Adult Congenital Diaphragmatic Hernia Presenting with Respiratory Symptoms: A Case Series

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ABSTRACT

Congenital diaphragmatic hernia (CDH) is typically recognized in the neonatal period due to severe respiratory compromise; however, a small proportion of cases present in adulthood with atypical manifestations. We present a case series of three adult female patients who presented primarily with respiratory symptoms and were subsequently diagnosed with CDH. Case 1 is a 60-year-old female presenting with progressive breathlessness and cough. Computed tomography (CT) revealed a 3.5 cm anterior diaphragmatic defect with herniation of the transverse colon and peritoneal fat. Case 2 is a 35-year-old female presenting with shortness of breath, left-sided chest pain, and abdominal discomfort. CT demonstrated a large left-sided defect with herniation of the stomach and bowel, causing mediastinal shift. Case 3 is a 66-year-old female with a two-year history of exertional breathlessness. CT imaging confirmed a right-sided defect with omental herniation. In all three cases, initial chest radiography was non-specific, mimicking other thoracic pathologies. Prompt diagnosis via CT imaging enabled accurate anatomical mapping and timely surgical referrals. This series highlights that adult CDH, though uncommon, should be considered in the differential diagnosis of unexplained cardiopulmonary symptoms. Early recognition utilizing CT prevents misdiagnosis and serious complications such as visceral strangulation.

Keywords: Congenital Diaphragmatic Hernia; Dyspnea; Tomography.

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Case 1

A 60-year-old woman presented to the pulmonary medicine outpatient department with progressive breathlessness for two months (Modified Medical Research Council [mMRC] grade II–III). She also reported a cough with expectoration for two weeks and a low-grade fever for two days. She had no history of chest or abdominal trauma, altered bowel habits, or prior thoracoabdominal surgery. Physical examination revealed reduced breath sounds over the right lower lung zone. A chest radiograph demonstrated a non-homogeneous opacity in the right lower zone, raising the suspicion of pulmonary consolidation. A CT scan of the chest and abdomen revealed a defect measuring approximately 3.5 cm along the anterior diaphragmatic curve, with herniation of peritoneal fat and a segment of the transverse colon into the anterior mediastinum. No intrinsic lung pathology was identified. A diagnosis of anterior congenital diaphragmatic hernia was established, and the patient was referred for elective surgical management.

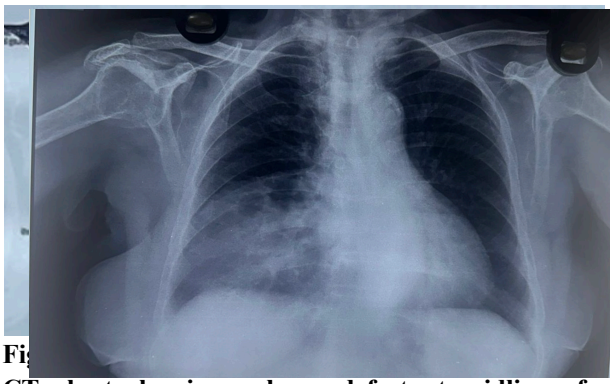
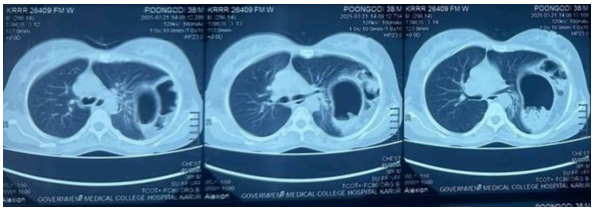


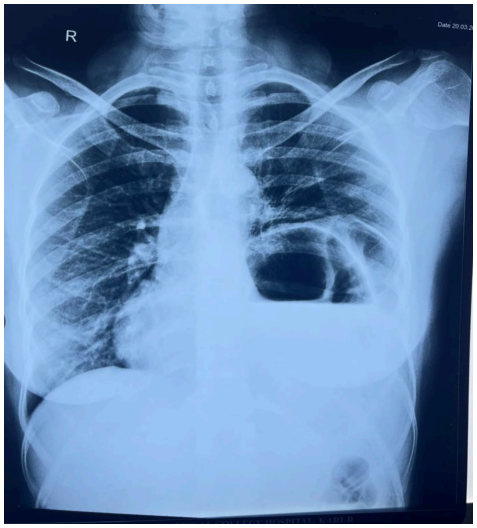
Fig. 2 (a) CT chest showing large left sided diaphragmatic defect (b) Chest x-ray showing bowel loops in the left side lower zone

Fig. 1 CT chest showing a large defect at midline of anterior diaphragmatic curve

Case 2

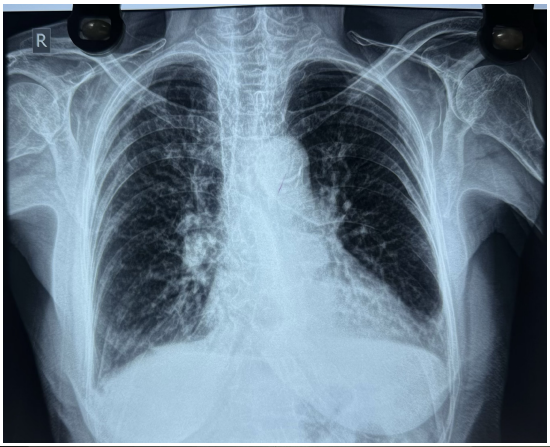
A 35-year-old woman presented with shortness of breath (mMRC grade II) and left-sided, non-radiating chest pain for two weeks, associated with vague abdominal discomfort for two months. She denied fever, cough, trauma, or prior surgery. Initial chest radiography revealed bowel loops occupying the left lower thoracic cavity with an elevated and discontinuous left hemidiaphragm. CT of the chest demonstrated a large left-sided diaphragmatic defect with herniation of the stomach, bowel loops, and omentum into the left hemithorax. This resulted in compression and partial collapse of the left lung with a mediastinal shift to the right. The findings were consistent with a large left-sided CDH, and the patient was referred for surgical repair following pulmonary evaluation.

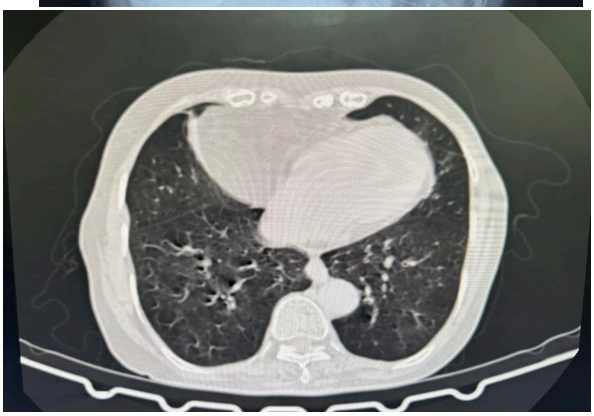




Case 3

A 66-year-old woman presented with a two-year history of gradually progressive exertional breathlessness (mMRC grade II–III). There was no associated chest pain, cough, gastrointestinal symptoms, or history of trauma. Chest radiography showed elevation of the right anterior hemidiaphragm with an ill-defined opacity. CT imaging revealed a defect measuring approximately 4 × 1.3 cm in the right hemidiaphragm, accompanied by the herniation of omental fat into the thoracic cavity. A diagnosis of right-sided CDH was established, and the patient was advised to pursue a surgical consultation.





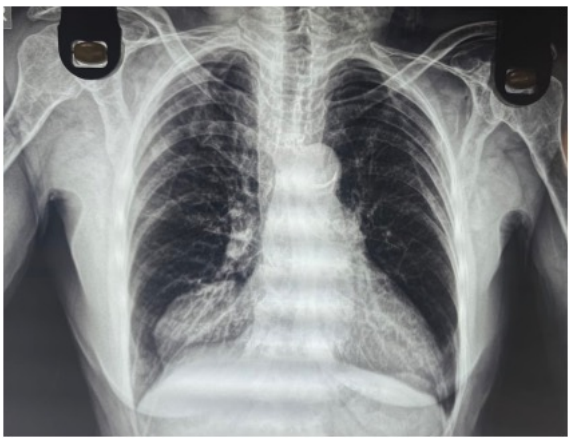


Fig. 3 (a) Pre-op X-ray (b) Post-op X-ray (c) CT Chest showing right diaphragm with herniation of omentum

Discussion

Congenital diaphragmatic hernia (CDH) is a developmental defect of the diaphragm permitting the herniation of abdominal viscera into the thoracic cavity [1]. The estimated incidence is approximately 1–5 per 10,000 live births, with most cases identified prenatally or in the perinatal period [2,3]. Adult CDH results from the incomplete closure of the pleuroperitoneal membrane or septum transversum during embryogenesis [8].

The defect is most often a Bochdalek hernia (posterolateral, typically left-sided) or a Morgagni hernia (anterior retrosternal, usually right-sided) [9,10]. While often asymptomatic from birth, adult presentations can be triggered by factors that raise intra-abdominal pressure, such as obesity, pregnancy, weight lifting, or trauma [5]. In adults, pulmonary development is typically adequate, and symptoms are more likely to result from lung compression, altered thoracic mechanics, or intermittent visceral herniation [1].

The clinical spectrum ranges from incidental radiological findings to acute respiratory or gastrointestinal compromise [5,9]. As seen in our series, patients predominantly presented with varying grades of dyspnea and chest discomfort [4]. The diagnostic challenge lies in differentiating CDH from other thoracic conditions; chest X-rays showing air-fluid levels, bowel loops in the thorax, or opacities can frequently mimic pleural effusion, pneumonia, or atelectasis [6]. Consequently, misdiagnoses, including Chronic Obstructive Pulmonary Disease (COPD) or gastric disorders, are common [4,6].

CT imaging is the gold standard for confirmation, as it accurately demonstrates the continuity of bowel or omentum through the diaphragmatic defect and allows for precise anatomical mapping [7]. If left untreated, complications such as visceral incarceration, strangulation, bowel obstruction, and respiratory failure can occur. Therefore, elective surgical repair—via laparotomy, thoracotomy, or minimally invasive approaches—is recommended even in asymptomatic patients to prevent these life-threatening events [7,9].

The main strength in this case series is the presentation of the varied and sometimes misleading respiratory symptoms and signs of adult congenital diaphragmatic hernia (CDH), with detailed clinical descriptions and good correlation with radiographic findings, especially with the use of computed tomography in the definitive diagnosis of the cases. The use of three cases with different locations and symptomatology is beneficial in the educational value of this case series, emphasizing the importance of considering CDH in atypical presentations of cardiopulmonary diseases and the challenges in diagnosing CDH, especially with inconclusive chest X-ray findings.

Nevertheless, the study has some limitations, including the small number of patients, which is not representative of the entire population. The study was conducted with female patients only.

Conclusion

Adult diaphragmatic hernias, though uncommon, must be considered in patients with unexplained cardiopulmonary or gastrointestinal complaints. CT imaging serves as the cornerstone for accurate diagnosis and intervention planning, preventing potential complications like volvulus or strangulation. Early recognition transforms clinical outcomes in these conditions that are often "hidden in plain sight".

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