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Imperforate Anus: A Review

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ABSTRACT

Imperforate anus is a birth defect where the opening to the anus is missing or blocked. The anus, also known as the rectum, is the opening at the end of the intestines through which stool leaves the body. Imperforate anus may end in a pouch, be too narrow, or open into part of the urinary system, female or male reproductive system, or other system of body. Symptoms may include absence of the first stool within 24 to 48 hours after birth, no anal opening, anal opening in an abnormal place, stool coming out from the vagina, base of penis, scrotum, or urethra, and/or swollen belly. Although the exact cause of imperforate anus is not fully understood, it is believed to be due to the abnormal development of the rectum when the embryo is forming inside the womb. Many forms of imperforate anus occur with other birth defects. Imperforate anus may also be part of a syndrome with multiple birth defects. Treatment may include colostomy and surgery to correct the defect. Prognosis depends on the severity and type of imperforate anus and the severity and type of any other birth defects.

INTRODUCTION

Imperforate anus is a congenital abnormality with a lack of an anal opening of proper size and location. Anorectal malformations include a wide spectrum of defects in the development of the lowest portion of the intestinal and urogenital tracts. Anorectal malformations are a spectrum of different congenital anomalies which vary from fairly minor lesions to complex anomalies.[1] Many children with these malformations are said to have an imperforate anus because they have no opening where the anus should be. The term imperforate anus is a misnomer that is commonly used to refer to a spectrum of anorectal malformations, ranging from a benign defect that requires a minor operation. Imperforate anus has an estimated incidence of 1 in 5000 births.[1,2] It affects boys and girls with similar frequency.[3] However, imperforate anus will present as the low version 90% of the time in females and 50% of the time in males. Since the mid-1980s, significant advances have been achieved in the field of pediatric surgery that allow for a better anatomic reconstruction of these defects, preserving other important pelvic structures. In 7th century Byzantine physician Paulus Aegineta described a surgical treatment for imperforate anus for the first time.[4] Yet, at least 30% of all patients born with these defects still have fecal incontinence even after a technically correct surgical repair, and another 30% have other functional defecation problems, mainly constipation and varying degrees of soiling.

PATHOPHYSIOLOGY

The embryogenesis of these malformations remains unclear. In 8% of patients, genetic factors are clearly associated with anorectal malformations.[5] The rectum and anus are believed to develop from the dorsal potion of the hindgut or cloacal cavity when lateral in growth of the mesenchyme forms the urorectal septum in the midline. This septum separates the rectum and anal canal dorsally from the bladder and urethra. The cloacal duct is a small communication between the two portions of the hindgut. Down growth of the urorectal septum is believed to close this duct by seven weeks' gestation. During this time, the ventral urogenital portion acquires an external opening; the dorsal anal membrane opens later. The anus develops by a fusion of the anal tubercles and an external invagination known as the proctodeum, which deepens toward the rectum but is separated from it by the anal membrane. This separating membrane should disintegrate at eight weeks' gestation.

CAUSES

Although the precise embryologic defect that causes anorectal malformations has not been determined, cloacal membrane formation and subsequent breakdown into urogenital and anal openings should occur by eight weeks' gestation. Defects in the formation or shape of the posterior urorectal septum account for many of the described abnormalities of imperforate anus. Mullerian ducts appear after this critical period; how they are incorporated into this development is unclear. No known risk factors predispose a person to have a child with imperforate anus. Which should be clearly emphasized to parents of affected children who may harbor feelings of guilt? A genetic linkage is sometimes present. Most cases of imperforate anus are sporadic without a family history of the condition, but some families have several children with malformations. Genetic studies are ongoing.

DIAGNOSIS

When an infant is born with an anorectal malformation, it is usually detected quickly as it is a very obvious defect. Doctors will then determine the type of birth defect the child was born with and whether or not there are any associated malformations. It is important to determine the presence of any associated defects during the newborn period in order to treat them early and avoid further sequelae. There are two main categories of anorectal malformations: those that require a protective colostomy and those that do not. The decision to open a colostomy is usually taken within the first 24 hours of birth. Sonography can be used to determine the type of imperforate anus.[6]

TREATMENT

Newborns with imperforate anus should not be fed and should receive intravenous hydration. If a urinary fistula is suspected, broad-spectrum antibiotics can be administered, although anaerobic coverage is unnecessary within the first 48 hours of life. Any cardiac murmurs identified upon physical examination should be evaluated using echocardiography prior to surgical intervention. Imperforate anus usually requires immediate surgery to open a passage for feces unless a fistula can be relied on until corrective surgery takes place. Depending on the severity of the imperforate, it is treated either with a perineal anoplasty [7] or with a colostomy. Children with anorectal malformations may undergo one or several of the following surgical procedures based on the child's presentation, physical examination findings, and imaging study findings.

Neonatal colostomy- A colostomy is performed in children who are not amenable to primary pull-through either because of malformation complexity or associated comorbidity. The colostomy is usually fashioned through a left lower quadrant incision. The colon is divided at the point where the descending colon meets the sigmoid colon, and both ends are brought to the abdominal wall. By fashioning the colostomy at this location, the entire sigmoid colon is kept in place; thus, when the pull-through is eventually performed, a large portion of the colon is available for the surgeon to bring down to the perineal skin. The mucous fistula (the downstream segment) should be very small, flush with the skin, and far enough from the proximal end to be outside the colostomy appliance (or under the flange) to avoid continued urinary soiling with feces. During this operation, the distal segment of the colon must be exhaustively irrigated to clean out the impacted meconium, which is always significant. This prevents post diversion urinary sepsis and allows for effective distal colostography.

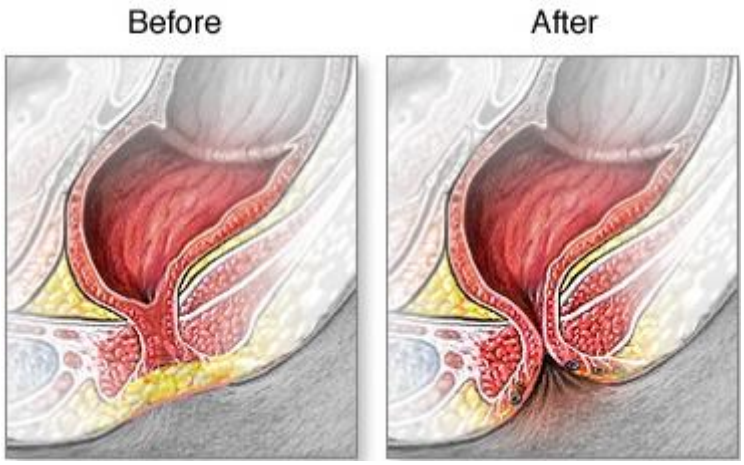

Figure No. 01: Neonatal colostomy

Primary neonatal pull-through without colostomy- Many pediatric surgeons opt for primary pull-through in children with perineal fistulas (or no fistulas) and close (< 1 cm) rectal pouches on 24-hour lateral pelvic radiography. Some pediatric surgeons who specialize in colorectal problems often offer the same procedure for girls with vestibular fistulas. Cystoscopy is usually performed to rule out associated malformation. This is performed immediately prior to the pull-through operation. A Foley catheter is inserted following the cystoscopy.

Posterior sagittal pull-through with colostomy- This approach is used in boys with rectourinary fistula (bulbar, prostatic, or bladder-neck fistula), in girls with cloaca or vestibular fistula, and in patients of either sex who do not have a fistula when the rectal pouch

is further than 1 cm on 24-hour lateral prone abdominal radiography. The approach is also used in children who may have malformations that were amenable to primary neonatal pull-through but were unable to undergo such a procedure because of extreme prematurity or other comorbidity.

Medication- Many children with anorectal malformations require medications for various reasons. Beyond perioperative medications, maintenance medications often include urinary antibiotic prophylaxis or treatment and/or laxatives. Urinary prophylaxis is used to mitigate the risk of urinary infection and urosepsis in children with risk factors for urinary infection such as urinary fistula, vesicoureteral reflux, or continent diversion. Common agents include oral amoxicillin, oral trimethoprim/sulfamethoxazole and gentamicin bladder irrigations. Common laxatives include senna products, milk of magnesia, and propylene glycol solutions.

Diet- After the obstruction is relieved using colostomy; primary pull-through, or dilation, children do not require special diet. The most common complication of imperforate anus repair is constipation or anal incontinence; therefore, diet can be a crucial part of management. Many patients may require laxatives, enemas, or other medications or irrigations in addition to dietary manipulations.[8] Children should avoid constipating foods, such as those included in the bananas, rice, applesauce, and toast (BRAT) diet. High-fiber and laxative foods (whole-grain foods and breads, dairy, fruits, vegetables, greasy foods, spicy foods) should be encouraged. Unfortunately, dietary manipulation is often of limited effectiveness because of the fussy nature of most children regarding diets. Fiber supplements and laxatives can be critically important in avoiding constipation, which can significantly affect prognosis.

CONCLUSION

In individuals with a normal anatomy, the large intestine (colon) empties into a pouch-like portion of bowel (rectum). Through complex nerve and muscle structures, the rectum releases stool through the anus out of the body. Imperforate anus is a malformation of the anorectal region that may occur in several forms. The rectum may end in a blind pouch that does not connect with the colon, or it may have openings to the urethra, bladder, or vagina. A condition of stenosis or narrowing of the anus or absence of the anus may be present.

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