

Images in clinical medicine



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Vaishnavi Yadav,  Pallavi Harjpal

Corresponding author: Vaishnavi Yadav, Department of Cardiovascular and Respiratory Physiotherapy, Ravi Nair Physiotherapy College, Datta Meghe Institute of Higher Education and Research, Sawangi (Meghe), Wardha, Maharashtra, India. vaishnavi.cardiopt@dmher.edu.in

Received: 13 Aug 2025 - **Accepted:** 08 Sep 2025 - **Published:** 04 Nov 2025

Keywords: Pulmonary hypoplasia, complete lung atelectasis, pulmonary agenesis

Funding: This work received no specific grant from any funding agency in the public, commercial, or non-profit sectors.

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Cite this article: Vaishnavi Yadav et al. A rare association of pulmonary aplasia with cervicomedullary junction anomalies: a clinical image. Pan African Medical Journal. 2025;52(95). 10.11604/pamj.2025.52.95.48957

Available online at: <https://www.panafrican-med-journal.com//content/article/52/95/full>

A rare association of pulmonary aplasia with cervicomedullary junction anomalies: a clinical image

Vaishnavi Yadav^{1,&}, Pallavi Harjpal²

¹Department of Cardiovascular and Respiratory Physiotherapy, Ravi Nair Physiotherapy College, Datta Meghe Institute of Higher Education and Research, Sawangi (Meghe), Wardha, Maharashtra, India, ²Department of Neurophysiotherapy, Ravi Nair Physiotherapy College, Datta Meghe Institute

of Higher Education and Research, Sawangi (Meghe), Wardha, Maharashtra, India

[&]Corresponding author

Vaishnavi Yadav, Department of Cardiovascular and Respiratory Physiotherapy, Ravi Nair Physiotherapy College, Datta Meghe Institute of Higher Education and Research, Sawangi (Meghe), Wardha, Maharashtra, India

Image in medicine

Pulmonary aplasia is a rare congenital anomaly characterized by the complete absence of the lung parenchyma and bronchial structures, resulting from failure of the lung bud to develop. Its estimated incidence is 1-3.4 per 100,000 live births, and it is often associated with malformations involving the cardiovascular, gastrointestinal, genitourinary, or musculoskeletal systems. We present the case of a 20-year-old female who reported a one-month history of tingling and numbness in both upper limbs, significantly limiting daily activities. She also experienced a cough with expectoration and progressive shortness of breath. Cervical spine magnetic resonance imaging (MRI) demonstrated partial block vertebrae at C2-C3 and C4-C5, kinking at the cervicomedullary junction

with cord thinning, subtle intramedullary edema, and the presence of a syrinx (Panel A, yellow arrow). Chest radiography revealed a dense opacity occupying the right hemithorax, accompanied by ipsilateral tracheal shift, hyperinflation of the contralateral lung, widening of intercostal spaces, and thoracic scoliosis (Panel B, green arrow). In the MRI, narrowing of the foramen magnum further indicated a significant craniocervical junction anomaly (Panel C, red arrow). This combination of abnormalities represents a multisystem developmental disorder, likely stemming from an early embryological insult involving both foregut derivatives and the paraxial mesoderm. The case highlights the need for a comprehensive, systematic evaluation for associated anomalies in patients with pulmonary aplasia, even when the primary presentation is neurological.

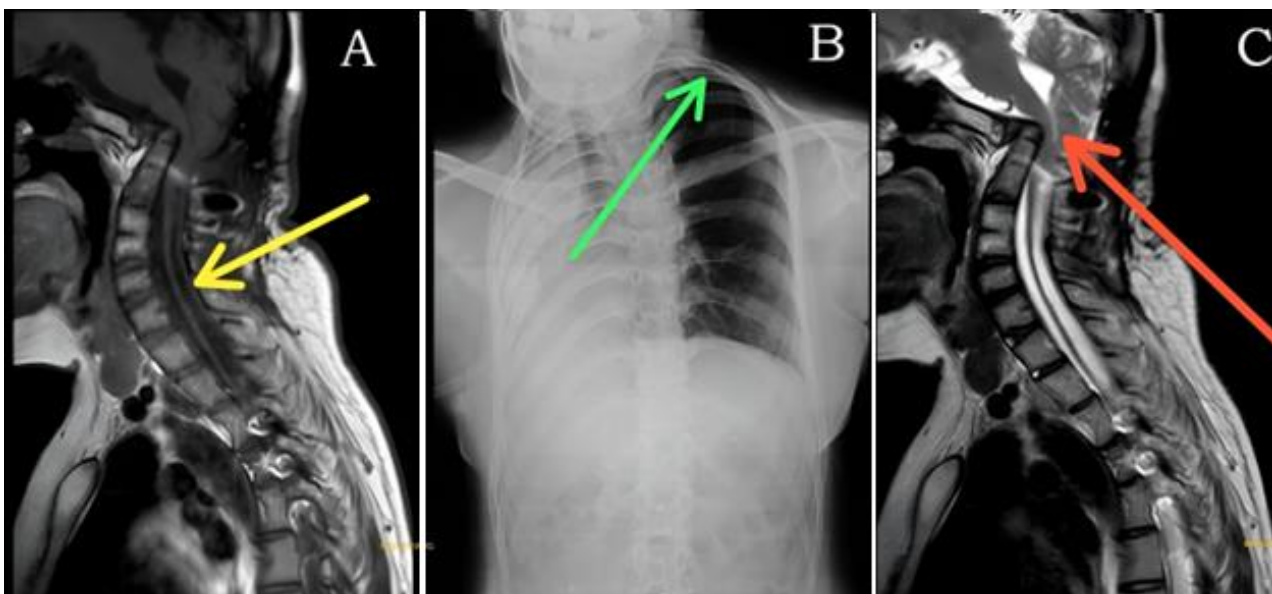


Figure 1: A) yellow arrow indicates subtle edema and syrinx formation; B) green arrow indicates lung hyperinflation and intercostal widening; C) red arrow indicates cervicomedullary kinking and thinning; narrowing of the foramen magnum was noted