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Congenital Diaphragmatic Hernia: A Case Report and Literature Review

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Abstract

Background: Congenital Diaphragmatic Hernia (CDH) is a rare developmental abnormality of the diaphragm with an incidence of approximately 2.5 cases per 10,000 births. Despite advances in treatment, mortality and long-term morbidity among survivors remain high.

Case Presentation: A newborn was delivered by planned caesarean section due to prenatal diagnosis of CDH, diagnosed at 18 weeks and 5 days (Correction 3.1) during routine ultrasound. The Observed-to-Expected (O/E) Lung area-to-Head circumference Ratio (LHR) was 52%. After birth, the newborn was in respiratory distress and required mechanical invasive ventilation. Surgical repair was planned after stabilisation for the second day of life. A subcostal laparotomy was performed, and an anteromedial hernia with mostly small intestine, the left colon flexure and the spleen as content was found. The abdominal contents were reduced, and the hernia was repaired with a suture. Due to the development of a severe pulmonary hypertension, extubation was only possible on the fifth postoperative day, but reintubation was needed. The patient remained in the intensive care unit for a total of 25 days and could be discharged 46 days after birth in a good condition.

Discussion: When treating patients with CDH, the most feared complication is pulmonary hypertension, which can be life-threatening and refractory to treatment. Surgical repair should be postponed until the newborn is medically stable.

Conclusion: CDH presents a diagnostic and therapeutic challenge. When diagnosed prenatally, patients should be referred to a tertiary centre for a multidisciplinary approach.

Key Words: Congenital Diaphragmatic Hernia, Prenatal Diagnosis, Respiratory Distress, Case Report.

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1- INTRODUCTION

Congenital Diaphragmatic Hernia (CDH) is a rare developmental abnormality of the diaphragm with an incidence of approximately 2.5 cases per 10,000 births and is characterised by an incomplete development of the diaphragm, which allows abdominal content to protrude into the thorax, compromising lung development (1, 2). The aetiology is unknown but thought to be multifactorial (3, 4). CDH is most frequent on the left side (80%–85%) but can also occur on the right side (10%–15%, rarer due to earlier closure of the pleuroperitoneal opening) and bilaterally (3%, though the incidence is greater in stillborn foetuses with CDH on post-mortem examination compared to live births). The posterolateral bochdalek hernia is the most frequent (70%–75%), followed by the anterolateral Morgagni hernia (23%–28%) and central hernias (2%-7%) (3, 5-7). Despite advances in treatment, mortality and long-term morbidity among survivors remain high (8).

2- PRESENTATION OF CASE

A male infant was delivered by planned caesarean section at 38 weeks and 5 days of gestation due to prenatal diagnosis of CDH. The mother was a healthy 25-year-old primigravida. Diagnosis was made at 18 weeks and 5 days during routine ultrasound (**Fig. 1**) showing CDH with dextrocardia and an Observed-to-Expected (O/E) Lung area-to-Head circumference Ratio (LHR) of 52% (observed LHR 0.88, expected 1.67).

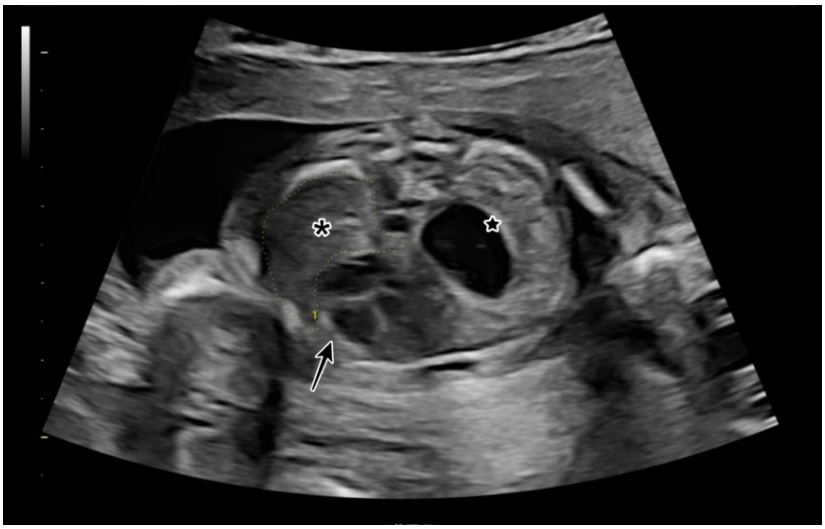

Fig. 1: Foetal ultrasound at 18 weeks and 5 days showing the right lung (asterisk), the heart in a dextrocardia position (arrow) and the herniating abdominal organs (star).

Amniocentesis performed at 20 weeks and 5 days showed a normal karyotype. Foetal echocardiography, performed at 24 weeks and 5 days confirmed dextrocardia without structural alterations and showed a hypoplastic left pulmonary artery and lung. A foetal magnetic resonance image (**Fig. 2**) was performed at 29 weeks to better describe the hernia and plan the surgical approach. Pregnancy occurred without complications. The patient was born with an APGAR score of 4/5/5 (1, 5, and 10 minutes, respectively), generalised cyanosis and SpO2 52% without spontaneous breathing movements, hypotonic, and with a heart rate of 70/min. He was manually ventilated and required a nasotracheal intubation followed by

mechanical ventilation. Hypotension (54/23 mmHg, MAP 33 mmHg) was rapidly corrected through volume resuscitation. He was admitted to the neonatal Intensive Care Unit (ICU) in a stabilised condition.

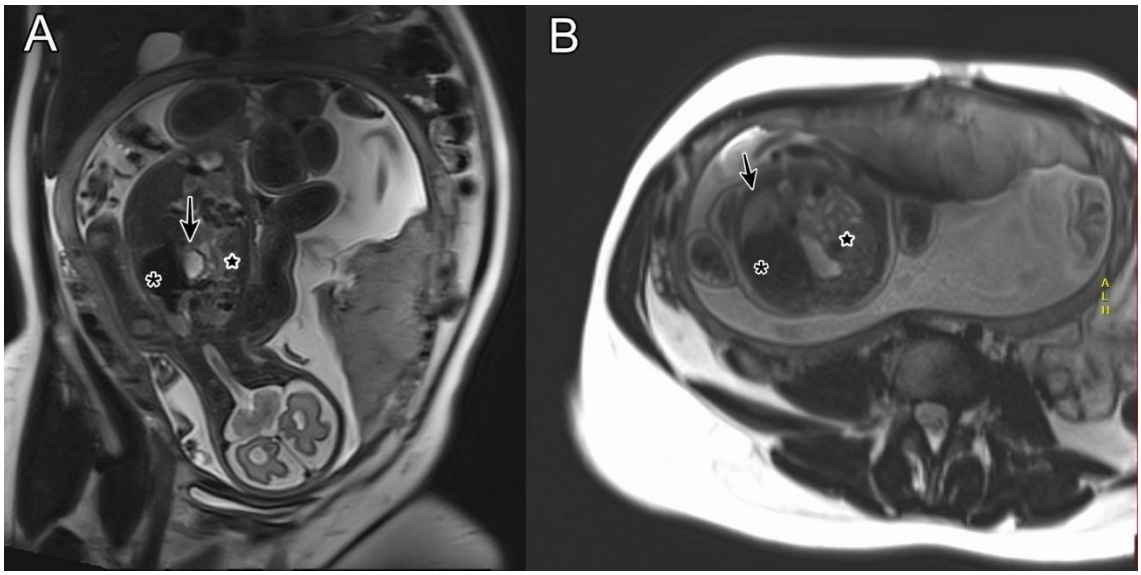

Fig. 2: Foetal MRI. Sagittal (A) and axial (B) planes showing left side CDH with intestinal content in the thoracic cavity (star), the heart (arrow), and the right lung (asterisk).

The chest radiograph (**Fig. 3A**) showed a large CDH on the left side with intestinal herniation reaching the first intercostal space, subtotal compression of the left lung and mediastinal deviation to the right side. Surgical repair was planned for the second day of life. Until then, the patient remained in the neonatal ICU.

A subcostal laparotomy was performed, and an anteromedial hernia with small intestine, the left colon flexure and the spleen as content was found. After reduction, the hernia was repaired with a suture. The procedure ended without complications, and the patient was transferred back to the neonatal ICU. A control radiography directly after surgery (**Fig. 3B**) showed a pneumothorax and effusion on the left side with a hypoplastic lung and a slight ingredient mediastinal deviation, which were treated conservatively.

Due to the development of severe pulmonary hypertension, extubation was only attempted on the fifth postoperative day. Afterwards, the patient presented clinical deterioration with respiratory distress and tachycardia; and needed to be re-intubated. A new chest radiograph was followed by computed tomography (**Fig. 4**), which did not show signs of hernia relapse but a pleural effusion on the left side without pneumothorax or infiltrates. The echocardiogram presented a severe pulmonary hypertension, which was treated with inhaled Nitric Oxide (iNO), sildenafil, and milrinone. After this treatment, the patient recovered well and was extubated 19 days postoperatively. He, then, remained stable and was transferred to the regular ward after 4 days. A control radiograph 28 days postoperatively showed an expanded left lung without pneumothorax (**Fig. 5**). The patient was discharged 46 days after birth in a good condition with sildenafil as therapy for the pulmonary hypertension. The follow-up appointment took place three months after discharge and showed no signs of pulmonary hypertension.

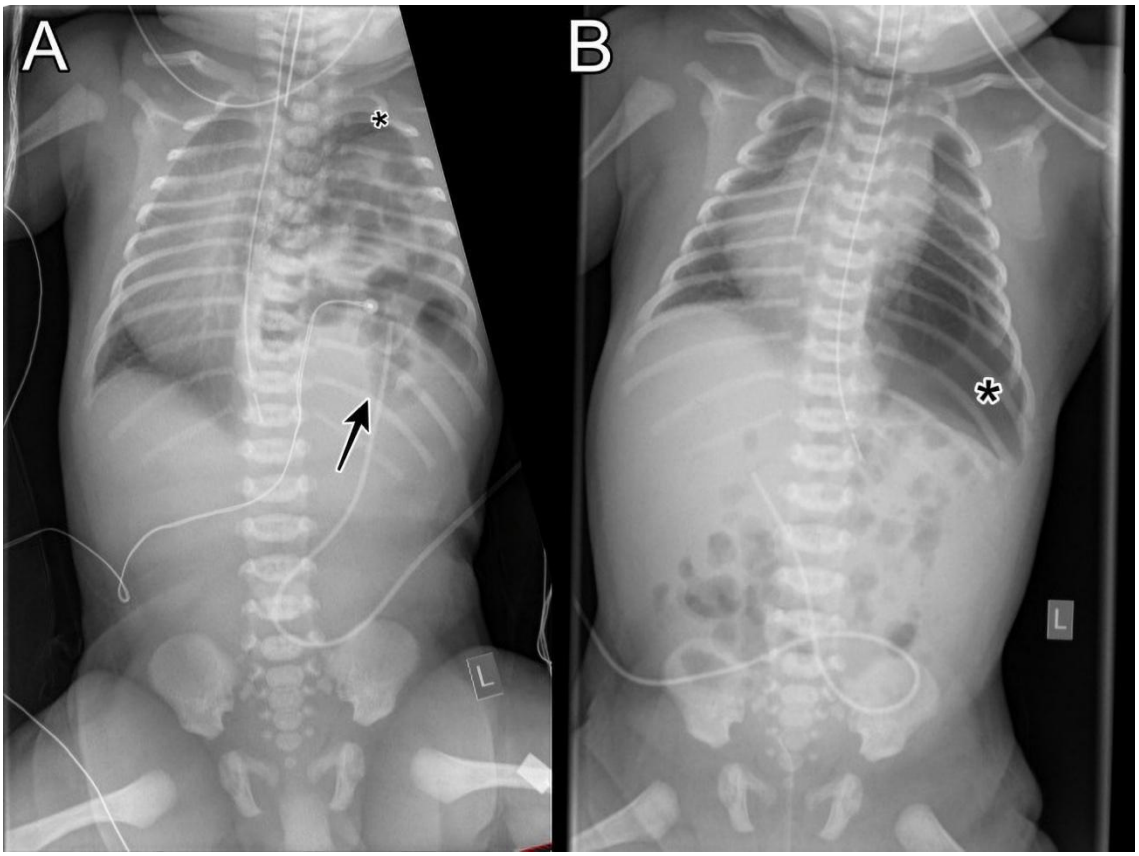

Fig. 3: Chest and abdominal radiography. Figure 3A, taken on the first day of life (preoperatively), shows left CDH with abdominal content in the thoracic cavity (arrow) and subtotal compressed left lung (asterisk) with mediastinal compression and deviation to the right side. Figure 3B, taken after surgery, shows successful repositioning of the abdominal organs in the abdominal cavity, a pneumothorax and effusion (asterisk), and a slight ingredient mediastinal deviation.

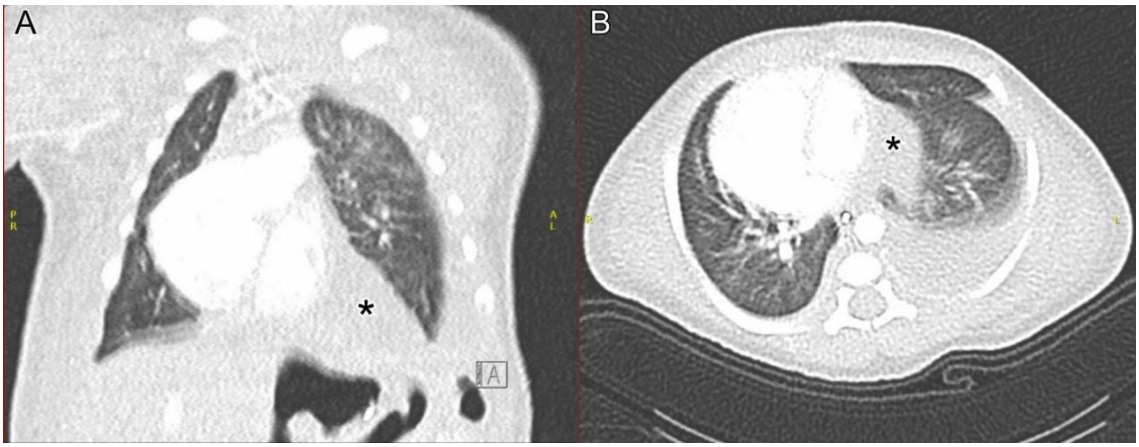

Fig. 4: Computed tomography. Coronal (A) and axial (B) planes showing the expansion of the left lung without signs of pneumothorax or hernia relapse and a pleural effusion of the left side (asterisk)

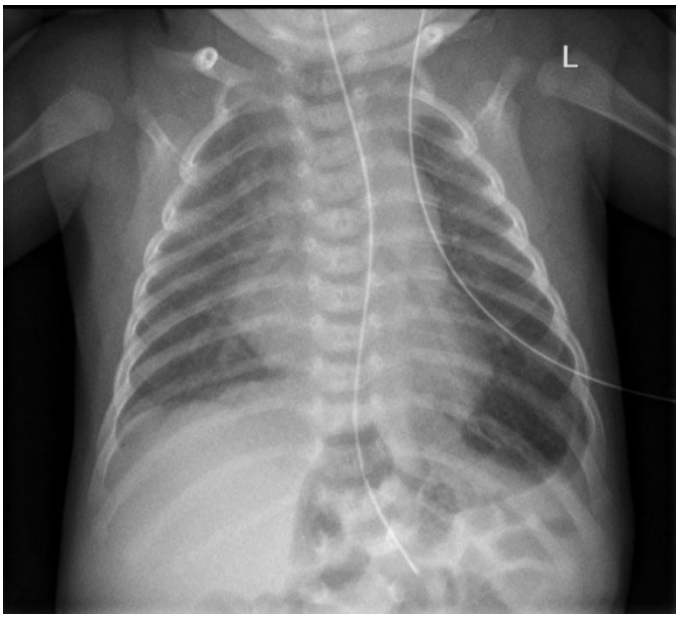

Fig. 5: Chest radiography showing an expanded left lung without signs of pneumothorax

DISCUSSION

This case presents an example of a successfully treated newborn with prenatal diagnosis of a left CDH. Approximately 20%–30% of CDH are not diagnosed prenatally, and in these cases, depending on its severity, the diagnosis may be established a few days after delivery or, in the case of small defects, only many years later (7).

During embryological development, the diaphragm normally closes during the eighth week of gestation, and a failure in this process results in a CDH. The aetiology of CDH is unknown but is believed to be multifactorial (3, 4). In approximately 50%–70% of cases, CDH occurs as an isolated defect, and the remaining cases are associated with structural abnormalities, chromosomal defects or gene disorders (3). Regardless of its aetiology, CDH leads to a compression of the lung, which shifts the heart to the contralateral side. Consequently, the lung parenchyma is susceptible to pulmonary hypoplasia and hypertension, contributing to morbidity and mortality (7).

Prenatal diagnosis is possible in approximately 60% of cases, mostly during routine ultrasound, at an average gestational age between 24 and 25 weeks. Ultrasound is considered the gold standard diagnostic tool for CDH based on direct (presence of abdominal organs in the thoracic cavity) or indirect signs (abnormal cardiac axis, mediastinal shift, polyhydramnios) (3, 9). Other radiographic and genetic tests can help identify the associated abnormalities and establish prognosis. Prenatal MRI can be used to assess the anatomy of the abdominal and thoracic cavities and the position of the abdominal organs and to estimate lung volume independent of maternal body habitus, foetal position and amniotic fluid volume (3, 9). Foetal echocardiography and karyotyping are used to screen for possible cardiac anomalies, left ventricular hypoplasia, or chromosomal abnormalities (3). Prenatal establishment of a diagnosis permits multidisciplinary counselling and referral to a tertiary care centre. In this case, a neonatal ICU doctor and a paediatric anaesthetist were allowed to be present during the birth.

When predicting outcomes prenatally, ultrasound-based LHR is the most established parameter. Since the original description of the LHR did not take gestational age into consideration, LHR is nowadays best expressed as the foetal O/E LHR. Severe pulmonary hypoplasia is suggested by O/E LHR ≤25% in left or <45% in right-sided CDH (3, 9, 10). Another prognostic predictor of the severity of CDH seems to be herniation of the liver, constituting an independent predictor of severity on left-sided hernias but with no predictive value on the right side, since the liver is almost always herniated there (11,12).

The most feared complication of CDH is pulmonary hypertension, which can be life-threatening and refractory to treatment. The use of iNO is one of the therapies of choice in infants (13, 14). In case of iNO-refractory pulmonary hypertension, sildenafil may be added or considered as an alternative to iNO (15). Another therapy option is milrinone, but this is normally used when cardiac dysfunction is associated with pulmonary hypertension (16). CDH is the most frequent indication for respiratory Extracorporeal Membrane Oxygenation therapy (ECMO) in neonates, with a survival rate of approximately 50% (17, 18).

The optimal timing for surgical treatment is controversial, but there is consensus in the literature that surgery should be postponed until the newborn is medically and physiologically stable (14, 19). Although a minimally invasive approach can be considered, the most consensual surgical approach is laparotomy with reduction of the abdominal organs and tension-free repair of the diaphragmatic defect (due to an increased risk of recurrence after minimally invasive approaches) (9). If defect closure is not possible, the use of a patch or an autologous wall muscle flap repair can be considered. The use of a prosthetic patch is associated with higher mortality; and, although less often used, autologous muscle flap repair has comparable or even superior outcomes to prosthetic patching (9,19).

Overall, and due to better diagnosis and treatment of infants with CDH, mortality rates are decreasing, but along with decreased mortality, comes an increase in CDH-associated morbidity. This may include chronic lung disease, recurrent infections, failure to thrive, adverse neurodevelopmental outcomes, musculoskeletal deformities, and gastroesophageal reflux; and these patients need long-term multidisciplinary follow-ups (9, 19).

In our case, a left CDH was diagnosed during a routine ultrasound at 18 weeks and 5 days of gestation. Since the O/E LHR was 52% without other factors of poor prognosis, the case was discussed by the interdisciplinary team and with the family, and the gestation was taken as term. After birth, and as soon as the newborn was stable, surgical repair was performed successfully by laparotomy with tension-free suturing of the diaphragm. After an initial complicated course, the newborn was discharged 46 days after birth in a good condition. There were no complications after discharge, and the follow-up after three months showed no pulmonary hypertension.

As a learning point, we believe that the description of this case shows the importance of prenatal diagnosis with a consequent early multidisciplinary approach to a complex congenital abnormality with high mortality, which ultimately led to the successful treatment of this newborn.

CONCLUSION

CDH presents a diagnostic and therapeutic challenge. When this condition is diagnosed prenatally, a

multidisciplinary approach and counselling are essential. Even though surgical treatment is successful in many patients, mortality and morbidity remain high; and further studies of this disease and development of treatment options should be pursued in order to change this scenario.

ETHICAL CONSIDERATIONS

This manuscript followed the CARE guidelines (https://www.care-statement.org).The parents were informed that data concerning the case would be submitted for publication and provided consent. All authors attest that they meet the current ICMJE criteria for Authorship.

CONFLICTS OF INTEREST

None.

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