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Case Report

Delayed presentation of congenital diaphragmatic hernia: A report of 2 cases ☆,☆☆

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ABSTRACT

Late presentation of congenital diaphragmatic hernia (CDH) presents usually after the neonatal period and often misdiagnosed for other respiratory pathologies. It is crucial to differentiate late presentation of CDH from other potential causes of respiratory distress and gastrointestinal symptoms. Herein, we present 2 cases of delayed presentation of congenital diaphragmatic hernia in infants. Initially, both cases were managed as respiratory conditions in outpatient settings, with no significant improvement. The correct diagnosis was eventually made through radiological evaluation at our tertiary centre, leading to successful surgical management. Delayed presentation of CDH beyond the neonatal period is rare, owing to the wide spectrum of clinical manifestations. Early diagnosis and surgical management are crucial to reduce morbidity and mortality, making a high index of suspicion essential for timely intervention.

Introduction

Congenital diaphragmatic hernia (CDH) affects approximately 1 in 2500 neonates and is characterized by abnormal diaphragmatic development, with a survival rate of around 67% [1]. CDH is most commonly associated with neonatal respiratory distress. However, delayed presentation occurs in 2.5% to 20% of all CDH cases [1]. The hernia predominantly occurs on the left side (80-85%), with 15%-20% on the right and 2% being bilateral [2,3]. In some patients, the symptoms manifest later or present with milder symptoms, while 25% may remain asymptomatic. Patients with delayed manifestations tend to have a better prognosis due to more developed lungs [1–3]. In this series, we present 2 children who exhibited long-term intermittent respiratory symptoms and were initially treated for

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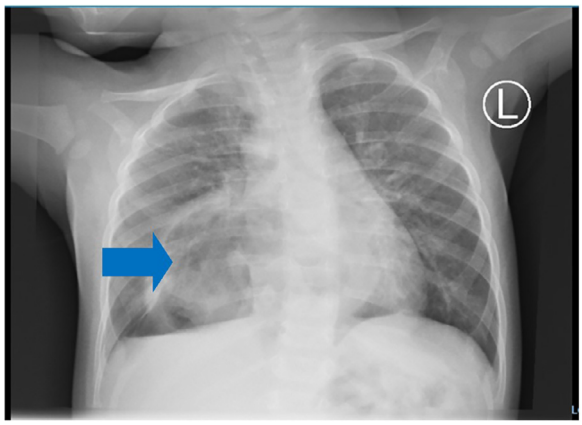

Fig. 1 – Plain chest X-ray showing multicystic mass-like lesion overlying the central cardiac shadow extending towards the right hemithorax (yellow arrow), suggestive of congenital diaphragmatic hernia.

upper respiratory tract conditions before receiving a correct diagnosis of CDH. This manuscript was prepared following the CARE guidelines (https://www.care-statement.org).

Case report

Case 1

A 7-month-old female presented with a 3-day history of cough, chest tightness, and difficulty sleeping comfortably at night. The cough was dry and not accompanied by fever. The mother noted a similar episode a few weeks prior, which improved somewhat with oral antibiotics. Despite the symptoms, the child maintained normal feeding and bowel habits, and her growth was reported as fair. She was born at term with no complications.

On physical examination, the child appeared healthy and well-nourished, with no signs of pallor, dyspnea, or cyanosis. Her vital signs were within normal limits, with oxygen saturation at 98% on room air. However, she exhibited bilateral lower chest indrawing and crepitations on auscultation, more pronounced on the right side. Laboratory tests were unremarkable, showing no leukocytosis, hemoglobin at 10.8 g/dL, and normal levels of serum potassium, sodium, creatinine, and liver enzymes.

A plain chest X-ray revealed features consistent with a right-sided congenital diaphragmatic hernia (Fig. 1), which was further confirmed by a chest CT scan (Fig. 2). An echocardiogram (ECHO) was performed and ruled out any associated cardiac anomalies.

The child was admitted and scheduled for diaphragmatic hernia repair. During surgery, a 3 cm posterolateral defect was identified, with abdominal contents herniating into the right hemithorax with no sac, limiting the expansion of the right lung (Fig. 3). The herniated contents were reduced, and the diaphragmatic defect was successfully closed primarily using running silk sutures.

Postoperatively, the child recovered well from anesthesia and progressed satisfactorily in the general ward. She received chest physiotherapy as part of her recovery plan and was discharged on day 6, showing no chest symptoms.

At a follow-up visit 3 weeks later, the surgical incision had healed, and the child was clinically stable. A control chest X-ray demonstrated adequate lung expansion (Fig. 4), confirming the success of the surgical intervention.

Case 2

A 2-year-old female child with a known history of cerebral palsy presented with an intermittent dry cough that the mother reported had worsened. The cough was accompanied by low-grade fevers, mostly occurring at night. However, the mother denied any history of difficulty in breathing. The child was feeding well and had normal bowel and urine motions. She had been regularly attending physiotherapy and occupational health clinics for her cerebral palsy.

On examination, the child appeared alert, awake, and playful, with a body weight of 8400 grams. She was not pale or jaundiced and was well-nourished. Her pulse rate was 140 beats per minute, and she was saturating at 96% on room air. Laboratory investigations revealed a normal complete blood

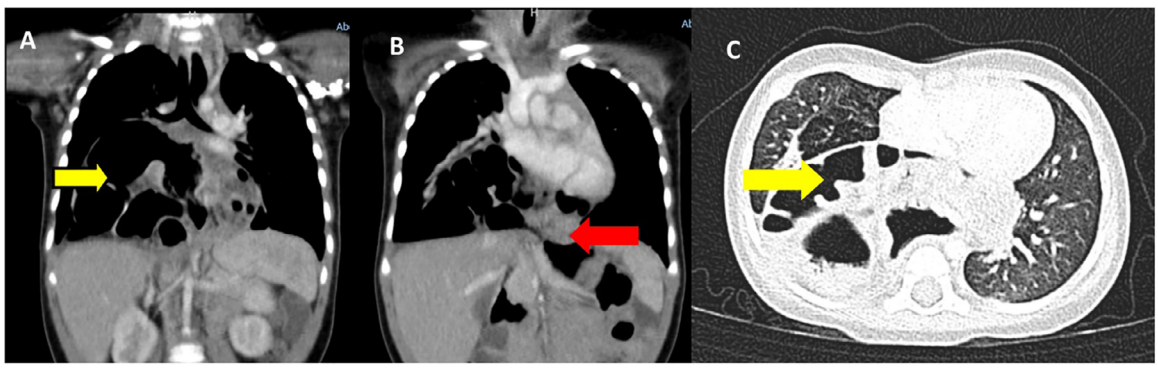

Fig. 2 – CT-scan of the chest revealing a 2 cm defect on the right central diaphragm (red) with herniation of the stomach, duodenum and multiple bowel loops into the right hemithorax (yellow arrows).

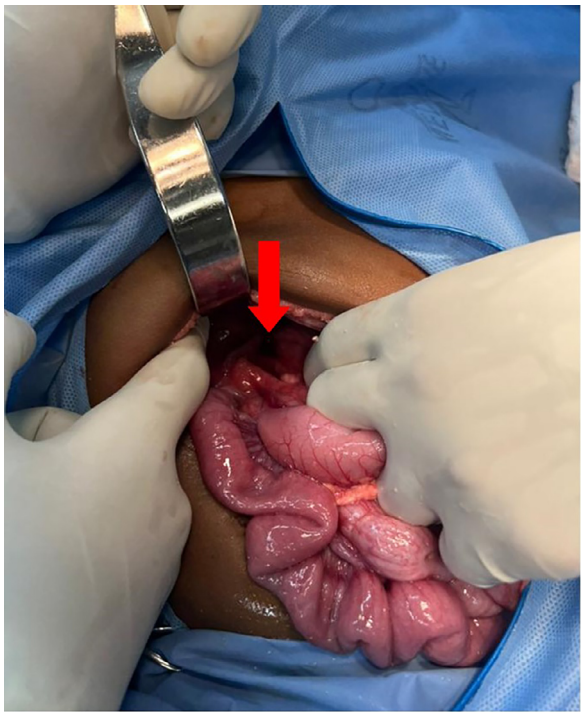

Fig. 3 – Intraoperative photograph showing Bochdalek diaphragmatic hernia (red arrow).

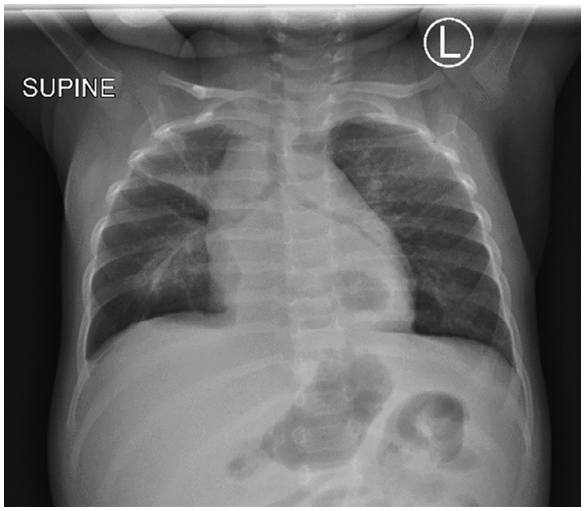

Fig. 4 – Post operative plain chest X-ray showing adequate lung expansion.

count, with a hemoglobin level of 12.4 g/dL, and normal biochemistry results.

Her plain chest X-ray revealed the presence of multiple bowel loops within the right hemithorax, suggestive of a right diaphragmatic hernia (Fig. 5). This diagnosis was further confirmed by a CT scan, which showed a right diaphragmatic defect with portions of the liver and intestines herniating into the right hemithorax (Fig. 6). The child underwent a laparotomy for the repair of the defect, during which a 3 × 5 cm Bochdalek hernia was identified, with multiple bowel loops herniated through it contained in a hernia sac, compressing the lung parenchyma. The herniated contents were successfully reduced, and the diaphragmatic defect was repaired primarily with interrupted silk sutures after excising the hernia sac.

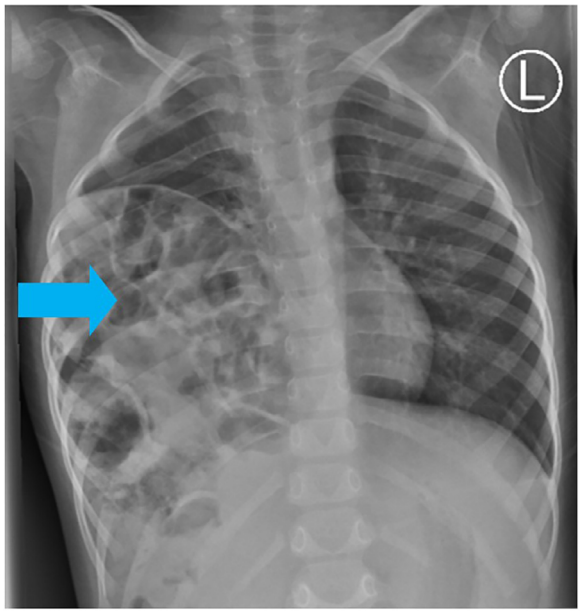

Fig. 5 – Plain chest X-ray showing multiple bowel loops in the right hemithorax (blue arrow) suggestive of congenital diaphragmatic hernia.

The child had an uneventful recovery, benefiting from chest physiotherapy, and was discharged on day 7 postsurgery. At her follow-up visit 3 weeks later, she remained asymptomatic and continued her regular visits to the physiotherapy and occupational therapy clinics for cerebral palsy management.

Discussion

There are 3 main types of congenital diaphragmatic hernia (CDH): Bochdalek hernia (70%), which is located in the posterolateral aspect of the diaphragm; Morgagni hernia (25%-30%), found in the anterior aspect; and the central type (2%-5%) [2]. Both cases in our series presented with left-sided Bochdalek CDH. Typically, the majority of CDH cases are diagnosed antenatally and present with respiratory distress during the neonatal period; however, this was not the case here, as both patients presented later [3]. If CDH is diagnosed later than 30 days it is considered late-onset CDH [4]. The exact causes of CDH are poorly understood, but some evidence suggests that genetic and environmental factors may play a role [5].

Generally, CDH results from the incomplete closure of the pleuroperitoneal canal during fetal development [5]. In adulthood, diaphragmatic hernia can develop due to various factors

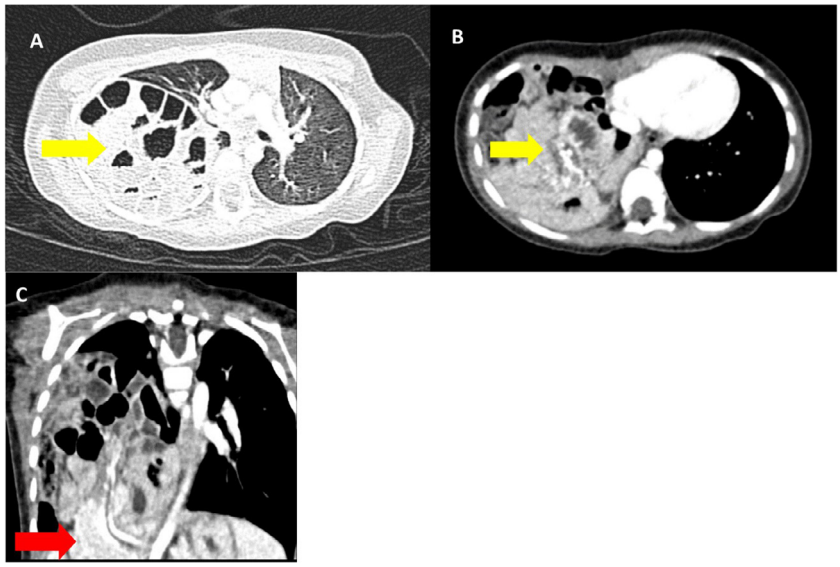

Fig. 6 – (A and B) Axial view CT-scan showing right posterolateral aspect of the diaphragm through which herniation of part of the stomach, liver, pancreas and bowel loops herniating into the right hemithorax (yellow arrows), (C) Coronal view showing 3.3 cm diaphragmatic defect (red arrow).

other than CDH. These factors include trauma, phrenic nerve palsy, and the delayed diagnosis of a hiatus hernia. These conditions can lead to a weakened or compromised diaphragm, allowing abdominal organs to herniate into the thoracic cavity [2].

Delayed presentation of CDH often occurs due to vague symptoms and the inability of caregivers to detect the underlying pathology, which can lead to misdiagnosis, mismanagement, and complications. Pneumothorax and pleural effusion are often the most common initial diagnoses made on a chest X-ray in cases of late-presenting CDH. However, distinguishing between CDH and tension pneumothorax is crucial, as the management of these conditions differs significantly. Late-presenting CDH can manifest with acute or chronic respiratory or gastrointestinal symptoms, including dyspnea, recurrent pulmonary infections, chest pain, postprandial fullness, or abdominal pain, making accurate diagnosis essential for appropriate treatment. Patients with right-sided CDH become symptomatic at a younger age compared to patients with left-sided defects [4,6].

As noted by Abdur-Rahman et al. [7], many patients are initially treated for upper respiratory tract infections, pneumonia, or even misdiagnosed with pneumothorax. This was also evident in our index cases, where the patients received partial treatment for chest infections before the correct diagnosis was made. Late-onset CDH presenting with gastrointestinal symptoms often involves a smaller hernia defect. The symptoms are typically due to varying degrees of obstruction caused by incarceration of the herniated organs. Moreover, late-presenting CDH is at higher risk for rare but serious complications, such as tension gastrothorax, gastric perforation, splenic torsion or rupture, intestinal obstruction, or bowel necrosis [4].

Typically, a plain chest X-ray can reveal multi-cystic lesions, which represent bowel loops in the thoracic cavity, as seen in our cases [7]. However, the X-ray findings can sometimes mimic pneumothorax, show an air-fluid level mistaken for pleural effusion, or be confused with diaphragmatic eventration, leading to misdiagnosis. In such cases, advanced imaging techniques like chest CT scans are essential for confirming the diagnosis and preventing further delays in management [5]. In many sub-Saharan countries, the incidence of CDH remains unknown due to the lack of access to basic imaging studies and the frequent late presentation of cases, which often leads to misdiagnosis and poorer outcomes. Consequently, clinicians are often compelled to rely heavily on physical findings and clinical examination to make a diagnosis. This underscores the need for improved diagnostic resources and early detection strategies in these regions [8].

Surgery remains the primary management approach for congenital diaphragmatic hernia (CDH), with options including transabdominal or transthoracic approaches and both open and minimally invasive techniques [9]. The main objectives of surgery are to reduce the abdominal contents into the peritoneal cavity and close the diaphragmatic defect to improve cardiopulmonary function [9]. Historically, surgery was performed as an emergency procedure; however, recent practices have shifted towards semi-elective surgery. This approach allows for the identification and management of any cardiovascular instability and pulmonary hypertension that may develop, thereby reducing the stress associated with the surgical procedure and potentially improving outcomes [10].

The outcome of CDH is significantly influenced by the severity of the diaphragmatic defect and the timing of treatment [11]. Since CDH symptoms can present intermittently, a normal chest X-ray does not necessarily rule out the diag-

nosis. Therefore, it is crucial for clinicians to consider CDH as a differential diagnosis when evaluating patients with respiratory and gastrointestinal symptoms [12]. In a study by Long et al., it was found that the overall survival rate of live-born infants was 75% at 1 year. The authors further highlighted that the major burden of mortality in this population occurs before surgical intervention, with 78% of all deaths happening before surgery. Notably, 93% of stable infants with CDH who underwent surgery survived to 1 year [13].

Currently, pediatric surgeons are increasingly adopting a strategy of delayed correction of the diaphragmatic defect in CDH patients. The timing of surgery is now guided by the optimization of the patient's clinical condition rather than a fixed time frame. However, clinical evidence supporting this approach is limited, and controversy remains over whether delayed repair is associated with improved survival. Some studies have concluded that the timing of CDH repair does not appear to influence 90-day survival rates, regardless of the severity of the disease [14].

Conclusion

Congenital diaphragmatic hernia can be detected antenatally or in the neonatal period. Management can start in utero in selected cases and in technologically advanced centers. Late presentation of CDH usually present with a wide range of chest, or abdominal, symptoms, therefore clinicians need a high degree of suspicion for diagnosis. Early diagnosis and management is vital to prevent complications like lung hypoplasia or gastric volvulus. In sub-Saharan Africa, diagnostic challenges persist due to the limited availability of imaging facilities in primary centers and the scarcity of expertise required to make accurate diagnoses. Timely antenatal visits and proper antenatal scans performed by trained sonographers are crucial for early detection. Early identification of conditions like congenital diaphragmatic hernia (CDH) allows for the planning of delivery and postnatal care at a tertiary-level center, significantly improving outcomes.

Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Author contributions

JL conceptualized and drafted the manuscript. BRK, JL and DM were the lead surgeons. VN and DM reviewed the medical records. SG reviewed and reported the radiology images. All authors have read and approved the final script.

Patient consent

Informed consent was obtained from the guardians of both cases. Accompanying images have been censored to ensure that the patient cannot be identified. A copy of the consent is available on record.

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