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Natural history of a complex congenital diaphragmatic hernia

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ABSTRACT

The main complication in the case of congenital diaphragmatic hernia (CDH), which results from the invasion of the abdominal viscera in the thoracic cavity, is represented by the insufficient development of the pulmonary parenchyma (pulmonary hypoplasia, with the increase of the vascular resistance, and finally pulmonary hypertension). Besides pulmonary hypertension, the fetus can also develop cardiac failure in various degrees. Usually, this occurs when the arterial canal closes. We present a completely prenatal diagnosed case of left diaphragmatic hernia of the stomach, liver, and intestinal loops associated with left pulmonary agenesis and stenosis of arterial duct. The important right shifting of the heart in the right hemithorax detected on second trimester ultrasound screening has complicated in the third trimester with pericarditis, pleurisy and heart failure. The magnetic resonance imaging (MRI) examination at 31 weeks of gestation confirmed the ultrasound findings and offered more detailed prognostic elements of the anomaly. Autopsy of the stillbirth sustained MRI diagnosis regarding the absence of the left lung parenchyma and stenosis of the arterial duct. The natural history of this complex malformation underlies the importance of the collaboration of multiple specialists in maternal-fetal medicine, fast fetal MRI, fetal therapy, prenatal genetics, and fetal pathology.

Keywords: congenital diaphragmatic hernia, ultrasound, MRI, cardiac failure, pulmonary hypertension, stenosis of arterial duct

INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a rare disorder with a mortality rate between 30% and 50% (1). Prevalence of the disease is around 3-3.6/10,000 live births (2). The Fetal Medicine Foundation considers the incidence to be around 1/4000 birth (3). The disease is known for causing pulmonary hypoplasia (4) in different degrees, which determines the alteration of pulmonary vascularization and pulmonary surfactant production (5). It also represents about 8% of all fetal malformations (6). A study published by the International Society of Ultrasound in Obstetrics and Gynecology (ISUOG) in 2002 that gathered data from 12 European countries through a rigorous screening program found a prenatal detection rate of 59% from a total of 187 cases that were diagnosed with CDH at birth (7). Authors consider this to be a high detection rate (8).

Three types of CDH are described as follows: posterolateral Bochdalek hernia (80-90% of cases), anterior Morgagni-Larrey hernia, and the central hernia. Of all the Bochdalek hernias, 85% occur on the left side, 10% on the right side, and only about 5% are bilateral (9). Fetal endoscopic tracheal occlusion (FETO) for severe isolated left congenital diaphragmatic hernia has been investigated in the multi-

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center TOTAL randomized trial, and the results have been published in July 2021, the study being stopped earlier for benefit. FETO is an investigational procedure for preventing or reversing pulmonary hypoplasia, hoping to restore lung development in fetuses with prognostically poor CHD. As a result of the procedure, 6-month postnatal survival was statistically significantly greater for fetuses with severe isolated left CDH assigned to FETO than expectant management (40% versus 15%); there was a smaller and uncertain benefit in fetuses with moderate abnormality. FETO increased the risk for preterm birth threefold (10,11). Even if these findings should help in counseling appropriate candidates who have a fetus with isolated left fetal CDH, the availability of FETO procedure in maternal-fetal units of our country is in the development process. We present the unusual natural history of a case of complex, nonsyndromic severe left congenital diaphragmatic hernia. Inclusion of the patient in this study followed after approval from the ethics committee and the signing of a formal consent by the patient.

CASE PRESENTATION

The case that we are discussing is about a fetus from the first-time pregnancy of a 29-year-old without a notable patient history. The pregnancy was confirmed early on (in the first weeks), the mother presented a negative non-invasive prenatal test for fetal aneuploidies, and the first-trimester ultrasound showed no notable findings. The defect was found in the second trimester, at 21 weeks + 3 days, and ultrasound was repeated every 2 weeks henceforth, in concordance with the actual guidelines. At 30 weeks, the MRI examination was also performed. The mother prematurely gave birth at 32 weeks + 3 days of gestation. The baby was stillborn.

The ultrasound examination was performed on high-end devices, with dedicated software for the obstetrics examination, equipped with a convex transducer with a wide band and frequency between 2 and 8 MHz and B mode eco-Doppler. We adapted the power as needed, generally using a lower setting to obtain a good image depth and special resolution (so we could better describe the existing lesions).

The second-trimester ultrasound screening discovered the displacement of the heart in right thorax and an important axis diversion, but with a normal origin of the large vessels. The left diaphragm was not visualized; the stomach was seen ascending in the thorax on the left side along with the left liver lobe. The intestinal position was normal at that time. Both kidneys were in normal position and had bilateral hydronephrosis, up to 8,2 mm (fig. 1, 2).

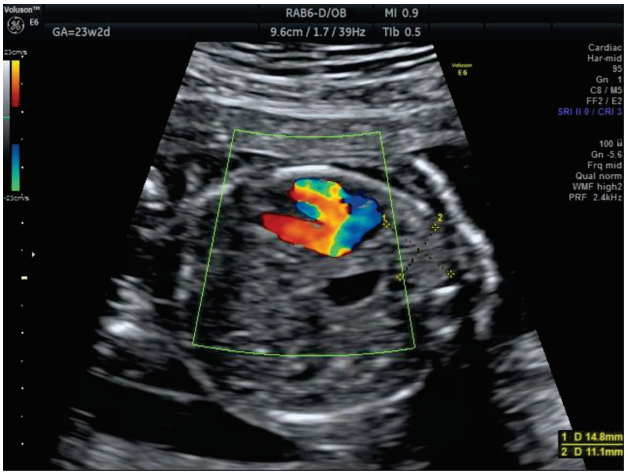

FIGURE 1. Ultrasound Doppler color image of the thorax containing the herniated stomach

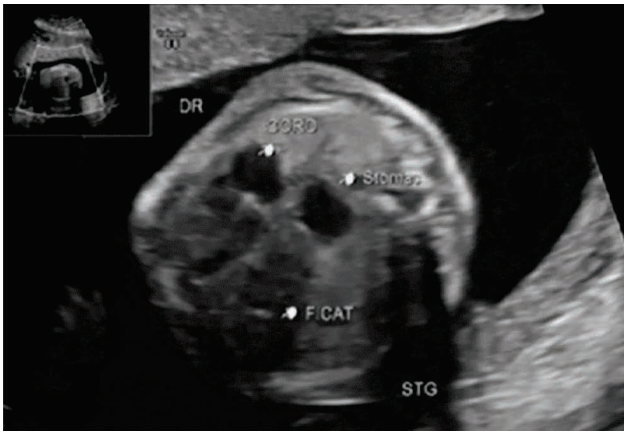

FIGURE 2. Ultrasound image of the thorax containing the herniated organs at this level

After the original ultrasound diagnosis was made, the patients were advised to undertake a follow-up MRI examination. This was made using a 1,5 Tesla MRI machine, without gadolinium contrast agents, as are the current international recommendations. In addition, we used body coils to enhance the image and the following sequences: Fast Imaging Employing Steady State Acquisition (FIESTA, FOW de 450/500 mm, TR of 5,2 ms, TE of 2,4 ms), Single Shot Fast Spin Echo (SSFSE, FOW de 450/500 mm, TR of 534.4 ms, TE of 160.2 ms), Diffusion Weighted Image (DWI, FOW de 450/500 mm, TR of 6.2 ms, TE of 3.1ms) and Liver Acquisition with Volume Acceleration (LAVA, FOW de 450/500 mm, TR of 6.2 ms, TE of 3.1 ms). The slice thickness was between 4 and 6 mm.

The MRI investigation performed at 31 weeks of pregnancy assesses the diaphragmatic defect that involves the whole left diaphragm. The whole left liver lobe was seen displaced in the thoracic cavity; the stomach was ascended with a median and slight right paramedian position. We could better see that all the small bowel intestinal loops are located in the left hemithorax. New findings were represented

by the identifying pericarditis and pleurisy on the right side. We could also identify ascending and transversal colon segments ascended in the thoracic cavity with a smaller caliber. There was no evidence of left pulmonary parenchyma. Also, the right pulmonary parenchyma was compressed, having a lower signal than the one corresponding to the gestational age (fig 3, 4).

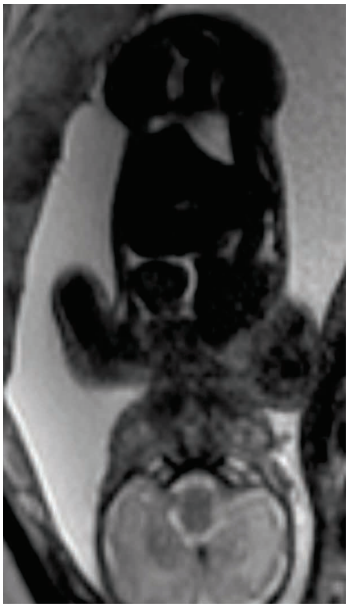

FIGURE 3. Coronal T2W MRI image that evidences the pericarditis

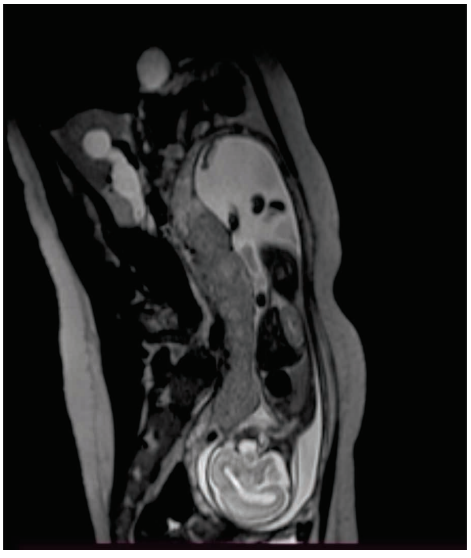

FIGURE 4. Coronal T2W MRI image that clearly shows the left liver lobe ascended in the thorax

The right pulmonary area was 312 mm^2, measured at the corresponding MRI slice to the 4-chamber view of the ultrasound. Cranial circumference was 267 mm. The lung to head ratio (LHR) (right pulmonary area/cranial circumference) was 1.16. The ratio between the calculated LHR and the estimated LHR was 38.57%, corresponding to a small pulmonary volume.

At the autopsy examination (fig. 5), it was evidenced that the heart was totally situated in the right hemithorax, the whole left liver lobe, as well as a good portion of the right liver lobe being accessioned through the left diaphragmatic defect. Almost all of the small intestinal loops were ascended in the left thoracic cavity, as well as the ascending and transverse parts of the colon. Left pulmonary parenchyma was not found (pulmonary agenesis). The pleurisy and pericarditis described on the MRI examination were also confirmed at the autopsy. Furthermore, the necrotic examination also found the stenosis of the arterial canal and established the small volume of the remaining right lung.

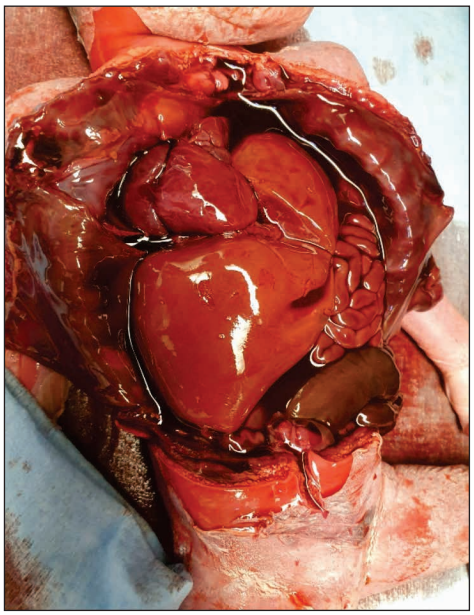

FIGURE 5. Autopsy image that shows the herniated organs ascended in the thorax

DISCUSSIONS

The analyzed case represents a severe form of complex left CDH with a progressive deterioration to the intrauterine demise of the fetus.

The early and completed diagnosis was in accordance with the specialty literature (9). The LHR showed a small lung volume that was in concordance with the autopsy finding and the poor evolution of the fetus (12). In our case, the LHR value of 1.16 sets the fetus in the 0.6-1.35 intervals that indicates a 57% survival rate (12,13).

Like in the majority of cases, the fetus suffered from the Bochdalek type of hernia, situated on the left side. The apparition of pleurisy worsened the prognosis, and the presence of pericarditis confirmed the fact that heart failure was probably the cause of death (14). The autopsy examination confirmed all the findings of the MRI, as well as the fact that heart failure appeared due to the closing of the arterial canal.

CONCLUSIONS

The correct evaluation and follow-up of the patient led to the early discovery of the malformation and established the correct prognostic. The MRI imaging completed the ultrasound findings; it brought new information regarding the severity and morphology of the defect and better characterized the hypoplastic right lung by calculating the LHR. Autopsy of the stillbirth sustained MRI diagnosis regarding the absence of the left lung parenchyma and stenosis of the arterial duct. The natural history of this complex malformation underlies the importance of the collaboration of multiple specialists in maternal-fetal medicine, fast fetal MRI, fetal therapy, prenatal genetics and fetal pathology. The development of a national specialized center for FETO is mandatory for the next step of maternal-fetal medicine in our country.

Conflict of interest: none declared
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