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CASE REPORT

Congenital Diaphragmatic Hernia - always need to be vigilant

Danielle de Lima Pimentel[1], Gabriel Vieira Kac[1], Jordanna de Paula Felipe Mendes[1], Lucas Monteiro Leite[1], Rhuanna Laurent Silva Ribeiro[1], Rinaldo Fábio Souza Tavares[2,3]

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Abstract

Objective: To alert pediatrics/neonatology residents about the importance of diagnostic suspicion of congenital diaphragmatic hernia (CDH) in newborns with respiratory distress, even if they are not immediately symptomatic at birth. **Case report:** We report here the case of a newborn with HDC who presented respiratory distress and cyanosis at 6 hours of age. He was in rooming-in with his mother and had no respiratory distress until then. She had received positive pressure ventilation in the delivery room. **Conclusion:** Morphological obstetric ultrasonography is essential for early diagnosis and planning of the clinical management of CDH cases from the very first care in the delivery room.

[1] Faculdade de Medicina da UFF, Departamento Materno-infantil - MMI - Neonatologia - Niterói - RJ - Brazil.
[2] Universidade Federal Fluminense - UFF, Departamento Materno-infantil - MMI - Neonatologia - Niterói - RJ - Brazil.
[3] Universidade Federal Fluminense - UFF, Faculdade de Medicina - Professor Assistente - Niterói - RJ - Brazil.

Correspondence to:
Rinaldo Fábio Souza Tavares.
Universidade Federal Fluminense - UFF, Departamento Materno-infantil. Rua Marquês do Paraná, nº 303, 2º andar, Prédio Principal, Niterói, RJ, Brazil. CEP: 24033-900. E-mail: rinaldotavares@id.uff.br

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INTRODUCTION

Congenital diaphragmatic hernia (CDH) is an embryonic malformation of a usually serious nature that requires hospitalization in a neonatal intensive care unit (ICU) and surgical intervention. In cases of CDH, there are failures in the development of the structures or in the fusion between them, forming a persistent communicating orifice that causes the protrusion of the abdominal contents into the thoracic cavity[1,2].

CDH can present in different ways: The first and most common is the Bochdalek hernia, corresponding to 70% to 75% of CDH cases, and the fissure appears in the posterolateral portion of the diaphragm[3]. The second most prevalent is the Morganni hernia, present in 20% to 25% of CDH cases, in which the fissure appears in the anteromedial portion of the diaphragm[3]. In the Bochdalek hernia, the most commonly found, there is a more typical presentation with respiratory difficulty in the first hours after birth[4].

After birth, spontaneous breathing, and especially the application of positive pressure ventilation, ends up promoting a marked distension of the stomach and intestine, resulting in a rapid increase in volume and pressure that the herniated organs apply to the lung. Thus, CDH can also be revealed not only by a decrease in abdominal volume at birth, but also by increased respiratory effort and vomiting[5]. In this context, it is important to emphasize that early symptoms of CDH are usually manifested when it is located on the left side; since obstruction of the hernia orifice by certain abdominal organs, such as the liver, could explain the delay in the onset of symptoms in right-sided hernias[4,5].

The abdomen may be excavated at birth, due to displacement of the abdominal viscera into the thorax. Another important finding in semiology is given by thoracic auscultation, which shows bowel sounds and the absence of lung sounds in the affected hemithorax. In patients with left-sided CDH, the heart may be displaced to the right due to mediastinal deviation. In less severe cases, respiratory difficulty may not be present until a few hours, days or even months after birth. In rare cases, it may go unnoticed and be accidentally discovered much later, usually in cases of smaller communications and/or "plugged" by massive organs such as the liver.

Associated anomalies are observed in approximately 50% of CDH cases and include chromosomal anomalies, congenital heart disease, and neural tube defects.

Diagnosis of diaphragmatic hernia can and should be performed in utero by means of prenatal ultrasound (USG). Prenatal ultrasound screening can identify more than 80% of CDH cases[6]. In cases of right-sided CDH, ultrasound diagnosis may be more difficult.

In these cases, magnetic resonance imaging may be requested to better characterize the tissue content[7]. In the delivery room, the diagnosis is suspected when respiratory distress and excavated abdomen are present. Confirmation usually occurs by chest/abdomen radiograph. The image reveals varied findings, such as intestinal loops or gastric bubble in the thoracic space, contralateral mediastinal deviation, atelectasis due to compression of herniated organs and presence of solid organs.

In extensive defects, numerous pneumatic intestinal loops, distended hemithorax and contralateral displacement of the heart and mediastinal structures are highlighted.

In general, children born with CDH have a mortality rate of 37.7%[8]. Syndromic neonates or those with other congenital anomalies have higher mortality rates (40.8% and 45.2%, respectively) in the first week of life when compared to those with isolated CDH (28.6%)[8].

CLINICAL CASE

A newborn female, born by cesarean section due to failure of vaginal delivery induction in a primiparous 16-year-old girl – with a gestational age of 39 weeks and 6 days, birth weight of 3470g. She presented gestational hypertension at the end of the pregnancy. The amniotic sac ruptured during the surgery with clear fluid and normodramnios. The pregnant woman had negative serology tests and prenatal care with 9 consultations in a private healthcare network. Early obstetric ultrasounds (USG) (first trimester) showed no evidence of malformations. She did not have a second or third trimester USG, including fetal morphological USG. The mother denies smoking, alcohol use or drug abuse during pregnancy. She denies using medications other than those prescribed (multivitamins and iron supplementation). The newborn received an Apgar score of 6 and 8 (first and fifth minutes) – requiring supplemental oxygen (O_2) and a fraction of inspired oxygen (FiO_2) of 30% on a mask with continuous positive airway pressure (CPAP) to improve cyanosis. After color improvement and as she remained stable, eupneic and without apparent malformations, she was referred to rooming-in.

The newborn in the rooming-in began to present, at 6 hours of life, respiratory distress and borderline saturation (90-94%), being admitted to the neonatal ICU at approximately 7 hours of life for investigation.

She was placed on nasal CPAP with 5 cm of water and FiO_2 30%; the first X-ray was requested below (figure 1):

Given the radiological changes found and the worsening clinical condition of the newborn, she was subjected to tracheal intubation and high-frequency oscillatory mechanical ventilation (HFOV).

She underwent corrective surgery on the 3rd day of life and, in the postoperative period, due to good clinical progress, she was placed on conventional mechanical ventilation - with the following postoperative radiological examination (figure 2):



Figure 1. First X-ray of the newborn at approximately 7 hours of age on nasal CPAP.



Figure 2. Post surgical correction - on the 3rd day of life.

She remained on TOT until the 7th day of life, when she was placed on nasal CPAP FiO2 30% with favorable evolution and room air at 12 days of life.

She was discharged from hospital on the 22nd day of life for outpatient follow-up.

DISCUSSION

It is estimated that Congenital Diaphragmatic Hernia (CDH) occurs in 1 in every 2500 to 3000 live births[9,10]. Affected newborns usually present clinical symptoms during the neonatal period, ranging from the first minutes to a few days of postnatal life. Rarely, they may present minimal symptoms or even be asymptomatic, being discovered accidentally later during childhood.

In those with a typical (neonatal) presentation, the severity of respiratory distress depends directly on the degree of pulmonary hypoplasia and pulmonary hypertension induced by it. In this neonatal context, hypoxia and acidosis contribute to maintaining or even worsening pulmonary hypertension. In some cases, this pulmonary hypertension is so severe that it may even be incompatible with life.

In addition to the respiratory condition, other concomitant malformations may add up and increase the morbidity and mortality of CDH. Other associated malformations are found in approximately 50% of CDH cases, including: chromosomal alterations, congenital heart disease and neural tube defects[2,10,11].

Approximately 10% of CDH cases will also present cardiac malformations[9]. The most commonly described defects are: ventricular septal defects, coarctation of the aorta, single ventricle, hypoplastic left heart and tetralogy of Fallot[2,10,11].

Clinical stabilization is essential before surgical correction. Once established, surgical correction is performed as soon as possible.

The care for patients with CDH begin in the delivery room, and therefore prenatal diagnosis through morphological ultrasound examination is essential for optimal management of these patients. An experienced neonatologist may even suspect the malformation when attending a newborn in the delivery room with ventilation difficulties, excavated abdomen and scarcity of vesicular murmurs on pulmonary auscultation, but this condition is not always evident initially[4,12].

In this case, the clinical examination did not suggest malformation and there was no morphological ultrasound examination during pregnancy. Therefore, the newborn was ventilated with positive pressure to the face in the delivery room.

When CDH is previously diagnosed, ventilating the newborn with positive pressure coupled to the face, such as through devices ("prongs") and/or masks, is avoided. If necessary, tracheal intubation is recommended to ventilate and prevent gas from escaping into the intestinal loops and thus avoid greater compression of the pulmonary and cardiac parenchyma. A gastric tube should also be inserted to prevent gas distension of the stomach and loops, and light continuous aspiration may even be performed.

In this case, the initial choice of high-frequency oscillatory ventilation (HFOV) follows the current recommendations in severe cases of CDH, in which pulmonary hypertension and persistent fetal circulation are normally present. As the clinical evolution revealed the absence of significant pulmonary hypoplasia and hypertension, this newborn was placed on conventional ventilation in the postoperative period of CDH correction.

Strategies for treating severe cases of CDH with significant pulmonary hypertension and persistent fetal circulation were not necessary, such as: deeper sedation and even neuromuscular blockade, vasoactive amines to maintain mean BP

above pulmonary pressure, inhaled nitric oxide as a pulmonary vasodilator, and in some cases indication for extracorporeal membrane oxygenation (ECMO).

The good evolution in the present case is due to the absence of significant pulmonary hypoplasia, demonstrated among other factors by adequate lung expansion (lung volume), as can be seen on the chest radiographs of the case and by the absence of other concomitant congenital anomalies.

CONCLUSION

Diaphragmatic hernia is a congenital malformation that continues to challenge the best clinical management in several neonatal units around the world. Early diagnosis is always desirable, through ultrasound examination during prenatal care (morphological ultrasound), providing the pregnant woman and her fetus with adequate neonatal ICU infrastructure and surgical team. The degree of morbidity and mortality associated with the condition depend on the location and size of the communication that can compromise normal fetal lung development. There is a wide range of possible clinical presentations, on the one hand, severe cases of pulmonary hypoplasia, severe pulmonary hypertension and persistent fetal circulation and potentially fatal. On the other hand, we can find in the literature asymptomatic cases that are accidentally discovered in older children. Between these two extremes we have the most typical presentations such as the present case, where the compression of intestinal loops present in the thorax compromises respiratory dynamics and causes symptoms in the immediate neonatal period, even though there was no significant pulmonary hypoplasia and the evolution was therefore "more benign". The interesting thing about the present case and what motivated its publication is the asymptomatic interval of a few hours, which may correspond to the moment when gaseous distension of the intestinal loops occurred and compromised respiratory dynamics. Positive pressure ventilation in the upper airway in the delivery room certainly contributed to clinical decompensation. This is why prenatal imaging diagnosis (not available in this case) is essential for better management from the delivery room onwards. Differential diagnosis of diaphragmatic hernia is essential in all newborns with respiratory distress, even if it did not begin immediately after birth. Prenatal ultrasound diagnosis is of fundamental importance for correct management of ventilation and care in the delivery room. Early diagnosis (prenatal) also allows the referral of this pregnant woman to more complex services that are prepared for multidisciplinary and comprehensive care, which are essential in these cases.

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