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(RESEARCH ARTICLE)

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Congenital diaphragmatic hernia (A report on 5 cases)

Leila Kartout, Ilham Elouardighi and Amina Barkat *

National Reference Center for Neonatology and Neonatal Intensive Care. Rabat Children's Hospital (RCH). Research Team on Maternal and Child Health and Nutrition. Faculty of Medicine and Pharmacy. Mohammed V Souissi University. Rabat. Morocco.

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Abstract

Introduction: Congenital diaphragmatic hernia (CDH) is defined by the failure of all or part of the diaphragmatic dome to develop; this anomaly results in the presence of certain abdominal organs within the thoracic cavity during critical stages of fetal lung development. In 80% of cases, this involves the posterolateral portion of the left dome, known as a Bochdalek hernia.

Materials and Methods: The aim of our study was to conduct a retrospective analysis of five cases of congenital diaphragmatic hernia collected between January 1 2023, and December 31 2025, at the National Reference Center for Neonatology and Neonatal Intensive Care in Rabat Morocco.

Results: Among our patients, the sex ratio was 4 with a clear predominance of females. Diaphragmatic involvement is usually unilateral, with a preference for the left side in nearly 80 to 90% of cases. In our study, there were two cases of associated malformations (one case of trisomy 21 and one case of type 1 omphalocele with agenesis of the corpus callosum). None of the newborns had a prenatal diagnosis, and the clinical presentation was dominated by respiratory symptoms, with one patient diagnosed at 15 days of life with bronchiolitis. A chest X-ray is the key test for establishing the diagnosis; it was performed on all newborns and was highly suggestive of the diagnosis. A chest CT scan was performed on two patients, aided in the diagnosis, and provided insight into the herniated organs. All patients received pre- and postoperative resuscitation to optimize their care. Surgical treatment which involved reducing the herniated organs and closing the diaphragmatic defect was performed on four patients; one patient died before the surgery. The short-term outcome was favorable for all patients who underwent surgery.

Conclusion: Congenital diaphragmatic hernia is a rare but serious condition. The prognosis remains poor and depends on the presence and severity of pulmonary hypoplasia and the presence of associated congenital malformations.

Keywords: Congenital Diaphragmatic Hernia; Respiratory Distress; Chest X-Ray; Surgical Treatment

1. Introduction

Congenital diaphragmatic hernia (CDH) is defined by the failure of all or part of the diaphragmatic dome to develop; this anomaly results in the presence of certain abdominal organs within the thoracic cavity during critical stages of fetal lung development. In 80% of cases, this involves the posterolateral portion of the left dome, known as a Bochdalek hernia.

* Corresponding author: A Barkat

1.1. Clinical Case 1

A full-term, healthy female newborn was admitted on day 14 of life with a history of bronchiolitis accompanied by rhinorrhea and respiratory distress rated 2/10 on the Silverman scale. A chest X-ray revealed a left intrathoracic air-fluid level [Figure 1]. The evaluation for congenital anomalies was negative, and the patient underwent surgery with a favorable outcome.

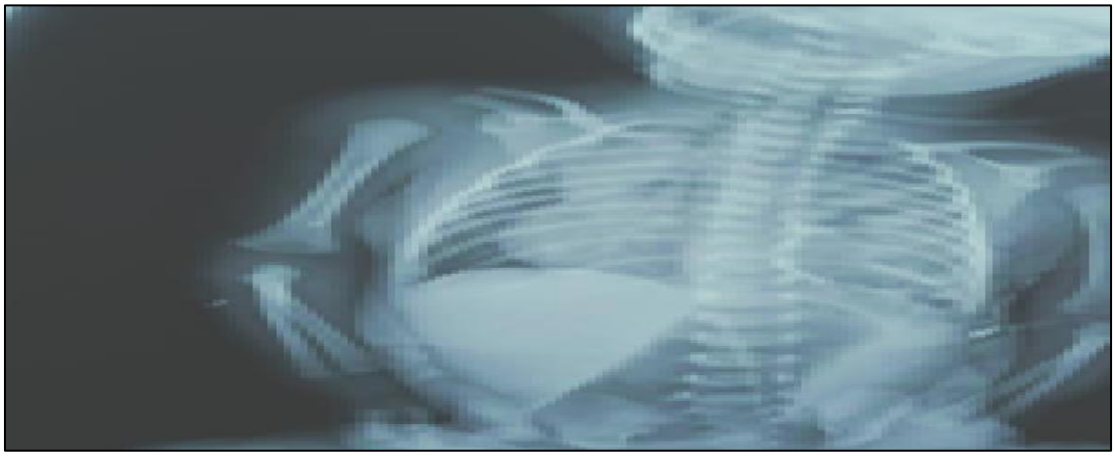

Figure 1 Left intrathoracic air-fluid level

1.2. Clinical Case 2

A full-term, eutrophic female newborn admitted to the neonatal intensive care unit (NICU) at 3 days of life for respiratory distress; on clinical examination, the infant was cyanotic, hypotonic, had Mongoloid facial features, and exhibited a respiratory distress score of 2/10 with abnormal heart sounds. The chest X-ray revealed an intra-thoracic opacity on the right side with digestive tract shadows [Figure 2]. A chest CT scan confirmed the presence of a Bouchdalek hernia. The newborn underwent surgery and is doing well.

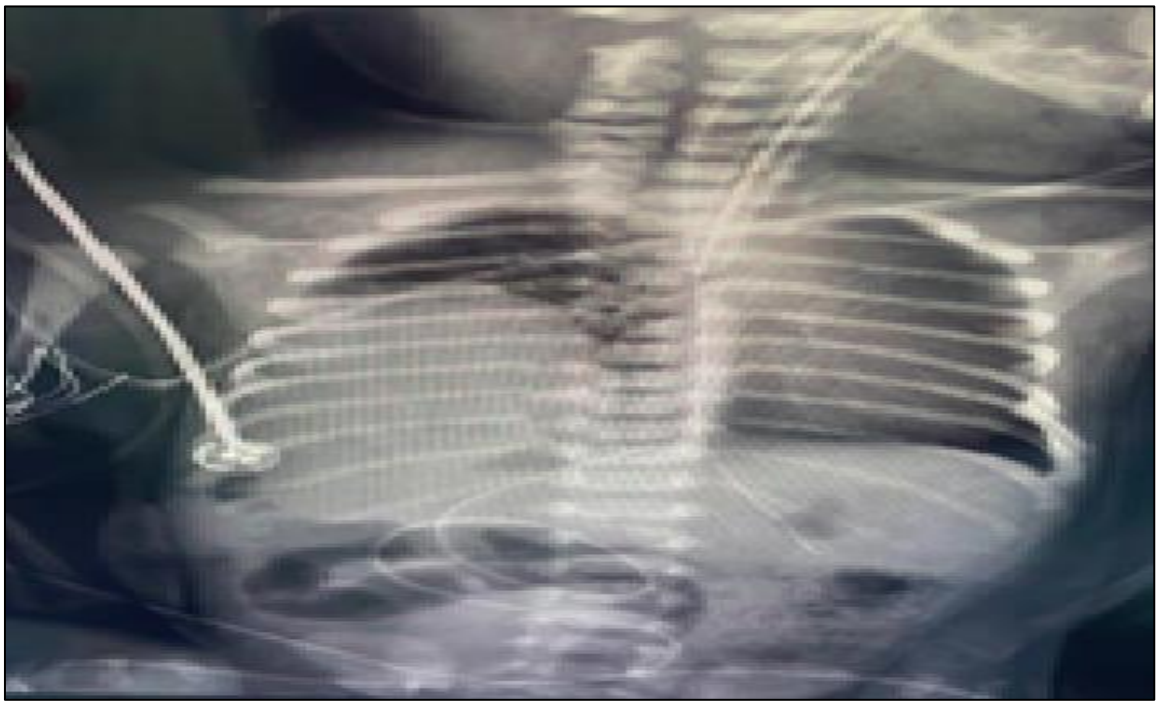

Figure 2 An intra-thoracic opacity on the right side with digestive tract shadows

1.3. Clinical Case 3

A full-term, normal-weight female newborn was admitted for respiratory distress; on physical examination, she presented with a distended left hemithorax and a rightward shift of heart sounds. A chest and abdominal X-ray was performed, revealing digestive tract opacities in the left intrathoracic space [Figure 3]. The newborn underwent surgery the day after admission and made a good recovery.

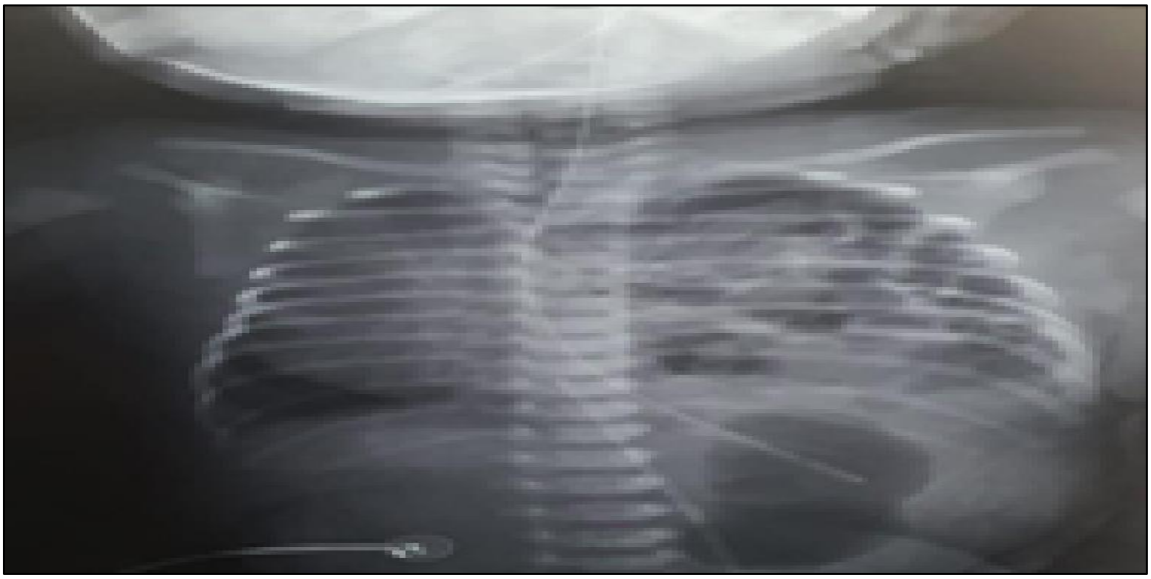

Figure 3 Digestive tract opacities in the left intrathoracic space

1.4. Clinical Case 4

A male newborn was admitted at birth for respiratory distress on a scale of 2/10 with type 1 omphalocele; on clinical examination, decreased vesicular breath sounds were noted in the right basal region. A chest and abdominal X-ray was performed, revealing a well-defined intra-thoracic opacity on the right side [Figure 4]. A chest CT scan revealed a right diaphragmatic hernia at hepatic segment VIII without pulmonary hypoplasia; a brain CT scan showed agenesis of the corpus callosum. The newborn died on day 3 of life prior to surgery.

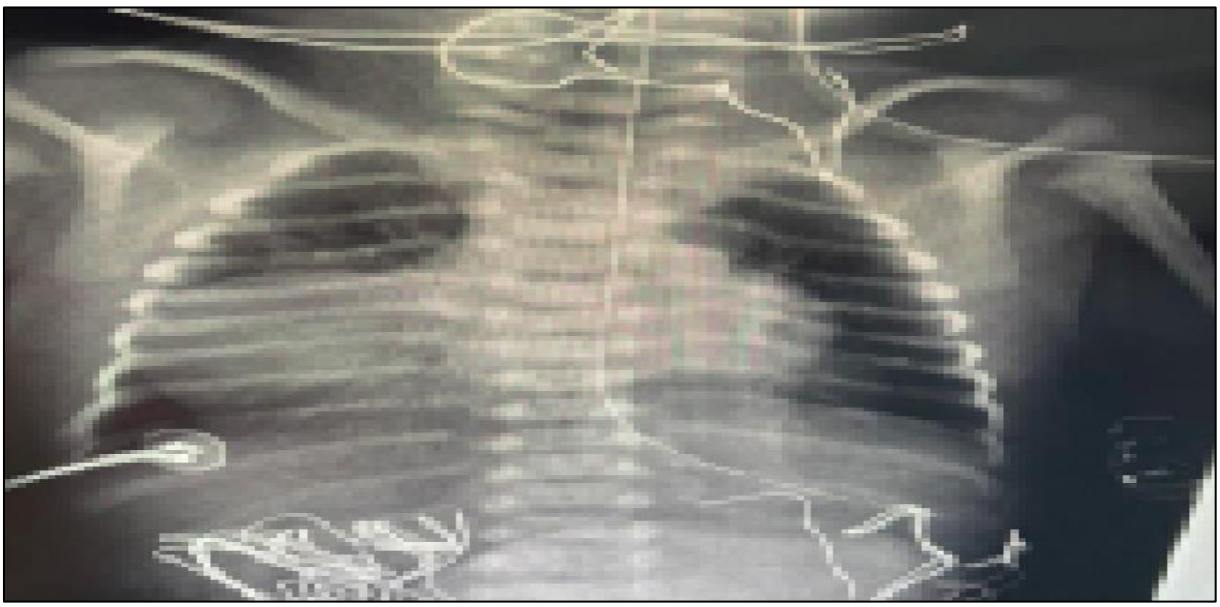

Figure 4 Intra-thoracic opacity on the right side

1.5. Clinical Case 5

A full-term, healthy female newborn was admitted for respiratory distress. On examination, she presented with grade 2/10 respiratory distress, a globular left hemothorax, and a flat abdomen. A chest and abdominal X-ray were performed, revealing digestive tract contents in the left intrathoracic space [Figure 5]. The newborn underwent surgery on day 10 of life following hemodynamic stabilization, with a favorable postoperative course.

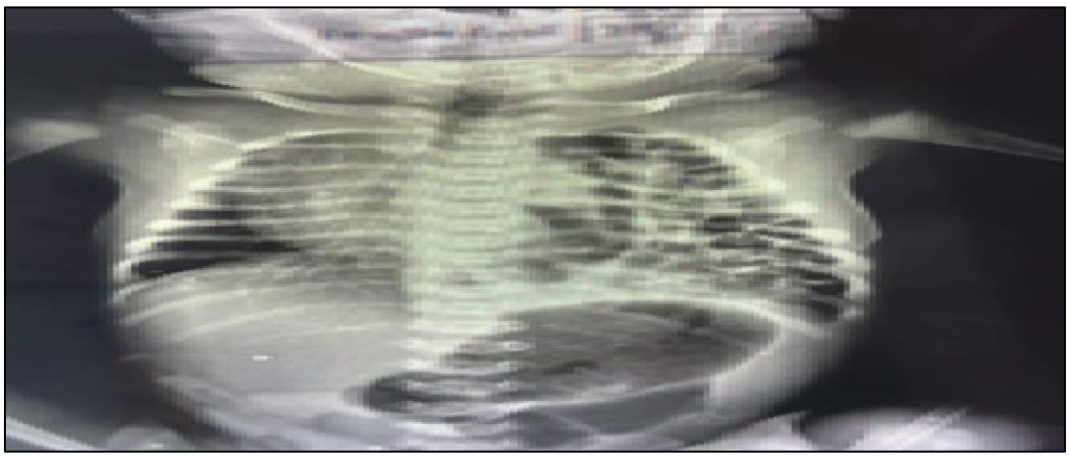

Figure 5 Digestive tract contents in the left intrathoracic space

2. Discussion

Congenital diaphragmatic hernia (CDH) is a rare malformation characterized by an opening in the diaphragm. This defect allows certain abdominal organs to migrate into the chest cavity, preventing optimal lung development. In the literature, the reported incidence of congenital diaphragmatic hernia ranges from 1 in 3000 to 1 in 5000 live births [2, 3]. It occurs 30 to 50% more frequently in boys than in girls. Diaphragmatic involvement is usually unilateral, with a predilection for the left side in nearly 80 to 90% of cases [2].

CDH is often associated with pulmonary hypoplasia, anatomical and functional abnormalities of the pulmonary vasculature, and underdevelopment of the left heart. The presence and severity of these malformations, as well as their association with other congenital malformations, increase the severity of the condition. [1, 4]. The diagnosis is usually made during pregnancy using an ultrasound. The age at diagnosis of CDH varies depending on whether it is a neonatal or delayed presentation.

Late-onset CDH accounts for 5 to 30% of cases [6]. This diagnostic delay is attributed to the wide clinical variability of the condition, which sometimes presents with misleading and diverse symptoms including, but not limited to, respiratory, gastrointestinal, and occasionally infectious signs thereby making diagnosis difficult. A chest X-ray is the key test for making the diagnosis [1].

The management of this condition is complex and requires a multidisciplinary team (obstetricians, pediatric intensivists, and surgeons). Treatment begins with a preoperative stabilization phase, which appears to play an important role in improving survival [1, 2]. This is followed by surgery, which involves repositioning the organs in the abdomen and closing (or repairing) the hole in the diaphragm.

The immediate postoperative course of CDH is generally uneventful. Early postoperative complications typically include surgical site infection, bleeding, early recurrence, chylothorax, intra-abdominal compartment syndrome, and pleural effusion. [5, 7].

3. Conclusion

Congenital diaphragmatic hernia is a rare but serious condition. The prognosis remains poor and depends on the presence and severity of pulmonary hypoplasia and the presence of associated congenital malformations.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict-of-interest to be disclosed.

Statement of informed consent

Informed consent was obtained from all individual participants included in the study.

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