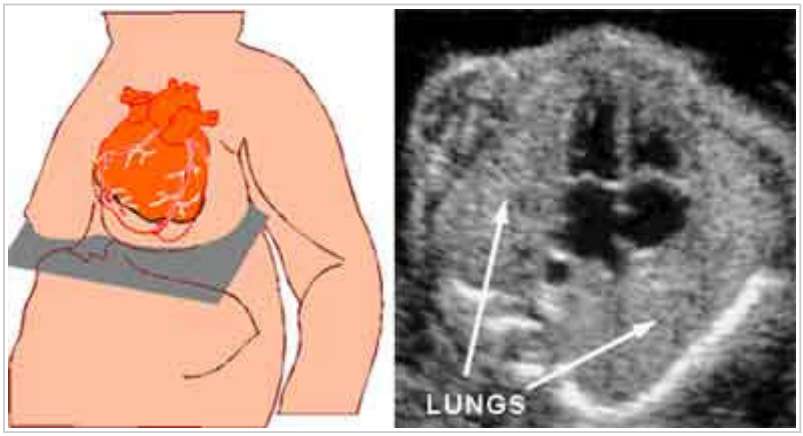
Diploma in Fetal Medicine & ISUOG Educational Series
The 18 - 23 weeks scan
Gianluigi Pilu, Kypros Nicolaides, Renato Ximenes & Phillipe Jeanty

Chapter 5

Pulmonary abnormalities

NORMAL SONOGRAPHIC ANATOMY

For the purpose an an ultrasound survey of fetal anatomy, examination of the lungs in the same section used for the four-chamber view of the fetal heart is sufficient. Under normal condition, the fetal lungs are uniformely echogenic. At 18-23 weeks, the central third of the thoracic area at the level of the four chamber view is occupied by the heart, and the remaining two thirds by the lungs, that are normally uniformely echogenic. This scanning plane can also be used for the measurement of the thoracic circumference, that is correlated with the development of the lungs.



A sagittal plane of the fetal trunk usually allows one to identify the diaphragm as a thin sonolucent line separating the abdominal from the thoracic cavity.

CYSTIC ADENOMATOID MALFORMATION (CAM)

Cystic adenomatoid malformation of the lung is a developmental abnormality arising from an overgrowth of the terminal respiratory bronchioles. The condition may be bilateral involving all lung tissue, but in the majority of cases it is confined to a single lung or lobe. The lesions are either macrocystic (cysts of at least 5 mm in diameter) or microcystic (cysts less than 5 mm in diameter). In 85% of cases, the lesion is unilateral with equal frequency in the right and left lungs and equal frequency in the microcystic and macrocystic types.

Prevalence

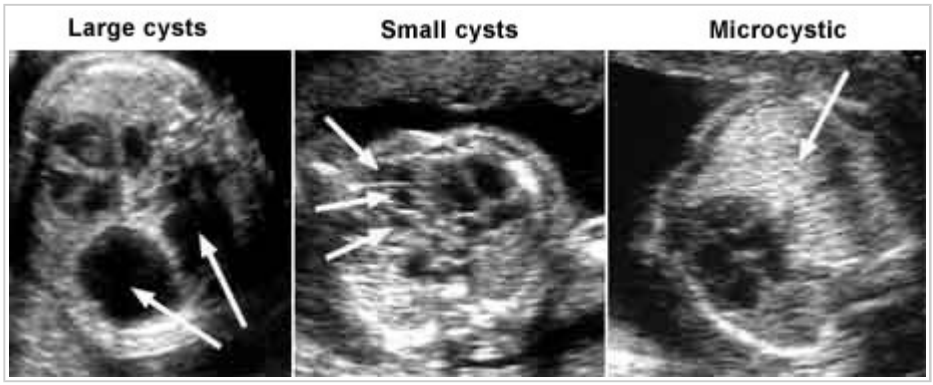
Cystic adenomatoid malformation of the lung is found in about 1 in 4000 births.

Etiology

This is a sporadic abnormality. In about 10% of cases, there are other abnormalities, mainly cardiac and renal.

Diagnosis

Prenatal diagnosis is based on the ultrasonographic demonstration of a hyperechogenic pulmonary tumor which is cystic (CAM type 1), mixed (CAM type 2), or solid – microcystic (CAM type 3). Microcystic disease results in uniform hyperechogenicity of the affected lung tissue. In macrocystic disease, single or multiple cystic spaces may be seen within the thorax. Both microcystic and macrocystic disease may be associated with deviation of the mediastinum.



When there is compression of the heart and major blood vessels in the thorax, fetal hydrops develops. Polyhydramnios is a common feature and this may be a consequence of decreased fetal swallowing of amniotic fluid due to esophageal compression, or increased fluid production by the abnormal lung tissue. Prognostic features for poor outcome include major lung compression causing pulmonary hypoplasia, polyhydramnios and development of hydrops fetalis irrespective of the type of the lesion.

Prognosis

Bilateral disease is lethal either in utero, due to progressive hydrops, or in the neonatal period. Isolated unilateral cystic adenomatoid malformation without hydrops is associated with a good prognosis; in about 70% of cases, the relative size of the fetal tumor remains stable, in 20% of cases there is antenatal shrinkage or resolution, and in 10% of cases there is progressive increase in mediastinal compression. In symptomatic neonates, thoracotomy and lobectomy are carried out and survival is about 90%. It is uncertain whether surgery is also needed for asymptomatic neonates.

Fetal therapy

Large intrathoracic cysts causing major mediastinal shift and associated hydrops can be treated effectively by the insertion of thoraco-amniotic shunts. The role of more invasive intervention, such as hysterotomy and excision of solid tumors in cases of fetal hydrops, remains to be defined. Although good results have been reported after such surgery in a small number of cases, the potential risks to the mother both during the pregnancy and in subsequent confinements should not be underestimated.

DIAPHRAGMATIC HERNIA

Development of the diaphragm is usually completed by the 9th week of gestation. In the presence of a defective diaphragm, there is herniation of the abdominal viscera into the thorax at about 10–12 weeks, when the intestines return to the abdominal cavity from the umbilical cord. However, at least in some cases, intrathoracic herniation of viscera may be delayed until the second or third trimester of pregnancy.

Prevalence

Diaphragmatic hernia is found in about 1 per 4000 births.

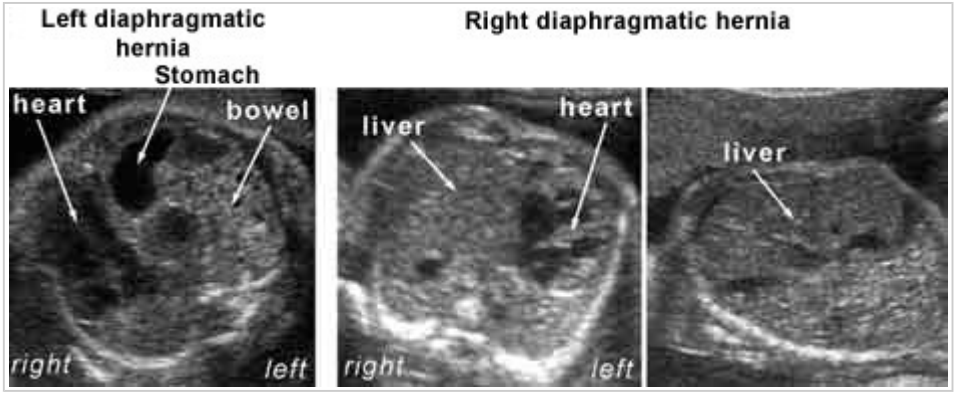
Etiology

Diaphragmatic hernia is usually a sporadic abnormality. However, in about 50% of affected fetuses there are associated chromosomal abnormalities (mainly trisomy 18, trisomy 13 and Pallister–Killian syndrome – mosaicism for tetrasomy 12p), other defects (mainly craniospinal defects, including spina bifida, hydrocephaly and the otherwise rare iniencephaly, and cardiac abnormalities) and genetic syndromes (such as Fryns syndrome, de Lange syndrome and Marfan syndrome).

Diagnosis

Prenatally, the diaphragm is imaged by ultrasonography as an echo-free space between the thorax and abdomen. Diaphragmatic hernia can be diagnosed by the ultrasonographic demonstration of stomach and intestines (90% of the cases) or liver (50%) in the thorax and the associated mediastinal shift to the opposite side. Herniated abdominal

contents, associated with a left-sided diaphragmatic hernia, are easy to demonstrate because the echo-free fluid-filled stomach and small bowel contrast dramatically with the more echogenic fetal lung. In contrast, a right-sided hernia is more difficult to identify because the echogenicity of the fetal liver is similar to that of the lung, and visualization of the gall bladder in the right side of the fetal chest may be the only way of making the diagnosis.



Polyhydramnios (usually after 25 weeks) is found in about 75% of cases and this may be the consequence of impaired fetal swallowing due to compression of the esophagus by the herniated abdominal organs. The main differential diagnosis is from cystic lung disease, such as cystic adenomatoid malformation or mediastinal cystic processes, e.g. neuroenteric cysts, bronchogenic cysts and thymic cysts. In these cases, a fluid-filled structure causing mediastinal shift may be present within the chest. However, in contrast to diaphragmatic hernia, the upper abdominal anatomy is normal.

Antenatal prediction of pulmonary hypoplasia remains one of the challenges of prenatal diagnosis because this would be vital in both counselling parents and also in selecting those cases that may benefit from prenatal surgery. Poor prognostic signs are, first, increased nuchal translucency thickness at 10–14 weeks, second, intrathoracic herniation of abdominal viscera before 20 weeks, and, third, severe mediastinal compression suggested by an abnormal ratio in the size of the cardiac ventricles and the development of polyhydramnios.

Prognosis

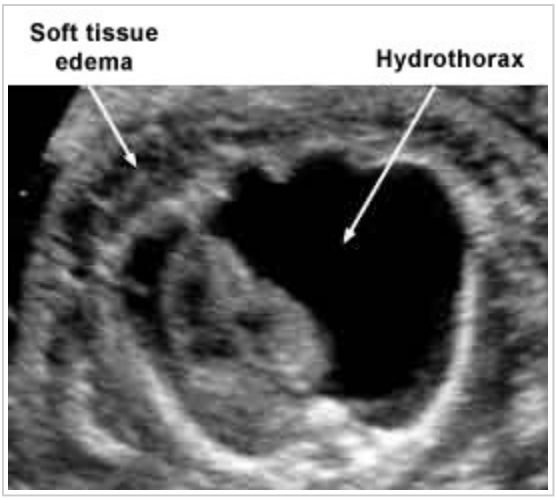
In the human, the bronchial tree is fully developed by the 16th week of gestation, at which time the full adult number of airways is established. The alveoli continue to develop even after birth, increasing in number and size until the growth of the chest wall is completed in adulthood. The growth of blood vessels supplying the acinus (intra-acinar vessels) parallels alveolar development, while the growth of pre-acinar vessels follows the development of the airways. In diaphragmatic hernia, the reduced thoracic space available to the developing lung leads to reduction in airways, alveoli and arteries. Furthermore, there is an increase in arterial medial wall thickness and extension of muscle peripherally into the small pre-acinar arteries, offering an explanation for the pulmonary hypertension and persistent fetal circulation observed after neonatal repair. Thus, although isolated diaphragmatic hernia is an anatomically simple defect, which is easily correctable, the mortality rate is about 50%. The main cause of death is hypoxemia due to pulmonary hypertension, resulting from the abnormal development of the pulmonary vascular bed.

Fetal therapy

Extensive animal studies have suggested that pulmonary hypoplasia and hypertension due to intrathoracic compression are reversible by in utero surgical repair. However, such therapy is likely to have limited success in the human because the bronchial tree is fully developed by the 16th week of gestation. For a fetus with a sonographically demonstrable large diaphragmatic hernia at 16–18 weeks, irreversible maldevelopment of the bronchial tree and vasculature is likely. However, in fetuses with a diaphragmatic defect which allows the intrathoracic herniation of abdominal viscera only after mid-gestation (when the bronchial tree and pre-acinar vessels are fully developed), prenatal correction, by allowing further development of the alveoli and intra-acinar vessels, may well prevent pulmonary hypoplasia and neonatal death. In a few cases of diaphragmatic hernia, hysterotomy and fetal surgery have been carried out but this intervention has now be abandoned in favor of minimally invasive surgery. Animal studies have demonstrated that obstruction of the trachea results in expansion of the fetal lungs by retained pulmonary secretions. Endoscopic occlusion of the fetal trachea has also been carried out in human fetuses with diaphragmatic hernia, but the number of cases is too small for useful conclusions to be drawn as to the effectiveness of such treatment.

PLEURAL EFFUSIONS

Fetal pleural effusions, which may be unilateral or bilateral, may be an isolated finding or they occur in association with generalized edema and ascites.



Prognosis

Irrespective of the underlying cause, infants affected by pleural effusions usually present in the neonatal period with severe, and often fatal, respiratory insufficiency. This is either a direct result of pulmonary compression caused by the effusions, or due to pulmonary hypoplasia secondary to chronic intrathoracic compression. The overall mortality of neonates with pleural effusions is 25%, with a range from 15% in infants with isolated pleural effusions to 95% in those with gross hydrops. The mortality rate in cases of antenatally diagnosed chylothorax is about 50%. Isolated pleural effusions in the fetus may either resolve spontaneously or they can be treated effectively after birth. Nevertheless, in some cases, severe and chronic compression of the fetal lungs can result in pulmonary hypoplasia and neonatal death. In others, mediastinal compression leads to the development of hydrops and polyhydramnios, which are associated with a high risk of premature delivery and perinatal death.

Fetal therapy

Attempts at prenatal therapy by repeated thoracocenteses for drainage of pleural effusions have been generally unsuccessful in reversing the hydropic state, because the fluid reaccumulates within 24–48 h of drainage. A better approach is chronic drainage by the insertion of thoracoamniotic shunts. This is useful both for diagnosis and treatment. First, the diagnosis of an underlying cardiac abnormality or other intrathoracic lesion may become apparent only after effective decompression and return of the mediastinum to its normal position. Second, it can reverse fetal hydrops, resolve polyhydramnios and thereby reduce the risk of preterm delivery, and may prevent pulmonary hypoplasia. Third, it may be useful in the prenatal diagnosis of pulmonary hypoplasia because, in such cases, the lungs often fail to expand after shunting. Furthermore, it may help to distinguish between hydrops due to primary accumulation of pleural effusions, in which case the ascites and skin edema may resolve after shunting, and other causes of hydrops such as infection, in which drainage of the effusions does not prevent worsening of the hydrops. Survival after thoracoamniotic shunting is more than 90% in fetuses with isolated pleural effusions and about 50% in those with hydrops.

SEQUESTRATION OF THE LUNGS

In lung sequestration, a portion of the lung develops without connection to the airways. The blood supply to the abnormal lung tissue is through arteries that arise from the descending aorta rather than from the pulmonary artery. This condition is classically divided in the radiological literature into intralobar (about 75%) and extralobar (about 25%), but the difference (which is based on the presence or absence of a separate pleural covering from the normal lung) cannot be accurately determined with prenatal ultrasound.

Prevalence

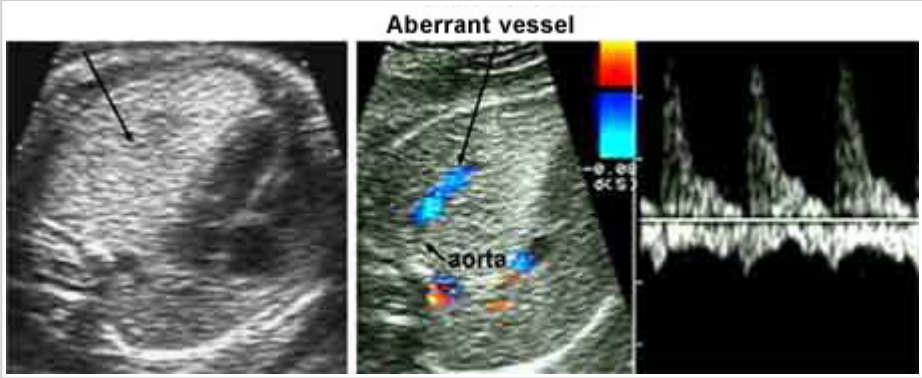
Sequestration of the lungs is rare and the prevalence is less than 5% of congenital pulmonary abnormalities.

Etiology

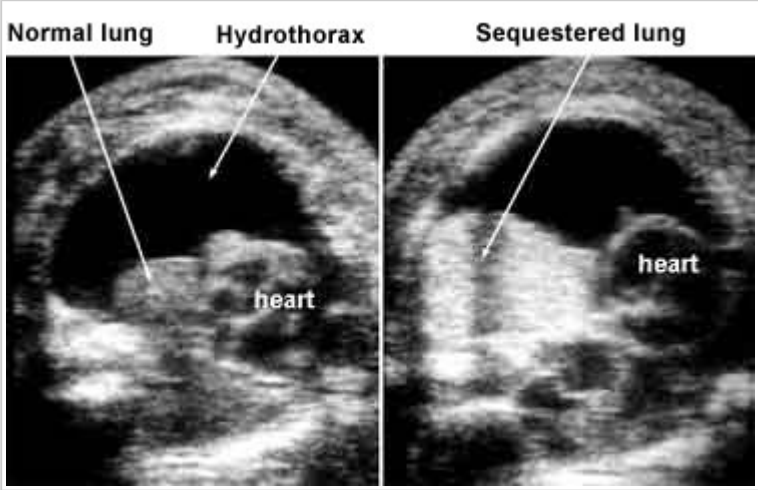
Sequestration of the lungs is a sporadic abnormality.

Diagnosis

The sequestrated portion of the lung appears as a homogeneous, brightly echogenic mass in the lower lobes of the lungs or in the upper abdomen (infradiaphragmatic sequestration). The diagnosis is confirmed by color Doppler demonstration that the vascular supply of the sequestered lobe arises from the abdominal aorta.



Large lung sequestration may act as an arteriovenous fistula and cause high-output heart failure and hydrops.



Intralobar sequestrations are usually isolated, whereas more than 50% of extralobar sequestrations are associated with other abnormalities (mainly diaphragmatic hernia and cardiac defects).

Prognosis

Postnatal outcome depends on the presence of associated abnormalities, and hemodynamic disturbances. In general, intralobar sequestration has an excellent prognosis, whereas extralobar sequestration has a poor prognosis because of the high incidence of other defects and hydrops.