978) Die chirurgische Behandlung der analen Inkontinenz mittels frei transplantierter autologer körpereigener Darmmuskulatur. Chirurg 49:320–321
57. Holschneider AM, Hecker WC (1984) Smooth muscle reverse plasty. A new method to treat anorectal incontinence in infants with high anal and rectal atresia. Results after gracilis plasty and free muscle transplantation. Prog Pediatr Surg 17:131–145
58. Hofmann-von Kap-herr S, Koltai I (1981) Neue Wege in der Behandlung der anorektalen Inkontinenz. Z Kinderchirurg 32:258–270
59. Hofmann-von Kap-herr S, Koltai IL, Tennant LJ (1985)Anal sphincter substitute using autologous smooth muscle in a fold-over, half-cylinder, double plasty (SFD-plasty): a new method of treatment of anorectal incontinence. J Pediatr Surg 20:134–137
60. Caldwell KP (1963) The electrical control of sphincter incompetence. Lancet 2:174–175
61. Fischer BH, Vonder Moser HA (1964) Electronic management of fecal incontinence. JAMA 207:1897–1898
62. Hopkinson BR, Lightwood R (1966) Electrical treatment of anal incontinence. Lancet 1:297–298
63. Dickson AS, Nixon HH (1968) Control by electronic stimulator of incontinence after operation for anorectal agenesis. J Pediatr Surg 3:696–701
64. Toudoire A, Prochaintz A (1970) Stimulator of the internal sphincter in childhood. Ann Chir Infant 11:349–352
65. Glen ES (1971) Intra-anal electrode: a stimulus to bowel and bladder control. J Pediatr Surg 6:138–142
66. Christiansen J, Lorentzen M (1987) Implantation of artificial sphincter for anal incontinence. Lancet I:244–245
67. Hajvassilou CA, Carter KB, Finlay IG (1997) Assessment of a novel implantable artificial anal sphincter. Dis Colon Rectum 40:711–717

68. Lehur PA, Michot F, Denis P, et al (1996) Results of artificial sphincter in severe anal incontinence. Report of 14 consecutive implantations. Dis Colon Rectum 39:1352–1355
69. Lehur PA, Glemain P, Bruley dec Varannes S, et al (1998) Outcome of patients with an implanted anal sphincter for severe fecal incontinence. A single institution report. Int J Colorectal Dis 13:88–92
70. Vaizey CJ, Kamm MA, Gold DM, et al (1998) Clinical, physiological and radiological study of a new purpose-designed artificial bowel sphincter. Lancet 351:105–109
71. Amae S, Wada M, Ohi R, et al. (2001) Development of an implantable artificial anal sphincter by the use of the shape memory alloy ASAIO J 47:346–350
72. Nishi K, Kamiyama T, Wada M, Amae S (2004) Development of an implantable artificial anal sphincter using a shape memory alloy. J Pediatr Surg 39:69–72
73. Nixon HH (1967) A modification of the proctoplasty for rectal agenesis. Pamietrik I-ss Zjazdu Naukowego Polskiego Towarzystwa Chirurgow Dzieciecyck, Warszawa 5–7, x
74. Davies MRQ, Cywes S (1984) The use of a lateral skin flap perineoplasty in congenital anorectal malformations. J Pediatr Surg 19:577–580
75. Millard DR, Rowe MI (1982) Plastic surgical principles in high imperforate anus. Plast Reconstr Surg 69:399–409
76. Mollard P, Maréchal JM, Jaubert de Beaujeu M (1975) Le repérage de la sangle du releveur au cours du traitement des imperforations anales hautes. Ann Chir Inf 15:461–468
77. Caouette-Laberge L, Yazbeck S, Laberge JM, Ducharme JC (1987) Multiple-flap anoplasty in the treatment of rectal prolapse after pull-through operations for imperforate anus. J Pediatr Surg 22:65–67
78. Yazbeck S, Luks FI, St.Vil D (1992) Anterior perineal approach and three-flap anoplasty for imperforate anus: optimal reconstruction with minimal destruction. J Pediatr Surg 27:190–195
79. Bass J, Rubin LZ, Walton JM, Cada M (2001) Combined posterior sagittal and three-flap anoplasty in the repair of anorectal anomalies. J Pediatr Surg 36:711–714
80. Becmeur F, Hofmann-Zango I, Jouin H, et al (2001) Three-flap anoplasty for imperforate anus: results for primary procedure or for redoes. Eur J Pediatr Surg 11:311–314
81. Dòria de Amaral F (1999) Treatment of anorectal anomalies by anterior perineal anorectoplasty. J Pediatr Surg 34:1315–1319
82. Belizon A, Levitt MA, Shoshany G, Rodriguez G, Peña A (2005) Rectal prolapse following posterior sagittal anorectoplasty for anorectal malformations. J Pediatr Surg. 40:192–196

32 Treatment of Chronic Constipation and Resection of the Inert Rectosigmoid

Marc A. Levitt and Alberto Peña

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32.1 Introduction

The colon absorbs water from the stool and serves a reservoir function. These processes depend on colonic motility, which is an area of physiology that is not well understood, and for which treatments of problems are limited. In normal individuals the rectosigmoid stores the stool, and every 24–48 h develops active peristaltic waves indicating that it is time to empty. A normal individual feels this sensation and decides when to relax the voluntary sphincter mechanism. Patients with anorectal malformations, as discussed in Chaps. 29, 30 and 33, lack a normal anal canal, have deficient sphincters, and have an accompanying motility disorder, usually hypomotility [1]. Their ability to have a voluntary bowel movement depends on these three factors. Solid stool allows for distension of the distal rectum, and proprioception allows the child to detect this. It is for this reason that loose stools make their ability to be fecally continent much less likely.

If a child is fecally continent (i.e., those with a good-prognosis anorectal defect, a normal sacrum, good sphincters, and an intact rectosigmoid), then management involves the treatment of constipation using laxatives, which help provoke peristalsis and overcome the dysmotility disorder. Patients in whom the rectosigmoid was resected, a common part of older operations for anorectal malformations, have hypermotility and require treatments that slow down the colon. Unfortunately, most of these patients are fecally incontinent because their lack of anal canal, and deficient sphincters cannot hold back the loose stool. They do not detect rectal fullness and thus cannot rely on proprioception for help with a voluntary bowel movement. For patients with fecal incontinence, a bowel management program is a way to artificially keep patients clean (Chaps. 29, 30 and 33). For the majority of patients (75%), management consists of avoidance and treatment of constipation, and toilet-training strategies.

Constipation in anorectal malformations is extremely common, particularly in the more benign types [2]. When left untreated, constipation can be extremely incapacitating, and in its most serious forms can produce a form of fecal incontinence known as overflow pseudoincontinence. Diet impacts colonic motility, but its therapeutic value is negligible in the most serious forms of constipation. It is true that many patients with severe constipation suffer from psychologic disorders, but a psychologic origin cannot explain the severe forms as it is not easy to voluntarily retain the stool when an otherwise autonomous rectosigmoid peristalses. Passage of large, hard pieces of stool may provoke pain and make the patient behave like stool retainers. This may complicate the problem of constipation; but it is not the original cause.

The clinician must decide which type of patient he or she is dealing with. Patients with a good prognosis are those more likely to have constipation, and while they are in disapers, aggressive, proactive treatment of their constipation is the best approach. Once they reach the age of toilet training, the child must have the capacity for voluntary bowel movements before employing treatment for constipation. Otherwise, theys require bowel management and enemas.

Most of these patients suffer from different degrees of dilatation of the rectum and sigmoid, a condition defined as megarectosigmoid, due to a hypomotility disorder that interferes with complete emptying of the rectosigmoid [1]. These children are born with a good-prognosis type of anorectal defect and underwent a technically correct operation, but did not receive appropriate treatment for constipation. They therefore developed fecal impaction and overflow pseudoincontinence. The impaction needs to be removed with enemas and colonic irrigations to clean the megarectosigmoid. The constipation is subsequently treated with the administration of large doses of laxatives. The dosage of the laxative is increased daily until the right amount of laxative is reached in order to completely empty the colon every day. If medical treatment proves to be extremely difficult because the child has a severe megasigmoid and requires an enormous amount of laxatives to empty, the surgeon can offer a resection of the sigmoid colon. After the sigmoid resection, the amount of laxatives required to treat these children can be significantly reduced or even eliminated. Before performing this operation it is mandatory to confirm that they are definitely suffering from overflow pseudoincontinence rather than true fecal incontinence with constipation. Failure to make this distinction may lead to an operation in which a fecally incontinent constipated child is changed to one with a tendency to have loose stool, which will make them much more difficult to manage (see Chaps. 29, 30 and 33).

When managed from the beginning, with aggressive treatment of constipation, children with a good prognosis should toilet train without difficulty. When constipation is not managed properly and a patient presents after many years, they behave much like children with idiopathic constipation, and may have overflow pseudoincontinence.

On occasion, the constipation in anorectal malformations is attributed to Hirschsprung's disease, and it is not uncommon that clinicians perform a rectal biopsy. In our experience, Hirschsprung's disease is no more common in patients with anorectal malformations than in the general population and we do not routinely biopsy these patients.

Constipation in anorectal malformations is a self-perpetuating disease. A patient who suffers from a certain degree of constipation and who is not treated adequately only partially empties the colon, leaving larger and larger amounts of stool inside the rectosigmoid, which results in greater degrees of megasigmoid. It is clear that dilatation of a hollow viscus produces poor peristalsis, which explains the fact that constipation leads to fecal retention, thereafter megacolon, which exacerbates the constipation. In addition, the passage of large, hard pieces of stool may produce anal fissures, which result in a reluctance by the patient to have bowel movements.

The clinician must accept the fact that the dysmotility associated with anorectal malformations is essentially incurable. It is manageable, however, but requires careful follow-up for life. Most importantly, it must be anticipated and treated early, even within weeks following the colostomy closure. Treatments cannot be given on a temporary basis; once they are tapered or interrupted, constipation recurs.

Some clinicians treat such patients with colostomies or colonic washouts via a catheterizable stoma or button device, and monitor the degree of colonic dilatation with contrast studies. Once the distal colon regains a normal caliber, the physician assumes that the patient is cured and the colostomy is closed or the washouts are discontinued with the predictable return of symptoms. Washouts are really only for patients with fecal incontinence who are incapable of having voluntary bowel movements and thus require a daily irrigation to empty. The patients described in this chapter are capable of emptying their colon with the help of adequate doses of laxatives.

Determining if the patient is continent or incontinent is the challenge. If incontinent, washouts with a bowel management regimen are appropriate. If continent, then aggressive management of the constipation after ensuring disimpaction is the appropriate treatment. These latter patients have a good-prognosis anorectal defect, good sacrum, and good sphincters.

Fecal impaction is a stressful event defined as a condition of retained stool for several days or weeks, crampy abdominal pain, and sometimes tenesmus. When laxatives are prescribed to such a patient the result is exacerbation of the crampy abdominal pain and sometimes vomiting. This is a consequence of an increased colonic peristalsis (produced by the laxative) acting against a fecally impacted colon. Therefore, disimpaction, proven by x-ray, must precede the initiation of laxative therapy.

Soiling of the underwear is an ominous sign of bad constipation. A patient who at an age of bowel control soils the underwear day and night and basically does not have spontaneous bowel movements may have "overflow pseudoincontinence." These patients behave as fecally incontinent individuals. When the constipation is treated adequately, the great majority of these pseudoincontinent children regain bowel control. Of course, this clinical presentation may also occur in a patient with anorectal malformation and true fecal incontinence. In such a patient with a poor-prognosis defect, poor sacrum, and poor sphincters, bowel management with a daily enema is needed (see Chaps. 29, 30 and 33).

When uncertain, we usually start the 3.5 to 4 year-old child having trouble with toilet training on a daily enema, and once clean with this regimen, and if they have the potential for bowel control, then try a laxative program. A contrast enema with a hydrosoluble material (never barium) is the most valuable tool that, in the constipated patient, usually shows a megarectosigmoid with dilatation of the colon all the way down to the level of the levator mechanism (Fig. 32.1). There is usually a dramatic size discrepancy between a normal transverse and descending colon and the very dilated megarectosigmoid. The size of the colon guides the dosing of the laxatives, and it seems that the more localized the dilatation of the rectosigmoid, the better the results of a sigmoid resection. The contrast study may show an absence of the rectosigmoid (Fig. 32.2), which may have been resected during the original operation, and correlates with hypermotility and usually fecal incontinence.

Some clinicans use rectal and colonic manometry in the evaluation of these patients; however, more objective techniques are needed. Manometry is performed by placing balloons at different levels of the colon and recording the waves of contraction [3] or the electrical activity [4]. Scintigraphy, a nuclear medicine tool, is also being used to assess colonic motility [5]. These are sophisticated tools but at present, their help as guides for therapeutic decisions is lacking. The key information the surgeon needs is to know if and where a colonic resection would provide benefit to the patient. Histologic studies of the colon in these patients mainly show hypertrophic smooth muscle in the area of the dilated colon and normal ganglion cells, but more sophisticated histopathologic investigations will hopefully soon yield results. Further investigations in this area will enhance our knowledge about colonic dysmotility in this patients, and thereby guide therapy.

32.2 Treatment

Patients with anorectal malformations and severe constipation in whom dietary measures or gentle laxatives do not work require a more aggressive regimen. It cannot be overemphasized that the treatment must start early. Drugs designed to increase the motility of the colon are best, as opposed to medications that are only stool softeners. As discussed, softening of the stool without improving the colonic motility will likely make the patient worse, because with soft stool they no longer have control, whereas they do reasonably well with solid stool that allow them to feel distension of the rectum.

Our protocol of treatment of these patients includes a trial of medical management [6]. If the patients respond to this treatment but require an enormous amount of laxatives to empty, then an operation is considered. The regimen uses the same medications as have been tried previously in many cases, but the protocol is different in that the dosage is adapted to the patient's response, the response is monitored daily with an abdominal radiograph, and the laxative dose adjusted if necessary. The patient has almost always been receiving less laxative than they required.

When a patient with anorectal malformations presents with constipation, the steps of treatment are as follows.

Fig. 32.1 Megarectosigmoid (Reprinted from Current Problems in Surgery, 39, Peña A., Levitt M. Colonic Inertia Disorders in Pediatrics, p 681, Mosby (2002), with permission from Elsevier.)

Fig. 32.2 Absence of rectosigmoid

32.2.1 Disimpaction

The disimpaction process is a vital and often neglected step. The routine first includes the administration of enemas until the patient is disimpacted (confirmed radiologically). If the patient remains impacted they are given a balanced electrolyte solution via a nasogastric tube in the hospital, and the enema regimen is continued. If this is unsuccessful, a manual disimpaction under anesthesia may be necessary. It is important to remember not to prescribe laxatives to a patient that is fecally impacted. To do so may provoke vomiting and severe abdominal pain. In addition, the patient will become reluctant to take laxatives because he or she is afraid of those symptoms.

32.2.2 Determination of the Laxative Requirement in a Disimpacted Patient

Once the patient has been disimpacted, an arbitrary amount of laxative is started, usually a senna derivative. The initial amount is based on the information that the parents give about the previous response to laxatives, and the subjective evaluation of the megasigmoid on the contrast enema. The empiric dose is given and the patient is observed for the next 24 h. If the patient does not have a bowel movement in the 24 hours after giving the laxative, it means the laxative dose was not enough, and it must be increased. An enema is also required in order to remove the stool produced during the previous 24 h. Stool in these extremely constipated patients should never remain in the rectosigmoid for more than 24 h.

The routine of increasing the amount of laxatives and giving an enema, if needed, is continued every night until the child has a voluntary bowel movement and empties the colon completely. The day that the patient has a bowel movement (which is usually with diarrhea), a radiograph should confirm that the bowel movement was effective, meaning that the patient has completely emptied the rectosigmoid. If the patient passed stool but did not empty completely, the dose of laxative must be increased.

Since this condition covers a wide spectrum, patients may have laxative requirements much larger than the manufacturer's recommendation. Occasionally, in the process of increasing the amount of laxatives, patients throw up before reaching any positive effect. In these patients, a different medication can be tried. Some patients vomit all kind of laxatives and and are unable to reach the amount of laxative that produces a bowel movement that empties the colon. Such a patient is considered intractable, and therefore a candidate for surgical intervention. Most of the time, however, the dosage that the patient needs in order to empty the colon completely, as demonstrated radiologically, can be achieved. At that dose, the patient should stop soiling because they are successfully emptying their colon each day, and because the colon is empty, they remain clean until the next voluntary bowel movement.

At this point, the patient and the parents have the opportunity to evaluate the quality of life that they have with that kind of treatment, understanding that this treatment will most likely be for life. There is an operation, a sigmoid resection, which provides symptomatic improvement, sometimes to the point that they do not need laxatives at all. Since this is a quality of life issue, it must be determined by the parents and the patient.

32.2.3 Surgical Treatment

32.2.3.1 Sigmoid Resection

For the last 14 years, we have been performing a sigmoid resection for the treatment of these conditions [7,8]. The very dilated megarectosigmoid is resected and the descending colon is anastomosed to the rectum. In a recent review of patients with anorectal malformations, 315 suffered from severe constipation and were fecally continent, but required significant laxative doses to empty their colon. Of these, 53 underwent a sigmoid resection. The degree of improvement varied. Following sigmoid resection, 10% of patients did not require any more laxatives, have bowel movements every day, and do not soil. Thirty percent of patients decreased their laxative requirement by 80%. The remaining 60% of patients decreased their laxative requirement by 40%. These patients must be followed closely because the condition is not cured by the operation. The remaining rectum is most likely abnormal, and without careful observation and treatment of constipation, the colon can redilate. It is vital however in its role as a reservoir, and to allow the patient to feel rectal distension.

It is vital however in its role as a reservoir, and to allow the patient to feel rectal distension.

The most dilated part of the colon is resected because it is most seriously affected. The nondilated part of the colon is assumed to have a more normal motility. Clearly, there must be a more scientific way to assess the dysmotile anatomy. Perhaps with emerging colonic motility techniques, these studies will help with surgical planning. It does seem that the patients who improve the most are those who have a more localized form of megarectosigmoid. Patients with more generalized dilatation of the colon do not respond as well. Perhaps in the future, these observations can be corroborated, and the results of resection better predicted by noninvasive modalities.

The administration of antegrade enemas through a continent appendicostomy or a button cecostomy is becoming popular [9]. Some clinicians use this approach, observe radiologically that the colon decreases in size over time, and then start laxatives. In our patients, we have only utilized antegrade enemas in incontinent patients who require a daily enema and seek more independence for their bowel management program [10]. An appendicostomy represents a useful alternative for patients who are treated with enemas only, since those antegrade enemas are only a different route of administration of enemas. It must be emphasized that the majority of patients with anorectal malformations and constipation can be treated with laxatives alone, provided it is in adequate doses, and on occasion benefit from a sigmoid resection. Therefore, most do not need washouts at all.

Distinguishing which patients require washouts because they cannot empty on their own from those who could empty if their constipation was adequately managed with laxatives is the key challenge for the clinician.

References

1. Peña A, Levitt MA (2002) Colonic inertia disorders in pediatrics. Curr Probl Surg 39:666–730
2. Peña A, Levitt MA (2005) Imperforate anus and cloacal malformations. In: Ashcraft KW, Holder TM, Holcomb W (eds) Pediatric Surgery, 4th edn. WB Saunders, Philadelphia, pp. 496–517
3. DeLorenzo C, Flores AF, Reddy SN, Hyman PE (1992) Use of colonic manometry to differentiate causes of intractable constipation in children. J Pediatr 120:690–695
4. Sarna SK, Bardakjian BL, Waterfall WE, Lind JF (1980) Human colonic electric control activity (ECA). Gastroenterology 78:1526–1536
5. Cook BJ, Lim E, Cook D, et al (2005) Radionuclear transit to assess sites of delay in large bowel transit in children with chronic idiopathic constipation. J Pediatr Surg 40:478–483
6. Peña A, Guardino K, Tovilla JM, Levitt MA, Rodriguez G, Torres R (1998) Bowel management for fecal incontinence in patients with anorectal malformations. J Pediatr Surg 33:133–137
7. Peña A, El-Behery M (1993) Megasigmoid – a source of pseudo-incontinence in children with repaired anorectal malformations. J Pediatr Surg 328:1–5
8. Levitt MA, Carney DE, Powers CJ, Tantoco JG, Caty MG (2003) Laparoscopically assisted colon resection for severe idiopathic constipation with megarectosigmoid. J Pediatr Endosurg Innov Tech 7:285–289
9. Marshall J, Hutson JM, Anticich N, Stanton MP (2001) Antegrade continence enemas in the treatment of slow-transit constipation. J Pediatr Surg 36:1227–1230
10. Levitt MA, Soffer SZ, Peña A (1997) The continent appendicostomy in the bowel management of fecally incontinent children. J Pediatr Surg 32:1630–1633
11. Peña A, Levitt MA (2002) Colonic inertia disorders in pediatrics. Current problems in surgery 39:681

33 Postoperative Treatment: Multidisciplinary Behavioral Treatment. The Nijmegen Experience

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33.1 Introduction

Most children born with anorectal malformations (ARM) suffer from defecation problems during childhood. For a majority of patients constipation disappears in adolescence and fecal incontinence also improves at that time [1]. Although problems tend to be worse in young children [2], even some adolescents and adults continue to have difficulties with defecation [3–5].

Chronic defecation problems have a negative impact on the child's somatic and psychosocial development. Chronic constipation leads to eating problems, chronic fatigue, inactivity, and urinary problems [6]. Incontinence leads to social and emotional problems like a negative self-image, lack of self-confidence, behavioral difficulties, and parent–child interaction problems [7–10].

Possible conservative treatments for fecal incontinence include dietary, medical, and behavioral treatment, all of which are often effective [11]. They are directed to prevent as much suffering as possible in young children and their parents. Long-term follow-up with adequate toilet training is required for all patients with an ARM [12].

We can discern the strict medical therapies, including the bowel management program, from the multidisciplinary behavioral treatment and the dietary contribution, both of which are too often neglected. An abnormality of the anorectal area does not mean it can never function properly and therefore automatically lead to a life with daily washouts. Proper functional rehabilitation with behavioral therapy can improve the incontinence or constipation.

As we try to avoid all unpleasant and often painful rectal interventions, we start with behavioral treatment if a child is still incontinent at 4 years of age. When a child has severe constipation and simple oral and consistently given medication therapy and dietary advice does not result in improvement, we start at an earlier age.

33.2 Multidisciplinary Behavioral Treatment

In the Nijmegen behavioral treatment protocol, developed at the Radboud University Nijmegen Medical Centre, defecation problems are considered to be the result of a dysfunctional interaction between the organ, which is impaired by the ARM, and behavioral factors [13]. To have a bowel movement, there has to be rectal filling. In healthy children this will lead to a sense of urge, which induces a defecation reflex and results in an empty bowel. For children to become continent, there has to be an interaction between the organ and the behavior of the child. The child has to perceive a sense of urge and to learn to react by withholding defecation for a short while, to go to the toilet or potty for voluntary evacuation. This will not only lead to an empty bowel, but also to empty diapers or trousers. In this learning process, several things can go wrong. For instance, a child who experiences pain with defecation will react to a sense of urge by withholding instead of relaxation. This will lead to incomplete defecation, which causes constipation. Children who have been constipated for a long time no longer feel any sense of urge, and in response will not do anything to have defecation, which results in persistence of the problems.

These processes do not only take place in healthy children, but also in children with ARM who are already more vulnerable because of their impairment.

Table 33.1 The Nijmegen Behavioral Treatment protocol. *ARM* Anorectal malformations

Knowing	• Child and parents are educated about defecation in general and in ARM, how children become continent normally, how defecation problems develop • They are educated about the working of and compliance to medication • Parents and child both learn what they can do to change the problems and the child learns skills of self-control
Daring	• Extinction of reactions of fear and aversion relating to urge sensations and defecation avoidance behavior like squeezing and withholding feces and avoiding the toilet. For example, by using desensitization techniques, by prescribing oral instead of anal medication, and by stopping invasive treatment
Wanting	• The child is motivated by taking small, reachable steps, being given direct rewards, and by having their effort rewarded at first and the result later on
Being able	• Sufficient emptying of the bowel by oral medication • Being able to defecate by learning an adequate straining technique
Doing	• Learning to try to defecate regularly and to empty their bowel 2–3 times a day after the meal by straining adequately 5–10 times each time • Learning to think about it themselves • Learning to react to an external sign first and, when possible, to an internal sign • Adjustment of exercises to the defecation rhythm • Compliance in taking medication
Continue doing	• Consolidation of behavior discipline in child and parents • Finishing medication if possible • Prevention of relapses

In the multidisciplinary treatment, a learning process is started in the child, with the parents as co-therapist, aimed at resolving the chronic defecation problems by teaching the child bowel self-control, primarily by training optimal defecation skills and subsequently toilet behavior. The therapeutic team consists of a pediatric surgeon, a child psychologist, and a pediatric physiotherapist, who work closely together. The pediatric surgeon determines whether the child's somatic condition is sufficient to reach some form of self-control over defecation. He/she facilitates the learning process by prescribing oral medication to allow sufficient emptying of the bowel. After disimpaction by enema or a rectal washout (if necessary), prescribing oral laxatives prevents new impaction. This treatment is continued as long as the child is withholding defecation or has not yet mastered enough self-control. For some children, medication will be necessary for a very long period of time.

The child psychologist educates the child an parents concerning defecation and manages the learning process, looking after the defecation and /or toilet behavior; he or she will establish whether the problems are caused by a lack of knowledge, defecation and/or toilet anxiety, or whether there is a motivational problem. Parental skills and cooperation in general, and in particular concerning the defecation problems, are investigated and attended to if necessary.

The pediatric physiotherapist assists in managing the learning process by teaching the child to use the lower part of the body in a pleasant way by playing motor games. If necessary, the child is taught to evacuate feces voluntarily even without a feeling of urge by teaching a correct straining technique. The child is instructed how to build up pressure in the right direction and to tense the abdominal wall muscles and relax the muscles of the pelvic floor selectively, and to combine both to defecate in the correct way [9].

The multidisciplinary treatment is carried out in accordance with a protocol. The different steps of the Nijmegen multidisciplinary behavioral treatment protocol are described in Table 33.1. This treatment protocol was studied in a prospective, controlled study with follow-up. Compared with a waiting-list control group, the experimental treatment group scored significantly better on two important measures (Templeton and percentages of feces in toilet; Table 33.2). Although young children had poorer scores than older children before treatment, no significant differences in the favorable outcome of treatment were found between both groups after treatment (Table 33.3). Nor was there any effect of type of ARM on treatment.

Table 33.2 Means and standard deviations (SD) of children in the experimental treatment and waiting-list control group on first and second assessment based on intention-to-treat analysis, and analysis of variance (ANOVA) on change of scores between both assessments, controlled for pretreatment scores. Reproduced with permission from van Kuyk et al. [14]

	Experimental treatment group *n* = 13		Waiting list control group *n* = 11		Significance ANOVA[a]
	First assessment	**Second assessment**	**First assessment**	**Second assessment**	
Templeton	2.2 (0.63)	1.6 (0.76)	2.0 (0.59)	2.0 (0.72)	0.03*
1 = good	-	7 (54%)	1 (9%)	2 (18%)	
2 = fair	9 (69%)	4 (31%)	8 (73%)	6 (55%)	
3 = poor	4 (31%)	2 (15%)	2 (18%)	3 (27%)	
Wingspread	2.7 (0.88)	2.3 (0.95)	2.9 (0.86)	2.6 (1.1)	0.53
1 = clean	1 (8%)	2 (15%)	-	3 (27%)	
2 = staining	3 (23%)	6 (47%)	3 (27%)	-	
3 = intermittent soiling	5 (38%)	3 (23%)	5 (46%)	6 (55%)	
4 = constant soiling	4 (31%)	2 (15%)	3 (27%)	2 (18%)	
Wingspread constipation	1.8 (0.69)	1.3 (0.60)	1.8 (0.72)	1.4 (0.49)	0.52
1 = no	4 (31%)	10 (77%)	3 (27%)	6 (55%)	
2 = sometimes	7 (54%)	2 (15%)	6 (55%)	5 (45%)	
3 = yes	2 (15%)	1 (8%)	2 (18%)	-	
Percentage of feces in toilet	41.5 (30.9)	57.9 (35.0)	43.5 (28.0)	38.5 (25.6)	0.049*
Number of days without soiling	3.7 (4.9)	5.8 (5.5)	3.7 (4.2)	2.8 (4.3)	0.09
Parental Judgment Incontinence Scale	17.5 (5.7)	13.2 (5.3) two missing	18.1 (6.1)	16.1 (7.3)	0.26

*$p \leq 0.05$

[a]ANOVA on change of scores (scores on the second assessment minus scores on the first assessment) between both groups, with scores at the first assessment as covariate

The results of multidisciplinary behavioral treatment remained stable over a mean follow-up period of 7 months (Table 33.4) [14].

This treatment is suitable for young children, thus preventing chronic problems in vulnerable children. Children can be treated individually as well as in groups. Group treatment seems to be very effective. Parents and children profit from each other. Groups are formed with six children of the same developmental age. The group meets six times for 2 hours each time, with a follow-up session after 6 months.

For children who do not profit from multidisciplinary behavioral treatment (for instance because of their anatomic limitations or because of psychiatric comorbidity or severe parent–child interaction problems), bowel management can be a good solution of their defecation problems. In our clinic, children start with behavioral treatment when possible, and if this is not effective enough in managing the problems, children begin bowel management.

With the new Krickenbeck classification [15], it has become easier to evaluate and compare the postoperative results after ARM repair, but it remains difficult to choose the best therapy for children with continence problems. By treating the children with multidisciplinary behavioral treatment, not only can further surgery for improving continence be avoided, but also the quality of life of these children can be improved, as they stop suffering from chronic incontinence and constipation. It is therefore a valuable adjunct to standard medical treatment of children with surgically repaired ARM.

Table 33.3 Means and SD of young and older children at pre- and posttreatment, and Student's *t*-test; also ANOVA on change of scores between young and older children, controlled for pretreatment scores. Reproduced with permission from van Kuyk et al. [14]

	Pretreatment			Posttreatment			
	Young children 2–4 years n = 10	Older children 5–12 years n = 14	Significance Student's t-test[a]	Young children 2–4 years n = 10	Older children 5–12 years n = 14	Significance Student's t-test[b]	Significance ANOVA[c]
Templeton	2.4 (0.52)	1.9 (0.57)	0.03*	1.7 (0.82)	1.4 (0.62)	0.46	0.78
1 = good	-	1 (7%)		5 (50%)	8 (57%)		
2 = f air	6 (60%)	11 (79%)		3 (30%)	5 (36%)		
3 = poor	4 (40%)	2 (14%)		2 (20%)	1 (7%)		
Wingspread	3.2 (0.71)	2.5 (0.89)	0.08	2.5 (0.98)	2.0 (0.87)	0.21	0.89
1 = clean	-	1 (7%)		1 (10%)	4 (29%)		
2 = staining	1 (10%)	5 (36%)		4 (40%)	6 (43%)		
3 = intermittent soiling	5 (50%)	5 (36%)		3 (30%)	3 (21%)		
4 = constant soiling	4 (40%)	3 (21%)		2 (20%)	1 (7%)		
Wingspread constipation	2.0 (0.83)	1.7 (0.58)	0.42	1.3 (0.63)	1.1 (0.31)	0.43	0.58
1 = no	3 (30%)	4 (29%)		7 (70%)	11 (79%)		
2 = sometimes	4 (40%)	9 (64%)		2 (20%)	3 (21%)		
3 = yes	3 (30%)	1 (7%)		1 (10%)	-		
Percentage of feces in toilet	27.8 (22.3)	52.9 (29.3)	0.03*	54.4 (30.4)	63.1 (33.2)	0.52	0.25
Number of days without soiling	1.1 (1.2)	5.6 (5.1)	0.01*	4.5 (5.0)	6.8 (5.8)	0.38	0.47
Parental Judgment Incontinence Scale	21.2 (4.2)	15.3 (5.5)	0.01*	16.2 (6.2)	11.8 (5.2) two missing	0.09	0.77

* $p \leq 0.05$

[a] Student's *t*-test on pretreatment scores of young and older children

[b] Student's *t*-test on posttreatment scores of young and older children

[c] ANOVA on change of scores (posttreatment scores minus pretreatment scores) between both groups, with pretreatment scores as covariate

Table 33.4 Means and SD of all completers at pretreatment and follow-up, and paired samples *t*-test. Reproduced with permission from van Kuyk et al. [14]

	First assessment (baseline) $n = 19$	Last assessment (follow-up) $n = 19$	Paired samples t-test[a]
Templeton	1.9 (0.55)	1.3 (0.48)	0.00*
1 = good	1 (5%)	13 (68%)	
2 = fair	15 (79%)	6 (32%)	
3 = poor	3 (16%)	-	
Wingspread	2.6 (0.81)	2.1 (0.71)	0.00*
1 = clean	1 (5%)	4 (21%)	
2 = staining	6 (32%)	10 (53%)	
3 = intermittent soiling	8 (42%)	5 (26%)	
4 = constant soiling	4 (21%)	-	
Wingspread constipation	1.9 (0.66)	1.3 (0.48)	0.00*
1 = no	5 (26%)	13 (68%)	
2 = sometimes	11 (58%)	6 (32%)	
3 = yes	3 (16%)	-	
Percentage of feces in toilet	48.8 (27.4)	77.9 (16.8)	0.00*
Number of days without soiling	4.5 (4.8)	8.2 (5.0)	0.01*
Parental Judgment Incontinence Scale	16.7 (5.6)	12.7 (5.2)	0.00*

* $P \leq 0.05$

[a] Paired samples *t*-test on scores of all children on baseline and on last assessment

References

1. Rintala RJ, Lindahl HG (2001) Fecal continence in patients having undergone posterior sagittal anorectoplasty procedure for a high anorectal malformation improves at adolescence, as constipation disappears. J Pediatr Surg 36:1218–1221
2. Langemeijer RA, Molenaar JC (1991) Continence after posterior sagittal anorectoplasty. J Pediatr Surg 26:587–590
3. Diseth TH, Emblem R (1996) Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. J Pediatr Surg 31:638–643
4. Rinatala R, Mildh L, Lindahl H (1992) Fecal continence and quality of life in adult patients with an operated low anorectal malformation. J Pediatr Surg 27:902–905
5. Rintala R, Mildh L, Lindahl H (1994) Fecal continence and quality of life for adult patients with an operated high or intermediate anorectal malformation. J Pediatr Surg 29:777–780
6. Benninga MA (1994) Constipation and Fecal Incontinence in Childhood. Doctoral Thesis, Universiteit van Amsterdam, Amsterdam
7. McGrath ML, Mellon MW, Murphy L (2000) Empirically supported treatments in pediatric psychology: constipation and encopresis. J Pediatr Psychol 25:225–254
8. Amendola S, De Angelis P, Dallóglio L, et al (2003) Combined approach to functional constipation in children. J Pediatr Surg 38:819–823
9. Benninga MA, Voskuijl WP, Akkerhuis GW, et al (2004) Colonic transit times and behaviour profiles in children with defecation disorders. Arch Dis Child 89:13–16
10. Kuyk EM van, Brugman-Boezeman AT, Wissink-Essink M, et al (2000) Defecation problems in children with Hirschsprung's disease: a biopsychosocial approach. Pediatr Surg Int 16:312–316
11. Hinninghofen H, Enck P (2003) Fecal incontinence: evaluation and treatment. Gastroenterol Clin North Am 32:685–706
12. Tsuji H, Okada A, Nakai H, Azuma T, Yagi M, Kubota A (2002) Follow-up studies of anorectal malformations after posterior sagittal anorectoplasty. J Pediatr Surg 37:1529–1533
13. Kuyk EM van, Brugman-Boezeman ATM, Wissink-Essink M, et al (2001) Defecation problems in children with Hirschsprung's disease: a prospective controlled study of a multidisciplinary behavioral treatment. Acta Paediatr 90:153–1159
14. Kuyk EM van, Wissink-Essink M, Brugman-Boezeman ATM, et al (2001) Multidisciplinary behavioral treatment of defecation problems: a controlled study in children with anorectal malformations J Pediatr Surg 36:1350–1356
15. Holschneider A, Hutson J, Peña A, et al (2005) Preliminary report on the International Conference for the Development of Standards for the Treatment of Anorectal Malformations. J Pediatr Surg 40:1521–1526

34 Continent Catheterizable Channels

Yunus Söylet

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34.1 Introduction

The concept of clean intermittent catheterization and its efficacy in emptying urinary reservoirs was first introduced by Lapides in 1972 and is accepted as the first step in the use of continent catheterizable channels (CCC). Subsequent important progress in CCC includes performance of the first continent cystostomy by Mitrofanoff in the 1980s, acknowledgment of low-pressure reservoirs from the beginning of the 1980s, recognition of the importance and efficacy of colonic washouts in the management of fecal continence, and the first continent cecostomy performed by Malone in 1990 [1–4]. CCC are used in anorectal malformations (ARM) for problems involving the gastrointestinal and/or urinary systems. The aim is to overcome fecal and urinary incontinence concomitantly. These circumstances also lead to competition for the appendix, since this has been the favored channel for both the Malone's antegrade colonic enema (MACE) and Mitrofanoff procedures. These limitations have lead to the creation of new techniques for these channels [5].

34.2 Indications

The Mitrofanoff and MACE procedures are widely used alone or in combination for different problems in both children and adults, such as ARM, spina bifida, sacral agenesis, exstrophy complex, postradiation cystitis, complex vesicovaginal fistulae, posterior urethral valves, prune-belly syndrome, cerebral palsy, sacrococcygeal teratoma, spinal-cord injury, perineal trauma, Hirschsprung's disease, intractable constipation, and different types of urethral failures, or in patients with perineal colostomy after rectal resection [6–19]. In the original description of Mitrofanoff and antegrade colonic enema (ACE) procedures, the conduit of choice is the appendix. In conditions in which the appendix is not available, tubular structures such as the ureter, fallopian tube, Meckel's diverticulum, and the vas deferens, or tubes created from the ileum, colon, stomach, bladder, rectus abdominis muscle, and prepuce can be used to create a Mitrofanoff channel. Ileal tubes are most commonly preferred if the appendix is not available. The use of the other organs except the ureters and bladder tubes are either sporadic or experimental (Table 34.1) [20–37].

In ARM, the ACE procedure is used with two indications: the treatment of fecal incontinence and/or the treatment of postoperative intractable constipation and developing megarectum/megacolon in which conservative management frequently fails [38]. The presence of sacrospinal anomalies in ARM can also lead to both urinary and fecal incontinence [39–42]. In this group of patients, the Mitrofanoff principle is used to provide urinary continence. In some

Table 34.1 Organs used in the construction of continent channels using the Mitrofanoff principle

Tubular organs	Tubularized organs
Appendix	Ileum
Ureter	Colon
Fallopian tubes	Stomach
	Bladder
	Prepuce
	Rectus abdominis muscle

patients with combined incontinence, the ACE and Mitrofanoff procedures are used together.

34.3 The Mitrofanoff Procedure

The Mitrofanoff procedure is also known as continent appendicovesicostomy according to the Mitrofanoff principle. The two indications for continent vesicostomy in ARM are the same as those for incontinent intestinal conduits: management of urinary incontinence and preservation of renal function. ARM patients usually have neurogenic bladders secondary to sacrospinal anomalies. In a small group of patients that have urethral injury secondary to their ARM reconstruction, the Mitrofanoff procedure may be helpful in the long-lasting management of urethral stricture. This is the third indication for the Mitrofanoff procedure, and has been reported in the management of urethral problems [43]. The Mitrofanoff procedure as the pioneer of CCC has helped to define the primary principles of these channels. Since the appendix is anastomosed to the bladder with an antireflux technique, there is no leak from the abdominal wall orifice, thus it is a continent vesicostomy. In this procedure, a catheter is advanced through a conduit to empty the urinary reservoir in a clean and intermittent fashion.

34.3.1 Technical Details

In the majority of cases a Mitrofanoff channel is constructed through a midline infraumbilical incision. If the patient will only have a continent vesicostomy constructed, the right laterovesical space is deeply freed before opening the peritoneum. The cecum and appendix are explored and the pedicle vessels of the appendix are carefully mobilized. The appendix is excised from the cecum with a cecal wall cuff. The aim in leaving the cecal cuff on the appendix is to enable a wide anastomosis on the abdominal wall. Once the cecal defect is closed, the tip of the appendix is opened obliquely, the lumen is irrigated with

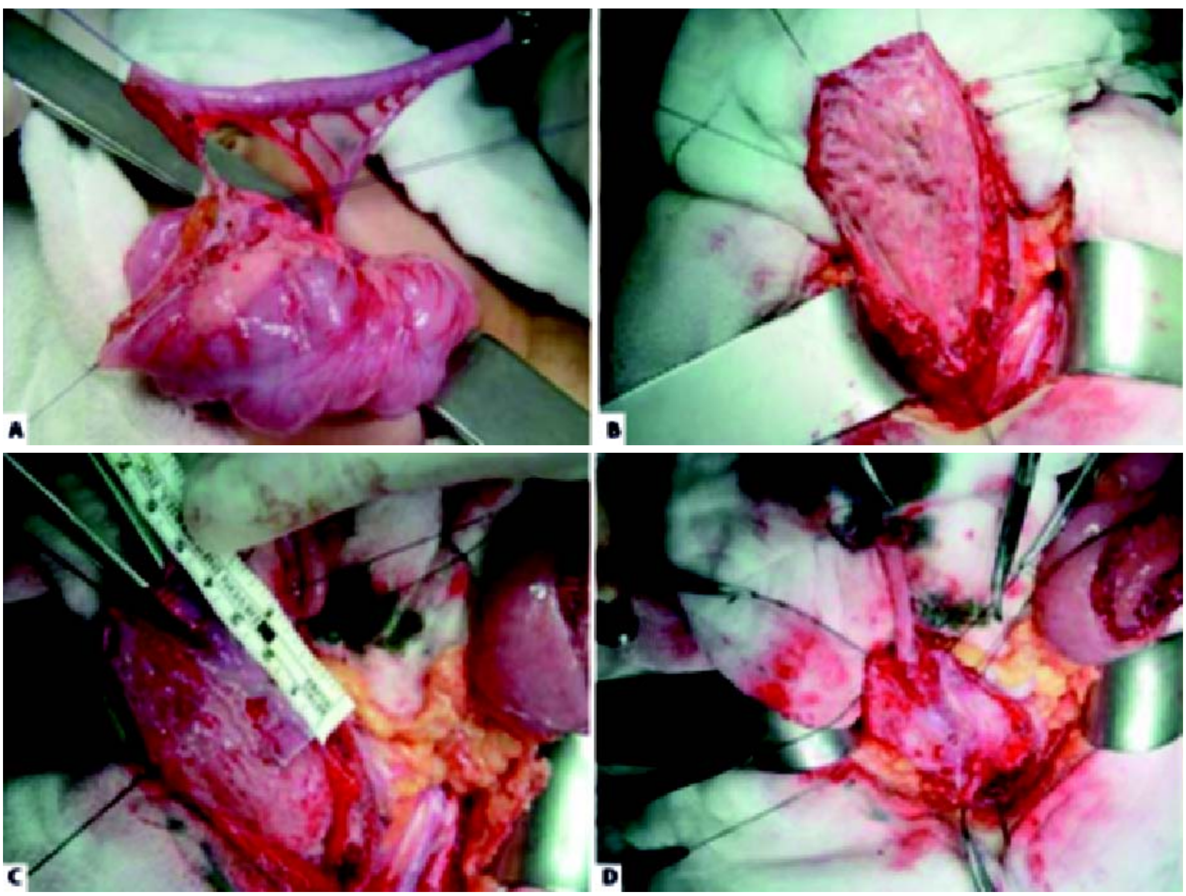

Fig. 34.1 (A–D) Submucous tunnel technique into the U-flap of thick-walled bladder for creating a Mitrofanoff channel

an antiseptic solution, and the patency of the lumen is checked with a catheter. If the bladder has a small capacity and thick wall (as it has most of the time), a U-shaped flap should be raised cranially for entrance. With the help of this flap, the appendix can be anastomosed to the bladder through a long tunnel (Fig. 34.1). The tunnel should at least be 2.5–3 cm long. Once the conduit–bladder anastomosis is completed, if no further procedures such as bladder augmentation, bladder-neck reconstruction, or ureteric reimplantation are necessary, the bladder is closed and the conduit is anastomosed either to the right lower quadrant or to the umbilicus. In the literature, two cases have been reported to have their appendix located into the left lower quadrant due to previous stomas created in the right lower quadrant. In these cases the cecum and the ascending colon were widely mobilized, the appendiceal vessels were dissected up to their origin from the superior mesenteric artery, and the conduit was moved to the left through a defect created in the bowel mesentery and anastomosed to the left abdominal wall [44].

In the majority of patients, augmentation cystoplasty is a part of this surgery. Augmentation provides a low-pressure reservoir, while the Mitrofanoff channel aids in emptying the reservoir regularly. The bladder neck should be constructed or closed so that it does not leak. It has been shown in many cases that even though vesicoureteric reflux may exist in the system, the creation of a low-pressure reservoir and guaranteeing regular emptying of the system will lead to disappearance of the reflux. Thus, in many patients with small bladders and limited space for a ureteric reimplantation, the reimplantation should not be carried out and morbidity is avoided [45].

In patients with urethral injuries as a result of ARM repair, if a long and complicated management is necessary, or if autoaugmentation is indicated for a different underlying pathology, it is appropriate to construct an appendicovesicostomy without opening the bladder. In the extravesical Mitrofanoff technique that we use in our patients, the abdomen is entered through a lower-abdominal midline incision, the right laterovesical space is opened, and a Lich-Gre-

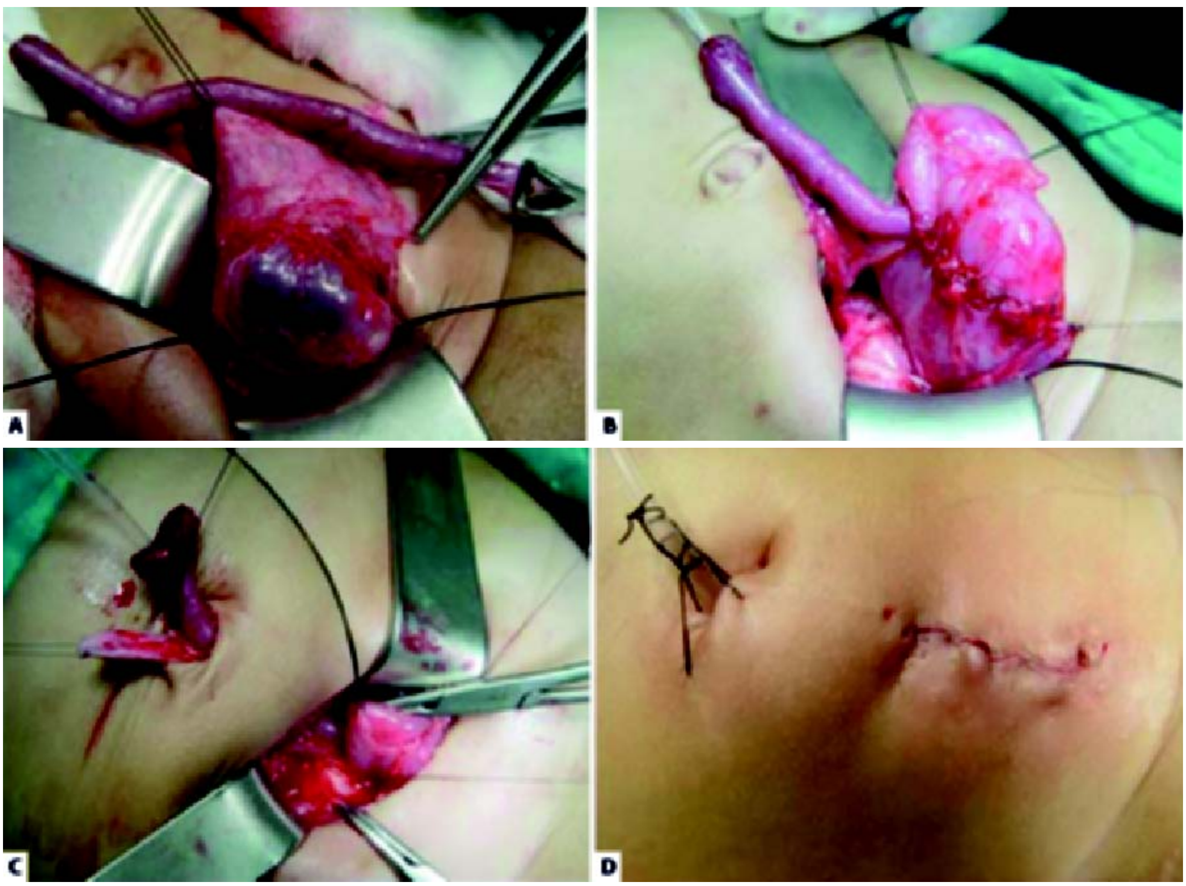

Fig. 34.2 (A–D) Extravesical Mitrofanoff technique

goire-like extramucosal tunnel is created in the right posterolateral wall of the bladder. The mucosa is entered close to the bladder neck and an appendicovesicostomy performed. The seromuscular layer is closed with sutures, the tunnel is completed, and the appendix is anastomosed to the skin (Fig. 34.2). In cases where the appendix is not available or has been used for the MACE procedure, the Mitrofanoff channel is most commonly created using the Monti technique [46]. Although this technique was described by Monti in 1997 [47], it was also described by Yang in 1993 [48]. When Yang reported this technique, however, he focused on the use of a needle as an aid for reimplants into the small bowel submucosa. In the Monti technique a 2.5-cm segment of the small bowel is detubularized longitudinally and retubularized transversally. A 2.5-cm segment of bowel provides a tube size of about 18–20 Fr. The length of the Monti tube is determined by the diameter of the bowel segment. The length will be at least 8 cm when the ileum is used and 10–12 cm when the colon is used. The transverse tubularization should be carried out first with mucosal apposition and then with a second row of serosal sutures [48]. Problems such as kinking, diverticula formation, and catheterization problems observed after longitudinal tubularization of ileal segments are rarely reported in Monti tubes [49]. Stomach and colonic tubes can be used for the same purpose [50]. The common pedicled bowel segment can be used in patients in whom bladder augmentation needs to be combined with an ileal Monti.

Two Monti tubes can be anastomosed to each other or elongated in a spiral fashion, similar to the construction of longer channels, creating a double Monti [51]. In addition, in both the Mitrofanoff and MACE procedures, longer tubes may be created using combinations of the appendix and Monti tube or appendix and cecal tube [52,53]. Rink created a continent catheterizable vesicostomy by modifying Casale's vesicostomy with an antireflux technique. Stomal continence was achieved in all patients with this technique, but 45% have required revision due to stomal stenosis. Thus, the use of the technique has been limited to large bladders requiring continent vesicostomies [54]. In a patient with microcolon-intestinal hypoperistalsis syndrome, a bladder tube created from the posterior wall of a huge, adynamic bladder has been used with success and no morbidity [55]. A salvage continent vesicostomy has been created in five children with enterocystoplasties and no appendix, using a bladder tube created completely extraperitoneally, and plicated with the bladder at its base in a Nissen-like fashion to add an antireflux property [56].

Minimally invasive techniques, although not widely popularized, have been used in the construction of continent urinary channel surgery. The first laparoscopic appendiceal–vesical anastomosis with a flap-valve mechanism has been reported by Hsu and Shortliffe [57]. There is also a case report on laparoscopic removal of a nonfuctioning kidney and anastomosis of the distal ureter to the skin as a continent channel in a patient who had previously undergone a ureteric reimplantation due to vesicoureteric reflux [58].

34.4 The MACE Procedure

Bowel management is necessary in the majority of ARM in the postoperative period. Fecal incontinence in some patients and intractable constipation in others are the main problems. Initially, conservative measures including aggressive potty training, dietary management, different medications, daily retrograde enemas, and biofeedback therapy in suitable patients are used to overcome these complications. Peña has reported a detailed bowel management program for ARM patients [59]. Reoperations for some patients can involve reconstructive surgical procedures such as gracilisplasty or, rarely, permanent stoma. Conservative measures should be initially undertaken in all patients. Patients that have success with daily retrograde enemas are good candidates for the MACE procedure. The most common cause of failure in the long term in patients who have success with daily enemas is the dislike they develop for enemas administered through the anus as they grow older. Many patients, especially teenagers, find this method tedious and bothersome. In this group of patients the antegrade colonic enema technique described by Malone in 1990 should be considered. The advantages are easy access to the colon compared to retrograde irrigations, on-way effective irrigation, smaller volumes for irrigation, psychological comfort, and combined management of urinary incontinence. Suitable anatomy, which means sufficient length of colon and absence of distal stenosis, motivation of patients and caregivers, trial of all conservative measures beforehand, technical success with rectal irrigations, and location by hand of the stoma site by patients, manipulation are factors influencing the success with the MACE procedure. Rigorous teaching and motivation before the procedure is mandatory [60–63]. Antegrade enema through a continent cecostomy is not a cure for intestinal problems with ARM, rather it is a more pleasant way for children to engage in a

Table 34.2 Continent cecostomy techniques. *MACE* Malone's antegrade colonic enema, *LACE* Left colonic antegrade colonic enema

Disconnection and reimplantation of the appendix
Orthotopic appendicostomy (+/- divided appendix)
Tubularized cecal/colonic flap
Transverse tubularized ileal tube (Monti)
Laparoscopic MACE (Appendicostomy only)
Cecostomy button
Percutaneous cecostomy catheter
LACE

bowel management protocol without the need for rectal enemas [8].

34.4.1 Technical Details

In the classical MACE technique, following effective bowel preparation and under the protection of broad-spectrum antibiotics, the appendix pedicle is mobilized, the blind tip is opened, the appendix is detached from the cecum, inverted, and is anastomosed to the cecal mucosa after being passed through a submucosal tunnel formed on the taenia. The seromuscular layer is closed on top to form an antireflux mechanism. The procedure is completed following cutaneous anastomosis. It is essential to suture the cecum or colon to the back of the anterior abdominal wall and not to leave a free intraperitoneal part of the conduit [4]. Multiple modifications of this original technique have been reported many times (Table 34.2). If the appendix is not available or has been previously used as a Mitrofanoff channel, ileal, colonic, or gastric Monti channels can be used to create an ACE. The cecum, transverse colon, left colon, and stomach tubes have been used in ACE construction. The left colon antegrade continence enema (LACE) procedure has been reported to have advantages by providing gravity-assisted evacuation, avoidance of the right and transverse colon, which has a large-volume capacity, a convenient stoma location in the left upper quadrant, shortening of the enema duration, a smaller fluid volume, and no ACE related abdominal pain. In the left colon, laterally or medially based tubes are used with either in situ flap-valve mechanisms or placement of a Monti tube in a submucosal tunnel along the colon taenia to form an antireflux mechanism [64–69]. The indications to perform a right or left colonic Monti-MACE procedure are: previous appendectomy, previous appendicovesicostomy, inadequate mesentery associated with the appendix, previous ileocecal augmentation, and obliterated appendicocecostomy [70]. Other modifications are based on similar indications.

In the MACE procedure, different antireflux techniques are used. In cecal imbrication (cecal wrap technique) the appendix is not detached from its base. An antireflux mechanism is developed in a similar fashion to Nissen's fundoplication around the in situ appendix (Fig. 34.3) [71]. In cases in which the ileocecal pouch reservoir has been created as a continent urinary diversion, a technique involving the reinforcement of the imbrication of the in situ appendix with a mesh has been reported [72]. In the extramucosal seromuscular taenial tunnel technique, analogous to the Lich-Gregoire technique, the seromuscular layer of the taenia is incised without opening the mucosa. On the distal part of the incision, the mucosa

Fig. 34.3 A, B Cecal imbrication around the in situ appendix

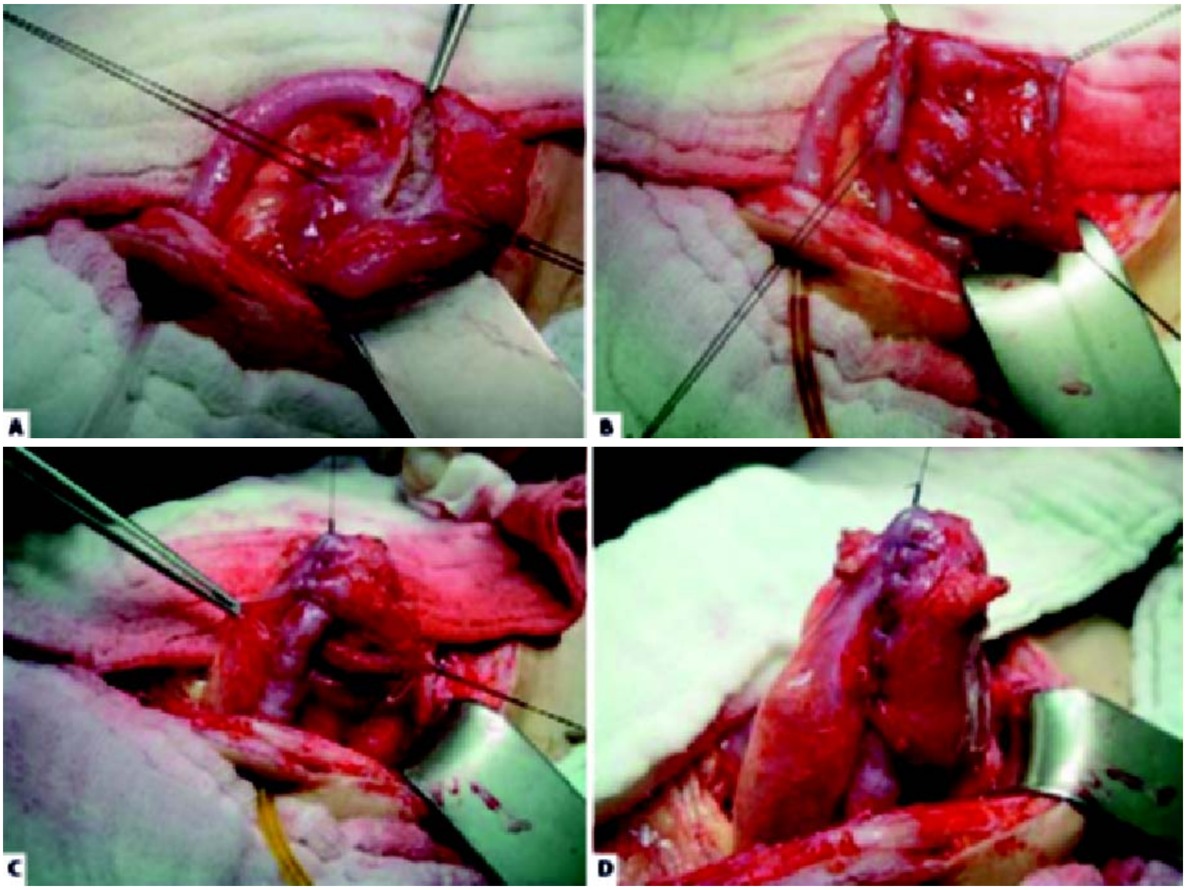

Fig. 34.4 (A–D) Malone's antegrade colonic enema (MACE) – extramucosal seromuscular taenial tunnel technique

is opened and the conduit is anastomosed to the cecum. The antireflux mechanism is created by covering the conduit with the seromuscular layer (Fig. 34.4). The application of the Monti channel to the ileocecal pouch involves similar technical aspects [73].

The intraluminal reimplantation technique involves the creation of a submucosal tunnel, similar to Cohen's reimplantation technique, and placement of the conduit into this tunnel. This technique is preferred in cases where bladder substitutes are constructed and the conduit needs to be anastomosed to the bowel (Fig. 34.5). The construction of a direct appendicostomy without supportive antireflux techniques have been reported mostly by authors using laparoscopic techniques, who state that it reduces the risk of conduit incontinence. On the contrary, Malone has reported stomal incontinence for gas and feces in all six cases in whom he performed a direct appendicostomy [62]. Lynch et al. report a 6.7% leakage rate in 28 patients with laparoscopically performed continent cecostomy and state that this is not a higher rate compared to those who have an additional antireflux procedure [74]. Similarly, van Savage et al. have reported no stomal leakage in 16 laparoscopically performed procedures. Their statement is that the continence mechanism is simply a function of the length of the appendix and the mucosal coaptation of the appendiceal lumen [75]. In the majority of cases that develop leakage, a surgical revision is necessary. In our series, a mild leakage was managed by submucosal injection, and in Mitrofanoff's series an artificial urinary sphincter application was beneficial in one case [76].

34.5 The Stoma Technique and Associated Problems

The most common complication in CCC construction is the stomal orifice at the anterior abdominal wall. The location of the stoma is related to the surgeon's choice as well as the type of conduit used. In our series, the fallopian tube and bladder stump were used as channels in two cases and were located as perineal stomas; this has provided technical applicability [77]. Duckett has also reported the use of perineal stomas

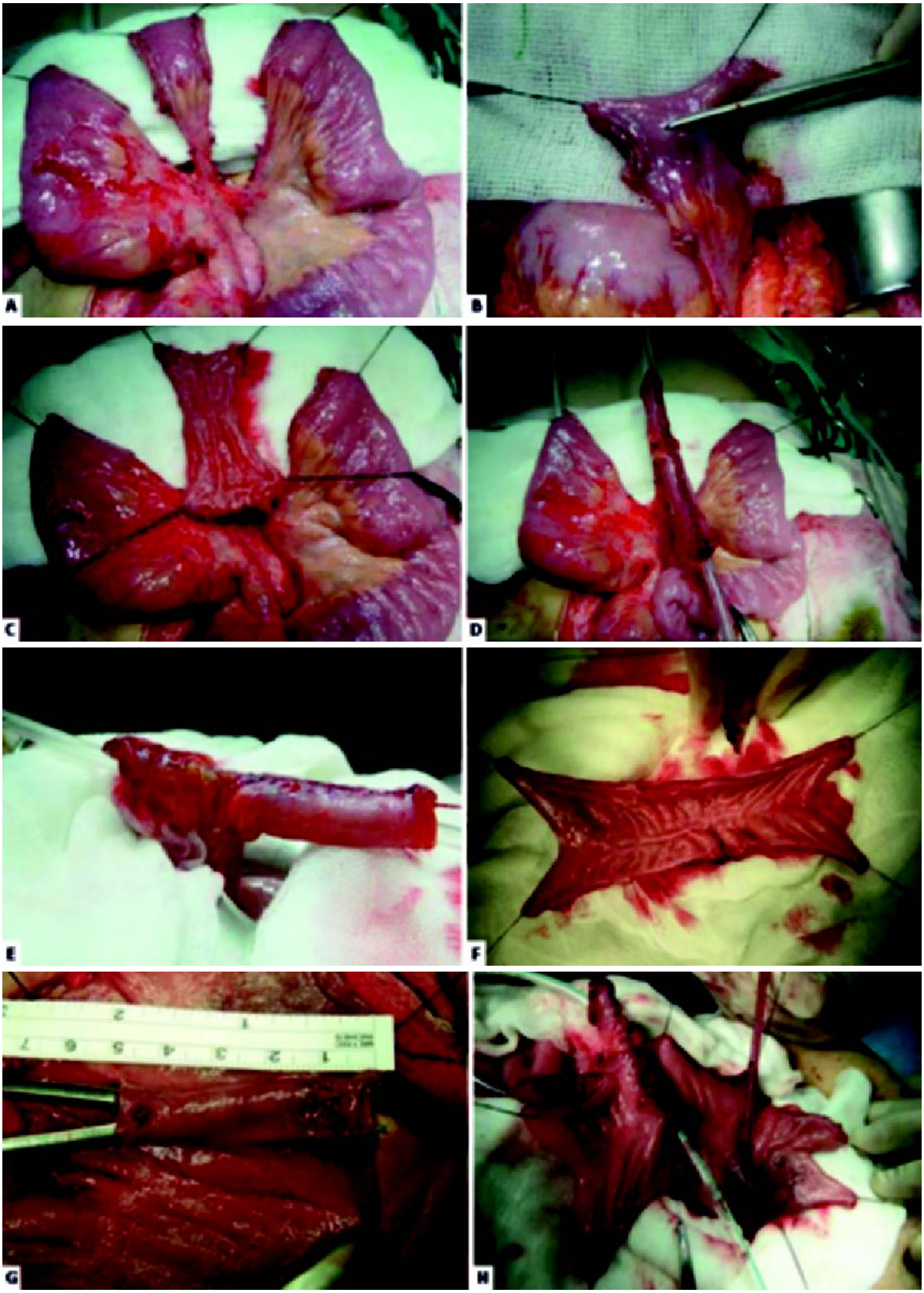

Fig. 34.5 (A–H) Reimplantation of Monti-MACE to the large bowel

in three cases that had ureter and ileal conduit construction [78].

Another group reports no difference in complications between umbilical and right lower quadrant abdominal stomas and prefers umbilical stomas due to better cosmesis, limiting lower quadrant stomas to ureteric conduits [79]. The right and left lower quadrants and umbilicus are preferred stomal sites. There is no significant difference in complications between umbilical and extraumbilical sites. On the other hand the umbilical stomas have superior cosmetic results [80–82].

Skin anastomosis techniques can vary. Multiple flap techniques aiming to avoid stenosis are frequently used. Mitrofanoff suggests maintaining an exteriorized small cuff of cecal mucosa to overcome the stomal stenosis complication reported as the most common problem (5–45%) [76]. V and Y type skin plasties [83,84], VQ, WQQ, and VQZ techniques and similar modifications all aim to create a more cosmetic and functional stoma [85].

Among the two most common CCC, the Yang-Monti channels and appendiceal channels are most frequently compared. Narayanaswamy has reported 60% catheterization difficulty and 28% pouch-like dilatation in Monti tubes in his series [86]. Studies involving similar comparisons among two conduits report no difference in the results. Obesity increases complication rates, as in any surgical procedure [87–90]. Bladder tube complication rates are around 30–40% and are higher in comparison to appendiceal and Monti channels [63,91].

Laterally based colonic conduits appear to have a higher rate of stomal stenosis compared to medially based colonic tubes (40% versus 12%), which may be attributed to the local blood supply properties of the colon [92].

MACE channels are more prone to stenosis compared to Mitrofanoff channels. The most possible cause may be less frequent catheterization. To overcome this problem, simultaneous catheterization of the Mitrofanoff and continent cecostomy channel is recommended [77]. Prevention of stomal stenosis can be overcome by skin plasties as well as prevention of catheter trauma. Snodgrass suggests parastomal Triamcinolone injection to prevent stenosis in Mitrofanoff stomas [93].

Appendicitis can lead to channel obliteration and loss of appendiceal channels [94,95]. Perforations have been reported in gastric and bladder tubes [96,97]. Mucosal prolapse and granulation of the tissue around the skin opening leading to a continuous bloody discharge and staining of underwear are not rare in our experience. In most cases, granulations can be managed with silver nitrate and prolapse with surgical revision.

Cecostomy tubes may be placed by either a percutaneous or an open approach, providing a comparable outcome to that of the original Malone procedure. This technique may be preferred not only in cases with a missing appendix, but also in those for whom the appendix needs to be reserved for urinary reconstruction, or to avoid a laparotomy in cases who do not require simultaneous reconstruction.

The disadvantage is the presence of a permanent catheter or button on the anterior abdominal wall. Shandling published his initial percutaneous cecostomy tube experience [98], only to notice later the difficulty of carrying a tube attached to the body and balloon tube complications. Thus, he suggested the insertion of a plastic trapdoor cecostomy tube 6–8 weeks after the procedure, which was better tolerated by patients. He reported no complications in his first eight-case series using plastic trapdoor cecostomy tubes [99]. Chait et al., in a series of 163 percutaneous cecostomy tubes, reported minor complications in 60% of cases and mentioned that they were easily managed with conservative measures. The failure rate was 10%, and 90% of patients reported an improved quality of life [100].

A different technique involving colonoscopy assistance for the insertion of a percutaneous cecostomy tube using the principles of percutaneous endoscopic gastrostomy (PEG) has been reported [101]. In the five-case cecostomy button series of Duel and Gonzales, three cases had localized infection and one patient required intravenous antibiotic administration due to cellulitis [102].

34.6 Postoperative Management

Catheters are left in situ for 2–3 weeks in the postoperative period of urinary and intestinal continent channels. Catheterization is initiated after this period. If the patient has a simultaneous augmentation cystoplasty, the reservoirs are continuously drained at nighttime for 2–3 months. Twenty-four-hour urinary output, and day and night urine volumes help to identify the frequency of catheterization.

Colonic enemas can start at between 1 and 30 days postoperatively. Colonic enemas may involve tap water, isotonic saline, salty water, phosphate, polyethylene glycol electrolyte, glycerine, mannitol, lactulose, or mineral oil, either alone or in combination. The volume of enemas will depend on the composition of the material used and other personal factors, and will vary between 30 and 1000 ml. The evacuation period may last between 10 and 120 min. Enema frequency may also vary from once a day to once a week. Dietary management, including medications that slow intestinal motility, is necessary as adjuvant therapy in almost all patients. If fecal incontinence continues despite conservative measures, the volume of enema should be increased, and the frequency decreased. Anal stenosis should be considered in cases with frequent but small-volume incontinence. Colicky abdominal pain is initially noted in 50% of cases, but this resolves spontaneously within a couple of months. Antispasmodic drugs have been found to be effective in these cases [103]. Some patients have reported spontaneous initiation of colonic motility simply with catheter insertion and no washouts [84]. Concentrated enemas leading to colonic evacuation through bowel irritation may lead to hyperphosphatemia, while high-volume nonirritable tap water and salt water enemas may cause fatal metabolic and electrolyte imbalances [62,104,105].

34.7 Results

There are two important scoring systems for MACE procedures. The first belongs to the Southampton group and the criteria are as follows: (1) complete success, where the patient is clean or has minor leakage with only irrigations, and no further measures are required; (2) partial success, where there is occasional leakage from the stoma or anus, and further measures are needed, but both child and family are satisfied; (3) unsuccessful, where there is continuous serious leakage. According to these criteria, a success rate of 62–98% is reported in the literature. A success rate of 89% in ARM drops to a level of 62% in intractable constipation. A second group of unsuccessful results have been reported in children younger than 5 years of age. These facts stress the importance of patient education and motivation [103].

Another scoring system has been suggested by the Melbourne group in patients with slow-transit constipation, wherein the scoring criteria are: the soiling score, pain frequency, pain severity, appetite score, and mood score prior to and after the MACE procedure. Evaluation also involves diagnostic tests and clinical findings [106].

In a study evaluating their 20-year, long-term experience, Mitrofanoff's team does seem to have found an answer for many important questions. They have established that all continent cystostomies remain functional after 20 years and have stated that the technique has long-lasting efficiency. They have found no catheterization problems during pregnancy and have reported successful delivery by cesarean section 16 years after the Mitrofanoff procedure. Patients requiring transplantation have not encountered any catheterization or reservoir problems during or after surgery. Late stomal stenosis has been attributed to the cessation of urine output in patients with renal insufficiency [76]. The first normal, complication-free vaginal delivery following MACE procedure was reported by Wren et al. [107]. An 8- to 10-year follow-up study of nine cases has reported intact channels in all and has estimated approximately 20,000 catheterizations without any problems. All patients except one (who was experiencing adolescent problems) reported that they were happy with their quality of life [108]. In another study reporting a 15-year experience of 50 patients, 96% continue catheterization, 10% have developed stomal stenosis, and 16% have required surgical revision due to stomal leakage; continence was achieved in 98% [109]. Simultaneous combined Mitrofanoff and ACE procedures are reported to provide 80% dual continence [110,111].

The main role of these long-term patent channels is to improve the quality of life for patients. Although some patients find CCC difficult, they report an increase in self-esteem, happiness, and social acceptance. Great support should be given to increase patient motivation, and patient groups should be organized to meet together at least once a year to prevent those patients who find the procedure lengthy and painful from abandoning their channels, despite the advantages [77,112–114].

References

1. Lapides J, Diokno AC, Silber SJ, Lowe BS (1972) Clean intermittent self catheterisation in the treatment of urinary tract disease. J Urol 107:458–461
2. Mitrofanoff P (1980) Cystostomie continente trans-appendiculaire dans le traitement des vessies neurologiques. Chir Pediatr 21:297–305
3. Shandling B, Gilmour RF (1987) The enema continence catheter in spina bifida: successful bowel management. J Pediatr Surg 22:271–273
4. Malone PS, Ransley PG, Kiely EM (1990) Preliminary report: the antegrade continence enema. Lancet 336:1217–1218
5. Cain MP (1999) Urologic reconstruction for total confidence – the MACE procedure and newer options for creating a Mitrofanoff channel. Dialogues Pediatr Urol 22:1

6. Sumfest JM, Burns MW, Mitchell ME (1993) The Mitrofanoff principle in urinary tract reconstruction. J Urol 150:1875–1878
7. Sheldon CA, Minevich E, Wacksman J, Lewis AG (1997) Role of the antegrade continence enema in the management of the most debilitating childhood rectourogenital anomalies. J Urol 158:1277–1279
8. McAndrew HF, Malone PSJ (2002) Continent catheterizable conduits: which stoma, which conduit and which reservoir? BJU Int 89:86–89
9. Lewitt MA, Soffer SZ, Peña A (1997) Continent appendicostomy in the bowel management of fecally incontinent children. J Pediatr Surg 32:1630–1633
10. Woodhouse CRJ, Gordon EM (1994) The Mitrofanoff principle for urethral failure. Br J Urol 73:55–60
11. Hodges AM (1999) The Mitrofanoff urinary diversion for complex vesicovaginal fistulae: experience from Uganda. BJU Int 84:436–439
12. Goepel M, Sperling H, Stöhrer M, Otto T, Rübben H (1997) Management of neurogenic fecal incontinence in myelodisplastic children by a modified continent appendiceal stoma and antegrade colonic enema. Urology 49:758–761
13. Hensle TW, Reiley EA, Chang DT (1998) The Malone antegrade continence enema procedure in the management of patients with spina bifida. J Am Coll Surg 186:669–674
14. Hill J, Stott S, MacLennan I (1994) Antegrade enemas for the treatment of severe idiopathic constipation. Br J Surg 81:1490–1491
15. Hakenberg OW, Ebermayer J, Manseck, Wirth MP (2001) Application of the Mitrofanoff principle for self-catheterization in quadriplegic patients. Urology 58:38–42
16. Yang CC, Stiens SA (2000) Antegrade continence enema for the treatment of neurogenic constipation and fecal incontinence after spinal cord injury. Arch Phys Med Rehabil 81:683–685
17. Krogh K, Laurberg S (1998) Malone antegrade continence enema for faecal incontinence and constipation in adults. Br J Surg 85:974–977
18. Sylora JA, Gonzales R, Vaughn, Reinberg Y (1997) Intermittent self-catheterisation by quadriplegic patients via a catheterizable Mitrofanoff channel. J Urol 157:48–50
19. Portier G, Bonhomme N, Platonoff I, Lazorthes F (2005) Use of Malone antegrade continence enema in patients with perineal colostomy after rectal resection. Dis Colon Rectum 48:499–503
20. Chitnis M, Chowdhary S, Lazarus C (2001) Application of the Malone antegrade continence enema principle in degenerative leiomyopathy. Pediatr Surg Int 17:470–471
21. Woodhouse CRJ, MacNeily AE (1994) The Mitrofanoff principle: expanding upon a versatile technique. Br J Urol 74:447–453
22. Figueroa TE, Sabogal L, Helal M, Lockhard JL (1994) The tapered and reimplanted small bowel as a variation of the Mitrofanoff procedure: preliminary results. J Urol 152:73–75
23. Marsh PJ, Kiff ES (1996) Ileocaecostomy: an alternative surgical procedure for antegrade colonic enema. Br J Urol 83:507–508
24. Abol-Enein H, Ghoneim MA (1996) A technique for the creation of a continent cutaneous urinary outlet: the serous lined extramural ileal valve. Br J Urol 78:791–792
25. Monti PR, Lara RC, Dutra MA, Rezende de Carvalho J (1997) New techniques for reconstruction of efferent conduit based on the Mitrofanoff principle. Urology 49:112–115
26. Duckett JW, Snyder HM III (1986) Continent urinary diversion: varions on the Mitrofanoff principle. J Urol 136:58–62
27. Boemers TM (2001) Mitrofanoff procedure with Meckel's diverticulum. Case report. BJU Int 88:799–800
28. Cain MP, Rink RC, Yerkes EB, Kaefer M, Casale A (2002) Long-term followup and outcome of continent catheterizable vesicostomy using the Rink modification. J Urol 168:2583–2585
29. Klauber GT, Cendron M (1994) Continent vesicostomy using a catheterizable posterior bladder tube: modification of the Mitrofanoff principle. J Pediatr Surg 29:71–73
30. Krstic ZD (1995) Preputial continent vesicostomy: preliminary report of a new technique. J Urol 154:1160–1161
31. Perovic S (1996) Continent urinary diversion using preputial penile or clitoral skin flap. J Urol 155:1402–406
32. Close CE, Mitchell ME (1997) Continent gastric tube: new techniques and long term follow up. J Urol 157:51–55
33. Mor Y, Kajbazadeh AM, German K, et al (1997) The role of ureter in the creation of Mitrofanoff channels in children. J Urol 157:635–637
34. Ashcraft KW, Dennis PA (1986) The reimplanted ureter as a catheterizing stoma. J Pediatr Surg 21:1042–1045
35. Ozgok Y, Kibar Y, Kilciler M, Ide T, Harmankaya C (2002) Reimplanted ureter as an alternative to the catheterizable Mitrofanoff tube. Eur Surg Res 34:266–270
36. Klauber GT, Cendron M (1994) Continent vesicostomy using a catheterizable posterior bladder tube: modification of the Mitrofanoff principle. J Pediatr Surg 29:71–
37. Celayir S, Dervisoglu S, Buyukunal SCN (1998) A modified Mitrofanoff procedure using the rectus abdominis muscle flap technique. A preliminary report in a rabbit model. Br J Urol 81:83–86
38. Malone PSJ, Curry JL (1999) The MACE procedure. Dialogue Pediatr Urol 22:2–5
39. Boemers TM, Van Gool JD, De Jong TPVM, et al (1994) Urodynamic evaluation of children with the caudal regression syndrome (caudal dysplasia sequence). J Urol 151:1038–1040

40. Holschneider AM, Kraeft H, Scholtissek CH (1990) Urodynamic investigations of bladder disturbances in imperforate anus and Hirschsprung's disease. Z Kinderchir 35:64–68
41. Emir H, Söylet Y (1998) Neurovesical dysfunction in patients with anorectal malformation. Eur J Pediatr Surg 8:95–97
42. Kılıç N, Emir H, Sander S, Elicevik M, Celayir S, Söylet Y (1997) Comparison of urodynamic investigations before and after posterior sagittal anorectoplasty for anorectal malformation. J Pediatr Surg 32:1724–1777
43. Freitas Filho FLG, Carnevale J, Melo Filho AR, Vicente NC, Heinisch AC, Martins JL (2003) Posterior urethral injuries and the Mitrofanoff principle in children. BJU Int 91:402–405
44. Wilcox DT, Gravamian R, Duffy PG (1996) Left sided appendiceal Mitrofanoff channel. Br J Urol 78:133–134
45. Söylet Y, Emir H, Ilce Z, Yesildag E, Buyukunal SN, Danişmend N (2004) Quo vadis? Ureteric reimplantation or ignoring reflux during augmentation cystoplasty. BJU Int, 94:379–380
46. Castellan MA, Gosalbez R, Labbie A, Monti PR (1999) Clinical applications of the Monti procedure as a continent catheterizable stoma. Urology 54:152–156
47. Monti PR, Lara RC, Dutra MA, de Carvalho RJ (1997) New techniques for reconstruction of efferent conduit based on the Mitrofanoff principle. Urology 49: 112–115
48. Yang WH (1993) Yang needle tunneling technique in creating antireflux and continent mechanisms. J Urol 150:830–834
49. Gosalbez R, Wei D, Gousse A, Castellan M, Labbie A (1998) Refashioned bowel segments for the construction of catheterizable channels (the Monti procedure): early clinical experience. J Urol 160:1099–1102
50. Woodhouse CRJ, Malone PR, Cumming J, Reilly TM (1989) The Mitrofanoff principle for continent urinary diversion. Br J Urol 63:53–57
51. Casale AJ (1999) A long continent ileovesicostomy using a single piece of bowel. J Urol 162:1743–1745
52. Cromie WJ, Barada JH, Weingarten JL (1991) Cecal tubularization: lengthening technique for creation of catheterizable conduit. Urology 37:41–42
53. Bruce RG, McRoberts JW (1998) Cecoappendicovesicostomy: conduit-lengthening technique for use in continent urinary reconstruction. Urology 52:702–704
54. Cain MP, Rink RC, Yerkes EB, Kaefer M, Casale AJ (2002) Long term followup and outcome of continent catheterizable vesicostomy using the Rink modification. J Urol 168:2583–2585
55. Klauber GT, Cendron M (1994) Continent vesicostomy using a catheterizable posterior bladder tube: modification of the Mitrofanoff principle. J Pediatr Surg 29:71–73
56. Hanna MK, Richter F, Stock JA (1999) Salvage continent vesicostomy after enterocystoplasty in the absence of the appendix. J Urol 162:826–828
57. Hsu THS, Shortliffe LD (2004) Laparoscopic Mitrofanoff appendicovesicostomy. Urology 64:802–804
58. Strand WR, McDougall EM, Leach FS, Allen TD, Pearle MS (1997) Laparoscopic creation of a catheterizable cutaneous ureterovesicostomy. Urology 49:272–275
59. Peña A (1992) Current management of anorectal malformations. Surg Clin North Am 72:1393–1416
60. Wilcox DT, Kiely EM (1998) The Malone (antegrade continence enema) procedure: early experience. J Pediatr Surg 33:204–206
61. Driver CP, Barrow C, Fishwick J, Gough DCS, Bianchi A, Dickson AP (1998) The Malone antegrade colonic enema procedure: outcome and lessons of 6 years experience. Pediatr Surg Int 13:370–372
62. Malone PSJ, Curry JI, Osborne A (1998) The antegrade continence enema procedure: why, when, and how? World J Urol 16:274–278
63. Clark T, Pope JC IV, Adams MC, Wells N, Brock III JW (2002) Factors that influence outcomes of the Mitrofanoff and Malone antegrade continence enema reconstructive procedures in children. J Urol 168:1537–1540
64. Perez M, Lemelle JL, Barthelme H, Marquand D, Schmitt M (2001) Bowel management with antegrade colonic enema using a Malone or a Monti conduit – clinical results. Eur J Pediatr Surg 11:315–318
65. Churchill BM, De Ugarte DA, Atkinson JB (2003) Left-colon antegrade colonic enema (LACE) procedure for fecal incontinence. J Pediatr Surg 38:1778–1780
66. Ahn SM, Han SW, Choi SH (2004) The results of antegrade continence enema using a retubularized sigmoidostomy. Pediatr Surg Int 20:488–491
67. Liloku RB, Mure PY, Braga L, Basset T, Moriquand PD (2002) The left Monti-Malone procedure: preliminary results in seven cases. J Pediatr Surg 37:228–231
68. Bruce RG, el-Galley RE, Wells J, Galloway NTM (1999) Antegrade continence enema for the treatment of fecal incontinence in adults: use of gastric tube for catheterizable access to the descending colon. J Urol 161:1813–1816
69. Calado AA, Macedo A, Barroso U, Netto JM, Ligouri R, Hachul M, Garrone G, Ortiz V, Sroug M (2005) The Macedo-Malone antegrade continence enema procedure: early experience. J Urol 173:1340–1344
70. Yerkes EB, Rink RC, Cain MP, Casale AJ (2002) Use of Monti channel for administration of antegrade continence enemas. J Urol 168:1883–1885
71. Gerharz EW, Vik V, Webb G, Woodhouse CRJ (1997) The in situ appendix in the Malone antegrade continence enema procedure for faecal incontinence. Br J Urol 79:985–986
72. Issa MM, Oesterling JE, Canning DA, Jeffs RD (1989) A new technique of using the in situ appendix as a catheterizable stoma in continent urinary reservoirs. J Urol 14:1385–1387

73. Cetinel B, Demirkesen O, Talat Z, Yaycioglu O, Solok V (2000) Application of continent retubularized ileal stoma (Monti procedure) to an ileocecal pouch. Urology 55:286
74. Lynch AC, Beasley SW, Robertson RW, Morreau PN (1999) Comparison of results of laparoscopic and open antegrade continence enema procedures. Pediatr Surg Int 15:343–346
75. Van Savage JG, Yohannes P (2000) Laparoscopic antegrade continence enema in situ appendix procedure for refractory constipation and overflow fecal incontinence in children with spina bifida. J Urol 164:1084–1087
76. Liard A, Seguier Lipszyc E, Mathiot A, Mitrofanoff P (2001) The Mitrofanoff procedure: 20 years later. J Urol 165:2394–2398
77. Tekant G, Emir H, Eroğlu E, Esentürk N, Büyükünal C, Danişmend N, Söylet Y (2001) Catheterisable Continent Urinary Diversion (Mitrofanoff principle) – clinical experience and psychological aspects. Eur J Pediatr Surg 11:263–267
78. Duckett JW, Lotfi AH (1993) Appendicovesicostomy (and variations) in bladder reconstruction. J Urol 149:567–569
79. Van Savage JG, Khoury AE, McLorie GA, Churchill BM (1996) Outcome analysis of Mitrofanoff principle applications using appendix and ureter to umbilical and lower quadrant stomal sites. J Urol 156:1794–1797
80. Khoury AE, Van Savage JG, McLorie GA, Churchill BM (1996) Minimizing stomal stenosis in appendicovesicostomy using the modified umbilical stoma. J Urol 155:2050–2051
81. Glassman DT, Docimo SG (2001) Concealed umbilical stoma: long term evaluation of stomal stenosis. J Urol 166:1028–1030
82. Van Savage JG, Yepuri JN (2001) Transverse retubularized sigmoidovesicostomy continent urinary diversion to the umbilicus. J Urol 166:644–647
83. Shaul DB, Harrison EA, Muenchow SK (2002) Avoidance of leakage and strictures when creating an invisible conduit for antegrade colonic enemas. J Pediatr Surg 37:1768–1771
84. Tam PKH (1999) Y-Appendicoplasty: a technique to minimize stomal complications in antegrade continence enema. J Pediatr Surg 34:1733–1735
85. Kajbafzadeh AM, Chubak N (2001) Simultaneous Malone antegrade continent enema and Mitrofanoff principle using the divided appendix: report of a new technique for prevention of stoma complications. J Urol 165:2404–2409
86. Narayanaswamy B, Wilcox DT, Cuckow PM, Duffy PG, Ransley PG (2001) The Yang-Monti ileovesicostomy: a problematic channel? BJU Int 87:861–865
87. Tackett LD, Minevich E, Benedict JF, Wacksman J, Sheldon CA (2002) Appendiceal versus ileal segment for antegrade continence enema. J Urol 167:683–686
88. Barqawi A, De Valdenebro M, Furness PD III, Kolye MA (2004) Lessons learned from stomal complications in children with cutaneous catheterizable continent stomas. BJU Int 94:1344–1347
89. Lemelle JL, Simo AM, Schmitt M (2004) Comparative study of the Yang-Monti channel and appendix for continent diversion in the Mitrofanoff and Malone principles. J Urol 172:1907–1910
90. Clark T, Pope JC IV, Adams MC, Wells N, Brock JW III (2002) Factors that influence outcomes of the Mitrofanoff and Malone antegrade continence enema reconstructive procedures in children. J Urol 168:1537–1540
91. Cain MP, Casale AJ, King SJ, Rink RC (1999) Appendicovesicostomy and newer alternatives for the Mitrofanoff procedure: results in the last 100 patients at Riley Children's Hospital. J Urol 162:1749–1752
92. Kurzrock EA, Karpman E, Stone AR (2004) Colonic tubes for the antegrade continence enema: comparison of surgical technique. J Urol 172:700–702
93. Snodgrass W (1999) Triamcinolone to prevent stenosis in Mitrofanoff stomas. J Urol 161:928
94. McAndrew HF, Griffiths DM, Pai KP (2002) A new complication of the Malone antegrade continence enema. J Pediatr Surg 37:1216
95. Sarin YK, Sinha A (2004) Acute appendicitis complicating Mitrofanoff procedure. J Pediatr Surg 39:1294–1295
96. Gosalbez R, Padron OF, Singla AK, Woodard JR, Galloway NT (1994) The gastric augment single pedicle tube catheterizable stoma: a useful adjunct to reconstruction of the urinary tract. J Urol 152:2005–2007
97. Paterson PJ, Jones BG (1996) Late conduit perforation in a modified classical Mitrofanoff continent urinary diversion. Br J Urol 78:474–475
98. Shandling B, Chait PG, Richards HF (1996) Percutaneous cecostomy: a new technique in the management of fecal incontinence. J Pediatr Surg 31:534–537
99. Chait PG, Shandling B, Richards HF (1997) The cecostomy button. J Pediatr Surg 32:849–851
100. Chait PG, Shlomovitz E, Connolly BL, Temple MJ, Restrepo R, Amaral JG, Muraca S, Richards HF, Ein SH (2003) Percutaneous cecostomy: updates in technique and patient care. Radiology 227:246–250
101. Rivera MT, Kugathasan S, Berger W, Werlin SL (2001) Percutaneous colonoscopic cecostomy for management of chronic constipation in children. Gastrointest Endosc 53:225–228
102. Duel BP, Gonzales R (1999) The button cecostomy for management of fecal incontinence. Pediatr Surg Int 15:559–561
103. Curry JI, Osborne A, Malone PSJ (1998) How to achieve a successful Malone antegrade continence enema. J Pediatr Surg 33:138–141
104. Schreiber CK, Stone AR (1999) Fatal hypernatremia associated with the antegrade continence enema procedure. J Urol 162:1433

105. Yerkes EB, Rink RC, King S, Cain MP, Kaefer M, Casale AJ (2001) Tap water and the Malone continence enema: a safe combination ? J Urol 166:1476–1478
106. Marshall J, Hutson JM, Anticich N, Stanton MP (2001) Antegrade continence enemas in the treatment of slow-transit constipation. J Pediatr Surg 36:1227–1230
107. Wren FJ, Reese CT, Decter RM (2003) Durability of the Malone antegrade continence enema in pregnancy. Urology 61:644
108. Fishwick JE, Gough DCS, O'Flynn KJ (2000) The Mitrofanoff procedure: does it last? BJU Int 85:496–497
109. Harris CF, Cooper CS, Hutcheson JC, Snyder HM III (2000) Appendicovesicostomy: the Mitrofanoff procedure – a 15-year perspective. J Urol 163:1922–1926
110. Mor Y, Quinn FMJ, Carr B, Mouriquand PD, Duffy PG, Ransley PG (1997) Combined Mitrofanoff and antegrade continence enema procedures for urinary and faecal incontinence. J Urol 158:192–195
111. Wedderburn A, Lee RS, Denny A, Steinbrecher HA, Kolye MA, Malone PSJ (2001) Synchronous bladder reconstruction and antegrade continence enema. J Urol 165:2392–2393
112. Yerkes EB, Cain MP, King S, Brei T, Kaefer M, Casale AJ, Rink RC (2003) Malone antegrade continence enema procedure: quality of life and family perspective. J Urol 169:320–323
113. Kokoska ER, Keller MS, Weber TR (2001) Outcome of the antegrade colonic enema procedure in children with chronic constipation. Am J Surg 182:625–629
114. Bau MO, Younes S, Aupy A, Bernuy M, Rouffet MJ, Yepremian D, Lottmann HB (2001) The Malone antegrade colonic enema isolated or associated with urological incontinence procedures: evaluation from patient point of view. J Urol 165:2399–2403

35 Vaginal Reconstruction for Congenital and Acquired Abnormalities

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35.1 Introduction

Most anomalies of the female genitalia are congenital and present at birth. These include vaginal atresia, vaginal obstruction due to imperforate hymen or a high vaginal septum, and anomalies of the internal and external genitalia in patients with intersex abnormalities. A smaller number of patients present at the time of puberty because of primary amenorrhea; this includes patients with the Mayer-Rokitansky syndrome and testicular feminization syndrome (TFS). The incidence of these anomalies varies between 1:4,000 live births and 1:10,000 live births. This wide range relates to differences in the definition and severity of these anomalies. The pediatric surgeon must have knowledge of a wide variety of presentations and the treatment for each entity. Surgical correction is predicated on the anatomy. Table 35.1 lists the common vaginal abnormalities and the surgical correction of each.

Table 35.1 Surgical correction of vaginal abnormalities

Vaginal abnormality	Surgical correction
1. Ambiguous genitalia	
a) Low insertion of vagina	Perineal flap vaginoplasty
b) High insertion of vagina (rare)	Passerini genito-vaginoplasty
2. Persistent urogenital Sinus	
a) Low insertion of vagina	Cutback vaginoplasty
b) Medium insertion of vagina	Urogenital mobilization
c) High insertion of vagina	Passerini genito-vaginoplasty
3. Absent vagina – with or without a uterus	
a) Mayer-Rokitansky syndrome	Colovaginoplasty
b) Testicular feminization syndrome	Colovaginoplasty
4. Cloacal anomaly	
a) Low common channel	Perineal vaginoplasty
b) Medium common channel	Urogenital mobilization
c) Long common channel	Intestinal vaginoplasty
5. Acquired abnormalities	
a) Resection for rhabdomyosarcoma	Colovaginoplasty

35.2 Anomalies of the Vagina

35.2.1 Embryology

Normal female genitalia result from the pairing, fusion, and recanalization of the Mullerian ducts; this process continues in a cephalad and caudal direction. The more cephalad portions form the fallopian tubes, and the fused portion, or uterovaginal primordium, forms the uterus and cervix. The vagina develops from paired solid outgrowths of the urogenital sinus called the sinovaginal bulbs. The fibromuscular portion of the vagina is formed from these bulbs as they grow caudally toward the end of the uterovaginal primordium. The precise boundary between the portion of the vagina contributed to by the uterovaginal primordium and the urogenital sinus is not defined. Defects can occur in development, fusion, and canalization. Failure of development leads to agenesis and failure of fusion leads to a variety of anomalies including duplication. Failure of canalization can lead to the formation of vaginal septa. The genital tubercle forms the clitoris and the genital swellings form the labia majora [15].

35.3 Ambiguous Genitalia

In order to evaluate patients who are born with abnormal external genitalia, a surgeon requires a systematic approach to diagnosis. A comprehensive evaluation includes a chromosomal analysis, family history, and laboratory studies, including urine testing for steroid levels and serum testing for electrolyte abnormalities. Physical examination should focus on establishing the presence or absence of gonadal symmetry. Ultrasound of the abdomen and contrast genitography complete the evaluation and can help accurately diagnose the various disorders and help guide their treatment.

35.3.1 Congenital Adrenal Hyperplasia (Female Pseudohermaphroditism)

Congenital adrenal hyperplasia is the most common diagnosis associated with ambiguous genitalia and has an incidence of 1:16,000 live births [16]. This disorder is characterized by a normal 46,XX karyotype. The phenotypic appearance of the patient is due to exposure to excessive endogenous androgens in utero. The amount of virilization can vary from mild clitoral hypertrophy to severe forms where the clitoris resembles a penis. The spectrum of phenotypes is illustrated in Fig. 35.1. This is due to deficiency of one of three enzymes: 21-hydroxylase, 11-hydroxylase, or 3-beta hydroxysteroid dehydrogenase. 21-Hydroxylase deficiency is seen in 75–91% of patients. The first goal of treatment is to address the steroid deficiencies. Glucocorticoid replacement is given if necessary and patients with the salt-wasting form of the syndrome are given fluorocortisone. Surgery is then advised depending on the amount of virilization present in the external genitalia.

35.3.1.1 Low Insertion of the Vagina

For most patients with the common form of this syndrome, low insertion of the vagina is present and surgical correction involves perineal vaginoplasty and/or clitoral recession. Every effort is made to preserve the neurovascular bundle in order to preserve vascularity and sensation. The procedure involves degloving of the enlarged clitoris and dissecting the erectile tissue to the bifurcation. Plication sutures are placed laterally and the tissue is folded under the pubis while the sutures are tied. In cases where the phallus is so large that reduction clitoroplasty is not possible (a rare event), the corporal bodies can be resected with preservation of the glans and neurovascular bundle.

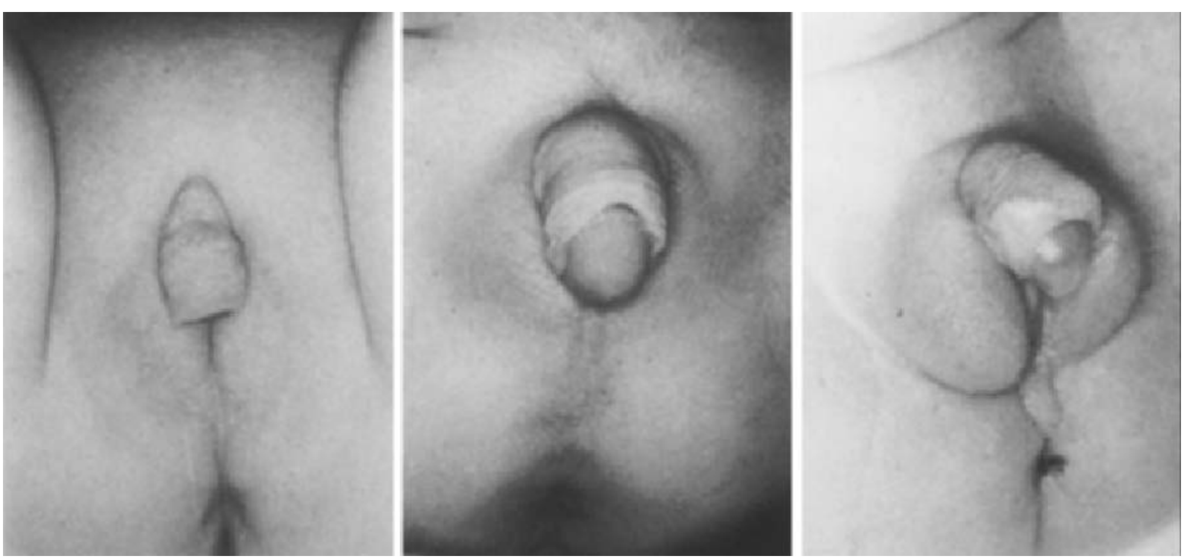

Fig. 35.1 Spectrum of phenotypes in patients with congenital adrenal hyperplasia

A large glans can be reduced by resecting a wedge of the ventral tissue and closing the skin edges. The vaginoplasty for low urogenital sinus abnormalities is illustrated in Fig. 35.2 and is performed by using a U- or Y-shaped incision posterior to the opening. The vagina is mobilized if necessary with traction sutures and the posterior anastomosis is performed with interrupted absorbable sutures.

35.3.1.2 High Insertion of the Vagina

Patients who are severely masculinized with a high insertion of the vagina, which is very rare, may require a complex clitorovaginoplasty. Historically, Hendren described a perineal pull-through vaginoplasty, as shown in Fig. 35.3 [6–8]. This involves separation of the vagina from the urogenital sinus and creation of two U-shaped flaps on the perineum above and below the area of the intended vaginal opening. The perineal flaps are required due to the inadequate length of the vagina, which enters the urogenital sinus high, close to the external sphincter of the urethra. In addition, the vagina itself is often diminutive. The final closure is an H-shaped configuration. Drawbacks to this procedure include an abnormal appearance to the perineum. We prefer the Passerini technique for this form of the adrenogenital syndrome because it results in a normal-appearing introitus [13]. Figure 35.4 illustrates the preoperative and postoperative appearance. Cystoscopy is performed and a Fogarty balloon catheter is placed in the vagina. An inverted Y incision is made starting at the base of the phallus, which is completely

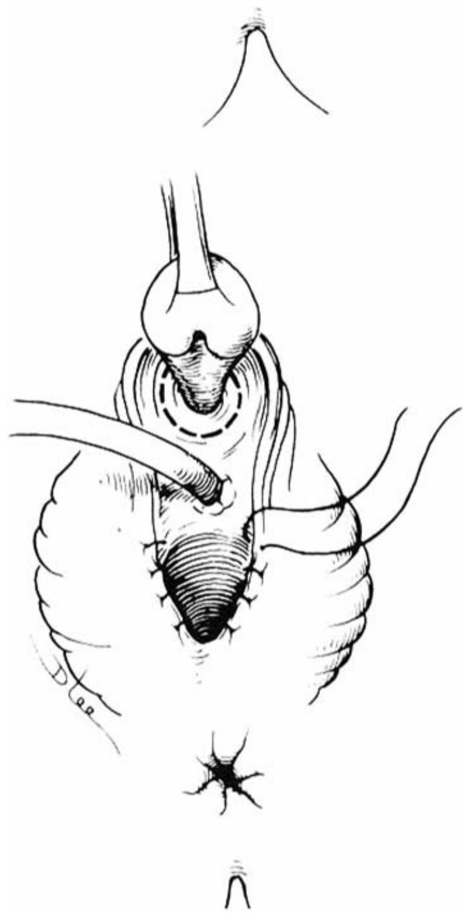

Fig. 35.2 Vaginoplasty for low urogenital sinus abnormalities

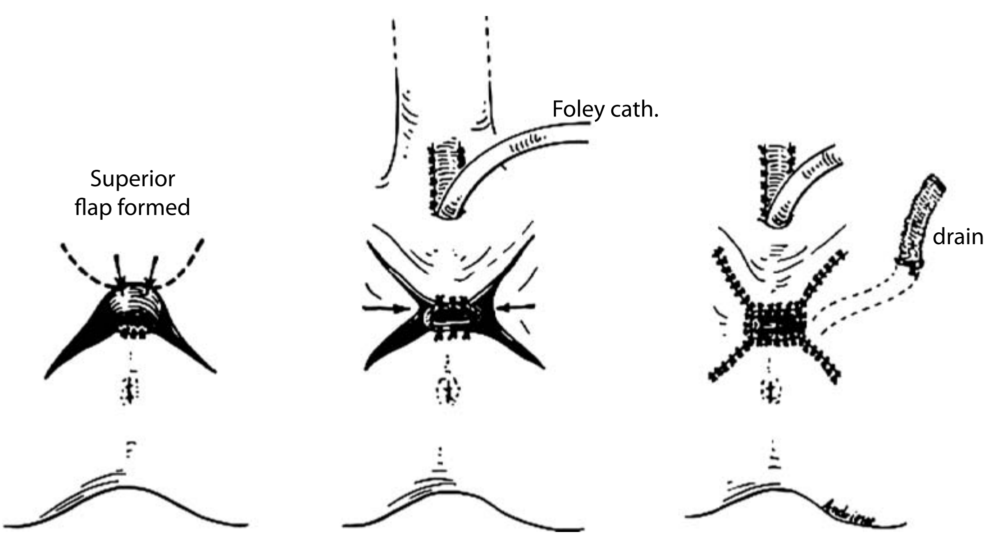

Fig. 35.3 Hendren flap vaginoplasty. *cath.* Catheter

degloved in the plane between Buck's fascia and the Dartos layer, mobilizing the urogenital sinus down to where the urethral meatus is to be located. This leaves flaps, which will be used to construct the distal vagina. The midline attachments of the perineal and pelvic floor musculature are divided posteriorly between the urogenital sinus and the rectum until the balloon is reached and the vagina exposed. The vagina is separated from the urethra and the opening is closed with interrupted absorbable sutures. Traction sutures are placed in the vagina. A reduction clitoroplasty is performed as described above except in cases where the phallus is too large, and then a resection of the corporal bodies and clitoral reduction is performed, taking care to preserve the dorsal neurovascular bundle. The urogenital sinus is split dorsally up to the point where it approaches the recessed clitoris, where a Y-flap is left and tacked down to form the urethral opening. The flaps that were created from the degloving are sutured to the open urogenital sinus and this is tabularized and anastomosed to the exposed vaginal opening. The skin edges are then closed and the vaginal opening is probed with a dilator.

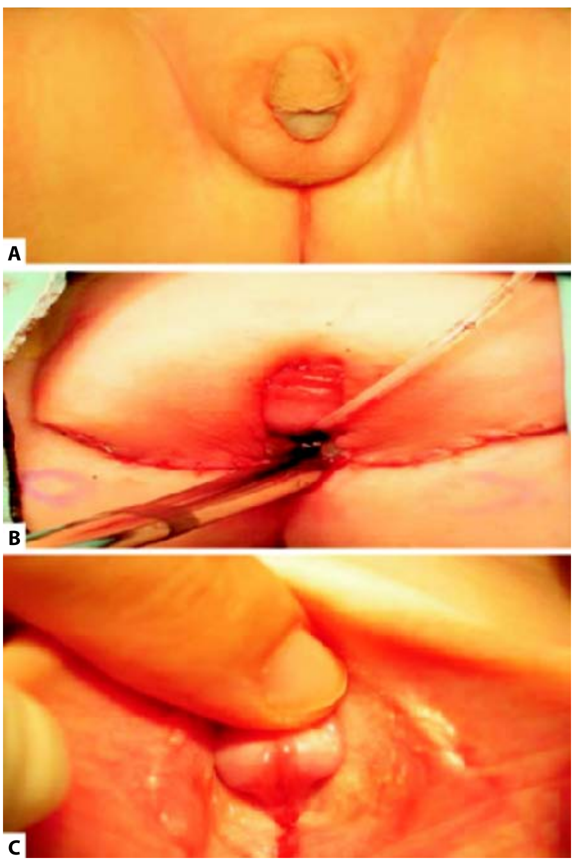

Fig. 35.4 Passerini genitovaginoplasty. **A** Preoperative anatomy. **B** Immediate postoperative result. **C** One month postoperative result

35.3.2 Mixed Gonadal Dysgenesis

Patients with this disorder have dysgenetic gonads and retained Mullerian structures, usually a streak gonad on one side and a dysplastic testis on the opposite side. Karyotypic analysis can reveal 46,XY or 45,XO/XY. Removal of the rudimentary gonads is necessary due to a risk of degeneration and malignancy. Most of these patients are raised as girls and surgical therapy includes bilateral gonadectomy, clitoral recession, and vaginoplasty, usually a simple perineal flap vaginoplasty.

35.3.3 True Hermaphroditism

Patients with true hermaphroditism are rare and have normal male and female gonadal tissue. Eighty percent have a karyotypic makeup consisting of 46,XX, but the chromosomal composition can be 46,XY or 46,XX/XY mosaic. The majority are raised as females and vaginal reconstruction follows the same guidelines as used for other forms of intersexuality.

35.4 Persistent Urogenital Sinus

Simple persistence of a urogenital sinus is defined as a common channel for the vagina and urethra, not associated with anorectal abnormalities or ambiguous genitalia. A genitogram should be performed to define the level of insertion of the vagina into the common channel. Low insertion may be treated with a cutback vaginoplasty. A medium level of insertion of the vagina may require total urogenital mobilization. A very high insertion is best treated with the Passerini technique.

35.5 Vaginal Atresia

Failure of the Mullerian ducts to reach the urogenital sinus contributes to congenital vaginal atresia. This can present as complete atresia, proximal atresia, or distal atresia, and each form has a different clinical picture. Complete atresia results in normal fallopian tubes and usually a rudimentary or bicornuate uterus. The vaginal plate is in the form of a fibrous band. In proximal atresia, the fallopian tubes as well as the cervix and uterus are hypoplastic or absent. Distal atresia results when there is a failure of the sinovaginal bulbs, arising from the urogenital sinus, to proliferate. In this presentation, the fallopian tubes, uterus, and cervix are normal.

35.5.1 Mayer-Rokitansky Syndrome

Classic total agenesis of the vagina has been described as Mayer-Rokitansky syndrome or Rokitansky-Küster-Hauser syndrome. These patients are characterized by a 46,XX karyotype and they account for 15% of females who present with primary amenorrhea. Complete absence of a vagina is present in 75% of patients, while 25% will have a short vaginal pouch. The presence of a uterus is variable, but usually a rudimentary bicornuate uterus is seen, which rarely produces menstrual blood, but may cause monthly cramping. Normal ovaries and fallopian tubes are present. One-third of these patients have urinary tract anomalies and about 12% have skeletal anomalies.

35.5.2 Testicular Feminization Syndrome

This entity is one form of androgen insensitivity syndrome, including defects in testosterone synthesis, androgen resistance syndromes, and 5-alpha reductase deficits. TFS is due to insensitivity of the androgen receptors in the external genitalia. The karyotype in these patients is 46,XY. Diagnosis is frequently made during routine inguinal herniorrhaphy in a girl when a gonad is found in the inguinal canal. Patients can also present with primary amenorrhea at puberty. Typically, examination reveals a very short vaginal vault. Treatment consists of bilateral gonadectomy to prevent masculinization at puberty and the rare case of malignancy. Reconstruction with an intestinal segment is performed at puberty. Some cases can be treated with progressive vaginal dilatation if there is a reasonable vaginal pouch.

35.5.3 Acquired Abnormalities

Rhabdomyosarcoma of the vagina or sarcoma botryoides has the typical presentation of a lobulated, grapelike mass protruding from the introitus, and occurs only rarely in girls over the age of 8 years [10]. The mass is irritating and there is a bloody discharge present. The tumor begins in the subepithelial layer and can expand rapidly and be multicentric. In the past, these tumors have been treated with radical pelvic exenteration. A less mutilating approach involving chemotherapy and local resection is now used. In rare cases, patients may not respond completely to chemotherapy and may require complete vaginal resection and hysterectomy. In one report of 17 females, surgical cure was achieved in 15 with subsequent successful menses and conception in a few [11]. When resection results in a cure, the vagina can be reconstructed at or after puberty with a colovaginoplasty.

35.6 Colovaginoplasty

Reconstruction of the vagina for congenital or acquired absence can be accomplished by several different procedures, which are summarized in Table 35.2. The goals of therapy should include fertility, if at all possible, adequate sexual function without the need for continual dilatations or lubrication, and elimination of the risk of malignant change in the intraabdominal gonads. Historically, the Frank procedure, the McIndoe procedure, and the laparoscopic traction approach described by Vecchietti have been used. The Frank procedure involves progressive nonsurgical dilatation with graduated dilators used for 20 min per day [4]. A 43% success rate was reported and failures were attributed to a lack of compliance. McIndoe described dissection of the space between the rectum and bladder with placement of a mold covered with a split-thickness skin graft [12]. Dilators are then used during epithelialization of the neovagina. Complications rates are small (10%) and include

Table 35.2 Techniques of vaginal reconstruction for congenital and acquired absence [5]. *STSG* split-thickness skin graft

Procedure	Technique	Advantages	Disadvantages
Frank	Graduated dilators	Noninvasive	Noncompliance
McIndoe	STSG-covered stent	No intraabdominal surgery	Requires dilators, lubricants
Vecchietti	Intraabdominal traction	Can be done laparoscopically	Requires dilators, lubricants
Intestinal vaginoplasty	Bowel as neovagina	No lubricants required	Major operation

failure of the graft to take, hematoma formation, and fistula occurrence. A drawback to this method is the requirement for lubricants. The Vecchietti procedure involves intraabdominal traction applied to the perineal membrane causing invagination over the course of 1 week [2]. This technique still requires the use of dilators. All of these techniques rarely produce a deep vaginal cavity. Because of this, we prefer intestinal vaginoplasty for reconstruction and have obtained very satisfactory results using a segment of colon as an interposition graft [17].

The technique of colovaginoplasty involves exploration of the abdomen and pelvis through a Pfannenstiel incision or by laparoscopy (Fig. 35.5). In cases of Mayer-Rokitansky syndrome, the uterine horns are excised and the ovaries are left in place so that the patient does not require exogenous hormone replacement. The patient can also harvest ova for in vitro fertilization using a surrogate mother if she desires. A 10- to 15-cm segment of sigmoid colon is then isolated, preserving the blood supply. One end is closed in two layers to form the vertex of the neovagina, and the other end is left open to act as the neovaginal orifice. The continuity of the colon is reestablished. The hymenal region of the vulva is incised and the vaginal tract is bluntly dissected between the bladder and rectum to the level of the peritoneal reflection. The open sigmoid loop is pulled down into position. A single-layer anastomosis is performed in the hymenal region.

Our series includes 36 patients who underwent intestinal vaginoplasty for a variety of anomalies and is depicted in Fig. 35.6. Fourteen patients had Mayer-Rokitansky syndrome, 10 had TFS, 8 had cloacal anomalies, 3 had vaginal rhabdomyosarcoma, and 1 patient had mixed gonadal dysgenesis. Some groups have described a small incidence of diversion colitis with this procedure, but we have not seen this [1,9,14]. All patients report adequate patency for intercourse and no need for lubrication. None of our patients have experienced excessive mucus production.

35.7 Cloacal Anomalies

If a persistent urogenital sinus is associated with an anorectal malformation the patient has a cloacal anomaly. This involves all three systems joining in

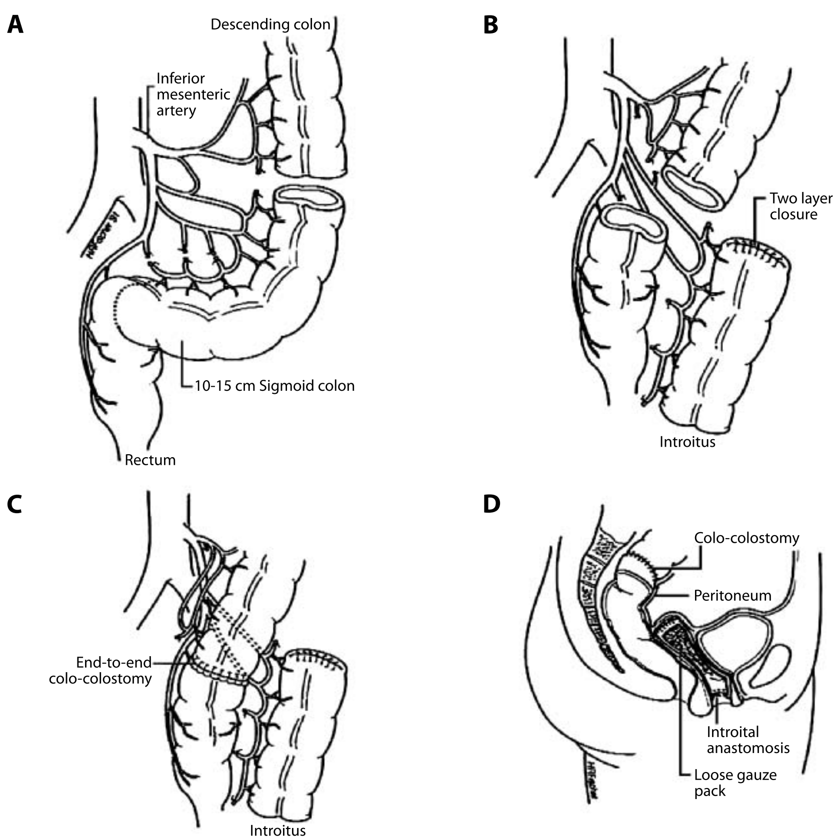

Fig. 35.5 Technique of colovaginoplasty. **A** Isolation of 10–15 cm of sigmoid colon. **B** The segment is positioned to anastomose to the hymenal ring. **C** The colocolostomy is completed. **D** The completed vaginoplasty is loosely packed with Vaseline gauze

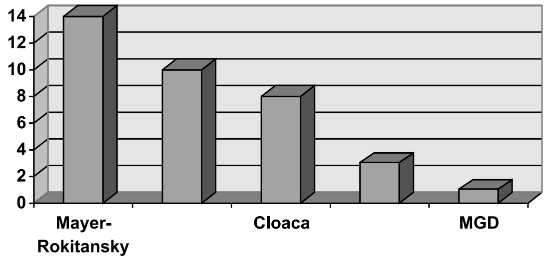

Fig. 35.6 Experience with intestinal vaginoplasty. *TFS* Testicular feminization syndrome, *Rhabdo* rhabdomyosarcoma, *MGD* Mixed gonadal dysgenesis

a common channel. The appearance externally is a closed perineum with a single opening. A low common channel is not usually associated with urinary obstruction, and a perineal vaginoplasty and pull-through anoplasty are adequate. A medium or long common channel is usually associated with urinary obstruction, and surgical correction is more complex. In the medium-length channel (2–4 cm), urogenital mobilization is adequate for vaginal reconstruction. In the long-channel variety (> 4 cm), an intestinal vaginoplasty is needed.

35.8 Congenital Vaginal Obstruction

Other forms of vaginal obstruction include imperforate hymen, labial fusion, and high transverse vaginal septum. Presentation can be similar for all forms of obstruction and patients often have a lower abdominal mass on examination. They can also present with urinary tract obstruction. With an imperforate hymen, the patient can present with hydrocolpos at birth or early infancy, or hydrometrocolpos at puberty. A bulging mass is seen on the perineum in the newborn and this can often be opened using a hemostat.

Labial fusion can present in one of two ways:

1. A primary skin bridge may be present and these patients can present with urinary tract infections; division may require anesthesia
2. Synechiae may be seen in girls typically between 1–5 years of age. This may due to chronic irritation or relative lack of estrogen stimulation. The membrane can be divided in an outpatient setting but may recur. It is useful to apply cream daily to the area post correction.

A high transverse vaginal septum is a rarer form of vaginal obstruction and can be associated with partial anterior vaginal agenesis or persistence of a common urogenital sinus [3]. Treatment is drainage in the acute setting followed by resection of the septum and perineal vaginal pull-through.

35.9 Summary

A large spectrum of anatomical vaginal abnormalities has been presented and an equally large spectrum of surgical procedures has been reviewed. Thorough evaluation of the anatomy of a newborn with ambiguous genitalia, persistent urogenital sinus, vaginal atresia, or cloaca is required before deciding which type of surgical correction is necessary. Indications for total replacement with a colovaginoplasty include total vaginal agenesis, as in the case of Mayer-Rokitansky syndrome, and absence of a uterus and proximal vagina, as in the case of TFS. A cloacal anomaly may require total replacement in the form of an intestinal vaginoplasty, usually utilizing a portion of the small bowel. Complex reconstructions like a Passerini clitorovaginoplasty are indicated for the highly virilized adrenogenital syndrome patient or a patient with a persistent urogenital sinus with a very high insertion of the vagina.

References

1. Baldwin JF (1904) The formation of an artificial vagina by intestinal transplantation. Ann Surg 40:398–403
2. Borruto F (1992) Mayer-Rokitansky-Kuster syndrome: Vecchietti's personal series. Clin Exp Obstet Gyencol 19:273–274
3. Bowman JA Jr, Scott RB (1954) Transverse vaginal septum; report of four cases. Obstet Gynecol 3:441–446
4. Fall FH (1940) A simple method for making an artificial vagina. Am J Obstet Gynecol 40:906–917
5. Graziano K, Teitelbaum DH, Hirschl RB, et al. (2002) Vaginal reconstruction for ambiguous genitalia and congenital absence of the vagina: a 27 year experience. J Pediatr Surg 37:955–960
6. Hendren WH, Crawford JD (1969) Adrenogenital syndrome: the anatomy of the anomaly and its repair. Some new concepts. J Pediatr Surg 4:49–58
7. Hendren WH, Donahoe PK (1980) Correction of congenital abnormalities of the vagina and perineum. J Pediatr Surg 15:751–763
8. Hendren WH, Atala A (1994) Use of bowel for vaginal reconstruction. J Urol 152:752–755
9. Hensle TW, Reiley EA (1998) Vaginal replacement in children and young adults. J Urol 159:1035–1038
10. Hilgers RD, Malkasian GD Jr, Soule EH (1970) Embryonal rhabdomyosarcoma (botryoid type) of the vagina. Am J Obstet Gynecol 107:484–502

11. Martelli H, Oberlin O, Rey A, et al. (1999) Conservative treatment for girls with nonmetastatic rhabdomyosarcoma of the genital tract: a report from the Study Committee of the International Society of Pediatric Oncology. J Clin Oncol 147:2117–2122
12. McIndoe AH (1950) Treatment of congenital absence and obliterative conditions of the vagina. Br J Plast Surg 2:254–267
13. Passerini-Glazel G (1989) A new 1-stage procedure for clitorovaginoplasty in severely masculinized female pseudohermaphrodites. J Urol 142:565–568
14. Pratt JH (1972) Vaginal atresia corrected by the use of small and large bowel. Clin Obstet Gynecol 59:639–649
15. Stenchever MA, Droegemuller W, Herbst AL, Mishell DR (eds) (2001) Comprehensive Gynecology, 4th edn. Mosby, St. Louis
16. Therrell BL Jr, Berenbaum SA, Manter-Kapanke V, et al. (1998) Results of screening 1.9 million Texas newborns for 21-hydroxylase deficient congenital adrenal hyperplasia. Pediatrics 101:583–590
17. Wesley JR, Coran AG (1992) Intestinal vaginoplasty for congenital absence of the vagina. J Pediatr Surg 27:885–889

36 Adult Sexual Function after Anorectal Malformation Repair

Melissa C. Davies and Christopher R.J. Woodhouse

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36.1 Introduction

Anorectal malformations (ARM) and cloacal anomalies are rare and complex malformations of the lower gastrointestinal and genitourinary tract. They affect approximately 1:3,500–1:5,000 live births [1]. They may occur in isolation or in association with abnormalities of the urogenital, cardiovascular and skeletal systems as well as with the vertebral, anal, cardiac, tracheoesophageal, renal and limb abnormalities (VACTERL) association. Treatment has traditionally focussed on achieving urinary and faecal continence with preservation of renal function. Diversion of the bowel may be required. It will often be possible to create a continent bladder, though emptying may require intermittent catheterisation. A continent rectum is more difficult and many children remain clean only by a process of "controlled constipation" or continue with a colostomy [2]. Faecal continence remains a challenge and is achieved in about 60% of patients.

The most severe form of cloaca is rare. It is often associated with other major congenital anomalies. The cloaca is a common channel comprising the urethra, vagina and rectum; there is almost never an anal sphincter and seldom a urethral one (Fig. 36.1). In intermediate forms the urethral sphincter may be normal. The urinary anomalies are potentially lethal and reconstructive surgery in childhood is technically very difficult. There is, therefore, little information

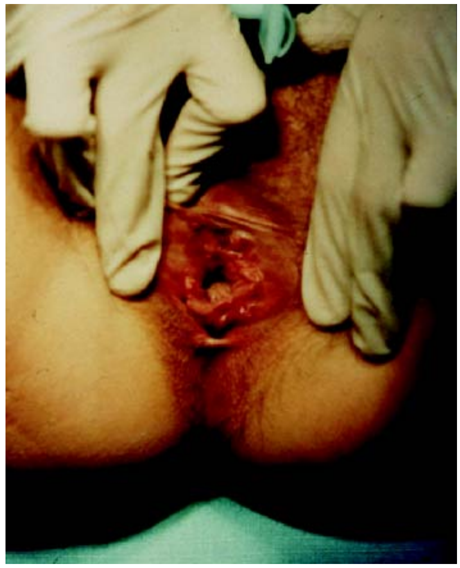

Fig. 36.1 Adult patient with an uncorrected cloaca

on long-term outcomes. This condition must be distinguished from cloacal exstrophy, which is a severe variant of ectopia vesicae.

With improved surgical techniques and paediatric intensive care facilities, many patients with ARM now live relatively normal lives, with a near-normal life expectancy. Therefore, sexual function and fertility are of increasing importance. Sexual function may be impaired for several reasons:

1. Associated malformations of the genital system
2. Iatrogenic injury at the time of surgical repair either to genital/reproductive structures, or to their nerve supply structures
3. Psychological problems as a consequence of ARM may impair relationships
4. Social isolation, which may be associated with urinary or faecal incontinence

Patients who have normal sexual function may also experience difficulties with fertility. In male patients these two aspects may be inextricably linked, as men with erectile difficulties also may fail to ejaculate and will require sperm extraction techniques. The extent of these problems in the adult ARM population is difficult to assess as many of the long-term outcome studies are carried out to include a paediatric population, where questioning about sexual activity and fertility would be inappropriate. Therefore, cohorts for late follow up have to be recruited from a variety of sources, including patient support groups, which may give an overestimate of late problems. Furthermore, research tends to concentrate on outcomes for faecal continence. Even when sexual function is considered, it seems to be little more than a footnote, sometimes without distinguishing between genders.

In spite of improved surgical techniques, there is still an appreciable amount of morbidity associated with ARM. The consequences of a serious medical condition combined with protracted and repeated hospital admissions will undoubtedly have an effect on both the patients and their families. Patients with ARM have been shown to have increased rates of significant emotional problems. In two recent series the rates of psychiatric diagnoses were 58% and 35%, respectively [3,4]. The higher rate may have been due to the twice-daily anal dilatations that the patients underwent as children. The intrusiveness of such a procedure may have had long-term consequences for self-esteem and body image, both of which are important in the context of sexual confidence. It may be prudent to consider this when defining bowel management regimes for young patients with ARM, where regular rectal enema or washout administration may be required to achieve faecal continence.

Bai et al. [5] have also postulated that the effects of faecal incontinence would have a knock-on effect in adult life, impacting on, for example, future occupation and relationships. It would seem likely that young adults who are faecally incontinent will have issues with close relationships and self-esteem. Furthermore, the stigma of incontinence may exacerbate any problems the patients may have, as society views people who are incontinent in a negative way, further limiting the role these patients feel able to play in society [6].

36.1.1 Female Genital Anomalies

The original descriptions of ARM noted the presence of a faecal fistula that was usually said to open into the vagina. It would seem that true rectovaginal fistula is, in fact, rare. Most of the fistulae actually open into the vestibule or represent a missed diagnosis of cloaca [7].

Aside from the fistula, the associated female genital anomalies have been poorly documented but are known to be common. Authors writing on surgical reconstruction in childhood may not be too concerned with the genitalia and surgeons may simply fail to record anomalies that are not directly relevant to their management [8]. Hall et al., in trying to quantify the incidence of genital anomalies from a retrospective chart review, found that the vagina had been assessed in only 72 out of 162 girls and the internal genitalia in only 51. They found that 22 (out of 72; 32%) had a vaginal anomaly and 18 (out of 51; 35%) had a uterine anomaly. All of the uterine anomalies were serious (Table 36.1 and Fig. 36.2) [9]. Subsequent studies have found the rate of female genital anomalies to be lower [10]; this may be in part explained by the delayed diagnosis of female genital anomalies, as many of these do not present until puberty.

In females with cloaca who survive into adult life, the pattern of genital anomalies is similar. Menstrual function is present in 68%, but of these, more than half had an obstructed outflow at a variety of levels. In 10 out of 41 girls, a diagnosis of absent or vestigial uterus was made on laparotomy in infancy, but 6 of them developed normal menstrual function at puberty, an observation that should make the paediatric surgeon very reticent in commenting on the internal genitalia [11]. The authors of this study further recommend that all females with cloaca undergo further gynaecological assessment early in puberty.

Table 36.1 Genital anomalies in female children born with anorectal malformations (ARM). Data from Hall et al. [9]

Lower genital tract anomalies (n = 72)	Number
Vaginal septum	13
Vaginal agenesis	5
Distal stenosis	1
Lower-third agenesis	1
Hymenal band	1
Total	**21 (29,1%)**
Upper genital tract anomalies (n = 51)	
Bicornuate uterus	8
Uterus didelphys	8
Hypoplastic uterus	2
Total	**18 (35%)**

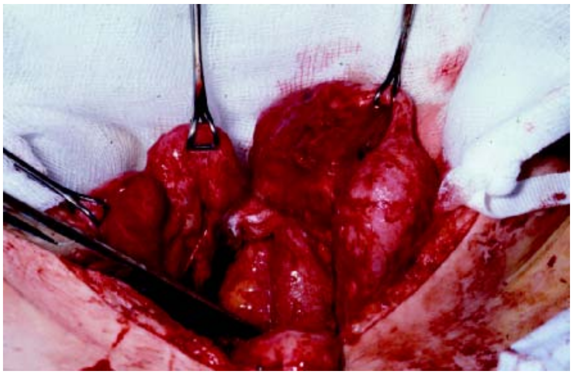

Fig. 36.2 Operative photograph of a uterus didelphys. There is a clamp on the right half and the left tube. The left half is rotated with the Fallopian tube and broad ligament lying posteromedially

36.1.2 Female Sexual Function

If the vagina is present, intercourse is possible unless there is a psychological problem, usually related to the fear of faecal incontinence. Unfortunately, the quality-of-life studies do not distinguish between men and women in giving these data. Sexual difficulties, particularly related to anal dysfunction, were found in 13% of those born with a low anomaly and 30% of those with a high or intermediate anomaly. The gender of the patients was not specified [12,13].

In those who have undergone vaginal surgery as part of their reconstruction, scarring is a common problem and can lead to stenosis and dyspareunia. The increasing recognition of the efficacy of vaginal dilatation alone has made this the initial treatment of choice [14], as failure will not have a detrimental effect on vaginal/perineal tissue. Nevertheless, there are cases where it is almost impossible to provide an adequate vagina without transferring healthy tissue such as bowel or skin.

Hall et al. report that 8 out of 17 patients for whom enough information was available had vaginal scarring and impaired vaginal function. It was not known what effect this had on intercourse. It was felt that much of the problem lay in the unoestrogenised tissue that was operated upon in infancy. They recommended the deferment of vaginoplasty to the post-pubertal period if possible [9]. In those patients who have undergone repeated vaginal operations, the failure rate of subsequent operations is unfortunately high. In a recent series, stenosis occurred in 40% of patients who had undergone more than one vaginal reconstruction [15]. If the vagina is too small, penetrative intercourse is impossible, but some patients prefer to use non-penetrative techniques rather than risk worsening their already tenuous anal control.

36.2 Pregnancy

Where intercourse is possible and the Mullerian structures normal, pregnancy is possible. Many of those looking after ARM patients are concerned about the potential damage that could occur during the course of pregnancy and childbirth. These include:

1. Possible deterioration in renal function – particularly if a degree of renal failure already exists.
2. Worsening urinary incontinence, or damage to urinary diversions during delivery.
3. Decreased faecal control as a result of pelvic floor injury.

In a series of 29 pregnancies in 20 women who had undergone lower urinary tract reconstructions for a variety of underlying conditions [16], it was reported that there was no significant deterioration in the renal function. Two-thirds of the patients underwent Caesarean section; the reasons postulated for this were reluctance to risk possible damage to the repair and the patient's continence mechanism, spinal anomalies that precluded spinal analgesia and the benefits of a planned procedure to ensure the availability of suitable staff.

Uterine anomalies, such as the didelphys, bicornuate or septate, may be symptomless and not present until the patient suffers recurrent miscarriage. These patients will require specialist gynaecological intervention. There has been some recent success in embryo implantation; overall, half of the women achieved a successful delivery. However, the pregnancy rate per embryo transfer is low: 19% for unicornuate and septate uteri and 11% for didelphys [17]. If the uterus fails to develop or is absent, there is nothing that can be done to enlarge it.

The issue of the potential for affected offspring may be a concern to females with an ARM. However, familial ARM is rare and at present no single gene has been implicated. There are numerous reports of cases with varying patterns of inheritance See Chaps. 2 and 3 [18–20]. Nevertheless, potential parents will often ask if screening is possible. At present it seems that the pick-up rate for all of the gastrointestinal atresias is low. When found, they are usually identified through the associated anomalies. In a large series from France of 118,265 foetal ultrasound scans, the sensitivity for ARM was 8.2%, and none of the 27 isolated cases was identified. The mean stage of pregnancy at which ARM was identified, was 27.5 weeks [21].

36.3 Vaginal Reconstruction

The problem of the woman with an absent or inadequate vagina is unresolved. Much of the reported work has been on patients with intersex conditions, but the same principles apply to the vaginal anomalies associated with ARM. A variety of techniques for reconstruction are available. None has been shown to produce an ideal substitute for the natural organ and it seems unlikely that any would have the same sexual sensation as the natural vagina. In an extensive review of the literature, it was found that follow-up assessment was usually confined to the observation that penetrative intercourse was possible, with no attempt to measure its quality [22]. There was little critical evaluation of female sexuality. In Peña's series there were 22 females over the age of 14 years. Only seven were considered to be gynaecologically normal. Six had a small or absent uterus and nine had an obstructed uterus at a variety of levels [7]. It is these patients who present a major surgical challenge. Although it is correct to evaluate as well as possible the quality of intercourse to decide which technique is the best, it must be remembered that surgery only enables penetrative intercourse to take place; it might be said that poor intercourse may be better than none at all.

If the vagina is normal and unscarred but narrow, every effort should be made to enlarge it by progressive self-dilatation. In motivated adults, it is possible over several months to lengthen a vagina from 5 mm to 10 cm with sufficient diameter for intercourse, with graded acrylic moulds (Fig. 36.3). The advantage of this technique is that the vagina maintains normal physiological function, including lubrication [23].

There is some conflict about the wisdom of routine dilatation of the vagina after genitoplasty in infancy. Krege et al. suggest that it should not be routine if only because of the psychological problems that it may cause (though they offer no evidence for this fear) [24]. Gearhart and, even more strongly, Hendren recommend dilatation to prevent postoperative stenosis [25]. In spite of this, all of the patients required further, albeit minor, surgery at puberty to allow intercourse.

In cases where the perineal tissues are scarred, dilatation therapy may not be an option. Occasionally, it may be possible to dilate the vagina under anaesthetic sufficiently that the woman can maintain its calibre with regular dilatation or intercourse. When dilatation is possible, the outcome for sexual intercourse appears satisfactory in the small number of cases that have been reported: in one series all of three women with congenital adrenal hyperplasia (CAH) were able to have satisfactory intercourse and two became pregnant. In contrast, 50% of patients (none of whom had CAH) who had various forms of reconstruction complained of bleeding with intercourse [23].

Fig. 36.3 Amielle dilators

If reconstructive surgery is required, careful definition of the extent of the problem is required. The patients should be adequately assessed and long-term follow-up is essential (Table 36.2). The external genitalia and vagina are best defined by examination under anaesthetic by all of the surgeons likely to be involved in the reconstruction – gynaecologist, plastic surgeon and urologist. The cystoscope can be used to inspect the vagina above a narrow introitus, often finding that the upper part of the vagina is normal. The internal genitalia are best defined by magnetic resonance imaging. The results are much better if there is a natural introitus, with clitoris and labia.

A narrow vagina may be augmented with bowel or skin. Bowel augmentation may be achieved with any suitable part of the large or small intestine. A piece of ileum equal in length to the existing vagina and with a long enough pedicle to reach the introitus is selected. It is opened on its antimesenteric border. The vagina is opened longitudinally either anteriorly or posteriorly and sutured "face to face" with the ileum. Follow up has been confined to establishing that intercourse takes place without undue difficulty. Up to 70% of women who had an intestinal vagina formed in infancy report the ability to have intercourse, with a 10% incidence of dyspareunia [26].

Table 36.2 Vaginoplasty assessment considerations; adapted from Davies et al. [15]. *MRI* Magnetic resonance imaging, *EUA* examination under anaesthesia

Assessment	Recommendations
Imaging	All patients should have pelvic MRI to assess: • Cervix – presence and patency • Distance between proximal and distal vagina • Presence of uterus +/- haematometra • Bony pelvis
Chromosome studies	Normally performed in neonates, but diagnosis should be confirmed
Psychology	Required pre- and post-operatively to: • Agree timing of surgery • Ensure expectations of surgery are realistic • Improve compliance with post-operative dilator therapy • Help deal with post-operative complications
EUA	Essential to assess: • Vagina • Urethra, bladder and rectum • Pliability of tissues • Presence and extent of scarring
Post-operative assessment	Should be ongoing and long-term and include: • Cosmesis • Menstruation • Sexual intercourse
Long-term follow-up	Possible long-term malignancy risk: • Annual vaginal examination • Early reporting of bleeding or discharge

Skin augmentation is usually performed by using skin from the medial aspect of the thigh. The technique was originally described by Sir Archibald McIndoe in 1938. There have been several modifications, but the principle remains the same: a cavity is created in the position of the vagina and lined with meshed split skin on a mould. The initial complication rate is high as the skin fails to take in about 65% of patients. Most will require at least one revision procedure and final surgery is best left until after puberty. About 75% of patients are able to have intercourse (and some of the remainder may be unwilling rather than unable). Self-dilatation with a mould is usually needed in periods of sexual inactivity [27]. A variety of other tissues have been used as free grafts to line a vagina that has been split open longitudinally. Many of the series are small and the results unpredictable [28].

In the small number of girls with normal internal genitalia and no vagina (Fig. 36.4), the timing of surgery is critical; however, menstruation can be suppressed temporarily with luteinising hormone releasing hormone agonists. Bowel, skin, amnion and other materials have been used to make replacement vaginas in these situations, but again, none is satisfactory. Skin on a pedicled flap is rather bulky and split skin has a poor take. The vagina is dry and the squamous lining desquamates, producing a foul discharge. Amnion has shown quite promising results, but availability is limited, especially in the era of HIV infection. Ileum may be too narrow, colon too large and both may produce copious smelly mucus, thus condemning the girl to a lifetime of wearing sanitary protection, which is far from ideal. There is a general tendency to make intestinal vaginas too long, which compounds the problem (Fig. 36.5).

The timing of reconstruction is very important. If surgery is carried out in young girls, dilatation may be required throughout childhood for an organ that, it may be hoped, will not be used for 14 years or more. Perhaps the most compelling argument against surgery in infancy is the risk of neoplasia. In a review, Schober identified five cases of squamous cell carcinoma of skin vaginas and four cases of adenocarcinoma of intestinal vaginas between 1927 and 1994.

The cases occurred in women between 25 and 30 years old and between 8 and 25 years after reconstruction [22]. The average time from surgery to diagnosis is estimated to be 17 years [29]. They are admittedly very rare cancers and none of the relatively large series of vaginal reconstruction report any cases. Nonetheless, the risk remains and a good case can be made for deferring elective surgery until a woman needs a vagina and can give her own consent.

Fig. 36.4 Clinical photograph of an adult female born with anorectal and vaginal agenesis

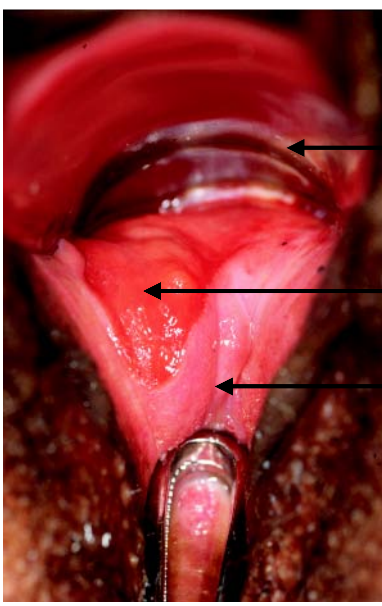

Fig. 36.5 Clinical photograph of a vagina constructed from the ileum in a woman born with anorectal agenesis. The appearance is good, but persistent coital haemorrhage has been a severe impediment to regular intercourse

36.4 Male Genital Anomalies

The incidence of male genital anomalies is lower than for female genital anomalies. However, an exact level is difficult to define because of limited and contradictory literature. Hoekstra et al. recorded all of the genitourinary anomalies associated with ARM in 150 children [30]. The genital anomalies in boys were relatively uncommon and mainly of little significance, especially when considering sexual function in later life (Table 36.3).

In a later series, the incidence of genital anomalies was identical, but the pattern was very different. The commonest was ambiguous genitalia and the gender of rearing may not have corresponded to the genotype. Cho et al. recorded 16 anomalies in 69 boys (23%), assuming all of those with ambiguous genitalia were male (Table 36.4) [31]. It would seem, therefore, that there is a wide range of possible genital anomalies in boys, many of which would have little impact on future sexuality or fertility.

Although spinal cord anomalies have a major impact on sexual function in both genders, it tends to be more severe in males, at least in those born with myelodysplasia. There is a high incidence of neurovesical dysfunction in children with ARM. The main cause is

Table 36.3 Genital anomalies in 80 boys born with ARM [30]

Genital anomalies (n = 80)	Number
Hypospadias	10
Scrotal deformity	3
Penile duplication	2
Cryptorchidism	2
Micropenis	1
Total	**18 (23%)**

Table 36.4 Genital anomalies in male children born with ARM. Data from Cho et al. [31]

Genital anomaly (*n* = 69)	Number
Ambiguous genitalia	7
Cryptorchidism	5
Bifid scrotum	4
Total	**16 (23%)**

the associated spinal abnormalities, especially hemivertebrae. The reconstructive surgery probably has little additional effect. The bladder problem is usually an upper motor neurone hyperreflexia due to lumbosacral anomalies. In a review of the literature, 29% of those with a high ARM and 6% with a low anomaly had urinary incontinence [32]. If an analogy can be drawn with the general myelodysplasia population, the impact on potency may not be great, especially if the neurological lesion is incomplete. All males with myelodysplasia with intact sacral reflexes and urinary continence are potent. With absent sacral reflexes, 64% with levels below D10 and 14% with levels above D10 are potent [33]. As most of those with ARM and neuropathy have a level well below D10, few should be impotent.

36.4.1 Male Sexual Function

There are few data on male sexual function. It would seem that no unit has been able to follow up its own patients into mature adult life. It is recorded that no adult patients have normal faecal continence, which is certainly a bad start for any individual wishing to share his life and bed with another [34]. Nonetheless, in this study there was no difference in arrangements for family life between patients and the normal population in The Netherlands, and the same proportions were cohabiting. There was no specific reference to sexual function, but it was striking that 24% of patients never had a lasting relationship [34].

Peña specifically records the sexual function of his older patients. All of 20 claimed to be potent, 14 enjoyed masturbation, 6 had had intercourse and one was married and has a child [7]. Iwai et al. give similar figures for all of their five male patients [35]. Rintala et al. have compared their own patients with low and high anomalies separately and compared them to healthy controls. In those with a low anomaly, 11 out of 83 patients (13%) had difficulties with intercourse but always related to concerns about anal function. No mention was made of any patient being physically unable to have intercourse [12]. In patients with an intermediate or high anomaly, 10 out of 33 (30%) had sexual dysfunction. Although it is not stated how many of these were male, three had erectile dysfunction and two had retrograde ejaculation. Six of the ten cited fear of anal incontinence as a significant limitation on their sexual activity [13].

Whilst sexual function may be easily assessed by questioning the patient, fertility requires closer investigation of the patient with sperm samples and imaging of the vas deferens. Therefore, in much of the published literature we see paternity being used as a means of assessing fertility. Impaired sexual function is seen in adult male patients who have undergone surgical procedures on the prostate and rectum, but appears to be a rare consequence of the surgery entailed in repairing ARM [36]. Pryor and Hendry report five patients with ARM who were infertile due to obstructed ejaculatory ducts. In some (they were part of a larger group) it was possible to produce a pregnancy either by surgery or by harvesting of sperm [37]. Recent advances in reproductive technology have increased the chances of those previously considered to be infertile and may be of particular importance in ARM patients. Intracytoplasmic sperm injection allows sperm to be extracted from the ejaculate, epididymis or testis. A single sperm can then be injected into an ovum and allow patients with very low sperm counts and those who are not able to ejaculate the possibility of fatherhood. Reasons for this group to be infertile or subfertile include:

1. Associated genital anomalies such as maldescent of the testes.
2. Associated sacral anomalies.
3. Iatrogenic injury to nerves or genital structures during surgical repair.

Testicular maldescent is reported to be present in 3–19% [38,39]; furthermore, the higher the level of ARM the higher the rate of cryptorchidism. Cortes et al. found that rates of cryptorchidism approached 50% in boys with associated renal or ureteric anomalies [40]. The rate of infertility attributable to undescended testes cannot be accurately determined and paternity rates reported in the literature for undescended testes range greatly [41].

In a study of 20 ARM patients investigated for infertility following an anorectal pull-through procedure it was found that iatrogenic injury accounted for at least half of the cases [36]. Other reported causes included recurrent epididymo-orchiditis [42] as a consequence of urine refluxing into the vas deferens [43]. Such urinary reflux may be idiopathic or a consequence of an associated vasal anomaly, such as vasal ectopia to the bladder, large prostatic utricle or posterior urethral valves [44]. Iatrogenic injury may also be implicated in urinary reflux.

36.5 Conclusions

ARM have adverse implications for fertility and sexual function for a variety of reasons. Full information about the diagnosis and its implications must be made available to the family and should be explained clearly so that the patients, if old enough, and their parents can fully appreciate the extent of the future problem. When dealing with adolescents and young adults with ARM it is important that issues regarding sexual function and fulfilment are addressed openly and without embarrassment. Many of these patients will have become used to visiting their paediatric surgeon or urologist where these adult problems are not fully addressed. It is important that they feel able to request help with issues of a more personal nature.

Another helpful resource that may be of particular help is the patient support group. These groups can provide useful additional information, often of a more practical nature; details of such organisations should be available in the clinics where these patients are seen. Another useful tool in aiding a patient's understanding of what are often quite difficult concepts is written literature, which should be up-to-date and written in clear easy-to-understand terms.

References

1. Peña A (1995) Anorectal malformations. Semin Pediatr Surg 4:35–47
2. Hendren WH (1986) Repair of cloacal anomalies: current techniques. J Pediatr Surg 21:1159–1176
3. Diseth TH, Emblem R (1996) Somatic function, mental health, and psychosocial adjustment of adolescents with anorectal anomalies. J Pediatr Surg 31:638–643
4. Ludman L, Spitz L, Kiely EM (1994) Social and emotional impact of faecal incontinence after surgery for anorectal abnormalities. Arch Dis Child 71:194–200
5. Bai Y, Yuan Z, Wang W, Zhao Y, Wang H, Wang W (2000) Quality of life for children with fecal incontinence after surgically corrected anorectal malformation. J Pediatr Surg 35:462–464
6. Sontag S (1977) Illness as metaphor. Doubleday, New York
7. Peña A (2000) Anorectal malformations: experience with the posterior sagittal approach. In: Mouriquand PDE (ed) Paediatric Surgery and Urology: Long-Term Outcomes., Penn: WB Saunders, Philadelphia, pp 376–386
8. Patankar JZ, Mali VP, Yashpal R, Neo GT, Prabhakaran K (2004) Anorectal malformation with congenital absence of vagina: a case report and review of the literature. Pediatr Surg Int 20:295–297
9. Hall R, Fleming S, Gysler M, McClorie GA (1985) The genital tract in female children with imperforate anus. Am J Obstet Gynecol 151:169–171
10. Metts JC, III, Kotkin L, Kasper S, Shyr Y, Adams MC, Brock JW III (1997) Genital malformations and coexistent urinary tract or spinal anomalies in patients with imperforate anus. J Urol 158:1298–1300
11. Warne S, Wilcox DT, Creighton S, Ransley PG (2003) Long-term gynecological outcome of patients with persistent cloaca. J Urol 170:1493–1496
12. Rintala R, Mildh L, Lindahl H (1992) Fecal continence and quality of life in adult patients with an operated low anorectal malformation. J Pediatr Surg 27:902–905
13. Rintala R, Mildh L, Lindahl H (1994) Fecal continence and quality of life in adult patients with an operated high or intermediate anorectal malformation. J Pediatr Surg 29:777–780
14. Laufer MR (2002) Congenital absence of the vagina: in search of the perfect solution. When, and by what technique, should a vagina be created? Curr Opin Obstet Gynecol 14:441–444
15. Davies MC, Creighton SM, Woodhouse CR (2005) The pitfalls of vaginal construction. BJU Int 95:1293–1298
16. Greenwell TJ, Venn SN, Creighton S, Leaver RB, Woodhouse CR (2003) Pregnancy after lower urinary tract reconstruction for congenital abnormalities. BJU Int 92:773–777
17. Heinonen PK, Kuismanen K, Ashorn R (2000) Assisted reproduction in women with uterine anomalies. Eur J Obstet Gynaecol Reprod Biol 89:181–184
18. Landau D, Mordechi J, Karplus M, Carmi R (1997) Inheritance of familial congenital isolated anorectal malformations: case report and review. Am J Med Genet 71:280–282
19. Boocock G R, Donnai D (1987) Anorectal malformation: familial aspects and associated anomalies. Arch Dis Child 62:576–579
20. Cozzi F, Wilkinson AW (1968) Familial incidence of congenital anorectal anomalies. Surgery 64:669–671
21. Stoll C, Alembik Y, Dott B, Roth MP (1996) Evaluation of prenatal diagnosis of congenital gastro-intestinal atresias. Eur J Epidemiol 12:611–616
22. Schober JM (1999) Feminising genitoplasty for intersex. In: Stringer MD, Oldham KT, Mouriquand PDE, Howard ER (eds) Paediatric Surgery and Urology: Long Term Outcomes, 1st edn. W.B. Saunders, London, pp 549–558
23. Costa EM, Mendonca BB, Inacio M, Arnhold IJ, Silva FA, Lodovici O (1997) Management of ambiguous genitalia in pseudohermaphrodites: new perspectives on vaginal dilatation. Fertil Steril 67:229–232
24. Krege S, Walz KH, Hauffa BP, Körner I, Rübben H (2000) Long term follow up of female patients with congenital adrenal hyperplasia from 21-hydroxylase deficiency, with special emphasis on the results of vaginoplasty. BJU Int 86:253–259

25. Bailez MM, Gearhart JP, Migeon CG, Rock JA (1992) Vaginal reconstruction after initial construction of the external genitalia in girls with salt wasting adrenal hyperplasia. J Urol 148:680–684
26. Hensle TW, Dean GE (1992) Vaginal replacement in children. J Urol 148:677–679
27. Hojsgaard A, Villadsen I (1995) McIndoe procedure for congenital vaginal agenesis: complications and results. Br J Plast Surg 48:97–102
28. Schober JM (1999) Quality of life studies in patients with ambiguous genitalia. World J Urol 17:249–252
29. Steiner E, Woernle F, Kuhn W, Beckmann K, Schmidt M, Pilch H, et al (2002) Carcinoma of the neovagina: case report and review of the literature. Gynecol Oncol 84:171–175
30. Hoekstra WJ, Scholtmeijer RJ, Molenaar JC, Schreeve RH, Schroeder FH (1983) Urogenital tract abnormalities associated with congenital anorectal anomalies. J Urol 130:962–963
31. Cho S, Moore SP, Fangman T (2001) One hundred three consecutive patients with anorectal malformations and their associated anomalies. Arch Pediatr Adolesc Med 155):587–591
32. Rintala R (1998) Anorectal malformations: an overview. In: Stringer MD, Mouriquand PDE, Oldham KT, Howard ER (eds) Pediatric Surgery and Urology: Long-term Outcomes, 1st edn. W.B. Saunders, London, pp 357–375
33. Woodhouse CRJ (2005) Myelomeningocele in young adults. BJU Int 95:223–230
34. Hassink EAM, Rieu PNMA, Brugman ATM, Festen C (1994) Quality of life after operatively corrected high anorectal malformation: a long term follow up study of patients aged 18 years and older. J Pediatr Surg 29:773–776
35. Iwai N, Yanagihara J, Tokiwa K, Deguchi E, Takahashi T (1988) Results of surgical correction of anorectal malformations. Ann Surg 207:219–222
36. Holt B, Pryor JP, Hendry WF (1995) Male infertility after surgery for imperforate anus. J Pediatr Surg 30:1677–1679
37. Pryor JP, Hendry WF (1991) Ejaculatory duct obstruction in subfertile males: analysis of 87 patients. Fertil Steril 56:725–730
38. Spouge D, Baird PA (1986) Imperforate anus in 700,000 consecutive liveborn infants. Am J Med Genet Suppl 2:151–161
39. Cortes D, Thorup JM, Nielsen OH, Beck BL (1995) Cryptorchidism in boys with imperforate anus. J Pediatr Surg 30:631–635
40. Bianchini MA, Fava G, Cortese MG, Vinardi S, Costantino S, Canavese F (2001) A rare anorectal malformation: a very large H-type fistula. Pediatr Surg Int 17:649–651
41. Woodhouse CR (2001) Prospects for fertility in patients born with genitourinary anomalies. J Urol 165:2354–2360
42. Shiraishi K, Takihara H (2004) Recurrent epididymo-orchitis caused by posterior urethral valve associated with imperforate anus. Int J Urol 11:58–60
43. Megalli M, Gursel E, Lattimer JK (1972) Reflux of urine into ejaculatory ducts as a cause of recurring epididymitis in children. J Urol 108:978–979
44. Oguzkurt P, Tanyel FC, Buyukpamukcu N (1998) Acute scrotum due to edidymo-orchitis associated with vasal anomalies in children with anorectal malformations. J Pediatr Surg 33:1834–1836

37 ARM: Aftercare and Impact from the Perspective of the Family

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37.1 Introduction

The birth of a child with an imperforate anus is not only a surgical challenge, but also a shocking event for a partnership and the whole family. Neither parents nor physicians, who have no regular contact with such cases, can imagine how this event (and the efforts for a definitive solution by pull-through surgery) will influence the social system around this handicapped individual. A diagnosis of anorectal anomaly (anorectal malformation, ARM) no longer means a death sentence, and over the last 6 decades, surgical treatment has became more and more successful, especially after the introduction of the current gold standard of posterior sagittal anorectoplasty (PSARP) [6]. For this reason, secondary issues have become more and more relevant. There are methods of improved follow-up [18,31] and coping with this special situation by both patients and families.

The view of a surgeon, of course, differs from that of a family. Unlike the surgeon, the families struggle with the daily care of the child after the pull-through surgery and live with the long-term consequences. Some surgeons even think that there are no further problems at all, if there are no surgical problems or complaints. An evaluation of the surgical results with the outcome of continence is usually performed only in their own hospital. Furthermore, the continence scores and their interpretation are quite divergent between different centres. Hopefully, the Krickenbeck standards [13] will help to rectify the current situation (Tables 37.1–37.3). On the other hand, unfortunately a lot of patients and parents tolerate major and minor disabilities or suppress the problems.

Table 37.1 Standards for diagnosis: International classification (Krickenbeck)

Major clinical groups	Rare/regional variants
Perineal (cutaneous) fistula	Pouch colon
Rectourethral fistula	Rectal atresia/stenosis
• prostatic	Rectovaginal fistula
• bulbar	H-fistula
Rectovesical fistula	Others
Vestibular fistula	
Cloaca	
No fistula	
Anal stenosis	

Table 37.2 International grouping (Krickenbeck) of surgical procedures for follow-up

Operative procedures	Perineal operation
	Anterior sagittal approach
	Sacroperineal procedure
	Posterior sagittal anorectoplasty (PSARP)
	Abdominosacroperineal pull-through
	Abdominoperineal pull-through
	Laparoscopically assisted pull-through
Associated conditions	Sacral anomalies
	Tethered cord

Table 37.3 International classification (Krickenbeck) for postoperative results

1. Voluntary bowel movements	Yes/no
Feeling of urge, capacity to verbalise, hold the bowel movement	
2. Soiling	Yes/no
Grade 1: Occasionally (once or twice per week)	
Grade 2: Every day, no social problem	
Grade 3: Constant, social problem	
3. Constipation	Yes/no
Grade 1: Manageable by changes in diet	
Grade 2: Requires laxatives	
Grade 3: Resistant to laxatives and diet	

In the last 2 decades, however, in several countries organisations of concerned people have been established (Table 37.4). For such rare conditions like ARM they have become an indispensable support and information network for the secondary issues that accompany this congenital and, in spite of successful surgery, chronic condition. Collaboration between parents and patients on one side and physicians on the other is a promising development that has made this article possible. This is the first time that a contribution from a self-help organisation has appeared in a standard textbook on anorectal malformations (ARM).

The German branch (Selbsthilfeorganisation für Menschen mit Anorektalfehlbildungen eV, SoMA eV) of support groups for people with ARM recently

Table 37.4 Eleven currently known support groups for anorectal malformations (in 2005)

Country	Name	Established	Members	Contact
Norway	NFA: Norsk Forening for Analatresi	1981	300	http://www.analatresi.no e-mail: have a look on website
Chile	Asociacion Nacional de ninos con malformaciones anorectales de Chile	1986	450	no website available e-mail: marioantoniovarela@vtr.net
USA	PTN: Pull-thru Network	1988	700	http://www.pullthrough.org e-mail: info@pullthrough.org
Germany	SoMA: Self-help-organisation for people with anorectal malformations	1989	500	http://www.soma-ev.de e-mail: info@soma-ev.de
Finland	AH-POTILAAT-RY: Finnish association for Anus atresia and Hirschsbrung disease patients	1991	100	http://www.ah-potilaat.org e-mail: info@ah-potilaat.org
The Netherlands	Dutch Association "Vereniging Anusatresie"	1993	350	http://www.anusatresie.nl e-mail: info@anusatresie.nl
Italy	AIMAR: Associazione Italiana per le Malformazioni AnoRettali	1994	500	http://www.romacivica.net/aimar/ e-mail: dalia.aminoff@agcm.it
Taiwan	Imperforate Anus Family Association	1994	200	http://www2.mmh.org.tw e-mail: mmhss@ms2.mmh.org.tw
Israel	Israelian Parents Organisation	1996	50	http://www.itmut.info e-mail: phili@itmut.info
Portugal	APMAR: Associacao Portuguesa de Malformacoes Anorectais e Patologias Associadas	2003	?	http://www.apamar.org e-mail: info@apamar.org
Australia	PCAA: Paediatric Continence Association of Australia	2004	200	http://www.pcaa.org.au

performed an exhaustive survey of member families. Some of the results of this ground-breaking research will be presented in this article. The focus of this contribution is on the application of non-surgical methods, contentment and continence function according to the Krickenbeck criteria and the impact on families as seen by parents.

Collaborative international studies will become possible when cooperation between professionals and support groups is improved further. A visionary goal is a standardised follow-up chart and regular registration of each individual in their country-specific network (Table 37.4).

37.2 Materials and Methods

This investigation was approved by an independent ethics committee at the Charité – University Medicine in Berlin. The study was conducted between the autumn of 2004 and spring 2005. It was a postal survey addressed to member parents involving a standardised questionnaire (total 23 pages). Parents were included if their children were between 3 and 17 years of age. The intention was to collect epidemiological data regarding type of ARM, type of post-surgical therapy, degree of contentment, persisting needs, psychiatric screening, quality of life (QOL), faecal continence and family impact, using descriptive statistics.

The final sample consisted of 104 (28%) polled families who are members of the parent network from all over Germany. Of course this is a selected sample of very committed parents, with one in three participating couples having an academic degree.

Evaluation of classification, aftercare methods and degree of contentment was carried out in collaboration with the Italian association for ARM (Associazione Italiana per le Malformazioni AnoRettali). The results of the Italian questionnaire were based on a sample size of 209 (with no age limits), which is about double our sample [1].

QOL and continence were measured using a paediatric disease-specific instrument, the Hirschsprung's disease/ARM quality-of-life instrument (HAQL) developed by Hanneman et al. [8]. We translated the HAQL into German according to the guidelines. The HAQL and a five-item index described by Ditesheim and Templeton [5] are the only QOL scores assessing faecal incontinence in children [29]. The strong relationship between incontinence and QOL was demonstrated by Rothbarth et al. [24] on two other scales in 32 adult patients. Hanneman et al. [8] evaluated the HAQL on 534 patients aged 6–17 years. Whereas we only used the parent version of the HAQL (initially only for families with 6- to 11-year-olds) consisting of 46 items with 12 additional items for children with stomas, this questionnaire covered the following domains: laxative and constipating diet (each two items), diarrhoea (two items), constipation (one item), faecal continence (eight items), urinary continence (four items), social functioning (three items), emotional functioning (six items), body image (two items) and physical symptoms (nine items). We transformed the answers into two of three qualitative outcome domains according to the Krickenbeck agreement: soiling and constipation (see Table 37.3).

To evaluate the impact on the family with a chronically ill child, the Anglo-American impact-on-family scale (IFS) [27] was originally developed with 33 items ordered in 4 levels. In 2001, Ravens-Sieberer et al. [22] validated a German version of this self-report scale on 273 families with a child affected by different chronic conditions, such as epilepsy, diabetes and neurological disabilities from mild psychomotor problems to severe mental retardation. We used this so-called FaBel (Familien-Belastung-Fragebogen) to estimate the impact of the specific disease (ARM) on the family in several areas. We stratified the score for each scale into four categories: "better" if the result was less then one standard deviation (SD) below the mean of the reference population (< mean – 1×SD) and "worse" or "much worse" if the score was raised more than one or two ("much worse") standard deviations (> mean + 1×SD or > mean + 2×SD, respectively), respectively, above the reference population of chronic disabled children. The results of our family study can thus be compared with a reference population (i.e. families with different chronic conditions). Five scales could be separated: financial burden and job problems (4 items), daily social impact, partnership, leisure time (15 items), personal strain, worries regarding the future (5 items), coping problems of the parents (3 items) and concerns regarding siblings (6 items). The first four scales built up the total impact score (27 items). This questionnaire is available in four languages, English, German, Spanish and Italian.

37.3 Results

37.3.1 Demographics and Classification

The sample consisted of 104 responses from parents of the German self-help organisation SoMA eV. In 78% of cases the mother completed the questionnaire, in 7% the father and in 15% both parents. In 19% of

Table 37.5 Methods of aftercare. Please note that this table does not include surgical follow-up and other professional issues (see Table 37.6)

Enemas	Diapers	51%	Alternative	Anal tampons	6%
	Enemas (rectal)	51%		Homeopathy	22%
	Microenemas (klysma)	18%		Osteopathy	9%
	Enemas (Malone)	3%		Acupuncture	1%
	Suppository	17%		Reflex zone therapy	6%
Medication	Laxative medication	19%		Something different	23%
	Constipating medication	11%		Physiotherapy	21%
	Bladder medication	18%		Diet	16%
	Other medication	7%		Nothing	7%
	Bladder catheter	18%		Biofeedback	6%

cases only one parent was living together with the child. About 30% of the families had no other children, while 42% have one further child. One-third of the parents have a tertiary academic degree.

The age range of the affected child was 3–17 years, with a median of 7 years (mean: 8 years). Most cases were aged between 5 (25th percentile) and 11 years old (75th percentile) and 60% were male.

For classification regarding the ARM we used the descriptive Wingspread classification [28] as well as the fistula classification [17]. The type of fistula was not known by the parent in 18% of cases. About 19% had a rectobulbar fistula and 20% a rectovesical or a bladder-neck fistula. A perineal fistula was reported in 13%. There was no fistula in 8%. In 7 out of 12 girls with a vaginal fistula there was no recognised cloacal malformation, although it was likely to be an unrecognised vestibular fistula (4 cases) or cloacal malformation (3 cases). Therefore, depending on the correct diagnosis, 8–12% of participants had a vestibular fistula and 16–19% had a cloacal anomaly.

About 5% had a different diagnosis from ARM, such as Hirschsprung's disease, or the diagnosis was unknown, and 5% could not define the level of the ARM according to Wingspread. According to the Wingspread classification, 17% had low ARM, 11% had an intermediate ARM and 46% a high ARM [28].

The data from Italy were quite similar, but only 8% had a rectovesical fistula, whereas in 18% no fistula was diagnosed. We also assessed in detail the associated malformations and, for example, VATER (Vertebral, Anal, Tracheo-Esophageal, Renal anomalies) or VACTERL (Vertebral, Anal, Cardiac, Tracheo-Esophageal, Renal and Limb anomalies) malformations were present in 24%.

37.3.2 Methods of Aftercare

The possible methods of aftercare available are given in Table 37.5. Only 25% of the families received regular follow-up, with 15% believing no aftercare was required; 60% did not receive any aftercare! The result of the received aftercare was judged to be good in 70% of cases, moderate in most other cases (27%). Two-thirds of the families perceived a great need for more follow-up treatment. Diapers and enemas are the most frequently applied methods (51%), and enema management has increased considerably in recent years. The 51% usage of diapers shows that treatment aiming for so-called social continence was inadequate and may need to be improved. The rates were lower only in patients with low ARM (28%) or perineal fistula (17%). In cases of vesical and bladder-neck fistula (62%) and vaginal fistula (83%) the diaper and enema rate was higher.

37.3.3 Degree of Contentment

The degree of contentment of the parents (Table 37.6) depended on one hand on the final outcome, but also on the way the surgeon dealt with and informed the parents and patients. Some parents reported that the surgeon was ignorant regarding basic maternal instincts like breast feeding, which was especially important to these families. In 66% the parents received no advice from nutritional specialists, although it is well known how important this issue is, especially in colonic diseases.

Another important issue was the communication of prognosis and diagnosis, which predetermined the expectations in the first place and therefore the later

Table 37.6 Degree of satisfaction with aftercare by different specialists

Follow-up by specialist	Not offered	Not satisfied	Satisfied
Surgeon/clinic performing pull-through	10%	32%	58%
Other surgeon	24%	17%	59%
Urologist	46%	15%	39%
Paediatrician	10%	25%	65%
Nursing staff	39%	12%	49%
Physiotherapist	44%	6%	50%
Ergotherapist	69%	3%	28%
Psychological support	83%	7%	10%
Dietician	67%	12%	21%
Stoma therapist	50%	6%	44%
Social worker	92%	4%	4%
Alternative medicine	55%	7%	38%
Other parents	21%	4%	75%

degree of contentment. That is why we asked the parents if and how they were informed about the malformation and its consequences. Almost half of the parents received no information (4%) or insufficient information (42%) about the malformation prior to surgery. Regarding the information about functional prognosis, 63% of the respondents answered this question by saying that they felt that they were either not informed (27%) or insufficiently informed (39%).

It was also necessary to consider the psychological strain on the parents in the hospital. It is worse when the surgeon could not imagine what huge psychological strain the initial weeks in hospital imposed upon the parents. In the long term only very few families (17%) were offered psychological support.

In more than 40% of the cases follow-up was either not offered by the attending surgeon or the follow-up supplied was not satisfactory. That level is too high and can be improved upon. Given that the surgery could also be a financial burden, the rate of referral to a social worker was too low. The initial attending surgeon should have in mind all the ancilliary services (see Table 37.6) and at least have proposed their use, as the information of further supporting facilities improved the degree of contentment of the families.

Concerning how valuable the support given by other parents is, it is surprising that 21% of the parents were not offered any support by other families. For example, the parents were not informed about self-help groups. Our members confirmed that even when they asked, they did not receive any information about how to contact us or other parents groups despite the high level of contentment demonstrating the importance of this kind of support.

37.3.4 Continence Function (HAQL)

In the outcome classification of Krickenbeck, three domains are separately evaluated: voluntary bowel movement, soiling and constipation. With the data of our survey we obtain from the simplified definition of "voluntary bowel movement" according krickenbeck-agreement (Table 37.3), because we see inconsistency in this item. But ist was possible to operationalise the soiling domain with data from the HAQL. The constipation domain of the new Krickenbeck classification works with several assumptions. For instance, that the constipation is treated and that the order of treatment is observed (diet→medication→irrigation). However, some parents perform colonic irrigation as preventive therapy as well as to manage soiling. This has to be considered, if judgements with this assessment method are made.

37.3.4.1 Soiling

Twenty-two percent of parents reported no soiling, but 48% (grade 1) experienced occasional soiling. Of the 30% of parents whose children had daily soiling, 9% reported social problems (grade 3) and 21% (grade 2) did not have major social problems.

Considering the degree of satisfaction with the treatment by the primary surgeon, we examined whether there was any relationship between continence results and parental satisfaction. Regarding constipation, the majority (80%) were satisfied in cases of no constipation or grade 1. However, in grades 2 and 3 the satisfaction with the first-line surgeon fell to 60%. High contentment was present when there was no soiling (80%), but the soiling grade (grade 1 and 3) were equally distributed (50%) between satisfied and unsatisfied parents. Even the parents in grade 2 with soiling every day but without social problems showed a high rate of contentment (83%). In conclusion, contentment with the primary surgeon depended only in part on the continence outcome.

These continence results have to be put in the context of the type of malformation, as even 80% of perineal fistula children had constipation. The majority of these were treated by dietary changes or laxatives. In children with perineal fistula the outcome for soiling was divided equally between none, occasional or daily soiling. With rectobulbar fistula, most cases needed enemas (grade 3) or at least laxatives (grade 2) because of constipation. Occasional or daily soiling was equally named without major social impact. The cases of so-called vaginal fistula, as well as vestibular fistula and rectovesical fistula usually needed enemas for constipation, and occasional soiling occurred.

In 43% of children with high ARM, bowel action frequency was more than four times a day on two or more days each week. This feature should not be classified as "diarrhoea" (39% of high ARM with thin stool at least occasionally). The risk for pseudo-incontinence has to be considered, especially in this specific type of malformation.

Table 37.7 Social, financial and psychological impact on the family

Impact-on-family scale (items)	Better	Similar*	Worse
Financial burden (4)	46%	33%	21%
Daily social impact (15)	15%	72%	13%
Personal strain (5)	5%	79%	16%
Parental coping problems (3)	12%	71%	17%
Concerns regarding siblings (6)	31%	52%	17%
Total impact score (27)	7%	79%	14%

*Mean reference population ± one SD

37.3.4.2 Constipation

Twenty-four percent of parents thought that there was no constipation, but 8% (or one-third) of them still perform enemas; 6% managed the constipation with diet (grade 1) and 19% with laxative medication (grade 2). The vast majority (41%) used different kinds of enemas (grade 3) to deal with the constipation. In 10% the constipation was not treated in any way.

37.3.5 Impact on the Family

In general, the total impact on the family was similar to that for other chronic conditions (Table 37.7). Only 7% had a significantly better and 14% a significantly worse total score (> mean + 1×SD). The financial impact was diverse. Half of the families perceived less financial burden than families of children with other chronic conditions, although there was a significantly worse subgroup (> mean + 2×SD) of 6%.

Looking at single items of the impact-on-family scale, some results were of specific interest: For instance, 66% had serious concerns regarding the future of the child (item number 26; 33% even strongly agreed). More than the half of the parents reported that travelling to hospitals strained them (item number 24) or that they felt like they were living on a roller coaster (item number 27). About 50% did not want further children because of the ill child (item number 15). Thirty percent of families had to give things up because of the child's illness (item number 20), but the vast majority (95%) still tried to treat the affected child as normal (item number 17). In complex cases in particular there needed to be support for coping with the situation, as 66% of parents of cloacal cases were worried about the future of their child.

37.4 Discussion

Failure of the initial pull-through surgery for ARM due to preventable reasons is now unacceptable. For instance, Peña et al. [19] and Moss [16] report cases where the neoanus was outside of the muscle complex, or where more complex situations (cloaca) were not recognised during the initial treatment. During the primary treatment, even the correct location of the stoma has to be considered. Therefore it is appropriate that parents have to be well informed and ask questions about receiving adequate surgery by an experienced surgeon. New or old surgical strategies with major modifications compared to the gold standard should only be evaluated under scientific circumstances (i.e. controlled and registered trials).

According to the EUROCAT group [4], the incidence of ARM in Germany ranges between 0.02% (Saxony-Anhalt) and 0.05% (Mainz). Only 30% of the ARM are isolated and not associated with other malformations. For instance, the prevalence of VATER and VACTERL of 24% in our group is similar to the 15% overall rate of VACTERL reported in the EUROCAT study, although they reported only 5% with cloacas, which is in contrast to our group (15%). We know of several cases were the diagnosis of cloaca was not made by the first-line surgeon, with serious results for the care of the child. Particularly complex cases like cloaca, which can sometimes be detected in prenatal ultrasound screening, should only be handled in specialised and experienced centres and not by every surgeon. In these cases social workers should be involved regularly because at least 60–70% of these cases later receive disability status. In this context we refer to our special subgroup analysis of 28 cloacal cases, which was presented at the 43rd Meeting of the German Society for Pediatric Surgery in Bremen this year [25].

One issue that the recent Krickenbeck conference tried to tackle is the lack of commonly accepted standards for classification [21] procedures and evaluation of outcome. Outcome results in particular, such as continence scores and QOL instruments, differ and are used divergingly [12]. Not only are the samples biased because of incomplete cases or results from only one centre, but also the judgement of "good" or "fair" results differs greatly and is often only evaluated by the physician but not by the patient or legal guardian, which would be more appropriate. The conclusions are therefore of limited value.

Regarding our results, further questions could be addressed. The HAQL as a disease-specific questionnaire reports many more details than are used in pragmatic, new standards for continence evaluation [13]. Of course faecal continence is a pivotal point for QOL. Therefore the patients need to be treated carefully, including life-long follow-up. There are other issues, however, that can be optimised. Interestingly, two publications using scales that were applied in our study (FaBel: [9]) raise the question: "Does healthcare meet the needs?" Satisfying needs for access to medical healthcare services and adequate surgery could be achieved; however, more psychosocial and paramedical care should be offered according to Hartman and colleagues [9]. Thyen et al. suggested that the family burden is a direct measure for unmet healthcare needs [30].

According to Trajanovska and Catto-Smith [29] there are only two QOL indices that also assess faecal incontinence. They missed in their review the QOL scale of Bai et al. [2], where the authors concluded that "somatic and psychological care and long-term follow-up are necessary to improve the quality of life". The strong relationship between QOL and continence function is already conditioned through the construction of the QOL scales, where continence function is a major domain in the instrument. Therefore the other domains (social acceptance or psychological problems) should be viewed as separate.

Just recently Funakosi et al. [7] pointed out, that half (6 out of 11) of the children with ARM in the 12- to 16-year age group suffered from significant depression. The 7- to 12-year age group with ARM were not so badly affected. Similarly, Pfeiffer et al. [20] explored by self-report 30 children with ARM (aged 10–16 years) and reported that recurrent, long hospitalisations alter personality development towards internalising disorders and low self-esteem. These self-reported results differ in some way from parental views analysed by Ludman and Spitz [14]. They analysed specific coping strategies and reasoned that with adolescents, covert denial or eventual acceptance of the disability continues, but in general the children are able to adapt themselves to society.

It is encouraging that already in 1983, Martinius [15] had considered the prevention of psychological disturbances in children with ARM. He stated that in 4- to 5-year-olds with anal atresia there were symptoms related to detachment anxiety, bonding, appetite and continence. Nowadays we should revise his conclusion that psychologically caused incontinence (encopresis) should be considered in cases where sphincter functioning can be proven. Today it is known that the physiological function of continence and rectal reservoir is a complex interaction that is not only defined by external sphincter function, but also by other muscles (levator), receptor density and innervation. At that time patients were treated by methods other than PSARP, for instance the Rehbein procedure, which itself caused a high level of incontinence. Furthermore, the concept of pseudo-incontinence should be considered in such cases.

It should be realised that dirty underwear is not only a warning sign, but also a burden. Occasional soiling may be a satisfying outcome for a malformation with a poor prognosis; nevertheless, it can be still a stressful experience for the affected person.

The expected benefit of biofeedback on children with defecation disorders is not scientifically founded, according to the Cochrane review by Brazelli and Griffith [3]. In accordance with these results, parents only rarely use this method, although it is recommended by some specialists. By contrast, according to the Cochrane review of 16 randomised trials with a total of 843 children, the benefit achieved by the combination of laxatives and behavioural interventions (e.g. toilet training) was scientifically proven. Standardised care after surgery with primarily non-invasive methods could be better established in the view of parents.

The tendency of some surgical experts to aim for continence through "experimental" surgery is not appreciated by most parents, because of repeated experiences with adverse outcomes. Re-do surgery for patients with injuries caused by accidents should be separate from the necessities of our patients with ARM because basic anatomy and requirements are not comparable. In addition, bowel control is not only a physical issue but also a question of strategy and psychology. This area of expertise differs from the surgical approach.

Owing to a lack of professional support and no standard guidelines, adults with persisting incontinence have to sort out through self study ways to deal with the continence problems. We support Hassink et al. [10] in calling for a stepwise protocol of procedures for earlier and more efficient ways of bowel control. Of course referral to self-help groups for adults should be a standard opportunity for everyone interested. There are some other further problems, like sexuality, which were not examined in our study but are still important. The care for adolescents and adults is more self-determined, and long term follow-up could be improved through routine cooperation between professionals and associations of affected people.

Hassink et al. [11] has already stated that "parents play a crucial role in the life of a child suffering from an anorectal malformation (ARM), since their guidance contributes to the degree to which the child learns to cope with his or her disability." The question can also be raised: Who guides the parents? In order to remove this deficiency, parents are considering the welfare of their children in parental networks.

37.4.1 A Call for Further Collaboration

Regrettably at the moment, it is quite exceptional if a paediatric surgeon informs the family of an affected child about the existence of self-help groups (see Table 37.4). But in our view it is a sign of a confident, competent and supportive surgeon, if he/she does so. This advisory information should just become a standard, with information on social concerns, possible psychological support or the exact description of the situation (exact classification of the deformity, treatment and continence function) given in the physician's letter. In ARM this self-evident information should be communicated regularly. Whether the family then becomes a member of a self-help group is an independent decision that every family has to make on its own. Such a communication could obviate an arduous and long search by many families for fellow sufferers. We have learned that even professionals in ARM surgery denied the existence of such initiatives against better knowledge. Why does this anxiety and prejudice exist? Substantial thought patterns are:

1. "My patients do not need aftercare." Unfortunately, this conceit prevails very often, that the surgeon's own patients require no self-help group, because they are already surgically optimally supported. At the same time it is common to forget that the surgical solution is a central, but not the sole factor in the treatment of congenital deformities.
2. "What can self-help groups do?" In many cases the surgeon has no idea about the sequelae of the malformation or the work of self-help groups and is afraid of the unknown. Both are simply a sign of ignorance.
3. "Only problematic cases are in self-help groups." Sometimes the surgeon fears that parents are negatively influenced by individual hard cases in self-help groups. Even if this were true, it would be especially desirable to increase the number of successful and satisfied patients in self-help groups. After all, a physician should not underestimate the capacity of the majority of his families.
4. "Self-help groups send my patients to another doctor." This statement assumes that self-help groups would direct patient currents. It is possible that this may occur through negative experience in individual cases, if certain discontented parents ask around for advice by other parents. However, this is always a subjective experience and never the official view of a self-help group. A self-help group or its members would never recommend a single physician, because this is too great a responsibility. The surgeon may think that parents have to take the decision by themselves as it is the life of their children. This impression emerges when the surgeon does not perceive the sorrow of the family and therefore the appropriate supportive work of self-help groups. The power of self-help groups is overestimated; only the quality of the treatment is decisive.

Wrong personal estimation and lack of knowledge about self-help groups by the surgeon directly affect the welfare of patients. Out of this consideration about supposed prejudices, the lack of information and cooperation with concerned families can itself be appraised as a massive quality deficit. Our central wish as a self-help organisation is not competition, but synergy for the welfare of the affected children.

37.4.1.1 Benefit for the Surgeon

Self-help groups are not only advantageous for the patient and their families, but also for the surgeon. It is really disappointing for a family when they finally discover after years of loneliness and troubles that there are others with similar problems. Then the parents question themselves: Didn't my surgeon know about the existence of these groups, which means that he was not at the state of the art? Or what was the reason? If surgeons are open to further support possibilities, they show that they are not narrow minded. A multidisciplinary approach proves that there is nothing to be hidden.

We have found that even between surgeons working in different places there may exist some hostility, which is not good for patients. Some knowledge is not shared. For instance, although we know the continence results for individual centres, every time it has been evaluated slightly differently. A scientific comparison to determine the best treatment is impossible without a common basis for discussion. A parent organisation may provide a natural exchange forum.

The number of treated patients is not generally a sign of the quality of a surgeon. Sadly, some families must suffer very experimental surgery with disastrous results. Even the best surgery with insufficient follow-up, could lead to catastrophic outcomes, with pseudo-incontinence [18,23]. Feedback from other cases through the parent organisations could improve the desire for a thorough follow-up, which would increase the patient compliance and overall experience of the physician.

Another advantage for the surgeon is to learn specific solutions for a rare disease. For instance, new treatment forms have been introduced by parent organisations. The enhanced bowel management programme, including not only medication but also methods like systematic colonic flushing, were first introduced into Germany by parents! We could not find any physician who would introduce it to us, so it was necessary to invite experts from other countries. Happily, nowadays a few centres already offer this more holistic follow-up.

We are grateful to surgeons if they give patient-focussed advice at our congresses or in our newsletters. Of course this can work in some way as advertisement, but we prefer local experts. By contrast, it is a major misbelief of some cooperative physicians to assume that a self-help group will and can control patients and will support only one doctor. Contentment, quality and exhaustive information are the main factors that influence patients.

Just recently, during the XIIth Colorectal Club meeting in Dublin [26], specialised paediatric surgeons emphasised that the benefit for both parties can be improved through intensified communication between patients and physicians in these self-help organisations. In this context it should not be forgotten that some of the self-help organisations were established by committed surgeons. Perhaps similar people can be identified for those countries without support groups?

37.4.1.2 Benefit for the Patient

It is reassuring when patients and parents come into contact with other families and no longer feel alone in the world with their problem. Children and adults with the same handicap can share intimate concerns, which could never be shared with uninvolved people in the same way. Since ARM is a rare condition, individual solutions are often necessary and only in such groups do concerned parents or patients meet so many different individuals!

The accumulation of different experiences brings a family's subjective one-sidedness into a more global perspective. To share often very simple tips and quirks with associates cannot be overestimated. It can be facilitating little things like dragging the catheter or modifying the daily routine. In the hurry of a hospital these things cannot be clarified. This information and education results in better aftercare for faecal continence and QOL, with finally a better outcome and greater degree of contentment.

A parent and patient organisation is a reservoir of lively information regarding all issues that could occur in the life of a child who is born with an imperforate anus.

37.5 Conclusion

Living with an ARM means for patients and their families not only bearing the hardships of one to three operations, but also the situation of patients with ARM is not satisfying concerning the diagnosis, the treatment and the aftercare. Based on the results of our surveys, study of the literature and our experience gathered from many consulting sessions, we conclude that the care of ARM should be in the hands of experienced specialists. Today there is a lack of information regarding the diagnosis and prognosis for patients. A wrong diagnosis can result in incorrect surgery. Therefore patients should have the possibility of a second opinion and the possibility to share experiences. Furthermore, a wrong prognosis can awaken false hope and this could result in bad, inappropriate or no aftercare and, at worst, in unnecessary operations.

There is a need for standard guidelines and their monitoring, which is also required for aftercare. The situation of faecal continence after ARM has to be considered in a very dedicated way. Patients with ARM and their families need support! They need qualified help to be continent or to reach social continence. Surgery has to look at the long-term consequences, if it is to be successful. Collaboration of all concerned disciplines is mandatory.

The impact of ARM on affected families is comparable to that of other chronic conditions. The entire family needs support to cope with the situation with the help of social workers, psychologists and similar specialists. Surgeons should supply the basic information about the physical problems. ARM require cautious handling, since they concern several highly intimate areas. This creates trust and prevents additional psychological stress on the patient.

We believe that surgeons should work together with self-help organisations and they should give patients and parents the opportunity to contact other fellow sufferers so that they do not feel alone with their disability. The authority and power of the treating physician has to be applied in a responsible way for the sake of our children, as it decides the child's and the family's quality of life – today and in the future.

References

1. Aminoff D, la Sala E (2004) Parents' perception on the care and follow-up received by health caregivers. Lecture given at the XIth International Meeting of the Pediatric Colorectal Club, New York, USA, May 22–23, 2004
2. Bai Y, Yuan Z, Wang W, Zhao Y, Wang H, Wang W (2000) Quality of life for children with fecal incontinence after surgically corrected anorectal malformation. J Pediatr Surg 35:462–464
3. Brazzelli M, Griffiths P (2001) Behavioural and cognitive interventions with or without other treatments for defecation disorders in children. The Cochrane Database of Systematic Reviews 2001, Issue 4. Art. No.: CD002240. DOI: 10.1002/14651858.CD002240
4. Cuschieri A, EUROCAT Working Group (2001) Descriptive epidemiology of isolated anal anomalies: a survey of 4.6 million births in Europe. Am J Med Genet 103:207–215
5. Ditesheim JA, Templeton JM Jr (1987) Short-term v long-term quality of life in children following repair of high imperforate anus. J Pediatr Surg 22:581–587
6. de Vries PA, Peña A (1982) Posterior sagittal anorectoplasty. J Pediatr Surg 17:638–643
7. Funakosi S, Hayashi J, Kamiyama T, Ueno T, Ishii T, Wada M, Amae S, Yoshida S, Hayashi Y, Matsuoka H (2005) Psychosocial liaison-consultation for the children who have undergone repair of imperforate anus and Hirschsprung disease. J Pediatr Surg 40:1156–1162
8. Hanneman MJ, Sprangers MA, De Mik EL, Ernest van Heurn LW, De Langen ZJ, Looyaard N, Madern GC, Rieu PN, van der Zee DC, van Silfhout M, Aronson DC (2001) Quality of life in patients with anorectal malformation or Hirschsprung's disease: development of a disease-specific questionnaire. Dis Colon Rectum 44:1650–1660
9. Hartman EE, Sprangers MA, Visser MR, Oort FJ, Hanneman MJ, van Heurn LW, de Langen ZJ, Madern GC, Rieu PN, van der Zee DC, Looyaard N, Aronson DC (2005) Anorectal malformations: does healthcare meet the needs? J Pediatr Gastroenterol Nutr 41:210–215
10. Hassink EA, Rieu PN, Severijnen RS, Brugman-Boezeman AT, Festen C (1996) Adults born with high anorectal atresia – how do they manage? Dis Colon Rectum 39:695–699
11. Hassink EA, Brugman-Boezeman AT, Robbroeck LM, Rieu PN, van Kuyk EM, Wels PM, Festen C (1998) Parenting children with anorectal malformations: implications and experiences. Pediatr Surg Int 13:377–383
12. Holschneider AM, Jesch NK, Stragholz E, Pformmer W (2002) Surgical methods for anorectal malformation from Rehbein to Peña – critical assessment of score systems and proposal for a new classification. Eur J Pediatr Surg 12:73–82
13. Holschneider A, Hutson JM, Peña A, et al (2005) Preliminary report on the International Conference for the development of standards for the treatment of anorectal malformations. J Pediatr Surg 40:1521–1526
14. Ludman L, Spitz L (1996) Coping strategies of children with faecal incontinence. J Pediatr Surg 31:563–567
15. Martinius J (1983) Bleibende psychische Störungen? Prophylaktische Möglichkeiten [Persistent mental–health problems? Prophylactic possibilities]. In: Hofmann-v.Kap-herr S (ed) Anorektale Fehlbildungen [Anorectal Malformation]. Gustav Fisher Verlag, Stuttgart, New York, pp 247–249
16. Moss RL (1998) The failed anoplasty: successful outcome after reoperative anoplasty and sigmoid resection. Pediatr Surg 33:1145–1147
17. Peña A (1990) Atlas of Surgical Management of Anorectal Malformations. Springer Verlag, New York
18. Peña A, Guardino K, Tovilla JM, Levitt MA, Rodriguez G, Torres R (1998) Bowel management for fecal incontinence in patients with anorectal malformations. J Pediatr Surg 33:133–137
19. Peña A, Hong AR, Midulla P, Levitt M (2003) Reoperative surgery for anorectal anomalies. Semin Pediatr Surg 12:118–123

20. Pfeiffer U, Bühligen U, Kleber B (2002) Persönlichkeitsentwicklung chronisch kranker Kinder am Beispiel Analatresie [Personality development of chronically sick children in the example of anal atresia]. 40. Jahrestagung der Deutschen Gesellschaft für Kinderchirurgie. Leipzig: 18.9.–21.09.2002. Published in Kinder und Jugendmedizin A119
21. Ratan SK, Rattan KN, Pandey RM, Mittal A, Magu S, Sodhi PK (2004) Associated congenital anomalies in patients with anorectal malformations – a need for developing a uniform practical approach. J Pediatr Surg 39:1706–1711
22. Ravens-Sieberer U, Morfeld M, Stein RE, Jessop DJ, Bullinger M, Thyen U (2001) Testung und Validierung der deutschen Version der "Impact on Family Scale" bei Familien mit behinderten Kindern [The testing and validation of the German version of the impact on family scale in families with children with disabilities]. Psychother Psychosom Med Psychol 51:384–393
23. Rintala RJ (2002) Fecal incontinence in anorectal malformations, neuropathy, and miscellaneous conditions. Semin Pediatr Surg 11:75–82
24. Rothbarth J, Bemelman WA, Meijerink WJ, Stiggelbout AM, Zwinderman AH, Buyze-Westerweel ME, Delemarre JB (2001) What is the impact of fecal incontinence on quality of life? Dis Colon Rectum 44:67–71
25. Schwarzer N, Jenetzky E, von Stralendorff L (2005) Folgen der Diagnose "Kloakale Fehlbildung" aus Elternsicht [Sequences of the diagnosis "Cloacal deformity" in parental perception]. 43 Jahrestagung der Deutschen Gesellschaft für Kinderchirurgie. Bremen: September 29–October 2, 2005
26. Schwarzer N (2005) XIIth International Meeting of the Pediatric Colorectal Club, Dublin, Ireland, July 10–11, 2005 in SoMA Aktiv, volume 10, pp 45–47
27. Stein RE, Riessman CK (1980) The development of an impact-on-family scale: preliminary findings. Med Care 18:465–472
28. Stephens FD, Smith ED (1986) Classification, identification, and assessment of surgical treatment of anorectal anomalies. Pediatr Surg Int 1:200–205
29. Trajanovska M, Catto-Smith AG (2005) Quality of life measures for fecal incontinence and their use in children. J Gastroenterol Hepatol 20:919–928
30. Thyen U, Sperner J, Morfeld M, Meyer C, Ravens-Sieberer U (2003) Unmet health care needs and impact on families with children with disabilities in Germany. Ambul Pediatr 3:74–81
31. van Kuyk EM, Brugman-Boezeman AT, Wissink-Essink M, Severijnen RS, Festen C, Bleijenberg G (2000) Biopsychosocial treatment of defecation problems in children with anal atresia: a retrospective study. Pediatr Surg Int 16:317–321

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