Case Report

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Delayed Presentation of Left-Sided Congenital Diaphragmatic Hernia in a Term Newborn: A Case Report

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ABSTRACT

Introduction: *Congenital diaphragmatic hernia (CDH) is a rare anomaly characterised by a diaphragmatic defect allowing abdominal organ herniation into the thoracic cavity, resulting in pulmonary hypoplasia and hypertension. Although most cases are detected antenatally, delayed presentation occurs in approximately 5-25% of cases.*

Case Report: *A male term newborn with normal prenatal imaging and genetic evaluation developed mild respiratory symptoms within the first hours of life, initially attributed to early-onset sepsis. On day three, acute respiratory deterioration with asymmetric chest expansion and absent left-sided breath sounds prompted urgent chest radiography, confirming left-sided CDH. Surgical repair on day five revealed herniation of the transverse colon, omentum, and spleen, with primary closure of a 3-4 cm defect. The postoperative course was uneventful.*

Conclusion: *This case highlights the diagnostic challenges of late-presenting CDH, underscoring the importance of maintaining a broad differential diagnosis in neonates with sudden respiratory deterioration, even following reassuring prenatal evaluation.*

Keywords

Congenital diaphragmatic hérnia, Delayed presentation, Neonatal respiratory distress.

Introduction

Congenital diaphragmatic hernia (CDH) is a rare congenital anomaly, with an estimated incidence of approximately 1-4 per 10,000 live births, characterized by a developmental defect of the diaphragm that allows abdominal organs to herniate into the thoracic cavity, resulting in varying degrees of pulmonary hypoplasia and pulmonary hypertension [1-3].

Advances in prenatal imaging have led to an increase in antenatal CDH diagnoses, most commonly during the second trimester via ultrasound and/or fetal magnetic resonance imaging, detecting over 60% of fetuses with right-sided defects and more than 80% of those with left-sided CDH [1,6]. Nevertheless, some cases may remain undiagnosed until after birth.

Case Report

A male newborn was delivered at 37 weeks and 2 days of gestation following a pregnancy with normal maternal serologies. Fetal ultrasounds had revealed short long bones, and an amniocentesis performed at 23 weeks of gestation, including array analysis and a skeletal dysplasia gene panel, showed no abnormalities.

Delivery was by cesarean section due to nonreassuring cardiotocography. There was prolonged rupture of the membranes with clear amniotic fluid, and maternal peripartum fever was documented. The Apgar scores were 10 at one and five minutes. The birth weight was 2665g. Physical examination at birth was unremarkable, with no dysmorphic features identified. A weak sucking reflex was noted.

Within the first 12 hours of life, the newborn developed mild jaundice, tachypnea, and intermittent nasal flaring, prompting a septic workup. C-reactive protein level was 1.46 mg/dL, and empirical ampicillin and gentamicin were initiated for suspected early-onset neonatal sepsis.

From days one to three of life, the newborn remained clinically stable with spontaneous ventilation without supplemental oxygen therapy. However, on day three, respiratory distress ensued shortly after feeding. A physical examination revealed marked tachypnea (respiratory rate 85 breaths per minute), global retractions, and moaning. Oxygen saturation dropped to 75% on room air. Asymmetrical chest expansion was observed, with hyperinflation of the left hemithorax. Cardiac auscultation revealed normal S1 and S2 sounds without murmurs. Pulmonary auscultation revealed markedly reduced breath sounds over the left hemithorax. The abdominal examination was unremarkable.

Chest radiography (Figure 1) revealed a congenital left-sided diaphragmatic hernia. A nasogastric tube was inserted for gastric decompression, and elective endotracheal intubation was performed without complication. The patient was transferred to the pediatric surgery unit, where surgical repair was performed on day five of life.

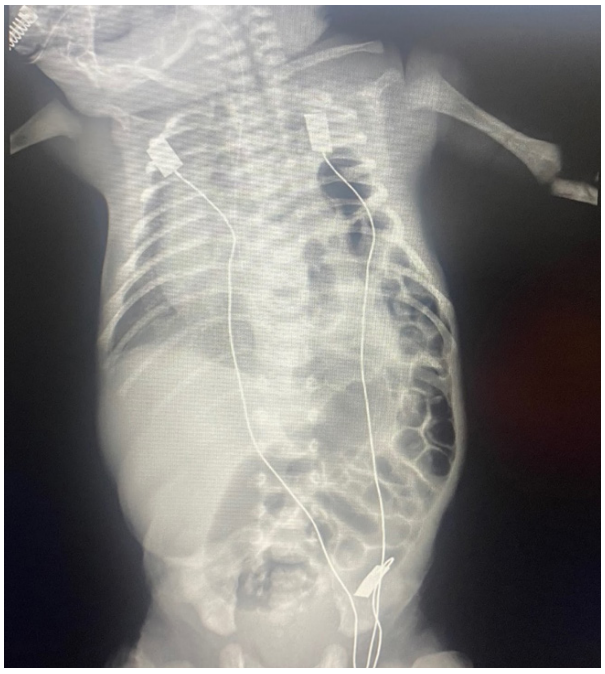

Figure 1: Chest X-ray on day 3 of life demonstrating left-sided congenital diaphragmatic hernia.

Inspection of the left thoracic cavity revealed herniation of the transverse colon, omentum, and spleen, with no evidence of ischemia or organ injury. The left lung appeared well-aerated, and the diaphragmatic defect, measuring 3-4 cm, was primarily closed.

The postoperative course was uneventful. The newborn was discharged on day eleven of life tolerating full enteral feeding and remains under regular multidisciplinary follow-up.

Discussion

CDH was first described in 1754 by Hunter and Mc Cauley as the protrusion of abdominal organs into the pleural cavity, or, conversely, of thoracic organs into the abdominal cavity, due to incomplete closure of the pleuroperitoneal canals [4]. The etiology remains unclear, but it is thought to be multifactorial, involving genetic, environmental, and nutritional factors [2,3].

Depending on the location of the diaphragmatic defect, CDH can be classified into different types. The most common form is the posterolateral *Bochdalek* hernia, accounting for 70-90% of cases and occurring predominantly on the left side (~80%) [1-3]. *Morgagni* hernias result from defects in the anteromedial portion of the diaphragm, representing 20-30% of cases [1-3]. Central hernias are rare (2-5% of cases) [2,3]. Bilateral diaphragmatic hernias are uncommon and associated with a poorer prognosis [1].

Thanks to advances in prenatal imaging, about two-thirds of CDH cases are detected antenatally [1,6]. However, some cases remain undetected until after birth [1]. Delayed presentation has been reported in 5-25% of cases [4]. Affected neonates typically present with acute respiratory distress, sometimes after an initial stable period of 24 to 48 hours, as was observed in this case [1]. Large defects manifest with severe respiratory distress, cyanosis, decreased breath sounds on the affected side, displaced cardiac sounds, and a scaphoid abdomen [1,4]. In contrast, smaller defects may present later with milder symptoms [4].

Differential diagnoses include bronchopulmonary sequestration, congenital pulmonary airway malformation, bronchogenic cysts, bronchial atresia and teratomas [1]. Identifying intra-abdominal contents within the thoracic cavity on a chest radiograph distinguishes CDH from other thoracic lesions [1,4].

Initial management includes early placement of a nasogastric tube for gastrointestinal decompression and prompt endotracheal intubation performed when respiratory distress is present [5]. Lung protective ventilation strategies are essential, targeting preductal oxygen saturations of 85-95%, peak inspiratory pressure below 25 cm H_2O, positive end-expiratory pressure of 3-5cm H_2O, and permissive hypercapnia ($PaCO_2$ of 45-60 mmHg) [2,5,7]. High-frequency ventilation may be considered if conventional ventilation is insufficient [5].

Advances in ventilatory support and surgical techniques have increased overall survival rates to 60-70% [2,7,8]. Surgical repair is generally delayed for at least 48 to 72 hours after birth to allow for cardiorespiratory stabilization and pulmonary vascular adaptation [2]. In cases of severe pulmonary hypertension, it may be further postponed until adequate hemodynamic control is achieved [5].

CDH occurs as an isolated defect in 50-70% of cases [1,2]. The

remaining cases are associated with additional anomalies, including congenital heart defects, urinary tract abnormalities, spinal defects, Pierre-Robin syndrome, esophageal atresia, omphalocele, and choledochal cyst [2,3]. Chromosomal abnormalities and single-gene mutations, such as those involving the *GATA4* and *LRP2* genes, have also been described [2]. Screening for associated anomalies and genetic counseling are therefore essential.

The prognosis is influenced by several factors, including gestational age, defect size, degree of pulmonary hypoplasia, presence and severity of pulmonary hypertension, liver herniation, and the presence of associated anomalies [1,2,6,8]. In this case, favorable prognostic factors included a moderate-sized diaphragmatic defect (~4 cm), left side herniation, and an absence of liver herniation.

Long-term multidisciplinar follow-up remains essential given the significant morbidity beyond the neonatal period [1,2,5,8].

Conclusion

This case highlights the diagnostic challenges of late-presenting CDH following reassuring prenatal imaging. Initial stability followed by acute respiratory deterioration illustrates how this condition may mimic early-onset sepsis, underscoring the importance of maintaining a broad differential diagnosis when confronted with sudden respiratory distress, particularly in the presence of asymmetric chest findings [4].

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