Anorectal Malformations

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Incidence
The incidence of imperforate anus is one in every 5,000 live births, with cloaca malformations accounting for 10%. Males (58%) are more commonly affected than females (42%).

Etiology
In the third week of gestation, the embryo consists of an amniotic cavity and a larger yolk sac separated by a trilaminar disc consisting of ectoderm (amniotic side), mesoderm (middle), and endoderm (yolk sac side). The disc then begins a cranio-caudal folding process that tubularizes a portion of the endoderm into what will eventually become the hindgut. The hindgut joins the allantois and the mesonephric ducts to form the cloaca. At the end of the cloaca, endoderm of the cloaca is in direct contact with surface ectoderm creating the cloacal membrane. During development, this membrane moves posteriorly and inferiorly.

Cloacal division into rectum and urogenital tract is initiated by the caudal movement of tissue between the allantois anteriorly and the hindgut posteriorly. This cranio-caudal movement stops at the verumontanum. At seven weeks, cloacal division is completed by lateral ingrowth of mesenchyme, thereby completing the urogenital septum and forming the perineum. The perineum divides the cloacal membrane into the urogenital membrane anteriorly and the anal membrane posteriorly. Mesenchymal swellings then surround the anal membrane. The anal pit, a depression in the ectoderm at the anal membrane, develops in the eighth week and the membrane perforates in the ninth week.

Anorectal malformations occur when this process fails. The exact etiology of failure is currently unknown.

Classification
Previously, infants with imperforate anus were classified based on the relationship of the rectal terminus to the levators. This was determined by inverting the infant and taking a transpelvic xray (invertogram). Rectal termini above the pubococcygeal line (a line drawn between the pubis and coccyx) are above the levators and designated high lesions. Termini between the pubococcygeal line and the lowest quarter of the ossified ischium (the “I” point) are translevator and designated intermediate lesions. Those below the “I” point traverse the levators and are designated low lesions.
More recently, a treatment-based classification system was proposed. In this system, infants are separated into two groups based on their need for a colostomy. Infants with a cutaneous fistula, anal stenosis, or anal membranes can undergo primary repair without protective colostomy. In contrast, those with rectourethral fistula, rectovesical fistula, anorectal agenesis without fistula, rectal atresia, vestibular fistula, vaginal fistula, or cloacal malformations require protective colostomy before primary repair.

Clinical Presentation

Most infants with imperforate anus are referred because no anal opening is identified (Fig. 79.1) on the newborn screening exam or because of failure to pass meconium. Although most are healthy, full-term infants, associated congenital anomalies are common. These anomalies include vertebral anomalies, limb anomalies, heart defects, and Downs syndrome. The presence of any one of these mandates ruling out the others. The two most common cardiac anomalies associated with anorectal malformations are tetralogy of fallot and ventricular septal defects. Duodenal atresia and Hirschsprung's disease (2%) are occasionally identified in infants with anorectal malformations.

Diagnosis

Imperforate anus is a clinical diagnosis. Inspect the perineum for meconium and/or a perineal fistula (Figs. 79.2 and 79.3). Determine gluteal and gluteal cleft development. Determine the presence or absence of an anal pit and the extent of sphincter development by eliciting an anal wink. This can be done by gently scratching the perianal skin. Preoperative tests are aimed at determining the presence of a fistula, the location of the rectal terminus, and if there are any associated lesions. To determine the presence and location of a cutaneous fistula, observe the newborn for 24-48 hours. Meconium on the perineum confirms a cutaneous fistula and a low lesion (Figs. 79.4 and 79.5). If there is no meconium, place a radiopaque marker on the perineum and obtain a prone, cross-table lateral x-ray of the pelvis. Air, acting as a contrast medium, will delineate the rectal terminus and differentiate a low vs. high lesion. To determine the relationship of the urinary tract to the rectum, strain the urine with diaper or gauze and obtain a urinalysis. Meconium in the urine documents a communication between the bowel and the urinary tract. A voiding cystourethrogram (VCUG) will document the fistula. Passage of a nasogastric tube (esophageal atresia), cardiac echo (cardiac anomaly), abdominal ultrasound (renal agenesis), plain films (vertebral anomalies), spine ultrasound and lumbar magnetic resonance imaging (tethered cord) will rule out associated anomalies. Female infants with a single perineal opening, or cloaca, need urgent evaluation of the urinary tract.

Treatment

The infant is kept NPO, on peripheral hyperalimentation and antibiotics until colostomy or primary repair is performed. Infants with a cutaneous fistula, anal stenosis, or anal membrane undergo a minimal posterior sagittal anorectoplasty (PSARP) or a transposition anoplasty (Pott's anoplasty). Infants with a flat bottom, meconium in the urine, or other fistula (i.e., urethral, vaginal, vestibular) undergo...
Fig. 79.1. High imperforate anus in a male. Median raphe is flat and without any signs of meconium extrusion.

Fig. 79.2. Young female with anterior ectopic anus. Arrows mark the posterior edge of the vaginal opening and the anus. These two structures are too close, and the anal opening lies outside the anal dimple.
Fig. 79.3. Low imperforate anus in a female with fistula visible at the posterior fourchette (vestibular fistula).

Fig. 79.4. Low imperforate anus in a male. Well developed raphe that will probably demonstrate fistula with meconium extrusion over first 1-2 days of life.
colostomy. Females with a cloaca also undergo colostomy, and if necessary, vaginostomy and/or urinary diversion. Postoperatively, the mucous fistula is irrigated (to remove impacted meconium) and a distal colostogram is performed to show the distal rectum and fistula (Fig. 79.6). The child is then followed closely to insure weight gain and adequate colostomy function. If all goes well, a PSARP is performed between 2 and 12 months of age.

Throughout the operation, an electrical stimulator is used to determine the exact location of the external sphincter and insure that dissection stays in the midline. The muscle complex and levators are divided, the rectum is identified and opened, and the fistula is identified. Because there is no dissection plane between the urethra and rectum at the fistula, a small portion of the anterior wall of the rectum is left on the urethra, and the rectum is separated from the urethra superiorly. The fistula is closed; the rectum is mobilized and pulled down to the perianal skin. The perineal body is reconstructed anteriorly and the rectum is secured within the muscle complex fibers. The anoplasty is completed with interrupted absorbable suture.

The first morning following the reconstruction, the child is fed and converted to oral pain medication. The Foley catheter and IV antibiotics are continued for five days. The child is discharged after spontaneous voiding. At two weeks, the anus is sized with a Hagar dilator in the office and the mother/caregiver is instructed on home dilatations. The colostomy may be closed six weeks later.
Outcomes

Results depend upon the level of the lesion and the sacrum. In general, infants with low lesions have an excellent outcome with constipation (40%), soiling (13%), and diarrhea (4%) accounting for the majority of complications. Infants with high lesions have a higher incidence of these complications: constipation (35%), soiling (54%), diarrhea (12%).

Selected Readings