Diploma in Fetal Medicine & ISUOG Educational Series
The 18 - 23 weeks scan
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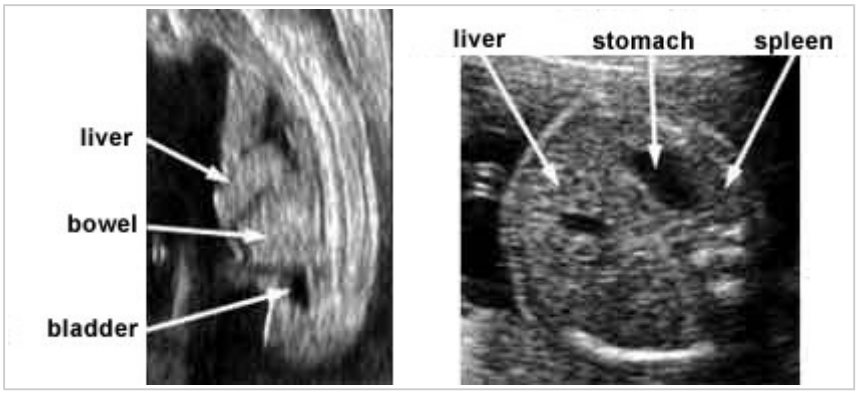
Chapter 7

Gastrointestinal tract

NORMAL SONOGRAPHIC ANATOMY

Sonographically, the fetal stomach is visible from 9 weeks of gestation as a sonolucent cystic structure in the upper left quadrant of the abdomen. The bowel is normally uniformly echogenic until the third trimester of pregnancy, when prominent meconium-filled loops of large bowel are commonly seen. The liver comprises most of the upper abdomen and the left lobe is greater in size than the right due to its greater supply of oxygenated blood. The gall bladder is seen as an ovoid cystic structure to the right and below the intrahepatic portion of the umbilical vein. The spleen may also be visualized in a transverse plane posterior and to the left of the fetal stomach. The abdominal circumference should be measured in a cross-section of the abdomen demonstrating the stomach and portal sinus of the liver. The visceral situs should be assessed, by demonstrating the relative position of the stomach, hepatic vessels, abdominal aorta and inferior vena cava.



ESOPHAGEAL ATRESIA

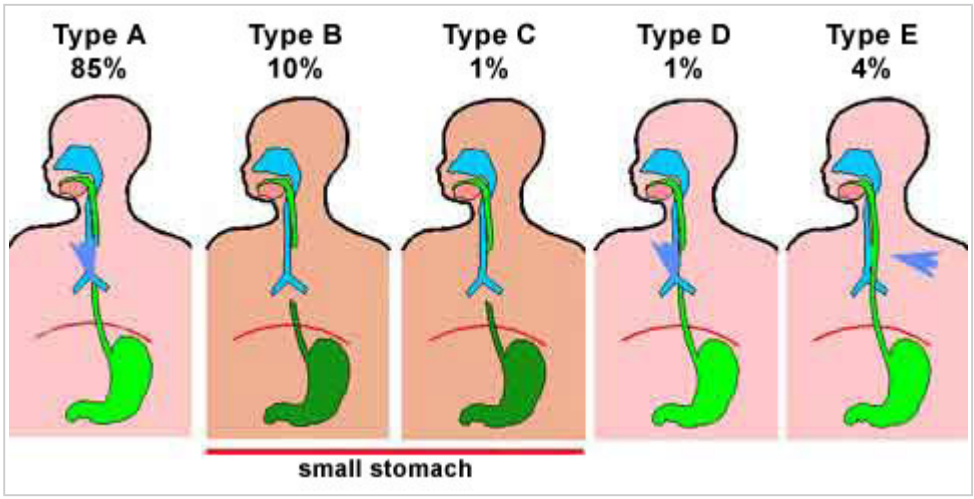
Esophageal atresia and tracheoesophageal fistulae, found in about 90% of cases, result from failure of the primitive foregut to divide into the anterior trachea and posterior esophagus, which normally occurs during the 4th week of gestation.

Prevalence

Esophageal atresia is found in about 1 in 3000 births.

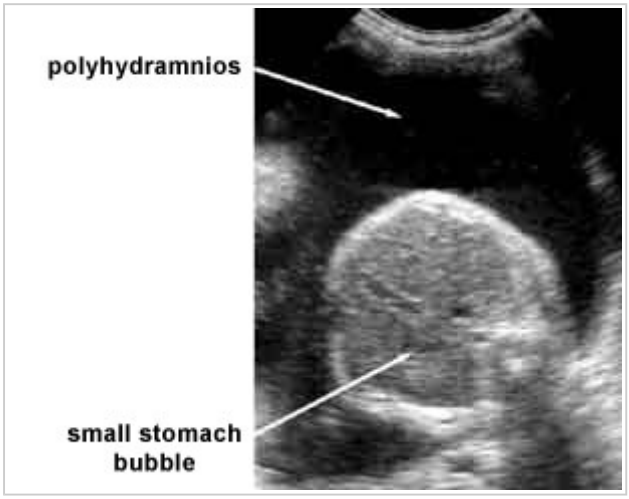
Etiology

Esophageal atresia and tracheoesophageal fistulae are sporadic abnormalities. Chromosomal abnormalities (mainly trisomy 18 or 21) are found in about 20% of fetuses. Other major defects, mainly cardiac, are found in about 50% of the cases. Tracheoesophageal fistulae may be seen as part of the VATER association (vertebral and ventricular septal defects, anal atresia, tracheoesophageal fistula, renal anomalies, radial dysplasia and single umbilical artery). In over 80% of cases, esophageal atresia occurs in association with a tracheo-esophageal fistula, allowing intake of amniotic fluid from the stomach, that may be therefore normally distended, particularly in early gestation.

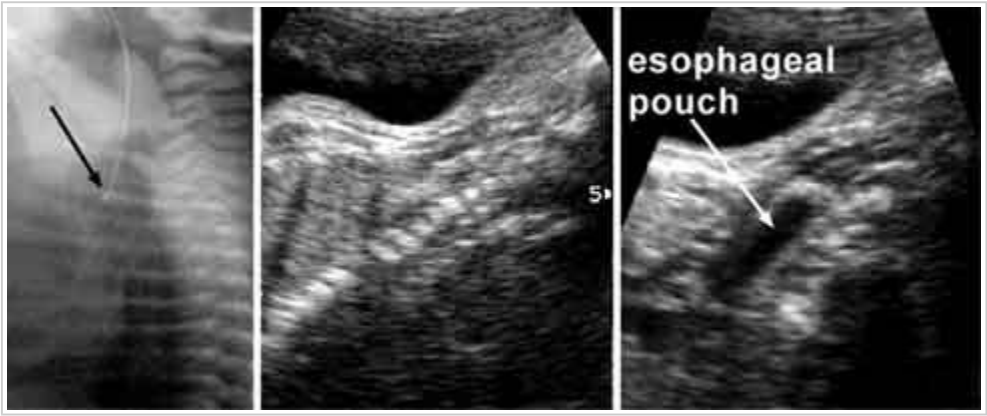


Diagnosis

Prenatally, the diagnosis of esophageal atresia is suspected when, in the presence of polyhydramnios (usually after 25 weeks), repeated ultrasonographic examinations fail to demonstrate the fetal stomach.



However, gastric secretions may be sufficient to distend the stomach and make it visible. If there is an associated fistula, the stomach may look normal. Occasionally (after 25 weeks), the dilated proximal esophageal pouch can be seen as an elongated upper mediastinal and retrocardiac anechoic structure. This is a dynamic finding, however, that occurs only at the time of fetal swallowing, and requires therefore prolonged sonographic visualization.



The differential diagnosis for the combination of absent stomach and polyhydramnios includes intrathoracic compression, by conditions such as diaphragmatic hernia, and muscular-skeletal anomalies causing inability of the fetus to swallow.

Prognosis

Survival is primarily dependent on gestation at delivery and the presence of other anomalies. Thus, for babies with an isolated tracheoesophageal fistula, born after 32 weeks, when an early diagnosis is made, avoiding reflux and aspiration pneumonitis, postoperative survival is more than 95%.

DUODENAL ATRESIA

At 5 weeks of embryonic life, the lumen of the duodenum is obliterated by proliferating epithelium. The patency of the lumen is usually restored by the 11th week and failure of vacuolization may lead to stenosis or atresia. Duodenal obstruction can also be caused by compression from the surrounding annular pancreas or by peritoneal fibrous bands.

Prevalence

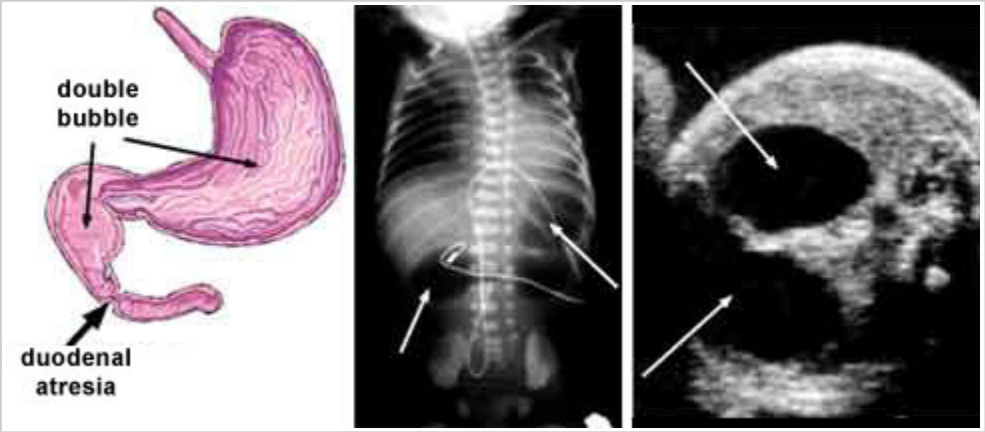
Duodenal atresia is found in about 1 per 5000 births.

Etiology

Duodenal atresia is a sporadic abnormality, although, in some cases, there is an autosomal recessive pattern of inheritance. Approximately half of fetuses with duodenal atresia have associated abnormalities, including trisomy 21 (in about 40% of fetuses) and skeletal defects (vertebral and rib anomalies, sacral agenesis, radial abnormalities and talipes), gastrointestinal abnormalities (esophageal atresia/tracheoesophageal fistula, intestinal malrotation, Meckel's diverticulum and anorectal atresia), cardiac and renal defects.

Diagnosis

Prenatal diagnosis is based on the demonstration of the characteristic 'double bubble' appearance of the dilated stomach and proximal duodenum, commonly associated with polyhydramnios.



However, obstruction due to a central web may result in only a 'single bubble', representing the fluid-filled stomach. Continuity of the duodenum with the stomach should be demonstrated to differentiate a distended duodenum from other cystic masses, including choledochal or hepatic cysts. Although the characteristic 'double bubble' can be seen as early as 20 weeks, it is usually not diagnosed until after 25 weeks, suggesting that the fetus is unable to swallow a sufficient volume of amniotic fluid for bowel dilatation to occur before the end of the second trimester of pregnancy.

Prognosis

Survival after surgery in cases with isolated duodenal atresia is more than 95%.

INTESTINAL OBSTRUCTION

Intestinal obstructions are either intrinsic or extrinsic. Intrinsic lesions result from absent (atresia) or partial (stenosis) recanalization of the intestine. In cases of atresia, the two segments of the gut may be either completely separated or connected by a fibrous cord. In cases of stenosis, the lumen of the gut is narrowed or the two intestinal

segments are separated by a septum with a central diaphragm. Apple-peel atresia is characterized by absence of a vast segment of the small bowel, which can include distal duodenum, the entire jejunum and proximal ileus. Extrinsic obstructions are caused by malrotation of the colon with volvulus, peritoneal bands, meconium ileus, and agangliosis (Hirschsprung's disease). The most frequent site of small bowel obstruction is distal ileus (35%), followed by proximal jejunum (30%), distal jejunum (20%), proximal ileus (15%). In about 5% of cases, obstructions occur in multiple sites. Anorectal atresia results from abnormal division of the cloaca during the 9th week of development.

Prevalence

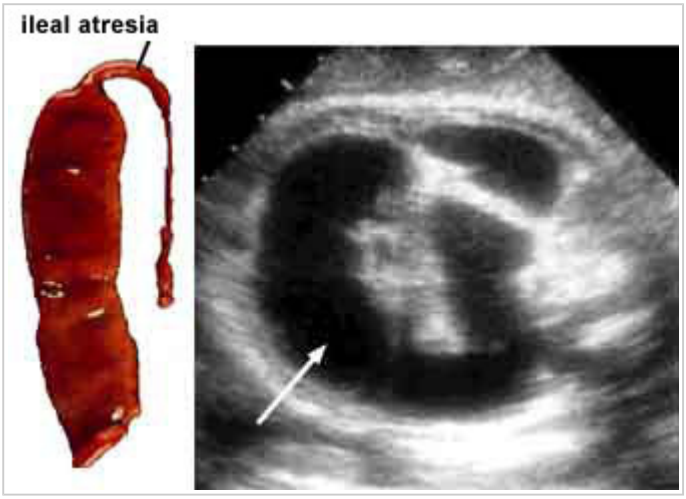
Intestinal obstruction is found in about 1 per 2000 births; in about half of the cases, there is small bowel obstruction and in the other half anorectal atresia.

Etiology

Although the condition is usually sporadic, in multiple intestinal atresia, familial cases have been described. Associated abnormalities and chromosomal defects are rare. In contrast with anorectal atresia, associated defects such as genitourinary, vertebral, cardiovascular and gastrointestinal anomalies are found in about 80% of cases.

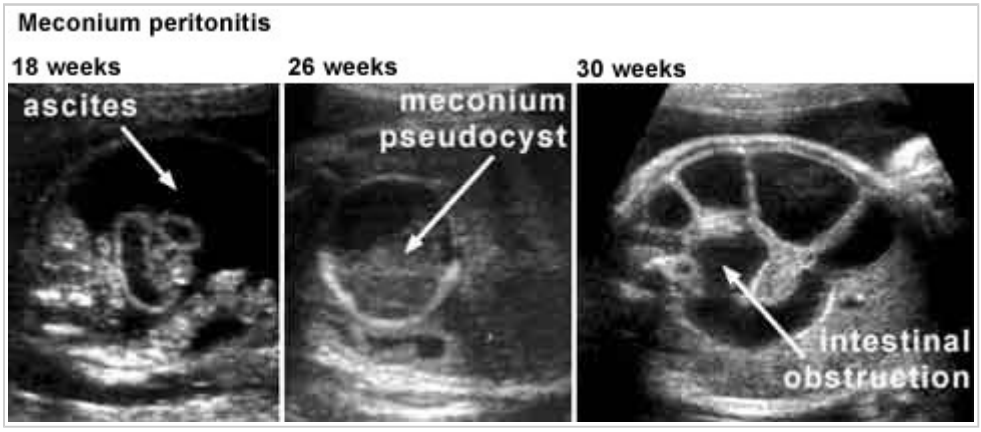
Diagnosis

The lumens of the small bowel and colon do not normally exceed 7 mm and 20 mm, respectively. Diagnosis of obstruction is usually made quite late in pregnancy (after 25 weeks), as dilatation of the intestinal lumen is slow and progressive. Jejunal and ileal obstructions are imaged as multiple fluid-filled loops of bowel in the abdomen.



The abdomen is usually distended and active peristalsis may be observed.

If bowel perforation occurs, transient ascites, meconium peritonitis and meconium pseudocysts may ensue.



Polyhydramnios (usually after 25 weeks) is common, especially with proximal obstructions. Bowel enlargement and polyhydramnios may be found in fetuses with Hirschsprung's disease, the megacystis– microcolon–intestinal

hypoperistalsis syndrome and congenital chloride diarrhea. When considering a diagnosis of small bowel obstruction, care should be taken to exclude renal tract abnormalities and other intra-abdominal cysts such as mesenteric, ovarian or duplication cysts. In anorectal atresia, prenatal diagnosis is usually difficult because the proximal bowel may not demonstrate significant dilatation and the amniotic fluid volume is usually normal; occasionally calcified intraluminal meconium in the fetal pelvis may be seen.

Prognosis

Infants with bowel obstruction typically present in the early neonatal period with symptoms of vomiting and abdominal distention. The prognosis is related to the gestational age at delivery, the presence of associated abnormalities and site of obstruction. In those born after 32 weeks with isolated obstruction requiring resection of only a short segment of bowel, survival is more than 95%. Loss of large segments of bowel can lead to short gut syndrome, which is a lethal condition.

HIRSCHSPRUNG'S DISEASE

Hirschsprung's disease is characterized by congenital absence of intramural parasympathetic nerve ganglia in a segment of the colon. It derives from failure of migration of neuroblasts from the neural crest to the bowel segments, which generally occurs between the 6th and 12th weeks of gestation. Another theory suggests that the disease is caused by degeneration of normally migrated neuroblasts during either pre- or postnatal life.

Prevalence

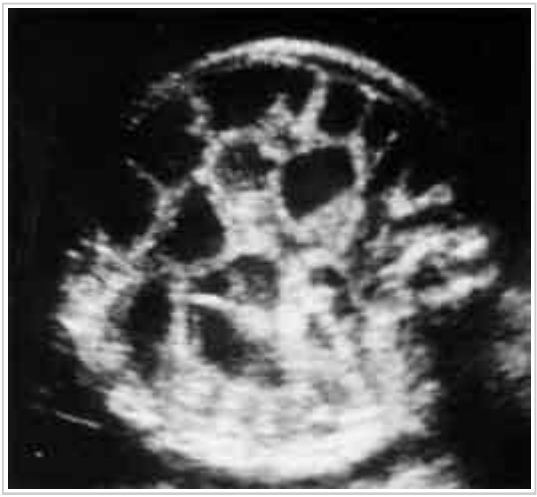
The disease occurs in about 1 in 3000 births.

Etiology

It is considered to be a sporadic disease, although in about 5% of cases there is a familial inheritance. In a small number of cases, Hirschsprung's disease is associated with trisomy 21.

Diagnosis

The aganglionic segment is unable to transmit a peristaltic wave, and therefore meconium accumulates and causes dilatation of the lumen of the bowel.



The ultrasound appearance is similar to that of anorectal atresia, when the affected segment is colon or rectum. Polyhydramnios and dilatation of the loops are present in the case of small bowel involvement; on this occasion, it is not different from other types of obstruction.

Prognosis

Postnatal surgery is aimed at removing the affected segment and this may be a two-stage procedure with temporary colostomy. Neonatal mortality is approximately 20%.

MECONIUM PERITONITIS

Intrauterine perforation of the bowel may lead to a local sterile chemical peritonitis, with the development of a dense calcified mass of fibrous tissue sealing off the perforation. Bowel perforation usually occurs proximal to some form of obstruction, although this cannot always be demonstrated.

Etiology

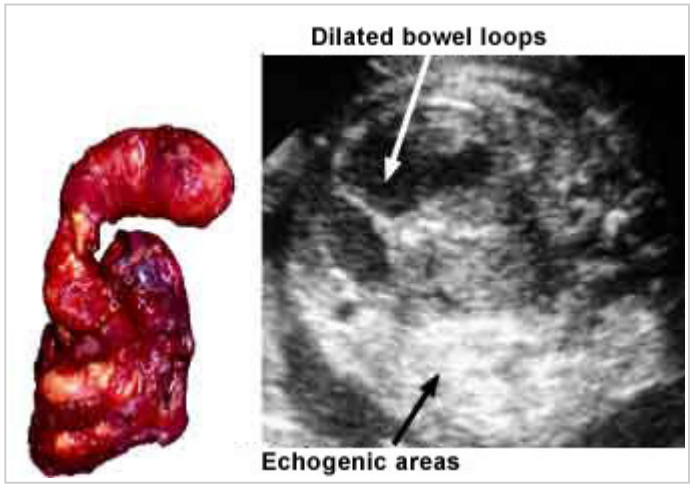
Intestinal stenosis or atresia and meconium ileus account for 65% of the cases. Other causes include volvulus and Meckel's diverticulum. Meconium ileus is the impaction of abnormally thick and sticky meconium in the distal ileum, and, in the majority of cases, this is due to cystic fibrosis.

Prevalence

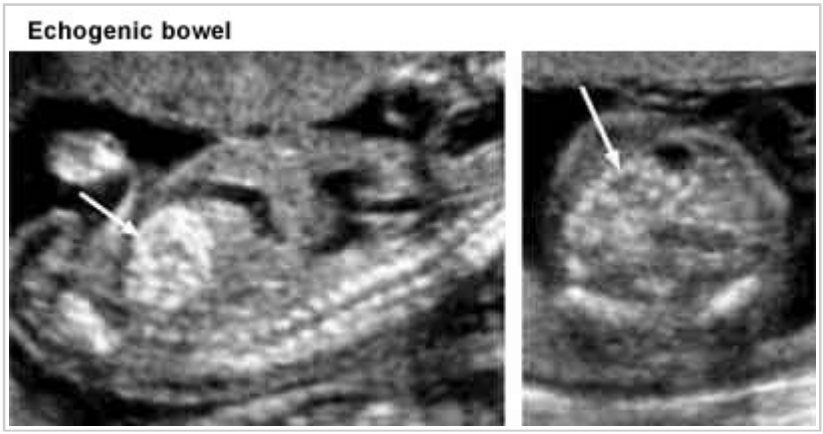
Meconium peritonitis is found in about 1 in 3000 births.

Diagnosis

In the typical case, meconium peritonitis is featured by the association of intra-abdominal echogenic area, dilated bowel loops and ascites.



The diagnosis should be considered if the fetal bowel is observed to be dilated or whenever an area of fetal intra-abdominal hyperechogenicity is detected.



The differential diagnosis of hyperechogenic bowel includes: intra-amniotic hemorrhage; early ascites; fetal hypoxia; meconium peritonitis; and cystic fibrosis.

Meconium ileus and hyperechogenic fetal bowel at 16–18 weeks of gestation may be present in 75% of fetuses with cystic fibrosis. The prevalence of cystic fibrosis in fetuses with prenatal diagnosis of intestinal obstruction may be about 10%. Therefore, when other causes of bowel hyperechogenicity have been excluded, DNA studies for cystic fibrosis should be considered.

Prognosis

Meconium peritonitis is associated with a more than 50% mortality in the neonatal period.

HEPATOSPLENOMEGALY

The fetal liver and spleen can be measured by ultrasonography. Causes of hepatosplenomegaly include immune and non-immune hydrops, congenital infection and metabolic disorders, and it is seen in Beckwith–Wiedemann and Zellweger syndromes. Hepatic enlargement may also be caused by hemangioma, which is usually hypoechogenic, or hepatoblastoma (the most frequent malignant tumor in fetal life), in which there are areas of calcification.

HEPATIC CALCIFICATIONS

Hepatic calcifications are echogenic foci in the parenchyma or the capsule of the liver.

Prevalence

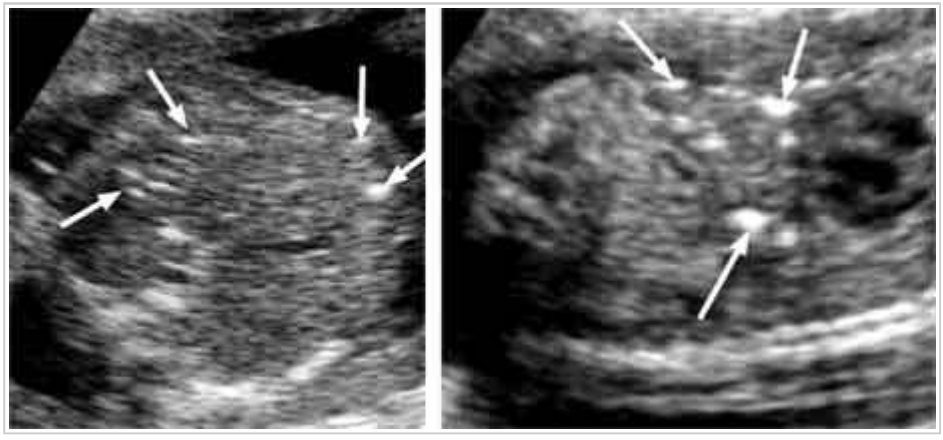
Hepatic calcifications are found at mid-trimester ultrasonography in about 1 per 2000 fetuses.

Etiology

The vast majority of cases are idiopathic but, in a few cases, hepatic calcifications have been found in association with congenital infections and chromosomal abnormalities.

Diagnosis

Solitary or multiple echogenic foci (1–2 mm in diameter) are observed within the substance of the liver or in the capsule.



Prognosis

This depends on the presence of associated infection or chromosomal defects. Isolated foci are of no pathological significance.

ABDOMINAL CYSTS

Abdominal cystic masses are frequent findings at ultrasound examination. Renal tract anomalies or dilated bowel are the most common explanations, although cystic structures may arise from the biliary tree, ovaries, mesentery or uterus. The correct diagnosis of these abnormalities may not be possible by ultrasound examination, but the most likely diagnosis is usually suggested by the position of the cyst, its relationship with other structures and the

normality of other organs.

Choledochal cysts

Choledochal cysts represent cystic dilatation of the common biliary duct. They are uncommon and their etiology is unknown. Prenatally, the diagnosis may be made ultrasonographically by the demonstration of a cyst in the upper right side of the fetal abdomen. The differential diagnosis includes enteric duplication cyst, liver cysts, situs inversus or duodenal atresia. The absence of polyhydramnios or peristalsis may help to differentiate the condition from bowel disorders. Postnatally, early diagnosis and removal of the cyst may avoid the development of biliary cirrhosis, portal hypertension, calculi formation or adenocarcinoma. The operative mortality is about 10%.

Ovarian cysts

Ovarian cysts are common and they may be found in up to one-third of newborns at autopsy, although they are usually small and asymptomatic. Fetal ovarian cysts are hormone-sensitive (human chorionic gonadotropin from the placenta) and tend to occur after 25 weeks of gestation; they are more common in diabetic or rhesus isoimmunized mothers as a result of placental hyperplasia. The majority of cysts are benign and resolve spontaneously in the neonatal period. Potential complications include development of ascites, torsion, infarction or rupture. Prenatally, the cysts are usually unilateral and unilocular, although, if the cyst undergoes torsion or hemorrhage, the appearance is complex or solid. Large ovarian cysts can be found in association with polyhydramnios, possibly as a consequence of compression of the bowel. Obstetric management should not be changed, unless an enormous or rapidly enlarging cyst is detected or there is associated polyhydramnios; in these cases, prenatal aspiration may be considered. A difficult differential diagnosis is from hydrometrocolpos, which also presents as a cystic or solid mass arising from the pelvis of a female fetus. Other genitourinary or gastrointestinal anomalies are common and include renal agenesis, polycystic kidneys, esophageal atresia, duodenal atresia and imperforate anus. Most cases are sporadic, although a few cases are genetic, such as the autosomal recessive McKusick–Kaufman syndrome with hydrometrocolpos, polydactyly and congenital heart disease.

Mesenteric or omental cysts

Mesenteric or omental cysts may represent obstructed lymphatic drainage or lymphatic hamartomas. The fluid contents may be serous, chylous or hemorrhagic. Antenatally, the diagnosis is suggested by the finding of a multiseptate or unilocular, usually mid-line, cystic lesion of variable size; a solid appearance may be secondary to hemorrhage. Antenatal aspiration may be considered in cases of massive cysts resulting in thoracic compression. Postnatal management is conservative and surgery is reserved for cases with symptoms of bowel obstruction or acute abdominal pain following torsion or hemorrhage into a cyst. Complete excision of cysts may not be possible because of the proximity of major blood vessels and in up to 20% of cases there is recurrence after surgery. Although malignant change in mesenteric cysts has been described, this is rare.

Hepatic cysts

Hepatic cysts are typically located in the right lobe of the liver. They are quite rare and result from obstruction of the hepatic biliary system. They appear as unilocular, intrahepatic cysts, and they are usually asymptomatic, although rarely may show complications such as infections or hemorrhages. In 30% of the cases of polycystic kidneys (adult type), asymptomatic hepatic cysts may be associated.

Intestinal duplication cysts

These are quite rare, and may be located along the entire gastrointestinal tract. They sonographically appear as tubular or cystic structures of variable size. They may be isolated or associated with other gastrointestinal malformations. Differential diagnosis includes other intra-abdominal cystic structures and also bronchogenic cysts, adenomatoid cystic malformation of the lung and pulmonary sequestration. Thickness of the muscular wall of the cysts and presence of peristalsis may facilitate the diagnosis. Postnatally, surgical removal is carried out.

Anomalies of the umbilical vein

Abnormalities of the umbilical vein, which are very rare, can be divided in three groups:

(1) Persistence of the right umbilical vein with ductus venosus and presence or absence of left umbical vein;

(2) Absence of the ductus venosus with extrahepatic insertion of the umbilical vein; and

(3) Dilated umbilical vein with normal insertion.

Normally, the umbilical vein enters the abdomen almost centrally at the level of the liver and courses on the left of the gallbladder. Persistence of the right umbilical vein is demonstrated by the fact that it is localized on the right of the gallbladder, bending towards the stomach. Color Doppler may help to diagnose these anomalies and may allow the differential diagnosis with other cystic abdominal lesions. Associated anomalies are frequent in anomalies of the first two groups and this influences the prognosis. These anomalies include cardiac, skeletal, gastrointestinal and urinary anomalies. The anomalies of the third group are rarely associated with other anomalies, and prognosis depends on the time at diagnosis and dimension of the varicosity.