Anorectal Malformations

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24.1 Introduction

Anorectal malformations (ARMs) comprise a spectrum of congenital defects with an incidence of 1:2500 that range in severity from mild anal stenosis alone to complex malformations involving the anorectum and urogenital tract. Both males and females are equally affected, and a detailed understanding of the anatomy of these defects is essential for the provision of optimal surgical care during the neonatal period. Improvements in neonatal and surgical care over the past decades have reduced mortality in ARM patients to around 3% in units with modern technology [1], and mortality is usually the result of uncorrectable associated anomalies rather than the ARM. Apart from cloacal abnormalities, prenatal suspicion of ARMs is uncommon, and the diagnosis is usually made shortly after birth. Because associated defects affect over half of patients, management in the neonatal period involves prompt screening based on a set algorithm. The modern operative management of ARMs involves anatomical reconstruction of the defect with minimal disturbance to the existing continence mechanisms. As ARMs represent relatively rare disorders, definitive repair should be performed in experienced tertiary units to ensure optimal healthcare outcomes.

24.2 Embryology of ARMs

In early embryology, the caudal region of the normal hindgut is called the cloaca. During the 7th week of gestation, cloacal division into ventral and dorsal components occurs with descent of the urorectal fold, forming the urogenital tract ventrally and the anorectal tract dorsally. The urorectal fold ultimately forms the perineal body between them. The dorsal part of the cloacal membrane (CM) ends at the position of the future anal opening at a fixed point near the tail groove in the normal rat model (Fig. 24.1) [2]. In ARMs, the critical factor for development may be an abnormally shortened cloacal membrane (Fig. 24.2) [2], which could lead to aberrant siting of the bowel termination during cloacal subdivision in ARMs, including urogenital connections. The site where the anal orifice should be, as marked by the location of the external sphincter apparatus in humans, is already established at a fixed point in the mouse model prior to cloacal subdivision [2]. In normal development, the cloacal membrane disintegrates where it meets the tip of the descending urorectal fold posteriorly, forming the anal ori-

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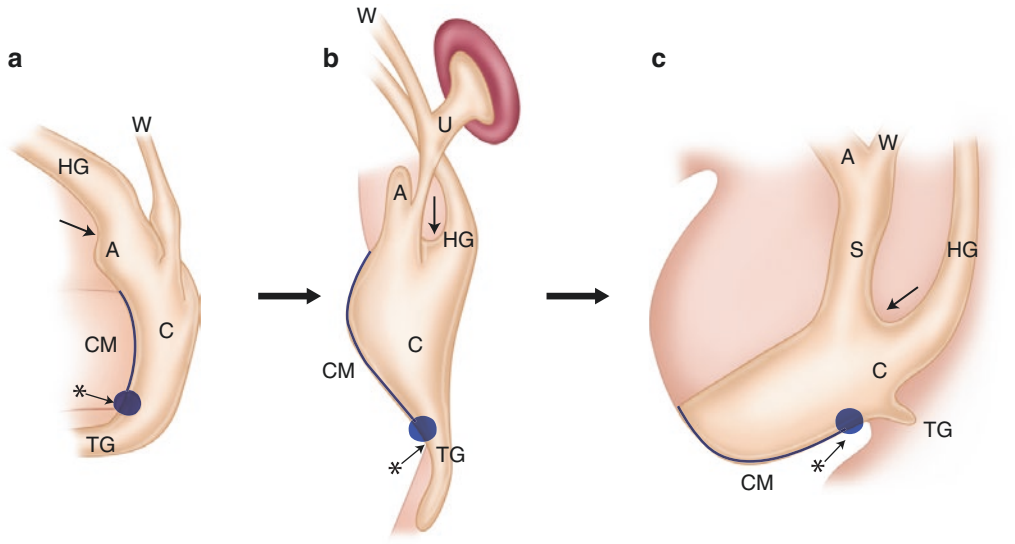

Fig. 24.1 Normal cloacal development in the rat model. Schematic diagram of a normal (**a** and **b**) and an abnormal (**c**) cloaca

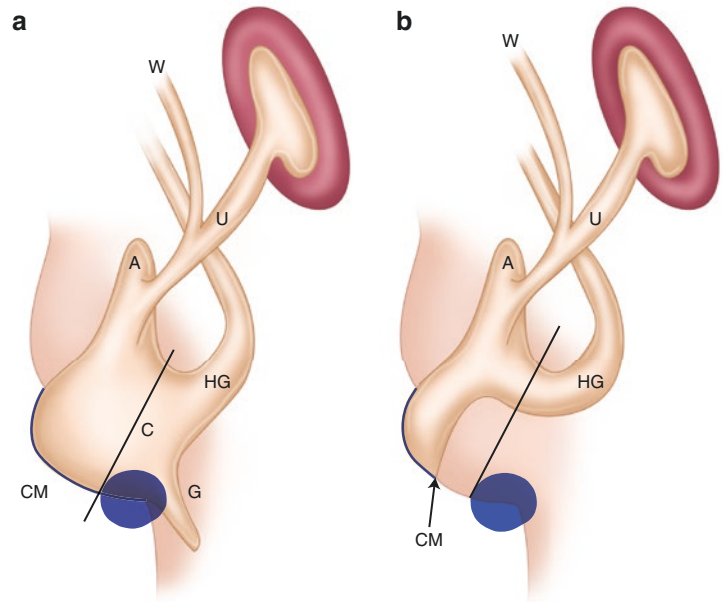

Fig. 24.2 Model of abnormal cloacal development. Schematic diagram of a normal (**a**) and an abnormal (**b**) cloaca. In the abnormal embryo, the cloacal membrane (CM) is too short (arrow). The cloacal membrane does not extend to the region of the tail groove (grey area). The dorsal cloaca is missing. In the normal embryo (**a**), the cloacal membrane is of normal length and extends to the region of the tail groove (grey area). Reproduced from Fig. 7 in Kluth [2]. Embryology of anorectal malformations. Semin Pediatr Surg 2010, 19: 201-208 and reproduced with kind permission from Elsevier

fice [3]. This initially closes with the ectoderm and is recanalized 2 weeks later. Abnormalities of recanalization of the anal orifice during the 9th week of gestation could assist to explain mild ARMs, including stenotic and membranous defects [3].

24.3 Etiologic Factors

The etiologic basis of ARMs is based on genetic and environmental influences. The most frequent chromosomal associations involve micro-

deletion of chromosome 22q11.2 (also known as DiGeorge or CATCH-22 syndrome) and Down syndrome (trisomy 21). A chromosomal abnormality is observed in approximately 10% of cases [3]. Townes-Brocks, Pallister-Hall, Opitz-Kaveggia, Johanson-Blizzard, Kaufman-McKusick, Lowe, oculo-auricular-vertebral (Goldenhar), fragile X and trisomy 8 syndromes are other reported associations [4]. A genetic aetiology is supported by a familial occurrence in up to 8% of cases [5] and among monozygotic twins. Autosomal dominant inheritance of mutations in *HLXB9* is responsible for 50% of patients with Currarino syndrome. Environmental risk factors for ARMs include prenatal exposure to caffeine, alcohol or drugs as well as maternal factors such as diabetes and epilepsy. Assisted reproductive techniques, primiparity, pre-eclampsia and maternal fever in early gestation have also been implicated [6].

24.3.1 Associated Anomalies

Malformations of the VACTERL (vertebral, anal, cardiac, tracheo-esophageal, renal, limb) and CHARGE (choanal atresia/coloboma, anal, renal, gastrointestinal and ear/hearing) sequence are recognized, non-random associations of ARMs. Over half of all patients with ARMs have at least one other associated congenital malformation, and these may affect over 90% of patients with severe ARMs. Approximately 10–15% fulfil the criteria for VACTERL association [6–8], having three or more anomalies from this sequence. A recent epidemiological survey identified a prevalence of 13% for cardiac defects (mainly atrial septal defect, ventricular septal defects and tetralogy of Fallot), 15% for skeletal defects, 10% for tracheo-oesophageal fistula, 25% for urologic abnormalities (most commonly vesicoureteric reflux, renal agenesis and dysplastic kidney), 13% for limb defects and 13% for genital anomalies [4, 7]. Uterine and vaginal abnormalities are common in cloaca patients and occur less frequently in females with milder types of ARMs. Additionally, craniofacial abnormalities including cleft palate are present in approximately 5% of patients.

24.3.2 Normal Anatomy of the Pelvic Floor

Knowledge of the normal anatomy of the pelvic floor and anal canal is necessary for understanding the pathologic anatomy of ARMs. The pelvic floor comprises a sheetlike diaphragm of striated muscles that support the pelvic organs and abdominal viscera. These muscles insert into the pubic bone anteriorly, the most inferior part of the sacrum posteriorly and the obturator membrane, ischium and ischial spine laterally. The ventromedial aspect forms a funnel-like sling around the urethra, vagina and anorectum, with fibres fusing medially at the perineal body and serving to close the urogenital and anorectal hiatuses by contraction [9]. The superior part of this muscle funnel is called the levator muscle, and its inferior fibres are continuous with the deep fibres of the external anal sphincter. Contraction of the puborectalis sling fibres of the levator complex maintains the anorectal angle (Fig. 24.3), preventing faecal descent during sudden increases in intra-abdominal pressure [10].

24.3.3 Anatomy of the Anal Canal

The voluntary or external anal sphincter (EAS) muscles of the anal canal are continuous superiorly with the fibres of the levator muscles, forming a striated funnel comprising of deep, superficial and subcutaneous components (Fig. 24.4). The EAS provides approximately 15% of resting continence but becomes activated during physical activities that increase the intra-abdominal pressure. It receives its motor and sensory supply from the inferior rectal branches of the pudendal nerve and the perineal branch of S4, which is also sensory to the skin of the anal canal to approximately 1 cm proximal to the dentate line [10]. The internal anal sphincter (IAS) fibres provide the remaining 80–85% of resting anal canal pressure and represent a thickened continuation of the inner smooth (visceral) muscle of the rectum [10]. The tone of the IAS is maintained by sympathetic pathways from the hypogastric plexus. The rectoanal inhibitory reflex of the IAS is principally intramural and

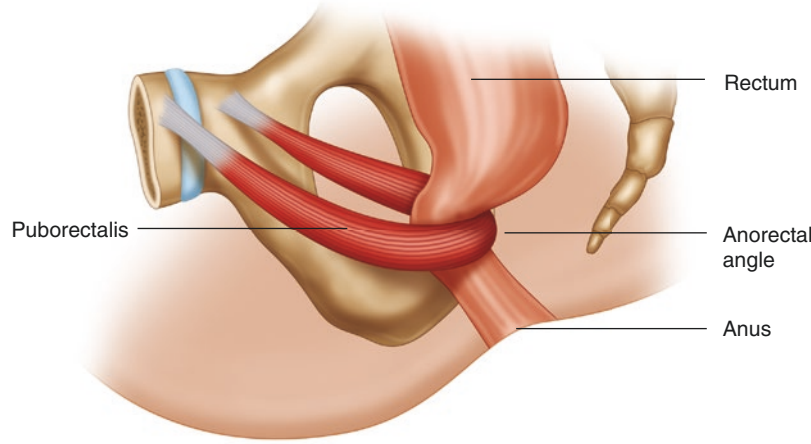

Fig. 24.3 The normal anorectal angle, formed by the puborectalis sling, which is part of the levator muscle complex

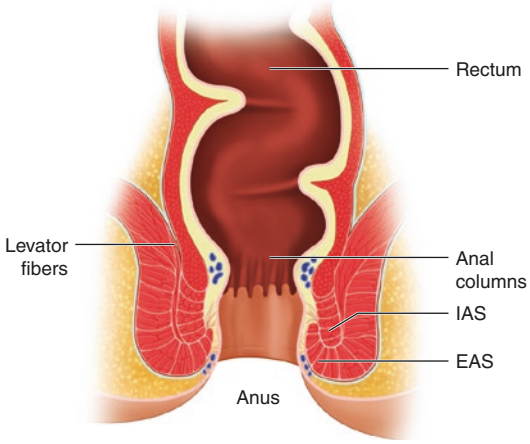

Fig. 24.4 Normal anatomy of the anal canal: the levator fibres are continuous with the deep portion of the external anal sphincter (EAS); the internal anal sphincter (IAS) is a thickened continuation of the smooth muscle of the rectum

mediated by nitric oxide synthase-containing neurons [11].

24.3.4 Pathologic Anatomy in Anorectal Malformations

Most of the structures observed in normal infants are present in anorectal malformations, but the anatomical relationships between structures involved in continence are displaced to varying degrees. In all ARMs, the levator muscles and external sphincter apparatus are more or less normally sited, but the bowel termination is increasingly displaced with increasing severity of malformation. Higher malformations are also associated with greater hypoplasia of the structures involved in faecal continence. Functional IAS tissue is present in the bowel termination of all ARMs, and the fistula contains all components of a normal anal canal, including stratified columnar epithelium, anal glands and anal columns [12]. In mild or 'low' malformations, the bowel termination is at least partially within the voluntary sphincter funnel, and the levator muscles are nearly normally developed.

24.3.5 Classification of ARMs

The main purpose of classification systems in ARMs has been to provide a platform for describing the anatomy, operative treatment and functional outcomes for different types of malformations. Several systems have been used over the past decades (Table 24.1), of which the most recent is the clinically oriented Holschneider (2005) [13] classification. This is a simplification of earlier models that divides

Table 24.1 Classification systems for ARMs

Holschneider (2005) [13]		Peña (1995) [14]		Stephens (1986) [15]	
Major clinical groups	Rare/regional variants	Males	Females	Males	Females
Perineal (cutaneous) fistula Rectourethral fistula: Bulbar Prostatic Bladder neck Vestibular fistula Cloaca No fistula Anal stenosis	Pouch colon Rectal atresia/ stenosis Rectovaginal fistula H-type fistula Others	Perineal fistula Rectourethral fistula: Bulbar Prostatic Rectovesical (bladder neck) Imperforate anus without fistula Rectal atresia	Perineal fistula Vestibular fistula Persistent cloaca: <3 cm common channel >3 cm common channel Imperforate anus without fistula Rectal atresia	*High* Anorectal agenesis Rectoprostatic fistula No fistula Rectal atresia *Intermediate* Bulbar fistula Anal agenesis *Low* Anocutaneous fistula Anal stenosis Rare malformations	*High* Anorectal agenesis Rectovaginal fistula No fistula Rectal atresia *Intermediate* Rectovaginal fistula Rectovestibular fistula Anal agenesis *Low* Anovestibular fistula Anocutaneous fistula Anal stenosis Cloaca Rare malformations

ARMs into major clinical groups and rare/regional variants. The preceding Peña [14] classification was based on the surgical approach and included division of ARMs into male and female groups. The Wingspread classification (1986) [15] considered anomalies based on the location of the rectal termination in relation to the levator plate. The major clinical groups described in the Krickenbeck classification are covered herein; the rare and regional types are uncommon, but the principles of surgical treatment are the same as for other types of ARMs.

24.3.6 Clinical Features of ARMs

In this communication, ARMs from the Krickenbeck classification with a bowel termination mostly *within* the external sphincter complex are considered *mild* types, and those with a bowel termination *outside* the EAS are considered *severe* anomalies.

24.3.7 Mild ARMs

24.3.7.1 Anterior Anus with or Without Anal Stenosis

Present exclusively in females, anterior anus represents the mildest type of ARM and is characterized by a normal-looking anus that is anteriorly sited (Fig. 24.5). In approximately half of patients, mild anal stenosis may be present. The bowel terminates natively mostly *within* (≥70%) the external sphincter complex. Anterior anus may be associated with a perineal groove, which is a mucosa-lined median cleft between the vestibulum and anus (Fig. 24.5) [16]. A perineal groove may also be present in the absence of an anorectal anomaly and in itself requires no treatment. The mucosal surface epithelializes gradually over time.

24.3.7.2 Perineal Fistula and Anal Stenosis in Males

In males, perineal fistula (Fig. 24.6) and anal stenosis essentially constitute variants of the same type of mild ARM [17]. In a male perineal fistula, there is a

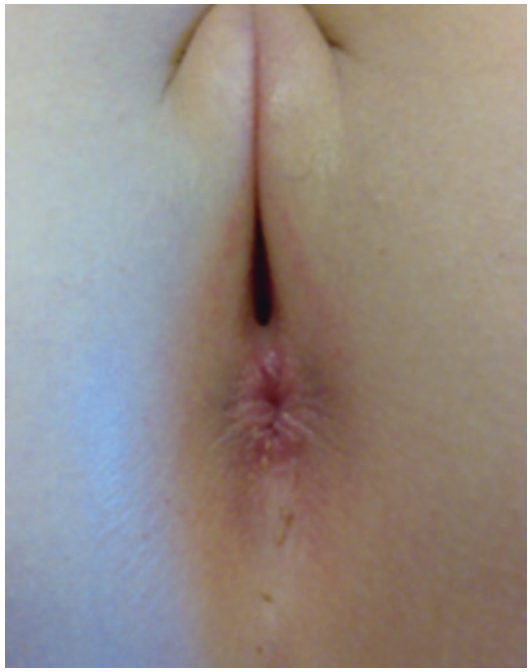

Fig. 24.5 Anterior anus with perineal groove. Reprinted from Fig. 3 Pakarinen and Rintala [16], with kind permission from Springer Science & Business Media

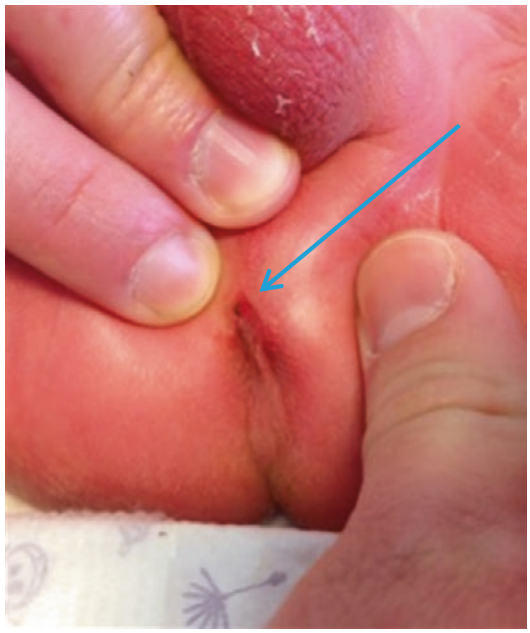

Fig. 24.6 Perineal fistula in a male (arrowed). There is a subcutaneous tract from the site of the bowel termination within the external sphincter complex

superficial subcutaneous tract in the midline for a variable distance along the perineum or scrotal raphe through which meconium extrudes (arrowed). The bowel termination is natively mostly *within* the external sphincter funnel, for which reason a perineal fistula in a male is considered to be a mild ARM. The bowel termination may be covered by a median bar or an anal membrane that is usually situated at the level of the dentate line [16]. Complete anal membranes are uncommon (2% of cases) and could also represent the least severe form of imperforate anus without a fistula [18].

24.3.8 Severe ARMs

24.3.8.1 Perineal or Vestibular Fistula in a Female

In contrast to a perineal fistula in a male, in females with a perineal or vestibular fistula, the bowel termination is anterior to and *outside* the support of the external sphincter funnel on the perineum (Figs. 24.7 and 24.8) [16, 19] or vestibulum (Fig. 24.9 and 24.10) [20]. Therefore, these are not considered mild ARMs. Rectovaginal fistula is a very rare variant, and most cases in which this is suspected are actually vestibular fistulas on careful clinical examination. In perineal and vestibular fistula, there are separate and usually normal openings for the urethra and vagina.

24.3.8.2 Cloaca

The most severe form of ARM in females is cloaca, which is characterized by a single perineal opening (Figs. 24.11 and 24.12). The urethra, vagina and anorectum terminate into a single common channel of variable length. Flat buttocks and natal cleft are suggestive of a longer common channel. Prenatal diagnosis of cloaca may be suspected in the presence of significant hydrometrocolpos. Nearly all cases of cloaca have associated anomalies.

24.3.8.3 Rectourethral Fistula

In males with no opening on the perineum (Fig. 24.13), the diagnosis is usually a rectourethral fistula, which is the most severe form of ARM in males. The fistulous connection of the terminal anorectum is most often at the level of

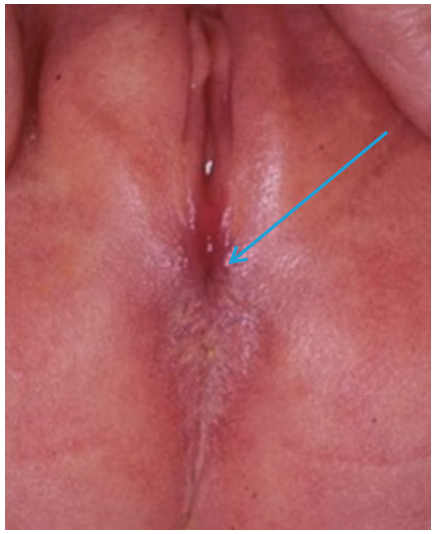

Fig. 24.7 Perineal fistula in a female (arrowed) reprinted from Pakarinen and Rintala [16], with kind permission from Springer Science & Business media

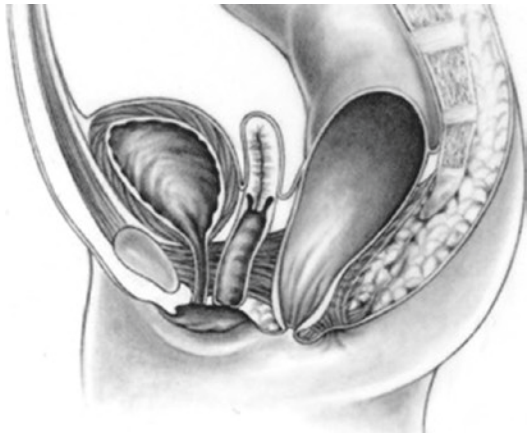

Fig. 24.8 Perineal fistula in a female; The diagram (black and white) from Levitt and Pena [19], with kind permission from BioMed Central

the bulbar or prostatic urethra but may be as high as the bladder neck (Fig. 24.14). Some patients may pass meconium via the urethra, which is diagnostic. Higher rectal terminations are associated with more significant hypoplasia of the native continence mechanisms, which is clinically apparent from a relatively flat and featureless bottom as with females with cloaca. The differential diagnosis when there is no perineal opening in males is imperforate anus without a fistula. Nearly all males with rectourethral fistula have associated anomalies.

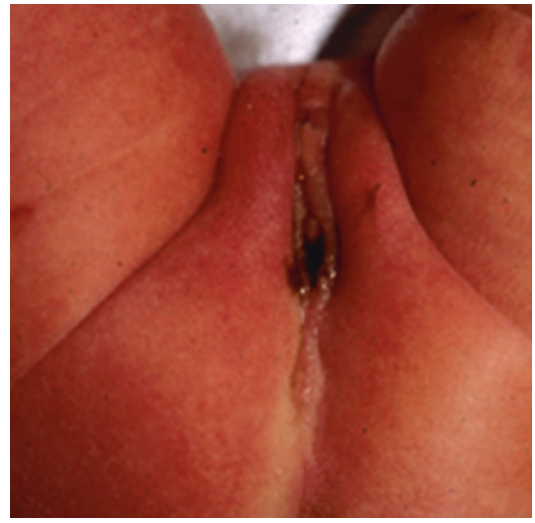

Fig. 24.9 Vestibular fistula in a female.

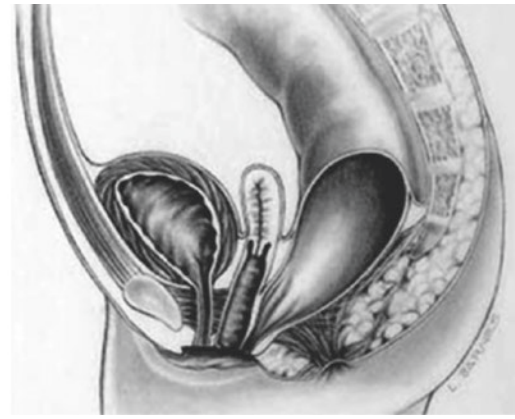

Fig. 24.10 Vestibular fistula in a female; Reprinted from Fig. 5 in Levitt and Pena [20], with kind permission from BioMed Central

24.3.8.4 Imperforate Anus Without a Fistula

Imperforate anus without a fistula is usually associated with well-developed sphincters, and this type of ARM occurs in males and females and is the most common type of ARM present in patients with Down syndrome. When the rectal termination is below the dentate line and immediately subcutaneous to the anal pit, the anomaly is termed anal agenesis. Less commonly, the anus may be normal-looking but terminates blindly at 1–3 cm of depth. In this type of ARM, termed rectal atresia, the rectal pouch commonly terminates above the levator plate [21].

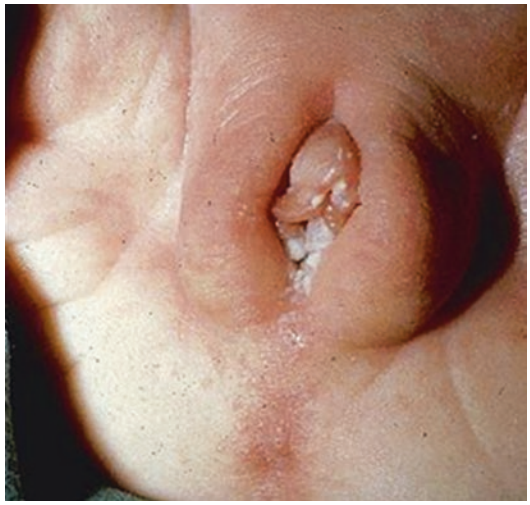

Fig. 24.11 Single channel cloaca.

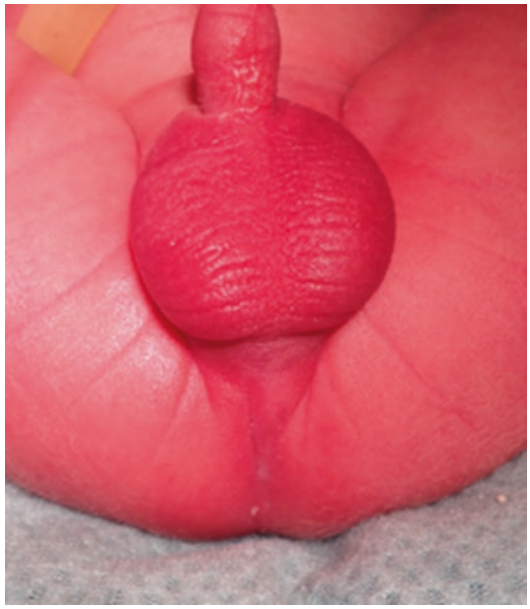

Fig. 24.13 No perineal opening in a male is usually a rectourethral fistula

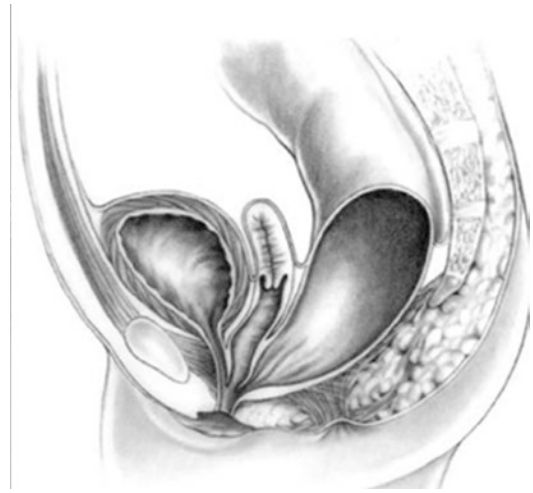

Fig. 24.12 Cloaca, single perineal opening; Reprinted from Fig. 7 in Levitt and Peña [19], with kind permission from BioMed Central

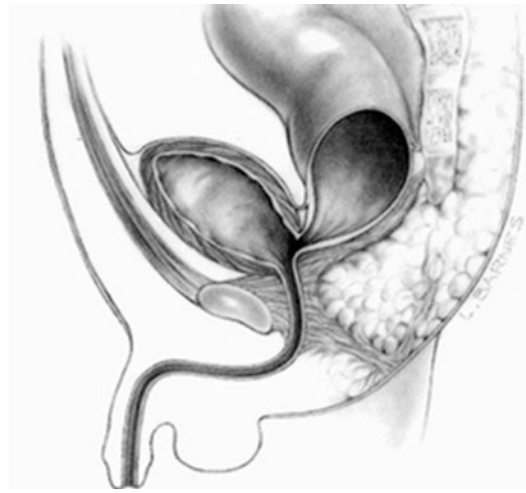

Fig. 24.14 No perineal opening in a male: usually rectobladder neck or rectourethral fistula. Reprinted from Fig. 6 in Levitt and Peña [19], with kind permission from BioMed Central

24.3.9 Initial Assessment

Clinical examination is often sufficient to make a provisional diagnosis of the severity of the ARM. In females, the diagnosis can be made from careful examination of the perineum to demonstrate the site of the termination of the anal canal: within the external sphincter complex in anterior anus, or fistulously on the perineum or vestibulum, or as a single channel cloaca. If there is any uncertainty in distinguishing an anterior anus from a perineal fistula in a female, electrical muscle stimulation under anaesthesia can clarify this [16]. In anterior anus, the anus is calibrated using Hegars (normal in a full-term neonate is Hegar 12–14). In males, meconium extruding onto the perineum indicates a mild ARM with a low rectal termination. If no fistula is appar-

ent, a nasogastric tube is passed, and the patient is kept nil by mouth. If there is still no meconium after 24–48 h of observation and gentle probing, it is safer to assume a high ARM as the working diagnosis and to proceed with a double-barrelled colostomy. Cross-table lateral X-rays and perineal ultrasound have limitations in their diagnostic sensitivity with regard to the level of the defect. Antimicrobial prophylaxis is appropriate in suspected urogenital connections or if there is vesicoureteric reflux.

24.3.10 Screening for Associated Anomalies

It is important to rule out serious associated malformations as part of the initial management. Normal passage of a nasogastric tube rules out oesophageal atresia, which is not uncommon in ARM patients. Cardiac and renal tract ultrasound and a chest/abdominal X-ray should be performed promptly for high anomalies. Screening investigations, during the hospitalization period include a micturating cystourethrogram, spinal column radiography (Fig. 24.15) and spinal ultrasound to assess for tethering. Chromosomal assessment and genetic consultation is indicated if a syndrome is suspected. Spinal cord magnetic resonance imaging can be performed at a later stage to rule out intraspinal anomalies.

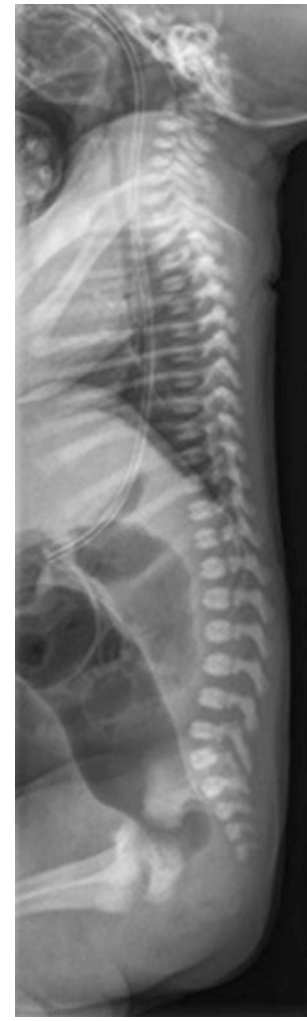

Fig. 24.15 Spinal column radiograph of a patient with a rectourethral fistula showing an extra vertebra with rudimentary ribs between T12 and L1 and four sacral segments

24.4 Initial Surgical Management

24.4.1 Mild ARMS

24.4.1.1 Anterior Anus

No operative management is indicated for patients with anterior anus because the anal canal terminates natively mostly within the external sphincter funnel. If anal stenosis is present, this usually responds well to serial Hegar dilatations up to Hegar 14, increasing the Hegar size at weekly intervals. Parents are taught to perform Hegar dilatations twice a day at home, attending outpatients for Hegar changes by a paediatric surgeon. In the absence of anal stenosis, observant, expectant management is sufficient. The long-term continence outlook with appropriate aftercare is good/normal in most cases [22].

24.4.1.2 Perineal Fistula, Anal Stenosis or Incomplete Anal Membranes in Males

In mild ARMs in males, the anal canal is usually located mostly within the external sphincter complex, and patients have a nearly normally developed anal canal [16]. In males with perineal fistula, the skin overlying the subcutaneous tract can be laid open over a thin probe inserted from the tip of the fistula to the centre of the sphincter complex to uncover the anus proper under general anaesthesia (Fig. 24.16), usually on the first day of life. Anal sphincter fibres are not divided

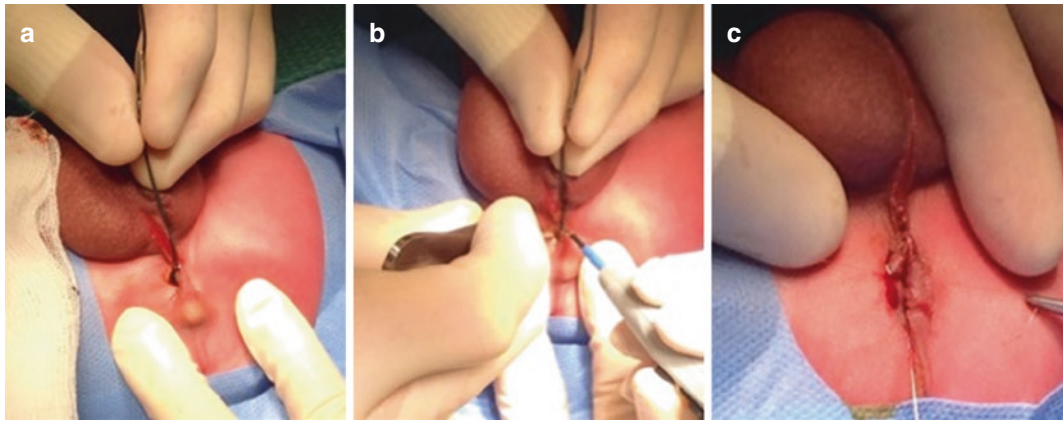

Fig. 24.16 Minimally invasive management of a perineal fistula in a male: (**a**) introduction of a thin probe through the subcutaneous fistula tract; (**b**) the skin overlying the fistula canal is laid open using diathermy up to the centre of the external sphincter complex; (**c**) anoplasty of the bowel terminus to the skin edges with a few absorbable interrupted sutures

during this procedure. The centre of the external sphincter complex may be delineated with electrostimulation. Standard anoplasty to suture the bowel termination to the skin edges is performed using a few interrupted absorbable sutures. Posterior sagittal anorectoplasty is also practised for males with perineal fistula, but there is no evidence to suggest superior outcomes compared to simply laying open the fistula tract. Urologic injuries should also be completely avoided with this minimally invasive approach.

If a mild ARM in a male is associated with a median bar or 'bucket handle' defect that interferes with faecal outflow, this should be excised. Males with anal stenosis and/or partial anal membranes can be treated with gradual Hegar dilatations, which can usually be performed without general anaesthesia. Complete anal membranes require surgical incision. If the patient has undergone a primary colostomy due to initial suspicion of a higher ARM, it is possible to incise the membrane under endoscopy control through the distal colostomy to transluminate the skin over the membrane [16]. The colostomy can be closed during the same procedure. Following anoplasty for perineal fistula, the anus usually approximates to Hegar 7–8 immediately post-operatively. Males with mild ARMs undergo a standard Hegar dilatation programme over 6 weeks up to Hegar 14, beginning 2 weeks post-operatively.

24.4.1.3 Anal Dilatation Programme

Parents are taught to perform twice daily dilatations with weekly outpatient visits for Hegar changes. Upon reaching Hegar size 14, a "2 + 2 + 2" programme is subsequently followed, comprising dilatations twice a day for the first 2 weeks, then every other day for 2 weeks and twice a week for the last 2 weeks. Anal strictures after a successful dilatation programme occur in only 2% of cases, and the likelihood requiring other anorectal surgery is low [17]. Funnel anus, a distinct type of anal stenosis characterized by a deep skin-lined funnel up to a stenotic ring (Fig. 24.17), is an exception [16]. As late presentation is common and although these patients may also be treated with serial dilatations, many go on to require excision of a megarectum. Funnel anus is most commonly associated with Currarino syndrome.

24.4.2 Severe ARMs

24.4.2.1 Perineal and Vestibular Fistulas in Females

The principles of surgical management of ARMs with an anal canal termination outside the external sphincter comprise restoration of the normal anatomical relationships between structures with minimal interference to existing continence mechanisms. Anterior sagittal anorectoplasty

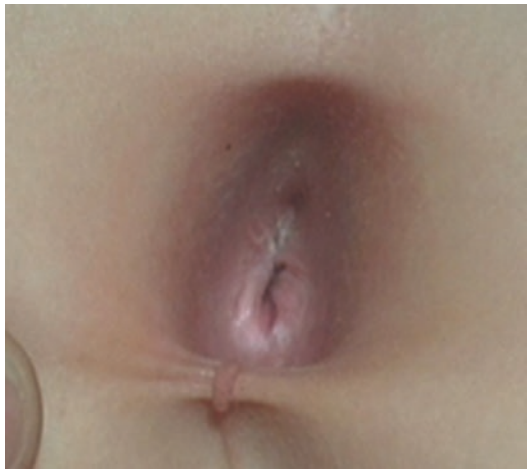

Fig. 24.17 Funnel anus. Reprinted from Fig. 2 in Pakarinen and Rintala [16], with kind permission from Springer Science & Business Media

(ASARP), also termed limited posterior sagittal anorectoplasty, is a well-established and minimally invasive operation for both perineal and vestibular fistulas in females [23]. In ASARP, a squash-racket incision is made around the fistula, extending this in the midline up to the centre of the external sphincter complex as marked pre-operatively by electrostimulation. The terminal anorectum is then carefully mobilized from its surrounding structures, taking particular care not to injure the posterior vaginal wall. Conservation of the fistula canal, which contains internal anal sphincter tissue in ARMs, is practised by the authors.

In ASARP, sufficient mobilization of the terminal anorectum to enable tension-free anastomosis to the centre of the external sphincter complex is technically important. The perineal body is then reconstructed in layers using absorbable sutures. ASARP may be safely performed as a single-stage procedure in otherwise well infants during the neonatal period [24, 25]. A covering sigmoid colostomy is currently advisable for older patients. Intravenous antibiotic prophylaxis is advisable during the immediate post-operative period. Most wound complications relating to ASARP are superficial infections that respond well to local hygiene and antibiotics. Patients undergo a standard anal dilatation programme up to Hegar 14 beginning 2 weeks post-operatively as for males with perineal fistula.

Posterior sagittal anorectoplasty is also practised for females with perineal and vestibular fistulas with comparable results to ASARP. Anal cutback has also been used to treat females with perineal fistula, but this does not equate to an anatomical repair as it leaves the external sphincter complex anteriorly deficient and the perineal body greatly shortened. Other procedures used in the past have been largely superseded by sagittal repairs.

24.4.2.2 Rectourethral Fistula

The current 'gold standard' approach to high urogenital connections including rectourethral fistula is posterior sagittal anorectoplasty (PSARP), which was first introduced by De Vries and Peña in 1982 [26]. PSARP largely modernized the safety and technical standards of the repair of severe ARMs by introducing an operative technique for anatomical reconstruction under direct vision. Serious operative complications reduced from 10 to 30% in classical operations of the past to 2% after PSARP [27]. Following a primary double-barrelled sigmoid colostomy in the immediate neonatal period, PSARP may be performed electively for rectourethral fistula at approximately 2–3 months of age but later if other congenital anomalies require repair first. The colostomy should be proximal enough to the retroperitoneal connections of the descending colon to reduce the risk of prolapse but most importantly so that enough length of distal bowel is left to reach the perineum during the definitive repair [20].

The level of the defect is ascertained pre-operatively as precisely as possible. The micturating cystourethrogram can demonstrate the fistula and its level of entry into the urogenital tract (Fig. 24.18), and a distal colostogram is complementary to this purpose. Filling the bowel with sufficient pressure to permit passage of contrast into the bladder through the fistula is technically important. Failure to demonstrate a urethral connection, however, does not completely rule out the possibility of one.

Classical PSARP involves a strict midline sagittal approach through the levator muscles and external sphincter with the patient in the prone position to access the bowel termination. Although nearly all urethral fistulas are acces-

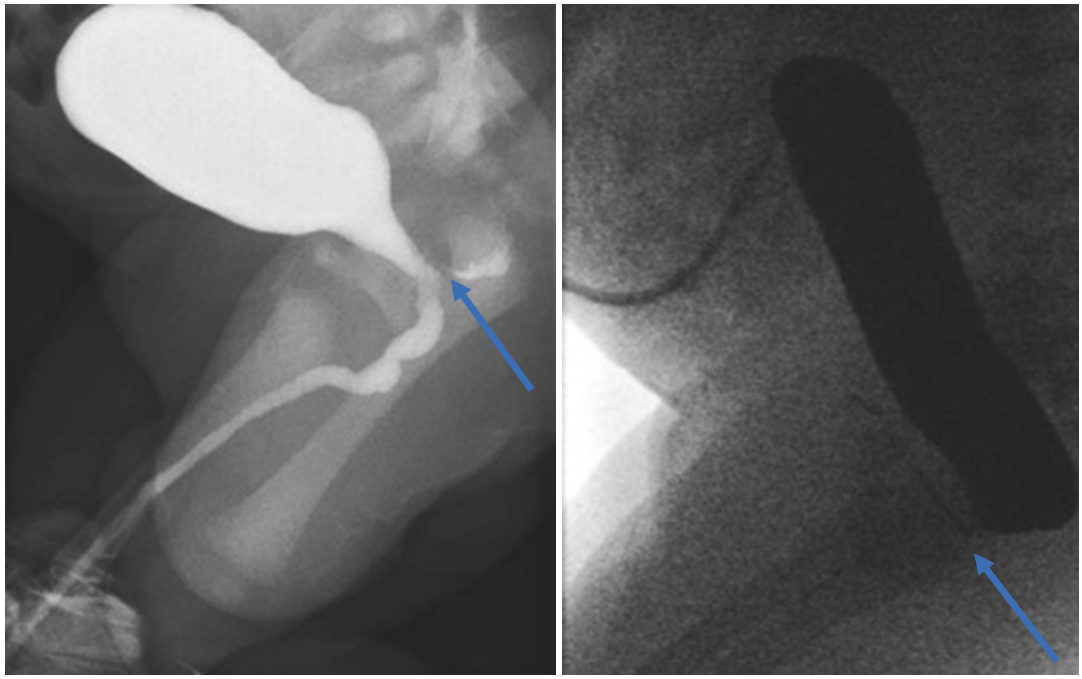

Fig. 24.18 Micturating cystourethrogram showing a rectourethral fistula at the level of the prostatic urethra (**a**) and the distal colostogram of the same patient (**b**) demonstrating the entry of contrast into the bladder through the fistula (arrowed)

sible through the posterior sagittal approach alone [20], laparoscopy is an attractive option for accessing high urogenital connections and permits excellent visualization of the fistula (Fig. 24.19). A combined technique, involving laparoscopic ligation of the fistula and a posterior sagittal incision for anastomosis of the bowel in the centre of the external sphincter complex, represents our current practice for patients' recto-prostatic or recto-bladder neck fistulas. In rectourethral bulbar fistulas, the posterior sagittal incision alone provides optimal access. Laparoscopic division are both anatomically challenging in these cases and may be complicated by a posterior urethral diverticulum due to incomplete excision of the fistula. As a limited modification of PSARP, the most distal part of the external sphincter may be conserved during posterior sagittal incisions. As with vestibular and perineal fistulas, an internal anal sphincter-saving technique that preserves the entire fistula canal is practised [27]. The recto-anal inhibitory reflex, indicative of functional

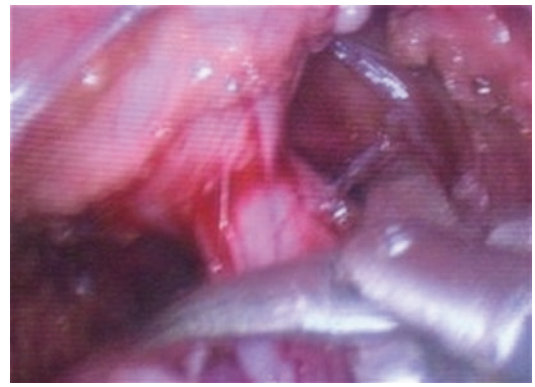

Fig. 24.19 Laparoscopic ligation of a high rectourethral (prostatic) fistula

internal anal sphincter tissue, has been demonstrated to be present in most patients post-operatively with this technique [28]. After PSARP, a standard anal dilatation programme is followed up to Hegar 14, after which colostomies can be closed.

Almost fully laparoscopic repairs for urethral fistula are also practised in some centres [29].

This technique involves laparoscopic dissection of the fistula and pull-through of the bowel via a small 1 cm incision at the centre of the external sphincter complex. Although minimally invasive, laparoscopic pull-throughs continue to be complicated by a high rate of rectal prolapse, and there are no long-term studies to suggest improved functional results. A benefit of the posterior sagittal incision is that it enables reconstruction of the normal anorectal angle through anatomical positioning of the bowel within the support of the sling muscles, which may be important for preventing rectal prolapse. Minor anal mucosal ectopy, which occurs in a few patients in the mid to long term after PSARP, is usually amenable to local corrective surgery.

24.4.2.3 Imperforate Anus Without a Fistula

The posterior sagittal approach is appropriate for patients with an imperforate anus without a fistula. In rectal atresia, the principles of operative management are otherwise the same as for urethral fistula. However, if a complete anal membrane is present (anal agenesis), this can be treated minimally invasively by incision as described previously. Colostomy closure can be performed at the same time. In imperforate anus without a fistula, the sphincter muscles are usually quite well developed, and the continence outcomes are mostly favourable.

24.4.3 Cloaca

Cloaca is a rare and complex type of anorectal malformation that requires considerable experience in its surgical management. In the immediate neonatal period, other life-threatening anomalies, including obstructive uropathy, cardiac anomalies and tracheo-oesophageal fistula, should be ruled out. Pre-operative investigations of the cloacal malformation should aim to establish the length of the rectourogenital confluence and the urethra and the anatomy of the bladder and gynaecologic structures for planning the reconstruction. Ultrasound of the urinary tract and imaging with contrast through the perineal opening may assist in this purpose (Fig. 24.20). However, not all structures may be visualized, and accurate interpretation of the findings is dependent on the experience of the radiologist.

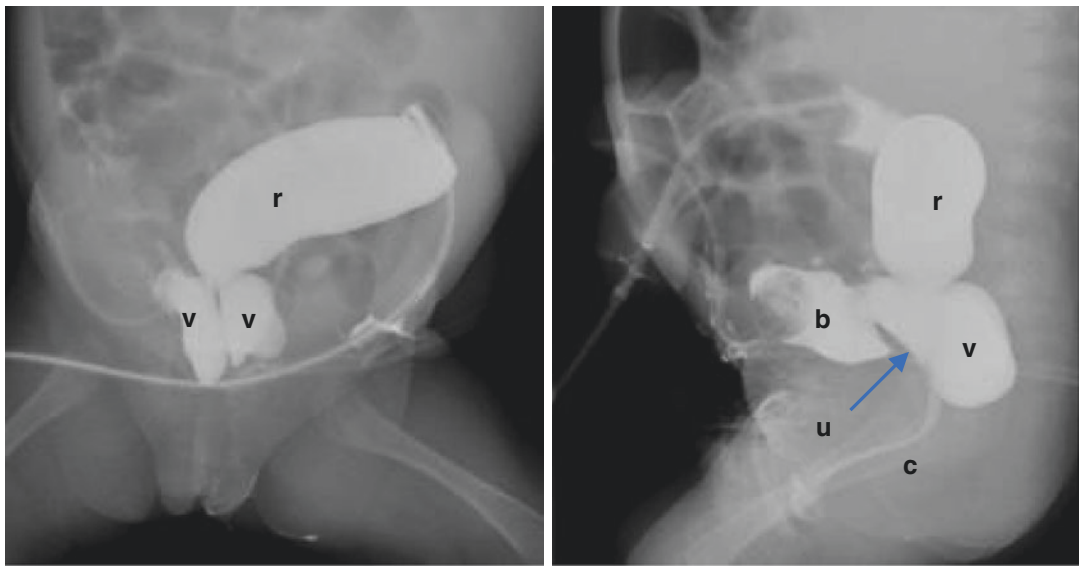

Fig. 24.20 Contrast study of a patient with a cloaca showing the rectum (r), duplex vagina (v), bladder (b), urethra (u) and urogenital confluence (c)

Endoscopy through the cloacal common channel using a paediatric cystoscope and warm flushing fluid is performed to measure the length of the common channel and urethra and visualize the number and size of vagina(s) and the position of the rectourogenital connection. A high-confluence cloaca is considered one with a common channel length in excess of 3 cm.

With regard to the initial colostomy, if there is (1) clear anatomy and (2) a patent vagina with no expectation of a possible need for vaginal reconstruction, a double-barrelled sigmoid colostomy may be performed. The colostomy should aim to leave as much distal bowel as possible for the definitive repair, so that the bowel can reach the perineum without tension. If there is (1) a very high confluence and (2) uncertainty of the need for sigmoid vaginal augmentation or (3) the anatomy is simply unclear, a transverse colostomy should be performed. A transverse colostomy does not preclude the later use of the sigmoid colon for vaginal reconstruction or hinder the subsequent pull-through. A contrast study of the distal bowel prior to the definitive repair should be performed and combined with the results of neonatal imaging when planning the definitive surgery.

Hydrometrocolpos is commonly present in cloaca patients, because urine may first pass into the vagina before being evacuated via the common channel [30]. Catheterization of the bladder is usually not possible without endoscopic aid, as the junction of the proximal urethra with the common channel is sharply angulated towards the pubic bone [30]. Catheter decompression of the hydrometrocolpos should be attempted as it can relieve urinary obstruction and prevent sepsis. If intermittent catheterization of either the bladder or vagina for urinary drainage is not feasible and severe urinary outflow obstruction or upper tract dysfunction is present, a vesicostomy is indicated. A colpostomy is only indicated in large hydrometrocolpos and often decompresses the upper urinary tract. However, a colpostomy may also preclude a posterior sagittal approach and total urogenital mobilization for vaginal pull-through.

24.4.4 Definitive Reconstruction

The timing and nature of the definitive reconstruction in cloaca patients are based on the clinical judgement and experience of the surgeon. The objectives include enabling faecal continence, urinary continence and a functional genital tract. Patients should be stable from a cardiovascular and urologic perspective, >5 kg of weight and over 6 weeks of post-gestational age. Through a posterior sagittal approach, the first structure to be opened in cloaca is the rectum, which is split in the midline; the vaginal and urethral communications are identified through the rectal opening. In high rectal communications, a laparotomy may be required. If there is a long common channel and a short proximal urethra, it may be opted to retain the common channel as the urethra, particularly if it is not too wide. The rectum and vagina are separated from their surrounding structures and pulled down to their anatomical positions on the perineum. The rectum is transected flush to the vaginal wall. The most difficult part of a cloacal repair this way is separation of the vagina from the urethra. Total urogenital mobilization, which was first introduced in 1996 [31], is an attractive option for cases with a short common channel for this reason. It involves mobilization of the urethrovaginal junction en bloc and bringing this down to the perineal skin, thereby only requiring separation of the rectum. In low confluences, posterolateral mobilization of these structures is sufficient, but in higher confluences, anterior retropubic dissection is also required. When required, vaginal reconstruction may be performed using bowel segment transposition with the (1) sigmoid colon or (2) ileum.

24.4.5 Basic Principles of Aftercare During the First Year of Life

Adequate aftercare of patients with ARMs is at least as important in securing optimal functional outcomes as a successful primary repair. Patients should be cared for in centres that are also able to provide high-quality, multidisciplinary manage-

ment of both the ARM and associated anomalies. In terms of bowel function, a tendency to constipation is a central feature that affects all types of ARMs. The aetiology may relate to developmental factors or to corrective surgery. The onset is commonly around the time the child begins to take solids, around 3–6 months of age. After surgical repair, patients should be reviewed in outpatients at regular intervals to ensure satisfactory stooling, and parents should be educated on the importance of attending to constipation. Most constipation in ARMs responds well to oral laxatives that include bulking agents (macrogols), lactulose and stimulant laxatives (natrium picosulphate). Treatment should be continued until the tendency to constipation completely resolves, which may not be for several years in some cases. Failure to address constipation can lead to severe complications, including megarectosigmoid from faecal impaction and secondary overflow incontinence [19]. Apart from an increasing level of the malformation, other negative prognostic factors for bowel function in ARMs are severe sacral defects, meningomyelocele and significant cognitive impairment. The families of patients with complex ARMs may benefit from additional psychological support to ensure appropriate adjustment to the child's condition and other disease-related stressors.

24.4.6 Long-Term Bowel Function

Parental counselling and open dialogue with regard to the functional outlook and possible problems can serve to establish a good foundation for the future years of follow-up. In mild ARMs, the functional outcomes are likely to become comparable to normal with adequate management of constipation, and toilet training from diapers can also be expected to occur at the normal age [17, 22]. In females with perineal or vestibular fistulas, at least 2/3 achieve normal bowel function over time, although minor continence disturbances may affect a proportion beyond the growth period [23]. In urethral fistula, the continence outcomes reduce with increasing level of fistula. They are good in approximately 40%, with the remainder reporting varying degrees of residual symptoms. Although the majority of patients with high bladder neck fistulas achieve social continence with modern care, secondary measures for bowel management such as an antegrade continence enema (ACE) conduits are likely to be required [27]. Secondary interventions aim to enable weaning from diapers before primary school and to ensure normal social integration during childhood. In cloaca, approximately half of patients attain faecal and urinary continence, and the remainder stay clean or dry by adjunctive measures, including bowel management, continent urinary diversion or intermittent catheterization [32]. However, most patients with ARMs report a normal quality of life after contemporary treatments in the long term [32, 33].

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