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## Elusive clinico radiological appearance of late presenting congenital diaphragmatic hernia—a case report

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## Case reports

# Elusive clinico radiological appearance of late presenting congenital diaphragmatic hernia—a case report

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## Abstract

Late presenting congenital diaphragmatic hernia (CDH) is a rare subset of diaphragmatic hernias presenting beyond 30 days of life, which may be challenging to diagnose. A 3-year-old girl was referred to our hospital with an acute history of respiratory and gastrointestinal symptoms, with a suspected diagnosis of left-sided pneumothorax. Initial radiological evidence pointed towards left hydropneumothorax. A chest X-ray and ultrasound of the chest seemed to confirm the diagnosis, hence intercostal drainage (ICD) tube was inserted. Post ICD insertion, mediastinal shift to the right was persistent and CT scan of the chest showed herniation of stomach and bowel loops into left hemithorax through a defect in the diaphragm, suggestive of late presenting CDH which was later corrected by a surgical repair. A high index of suspicion for late presenting CDH as a differential diagnosis for children presenting with varied respiratory and gastrointestinal symptoms would help in early surgical management and prevention of complications.

**Key words:** Congenital diaphragmatic hernia, hydropneumothorax, late presenting congenital diaphragmatic hernia

## Introduction

A congenital diaphragmatic hernia is an abnormality in the structure or continuity of the diaphragm with an estimated incidence of 1/2000 to 1/5000 live births.<sup>1</sup> The diagnosis is usually made antenatally or soon after birth, as most cases of CDH present in the neonatal period<sup>2</sup> and associated with mortality rates of almost 29-50%.<sup>3</sup>

However, literature is replete with cases of CDH<sup>4</sup> presenting in older children and adults. Late presenting CDH (beyond one month of age)<sup>2, 5</sup> is

milder and may even be asymptomatic. It accounts for 5-25% of the cases<sup>6</sup> and generally tends to have a better prognosis with prompt diagnosis.<sup>3</sup>

Misleading clinical features is the commonest reason why late presenting CDH is generally misdiagnosed even in a tertiary care setting with the best of radiological equipment and specialist facilities.<sup>4</sup> Here, we present a rare case report of a 3-year-old girl child with late presenting left-sided CDH with both respiratory and gastrointestinal symptoms and no history of trauma, who was initially diagnosed as hydropneumothorax, before surgically correcting the defect.

**Case** A 3-year-old female child was referred to the emergency department of our hospital as a suspected case of left-sided pneumothorax. She presented with a history of fever, cough, coryza for two days, and pain in the abdomen with multiple episodes of vomiting of one-day duration. The child was treated with home-based remedies and when the symptoms did not subside, was taken to a local paediatrician, who observed decreased breath sounds

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on the left side and considering the possibility of pneumothorax, the child was immediately referred. History of several episodes of similar semiology on and off for one year, treated symptomatically on an outpatient basis at various local health facilities, without subjecting the child to any investigations, after which child was normal. No significant history of trauma was present.

The child was born by normal vaginal delivery and had an uneventful birth history with no requirement of resuscitation or respiratory support at birth. Postnatally, the child had attained appropriate growth and developmental milestones as per age.

On examination, the child appeared sick, with a temperature of 100 degrees Fahrenheit, heart rate of 150/min, and respiratory rate of 52/min but maintaining saturation at room air. She had no signs of dehydration. Her anthropometry was within normal limits for age (weight—13 kg, height—94 cm). She had bilateral subcostal and intercostal retractions and air entry was absent on the left side anteriorly. Mild diffuse tenderness was present on abdominal examination without any organomegaly. Blood investigations done were fairly within normal limits apart from slightly elevated total counts with a neutrophilic preponderance (Haemoglobin—11.5 g/dl, total leucocyte count—14000 cells/cumm, neutrophil—60%). Chest X-ray (Fig 1a) appeared like a left hydropneumothorax with the collapse of the left lung, as there was an air-fluid level and shift of the mediastinum to the right side. Ultrasound of the chest revealed moderate to gross left hydropneumothorax with multiple internal echoes

within the fluid component with no evidence of loculation or septation. Ultrasound of the abdomen showed no sonological abnormality.

The child was referred to paediatric surgery for ICD insertion. Before ICD insertion, syringing revealed a small quantity of air followed by haemorrhagic fluid raising the suspicion of malignancy or tuberculosis for which the fluid was sent for laboratory analysis. However, it came negative. Following ICD placement, the air column was moving adequately which further reiterated our diagnosis. Post ICD, X-ray taken revealed that air-fluid levels had resolved and yet the mediastinal shift to the right was persistent; the left lung did not seem to re-expand as seen in Fig 1 b and 1 c. CT scan of the chest showed left diaphragmatic defect with herniation of stomach and bowel loops into left hemithorax with passive left lung collapse as seen in Fig 2. Nasogastric (NG) tube was inserted the next morning. Thereafter immediate laparotomy was scheduled to repair the diaphragmatic defect.

Intraoperatively, a posterolateral defect measuring 5x1cm was noted with the hernia sac containing the stomach, omentum, spleen, splenic flexure, and descending colon almost heading on to strangulation (Fig 3). The contents were reduced and placed back in the abdomen followed by primary closure of the defect. Chest X-rays post-surgery (Fig 4) showed the continuity of diaphragm restored, bowel loops back in anatomical position and the child had normal chest expansion clinically. The child had an uneventful post-operative recovery and was discharged on day ten (Fig 5).

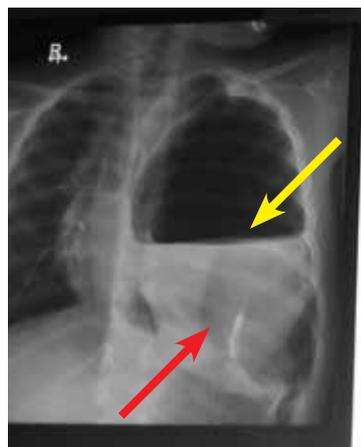


Fig 1a: On presentation

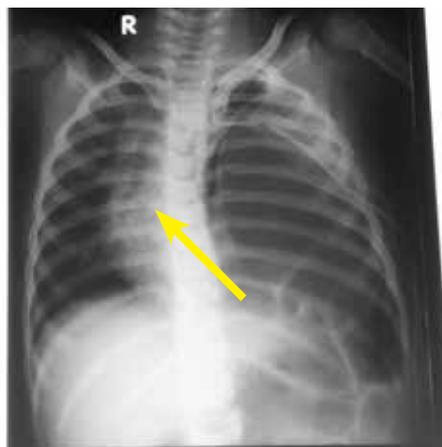


Fig 1b: Post ICD insertion (immediate)



Fig 1c: 12 hrs post ICD insertion

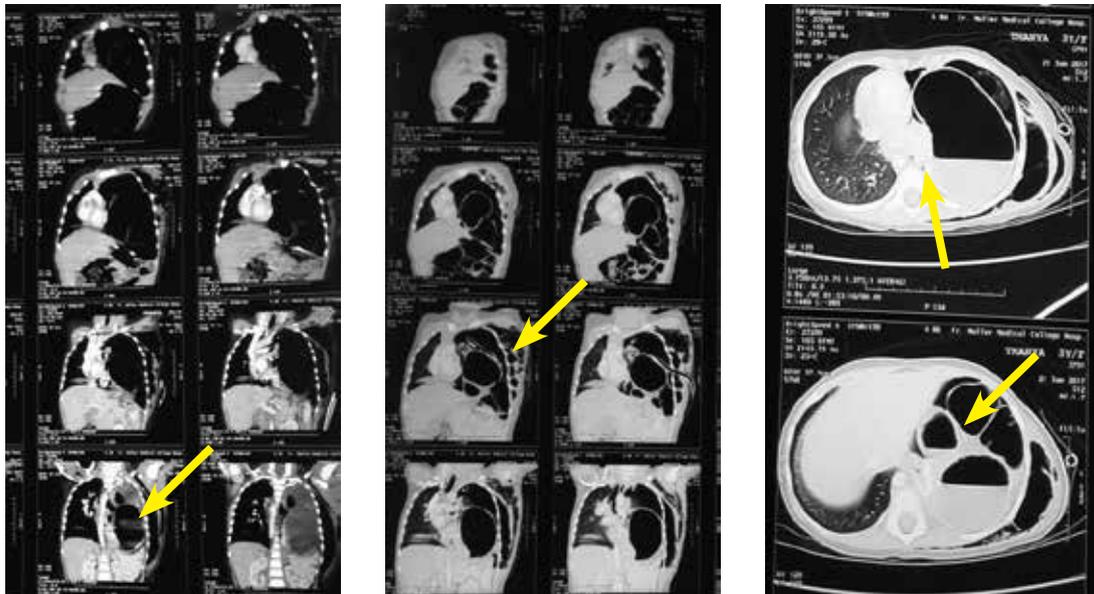


Fig 2: CT scan of the chest – multiple sections



Fig 3: Intraoperative picture showing defect and herniation of bowel



Fig 5: Well approximated left lower subcostal scar post staple removal (post-op day 10)



Fig 4: Postoperative chest X-ray after surgical repair of the diaphragmatic defect

### Discussion

Diaphragmatic hernias presenting in children can either be congenital or acquired (secondary to blunt or penetrating trauma to the abdomen).<sup>7</sup> No history of trauma and absence of intraoperative findings suggestive of trauma supports a congenital etiology in our child. Similar reports have been published by Mei-Zahav, et al.<sup>6</sup> and are relevant, especially when the cause of herniation is unclear and atypical in presentation.

Misdiagnosing late presenting CDH as hydropneumothorax is not uncommon in an acute

setting as the latter is a medical emergency and tends to get over-diagnosed<sup>8,9</sup> which happened in our case too. Going back to the chest X-ray, we can see intrathoracic bowel loops, which require keen observation. A simple cough and cold would end up as a missed congenital anomaly was never expected even by the parents.

ICD tube insertion itself may contribute to iatrogenic mortality<sup>3</sup> in CDH cases. Ideal management should have been the insertion of NG tube followed by a chest X-ray, which would not only confirm the diagnosis but also serve as an initial treatment of choice. Bag and mask or blow-by oxygen should be avoided although respiratory support is essential in initial stabilization of a patient with respiratory distress.<sup>1</sup>

Surgical management is necessary for correcting the defect as well as preventing complications.<sup>2</sup> Surgical repair is easier in late presenting CDH than neonatal CDH cases as it rarely requires a patch or flap.<sup>6</sup> Prognosis is generally favourable mainly due to good lung development.<sup>1</sup>

### Conclusion

A high index of suspicion and the possibility of late presenting CDH as a differential diagnosis for infants and children presenting with varied respiratory and gastrointestinal symptoms would help in early surgical management and prevention of complications. Also, it is always ideal to insert a naso or orogastric tube before taking an X-ray to aid prompt diagnosis.

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