Avoiding misdiagnosis in postnatal presentation of congenital diaphragmatic hernia: A report of two cases and review of radiologic features

Sachin Kumbhar, MD, Sabina Siddiqui, MD, Pooja Thakrar, MD

Department of Pediatric Radiology, Children's Hospital of Wisconsin, 9000 W. Wisconsin Ave., MS-721 Milwaukee, WI 53226 USA
Department of Pediatric Surgery, Children's Hospital of Wisconsin, 9000 W. Wisconsin Ave., C-320, Milwaukee, WI 53226 USA

ABSTRACT

Diagnosis of a congenital diaphragmatic hernia after the first day of life can be challenging. The clinical symptoms are often nonspecific, and initial radiographic findings in these patients may mimic many other acute chest conditions, including pneumonia, pleural effusion, and pneumothorax. In turn, diagnostic uncertainty may expose the patient to unnecessary and even potentially harmful interventions such as thoracostomy tube placement. As such, it is imperative that radiologists remain aware of this uncommon entity and recognize imaging findings which may provide clues to its diagnosis.

Keywords:
Congenital diaphragmatic hernia
Late presentation
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Introduction

Between 75% and 95% of congenital diaphragmatic hernias (CDH) are detected on prenatal sonography or declare themselves within the first month of life [1–4]. The remaining patients present later in life with a wide variety of respiratory and/or gastrointestinal symptoms. As there are no symptoms specific to CDH, accurate diagnosis in children with delayed presentations can be challenging. Misdiagnosis as pneumothorax, pneumonia, pleural effusion, or congenital pulmonary airway malformation is not infrequent and can lead to unnecessary or potentially harmful interventions. For example, thoracostomy catheter placement in patients with CDH who have been incorrectly diagnosed with a pneumothorax can increase morbidity through complications such as bowel perforation and mediastinitis.

We present 2 cases of late-presenting CDH and review its radiologic spectrum. We aim to make readers aware of this entity, to discuss how its radiologic findings can mimic other diagnoses, and to offer suggestions to facilitate early diagnosis.

Case 1

An 8-month-old 8.1-kg, full-term female infant presented to our emergency department with vomiting. She had been...
well until 1 day prior to presentation, when she had begun vomiting after every feed. Some of the episodes of vomiting had included bilious contents. The patient had been delivered uneventfully at term without any perinatal complications. On examination, she was alert and active. Her vital signs were within normal limits with a respiratory rate of 44, and she was breathing comfortably on room air. Her lungs were clear to auscultation. Because of the bilious nature of her emesis, the decision was made to perform an upper gastrointestinal series to evaluate for malrotation/midgut volvulus.

A preliminary image of the chest revealed opacification of the left lower hemithorax with mediastinal shift to the right (Fig. 1). Contrast administration demonstrated a distended, abnormally oriented stomach which was inferiorly and mediately displaced. The proximal small bowel extended into the left upper quadrant of the abdomen and demonstrated a focal area of narrowing. Minimal contrast passed further distally into nondilated loops of bowel in the left hemithorax (Fig. 2). A diagnosis of left congenital diaphragmatic hernia was made.

The patient was taken to surgery for repair 2 days later. The small bowel and right colon were reduced from the left chest into the abdomen. A 2.5 x 1 cm hernia defect in the posterolateral left hemidiaphragm was closed primarily. Inspection of the bowel revealed anticipated malrotation but with dense Ladd’s bands. The Ladd’s bands were lysed, and the mesentery was widened. The patient tolerated the procedure well and was discharged to home a week later. She was doing well at her 1-month follow-up visit.

**Case 2**

A 2-month-old 5.6-kg, full-term male infant presented to an outside emergency department with respiratory distress, altered mental status, and bloody stools. He had been doing well until approximately 3 hours prior to presentation, at which time he had become pale and lethargic and had started grunting. He was noted to have been fussy earlier in the day and had had an episode of nonbilious emesis after his noon feed. There was no history of fever, rhinorrhea, or cough. His mother had obtained routine prenatal care and had had an uneventful pregnancy and delivery. He had been in good health at his 1-month well-child visit.

On examination, the infant appeared lethargic and had a weak cry. He was tachycardic and made grunting sounds while breathing. No rales or rhonchi were audible on auscultation. The abdomen was distended with generalized tenderness. The mucous membranes were dry, and capillary refill was delayed. Intravenous fluids were started, and the patient was referred to our institution for further care.
Frontal and left lateral decubitus radiographs of the chest and abdomen were performed. There was marked distention of the stomach with a paucity of bowel gas in the abdomen. The left hemithorax was nearly completely opacified (Fig. 3). A differential diagnosis of left pleural effusion, left chest mass, and left-sided diaphragmatic hernia was given. A CT scan of the chest, abdomen, and pelvis was subsequently performed. The CT demonstrated herniation of bowel and the left kidney into the left chest. There was a moderate left pleural effusion with partial left lung collapse and rightward mediastinal shift (Fig. 4). A fluoroscopic upper gastrointestinal series performed the following morning confirmed a left-sided diaphragmatic hernia with herniation of small and large bowel loops into the left chest (Fig. 5).

The patient was taken for repair of the hernia the next day. The small bowel, part of the stomach, and the transverse and descending colon were reduced from the chest into the abdomen. An approximately 5 x 3.5 cm defect in the posterolateral left hemidiaphragm was closed primarily. Dense Ladd’s bands were lysed, and the mesentery was widened. The patient did well postoperatively and was discharged to home 4 days later. He was doing well at his 4-month well-child visit.

Discussion

Late-presenting CDH is more commonly left-sided [1]. Approximately 80% of these children present acutely. Presentation may be with respiratory and/or gastrointestinal symptoms. Chronic presentation is more commonly associated with right-sided CDH [5]. A small percentage of children with CDH are asymptomatic, and the abnormality is incidentally detected on chest radiograph [3,6]. As presenting symptoms are nonspecific, imaging plays an important role in the diagnosis of CDH. Imaging findings may be straightforward, but it is not unusual for the radiologic appearance of CDH to mimic other diseases [1,7]. A vigilant radiologist may be able to raise concern for CDH when imaging findings are atypical for other diseases or are discordant with the presenting symptoms.

A chest radiograph is often the first imaging study performed in children with CDH, particularly those with respiratory symptoms. Abdominal radiographs are more often obtained in patients with gastrointestinal symptoms at presentation [3]. The diagnosis of CDH is clear when gas-filled loops of bowel are seen in the chest, with or without associated mediastinal shift. However, normal chest radiographs early in life have been reported in patients later found to have CDH [8], and in many patients, the classic imaging appearance of CDH is simply lacking. A paucity of gas within herniated bowel loops or a hernia containing predominantly solid organs can lead to a more solid or consolidative appearance and misdiagnosis as pneumonia, pleural effusion, or chest mass [2].

On the opposite end of the spectrum, too much gas in the herniated portion of the bowel may also lead to misdiagnosis. Excessive gas within a hollow viscus in the chest may be mistaken for a pneumothorax or congenital lobar emphysema. Some authors have reported errant thoracotomy catheter placement in patients thought to have pneumothoraces [2,9]. Berman et al suggest that performing a chest radiograph after inserting a nasogastric tube is helpful to arrive at an ac-
In an abnormal location [11]. Both of our cases had marked gaseous distention of the stomach with a paucity of intra-abdominal bowel gas. An opacified lower hemithorax, if detected on these abdominal radiographs, may be a clue to the underlying diagnosis of CDH.

A literature review conducted by Baglaj et al suggests than an upper gastrointestinal series is the best modality for diagnosis of CDH presenting postnatally. A CDH may contain only stomach, only small bowel, or only large bowel. As such, if the index of suspicion is high, a complete small bowel follow-through and/or contrast enema should be performed if the stomach is found to be within the abdomen on the upper gastrointestinal study [3]. In a very small percentage of children with CDH, the hernia may contain only solid organs [1,3]. The upper gastrointestinal series and contrast enema would likely appear normal in such cases. Ultrasound and computed tomography (CT) are other useful diagnostic modalities which can aid in the diagnosis of CDH by demonstrating bowel and/or solid abdominal organs in the chest [12].
Conclusions

Late-presenting CDH can manifest as a variety of respiratory or gastrointestinal symptoms, none of which is specific. Chest and/or abdominal radiographs are often the first imaging studies to be performed in these patients but may not demonstrate the classic appearance of gas-containing viscerae in the chest. Radiologic features that could indicate the possibility of CDH include the presence of lower hemithoracic opacification in a child with gastrointestinal symptoms or an atypical appearance of the gastric bubble in a child with respiratory symptoms. In the presence of such atypical radiographic findings, a fluoroscopic exam or cross-sectional imaging study should be performed to clinch the diagnosis. Performing radiographs after placement of a nasogastric tube can also be helpful to avoid misdiagnosing a tension gastrothorax as a pneumothorax.

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