Diaphragmatic Eventration in a Neonate: A Challenging Diagnosis

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Keypoints

What is known:

- Diaphragmatic eventration, a rare anomaly, can be either congenital or acquired.

- Diagnosis is normally made when a hemidiaphragm is elevated on a frontal or lateral chest radiograph. Ultrasound is useful to confirm the diagnosis and to differentiate from a diaphragmatic hernia.

Introduction

A 3-week-old female neonate presented to the emergency department with cough, nasal obstruction, and dyspnea in the past 24 hours. She was born by vacuum-assisted delivery, after a 39-week gestation. The infant was referred for pediatric cardiology evaluation, and echocardiogram showed a persistent left superior vena cava draining into the coronary sinus. Her perinatal period was uneventful.

Physical examination revealed hypoxemia. Even though there were obvious signs of breathing difficulty, pulmonary auscultation was normal. The chest radiograph showed a right lower lobe opacity (Fig. 1).

At this stage, differential diagnoses included pleural effusion, diaphragmatic hernia, diaphragmatic eventration, and less probably, congenital pulmonary lesions. Complete blood count and C-reactive protein were normal. A few days after hospitalization, the blood culture was sterile, and she tested positive for

What is added:

- As observed in this case, diagnosis is incidental in most patients, and since the outcome is often benign, an excessive investigation should be avoided.

rhinovirus, supporting the hypothesis of viral upper respiratory infection.

An ultrasound suggested focal elevation of the right hemidiaphragm associated with a liver protrusion, confirming the suspicion of concomitant diaphragmatic eventration. After one week of surveillance and general supportive care, the infant was discharged home.

Eventration of the diaphragm, an uncommon disorder, consists of thinned diaphragmatic muscle leading to an abnormal contour of the diaphragmatic dome, with no disruption in continuity.^{1,2}

This condition is a consequence of either inadequate development (congenital) or atrophy (acquired) of the diaphragm.³ Early clinical presentation and a history of congenital heart defect may be suggestive of a congenital etiology.⁴ In neonates, clinical presentation ranges from asymptomatic to life-threatening respiratory distress, due to the elevation of intra-abdominal organs, resulting in compression of the lungs.⁵ Often, as described in this clinical case, the diagnosis is made by an occasional



Figure 1. Chest radiograph, frontal (a) and lateral (b) projections, shows elevated right hemidiaphragm.

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finding only after a chest radiograph is performed for other reasons.^{2,4} Ultrasound is of utmost importance to distinguish an intact diaphragm, suggestive of eventration, from a diaphragmatic defect, as observed in a diaphragmatic hernia.

In asymptomatic patients, treatment is generally conservative.⁵ We aimed to raise awareness about this anomaly to prevent unnecessary medical investigations.

Keywords: Diagnosis, Differential; Diaphragmatic Eventration/complications; Diaphragmatic Eventration/ diagnosis; Infant, Newborn

Author Contribuitions

ATR participated in the study conception or design. ATR participated in acquisition of data. ATR participated in the analysis or interpretation of data. ATR participated in the drafting of the manuscript. AB, JS and FR participated in the critical revision of the manuscript. All authors approved the final manuscript and are accountable for all aspects of the

work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Protection of human and animal subjects

The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki 2013).

Provenance and peer review

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Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data

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