Late Presentation of Congenital Diaphragmatic Hernia: A Diagnostic Dilemma-A Case Report

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Abstract:
Congenital diaphragmatic hernias are commonly symptomatic within 24 hours after birth, but late presentation is not uncommon. Late presentation of congenital diaphragmatic hernia poses diagnostic difficulties as clinical picture are vague, and more commonly presented with non-specific gastrointestinal and respiratory symptoms. Due to the vague and non-specific clinical presentation, clinician faces a diagnostic dilemma resulting in delay in diagnosis and many a times an inappropriate management. This article reports 2 cases of late-presenting congenital diaphragmatic hernia (over the period of 6 months from September 2014 to February 2015) in National Institute of Disease of Chest and Hospital (NIDCH). In first case, she was diagnosed as right-sided tubercular pleural effusion and was treated with CAT-1 anti-tubercular therapy for 6 months without any clinical improvement. Later CT scan of chest was done and diagnosed as a case of congenital diaphragmatic hernia. The second case was diagnosed as a left-sided hydropneumothorax and treated with left tube thoracostomy. During removal of the intercostal chest tube, some fatty tissue was pulled out of the thoracostomy site. In NIDCH, she was diagnosed as a case of diaphragmatic hernia by barium follow-through. Both cases were diagnosed as Bochdalek hernia during the repair of the hernia defect via thoracotomy.

Keywords: Congenital diaphragmatic hernia, Bochdalek hernia, late presentation

Introduction:
Congenital diaphragmatic hernia is a developmental defect of the diaphragm that allows abdominal viscera to herniate into the chest. Bochdalek hernia (herniation of abdominal content through diaphragm in the posterolateral aspect) and Morgagni hernia (herniation of abdominal content through diaphragm in the anterior aspect) are the two types of congenital diaphragmatic hernia. Of them, Bochdalek’s hernia is the common one. Apart from that, diaphragmatic hernia can be acquired types. Acquired diaphragmatic hernias are common than the congenital hernias. Hiatus hernia is the common type of acquired diaphragmatic hernia.

Congenital diaphragmatic hernia occurs in 1 per 3000 live births.¹ Most of the patients with congenital diaphragmatic hernia are symptomatic within the first 24hrs of life, and present with respiratory distress. Child presenting on the early life has more respiratory symptoms, and earlier the onset of symptoms and signs, the more severe is the pulmonary disease. However the late presentation is not uncommon and can represent 5-25% of patients with CDH.² Late presentation is more with gastrointestinal and respiratory symptoms but are not specific. Patients presenting with gastrointestinal symptoms have been shown to be significantly older than those presenting with respiratory symptoms.³

As the presentation in older children is vague, inconsistent and chronic, clinician faces a diagnostic difficulties in these group of patients resulting in diagnostic delay, inappropriate treatment and potential fatal outcome.⁴ Patient may also present with complications like obstruc-
tion, strangulation or perforation of intrathoracic bowel or injury to any organs present inside the thoracic cavity due to trauma or during tube thoracostomy/thoracocentesis. Also these subsets of patients have milder form of pulmonary disease and some may go unnoticed for life. We herein report 2 patients of CDH with late presentation, causing diagnostic dilemma and delay, inappropriate management but finally been managed appropriately with favorable outcomes. The main objective of this case report is to highlight the diversities of the clinical presentation of late-presenting congenital diaphragmatic hernia, so that it may be beneficial for the physician for correctly diagnosing them in the future and these patients can be managed appropriately and timely.

Case 1:

A 10-year old girl hailing from Kisoregonj, presented with complaints of shortness of breath and non-productive cough for 2 months. Shortness of breath was mild to moderate, exertional and gradual on onset. She had no orthopnea or paroxysmal nocturnal dyspnea. She had respiratory problem since 10 months of her age and had been treated several times as a case of bronchopneumonia but her problem did not subsided completely. She was also given anti-tubercular chemotherapy 1 year back after diagnosing as case of right-sided pleural effusion (Fig-1) without clinical and radiological improvement. She had constipation and intermittent vomiting since her childhood. Her antenatal and perinatal history was uneventful. On examination, she was normal looking with average built and normal nutritional status. Her other general examination revealed normal findings. Clinically, chest examination was suggestive of right-sided pleural effusion. Bowel sound was heard on right lower chest.

ECG, echocardiography and other routine investigations were normal. Chest X-ray was suggestive of right-sided pleural effusion (Fig-1). CT scan of chest showed inhomogenous isodense lesion mostly occupying the mid and the lower right hemithorax with multiple air pocketing inside the lesion (Fig-2). Barium follow through showed almost whole of the jejunum and ileum and part of the large intestine occupying the right lower chest and upper abdomen with hugely dilated stomach (Fig-3). Patient underwent primary repair of the diaphragmatic hernia defect in two layers by right posterolateral thoracotomy incision. Jejunum, ileum and part of ascending colon, and also liver were found occupying the right hemithorax with no hernial sac. The lower lobe of right lung appeared hypoplastic. There was defect about 6X4cm in size located posterolateral part of the right diaphragm. Post-operative period was uneventful and patient was discharged on 14th POD. At the time of her discharge, she was hemodynamically stable and her wound was healthy.

Case 2:

A 21 years old lady hailing from Bogra, presented with complaints of shortness of breath and left-sided chest pain for 1 and ½ months. Chest pain was intermittent, gradual on onset, mild, non-radiating, and dull aching in nature. It had no aggravating factors and relieved after taking analgesics. Shortness of breath was mild to moderate, exertional and gradual on onset. She had no history of trauma to the chest. She had history of non-radiating intermittent colicky periumbilical pain, but no history of nausea, vomiting or change in bladder or bowel habits. She had history of failed appendectomy 8 years back, during which surgeon could not locate her appendix via standard McBurney’s incision. With these complaints, she visited a tertiary hospital where she was diagnosed as a case of left-sided hydro pneumothorax (Fig-4) and tube thoracostomy was done, without any improvement. During removal, some fatty tissue also came out with the tube, which was again reduced inside the chest and she was referred to NIDCH.

On examination, she was normal looking with average built and normal nutritional status. All other general examination parameters were normal. Clinically, chest examination was suggestive of left-sided hydro pneumothorax. Bowel sound was heard on interscapular region of left chest.

All routine investigations were within normal limits. Chest X-ray was suggestive of left-sided hydro pneumothorax (Fig-3). CT scan of chest showed inhomogenous isodense lesion mostly occupying the mid and the lower
left hemithorax with multiple air pocketing inside the lesion (Fig-5). Barium follow through showed almost whole of the jejunum and ileum and part of the large intestine occupying the left lower chest and upper abdomen with hugely dilated stomach (Fig-VI). Patient was treated surgically as previous case and postoperative period was uneventful. Patient discharged on 12th POD. At the time of discharge, she was taking normal diet with no abdominal complaints and wound was healthy.

**Fig-1:** CXR showing dense homogenous opacity in lower and part of mid zone of right hemithorax, diagnosed as a case of right-sided pleural effusion.

**Fig-2:** CT scan of the chest showing showed inhomogenous isodense lesion mostly occupying the mid and the lower right hemithorax with multiple air pocketing inside the lesion.

**Fig-3:** Barium meal follow through contrast x-ray showing almost whole of the jejunum and ileum and part of the large intestine occupying the right lower chest and upper abdomen with hugely dilated stomach.

**Fig-4:** X-Ray of chest showing dense homogenous opacity on left lower zone with hyperlucent area devoid of bronchovascular marking with collapsed margin, diagnosed as case of left-sided hydropneumothorax.
Discussion:

First description of a congenital diaphragmatic hernia (CDH) is attributed to Lazare Rivere, at the beginning of the 17th century from autopsy of an adult. 5 First report of a CDH in newborn was made by George Macualay, in 1754, although it was also an autopsy finding. 5 In 1848, Vincent Alexander Bochdalek, reported 2 cases, and he described the location of the diaphragmatic defect as being in the posterolateral aspect of the diaphragm, hence the origin of the terms Bochdalek hernia and Bochdalek foramen. 5 The incidence of CDH hernia is 1 in 3000 live births 1 and more on left side as the right diaphragm develops slightly earlier than the left, and also liver protects the herniation on right side. However, in our cases, one patient had left sided CDH and another on right side. In review study of Baglaj 6, CDH has male predominance of 2:1, but our both patients were female.

Patients can present with a wide range of non-specific gastrointestinal or respiratory symptoms. Patients presenting with gastrointestinal symptoms have been shown to be significantly older than those presenting with respiratory symptoms. 3 Baglaj, in his review of 125 articles 6 including 362 children, reported that 43% present with respiratory symptoms, 33% with gastrointestinal symptoms, 13% with a mixture of the 2 groups of symptoms and 11% being asymptomatic. In our cases, both patients had mostly respiratory symptoms. In second case, nonspecific gastrointestinal symptoms were present and she also had history of failed appendectomy. These findings are consistent with the Baglaj 6, with more common respiratory symptoms in late presentation.

No single symptom is pathognomonic to the late-presenting CDH, posing a diagnostic difficulty. There are number of case reports like ours, in the literature, where late-presenting CDH have undergone unnecessary emergency tube thoracostomy. 7, 8 Both these cases were misdiagnosed and hence mismanaged. Commonly
employed investigations for diagnosis of CDH includes chest X-ray (with or without nasogastric tube insertion), contrast radiological studies, computer tomography (CT) scan, thoracoscopy and laparoscopy. In our first case, diagnosis was confirmed after the CT scan of the chest and in second case diagnosis confirmed after barium meal follow through contrast study.

There is a lower incidence of associated anomalies in late-presenting CDH as compared with neonatal CDH, including cardiac and great vessel anomalies, pulmonary anomalies (lung sequestration, accessory lobe, lung cyst), funnel chest, chromosomal anomalies, urogenital anomalies (hypospadias, horseshoe kidney) and meningomyelocele. In our cases, there were no any such kind of congenital anomalies found. So the overall prognosis is good in late-presenting CDH, and once the repair is done, they could have normal life.

Finally, the diagnosis of CDH in later ages is uncomman. Our first case was reportable for being the right-sided CDH. Our second patient was the oldest case of CDH during the review of the literature and making it a reportable case. Hence, even though late-presentation is uncommon, CDH should be considered in the differential diagnosis of any patient with usual respiratory and gastrointestinal symptoms, abnormal chest radiographic findings, and patient not improving with the usual treatment as seen in these patients. The incidence of CDH in Bangladeshi population is still unknown and also the adult patient with CDH, making a field for further study in our Perspective.

References:
