A rare case of severe congenital diaphragmatic hernia

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ABSTRACT

Background. Diaphragmatic hernia is a complex congenital anomaly, often associated with other structural defects with lethal potential. Congenital diaphragmatic hernia (CHD) often causes a chain of multiple complications since intrauterine development, the most frequent of which is pulmonary hypoplasia complicated, in turn, by the installation of persistent pulmonary hypertension in the newborn after birth. Although the medical and surgical management of CDH has improved considerably, morbidity and mortality rates remain high.

Case report. We presented the case of a newborn diagnosed with a massive Bochdalek-type left diaphragmatic hernia, with the ascension into the thoracic cavity of the intestine, spleen, stomach, left lobe of the liver, the antenatal diagnosis was established one month before birth, with fatal evolution within 48 hours of at birth despite the intensive support applied.

Conclusions. The presented case is a very rare case of severe CHD with a fatal outcome mainly due to the wide diaphragmatic defect that allowed the thoracic ascension of the spleen, stomach, intestinal loops, and the right liver lobe associated with hypo/aplasia of the left lung, severe displacement and compression of the heart and severe pulmonary hypertension.

Keywords: congenital diaphragmatic hernia, pulmonary hypoplasia, newborn, preoperative stabilization

INTRODUCTION

Congenital diaphragmatic hernia (CDH) (1-4 cases per 10,000 live births) represents a closing defect of the diaphragm through which the abdominal organs penetrate the thoracic cavity, compressing the lungs and diverting the mediastinum to the opposite side [1]. The diaphragm is a musculotendinous formation that develops between the third and ninth weeks of fetal life in the form of a dome, separating the thoracic cavity from the abdominal cavity [2,3]. The most common form of CDH is the Bochdalek hernia (80-85%), where the defect is found in the posterior part of the diaphragm and, in 70-75% of the cases, is located on the left side. CHD occurs on the right side in 10-15% of cases. Bilateral hernias are extremely rare and frequently fatal (5-10%) [2,4,5]. In Morgagni hernia, a less frequent type of CDH (23-28%), the hernial orifice is located in the anterior part of the diaphragm[2,4], as seen in Figure 1.

Often, CDH is diagnosed during pregnancy through routine ultrasound examination [3,6]. If not identified prenatally, the defect is frequently diagnosed immediately after birth [3,6]. A clinical picture suggestive of
CHD includes severe respiratory failure with early onset (due to inefficient gas exchange secondary to lung hypoplasia), depressed abdomen (as abdominal organs migrate into the thorax), abnormal location of heart sounds, intestinal noises heard over the thorax, absent/diminished pulmonary sounds [7–9]. The initial management of CHD is complex: stabilization (adequate respiratory support, abdominal decompression through a nasogastric tube, cardiac and circulatory support) and establishing the optimal moment for surgical correction [3,10]. There is no standardized conduct regarding the optimal moment for intervention; each center has its protocol based on its experience. Recently, the optimal moment suggested for surgical correction is when, with medical treatment and respiratory support, the pressure in the pulmonary artery is controlled [6]. Finally, the outcome depends on the severity of the lung hypoplasia and associated cardiac congenital anomalies [1,11].

**CASE REPORT**

We are presenting the case of a male infant with a birth weight of 3318 g (75-90th centile), length of 53 cm (75-90th centile), head circumference of 35 cm (50-75th centile), born from cranial presentation through C-section, performed for recent uterine scar, at 37 weeks gestation. The mother was 19 years old, and a previous pregnancy ended with the delivery by C-section of a healthy girl, 2400 g, with no other significant obstetrical history. Affirmatively, both parents are healthy, and no history of congenital defects was found in their families. The first obstetrical evaluation of the pregnant woman was one month before delivery when she was admitted with imminent preterm delivery. The ultrasound examination performed at that moment revealed the presence of fetal CHD. The pregnant woman was discharged after cessation of uterine contractions, with corrective treatment for gestational anemia.

At birth, the neonate presented severe hypoxia, with an Apgar score of 3/1 minute, 5/5 minutes. Intubation and positive pressure ventilation were initiated from birth, using 100% oxygen. According to the recommended protocol, a nasogastric tube was placed for abdominal decompression. POCUS (point of care ultrasound) lung at the delivery room confirmed extensive hernia on the left side, with severe heart shift to the lower part of the right hemithorax. After the initial stabilization in the delivery room, the newborn was transferred to the neonatal intensive care unit (NICU) for continuous care and vital signs monitoring. High-frequency oscillation ventilation (HFOV) was chosen for respiratory support. From the beginning, the infant needed high respiratory support – amplitudes of 23-25 bar, frequency of 8 Hz, mean airway pressure of 12 cm H₂O, and 100% oxygen concentration (FIO₂). Parenteral nutrition was initiated on the umbilical venous line with prophylactic antibiotic therapy (penicillin plus amikacin). Another lung and abdominal ultrasound evaluation confirmed intestinal loops in the thoracic cavity; the spleen and left liver lobe were also displaced into the thorax. Echocardiography showed a heart rotation with no defects except patent foramen ovale and
ductus arteriosus. Severe tricuspid regurgitation suggested severe pulmonary hypertension. No abnormalities were found on the head ultrasound. A thoracic X-ray (Figure 2) confirmed the results of lung and abdominal ultrasound, and the final diagnosis was: massive left CHD, with intestinal loops, stomach, spleen, and 1/3 of the left hepatic lobe herniated into the thorax and severe displacement of the heart to the base of the right axilla. Increased markers of perinatal hypoxia (creatine phosphokinase and lactate dehydrogenase) were the only abnormal lab test results found.

Selective pulmonary vasodilator (sildenafil) was used to treat pulmonary hypertension associated with lung hypoplasia. Pediatric surgical service was contacted, and the transfer of the infant was approved once respiratory and hemodynamic stabilization had been reached.

Despite the intensive care, the clinical status of the infant was continuously extremely severe, with an increased need for oxygen. The maximal peripheral saturation of oxygen (SpO₂) obtained was around 70-80% for a short period. Gradually, despite maximal respiratory support, SpO₂ continued to decrease. Unfortunately, stabilization of the infant did not succeed, and death was pronounced after almost 48 hours of intensive care. Migration of the abdominal organs in the thorax, left lung hypoplasia, and heart displacement were confirmed by the autopsy (Figure 3).
DISCUSSION

We presented the case of a newborn diagnosed one month before delivery with massive left CHD type Bochdaleck with stomach, gut, spleen, and left liver lobe migration into the thoracic cavity and fatal course in 48 hours after delivery despite intensive care and support.

The global prevalence of CHD is 2.6/10,000 births, with the mortality rate being 37.7%, in most cases during the 2-6 days of life [12]. Severe CHD cases occur in 1-4/10,000 cases [3,13]. The management of CHD is complex. If the defect is discovered prenatally – through fetal ultrasound or magnetic resonance imaging – the location of the defect, the liver’s position (into the abdomen or the thorax), and congenital cardiac defects are the main predictors of the outcome. Heart congenital abnormalities may be associated in 1/5 of the cases, while urogenital anomalies may be found in 1/4 of the cases [14]. If the birth is expected before 34 weeks of gestation, a prophylactic cure of corticosteroids is recommended [6,10].

After delivery, in CHD prenatally diagnosed, intubation and positive pressure respiratory support are recommended, aiming for SpO₂ of 80-95%, using maximal inspiratory pressures of 25 mmHg [3]. After stabilization in the delivery room, protective positive pressure ventilation and hypercarbia are recommended. Recently, HFOV has been the preferred mode of ventilation for these infants [3] as it uses very small tidal volumes, smaller than the dead space, delivered with supraphysiological frequencies over the continuous distending pressure created by the small tidal volumes [3,15].

Pulmonary hypertension, secondary to lung hypoplasia, can be managed using nitric oxide or, when nitric oxide is not available, as in our case, with sildenafil; milrinone may be added if cardiac dysfunction is associated [10]. Blood pressure must be supported, if necessary, with inotropes and/or vasoactive in trying not to aggravate the right-to-left shunting and secondary hypoxemia/hypoxia [8]. Definitive CHD management implies repositioning the abdominal viscera and surgical repairmen of the diaphragmatic defect. The surgical correction is performed after hemodynamic and respiratory stabilization: a pressure of 8-20 mmHg in the pulmo-

nary artery, normal systemic blood pressure, SpO₂ between 80-95%, lactate <3 mmol/L, urine output > 1 mL/kg/hour. Reaching all these parameters may be extremely difficult or even impossible in severe CHD, as in the presented case [3,10]. Also, CHD outcome depends to a great extent on the severity of lung hypoplasia as reduced alveolar and vascular surface negatively impacts the efficient gas exchange and aggravates hypoxia and hypoxemia [8]. In our case, the severity of the diaphragmatic defect and lung hypoplasia were decisive for the fatal outcome.

A better prognosis of CHD may be associated with cases prenatally diagnosed as this allows scheduling the birth in centers with experience in CHD management, preoperative stabilization is more manageable, and adverse effects of postnatal transportation of a critical infant are avoided, minimizing complications and optimizing the chances for survival [8].

CONCLUSION

In conclusion, we presented a rare case of CHD, extremely severe, with a fatal course due to the sizeable diaphragmatic defect that allowed stomach, intestinal loops, spleen, and left hepatic lobe migration into the thoracic cavity associated with hypoplasia of the left lung, severe displacement and compression of the heart, and severe pulmonary hypertension.

*Patient consent:*

The patient’s mother informed content was obtained before publishing the data.

*Conflict of interest:*

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