



**Review Article**

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# Congenital Diaphragmatic Hernias: A Review Article



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## Abstract

Congenital diaphragmatic hernia (CDH) is a rare anomaly that occurs through foramen of Bochdalek (common), foramen of Morgagni (rare), or esophageal hiatus (rare). It is commonly unilateral (right or left) and rarely bilateral. Genetic factors are blamed for the pathogenesis of CDH and other factors as fetal male gender, maternal age above 40, vitamin deficiencies, smoking, alcohol and chemicals during pregnancy are additional contributors to the incidence of this anomaly. CDH commonly presents in neonates and rarely later in life. It may be associated with respiratory, gastrointestinal and urinary complications. Pulmonary agenesis, pulmonary hypoplasia and persistent pulmonary hypertension are the main determinants of mortality and morbidity in neonates with CDH. Reduction of the hernia and repair of the diaphragmatic defect are achieved through either transthoracic or transabdominal approach.

**Keywords:** Diaphragmatic Hernias; Congenital; Morgagni Hernia; Bochdalek Hernia; Hiatus Hernia; Clinical Associations

**Abbreviations:** AGV: Acute Gastric Volvulus; BH: Bochdalek Hernia; CDH: Congenital Diaphragmatic Hernia; CDHs: Congenital Diaphragmatic Hernias; CHA: Congenital Heart Anomaly; CMH: Congenital Morgagni Hernia; CMLH: Congenital Morgagni-Larrey's Hernia; CPEH: Congenital Paraesophageal Hernia; ECMO: Extracorporeal Membrane Oxygenation; EPS: Extralobar Pulmonary Sequestration; GEJ: Gastroesophageal Junction; L-CDH: Left Congenital Diaphragmatic Hernia; MRI: Magnetic Resonance Imaging; PPFs: Pleuro Peritoneal Folds; R-CDH: Right Congenital Diaphragmatic Hernia; VT: Ventilatory Time

## Introduction

Congenital diaphragmatic hernia (CDH) is an uncommon congenital anomaly of the diaphragm resulting in herniation of the abdominal viscera into the thoracic cavity. It develops from incomplete closure and fusion of the fetal pericardioperitoneal canals and pleuroperitoneal folds (PPFs) or abnormality of the esophageal hiatus. Congenital diaphragmatic hernias (CDHs) occur mainly through two diaphragmatic defects: rarely in the foramen of Morgagni and commonly in the foramen of Bochdalek. CDH through the esophageal hiatus is also rarely recognized. All the three types of CDH are repaired surgically. Pulmonary hypoplasia and persistent pulmonary hypertension are the two main determinants of neonatal mortality and morbidity in cases with CDH [1-7]. CDHs are mostly diagnosed early in life; later diagnosis is rare especially if the hernia is asymptomatic. Despite advances in therapeutic modalities, CDH in the newborn represents challenges to the multi-disciplinary teams involved in CDH management [8,9].

Early intubation of antenatally scan-diagnosed CDH in the newborn is crucial in preventing respiratory deterioration and persistent pulmonary hypertension [10,11]. Observed-to-expected lung-to-head ratio, liver position, and total lung volume measured by magnetic resonance imaging (MRI) are the prognostic predictors most often used, and they correlate with

neonatal mortality and morbidity [12]. Extralobar pulmonary sequestration (EPS) can be found incidentally in CDH; it usually arises in the chest or the abdomen and rarely in the diaphragm [13]. Hepatopulmonary fusion is a rare malformation associated with R-CDH and is only discovered during surgical repair of the diaphragmatic defect [14]. The association of 46,XY sex disorder with CDH is rare; it has been described with or without other congenital anomalies as external ambiguous genitalia, truncus arteriosus, bifid thymus, gut mal rotation, and limb anomalies [15].

Neonates with CDH have a high prevalence of congenital malformations [16]. The prognosis for infants with CDH associated with cardiovascular malformation is poor. The severity of the cardiovascular malformation is more important as a predictive factor for mortality and morbidity than the severity of CDH itself [17]. Minor forms of congenital heart anomaly (CHA) have no negative impact on the survival of infants with CDH. However, mortality appears to be significantly higher in infants with CDH associated with major forms of CHA [18]. The possibility of a diaphragmatic hernia is suspected when a cardiac mass with specific echocardiographic features is observed [19]. Large CDH defect sizes increase mortality while the association between defect sizes and morbidity is not fully elucidated [20]. Growth patterns during the first year of life were described in

infants with CDH. Poor growth was a common early finding in CDH patients, which improved during infancy [21].

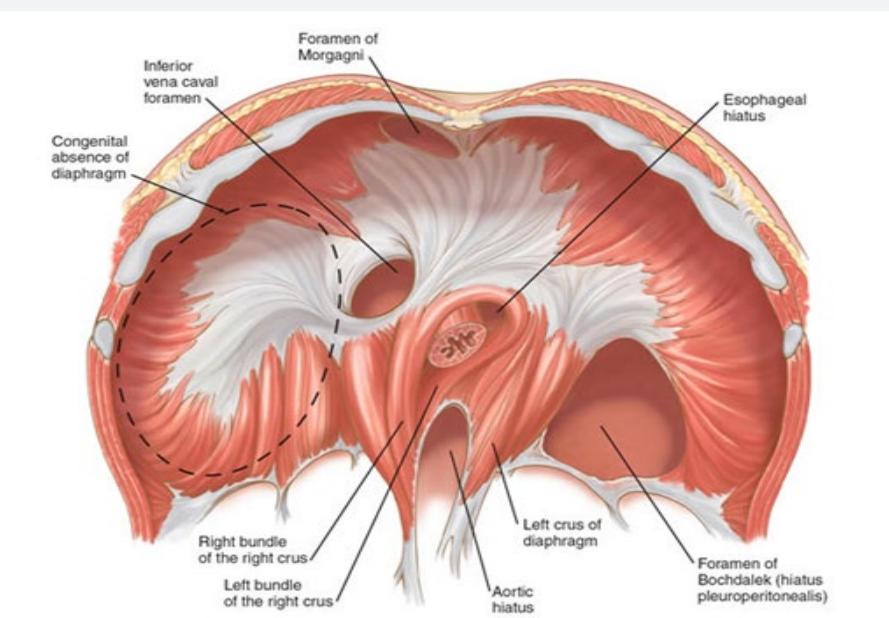
Furthermore, it was emphasized on the importance of close follow-up and intense nutritional management in CDH infants [22]. A clinical prediction rule, for neonates with CDH, was designed using predictors generated from: very low birth weight, absent or low 5-minute Apgar score, presence of chromosomal or major cardiac anomaly, and pulmonary hypertension. This clinical model discriminated between neonates at high, intermediate or low death risks [23]. Low five minute Apgar score, and high alveolar-arterial gradient are significant predictors of neonatal mortality in CDH. R-CDH is associated with high mortality (50%) and prolonged length of stay than L-CDH [24].

Ventilatory time (VT) is an important marker to identify subjects at risk for short-term neurodevelopmental impairment in CDH survivors [25]. Fetal ultrasonography and MRI are essential antenatal outcome predictors in neonates with CHD [26]. Fetal lung area-to-head circumference ratio and observed-to-expected lung-to-head ratio measurements can accurately predict postnatal survival and the need for extracorporeal membrane oxygenation (ECMO) therapy in fetuses with left-

sided congenital diaphragmatic hernias [27]. CDH presentation in adults is extremely rare. Patients with late presentation of CDHs complain of a wide variety of symptoms and diagnosis has proved to be difficult [28]. The late-presenting Bochdalek hernias often constitute difficulties in diagnosis that may lead to inappropriate treatment; an example is the herniated stomach that is mistaken for a tension pneumothorax [29].

### Etiology, Incidence and Risk Factors Of CDH

The diaphragm is derived from multiple embryonic mesodermal sources, but how these structures give rise to the diaphragm is not definitely known. The exact etiology of CDH is still unknown but some researches point out genetic factors as a possible cause of the defect [30,31]. Although numerous chromosomal aberrations and gene mutations are associated with CDH, the etiology of the diaphragmatic defect is identified in less than 50% of patients [32]. Attention has to be paid to chromosomal abnormality in cases of CDH associated with abdominal wall closure defect in the presence of intact septum transversum [33]. In mouse models, it was demonstrated that the transient embryonic pleuroperitoneal folds (PPFs) were the source of the diaphragm's muscular tissue and that migration of PPF cells controlled diaphragm morphogenesis.



**Figure 1:** Foramina and congenital defects of diaphragm.

Furthermore, mutations in PPF-derived muscle connective tissue fibroblasts resulted in the development of biomechanically weak and more compliant non-muscular localized parts of the diaphragm, leading to CDH. [34]. The incidence of CDH is reported to be 1 in 2500 births, with left congenital diaphragmatic hernia (L-CDH) being seven times commoner than right-side

one (R-CDH). Many cases of CDH are discovered prenatally or during the immediate postnatal period while 5-25% of cases can be late-presenting with respiratory or gastrointestinal problems or complications such as gastric volvulus, obstruction, perforation, peritonitis or necrosis [35]. In USA, the incidence of CDH was estimated to be 1.93/10,000 births. Risk factors for the

development of CDH included foetal male gender, maternal age above 40, Caucasian ethnicity, smoking and alcohol use during pregnancy. As compared to foetuses without CDH, foetuses with CDH were at an increased risk of stillbirth, preterm birth, and intrauterine growth restriction. In CDH, neonatal mortality during the first year amounted to 45.89% [36](Figure 1).

### Right- Versus Left-Sided CDH

Regarding the sidedness of CDH, Morgagni hernia is typically detected on right side anteriorly while Bochdalek hernia on left side posteriorly, because of the protective roles of the liver and heart on either side respectively. Hiatus hernias range from herniation of a small portion to herniation of the whole stomach into the left thoracic cavity; very rarely into the right thoracic cavity [37]. It was found that the severity of left heart hypoplasia correlated with the severity of CDH. Both right and left CDH had decreased the left ventricular volume and in addition, L-CDH compressed the left heart [38]. Right ventricular dimensions were reported to be significantly reduced in fetuses with isolated R-CDH [39]. R- CDH seems to have a poorer outcome than that reported for fetuses with L-CDH with similar lung size before birth [40].

R-CDH requires prosthetic mesh repair more frequently than L-CDH because of larger defect size or complete agenesis. Recurrent herniation is significantly higher in the R-CDH. Survivors of R-CDH do not have a significantly different neurodevelopmental outcome when compared to L-CDH survivors. Compared to L-CDH, fetuses with R-CDH are less likely to be diagnosed prenatally and have a higher need for extracorporeal membrane oxygenation [41,42]. R-CDH is not associated with increased mortality, but with increased severity of pulmonary hypoplasia necessitating increased requirement for pulmonary vasodilatory therapy and for tracheostomy [43].

### Hernia of Morgagni-Larrey

Giovanni Battista Morgagni (1682-1771) was the first to describe anatomical structures like the trigonum sternocostale dextrum (the Morgagni's foramen), the appendix testis (the Morgagni's hydatid), the vertical folds of distal rectum (the Morgagni's columns) and many others. For this, it was said of him: "If all the anatomical findings made by Morgagni should bear his name, probably one third of human body would be called Morgagni's" [44]. Hernia of Morgagni is a congenital herniation of abdominal contents into the thoracic cavity through a retrosternal (retrocostoxiphoid) diaphragmatic defect (foramen of Morgagni). It is also termed parasternal diaphragmatic hernia of Morgagni-Larrey (retrochondrosternal, retrocostoxiphoid, retrosternal, subcostal, substernal or subcostosternal hernia) [45]. Morgagni-Larrey hernia is the rarest of all CDHs [46].

On CT examination, unusual giant bilateral Mogagni hernia, extending to both thoracic apices, was detected. [47,48]. The incidence of congenital Morgagni's hernia (CMH) among all

CDHs is 3-4% (less than 5%) and about 90% of the hernias occur on the right side, 8% are bilateral and 2% are on the left side [49]. CMH is more recordable from women with advancing age. Embryonic disorder of diaphragmatic differentiation is the major etiological factor but vitamin deficiencies and some chemical substances are also predisposing. CMH, in the pediatric age group, commonly presents with recurrent chest infection and has a high incidence of associated anomalies, commonly congenital heart disease and Down syndrome [50]. A case of CMH has been detected in a 3-month-old child affected by Marfan's syndrome [51]. In CMH, the hernial sac is usually small and is surgically repaired through either abdominal or lateral thoracotomy approach. Median sternotomy is rarely used as an approach for repair these hernias [52].

Transabdominal approach is preferred to transthoracic one for the surgical repair of Morgagni hernias [53]. There is a high rate of recurrence after laparoscopic repair of Morgagni hernia without the use of a prosthetic polypropylene mesh [54]. Hernia of Morgagni may be associated with mediastinal lipoma in an adult [55]. It may consist of the thoracic protrusion of fat and/or abdominal viscera through a congenital diaphragmatic defect. It may be asymptomatic, mimicking a large intrathoracic lipoma. Improper diagnosis can cause catastrophe during surgical repair [56,57]. Bilateral Morgagni hernias associated with left Bochdalek diaphragmatic hernia and Down syndrome are very rare congenital anomalies [58]. Congenital Morgagni-Larrey's hernia (CMLH) is known to be associated with a high incidence of bilaterality and associated anomalies like congenital heart disease, Down's syndrome, gut malrotation, inguinal hernia, umbilical hernia, and pyloric stenosis [59-62]. Gastric volvulus and intestinal obstruction are described as complications of CMH [63, 64].

### Hernia of Bochdalek

Professor Vincenz Alexander Bochdalek was a clever anatomist and pathologist who studied and worked at Charles-Ferdinand University in Prague from the 1820s to the early 1870s. His name is permanently linked to certain anatomico-pathological structures, particularly the vertebrocostal (lumbocostal) triangle in the posterior part of the diaphragm (foramen of Bochdalek) and the hernia through this foramen (posteriorlateral hernia of Bochdalek) [65,66]. Bochdalek hernia (BH) is the commonest of all CDHs. It occurs in 1 in 2000 to 3000 newborns [67]. It usually presents in neonates with respiratory failure and very rarely in adults, needs to be early corrected and repaired, and is associated with a high mortality [68-73]. No correlation is found between asymptomatic incidentally discovered BH and age, gender, or body mass index in adults [74].

Symptomatic BH in adults may lead to gastrointestinal dysfunction or severe pulmonary disease. [75] Bilateral occurrence is rare; a case of bilateral BH was associated with severe aortic tortuosity and aberrant right subclavian artery [76].

BH may be associated with acute gastric volvulus (AGV) [77,78], intestinal obstruction, perforation, gangrene and empyema [79,80]. Herniated stomach, spleen and colon [81], and ectopic intrathoracic kidney [82-84]. Other rare associations are septic ureteric obstruction [85], and intermittent heart block [86].

### Congenital Hiatus Hernia

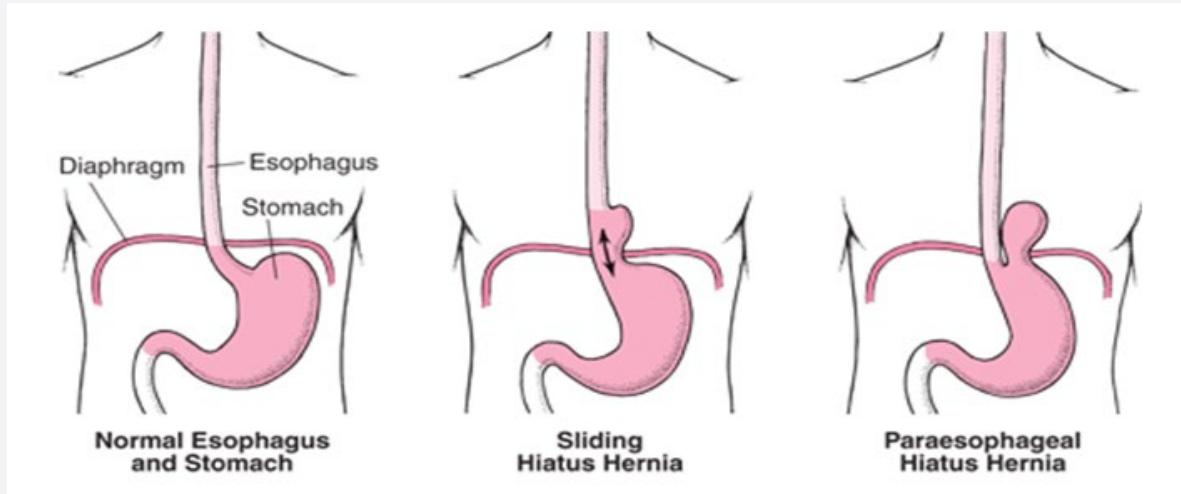
The pathogenesis of hiatus hernia is attributed to three theories:

1. An increase in the intra abdominal pressure forcing the gastroesophageal junction (GEJ) into the thorax;
2. Shortening of the esophagus due to fibrosis or excessive vagal nerve stimulation; and
3. Widening of the esophageal hiatus in response to congenital or acquired molecular and cellular changes in the diaphragm [87].

Congenital paraesophageal hernia (CPEH) is a rare condition in childhood. It usually presents with non-specific symptoms of recurrent attacks of chest infection and/or vomiting; it may be associated with Marfan's syndrome [88,89]. Most reported cases of CPEH refer to adult patients. Familial occurrence of sliding hiatus hernias is postulated in some cases [90]. A case of a boy with hiatus hernia that developed eight days following a transabdominal repair of the left BH is mentioned in the

literature [91]. An intrathoracic gastric volvulus complicating CPEH is rare; rapid diagnosis and treatment are essential to avoid dreadful complications as gastric dilatation, gangrene and perforation that may lead to cardiopulmonary arrest [92]. In an old cadaver, a congenital hiatus hernia associated with non rotation of the midgut was observed. The hernial sac originated from the peritoneum near the GEJ, and contained the midgut. The duodenum was abnormally placed and the common hepatic duct was variably drained [93]. CPEHs differ from the more common sliding hiatus hernias in anatomy, pathology, symptoms, complications, and management [94].

In literature, there are case-reports of intrathoracic stomach secondary to congenitally short esophagus in a preterm infant [95], hiatus hernia associated with unilateral pulmonary agensis and malformations of big vessels [96], CPEH causing severe hyporegenerative anemia [97], massive hiatal hernia containing the colon, intestine, and stomach mimicking chest pathologies [98], and gastric herniation through the aortic hiatus [99]. A case of a child was diagnosed to have congenital bilateral cervical lung herniation associated with retrosternal (Morgagni), posterolateral (Bochdalek), esophageal hiatal, and inguinal hernias, together with multiple urinary bladder diverticula. These multiple herniations were thought to be secondary to defective embryonic celomic mesoderm [100,101](Figure 2)



**Figure 2:** Types of congenital hiatus hernia.

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