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Congenital Abdominal Anomalies

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Abstract

Introduction: Abdominal anomalies that appear during intrauterine life are complex due to many organs that are affected. In cases, the ultrasound appearance is a cystic image with different content and the differential diagnosis is often difficult. **Body—research methods:** the organs affected by abdominal congenital anomalies involve the gastrointestinal tract (stomach, duodenum, small bowel or colon, and gall bladder), the kidney and urinary tract, the peritoneal cavity (ascites), suprarenal glands, and tumors of the reproductive system (especially the ovaries). In order to identify the affected structures, it is mandatory to know the normal aspect of the abdominal content at different gestational ages. The diagnosis may be very difficult, but its accuracy is important, considering the need of further counseling the couple. In minor conditions, without chromosomal anomalies or associations, the outcome is usually good, and there are even possibilities of in utero treatment. In severe conditions, with poor outcome, the couple can choose to terminate the pregnancy, after counseling is provided. **Conclusion:** abdominal congenital anomalies are common findings in ultrasound screenings for anomalies in all the trimesters of pregnancy and their recognition is important for subsequent management.

Keywords: gastrointestinal anomalies, cystic anomalies, congenital anomalies of the kidney and urinary tract

1. Introduction

The chapter will be structured on four main sections: the normal ultrasonographic appearance of the fetal abdominal cavity and abdominal wall, the absence of the normal abdominal structures, abnormal structures present in the abdomen (cysts, tumors, etc.), and abdominal wall anomalies.

Abdominal anomalies that appear during intrauterine life are complex due to the number of different organs that are affected. In most cases, the ultrasound appearance is of a cystic image with different content, and the differential diagnosis is often difficult.

2. Congenital anomalies overview

Congenital anomalies of the abdominal contents involve organs of the gastrointestinal tract, kidneys and urinary tract, suprarenal glands, and genital organs (mainly the ovaries).

Anomalies in the fetal abdomen are diagnosed either by the lack of visualization of normal structures or evidence of abnormal images during the anomalies screening ultrasound examination. Therefore, obtaining and documenting standard normal images of the abdomen is of crucial importance.

This chapter is structured in 4 subsections:

2.1. Normal aspect of the fetal abdomen.

2.2. Anomalies consisting of absent structures.

2.3. Abnormal structures present in the abdomen: cystic or echoic masses.

2.4. Abdominal wall anomalies.

2.1. Normal ultrasound images of the abdomen

At the midtrimester routine ultrasound scan, there are a couple of images that must be obtained according to ISUOG guidelines [1], images that can rule out most of the abdominal pathological conditions.

First step of the examination should be visceral situs assessment, by demonstrating the position of the stomach, liver veins, abdominal aorta, and superior cava vein as seen in **Figures 1** and **2**.

The abdominal transverse section at the umbilical vein–portal sinus is the level for abdominal circumference measurement and a standard image to identify some important structures:

- fetal stomach—an anechoic structure of bean-like form. It can be seen from 9 weeks, but at 14 weeks, the different parts of the stomach can be seen and gastric peristalsis can be detected



Figure 1. Transverse section of the fetal abdomen—the structures that can be seen are: stomach (s), umbilical vein (uv), liver (l), and suprarenal gland (sr).

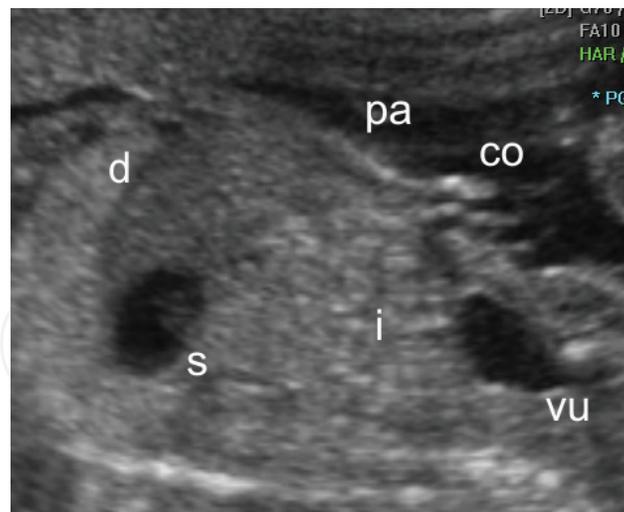


Figure 2. Sagittal section of the fetal abdomen—the continuity of the anterior abdominal wall is obvious (pa), the cord insertion (co), stomach (s), urinary bladder (vu), diaphragm (d), and bowel (i).

after 16 weeks of gestation. Nomograms of stomach size have been imagined at different gestational age [2]. Hyperechoic mass of uncertain origin may be seen inside the stomach and may disappear at the following examinations without pathological significance [3].

- fetal liver is an hypoechogenic structure that occupies the section on the right of the stomach.
- gall bladder is an ovoid, hypoechoic structure on the right and under the intrahepatic part of the umbilical vein.
- the umbilical vein has a caudal trajectory, penetrates the liver and connects with the left portal vein. Actually, the umbilical vein is the left portal vein, because the right portal vein regresses at 6–7 weeks.
- the anterior abdominal—skin, subcutaneous cellular tissue, and muscles.

The sagittal section at the abdominal level is essential for assessing the integrity of the anterior abdominal wall, because the insertion of the umbilical cord and the wall above and below the insertion can be visualized. Differentiating between the internal structures of the abdomen can be difficult, because the liver and intestinal loops have similar echogenicity. Between 8 and 10 weeks of gestation, the physiological gut herniation can be visualized by ultrasound, as a hyperechoic mass at the base of the umbilical cord. Reintegration occurs between 10 and 12 weeks and it is complete at approximately 11 weeks and 5 days.

At a lower transverse section of the abdomen, the kidneys, small bowel, and colon can be seen as the following:

- the kidneys are located on both sides of the spine; their adult similar shape can be recognized from 12 weeks but the medular/cortical differentiation is obvious starting from 15 weeks of gestation. The fluid filled structure of the calyces is a useful landmark to identify the kidneys. Corticomedullary differentiation (CMD) is determined according to echogenicity of the cortex and the medulla, considering that during the prenatal period, the

normal renal cortex is as echogenic as the liver or spleen and that the normal renal medulla is relatively hypoechogenic, which leads to a well-defined CMD [4].

- the small bowel is visible as a uniform, echogenic mass that can be differentiated from the colon by its central layout, aspect, and peristaltic changes, until the third trimester when its aspect is of round anechoic image with peristalsis (large intestinal loops filled with meconium) [5].
- the colon appears as a tubular structure located at the periphery of the abdomen, meanwhile the small intestine is centrally placed. The haustration of the wall serves to differentiate from other anechoic or hypoechoic structures. Meconium production begins at 16 weeks of pregnancy, giving the ultrasound-like appearance of the intestinal wall.

The echogenicity of the colon is established in comparison to other structures such as the liver or bladder:

- grade 0: the abdomen is uniformly homogeneous and the colon cannot be visualized.
- grade 1: the colon is hypoechogenic, isoechoic with the stomach and the bladder, haustrations being present.
- grade 2: the colon's echogenicity is higher than the bladder, but lower than the liver and occurs after 29 weeks.
- grade 3: echogenicity is similar to the liver's and occurs after 34 weeks.

Coronal or sagittal section of the fetal abdomen allows the visualization of the urinary bladder. It can be identified starting at 11–12 weeks of gestation as an anechoic mass with a thin wall, delineated by two umbilical arteries.

2.2. Absence of normal abdominal structures

The most common condition is the nonvisualization of the stomach, kidneys, or urinary bladder.

Nonvisualization of the gall bladder occurs in rare chromosomal diseases, mucoviscidosis, and in 20% of the fetuses with biliary atresia [6].

Absent stomach or small stomach can be associated with mechanical or functional absence of stomach filling:

- absent or reduced fetal swallowing: facial clefts, neuromuscular conditions, infections, SNC conditions, and fetal hydrops.
- mechanical obstruction or compression: esophageal atresia, diaphragmatic hernia, and thoracic tumors.
- reduced amniotic fluid production: oligohydramnios of any cause.

2.2.1. Esophageal atresia

There are five types of esophageal atresia, but the most common one after de Jong is the type with esophageal stump and trachea-esophageal connection below the interruption. In such cases, it is important to look after other indirect signs, like polyhydramnios and small for gestational age. Many of the anomalies are nonisolated [7]. Still, 44% of the anomalies are not diagnosed during the prenatal period [7].

Esophageal atresia is a condition defined by the lack of continuity between the proximal and the distal esophagus. The missing portion is the middle one and the main cause is poor blood flow. The anomaly appears during the 8th week of pregnancy when the primitive intestine fails to divide forming the trachea anteriorly and esophagus posteriorly. According to de Jong, in 90% of cases, esotracheal fistulae are associated. There are five types of esophageal atresia considering the anatomical location of the defect and the existence of the tracheoesophageal fistula [7]:

- type A: without fistula
- type B: proximal fistula
- type C: distal fistula (88% of cases)
- type D: double fistula, proximal, and distal
- type E: existing fistula without concomitant atresia

The incidence of this anomaly is 1:3500 live born and usually is nonisolated, being associated with CNS anomalies, VACTERL association, T21, T18, diaphragmatic hernia, and abdominal wall anomalies. [7]. Twenty percent of cases of the associations are with chromosomal anomalies (trisomy 13, 18, 21) [8] and cardiac defects may be present in 50% of cases. VACTERL syndrome is comprised of vertebral defects, ventricular septal defect, anal atresia, esotracheal fistula, renal anomalies, radial aplasia, and single umbilical artery [9].

Ultrasound diagnosis of esophageal atresia can be missed in pregnancy according to Brandberg and 44% of the cases cannot be diagnosed.

The prenatal diagnosis of esophageal atresia is difficult due to the absence of specific signs. The rate of detection varies between 8 and 24% [10, 11], and the lack of detection is due in 85% of cases to the tracheoesophageal fistulae association.

Prenatal esophageal atresia must be suspected if polyhydramnios is present after the 25th week of gestation and if the gastric pouch is absent. Sometimes, gastric secretions are sufficient to distend the gastric pouch, and make the diagnosis more difficult; if an esotracheal fistula is present, the gastric pouch may appear normal. Sometimes, the distal dilated portion of the esophagus may be observed as a hypoechoic area in the mediastinum, behind the heart (blind pouch aspect). Although the sign has a great diagnostic value, it is rarely present before 25 weeks of gestation [8, 12, 13]. The detection rate is improved if an antenatal MRI is performed.

The differential ultrasound diagnosis can be made with other conditions with absent stomach, the most frequent are:

- facial cleft;
- congenital diaphragmatic hernia;
- neuromuscular conditions [9].

The risk of association with other anomalies, chromosomal and nonchromosomal, is high rising to 30–40% of cases: trisomy 21 and 18, VACTERL association. Perinatal approach implies karyotype analysis when the diagnosis is made. The birth should take place in a tertiary center with a neonatal intensive department considering the high risk of fetal hypotrophy (40% of cases) and prematurity (due to polyhydramnios). Surgical reconstruction of the esophagus is possible. The prognosis depends on the association with other congenital anomalies and on the gestational age at birth. After 32 weeks of gestation, if the prenatal diagnosis is achieved, reflux and aspiration pneumonia can be prevented and the survival rates are over 95%. Long time prognosis is affected by the high rate of postoperative complications [9]. The postoperative mortality is approximately 9%, with a intrauterine fetal mortality of 22%. Retrospective studies have shown a neonatal mortality of 75%.

2.2.2. Absence of the kidney in the renal fossa

It can be caused by renal agenesis or ectopic kidney. True isolated renal agenesis is confirmed by the absence of renal artery and is associated with oligoamnios if it is bilateral. Unilateral renal agenesis can be isolated but association as VACTERL is common [14]. Failing to visualize the urinary bladder can be caused more frequently by bilateral renal agenesis or exstrophy.

2.2.3. Renal agenesis

It is defined as bilateral absence of kidneys with an incidence variable from 1 to 3/10000 births and it is more frequent in male fetuses—**Figures 3 and 4**. Renal agenesis can be sporadic (due to teratogens or diabetes mellitus), isolated, or part of syndrome association, the most frequent found being:

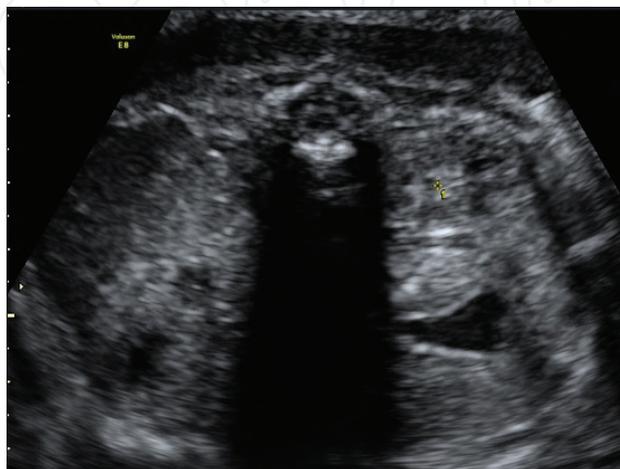


Figure 3. Renal agenesis—transverse abdominal section—no renal structure on the right side.

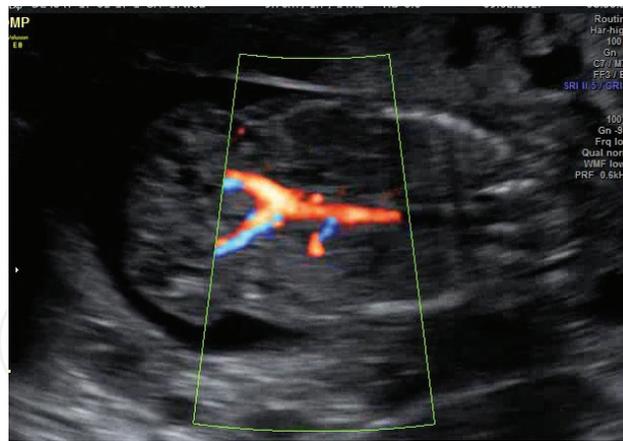


Figure 4. Unilateral renal agenesis—coronal section depicting only one renal artery.

- Cat’s eye syndrome, characterized by iris coloboma, anal atresia, preauricular tags, and renal anomalies.
- 4p-Syndrome is characterized by multiple heterotopies of cells (in adrenals, brain, pancreas, and skin), renal agenesis, cardiac, facial, and genital anomalies.
- Fraser syndrome with: cryptophthalmos, syndactyly, auditory canal atresia, cleft palate, renal agenesis, and genital anomalies.
- a very severe syndrome: cerebro-oculo-facio-skeletal syndrome, characterized by: microcephaly, microphthalmia, narrow palpebral fissures, high nasal bridge, large ears, micrognathia, kyphosis, scoliosis, flexion contraction of the extremities, and rocker-bottom foot.
- in some cases, Müllerian duct anomalies can be associated.

Oligohydramnios is a constant finding in bilateral renal agenesis, and it can be seen from 14 weeks of gestation. “Potter syndrome” associates the following abnormalities:

- pulmonary hypoplasia.
- typical face—low set ears, epicanthus, parrot-beak nose, and receding chin.
- abnormal position of the hand and foot and bowed legs, clubbed feet, hip dislocation.
- intrauterine growth retardation.
- absence of urinary bladder visualization.

Ultrasound diagnosis of bilateral renal agenesis is based on three main elements:

- oligohydramnios.
- bilateral absence of the fetal kidney (sometimes the adrenal glands can be confused with the kidneys therefore the visualization of the normal cortical and medular structures is useful for exclusion).
- absence of the urinary bladder. If the urinary bladder is present the bilateral adrenal agenesis is excluded.

The differential diagnosis should include other anomalies of the kidney, associated with oligohydramnios as polycystic kidney disease, multicystic kidneys, etc.

When bilateral renal agenesis is suspected, a detailed examination of the fetal anatomy should be performed considering the high risk of other anomalies association [15]:

- Cardiovascular malformations (14%): tetralogy of Falloch, ventricular septal defect, atrial septal defect, hypoplastic left ventricle, coarctation of the aorta, dextrocardia, single ventricle, transposition of the great vessels, total anomalous pulmonary venous drainage, tricuspid atresia, and hypoplastic aorta.
- Musculoskeletal malformations (40%): absent radius and fibula, digital anomalies, lumbar hemivertebrae, cleft palate, sacral agenesis, and diaphragmatic hernia.
- Central nervous system malformations (11%) include hydrocephaly, microcephaly, meningocele, cephaloceles, holoprosencephaly, and iniencephaly.
- Gastrointestinal malformations (19%): duodenal atresia, imperforate anus, esotracheal fistula, intestinal malrotations, absent stomach or gallbladder, and omphalocele.
- Single umbilical artery.

The prognosis is usually lethal due to the association of pulmonary hypoplasia and growth retardation, the fetuses die in utero or in the first days of life.

2.2.4. Bladder exstrophy

Bladder exstrophy as well as cloacal exstrophy arise from the abnormal growth of the caudal fold, resulting in a anterior abdominal wall defect. The absence of the anterior abdominal wall and the anterior bladder wall will expose the posterior bladder wall to the amniotic fluid. Small defects result in epispadias, but a larger one might expose the posterior bladder wall. The incidence of bladder exstrophy is 1:30,000 births, with male fetuses being more affected [16]. These anomalies are frequently sporadic, although familial transmissions have been reported [16].

Positive and differential ultrasonographic diagnosis of bladder exstrophy should be suspected whenever the bladder cannot be visualized, even though the amniotic fluid volume is normal (the cycle of the bladder filling is 15 min)—**Figure 5**. Others ultrasound signs useful are:

- a solid hyperechoic mass that comes in close contact with the umbilical arteries.
- the absence of the bladder within the fetal pelvis.
- normal volume of the amniotic fluid.
- genital ambiguity (duplication and division of the penis).

Bladder exstrophy must be differentiated from gastroschisis and omphalocele. In those conditions, despite the abdominal wall defects, the bladder is still present within the fetal pelvis [17]. The risk of association with chromosomal and nonchromosomal syndromes is low. Perinatal counseling depends on the association with other anomalies. If the prenatal diagnosis of bladder exstrophy is made, therapeutic abortion must be offered as an option.



Figure 5. Bladder exstrophy—no urinary bladder can be seen.

The prognosis depends on the grade of urinary incontinence and the associated genital anomalies [17] with a survival rate of 80% in the cases where surgery interventions are needed to reconstruct the bladder wall and the genitalia. Although, it has been suggested that in the case of genital ambiguity, the sex should be deemed female, later follow-up has shown that the patients, male or female, can lead a normal life with a normal IQ and in some cases, corrective and fertility surgery can improve the quality of life.

2.3. Abnormal structures present in the abdomen: cystic or hypoechoic masses

The presence of cystic lesions in the fetal abdomen can have many underlying conditions. The diagnosis can be made upon their appearance: unique or multiple, the situs of the abnormal structure, and its relations with the abdominal viscera, the aspect of the wall and content. In most cases, the correct prenatal diagnosis of these structures is difficult to make and their origin can be determined only after birth.

Single cystic masses can be:

- megacystis,
- ovarian cysts,
- intestinal cysts (mesenteric or duplication intestinal cysts),
- choledocal cysts,
- hepatic cysts,
- kidney solitary cyst.

Multiple cystic lesions can be more frequent:

- double bubble image in duodenal atresia,
- bowel dilatations in obstruction conditions,
- multiple intestinal cysts,

- bowel volvulus,
- polycystic kidney,
- hydronephrosis,
- dilated megaureter in uretero-pelvic junction obstruction,

Other complex aspects involving hypo and hyperechoic structures can be found in:

- ascites,
- meconium peritonitis,
- liver calcification.

The differential diagnosis of the cystic abdominal proliferations includes:

- mesenteric cysts,
- omental cysts,
- choledochal cysts,
- renal cysts,
- intestinal obstruction,
- intestinal duplication,
- meconium peritonitis-related cysts,
- anterior meningomyelocele,
- ovarian cysts,
- hydrometrocolpos,
- megacystis,
- the persistence of the cloaca.

2.3.1. Duodenal atresia

It is defined as the obstruction of the bowel produced by a degenerated middle segment of the duodenum. It can be also extrinsic due to a malformed annular pancreas or fibrous peritoneal bands that cause compression. The most frequent affected segments are the 2nd and the 3rd segment of the duodenum. With an incidence is 1:10,000 births, it is the second main cause of small bowel obstruction [18].

The diagnosis is suspected in the presence of polyhydramnios and it is based on the “double bubble” sign (**Figure 6**) formed by the stomach and the proximal part of the duodenum, both appearing dilated. This sign may be present from 19 weeks, but usually it is seen in the second part of the pregnancy or even later due to the fetal capacity of swallowing small amounts



Figure 6. Duodenal atresia—double bubble sign.

of liquid and delaying the dilatation of the intestine. Sometimes, the only consistent sign at repeated scans is a dilated stomach. Differential ultrasonographic diagnosis should be made with other cystic structures present in the upper or inferior abdomen:

- persistent right umbilical vein or varicose veins pertaining to the umbilical vein.
- choledocal cyst.
- hepatic cyst.
- intestinal duplication cyst.
- ovarian cysts.
- kidney cyst or multicystic dysplastic kidney.
- mesenteric cysts.
- urachal cyst.
- ureterocele.
- pyelectasis.

Duodenal atresia has a high risk (62%) of association with chromosomal anomalies and associated anomalies mainly with Down syndrome. Duodenal atresia is a sporadic anomaly, but it has been demonstrated that it can be transmitted in an autosomal recessive manner. Almost half of the fetuses with duodenal atresia manifest other anomalies: skeletal defects (vertebral and rib abnormalities, sacral agenesis, esotracheal fistula, intestinal malrotation, Meckel's diverticulum, and anorectal atresia), renal and cardiac defects. Because of the close vicinity with the Vater Ampulla, in 1% of cases biliary duct anomalies are present. Gall bladder atresia is impossible to diagnose before birth, but it can be suspected, if duodenal atresia is present and the gall bladder cannot be visualized [18].

The prognosis of isolated duodenal atresia is favorable, the rate of survival after corrective surgery is over 95%. The incidence of early postoperative mortality is between 3 and 5%, and the long term postoperative mortality rate is 6%. Considering the high risk of chromosomal anomalies association, the karyotype analysis is mandatory whenever duodenal atresia diagnosis is made prenatally, and also a careful examination of the fetus anatomy. Polyhydramnios can cause preterm birth and although vaginal delivery is allowed, it must take place in a tertiary unit to assure proper care for the neonate.

2.3.2. Intestinal obstruction

Intestinal obstruction is defined by the total or partial intestinal obstruction, which can occur in an intrinsic or extrinsic manner. The intrinsic lesions are caused by intestine stenosis or atresia. In the case of atresia, the two intestinal segments can be completely separated or tied by a fibrous band. In the case of intestinal stenosis, the two segments can be separated by a septum or a central diaphragm or the intestinal lumen is very narrow. “Apple-peel” atresia is characterized by the absence of an important intestinal segment that can include the distal duodenum, the entire jejunum, or the proximal ileum—**Figure 7**. Conditions that can produce extrinsic obstruction are most frequently represented by:

- intestinal malrotations with volvulus,
- peritoneal adhesences,
- meconium ileus.
- Hirschsprung’s disease.

The intestinal areas affected in the order of frequency are: the distal ileum (35%), the proximal jejunum (30%), distal jejunum (20%), proximal ileum (15%), or it can be multicentric in approximately 5% of cases. Anorectal atresia results from an abnormal cloacal division in the 9 weeks of gestation [5]. The incidence of intestinal obstruction is 1:2000 births. In half of the cases, obstruction of the small intestine is present, and the other half is made up by anorectal atresia.



Figure 7. Duodenal stenosis—in utero image.

Intestinal obstruction is usually sporadic, but it can have a genetic transmission when the affected areas are multiple. Anorectal atresia is associated in 80% of cases with vertebral defects, genitourinary, cardiac and other gastrointestinal abnormalities. The association with chromosomal or other anomalies is rare.

Ultrasound diagnosis is usually possible beyond 25 weeks of gestation, due to the slowly dilatation progression. The size of the intestinal lumen does not exceed 2–7 mm. The jejunum and ileum obstructions aspect is of dilations of the intestinal loops with active peristalsis. The fetal abdomen is fully distended and the majority of cases also present polyhydramnios, especially in the case of proximal obstructions. Similar images of the fetal abdomen can be found in Hirschsprung's disease. Transitory ascites or meconium peritonitis can appear when intestinal perforations occur [5]. When intestinal obstruction is diagnosed, the work-up to exclude other intestinal tract anomalies, ovarian cysts, mesenteric cysts, or intestinal duplication cysts is mandatory. Anorectal atresia is difficult to diagnose prenatally due to the great distension of the proximal bowel and the normal amount of amniotic fluid. Occasionally, calcification of the fetal meconium may be present within the fetal pelvis.

The prognosis depends on the type and location of the obstructed site and on the association of other anomalies. Considering that polyhydramnios that can occur, preterm births are common and the birth should take place in a tertiary unit. After birth, the affected babies develop emesis and abdominal distention, and can require immediate surgery repair. For isolated obstructions, requiring small resections of the intestine, the survival rates are over 95%. The resection of a large intestinal segment can lead to a lethal syndrome—short gut syndrome.

2.3.3. *Hirschsprung's disease*

It is caused by the congenital absence of intramural parasympathetic ganglia of the colon. The deficiency is caused by the lack in neuroblast migration from the neural crest, which occurs between the 6th and the 12th week of development. Also, the degeneration of the already migrated neuroblasts, which can occur before or after birth, may be involved. The incidence is approximately 1:3000 births. This disease is usually sporadic, but in 5% of cases there is a familial transmission. In a small number of cases, Hirschsprung's disease is associated with Down syndrome.

Ultrasound diagnosis can be suspected in the presence of the dilatation of the intestinal lumen produced by abnormal peristaltic movements. At the ultrasound scan, images are similar to those found in anorectal atresia, in which the colon is also affected. Similarly, when the small intestine loops are affected, the images are of an obstruction with dilated intestinal loops and polyhydramnios. The prognosis depends on the postnatal postoperative evolution, which aims to resect the affected intestinal segment, and can include a temporary colostoma with a neonatal mortality rate of 20%. Considering the prematurity risk due to associated polyhydramnios and the need for postnatal surgery, the birth must take place in a tertiary unit, but cesarean birth is not mandatory.

2.3.4. *Choledochal cysts*

They are defined as cystic dilatations of the common bile duct; they are rare and are usually of unknown etiology. The diagnosis is made in the presence of a transonic image located in

the upper right abdominal quadrant. The differential diagnosis includes intestinal duplication cysts, hepatic cysts, situs inversus, and duodenal atresia. The absence of polyhydramnios and the presence of intestinal peristalsis are useful for excluding intestinal obstruction. Common complications that intervene after birth are: biliary cirrhosis, hypertension, lithiasis, and rarely — adenocarcinoma. The complications can be prevented by early postnatal diagnosis and surgical resection. Surgical mortality rate is about 10% [19].

2.3.5. Mesenteric and omentum cysts

They are usually an accumulation of lymphatic fluid produced by lymphatic hamartomas or blockage of the lymphatic drainage. Ultrasound diagnosis can be suspected in the presence of a hypoechoic lesion located usually on the midline. The cysts can vary in size, their content can be unilocular or multilocular, or filled with echoic masses with solid content caused by secondary hemorrhage. The cysts can be filled with serous, hemorrhagic, or chylous fluid.

The prognosis of the cyst depends on the possible complications: intestinal obstruction, torsion, and hemorrhage. Malignant transformation of the cysts is rare. Prenatally, the cysts require drainage by puncture, only when they are of important size and can cause compression and secondary hydrops. After birth, the management is surgical but a complete resection of the cysts is difficult because of the close vicinity with large vessels and because of the 20% rate of recurrence.

2.3.6. Hepatic cysts

Hepatic cysts are a rare finding in the prenatal life. Usually, they are located in the right hepatic lobe and can produce obstruction of the intrahepatic biliary system. Their ultrasound aspect is of a unilocular anechoic round structure. Even after birth, they are asymptomatic and complications like infections and hemorrhage rarely occur. In approximately 30% of cases, hepatic cysts are associated with polycystic renal disease [19].

2.3.7. Duplication intestinal cysts

Duplication intestinal cysts are rare but located throughout the entire gastrointestinal tract. They appear in the form of tubular-like structures of variable sizes, and can occur singularly or associated with renal malformations, adenomatous cystic lung malformations, and lung sequestration. The thickening of the cystic wall and the peristaltic movements make the diagnostic process easier. The surgical excision of the cysts is the only therapeutic choice.

2.3.8. Ovarian cysts

Ovarian cysts are the most frequent fetal tumors and they are a relatively frequent finding during fetal autopsy (approximately 1/3 of fetuses). The cysts are usually asymptomatic and small. Ovarian fetal cysts are hormone-dependent (influenced by the human chorionic gonadotropin hormone, estrogens, and placenta hormones) and appear after 25 weeks of gestation [20].

Ovarian cysts (**Figure 8**) appear as anechoic round images located laterally to the urinary bladder. They are unilateral and unilocular and appear most frequently in isoimmunization-affected fetuses or those coming from pregnancies associated with diabetes, probably due to placental hyperplasia.

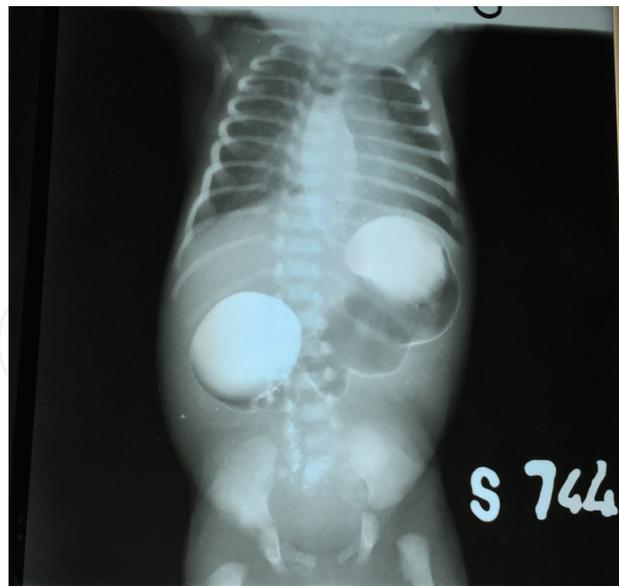


Figure 8. Duodenal stenosis—same case after birth.

The clear majority of the ovarian cysts is benign and resolves spontaneously after birth. The differential diagnosis has to be made with hydrocolpos, a condition that manifests itself as a hypoechoic or hyperechoic pelvic mass in a female fetus [20]. The prognosis is usually good but complications may appear like: torsion of the cysts, ascites, intracystic hemorrhage, rupture of the cysts, or cyst infarction. In such cases, the inner aspect of the cyst can change and speckles or hyperechoic structures inside the cyst can appear. When the cysts are large, intestinal compression could induce polyhydramnios. In such cases, cyst puncture and amniotic drainage should be considered. There is no need for cesarean section, but when the risk of premature birth is high due to polyhydramnios, birth should take place in a tertiary unit.

2.3.9. Congenital hydronephrosis

It is defined by a dilatation of the renal pelvis and calyceal system that exceeds the normal size according to the age of gestation (anteroposterior diameter of the renal pelvis over 4 mm at 20 weeks and over 7 mm at 32 weeks)—**Figures 9 and 10**. The most frequent cause is the ureteropelvic junction obstruction. When dilatation of the ureter occurs, the obstruction is lower usually at the vesicoureteral junction [21]. It is more common in males than in females, with a sex ratio of 5:1. Ureteropelvic junction obstruction is usually sporadic, but can also be inherited.

Anatomic causes that produce ureteropelvic junction obstruction are: fibrous adhesions, bands, kinks, ureteral valves, aberrant lower pole vessels, abnormal ureteral insertion, and unusual shapes of the pyeloureteral outlet. They can be associated with urinary anomalies and extra-urinary anomalies: vesicoureteral reflux, bilateral ureteral duplication, bilateral obstructed megaureter, contralateral nonfunctioning kidney, contralateral renal agenesis, Hirschsprung's disease, cardiovascular abnormalities, neural tube defects, sagittal synostosis, mandibular hypoplasia, esophageal atresia and distal fistula, imperforate anus, syndactyly, congenital hip dislocation, and adrenogenital syndrome.

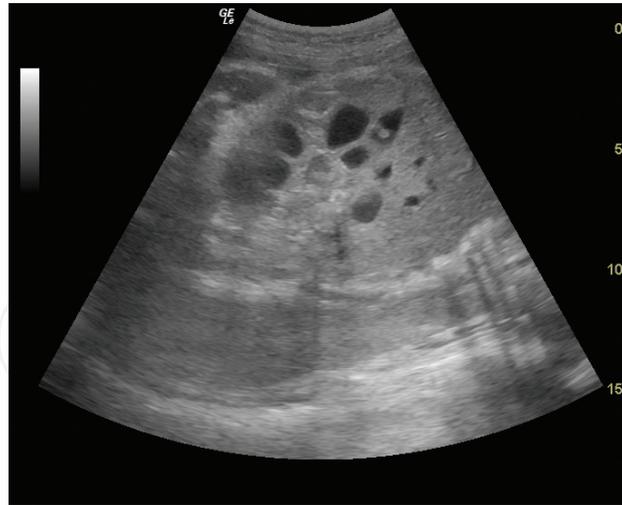


Figure 9. Intestinal obstruction—small bowel dilatations above the obstruction level.

Ultrasound diagnosis is based on the evidence of the dilated renal pelvis and on the measurement of the anteroposterior diameter of the dilatation on transverse abdominal section. Former criteria of hydronephrosis dilatation included also the ratio between the maximum transverse pelvic diameter and the renal diameter at the same level with ratios above 50% being the cut-off for hydronephrosis. Most UPJ obstructions are unilateral and bladder should fill normally. In case of severe oligohydramnios, association of unilateral hydronephrosis contralateral renal agenesis or dysplasia should be suspected. Differential diagnosis includes multicystic dysplastic kidneys, polycystic kidneys, and other condition associated with anechoic abdominal masses [22].

The prognosis of ureteropelvic obstruction is generally good, but requires serial ultrasound scans to evaluate the progress of the dilatation. In most cases, the surgery is not needed, but the postsurgery mortality is absent [23].

Concerning the prenatal management, there is no need for premature intervention and vaginal birth is preferred (**Figures 11 and 12**).

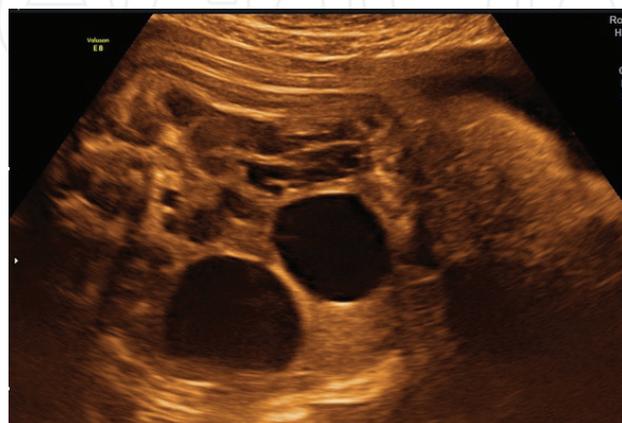


Figure 10. Ovarian cyst—two anechoic round structures in the fetal pelvis with no Doppler signal.

2.3.10. Multicystic kidney disease

It is a congenital disorder characterized by cystic lesions that correspond primarily to dilated collecting tubules. The disease can be bilateral, unilateral, or segmental and it is known also as Potter's type II syndrome, cystic kidney disease, and multicystic dysplastic kidneys.

It can be isolated or associated with urinary or extraurinary disorders such as: malrotations, ureteropelvic junction obstruction, horseshoe kidney, cardiovascular malformations, CNS abnormalities (anencephaly, hydrocephalus, iniencephaly, spina bifida, occipital meningocele), diaphragmatic hernia, cleft palate, microphthalmia, duodenal stenosis and imperforate anus, esophageal fistula, and bilateral absence of radius and thumb. Association with liver cysts or pancreatic cysts is not characteristic for Potter II syndrome [24].

Ultrasound diagnosis of multicystic kidneys (**Figure 11**) is made in the presence of an enlarged unilateral or bilateral kidney with multiple round anechoic structures peripheral located and variable in size. In some cases, the kidneys can be small but the renal sinus cannot be identified. Oligohydramnios is a common association, but in some cases, when the lesion is unilateral or the obstruction is incomplete, amniotic fluid may have normal volume.

Differential diagnosis includes infantile polycystic kidney disease, single cyst, ureteropelvic junction obstruction, Wilms tumor and hamartoma that has undergone necrosis, but can be difficult to diagnose prenatally.

The prognosis of bilateral disease is poor and termination of pregnancy can be offered. Unilateral isolated disease prognosis can be favorable, but delivery should take place in a tertiary unit (**Figures 13 and 14**).

2.3.11. Autosomal dominant polycystic kidney disease (ADPKD)

Adult polycystic kidney disease (APKD) is an autosomal dominant condition characterized by the presence of multiple cysts of variable size in the renal parenchyma. The cysts can replace the parenchyma and are produced by the dilatation of the collecting tubules and other tubular segments

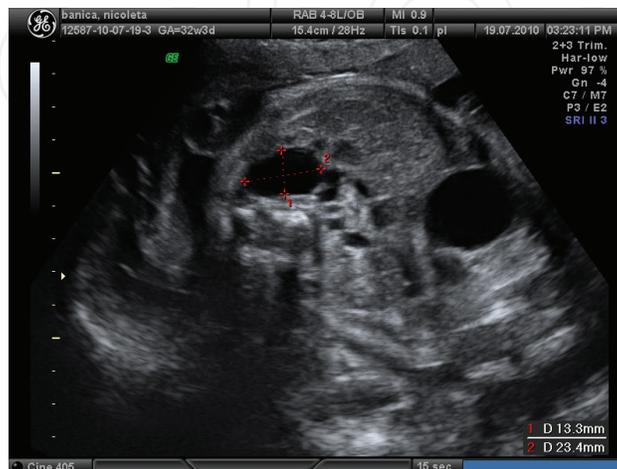


Figure 11. Hydronephrosis—significant dilatation of the renal pelvis and reduced thickness of the renal functional tissue.



Figure 12. Ureteral dilatation consecutive to uretero-vesical obstruction.

of the nephrons. Potter's type III polycystic is one of the entities responsible for ADPKD in which cysts of variable sizes (some up to several centimeters) coexists with normal renal structure. Being an autosomal dominant disease, the risk of transmission to the off springs of the altered gene on chromosome 16 is 1/2 pregnancies. APKD is one of the most common genetic disorders and the third most prevalent cause of chronic renal failure [25]. Epigenetic factors and penetrance of the gene determine the variability in symptoms, the disease manifesting late in the fourth decade. Intrauterine manifestation of the disease is not uncommon [26]. APKD is associated with cysts affecting other organs: liver, pancreas, spleen, lungs, testes, ovaries, and epididymis.

Prenatal diagnosis is difficult and often late due to the nonspecific appearance of the kidneys; only a few of these diagnoses can be recognized in the prenatal period, by family history, amniotic fluid volume, associated abnormalities, and genetic testing [4].

Ultrasound diagnosis of the APKD can be suspected in the presence of moderately enlarged hyperechogenic kidneys, with increased corticomedullary differentiation (CMD), in association with a normal amount of amniotic fluid. The diagnosis is usually late and should prompt investigation of both parents.



Figure 13. Multicystic kidney—multiple anechoic round structures occupying the whole renal area with no remaining normal renal structure.

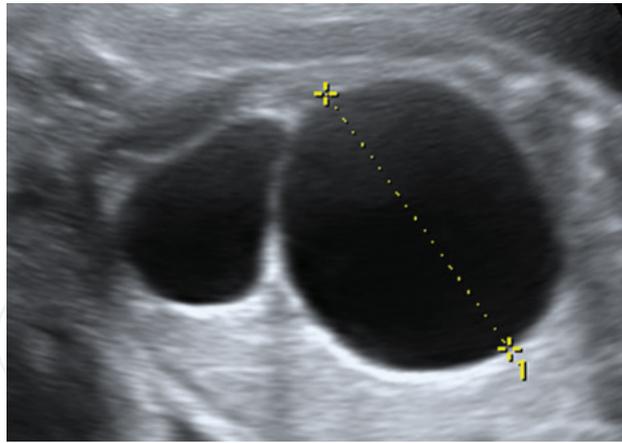


Figure 14. Giant kidney cyst.

The prognosis depends on other associated anomalies. The parents must be informed that the immediate outcome can be good, but the natural history of the disease includes the following: from completely asymptomatic to lower abdominal pain, renal enlargement, renal insufficiency, uremia and hypertension, with a mean age of onset of symptoms in the fourth decade of life.

Obstetrical management is based on the gestational age at diagnosis; in the first trimester, pregnancy termination can be offered. Family members should undergo ultrasound examination as well. The fetus must be thoroughly scanned to exclude other anomalies. Otherwise, the obstetrical attitude is not altered and vaginal birth is the first choice (**Figure 15**).

2.3.12. *Infantile polycystic kidney disease*

Infantile polycystic kidney disease (IPKD) is an autosomal recessive disorder characterized by bilateral and symmetrical enlargement of the kidneys. The disease is known also as polycystic kidney disease type I, infantile polycystic disease of the liver and kidney. Unlike the adult polycystic kidney disease, there is no renal parenchyma, only dilated collecting tubules [27]. As a recessive autosomal inherited condition, the risk of transmission is 25%. The disease is always bilateral. Other anomalies that can be associated: liver cysts, biliary duct hyperplasia, and dilatation of the biliary tree and portal hypertension.

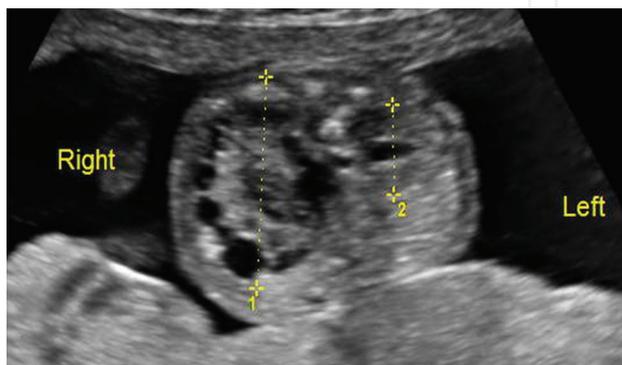


Figure 15. Autosomal polycystic kidney disease.

Ultrasound diagnosis is based on three elements:

- bilaterally enlarged kidneys of typical hyperechogenic texture.
- oligohydramnios.
- absent fetal bladder.

The disease can be diagnosed late in pregnancy or may progress gradually during pregnancy; therefore, repeated scans are mandatory.

The prognosis depends on the clinical variety of IPKD, which can vary from mild to severe with intrauterine death. After birth, the most important complications are respiratory, as a consequence of the pulmonary hypoplasia. Death later in life is the result of renal failure.

The management depends on the diagnosis age; if the diagnosis is made before viability, termination of pregnancy should be offered to the parents. Also in cases with severe oligohydramnios and absent bladder, the termination of pregnancy can be offered even in the third trimester.

2.3.13. *Megacystis*

It represents an enlarged urinary bladder usually due to a bladder outlet obstruction [lower urinary tract obstruction (LUTO)], which may appear also in nonobstructive conditions (megacystis-microcolon-intestinal hypoperistalsis syndrome). It can be detected from the first trimester scan in about 1 in 1500 pregnancies [28].

Prognosis, management, prediction of resolution, and postnatal outcome depend on the subsequent cause. Fetal lower urinary tract obstruction (LUTO) has an incidence of 2.2/10000 births and it is usually diagnosed during the late first or early second trimester of pregnancy [28, 29]. The most common condition in the LUTO spectrum includes urethral valves, urethral atresia, and urethral stenosis.

2.3.14. *Posterior urethral valves*

It is a condition that causes lower urinary tract obstruction due to a membrane-like structure in the posterior urethra of male fetuses. The disease is usually sporadic, and has a heterogeneous embryologic origin. Young type I syndrome valves seem to result from an exaggerated development of the urethrovaginal folds with an abnormal insertion of the distal end of the Wolffian ducts. Other valves, like in Young type III, develop because of abnormal canalization of the urogenital membrane [30]. Distention of the bladder (megacystis) leads to vesicoureteral reflux and hydronephrosis and can cause renal dysplasia [30].

Posterior urethral valves can be associated with other anomalies of the urinary tract (sequence) megacystis, megaureter, hydronephrosis, paraureteral diverticula, and dilatation of proximal urethra, cryptorchidism, and hypospadias. There are also extraurinary anomalies that can be associated: tracheal hypoplasia, patent ductus arteriosus, total anomalous pulmonary vein drainage, mitral stenosis, scoliosis, skeletal anomalies in lower extremities, and imperforate anus. The most frequent chromosomal abnormalities that can be associated are: trisomies 18 and 13, del 2q, and 69 XXY.

Ultrasound diagnosis can be made in the presence of megacystis, hydroureter, and hydro-nephrosis in a male infant. In female fetus, lower urinary tract obstruction includes agenesis of the urethra, megacystis-microcolon-intestinal hypoperistalsis syndrome, and variants of the caudal regression syndrome. When the rupture of the megacystis intervenes, urine can extravasate into the peritoneal cavity. Oligohydramnios can also occur.

The differential diagnosis is difficult in prenatal life, and includes ureteropelvic junction obstruction, ureterovesical junction obstruction, primary megaureter, and massive vesicoureteral reflux, absence of the urethra or detrusor hypertrophy.

Prognosis depends on the renal reserve, which can be difficult to assess prenatally. Another factor that can influence the outcome is the timing of the occurrence of urinary obstruction, and this is a critical factor: the earlier its appearance—the worse the prognosis. Complications include: pneumomediastinum and pneumothorax, related to pulmonary hypoplasia, associated congenital anomalies, renal failure, and surgical complications after decompressive surgery. Termination of pregnancy can be offered if the prognosis is poor and if other anomalies are associated. In utero vesicoamniotic shunts are possible, but the results at 2 years of life are not very encouraging [31].

2.3.15. *Meconium peritonitis*

Intestinal perforation in fetuses during pregnancy can lead to a sterile, chemical, and localized type of peritonitis. At the site of the perforation, a reactive process of calcification and fibrosis occurs in order to stop the progression. The etiology is in over 50% of cases due to intestinal stenosis, atresia, or meconium ileus. Other cited situations are volvulus or Meckel's diverticulitis. Meconium ileus is associated with cystic fibrosis and results from the blockage of the distal ileum with compacted meconium. The incidence is 1:3000 births.

The diagnosis is suggested by the dilatation of the intestinal loops or the presence of hyperechoic areas situated in the abdomen of the fetus. Over 80% of the fetuses that developed meconium peritonitis also have intra-abdominal calcifications. Ascites is an important sign of intestinal peritonitis. Other ultrasonographic signs suggesting meconium peritonitis include: polyhydramnios, meconial pseudocysts, thickening of the abdominal wall or pleural effusions.

The differential diagnosis of hyperechoic abdominal masses include: intra-amniotic hemorrhage, early ascites, fetal hypoxia, meconium peritonitis, and cystic fibrosis.

Meconium ileus and hyperechoic images of the intestinal loops at 16–18 weeks of gestation can be present in over 75% of cystic fibrosis affected fetuses. The incidence of cystic fibrosis in fetuses diagnosed before birth with intestinal obstruction is approximately 10%, consequently when other causes of intestinal blockage have been excluded, a genetic cystic fibrosis test is recommended.

The prognosis is poor in this case; approximately, 50% of fetuses suffering from meconium peritonitis pass away in the neonatal period.

2.3.16. *Liver calcifications*

Most of them are idiopathic, but can be associated with congenital infections and chromosomal anomalies. The incidence rate is 1:2000 fetuses, especially in the 3rd trimester. The

ultrasonographic image is of hyperechoic nodules situated in the liver parenchyma or liver capsule. The diagnosis is easy in the presence of solitary or multiple hyperechoic images (size between 1 and 2 mm) situated in the liver parenchyma or capsule. The prognosis depends on the association with infections or chromosomal anomalies. Isolated calcifications do not have pathological significance.

2.4. Abdominal wall anomalies

The evaluation of the integrity of the abdominal wall is part of routine midtrimester scan, although it can be performed starting with the first trimester examination. Conditions as: oligoamnios, placenta, or uterine wall proximity, excessive movement of the fetus can obstruct the proper visualization of the abdominal anterior wall. It is very important that the integrity of the abdominal wall is assessed by showing the insertion of the umbilical cord and visualization of the bladder in the pelvis, so that that bladder and cloacal exstrophy can be excluded.

The abdominal wall anomalies will be discussed separately in a subsequent chapter.

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