Abdominal and abdominal-wall abnormalities

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INTRODUCTION

Abdominal-wall abnormalities are one of the commoner fetal abnormalities demonstrated by ultrasound. They were recognized early in the development of fetal ultrasound because disturbance of the abdominal-wall contour in the region of the cord insertion was demonstrated even by static B-mode scanning before real time was available. Since many of these abnormalities cause a rise in the serum alpha-fetoprotein, a specific search for abdominal-wall abnormalities as a cause for this has been undertaken in virtually all ultrasound departments using routine serum alpha-fetoprotein screening. Even though the primary aim of this screening was to exclude neural-tube defects, anterior abdominal-wall defects as a group are the second commonest anatomical cause for raised serum alpha-fetoprotein. The spectrum of anterior abdominal-wall defects extends from the very minor exomphalos with bowel herniating into the base of the cord to the most major defects, including the pentalogy of Cantrell and body-stalk defect or early amnion rupture sequence.
NORMAL APPEARANCES

The anterior abdominal wall is best demonstrated in axial section. The anterior abdominal wall is clearly outlined by amniotic fluid on its external surface with the site of the cord insertion being clearly demonstrated. The lowest part of the anterior abdominal wall is sometimes obscured by the flexed legs with assessment of the anterior abdominal wall below the cord insertion sometimes being extremely difficult, whatever approach is utilized. A midline, sagittal view of the fetus will sometimes give better views of this area but this is not always feasible. The internal aspect of the anterior abdominal wall can be difficult to see clearly because of the similar echodensities of the anterior abdominal wall and adjacent liver and bowel. This becomes more clearly identified as the pregnancy advances.

NORMAL VARIANTS, PITFALLS AND ARTEFACTS

By 12 weeks' gestation, the bowel has returned to the abdomen so that any herniated abdominal contents are abnormal after this time. The exclusion of abnormality of the anterior abdominal wall is difficult in certain circumstances, particularly where the anterior abdominal wall of the fetus lies adjacent to the maternal uterus or placenta. This is not a major difficulty with adequate amounts of liquor in the first and second trimesters since the fetus will usually turn given time so that the anterior abdominal wall can be checked. With severe oligohydramnios and anhydramnios and, in particular, with the reduced mobility of the fetus in the third trimester, exclusion of an anterior abdominal-wall defect can be very difficult. With extreme oligohydramnios, the distortion of the anterior abdominal wall can often mimic a moderate-sized, anterior abdominal wall defect. Defects in the infra-umbilical portion of the abdominal wall are difficult to exclude with flexion of the fetal femora. Since many of these defects are associated with bladder abnormalities, presence of a normal bladder, even in the absence of clear demonstration of the anterior abdominal wall, excludes cloacal extrophy and thus suggests an intact lower abdominal wall, but will not exclude exomphalos or gastroschisis.

ANTERIOR ABDOMINAL-WALL APPEARANCES

List of conditions

- Exomphalos
- Gastroschisis
- Limb–body-wall complex/early amnion rupture sequence
- Ectopia cordis (including pentalogy of Cantrell)
- Cloacal and bladder extrophy.

Exomphalos

Exomphalos occurs in about 0.1 to 0.3% of pregnancies,1,2 the incidence increasing with maternal age.

Exomphalos is an incomplete return of the abdominal contents to the abdominal cavity in early pregnancy. In its minor form this can consist of herniation of a small loop of bowel into the base of the cord and can be extremely difficult to diagnose. Any transonic area at the base of the cord that is not a blood vessel is almost certainly a small loop of bowel in a minor exomphalos. The umbilical arteries are seen coursing around the sac and its contents in contradistinction to gastroschisis. The most major exomphalos may, indeed, be larger in cross-sectional area than the abdomen. The sac of an exomphalos, which consists of peritoneum and amnion, may contain any of the abdominal organs, although bowel is most commonly present, but the stomach, liver and spleen may be partially or entirely present within the exomphalos. Exomphalos containing only bowel are in the minority (10–25%).3,4,5 The relatively thick sac wall restricts the 'leakage' of alphafetoprotein and levels are not as raised as in gastroschisis.6 There is frequently some free fluid within the larger exomphalos, which obviously communicates freely with the peritoneal cavity (Fig. 11.1). As the bowel is contained within a sac, it does not become exposed to amniotic fluid and thus does not become thickened. The relatively wide neck to the sac (Fig. 11.2) seen in exomphalos also contributes to the reduced incidence of bowel atresias compared with gastroschisis.

Other abnormalities are frequently associated with exomphalos. Cardiac malformations are seen in up to 50%, limb abnormalities in about 30% and...
chromosome abnormalities in 28–36%, mainly trisomies 13 and 18.\textsuperscript{7} The association of exomphalos with bladder exstrophy imperforate anus and spinal defects is well recognized (OEIS Complex) and is one of the rarer causes of nuchal thickening.\textsuperscript{8}

Some authors quote a chromosome abnormality rate of 61% at 11–14 weeks' gestation.\textsuperscript{2} Chromosome abnormalities are more frequently associated with small exomphalos containing only bowel (67% compared with 16% if liver is also present).\textsuperscript{1} It is important to differentiate a large exomphalos from limb–body–wall complex (see below). Exomphalos is a feature of the Beckwith–Wiedemann syndrome and is seen in 10% of cases. Other features include macroglossia, gigantism and cystic kidneys. Any fetus with exomphalos should be thoroughly examined for other abnormalities, particularly cardiac malformations. With larger exomphalos, the heart is often difficult to examine comprehensively. This is due to the tendency of the heart to rotate, as much of the abdominal contents lie outside the normal abdomen. The fetus also tends to lie on its side, which makes an anterior approach to the heart quite difficult. The exomphalos itself may also impede the view of the heart to some extent. Umbilical cord cysts are sometimes associated with exomphalos and in such cases there is a very high risk (44%) of trisomy 18. Diagnosis as early as 12 weeks has been described with 3D ultrasound.\textsuperscript{10}

Ultrasound diagnosed exomphalos in 66–75% of cases\textsuperscript{7,11} in two large British series from the late 1980s to early 1990s, but accuracy has increased in later years to as high as 86% in one American series.\textsuperscript{12} Misdiagnosis of exomphalos as gastroschisis occurred in 5%.\textsuperscript{7} In view of the high risk of chromosome abnormalities all patients carrying a fetus with exomphalos should be offered karyotyping.

Polyhydramnios has been demonstrated in 30% of fetuses with exomphalos,\textsuperscript{13} but the cause, whilst sometimes related to other abnormalities, is not always clear.

The fetus with exomphalos should be examined at intervals throughout the pregnancy, although complications such as bowel atresia are quite uncommon. Rupture of the sac in utero is rare.\textsuperscript{5,14,15} There is no evidence of improved outcome if the fetus is delivered by caesarean section\textsuperscript{6,17} and this should be reserved for those pregnancies with obstetric indications for such delivery.

In the absence of other abnormalities, the outcome for a fetus with exomphalos is good, with surgical repair being feasible in most cases, although sometimes complex, staged, abdominal-wall repairs are required. The survival in exomphalos is strongly related to the incidence of associated anomalies and this has to a large extent prevented an improvement in survival in recent years. In the presence of a major associated anomaly, the survival can be as low as 20%. Interestingly, whilst large size is associated with a poorer prognosis, this is not a major factor.

Although in most cases the relative size of the exomphalos compared to the abdominal cross-section remains unchanged, relative increase in size has been observed in some cases. Relative decrease
in size of the exomphalos has also been demonstrated in other cases (Fig. 11.3).

As there is usually an intact sac covering exomphalos at birth, immediate surgery within hours is not always necessary and, indeed, with small exomphalos, delayed repair is often feasible.

**Gastroschisis**

This condition, which has no known genetic associations, is a herniation of abdominal contents, usually to the right of the cord insertion (Fig. 11.4). There is some dispute as to the underlying cause, some authorities suggesting that this is due to rupture of an exomphalos, and others suggesting that it is a developmental defect of the abdominal wall due to abnormal involution of the right umbilical vein.\(^1\) A gastroschisis never has a surrounding membrane and usually contains only bowel, predominantly small bowel (Fig. 11.5). As the bowel is in direct contact with the amniotic fluid, both serum and amniotic fluid alphafetoprotein levels are elevated more than in exomphalos.\(^6\) The stomach and large bowel do occasionally herniate from the abdomen, but this is less common. It is unusual for liver, spleen or bladder to herniate. In early pregnancy, the multiple loops of bowel can be seen outlined by the amniotic fluid, with bowel-wall thickness and lumen dimensions being normal. In later pregnancy, complications such as bowel-wall thickening, shortening and bowel dilatation may supervene. The thickening is thought to be a chemical peritonitis related to the exposure of the bowel to fetal urine in the amniotic fluid.\(^2\) By definition the bowel of gastroschisis is non-rotated. Intestinal atresias or stenosis secondary to intestinal ischaemia are reported in up to 30\%.\(^2\) Bowel lumen diameter of greater than 17 mm is very suspicious of significant bowel dilatation.\(^2\) Babcock et al\(^2\) showed that a maximum small bowel diameter of more than 11 mm was related to postnatal bowel complications, but operator variation in measurement makes utilization of this figure in practice difficult. Abnormal ultrasound appearance

![Fig. 11.3](image1) (A) Axial section of fetus, showing fetal trunk (FT) with exomphalos (E). Note that the exomphalos is almost the same size as the fetal trunk. (B) Same fetus 4 weeks later, showing relative decrease in the size of the exomphalos (E). At birth, the exomphalos was very small and repair was delayed until 3 months of age. Note the umbilical cord (UC) entering the apex of the sac, a diagnostic feature of exomphalos. FT, fetal trunk.

![Fig. 11.4](image2) Gastroschisis containing free loops of bowel. Umbilical cord (UC) seen to the right of the loops of bowel, differentiating this from an exomphalos.
of bowel is associated with more difficult repair and higher incidence of overall complications.\textsuperscript{24} Gastroschisis is not usually associated with other abnormalities, although every fetus with gastroschisis should be examined by ultrasound to exclude other abnormalities. Karyotypic abnormalities are extremely uncommon and most centres would not recommend karyotyping for gastroschisis. In the absence of additional abnormalities, the antenatal outlook for fetuses with gastroschisis is good. The antenatal detection rate of gastroschisis in two series has been quoted as 71.6\%\textsuperscript{25} and 70\%.\textsuperscript{11}

About 48\% of fetuses with gastroschisis will be small for dates.\textsuperscript{26} Difficulties in management occur because ultrasound abdominal circumference measurements are not usually valid in this group and as this is the cornerstone of assessment of fetal size in utero, accurate assessment becomes difficult. Cardiotocograph monitoring is also affected by the bowel in the amniotic cavity, possibly mediated through tension on the vagus nerve. Whilst caesarean section is not usually considered advantageous over vaginal delivery in this condition,\textsuperscript{24} many fetuses with gastroschisis are delivered by caesarean section for obstetric reasons related to the difficulty in monitoring both fetal size and fetal wellbeing.

Misdiagnosis of gastroschisis as exomphalos occurred in 14.7\% of cases in one series from the late 1980s to early 1990s.\textsuperscript{25} Confusion with limb–body–wall complex can be avoided by identi-

fying the other abnormalities usually seen in the latter condition (see below).

Outcome is not affected by delivery in a tertiary obstetric centre as compared with a district general hospital,\textsuperscript{127} although delivery within easy reach of a neonatal surgery centre is advisable. There is no evidence that labour or ruptured amniotic membranes affects neonatal outcome.\textsuperscript{28} The only ultrasound features that predict a poorer outcome are polyhydramnios\textsuperscript{29} and a dilated fetal stomach.\textsuperscript{30} CTG monitoring may improve outcome. Also\textsuperscript{29} postnatal management requires immediate enclosure of the bowel in ‘clingfilm’ to retain moisture followed by early transfer to the neonatal surgical unit for early closure. This can almost always be achieved in one operation, but sometimes requires complex staging procedures. Because of bowel–wall thickening and the incidence of atresias, lengthy intravenous feeding is often required and an overall, neonatal mortality rate of 10\% is a combination of the early neonatal deaths caused by bowel ischaemia and later problems relating to short bowel and the complications of intravenous feeding. More recent series have suggested survival in isolated gastroschisis as high as 92\%.\textsuperscript{31} The majority of survivors will be on full feeding within 4 weeks of delivery, but some may require many months of intravenous feeding. Bowel dilatation in utero of 18 mm or more is associated with significant delay in establishing oral feeding.\textsuperscript{32} It is extremely difficult to predict when bowel is at risk in utero. Colour flow Doppler, whilst helpful, has not answered all the questions and the balance between preterm delivery with all its problems and the risks of bowel ischaemia, the symptoms and signs of which in utero are very non-specific, is a difficult one to achieve.

Spontaneous resolution of gastroschisis and closure of the anterior abdominal-wall defect has been described.\textsuperscript{33} This, however, carries a poor prognosis as it is due to ischaemia and subsequent absorption of the extruded bowel loops, resulting in virtual complete absence of the small bowel.

**Limb–body–wall complex**

This is a very rare condition also known as body stalk anomaly. Some authors consider this to be a variant of early amnion rupture. Some consider it to be an extreme form of the amniotic band syndrome. The cord is shortened often, with only a
single artery and a very large, anterior abdominal-wall defect is seen, usually affecting the left side. There is often an associated, extensive, lower-spine neural-tube defect.\textsuperscript{34} Spinal dysraphism is common and association of anterior abdominal-wall defect with scoliosis and spinal defect should suggest the diagnosis. Other primary features are exencephaly or encephalocoele, facial cleft and limb defects, thoracic and abdominal abnormalities entangled with the membranes. All such pregnancies have failure of fusion between the amnion and the chorion. The outcome is uniformly fatal.

Pathogenesis is uncertain, and theories of causation include body-stalk dysmorphogenesis and early amnion rupture owing to a vascular disruption. The umbilical cord is short. Karyotypic abnormalities have not been described. The condition has been linked with cocaine abuse\textsuperscript{35} and diagnosis as early as 9 fetal weeks has been reported.\textsuperscript{36}

\section*{Ectopia cordis}

This is a rare, body-wall abnormality where the heart has herniated through a defect in the chest or thoraco-abdominal wall. When there is an abdominal element, this is often known as the pentalogy of Cantrell (ectopia cordis, diaphragmatic defect, exomphalos, pericardial defect and intracardiac abnormality).\textsuperscript{37,38} The defect is thought to be due to lack of fusion of the lateral body folds. Ectopia cordis is easy to demonstrate by ultrasound as the pulsating heart is clearly identified outside the confines of the chest. The diagnosis can be difficult if amniotic fluid volume is reduced.

The rotation and occasional displacement of the heart seen with a large exomphalos should not be confused with pentalogy of Cantrell.

\section*{Cloacal and bladder extrophy}

In cloacal extrophy as well as an exomphalos, there is a cloacal abnormality, which opens on to the anterior abdominal wall. This can sometimes be a very difficult condition to diagnose, but should be suspected in any abdominal-wall defect where a normal urinary bladder is not demonstrated within the fetal abdomen (Fig.11.6). Associated hydronephrosis is a common finding as are lower spinal anomalies.\textsuperscript{39} Less commonly, gastrointestinal, central nervous system and cardiac anomalies are seen. The thorax is usually narrow and there is a large sacral meningomyelocele.\textsuperscript{35,40} Bilateral club-feet are a common association.\textsuperscript{41} This is a very serious condition requiring multiple surgical procedures post-natally with complicated urinary bowel and genital implications. Long-term outcome is not good with a 55\% mortality.\textsuperscript{42}

Bladder extrophy is a less severe condition, which, because the lower abdominal wall is often partially obscured by the femora, is more difficult to demonstrate. The urachus has been mistaken for the fetal bladder in one case of extrophy,\textsuperscript{43} although isolated case reports have been made.\textsuperscript{44} The diagnostic sign of this condition is absence of an intra-abdominal bladder with a low anterior abdominal-wall mass, the umbilical arteries running on either side of the mass.\textsuperscript{45} The outcome following surgery for bladder extrophy is very good.
vena cava is closely related to the posterior aspect of the liver. The spleen is of the same density as the liver, but is often difficult to differentiate from it. The normal fetal stomach can usually be recognized from early second trimester onwards, although not always on a single examination as its size varies with time. A range of normal values for gastric area that correlates with gastric volume has been produced but is not used routinely. Repeat scanning usually enables it to be identified over a period of hours or on separate days. The kidneys are easily recognized by their position and slightly brighter cortex compared to the renal medulla and adjacent liver. The bowel has a slightly increased echodensity compared to the liver and a ‘coarser’ texture.

Some small fluid-filled loops of small bowel can often be identified in the second and third trimester. The large bowel in the third trimester often has a relatively transonic appearance and is recognized by its haustral appearance and its position in the abdomen. This should not be mistaken for dilated loops of small bowel that would be pathological. The normal calibre of the fetal large bowel is up to 5 mm at 20 weeks and up to 20 mm at term. A full range of large bowel diameters has been produced. The normal pelvic colon can be mistaken for a pelvic mass or ovarian pathology. The fetal bladder is of variable size and transonic in appearance. Its position in the pelvis confirms its nature. Fetal ureters are not identified in utero when normal. The fetal adrenal glands can be clearly identified in many cases in the second and third trimesters. Their position above the kidneys and differentiation between the more echodense medulla and less echodense cortex is typical.

**INTRA-ABDOMINAL ABNORMALITIES**

**Liver**

Echo bright areas in the liver, either solitary or multiple, are recognized. In many of these, the underlying cause is not determined. If they are uncalcified, they may well represent small haemangiomas. Calcified lesions suggest the possibility of previous viral infection, particularly cytomegalovirus, toxoplasmosis or varicella (Fig. 11.7). In the absence of infection, isolated calcified foci in the liver have a good outcome.

Liver tumours, particularly large haemangiomas, haemangioendotheliomas and hepatoblastomas, have been demonstrated in utero. These manifest themselves as liver enlargement with mixed echogenicity. Large haemangiomas may be the cause of extensive arteriovenous shunting and developing hydrops, but are very rare. Whilst these are benign lesions, the outlook is poor if hydrops is present. In utero treatment with maternal cortico steroids has been reported to reduce the size of the lesion. Liver cysts, often lymphangiomas, have been described antenatally and if unilocular, they must be differentiated from choledochal cysts and a normal gall bladder. Some are multilocular and they must be differentiated from a duplication cyst or mesenteric cyst. One further differential diagnosis of a cystic mass within the liver is a varix of the intra-abdominal portion of the umbilical vein. This can usually be diagnosed as it communicates with the umbilical or portal vein and shows venous flow on Doppler or colour flow (Fig 11.8).

**Gall bladder**

The fetal gall bladder is very variable in size and whilst absence of the gall bladder can be associated with polysplenia syndromes and biliary atresia, such diagnoses are rarely made antenatally. A subjectively enlarged gall bladder has been reported in association with chromosomal disease. Calcific foci in the fetal gall bladder are well recognized. These are due to small gallstones and are usually incidental.
findings (Fig. 11.9). Most of these have no postnatal implications. Non-visualization of the gall bladder is unusual if persistently sought. In fetuses with non-visualization the fetal anomaly risk in one series has been shown to be 41%.\(^5\) Gall bladder duplication is rare but should be differentiated from choledochal cyst as it is a benign condition.\(^6\)

**Choledochal cysts**

Transonic areas in the portahepatis or subhepatic area separate from the gall bladder are rare. Such an abnormality should be considered to be a choledochal cyst when a separate gall bladder can be identified and the duodenum is seen separately from this mass (Fig. 11.10).\(^6\) Renal-tract abnormalities and adrenal cysts rarely cause confusion. A duplication cyst of the duodenum can give similar appearances as can a simple, hepatic cyst. Some choledochal cysts, however, will be seen communicating with the branching biliary structures and this further confirms the diagnosis. All cases so far diagnosed antenatally have been female\(^5\) and have been diagnosed between 15 and 37 weeks. In 56% of cases, the cyst was seen to grow in utero.

**Spleen**

In utero splenic abnormalities are very unusual, but transonic splenic cysts have been described as isolated findings, and normally have a good outcome (Fig. 11.11). Complete resolution postnatally within 6 months has been described.\(^6\) Splenomegaly caused by cytomegalovirus infection has been described antenatally.\(^6\)
Intra-abdominal cysts

Cystic abdominal lesions in the fetus are quite common. When such an abnormality is suspected, confirmation of normality of organ systems is essential. This includes normality of the gall bladder, kidneys, bladder, stomach, duodenum and large bowel if possible. Determination of gender also helps. In females, the commonest cause of a transonic abdominal mass with thin walls is an ovarian cyst (Fig. 11.12). These are usually seen in the third trimester. They can occur in any site within the abdomen and be up to 10 cm in diameter. Such cysts are almost always follicular and are related to hormonal stimulus from the pregnancy. Unless very large, these do not usually require in utero management, although in utero aspiration has been described. This should be considered if the cyst is thought likely to cause obstruction to delivery or be causing diaphragmatic elevation and subsequent pulmonary compromise. The vast majority of these, however, cause no difficulty in pregnancy or delivery. Postnatally the majority will resolve although this may take up to 6 months. If they are large and thought to be causing compression, then postnatal aspiration may be appropriate with postnatal ultrasound follow-up to check for resolution. Any recurrence or enlargement in size should raise the possibility of an alternative cause. Aspirate should be clear and straw-coloured in an uncomplicated cyst. Some authorities would advise early surgical removal of these cysts to preserve fertility, but other authorities would suggest that this is unlikely to affect the long-term outlook for ovarian function in later life. Auto-amputated cysts have been described. Cysts larger than 50 mm may be candidates for postnatal aspiration or removal. Ovarian cysts that show solid areas (Fig. 11.13) or debris (Fig. 11.14) usually indicate torsion or haemorrhage and should be removed postnatally. Polyhydramnios occurs in some pregnancies.

The second commonest cause (Fig. 11.15) is a duplication cyst. This is usually unilocular and, with high resolution, it is occasionally possible to

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**Table 11.1** Intra-abdominal cysts (excluding renal tract)

<table>
<thead>
<tr>
<th>Location</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Liver</td>
<td>Simple cyst/lymphangioma</td>
</tr>
<tr>
<td></td>
<td>Choledochal cyst (exclude large gall bladder)</td>
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<tr>
<td></td>
<td>Varix of umbilical vein</td>
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<tr>
<td>Adrenal</td>
<td>Simple cyst</td>
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<tr>
<td></td>
<td>Haemorrhage (evolving)</td>
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<tr>
<td></td>
<td>Neuroblastoma</td>
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<tr>
<td>Bowel</td>
<td>Duplication cyst</td>
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<tr>
<td></td>
<td>Mesenteric cyst/lymphangioma</td>
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<tr>
<td></td>
<td>Meconium peritonitis (exclude dilated bowel loops)</td>
</tr>
<tr>
<td>Ovarian</td>
<td>Follicular cyst</td>
</tr>
<tr>
<td>Uterus/vagina</td>
<td>Hydrometrocolpos</td>
</tr>
</tbody>
</table>

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**Fig. 11.11** Splenic cyst. Axial section showing splenic cyst (C), within left upper abdomen. This resolved in the neonatal period.

**Fig. 11.12** Oblique section of fetal abdomen in the third trimester, showing a cyst adjacent to the fetal bladder (FB). Postnatal follow-up confirmed this to be a follicular ovarian cyst.
demonstrate a multilayered wall. These cysts may be up to several centimetres in diameter and may be recognized in the second or third trimester. As they are closely related to bowel, they may sometimes cause compression of bowel in the postnatal period. They do not require intervention in utero and if they are not causing symptoms ex utero, urgent surgery is not usually required. All should, however, be removed in infancy even if not causing symptoms. Interestingly, since many of these cysts are relatively flaccid, they may well not be palpable in the neonatal period. They have been diagnosed as early as 12 weeks.

Mesenteric cysts, which are usually lymphangiomas, may also be seen in utero as transonic lesions, but are usually multilocular and not usually confused with the two former diagnoses (Fig. 11.16). These cysts frequently require operation after delivery and cannot always be entirely removed.
A pseudo-cyst related to meconium peritonitis can sometimes be diagnosed, but additional features including dilated bowel, peritoneal or bowel calcification and ascites often help to differentiate the cause (Fig. 11.17).

Posterior abdominal-wall tumour

Masses arising from the tissues of the posterior abdominal wall are extremely rare, but may arise from striated muscle or fibrous tissue. The appearances are those of a soft-tissue mass of slightly different density to the liver and spleen, usually extending into the abdomen, but sometimes seen extending outwards and distorting the abdominal circumference. They can be suspected by excluding other organs of origin such as liver, adrenal, kidney or bowel as the source of origin of the mass, but the ultimate diagnosis usually can only be confirmed postnatally, especially if the mass extends into the abdominal cavity.
There is no established routine of management of these lesions. The ultimate diagnosis is usually made only after delivery, although termination of a pregnancy for a lesion occurring earlier in pregnancy is feasible.

As most of these lesions are aggressive in character, the long-term outlook is poor, particularly if adjacent structures such as the spine are involved. Cystic enteric duplication cysts have been described as a cause of benign retroperitoneal mass.83

**Adrenal masses**

The normal adrenal gland in the fetus is relatively larger than the adrenal gland in the older child and adult. The glands are relatively inconspicuous with normal tissue density slightly higher than the adjacent liver, spleen or kidney. The density of the medulla is usually higher than the density of the cortex, and the glands are most easily recognized in the transverse section. The typical Y-shaped appearance of the adrenal gland in longitudinal section is best appreciated in cases of renal agenesis where the adrenal gland may fill the renal fossa and be mistaken for renal tissue. Appreciation of the similarity in appearances is usually sufficient to enable differentiation of a normal adrenal gland in cases of suspected renal agenesis.

**Adrenal haemorrhage**

Haemorrhage into the left adrenal gland in particular is an uncommon event seen in the second or third trimester.81,84 It is sometimes associated with fetal distress and growth retardation. Adrenal haemorrhage presents as a mass above the kidney, distorting the normal adrenal anatomy. The mass is usually hypoechoic, but can be mixed in character and will usually change in appearance over a period of time. The echogenicity varies with time and the size usually reduces. The condition has been described in association with renal vein thrombosis, particularly on the left side, and close examination of the kidneys is necessary. The condition usually has a benign outcome with the mass resolving in utero or early neonatal life without any long-term sequelae if the haemorrhage is isolated. Occasionally, a calcified residual lesion is seen. Adrenal function is not usually affected. Haemorrhagic cysts of the adrenal cortex are described in Beckwith–Wiedemann syndrome.85 Differentiation from neuroblastoma can sometimes be achieved using colour Doppler.86

**Neuroblastoma**

The other, well-described adrenal mass is a neuroblastoma. It is usually only seen in the third trimester. The appearance is variable, but it should be suspected in a suprarenal mass that is increasing in size with time. Evidence of spinal involvement should be sought, but is difficult to exclude. Secondary deposits in the liver, whilst subtle and often of density only slightly more than the surrounding liver, should be sought. Antenatally diagnosed neuroblastomas and neonatal cases have a good long-term prognosis, sometimes with spontaneous regression, but the diagnosis, which may be suspected antenatally, cannot be confirmed until the postnatal period. Unless the mass is enlarging rapidly, early delivery is probably not justified. Some neuroblastomas will have a cystic component and can entirely mimic an adrenal haemorrhage in utero.87 The diagnosis can only be confirmed biochemically and histologically after delivery.87 Other neuroblastomas will be solid, isoechoic or complex.88 Whilst some tumours do well, others do not. The DNA index may be the most important predictor.89 Hydrops is a complication seen in the more advanced stages of the disease (Stages IV and IVS).90 Accurate antenatal staging is impossible. Metastases to the umbilical cord and placenta are described.90 Masses can become so large that dystocia results (Fig. 11.18).

**Extralobar pulmonary sequestration**

Intrathoracic pulmonary sequestrated segments are easily recognized. Intra-abdominal sequestrated lung is a more difficult diagnosis to make, presenting usually as a solid suprarenal mass most often on the left side. It is impossible to differentiate from an adrenal lesion in utero. The relative lack of change in size with time makes haemorrhage unlikely, although the differential diagnosis from neuroblastoma cannot be made with certainty in utero.91 Most of these lesions are hyperechoic,98 but some are mixed with small cystic areas present (Fig 11.19). These lesions have been described in
association with diaphragmatic hernia. Histology can also be mixed with elements of congenital cystic adenomatoid malformation present.

**Uterus and vagina**

The normal uterus and vagina, like the ovary, are not visible in utero because of similarity of echo-texture to the surrounding structures. The labia can, however, be confidently seen in most females from 15 weeks onwards. Vaginal atresia or an imperforate hymen may present in utero as a cystic pelvic mass (hydrometrocolpos). This is a unilocular cyst arising from the pelvis. On high-quality scans it is sometimes possible to determine that the echogenicity of the fluid is greater than that of urine in the adjacent bladder. Frequently, because of the size of the mass, the adjacent bladder is compressed and differentiation of this mass from a distended bladder or other causes of pelvic mass such as an ovarian cyst can be difficult. There is frequently dilatation of the ureters and pelvicalyceal systems with this condition, which is unusual with an ovarian cyst, but this does not differentiate this condition (hydrometrocolpos) from a dilated obstructed or neurogenic bladder. Associated anomalies have been described, including anorectal atresia, other bowel atresias and polycystic kidneys. It is impossible to determine antenatally whether the cause of the abnormality is a simple, imperforate hymen that would require only a minor surgical procedure or vaginal atresia requiring major reconstructive surgery in later life. Amniotic fluid volume in hydrometrocolpos is usually normal, but can be reduced, sometimes critically so.

**Bowel abnormalities**

**Oesophageal atresia**

The normal oesophagus can occasionally be demonstrated in utero with high-resolution equipment (Fig. 11.20). A dilated upper oesophagus can be demonstrated in some types of oesophageal atresia, but is a late manifestation. Oesophageal atresia may be suspected if the fetal stomach is either small or not easily demonstrated on more than one occasion in utero in the second trimester. Caution must be exercised, however, in attributing this to oesophageal atresia as a number of other conditions with poor swallowing can also cause the same appearance including primary microgastria (small stomach). A small or absent stomach is the hallmark of oesophageal atresia without a tracheoesophageal fistula (Type A). Some cases of oesophageal atresia with fistula will also have a small stomach, but many cases of oesophageal atresia with
a tracheo-oesophageal fistula will have normal ultrasound appearances in the second trimester, because fluid can pass into the stomach via the trachea and distal tracheo-oesophageal fistula. Polyhydramnios developing in the third trimester may raise the possibility of this diagnosis even when the second trimester scan was normal. About two-thirds of cases of tracheo-oesophageal fistula and oesophageal atresia will have polyhydramnios and one-third will have a small, fetal stomach. Confident diagnosis of oesophageal atresia with a fistula has been described using systematic scanning of the neck and upper chest in three planes.\(^{105}\) This is only applicable in a high-risk situation and was only successful after 23 weeks, which is the common time for routine scanning, is very unusual. Differential diagnosis of polyhydramnios with absent stomach in the third trimester includes craniofacial anomalies, neuromuscular abnormalities and misplaced stomach, e.g. diaphragmatic hernia or situs inversus. Associated abnormalities are common including the VATER association (see Table 11.2). Cardiac, skeletal, genitourinary and other gastrointestinal abnormalities, central-nervous-system anomalies and facial anomalies have been described. Chromosome anomalies, especially trisomies 18 and 21, are also associated. The prognosis in the absence of other anomalies is good with a mortality rate of less than 10% for liveborn infants.\(^ {106}\) Much of the mortality is related to complications of polyhydramnios and prematurity. There is no evidence that prenatal diagnosis improves neonatal outcome.\(^ {107}\)

### Stomach

The normal stomach can usually be demonstrated on a mid-trimester scan. Its size is variable from time to time and patient to patient, but it is an easily identified transonic structure to the left of the midline in the upper abdomen. Other surrounding structures such as the left kidney and the liver should be identified to exclude the possibility of a renal cyst, hydrenephrosis or a large gall bladder in situs inversus as a cause of a cystic structure in the left upper abdomen. Other uncommon causes of cysts in the left upper quadrant include duplication cysts, mesenteric cysts and ovarian cysts. In situs inversus, the stomach will be on the right side, but the heart will also, in the complete form, be on that side. In partial situs inversus, the abdominal situs and cardiac situs will be different and this is commonly associated with complex isomorphic cardiac malformations.

A small stomach can be an isolated finding (microgastria)\(^ {108}\) or associated with other malformations such as oesophageal atresia, tracheo-oesophageal cleft or any cause of diminished or absent fetal swallowing such as major neuromuscular disorders.

### Duodenal atresia

The normal duodenum can only occasionally be demonstrated in the second and third trimester. Dilatation of the duodenum, which can be as large as or larger than the stomach, in its most marked forms suggests a diagnosis of duodenal atresia or stenosis (Fig. 11.21). The typical appearance is of a ‘double bubble’ with the stomach and duodenum each forming similarly sized, fluid-filled structures. Duodenal peristalsis with variation in size of the two bubbles relative to one another can sometimes be seen. It is important not to mistake a normal gall bladder for a slightly enlarged duodenum. Continuity between the two bubbles in duodenal obstruction prevents this mistake being made.

This diagnosis is rarely made before the latest part of the second trimester and when made in the third trimester is often associated with polyhydramnios. An isolated diagnosis of the condition in the first trimester has been made.\(^ {109}\) Duodenal atresia can be an isolated abnormality or part of the VATER association. Thirty percent of duodenal atresias occur in fetuses with Down’s syndrome.

Duodenal obstruction is usually caused either by complete obliteration of the duodenal lumen or by a web in the lumen of the duodenum. It is impossible to determine the cause antenatally. The postnatal association of Ladd’s bands and malrota-

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<th>Table 11.2 Features of VATER association</th>
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Cardiac abnormalities are also a common associated feature (Smith DW. The VATER association. Am J Dis Child 1974; 128:767–770.)
tion of the bowel also cannot be identified antenatally. Apart from the association with Down’s syndrome and the VATER association, other concurrent isolated abnormalities have been described in all systems. About 50% of duodenal atresia fetuses have other abnormalities. In the absence of other abnormalities the postnatal outlook is excellent.\textsuperscript{110}

Duodenal atresia will occasionally be associated with oesophageal atresia to produce a characteristic ‘C’-shaped cystic mass within the upper abdomen (Fig. 11.22).\textsuperscript{111,112}

### Jejunal and ileal atresia

Separate loops of normal jejunum and ileum are not visible on antenatal scanning, although with modern, high-resolution scanners, the loops of bowel can be clearly differentiated from the other abdominal organs. If multiple, fluid-filled, bowel loops are demonstrated persistently on antenatal scanning, a small bowel atresia must be suspected. Small bowel atresias are rare (probably less than 1 in 5000 deliveries). Atresias of the jejunum (Fig. 11.23) are almost twice as common as those of the ileum.\textsuperscript{113} The exact site and cause of the atresia cannot be determined, but, in general terms, the more loops of bowel present and the more dilated they are, the more distal the atresia is likely to be. Multiple atresias are well described. Association with malrotation cannot be determined antenatally.

Associated abnormalities of the bowel are described (malrotation, gastrochisis, duplication, meconium ileus) but outside the gastrointestinal tract abnormalities are uncommon. The gastrointestinal tract associations in many situations are thought to be the cause of the atresia rather than the effect. In the absence of other abnormalities, the outlook for small bowel atresias is excellent, ileal atresias slightly...
better than jejunal because of the association of polyhydramnios and prematurity with some jejunal atresia. Differentiation from ureteric dilatation is essential. Differentiation from large bowel dilatation is usually possible by considering the position of the bowel loops. This diagnosis is rarely made before the late second trimester and more usually in the third trimester.114 As well as atresia of the small bowel, associated conditions, such as meconium ileus, should also be considered.

Meconium ileus

Meconium ileus is a common manifestation of cystic fibrosis in the fetal and neonatal period. The condition is one in which the bowel content becomes extremely thick and tenacious causing occlusion of the bowel that typically starts in the terminal ileum. Dilatation of the bowel proximal to the occlusion usually occurs. Complications such as volvulus and atresia are well recognized. The condition is, however, not often suspected antenatally, but should be suspected if the bowel is ‘bright as bone’ and particularly if this bright bowel is associated with dilatation or abdominal calcification. The brightness in the bowel is due to increased reflectivity of the inspissated meconium in the terminal ileum.115–118 The diagnosis can be confirmed by genotyping the parents and if both have one of the cystic fibrosis genes (most commonly Delta F 508), then the diagnosis is confirmed. About 80% of cases of cystic fibrosis presenting as meconium ileus can be diagnosed by parental genotyping. The condition is inherited in an autosomal recessive manner, each succeeding pregnancy having a 1 in 4 risk. The prognosis for neonates with meconium ileus is good, but the long-term prognosis for the individual is related to the outlook for cystic fibrosis, which, whilst improving, causes significant morbidity with increasing mortality from the late teens onwards.

Meconium peritonitis

This condition occurs when perforation of the bowel occurs in utero. It is manifested by calcification in the peritoneum typically peripherally arranged in the abdomen or associated with an intra-abdominal cyst, ascites or meconium calcification. (Fig. 11.17). It can be associated with meconium ileus (10%),119 but other causes of perforation are more common. The calcification is caused by a chemical reaction between the peritoneum and irritant meconium. The prognosis depends on the underlying cause, but, in the absence of cystic fibrosis, is generally good.120–123 Cases have been described in association with cocaine abuse,124 in the rubella syndrome125 and associated with cytomegalovirus infection,126 but most are associated with volvulus, atresia or meconium ileus.127 Prenatal appearances will predict postnatal outcome.128

Large bowel pathology

The normal large bowel is not always clearly identified in the second trimester. In the third trimester, the appearances are very varied and frequently the meconium is of low echogenicity so that the colon stands out from the other abdominal contents and can be mistaken for dilated small bowel or ureters. The typical haustral pattern and peripheral position of the bowel in the abdomen almost always enables the normal large bowel to be differentiated from dilated small bowel. Dilatation of the large bowel has been described in imperforate anus,129–131 but this condition is more frequently diagnosed postnatally following a normal antenatal scan.
Intraluminal, large-bowel calcification has been described antenatally and postnatally in imperforate anus with communication between the large bowel and the urinary tract. The reaction between urine and meconium causes the calcification. Imperforate anus is often associated with the other anomalies of the VATER association (Table 11.2), the renal and sacral abnormalities being the most frequent.

**Persistent cloaca**

This results from lack of development of the cloacal septum, so that the genital, gastrointestinal and urinary tracts all open into a single structure. Several cases have been reported antenatally, usually presenting as a septated pelvic mass with oligohydramnios and poor fetal growth. The diagnosis should be considered in any female fetus presenting with hydronephrosis, a cystic pelvic mass and a bladder that is difficult to define separate from the mass. Prognosis is good in the absence of life-threatening, associated anomalies, although other such anomalies are common leading to termination in a large number of cases.

**Hirschsprung’s disease**

This has only occasionally been diagnosed in utero with variable appearances suggesting both large and small bowel dilatation. Differentiating this from other causes of bowel dilatation is virtually impossible. Colon atresia has not yet been described antenatally.

**Bright bowel**

The observation of increased echogenicity of the bowel above the normal is a non-specific finding. The standard is that the bowel should be as ‘bright as bone’ to be considered abnormal (Fig. 11.25). This appearance is associated in some cases with Down’s syndrome, intra-uterine growth retardation, cystic fibrosis, swallowed blood and viral infections, but in the majority of cases no underlying cause is found. If this appearance is seen in the second trimester, some would consider invasive testing for karyotype to be advisable, together with testing to exclude cystic fibrosis. Long-term follow-up to exclude intra-uterine growth retardation would also be considered appropriate.

**Megacystis microcolon intestinal hypoperistalsis syndrome (MMIH)**

This rare condition can be suspected in utero if the fetal bladder is particularly large and there is hydronephrosis. Dilated bowel may also be seen in this circumstance, but is not usually a predominant feature. It is an important condition to consider as a differential diagnosis of posterior urethral valves because invasive management with drainage of the bladder in this condition is futile as the long-term outlook for affected babies is extremely poor. Differential diagnosis is helped by the fact that the vast majority of fetuses with this condition are female whereas posterior urethral valves are an entirely male condition. Recent evidence suggests an autosomal recessive mode of inheritance in some cases.

**Sacrococcygeal teratomas**

Whilst the majority of these lesions arising from the coccyx produce a mass at the lower end of the spine or in the buttock, the intrapelvic form of these
lesions can appear isolated and should be considered with any mixed solid and cystic mass arising in the pelvis.

According to the accepted classification, these are type IV tumours. An intra-abdominal component has been seen in 40% of cases, but rarely has the tumour been entirely intrapelvic or intra-abdominal. The masses can be entirely cystic and difficult to differentiate from hydrometrocolpos, a dilated urinary bladder or an anterior meningocele. Polyhydramnios is a common feature, especially in later diagnosed cases and hydrops occurs in some as a result of high output cardiac failure secondary to the vascular nature of some of these tumours. Associated musculoskeletal abnormalities have been described. The outlook for these cases must be guarded since a confident diagnosis of type IV tumour is unlikely in utero and an entirely intra-abdominal or intrapelvic tumour has a greater risk of malignancy than the commoner lesions presenting as external masses. Overall mortality of 32% for the lesion relates to the complications of polyhydramnios and hydrops as much as to the lesion itself. More recent series have, however, suggested an even poorer prognosis with a 62% perinatal mortality, although all those who died were born before 34 weeks. As these tumours produce alphafetoprotein, maternal serum alphafetoprotein should be elevated, although only occasionally is this a cause of raised alphafetoprotein assay in the second trimester.

Interventions including cyst aspiration, fetal surgery and radio frequency ablation, as well as amnio infusion or reduction have been shown to improve prognosis, but 81% of cases have serious antenatal complications.

Fetal MRI is a useful addition, improving counselling of parents regarding complications and outcome.

**Ascites**

Any fluid seen surrounding the bowel is abnormal. A very thin, black line around the inner aspect of the abdomen is, however, a normal appearance and should not be confused with small amounts of ascites. Small amounts of ascites can be sometimes identified adjacent to the right lobe of the liver. Isolated ascites is a frequent, early manifestation of hydrops fetalis and the causes of this should be sought both by ultrasound, excluding cardiac lesions and other fetal abnormalities, and by appropriate invasive testing to exclude the other causes, although persistent isolated ascites is most commonly associated with intra-abdominal pathology. Ascites caused by intra-abdominal pathology is quite uncommon. However, the two most frequent associations are urinary ascites secondary to obstructive uropathy and meconium peritonitis. Rare associations are with liver disease and certain metabolic storage diseases. It is usually (92%), but not always possible to determine the cause of the ascites. In general terms, when associated with hydrops or abnormalities or when detected before 23 weeks, the outlook is poor.

**Intra-abdominal calcification**

Calcification within the fetal abdomen is rare and has many individual causes. If the organ of origin can be determined, then the cause of calcification can often be determined (see Table 11.3).

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References

Exomphalos

Gastroschisis

**Limb–body-wall complex**


**Cloacal and bladder exstrophy**


**Normal anatomy**


**Liver**


**Gall bladder**


**Choledochal cysts**


**Spleen**

Intra-abdominal cysts

Posterior abdominal wall tumours

Adrenal masses

Extra-lobar pulmonary sequestration

Uterus and vagina
11. Abdominal and abdominal-wall abnormalities


Oesophageal atresia

Stomach

Duodenal atresia

Jejunal and ileal atresia

Meconium ileus

Meconium peritonitis
Large bowel pathology

Persistent cloaca

Hirschsprung’s disease

Bright bowel

Megacystis microcolon intestinal hypoperistalsis syndrome

Sacrococcygeal teratoma