19.2 Complex Urogenital and Anorectal Malformations

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PERSISTENT CLOACA

Cloacal malformations represent a spectrum of developmental defects in which the urinary tract, the vagina, and the rectum converge above the level of the perineum, creating a single perineal opening, the cloaca (Latin for "sewer") (Fig. 19.2-1).[1,2] Because of its prognostic and therapeutic implications, Peña[3] and Peña et al.[4] subdivided these anomalies into two major subgroups based on the length of the common channel. Cloacal malformations with a short common channel (less than 3 cm) have a lower incidence of associated defects and less difficult repair. Cloacal malformations with a long common channel (longer than 3 cm) have a high incidence of associated defects, complex anatomy, more difficult repair, and inferior functional results.

Incidence: The condition is seen typically in females with an incidence of 1 in 40,000 to 50,000 births.

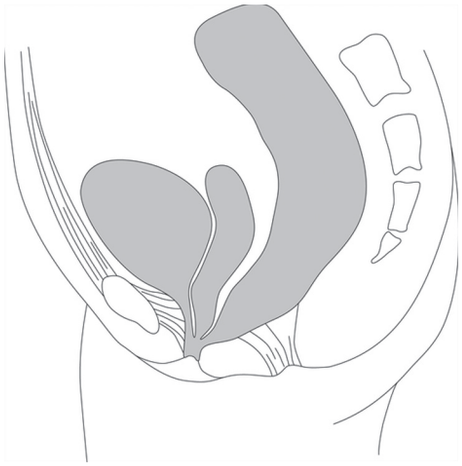

FIGURE 19.2-1: Diagram of a cloaca. Sagittal view of the pelvis. The rectum, vagina, and urethra are fused together, forming a single common channel opened to the perineum.

Genetics and Embryology: The most widely accepted theory is that cloacal malformations are related to failure in development of the urorectal septum. This is a structure that normally fuses with the cloacal membrane by the 6th week of gestation and subdivides the primitive cloaca into the urogenital sinus in front and the rectum posteriorly (Fig. 19.2-2). Development of these structures may be arrested at any stage, leading to a wide spectrum of defects unified under the term urorectal septum malformation sequence (URSM) (Fig. 19.2-3).[5,6] In this classification, persistent cloaca are termed partial URSM.

The aberrant drainage of urine can lead to bladder obstruction, hydrocolpos, and colonic dilatation. It is also thought that abnormalities in cloacal septation and urogenital sinus formation could interfere with normal mesonephric and paramesonephric duct development, explaining the high incidence of genital tract duplication or agenesis and increased anomalies of number and position of the kidneys in these patients. Associated lower spinal cord, lumbosacral spine, lower limbs, and bladder anomalies as well as ambiguous genitalia are also suggesting multiple disturbances during the embryonic development of the caudal pole.[2,7]

Etiology: The etiology is unknown. There are several case reports in monozygotic twins, in association with maternal drug abuse and one reported case of apparent genetic transmission.[8,9]

Diagnosis: Ultrasound diagnosis of cloacal malformations is challenging and usually considered when a pelvic cyst is seen in a female fetus (Fig. 19.2-4). In this situation, a differential diagnosis should include hydrocolpos or megacystis in the setting of cloaca, isolated hydrocolpos, urogenital sinus, obstructive uropathy, and other pelvic cystic lesions such as ovarian cyst, enteric duplication cyst, bowel atresia, cystic type IV sacrococcygeal teratoma, and anterior meningocele.[10,11] In addition, the sequence of findings on serial sonograms of transient ascites (from escape of urine via the fallopian tubes), progressive hydrocolpos, hydronephrosis, oligohydramnios with or without development of meconium peritonitis and peritoneal calcifications has been described as suggestive of this malformation, but is not clearly specific.[8,12–14]

The most specific sonographic signs supporting this diagnosis and the communication between the urinary system and the distal bowel in a female fetus are rectal fluid dilatation with intraluminal calcifications[8,13,15] and the detection of calcified meconium in the urinary system.[16] Unfortunately, these signs are not always present or easily detected.

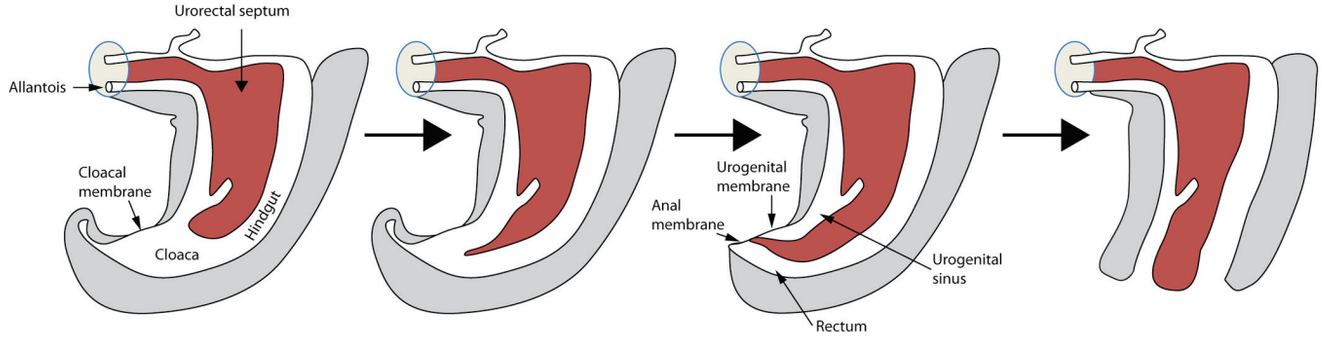

FIGURE 19.2-2: Normal development of the urorectal septum. The urorectal septum grows in a caudal direction to separate the cloaca into the dorsal rectum and the ventral urogenital sinus. The urorectal septum also divides the cloacal membrane into the urogenital and anal membranes. These structures will grow caudal, so they will be no longer be part of the ventral wall. Both membranes will subsequently rupture.

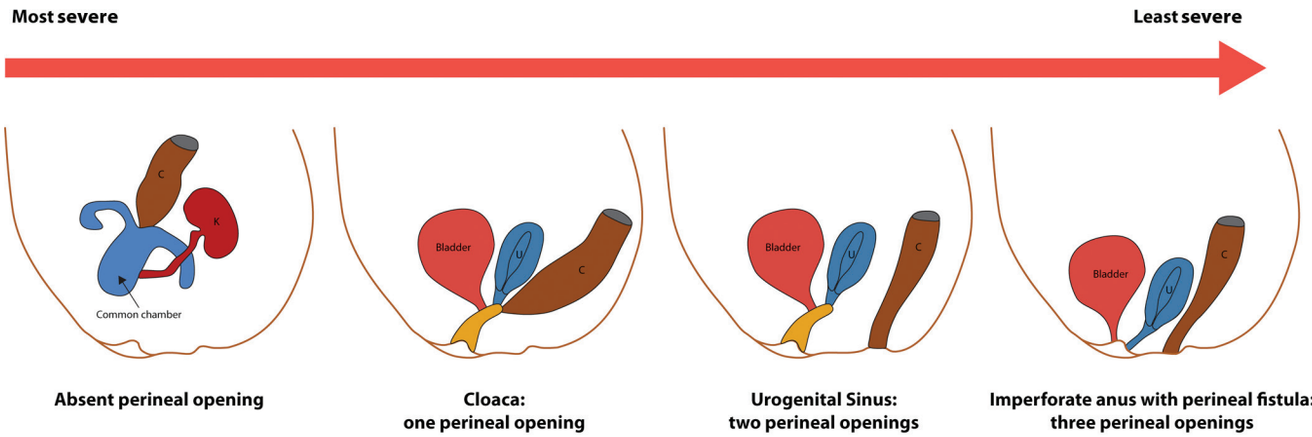

FIGURE 19.2-3: Spectrum of abnormalities related to the urorectal septum. Variations of these forms may also occur. C, colon; K, kidney; U, uterus.

In recent years, several authors have defined the normal appearance of the colon with fetal MRI. Starting around 20 to 21 weeks of gestation, meconium is expected to fill the rectum, and by 26 weeks, much of the colon is visualized with its characteristic bright T1 and dark T2 signal. The rectum is close to the bladder and its cul-de-sac at least 10 mm below the bladder neck.[11,17,18] MRI provides a resource to assess the rectum and its content and differentiate isolated hydrocolpos and urogenital sinus from cloacal malformation. Isolated hydrocolpos and urogenital sinus will present with a normal rectum.[19,20] Cloaca can have fluid signal instead of meconium in the rectum and/or rectal dilatation located high in the cul-de-sac, not extending below the bladder base. Content signal ranges from normal meconium to increased fluid signal in a series of long common channel cloacas. In addition, abnormal signal can be detected layering withing the bladder due to the presence of meconium.[21]

Differential Diagnosis: The differential diagnosis includes other etiologies presenting with megacystis and/or hydrocolpos. Megacystis can be seen in the setting of

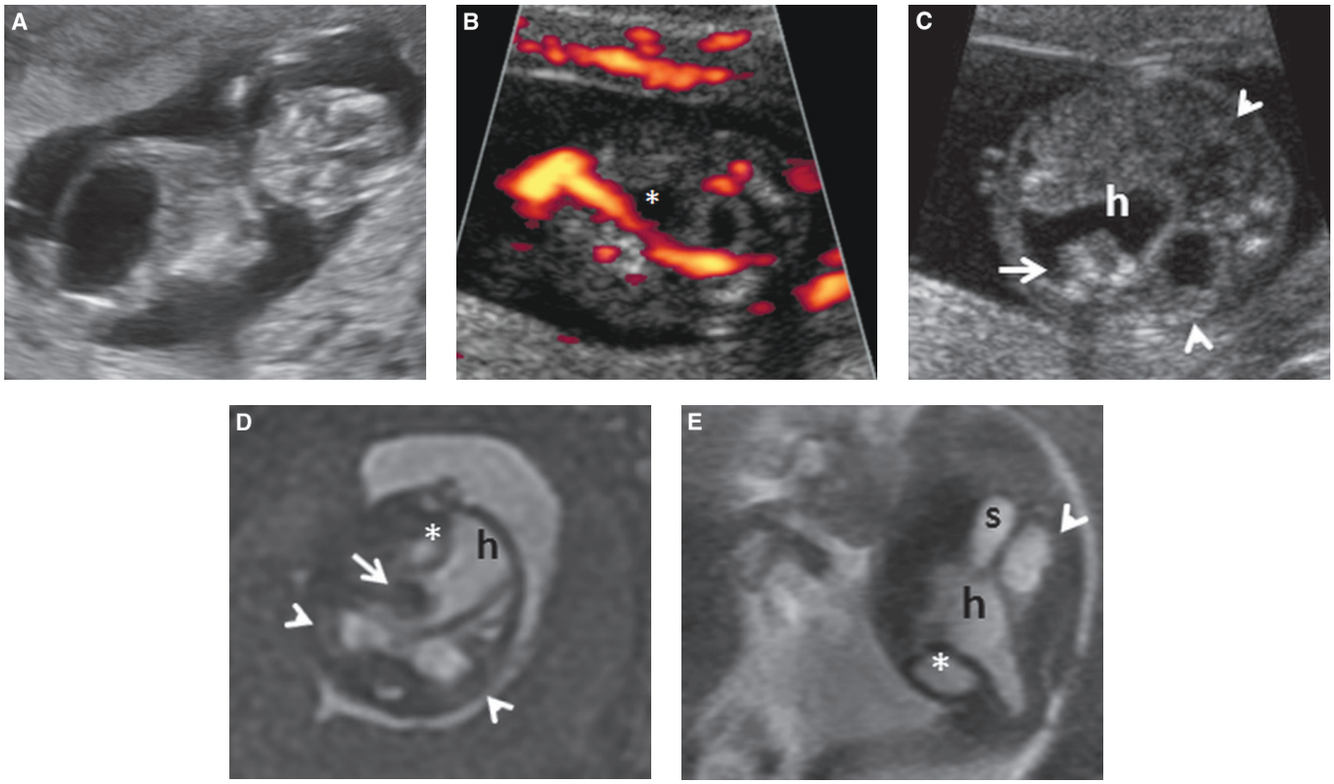

FIGURE 19.2-4: Cloacal malformation with hydrocolpos. **A:** Coronal US scan at 13 weeks GA shows a large abdominopelvic cyst. Axial US **(B)**, axial 2D SSFP and sagittal fetography **(C)** fetal MRI sequences on same fetus at 17w GA. There is bilateral hydronephrosis, large hydrocolpos *(h)* containing intraluminal echogenic foci with posterior shadowing (*arrow* on **C**) or dark T2 material as expected for meconium (*arrow* on **D**). The bladder is anterior to the hydrocolpos *(asterisk)* and the stomach *(s)* anterior to the left kidney (*arrowhead* on **E**).

mechanical obstruction such as urethral atresia, or in the setting of megacystis–microcolon intestinal hypoperistalsis syndrome and prune belly syndrome. Urethral atresia is typically associated with early development of oligo/anhydramnios, typically with normal rectal anatomy. Megacystis–microcolon intestinal hypoperistalsis syndrome, a process more frequently affecting females, is typically associated with polyhydramnios during the third trimester, and fetal MRI will be extremely helpful in defining the presence of microcolon.[11] Megacystis is also seen in prune belly syndrome, characterized by deficient abdominal musculature, urinary tract abnormalities, and cryptorchidism in males or major genital tract malformations in females including cloacal malformation.[21] Hydrocolpos due to underlying obstruction or urogenital sinus has a normal rectum and fetal MRI is helpful in differentiating it from cloacal malformation.[19]

It will be important to remember that transient ascites and meconium peritonitis (from urine and meconium scape through the tubes) can be seen with cloacal malformation in the absence of bowel perforation. Fetal MRI will help to define the appearance of the bowel and the pelvic structures to support one of the two possibilities.[11]

Prognosis: The prognosis is dictated by the presence of oligohydramnios and renal dysplasia, the type of cloaca, and the presence of other associated malformations. Amniotic fluid volume is easily tracked with ultrasound. Attempts to define the type of cloaca and potential additional malformations have been described with the use of fetal MRI during the third trimester in a series of six fetuses.[21] In this series, long common channel cloacas were linked to dilated and high rectums. Malformations consistent with VACTERL association were defined in one fetus.

With early diagnosis, management of associated anomalies, and efficient meticulous surgical repair, patients can have the best chance for a good anatomic outcome.[4] Functional results will depend as well on the degree of sacral development, nerve supply, and spinal cord anomalies. In cases with incontinence despite excellent anatomic repair, effective bowel and bladder management programs have been devised to improve the patient's quality of life.[22]

Management: Once the diagnosis is suspected with ultrasound, fetal MRI will help characterize the defect, assess renal parenchyma and help identify potential associated malformations. Fetal echocardiogram is recommended to rule out congenital heart defect as well. This approach will provide more accurate information, and a comprehensive multidisciplinary counseling session with the parents will be possible in the presence of pediatric and colorectal surgeons, pediatric urologist, neonatologist, and pediatric nephrologist.

Attempts to restore the amniotic fluid volume have been described in cases of bladder outlet obstruction with the use of vesicoamniotic shunts and serial amnioinfusions, and could be applied to cloacas with megacystis.[13] However, these options will not prevent renal dysfunction and vesicoamniotic shunts might lead to bladder dysfunction, in some cases severe enough to preclude kidney transplantation.[23] Open fetal vesicostomy would be a more effective way to decompress the bladder, preventing further renal damage but could precipitate preterm labor.[24]

Delivery is recommended at a tertiary center, and these infants need urgent clinical and radiological evaluation by experienced pediatric surgeons and radiologist with crucial decisions taken during the first 24 hours.[4,13,25]

Recurrence Risk: The condition is sporadic, with low recurrence risk.

PERSISTENT UROGENITAL SINUS

Urogenital sinus is the persistence of an embryonic state in which there is a single-exit chamber for the bladder and the vagina in the presence of a normal rectum and anus. On physical exam, there are two perineal openings, one anterior for the urogenital sinus and the other for the anus.[1]

Incidence: The condition affects females and is rare.[1] The exact incidence is not well defined in the literature.

Embryology: The anomaly is the result of abnormal development of the lower vagina and failure of urethrovaginal septation. In this situation, urine flow can be impaired, leading to potential development of cystic dilatation of the vagina, also known as hydrocolpos (or hydrometrocolpos if the uterus is also involved). The hydrocolpos can compress the bladder forward, and can result in obstructive uropathy. Rarely, urine can reach the peritoneum through the fallopian tubes and present as urinary ascites.

Etiology: The most common etiology for persistent urogenital sinus is adrenogenital syndrome (congenital adrenal hyperplasia) or other in utero exposure to androgenic stimuli, with expected clitoral enlargement/ambiguous genitalia. It may also occur as pure urogenital sinus with normal external genitalia, or associated with gonadal dysgenesis and hermaphroditism.[1,26]

Diagnosis: Prenatal cystic dilatation of the vagina will compress and displace the bladder anteriorly. A pelvic cystic mass with fluid–fluid levels, representing thick, mucoid cervical or vaginal secretions, should be highly concerning for hydrocolpos.[27] Associated findings in the setting of urogenital sinus can include hydroureteronephrosis, urinary ascites, and in severe cases renal dysplasia and oligohydramnios (Fig. 19.2-5).[20,28]

Enlarged adrenal glands with a cerebriform pattern, typically seen in neonates with congenital adrenal hyperplasia, might not be detected in utero until the late third trimester.[29] Babies

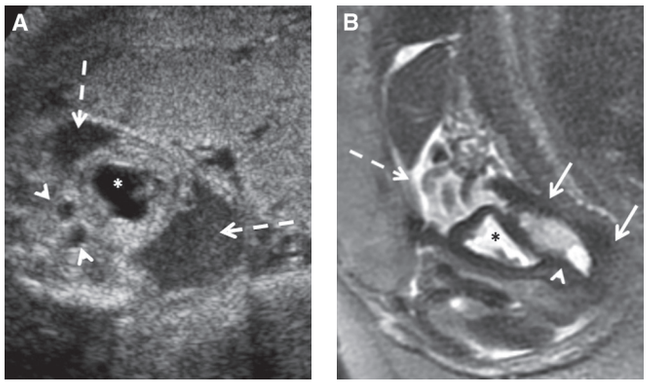

FIGURE 19.2-5: Fetus at 26 weeks gestation with urogenital sinus and severe oligohydramnios. **A:** Axial US image. **B:** Sagittal heavily T2w hydrography sequence (MR fetography). There is a septated hydrocolpos *(arrowheads)* posterior to a thick-walled bladder *(asterisk)*. There is ascites *(dashed arrow)* and normal meconium-filled (dark T2 signal) rectum *(arrows)* posterior to the hydrocolpos.

with urogenital sinus can present with normal or ambiguous genitalia.

Fetal MRI can detect the cervical imprint on the vagina confirming the diagnosis of hydrocolpos and will define the presence or absence of meconium in the rectum or potential abnormal content. With normal meconium in the rectum and hydrocolpos, urogenital sinus or vaginal obstruction should be suspected. If there is absence of meconium in the rectum or the rectum is identified but with fluid content, enteroliths, or dilatation, the underlying malformation is cloaca.[19,21,30]

Differential Diagnosis: One difficulty in diagnosing pelvic cystic masses in female fetuses is the potential confusion of the bladder with the hydrocolpos and an initial consideration of megacystis. However, the fetal bladder is outlined by the umbilical arteries, has an anterior midline position, and its volume is expected to change, allowing its correct identification.[19] In the setting of hydrocolpos, the possibility of cloaca, urogenital sinus, or obstructive vagina should be entertained. Other pelvic cystic lesions would be considered in the differential such as ovarian cyst, anterior meningocele, type IV sacrococcygeal teratoma, enteric duplication cyst, lymphatic malformations, and ectopic multicystic dysplastic kidney.[27]

Prognosis: The prognosis will be dictated by the degree of obstructive uropathy, and potential associated malformations. In survivors, the long-term outlook is reasonably good after reconstructive surgery. In the neonatal period, a correct prenatal diagnosis, absence of renal dysplasia, and coordinated work between the involved teams (obstetricians, neonatologists, radiologist, and pediatric surgeons) will significantly impact the clinical outcome.[28]

Management: Multidisciplinary counseling and delivery in a tertiary center is indicated. In the neonatal period, vaginal drainage will alleviate the abdominal distension and potential cardiorespiratory compromise and will improve any obstructive uropathy associated.[4] Metabolic assessment and treatment in cases of congenital adrenal hyperplasia and chromosomal studies will be indicated in cases of ambiguous genitalia.[1]

In families with known history of congenital adrenal hyperplasia, early medical treatment can start in utero with maternal glucocorticoid administration before the development of the urogenital sinus (6th to 7th week of gestation) and be maintained after confirmation of gene mutation and female karyotype (usually by the 13th week of gestation).[29]

Recurrence Risk: Persistent urogenital sinus, in the absence of congenital adrenal hyperplasia, is usually a sporadic condition with low recurrence risk. On the other hand, congenital adrenal hyperplasia is an autosomal recessive disorder with an incidence of approximately 1:14,000 live births.[31] When other malformations are associated including postaxial polydactyly, then an underlying genetic condition should be considered such as McKusick–Kaufman, Bardet–Biedl, Ellis van Creveld syndromes (with autosomal recessive inheritance), or Pallister–Hall syndrome (with autosomal dominant inheritance).[32]

HYDROCOLPOS

Hydrocolpos is the cystic dilatation of the vagina. Hydrometrocolpos is distention of the uterus and vagina. We use the term hydrocolpos here to refer to both conditions.

Incidence: The condition is rare. The prevalence of congenital hydrocolpos is less than 1 per 30,000 births and represents 15% of the abdominal masses in female fetuses.[19]

Embryology: There are two types of hydrocolpos. The urinary type is related to a urogenital sinus or cloacal malformation. The secretory type is related to vaginal obstruction because of abnormal development of the lower vagina presenting as imperforate hymen, vaginal agenesis or hypoplasia, and transverse septum.

Etiology: Most cases of hydrocolpos are sporadic but can also be seen with genetic syndromes such as McKusick–Kaufman, Bander–Belt, Langer–Giedion (tricho–rhino–phalangeal syndrome), Pallister–Hall, and Mayer–Rokitansky–Küster–Hauser syndrome/MURCS (Müllerian duct, renal agenesis/ectopia, and cervical thoracic somite dysplasia) association (Fig. 19.2-6).[32]

Diagnosis: A pelvic cystic mass posterior to the bladder, with or without fluid–fluid levels, should be highly concerning for hydrocolpos in a female fetus. When there are Müllerian duct anomalies, it may appear as a septated cyst. Hydroureteronephrosis and ascites can be associated.[19,20,33] Fetal MRI can define the appearance of the rectum after 21 weeks of gestation.[17] With a normal rectal cul-de-sac filled with meconium, cloacal malformation is considered unlikely, and hydrocolpos could be related to urogenital sinus or vaginal obstruction. If there is absence of meconium in the distal rectum or the rectum is identified but with fluid content, enteroliths, or dilatation, the underlying malformation is a cloaca.[19]

At birth, they could present as a large abdominal mass with hydroureteronephrosis and possible respiratory distress from elevation of the diaphragm. If diagnosis and treatment are delayed, infection may follow, resulting in pyocolpos and sepsis.[4,33]

Differential Diagnosis: Differential diagnosis includes other cystic masses, such as ovarian cyst, enteric duplication cyst, megacystis, duplicated urinary bladder, anterior meningocele, and type IV sacrococcygeal teratoma.

In the setting of hydrocolpos, the possibility of cloaca, urogenital sinus, or obstructive vagina should be entertained. Complete assessment of the fetus will also help to define the defect as potentially isolated or in the setting of a syndromic association or genetic disease.

Prognosis: The prognosis will be dictated by the type of underlying defect (hydrocolpos versus urogenital sinus or cloacal malformation), the degree of obstructive uropathy present, and potential associated malformations or genetic syndromes. In isolated forms with preserved renal function, the outlook is excellent after final surgical reconstruction.[32,34]

Management: A complete assessment of the fetal anatomy is recommended to rule out other structural defects. Amniocentesis could be considered when there is concern for a major genetic anomaly. Multidisciplinary counseling and delivery in a tertiary center is indicated. In the immediate neonatal period, vaginal drainage will alleviate the abdominal distention and potential cardiorespiratory compromise and will improve urinary obstruction, if present.[4] In order to rule out syndromes, high-resolution chromosome studies as well as long-term clinical follow-up will be needed.[32]

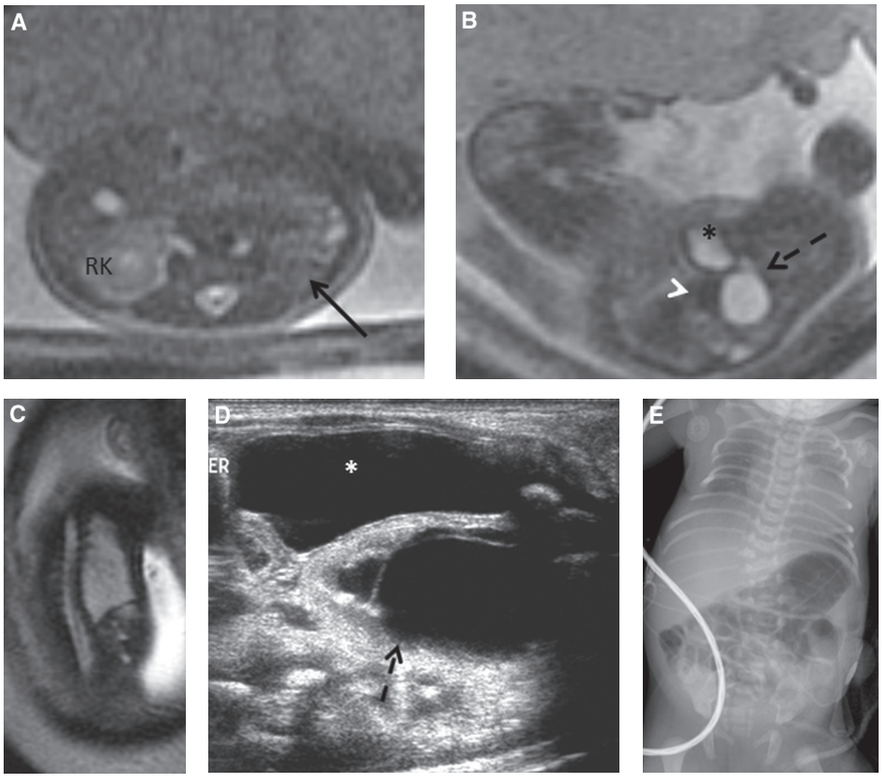

FIGURE 19.2-6: Hydrocolpos in the setting of MURCS association, also known as type II Mayer-Rokytansky-Küster-Hauser (MRKH) syndrome. Axial **(A,B)** and coronal **(C)** T2-weighted fetal MRI at 29 weeks GA. **A:** There is left kidney renal agenesis with bowel filling the left renal fossa *(arrow)*. A normal right kidney *(RK)* is present. **B:** Posterior to the bladder *(asterisk)*, normal rectum *(white arrowhead)*, and hydrocolpos *(black dashed arrow)* are noted. **C:** There is rightward thoracic scoliosis. Pelvic US **(D)** and chest-abdomen x-ray **(E)** on first day of life showing hydrocolpos *(black dashed arrow)* and focal segmentation vertebral anomaly at the lower thoracic spine accounting for thoracic scoliosis and short fourth ribs. There is also cardiomegaly and increased lung vascularity because of truncus arteriosus.

Recurrence Risk: Hydrocolpos is usually a sporadic condition with low recurrence risk. However, some syndromic forms are genetically transmitted.[32]

CLOACAL EXSTROPHY

Cloacal exstrophy, also known as the OEIS complex (omphalocele, cloacal exstrophy, imperforate anus, spinal defects)[35] is one of the rarest and most complex anorectal and urogenital malformations. The entity represents an anterior wall defect that includes the persistence and exstrophy of a common cloaca that receives ureters, ileum, and a rudimentary blind-ending hindgut (Fig. 19.2-7). It is commonly associated with omphalocele, spinal dysraphism, and incompletely formed external genitalia and is always associated with imperforate anus.[36] Typically in females, there are usually two vaginal openings, widely separated and entering the lower aspect of each hemibladder and bilateral hemiuteri as well as two widely separated clitoral halves, while in males there is usually a rudimentary hemipenis on each side caudal to the exstrophy.[1] The terminal ileum could prolapse, giving the appearance of an elephant's trunk (Fig. 19.2-8).[37]

Incidence: The reported incidence is 1 in 200,000 to 400,000 live births.[1] The condition can affect both genders, with most recent studies suggesting no predilection toward either gender[38] or female preponderance.[39] It has also been described as increased

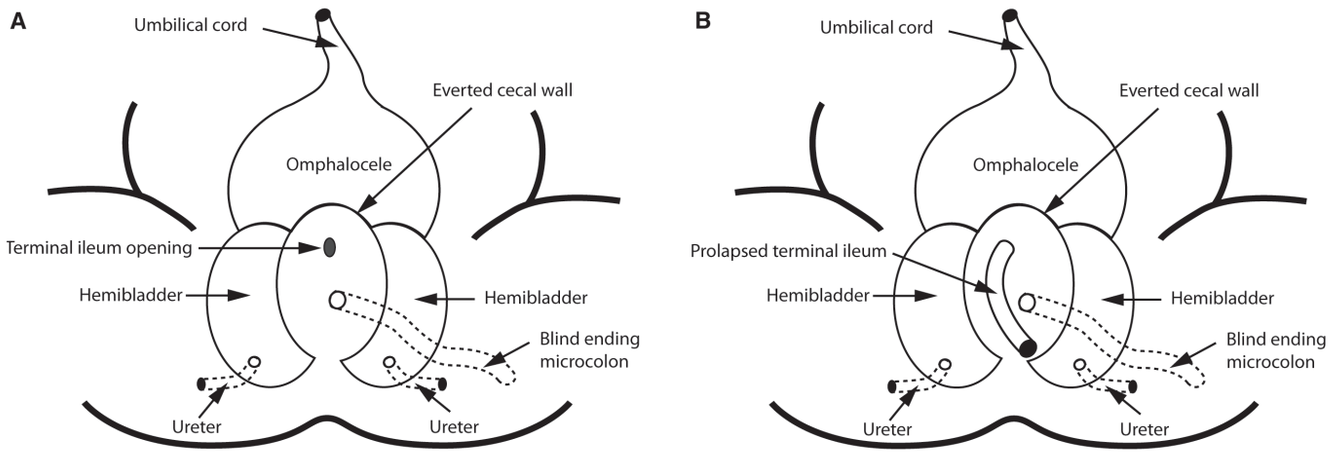

FIGURE 19.2-7: Diagrams of typical anatomical appearances of cloacal exstrophy without **(A)** and with **(B)** prolapse terminal ileum. (Reproduced with permission from Calvo-Garcia MA, Kline-Fath BM, Rubio EI, et al. Fetal MRI of cloacal exstrophy. *Pediatr Radiol*. 2013;43:593–604.)

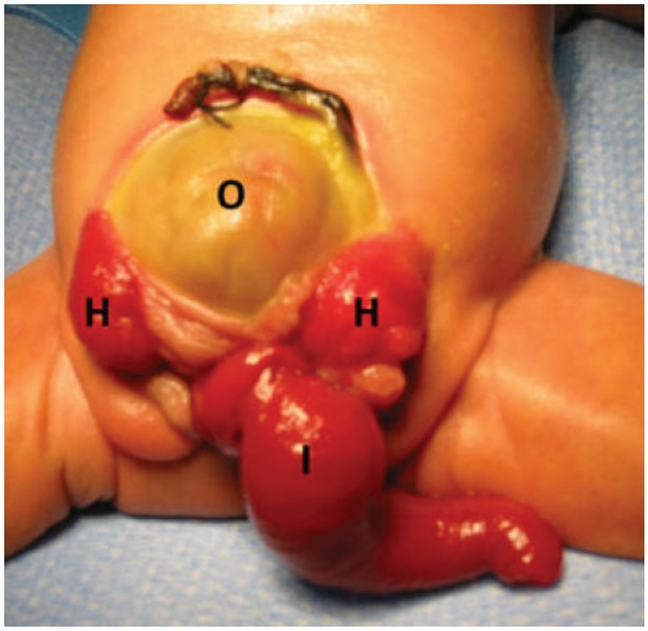

FIGURE 19.2-8: Cloacal exstrophy with prolapsed terminal ileum photograph. There is a low-lying omphalocele *(O)*. Extending from the back of the cecal plate is a prolapsed terminal ileum *(I)*. The exstrophied hemibladders *(H)* are seen on each side. (Reproduced with permission from Bischoff A, Calvo-Garcia MA, Baregamian N, et al. Prenatal counseling for cloaca and cloacal exstrophy: challenges faced by the pediatric surgeons. *Pediatr Surg Int.* 2012;28:781–788.)

incidence of cloacal exstrophy in multiple gestations, especially monozygotic twins.[39,40]

Embryology: Cloacal exstrophy is a severe and rare malformation thought to be a developmental field defect affecting the mesoderm, which later contributes to infraumbilical mesenchyme, urorectal septum, and caudal vertebrae during early embryogenesis (Figs. 19.2-2 and 19.2-9).[39]

Before the 5th week of embryonic development, the urinary, genital, and gastrointestinal tracts empty into a common chamber, the cloaca. At the caudal end of the cloaca, ectoderm lies directly over endoderm, forming the cloacal membrane, which at this stage forms part of the ventral wall. During the 6th week of development, the mesoderm grows toward the midline, forming the infraumbilical abdominal wall. Simultaneously, the urorectal septum extends caudally toward the cloacal membrane, and by the 8th week of gestation, the cloaca is divided into an anterior chamber, the primitive urogenital sinus, and a posterior chamber, the rectum. There is also caudal retraction of the cloacal membrane, so it is no longer a part of the abdominal wall and eventually ruptures at the end of the 10th week of gestation. Abnormal mesodermal migration between the ectodermal and the endodermal layers of the cloacal membrane or failure of the mesoderm to grow between those two layers is thought to cause its premature rupture. Some authors think that the stage of development when the abnormality occurs determines the type of defect: cloacal exstrophy, bladder exstrophy, or isolated epispadias. Early damage would affect the

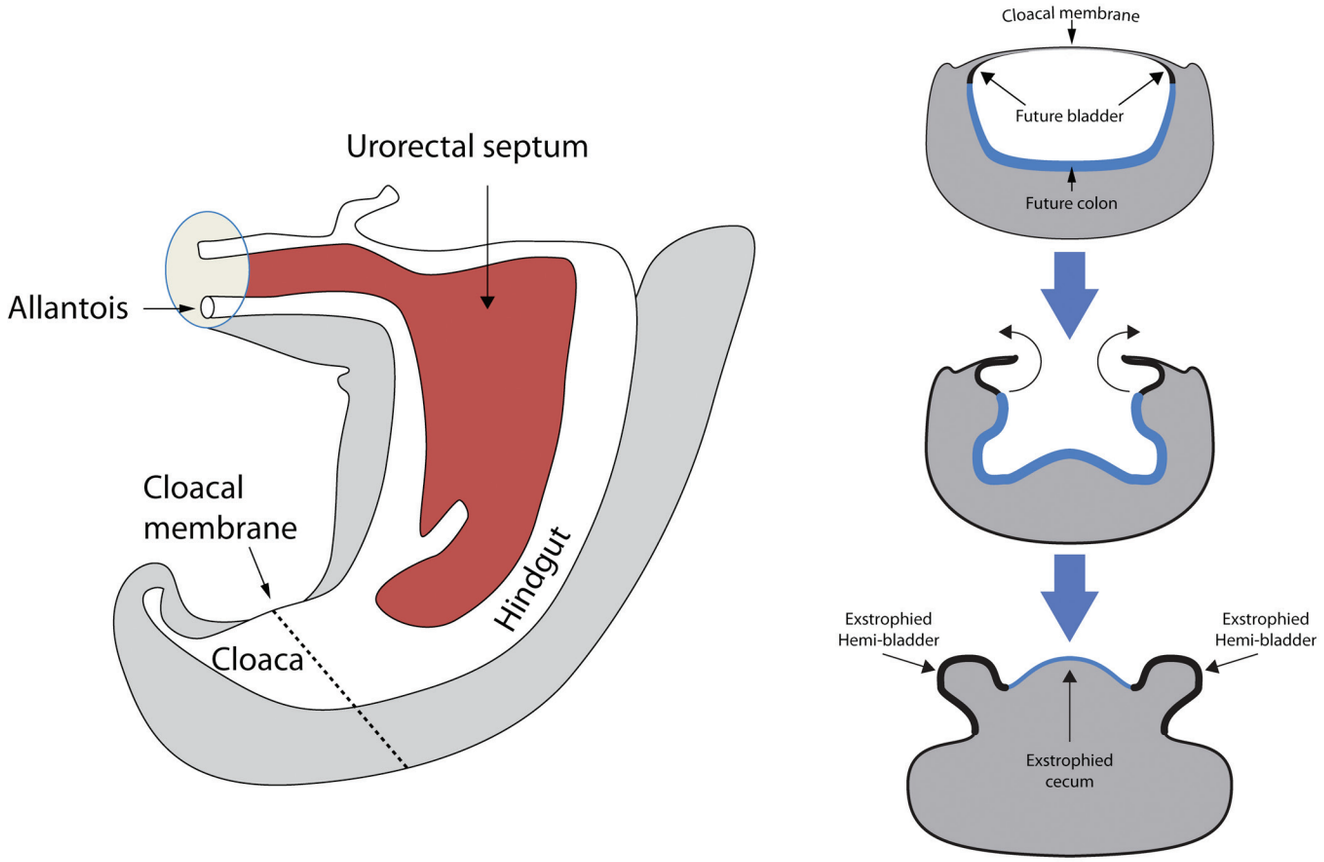

FIGURE 19.2-9: Diagram of events leading to cloacal exstrophy. The urorectal septum fails to grow caudal. Subsequently, there is premature rupture of the cloacal membrane while it is still part of the infraumbilical ventral wall. There is a resultant infraumbilical abdominal wall defect with everted bladder plates and cecal wall.

mesenchyme that contributes to the infraumbilical mesoderm, the urorectal septum, and the lumbosacral somites. This results in failure of cloacal septation. This leads to persistent cloaca and rudimentary hindgut with imperforate anus. A breakdown of the cloacal membrane, causes exstrophy of the cloaca, failure of fusion of the genital tubercles, and diastasis of the pubic rami. Occasionally there is also an omphalocele, and incomplete development of the lumbosacral vertebrae with commonly a closed neural tube defect. This constitutes what has been classically recognized as cloacal exstrophy. A later failure in migration of the infraumbilical mesoderm, after the caudal movement of the urorectal septum has been completed will result in bladder exstrophy.[41]

Etiology: The underlying cause remains unknown, but genetic and environmental factors are thought to play a role.[42] However, no single environmental exposure or consistent genetic defect has yet been identified.[38]

Diagnosis: Prenatal diagnosis with ultrasound has been suggested with the following criteria: failure to visualize the bladder in the presence of normal amniotic fluid volume, large midline infraumbilical anterior wall defect, omphalocele, cystic anterior wall structure, and/or neural tube defect.[43] In 1999, Hamada et al.[37] reported the "elephant trunk-like" image as a new criterion for the diagnosis. This sign represents the prolapse of the terminal ileum through the exstrophy, and it is the most specific sonographic sign supporting this diagnosis (Fig. 19.2-10). However, it is inconsistently found as the prolapse can be intermittent or potentially obscured by an adjacent omphalocele or umbilical cord.[44]

In the case of delayed rupture of the cloacal membrane, a protruding infraumbilical cyst will be seen outlined by the umbilical arteries.[45] Later in the pregnancy, usually it breaks down, and by the time of delivery, a classic cloacal exstrophy is observed.

When a classic elephant trunk-like sign is not seen, fetal MRI will be able to define the infraumbilical anterior wall defect with or without associated omphalocele. It will provide improved renal and spine assessment, and helps confirm a primitive hindgut with imperforate anus in the absence of meconium signal in the expected distribution of the colon and rectum.[46]

Differential Diagnosis: In the absence of an elephant trunk-like sign, a prenatal diagnosis with ultrasound can be challenging, and several cases have been misdiagnosed as omphalocele, gastroschisis, or sacrococcygeal teratoma.[43,47] In other cases, a differential diagnosis with bladder exstrophy is considered.[48] The first clues for the correct diagnosis would be the absence of the bladder despite normal amniotic fluid and a protuberant anterior pelvic contour with or without associated omphalocele. In that situation, cloacal or bladder exstrophy should be considered, and fetal MRI performed after 21 weeks, when meconium is expected to fill the rectum, could provide a definitive diagnosis.

Prognosis: With modern surgical and medical treatment and in the absence of other severe malformations, the long-term survival rate is 83% to 100%. However, the challenge remains in how to improve the quality of life of these children, and in order to optimize functional results, it is paramount that a correct unified surgical management plan be instituted from birth.[49] Lifelong colostomy could be necessary, but pull-through procedures of the distal colon to the perineum might be successful together with the use of daily enemas. The majority of the patients achieve urinary continence with modern surgical reconstructive techniques, but most of them require intermittent bladder catheterization. Renal function has also a great impact on long-term outcome. Sexual function might be impaired, and reproduction is unlikely.[40]

Management: Once a confident diagnosis is established, parental counseling with a multidisciplinary team, including colorectal surgeons, pediatric urologists, pediatric neurosurgeons, and neonatologists, must be provided. After thorough patient education and counseling, parents can make an informed decision about available options. If parents choose postnatal treatment, referral to centers with experience in treating cloacal exstrophy is recommended.[40,42]

Recurrence Risk: The vast majority of cases are sporadic without a recognized associated chromosomal abnormality. There is, however, a higher reported incidence of cloacal exstrophy in monozygotic twins and in families in which one member is affected.[50]

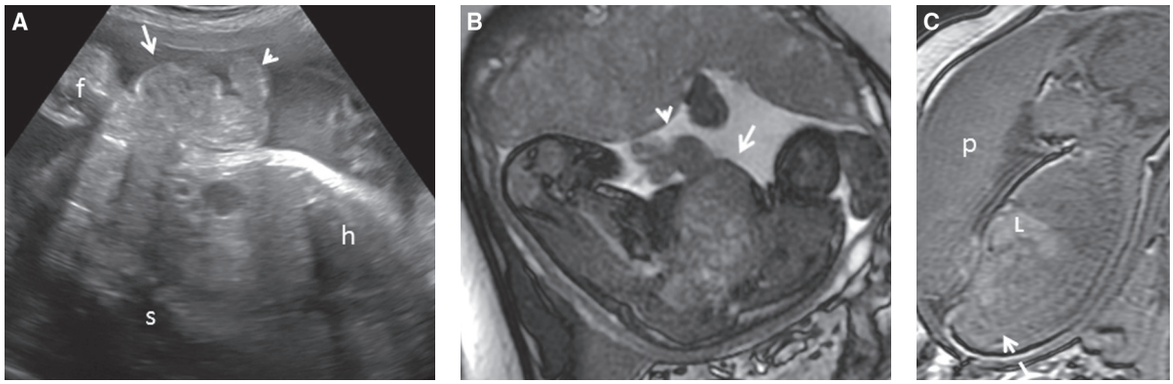

FIGURE 19.2-10: US and fetal MRI at 33 weeks of gestation. **A:** Sagittal oblique US image shows a protruding "mass" *(arrow)* in the infraumbilical abdominal wall with a tubular extension *(arrowhead)* representing the "elephant trunk-like" image. Spine *(s)*, femur *(f)*, and heart *(h)* are labeled. **B:** Axial 2D FIESTA. There is absent bladder despite normal amniotic fluid volume and protruding anterior pelvic contour *(arrow)* with a tubular extension *(arrowhead)*, the prolapsed terminal ileum. **C:** Sagittal midline T1-weighted MRI shows lack of bright meconium signal in the rectum *(dashed arrow)*. Liver *(L)* and placenta *(p)* are labeled.

BLADDER EXSTROPHY

Bladder exstrophy is characterized by a defect involving the infraumbilical abdominal wall and anterior wall of the urinary bladder. As a result, the open bladder mucosa becomes exposed and everted through the wall defect (Fig. 19.2-11). The umbilical cord is low set and the pubic symphysis is always widened. In males, there is complete epispadias with the urethral plate open to the tip of the phallus, resulting in a short, wide, anteriorly displaced penis. In females, there is an open urethral plate, a bifid clitoris, and wide separation of the labia.[42,51] In the most severe form, it may be accompanied by omphalocele, inguinal hernia, undescended testis, ventrally located anal orifice, relaxed anal sphincter, and intermittent rectal prolapse.[52] Imperforate anus, spine defects, and spina bifida, more typical in cloacal exstrophy, have been found only in a low number of cases.[39]

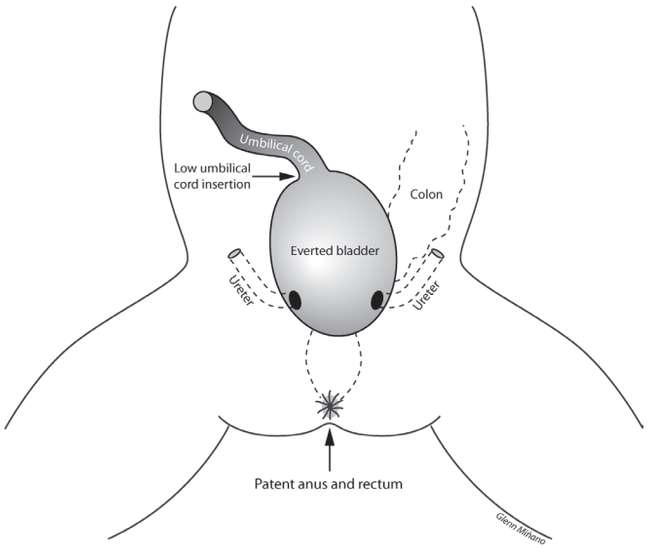

FIGURE 19.2-11: Diagram of typical anatomical appearance of bladder exstrophy. The everted and opened bladder is exposed through an infraumbilical wall defect. There is a patent anus with normal rectum. (Reproduced with permission from Calvo-Garcia MA, Kline-Fath BM, Rubio EI, et al. Fetal MRI of cloacal exstrophy. *Pediatr Radiol*. 2013;43:593–604.)

Incidence: It occurs in 1 per 30,000 live births and is more common in males, with a 2:1 male-to-female ratio.[42]

Embryology: Bladder exstrophy represents a slightly later abnormality in embryogenesis than does cloacal exstrophy. Both processes start in the 4th week of gestation because of failure of mesenchymal cells to migrate between the ectoderm of the abdominal wall and the endoderm of the cloacal membrane. As a result, the cloacal membrane ruptures prematurely. If the defect occurs after the urorectal septation of the cloaca, it will result in bladder exstrophy.

Etiology: The etiology remains unclear, but is thought to represent a multifactorial disorder in which environmental and genetic factors are likely to play a role.[53]

Diagnosis: Usual sonographic findings include persistent nonvisualization of the fetal bladder despite a normal amniotic fluid volume with a lower abdominal bulge, low umbilical insertion, small penis with anteriorly displaced scrotum, and widening of the iliac crests (Fig. 19.2-12).[51] Color Doppler can help identify the umbilical arteries running alongside the bulging mass, also noted in the case of cloacal exstrophy and linking the bulging mass with the bladder.[47,54] Three-dimensional (3D) ultrasound has also been reported to help in the diagnosis of bladder and cloacal exstrophy.[55]

Similar findings will be noted with fetal MRI.[56] In addition, normal appearance of the rectum and colon will allow a more definitive diagnosis, excluding cloacal exstrophy, especially in cases where omphalocele or other malformations are noted.

Differential Diagnosis: The main differential diagnosis is cloacal exstrophy. The absence of associated malformations such as omphalocele or spinal defects would make cloacal exstrophy less likely. However, bladder exstrophy has been reported in a low number of cases with those same associated defects, and cases of cloacal exstrophy might not show obvious spinal defects. Fetal MRI will be able to differentiate between the two in those situations.

Other abdominal wall defects, such as omphalocele and gastroschisis, need to be considered, but they are easily differentiated, and there should be a normally filled bladder.

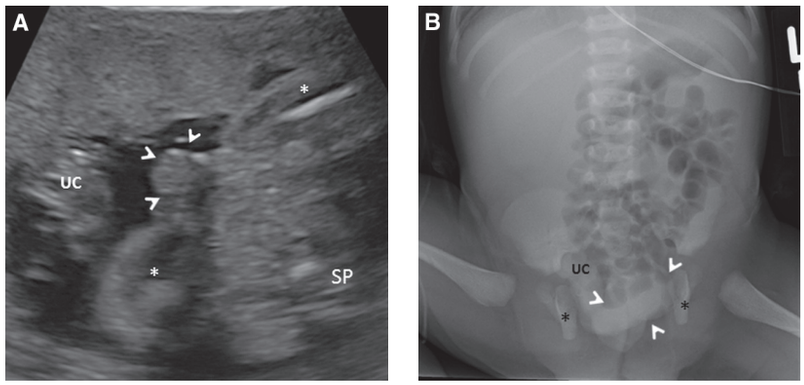

FIGURE 19.2-12: Bladder exstrophy. **A:** Axial US image at 24 weeks of gestation shows a protruding anterior pelvic wall "mass" *(arrowheads)* and lack of visualization of fluid-filled bladder in the presence of normal amniotic fluid volume. The spine *(SP)* and thighs *(white asterisks)* are denoted for orientation. Umbilical cord *(UC)*. **B:** Postnatal frontal abdomen x-ray on the first day of life. Note also the pubic symphysis diastasis *(black asterisks)*.

Prognosis: The prognosis for bladder exstrophy has improved quite significantly in recent years, and the overall outcome is relatively good with expected urinary continence and normal sexual function in many patients.[57]

Management: Prenatal diagnosis will allow appropriate parental counseling,[57] as well as optimal perinatal management and prompt postnatal surgical intervention. Early intervention, in the first 48 hours of life, minimizes damage to the exposed bladder and permits early closure and possibly avoids the need for pelvic osteotomy.[51]

Recurrence Risk: The recurrence risk for offspring of affected individuals is 1 in 70. Among siblings, the estimated recurrence risk has been reported to be between 0.3% and 1%.[58–60]

PATENT URACHUS

The urachus is a vestigial remnant of the allantois, extending from the anterosuperior aspect of the bladder toward the umbilicus. Complete or partial persistence of its lumen gives rise to urachal disorders. The anomalies of the urachus are classified according to the persisting patent segment and include congenital patent urachus, urachal cyst, umbilico-urachal sinus, and vesico-urachal diverticulum (Fig. 19.2-13).[61]

Incidence: Urachal anomalies are uncommon, with a male-to-female ratio of 2 to 1.[62] Patent urachus accounts for about 50% of all cases, urachal cyst 30%, umbilical-urachal sinus about 15%, and vesico-urachal diverticulum 3% to 5%.[63]

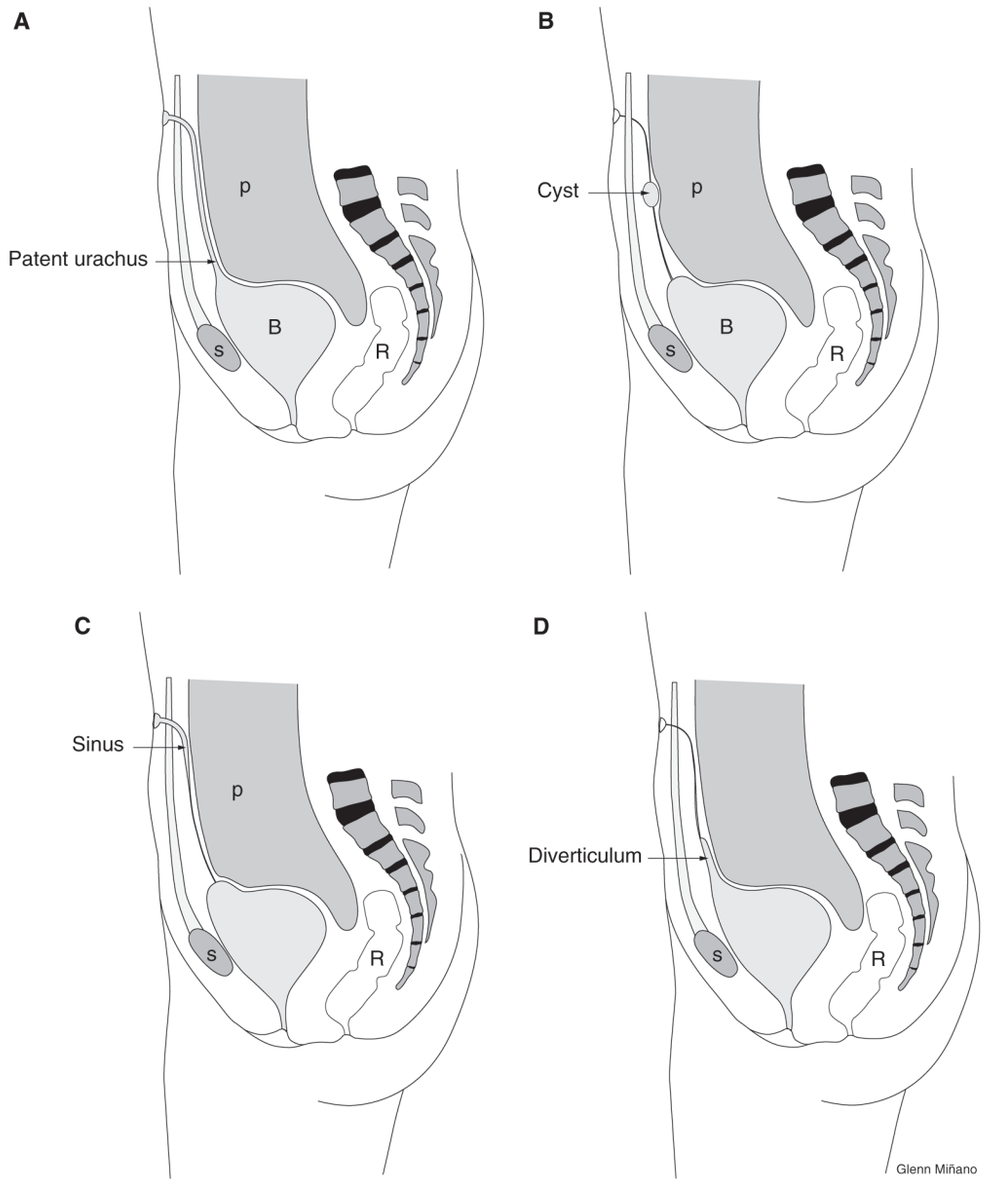

FIGURE 19.2-13: Types of congenital urachal anomalies. **A:** Patent urachus. **B:** Urachal cyst. **C:** Urachal sinus. **D:** Urachal diverticulum. B, bladder; p, peritoneal cavity; r, rectum; s, symphysis pubis.

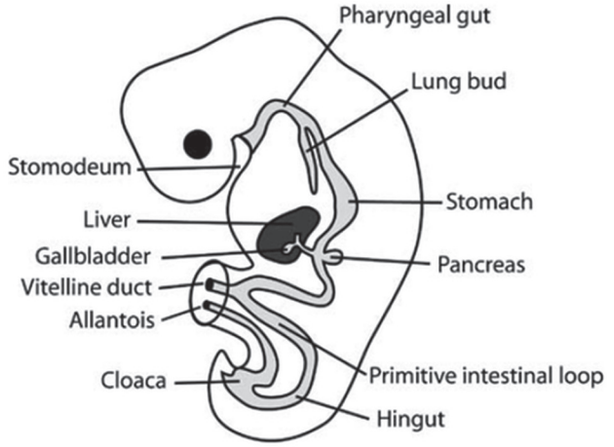

FIGURE 19.2-14: Embryo with primitive umbilical cord. (Reproduced with permission from Bunch PT, Kline-Fath BM, Imhoff SC, et al. Allantoic cyst: a prenatal clue to patent urachus. *Pediatr Radiol*. 2006;36:1090–1095.)

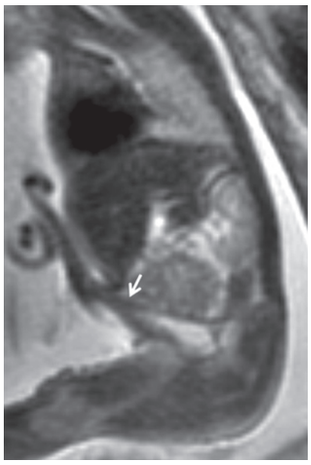

FIGURE 19.2-15: 25-Week-old fetus with patent urachus. T2w sagittal MR image demonstrates elongation of the bladder dome toward the abdominal cord insertion *(white arrow)*.

Embryology: The urachus is the intraabdominal remnant of the embryonic allantois (Fig. 19.2-14). The allantois appears on approximately the 16th day of gestation as a small diverticulum from the caudal wall of the yolk sac that extends into the connecting stalk. The allantois is involved with development of the dome of the urinary bladder and the umbilical vessels, and forms a hollow tube that connects the superior aspect of the urogenital sinus with the anterior abdominal wall at the umbilicus. As the bladder enlarges, the allantois becomes obliterated to form a thick tube, the urachus, also known as the median umbilical ligament, situated in the prevesical space, between the peritoneum behind and the transversalis fascia in front. Defective closure may happen and leads to congenital pathology of the urachus.[62]

Etiology: The etiology of urachal anomalies is unknown. Urachal anomalies, particularly patent urachus, have been reported, in up to one-third of these cases, in the setting of bladder outlet obstruction as a "pop-off" mechanism to protect the upper urinary tract.[61]

Diagnosis: A patent urachus results from persistent communication between the bladder lumen and the umbilicus. The bladder dome will show elongated appearance toward the abdominal cord insertion (Fig. 19.2-15). In some cases, it could lead to the development of a true cyst at the base of the umbilical cord, which communicates with the bladder dome and will lead to separation of the umbilical cord vessels (Fig. 19.2-16). The cyst is known as allantoic, or vesico-allantoic cyst. Ultrasound with color Doppler, 3D, 4D ultrasound, and fetal MRI can characterize these lesions. Some of these cysts can be very large or progress to diffuse cord edema.[64]

Urachal cysts are noted as anterior midline cysts, cranial to the bladder without dynamic changes in size or shape.[65]

Patent urachus with in utero interval resolution of allantoic cyst has been described, and in some cases in association with subsequent bladder prolapse.[66,67]

Differential Diagnosis: Allantoic cysts in the setting of patent urachus will present as a cyst near the fetal anterior abdominal wall. Omphalomesenteric cysts are also true umbilical cord cysts and present in this same location, but they do not separate the cord vessels and do not communicate with the bladder dome. Umbilical cord pseudocysts will affect more distal segments of the cord.[64] Color Doppler ultrasound will be able to differentiate from umbilical vein varix or umbilical artery aneurysm.[62,68]

The differential diagnosis of a urachal cyst includes ovarian cyst, mesenteric cyst, omphalomesenteric duct remnant, and bowel atresia.

Prognosis: Allantoic cysts have no increased risk for chromosomal anomaly. One-third of these cases occur in association with bladder outlet obstruction, and its presence supports a patent urachus with urine leakage from the umbilicus noted in the neonatal period.[64]

Potential umbilical cord vessel compression, umbilical cord hematoma or torsion leading to fetal distress and even fetal death has been described as potential rare complications in the setting of umbilical cord cysts.[69]

All urachal malformations have a high incidence of recurrent infection and stone formation and may develop benign and malignant neoplasms.[61,63]

Associated anomalies are uncommon but include omphalocele and omphalomesenteric remnants[64,66,70] as well as other genitourinary conditions such as hypospadias, undescended testicles, crossed renal ectopia, vesicoureteral reflux, hydronephrosis, and lower urinary tract obstruction.[61,66]

Management: In the presence of an allantoic cyst or any umbilical cord cyst, some authors recommend close prenatal surveillance, including assessment of the size of the cyst, Doppler analysis of the umbilical circulation and delivery as soon as fetal maturity is demonstrated.[69]

Cesarean section can be considered in cases of large allantoic cysts in order to better define the site of cord ligation.[64]

After delivery, surgical consult of all urachal malformations is recommended.[63]

Recurrence Risk: These conditions are sporadic with a low recurrence risk.[64]

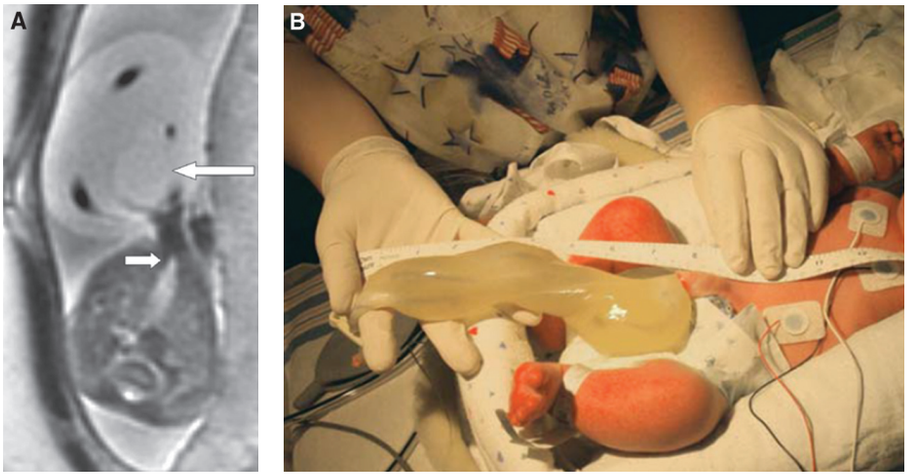

FIGURE 19.2-16: Patent urachus with allantoic cyst. **A:** Fetal MRI axial T2-weighted image shows elongation of the bladder dome toward the abdominal cord insertion *(short arrow)* with a cyst *(long arrow)* at the base of the cord. Note the splaying of the umbilical vessels *(dark structures)*. **B:** First day of life photograph. There is a large edematous mass in the umbilical cord. (Reproduced with permission from Bunch PT, Kline-Fath BM, Imhoff SC, et al. Allantoic cyst: a prenatal clue to patent urachus. *Pediatr Radiol*. 2006;36:1090–1095.)

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