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Congenital Diaphragm Hernia: Case Report

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Introduction

Congenital diaphragmatic hernia (CDH) is a congenital anomaly of unknown etiology characterized by the protrusion of abdominal organs into the thoracic cavity due to a defect in the development of the diaphragm during the early fetal stage [1-4]. Its epidemiological context is complex, with a prevalence of approximately 2.5 cases per 10,000 live births, which could be modified by variability in data collection and its hidden mortality in unborn cases due to its high mortality rate [1, 3, 4]. Its prevalence does not appear to be associated with maternal age [5], and most studies have not observed an association with sex, compared to some that report a slightly higher prevalence in males [2, 6, 7].

CDH is caused by a failure of the pleuroperitoneal duct to close between weeks 9 and 10 of gestation, which facilitates herniation of the midgut and, in some cases, the stomach, liver, or spleen. It is associated with high perinatal morbidity and mortality, primarily influenced by pulmonary hypoplasia and secondary pulmonary hypertension [2, 9].

Fetal lung development depends on mechanical forces (stretching or stretching) regulated by growth factors, especially those derived from fibroblasts (FGFs) [8, 9, 12]. The presence of a herniated mass alters these forces, causing pulmonary hypoplasia and impaired vascular development, accompanied by pulmonary hypertension. These conditions are the main causes of the high morbidity and mortality associated with CDH. Furthermore, up to 57% of cases present with other malformations, such as alterations in the central nervous system, cardiovascular system, genitourinary system, and chromosomal system, which worsens the prognosis and increases mortality to 80-90% [1, 2, 5].

CDH is classified according to the location of the defect, with Bochdalek hernia being the most common type (75%) with the majority located on the left posterolateral side (85%). The remaining cases include retrosternal or parasternal hernias, such as Morgagni and Larrey, as well as central hernias involving the tendinous part of the diaphragm [1-3]. Its diagnosis is mainly made in the prenatal or neonatal period, and despite the difficulties, the prognosis has improved in recent years thanks to advances in detection, neonatal management and treatments such as fetoscopic tracheal occlusion (FETO) [1, 3, 4].

The purpose of this publication is to present a clinical case of CDH, addressing key aspects such as its etiology, diagnosis, treatment, and prognosis, emphasizing the importance of a timely diagnosis to reduce associated morbidity and mortality.

Clinical Case

We present the case of a 35-week preterm newborn born prematurely, LBW 2800, height: 49 cm, cc: 33 cm, male, eutrophic, with evolving RDS and an APGAR score of 7-3-6-7. He was hospitalized for management and evaluation. A chest X-ray (Figure 1) was requested in portable anteroposterior projections (a), which showed a reduction in lung parenchyma in the right hemithorax and a shift of the mediastinum and heart to the left. A chest CT scan (Figures 2 and 3) was also performed, confirming the diagnosis of right CDH. After evaluation by a pediatric surgeon, corrective surgery was scheduled. A hernioplasty was performed, revealing a right lateral paraesophageal diaphragmatic defect, approximately 3 centimeters in size, through which the stomach, loops of small intestine, and colon ascended into the thoracic cavity. The abdominal cavity was reduced. The liver was displaced, and the stomach, small bowel loops, and colon were reduced downward. The defect was sutured directly, leaving no chest drain. The infant had a good postoperative course.

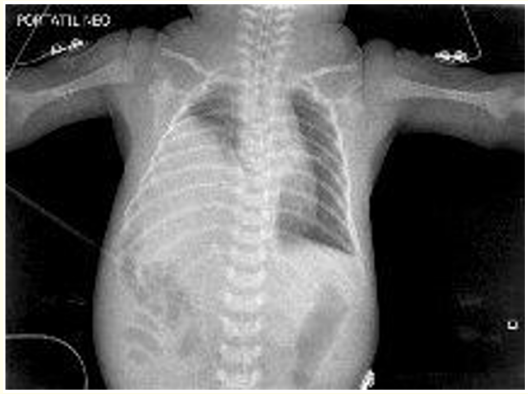

Figure 1

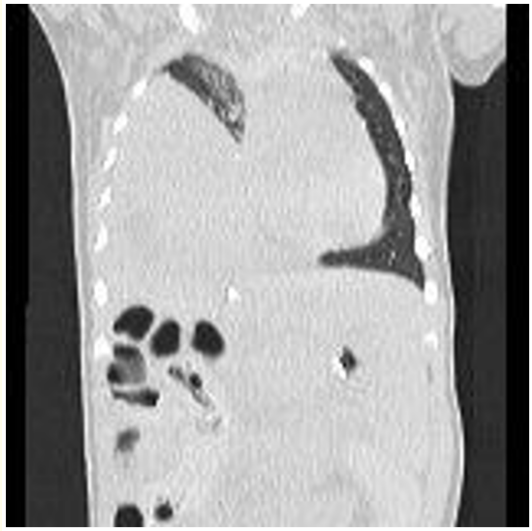

Figure 2

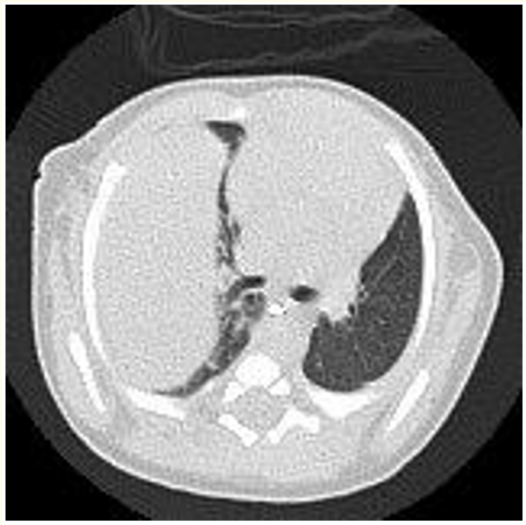

Figure 3

Discussion

The etiology of CDH remains unknown and is likely multifactorial. It occurs as an isolated defect in 50%-70% of cases, while the remaining 30%-50% are associated with structural, chromosomal, or genetic abnormalities [1, 7]. The most common malformations affect the cardiovascular system (ventricular septal defects, atrial septal defects, tetralogy of Fallot), the central nervous system (neural tube defects and hydrocephalus), and the musculoskeletal system (polydactyly, syndactyly). Cardiovascular abnormalities occur in up to 40% of cases, while central nervous system and extremity abnormalities occur in 5%-10% [1, 4, 10].

Chromosomal abnormalities such as aneuploidies and rearrangements are present in 10%-35% of non-isolated cases, with trisomies 13, 18, 21, and 45X being the most common [11]. In addition, up to 13% of cases have copy number variants (CNVs), with Pallister-Killian syndrome being one of the most common. Around 10% of cases have an underlying genetic syndrome, most notably Fryns syndrome. More than 20 genes have been associated with CDH, including COUP-TFII, FOG2, GATA4, WTI, and SLIT3 [7, 11, 12].

There is growing evidence suggesting that specific environmental factors and pathways influence the development of CDH. Vitamin A deficiency, exposure to thalidomide, and anticonvulsants have been proposed as possible triggers. Furthermore, mycophenolate mofetil and allopurinol have been associated with CDH by affecting purine biosynthesis [2, 7].

Approximately 60% of CDH cases are diagnosed prenatally [1, 5]. Diagnosis is based on prenatal ultrasound and in the vast majority of cases the abnormality is detected during screening for anomalies, so the average gestational age at diagnosis is approximately 22-24 weeks [7]. Direct signs include the presence of abdominal organs within the thoracic cavity, and indirect signs include polyhydramnios, abnormal cardiac axis or mediastinal shift [3, 4]. Left-sided CDH is identified by the presence of fluid-filled stomach and intestine in the thoracic cavity, whereas right-sided CDH is more difficult to detect due to the similar echogenicity of the herniated liver and lung, although identification of the gallbladder in the thorax confirms the diagnosis [1, 7].

A correct prenatal evaluation identifies prognostic factors, including diagnosis before 25 weeks of gestation, the presence of associated anomalies, the presence of significant hepatic herniation, severe pulmonary hypoplasia, right-sided CDH, polyhydramnios, and poor left ventricular development, among others, all of which are related to poor pre- or postnatal outcomes [2, 3, 7]. Although ultrasound

can detect it, fetal magnetic resonance imaging allows a more precise quantification of hepatic herniation, and also helps to define the anatomy, assess lung volume, and detect anomalies [4, 7]. Fetal echocardiography rules out cardiac anomalies, and fetal karyotype allows the identification of possible chromosomal abnormalities [4].

Approximately 40% of CDH cases are not diagnosed prenatally [1, 5]. Symptoms are related to pulmonary hypoplasia and hypertension; in severe cases, they usually appear immediately after birth, while in the rest, most often, they present within the first 24 hours with tachypnea, chest retraction, tachycardia, and cyanosis. Physical examination may reveal a barrel chest, a collapsed abdomen, and absent breath sounds on the ipsilateral side. The diagnosis is made by observing radiographic gas shadows from the stomach and intestines in the chest along with mediastinal shift. The presence of a nasogastric tube in the chest cavity supports the diagnosis [1, 2]. Some patients are asymptomatic, and CDH is detected late by radiological studies for other reasons. In these cases, the absence of gastric gas in its usual location suggests the presence of left-sided CDH [2]. In the postnatal period, the prognosis is strongly related to the timing of disease onset; if symptoms begin within the first 24 hours of birth, survival is close to 50% or less, improving to over 90% in later cases. Differential diagnosis includes congenital cystic lesions of the lung (cystic adenomatoid malformation, bronchopulmonary sequestration, and bronchogenic and enteric cysts) [2-4, 7].

Regarding treatment, prenatal intervention using FETO is not recommended in cases of CDH without hepatic herniation [2]. The FETO method seeks to stimulate lung development through pulmonary hyperplasia and tracheal closure, which retains pulmonary fluid and promotes cell growth and vascularization. However, it also reduces the number of type 2 pneumocytes and surfactant production, so the balloon is removed between 33 and 34 weeks of gestation [2, 7].

CDH is not considered a surgical emergency, so if diagnosed in the neonatal period, surgery is deferred until respiratory and hemodynamic stabilization is achieved in the Intensive Care Unit [1, 4, 7]. Management of pulmonary hypertension includes low-pressure mechanical ventilation to prevent pneumothorax and muscle paralysis to facilitate ventilation. Hypercapnia should be prevented and treated, as it worsens pulmonary hypertension. If conventional ventilation fails, high-frequency ventilation, vasodilators, exogenous surfactant, and extracorporeal membrane oxygenation (ECMO) can be used [1-4, 7].

CDH surgery is performed after the baby has been stabilized using criteria such as mean arterial pressure (MAP), preductal saturation, lactate, and urine output [1]. The procedure is performed through an abdominal incision, repositioning the herniated organs and closing the defect, either directly or with a graft if necessary. Complications include infection, patch erosion, thoracic deformity, restrictive lung disease, and recurrence [2, 7].

Approximately 7% of patients undergoing primary repair experience recurrence within the first year. Several criteria can be considered to predict prognosis before surgery: Blood pH <7, partial pressure of carbon dioxide >100 mmHg, the presence of a stomach in the chest, and the development of pneumothorax are indicators of poor prognosis [2].

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