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MINISYMPOSIUM: FETAL/NEONATAL IMAGING

Fetal anterior abdominal wall defects: prenatal imaging by magnetic resonance imaging

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Abstract Abdominal wall defects range from the mild umbilical cord hernia to the highly complex limb–body wall syndrome. The most common defects are gastroschisis and omphalocele, and the rarer ones include the exstrophy complex, pentalogy of Cantrell and limb–body wall syndrome. Although all have a common feature of viscera herniation through a defect in the anterior body wall, their imaging features and, more important, postnatal management, differ widely. Correct diagnosis of each entity is imperative in order to achieve appropriate and accurate prenatal counseling and postnatal management. In this paper, we discuss fetal abdominal wall defects and present diagnostic pearls to aid with diagnosis.

Keywords Abdominal wall defect · Bladder · Cloaca · Exstrophy · Fetus · Magnetic resonance imaging · Limb–body wall defect · Umbilical cord insertion

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Introduction

Anterior abdominal wall defects range from the mild umbilical cord hernia to the severe and uniformly fatal limb–body wall syndrome. The overall estimated incidence of abdominal wall defects is approximately 6 per 10,000 births [1]. The embryology is complex but exquisitely reviewed by Pakdaman et al. [1]. Although there are significant differences among entities, all involve herniation of one or more fetal intra-abdominal contents through a defect in the ventral abdominal wall. The most important first step in the characterization of the defect is the location of the umbilical cord insertion and its relationship to the defect. Evaluation of the abdominal wall, gastrointestinal and genitourinary system then follows, continued by an analysis of the spine and remaining fetal anatomy. Many times the prenatal diagnosis is straightforward, aiding in patient counseling and perinatal planning. In certain instances, however, the diagnosis is not immediately evident. In this paper, we review the MRI spectrum of anterior abdominal wall defects by degree of increasing complexity and present diagnostic pearls to aid with the correct recognition of each entity. We present and analyze cases that were challenging to characterize prenatally and provide clues to avoid misdiagnosis.

Umbilical cord hernia

Relatively little has been written about this entity. The simplest of the abdominal wall defects results from a persistent defect of the abdominal wall fascia at the site of the umbilical cord, with intact skin covering it and a relatively small protrusion of the bowel (Fig. 1). Unlike omphalocele, which is only covered by an amniotic membrane, an umbilical cord hernia is always covered by intact skin and subcutaneous tissues [2]. In

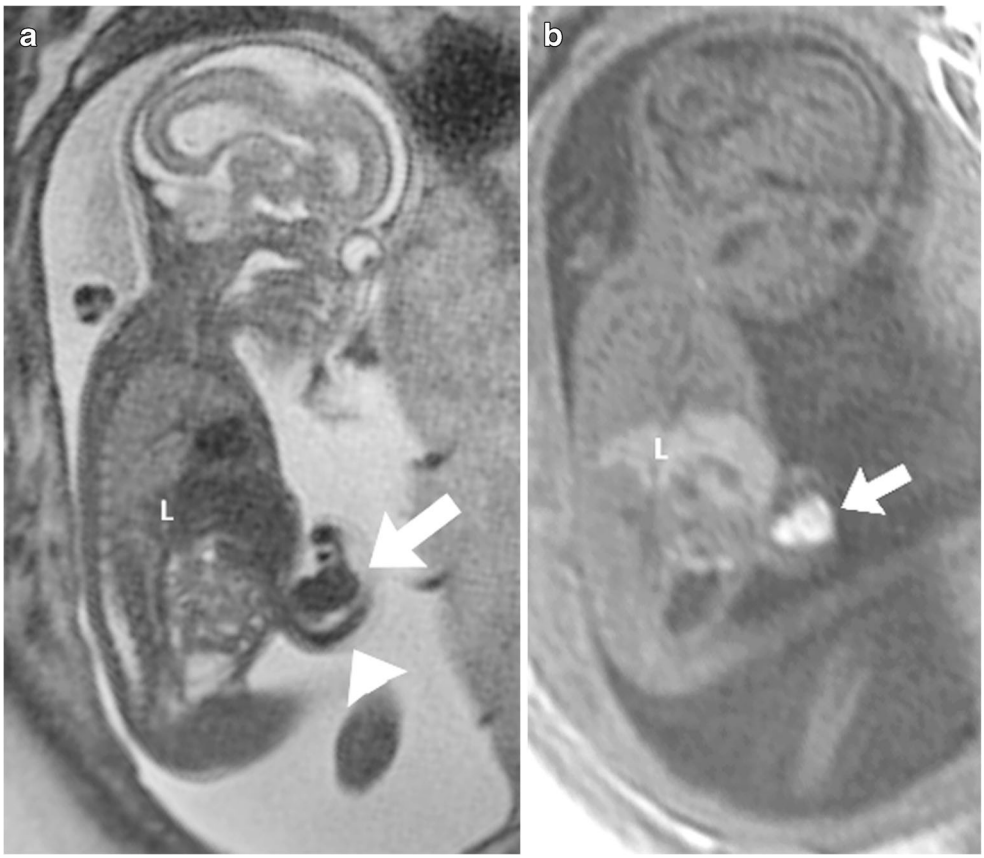

Fig. 1 Umbilical cord hernia in a 19-week fetus. **a, b** Sagittal single-shot turbo spin-echo (TSE; **a**) and T1-weighted (**b**) MR images show the herniated meconium-containing loop of small bowel (*arrows*), demonstrating its characteristic low signal on single-shot TSE and high signal on T1-W imaging. Umbilical cord vessels are denoted (*arrowhead*). *L* liver

the earlier gestational weeks, however, it might be difficult to distinguish skin from membrane by MR, although this difference becomes more apparent in the second and third trimesters. The prognosis is excellent. One important clinical consideration when managing this entity is knowledge of its existence, particularly when clamping the cord immediately after birth, so as to avoid bowel injury.

Gastroschisis

The word "gastroschisis" is derived from the ancient Greek *gastro-* (stomach) and *schisi* (split, open). It refers to the herniation of midgut bowel into the amniotic cavity through a most commonly right-side para-umbilical full-thickness wall defect [3]. The defect is usually <2 cm and is not covered by the amniotic membrane, so the loops of bowel float freely within the amniotic cavity (Fig. 2). The incidence of gastroschisis is approximately 1 in 4,000 live births, although it is increasing worldwide. Its etiology is uncertain and likely multifactorial. Some authors propose an ischemic event that results in a focal weakness of the abdominal wall, others propose an error in the early lateral folding of the embryo, and yet others propose the rupture of an umbilical cord hernia as the etiology of gastroschisis [4, 5]. Young maternal age is a factor contributing to its etiology, with an incidence among teenage mothers seven times greater than in mothers older than 25 years [1]. The incidence of associated congenital abnormalities is rare in fetuses with gastroschisis. The postnatal outcome is usually favorable, with the condition of bowel at birth being the most important indicator of postnatal morbidity.

Babies with uncomplicated gastroschisis have a survival of >95% [6]. Dilated, echogenic bowel is usually an indication of bowel at risk; constant vigilance by means of frequent US surveillance and possible early delivery should be considered if these findings are encountered (Fig. 3). Survival decreases to 70% if ischemia, atresia or perforation of the bowel is encountered at birth. The widely preferred mode of delivery for fetuses with gastroschisis is vaginal [7, 8]. As for timing of delivery, there is an ongoing debate on the potential benefits of early delivery in fetuses with gastroschisis. Groups in favor claim that the longer the bowel is exposed to the amniotic fluid, the worse the damage and the longer the hospital stay, advocating for elective delivery at 34 weeks of gestation. Groups against believe that fetuses should not be exposed to the risks of early delivery and prematurity to protect the bowel from a damage that might not occur [9, 10]. In our clinical practice, we wait for the spontaneous onset of labor; this is because a review of our gastroschisis patients has shown no difference in outcome between patients born at later preterm versus those born at term in terms of time to enteral feedings,

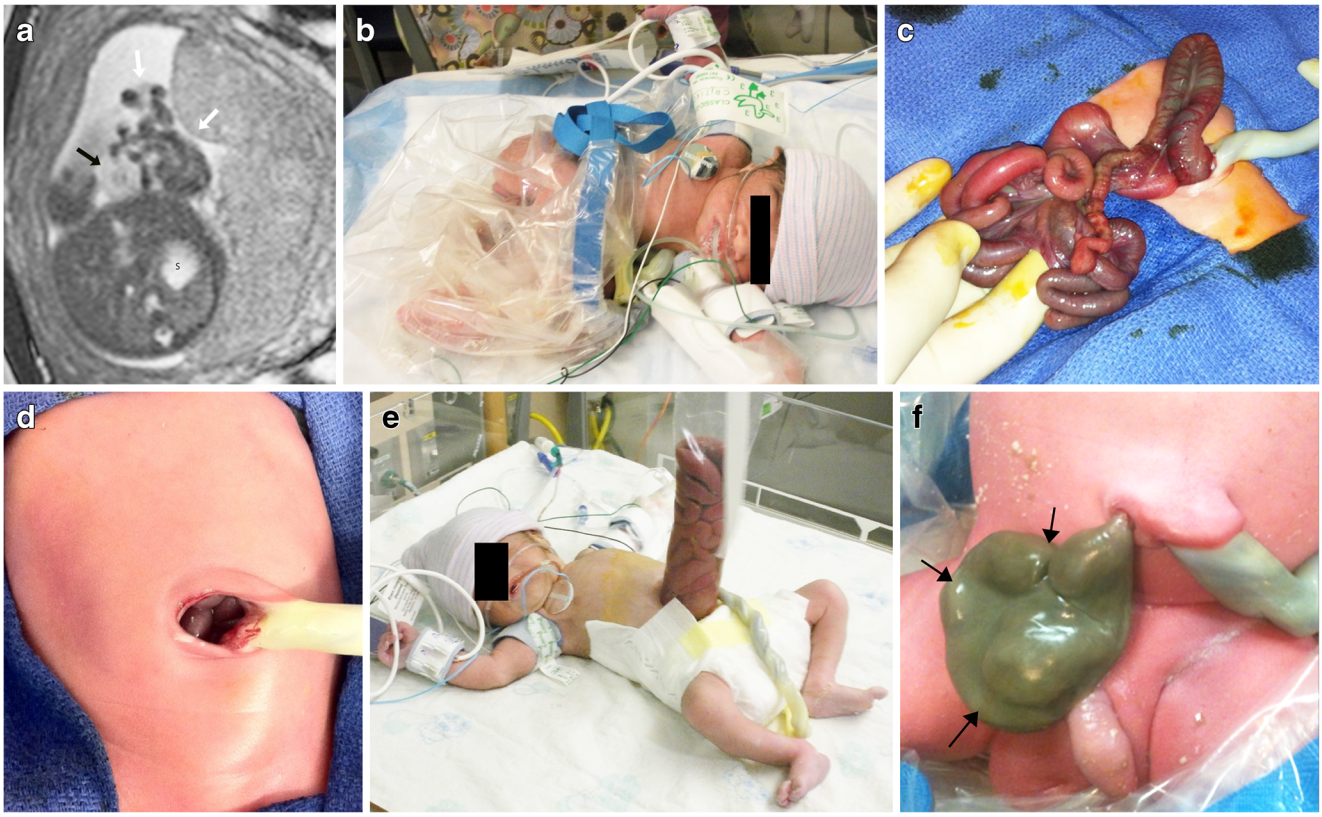

Fig. 2 Gastroschisis in a 22-week fetus. **a** Axial single-shot turbo spin-echo MR image demonstrates free-floating, non-distended bowel loops (*white arrows*) herniating through a parasagittal defect located to the left of the cord insertion (*black arrow*: umbilical cord). Left-side gastroschisis is much less frequent than right-side gastroschisis. *S* stomach. **b** Clinical photograph after birth shows that the lower body of the child with gastroschisis is placed in a Lahey bag to preserve humidity and temperature. **c** Clinical photograph shows primary reduction and closure of gastroschisis. **d** Photograph shows the remaining defect, which is covered by a dressing and closes spontaneously over the following few days. **e** Alternatively, the bowel can be placed into a preformed silo, as in this photograph, with a spring-loaded ring held underneath the abdominal wall. **f** In this case the gastroschisis defect closed before birth, causing incarceration, ischemia and necrosis of the eviscerated bowel (*arrows*). This event almost invariably leads to short-bowel syndrome

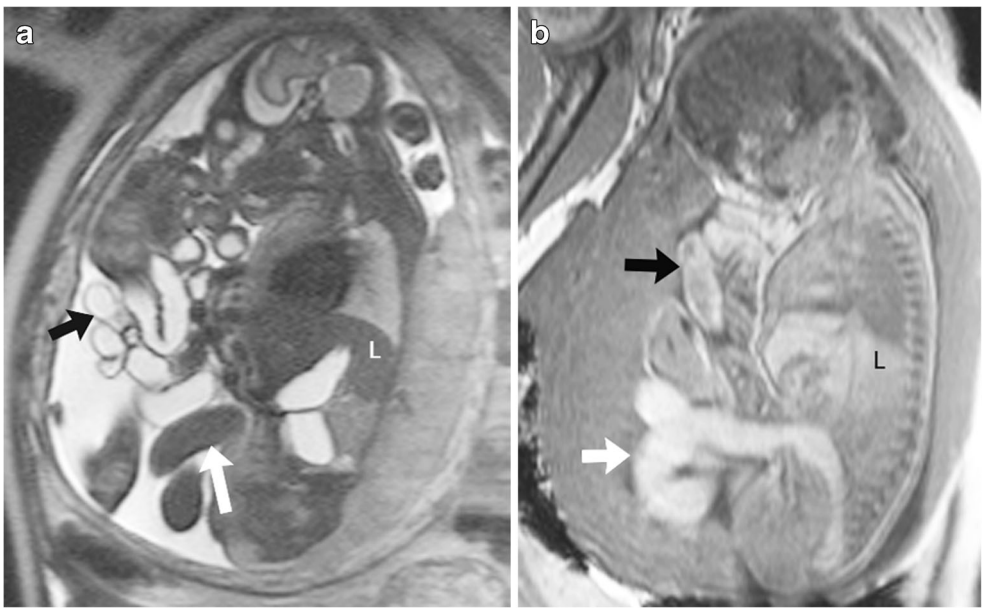

Fig. 3 Complex gastroschisis in a 34-week fetus. **a, b** Sagittal single-shot turbo spin-echo (TSE) (**a**) and T1-weighted (**b**) MR images demonstrate free-floating loops of small bowel (*black arrow*) and large bowel (*white arrow*). Note how the loops of small bowel appear distended and mildly thick-walled, best seen in the TSE image (**a**). The T1-W image demonstrates the meconium-containing herniated distal colon, which then enters the abdominal cavity, taking its expected anatomical place posterior to the bladder. *L* liver

time to stop parenteral nutrition, length of hospital stay and mortality (in press).

Immediately after birth the extruded bowel must be protected with a Lahey bag (Fig. 2). If the bowel is healthy, a primary reduction and closure at birth is an option. The most commonly used repair method, however, consists of placing the bowel in a preformed silo that is squeezed on a daily basis until the bowel is completely reduced, followed by a surgical closure. If the bowel is compromised (e.g., necrosis, perforation), a bowel resection, ostomy or other type of bowel-repair surgical maneuver might be needed. Complex gastroschisis cases usually undergo a silo placement and delayed closure. Intestinal atresias found in the bowel of a child with gastroschisis are generally not repaired at the time of birth but rather 4–6 weeks later, once the swelling of the bowel wall has subsided.

Infants with gastroschisis do not undergo the normal embryologic process of intestinal rotation and have their bowel in a non-rotation configuration, which does not pose a significant risk of midgut volvulus. Moreover, the adhesions that develop after the bowel manipulation make a midgut volvulus even less likely in these babies [11, 12].

In some rare cases the abdominal wall defect closes before birth, causing strangulation of the eviscerated bowel. This can lead to ischemia, necrosis and even amputation of the externalized bowel, an entity termed "vanishing gastroschisis." In less extreme cases the necrotic bowel might still be attached at the time of delivery and present at birth as a conglomerate of scarred tissue with no resemblance to normal bowel (Fig. 2). These are usually catastrophic clinical situations in which different degrees of short bowel will be encountered.

The differential diagnosis of the fetus with free-floating bowel includes, aside from gastroschisis, a ruptured omphalocele and the limb–body wall complex. The main difference between the ruptured omphalocele and gastroschisis is the insertion of the umbilical cord, which in gastroschisis is medial to the defect, whereas for omphalocele the herniated contents do so through the base of the umbilical cord. Differentiation between gastroschisis and limb–body wall complex is usually not a diagnostic dilemma because the latter involves additional severe abnormalities rather than simple herniated loops of bowel, as discussed in the limb–body wall complex section.

Omphalocele

Omphalocele (or exomphalos) is a defect of the anterior abdominal wall with herniation of abdominal contents through the base of the umbilical cord. Its incidence is approximately 1 per 5,000 live births. The defect is covered by a three-layer membrane formed by peritoneum, Wharton's jelly and amnion [13]. The etiology is uncertain, but two theories have been proposed, one in which the lateral folding of the abdominal wall is deficient, and one in which the bowel does not completely return to the abdominal cavity after the physiological herniation that occurs prior to the 12th week of gestation [3]. Given that bowel in the umbilical cord is a normal phenomenon during the first 10 weeks of gestation, a diagnosis of omphalocele should not be made before the 12th week of gestation. Omphaloceles are classified as *small* when the base of the defect is less than 5 cm in diameter; *giant* when the base is more than 5 cm in diameter, or more than 75% of the liver is herniated through the defect; and *ruptured* (Figs. 4, 5, 6, 7, 8, 9 and 10). This classification, however, varies among centers. Some pediatric surgeons define an omphalocele as small if it is amenable to a primary closure shortly after birth and giant if the size is such that a primary closure is not feasible [14–17]. In the case of giant omphaloceles the thoracic cavity might be abnormal in shape and reduced in size, leading to pulmonary hypoplasia and resulting in potential postnatal respiratory insufficiency. MR has been shown to aid in predicting the outcome of the fetus with isolated giant omphaloceles, with those showing lung volumes of less than 50% of expected capacity demonstrating increased postnatal morbidity, including lower Apgar scores at birth, prolonged ventilator support and longer hospitalization periods [18]. At our institution the overall survival in fetuses with isolated giant omphaloceles is excellent (94%). When additional serious abnormalities are present, however, mortality increases dramatically, with reported values as high as 80–100% [19].

The key to identifying an omphalocele is to evaluate the insertion of the umbilical cord, which should be at the base/apex of the omphalocele. This is in contradistinction to gastroschisis (paramedian defect) and bladder exstrophy (infra-umbilical defect, see section on bladder exstrophy). A surrounding membrane is another distinguishing feature of omphaloceles, although this is sometimes difficult to visualize, particularly if plastered against the extruded liver. The presence of ascites, sometimes seen with omphaloceles, might delineate the membrane (Fig. 5). In certain cases, however, the membrane ruptures, resulting in spillage of abdominal contents (Fig. 7); differentiation between a ruptured omphalocele and gastroschisis in this case depends on the insertion of the umbilical cord and the type of intra-abdominal organs eviscerated through the defect.

Once an omphalocele is diagnosed, a careful search for additional abnormalities should be undertaken because this type of abdominal wall defect is seen in conjunction with other abnormalities in 54% of cases, the severity of which determines postnatal prognosis. Chromosomal abnormalities, predominantly trisomies 13 and 18, are seen in 30–40% of patients with omphaloceles, and Beckwith–Wiedemann

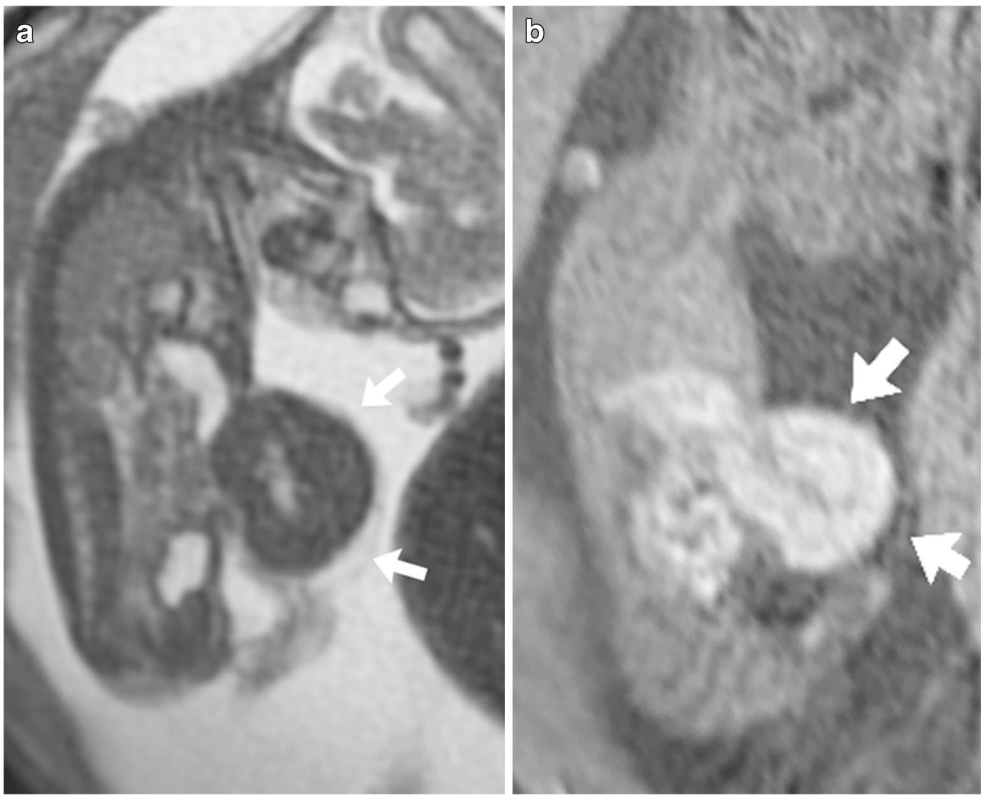

Fig. 4 Liver-only omphalocele. **a, b** Sagittal steady-state free precession (**a**) and T1-W gradient recalled echo (**b**) MR images demonstrate liver-only omphalocele (*arrows*) in a 20-week fetus

syndrome is found in 5–10% of cases (Fig. 8) [20–22]. Of note, small (bowel only) omphaloceles have an increased incidence of chromosomal abnormalities compared to giant omphaloceles.

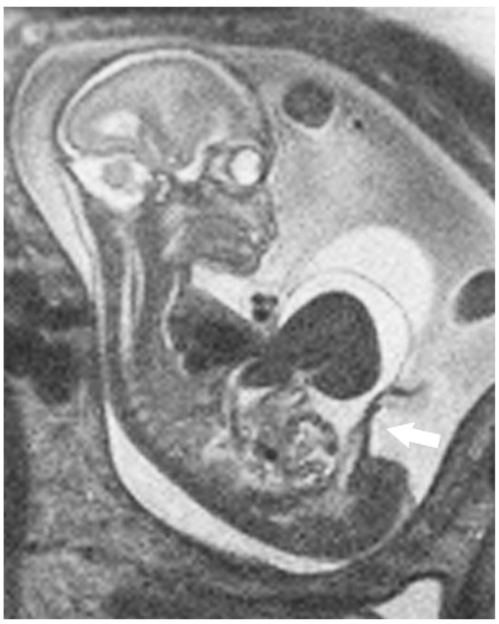

Fig. 5 Giant omphalocele**.** Sagittal single-shot turbo spin-echo MR image shows wide-mouth giant omphalocele containing most of the liver, which is surrounded by ascites in this 20-week fetus. Note the umbilical vessels (*arrow*)

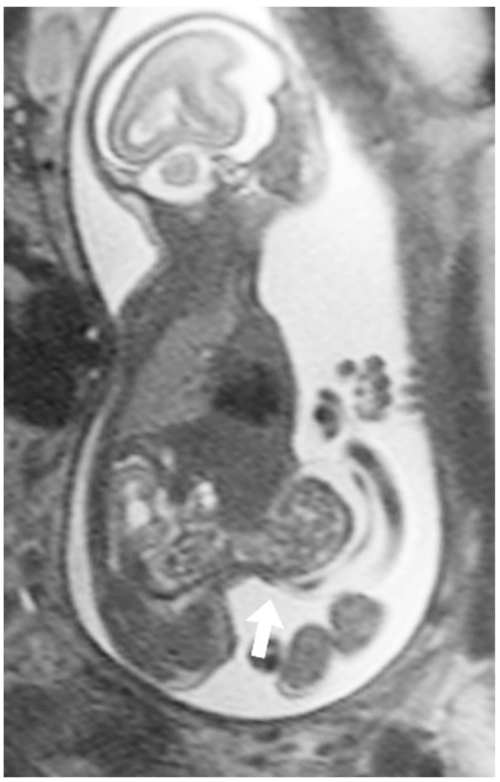

Fig. 6 Bowel-only omphalocele. Sagittal single-shot turbo spin-echo MR image shows bowel-only omphalocele in a 20-week fetus. Note the umbilical vessels (*arrow*)

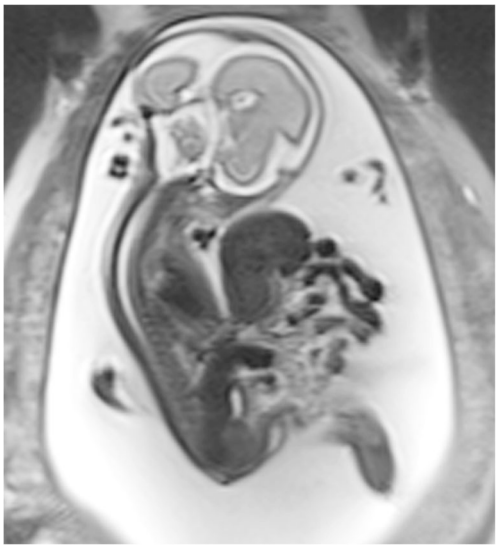

Fig. 7 Ruptured omphalocele. Sagittal single-shot turbo spin-echo MR image shows ruptured omphalocele in a 26-week fetus. Note extruded liver and bowel (small and large) and lack of membrane covering the defect

Most infants with omphaloceles are born at term unless there are complicating features. Fetuses with small omphaloceles can be delivered vaginally whereas those with giant omphaloceles should be delivered by cesarean section. The postnatal management of the neonate with an omphalocele depends on the size of the defect and the condition of the child. Small omphaloceles are generally amenable to primary closure [6]. Giant omphaloceles might be managed by a staged closure technique (if the child is medically stable) or by a delayed closure ("paint and wait") technique. In the staged closure technique, a mesh is sutured to the fascia to form a silo, which is reduced over the course of 1–2 weeks followed by a definitive fascial closure. In the "paint and wait" technique, the membrane is covered by xeroform gauze for months, allowing an eschar to form over the intact amnion sac, which then epithelializes over time; the actual fascial closure is often postponed for 1–4 years (Fig. 9).

Exstrophy-epispadias complex

Bladder and cloacal exstrophy are abdominal wall defects that present in a spectrum from most severe (cloacal exstrophy) to least severe (epispadias). Cloacal exstrophy involves the entire lower half of the body including the hindgut, bladder and genitalia, and is associated with a series of other defects. Bladder exstrophy involves the bladder and external genitalia,

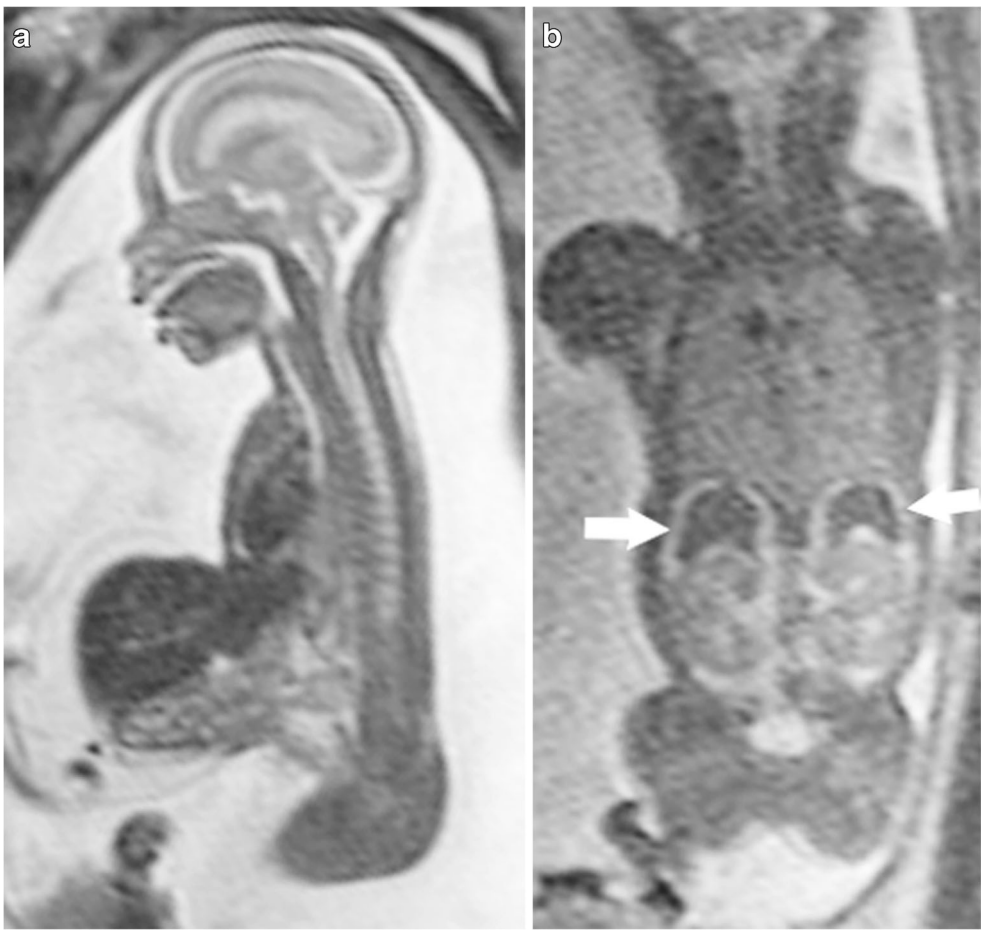

Fig. 8 Omphalocele spectrum. **a, b** Sagittal (**a**) and coronal (**b**) single-shot turbo spin-echo MR images in a 19-week fetus with Beckwith–Wiedemann syndrome. Note the liver- and bowel-containing omphalocele in image (**a**) and marked adrenomegaly (*arrows* in **b**)

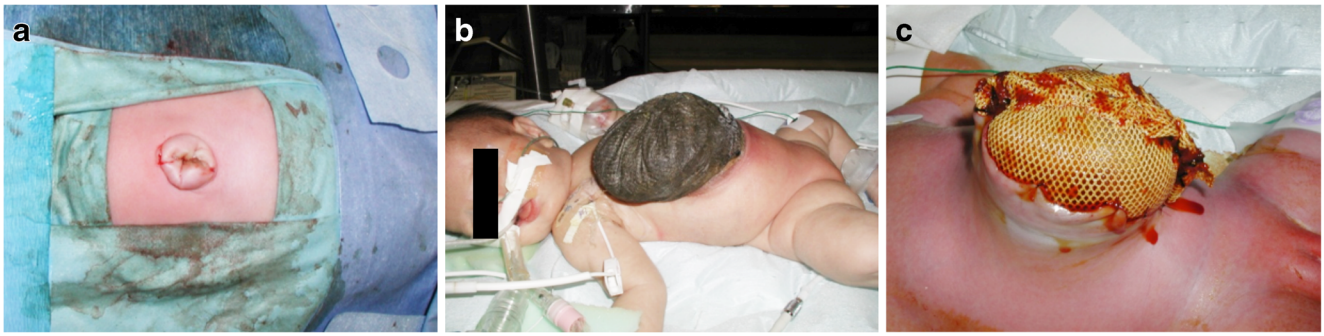

Fig. 9 Repair of omphalocele. Postnatal clinical photographs demonstrate different approaches to omphalocele repair: (**a**) primary closure of a small omphalocele, (**b**) "paint and wait" delayed closure technique and (**c**) staged closure technique

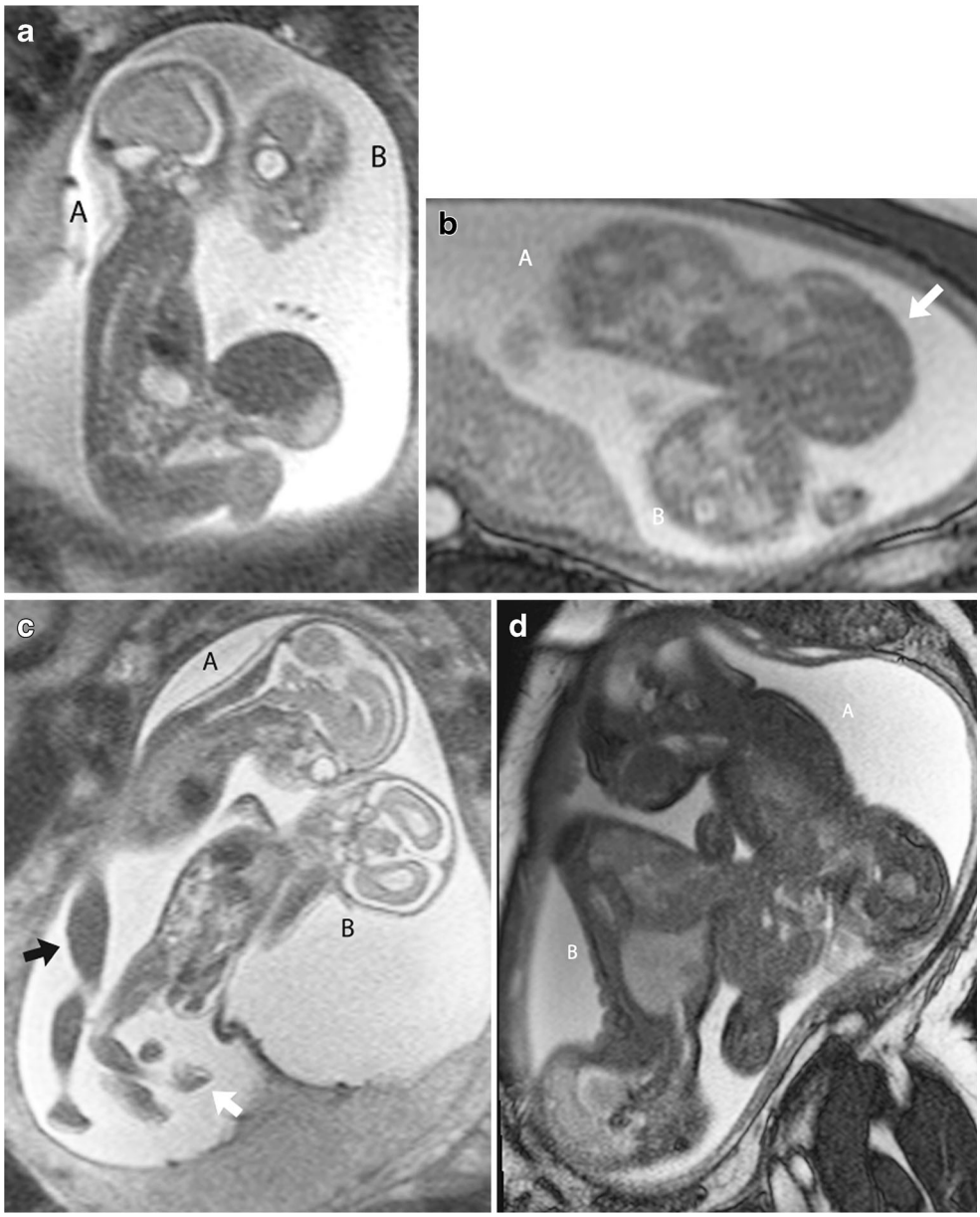

Fig. 10 Conjoined twins sharing a liver and bowel omphalocele. **a, b** Coronal single-shot turbo spin-echo (**a**) and axial steady-state free precession (**b**) MR images demonstrate the vertex fetuses (marked as A and B) at 19 weeks of gestation. The shared extruded liver (*arrow*) is best seen in the axial image (**b**). **c** Single-shot turbo spin-echo MR image demonstrates evidence of arthrogryposis and muscle wasting in the B fetus, denoted by increased T2 signal in the soft tissues of the lower extremity and decreased muscle mass when compared to those of fetus A (*black arrow*). Unlike fetus A, fetus B had a small chest cavity, a club foot (*white arrow*) and persistently flexed upper extremities (not shown). **d** At 32 weeks of gestation, the mother returned to our institution for follow-up, at which time fetus A was noted to be in breech presentation while fetus B remained vertex (sagittal steady-state free precession image). Given that B demonstrated extensive muscle wasting and contractures, it was postulated that A had rotated around the axis of the omphalocele while B remained in unchanged position. Three days later the mother had intrauterine fetal demise of both twins, possibly secondary to vascular compromise around the shared hepatic pedicle

while epispadias involves only the urethra and bladder neck. Isolated epispadias, the least severe of these entities, is usually not imaged by MR prenatally and is not discussed further in this paper.

Bladder exstrophy

Bladder exstrophy occurs in 1 per 33,000 live births and is more common in males than females (5:2) [23]. It is caused by inappropriate retraction of the cloacal membrane with subsequent eversion of the bladder plate. The postnatal clinical findings in bladder exstrophy are notable for an everted, open posterior bladder plate and an epispadic urethra. In boys, the corpora cavernosa of the penis are shorter and wider than normal, with wide separation of the corporal attachments; in girls the clitoris is bifid, there is wide separation of the labia, and there may be uterine and vaginal anomalies [24]. The umbilical cord is low-set and the symphysis pubis is always widened. The entire pelvic anatomy is altered, with externally and anteriorly rotated pelvic bones, a flatter and wider puborectal sling and divergence of the levator ani muscles. The anus is usually anteriorly displaced [25].

The hallmark of bladder exstrophy on prenatal imaging is an absent urinary bladder in the presence of normal amniotic fluid and lung volumes (Fig. 11). Direct eversion of the bladder plate is difficult to see prenatally. Instead, a soft-tissue mass representing the mucosa is noted to be protruding from the infraumbilical abdominal wall. The distal ureters join the mass, and the urine drains directly into the amniotic cavity. The gastrointestinal tract is otherwise normal in bladder exstrophy, findings that can be well evaluated by means of fetal MR: in bladder exstrophy, fetal MRI shows a normal column of T1-hyperintense meconium reaching the pelvic floor after week 20 of gestation [26], unlike in cloacal exstrophy.

Differentiating an omphalocele from a case of bladder exstrophy relies, in part, on the presence or absence of a bladder (present in an omphalocele, absent in bladder exstrophy) and the position of the umbilical cord (apical/base in omphalocele, and superior to the defect in bladder exstrophy). The differential diagnosis of the fetus with non-visualization of the bladder in the presence of normal amniotic fluid includes bladder exstrophy and cloacal exstrophy. With bladder exstrophy, a normal hindgut should be identified, whereas in cloacal exstrophy the hindgut is abnormal (see next section).

The prenatal diagnosis of bladder exstrophy is important for guiding postnatal care. Vaginal delivery is not contraindicated. The primary objectives in bladder exstrophy repair are to close the bladder, to create outlet resistance that is sufficient to enlarge the bladder but not high enough to impair its function, to reconstruct the genitalia for appearance and function, and ultimately to achieve urinary continence [27]. The surgical repair involves bladder closure, with dissection to free the bladder from its superficial attachments in order to place it deep into the pelvis; epispadias repair; and genitoplasty. Bilateral pelvic osteotomies are also crucial to loosen the pelvic bones and allow repair of the pubic diastasis. While traditionally surgery was performed in the first 72 h after birth, at our institution we prefer to delay the surgery until 2–4 months of age. This waiting period allows for parental bonding and for normal growth and development of organs before the complex surgery. Referral to a tertiary hospital

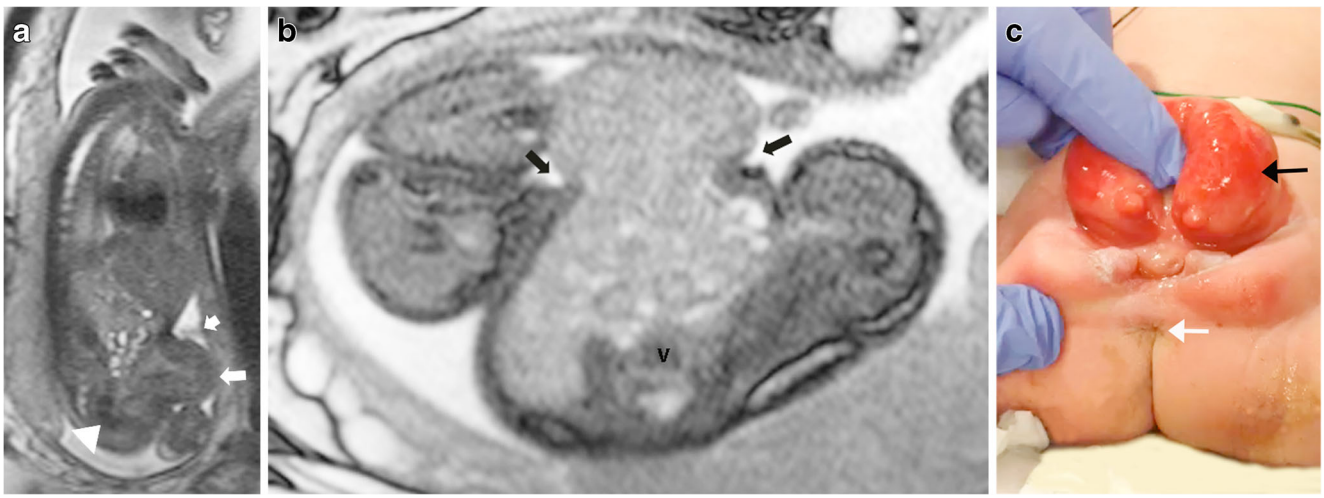

Fig. 11 Bladder exstrophy in a 29-week female fetus. **a** Sagittal single-shot turbo spin-echo MR image through the midline demonstrates the extruded and everted bladder (*long arrow*), a normal hindgut manifested by the normal morphology and position of the colon (*arrowhead*), and the exit site of the umbilical vessels, superior to the abdominal defect (*short arrow*). Of note, the kidneys and amniotic fluid volume were normal (not shown). **b** Axial steady-state free precession MR image clearly depicts the intersection between the normal abdominal wall and the extruded bladder (*arrows*). *V* vertebra. **c** Clinical photograph of the child after birth. Note the everted bladder (*black arrow*) and the anteriorly placed anus (*white arrow*)

with expertise in this rather rare and highly complex anomaly is strongly suggested [27].

Cloacal exstrophy

Cloacal exstrophy, also known as the OEIS (omphalocele, cloacal exstrophy imperforate anus, spinal defects) complex, occurs in approximately 1 per 200,000–400,000 live births, with no gender predilection. The etiology is unknown but genetic and environmental factors might play a role. As in bladder exstrophy, the entity involves an infra-umbilical wall defect, non-visualization of the bladder, and normal amniotic fluid volume. In the case of cloacal exstrophy, the defect includes exstrophy of the rudimentary hindgut and prolapsing of the ileum from the cecal plate, which leaves the bladder as two separate halves on either side of the bowel. These bladder plates each receive one ureter into the back wall.

In normal embryonic development, the gastrointestinal and genitourinary tracks empty into a common cloaca before the fifth week of gestation (Fig. 12). At the distal end of the cloaca lies the cloacal membrane. During the sixth week of gestation, the urorectal septum extends caudal toward the cloacal membrane, and by the eighth week the cloaca is divided into an anterior chamber (the primitive urogenital sinus) and a posterior chamber (the rectum; Fig. 12). In the fetus with cloacal exstrophy, however, there is an early breakdown of the cloacal membrane with eversion of the hemibladders and hindgut (Fig. 12) [28]. The existing bowel herniates between the two bladder halves, creating the characteristic "elephant trunk" appearance. This elephant trunk can be seen on fetal MR as a meconium-filled, T1-hyperintense blind-ending segment of bowel protruding between the hemibladders (Figs. 13 and 14). Anal atresia is invariably seen, with absence of an anal dimple [29]. An omphalocele is usually seen cranial to the extruded cloaca (with the cord in this case inserting in the usual anatomical location expected for an omphalocele and superior to

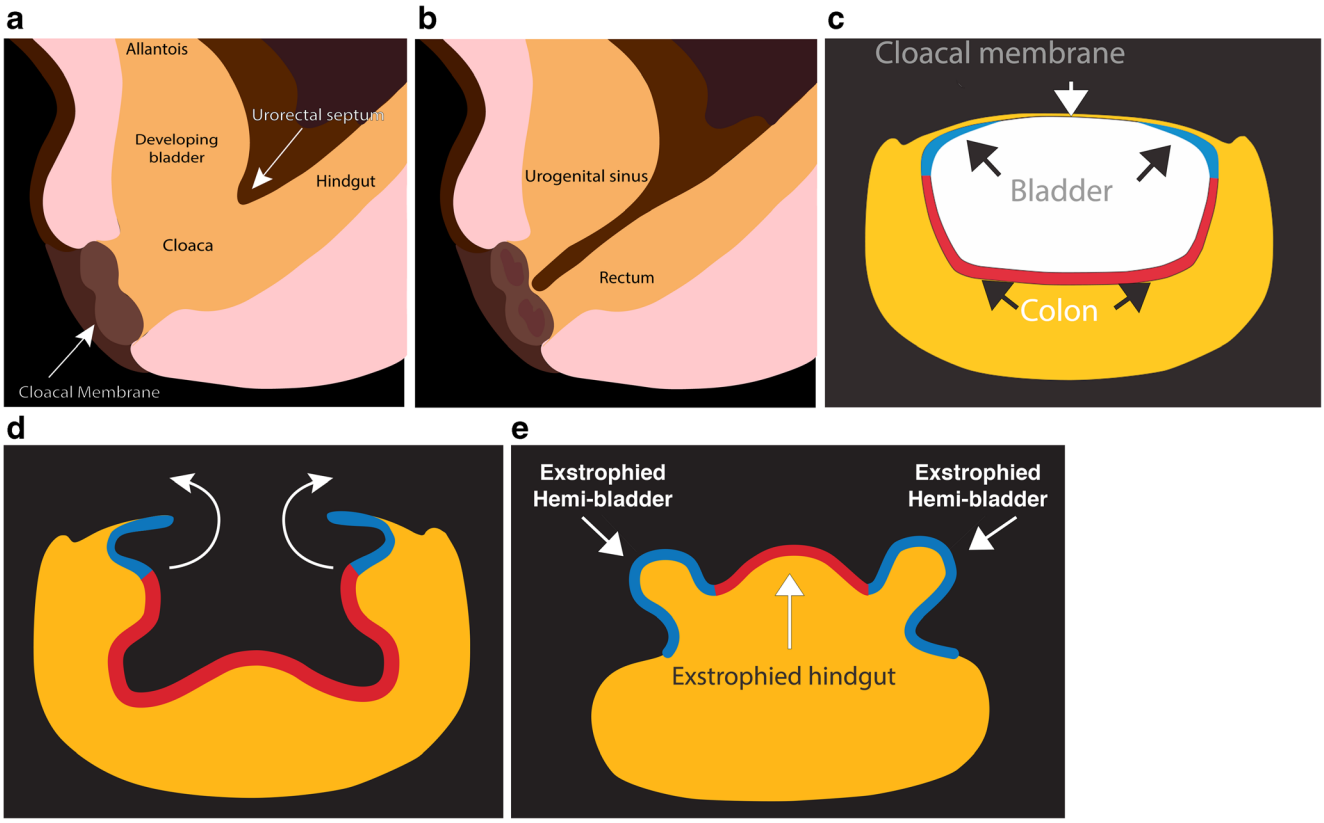

Fig. 12 Cloacal exstrophy. **a** Diagram of normal early gestation (week 5), when the cloaca is divided into a dorsal and a ventral part by a wedge of mesenchyme called the urorectal septum. **b** The urorectal septum then grows caudal toward the cloacal membrane, which ruptures, leading to the creation of two distinct parts by the eighth week of gestation: the rectum and cranial parts of the anal canal dorsally, and the urogenital sinus ventrally. The urogenital sinus in turn gives rise to the bladder and urethra, and in females, portions of the vagina. **c**–**e** Sequential diagrams demonstrate transverse sections at the level of the hindgut in a fetus with cloacal exstrophy. In cloacal exstrophy the urorectal septum fails to grow caudally, leading to premature rupture of the cloacal membrane and exstrophy of the persistent cloaca and rudimentary hindgut. Images (**a**) and (**b**) reprinted with permission from Elsevier; images previously published in [1]. Images (**c**), (**d**) and (**e**) reprinted with permission from Wolters Kluwer Health

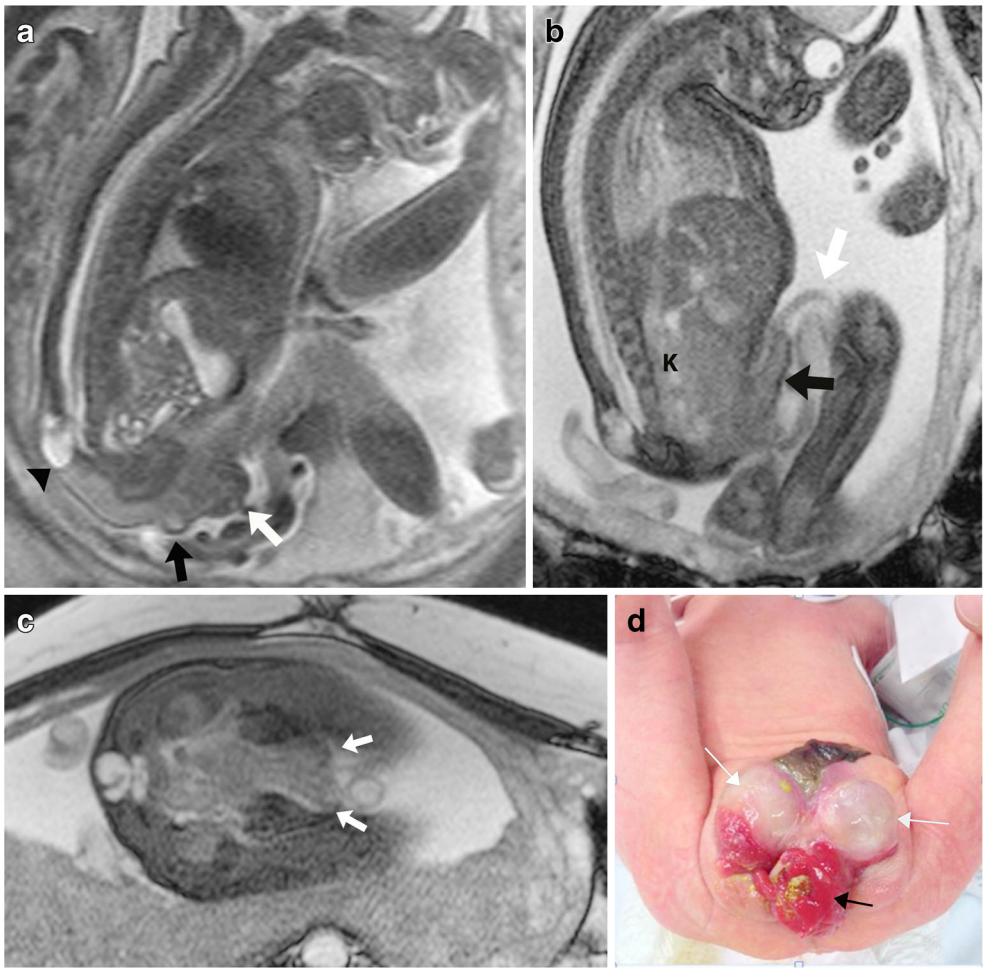

Fig. 13 Cloacal exstrophy in a 28-week-old genotypically male fetus. **a** Sagittal single-shot turbo spin-echo MR image demonstrates one of the two everted hemibladders (*white arrow*), a dysmorphic scrotal sac (*black arrow*) and a lipomyelomeningocele (*black arrowhead*) with a tethered cord. **b** Sagittal steady-state free precession MR image demonstrates the characteristic midline "elephant trunk" (*black arrow*), or bowel that extruded between the two hemibladders. The right kidney (*K*) was positioned anterior to the spine, which terminated abruptly at the mid-sacral level. Note the umbilical vessels (*white arrow*). In this case an omphalocele was not present, suggesting that this was a forme fruste of cloacal exstrophy. **c** Axial steady-state free precession MR image through the pelvis demonstrates the two bladder plates (*arrows*). **d** Clinical photograph after birth; black arrow indicates the terminal ileum, white arrows the hemibladders

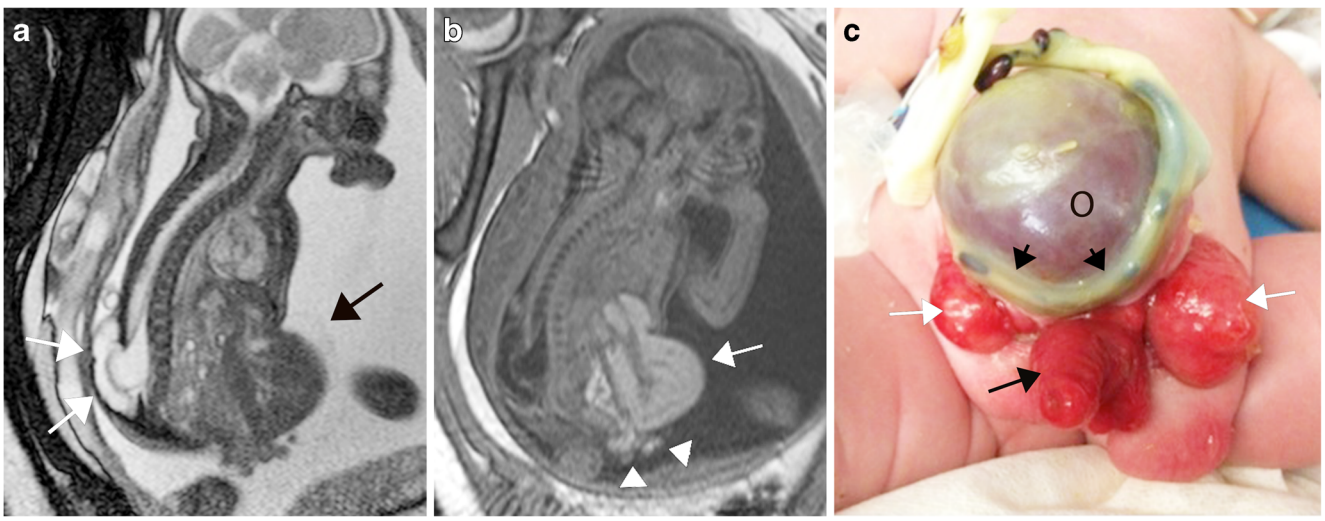

Fig. 14 Cloacal exstrophy in a 28-week-old genotypically male fetus. **a** Sagittal steady-state free precession MR image demonstrates a membrane-covered omphalocele (*black arrow*) containing liver and bowel. Lack of bladder visualization is consistent with the diagnosis of exstrophy. Note the large posterior neural tube defect consisting of a terminal myelocystocele (*white arrows*). The sacral elements were abnormally hypoplastic. **b** Sagittal T1-W MR image demonstrates to better advantage the T1-hyperintense extruded liver (*arrow*) and meconium-containing bowel (*arrowheads*). Note the absence of a normal T1-hyperintense column of meconium-containing colon, which would be positioned immediately anterior to the spine. **c** Clinical photograph after birth. Note the omphalocele (*O*), the terminal ileum (*long black arrow*) and the hemibladders (*white arrows*). The cord insertion is at the base of the omphalocele, superior to the exstrophy complex (*short black arrows*)

the cloacal complex). The pubic diastasis seen in cloacal exstrophy is more severe than in classic bladder exstrophy, although these findings are difficult to see at fetal MR [30]. Spinal defects are common, including incomplete development of the lumbosacral segments, tethered cord and closed neural tube defects. A persistent umbilical cyst is sometimes seen during the early second trimester if there is delayed rupture of the cloacal membrane. Associated malformations might also involve the kidneys (agenesis, cystic dysplasia, ectopia) and include lower-extremity defects, ascites, narrow thorax and a single umbilical artery [31].

Although the ultimate goals for repairing cloacal exstrophy are the same as for bladder exstrophy, the initial management is quite different. In the first few days of life, the hindgut is dissected free of the bladder plates and inferior abdominal wall, the small intestine is re-tubularized, and a colostomy is brought up to the abdominal wall. It is important to salvage as much hindgut as possible because it can later be used for bladder reconstruction, if needed [32]. At the time of the initial surgery for the colostomy, the posterior walls of the two hemibladder plates are brought into continuity to create a template for later bladder closure. The second stage of closure, which usually occurs after 1–2 years, is to close the bladder and to perform urethral reconstruction and epispadias repair. This requires osteotomies that are usually done in a staged approach, with gradual re-approximation of the pubic bones. Unlike in classic bladder exstrophy, continence is not usually attainable with bladder closure and urethral reconstruction alone, but more often requires bladder closure, augmentation and diversion.

In regard to sexual identity and function, genotypic females with cloacal exstrophy might require vaginal reconstruction, which can be achieved by bringing existing vaginal structures to the perineum or creating a neovagina by means of skin flaps or intestine. The treatment of the genotypically XY male with cloacal exstrophy has evolved through time. In these infants the phallus is usually divided and small, and the hemicorpora might be unequal in size. In the second part of the last century, studies advocated female gender assignment for male neonates with minimal phallic structures, the surgical approach involving orchiectomy and removal of the rudimentary existing phallus [33]. A clitoris was made out of the glans of the penis, labial walls were created out of the scrotal walls and vaginal reconstruction was undertaken. Female gender reassignment for XY babies with cloacal exstrophy has been abandoned. The current strategy for XY patients is based on penile reconstruction, where the bifid phallus and split corpora are brought together in the midline and penile growth is stimulated by means of testosterone administration.

Differentiating classic bladder exstrophy from cloacal exstrophy can sometimes be challenging, noting that an erroneous diagnosis can drastically change the expected postnatal management and course. The ability to distinguish between bladder exstrophy, cloacal exstrophy and omphalocele can be further hindered in fetuses with classic bladder exstrophy who have large bladder plates and wide pubic diastasis because in this setting the everted bladder can be mistaken for an omphalocele. Carefully tracing the course of the umbilical cord insertion and assessing the presence or absence of a bladder and the appearance of the hindgut and spine are essential in order to reach the correct diagnosis.

Pentalogy of Cantrell

First described by Cantrell et al. [34] in 1958, the full pentalogy includes (1) midline supra-umbilical abdominal defect, (2) defect of the lower sternum, (3) deficiency of the diaphragmatic pericardium, (4) deficiency of the anterior diaphragm and (5) cardiac abnormalities. The estimated incidence is about 6 per 1 million live births, with a male predominance of 3:1. Most cases are sporadic but familial cases have been reported. Chromosomal anomalies such as trisomies 13 and 18 have been reported in conjunction with pentalogy of Cantrell. Its cause is thought to be an error in formation and migration of the ventral mesoderm during the second to fifth weeks of gestation, with failure of fusion of the transverse septum of the diaphragm and lateral folds of the thorax resulting in diaphragmatic and cardiac defects [34, 35]. The abdominal wall defect most commonly seen is an epigastric omphalocele (Fig. 15). Cardiac defects are often present, with ventricular septal defect being the most common, followed by atrial septal defects, left ventricular diverticulum, pulmonary atresia, tetralogy of Fallot, dextrocardia and transposition of the great arteries, among others. Pentalogy of Cantrell carries a high mortality, with survival rates calculated to be less than 40%.

Ectopia cordis (from Greek *ektopos* or "out of place" and the Latin *cordis* or "heart") is a rare congenital defect in which the heart is partly or completely positioned outside the thoracic cavity, with the heart extruded through a sternal defect, usually an inferior sternal cleft. It was first reported in 1671 by the Danish Neil Stensen, who described "the sternum was split and the heart, liver and spleen, most of the intestine and right kidney have passed out through the split being thus uncovered" [36]. The ectopic heart might be situated anywhere from the neck to the abdomen, but most commonly protrudes outside the thoracic cavity through a defect in the sternum. Complete ectopia cordis is considered a neonatal emergency and is typically fatal, with the more common partial ectopia cordis demonstrating an overall better outcome.

Postnatal management of pentalogy of Cantrell depends on the concomitant abnormalities, with children with severe forms of the disease undergoing single or staged surgical repair after birth.

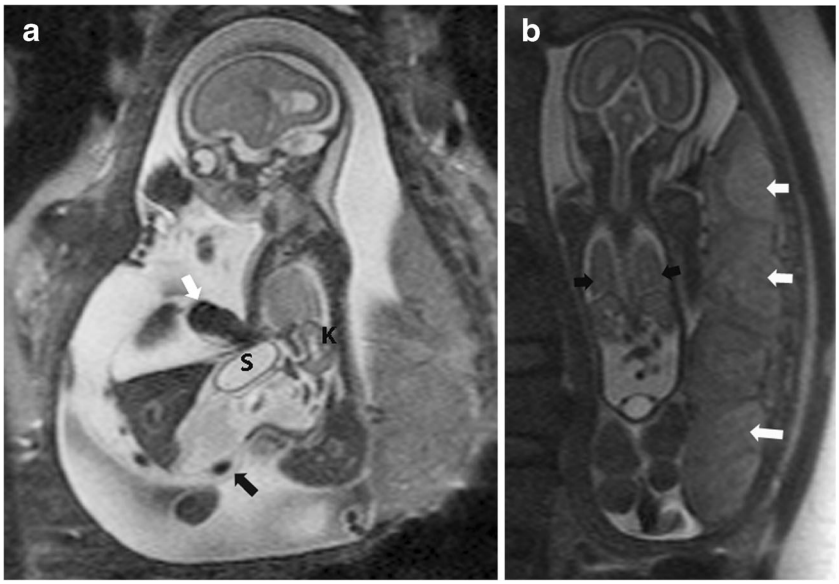

Fig. 15 Pentalogy of Cantrell in a 22-week fetus. **a** Sagittal single-shot turbo spin-echo MR image demonstrates the dysmorphic and extruded heart (*white arrow*), the stomach (*S*) and the deformed triangular-shape liver. Note the cord exiting through the base of the membrane-covered abdominal wall defect (*black arrow*). The inferior pole of the kidney (*K*) is mildly displaced; notice the adrenal gland, displaced anterior to the kidney. **b** Coronal single-shot turbo spin-echo MR image demonstrates partially collapsed lungs (*black arrows*) surrounded by pleural fluid; placentomegaly and abnormally prominent cotyledons (*white arrows*) for this gestational age are also noted

In the differential diagnosis of the fetus with an abdominal wall defect, pentalogy of Cantrell is the most likely diagnosis when encountering an omphalocele of the upper abdominal wall in association with ectopia cordis. If these findings are seen in unison with scoliosis or other spinal or limb anomalies, limb–body wall complex should then be considered.

Limb–body wall complex

Limb-body wall complex, also known as body stalk anomaly, amniotic band disruption complex and amniotic rupture sequence, refers to a pattern of severe fetal polymalfomations characterized by the presence of at least two of the following anomalies: (1) thoraco- or abdominoschisis, sometimes with complete evisceration of the abdominal contents; (2) limb reduction and malformations including clubfoot as well as partial or complete amputations; and (3) exencephaly or encephaly with facial defects [37]. Multiple other abnormalities might be encountered including ectopia cordis, neural tube defects, severe scoliosis, spinal anomalies and visceral malformations [38]. Partial adherence of fetal parts to the placenta or uterine wall might be encountered (Fig. 16). The eccentric body wall defect is typically large and lateral in location, usually on the left side (unlike gastroschisis, where right-side defects are the norm). There might be persistence of the extra-embryonic coelom with separation of the amnion and chorion [1, 3]. A short and uncoiled umbilical cord might be also seen, its appearance in part secondary to the lack of fetal mobility (umbilical cord coiling is thought to be caused by fetal movement during the pregnancy).

Limb–body wall defect occurs rarely (1:14,000–1:42,000 pregnancies) and is uniformly fatal. The etiology is unknown, although several theories have been set forward. One such theory invokes an early rupture of the amnion, resulting in the creation of fibrous bands that in turn cause traumatic lesions of the fetal body [38]. Other proposed explanations include a vascular insult during the first 4–6 weeks of gestation leading to failure of ventral body wall closure, and teratogenic exposure (including cocaine). None of the proposed theories, however, explains fully the phenotype encountered, suggesting that the true causative agent might be multifactorial.

The fetus with limb–body wall defect usually has a normal karyotype and no suspected genetic etiology, thus minimizing the risk of a subsequently affected pregnancy. There is no fetal treatment and there is invariably early postnatal death; management is usually supportive. Recognition and differentiation between this universally fatal entity and other abdominal wall defects is crucial. In the differential diagnosis of this polymalformative sequence the other entity that could be entertained is pentalogy of Cantrell. Of these, limb–body wall defect is

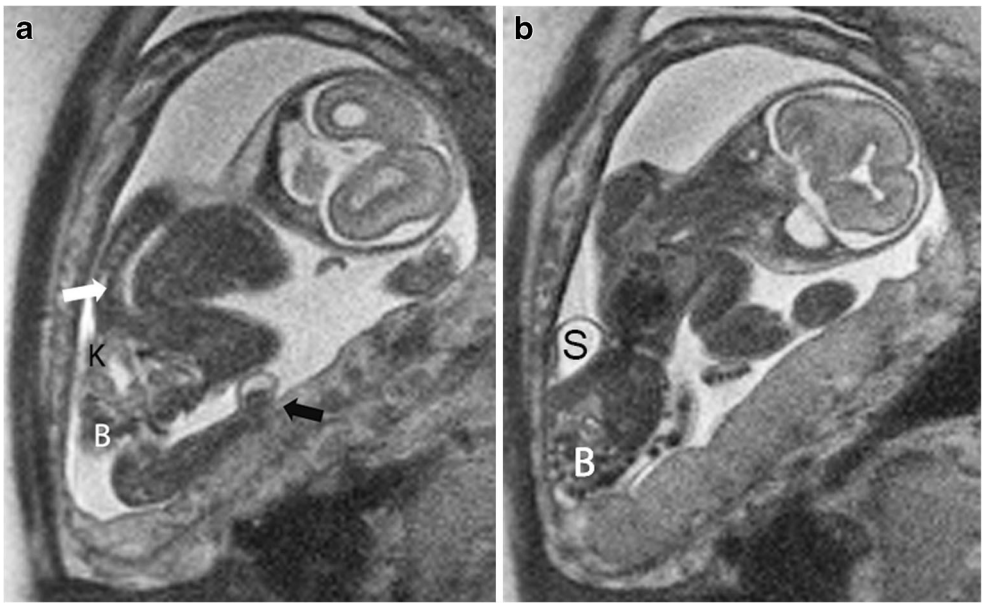

Fig. 16 Limb–body wall defect in a 20-week fetus (single-shot turbo spin-echo image). **a** Note the marked scoliosis (*white arrow*), the extruded kidney (*K*), the matted and herniated bowel (*B*) and the club foot (*black arrow*). **b** Stomach (*S*) and liver are also extra-abdominal without membrane covering them. The extracorporeal components are matted together, with bowel and part of liver adhered to the uterine wall. This fetus also had a short and unwound cord (not shown), frequently seen in cases of limb–body wall defect

associated with scoliosis, which suggests the diagnosis. Only in this entity can the loops of bowel be matted and adhered to the placenta, or can fetal segments be adhered to the chorion through a ruptured amnion. The defect is usually to the left of the cord, whereas in gastroschisis it is usually to the right, in exstrophy it is infra-umbilical, and in omphalocele the cord insertion is at the base of the defect. The presence of a short and unwound cord is another indicator of limb–body wall defect sequence.

Teaching points

1. When encountering a suspected abdominal wall defect, careful and detailed evaluation of the umbilical cord insertion is warranted, its site being a helpful clue to the correct diagnosis:

    a. Apex/base of defect: omphalocele
    b. Paramedian: gastroschisis/limb–body wall defect
    c. Superior to the defect: exstrophy.

2. Factors to evaluate include:

a. Membrane-covered defect vs. uncovered (omphalocele vs. gastroschisis, ruptured omphalocele, limb–body wall defect)
b. Presence vs. absence of bladder with normal amniotic fluid (omphalocele, gastroschisis, limb–body wall defect, pentalogy of Cantrell vs. bladder exstrophy or cloacal exstrophy)
c. Normal amniotic fluid, absent bladder with normal vs. abnormal gastrointestinal tract (bladder exstrophy vs. cloacal exstrophy).

3. Careful evaluation of the gastrointestinal tract is warranted: an abnormal hindgut in the absence of a bladder would indicate cloacal exstrophy; if there is normal distal bowel in the absence of a bladder, a diagnosis of bladder exstrophy is suggested.
4. Always search carefully for additional abnormalities. Many of these entities are associated with additional malformations and syndromes (i.e. omphalocele and Beckwith–Wiedemann), and a thorough fetal survey should always be undertaken.

Conclusion

Abdominal wall defects are challenging diagnostic anomalies that range from the very benign umbilical cord defect to the highly complex exstrophies, limb–body wall complex and pentalogy of Cantrell. Careful prenatal analysis of the umbilical cord insertion and all other anatomical features by means of US and MRI is the key to determining the correct diagnosis. Accurate diagnosis is imperative for appropriate prenatal counseling, delivery planning and postnatal treatment of the fetus carrying an abdominal wall defect.

Compliance with ethical standards

Conflicts of interest None

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