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CASE REPORT

A Rare Case of Late Presentation of Congenital Diaphragmatic Hernia

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ABSTRACT

Congenital diaphragmatic hernia is a well-described condition that occurs in about 1 in 5,000 live births. A majority of the patients are diagnosed either antenatal or will present in the first few hours of life with respiratory distress. Presentation in adults is extremely rare and accounts for about 5 to 25% of diaphragmatic hernias. Patients, who present with late diaphragmatic hernias, complain of a wide variety of symptoms and diagnosis can be difficult. It consists of herniation of bowel, and occasionally solid organs, into the chest. It is more common on the left side (seen in 80% of cases), as the liver provides a relative barrier on the right side. The major clinical problem is pulmonary hypoplasia, a result of the lung having failed to develop *in utero* as the thoracic cavity is filled with abdominal contents. These congenital diaphragmatic defects have also been described in the adult population, and the widespread use of computed tomography has led to the recognition that these hernias are not uncommon and are often asymptomatic.

Keywords: Abdominal hernia, Computed tomography, Congenital diaphragmatic hernia, Late presentation.

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INTRODUCTION

Herniation of the abdominal viscera into the thoracic cavity can occur through one of the two routes: (1) Through the diaphragmatic hiatus (sliding and paraesophageal hernia) or (2) through a congenital defect in the diaphragm muscle (Bochdalek and Morgagni hernia). Hiatal hernia develops from weakness or tears in the phrenicoesophageal ligament. The diaphragm is formed between the 8th and 10th weeks of fetal life, and the pleuraperitoneal canals close, dividing the coelomic cavity into thoracic and abdominal cavities. During the same period, the gastrointestinal tract elongates into the umbilical pouch and rotates on its return to the abdominal cavity. The left pleuraperitoneal canal closes slightly later than the right. The interrelationship of these two important developmental events results in the clinical spectrum of diaphragmatic hernias: (1) Bochdalek hernia (posterolateral defect), (2) Morgagni hernia (retrosternal defect), and (3) intrapericardial hernia (defect in central tendon).

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CASE REPORT

A 70-year-old female patient presented with pain and distension of the abdomen and constipation for 7 days. The abdominal pain was sudden in onset and of moderate to severe degree. The pain was of colicky type, continuous, nonradiating and did not relieve on taking medication. No significant aggravating factors were found. Abdominal distension has gradually progressed over 7 days. No history of fever, vomiting, and /trauma to the abdomen was observed. On general examination, the pulse was 90 bpm and blood pressure was 130/90 mm Hg. Systemic examination revealed abdominal distension, tenderness in umbilical, hypogastric, and right and left iliac fossae. On auscultation, increased bowel sounds were heard. Respiratory system, cardiovascular system, and central nervous system were within normal limits. Complete blood count, renal function test, and serum electrolytes were normal.

Plain chest X-ray (CXR) revealed the trachea and mediastinum shifted to the right side (Fig. 1). Left pleural effusion was noted. Herniation of bowel loops in left hemithorax was noted. Computed tomography (CT) revealed a defect of 2.7 cm in the left hemidiaphragm through which there was herniation of splenic flexure of the colon (Figs 2A to C). Distal ilium, ascending colon, and transverse colon were dilated. Pleural effusion and hypoplastic lung were noted on the left side. Oral contrast was given to rule out herniation of stomach.

DISCUSSION

Congenital diaphragmatic hernia (CDH) was first described in the 17th century and was associated with a high mortality rate.[1] Its estimated incidence has been

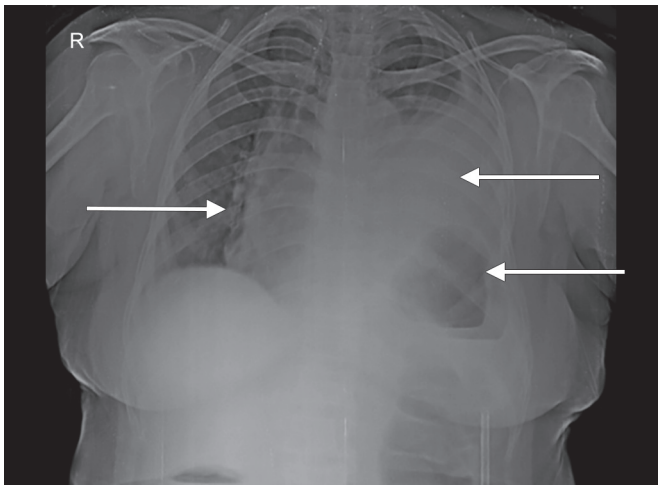


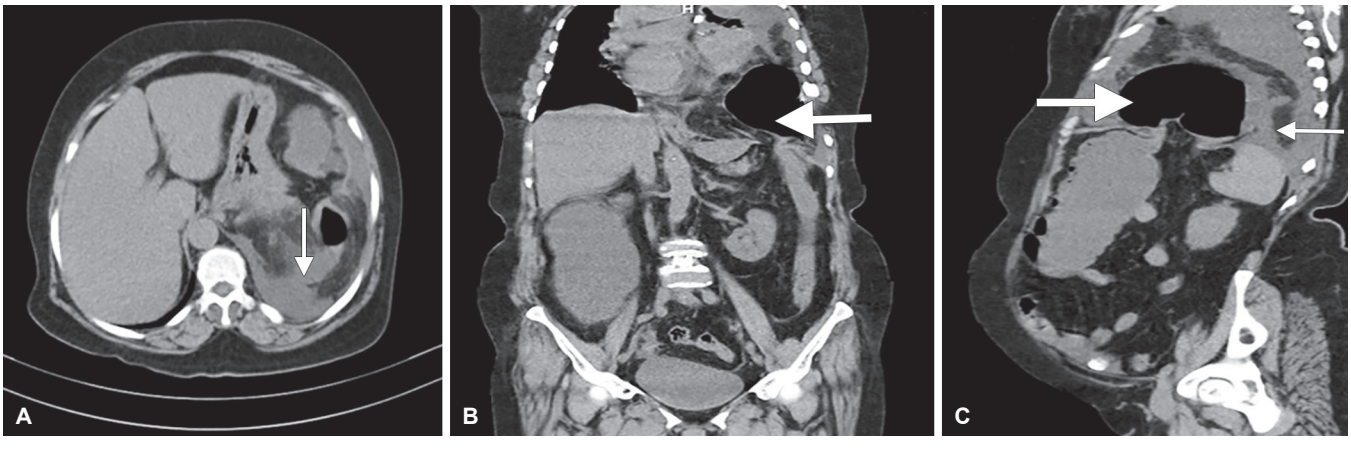
Fig. 1: Chest radiograph (PA view) shows trachea and mediastinum shifted to the right side (arrow), left pleural effusion (arrow), and herniation of bowel loops in the left hemithorax (long arrow)

reported to be 1 in 2,000 to 5,000 live births.[2] The CDH usually presents with respiratory distress during the neonatal period and is associated with pulmonary hypoplasia. The prognosis depends on the degree of pulmonary hypoplasia, pulmonary hypertension, and other anomalies.[2] Late presentation of CDH is uncommon, accounting for 5 to 30% of all CDH cases in several studies.[3] Beyond infancy, it is an unusual presentation. Other causes of diaphragmatic hernia are: Trauma, phrenic nerve palsy, and long standing acquired hiatus hernia. The CDH Study Group studied the demographics, clinical manifestations, and outcome of late onset CDH[4] and found the incidence to be 2.6%. The male to female ratio was 1.8 with a mean age at diagnosis of 372 days.[4] Presenting symptoms of late onset CDH could be classified as: (1) Respiratory (upper respiratory tract infection, pneumonia, respiratory distress, cough, wheezing, etc.), (2) gastrointestinal (vomiting, abdominal pain, failure to thrive, constipation, etc.), (3) both, or (4) asymptomatic.[4]

Gastrointestinal symptoms predominate in left-sided hernias whereas respiratory symptoms are more common in right-sided hernias.[4] The CDH Study Group also concluded that the prognosis of late-onset CDH is excellent once the correct diagnosis is made. However, making the correct diagnosis is challenging because of its diverse clinical presentations. High index of clinical suspicion and a CXR are important in making a diagnosis. Nevertheless, the variety of organs and the size of herniation may make CXR interpretation difficult and late presenting CDH may mimic pneumonia, tumor, and diaphragmatic eventration. The hernia may be intermittent and the presence of a normal CXR does not exclude the diagnosis.[5] The CDH is a displacement of the abdominal organs into the thoracic cavity through a weak area or a distinct defect in the diaphragm. The causes of precipitating visceral herniation may be related to mechanical or pressure changes in the thoracoabdominal cavities.

In the case being discussed, we initially suspected herniation of stomach but that was ruled out by giving oral contrast and the postcontrast scan revealed stomach in the left hypochondrium; there is herniation of the splenic flexure into the left hemithorax and dilatation of the distal ileum, ascending and transverse colon. Left lung was hypoplastic resulting in the shift of mediastinum to the right side. As already discussed, the patient presented with abdominal symptoms (left-sided hernias) rather than respiratory symptoms (right-sided hernias).

The most frequent types of diaphragmatic hernia are: (1) Left posterolateral (Bochdalek hernia) and (2) sternocostal (Morgagni hernia). Bochdalek hernia, resulting from inadequate closure of the posterolateral pleuroperitoneal membrane, is the most frequently seen CDH. Defects occur more frequently on the left side than on the right side of the diaphragm, and the abdominal contents including stomach, bowel loops, liver, spleen, or fat tissues may be



Figs 2A to C: Axial, coronal, and sagittal CT scan images show defect in diaphragm (arrow) with herniation of bowel loops, dilated ascending loop (arrow), and collapse descending loop (arrow)

displaced into the thoracic cavity. A posterolateral hernia on the right side is very rare and this is probably attributable to the protection provided by the liver. Foramen of Morgagni hernias are rare diaphragmatic hernias, usually occurring on the right and located in the anterior mediastinum because of the retrosternal location of the foramen of Morgagni, described as an anterior diaphragmatic defect. In adults, foramen of Morgagni hernia is also associated with obesity, trauma, weight lifting, or other causes of increased intra-abdominal pressure. The most frequent cause of herniation of the abdominal viscera in adults seems to be trauma, whereas in babies or newborns it is most often attributable to congenital absence or defective fusion of the septum transversum or the pleuroperitoneal membrane.

The detection of diaphragmatic hernia is made with prenatal ultrasonography in 50 to 90% of cases.[6,7] When diagnosis is made *in utero*, amniocentesis is often performed for detecting chromosomal aberrations and may help to estimate lung maturity.[8,9] After birth, a diagnosis can readily be made based on of symptoms and physical signs. A plain X-ray of the thorax and abdomen provides details of the position of the herniated viscera. Blood gases and pH status reflect the efficiency of gas exchange. A physical examination may be sufficient, but passing a nasogastric catheter into the stomach before X-rays may help to locate it or to detect any esophageal displacement.[10] In some rare cases, herniation of viscera through the diaphragm is an incidental finding in adult patients undergoing CXR or CT scans for other symptoms not related to this pathology.

CONCLUSION

This case highlights the diagnostic difficulties that clinicians face when dealing with late presentation of CDH due to its rarity and diverse presentations. Hitherto, early presentation of CDH has been reported. Late presentation of CDH is uncommon. Plain CXR and CT are the modalities used in detection and diagnosis of CDH. The role of oral contrast cannot be underestimated in the evaluation of the type of bowel loop herniated-stomach, small bowel, or large bowel.

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