

Anorectal Malformations

BIRTH DEFECT RESEARCH FOR CHILDREN



What are Anorectal Malformations?

Anorectal malformations are a group of birth defects that involve the external opening of the anus preventing the normal passage of stool. The anal opening may be smaller than normal size; in an abnormal location or not visible upon examination.



Anorectal Malformations



There are many types of anorectal malformations. Some of the more common types include:

Anal Stenosis / Atresia - a narrowing or complete absence (closure) of the anal opening.

Anoperineal / Rectoperineal fistula - a small tube-like opening that creates an abnormal passageway between the colon and the anal opening.

Ectopic Anus - an anal opening that is not in the normal place.

Perineal anus - an anal opening within the perineum (space between anus and genital region).

Imperforate anus - the anal opening is completely closed or narrowed to only a small passage. A complete imperforate anus is often associated with atresia (absence) of the lower rectum.

“high type” - the rectum ends above pelvic nerve and muscle structures.

“low type” - the rectum extends completely or partially through the pelvic nerve and muscle structures.

How Many Children Have Anorectal Malformations?

One out of every 5,000 babies is born with an anorectal malformation. All racial, social, ethnic, and economic groups are affected equally. However, males are affected more often than females.

What Causes Anorectal Malformations?

No definite cause for anorectal malformations has been established. Some studies have suggested a possibility of autosomal recessive inheritance in a small number of families.

Are There Associated Findings?

Additional birth defects occur in 48% of all patients with anorectal malformations. The most common associated findings are:

skeletal - genitourinary
esophageal atresia - tracheoesophageal fistula
CNS - cardiovascular
VATER association

Helping A Child With An Anorectal Malformation

Diagnosis is usually made shortly after birth when the newborn is given a routine physical exam.

Treatment involves surgical reconstruction of the anus. If the rectum connects with other organs, repair of these organs will also be necessary.

Surgery for infants with imperforate anus depends upon the severity of the condition and involves creating an opening for passage of stool. The baby is put into a deep sleep by general anesthesia during surgery.

A low imperforate anus can be repaired in the newborn period by a procedure called a perineal anoplasty.

A high type imperforate anus surgery involves a colostomy (temporary opening of the large intestine into the abdomen to allow passage of stool). Then, the infant is given time to grow until definitive repair can be done with a pull-through operation in which the rectum is “pulled down” and sewn into a newly-made anal opening in the perineum. After surgery, the newly-formed anus needs to be dilated regularly for several months until a soft, mature scar is obtained. The colostomy can then be closed.



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Risks for any anesthesia are breathing problems and reactions to medications. Other risks include damage to supporting muscles in the pelvis or the urethra, and temporary postoperative paralysis of the bowel.

Prognosis following a successful surgery is usually good. Normal bowel function can be established in most cases. The long-term prognosis is excellent if no other pelvic abnormalities are found.

Be sure to call your health care provider if a child treated for imperforate anus develops abdominal pain or fails to develop bowel control by the age of three.

The cost of any surgery varies significantly between surgeons, medical facilities, and regions of the country. Children who need more extensive surgery will require more expensive treatment. Examples of estimated costs are:

- 1) Surgeon's fee: avg. \$1,500-\$3,000
- 2) Anesthesiologist's fee: avg. \$350-\$400/hour
- 3) Hospital charges: basic rate \$1,500-\$1,800 (ICU and private rooms more)
- 4) Medication charges: \$200-\$400
- 5) Additional charges: assistant surgeon, diagnostic procedures, and complications.

Fact Sheet by:

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