Case Report

**Congenital diaphragmatic hernia: an unusual presentation**

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**ABSTRACT**

Congenital diaphragmatic hernia is a developmental defect of the diaphragm that allows herniation of abdominal contents into the thoracic cavity. It is usually diagnosed in the neonatal period. Late presenting cases are uncommon and difficult to diagnose, due to their rarity and diversity of semiology, with clinical presentation overlapping with other more frequent pathologies. We report a case of a 14-month-old male, with history of several episodes of bronchial hyperresponsiveness, that presented with a two-day history of nasal obstruction, emetizing cough and respiratory distress, without other associated symptoms. On physical examination we observed a 95% oxygen saturation, tachycardia, pallor, signs of respiratory distress, decreased breathing sounds in the left hemithorax and excavated abdomen. Chest radiography revealed the presence of intestinal loops in the left hemithorax, with contralateral mediastinal deviation, suggesting diaphragmatic hernia. Surgical correction was performed with direct closure of the posterolateral defect. This case highlights the importance of clinical evaluation and considering this entity in cases of persistent gastrointestinal or respiratory symptoms, in all age groups.

**Keywords:** Diaphragmatic hernia, Bochdalek hernia, Infants

**INTRODUCTION**

Congenital diaphragmatic hernia is a developmental defect of the diaphragm that allows herniation of abdominal contents into the thoracic cavity.1,2 The defect is usually observed on the left side (80-90%) and less frequently on the right side (15%) or bilaterally (1-2%).1-3 Furthermore, the posterolateral location (known as Bochdalek hernia) is more common than the anterior (known as Morgagni hernia) or the central hernia.1,4 It may present itself as an isolated finding or associated with other anomalies, such as chromosomal abnormalities (trisomies 18, 13 and 21), congenital heart disease, gastrointestinal or genitourinary anomalies and neural tube defects.1-3

The defect can vary from a small opening to a complete absence of diaphragm; therefore, the clinical presentation can be equally variable.1,5 The most common clinical presentation includes respiratory distress and cyanosis in the first hours or days of life.1,3,5 Nevertheless, a minority of patients show few or no symptoms and the defect will only manifest later in life.3

**CASE REPORT**

A 14-month-old male with history of several episodes of bronchial hyperresponsiveness was brought to the emergency department with a two-day history of nasal obstruction, emetizing cough and respiratory distress, without fever or other associated symptoms. He was observed in the emergency department the day before and was discharged with the diagnosis of upper respiratory tract infection.
Regarding his medical history, he was a full-term newborn with appropriate weight for gestational age and had an uneventful perinatal period. His growth chart showed weight at the 50th percentile, length at the 85th percentile and head circumference at the 50-85th percentile. Neurodevelopment milestones were achieved at a normal age range. Apart from the history of bronchial hyperresponsiveness there were no more relevant events.

**DISCUSSION**

The herniated viscera causes lung compression and compromises vascular development during the fetal stage, resulting in variable degrees of pulmonary hypoplasia and pulmonary hypertension.\(^1\,^2\) The defect can vary from a small opening to a complete absence of diaphragm; therefore, the clinical presentation can be equally variable.\(^1\,^2\)

Late presenting cases are uncommon and often misdiagnosed, as the signs and symptoms are not specific and overlap with those of more frequent pathology.\(^3\,^4\) Our patient had history of episodes of respiratory distress which led at first to misdiagnosis of bronchial hyperresponsiveness. Clinically these cases may present acutely or insidiously with recurrent/ intermittent symptoms.\(^6\,^7\) The clinical picture may include gastrointestinal complaints (such as abdominal pain, vomiting and/or constipation), respiratory distress, chronic cough, recurrent respiratory infections, failure to thrive or even being assintomatic.\(^3\,^5\) Due to its diverse presentation, diagnosis is challenging and a high index of suspicion is necessary.\(^4\,^5\)

Similar to the case presented, herniation may be intermittent, and the presence of a previous normal chest radiography does not exclude the diagnosis.\(^5\,^6\) A possible explanation is the fact that a pre-existing congenital defect could be blocked by an adjacent solid organ and, in cases of increased intra-abdominal pressure, abdominal contents herniate into the thorax.\(^8\)

Physical examination can show a barrel-shaped chest, a scaphoid-appearing abdomen, absence of breath sounds and/or presence of peristaltic sounds on the ipsilateral side and signs of respiratory distress.\(^2\) If the defect is on the left, the heartbeat could be displaced to the right because of a shift in the mediastinum.\(^2\)

Diagnosis is usually made in the prenatal screening by ultrasound, complemented with magnetic resonance imaging if necessary.\(^2\,^3\) After birth, chest radiography confirms the diagnosis and shows herniation of abdominal contents into the hemithorax with lung compression, contralateral displacement of mediastinal structures, as well as reduced size of the abdomen with decreased or absent intra-abdominal bowel.\(^3\) In some cases, initial radiographic findings have been misinterpreted as being caused by a pneumothorax or pleural effusion and chest tubes have been inappropriately placed.\(^4\,^6\) It is helpful to place a
nasogastric tube, as it will appear within the thoracic cavity, deviated from its expected location, supporting the diagnosis.\textsuperscript{3,4,6,7} Computed tomography imaging is recommended to confirm the diagnosis and assess possible complications.\textsuperscript{4,5,7}

Patients with neonatal presentation frequently face significant mortality and long-term morbidity with respiratory, nutritional, musculoskeletal, and neurodevelopmental problems, as well as possible hernia recurrence.\textsuperscript{1,3} The outcome is worse when associated anomalies coexist, so long-term follow-up by a multidisciplinary team is recommended.\textsuperscript{1,3,4} Late presenting cases usually have a better prognosis, since lung development occurs appropriately and there is not simultaneous pulmonary hypoplasia or pulmonary hypertension.\textsuperscript{2,4,5,7} However, once diagnosis is made, it is imperative to achieve bowel decompression in order to prevent complications such as gastric volvulus, bowel strangulation or respiratory complications.\textsuperscript{4,5}

\section*{CONCLUSION}

Late presenting cases are uncommon and difficult to diagnose, due to their rarity and diversity of semiology, with clinical presentation overlapping with other more frequent pathologies. A previous normal chest X-ray does not exclude the diagnosis. This case highlights the importance of clinical evaluation and of considering this entity in cases of persistent gastrointestinal or respiratory symptoms, in all age groups.

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