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Congenital Hernia of the Diaphragmatic Dome with Neonatal Revelation, Experience from the Neonatology and Neonatal Intensive Care Unit at the University Hospital Center of Oujda

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Abstract

Congenital hernia of the diaphragmatic dome (CHDD) is an embryonic malformation in which all or part of the diaphragmatic dome fails to develop properly. In the majority of cases (80% to 90%), this malformation affects the left posterolateral part of the diaphragm, while in 10% to 15% of cases it affects the right. Bilateral cases are extremely rare, accounting for less than 1% of cases. This malformation is estimated to occur at a frequency of around 1 in 3500 births, with a male predominance. The diaphragmatic defect causes the abdominal organs to rise into the thoracic cavity during critical phases of lung development. These anomalies result in bilateral pulmonary hypoplasia, a reduced number of pulmonary vessels, and pulmonary arterial hypertension (PAH). The combination of these anatomical and functional anomalies, in varying degrees, explains the wide variability of symptoms at birth. Diagnosis is usually made prenatally by ultrasound, which enables severe forms of the disease to be detected and appropriate management initiated. The prognosis remains generally grave, with a neonatal mortality rate of between 30% and 60% depending on the study, and around half of all children will have long-term sequelae.

Keywords

Congenital Diaphragmatic Hernia, Prenatal Diagnosis, Pulmonary Hypoplasia, Pulmonary Hypertension, Medical-Surgical Treatment

1. Introduction

Congenital diaphragmatic dome hernia (CDHD) is an anomaly characterized by a defect in the complete or partial formation of the diaphragmatic dome, resulting in a variable deficit. It has a prevalence of around 1 in 3500 births [1] [2]. Diagnosis is usually made prenatally during an ultrasound scan, which enables appropriate management to be recommended depending on the severity of the CDHD. The defect causes the abdominal viscera to migrate into the thoracic cavity. This anomaly is associated with bilateral pulmonary hypoplasia, a reduced number of pulmonary vessels, and pulmonary hypertension. The coexistence of these anatomical and functional anomalies explains the diversity of symptoms encountered at birth [1].

In 80% of cases, the diaphragmatic breach is located in the posterolateral part of the left dome, commonly known as Bochdalek's hernia. Other locations include right-sided hernias (10% - 15%) and bilateral hernias (1%). Diaphragmatic breaches may be isolated or associated with other anomalies, such as pulmonary, intestinal, cardiac malformations and/or chromosomal abnormalities [2].

The prognosis remains grave, with an overall neonatal mortality rate of 30% - 60%, depending on the study. More than half of children with CDH lead entirely normal lives, while others may experience various difficulties (respiratory, nutritional, orthopedic, and pulmonary hypertension), which tend to improve with time [1].

The therapeutic management of congenital diaphragmatic hernias (CDH) varies according to when they are discovered. Forms discovered in the neonatal period require urgent intervention involving the collaboration of neonatologists, anaesthetists and paediatric surgeons [3]. Unlike when diagnosed at birth, when respiratory distress can be directly life-threatening, late discovery of the disease is characterized by generally moderate respiratory and digestive symptoms, reducing post-operative morbidity and offering a favorable prognosis [4]. Our study aims to investigate the epidemiological, clinical, diagnostic, therapeutic and evolutionary aspects of congenital diaphragmatic hernias with neonatal revelation.

2. Materials and Methods

This is a retrospective study of the records of 9 cases of congenital diaphragmatic hernia collated between the months of June 2018 and May 2024 in the neonatology and neonatal intensive care unit of the Mohammed VI University Hospital in Oujda, based on exploration of medical records, registers of entrants and operative reports. All neonates with congenital diaphragmatic hernia admitted to the neonatal intensive care unit during the study period were included, whether or not they underwent surgery. All neonates with congenital diaphragmatic hernia outside our study period, cases of diaphragmatic ventration and hiatal hernia, as well as congenital diaphragmatic hernia diagnosed after the neonatal period, were excluded from the study. All files were processed using an operating sheet, and data entry was performed using Microsoft Office Excel 2016.

3. Parameters Studied

Analysis of the medical records enabled the following data to be analyzed: Patient age and sex, gestational age in weeks, age at symptom onset, patient growth metrics, patient's background, maternal history, pregnancy follow-up, course of delivery, patient clinical presentation, imaging examinations, including antenatal ultrasound, chest X-ray, transthoracic ultrasound (TTE) and abdominal ultrasound, associated malformations or chromosomal abnormalities, treatment received by patients, herniated organs, post-operative course, evolution and duration of hospital stay.

4. Results

In the neonatology and neonatal intensive care unit, we collected 9 confirmed cases of HCCD that met the inclusion criteria, over a 6-year period from June 1, 2018 to May 31, 2024, representing an incidence of 1.5 cases per year. All patients studied were newborns, born at term, without intrauterine growth retardation (IUGR) or low weight for gestational age. The average weight in our series was 3280 grams. Our series included 6 boys and 3 girls, resulting in a sex ratio of 2, indicating a clear male predominance. Five of our patients exhibited symptoms of respiratory distress immediately at birth, while the remaining four developed symptoms after more than 5 days.

Maternal age ranged from 22 to 34 years. All newborns studied were born to multiparous mothers with at least one healthy child, and no history of child death.

Two patients were referred from peripheral health facilities. In our series, newborns from consanguineous marriages accounted for 28.5%. Additionally, 28.5% of the pregnancies were poorly attended, despite medicalized deliveries.

Two pregnancies in our series (28.5%) were poorly attended, with a medicalized delivery. Delivery was by caesarean section for 57% of the mothers.

Antenatal diagnosis was established with certainty in one of our patients (11.1%).

In our series, HCCD was diagnosed within the first 24 hours of life in 4 newborns (44.4%), while the remaining 4 were diagnosed after 5 days of life.

The presenting symptoms were dominated by respiratory issues, which accounted for 100% of manifestations, characterized by neonatal respiratory distress and bouts of cyanosis.

Vomiting occurred in two patients (22.2%). There were no transit disorders, and meconium was emitted on time in all patients. All neonates showed respiratory instability, with ambient oxygen saturation levels ranging from 60% to 90%. Clinical assessments for malformations were negative in all patients.

Radiological diagnosis relied on chest radiography, performed on all patients, which strongly suggested the diagnosis.

The most suggestive and frequent semiological signs in all patients included Intrathoracic and mediastinal deviation to the right.

Abdominal and transfontanellar ultrasounds were conducted in 5 patients (55.5%) as part of the malformation work-up, and no abnormalities were detected.

Doppler echocardiography was performed on 2 patients in our series (22.2%). One was normal, while the other showed a right-sided heart with pulmonary arterial hypertension (PAH). Neither Chest CT nor MRI scans were performed in any of our patients.

A systematic biological work-up was performed on admission to detect preoperative abnormalities, in particular hydroelectrolyte imbalances, haemostasis disorders and infectious syndrome. In our series, two cases presented a positive infectious work-up on admission, suggesting a maternal-fetal infection of undetermined origin, which further aggravated their condition. However, none of the patients in our series benefited from gasometry.

All neonates were hospitalized in the neonatal intensive care unit prior to surgery, with an average length of stay of 1.6 days, ranging from 1 to 3 days. In our series, 8 patients underwent surgery. 4 neonates underwent surgery 24 hours after hospitalization, 3 neonates after 48 hours of hospitalization, and 1 patient underwent surgery 72 hours after admission. All patients had benefited from preoperative stabilization for 24 to 48 hours. For the management of pulmonary arterial hypertension (PAH), sildenafil was administered in 2 of our patients (22.2%).

Surgical treatment aimed to reintroduce the digestive contents into the abdomen and close the edges of the defect. Consequently, 8 of our patients underwent surgery: 7 had a posterolateral Bochdalek hernia (87.5%), while 1 patient had a retro-costo-xiphoid hernia. Unfortunately, 1 patient died before surgery could be performed.

The approach used in all patients operated on in our series was laparotomy, with a left subcostal incision in 66% of cases and a left supraumbilical transverse incision in 33%.

Surgical exploration revealed a large diaphragmatic defect in 4 patients in our series, accounting for 50% of those operated on. In terms of pulmonary status, left-sided pulmonary hypoplasia was present in 5 patients, which accounts for 62.5% of cases.

The organs herniated were the transverse colon, stomach, small intestine, and right colon, followed by the spleen and pancreas (**Table 1**, **Table 2**). Liver and kidney hernias were not observed in the patients who underwent surgery on in our series.

Following surgery, one patient had an uncomplicated immediate postoperative course, whereas 7 patients experienced complications that ultimately led to death. These complications primarily included pneumothorax, pulmonary hypertension, and healthcare-associated infections.

In our study, mortality in patients with congenital diaphragmatic hernia generally resulted from a combination of factors, primarily severe respiratory failure, and septic shock. It should be noted that five patients in our series had pulmonary hypoplasia. In addition, three patients developed pneumothorax, treated by pleural drainage, and only one case was diagnosed with severe pulmonary arterial hypertension (PAH). Another contributing factor to mortality was septic shock, occurring either as a result of nosocomial infection or maternal-fetal in-

fection. This complication exacerbated patients' respiratory distress (Figure 1).

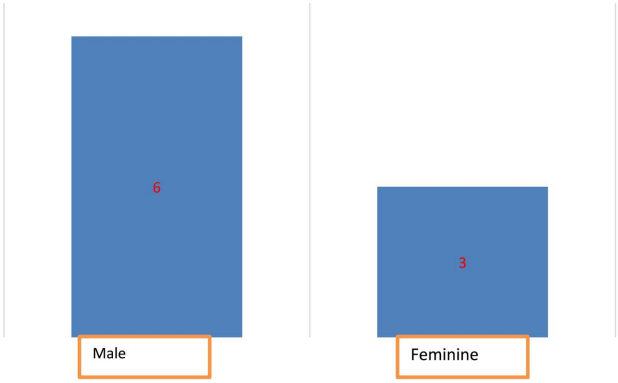

Figure 1. Distribution of patients by gender.

Table 1. Summary table of data from 9 patients with congenital diaphragmatic hernia.

Presentation of patients	Case 1	Case 2	Case 3	Case 4	Case 5
Prenatal diagnosis	Done by prenatal ultrasound without further details	No	No	No	No
The onset of symptoms	1st day of life	1st day of life	1st day of life	1st day of life	8th day of life
Symptomatology indicative of the diagnosis	Signs of respiratory distress	Signs of respiratory distress	Signs of respiratory distress	Signs of respiratory distress	Respiratory distress + vomiting
thoracoabdominal radiography	Intrathoracic gastrointestinal clarity	Intrathoracic gastrointestinal clarity	Intrathoracic gastrointestinal clarity	Intrathoracic gastrointestinal clarity	Intrathoracic gastrointestinal clarity
Laboratory tests	Normal	Normal	biological inflammatory syndrome	Normal	Normal
Associated malformations or chromosomal anomalies	no apparent malformations	Left hypoplastic lung.	The entire left lung is hypoplastic	Absent	Left hypoplastic lung
Type of hernia		Left posterolateral	Retrocosto-xiphoid hernia of Moragani-Larrey	Left posterolateral	Left posterolateral
Surgical treatment	Not done due to the instability of the newborn upon admission	2nd day of life: A large diaphragmatic defect	2nd day of life.	3rd day of life: A large diaphragmatic defect	4 days after being hospitalized
Postoperative progress	Death before surgery	Death on postoperative day 5 due to cardiorespiratory arrest following severe respiratory failure.	Death on postoperative day 5 due to cardi-orespiratory arrest fol-lowing severe respira-tory failure.	Good recovery	Death on posto perative day 5 due to cardiorespiratory arrest following severe respiratory failure.

Continued

Presentation of patients	Case 6	Case 7	Case 8	Case 9
Prenatal diagnosis	No	No	No	No
The onset of symptoms	8th day of life	6th day of life	1st day of life	7th day of life
Symptomatology indicative of the diagnosis	Signs of respiratory distress + vomiting.	Signs of respiratory distress.	Signs of respiratory distress.	Signs of respiratory distress.
thoracoabdominal radiography	Intrathoracic gastrointestinal clarity.	Intrathoracic gastrointestinal clarity.	Intrathoracic gastrointestinal clarity.	Intrathoracic gastrointestinal clarity.
Laboratory tests	Normal	biological inflammatory syndrome.	Normal	Normal
Associated malformations or chromosomal anomalies	Not mentioned in the operative report	Total left pulmonary hypoplasia.	left pulmonary hypoplasia.	no apparent malformations.
Type of hernia	Left posterolateral	Left posterolateral.	posterolateral	Left posterolateral
Surgical treatment	Surgically operated 2 days after admission Large diaphragmatic defect.	Surgically operated 2 days after admission.	4 days after being hospitalized.	5 days after being hospitalized: A large diaphragmatic defect
Postoperative progress	Death due to cardiopulmonary arrest from suffocating pneumothorax.	Death 24 hours after surgery due to cardiopulmonary arrest from septic shock.	Death 48 hours after surgery due to cardiopulmonary arrest from septic shock.	Death 3 days after surgery due to cardiopulmonary arrest from septic shock.

Table 2. The distribution of patients operated on according to the herniated viscera identified intraoperatively.

Herniated viscera	number	Percentage
Transverse colon	6	75%
Stomach	5	62.5%
Small intestine	4	50%
Right colon	4	50%
Rate	3	37.5%
Pancreas	2	25%

5. Discussion

In our study, the incidence was 1.5 cases per year, and the sex ratio (M/F) was 2, with a very marked predominance of Bochdalek hernias. The most frequent symptoms were respiratory, followed by digestive. The diagnosis was evoked antenatally in one patient, and thoracoabdominal radiography was suggestive of the diagnosis in all patients. 8 patients underwent laparotomy. The immediate postoperative course was uncomplicated in only 1 patient, while death occurred in 7. Post-operative complications leading to death were pulmonary hypertension, pneumothorax, and septic shock.

It is difficult to pinpoint the exact epidemiological characteristics of HCCD.

This rare malformation has a poor prognosis and a significant underlying mortality rate [5].

Its reported frequency in the literature is around 1 in 3500 live births [1] [2] [6], as is its male predominance, with a higher incidence in boys than in girls, exceeding 50% [7] [8]. Our series' results are consistent with these findings. In our context, there are no studies that specify with exactness the frequency of this condition.

The involvement of the diaphragm is typically observed on the left side in approximately 80% - 90% of cases, on the right side in 10% - 15%, and is exceptionally bilateral, occurring in less than 1% of cases [2] [6]-[9]. This pattern is also supported by our series' findings. The frequency of malformations associated with HDC varies depending on the study's sample size. It has been reported to range from 28.2% to 40% in larger studies and up to 80% in smaller-scale studies [10]-[12]. Genetic abnormalities are present in 10% of cases, of which 27% are associated with syndromes and 73% with chromosomal abnormalities [13] [14]. Abdominal and transfontanellar ultrasounds were performed as part of the malformation evaluation in 5 of our patients, accounting for 55.5%. No abnormalities were observed. The diagnosis of HCCD can be made during pregnancy by imaging studies such as routine prenatal ultrasounds. Alternatively, it may be diagnosed postnatally in the presence of neonatal respiratory distress or, in some cases, later due to atypical symptoms or even during routine medical follow-up examinations [8].

In our series, the antenatal diagnosis was established with certainty in one of our patients, *i.e.* 11.1%. Front and side thoracic radiography strongly suggest the diagnosis of HCCD due to the presence of intra-thoracic digestive tract visualization, which is a hallmark sign of this condition [15] [16]. Every patient in our series underwent a thoracoabdominal radiograph, which enabled the diagnosis to be made. Other essential examinations include cardiac echocardiography to detect pulmonary hypertension, assess cardiac function, and rule out associated cardiac malformations [17]. In our series, Doppler echocardiography was performed in 2 patients (22.2%); one was normal, while the other showed right-sided heart abnormalities with pulmonary arterial hypertension (PAH). Diaphragmatic ultrasound or fibroscopy may be recommended to distinguish HCCD from diaphragmatic eventration, while CT and MRI are rarely necessary in this context [17].

Stabilization of patients before surgery focuses on ensuring adequate oxygenation, reducing airway pressures, and age-appropriate blood pressure [18]. For managing pulmonary arterial hypertension (PAH), sildenafil was administered to 2 of our patients, accounting for 22.2%. Several surgical approaches are possible, but laparotomy is generally preferred [19]. In our series, all patients underwent laparotomy. The organs herniated vary depending on the side of the lesion. For instance, the stomach was involved in 40 to 63.2% of cases, the colon or entire intestine in 57.9%, the spleen or liver in 36.8%, and the kidney in 5.2% [4] [20]. Our findings generally align with these patterns, with the exception that liver and kid-

ney hernias were not observed in the patients operated on in our series.

Post-surgical management of HCCDs generally involves addressing immediate complications. Frequent early postoperative complications include surgical site infection, bleeding (especially when ECMO is used), early recurrence (particularly after laparoscopic repair of large hernias), chylothorax (associated with repairs using a prosthesis or ECMO), intra-abdominal compartmental syndrome, as well as pleural effusion (frequently encountered but rarely requiring drainage) [21]. In our series, immediate postoperative complications were dominated by pneumothorax and healthcare-associated infections. However, some series report the absence of immediate postoperative complications [22]-[24].

Mortality associated with HCCD is influenced by several factors, including lesion size, birth weight, blood gas results, presence of pulmonary hypertension, and association with chromosomal abnormalities and cardiac malformations [21] [25] [26]. Some studies indicate a survival rate of up to 90% [25]. However, population-based studies indicate a survival that generally varies between 42% and 68%. This variation could be explained by the hidden mortality of HCCD, including deaths in utero and those occurring before hospital admission [17] [25]. In contrast to the situation where it is diagnosed at birth, a period when respiratory distress can directly compromise vital prognosis, late discovery of the disease manifests itself in generally less severe respiratory and digestive symptoms, reducing postoperative morbidity and improving prognosis [4]. By assessing the prenatal and postnatal risk associated with infants with CDH, families and healthcare professionals can educate and advise families while developing a strategy based on best practices and the level of risk. This approach includes postnatal interventions such as extracorporeal life support (ECLS), and more recently, exploring the potential for prenatal interventions like fetoscopic endoluminal tracheal occlusion (FETO) and the use of new pharmacological therapies [27]. In our context, the lack of technical resources may explain the high mortality in our series.

6. Conclusion

Congenital diaphragmatic dome hernia is a rare condition. The most common form is Bochdalek's hernia or left posterolateral hernia. CCHD is more common in male children. Prenatal ultrasound diagnosis enables early and optimal management. After birth, X-rays are sufficient to raise the suspicion of the diagnosis. The prognosis depends on the severity of pulmonary hypoplasia and pulmonary hypertension. At present, it is recommended not to opt for emergency surgery, but rather for preoperative stabilization to improve prognosis. Unlike when diagnosed at birth, when respiratory distress can be directly life-threatening, late discovery of the disease is characterized by generally moderate respiratory and digestive symptoms, reducing post-operative morbidity and offering a favorable prognosis. The availability of technical resources can improve the prognosis, which explains the higher mortality in developing countries compared to developed countries.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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