Prenatal Diagnosis and Obstetric Management

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In Europe ultrasound examinations are commonly performed in virtually all pregnancies, usually between 11 and 13 weeks' gestation and around 20 weeks' gestation [1]. One of the main objectives of these investigations is the detection of fetal anomalies. The uptake of anomalies is variable in different studies, and indeed, the value of universal screening for anatomic malformations is debated [1–3]. The detection rate much varies depending upon different factors and the affected organs in particular. Sonographic investigation of the fetal gastrointestinal tract suffers from many limitations mostly because the fetal bowel is almost completely empty in early gestation. Furthermore, the esophagus and anorectal tract are incompletely seen. As a consequence of this, most intestinal obstructions are not identified until late in gestation or even after birth (Fig. 1.1).

Nevertheless, the identification of abnormal fetal sonographic findings of the gastrointestinal tract does occur, and in these cases, pediatric specialists are usually consulted to discuss the management strategy in the perinatal period and the prognosis. Such consultations have a particular relevance when the diagnosis is made in early gestation and the couples are considering the option of a pregnancy termination. It seems important to stress that caution is necessary when discussing the implications of antenatal diagnosis. The accuracy of sonography is limited, and anomalies identified in utero tend to have a different outcome than those that are identified after birth. Ancillary methods are now available for prenatal diagnosis in selected cases, including genetic testing and magnetic resonance, and multidisciplinary discussion is certainly indicated.

In the following pages, we will briefly review the state of the art of prenatal diagnosis of the anomalies of the gastrointestinal tract, focusing upon the information that seem relevant for the pediatric surgeons who work in close contact with obstetric departments.

1.1 Normal Sonographic Appearance of the Fetal Gastrointestinal Tract

Fetuses start swallowing amniotic fluid early in gestation, and the fluid-filled stomach is visible as early as 9 weeks of gestation as a C-shaped sonolucent structure in the upper left quadrant of the abdomen. The bowel has normally a uniform echogenic appearance until the third trimester of pregnancy when meconium-filled loops of large bowel are commonly seen. The liver is large prenatally and comprises most of the upper abdomen. The gallbladder is usually seen since midgestation as an ovoid cystic structure to the right and below the intrahepatic portion of the

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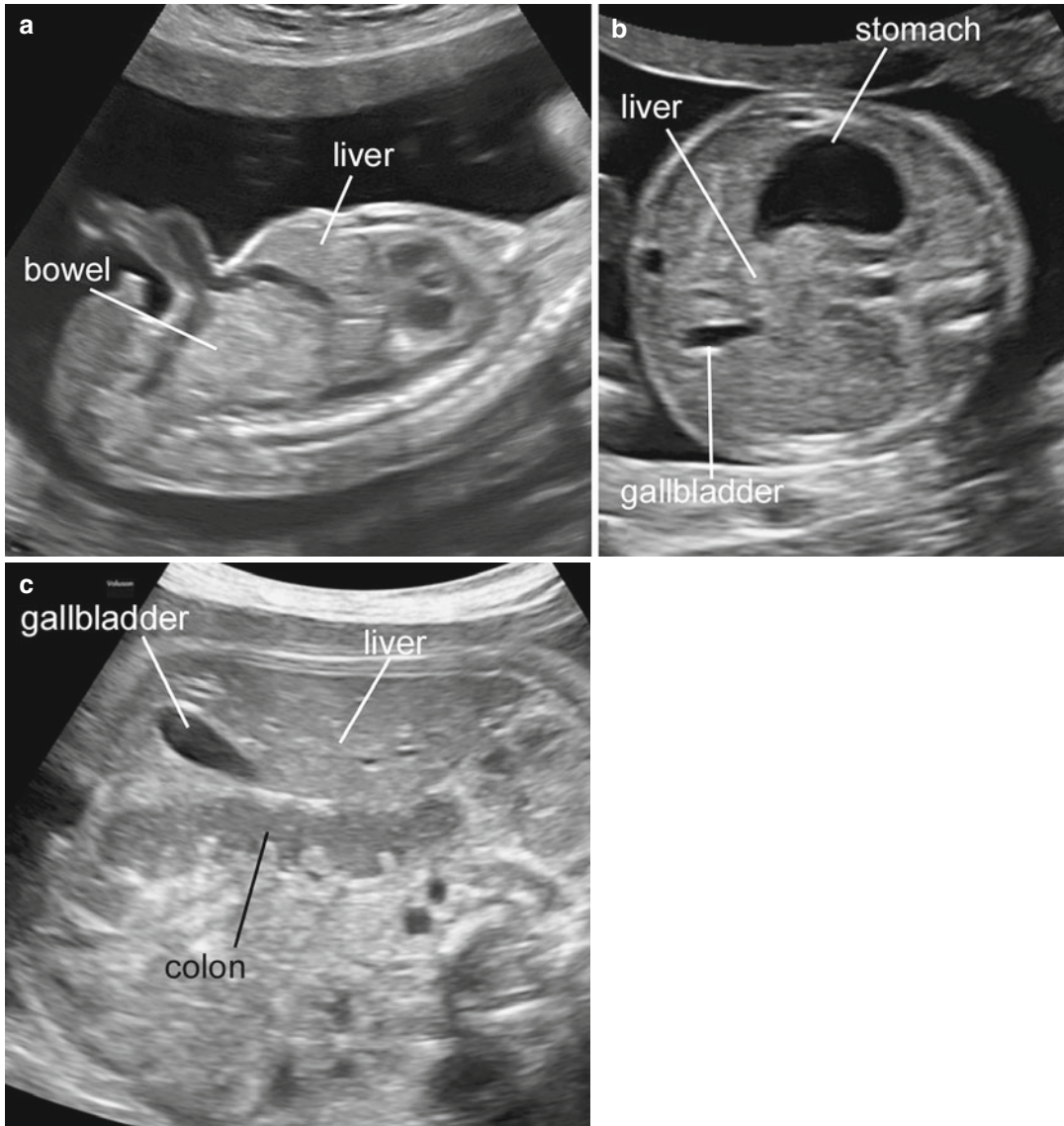e-mail: gianluigi.pilu@aosp.bo.it

Fig. 1.1 The normal appearance of the fetal gastrointestinal system at 20 weeks (**a**, **b**) and close to term gestation (**c**); at 20 weeks the liver is large and occupies the entire upper abdomen, and the stomach and gallbladder are fluid filled and easily visible; the bowel is empty and appears sonographically homogeneous; in advanced gestation, the meconium-filled large bowel can be seen

umbilical vein. The spleen may also be visualized posterior and to the left of the fetal stomach. The proximal and distal esophagus can be at times visualized, when the fetus is in a favorable position and particularly in the course of swallowing. However, it is impossible to visualize the entire length. The anal complex can also be seen although usually only in late gestation (Fig. 1.2).

1.2 Esophageal Atresia

As the esophagus is poorly and anyhow incompletely visualized with fetal sonography, most cases of atresia escape antenatal detection [4]. The majority of cases are associated with a tracheoesophageal fistula that allows distal transit of fluid and filling of the stomach. In
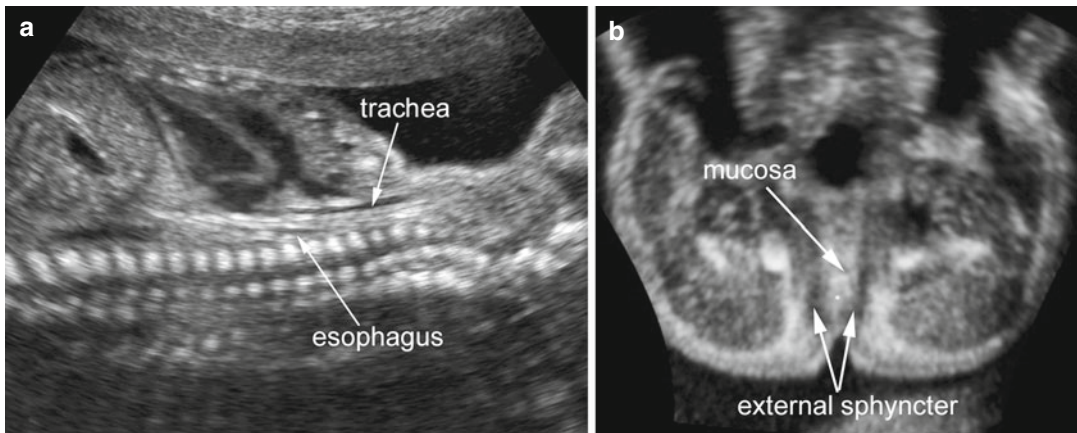
Fig. 1.2 It is difficult to demonstrate sonographically the fetal esophagus and the anorectal complex. When the fetus is in a favorable position and the quality of the images is adequate, at least the distal esophagus coursing posterior to the trachea (**a**), and the echogenic anorectal mucosa, surrounded by the sonolucent external sphincter (**b**) may be seen

late gestation, however, the size of the fistula does not allow adequate transit and as a consequence of this fluid accumulated into the amniotic cavity, and the stomach appears minimally distended.

The diagnosis of esophageal atresia is suspected when, in the presence of polyhydramnios (usually only in the third trimester), repeated ultrasonographic examinations demonstrate a small stomach bubble. In most cases the condition can only be suspected and the final diagnosis is only possible after birth. The only exception is in cases in which during swallowing the dilated proximal esophageal pouch is seen, as an elongated upper mediastinal and retrocardiac anechoic structure. This finding however is present only after 28 weeks and transiently [4].

The differential diagnosis for the combination of a small stomach bubble and polyhydramnios includes intrathoracic compression, by conditions such as diaphragmatic hernia, and muscular-skeletal anomalies causing inability of the fetus to swallow. Fetal magnetic resonance has also been reported to be of help in these cases [4].

In one of the largest available series, polyhydramnios was present in 50 % of cases, and the atresia was suspected or diagnosed antenatally in about one-third of cases, at a median gestational age of 31 weeks. As expected type 1 atresia was more frequently suspected (polyhydramnios in 100 % of cases, small gastric bubble in over 80 % of cases) than cases with a tracheal fistula (polyhydramnios 50 % of cases, small gastric bubble in 25 %) [4].

Esophageal atresia and tracheoesophageal fistula are often associated with other major defects, including chromosomal anomalies, malformations, and syndromic associations, that are not always obvious on prenatal examinations (Fig. 1.3).

1.3 Duodenal Atresia

Prenatal diagnosis is based on the demonstration of the characteristic "double bubble" appearance of the dilated stomach and proximal duodenum, commonly associated with polyhydramnios. 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 sufficient volume of amniotic fluid for bowel dilatation to occur before the end of the second trimester of pregnancy.

In a review of the literature, prenatal diagnosis was made in 77 % of cases. Other malformations were often present and trisomy 21 was found in about one-third of cases [5] (Fig. 1.4).
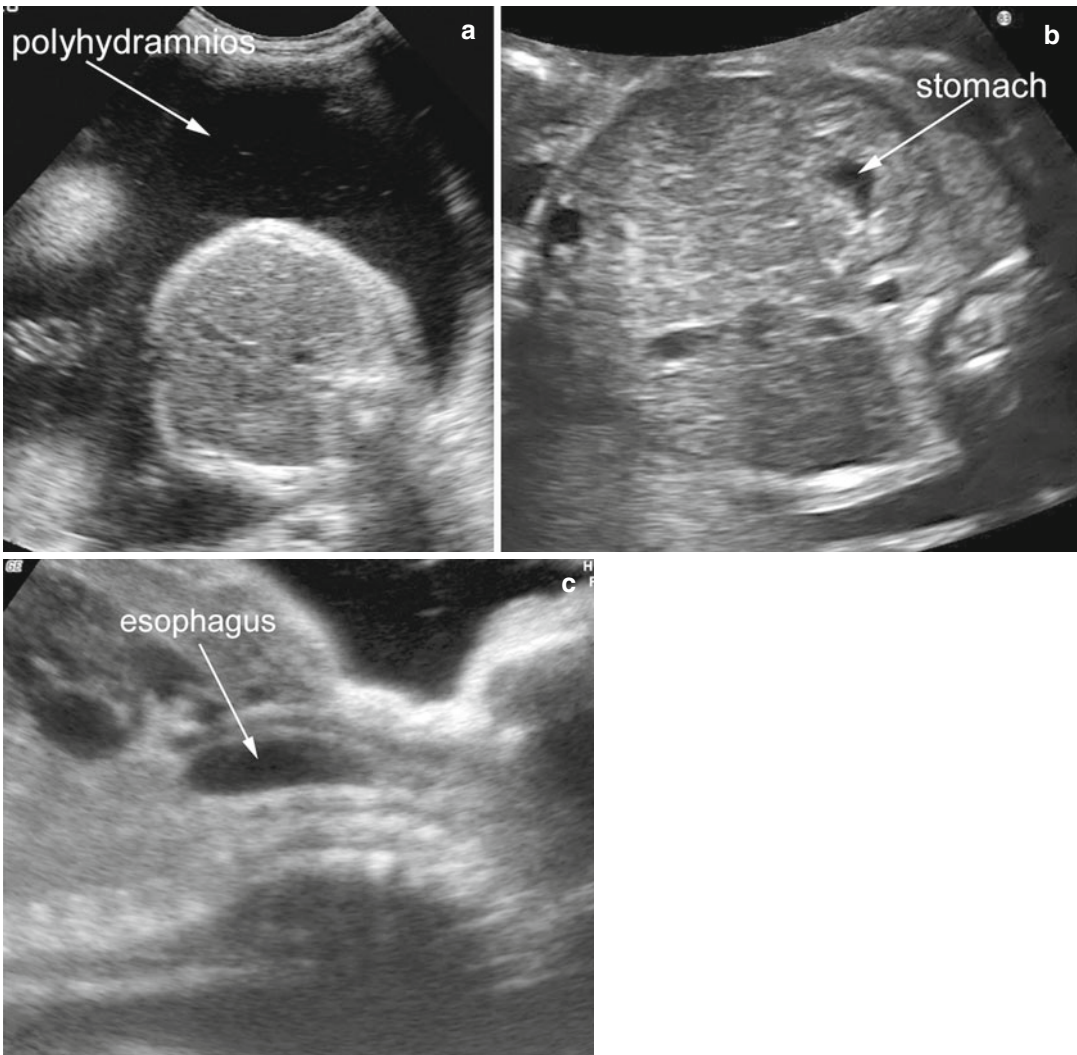
Fig. 1.3 Esophageal atresia. The amniotic fluid is much increased (**a**), the stomach bubble is small (**b**), and the proximal esophagus forms a pouch when the fetus swallows (**c**)

1.4 Pyloric Atresia

Few cases of pyloric atresia, often in association with other malformations, have been described. The typical finding includes polyhydramnios and a large stomach that usually appear only in the third trimester of gestation. The index of suspicion is increased when dilatation of the esophagus is also seen [5].

1.5 Intestinal Obstruction

In early gestation the bowel is virtually empty and it has a homogeneous echogenic texture. Only in the third trimester of gestation it becomes possible to visualize the large bowel, distended by echogenic meconium. The appearance is very variable and the size of the colon has been reported to vary between 7 and 20 mm. Individual
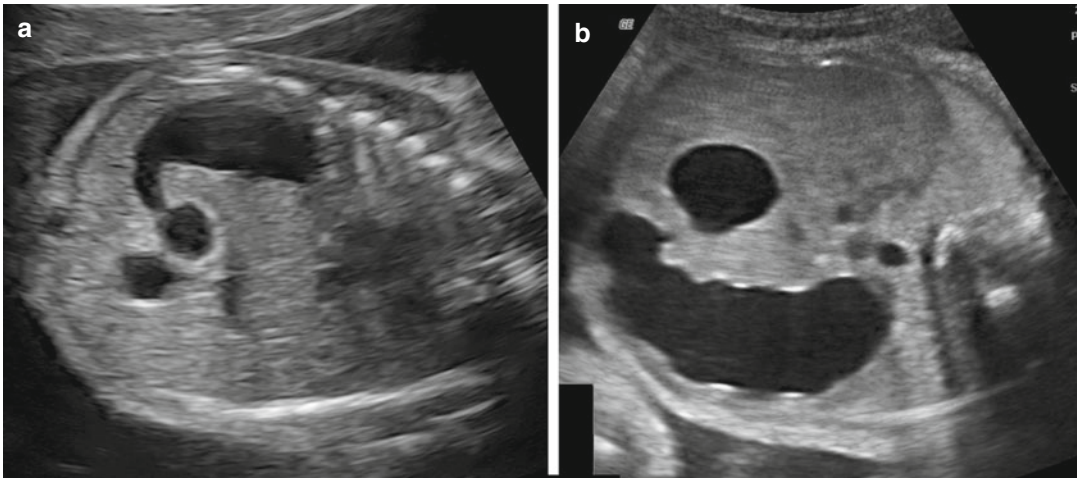
Fig. 1.4 Duodenal atresia; double bubble sign in the second (**a**) and third trimester (**b**)
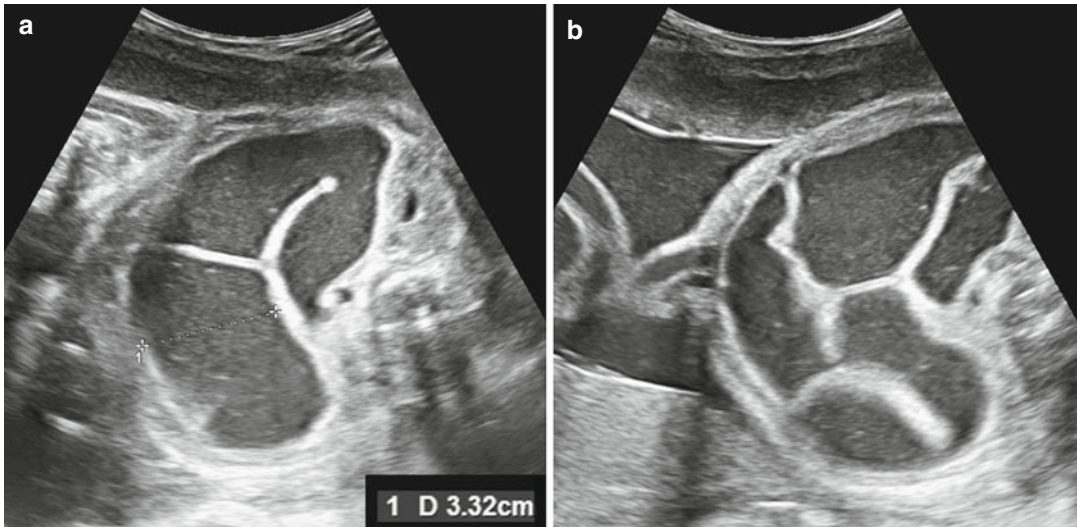

Fig. 1.5 Ileal atresia; gross dilatation of bowel loops. During real-time examination, increased perystalsis (**a**, **b**) was seen.

small bowel loops may be seen at times, usually with a maximum diameter of few mm.

Small bowel obstruction is usually visible only late in pregnancy and almost invariably after 25 weeks of gestation. The diagnosis is easy when distended, fluid-filled, and peristaltic bowel loops are seen. The presence of polyhydramnios increases the index of suspicion. However, at times it may be difficult to differentiate bowel obstruction from normally prominent large bowel. It is not rare that the final diagnosis can only be made after birth. The whirlpool sign suggesting the presence of a volvulus has been described in fetuses.

In a review of the literature, about 50 % of small bowel obstruction were detected antenatally, and false-positive diagnosis was frequent (the specificity was as low as 30 %). The detection rate was greater with jejunal than ileal obstruction (60 % and 25 %, respectively). Cystic fibrosis has been reported in up to 13 % of cases [5] (Fig. 1.5).

1.6   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. In many cases of meconium peritonitis, ultrasound will only reveal findings of intestinal obstruction. However, a specific diagnosis can be made if dilated bowel loops are found in association with ascites and calcium deposit. It may also be possible to document at times a typical sequence of events: ascites associated with segmental dilatation of bowel loops, followed by disappearance of the fluid and progression of bowel enlargement, usually in the presence of intra-abdominal calcifications. Cystic fibrosis has been reported in up to 20 % of these cases [5].

1.7   Abdominal Cysts

Abdominal cysts 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 [6].

1.7.1   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 differentiate the condition from bowel disorders.

1.7.2   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 (hCG 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 the 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 on 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 few cases are genetic, such as the autosomal recessive McKusick-Kaufman syndrome with hydrometrocolpos, polydactyly, and congenital heart disease (Fig. 1.6).

1.7.3   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 in the midline, cystic lesion of variable size; a solid appearance may be secondary to hemorrhage.
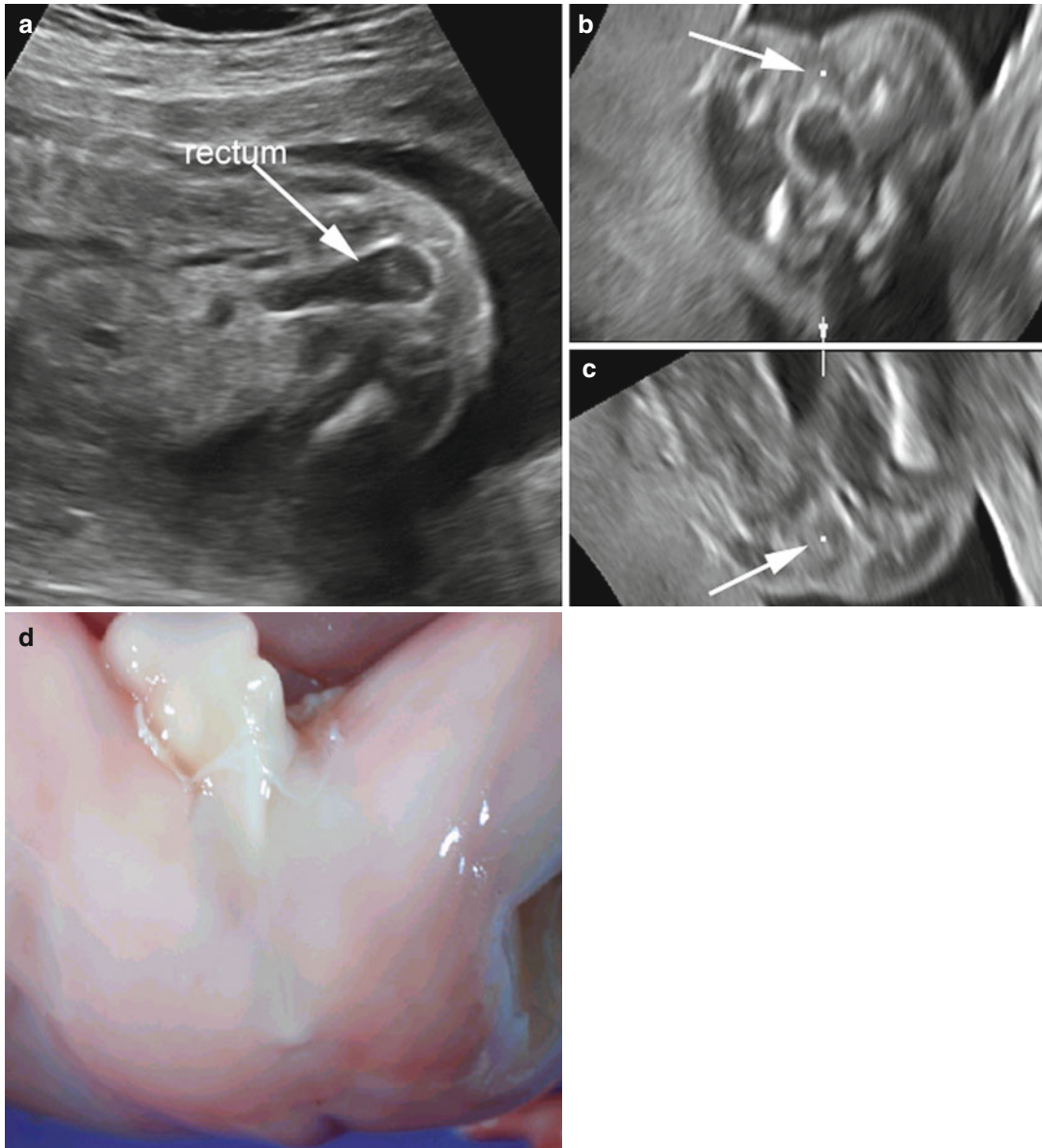
Fig. 1.6 Imperforate anus. In this fetus at 20 weeks of gestation with multiple anomalies, the diagnosis was suggested by the enlargement of the rectum (**a**) in association with the failure to demonstrate the echogenic anal mucosa (**b**, **c**); the diagnosis was confirmed at the time of autopsy (**d**)

1.7.4 Intestinal Duplication Cysts

They 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 broncho-genic cysts, adenomatoid cystic malformation of the lung, and pulmonary sequestration. The thickness of the muscular wall of the cysts and

the presence of peristalsis may facilitate the diagnosis. Postnatally surgical removal is carried out.

1.8 Anorectal Malformation

The fetal anus and rectum are poorly demonstrated by ultrasound and are not a part of the standard examination of fetal anatomy. As a general rule, anorectal malformations are not amenable to antenatal diagnosis. In a handful of cases, however, anorectal atresia has been suspected antenatally, usually by the observation of a dilatation of the upper rectum and failure to demonstrate the anorectal complex that is normally formed by the anechoic ring of the external sphincter muscle surrounding the echogenic mucosa (Fig. 1.6) [7].

1.9 Obstetrical Management of Fetal Intestinal Anomalies

Intestinal anomalies are invariably treated after birth, and standard obstetric management is usually not changed. As a general rule, delivery may occur at the term and vaginally. A possible exception is represented by the presence of polyhydramnios that is frequently found with esophageal and duodenal atresia, less frequently with small bowel obstruction. An excessive amount of amniotic fluid may result in overdistension of the uterus and trigger premature labor or rupture of the membranes. The administration of tocolytic drugs in these cases is of limited efficacy, and in the presence of severe polyhydramnios, most perinatologists would favor serial evacuative amniocenteses. When there is potential for prematurity, maternal administration of steroids should also be considered to prevent neonatal respiratory distress.

Of special concern is the issue of fetal pain. It is difficult for perinatologists to observe with ultrasound fetuses with obstructed bowel that demonstrate gross intestinal dilatation with intense peristalsis and overdistension of the abdomen and vomit incessantly and not to think the agony they are experiencing. Little is known about fetal pain, and the ultimate psychological consequence of long-enduring distress, but studies on premature infants do suggest that this may have a major impact [8]. Unfortunately, an effective approach to pain therapy in utero is not available yet, but certainly this problem will have to be addressed in the future.

Conclusions

In most cases, intestinal anomalies will escape prenatal detection and will be recognized only postnatally. The pediatric surgeon may however be consulted prior to birth in the presence of abnormal fetal sonographic findings. At times the diagnosis will be clear-cut (double bubble sign and polyhydramnios indicating duodenal atresia, grossly dilated and peristaltic bowel loops suggesting small bowel obstruction, a whirlpool sign suggesting a volvulus). In other cases, there will be many uncertainties. It is important to stress that the accuracy of antenatal imaging technique in the identification and differentiation of gastrointestinal anomalies is limited. In many cases, a definitive diagnosis will only be possible after birth.

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