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Case Series

Anatomy Section

Unveiling the Spectrum: Case Series on Congenital Diaphragmatic Hernia Variations in Second Trimester Foetal Autopsies

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ABSTRACT

Congenital Diaphragmatic Hernia (CDH) is a developmental defect characterised by abnormal diaphragm formation, leading to the herniation of abdominal contents into the thoracic cavity. CDH occurs in approximately 1 in 2,500 live births and can significantly affect pulmonary development and overall foetal health. The present series aimed to document the spectrum of presentations observed in second-trimester autopsies, which may aid in understanding the variability of the condition and its implications for foetal outcomes. Presentations of CDH were observed in three foetuses with gestational ages ranging from 14 to 28 weeks. Among the three CDH cases discussed, each presented a different type: one was bilateral CDH, one was left-sided CDH and one was right-sided CDH. The first specimen exhibited bilateral CDH of Bochdalek's type on the right, with an absent hemi-diaphragm on the left. The foetus had massive herniation of abdominal contents into the right-side of the thoracic cavity, resulting in a significant mediastinal shift to the left. The second specimen displayed left-sided Bochdalek's type of CDH, which was associated with herniation of the intestinal loops into the left-side of the thoracic cavity, resulting in a mediastinal shift to the right. The third specimen presented right-sided Morgagnian type of CDH, where only a small portion of the left lobe of the liver had herniated into the thoracic cavity and there was no mediastinal shift. The variability in presentations of CDH in the present case series underscores the complexity of the condition. Factors influencing outcomes include the timing of the hernia diagnosis, the degree of lung hypoplasia and the presence of associated anomalies. The identification of CDH through ultrasound allows for better prenatal counseling and management strategies for affected pregnancies.

Keywords: Antenatal, Congenital anomaly, Diaphragm, Lung hypoplasia, Prenatal diagnosis

INTRODUCTION

The CDH is a developmental abnormality of the thoracoabdominal diaphragm, in which various parts of the diaphragm are poorly formed to varying degrees, ranging from a negligible defect to complete absence of the diaphragm [1]. Such a defect allows the densely packed abdominal contents of the developing foetus to enter the thorax. The herniation of abdominal viscera, including intestinal loops, stomach, or liver, exerts pressure on the developing lung, resulting in mediastinal shift and ultimately affecting the development of cardiopulmonary organs and compromising their function [2].

Usually, CDH is detected during antenatal ultrasonographic screening. Most cases of CDH of lesser severity are compatible with life and can be delivered at full term, although they often present with respiratory complications due to pulmonary hypoplasia and pulmonary hypertension [3]. There can also be associated gastrointestinal symptoms [4]. Minor CDH defects can be surgically corrected, either during the antenatal or postnatal period. However, there are instances where the defects are severe and prove fatal. The etiology and pathophysiology of CDH remain uncertain. Research indicates that the mortality rate for bilateral CDH is 65%, compared with only 33% for unilateral CDH [5].

The CDH occurs in about 1 in 2,500 to 3,000 live births and can be classified into various types based on the location of the defect. Bilateral CDH is extremely rare, comprising less than 1% of all CDH cases [6]. Most affected foetuses die in utero, with less than 35% surviving [6-10]. CDH can either be an isolated anomaly or occur in conjunction with other malformations. It accounts for about 8% of all major congenital malformations and is considered one of the major surgical emergencies in newborns [9].

The most common type of CDH occurs through the posterolateral foramen of Bochdalek, with an 80% predominance on the left-side. Only 1-5% of CDH cases occur through the Foramen of Morgagni, which is retrosternal at the sternocostal triangle, known as the space of Larey. The Morgagni type is more commonly found on the right-side in the anterior mediastinum [11].

CASE SERIES

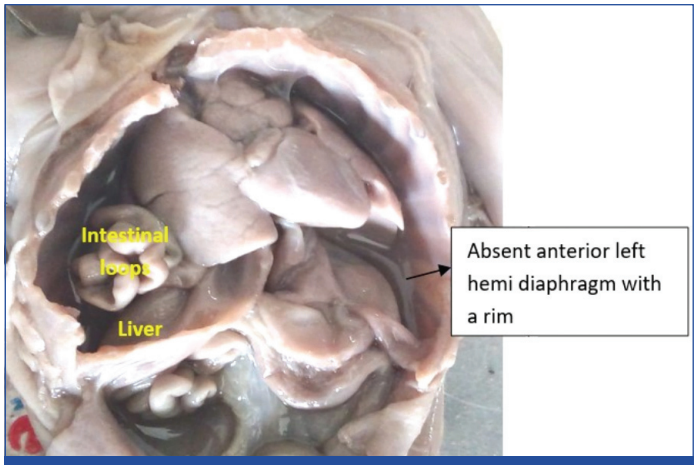
Specimen-1

Bilateral CDH: Right-sided Bochdalek's hernia and left-sided anterolateral hernia: This specimen was a 14-week-old female foetus. This foetus was the second conception for the mother and was aborted spontaneously. The first child had no anomalies. There was no history of consanguineous marriage, adverse obstetric history, or significant medical history regarding the parents. An anomaly scan had not yet been performed.

On autopsy, the foetus was found to have bilateral CDH. On the right side, there was herniation of intestinal loops and part of the liver through a large posterolateral defect in the right hemidiaphragm, specifically through Bochdalek's foramen. As a result, there was a massive mediastinal shift to the left. The left hemidiaphragm was partially absent along the anterior half, leaving no demarcation between the left thoracic and abdominal cavities. The abdominal viscera on the left-side were observed in-situ and had not herniated into the thoracic cavity [Table/Fig-1]. There were no associated anomalies.

Specimen-2

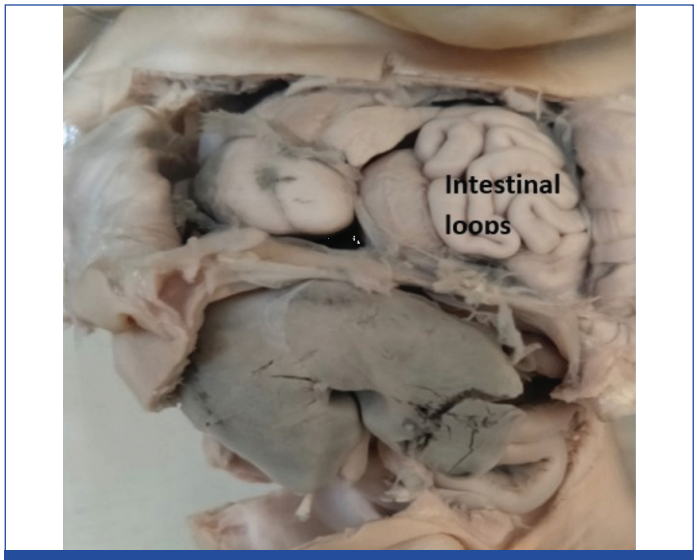
Left-sided Bochdalek's hernia: This specimen was a 19-week-old male foetus. This foetus was the first conception for the



[Table/Fig-1]: A 14-week-old foetus with bilateral CDH. Part of liver and intestinal loops is seen in right hemithorax. On the left, there is absence of left hemidiaphragm.

mother and was aborted spontaneously. There was no history of consanguineous marriage, adverse obstetric history, or significant medical history regarding the parents. An anomaly scan had not yet been performed.

Upon opening the abdominal and thoracic regions of this foetus, the authors observed left-sided CDH containing intestinal loops. The herniation was through the left Bochdalek's foramen, present posterolaterally. As a result, there was a massive mediastinal shift to the right, accompanied by a severely hypoplastic right lung [Table/Fig-2]. There were no other associated anomalies.

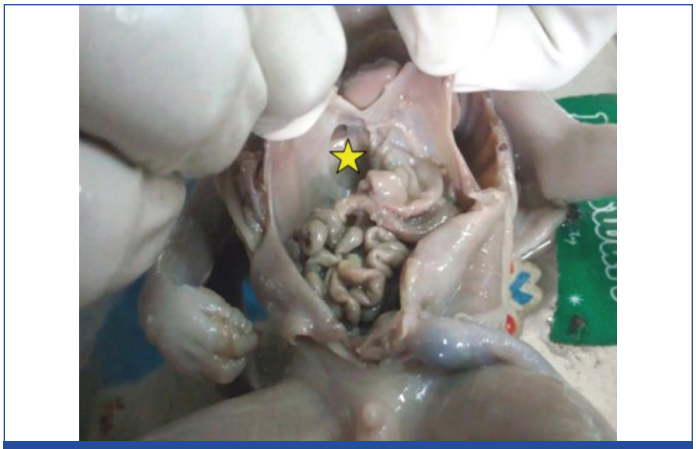


[Table/Fig-2]: A 19-week-old foetus with left-sided Bochdalek's hernia. Intestinal loops are seen in the left hemithorax with a massive mediastinal shift to the right.

Specimen-3

Right-sided anterolateral hernia: This specimen was a 20-week-old male foetus. This foetus was the first conception for the mother and was aborted spontaneously. There was no history of consanguineous marriage, adverse obstetric history, or significant medical history regarding the parents. An anomaly scan had not yet been performed.

On autopsy, part of the left lobe of the liver was seen partially herniating through the diaphragm into the right side of the thoracic cavity. Upon removal of the liver for better visualisation, the authors could identify a large anterior defect involving the anterolateral part of the right hemidiaphragm, also affecting a portion of the central tendon, thus creating an anterolateral hernia through the right foramen of Morgagni [Table/Fig-3]. There was no mediastinal shift. There were no other associated anomalies.



[Table/Fig-3]: A 20-week foetus with right-sided anterolateral hernia.
*represents the defect in the diaphragm, involving parts of the central tendon and right hemidiaphragm

DISCUSSION

The CDH is the herniation of abdominal contents through a defect in the diaphragm and is associated with varying degrees of pulmonary hypoplasia. CDH can present as 'isolated cases' (i.e., the only malformation is the diaphragmatic hernia) or as 'non isolated cases' (i.e., associated with other anomalies [12].

The major causes of CDH recorded in various research articles span a wide range, from chromosomal aberrations to nutritional deficiencies, particularly of vitamin A [13]. The most frequently encountered anomalies are trisomy 18 and tetrasomy 12p [14,15].

Embryological basis: The diaphragm arises from the fusion of the following embryonic structures: the septum transversum, the dorsal mesentery of the esophagus and the dorsal and dorsolateral body wall. The most important structures are the septum transversum and the pleuroperitoneal membranes. The septum transversum develops as the most superior part of the cranial mesenchyme during the fourth week of intrauterine life and contributes to the partition of the coelom, as well as part of the diaphragm, stomach and duodenum; however, it does not completely separate the thoracic and abdominal cavities. The two pleuroperitoneal membranes fuse with the dorsal mesentery of the esophagus and the septum transversum completes the partition between the thoracic and abdominal cavities, forming the primordial diaphragm. Typically, the diaphragm is complete by the eighth week of gestation. Recent studies have associated mutations in the GATA Binding Protein 6 (GATA6) gene and even over-expression (triplication) of Endothelin Receptor type A (EDNRA) messenger Ribonucleic Acid (mRNA) with the development of CDH [16-18].

Clinical implications and outcome: The wide range of presentations of CDH, including its site, the herniation of any abdominal organ into the thoracic cavity, the degree of mediastinal shift and the extent of pulmonary hypoplasia and pulmonary hypertension, are crucial factors determining the outcome of CDH. Diagnosis during the prenatal period generally induces a better prognosis; prenatal assessment of the Lung-to-Head Ratio (LHR) and the position of the liver through ultrasound are used to diagnose and predict outcomes. Early diagnosis, along with an increased understanding of this disease, is essential for a timely approach to managing critically ill infants and offers the potential for improved outcomes and significant reductions in morbidity [12].

As mentioned, many cases of CDH go undetected antenatally and are diagnosed only when patients present with complaints after birth. CDH repair is one of the most commonly performed surgical procedures in the neonatal period. Despite advances in neonatal intensive care, anaesthesia and surgery, the postoperative mortality rate for CDH remains over 30% [19], primarily attributed to pulmonary hypertension.

Ağaçayak E et al., reported a case of bilateral CDH in which the left lobe of the liver was found in the right thoracic region, resulting in mediastinal shift [14]. This case also involved herniation of the stomach on the left-side. Supriya et al., documented a case of right-sided diaphragmatic hernia, in which small intestinal loops and

mesentery were observed in the thorax [12]. Chae et al., reported a case of antenatal CDH involving the stomach and left hemicolon in the left half of the thorax, leading to strangulation [20].

Mwamanenge NA et al., reported a six-week-old baby with a left-sided Bochdalek hernia, which included herniation of bowel loops, spleen and part of the liver into the thoracic cavity. This condition was diagnosed postnatally when the baby was investigated for respiratory distress [21]. Martins J and Oberhauser M reported a case of 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 [22]. Basak A and Debnath D reported a series of post natal cases of CDH, all presenting with respiratory distress [23]. The left-sided posterolateral type of CDH was found in four of the five reported cases, with varying abdominal contents herniating into the thoracic cavity, including the liver, spleen and intestinal loops, resulting in differing extents of mediastinal shift. The type of CDH is not mentioned for the remaining case.

CONCLUSION(S)

The present case series highlights the diverse presentations of CDH observed in foetal autopsies, emphasising the complexity and variability of this condition. The present study findings demonstrate that CDH can manifest in multiple forms—left-sided, right-sided and bilateral—with varying degrees of associated pulmonary hypoplasia and other congenital anomalies. The differences in clinical outcomes underscore the importance of accurate prenatal diagnosis and comprehensive counseling for expectant parents. By enhancing the authors understanding of the spectrum of CDH presentations, they can improve prenatal care strategies, ultimately aiming for better management and outcomes for affected infants.

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