Case Report

Congenital Diaphragmatic Hernia Diagnosed Postnatally Despite Prenatal Suspicion of Duodenal Atresia: A Case Report and Review of Perinatal Management Strategies

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ABSTRACT

Background

Congenital diaphragmatic hernia (CDH) is a developmental malformation characterized by incomplete diaphragm formation and is associated with significant neonatal morbidity and mortality. Auxiliary imaging modalities, such as ultrasound and magnetic resonance imaging, as well as molecular studies, have improved both treatment and prognosis. However, atypical presentations may delay diagnosis and hinder perinatal care planning.

Case Presentation

We report the case of a term neonate who was prenatally suspected to have duodenal atresia and presented at birth with severe respiratory distress requiring immediate medical intervention. Initial stabilization included positive pressure ventilation, which led to progressive clinical deterioration. A chest radiograph performed shortly after birth revealed findings consistent with a left-sided Bochdalek hernia. The patient underwent prompt endotracheal intubation and surgical repair in a tertiary care center. Despite the extensive diaphragmatic defect, the clinical course was favorable, and the patient was discharged without complications 24 days later. ***Discussion:*** This case highlights the diagnostic challenges in prenatal imaging of CDH, particularly when other gastrointestinal anomalies are suspected. It underscores the importance of maintaining a broad differential diagnosis in cases of prenatal abdominal findings and supports the need for postnatal reevaluation when initial diagnoses are inconclusive. The favorable outcome in this patient was largely attributable to the availability of a multidisciplinary team and timely surgical intervention. Current literature supports that early referral to specialized centers and adherence to standardized ventilatory and surgical protocols are critical for improving survival in CDH.

Conclusion

This report reinforces the importance of high-resolution prenatal imaging, multidisciplinary perinatal planning, and early postnatal intervention in the management of CDH. Even in the absence of a definitive prenatal diagnosis, coordinated care in a specialized setting can significantly enhance outcomes. The findings contribute to clinical strategies for handling atypical CDH presentations and optimizing neonatal care pathways.

Keywords

perinatal management; Bochdalek hernia; congenital diaphragmatic hernia; prenatal diagnosis, neonatal surgery.

AUTHORS'S CONTRIBUTION

Data gathering and idea conception: Luis Alberto Pérez Covarrubias, María Valeria Jiménez-Báez. The original idea and clinical focus of this study were proposed by Pérez-Covarrubias, Jiménez-Báez contributed substantially to the case documentation and literature review.

Study design: María Valeria Jiménez Báez, Luis Alberto Pérez Covarrubias, Ernesto Baltazar Herrera, Víctor Manuel Espinoza Gutiérrez. The study design and methodology were developed with input from the cardiology and pediatrics departments to ensure multidisciplinary accuracy in clinical interpretation.

Data gathering: Luis Alberto Pérez Covarrubias, Ernesto Baltazar Herrera, Adrián Valle-Partida. Clinical data were compiled from hospital records and imaging archives, with rehabilitation and perinatal insights contributed by Valle-Partida.

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INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a developmental defect caused by incomplete diaphragm formation, resulting in herniation of abdominal organs into the thorax and subsequent pulmonary hypoplasia and persistent pulmonary hypertension (PPHN), which are strongly linked to increased neonatal morbidity and mortality[1]. The diaphragm develops between the fourth and twelfth weeks of gestation, a period considered critical for the emergence of secondary embryological anomalies.

The most common form of CDH, accounting for approximately 75% of cases, is the Bochdalek hernia, typically located in the posterolateral region and predominantly on the left side in up to 85% of cases. In contrast, anteriorly located hernias—referred to as Morgagni hernias—and central diaphragmatic hernias are less common, with a combined estimated incidence ranging from 2% to 28% of cases[2].

The size of the diaphragmatic defect is directly proportional to the extent of abdominal organ herniation—such as the stomach, spleen, liver, and small intestine—into the thoracic cavity, thereby significantly interfering with normal lung development. The estimated incidence of CDH ranges from 1 in every 3,000 to 5,000 live births. Post-diagnosis survival exceeds 60%, but outcomes largely depend on the severity of the defect and the availability of medical resources[3,4].

An accurate prenatal diagnosis is essential for effective perinatal care planning. Therefore, obstetric ultrasound should be performed during the second trimester of pregnancy to identify the presence of abdominal organs within the thoracic cavity and to assess lung development. Additionally, if the ultrasound findings are positive, fetal magnetic resonance imaging (MRI) contributes to estimating total lung volume and enhances the detection of associated anomalies, Both studies improve diagnostic accuracy and neonatal prognosis[5,6].

The Congenital Diaphragmatic Hernia Study Group (CDHSG) classification correlates clinical outcomes with defect size, with larger defects being associated with a higher risk of respiratory failure, chronic lung disease, and the need for extracorporeal membrane oxygenation (ECMO)[7].

Screening for CDH facilitates timely referral to high-complexity hospitals capable of providing comprehensive and multidisciplinary management, which improves survival rates and reduces complications associated with the condition[8,9].

This report aims to present a clinical case of congenital diaphragmatic hernia diagnosed and managed at the General Hospital of Playa del Carmen. The report highlights critical aspects of prenatal diagnosis, perioperative management, and clinical outcomes, emphasizing the importance of early identification and multidisciplinary care within a secondary-level hospital to optimize neonatal prognosis.

CASE REPORT

A female neonate was born to a 24-year-old mother with adequate prenatal care (six visits since the first trimester). During the third trimester, the mother had a urinary tract infection treated with an unspecified medication. Ultrasound at 15 weeks showed breech presentation, probable dextrocardia, and a “double bubble” sign suggestive of duodenal atresia. Due to fetal bradycardia (95 bpm), an abdominal cesarean section was performed.

On September 8, 2022, the infant was born with generalized cyanosis, poor respiratory effort, and heart rate below 100 bpm. One cycle of positive pressure ventilation (PPV) was administered with a favorable response, followed by supplemental oxygen via free flow, although cyanosis recurred upon removal. Patency of the choanae, esophagus, and anus was confirmed; the umbilical cord had two arteries and one vein without bleeding. Apgar scores were 4 and 7 at 1 and 5 minutes, respectively; Silverman-Anderson score was 3/4, indicating respiratory distress. Gestational age by Capurro method was 37 weeks. Weight was 2560 g,

length 48 cm, and head circumference 31 cm.

Physical examination revealed a distended left hemithorax, normal heart sounds on the right, absent breath sounds on the left without added sounds, and a soft, non-distended abdomen without palpable masses or bowel sounds.

Due to severe respiratory distress, the patient was intubated with a 3 Fr endotracheal tube and placed on mechanical ventilation (PIP 16 cmH2O, PEEP 5, rate 40/min, FiO2 30%), achieving oxygen saturation above 90%. Arterial blood gas showed pH 7.32, PCO2 34 mmHg, PO2 16.4 mmHg, HCO3 18.7 mmol/L, base excess -8.6 mmol/L, and lactate 2.4 mmol/L.

Chest X-ray revealed a diaphragmatic hernia (Figure 1). Echocardiogram showed a structurally normal heart, patent ductus arteriosus (PDA) with left-to-right shunt, gradient 21 mmHg, preserved ventricular function, and suprasystemic pulmonary pressure.

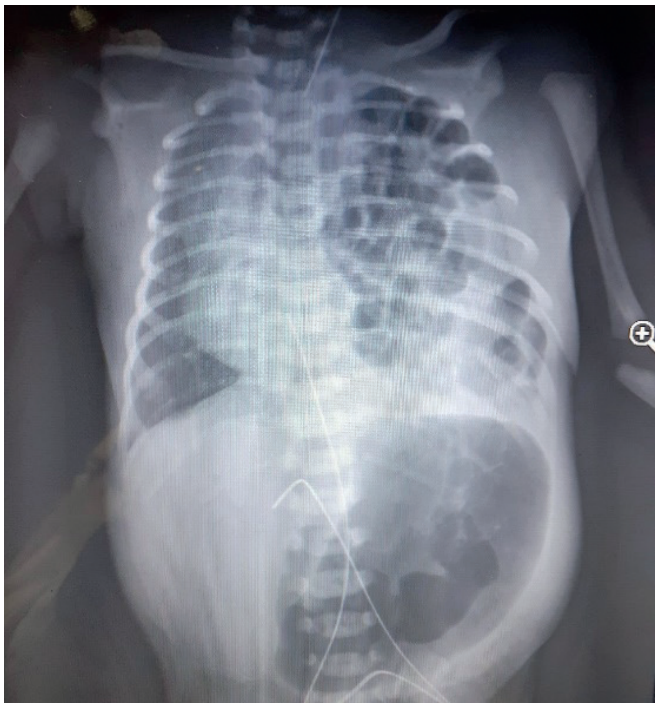

Figure 1. Chest X-ray
It observed revealed a diaphragmatic hernia

No surgical contraindications were identified. Pediatric surgery observed bilateral ventilation despite bowel sounds in the left hemithorax, indicating mild pulmonary hypoplasia with a favorable prognosis. A globular abdomen suggested a large diaphragmatic defect permitting free movement of intestinal loops between the thoracic and abdominal cavities. Given the patient's hemodynamic stability, surgical reduction of the herniated bowel and diaphragmatic repair were planned to enhance pulmonary development and mitigate pulmonary hypertension.

On September 9, an open supraumbilical laparotomy was performed. A 5 cm posterolateral defect was identified in the left hemidiaphragm containing approximately 30% of the small intestine and ascending colon without vascular compromise. Intestinal loops were reduced, and primary closure was performed using 4-0 Prolene sutures anchored to the diaphragmatic tendon to prevent tearing. Adequate closure without recurrence was confirmed, classifying it as a Bochdalek hernia. Intraoperative chest X-ray demonstrated satisfactory closure and left lung re-expansion **(Figure 2)**.

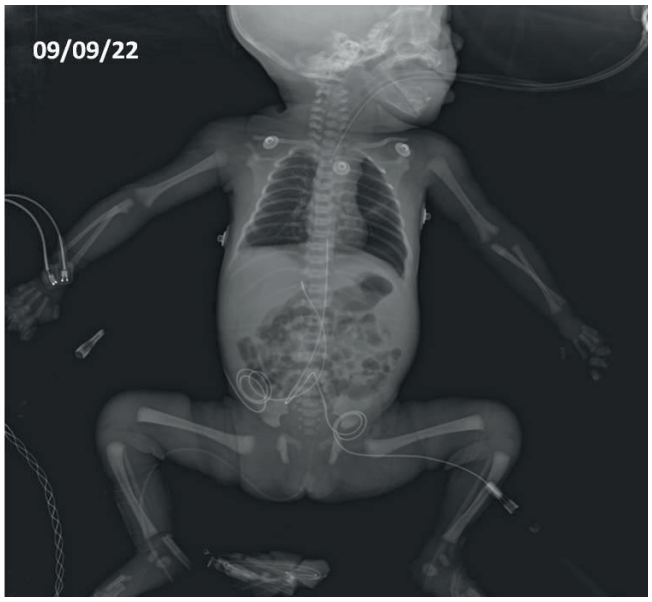

Figure 2. Chest X-ray control
It demonstrated satisfactory closure and left lung re-expansion

Postoperatively, the patient was admitted to the neonatal intensive care unit with hypothermia, non-perfusing mean arterial pressure, and bradycardia, requiring dopamine and dobutamine at 10 mcg/kg/min. Milrinone maintenance was initiated, and mechanical ventilation continued (PIP 20, PEEP 5, rate 50/min, FiO2 60%) with oxygen saturation above 95%. Antibiotic therapy with ampicillin and amikacin was started after blood cultures.

Follow-up echocardiogram on September 13 showed hemodynamic stability, decreased pulmonary pressure, opening of the left ventricle with a 3 × 3 mm mid-

trabecular ventricular septal defect (VSD) and right-to-left shunt with a 9 mmHg gradient, likely masked by extrinsic intestinal compression. The small VSD suggested possible spontaneous closure. PDA with left-to-right shunt persisted. Pharmacologic closure and loop diuretic therapy were initiated. Ventricular function remained preserved with pulmonary systolic pressure at 53 mmHg.

A left pleural effusion was detected (Figure 3), and a chest tube was placed, draining 40 ml of serosanguinous fluid. The tube was removed nine days later with no radiographic evidence of effusion (Figure 4).

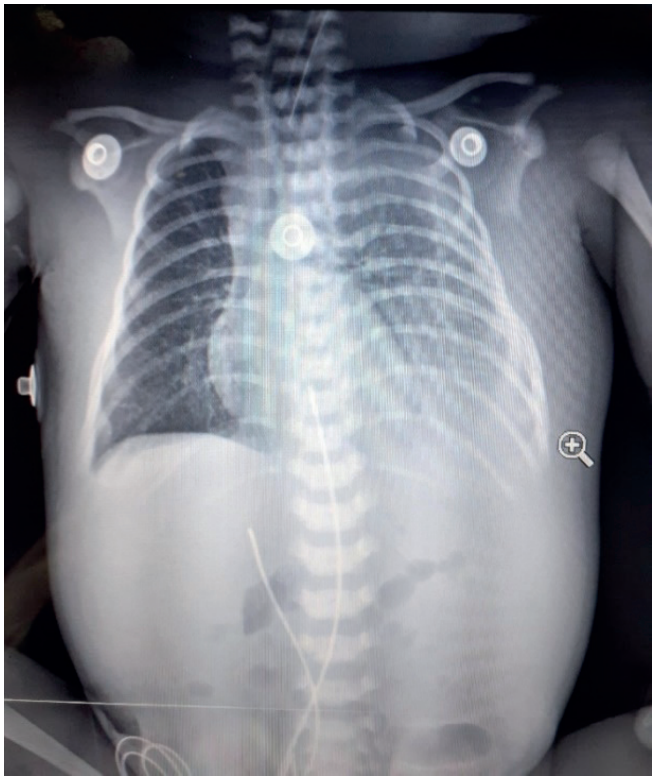

Figure 3. Chest Tube

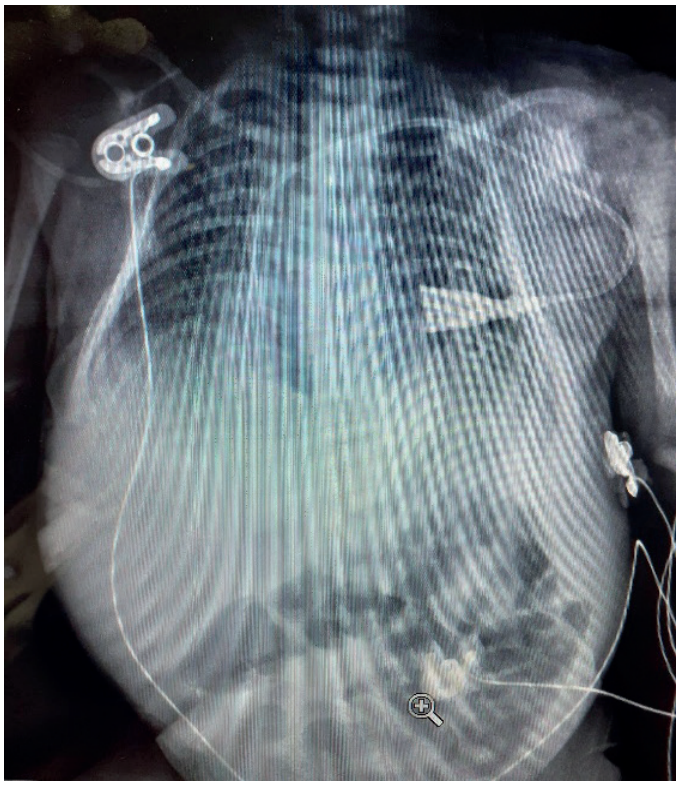

Figure 4. Chest removed tube

Left pleural effusion was detected. chest tube was placed, draining 40 ml of serosanguinous The tube was removed nine days later with no radiographic evidence of effusion

The patient required initially high ventilatory settings, with gradual weaning and scheduled extubation on September 27. She remained on a headbox for three days and free-flow oxygen for one day, discontinuing supplemental oxygen on October 1 with saturations above 95% and no respiratory distress.

Empirical antibiotics (ampicillin and amikacin) were discontinued after five days due to poor clinical response, and therapy was switched to cefotaxime and vancomycin, completing 14 days. Blood cultures were negative, while pleural fluid cultures grew Staphylococcus epidermidis sensitive to vancomycin.

The patient developed low cardiac output postoperatively, managed with dopamine (7 days), dobutamine (12 days), and norepinephrine (3 days).

The clinical course was favorable, and the patient was discharged 24 days after surgery, with outpatient follow-up by pediatric surgery.

DISCUSSION

The underlying pathogenesis of congenital diaphragmatic hernia (CDH) remains incompletely characterized. Published evidence continues to evolve, yet key aspects of prenatal and neonatal management remain areas of ongoing investigation. A widely accepted theory emphasizes the role of the

pleuroperitoneal folds (PPFs) as critical transitional embryologic structures essential for diaphragm formation, contributing to the development of both muscular and tendinous components[9]. Disruptions in the migration, proliferation, or differentiation of myofibroblastic precursors originating from the PPFs—as well as increased localized apoptosis—have been proposed as central mechanisms in the etiology of CDH[1].

CDH has an estimated incidence of 1.4 per 10,000 live births, with 70–85% of cases presenting as a left-sided posterolateral defect (Bochdalek hernia) (5). This anatomical distribution aligns with embryological vulnerability and has prognostic relevance, particularly with respect to the timing and technical feasibility of surgical repair[10,11]. Less common variants, such as anterior (Morgagni) and central hernias, must also be considered in the differential diagnosis, especially in the context of atypical findings on prenatal imaging[12].

Pulmonary hypoplasia and abnormal remodeling of the pulmonary blood vessels caused by early herniation of abdominal organs into the chest are the main causes of neonatal death. Mortality rates can reach up to 30%, even in specialized centers following standard protocols[13,14]. These outcomes underscore the necessity for comprehensive multidisciplinary approaches and long-term follow-up strategies. Fetal surgery, including early interventions such as fetoscopic procedures, continues to be explored as a potential modality to mitigate pulmonary underdevelopment[15].

Prenatal diagnosis—most commonly through second-trimester obstetric ultrasonography—is essential in all pregnancies to enable early referral to tertiary care centers. Advanced fetal assessment tools such as lung-to-head ratio (LHR), fetal magnetic resonance imaging (MRI), and genetic testing are critical for accurate prognostication and detection of associated anomalies, which occur in up to 40% of CDH cases[1]. As demonstrated in our case, limitations in access to these diagnostic resources remain a significant barrier in many global settings, particularly in low-resource regions where comprehensive prenatal evaluations and multidisciplinary planning are often unattainable.

At birth, neonates with CDH require highly specialized resuscitation protocols. Bag-mask ventilation is contraindicated due to the risk of gastrointestinal distension and exacerbation of pulmonary compromise. Instead, immediate endotracheal intubation and pressure-limited mechanical ventilation (<25 cmH_2O) are recommended to minimize the risks of barotrauma and volutrauma[10].

What are the survival prospects for these patients? In high-resource tertiary care settings, survival rates of up to 70% have been reported, directly linked to timely prenatal diagnosis, structured perinatal planning, and access to advanced ventilatory and surgical care[8]. Some studies suggest improved outcomes in cases diagnosed after 25 weeks of gestation, though this may reflect selection bias rather than a true prognostic advantage[13].

Fetoscopic endoluminal tracheal occlusion (FETO) has emerged as a potential intervention for selected high-risk cases. This procedure promotes lung growth by increasing intrapulmonary pressure and stimulating pulmonary vascular development[7,8,10]. However, FETO remains limited to select cases due to procedural risks and resource constraints, and its integration into routine management has not yet been fully established.

Postnatal care of CDH patients requires an integrated multidisciplinary approach, involving neonatologists, pediatric surgeons, cardiologists, and radiologists in centers equipped with neonatal intensive care units (NICUs), high-frequency ventilation, and extracorporeal membrane oxygenation (ECMO), as indicated. Timely transfer to specialized centers is critical for optimizing outcomes[14,15,16].

This clinical case presents a unique scenario in which prenatal imaging suggested duodenal atresia, and CDH was only diagnosed postnatally following respiratory decompensation. This highlights the importance of maintaining a broad differential diagnosis in the presence of prenatal gastrointestinal anomalies and of conducting a thorough postnatal evaluation in neonates with unexplained respiratory distress. Notably, despite the large anatomical defect, the patient had a favorable outcome, likely due to early recognition, timely surgical intervention, and availability of a multidisciplinary care team.

A key strength of this case was the immediate availability of comprehensive specialized care—including neonatal stabilization, cardiological assessment, and surgical repair—which collectively contributed to the patient's positive clinical course. Conversely, the lack of a definitive prenatal diagnosis of CDH limited pre-delivery planning and represents an area for improvement.

This case adds valuable insight to the existing literature, demonstrating that even in the absence of a conclusive prenatal diagnosis, comprehensive and timely management can result in favorable neonatal outcomes. It further underscores the need to enhance prenatal diagnostic capabilities and multidisciplinary preparedness in secondary-level hospitals, offering practical guidance applicable to resource-limited healthcare settings.

CONCLUSIONS

Congenital diaphragmatic hernia (CDH) remains a complex condition associated with significant neonatal morbidity and mortality. However, timely prenatal diagnosis using high-resolution ultrasonography is essential for comprehensive perinatal management. Early detection allows for anticipation and management of life-threatening complications such as pulmonary hypoplasia and persistent pulmonary hypertension, enabling the assembly of specialized multidisciplinary teams and ensuring delivery at centers equipped with NICUs, advanced ventilatory support, and surgical expertise.

Providing families with accurate prognostic information and therapeutic options through prenatal counseling fosters informed and timely decision-making, which directly influences survival rates and long-term quality of life. Furthermore, early referral ensures optimal use of medical resources and reduces postnatal complications.

Finally, it is imperative that health systems implement policies that promote equitable access to advanced diagnostic technologies and specialized care for high-risk pregnancies, thereby improving outcomes for both mothers and neonates.

Informed Consent

All procedures conducted in this clinical study complied with the ethical standards outlined in the Declaration of Helsinki, as well as with national and institutional regulations governing research involving human subjects. Prior to participation, written informed consent was obtained from all study participants or, in the case of minors, from their parents or legal guardians, along with the assent of the children when appropriate.

Participants were thoroughly informed about the nature, objectives, potential risks, and benefits of the research, and about how their personal data would be handled. The informed consent documents were written at an eighth-grade reading level to ensure clarity and comprehension by the target population. A colleague reviewed the document prior to IRB submission to confirm its readability. All participants or their guardians were given sufficient time to ask questions and to make a voluntary, unpressured decision regarding participation in the study.

The study was conducted by the ethical principles established in the Declaration of Helsinki. It was approved by the Ethics Committee of the Playa del Carmen General Hospital, issued on Juny 2, 2024. The confidentiality and anonymity of patients were guaranteed at all times, omitting any personal data that could allow their direct or indirect identification

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