

Pediatric Congenital Diaphragmatic Hernia

Irina Suley Tirado Pérez^{1*},
Ulfran De Jesús Castro Salas²
and Andrea Carolina Zarate
Vergara ACZ³

Abstract

Congenital diaphragmatic hernia is a condition characterized by a defect in the diaphragm that leads to the protrusion of the abdominal contents in the thoracic cavity which interferes in the pulmonary development. Diaphragmatic eventration is a very infrequent condition in children, asymptomatic or commonly present with low or gastrointestinal respiratory manifestations and whose diagnosis is based on an incidental finding when requesting images to verify a diagnostic suspicion occurring in most cases of late occurrence. It can be of congenital or acquired cause. The prognosis in general is good, with probable complications being chronic lung infection, diaphragmatic rupture, ulcers and stomach volvulus. Treatment is a topic depend size and location of the defect, size and location of the defect, some pediatric surgeons do not operate if the patient is asymptomatic and only plicate the symptomatic patients. Due to its importance in pediatric population, through this is intended to make a quick review of the updated literature on the subject contributing to have relevant information regarding the issue cooperating with the diagnosis and appropriate treatment in our practice.

Keywords: Congenital Diaphragmatic Hernia (CDH); Diaphragmatic eventration; Diaphragmatic hernia; Congenital anomalies

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Introduction

Congenital diaphragmatic hernia (CDH) is a defect that occurs in 1 in 3,000 live births, of which approximately 60% occur in isolation without other congenital anomalies. This is a rare congenital anomaly of the diaphragm that occurs due to poor embryogenesis with atrophy of the diaphragm muscle fibers and loss of muscle tone. It is more frequent on the left side, and bilateral cases have been reported, male predominance is also recognized. It is associated with gestation infections such as rubella and cytomegalovirus, Jarcho-Levin's spóndyl-thoracic dysplasia, malformations such as pulmonary hypoplasia, pulmonary sequestration, congenital heart disease, renal ectopia, gastric volvulus, chromosomal diseases, various myopathies and spinal cord abnormalities. Often the diagnosis of diaphragmatic eventration is a finding when performing chest X-rays in a respiratory distress syndrome or in febrile conditions. Considering that many patients are asymptomatic but at the same time with bronchial segmentation and alveolar multiplication extending up to 8 years of age and that the partial occupation of the hemithorax is directly detrimental to the complete development of that lung, raises the discussion about the indication and timing of the surgical correction of this pathology. The most

- 1 Master Pediatric Palliative Care, Pediatric Intensive Care Student, University of Santander, Colombia
- 2 Pediatric Teacher, University of Santander, Colombia
- 3 Pediatric Intensive Care Student, University of Santander, Colombia

Corresponding author:

Irina Suley Tirado Pérez

✉ irinasuley@gmail.com

Magister Pediatric Palliative Care, Center for Research and Teaching, Graduate Student of Pediatric Intensive Care Level I, University of Santander, Columbia.

Tel: 3145598697

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frequent defect occurs in the left posterolateral region of the diaphragm (Bodahlek hernia), but may be right in 15% of cases or bilateral in about 1-2%. Complete agenesis of the diaphragm and eventration are very rare manifestations. The prenatal diagnosis of CDH is has been increased thanks to prenatal ultrasound screening programs, from being a diagnosis of neonatal urgency to an increasingly well-known condition since the fetal stage. Immediate treatment at birth includes bowel decompression, avoidance of mechanical ventilation to the extent possible. The main management focus includes non-invasive ventilation, hemodynamic monitoring and treatment of pulmonary hypertension followed by surgery. Although inhaled nitric oxide is not approved by the FDA for the treatment of CDH-induced pulmonary hypertension, it is commonly used. Extracorporeal membrane oxygenation (ECMO) is typically considered after the failure of conventional medical treatment for neonates with gestational age ≥ 34 weeks or weighing >2 kg with CDH that do not have large associated lethal abnormalities. Multiple factors

such as prematurity, associated anomalies, persistent pulmonary hypertension severity, type of repair, and need for ECMO may affect the survival of a baby with congenital diaphragmatic hernia. With advances in CDH management, overall survival has improved and it has been reported 70-90% in children not undergoing ECMO and up to 50% in children undergoing ECMO.

Discussion

The incidence of CDH has been reported 1:2,000 - 5,000 live births, Neonatal CDH is a well-recognized entity, but its presentation beyond the neonatal period varies, giving rise to erroneous clinical and radiological diagnoses. In contrast to the high neonatal mortality and morbidity rates for CDH, the prognosis for late CDH hosts if diagnosed early is generally favorable [1,2]. CDH is the result of incomplete closure of the normal pleuroperitoneal canal during fetal development. Most cases are diagnosed before birth or in the neonatal period. However, 5% to 45.5% of the CDH may appear asymptomatic during the neonatal period, to manifest itself in adulthood. The congenital defect is identical in neonates and older patients, but the approaching and complicating symptoms of CDH in older patients differ considerably from those found in newborn patients. Although the exact etiology of most cases of congenital diaphragmatic hernia remains unknown, there is increasing evidence that genetic factors play an important role in the development of CDH.

Chromosomal abnormalities have been identified as an important etiology for non-isolated CDH. In most published cases, chromosomal abnormalities were identified using a combination of anal-chromosome bands-GYsis and/or FISH. The use of new technologies, such as comparative genomic hybridization based matrices (arrayCGH) - is likely to increase the number of chromosomes identified in individuals with CDH and may aid in the identification of CDH-related genes. Trisomies 13, 18, and 21 and 45, are the common aneuploidias described in association with CDH [1,3]. Embryologically, the etiology of CDH is postulated as the abnormal migration of myoblasts from the superior cervical somites in two of the four embryological structures that contribute to the development of the diaphragm as septum transversum (Beginning at week 4 of gestation) and the pleuroperitoneal membrane 8-12 weeks of gestation). Thomas hypothesized the involvement of altered myoblast growth in the pleuroperitoneal membrane, when the abdominal viscera return to the peritoneal cavity prematurely [2,4]. The CDH is more common among men a 2: 1 ratio generally, although several factors depend like the reference population; the diaphragmatic abnormality can be localized or diffuse. In the diffuse variety, the diaphragm consists of a thin diaphanous membrane that is peripherally attached to the normal muscle membrane. Partial defects mainly affect the right hemidiaphragm (65%), while diffuse defects tend to be unilateral and most commonly affect the left side. The first clinical manifestations of CDH are not obvious and are not specific, and could often lead to misdiagnosis in a considerable number of patients. A large population of these cases with asymptomatic, non-iatrogenic diaphragmatic manifestations occur in 7 to 35 per 100,000 people and were observed in 1 for every 10,000 health recruits [2,4].

The main symptoms in CDH are due to intra-abdominal elevation causing compression of the lower lobe of the lung causing dyspnea, cyanosis, acute respiratory distress, vascular dysfunction, and cardiac symptoms. Anorexia, nausea, and vomiting may also be associated. It is hypothesized that CDH causes these problems in 5 ways:

1. Inability to ventilate the ipsilateral lung;
2. Direct compression of the ipsilateral lung;
3. Pneumonia due to chronic atelectasis;
4. Pendelluft due to ventilation between the affected and the unaffected lung;
5. Paradoxical movement of the diaphragm to make ventilation of the contralateral lung inefficient [4-7].

In infants, the mediastinum is very mobile, and the paradoxical movement of the affected diaphragms causes a displacement of the heart and mediastinum towards the contralateral side thorax. In unilateral CDH, the vital capacity and the total lung present a decrease from 20% to 30%, resulting in chest tightness, respiratory tract infections and cough with or without expectoration. Bilateral diaphragmatic eventration reduces lung function even more seriously, especially in the supine position. Although these children can tolerate bilateral eventral diaphragmatic events, they are prone to chronic respiratory failure [5-7].

CDH can be isolated or associated with other developmental defects. Significant associated findings with congenital heart disease, pectus excavatum, cleft palate, hypospadias, cryptorchidism, and congenital torticollis. Results revealed hypoplastic pulmonary (10%) and congenital heart disease (9%), as the major anomalies associated with this study [5-7]. Typical clinical manifestations may lead to the diagnosis of CHD, which is often confirmed by chest X-ray and fluoroscopy. CHD symptoms in children usually require surgical treatment. Indications For surgery include:

1. Rapid breathing without improvement under conservative management;
2. Two or more ipsi-lateral recurrent pneumonias;
3. Life-threatening pneumonia;
4. Inability to ventilate mechanical ventilation;
5. Respiratory distress related to paradoxical of the diaphragm.

The plication of the diaphragm is surgery option for CDE, which was first successfully performed in an adult in 1923 By Jean Louis Petit and the child in 1946 by Bisgard. The goal of the surgery is to flatten the dome of the diaphragm and provide the lung with a larger volume for expansion. Since its original description, diaphragm plating has been performed with numerous modifications, including the minimally invasive video assisted thoracic surgery approach [5,6].

Important points are as follows:

- Congenital diaphragmatic hernia (CDH) is a defect that occurs in 1 in 3,000 live births, of which approximately 60% occur in isolation without other congenital anomalies. However, 5-45.5% of the CDH may appear asymptomatic during the neonatal period.
- The CDH is more common among men a 2: 1
- Chromosomes more in association with CDH: Trisomies 13, 18, and 21 and 45.

Prenatal diagnosis by ultrasound detects more than 50% of CDH cases at a mean gestational age of 24 weeks. Images diagnosed as chest X-ray, computerized axial tomography and magnetic resonance with usefulness in the diagnosis of this pathology. Three-dimensional ultrasound, fetal echocardiography and fetal magnetic resonance (MR) are other methods of prenatal diagnosis used in assessing the severity and outcome of the CDH. Left a single side of the CDH can be characterized by the presence of heterogeneous mass which may be the stomach full of fluid or intestines. In contrast, isolated on the right side of CDH is extremely difficult to diagnose by ultrasound if the liver is the only organ that has herniated. Indirect signs, such as a change in the cardiac axis, identification of the gallbladder and vasculature in the liver using Doppler can aid in the diagnosis. MRI has been found to be useful in detecting abnormalities in the fetus and can be a valuable adjunct to assess liver position and lung volume estimation. Associated cardiac and neural tube defects can affect the outcome of newborns with CDH [8,9]. It was found that the diaphragmatic plication re-expanded the structures most commonly herniated are: small intestine, stomach, part of the descending colon, left kidney and left lobe of the liver occupying the left side of the thorax causing pulmonary hypoplasia. The severity of this will depend on the moment of the fetal life in which the herniation and compression occurred; there is a deviation of the mediastinum towards the left that can cause compression and hypoplasia of the contralateral lung. The pulmonary artery is diminished in size in proportion to the size of the lung; the pulmonary ramification is also diminished. The capillary alveolar relation is normal, but due to the decrease in the number of alveoli the total vascular area is diminished causing pulmonary hypertension, in addition, the existing distal arteries may have little smooth musculature. The first cause of death in these patients is progressive hypoxemia and acidosis. Pulmonary hypertension is a combination of irreversible factors (hypoplasia and dysplastic arteries) and reversible (constriction of relatively normal arteries). The effect of lung compression (due to abdominal chest content on respiratory function) is controversial. Abdominal content causes respiratory failure when there is massive gas distension in the stomach and intestine, pneumothorax is a common complication of mechanical ventilation in patients with hypoplastic lungs. The ratio lecithin sphingomyelin and phosphatidyl glycerol are normal [5-7].

Important points are as follows:

- The first clinical manifestations of CDH are not obvious and are not specific.

- The severity of this will depend on the moment of the fetal life in which the herniation and compression occurred.
- The main symptoms in CDH are due to intra-abdominal elevation causing compression: dyspnea, cyanosis, acute respiratory distress, vascular dysfunction, and cardiac symptoms. Anorexia, nausea, and vomiting may also be associated.
- The first cause of death in these patients is progressive hypoxemia and acidosis.

In the differential diagnosis, the history of perinatal trauma should be ruled out to exclude posttraumatic diaphragmatic paralysis, the symptomatology of which is similar to that of diaphragmatic eventration; Among other causes to rule out congenital lobar emphysema and, if present on the right side, right pleural effusion, hernias, tumors or hepatic cysts. In case of diaphragmatic paralysis, the definitive diagnosis is made with the diaphragmatic biopsy, to verify the interruption of the striated muscle in the tissue. [4,8]. The prognosis of this pathology is good; Although the incidence of chronic lung disease is 33% to 52% at discharge, the majority of surviving infants have CDH clinical improvement over time. However, almost 50% of adult survivors have deterioration in lung function tests. Survivors with CDH are at risk for much morbidity that may affect development and function. Babies with large defects, those who have received ECMO, or those with a patch repair are at increased risk. These unique patients, especially those at increased risk, require long-term follow-up by a multidisciplinary team of medical, surgical, and developmental specialists to identify and treat co-morbidities prior to additional disability outcomes. Preventive pediatric health care is recommended in accordance with the guidelines developed by the American Academy of Pediatrics for all children, including those with CDH. To emphasize the importance of monitoring specific morbidity associated with CDH, additional suggestions are provided. These are most applicable to children with extraordinary medical and surgical complications associated with CDH and should be individualized according to the specific needs of each children [9].

Important points are as follows:

- Prenatal diagnosis by ultrasound detects more than 50% of CDH cases at a mean gestational age of 24 weeks.
- Images diagnosed as chest X-ray, computerized axial tomography and magnetic resonance with usefulness.
- The prognosis of this pathology is good in pediatric.
- But Babies with large defects, those who have received ECMO, or those with a patch repair are at increased risk.
- Treatment is surgery.

Conclusion

Despite the unclear etiology of CDH in recent years, there has been a greater trend for survival in CDH patients largely due to medical and surgical advances in CDH management; the overall

survival reported is 70- 90%. Once diagnosed, an early surgical intervention is necessary for the prevention of complications. Patients with CDH final presenters have a more favorable prognosis due to less severe or absent pulmonary hypoplasia. Multiple factors such as prematurity, ECMO, especially associated cardiac abnormalities, need for transport, severity of pulmonary

hypertension, type of repair can affect the outcome and survival of a patient with CDH. Early diagnosis and appropriate treatment contribute to a good prognosis.

Conflicts of Interest

None.

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