2) Anorectal Malformation (Imperforate anus) - Dr. Mahdi

Embryology of the imperforate anus

- Between 4-6 weeks, the cloaca becomes the common depository for the developing urinary, genital and rectal systems.
- The cloaca is quite promptly divided into an anterior urogenital sinus and a posterior intestinal canal by the urorectal septum.
- Two lateral folds of cloacal tissue join the urorectal septum to complete the separation of the urinary and rectal tracts.

Imperforate anus

- Includes agenesis and atresia of the rectum and anus
- Etiology: unknown
- Incidence: 1 in 4,500
- SEX: 60% male

Classification

- Anorectal malformations represent a wide spectrum of defects.
- The terms “low,” “intermediate,” and “high” are arbitrary and not useful in therapeutic or prognostic terms

### Males

- Cutaneous (perineal fistula)
- Rectourethral fistula
- Bulbar
- Prostatic
- Recto–bladder neck fistula
- Imperforate anus without fistula
- Rectal atresia

### Females

- Cutaneous (perineal fistula)
- Vestibular fistula
- Imperforate anus without fistula
- Rectal atresia
- Cloaca
- Short common channel
- Long common channel
- Complex malformations

MALE ANORECTAL DEFECTS

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.
- 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.
- This diagnosis is established by perineal inspection. No further investigations are required.
- The terms covered anus, anal membrane, anteriorly mislocated anus, and bucket-handle malformations all are refer to perineal fistulas.

2. Rectourethral Fistulas

- Imperforate anus with a rectourethral fistula is the most frequent defect in male patients.
- The fistula may be located at
- The lower (bulbar) part of the urethra
- Lower urethral fistulas are usually associated with good-quality muscles, a well-developed sacrum, a prominent midline groove, and a prominent anal dimple.
- The higher (prostatic) part of the urethra
- Higher urethral fistulas are more frequently associated with poor-quality muscles, an abnormally developed sacrum, a flat perineum.
3. Recto-Bladder Neck Fistulas

- In this defect, the rectum opens into the bladder neck.
- The patient usually has a poor prognosis for bowel control because the levator muscles, the striated muscle complex, and the external sphincter frequently are poorly developed.
- The sacrum is often deformed and short.
- The perineum is often flat, which is evidence of poor muscle development.
- About 10% of males with anorectal atresia fall into this category.

4. Imperforate Anus without Fistula

- Interestingly, most patients with this unusual defect have a well-developed sacrum and good muscles, and have a good prognosis in terms of bowel function.
- The rectum usually terminates approximately 2 cm from the perineal skin.

5. Rectal Atresia

- In this extremely unusual defect in male patients (~1% of the entire group of malformations), the lumen of the rectum is totally (atresia) or partially (stenosis) interrupted.
- The upper pouch is represented by a dilated rectum, whereas the lower portion is represented by a small anal canal that is in the normal location and is 1 to 2 cm deep.
- These two structures may be separated by a thin membrane or by dense fibrous tissue.
- The repair involves a primary anastomosis between the upper pouch and anal canal and is ideally approached posterior sagittally.
- 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 and have almost normal voluntary sphincters.

FEMALE ANORECTAL DEFECTS

1. Rectoperineal Fistulas

- From the therapeutic and prognostic viewpoint, this common defect is equivalent to the perineal fistula described in the male patient.
- The rectum is well positioned within the sphincter mechanism, except for its lower portion, which is anteriorly located.
- The rectum and vagina are well separated.

2. Rectovestibular Fistulas

- Rectovestibular fistula is the most common defect in girls and has an excellent functional prognosis.
- The diagnosis is based on clinical examination.
- A meticulous inspection of the neonatal genitalia allows the clinician to observe a normal urethral meatus and a normal vagina, with a third hole in the vestibule, which is the rectovestibular fistula.

3. Imperforate Anus without Fistula

- This defect in female patients carries the same therapeutic and prognostic implications as described for male patients.
4. Rectal atresia

5. Persistent Cloaca

- 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 fuse, creating a single common channel.
- 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.
- This distance has technical and prognostic implications.
  1. Short common channel less than 3 cm
  2. Long common channel more than 3 cm

6. Complex Malformations

- Unusual and bizarre anatomic arrangements can be seen.
- Each case represents a unique challenge to the surgeon, with different prognoses and therapeutic implications.
- No general guidelines can be drawn for the management of these patients. Each case must be individualized.

ASSOCIATED DEFECTS

1. Sacrum and Spine: Sacral deformities appear to be the most frequently associated defect.
2. Genitourinary Defects: The frequency of associated genitourinary defects varies from 20% to 54%.
3. Anal atresia may occur as a part of the VACTERL group of anomalies
   - V: Vertebral body segmentation defect
   - A: Anal atresia
   - C: Cardiovascular (PDA, VSD)
   - TE: Tracheo esophageal fistula
   - R: unilateral Renal agenesis
   - L: Limb anomaly (radial ray hypoplasia)
   - So, very careful examination of the baby must be made to exclude these anomalies
Management

**Newborn Male Anorectal Malformation**

During the first 24 hours, the neonate should

- Take nothing orally
- receive intravenous fluids
- and antibiotics
- and be evaluated for associated defects that may represent a threat to life. These include:
  - NGT to exclude esophageal atresia
  - Echocardiogram to exclude cardiac malformations, esophageal atresia.
  - Radiograph of the lumbar spine and the sacrum
  - Spinal ultrasonogram to evaluate for a tethered cord.
  - Ultrasonography of the abdomen will evaluate for renal anomalies.
  - Urine analysis

After 24 hours

- Re evaluate
- If the neonate has signs of a perineal fistula, treatment is by doing an anoplasty, without a protective colostomy, this can be performed during the first 48 hours of life.
- if there is no meconium on the perineum, we recommend obtaining a cross-table lateral radiograph with the patient in the prone position
  - If air in the rectum is located below the coccyx, and the patient is in good condition with no significant associated defects, one may consider performing a posterior sagittal operation with or without a protective colostomy
  - Conversely, if the rectal gas does not extend beyond the coccyx, or the patient has meconium in the urine, an abnormal sacrum, or a flat bottom, a colostomy should be done.
  - Then perform a posterior sagittal anorectoplasty 1 to 2 months later, provided the neonate is gaining weight appropriately.

**Newborn Female Anorectal Malformation**

During the first 24 hours, the neonate should

- Be examined carefully (perineal examination)
- Take nothing orally
- receive intravenous fluids
- and antibiotics
- and be evaluated for associated defects that may represent a threat to life. These include:
  - NGT to exclude esophageal atresia
  - Echocardiogram to exclude cardiac malformations, esophageal atresia.
  - Radiograph of the lumbar spine and the sacrum
  - Spinal ultrasonogram to evaluate for a tethered cord.
  - Ultrasonography of the abdomen will evaluate for renal anomalies.
Complications

Complications of posterior sagittal anorectoplasty

There are several complications related to operative intervention and repairs of anorectal malformations in general.

1. Wound infection in the immediate postoperative period can occur and, fortunately, usually affects only the skin and subcutaneous tissue. All heal secondarily without functional sequelae.
2. Anal strictures may be the consequence of failure to follow the protocol of dilatations. When trying to prevent discomfort for the patient, some surgeons dilate the anus once a week, frequently under anesthesia. This protocol can eventually create a severe, intractable fibrous stricture. Strictures can also occur with devascularization of the rectum during the rectal mobilization.
3. Constipation is the most common functional disorder observed in patients who undergo a posterior sagittal anorectoplasty. Patients with the best prognosis have the highest incidence of constipation. Patients who underwent tapering do not have more constipation than those without a tapering procedure. Patients with a very poor prognosis, such as with bladder neck fistula, have a low incidence of constipation

Differential Diagnosis of Conditions That May Be Associated with Failure to Pass Meconium in the Newborn

- Hirschsprung's disease >> Tight anus, empty rectum, transition zone
- Meconium plug syndrome >> Meconium plugs
- Meconium ileus >> Abdominal distention at birth, cystic fibrosis
- Anorectal malformation >> Absent anus, tight anus or fistula
- Small left colon syndrome >> Transition zone* at splenic flexure
- Hypoganglionosis >> Transition zone*
- Neuronal intestinal dysplasia type A >> Transition zone*, mucosal inflammation
- Neuronal intestinal dysplasia type B >> Megacolon
- Megacystis-microcolon-intestinal hypoperistalsis syndrome >> Microcolon, megacystis

* -- Transition zone (from small- to large-diameter bowel) refers to radiographic visualization on contrast study.