Anorectal Malformations

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KEYWORDS

• Anorectal malformation • Imperforate anus • Perineal fistula
• Neonatal bowel obstruction • VACTERL association • Congenital anomaly

KEY POINTS

- Workup of patients with anorectal malformations should begin with a thorough physical examination.
- Ruling out significant associated anomalies is necessary before initial surgical intervention.
- The type of lesions, high versus low, has a significant impact on continence and long-term outcomes.
- Associated gynecologic problems are not uncommon and may not be picked up until puberty or later.

HISTORY

The first description of humans born with anorectal malformations (ARM) was described by Aristotle in the early third century BCE, and the earliest reports of treatment for such defects began to appear in the second century CE by Soranus, who described cutting the thin perineal membrane and then dilating it. There was little progress in the treatment or description of ARM until the late seventeenth century, when Saviard attempted to treat a high lesion by plunging a trocar through the perineum. About a century later the first dissection to a blind-ending colon was described by Bell, who also described the associated anatomic defects of rectovesical and rectovaginal fistulas.[1] However, it is likely that during this time most of these infants died, and surgeons were unable to achieve successful treatment. In the late 1700s inguinal colostomies were first reported; however, most of these procedures resulted in death and this procedure was thus used only as a final option.[2] In 1835 Amussat[3] performed the first proctoplasy, but destroyed most of the sphincter complex during the procedure. From the mid-1900s to the early 1980s passage of the rectum through a puborectalis ring was used, as described by Stephens.[4] A potential disadvantage of this

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approach was that it involved a blind dissection around the urethra and prostate gland. The posterior sagittal anorectoplasty was described by DeVries and Peña[5] in 1982, and is still used today. In 2000 Georgeson and colleagues[6] described the laparoscopic approach to imperforate anus.

EPIDEMIOLOGY

Anorectal malformations are one of the causes of bowel obstruction in the neonate. However, the incidence is rare, occurring in about 1 in every 4000 to 5000 live births, with about one-third being isolated and the remainder associated with other congenital abnormalities.[7] There is a slight male predominance and certain familial associations have been described[8,9]; however, there has been no discernible identifier regarding a possible cause. In addition, ARM have been associated with various genetic conditions (see later discussion). The most common lesion seen in males is a rectourethral fistula, with the most common in females being a rectovestibular fistula.

EMBRYOLOGY

Anorectal malformations occur secondary to abnormalities in the development of the hindgut, which gives rise to the descending colon, rectum, and the upper part of the anal canal, the lining of bladder, and the urethra. The hindgut will enter into the posterior region of the cloaca. The fetal cloaca is an endoderm-lined cavity covered at its ventral portion by ectoderm. The junction of the endoderm and ectoderm is termed the cloacal membrane. During development the urorectal septum will divide the area between the hindgut and the allantois, ultimately forming the perineal body. The allantois is an endodermal outpouching of the yolk sac, with its specialized mesenchyme that lies close to the cloaca and becomes the umbilical cord. During the seventh week of development the cloacal membrane separates, creating the anal opening for the hindgut and ventral opening for the urogenital sinus, with the perineal body forming between the two. The posterior aspect closes with ectoderm and then is recanalized 2 weeks later. An evolving theory on the etiology of these defects has occurred, with current thinking being that the development of imperforate anus is caused by lack of recanalization during the ninth week of gestation and ectopic positioning of the anal opening in the cloaca. The extent of anorectal defects relates to the degree of development in the posterior aspect of the cloaca. Smaller defects will lead to distal presentations such as covered anus and anocutaneous fistula, whereas larger defects will lead to more proximal anomalies such as urogenital fistula or cloacal malformations.[10,11]

GENETICS

In addition to the multitude of genetic syndromes that have been associated with ARM, recent investigations have evaluated possible genetic causes of ARM. Overall, the incidence of anorectal malformations in the setting of a genetic disease is about 5% to 10%. Trisomy 21 and a microdeletion of the chromosome 22q11.2 are the 2 most frequent. However, anorectal malformations have been reported in association with mutations in almost all chromosomes.[12] Towne-Brock syndrome, FG syndrome (Opitz-Kaveggia syndrome), Johanson-Blizzard syndrome, Kaufman-McKusick syndrome, Trisomies 8 and 21, and Fragile X syndrome are some of the genetic syndromes that have been shown to have associated ARM.[11] Trisomy 21 differs from most of the other genetic diseases in that the associated ARM is usually without an associated fistula. In addition to genetic disorders, recent literature has begun to