A 27-years-old woman, G3P2L2, came for a routine second trimester anomaly scan. Gestational age was determined to be 24 weeks. USG images of the fetal thorax [Figures 1 and 2] are shown. What is the complete antenatal diagnosis?

What is your diagnosis?

**Diagnosis: Left-sided congenital diaphragmatic hernia with intrathoracic herniation of the left lobe of liver**

USG of the fetal thorax reveals mediastinal shift to the right [Figure 3]. The gastric bubble is seen in the left hemithorax, posteromedially, adjacent to the spine [Figure 3]. Posterolaterally, adjacent to the gastric bubble, small echogenic foci are seen corresponding to the bowel loops [Figure 4] and anteriorly in the left hemithorax, a homogenous hypo/isoechoic structure is seen, corresponding to the left lobe of the liver [Figure 4]. The patient opted for termination of pregnancy. Autopsy of the fetus was performed [Figures 5 and 6], which confirmed the diagnosis.

Congenital diaphragmatic hernia (CDH) is a developmental abnormality, resulting in a defect in the diaphragm through which the abdominal viscera herniate into the chest. CDH is the most common developmental anomaly of the diaphragm. The incidence is between 1 in 2000 and 1 in 5000 live births. The sex ratio is 1: 1. The majority of CDH (75–90%) are left-sided, 10% are right sided, and <5% are bilateral.[1]

The muscular diaphragm forms between the 6th and 14th weeks of gestation by fusion of the septum transversum, the
Radiological Quiz & Answer

Figure 3: Transverse USG of the fetal thorax - four chamber cardiac view shows the echogenic fetal spine (a), the gastric bubble (b) as an anechoic cystic structure and the four-chamber cardiac view (c)

dorsal mesentery of the esophagus, the innermost muscle layer of the thoracic cage, and the descending cervical myoblasts. There is a communication between the pleural and peritoneal cavities (the pleuroperitoneal canal) up to

Figure 4: Transverse USG of the fetal thorax - four chamber cardiac view captured five minutes later shows multiple small echogenic foci, representing the intestines (d), as well as the boundary between the fetal intestine and the left lobe of liver (e). The left lobe of the liver (f) is seen as a homogenous structure isoechoic to the fetal liver in the anterior part of the left hemithorax. sp-spine

Figure 5: Dissection photograph of the fetal thorax and abdomen shows the various structures: 1 - right lobe of the liver, 2 - right hemidiaphragm, 3 - fetal heart, 4 - right lung, 5 - thymus, 6 - hypoplastic left lung, 7 - hernial sac in the left hemithorax with contents

Figure 6: Dissection photograph of the fetal thorax and abdomen after opening the hernial sac, shows absence of the left hemidiaphragm and continuation of the left lobe of the liver and small intestinal coils from the abdomen into the left hemithorax. 8 - left lobe of the liver, 9 - small intestinal coils

the 8th week of gestation, and the most widely held view regarding the etiology of CDH, is that there is a failure of this canal to close.[2]

CDH is accompanied by pulmonary hypoplasia (most obvious on the ipsilateral side but also seen on the contralateral side[2]), lung immaturity and left heart hypoplasia (in left-sided hernias[2]).[1] The intra-acinar arteries have an abnormally high smooth muscle content.[2] These factors lead to persistent pulmonary hypertension in the newborn.[1,3] The presence of pulmonary hypoplasia
is the most important determinant of fetal survival.\textsuperscript{[3]} In general, the mortality rate in large studies has been about 50%.\textsuperscript{[1]}

The detection rate of CDH by USG ranges from 18 to 87% (59%).\textsuperscript{[3]}

The usual contents of a left-sided diaphragmatic hernia are loops of the small intestine; the stomach, colon and spleen are frequently present, whereas the pancreas and liver are rarely present.\textsuperscript{[1]} In two-thirds of the cases of left-sided (posterolateral) Bochdalek hernia, the left lobe of the liver herniates into the chest to varying degrees.\textsuperscript{[3]} Identification of liver herniation is important as it is one of the indicators of a less favorable prognosis and generally signifies a large defect and one much more difficult to repair. If the stomach is seen posteriorly or in the mid-chest, it implies herniation of the liver. The presence of vessels leading to the left lobe of the liver in the chest, as well as a curvature of the umbilical vein to the left are both indicators of liver herniation.\textsuperscript{[3]} In 10–20% of patients with Bochdalek hernias, the presence of a parietal-pleuroperitoneal sac (hernial sac) can be demonstrated.

References


Source of Support: Nil, Conflict of Interest: None declared.