CASE REPORT

Fetal Massive Pericardial Effusion as a Sign of Bilateral Diaphragmatic Agenesis: A Case Report

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ABSTRACT

Congenital diaphragmatic hernia (CDH) is an unusual fetal malformation that requires early diagnosis and treatment. Commonly, presentations are in the left side (85%), followed by right (13%) and only very few (2%) bilateral. The last one is a severe condition associated with high mortality rates, and very rarely these cases survive for surgical treatment. We describe the case of a 34-week male fetus with pericardial effusion in transabdominal ultrasound. Pericardiocentesis was performed, and the presence of liver was noticed at the thoracic cavity, confirming a diaphragmatic hernia. The newborn presented acute respiratory distress and died 1 hour after birth. Necropsy revealed complete diaphragmatic agenesis, pulmonary hypoplasia, and incomplete intestinal malrotation. Survival of these patients depends on cardiopulmonary function. Bilateral agenesis of the diaphragm associated with incomplete intestinal malrotation is a rare entity, and its significance requires further research.

Keywords: Diaphragmatic, Fetal diaphragm agenesis, Fetal massive pericardial effusion, Fetal medicine, Fetal pericardial effusion, High risk pregnancies, Maternal fetal medicine, Perinatal outcome, Prenatal diagnosis, Ultrasonography.

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CASE DESCRIPTION

A 22-year-old woman with a 34-week and 2-day pregnancy and a 36-hour premature rupture of membranes was admitted at labor room. Expectant management with antibiotic and corticosteroid therapy was provided. The patient was previously diagnosed with fetal hydrothorax and an ultrasound was performed, showing active heart, surrounded by liquid in the entire thoracic cavity apparently as a result of a pericardial effusion, which was classified as severe and considering the possibility of a diaphragmatic hernia (Fig. 1). Pericardiocentesis was performed due to imminent mortality risk, extracting 45 mL of nonhematic, yellow, serous fluid. Lung tissue severely compressed (Fig. 2). During the procedure, the presence of liver was noticed at the chest cavity, confirming a diaphragmatic hernia, a right one by the presence of liver.

Twelve hours after the procedure, the patient started labor, concluding in vaginal delivery of a 2,355 g male newborn, with nondysmorphic facies and growth adequate for gestational age, and APGAR score 2 at first minute and 5 at 5 minutes. Intubation was performed and chest X-ray confirmed the presence of liver occupying most of the thorax (Fig. 3). The patient died 60 minutes after delivery. Necropsy was performed, revealing the absence of the entire diaphragm muscle, concluding as a bilateral diaphragmatic agenesis associated with a massive pericardial effusion.

NECROPSY

Informed consent form was granted for necropsy. Internal examination revealed distended pericardium showing herniation of the hepatic dome (Fig. 4), at the opening it revealed residual hydropericardium of 5 mL, heart displaced to the left side, with atrioventricular agreement and normal pulmonary and systemic drainages. Elevated hepatic dome, covered by a thin membrane (Fig. 5) without evidence of diaphragmatic muscle fibers except in the crural region (no communication of thoracic and abdominal cavity) was observed. The dome was deformed by the cardiac impression and had a throttling ring. Lungs were displaced to

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Fig. 1: Thoracic axial view

Fig. 2: Thoracic oblique view. Severe compression of lung tissue

Fig. 3: Neonatal X-ray

Fig. 4: 1, lungs; 2, thymus; 3, heart; 4, liver; 3 and 4 inside pericardium

Fig. 5: 1, pericardium; 2, liver; 3, residual crural diaphragm; 4, fibrous membrane covering the hepatic dome

the back, normal lobulated, pulmonary surface showing diffuse tan white plaques with the predominance of the right lung. The lung/body weight ratio was 0.01, confirming lung hypoplasia (normal: >0.012). In the abdominal cavity, incomplete intestinal malrotation was found, with Treitz on the left. Other organs were with normal anatomical characteristics. The histological examination of the internal organs showed congenital pneumonia and hyaline membrane, with rest of the organs without specific findings.

DISCUSSION

The prevalence of congenital diaphragmatic hernia (CDH) is approximately 1–4 cases per 10,000 live births, being 85% in the left side, 13% right side, and 2% bilateral.^{1–3} Most of the cases are sporadic.^{4,5} In an analysis of data from 31 population-based European registries over a 29-year period and including over 12 million births, the prevalence for CDH overall and isolated CDH was 2.3 and 1.6 per 10,000 live births, respectively.³ Some studies have found a higher prevalence in male fetus.^{1,3,6} The prevalence does not appear to be associated with maternal age.³

Diagnosis of CDH is based on transabdominal ultrasonography. Prenatal signs vary from an isolated diaphragm hernia to a pericardial effusion frequently misdiagnosed as hydrothorax, which

can be misleading due to the close appearance. Fetal pericardial effusion is defined as the gathering of fluid in the pericardial layer, considering 2 mm thickness as abnormal. One of the causes of fetal pericardial effusion is CDH.^{4,5}

Fetus with bilateral diaphragm hernia will have severe physiological conditions due to the entire chest compromise, the inability of adequate pulmonary development, and subsequent hypoplasia. In addition, this condition is associated with other organ dysfunction and chromosomopathies.⁷

Pericardial effusion is defined on ultrasound examination when the heart is partially or completely surrounded by liquid that is seen in all projections. If it measures less than 4 mm, it is considered small, and if it measures >4 mm, it is considered large. In cases of massive pericardial effusion, as this case, a posterior displacement of the lungs can be found on the four-chamber view (Fig. 2). The main differential diagnosis is pleural effusion, which appears on sonography, a fluid layer surrounding the lungs.⁸

Bilateral diaphragmatic hernia was an extremely unusual finding as the etiology of the pericardial effusion. This prenatal diagnosis was confirmed by the presence of the liver on the chest cavity after pericardiocentesis. Even the probability of an isolated right diaphragmatic hernia is higher, in this case bilateral diaphragmatic hernia agenesis was still considered as possible.

In this case, the pericardial effusion required further etiology determination and also immediately treatment because of the increased risk of perinatal morbidity and mortality. Prognosis is worse when associated with congenital malformations or fetal hydrops.⁹

In this case, a pericardiocentesis was timely done to prevent further hypoxia, while the fetus has continuous oxygen supply from umbilical cord, preventing further perinatal additional procedures depending on the newborn lungs, which condition at the moment of the procedure was unknown (either they could be functional or aplasic). Also, at the pericardiocentesis, the diagnosis was confirmed with the visualization of liver tissue in ultrasound, confirming the diaphragmatic hernia before birth. The etiology of the hernia was confirmed with the anatomical pathology examination.

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