International Journal of Life Sciences, Biotechnology and Pharma Research Vol. 14, No. 4, April 2025 Online ISSN: 2250-3137
Print ISSN: 2977-0122

DOI: 10.69605/ijlbpr_14.4.2025.119

CASE REPORT

Congenital Diaphragmatic Hernia: A Case Study

[1]Dr. Lakshya Sharma, [2]Dr. Karan Vaishya

[1,2]Junior Resident, Department of Radiodiagnosis, People's College of Medical Sciences and Research Centre Bhopal, MP, India

Corresponding author
Dr. Lakshya Sharma
Junior Resident, Department of Radiodiagnosis, People's College of Medical Sciences and Research Centre Bhopal, MP, India

Received: 11 March, 2025 Accepted: 27 March, 2025 Published: 08 April, 2025

ABSTRACT

Congenital diaphragmatic hernia (CDH) is a serious developmental anomaly marked by a defect in the diaphragm that allows abdominal organs to herniate into the thoracic cavity, leading to compromised lung development and pulmonary hypoplasia. In this case, a 33-week pregnant woman underwent routine obstetric ultrasound, which revealed hallmark features of left-sided CDH, including the herniation of the stomach into the thorax, rightward displacement of the heart, hypoplastic left lung, and associated polyhydramnios. These findings confirmed the diagnosis, with other thoracic anomalies ruled out based on diaphragm discontinuity and the presence of abdominal contents within the chest cavity. Prognostic markers such as gestational age at diagnosis, laterality of the defect, and contralateral lung size were assessed, suggesting a potentially poor outcome. Although the exact lung-to-head ratio (LHR) was not measured, imaging implied significant lung underdevelopment. The case underscores the value of prenatal imaging in early detection and risk stratification of CDH, while highlighting the importance of further evaluation through fetal MRI, echocardiography, and genetic testing. Timely multidisciplinary intervention and delivery planning at a tertiary care center remain crucial for optimizing neonatal outcomes.

Keywords: Congenital diaphragmatic hernia, Prenatal diagnosis, Lung-to-head ratio, Pulmonary hypoplasia, Polyhydramnios

INTRODUCTION

Congenital diaphragmatic hernia (CDH) is a serious developmental anomaly that arises due to a defect in the formation of the diaphragm during embryogenesis, leading to the herniation of abdominal contents into the thoracic cavity. This condition significantly disrupts normal fetal thoracic anatomy, most notably by compressing the developing lungs, resulting in pulmonary hypoplasia and associated vascular abnormalities. CDH represents a substantial clinical challenge due to its variable presentation, unpredictable prognosis, and complex perinatal management requirements [1-6].

The incidence of CDH is estimated to occur in approximately 1 in 2,500 to 1 in 4,000 live births. Although it may be diagnosed in isolation, CDH can also coexist with chromosomal anomalies, syndromic conditions, and other structural malformations, most commonly affecting the cardiovascular and central nervous systems. The pathogenesis of CDH is multifactorial and remains incompletely understood. While the classic explanation attributes the defect to a failure of closure of the pleuroperitoneal canals during the embryonic period, more recent research suggests that disruptions in molecular signaling pathways—particularly involving retinoic acid and vitamin A metabolism—may contribute significantly to abnormal diaphragmatic and pulmonary development [2-4].

CDH is typically classified based on the anatomical location of the defect. The majority of cases are left-sided, accounting for nearly 80–85% of diagnoses [5]. Right-sided defects are less common and often more difficult to detect prenatally, while bilateral hernias are rare and usually associated with the poorest outcomes. In left-sided CDH, abdominal organs such as the stomach, intestines, spleen, and occasionally liver herniate into the left hemithorax. This displacement leads to mediastinal shift and significant restriction of lung development on the affected and contralateral sides, although the contralateral lung may still be more functional [1].

Pulmonary hypoplasia, the most critical component of CDH, results not only from the mechanical compression of developing lungs by the herniated viscera but also from intrinsic underdevelopment of

DOI: 10.69605/ijlbpr_14.4.2025.119

the pulmonary parenchyma and vasculature. This condition can lead to persistent pulmonary hypertension of the newborn (PPHN), a major cause of morbidity and mortality in affected neonates. Moreover, structural abnormalities of the pulmonary arteries and arterioles may contribute to increased pulmonary vascular resistance, further exacerbating hypoxemia and right heart strain after birth [5].

Advancements in prenatal imaging have significantly improved the detection and evaluation of CDH. High-resolution obstetric ultrasound is the primary tool for diagnosis, often revealing the characteristic intrathoracic stomach bubble, mediastinal shift, and absence of abdominal contents in their usual location. One of the most informative metrics derived from prenatal ultrasound is the lung-to-head ratio (LHR), a surrogate measure for lung volume that provides a prognostic indicator of survival. While LHR values above 1.4 are generally associated with favorable outcomes, lower values, particularly those under 1.0, suggest severe hypoplasia and increased postnatal risk [7]. Additionally, fetal MRI may be employed to further assess lung volumes and the extent of organ herniation with greater anatomical detail [1, 5].

Despite early diagnosis, the postnatal course for infants with CDH remains highly variable. Immediate neonatal management is aimed at stabilizing the infant while minimizing barotrauma to the hypoplastic lungs. Strategies such as gentle ventilation, permissive hypercapnia, and the use of pulmonary vasodilators have become cornerstones of supportive care [5, 8, 9]. In severe cases, extracorporeal membrane oxygenation (ECMO) may be considered to provide cardiopulmonary support while awaiting physiological stabilization prior to surgical intervention [8].

The timing of surgical repair is contingent on the clinical stability of the newborn. Historically, early surgical intervention was favored, but current practices recommend delaying surgery until the infant is hemodynamically stable and adequately oxygenated. Surgical techniques vary depending on the size and location of the diaphragmatic defect. Smaller defects may be closed primarily, while larger or complete agenesis of the diaphragm may require patch repair using synthetic or biological materials [8].

Outcomes in CDH have improved with the development of multidisciplinary management protocols, but survival rates still vary widely, ranging from 50% to 80% depending on the severity of lung hypoplasia, presence of associated anomalies, and access to specialized care [6, 8]. Long-term morbidities remain a concern and include chronic lung disease, feeding difficulties, neurodevelopmental delays, and gastroesophageal reflux disease [5, 6].

CASE REPORT

A 33-week pregnant woman presented for a routine obstetric ultrasound. There was no significant past medical or obstetric history, and prior ultrasounds had been unremarkable. The patient was asymptomatic, and the gestational progress had been normal up to this point.

Imaging Findings

Prenatal ultrasound revealed classic signs suggestive of a left-sided congenital diaphragmatic hernia. A stomach bubble was visualized within the thoracic cavity, indicating herniation of abdominal contents through a diaphragmatic defect. This herniation resulted in a rightward displacement of the heart, confirming the presence of a mass effect within the left hemithorax. Additionally, the left lung appeared hypoplastic, consistent with compression and underdevelopment due to the occupying abdominal organs. Polyhydramnios was also noted, likely a consequence of impaired fetal swallowing caused by the displacement of gastrointestinal structures into the thoracic cavity.

Differential Diagnosis

Although imaging findings were highly suggestive of congenital diaphragmatic hernia (CDH), other congenital thoracic anomalies were considered in the differential diagnosis. These included congenital cystic adenomatoid malformation (CCAM), bronchopulmonary sequestration, bronchogenic cysts, bronchial atresia, and mediastinal teratomas. However, these alternatives were ruled out based on the specific presence of a stomach bubble within the thoracic cavity and clear evidence of diaphragm discontinuity—hallmark features that are more specific and characteristic of CDH.

Prognosis and Assessment

Several prognostic markers were assessed to estimate the likelihood of survival and to guide appropriate perinatal management in this case. The hernia appeared large, with multiple abdominal organs herniated into the thoracic cavity, which is typically associated with a poorer prognosis. The gestational age at diagnosis was 33 weeks, considered relatively late and potentially favorable compared to earlier detection, which often correlates with more severe outcomes. The contralateral lung, however, appeared small, indicating a higher risk of pulmonary hypoplasia. The condition was unilateral and left-sided, which is more common and generally associated with better outcomes compared to bilateral CDH. The Lung-to-Head Ratio (LHR), calculated by dividing the area of the contralateral lung by the head circumference, is a key predictive metric. An LHR greater than 1.4 is typically linked to good survival rates, while an LHR less than 1 suggests poor prognosis. In this case, the exact LHR measurement would be necessary to assess risk more precisely. Additionally, the Composite Prognostic Index (CDH-CPI), which integrates ten prenatal parameters, may offer a more comprehensive prediction of survival and

DOI: 10.69605/ijlbpr_14.4.2025.119

the potential need for advanced interventions such as Extracorporeal Membrane Oxygenation (ECMO).

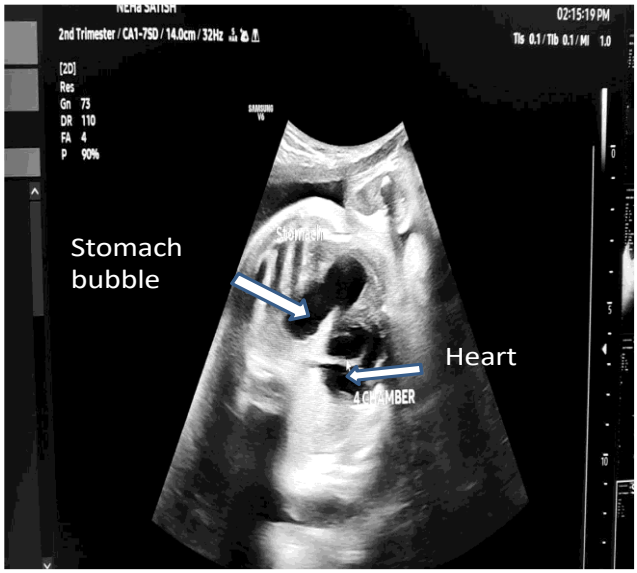

Figure 1. Congenitaldiaphragmaticherniawithstomach bubbleinthorax

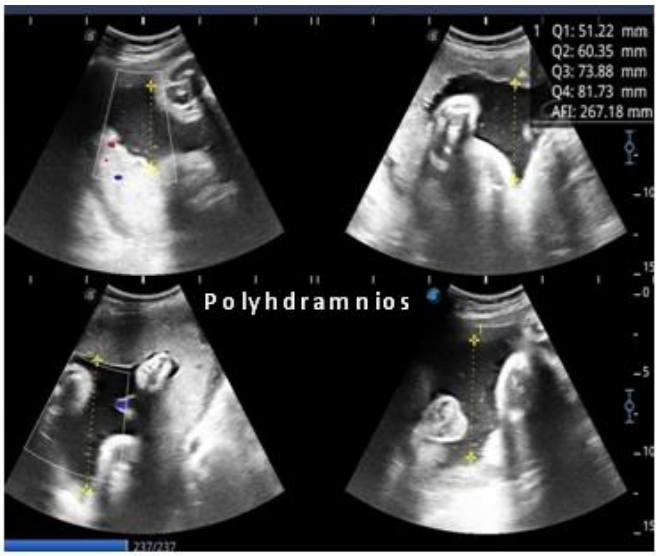

Figure 2. Prenatal Ultrasound Images Demonstrating Left-Sided Congenital Diaphragmatic Hernia

DISCUSSION

Congenital diaphragmatic hernia (CDH) remains a complex and life-threatening congenital anomaly with multifactorial etiology and variable prognosis. In this case, the prenatal ultrasound findings were classic for a left-sided CDH, which is the most common form and accounts for approximately 80–85% of cases (Chandrasekharan et al., 2017)[5].The hallmark features included herniation of abdominal contents (specifically the stomach) into the thoracic cavity, rightward displacement of the heart, hypoplastic left lung, and polyhydramnios—reflecting impaired fetal swallowing due to gastrointestinal displacement.

The gestational age at diagnosis in this case was 33 weeks, which is relatively late and generally considered more favorable than diagnoses made in the early second trimester. Early diagnosis, while facilitating advanced planning, often correlates with more severe anomalies and higher morbidity (Basak & Debnath, 2023). Basak and Debnath reported in their series that later gestational diagnoses were often associated with better outcomes, although lung hypoplasia remained a significant determinant of postnatal survival[6].

The Lung-to-Head Ratio (LHR) remains a critical tool for prenatal prognostication. Lipshutz et al. (1997) demonstrated that an LHR >1.4 was predictive of improved survival rates, whereas LHR <1 was associated with poor outcomes. In our case, while the exact LHR was not calculated, the observed small contralateral lung suggests an LHR potentially <1, warranting close follow-up and consideration of poor prognostic implications[7].

Further assessment with fetal MRI and fetal echocardiography can enhance diagnostic accuracy by quantifying total fetal lung volume and detecting

DOI: 10.69605/ijlbpr_14.4.2025.119

coexisting cardiac anomalies, which occur in up to 20–25% of CDH cases (Canadian CDH Collaborative, 2018)[8]. These additional imaging modalities are crucial, particularly when ECMO planning and surgical timing are considered.

The current consensus on CDH management emphasizes a multidisciplinary approach, as outlined in the Canadian Congenital Diaphragmatic Hernia Collaborative's guidelines (Puligandla et al., 2018). Prenatal counseling, delivery at tertiary centers with neonatal surgical capability, and individualized management plans based on risk stratification are paramount. While this case presented no immediate signs of fetal distress, postnatal stabilization strategies—including gentle ventilation and permissive hypercapnia—would be crucial to avoid barotrauma to the hypoplastic lungs[8].

In the postnatal setting, pulmonary hypertension remains a major complication and determinant of survival. Several studies have explored pharmacologic interventions:Noori et al. (2007) showed that sildenafil improved pulmonary hemodynamics in neonates with CDH-related pulmonary hypertension [9].Patel (2012) reported positive responses in cardiac function with milrinone in a case series of six neonates [10].Lawrence et al. (2019) demonstrated the use of prostaglandin E1 to maintain ductal patency and support right heart function in severe cases[11].

Surgical Timing is another debated aspect of CDH care. Chatziioannidis et al. (2014) stressed the importance of stabilizing the neonate before surgery rather than rushing to immediate repair, particularly in unstable or ECMO-dependent infants [12]. On the other hand, cases of late-presenting CDH, such as those reported by Kim and Chung (2013), may show different clinical trajectories with acute onset respiratory distress after birth or later in infancy, underscoring the heterogeneity of this condition [13].

CONCLUSION

In conclusion, this case highlights a left-sided congenital diaphragmatic hernia characterized by herniation of the stomach into the thoracic cavity, resulting in rightward displacement of the heart and hypoplasia of the left lung, accompanied by polyhydramnios. These findings underscore the importance of comprehensive prenatal evaluation. Advanced imaging modalities such as fetal MRI and fetal echocardiography play a crucial role in assessing the severity of lung hypoplasia and detecting associated cardiac anomalies. Additionally, genetic testing is essential for identifying any underlying chromosomal abnormalities, which can further influence prognosis and guide perinatal management strategies.

REFERENCES

1. Kosiński P, Wielgoś M. Congenital diaphragmatic hernia: pathogenesis, prenatal diagnosis and management - literature review. Ginekol Pol. 2017;88(1):24–30.
2. Montedonico S, Nakazawa N, Puri P. Congenital diaphragmatic hernia and retinoids: searching for an etiology. Pediatr Surg Int. 2008 Jul;24(7):755–61.
3. Coste K, Beurskens LW, Blanc P, Gallot D, Delabaere A, Blanchon L, et al. Metabolic disturbances of the vitamin A pathway in human diaphragmatic hernia. Am J Physiol Lung Cell Mol Physiol. 2015 Jan 15;308(2):L147–57.
4. Clugston RD, Zhang W, Alvarez S, de Lera AR, Greer JJ. Understanding abnormal retinoid signaling as a causative mechanism in congenital diaphragmatic hernia. Am J Respir Cell Mol Biol. 2010 Mar;42(3):276–85.
5. Chandrasekharan PK, Rawat M, Madappa R, Rothstein DH, Lakshminrusimha S. Congenital diaphragmatic hernia – a review. *Matern Health Neonatol Perinatol*. 2017;3:6.
6. Basak A, Debnath D. Congenital diaphragmatic hernia: a case series in tertiary care centre. *Int J Contemp Pediatr*. 2023;10:728–32.
7. Lipshutz GS, Albanese CT, Feldstein VA, Jennings RW, Housley HT, Beech R, et al. Prospective analysis of lung-to-head ratio predicts survival for patients with prenatally diagnosed congenital diaphragmatic hernia. *J Pediatr Surg*. 1997;32:1634–6.
8. Canadian Congenital Diaphragmatic Hernia Collaborative, Puligandla PS, Skarsgard ED, et al. Diagnosis and management of congenital diaphragmatic hernia: a clinical practice guideline. *CMAJ*. 2018;190(4):E103–12.
9. Noori S, Friedlich P, Wong P, Garingo A, Seri I. Cardiovascular effects of sildenafil in neonates and infants with congenital diaphragmatic hernia and pulmonary hypertension. *Neonatology*. 2007;91(2):92–100.
10. Patel N. Use of milrinone to treat cardiac dysfunction in infants with pulmonary hypertension secondary to congenital diaphragmatic hernia: a review of six patients. *Neonatology*. 2012;102(2):130–6.
11. Lawrence KM, Berger K, Herkert L, Franciscovich C, O'Dea CLH, Waqar LN, et al. Use of prostaglandin E1 to treat pulmonary hypertension in congenital diaphragmatic hernia. *J Pediatr Surg*. 2019;54(1):55–9.
12. Chatziioannidis I, Mouravas V, Babatseva E. When is the appropriate timing of surgical repair for congenital diaphragmatic hernia? *Hippokratia*. 2014;18(4):381.
13. Kim DJ, Chung JH. Late-presenting congenital diaphragmatic hernia in children: the experience of single institution in Korea. *Yonsei Med J*. 2013;54(5):1143–8.