Case Report

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TANAFFOS

When a Simple Bronchiolitis Reveals the Unexpected

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Background: Pulmonary agenesis is a rare congenital anomaly characterized by the complete or partial absence of lung tissue, bronchus, and pulmonary vasculature. Advancements in prenatal imaging have improved early detection; however, some cases may still go undiagnosed until after birth and can be discovered fortuitously.

Case Presentation: We present a case of a two-month-old male infant with an uneventful perinatal history who presented with respiratory distress initially diagnosed as bronchiolitis. Imaging revealed complete absence of the left lung, main bronchus, and vasculature, with compensatory hyperinflation of the right lung. Based on these findings, a diagnosis of left pulmonary agenesis was made. The infant improved with symptomatic treatment for bronchiolitis and did not require oxygen. Further investigations found no associated congenital anomalies.

Conclusion: This case emphasizes the diagnostic challenges of pulmonary agenesis in neonates, the limitations of prenatal screening, and the importance of comprehensive evaluation. In isolated cases without additional malformations, prognosis is favorable, and conservative management with follow-up is often sufficient.

Keywords: Pulmonary agenesis; Bronchiolitis; Congenital malformation

INTRODUCTION

Pulmonary agenesis is an exceptionally rare congenital anomaly, defined by the complete or partial absence of lung tissue, bronchus, and pulmonary vasculature (1). While advancements in prenatal imaging have improved early detection, certain cases may remain undiagnosed until postnatal life. This case report presents an incidental diagnosis of isolated left pulmonary agenesis in a two-month-old infant initially admitted for respiratory distress, highlighting the diagnostic challenges and the importance of comprehensive evaluation in neonatal respiratory presentations.

CASE SUMMARIES

We report the case of a two-month-old male infant, born to non-consanguineous parents, with no known family history of congenital anomalies or genetic disorders. Pregnancy was monitored regularly and remained uneventful, with all prenatal ultrasounds reported as normal. There was no history of maternal infections, teratogenic exposures, or vitamin deficiencies during gestation. The infant was born at term via spontaneous vaginal delivery, with a birth weight of 3.2 kg, appropriate for gestational age. The Apgar scores were 9 and 10 at 1 and 5 minutes, respectively, indicating good adaptation to extrauterine life. Initial postnatal examinations revealed no abnormalities, and the infant was discharged home in good health

The patient was hospitalized at two months of age for wheezing and dyspnea. On physical examination, the infant was febrile, with a temperature of 39.5°C, a

respiratory rate of 70 breaths per minute, subcostal retractions, and an oxygen saturation of 99% on room air. Cardiovascular examination was unremarkable. Pulmonary auscultation revealed bilateral wheezing with significantly decreased vesicular breath sounds on the left side.

A chest X-ray showed complete opacification of the left hemithorax, with mediastinal shift to the left and hyperinflation of the right lung exerting a contralateral mass effect (Figure 1). Laboratory tests did not indicate an inflammatory syndrome (C-reactive protein: 5 mg/L). Given these findings, a chest computed tomography (CT) angiography was performed for further evaluation. The CT scan revealed absence of the left lung, left main bronchus, left pulmonary artery, and pulmonary vein, along with compensatory hyperinflation of the right lung extending across the midline to occupy both hemithoraces. The mediastinum was significantly displaced to the left. No anomalies were noted in the right lung parenchyma, bronchi, or major thoracic vessels (Figure 2). Based on these findings, a diagnosis of left pulmonary agenesis was made.

The infant was managed symptomatically for acute bronchiolitis. Treatment included saline nasal irrigation, antipyretics, and fractional feeding. No oxygen supplementation was required. Clinical improvement was observed within 48 hours, and respiratory distress resolved progressively.

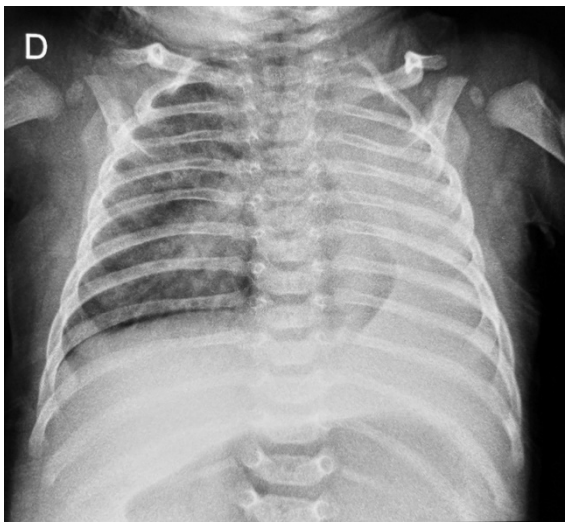


Figure 1. Chest X-ray showing complete opacification of the left hemithorax, with hyperinflation of the right lung and mediastinal shift to the left

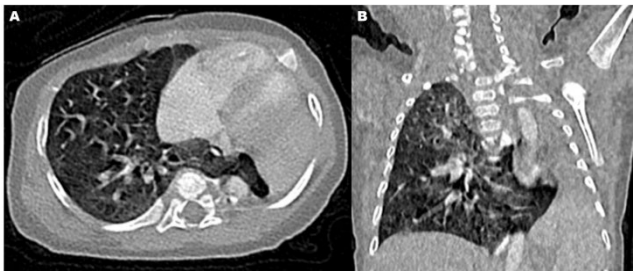


Figure 2. Axial (A) and coronal (B) views of chest CT angiography showing marked hyperinflation of the right lung extending into both hemithoraces, with complete mediastinal shift to the left, and the absence of the left lung, left main bronchus, left pulmonary artery, and left pulmonary vein

Further investigations were conducted to assess for associated congenital anomalies. A transthoracic echocardiogram showed normal intracardiac anatomy and function, except for agenesis of the left pulmonary artery, consistent with the CT findings. Transfontanellar and abdominal ultrasonography were normal. Genetic evaluation, including karyotyping, revealed a normal male 46, XY chromosomal complement, with no detectable genetic abnormalities.

DISCUSSION

Pulmonary agenesis is a rare congenital malformation classified into three types: Type 1 (Agenesis) is characterized by the complete absence of pulmonary parenchyma, bronchus, and pulmonary artery on the affected side. Type 2 (Aplasia) presents with complete absence of pulmonary parenchyma but with a rudimentary bronchus. Type 3 (Hypoplasia) involves the presence of variable amounts of pulmonary parenchyma, bronchi, and supporting vasculature (1). This condition arises between the 4th and 5th weeks of gestation, during the embryonic phase, before the pseudoglandular period when the primitive lungs emerge as a diverticulum from the foregut (2).

While the exact etiology remains unclear, several factors have been implicated, including genetic mutations, viral infections, and maternal vitamin A deficiency during pregnancy (3). Advances in imaging techniques, particularly prenatal ultrasound and fetal magnetic resonance imaging, have significantly improved early

detection rates and enabled better perinatal management (4). However, in our case, the diagnosis of pulmonary agenesis was fortuitous, despite the performance of routine prenatal ultrasounds, underscoring the potential for this anomaly to escape early detection.

The differential diagnosis includes total lung atelectasis, congenital cystic adenomatoid malformation, and congenital diaphragmatic hernia. Pulmonary agenesis is frequently associated with cardiovascular, gastrointestinal, urogenital, skeletal and cytogenetic anomalies, necessitating systematic screening (5).

Prognosis depends on multiple factors, including laterality, genetic abnormalities, and the presence of associated congenital anomalies. Right-sided agenesis is associated with a worse prognosis due to the larger functional demand placed on the remaining lung. However, isolated cases of pulmonary agenesis without additional malformations generally have a favorable outcome and can be managed conservatively with regular follow-up (6).

CONCLUSION

This case underscores the importance of considering congenital pulmonary anomalies in the differential diagnosis of persistent or severe respiratory symptoms in infants. Isolated pulmonary agenesis, though rare, may present subtly and evade detection during routine prenatal screening. A thorough clinical assessment, supported by advanced imaging modalities, is crucial for accurate diagnosis. In the absence of associated anomalies, the prognosis remains favorable, and conservative management with routine follow-up is often sufficient. This report adds to the limited literature on isolated pulmonary agenesis and emphasizes the value of early postnatal evaluation in ensuring optimal outcomes.

Ethical Considerations

This study, involving a human participant, was planned, conducted, and reported in accordance with the World Medical Association (WMA) Declaration of Helsinki. Written informed consent was obtained from the patient's parents for publication of this case report and any accompanying images.

Artificial Intelligence (AI) Disclosure Statement

During the preparation of this study, the authors used ChatGPT to review and correct spelling and grammar mistakes. After using this AI tool, the authors revised and edited the content and take full responsibility for the publication.

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