

Imperforate Anus With Renal Ectopia

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This case study describes the sonographic findings of an infant born with an imperforate anus. Imperforate anus occurs in about 1 in 4000 to 5000 live births. It can be associated with VACTERL or VATER syndrome, which includes vertebral, anal, cardiac, tracheoesophageal, renal, and limb malformations. Prognosis depends on the level of the imperforate anus and the extent of the associated anomalies.

Key words: imperforate anus, VACTERL syndrome, infant

Case Report

A diabetic mother delivered a term male infant who was admitted into the neonatal intensive care unit secondary to respiratory depression requiring intubation. Clinical assessment noted an anal dimple and chordee of the penis. Sonography, along with other diagnostic imaging tests, was performed to assess the anal dimple and to evaluate for anomalies associated with VACTERL or VATER syndrome. This syndrome includes vertebral, anal, cardiac, tracheoesophageal, renal, and limb malformations.¹ Three of these anomalies must be present to be characterized as VACTERL or VATER.²

A retroperitoneal sonogram demonstrated absence of a kidney in the right renal fossa. The left and right kidneys were both present within the left renal fossa and fused at the inferior poles. This finding was consistent with crossed fused renal ectopia (Figure 1). Sonographic assessment of the perineum demonstrated a 1.5-cm distance between the sac of the rectum and the anal dimple, indicating a high-level imperforate anus (Figure 2). A spinal sonogram visualized a small cyst at the tip

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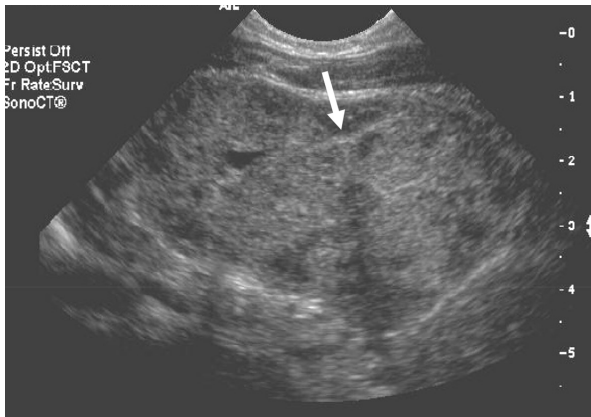


FIGURE 1. Transverse image of the left renal fossa demonstrating fused inferior poles of the left and right kidney. The arrow points to the interface between the right and left kidney.

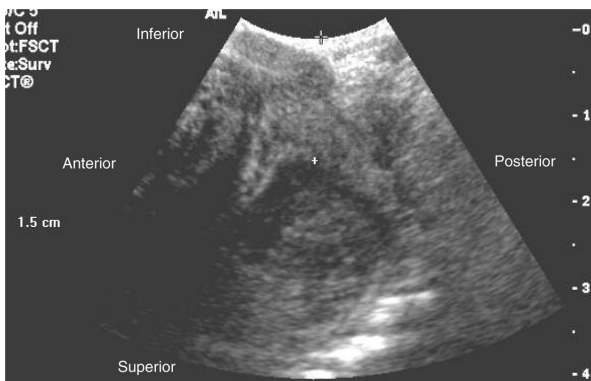


FIGURE 2. Perineal scan of the imperforate anus in the longitudinal plane. The cursors show a 1.5-cm distance between the sac of the rectum and the anal dimple on the skin.

of the spinal cord consistent with persistent ventriculus terminalis cyst (Figure 3). This is most likely a normal embryologic remnant.³ Spinal radiographs and an osseous survey found no vertebral or limb anomalies. The echocardiogram was also normal. A genetic consult was also obtained, but based on the imaging findings and clinical evaluation, VACTERL syndrome was ruled out, and chromosomal analysis was not recommended.

Three days postpartum, a colostomy was performed due to the imperforate anus. A barium enema performed postcolostomy demonstrated a rectourethral fistula from the distal rectal sac to the bulbar/proximal penile urethra (Figure 4). The infant was placed on antibiotics to prevent a urinary tract infection from the fistula. The patient was

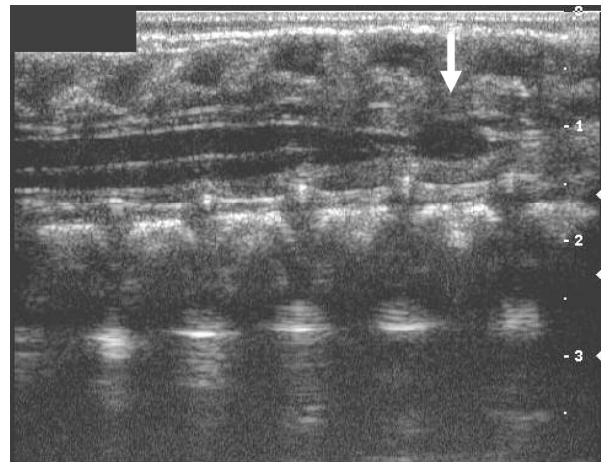


FIGURE 3. Longitudinal image of the spinal cord. Arrow points to a persistent ventricularis terminalis cyst at the tip of the spinal cord.

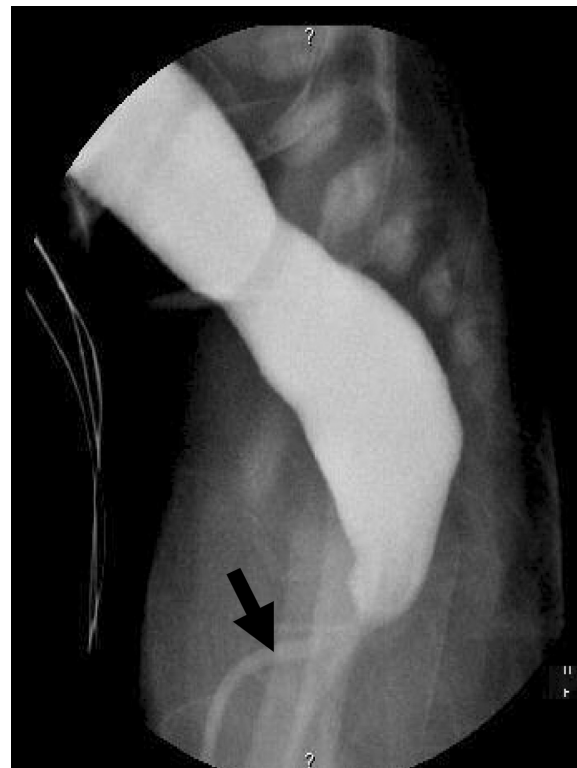


FIGURE 4. Lateral image of a barium enema. The arrow indicates a rectourethral fistula from the distal rectal sac to the bulbar/proximal penile urethra.

discharged with instructions to return to the pediatrician if the ostomy became red or inflamed or the infant developed a fever. Another barium enema is to be performed at a later date to determine if the

fistula spontaneously closed or if further surgical correction is needed.

Incidence and Classifications

Imperforate anus is a congenital anomaly that occurs in approximately 1 in 4000 to 5000 live births.⁴ Anorectal malformations are slightly more common in boys. The most common defect in both boys and girls is an imperforate anus with a fistula between the distal bowel and the urethra in boys or the vestibule of the vagina in girls.^{5,6} Classification of anorectal anomalies is based on the level at which the blind-ending rectal pouch ends relative to the levator ani musculature.⁵⁻⁸ High lesions end above the levator ani muscle and more commonly include the presence of a fistula.^{5,6} Intermediate lesions are characterized by the rectal pouch ending at the level of the levator with or without a fistula. In low lesions, the rectal pouch has completely traversed the levator musculature, and a fistula or an opening usually is evident on the skin at midline.^{5,6}

Embryology of Imperforate Anus

Imperforate anus is defined as the congenital absence of an anal opening due to the presence of a membranous septum (persistence of the cloacal membrane) or a complete absence of the anal canal.⁹ During the fourth to sixth week of gestation, the cloaca is a close-ended, expanded portion of the hindgut.¹⁰ As part of the urinary bladder formation and separation from the hindgut, the urorectal septum grows into and divides the cloaca into the urogenital sinus, which will separate into the future bladder and rectum. Failure of this process results in an imperforate anus. There can also be failure of the endodermally derived lining of the rectum to connect to the ectodermally derived lining of the anus. Most anorectal anomalies result from abnormal development of the urorectal septum, resulting in incomplete normal separation of the cloaca into urogenital and anorectal portions.¹⁰

Diagnosis

When evaluating an imperforate anus, it is important to determine where the distal end of the

hindgut terminates.^{7,11} There are pitfalls in attempting to accomplish this with plain radiographs, including those that are taken in the cross-table prone position.¹¹ For example, the colon can incorrectly appear to end in a high position if the air column fails to progress to the end of the colon because of impacted meconium.¹¹ Sonography can improve findings by directly visualizing the end of the hindgut pouch. In addition, the sacral level can be assessed with high-resolution, real-time sonography using an 8- to 12-MHz transducer in the sagittal plane through the anterior abdominal wall with the patient in a supine position. A perineal approach at the site of the anal dimple can also be obtained.^{2,7,11} With either approach, the distance between the skin surface of the perineum and the blind-ending rectum is measured. A distance of 1.0 cm or less between the pouch and the perineum suggests a low-type imperforate anus, a distance of 1.0 to 1.5 cm indicates an intermediate-type imperforate anus, and a distance of 1.5 cm or greater implies a high-type imperforate anus.^{7,12} A high-type imperforate anus can be mistaken for a low type if the sonogram is performed while the patient is crying because this displaces the pouch caudally.⁷ Gentle scanning pressure should be used to avoid compressing the area of interest.

Fluoroscopy is frequently used to rule out fistulas in patients with imperforate anus. A voiding cystourethrogram may be performed by inserting a catheter and filling the bladder with contrast. When draining the contrast from the bladder, fluoroscopy is able to view a fistulous connection between the urethra and bladder.

A colostomy barium enema may also visualize a fistula when an irrigation tip is inserted into the stoma and barium flows into the large bowel.^{13,14} In this particular case, barium was not only visualized in the bowel but also in the urethra, which was consistent with a rectourethral fistula. After these procedures are performed and the diagnosis is made, a decision can be made on the type of surgery to be performed.

Surgical Management

The surgical approach is determined by the position of the rectal pouch in relation to the levator

sling.⁵ Low lesions may have a primary, single-stage repair procedure on the perineum without need for a colostomy.⁵ Three approaches may be used depending on the severity. For anal stenosis with a normal location of the anal opening, simple dilatation is performed daily for several months.^{5,6} If there is a small distance between the anal opening and the center of the external sphincter, a cutback anoplasty is performed.⁴⁻⁷ The more severe low lesions may require the abnormal anal opening to be repositioned and the perineal body reconstructed.^{5,6,15}

Infants with intermediate or high lesions require a colostomy as the first part of a three-stage reconstruction. The second-stage procedure usually is performed three to six months later. It consists of correcting the fistula, repositioning the rectum, and reconstructing the perineal musculature. The third and final stage is performed a few months after the second stage, and it consists of colostomy closure.^{4,5} Anal dilatations continue for several months after the colostomy closure.⁵

Prognosis

The major goal with an imperforate anus is to correct the defect and achieve fecal continence. Success is dependent on the level of the rectal pouch and the normalcy of the sacrum. The best results are seen in patients with low lesions and a normal sacrum, with incontinence reported to occur in up to 40% of patients at long-term follow-up.⁵ In patients with intermediate lesions and a normal sacrum, soiling was reported to occur in 50% to 75%.⁵ In high lesions, the rate of incontinence is nearly 100%.⁵ Patients who have continued problems with either constipation or soiling may begin a bowel-training program with daily enemas to keep the lower rectum decompressed while controlling the need to defecate.⁵ Success or failure in achieving continence cannot be judged until after age 10 years.

Overall prognosis is also affected by the number of other anomalies. Imperforate anus is associated with VATER or VACTERL syndrome.^{1,4} This acronym refers to a combination of several anomalies: vertebral, anal, cardiac, tracheo-esophageal, renal, and limb. The more common VATER or

VACTERL findings include V (caudal regression, scoliosis, and hemivertebrae), A (anal atresia), C (ventral septal defect), TE (tracheo-esophageal fistula), R (hydronephrosis, unilateral polycystic kidneys, or renal ectopia), and L (polydactyly, underdeveloped or absent radius). At least three of these should be present for a diagnosis of this disorder.⁸

The patient in this case study was found only to have a renal anomaly and crossed fused ectopia, along with the imperforate anus. Cross-fused ectopia results from fusion of metanephrogenic blastema during embryologic development. The kidneys will be located on the same side and joined at any location.¹⁶ The ventriculus terminalis cyst of the spinal cord was not diagnosed as a vertebral anomaly. This small cyst, located at the distal end of the spinal cord, is considered a residual lumen of the conus medullaris and a normal anatomic variant when no other spinal pathology is found.³

Conclusion

Anorectal anomalies result from abnormal development of the urorectal septum. Classifications of anorectal anomalies are based on the level at which the blind-ending rectal pouch ends relative to the levator ani musculature. Imperforate anus is associated with the VATER or VACTERL syndrome. Although this patient had high-level imperforate anus along with renal ectopia, VACTERL syndrome was not diagnosed.

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