Anorectal Malformations

22

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22.1 Introduction

Anorectal malformations (ARM) are congenital defects in which the terminal part of the hindgut is abnormally placed and lies outside (partially or completely) the sphincter mechanism.

22.1.1 Epidemiology

- Incidence ~1 in 5000.
- Frequently associated to Down syndrome and Cat Eye syndrome.
- M > F (60:40).
- Second child involvement is rare (<1%).
  - Exception: In those cases of ARM associated with pre-sacral masses second child involvement is extremely high.

About 5% of infants have no fistula (usually associated with Down syndrome) but the vast majority does have a connection between the distal rectum and the adjacent urogenital tract or the perineal skin.

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22.1.2 Embryology

By 21 days, there is a common chamber (cloaca[1]) occluded by a membrane but visible from the outside as an ectodermal pit—the *proctodeum*. At ~33 days, the posterior hindgut is then separated from the anterior urogenital sinus by mesenchymal ingrowth from the urorectal septum. This cloacal membrane breaks down at about 46 days. The process is regulated by differential expression of the gene Sonic Hedgehog (*SHH*), and other target genes such as *BMP-4* and the *HOX* genes.

22.1.3 The Anatomical Classification

Males	Females
Rectoperineal fistula	Rectoperineal fistula
Rectobulbar fistula	Rectovestibular fistula
Rectoprostatic fistula	Cloaca
Recto bladder neck fistula	
Imperforate anus without fistula (i.e., rectal atresia)	

22.1.4 Associated Anomalies

Associated malformations are much more commonly associated with complex defects (e.g., cloacas, recto-bladder neck fistula).

- Urologic— ~50%.
  - Most common: Absent or nonfunctional kidney, hydronephrosis, vesicoureteral reflux.
- Vertebral— ~30%.
  - Mainly hemivertebrae.
- Cardiac— ~30% (only one-third of them require treatment).
- Hydrocolpos in cloaca patients— ~30%.
- Spinal cord— ~25%.
  - Tethered cord.
- Other gastrointestinal.
  - Esophageal atresia ~8%.
  - Duodenal atresia 3–6%.

22.1.5 Clinical Features

Perineal examination is the most important part of diagnosis and frequently it allows the diagnosis of the specific type of anomaly to be made.

[1] Cloaca—Latin "sewer." The Cloaca Magna was the main sewer in Imperial Rome.

22.1.5.1 **Males** (Fig. 22.1)

- Prominent, well-formed midline groove.
  - Most likely is "benign" and has a good prognosis.
- Anal dimple.
  - Type of malformation is usually a bulbar fistula, or no fistula.
- Bucket handle malformation.
  - Implies that there is a perineal fistula.
- Flat bottom (no midline groove, no anal dimple).
  - Implies that most likely it is a complex malformation.
- Bifid scrotum.
  - Implies that there is a recto-bladder neck or recto-prostatic fistula.
- Meconium in urine.
  - Implies that there is a recto-urinary fistula.

22.1.5.2 **Females** (Fig. 22.2)

The perineal (genital) inspection of female infants allows a precise diagnosis of the specific type of ARM in most cases. It is necessary for the clinician to separate the labia of the genitalia in order to be able to make a precise diagnosis.

- Single perineal orifice.
  - Cloaca.

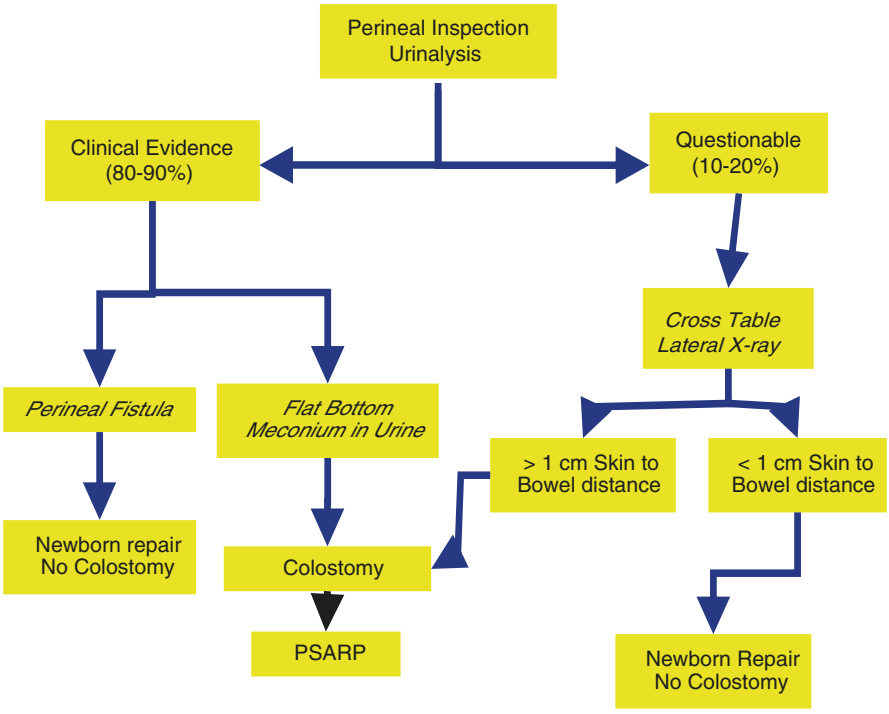

Fig. 22.1 Decision algorithm in boys

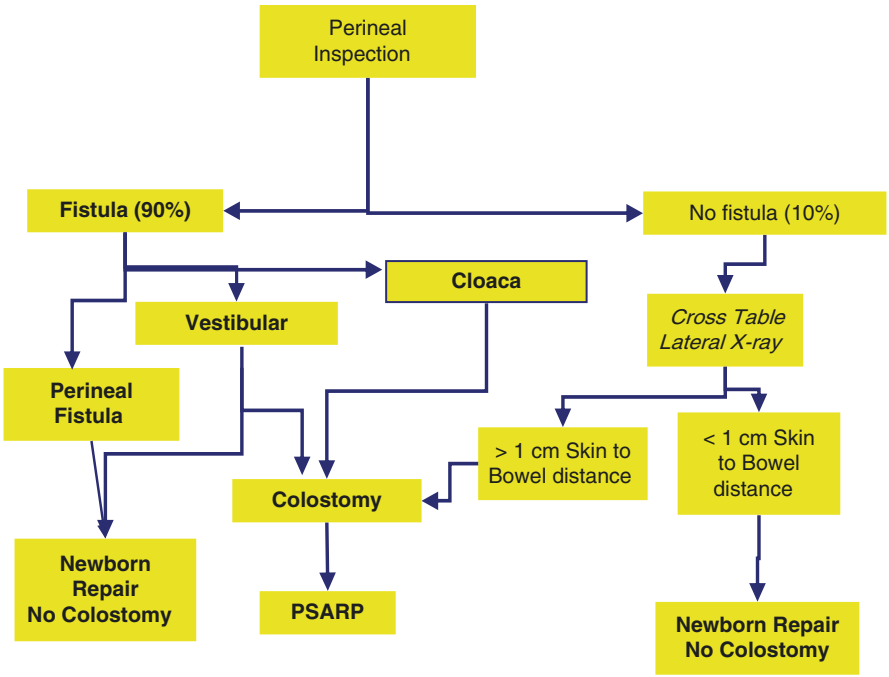

Fig. 22.2 Decision algorithm in girls

- Bowel orifice located in the vestibule.
  - Vestibular fistula.
- Bowel orifice located at the perineal body.
  - Recto-perineal fistula.
- Palpable separated pubic bones.
  - Covered cloacal exstrophy.
- Meconium coming from inside the vagina.
  - Recto-vaginal fistula (almost nonexistent).

22.2 Neonatal Management

The first 24 hours represents the "window of opportunity" to rule out major associated malformations which may represent a danger to the infant's life.

- Immediately after birth:
  - Remain fasting.
  - N/G tube must be inserted.
  - IV access must be established.
  - Prophylactic antibiotics.

Once all this is done:
TWO MAJOR Questions must be answered:

- Does the infant have a serious malformation that represents a "risk to life" situation?
- Does the infant need a colostomy or a primary repair?

These questions must be answered in the order presented here. In order to do that, the following studies must be performed, DO NOT PERFORM SURGICAL PROCEDURES BEFORE ANSWERING THESE QUESTIONS.

- Renal ultrasound in males.
  - Rule out hydronephrosis and/or absent kidney.
- Renal and pelvic ultrasound in females.
  - Rule out hydronephrosis, and/or absent kidney, and presence of hydrocolpos in those with cloacas.
- X-ray film of the sacrum, to estimate the sacral ratio and try to predict the future functional prognosis (See Fig. 22.3).
- Lumbosacral ultrasound to rule out the tethered cord.
- ECHO cardiogram.
- "Babygram"
  - X-ray film of the entire body to rule out hemivertebrae, esophageal, and duodenal atresia.

22.2.1 Surgical Management—Colostomy or Primary Repair?

After 24 hours of life, all infants will have abdominal distention. This is a prerequisite to creating a valid image—the *"cross-table lateral film of the pelvis and abdomen."* This is taken with the infant in prone position, with the pelvis elevated. The

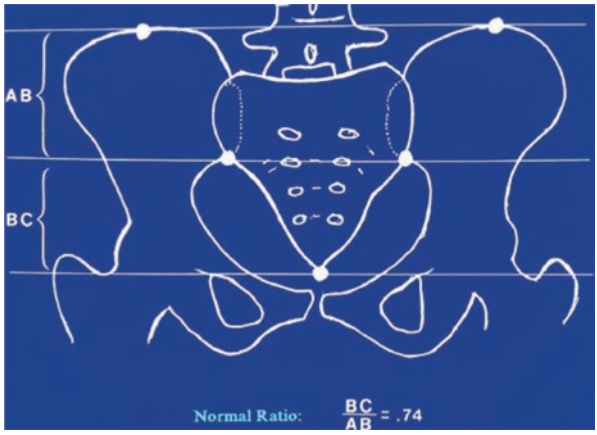

Fig. 22.3 Sacral ratio

purpose of this film is to determine the location of the most distal part of the rectum, as shown by intraluminal air in the rectum. Most importantly, it will determine the location of the rectum as related to the most distal part of the sacrum and will help the surgeon to determine whether or not the rectum is reachable via the posterior sagittal approach.

This film is mainly valuable when the baby does not have any of the clinical signs described above.

22.2.2 Primary Perineal Approach

Depending on the experience of the surgeon, a primary perineal approach is indicated in cases of full-term infants without any important associated defects, and clearly suffering from:

- Perineal fistula.
- Vestibular fistula.
- Rectal gas is present below the most distal part of the sacrum in the cross-table lateral film.

22.2.3 Colostomy

- This procedure is indicated in all other types of anorectal malformations.

The ideal colostomy is created in the descending colon and is mandatory to separate the stomas enough, to allow the placement of a stoma bag covering only the functional, proximal stoma. The proximal stoma is opened in the center of a triangle formed by the umbilicus, the iliac crest, and the lower rib. The nonfunctional stoma (mucus fistula) is opened medially and lower.

In our opinion, loop colostomies are contraindicated due to the fact that they represent a risk for fecal contamination of the urogenital tract.

In our opinion, transverse colostomies are also contraindicated, because they do not allow removal of meconium from the distal colon; may produce hyperchloremic acidosis due to absorption of urine from the bowel; and, contribute to the formation of a megarectum.

In our opinion, sigmoid colostomies are undesirable, because they sometimes interfere with the performance of the subsequent pull-through.

Following formation of the colostomy, the infant must be checked and followed to be sure that they are growing and developing normally. Once this is demonstrated and depending on the surgeon's experience, the infant can be subjected to the main repair.

22.2.4 High-Pressure Distal Colostogram

The main repair of an ARM should never be done without a precise anatomic diagnosis done by a high-pressure distal colostogram. The study is done under fluoroscopy, using ONLY water-soluble contrast material. A radio-opaque marker is placed at the anal dimple. A Foley catheter is inserted through the mucous fistula, the balloon of the catheter is inflated with 5 ml of water and the contrast is injected with a syringe by hand. The infant is placed in a supine position and the injection continues until the most distal part of the rectum is reached. Place the infant in a lateral position, being sure that in the fluoroscopy screen one can see the sacrum, the anal marker, and the entire pelvis. This must be a perfect lateral position, as shown by the images of both hips being superimposed. The injection continues until the contrast stops, forming a horizontal line, which may be misinterpreted by an inexperienced radiologist as being the most distal end of the rectum, making the wrong diagnosis of a "high" malformation. That line actually represents the effect of the compression produced by the tone of the funnel-like muscle sphincter mechanism that surrounds the lowest part of the rectum. At that point, the provider must continue injecting the contrast using reasonable hydrostatic pressure, which eventually will show the true most distal part of the rectum. The injection continues in order to demonstrate the fistula location and the passing of contrast through the urethra or back into the bladder. As a bonus, the study will also allow one to rule out the presence of vesicoureteral reflux.

22.2.5 Main Repair

The main repair of an ARM should never be a surgical "exploration." A technically correct pre-op evaluation allows the performance of a planned procedure and avoids technical mistakes.

The type of operation will be determined by the specific anatomic diagnosis:

- Rectourethral bulbar fistula.
  - PSARP (Posterior Sagittal Ano-Recto-Plasty).
- Rectourethral prostatic fistula.
  - Depending on the surgeon's experience, it can be repaired via PSARP or laparoscopically assisted.
- Recto-bladder neck fistula.
  - Abdominal approach (laparotomy or laparoscopy).
  - N.B. this malformation only occurs in 10% of males.

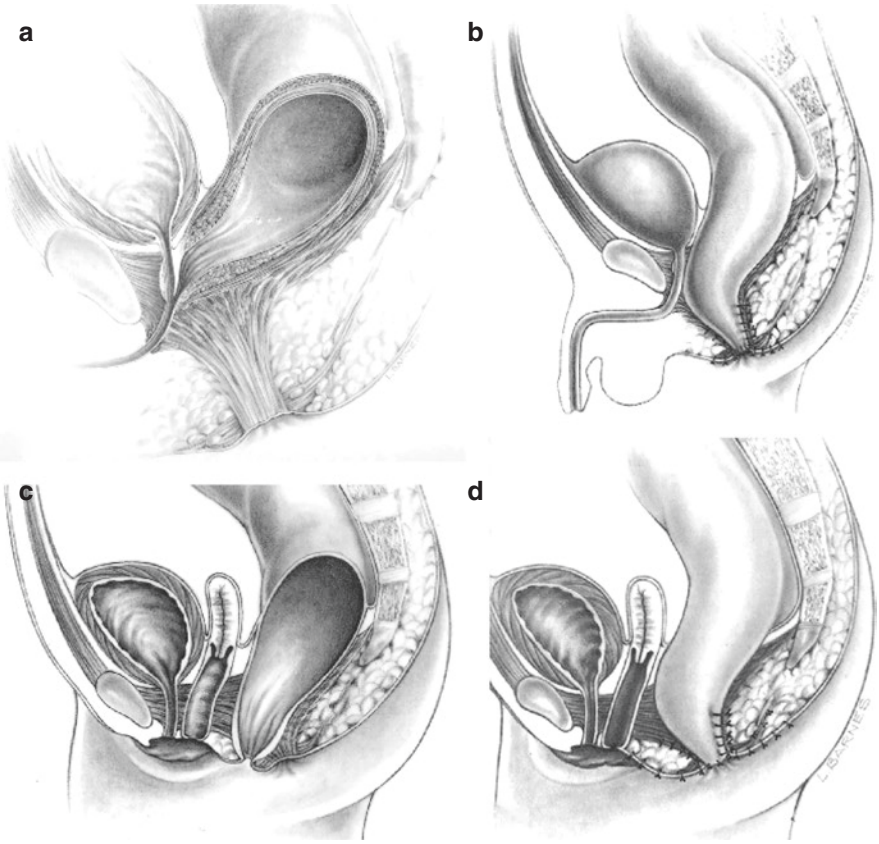

Fig. 22.4 PSARP in boys (**a**, **b**) – PSARP in girls (**c**, **d**)

22.2.5.1 PSARP[2]

(Important steps in male patients with rectourethral bulbar or prostatic fistulae (Fig. 22.4a and b)

- Patient in a prone position with the pelvis elevated.
  - ALWAYS put a Foley catheter in the bladder.
- Posterior midline incision running from the coccyx to the perineal body.
  - Preferentially use an electrical stimulator to identify the muscle structures and divide them in the middle.
  - Divide all the sphincter mechanisms in the midline.
- Open the posterior rectal wall and extend the incision all the way down to the fistula.
  - Place multiple fine sutures taking the rectal mucosa cephalad to the fistula.

[2] Classic Paper—deVries PA, Peña A. Posterior sagittal anorectoplasty. J Pediatr Surg. 1982; 17: 638–43.

  - Create a plane of dissection between the anterior rectal wall and the posterior urethra.
  - Close the fistula with three fine absorbable sutures.
- Perform a circumferential dissection of the rectum to bring it down. Remain as close as possible to the rectal wall during the dissection, but do not injure the rectum.
  - Determine the limits of the sphincter mechanism.
  - Place the rectum within the limits of the sphincter.
- Reconstruct the perineal body if necessary.
- Anoplasty to be done with sixteen circumferential stitches of fine absorbable sutures.
- Suture the levator and muscle complex behind the rectum, preferably taking a bite of the posterior rectal wall.

Foley catheter must remain in place for one week. If the catheter accidentally comes out, do not try to replace it, such an attempt may injure the repair and most likely the patient will pass urine uneventfully.

PSARP in cases of recto-prostatic fistula. Same as above, but must look for the rectum higher (in front of the sacrum).

22.2.5.2 Recto-Bladder Neck Fistula

- Approach the patient via laparotomy or laparoscopy.
- Ligate the fistula.
- Inspect the blood supply of the recto-sigmoid carefully.
- Ligate the necessary vessels to allow the rectum to reach the perineum without tension, being sure to preserve the necessary arcades of the colon's blood supply to guarantee that the rectum receives, a good blood supply.
- Approach the pelvis via a posterior sagittal incision to create the correct space to pull the rectum down.

Pull the bowel down and reconstruct as described for rectourethral ***fistulae.***

22.2.5.3 PSARP For Females with Recto-Vestibular Fistula (Fig. 22.4b and c)

- Prone position.
  - Make a posterior sagittal incision as previously described.
  - Divide the sphincter mechanism and identify the posterior rectal wall.
  - Apply multiple fine sutures in a circumferential manner at the bowel opening in the vestibule.
- Applying uniform traction on the sutures dissect first the posterior rectal wall, follow by the lateral walls of the rectum.
  - Using a delicate and meticulous technique create a plane of separation between the anterior rectal wall and the posterior vaginal wall (this is the most delicate part of the operation).

- Determine the limits of the sphincter.
- Reconstruct the perineal body.
- Anoplasty and wound closure as previously described.

22.2.5.4 Perineal Fistula in Females

The technique has been described as a "miniPSARP." The operation is very similar to the one described for a recto-vestibula fistula. However, the separation of the rectum from the vagina is much easier, because there is a plane of separation between them.

22.2.5.5 Perineal Fistula in Males

This malformation can be repaired following the same principles described for females. However, the surgeon must keep in mind that the anterior rectal wall is intimately attached to the posterior urethra and there is no real plane of separation. The most common and feared intraoperative complication in this malformation is the urethral injury. Insertion of a Foley catheter is mandatory to perform this procedure.

22.2.5.6 Cloaca

Rectum, vagina, and urethra are fused together forming a common channel of variable length, and opening at the same location as the normal female urethra. The treatment of these malformations depends on the length of the common channel (distance between the external single perineal orifice and the point of bifurcation or trifurcation of the common channel).

Common Channel <3 cm

- This malformation is repaired via a posterior sagittal anorectoplasty.
- The rectum is separated from the vagina following the same principles described in other malformations.
- Both urethra and vagina are mobilized using the maneuver called "***Total Urogenital Mobilization.***"
- The limits of the sphincter are determined.
- The perineal body is reconstructed.
- The rectum and anus are reconstructed as previously described.

Common Channel >3 cm

These malformations require a posterior sagittal approach and a laparotomy. The three structures (urethra, vagina, and rectum) must be separated transabdominally, under direct vision. These complex operations must be done by surgeons fully dedicated to the repair of complex malformation, working in a center with a high volume of cases. The surgeons must have a special training in urology, in addition to pediatric surgery or to work in conjunction with a pediatric urologist with experience in the treatment of these defects.

Further Reading

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