Magnetic Resonance Imaging of Anorectal Malformations

Daniel J. Podberesky, MD[a,*], Alexander J. Towbin, MD[a], Mohamed A. Eltomey, MD[b], Marc A. Levitt, MD[c]

KEYWORDS

• Anorectal malformation • Cloaca • Cloacal malformation • MR imaging • Children

KEY POINTS

- Anorectal malformation (ARM) comprises a diverse spectrum of congenital malformations of the anus and rectum with an incidence of approximately 1 in 5000 newborns.
- Fetal MR imaging plays an increasingly important role for ARM patients; it helps in providing appropriate parental counseling and enables proper antenatal and postnatal management.
- With the advent of faster imaging sequences and more advanced imaging techniques, MR imaging is increasingly being relied on to aid definitive surgical correction planning.
- MR imaging is increasingly being called on to aid in the evaluation of postoperative complications following the original corrective surgery in ARM patients.
- MR imaging has the potential to serve as an ionizing radiation-free, one-stop shop for the imaging evaluation of ARM patients

INTRODUCTION

Anorectal malformation (ARM) comprises a diverse spectrum of congenital malformations of the anus and rectum. These malformations can range in severity from minor and easily treated with excellent prognosis, such as rectoperineal or rectovestibular fistulae, to those that are complex and difficult to manage with relatively poor prognosis, such as cloacal malformation and caudal regression syndrome. Overall, congenital ARM affects approximately 1 in 5000 newborns, with a slight male predominance.[1] The incidence of cloacal malformations has frequently been reported in the literature as approximately 1 in 40,000 to 50,000 newborns,[2] although the incidence is likely more frequent (approximately 1 in 20,000) because many of these patients were previously erroneously diagnosed with a rectovaginal fistula.[3]

> Anomalies of the gastrointestinal, genitourinary, skeletal, nervous, and cardiovascular systems are frequently associated with ARM.

Associated anomalies are frequently found in patients with congenital ARM. Although all organ systems can be affected, abnormalities of the gastrointestinal, genitourinary, skeletal, nervous, and cardiovascular systems are most common.[4] Anomalies of the gastrointestinal tract outside of

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[a] Department of Radiology, Cincinnati Children's Hospital Medical Center, University of Cincinnati College of Medicine, 3333 Burnet Avenue, MLC 5031, Cincinnati, OH 45229, USA; [b] Department of Radiology and Medical Imaging, Tanta University, Elbahr Street, Medical Compound, Tanta 31111, Egypt; [c] Department of Surgery, Colorectal Center, Cincinnati Children's Hospital Medical Center, University of Cincinnati College of Medicine, 3333 Burnet Avenue, MLC 2023, Cincinnati, OH 45229, USA
* Corresponding author.
E-mail address: daniel.podberesky@cchmc.org

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the primary ARM include esophageal atresia and duodenal atresia. Anomalies of the genitourinary tract include but are not limited to absent, dysplastic, or horseshoe kidney; hypospadias; bifid scrotum; vesicoureteral reflux and hydroureteronephrosis; undescended testes; vaginal abnormalities; and Müllerian structure anomalies. Common skeletal abnormalities include sacral agenesis or dysgenesis, spinal dysraphism, vertebral segmentation and fusion anomalies, and scoliosis. Specific spine abnormalities encountered in ARM patients include tethered spinal cord, meningoceles and myelomeningoceles, intradural lipomas, and diastematomyelia. Cardiovascular anomalies are present in 12% to 22% of ARM patients, with tetralogy of Fallot and ventricular septal defect most commonly encountered.[4]

Three specific associations typically encountered with congenital ARM are the Currarino triad; caudal regression syndrome; and the syndrome of vertebral defects, anorectal anomalies or atresia, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb defects (VACTERL). In 1971, Currarino and colleagues[5] first described the triad of ARM, partial agenesis of the sacrum (typically a sickle-shaped sacrum), and presacral mass (typically a teratoma or anterior meningocele). The triad is inherited in an autosomal dominant pattern secondary to a mutation in the HLXB9 homeobox gene.[6]

Caudal regression syndrome is a rare disorder that affects the lower half of the body to varying degrees, including the lower extremities, low back or spine, genitourinary tract, and gastrointestinal tract, including ARM (typically imperforate anus). Approximately 15% to 25% of cases of caudal regression syndrome occur in children of a diabetic mother.[7]

VACTERL is a nonrandom cluster of a group of congenital anomalies. Typically, at least three malformations are required to be diagnosed with the association. The VACTERL association occurs in approximately 1 in 10,000 to 1 in 40,000 newborns, with approximately 55% to 90% of these patients having an ARM.[8]

Currarino triad = ARM, partial agenesis of the sacrum, and presacral mass.

VACTERL = vertebral defects, anorectal anomalies, cardiac defects, tracheoesophageal fistula, renal anomalies, and limb defects.

As with most complex congenital malformations, imaging has long played a critical role in the evaluation and management of patients with ARM. From the first invertogram radiograph described by Wangensteen and Rice[9] in 1930, to contrast fluoroscopic examinations such as contrast enemas, distal high-pressure colostograms, and voiding cystourethrograms, to cross-sectional modalities of today such as ultrasound, CT, and (most recently) MR imaging, diagnostic imaging helps provide the surgeon with the information needed to correct the malformation. To accomplish a successful postoperative outcome, an accurate preoperative imaging assessment is required. This includes assessment of the level and type of malformation, the presence of a fistula, the developmental state of the sphincter muscle complex, and the presence of associated anomalies. In the postoperative patient, accurate imaging is required for identification of postoperative complications, potential reoperative planning, other associated anomalies that may have initially been inconspicuous, and predicting morbidity and quality of life. MR imaging is ideally suited to fulfill these requirements because of its lack of ionizing radiation, excellent intrinsic contrast resolution, and multiplanar imaging capabilities. Disadvantages of MR imaging in ARM patients include the frequent need for sedation, relative high cost, and relative lack of expertise and access to the technique. Despite these disadvantages, MR imaging is increasingly being used by radiologists in the diagnostic work-up of ARM patients, including in the prenatal state, before definitive surgical repair, and postoperatively.

EMBRYOLOGY, CLASSIFICATION, AND ANATOMY

A basic understanding of the normal and pathologic embryology and anatomy of the anorectum, particularly the sphincter mechanism, is helpful when interpreting MR imaging studies in the ARM patient (**Fig. 1**). The process of normal and abnormal development of the hindgut is not fully understood, although various theories have been offered over the years.[10–15] Cranially, the hindgut is in continuity with the midgut; caudally, it is in direct contact with the ectoderm, thus forming the cloacal membrane. When development progresses, the caudal part of the hindgut, the cloaca, differentiates into two separate organ systems, the urogenital tract and the anorectal tract.[16] Normal anorectal and genitourinary development depends on the normal development of the dorsal cloaca and the cloacal membrane, the latter having a crucial role in the pathogenesis of ARM.[17,18] Cloacal membrane defects are thought to also affect development of the genitourinary system and mesenchymal tissue leading to genital malformation and abnormal pelvic floor and sphincter muscle development.[16]