Congenital diaphragmatic hernia in an infant: Don't miss the diagnosis!

Materne C [1], Farhat N[2], Pieltain C[3], Seghaye MC[2]

Departments of Pediatrics [1], Pediatric cardiology[2] and neonatology[3] of University Hospital Liège.

Introduction

Late-presenting Congenital Diaphragmatic Hernia (CDH) is a rare entity. Diagnosis and management remain a challenge. We hereby report a case of a two-month-old infant in whom CDH was diagnosed.

Case presentation

Neonatal antecedant – Pregnancy

- Short long bones (3rd percentile)
- Hydramnios
- Large muscular ventricular septum defect.

Neonatal antecedant – Birth

- Admission to the neonatal non intensive care unit for social reasons
- Physical exam : normal except of dysmorphic features.
  - ↘ Micro-array : normal.

At the age of 2 months, the baby was admitted to the hospital for failure to thrive. She complained of occasional vomiting and feeding difficulties.
During hospitalization, she developed respiratory distress additionally to the gastro-intestinal discomfort. Auscultation revealed decreased air entry on the left side of the chest. Chest Xray showed a left posterior-lateral diaphragmatic hernia (Figure A.) Surgical repair was performed. Post-operative outcome was favorable with improvement of gastro-intestinal symptoms and satisfactory weight gain.

In the classic form of CDH the diagnosis is often made on prenatally or at birth. The typical presentation consists of a classic symptomatic triad : respiratory distress, deviation of heart sounds and flat abdomen. The estimated overall prevalence is 1 case per 2500 to 3500 live births among whom 5-30 % are late-presenting CDH. Physiopathology and mechanism of late-presenting CDH remain unknown.

Commonly, diagnosis is suspected in case of acute respiratory distress or gastrointestinal symptoms (GI) such as volvulus. But sometimes the manifestation is non specific, such as pathological gastro-intestinal reflux, vomiting or recurrent pneumonia. Surgical repair with or without patch closure of the diaphragm is indicated.

Illustrations

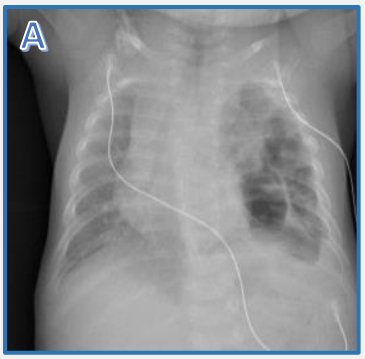
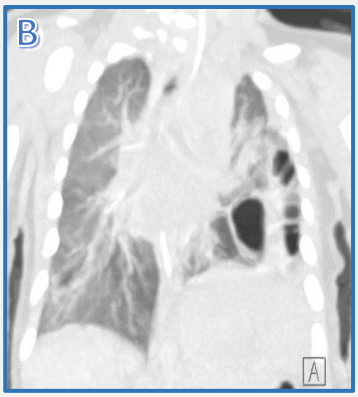


Figure A : Highlighted on chest x-ray a right mediastinal shift associated with digestive-like aerial light filling almost entirely the left pulmonary hemifield. Left lung appears atelectased at the apex and in the supra-hilar region



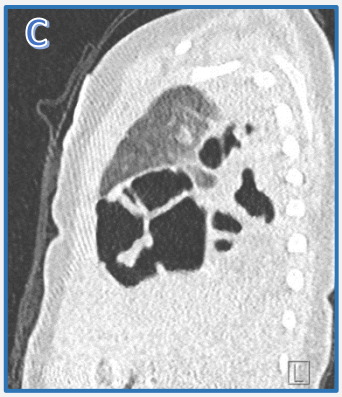


Figure B and C : CT Scanner thoracic revealing left diaphragmatic hernia with intrathoracic position of digestive loops and peritoneal fat. Presence of a slight mediastinal shift on the right side. Bearing of the left diaphragmatic dome. Subtotal left inferior lobar atelectasis. Left, upper and lower right apical lamellar atelectasis.

Conclusion

This case illustrates a late-presenting CDH with gastrointestinal symptoms and failure to thrive. Late-presenting CDH is a rare and often misdiagnosed condition. Physicians should suspect it when GI and/or respiratory symptoms do not respond to usual management.