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Case Report

Unmasking the silent hernia: the late presentation of congenital diaphragmatic hernia — a case report

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ABSTRACT

Congenital diaphragmatic hernia (CDH) is a rare congenital anomaly characterized by herniation of abdominal contents into the thoracic cavity through a defect in the diaphragm. While commonly diagnosed prenatally or in the neonatal period, late-presenting CDH can occur and may mimic other thoracic emergencies such as tension pneumothorax, complicating diagnosis and management. We report a case of an 11-month-old female infant with a 1-week history of fever, cough, and sudden onset of rapid breathing. Infant was treated at a local hospital with IV antibiotics, then was referred to our hospital due to lack of clinical improvement. After clinical assessment, CDH was suspected and was confirmed with chest X-ray. Later, the infant underwent a successful CDH repair procedure with an uneventful post-operative course, subsequently got discharged in stable condition with no significant long-term complications. This case emphasizes the importance of early recognition and maintaining a high index of suspicion for CDH in infants presenting with recurrent lower respiratory symptoms or pneumonia unresponsive to antibiotic treatment.

Keywords: Congenital diaphragmatic hernia, Pneumonia, Infant

INTRODUCTION

Congenital diaphragmatic hernia (CDH) is an uncommon developmental defect in which abdominal viscera protrude into the thoracic cavity through an abnormal opening in the diaphragm.[1,2] This condition results from abnormal embryologic development of the diaphragm, most commonly due to incomplete fusion of the pleuroperitoneal membranes.[3] Approximately 85% of cases occur on the left side, whereas bilateral defects are rare.[4] Although many cases are detected during prenatal ultrasonography or present shortly after birth with respiratory distress, a small proportion remain asymptomatic until later in infancy or childhood. The timing and severity of symptoms depend on the size of the diaphragmatic defect and the extent of herniation of abdominal contents into the thoracic cavity. Because late-presenting CDH often mimics more common respiratory or gastrointestinal disorders, diagnosis may be delayed or initially overlooked.[5] Early recognition is essential, as timely surgical management is associated with favourable outcomes.[6] We report a case of late-presenting CDH that was initially diagnosed as pneumonia, emphasizing the importance of considering CDH in the differential diagnosis of persistent or atypical respiratory symptoms.

CASE REPORT

A 11-month-old infant hailing from Chitradurga district, born as a 3rd issue to a 2nd degree consanguineous married couple, immunized appropriately as per NIS, developmentally normal, with no significant past/birth/family history, was presented to our paediatric emergency unit with a history of fever and cough for 1 week and with sudden onset of rapid breathing. Infant was treated at a local hospital as a case of lower

respiratory tract infection with IV Antibiotics for about 1 week, then was referred to our hospital due to lack of clinical improvement. On receiving, infant was noted to be afebrile, having tachycardia (162 beats per minute), tachypnoea (64 breaths per minute) with increased work of breathing (nasal flaring with subcostal and intercostal retractions) with oxygen saturation of 84-86% on Room air. No signs of shock present. On auscultation, air entry on the left side of the thorax was noted to be absent, with occasional bowel sounds heard.

The infant was immediately shifted to PICU and was put on oxygen support (Nasal prongs with 2 litres O_2). Relevant blood investigations were sent, and a chest X-ray (Figure 1) was done, suspecting CDH, which confirmed the presence of bowel loops in the left thorax. Higher IV Antibiotics were started, and routine monitoring was done.

A diagnosis of diaphragmatic hernia with mediastinal shift was made, and the on-call paediatric surgeon was consulted immediately. The child was prepared for emergency laparotomy and repair, under general anaesthesia (Figure 2 and 3). Immediately after the surgery, the patient was shifted to PICU for observation. On the 4th post-operative day, the patient was discharged. On follow-up, the patient was stable with no distress or other setbacks noted.

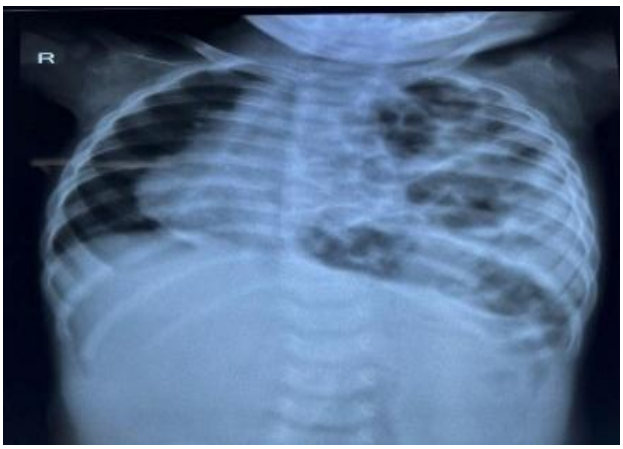


Figure 1: Chest X-ray image showing left-sided air-filled cystic structures pushing the mediastinum to the right side with absent left hemidiaphragm outline.

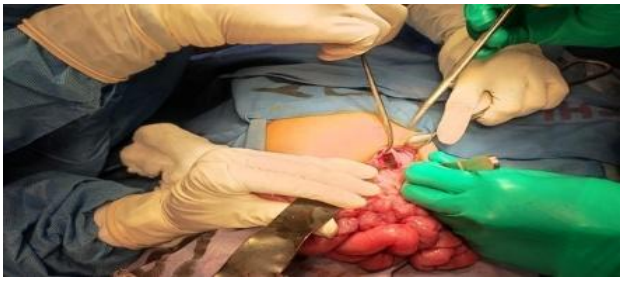


Figure 2: Intraoperative picture showing left-sided posterolateral diaphragmatic defect.

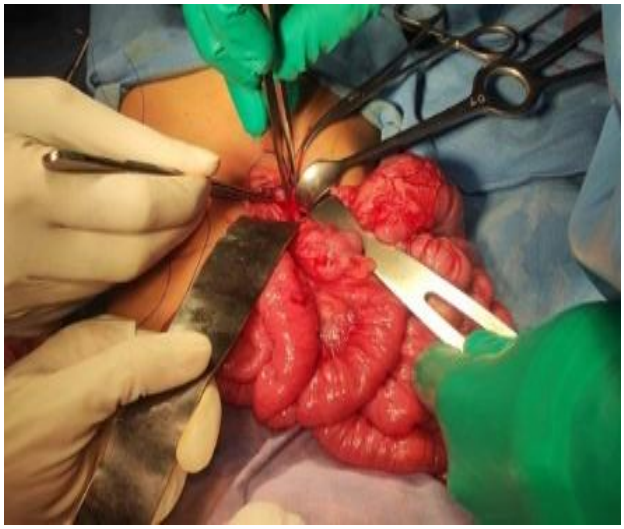


Figure 3: Intraoperative picture showing suturing of the defect after reduction of herniated gastrointestinal organs from chest cavity.

DISCUSSION

CDH is typically diagnosed during the prenatal period or in the immediate neonatal period because of respiratory compromise resulting from pulmonary hypoplasia. However, a minority of patients remain asymptomatic beyond the neonatal period and present later in infancy or childhood. In one single-institution series spanning three decades, only 7 of 37 patients (18.9%) presented late, with ages at diagnosis ranging from 2.5 months to 16 years.[6] Delayed presentation is uncommon and often leads to diagnostic uncertainty, since symptoms are nonspecific and overlap with more frequently encountered respiratory and gastrointestinal disorders, a difficulty borne out by the considerable diversity of presentations described in previously reported cases (Table 1).[1-3,5,7-12]

Our patient presented at 11 months of age with fever, cough, and progressive respiratory distress and was initially managed as a lower respiratory tract infection. Failure to improve despite intravenous antibiotics prompted further evaluation, and clinical examination demonstrating absent air entry over the left hemithorax with audible bowel sounds raised suspicion of a diaphragmatic defect, which was subsequently confirmed by chest radiography. This sequence closely parallels other infants in whom CDH was mistaken for an antibiotic-unresponsive respiratory infection, including a 6-week-old infant with persistent lower respiratory tract infection and a 10-month-old infant with respiratory distress and reduced oral intake, in both of whom, as in our patient, the correct diagnosis was reached only after standard treatment for presumed infection had failed.[5,10] This case therefore reinforces the importance of re-evaluating the diagnosis in children who do not respond to appropriate treatment for presumed pneumonia.

The mechanism underlying late-presenting CDH differs from that of neonatal CDH. In these patients, the diaphragmatic defect is usually small and may be temporarily occluded by adjacent abdominal organs or omentum, delaying herniation into the thoracic cavity.[1,4] A subsequent rise in intra-abdominal pressure from infection, crying, coughing, or physical exertion can then precipitate herniation, producing the sudden onset of respiratory or gastrointestinal symptoms. A comparable pressure-related trigger has been invoked to explain the recurrent, exertion-associated abdominal and chest symptoms of an adolescent with intermittent left upper quadrant pain[3] and the acute tension gastrothorax of a school-aged boy.[11] Consequently, the clinical picture varies widely with the extent of organ herniation and the degree of pulmonary compression, ranging from insidious respiratory symptoms in infancy to acute thoracoabdominal emergencies in older children.

Our review of previously reported cases (Table 1) demonstrates considerable heterogeneity in age at presentation and clinical manifestations, and comparing our findings with these reports helps situate the present case within the broader clinical spectrum of late-presenting CDH. Infants under one year of age, similar to our patient, most often presented with a predominantly respiratory picture that was initially mistaken for pneumonia or bronchiolitis, as in a 6-week-old infant with antibiotic-unresponsive lower respiratory infection, a 7-month-old infant whose vomiting and sudden respiratory distress were ultimately found to result from a strangulated bowel within the hernia sac, a 6-month-old infant with recurrent aspiration pneumonia across three prior admissions before the defect was identified, and 9-month-and 10-month-old infants who presented with acute respiratory distress and with respiratory distress accompanied by reduced oral intake, respectively.[5-10] In contrast, older children and adolescents more often presented with abdominal pain, vomiting, or mixed respiratory-gastrointestinal symptoms, as illustrated by a 9-year-old boy with 1p36 deletion syndrome, a 17-year-old girl with recurrent left upper quadrant pain and postural chest tightness, an 8-year-old boy with tension gastrothorax, and a 13-year-old boy initially misdiagnosed with a pleural effusion.[2,3,11] This pattern suggests that the dominant symptom complex of late-presenting CDH shifts from a respiratory presentation in infancy toward an abdominal one with increasing age, likely reflecting differences in the volume and type of herniated viscera and in the ability of older children to localise and describe pain. Two reports parallel our patient particularly closely in that an alternative diagnosis was actively pursued before CDH was recognised: a 2-year-old child initially treated for a presumed tension pneumothorax, and the 6-month-old infant with recurrent aspiration pneumonia noted above.[1,8] Similar to these and several other published reports, our patient initially received treatment for a respiratory infection before the correct diagnosis was established, a recurring pattern that underscores why late-presenting CDH should remain an important differential diagnosis in children with persistent respiratory symptoms, particularly when clinical improvement is not observed with standard medical therapy.

Chest radiography remains the initial imaging modality of choice and is often sufficient to establish the diagnosis when bowel loops or air-fluid levels are visualised within the hemithorax, particularly in the presence of mediastinal shift. Nevertheless, radiographic findings may occasionally resemble pneumonia, pleural effusion, pneumothorax, or congenital pulmonary airway malformation, increasing the risk of inappropriate intervention; this pitfall is well illustrated in the literature, where late-presenting CDH has been misread as a tension pneumothorax, a simple pleural effusion, and a tension gastrothorax, in each instance risking needle decompression or chest tube insertion before the correct diagnosis was made.[1,11,12] Careful interpretation of imaging in conjunction with physical examination findings, including diminished breath sounds and bowel sounds within the chest, as proved decisive in our patient, can facilitate timely diagnosis and prevent such unnecessary procedures.

Definitive management of late-presenting CDH consists of surgical reduction of the herniated abdominal contents and repair of the diaphragmatic defect. Unlike neonates with severe pulmonary hypoplasia, who often require prolonged preoperative stabilisation, children with delayed presentation generally have better-developed lungs and favourable postoperative outcomes following timely surgical correction; in the single-institution series discussed above, all seven late-presenting patients underwent successful primary repair with no postoperative mortality and no recurrence over a median follow-up of 3.8 years.[6] Our patient underwent emergency laparotomy with successful repair and had an uneventful postoperative recovery, consistent with the benign courses reported in comparable infant cases once the diagnosis was established.[5,8-10] By contrast, delay in recognition has occasionally led to bowel strangulation requiring resection, as in the 7-month-old infant described above, who needed enterectomy and a staged jejunostomy before eventually recovering well; this contrast suggests that the favourable outcome in our patient reflects, at least in part, the comparatively short interval between symptom onset and surgical correction.[7]

Although individually uncommon, each reported case of late-presenting CDH adds to a growing body of evidence that broadens the recognised clinical spectrum and diagnostic pitfalls of this condition across the paediatric age range.[1-3,5,7-12] Considered together with these reports, our case reinforces the need for clinicians to maintain a high index of suspicion when evaluating infants with unexplained or persistent respiratory distress, especially when physical examination or radiographic findings are atypical, or the response to standard antimicrobial therapy is poor.

Table 1: Summarizes a few similar reported cases of late presentation of CDH in the literature, showcasing diverse patient demographics and clinical presentations.

Reported cases	Patients' relevant demographics	Clinical features
Ashagre, Wintana[1]	2-year-old boy with no significant past history.	Sudden choking episode followed by vomiting initially and then had progressive hurried breathing.
Zihra, Rehmaan[2]	9-year-old boy with 1p36 deletion syndrome and ADHD	Abdominal pain, multiple episodes of non-bilious vomiting, decreased oral intake, and sudden onset of shortness of breath.
Vivienne Vinton, Molly Posa[3]	17-year-old female with recurrent episodes of acute left upper quadrant pain.	Severe abdominal pain (LUQ) with chest tightness when lying flat, nausea, and one episode of non-bloody, non-bilious vomiting.
Mwamanenge, Mussa[5]	6-week-old infant with no marked past history.	Lower respiratory infection features not responding to antibiotics.
Flausino, Maes Manara[7]	7-month-old infant with no obvious past history.	Vomiting and sudden onset of respiratory distress.
Ghabisha, Faisal[8]	6-month-old infant with a history of recurrent aspiration while feeding and repeated hospital admissions three times in the past for suspected aspiration pneumonia.	Low-grade fever, cough, poor feeding, and hurried breathing.
A Sridhar, S Nichani[9]	9-month-old infant with no significant past history.	Acute onset of hurried breathing, grunting, and excessive screaming for a 1–2-hour duration.
Mohammad G Ibdah[10]	10-month-old infant with no notable past history.	Sudden onset of respiratory distress and vomiting with decreased oral intake and activity.
Aabha A Anekar[11]	8-year-old boy with no obvious medical history.	Sudden onset of respiratory distress with chest pain and 3 episodes of non-bilious vomiting.
Hyun Beak Shin, Yeon-Jun Jeong[12]	13-year-old boy with no considerable past history.	Abdominal pain (LUQ) and vomiting.

CONCLUSION

Late-presenting CDH is a rare entity and often presents with nonspecific respiratory or gastrointestinal symptoms in infancy and childhood.

Because its presentation often mimics common conditions such as pneumonia, diagnosis may be delayed unless clinicians maintain a high degree of clinical suspicion. Careful physical examination together with appropriate interpretation of chest radiographs is essential for early recognition. Prompt surgical repair following diagnosis is associated with excellent outcomes and minimizes the risk of complications related to delayed treatment.

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