

# Ultrasound and Diagnosis of Fetal Anomalies

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## ABSTRACT

**Background:** Congenital abnormalities account 20 to 25% of perinatal deaths. Now, many genetic and other disorders can be diagnosed early in pregnancy. Screening examinations during pregnancy are an essential part of prenatal care. Among the various screening tests that are now offered to pregnant women, ultrasound (US) has the broadest diagnostic spectrum. There is no modality that can detect as many abnormalities during pregnancy as US. A priority goal in screening is the early detection of major fetal anomalies, which are defined as malformations that affect fetal viability and/or quality of life. During the past 10 years, some multicentric studies in Europe and USA show the successfulness of US diagnostics in detecting congenital abnormalities, even in women with low-risk pregnancy. The term sonoembryology designates the description of the embryonic anatomy, the normal anatomic relations and the development of abnormalities as visualized by ultrasound. To confirm the presence of normal anatomy or to make the diagnosis of an anomaly, we need knowledge of the normal embryonic development, including the appearance of the normal embryo.

**Definition of fetal anomalies:** Any deviation from the normal range during morphogenesis, constitutes an anomaly. Major anomalies are malformations that affect viability and/or the quality of life and require intervention, and minor anomalies are malformations that are definitely present, but are minimal and usually have no functional significance (e.g. ear tags).

Incidence data on major congenital anomalies vary considerably, depending on the type of detecting system used. The passive detection system reports 2 to 3% of newborns, meanwhile the active detection system, in which newborns are systematically examined by trained obstetricians, reports the incidence of congenital defects in 7.3% of all newborns.

**Etiology:** About 20% of anomalies in live-born infants are based on a defective gene, 10% are due to chromosomal abnormalities and 10% are mainly due to exogenous injury to the conceptus. Some 60% of all congenital anomalies are indeterminate or multifactorial causes (hereditary factors and environmental influences).

US can detect about 74% of major birth defects and possibly a higher number, when conducted by a well-trained specialist. We have demonstrated in this paper some fetal anomalies found by US during our practice in Tirana and Prizren. There are some anomalies of the CNS, neural tube defects, anomalies of the head, neck and spine, thoracic and cardiac anomalies, gastrointestinal and urinary tract anomalies as well as some extremities anomalies.

**Conclusion:** US diagnostic is a very useful method for evaluating the fetal health, fetal anomalies, anomalies of placenta and amniotic fluid as well as umbilical cord.

**Keywords:** Ultrasound, Fetal anomalies, Congenital abnormalities.

## INTRODUCTION

Congenital abnormalities account 20 to 25% of perinatal deaths. Now, many genetic and other disorders can be diagnosed early in pregnancy. Screening examinations during pregnancy are an essential part of prenatal care. Among the various screening tests that are now offered to pregnant women, US has the broadest diagnostic spectrum. There is no modality that can detect as many abnormalities during pregnancy as US. A priority goal in screening is the early detection of major fetal anomalies, which are defined as malformations that affect fetal viability and/or quality of life. During the past 10 years, some multicentric studies in Europe and USA show the successfulness of US diagnostics in detecting congenital abnormalities, even in women with low-risk pregnancy.

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### Neural Tube Defects (NTD)

Examples of NTD are anencephaly, exencephaly, encephalocele, iniencephaly and spina bifida. Based on EUROCT working group, the incidence of NTD in Europe is 11.5 per 10,000 infants (live-born infants, still-born infants and abortions).

*Anencephaly* is the absence of the superior vault and cerebrum. It is the most common and severe anomaly of the central nervous

system and the easiest to diagnose with US. The incidence is 1:100 births with 4:1 preponderance of females over males.<sup>5</sup>

Anencephaly can be detected by US at 11 weeks of gestation. The most characteristic feature at US is the absence of a superior vault and intracranial structures, large bulging eyes and marked disproportion between the head and trunk (Fig. 1).

In about 50% of cases, anencephaly is accompanied by spina bifida, palatoschisis, pes equinovarus, omphalocele and polyhydramnios.<sup>6</sup>

*Exencephaly or acrania*, all or most of the superior vault is absent while brain tissue is present (Fig. 2). It is less common than anencephaly. Evidence indicates that exencephaly is a precursor stage of anencephaly.<sup>7</sup>

*Cephalocele* is a defect in the bony skull through which the meninges (superior meningocele) and brain substance (encephalomeningocele) may protrude. The incidence is 1:2000 live births. Sonographically appears as a sac-like protrusion at the back of the skull (Fig. 3). A sac that contains only meninges will appear purely cystic, while a sac containing brain and meninges will also contain solid structures depending on the



Fig. 1: Anencephalus

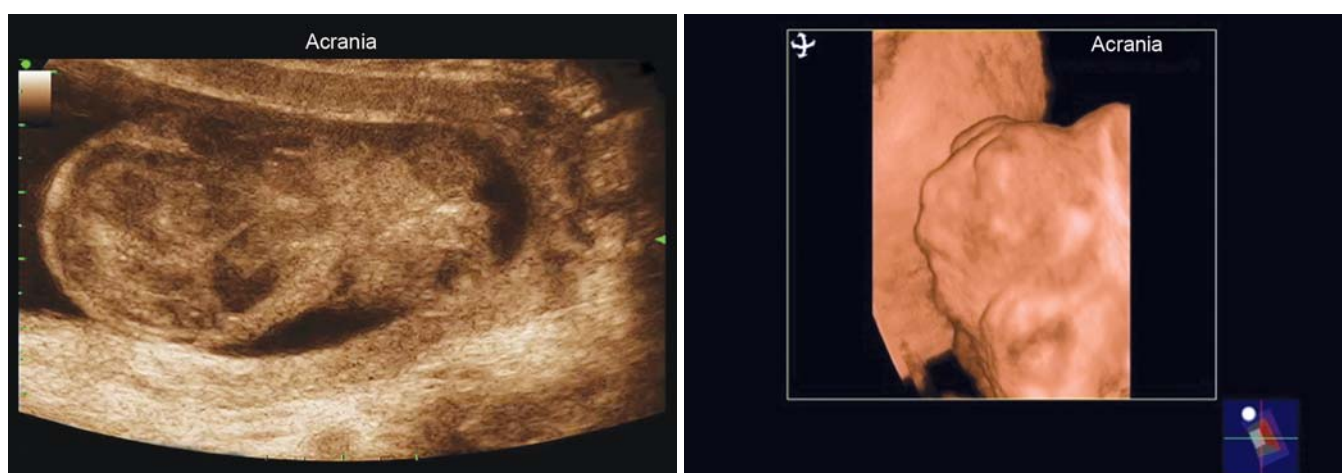


Fig. 2: Acrania



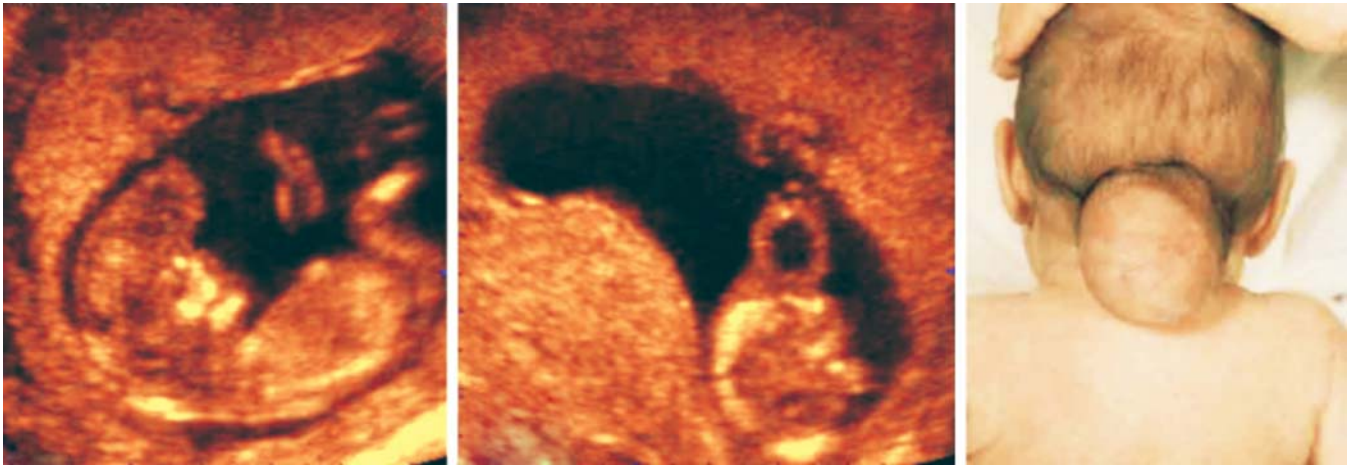


Fig. 3: Encephalocele

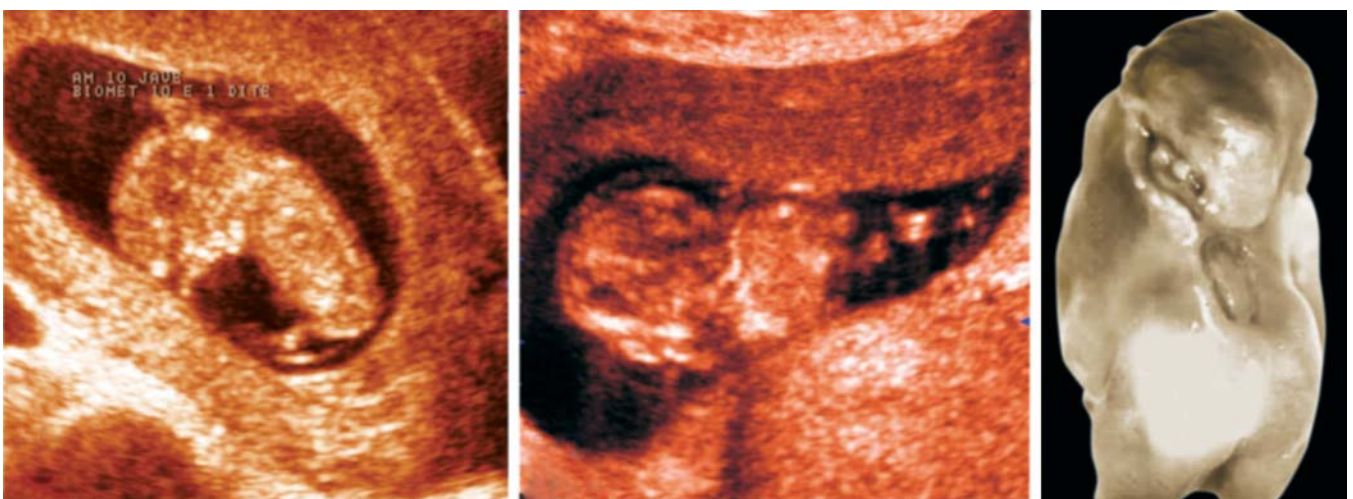


Fig. 4: Iniencephaly

proportion of herniated brain. Associated anomalies are hydrocephalus, microcephaly and spina bifida.

*Iniencephaly* is a malformation characterized by encephalocele, marked dorsiflexion of fetal head and cervical rachischisis. The incidence is very rare and the etiology is uncertain. The US diagnosis is based on detection of craniorachischisis with a short spine and marked dorsiflexion of the fetal head (Fig. 4).

Associated anomalies are found in 48% of fetuses with iniencephaly, such as anencephaly, holoprosencephaly, polymicrogyria, vermian agenesis, hydrocephalus, cyclopia, cleft lip and palate, cardiovascular disease, diaphragmatic hernia, abdominal wall defects, situs inversus, anal atresia, etc.

*Spina bifida* is a combined developmental defect, involving the spinal canal and its contents are characterized by partial or complete absence of the vertebral arches (Fig. 5). Several forms are distinguished according to the involvement of the spinal cord, meninges and skin:

- Complete spina bifida (rachischisis)
- Partial spina bifida:
  - Myelocele
  - Spina bifida cystic (meningocele, myelomeningocele) (Fig. 6).

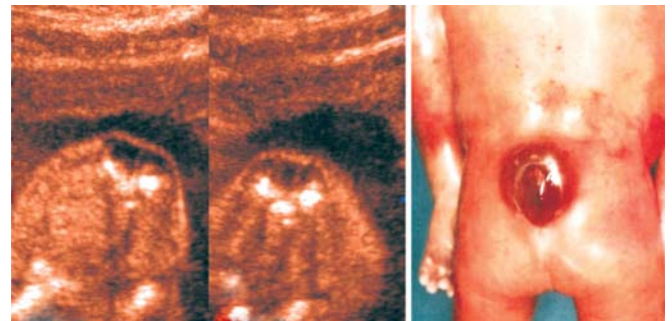


Fig. 5: Spina bifida

Posterior defects of neural tube closure are among the most common fetal anomalies. Spina bifida aperta is particularly important in prenatal diagnosis. The incidence in UK is 0.5%. Cleft in the spinal canal are among the defects that require great care and experience in their US detection. The majority of spinal defects are still overlooked in routine prenatal screening examinations.

US signs that suggest spina bifida are:

- Head growth decreases while trunk growth progresses normally
- Lemon sign (Fig. 7) and banana sign



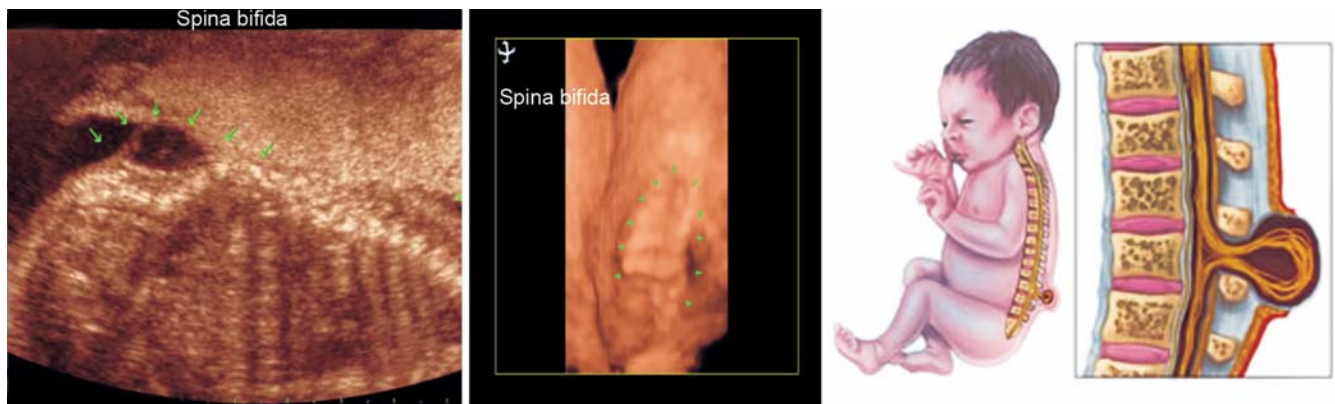


Fig. 6: Spina bifida cysticum

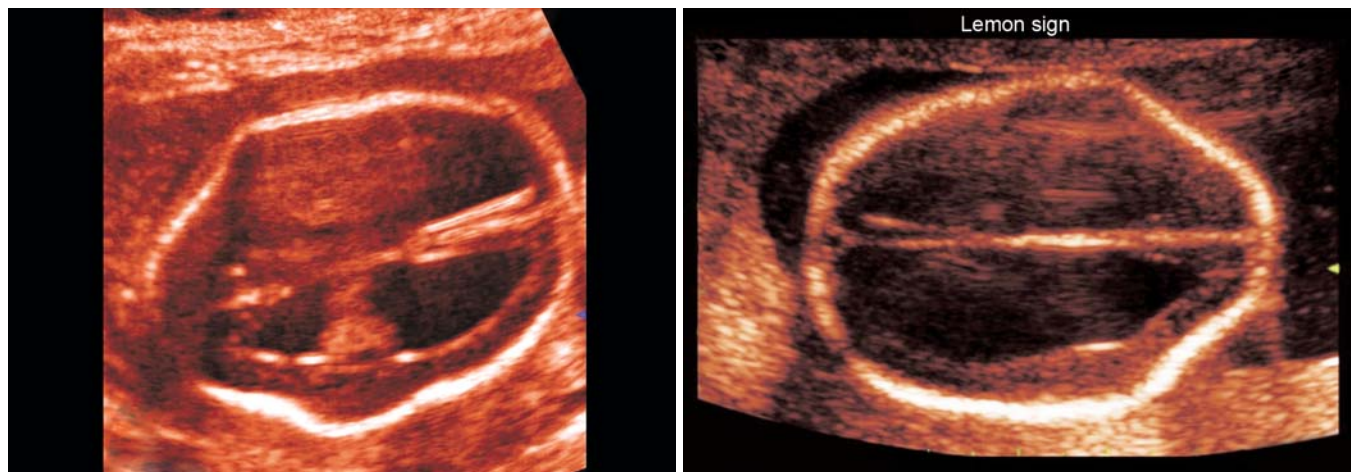


Fig. 7: Lemon sign

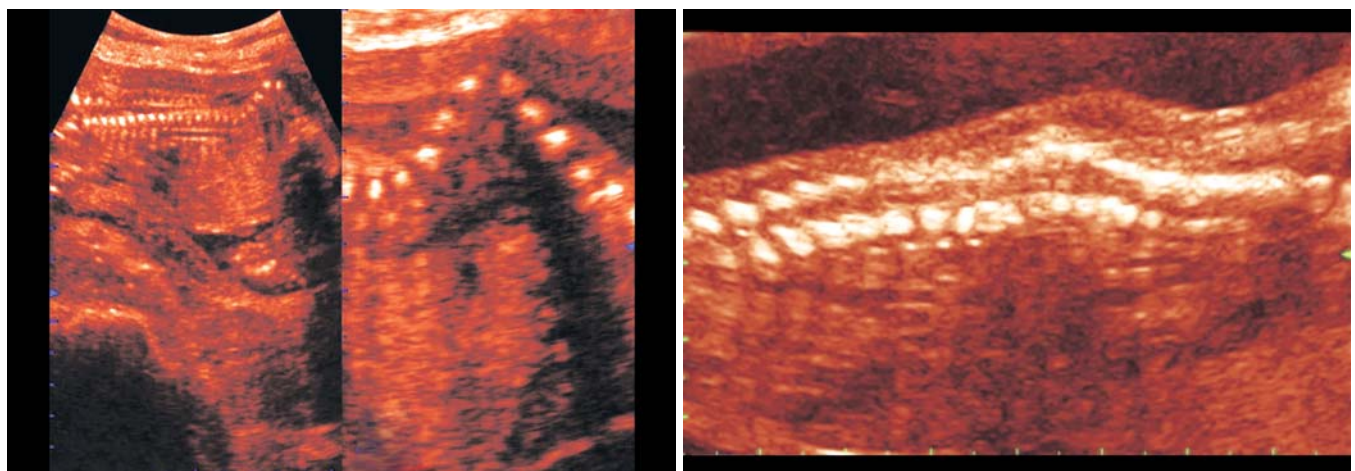


Fig. 8: Angulation of the fetal spine

- Foot deformities (pes equinovarus and impaired leg movements)
- Angulation of the fetal spine (Fig. 8).

*Sacroccygeal teratoma* is a cystic germ-cell tumor located in the sacroccygeal region (Fig. 9). The US diagnosis is:

- Nonhomogeneous echo structure (cystic and solid areas)
- Fetal hydrops in 25% of fetuses
- Polyhydramnios in 70% of cases

- Secondary signs (bladder displacement by tumor and ureteral obstruction leading to hydronephrosis)
- Associated anomalies in 18% of cases (musculoskeletal system).

### CNS Anomalies

*Hydrocephalus* denotes the increased intracranial accumulation of cerebrospinal fluid (CSF) with expansion of the CSF spaces.



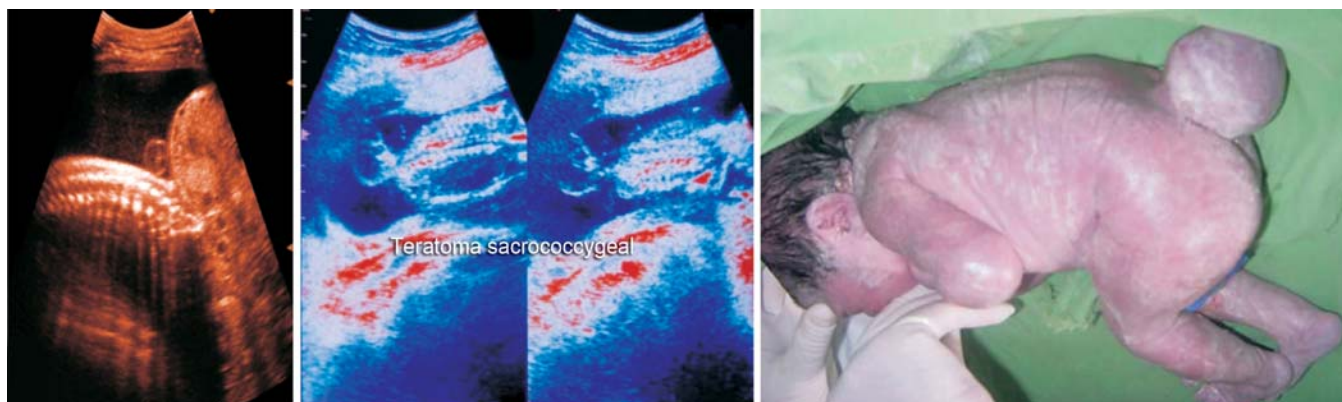


Fig. 9: Sacrococcygeal teratoma

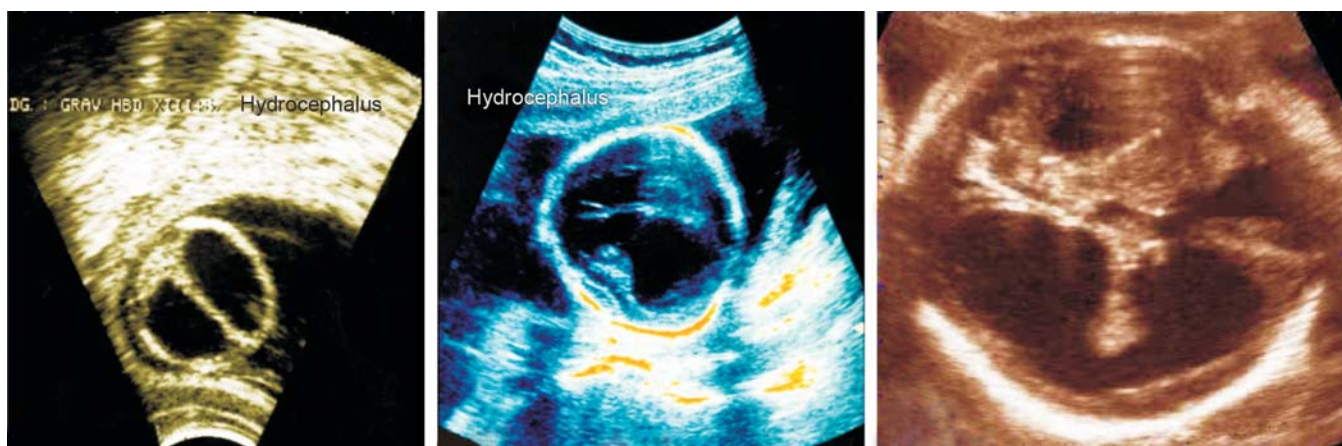


Fig. 10: Hydrocephalus

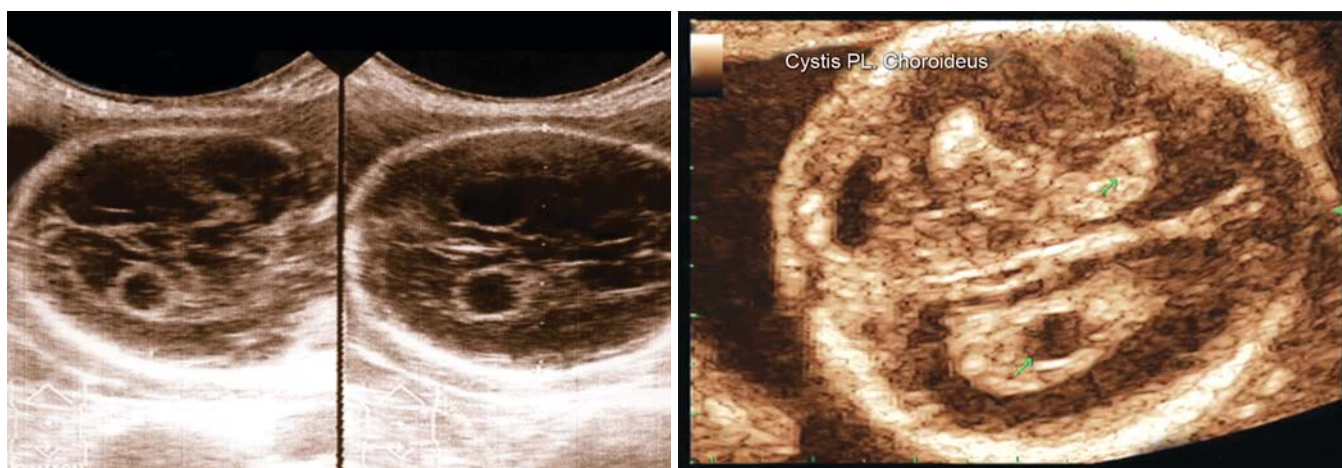


Fig. 11: Choroid plexus cyst

The incidence is 0.3 to 3 per 1000 live births. There are three forms:

- Internal hydrocephalus (expansion of the ventricular system)
- External hydrocephalus (expansion of the subarachnoid space)
- Communicating hydrocephalus (internal and external hydrocephalus).

The US diagnosis is possible in late pregnancy by noting excessive growth of the fetal head (Fig. 10). Now, with modern

US equipment, it is possible to detect hydrocephalus early in gestation, long before the skull becomes enlarged. 1/3 of cases is associated with neural tube defects. Other associated anomalies are heart defects, gastrointestinal and renal anomalies, cleft lip and palate, etc. The prognosis depends on the cause and extent of hydrocephalus as well as the presence of associated anomalies.

*Choroid plexus cyst(s):* Unilateral or bilateral cyst formation within the choroid plexus. This kind of cysts are found



sonographically in about 1% of fetuses between 16 and 24 weeks. Choroid plexus cysts appear sonographically as hypoechoic, usually round structures within the choroid plexus. The cysts may be unilateral or bilateral. Most cysts observed between 16 and 24 weeks, are no more detectable at 26 to 28 weeks (Fig. 11).

## Anomalies of the Neck

*Cystic hygroma* is an anomaly of the lymphatic system that appears sonographically as a thin-walled, uni- or multiloculated cystic mass. About 80% are located on the posterolateral part of the fetal neck. Other sites are the axilla, mediastinum and anterior chest wall. Cystic hygroma occurs in 0.3 to 2% of fetuses. A typical feature is the nuchal ligament which appears as a dorsal cord on transverse scans (Fig. 12). Differential diagnosis is required from cervical meningocele, myelomeningocele and hemangioma. Cystic hygromas are associated with chromosomal abnormalities in 50 to 80% of cases. The most common is Turner syndrome (monosomy XO).

## Facial Anomalies

*Cleft lip and palate* is a cleft formation in the lip and/or maxilla and/or palate. There are two forms: Lateral and median clefts.

The incidence is 1:1000. The defect is present in 13% of all infants with congenital anomalies. Cleft lip and palate can be diagnosed sonographically in a coronal or sagittal scan at the level of the maxilla (Fig. 13). 3D US can provide an accurate tomographic survey of cleft lip defects.

*Cyclopia* is a facial anomaly with a common orbit and only one eye.

*Synophthalmia* is a facial anomaly with a common orbit and two closely adjacent eyes (Fig. 14).

The incidence of these anomalies is very rare. The disorder is mainly found in association with holoprosencephaly. By US to targeting imaging of the fetal face will disclose the anomaly. The common orbit is best appreciated in a transverse scan through the upper half of the face.

## Thoracic Anomalies

*Hydrothorax* is unilateral or bilateral effusion within the pleural cavity. The incidence is 1:10,000 births. By US, we can see fluid collection in the chest between the lung and bony thorax (Fig. 15). As the hydro/chylothorax increases, the pleural cavity fills with a hypoechoic fluid causing marked compression of

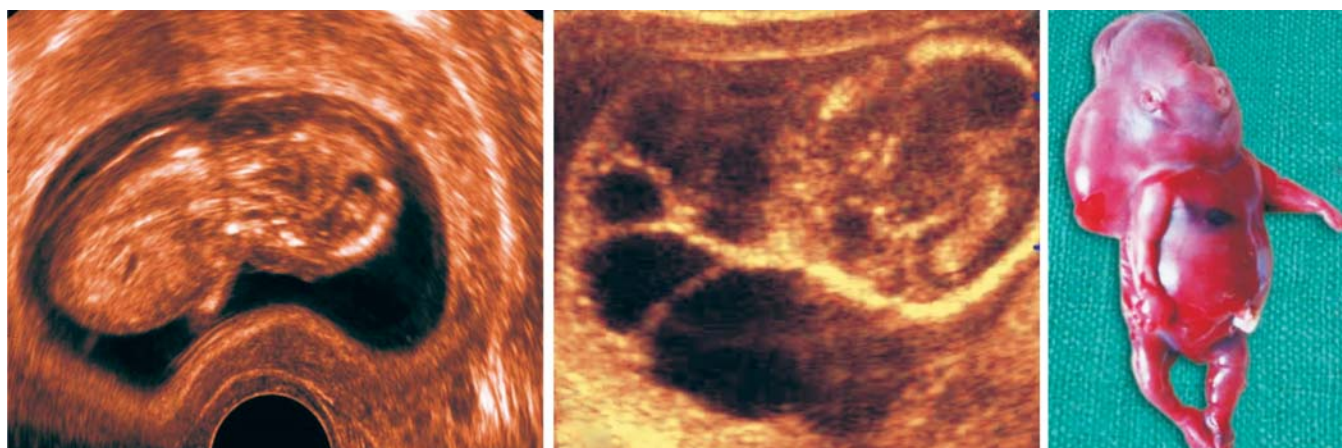


Fig. 12: Cystic hygroma



Fig. 13: Cleft lip and palate





Fig. 14: Synophthalmia

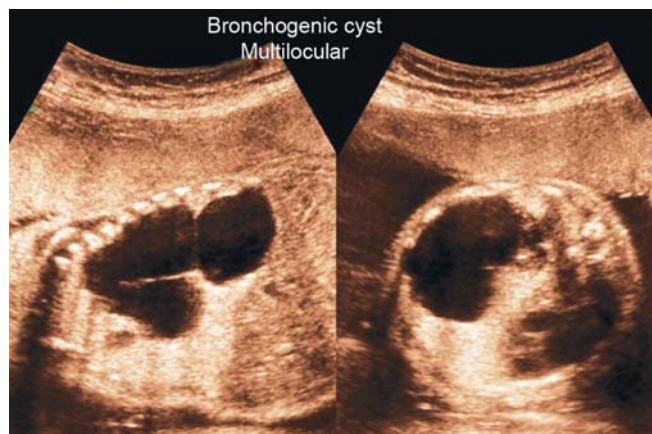


Fig. 16: Bronchogenic cyst

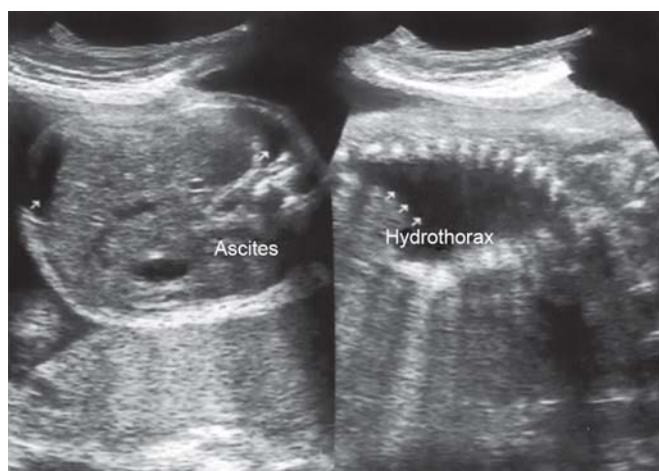


Fig. 15: Hydrothorax



the lung and cardiac tissue. Associated anomalies are trisomy 21, anomalies of the blood vessels or lymphatics. Hydrothorax is usually associated with polyhydramnios.

*Bronchogenic cysts* are isolated cysts occurring within the thorax. The incidence is very rare. By US, we can see an isolated, hypoechoic, smoothly margined structure in the lung region (Fig. 16). Sometimes occur in the area of mediastinum.

*Thoracopagus* is an anomaly of twins characterized by a fusion of the thoracic part of the both fetuses. By US, we can see two adjacent spinal columns with a common thorax (Fig. 17). The pelvic regions are not fused.

*Cephalothoracopagus* is an anomaly of twins characterized by a fusion of the head and trunk (Fig. 18).

*Cardiac rhabdomyoma* is the most frequent cardiac tumor (60% of cases) (Fig. 19). It may be multiple and often affect the interventricular septum. Sometimes the remission is spontaneous. The tumor is detected incidentally in the fetus, but, often there are associated supraventricular extrasystoles or nonimmune hydrops due to circulatory obstruction by the tumor. Rhabdomyoma can be associated with tuberous sclerosis (Bourneville-Pringle disease), which is associated with mental retardation and seizures.

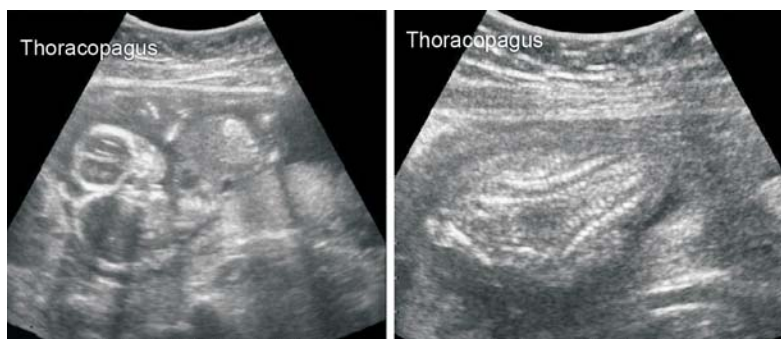


Fig. 17: Thoracopagus



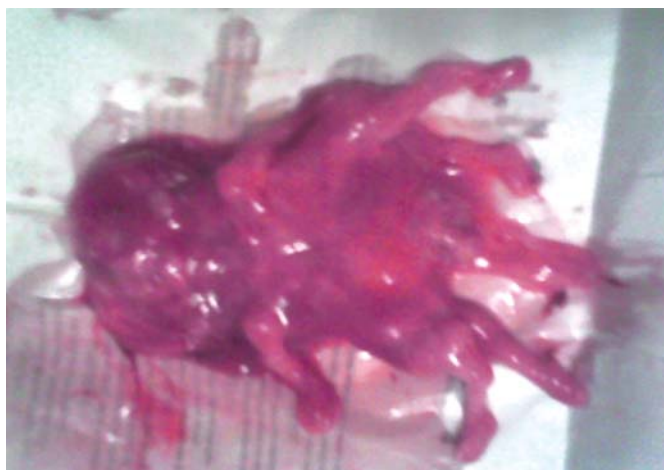


Fig. 18: Cephalothoracopagus

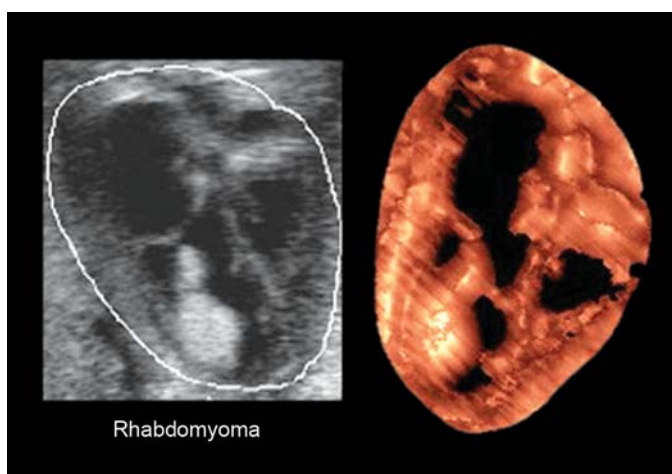


Fig. 19: Cardiac rhabdomyoma

### Anomalies of the Gastrointestinal Tract and Anterior Abdominal Wall

*Omphalocele* is a ventral abdominal wall defect with a midgut hernia sac containing abdominal viscera. The incidence is 1:10,000 births. Both longitudinal and transverse US scans show a hernia sac located in front of the abdominal wall and connected to the abdomen by a base of variable width (Fig. 20). Depending on the size of the defect, the sac may contain only bowel loops or it may contain stomach and liver. The fetus with omphalocele has an abdominal circumference too small in relation to the fetal age. The early US diagnosis between 10 and 14 weeks of gestation is possible. But, it should be noted that the physiologic umbilical hernia is still present up to 12 weeks of gestation and may be misinterpreted as omphalocele. Serial scans should be obtained in equivocal cases.

About 45 to 60% of live-born infants with omphalocele have associated anomalies, like neural tube defects, skeletal malformations, cardiovascular and gastrointestinal anomalies.

*Gastroschisis* is an open sporadically occurring abdominal wall defect with extruded loops of bowel. The incidence is less common than omphalocele, 1:30,000 births. Since there is no

hernia sac, the bowel loops extruded from the paraumbilical defect on the right side of the abdomen, float freely in the amniotic fluid (Fig. 21). The condition can be diagnosed by the end of the first trimester with transvaginal US.

*Esophageal atresia* is an anomalous closure of the esophagus that may or may not be associated with a tracheoesophageal fistula. The incidence is reported from 1:1500 to 1:5000 live births. Diagnosis is difficult with prenatal US. Polyhydramnios and nonvisualization of fetal stomach are helpful suggestive signs (Fig. 22). In 64% of cases, esophageal atresia is associated with other anomalies of gastrointestinal tract, heart, urogenital tract, skeletal system and CNS. Polyhydramnios is considered typical for esophageal atresia, but it is not a sensitive marker because it appears after 24 weeks of gestation and also occurs in numerous other disorders.

*Duodenal atresia* is the congenital closure of duodenum. The incidence is 1:10,000 live births. The US sign of duodenal atresia is the “double-bubble sign”, which consists in two adjacent fluid-filled cavities in the upper abdomen representing the dilated stomach and the distended proximal duodenum between the pylorus and the stenosis (Fig. 23). More than half



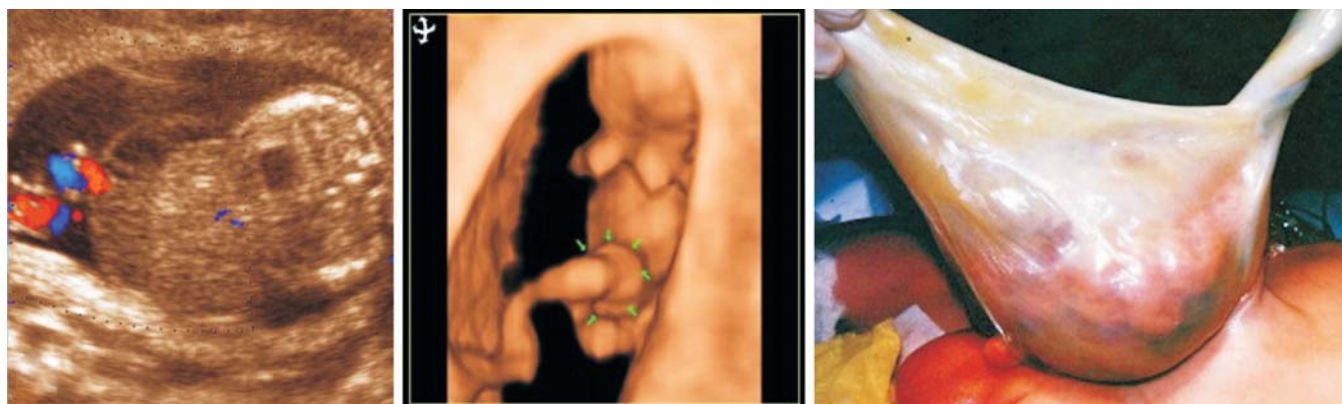


Fig. 20: Omphalocele

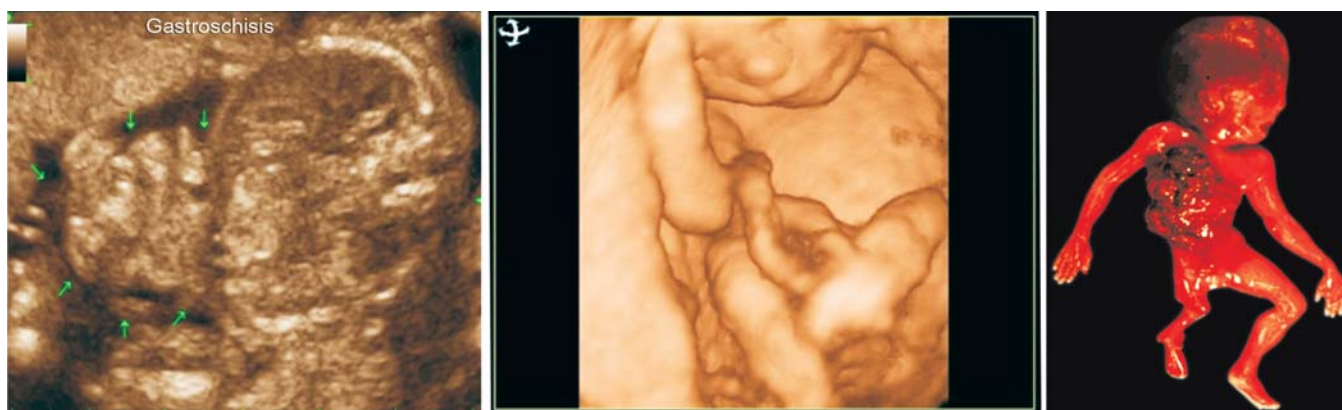


Fig. 21: Gastroschisis



Fig. 22: Esophageal atresia

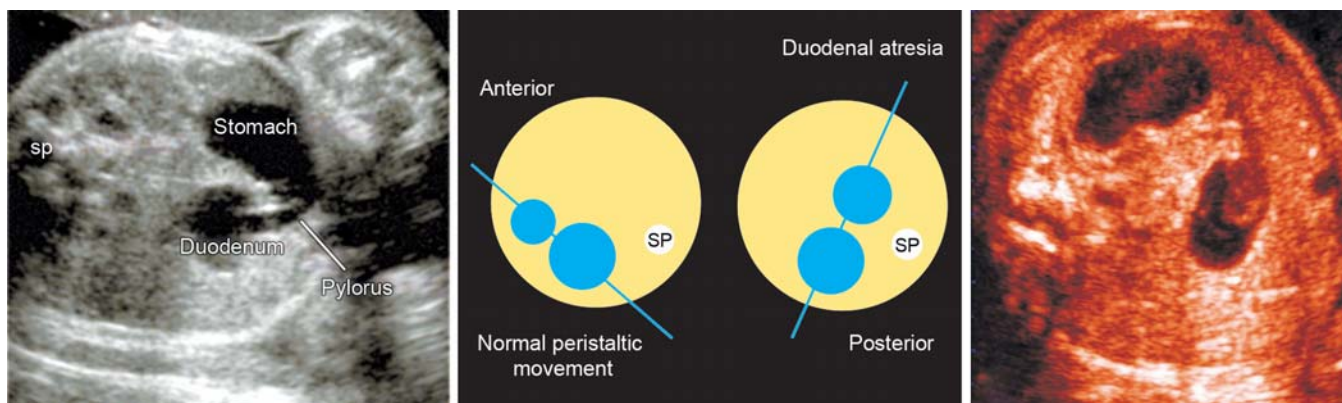


Fig. 23: Duodenal atresia

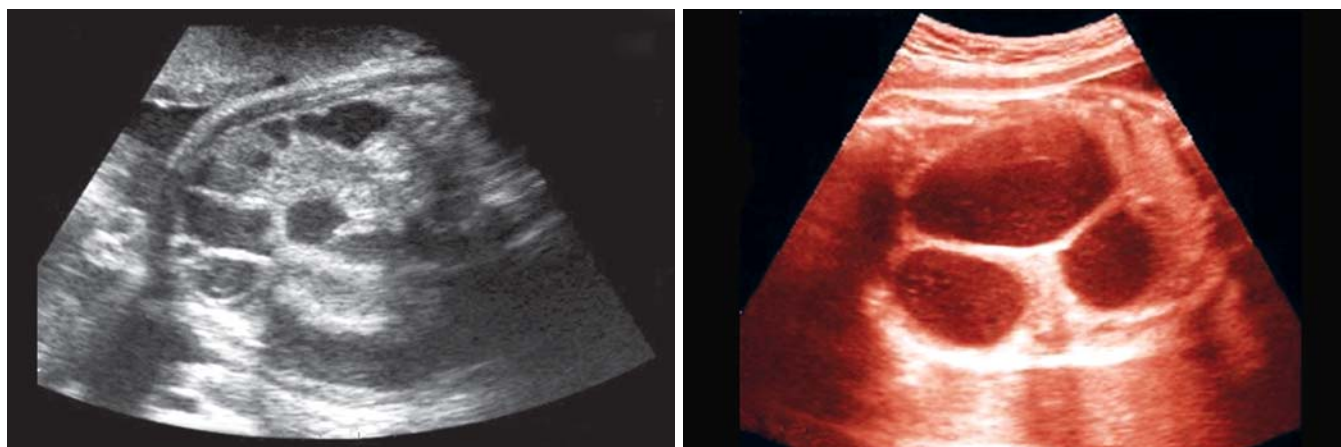


Fig. 24: Small-bowel atresia

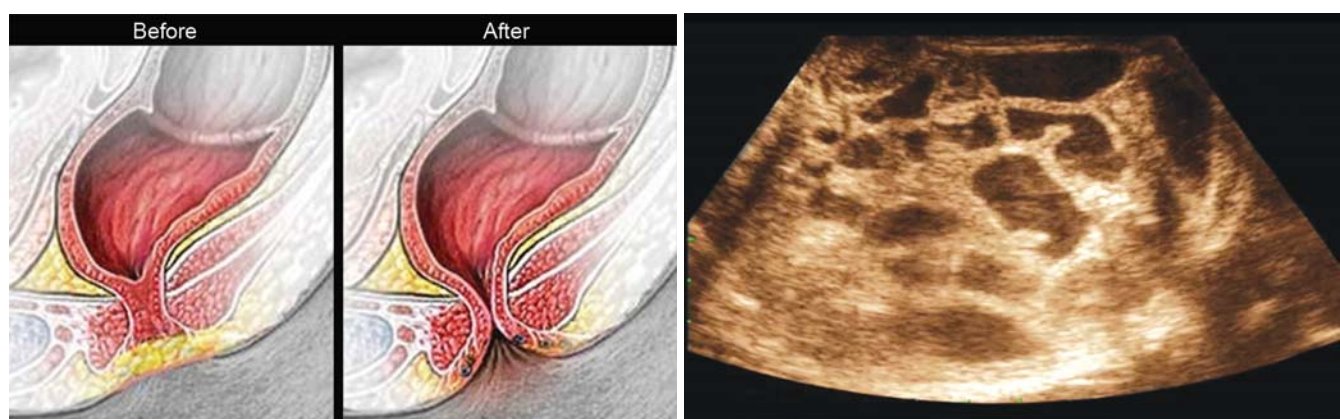


Fig. 25: Anal atresia

of fetuses with duodenal atresia have associated cardiac, renal, musculoskeletal or CNS anomalies. Trisomy 21 is present in 30 to 43% of cases.

*Jejunal and ileal atresia* is the complete distal closure of the small-bowel lumen at the level of the jejunum or ileum. The incidence is 1:6000 and most cases are jejunum atresia. By US, the small-bowel atresia usually appears as multiple cystic areas within the fetal abdomen (Fig. 24). These areas may change their shape during prolonged observations as a result of peristalsis. Most cases are diagnosed at the third trimester.

*Anal atresia* is the congenital closure of the anus. The incidence is 1:2000 to 1:3000 births. By US, we can see enlarged fluid-filled loops of large bowel (Fig. 25). Others have normal appearing colon loops. Thus US cannot exclude anal atresia with complete confidence.

### Anomalies of the Kidneys and Urinary Tract

*Polycystic renal diseases* are the most frequent congenital kidney anomalies. Autosomal-recessive polycystic kidney disease (ARPKD) corresponds to infantile polycystic kidney disease or potter type I cystic kidney disease (Fig. 26). The incidence is between 1:6000 and 1:40,000 births. The prenatal US findings are highly variable. Some affected fetuses display

only the image of a 'sponge-like kidneys' and some others show none at all. It is common to find oligohydramnios or anhydramnios in the early second trimester.

*Megacystis* or big bladder is an anomaly caused by the obstruction of the lower segment—posterior urethral valve (Fig. 27). This rare syndrome is difficult to distinguish from Prune-Belly syndrome, but a presumptive diagnosis can be made when a dilated urinary bladder is found in association with distended bowel loops (Fig. 28).

*Hydronephrosis* is defined as enlargement of the renal pelvis to more than 15 cm in its anteroposterior dimension. It is the most common fetal kidney anomaly. The US evaluation of the expanded pyelon is performed in the anteroposterior manner (Fig. 29).

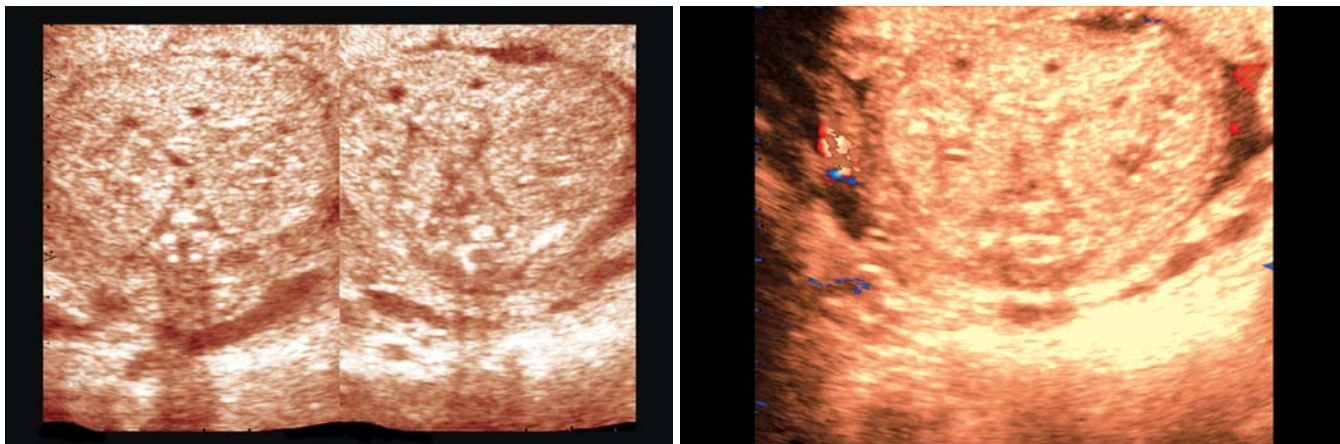
### Anomalies of the Extremities

*The short-rib polydactyly* syndromes are a group of osteochondrodysplasias, characterized by a narrow thorax, brachymelia and polydactyly (Fig. 30). Various organ malformations may also be found.

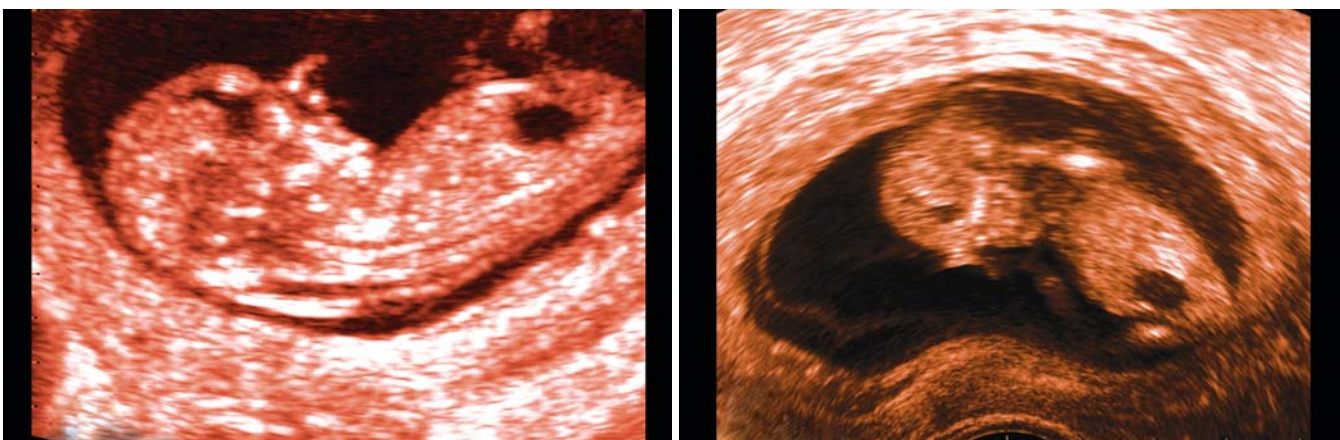
Amelia denotes the absence of one or more entire limb(s) (arm or leg). A small soft-tissue protuberance may be present at the site of missing limb (Fig. 31).

Phocomelia is a congenital condition in which one or more limbs are missing, with the hand and/or foot attached directly to the trunk of the body (Fig. 32).





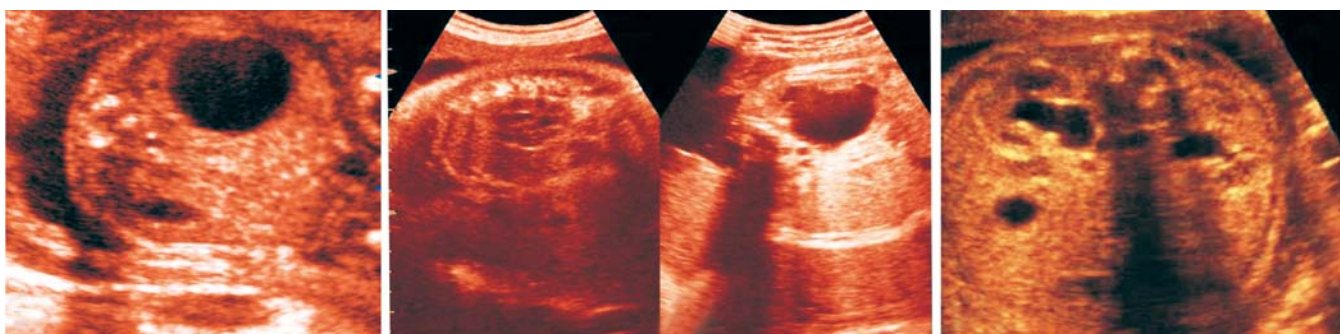
**Fig. 26:** Polycystic kidney



**Fig. 27:** Megacystis



**Fig. 28:** Prune-belly syndrome



**Fig. 29:** Hydronephrosis





Fig. 30: Polydactyly



Fig. 31: Amelia of upper extremities

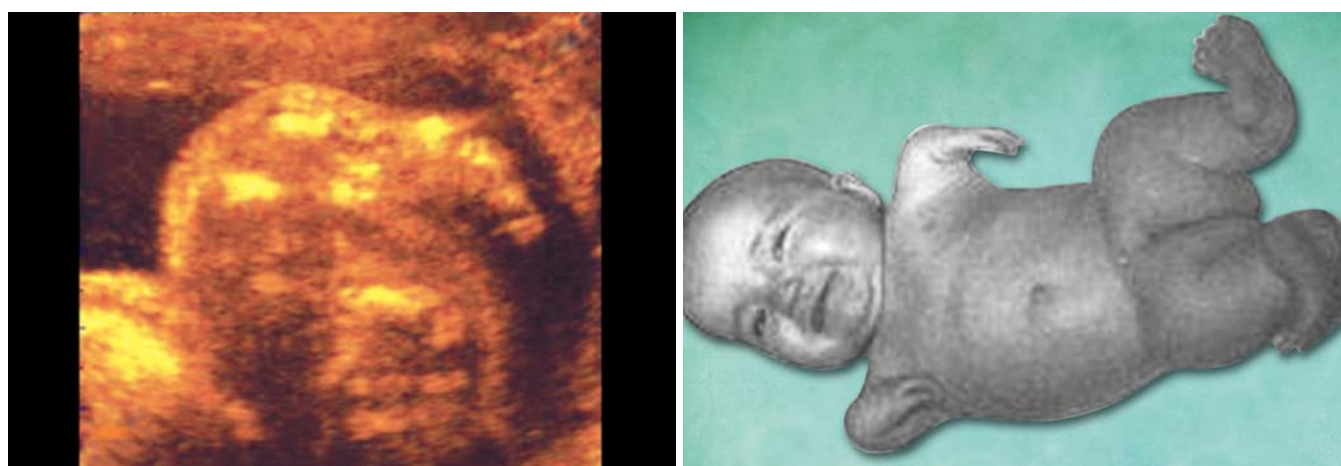


Fig. 32: Phocomelia

## CONCLUSION

US diagnosis is a very useful method for evaluating the fetal health, fetal anomalies, anomalies of placenta and amniotic fluid as well as umbilical cord.

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