Delayed Presentation of Congenital Diaphragmatic and Hiatal Hernias in Childhood: Two Atypical Cases

Dr Zeenat Bhikhoo, Dr Jessica L. A. Mills
Division of Pediatric Surgery, IWK Health Centre and Dalhousie University, Halifax, Nova Scotia, Canada

Background

Congenital diaphragmatic hernia (CDH) and hiatal hernia (HH) usually present in neonates with respiratory distress or feeding difficulties. Late presentations beyond infancy are rare (5–25%) and often misdiagnosed as eventration, pneumonia, or mediastinal mass. Both can cause gastric volvulus, strangulation, or respiratory compromise if unrecognized.

Objectives

To highlight two atypical, delayed presentations of diaphragmatic and hiatal hernias in older children and emphasize the need for high clinical suspicion and timely surgical intervention.

Case 1: Late presenting Congenital Diaphragmatic Hernia with Gastric Volvulus

A 15 year old male with neurofibromatosis type 1 presented with acute epigastric pain and vomiting after years of misdiagnosis as eventration. CT showed gastric volvulus with a left posterolateral defect. Surgical reduction, primary repair, and gastropexy were performed with uneventful recovery.

Case 2: Large Hiatal Hernia Mimicking a Thoracic Cyst

A 10 year old male followed for cardiac arrhythmia presented with retrosternal pain and vomiting. CT and UGI revealed a large hiatal hernia with partial gastric volvulus. Laparoscopic reduction, crural repair, and fundoplication were performed. A relook procedure for adhesions was required; full recovery achieved.

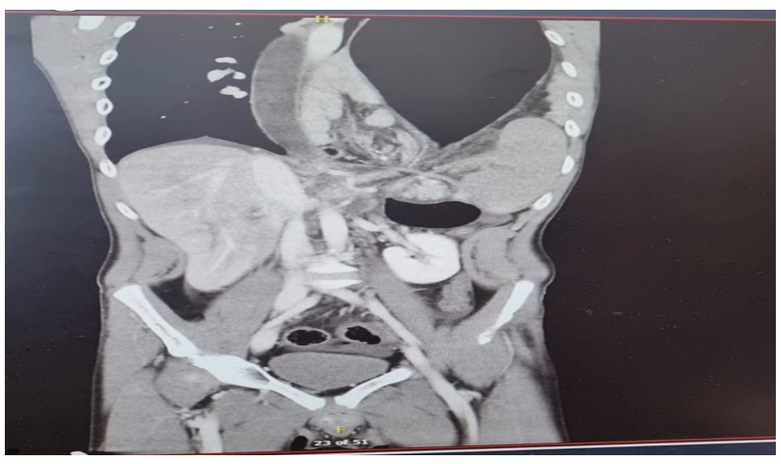

Figure 1: CT showing stomach in thoracic cavity

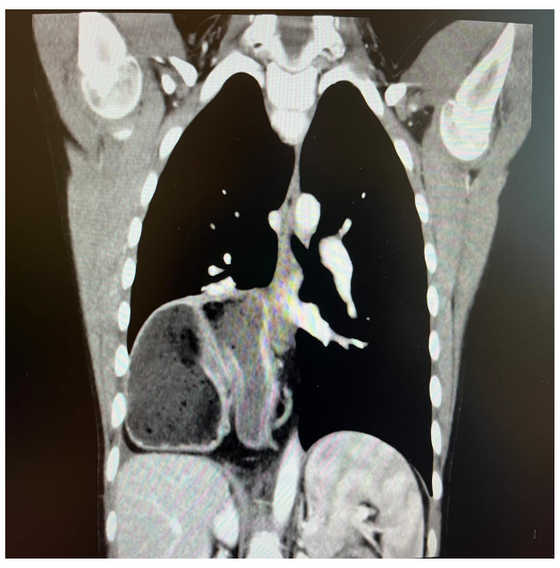

Figure 2: CT showing large hiatal hernia

Discussion

CDH can mimic eventration; HH can mimic mediastinal cysts. Cross‑sectional imaging is key for differentiation. Associations include neurofibromatosis and connective tissue disorders. Early surgical intervention prevents complications and improves outcomes. Prognosis is excellent with low recurrence rates.

Conclusion

Delayed presentations of CDH and HH are rare but clinically significant. High suspicion in older children with atypical symptoms and timely surgical repair result in excellent outcomes.

Keywords: Congenital diaphragmatic hernia, Hiatal hernia, Gastric volvulus, Late presentation, Paediatric surgery