Obstetrics

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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...
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

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.[3] In general, the mortality rate in large studies has been about 50%.[1]

The detection rate of CDH by USG ranges from 18 to 87% (59%).[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.[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.[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.[3] In 10–20% of patients with Bochdalek hernias, the presence of a parietal-pleuropitoneal sac (hernial sac) can be demonstrated.

References


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