



Ectopia Cordis Interna in an Asymptomatic Adult Male: A Rare Case Report

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Abstract

Ectopia cordis is a rare congenital malformation in which the heart is partially or completely located outside the thoracic cavity. Most cases present in infancy with severe cardiorespiratory compromise. We report an exceptionally rare presentation of ectopia cordis interna, in which a structurally normal heart is located entirely within the upper abdomen, discovered incidentally in an asymptomatic young adult male undergoing evaluation for a respiratory infection. This case highlights the importance of recognising unexpected mediastinal abnormalities on routine imaging and underscores the need for careful correlation between clinical findings and radiological appearances.

Keywords: Ectopia Cordis Interna; Congenital Cardiac Anomaly; Absent Cardiac Silhouette; Incidental Radiographic Finding; Adult Congenital Heart Disease; Mediastinal Abnormalities; Cross-Sectional Imaging; Asymptomatic Presentation

Introduction

Ectopia cordis is a spectrum of congenital anomalies characterised by abnormal positioning of the heart outside its usual intrathoracic location. The condition is typically diagnosed at birth and is frequently associated with thoracoabdominal wall defects, intracardiac malformations, and high neonatal mortality. Survival into adulthood is exceedingly rare [1-3].

Ectopia cordis interna, a variant in which the heart is located within the abdominal cavity while maintaining intact thoracic structures, is exceptionally uncommon [4-5]. Adult presentations are almost unheard of. This case provides a unique opportunity to explore the anatomical, clinical, and diagnostic considerations associated with this anomaly and reinforces the importance of maintaining diagnostic curiosity when encountering unexpected radiographic findings.

Case Presentation

A 28-year-old previously healthy male presented to primary care with a 4-day history of productive cough and low-grade fever. He denied dyspnoea, chest pain, palpitations, syncope, or exercise intolerance. His medical history was unremarkable, and he had no known congenital abnormalities.

On examination, he was afebrile with stable vital signs. Chest auscultation revealed mild crackles in the right lower zone. Cardiac auscultation was notable for poorly localised heart sounds, which were faint but regular. This was initially attributed to chest wall resonance and body habitus. However on inspection, palpation and auscultation, a normal beating heart was seen and felt in the epigastrium and the heart sounds were normal.

Investigations

Routine blood tests showed mild leukocytosis and elevated C-reactive protein to 35 g/dl.

A standard posteroanterior chest radiograph demonstrated absence of the expected cardiomeastinal silhouette, no visible cardiac borders or aortic contour, clear lung fields except for patchy right lower-zone infiltration, normal bony thorax and diaphragm. The absence of a cardiac silhouette prompted further evaluation.



Figure 1: Posteroanterior Chest Radiograph.

Posteroanterior chest radiograph demonstrating complete absence of the expected cardiomeastinal silhouette. No cardiac borders, aortic knuckle, or mediastinal contours are visible. Lung fields are clear except for patchy right lower-zone infiltration consistent with infection. The bony thorax and diaphragm appear normal. These findings prompted further evaluation for congenital mediastinal anomalies.

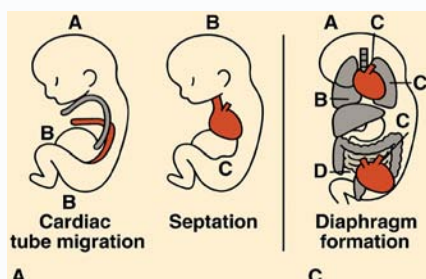


Figure 2: Figure X. Embryological stages of cardiac development leading to ectopia cordis interna.

Sequential schematic illustrations showing key stages in embryonic heart development:

- Panel A: Cardiac tube migration — The primitive cardiac tube (B) forms ventrally to the neural tube (A) and begins its descent toward the thoracic cavity.
- Panel B: Septation — The developing heart (C) undergoes internal division into atria and ventricles, establishing the four-chambered structure.
- Panel C: Diaphragm formation — The diaphragm (D) forms as a muscular partition between thoracic and abdominal cavities. In ectopia cordis interna, abnormal cardiac migration or delayed diaphragm closure may result in the heart (C) residing entirely within the abdomen.

Labels:

- A) Neural tube and cranial structures
- B) Cardiac tube or developing lungs
- C) Heart (migrating, septating, or positioned)
- D) Diaphragm or thoracoabdominal boundary

Ultrasound, colour doppler and echocardiography in the thorax and abdomen revealed a structurally normal heart located entirely within the upper abdomen, inferior to the diaphragm and anterior to the stomach, intact thoracic cavity with no pericardial sac present, normal great vessel continuity, with elongated vascular pedicles traversing the diaphragm, no associated thoracoabdominal wall defects and right lower-lobe consolidation consistent with infection. This imaging confirmed ectopia cordis interna. Patient was offered CT scan imaging but was refused as he was asymptomatic and was poor to afford the cost.

Differential Diagnosis

The differential diagnosis for an absent cardiac silhouette on chest radiograph included technical factors (rotation, under-penetration), congenital absence of the pericardium, diaphragmatic hernia with

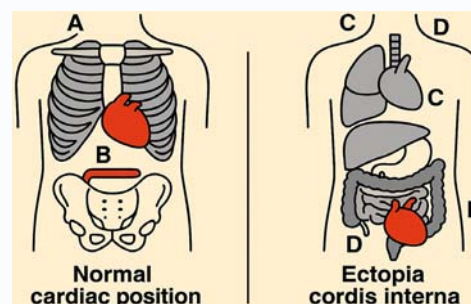


Figure 3: Comparative anatomy of normal cardiac position and ectopia cordis interna.

Side-by-side schematic illustrations demonstrating cardiac positioning:

- Left panel: Normal cardiac position — The heart (C) is located within the thoracic cavity, nestled between the lungs (B) and protected by the ribcage (A). The pelvis (D) is shown for anatomical reference.
- Right panel: Ectopia cordis interna — The thoracic cavity is empty, with the heart absent from its expected location. The lungs (C) remain in place, while the heart is positioned within the upper abdominal cavity, adjacent to the intestines (D). The ribcage (A) is intact.

Labels:

- A) Ribcage
- B) Lungs (left panel only)
- C) Heart (normal vs absent from chest)
- D) Pelvis or intestines (depending on panel)

mediastinal shift, post-pneumonectomy state, true ectopia cordis variants and imaging artefact.

Treatment

The patient was treated conservatively for community-acquired pneumonia with oral antibiotics. No intervention was required for the congenital anomaly, as he remained asymptomatic with normal cardiac function.

Outcome and Follow-Up

The patient made a full recovery from his respiratory infection. He was referred to cardiology for baseline assessment, which confirmed normal cardiac structure and function. He was counselled regarding the congenital anomaly and advised to seek medical attention if he developed symptoms such as exertional dyspnoea, palpitations, or syncope. Annual follow-up was recommended.

Discussion

Ectopia cordis is a rare congenital anomaly with an incidence of approximately 5.5–7.9 per million live births [6]. Most cases involve thoracic or thoracoabdominal displacement of the heart and are associated with severe structural defects [7]. Survival into adulthood is extremely rare.

Ectopia cordis interna, in which the heart is located within the abdominal cavity, represents an even rarer variant. The embryological basis is thought to involve failure of normal thoracic cavity formation or abnormal migration of the cardiac primordium [8-10]. The absence of associated thoracoabdominal wall defects in this case is particularly unusual.

This case underscores several important clinical lessons:

- Unexpected radiographic findings should always prompt further investigation, even in clinically stable patients.
- Congenital anomalies may remain undiagnosed into adulthood, particularly when asymptomatic.

- CT imaging plays a crucial role in defining anatomical relationships and excluding associated structural abnormalities.
- Long-term prognosis in isolated ectopia cordis interna is uncertain due to the rarity of adult cases.

This manuscript describes the incidental discovery of a rare congenital anomaly—ectopia cordis interna—in a clinically stable young adult male presenting with a chest infection. The case is unique in its asymptomatic presentation, absence of associated thoracoabdominal wall defects, and the complete intra-abdominal location of the heart confirmed on cross-sectional imaging. To our knowledge, adult presentations of this anomaly are exceptionally rare and have not been widely documented in the literature.

We believe this case offers valuable educational insights into:

- The importance of correlating radiographic findings with clinical presentation
- The role of cross-sectional imaging in evaluating unexpected mediastinal abnormalities
- The potential for congenital anomalies to remain undiagnosed into adulthood

Learning Points

- Ectopia cordis interna is an exceptionally rare congenital anomaly that may remain undiagnosed until adulthood.
- Absence of a cardiac silhouette on chest radiograph warrants urgent cross-sectional imaging to determine cardiac position and associated anomalies.
- Asymptomatic adults with ectopia cordis interna may not require intervention, but cardiology follow-up is advisable.
- Clinicians should maintain diagnostic curiosity when imaging findings do not correlate with clinical expectations.

Conclusion

Ectopia cordis interna is an exceptionally rare congenital anomaly, and its incidental discovery in an asymptomatic adult underscores the remarkable variability in congenital cardiac presentations. This

case demonstrates that even profound anatomical deviations may remain clinically silent and undetected well into adulthood. It also highlights the importance of maintaining diagnostic vigilance when radiographic findings do not align with expected anatomical patterns. Cross-sectional imaging is essential for accurate characterisation of unusual mediastinal appearances, and long-term follow-up is advisable given the limited understanding of the natural history of this anomaly. Ultimately, this case reinforces the value of routine imaging as a gateway to uncovering unexpected congenital conditions that may otherwise remain hidden.

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