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Fetal abdominal wall defects

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The most common fetal abdominal wall defects are gastroschisis and omphalocele, both with a prevalence of about three in 10,000 births. Prenatal ultrasound has a high sensitivity for these abnormalities already at the time of the first-trimester nuchal scan. Major unrelated defects are associated with gastroschisis in about 10% of cases, whereas omphalocele is associated with chromosomal or genetic abnormalities in a much higher proportion of cases. Challenges in management of gastroschisis are related to the prevention of late intrauterine death, and the prediction and treatment of complex forms. With omphalocele, the main difficulty is the exclusion of associated conditions, not all diagnosed prenatally. An outline of the postnatal treatment of abdominal wall defects is given. Other rarer forms of abdominal wall defects are pentalogy of Cantrell, omphalocele, bladder exstrophy, imperforate anus, spina bifida complex, prune–belly syndrome, body stalk anomaly, and bladder and cloacal exstrophy; they deserve multidisciplinary counselling and management.

Introduction

The most common abdominal wall defects include ectopia cordis, bladder exstrophy gastroschisis, and omphalocele. The first three are classified in the group of ventral body wall defects, share a similar origin, and are likely to be caused by an abnormal closure of the later body wall folds that approach each other in the midline and close by the end of the sixth postmenstrual week. Alternative hypotheses for the cause of gastroschisis, such as amniotic membrane rupture at the insertion of the cord, abnormal apoptotic patterns during regression of the right umbilical vein, or vascular damage to the

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base of the umbilicus caused by disruption of the omphalomesenteric artery, provide less convincing explanations [1]. Omphalocele is a separate entity, whose origin is attributed to a failure of physiologically herniated bowel loops to return from the umbilical cord to the abdominal cavity by the 12th postmenstrual week [1].

Epidemiology

In this chapter, we mainly concentrate on gastroschisis and omphalocele: they are the two most common anomalies. In 2011, based on the European registries included in the EUROCAT network [2], the total prevalence of gastroschisis was 3.09 per 10,000 births, with a live birth prevalence of 2.63 per 10,000 [2]. The corresponding figures for omphalocele were 3.29 and 1.13 per 10,000. The prevalence of gastroschisis has increased from 1980 to 2011, whereas that of omphalocele has remained stable (Fig. 1). A similar trend for an increasing prevalence of gastroschisis has been reported in North American populations, involving particularly pregnancies from younger women of non-Hispanic white maternal race and ethnicity [3]. It is speculated that these different trends are related to the cause of the two conditions. Although omphalocele seems to be mainly genetically determined, some evidence suggests that poor socioeconomic status and prenatal care, as well as teratogens (e.g. recreational drugs, salicylates, paracetamol, and pseudoephedrine) may be important contributors to the development of gastroschisis [4,5].

Another important historical trend has been the anticipation of the prenatal diagnosis of abdominal wall defects. This is because omphalocele and gastroschisis are easily diagnosed at the 11–14 weeks nuchal scan: a recent study based on over 45,000 pregnancies reported a sensitivity of 100% for both abnormalities [6], whereas a systematic review of the literature found a sensitivity near to 90% [7]. The increasing diffusion of first-trimester screening for chromosomal abnormalities has, therefore, increased the proportion of cases of these two abnormalities diagnosed at the nuchal scan. In the EUROCAT network, during the period 2007–2011, 22% of the chromosomally normal cases of gastroschisis were diagnosed before 14 weeks, and 50% between 14 and 23 weeks. The corresponding figures for euploid omphalocele were 35% and 30%. The overall prenatal detection rate was 91.6% for gastroschisis and 83.3% for omphalocele [2].

Gastroschisis

Gastroschisis is observed on ultrasound as a full-thickness defect in the abdominal wall, in most cases to the right of the insertion of the umbilical cord. It is only rarely located to the left of cord

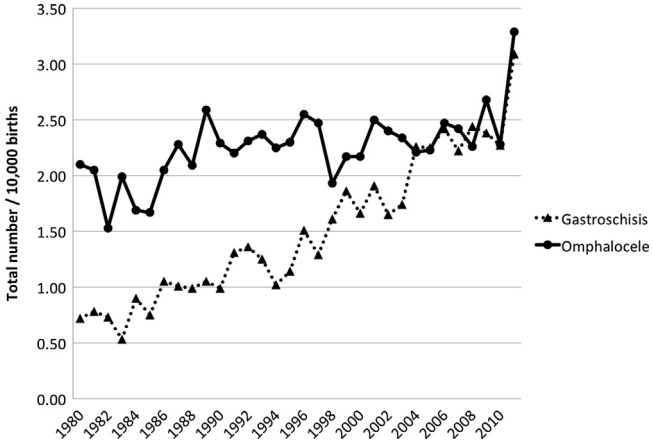

Fig. 1. Total prevalence of gastroschisis and omphalocele (cases per 10,000 births) in the EUROCAT network, 1980 to 2011.

insertion. Bowel loops and occasionally parts of other abdominal organs herniate outside the abdominal wall with no covering membrane or sac (Fig. 2).

A diagnosis of gastroschisis should prompt a careful search for other abnormalities. No close association exists with aneuploidies; therefore, most centres do not actively offer fetal karyotyping in these fetuses. Mastroiacovo et al. [8] analysed 3322 cases of gastroschisis from 24 birth-defect registries worldwide, and found that 469 (14.1%) cases were registered as 'non isolated,' including 41 chromosomal syndromes, 24 other syndromes, and 404 multiple congenital anomalies. Among cases of multiple congenital anomalies, two patterns emerged, with 26 cases resembling limb–body wall complex and 26 others resembling the omphalocele, bladder exstrophy, imperforate anus, spina bifida (OEIS) complex (see below in the omphalocele paragraph). In both situations, omphalocele rather than gastroschisis is more commonly reported, and the investigators argued that these cases might represent misdiagnoses of the abdominal wall defect. Therefore, combining their review with other published reports, the investigators stated that the best estimate of the proportion of gastroschisis associated with major unrelated defects is about 10%. Similar figures were reported in another registry-based study [9], and in a recent prenatal series [10]. Arthrogryposis may also be present in a minority of these fetuses [11].

In recent studies, survival of gastroschisis approaches 95% [12,13]. Morbidity related to intestinal complications, however, can be relevant. It has, therefore, been proposed that gastroschisis should be classified as simple (if isolated) or as complex (if associated with intestinal atresia, perforation, stenosis, or volvulus) [14].

Prediction of outcome

Ideally, it would be useful to have reliable prenatal predictors of gastroschisis complications, to provide parents with a more accurate prognostic assessment, and identify candidates for investigational preventive treatments. One main outcome of interest is that gastroschisis is known to be associated with a risk of intrauterine death of about 5% [15]. This increased risk is supposed to be associated with compression of the umbilical vessels by herniated viscera, as well with the increased incidence of fetal growth restriction or small-for-gestational age weight in fetuses with this condition [16]. The prenatal detection of fetal growth restriction has actually been shown to predict a higher risk of adverse outcomes [17]. Diagnosis of fetal growth restriction, unless extreme, is not an easy task in gastroschisis, as abdominal circumference measurements are affected by the condition. It has been

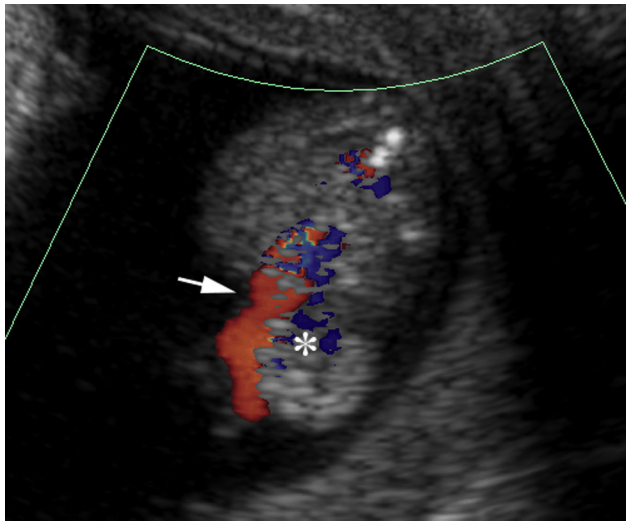

Fig. 2. Transverse view of the fetal abdomen at 12 weeks of gestation, showing gastroschisis: herniated bowel loops (*) can be seen lateral to umbilical cord insertion (arrow).

debated whether using fetal weight estimation formulas that do not include abdominal circumference is helpful [18,19]. A recent report also suggested that fetuses with gastroschisis associated with bladder herniation (a finding present in about 6% of cases during gestation) might present a higher risk of fetal distress and perinatal death [20].

Another main outcome of interest is that complex gastroschisis, which is present in slightly more than 10% of cases, involves a higher risk of postnatal complications [12,21]. Unfortunately, it is not even clear what causes the increased incidence of bowel complications in gastroschisis: a considerable amount of data suggest that components of the amniotic fluid, particularly in the third trimester, are able to cause inflammation of the exposed bowel, possibly leading to perivisceritis [22]; the bowel may become constricted at the level of the abdominal defect, suffering ischaemic injury; bowel atresia is associated with gastroschisis in 5–15% of cases [11–13,23]. A number of prenatal ultrasound findings have been reported in association with complex gastroschisis: intra- and extra-abdominal bowel dilatation; stomach dilatation or herniation; and thickening of the bowel wall. The presence of intra-abdominal bowel dilatation (variably defined as a luminal diameter more than 6, 10 or 14 mm) seems to be the only finding consistently associated with complex gastroschisis [13,23,24].

Prevention of adverse outcomes

The limited knowledge of the mechanisms causing adverse outcomes in gastroschisis does not help in their prevention. Experimental studies on animal models and preliminary data from humans studies suggest that regularly exchanging the amniotic fluid by aspiration and replacement with saline (amnioexchange) may improve the neonatal outcomes [22]. A randomised-controlled trial comparing saline amnioexchange (performed every two weeks from 30 weeks) to standard management, is currently near to completion [25].

Given the increased risk of intrauterine fetal death, it is also generally recommended that pregnant women with gastroschisis should be considered high risk, deserving some form of increased fetal monitoring. The modalities of such monitoring lack consensus: the most common options are cardiotocography, suggested even daily in the third trimester [26], and umbilical and middle cerebral artery Doppler [16]. Daily fetal heart rate home monitoring has also been successfully used, by training pregnant women to record fetal heart rate for a 30-min period and transmit the trace by fax or email to the supervising centre [27].

Timing and mode of delivery

Some evidence of limited quality on when and how to deliver pregnancies complicated by fetal gastroschisis has been published. Many centres advocate planned preterm birth at 36–38 weeks, but studies on this subject have reported variable and conflicting results. Moreover, most studies report a mean gestational age at spontaneous delivery around 36–37 weeks. A recent systematic review could include only one small randomised trial [28], and drew no firm conclusions about preterm birth for infants with gastroschisis [29]. Studies on caesarean section are not conclusive and do not show any consistent advantage over vaginal delivery; it must be remembered that trauma to the abdominal viscera can occur with either modality, and delivery must therefore be careful [30]. Most investigators recommend delivery in a tertiary centre with neonatal intensive care and paediatric surgery facilities. Although this is likely to reduce morbidity, no good evidence is available on the subject.

Postnatal management

The exposed bowel loops can cause significant water loss by evaporation. Immediately after delivery, intravenous fluid resuscitation should be started. The herniated loops should be wrapped in warm saline-soaked gauze, and covered with plastic wrap to reduce water and heat losses. The newborn should be positioned on the right side, with the packed bowel placed centrally on the abdomen to prevent kinking of the mesentery [31].

The ideal treatment of gastroschisis is immediate repositioning of the herniated bowel into the abdominal cavity, with closure of the abdominal wall (primary reduction and repair). If the patient is

unstable, however, or if reduction is likely to cause an abdominal compartment syndrome, staged repair is preferred: it consists of applying a plastic silo around the bowel, which allows to progressively push the bowel into the abdominal cavity over days, until definitive closure is possible. Prefabricated silos are available, with a circular retention spring that can be placed into the abdominal defect without sutures or general anesthesia [31]. Immediate closure may be associated with a higher risk of respiratory complications, whereas delayed closure usually involves a longer time to reach full enteral feeds. Most evidence, however, comes from retrospective studies, which may be biased by the policies of individual centres, as well as by association with complex gastroschisis: when bowel atresia is present, primary anastomosis is often impossible owing to bowel thickness and extent of the peel of the serosa. These children are therefore more likely to remain on parenteral nutrition for some weeks until repair is possible, with the inherent risk of increased infectious and cholestatic complications [31,32].

Omphalocele

In omphalocele (also called exomphalos), a midline defect with intra-abdominal contents is present, which herniate within a peritoneal sac into the amniotic cavity through the base of the umbilical cord. The membrane covering the sac consists of peritoneum on the inner side, amnion on the outer side, and Wharton's jelly between the layers. The size of the wall defect is variable, and the umbilical vessels insert onto the sac's membrane rather than on the intact abdominal wall. The sac usually contains bowel loops, but also the liver and other organs may herniate (Fig. 3).

Spontaneous rupture of the omphalocele sac *in utero* has been sparsely reported, with bowel loops floating freely in the amniotic fluid and mimicking gastroschisis. This condition is rare, and can be differentiated from gastroschisis because, in the latter, the umbilical cord always inserts on a normal segment of abdominal wall. Another condition that can be confused with omphalocele is umbilical cord hernia. Cord hernia is characterised by a normal insertion of the cord into the umbilical ring with intact skin covering, whereas, in omphalocele, there is a large defect of the umbilical ring area without muscles and skin [33]. In one of the largest studies of prenatally diagnosed omphalocele, the prevalence of omphalocele at the time of the nuchal scan (11–14 weeks) was 1:381 (0.08%) [34]. The prevalence was, however, related to crown-rump length (CRL) and sac content: when only bowel loops were herniated, the prevalence ranged from 1 out of 98 for a CRL of 45–55 mm to 1 out of 2073 for a CRL of 65.0–84.0 mm. For cases of omphalocele where also the liver was herniated, the prevalence was 1 out of 3360. The same study observed that, in 53 euploid fetuses with omphalocele containing only bowel at the nuchal scan, 49 (92.5%) had spontaneous resolution of the omphalocele by 20 weeks, and four fetuses had persistence until delivery. These findings have been interpreted as a delay in the

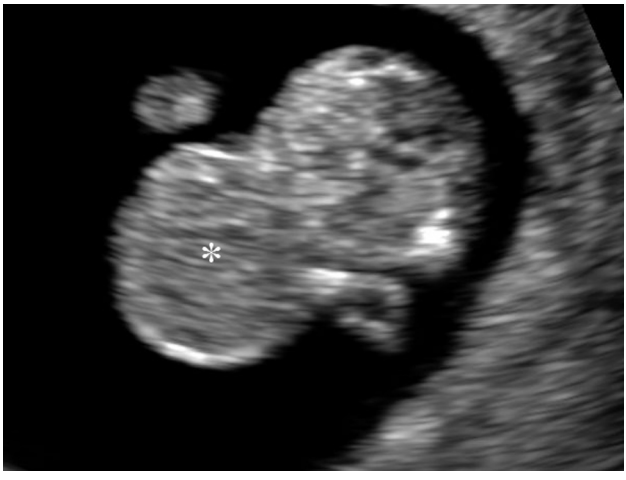

Fig. 3. Transverse view of the fetal abdomen at 12 weeks of gestation, showing a large omphalocele (*).

recovery of physiological herniation of the bowel, which is usually complete by 11 weeks; this delay is more frequent in aneuploid than euploid fetuses [34]. Spontaneous resolution of first-trimester omphalocele was reported, but with lower frequency also in other studies; spontaneous resolution has not been observed in cases with herniated liver [35,36].

Associated abnormalities

The finding of an omphalocele on prenatal ultrasound should prompt a careful search for associated anomalies and discussion of prenatal invasive testing. As introduced above, the presence of omphalocele indicates an increased risk of aneuploidy: in a recent first trimester study, fetuses with omphalocele were found to carry a chromosomal abnormality in 54–57% of cases [34–36]. The most common aneuploidy is trisomy 18, which is present in 80% of fetuses with omphalocele and associated malformations, and in 54% of fetuses with isolated omphalocele but increased nuchal translucency [36]. The risk of aneuploidy does not change if the omphalocele contains only bowel or also the liver [34,35], but correlates with nuchal translucency thickness. A normal nuchal translucency is therefore a reassuring sign, but the residual risk of aneuploidy may still be as high as 28% [35]. The role of comparative genomic hybridization (CGH)-based microarrays is still uncertain, given the small number of cases of omphalocele included in prenatal series [37].

Given the increased risk of intrauterine death with many chromosomal abnormalities, the association with aneuploidy will be less strong when the diagnosis of omphalocele is made in the second or third trimester of pregnancy, or at birth. In the EUROCAT registry, which reflects screening policies who differ among countries and who have changed in the past 30 years, the overall prevalence of aneuploidy is 23% [2].

Omphalocele can be associated with a number of abnormalities. Therefore, even if apparently isolated at the nuchal scan, further ultrasound follow up in the second trimester is recommended. The list of the possible associations is long. Pentalogy of Cantrell, which is usually diagnosed already in the first trimester, is a complex anomaly involving omphalocele and a spectrum comprising an anterior diaphragmatic hernia, sternal clefting, ectopia cordis, and an intracardiac defect [38]. Not all five elements are always present, but survival is on average less than 40%, falling to 5–10% in the case of severe ectopia cordis [39]. The cause of these midline supraumbilical defects has yet to be identified: although sporadic in most of the described infants, X-linked recessive inheritance was suggested for some families and genes located on the X-chromosome (Xq25-q26.1) may be involved in some of the reported cases [40].

Postnatally, Beckwith–Wiedemann syndrome is a growth disorder characterised by macroglossia, macrosomia, omphalocele, hypoglycaemia leading to seizures, visceromegaly, hemihyperplasia, renal abnormalities, ear creases and pits, nevus flammeus, and embryonic tumours (e.g. Wilm's tumour, hepatoblastoma, neuroblastoma, and rhabdomyosarcoma) [41]. Although omphalocele is not invariably present, when observed prenatally it should guide a targeted ultrasound examination searching for all possible prenatal features of the syndrome, which include additionally polyhydramnios, placentomegaly, and placental mesenchymal dysplasia (observed on ultrasound as multiple cystic changes in the placenta). In 2005, a diagnostic model was proposed, combining ultrasound, pathological, cytogenetic and molecular findings. The presence of two major criteria (abdominal wall defect, macroglossia, macrosomia) or one major plus two minor criteria (nephromegaly or dysgenesis, adrenal cytomegaly, aneuploidy or abnormal loci, and polyhydramnios) should enable diagnosis of Beckwith–Wiedemann syndrome [42]. This model, however, has neither been validated prospectively, nor takes into account newer molecular tools for diagnosis from fetal samples. Beckwith–Wiedemann syndrome involves dysregulation of imprinted growth regulatory genes on chromosome 11p15.5 (also known as the Beckwith–Wiedemann syndrome critical region) [43]. Regulation may be disrupted by any one of numerous mechanisms. Comprehensive laboratory evaluation includes karyotype analysis, DNA methylation tests, and genomic analysis of chromosome 11p15.5. About 85% of individuals have no family history of Beckwith–Wiedemann syndrome, whereas about 15% have a family history consistent with autosomal dominant transmission. Children of subfertile parents conceived by assisted reproductive technology may have an increased risk for imprinting disorders, including Beckwith–Wiedemann syndrome. Cytogenetically detectable abnormalities involving chromosome 11p15 are found in 1% or fewer of affected individuals. Molecular genetic testing can identify epigenetic and genomic

alterations of chromosome 11p15 in individuals with Beckwith–Wiedemann syndrome: (1) loss of methylation on the maternal chromosome at imprinting centre 2 (IC2) in 50% of affected individuals; (2) paternal uniparental disomy for chromosome 11p15 in 20%; and (3) gain of methylation on the maternal chromosome at imprinting center 1 (IC1) in 5%. Methylation alterations that are associated with microdeletions or microduplications in this region are associated with high heritability. Experience with prenatal diagnosis of methylation abnormalities involved in Beckwith–Wiedemann syndrome, however, is quite limited [44]. Sequence analysis of *CDKN1C* identifies mutations in about 40% of familial cases and 5–10% of cases with no family history of the syndrome.

In the omphalocele, bladder exstrophy, imperforate anus, spina bifida complex (OEIS) complex, the prenatal findings are omphalocele, a skin-covered lumbosacral neural tube defect, impossibility to visualise the bladder, and limb defects. Anal atresia, bladder exstrophy, and abnormal genitalia are more difficult to visualise. The OEIS complex is usually identified after 16 weeks, even if reports of earlier diagnoses exist [45,46]. The underlying cause remains unknown, although genetic and environmental factors are likely to play a role.

Omphalocele has been found to be associated with a number of other conditions: neural tube defects, defects of the diaphragm, fetal valproate syndrome, and a number of single gene disorders and syndromes. A comprehensive review of such associations is given in the article by Chen [47]. The presence of associated anomalies on ultrasound, or a significant family history, may raise the suspicion for one of these conditions.

Prediction of outcome

Given the impressive list of conditions associated with omphalocele, only some of which are amenable to prenatal diagnosis, it is clear that the prognosis is driven by the presence and nature of the associated features. How accurate then is a prenatal diagnosis of isolated omphalocele? This is affected by the gestational age at examination, as well by the availability and use of more or less sophisticated molecular diagnosis tools. In a recent study from the Netherlands, which is a good example of contemporary practice, 12 out of 31 apparently isolated cases at prenatal diagnosis (39%; 95% confidence interval 22–58%) were found to have associated anomalies after delivery [48]. In a similar study from the USA, five out of 19 fetuses with omphalocele prenatally isolated or associated with minor anomalies (e.g. renal pelvis dilation, single umbilical artery, and umbilical cord cyst) were found to have additional abnormalities after birth (26%; 95% confidence interval 9–51%) [49]. In both studies, karyotype was offered in all cases, and fetuses with aneuploidies were excluded. It is of interest that, in the study by Porter et al. [49], preterm birth complicated more than one-third of cases, and the only neonatal deaths occurred because of complications of prematurity. In another Dutch study, 12 (14%) out of 88 fetuses with a prenatal diagnosis of omphalocele were alive and well at paediatric follow up, including two with multiple associated anomalies that could be corrected [50]. In the largest prenatal study available, including cases from a single institution in the UK observed from 1991 to 2002, only 55 out of 445 fetuses with persistent omphalocele were liveborns, and 44 made it to surgery [51].

A peculiar group of fetuses with omphalocele is that of giant omphaloceles (i.e. those omphaloceles with a large defect, significant herniation of the liver, or both). Wide variation among definitions of giant omphalocele exist: one of the most authoritative (and restrictive) ones defines it as a defect containing most (>75%) of the liver [52]. Among the other definitions proposed, some are based on absolute size, and some on the relative size to the fetus [50]. Intuitively, the larger the defect, the higher the risk of postnatal complications, such as pulmonary hypoplasia leading to respiratory insufficiency and prolonged ventilatory support, often with delayed closure of the abdominal wall defect. Among these infants, an increased prevalence of deficits in developmental achievements has to be expected [52].

Timing and mode of delivery

No randomised-controlled trials have been conducted on mode of delivery in fetuses with omphalocele, and the decision for a caesarean section should be dictated by obstetric indications. Many clinicians decide to deliver fetuses with large defects by caesarean section, as they fear sac rupture or liver damage during delivery [31]. As the definition of a ‘large’ omphalocele is not standardised,

retrospective studies are difficult to compare. In the recent study by Kleinrouweler et al. [50] 17 out of 21 (81%) deliveries were vaginal, with four caesarean sections based exclusively on obstetric indications and not on features such as size or liver herniation (present in 47% of vaginal deliveries). No complications such as sac rupture or liver haemorrhage were observed. Preterm delivery in omphalocele is not indicated.

Postnatal management

The immediate postnatal management of newborn babies with omphalocele should aim to stabilize vital functions and search for associated anomalies that might affect treatment or monitoring (e.g. congenital heart disease, pulmonary hypoplasia). Although fluid and heath losses are less than with gastroschisis, the omphalocele should be covered with saline-soaked gauze to minimise them. Neonatal hypoglycaemia should alert for Beckwith–Wiedemann syndrome. Treatment is driven by gestational age, size of the defect, and associated anomalies if present [31]. If the abdominal wall defect is small or medium-sized (up to about 4 cm in diameter), treatment usually is by primary closure and does not usually involve technical problems. Larger defects, on the contrary, can pose major challenges: most cases are not considered amenable to primary repair owing to the lack of abdominal domain. Non-operative techniques use agents (e.g. povidone–iodine, sulfadiazine, neomycin, silver-impregnated dressings, neomycin, polymyxin, and bacitracin ointments), helping the amnion sac to form an eschar, which then epithelialises in 4–10 weeks. These methods are used when the defect is too large for primary repair, and also when cardiac or respiratory comorbidities preclude surgery. Most women who have given birth to babies with omphalocele will require a later repair of the ventral hernia defect, which can be carried out during infancy. Finally, neonatal staged closure implies the closure of the abdominal wall with multiple procedures. This can by achieved by either using the amnion sac with serial reductions, or by replacing the sac with a mesh and closing it gradually over time [32].

Other abdominal wall abnormalities

We have already mentioned some rare and complex abnormalities involving the abdominal wall, such as pentalogy of Cantrell and the OEIS complex.

Body stalk anomaly is characterised by herniation of the abdominal contents into the extraembryonic coelom through a large wall defect, and by a short or absent umbilical cord. The ultrasound features change with gestation: in early pregnancy, the upper part of the embryo is clearly situated in the amniotic cavity, whereas the lower part is located in the extraembryonic coelom, with abdominal contents protruding into it (Fig. 4); later in pregnancy, the umbilical cord becomes short or absent [53,54]; kyphoscoliosis, cranial defects, and limb defects can be also be observed. In case of limb defects, the term limb–body wall defect or complex has been used: however, body-stalk anomaly and limb–body wall complex seem to be part of the same spectrum of primary ectodermal failure in the early embryonic disc [55].

In prune-belly syndrome, a combination of deficient abdominal wall muscles, urinary tract abnormalities, and cryptorchidism in males exists. Early in gestation, this condition should be suspected in case of a large megacystis. It is still unclear whether the hypoplasia of abdominal wall muscles is secondary to the stretching caused by megacystis, or if an unknown underlying cause may explain both [56].

In bladder exstrophy, mesenchymal cells fail to migrate between the abdominal ectoderm and the cloaca. Consequently, no muscular or connective tissue covers the anterior abdominal wall. Five ultrasound findings are described for prenatal diagnosis: (1) non-visualisation of the bladder; (2) a lower abdominal bulge formed by the exstrophied bladder; (3) small penis with anteriorly displaced scrotum; (3) low umbilical insertion; and (5) widening of the iliac crests [57]. Not all signs, however, are always present, and an urachal cyst may mimic the presence of the bladder; consequently, the accuracy and sensitivity of ultrasound are relatively low, particularly if no associated findings (such as in the OEIS complex) are present [58]. Cloacal exstrophy is associated with rupture of the cloacal membrane before fusion with the urorectal septum. As a consequence, these fetuses present with two exstrophied

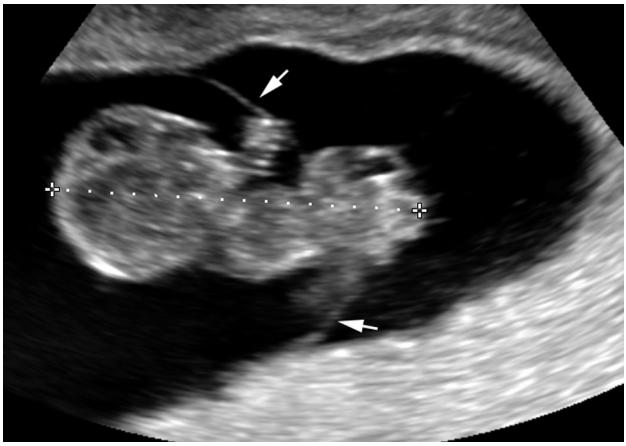

Fig. 4. Body stalk anomaly at 9 weeks. Arrows delineate the amniotic membrane. The upper part of the fetal body is on the left of the amniotic membrane, within the amniotic cavity. The dysmorphic lower part of the body is on the right, in the extraembryonic coelom.

Table 1
Ultrasound findings in fetal abdominal wall defects.

Abnormality	Covering membrane	Site of defect	Umbilical cord insertion	Additional findings
Omphalocele	Yes	Umbilical insertion	Omphalocele membrane	
Gastroschisis	No	Right of umbilical insertion	Normal insertion	
Umbilical hernia	Yes	No umbilical ring defect	Normal insertion	
Pentalogy of Cantrell	Yes	Above umbilical insertion	Omphalocele membrane	Anterior diaphragmatic hernia, sternal clefting, ectopia cordis, and intracardiac defect.
OEIS complex	Yes	Umbilical insertion	Omphalocele membrane	Bladder exstrophy, imperforate anus, and spina bifida.
Body-stalk anomaly	Herniated organs in extraembryonic coelom	Whole anterior abdominal wall	Cord absent or shortened	Kyphoscoliosis, cranial defects, and limb defects.
Bladder exstrophy	Not applicable	Below umbilical insertion	Low insertion	Non-visualisation of bladder, lower abdominal bulge (exstrophied bladder), small penis with anteriorly displaced scrotum (if male), and widening of the iliac crests.
Cloacal exstrophy	Not applicable	Below umbilical insertion	Low insertion	Renal anomalies, neural tube defect, omphalocele, vertebral anomalies, non-visualisation of the bladder, distended bladder, hydrocolpos, dilated or echogenic bowel, umbilical cord cyst, separated pubic bones, and 'elephant trunk' sign.

OEIS, omphalocele, bladder exstrophy, imperforate anus, spina bifida complex.

hemibladders separated by a foreshortened hindgut or cecum, sometimes with a constellation of associated findings. In a recent postnatal series of 11 cases, the following prenatal ultrasound findings were variably present: hydronephrosis, neural tube defects, omphalocele, vertebral anomalies (particularly involving the sacrum), non-visualisation of the bladder, distended bladder, hydrocolpos, dilated or echogenic bowel, umbilical cord cyst, separated pubic bones, and the 'elephant trunk' sign (protrusion of ileum through the defect, resembling the trunk of an elephant floating in amniotic fluid) [59]. The postnatal correction and prognosis of bladder and cloacal exstrophy depends on a number of anatomical details, not all accurately investigated prenatally, and counselling and delivery in a centre with experience in their treatment are of paramount importance [58,59].

Conclusion

The most common abdominal wall defects are gastroschisis and omphalocele, both with a prevalence of about 3 in 10,000 births. The prevalence of gastroschisis has increased in the past 30 years, whereas that of omphalocele has remained basically stable. Prenatal ultrasound has a high sensitivity for these abnormalities already at the time of the first trimester nuchal scan. A summary of ultrasound findings in fetal abdominal wall defects is given in Table 1.

Major unrelated defects are associated with gastroschisis in about 10% of cases, whereas omphalocele is associated with chromosomal or genetic abnormalities in a much higher proportion of cases. Challenges in the management of gastroschisis are related to the prevention of late intrauterine death, and the prediction and treatment of complex forms (those associated with intestinal atresia, perforation, stenosis, or volvulus). With omphalocele, the main difficulty is counselling the woman about the possible associated conditions, not all easily diagnosed prenatally. Other rarer forms of abdominal wall defects (e.g. pentalogy of Cantrell, OEIS complex, prune-belly syndrome, body stalk anomaly, and exstrophies) deserve multidisciplinary counselling and management.

Conflict of interest

None declared.

Practice points

- Prenatal ultrasound has a high sensitivity for omphalocele and gastroschisis.
- In most western countries, the prevalence of gastroschisis has been increasing over the past few decades.
- Gastroschisis is associated with a risk of intrauterine death of 5%.
- Gastroschisis has a high survival, but bowel-related morbidity is high in cases of complex gastroschisis (associated with intestinal atresia, perforation, stenosis, or volvulus).
- Omphalocele is often associated with chromosomal or genetic abnormalities.
- Large (giant) omphaloceles can present with pulmonary hypoplasia.
- In the absence of randomised-controlled trials, mode of delivery of abdominal wall defects should be dictated by obstetric indications.

Research agenda

- Trials on fetal well-being assessment in gastroschisis.
- Role of comparative genomic hybridisation array and other molecular techniques to exclude associated syndromes in omphalocele.
- Randomised trials evaluating mode of delivery and postnatal surgical management in omphalocele and gastroschisis.

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