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Case Report

Unilateral Lung Agenesis -A Rare Differential

Siddarth Mathur, Ramakant Dixit, Mukesh Goyal, Hasha Thankam Somson

Department of Respiratory Medicine, JLN Medical College, Ajmer, Rajasthan, India.

Abstract

Unilateral lung agenesis is an uncommon developmental anomaly stemming from the incomplete formation of the primitive lung bud. Its manifestation and onset vary widely, from asymptomatic instances to diagnoses in adulthood prompted by recurring respiratory infections. Roughly half of these cases coincide with other congenital anomalies. A 23 years female diagnosed as a case of treated pulmonary tuberculosis with right fibrocavitatory disease on the basis of chest x-rays despite negative smear microscopy of sputum for acid-fast bacilli (AFB) referred to us with a complaint of shortness of breath on exertion. She was evaluated further and on contrast, enhanced CT chest (CECT) was detected to have right lung agenesis, which was confirmed on fiber optic bronchoscopy.

Keywords: Hypoplastic lung, Lung agenesis, Bronchoscopy.

Introduction

The discovery of pulmonary agenesis dates back centuries to De Pozze's initial description in 1673, with Muhammad recording the first case in India in 1923. [1] This congenital defect arises when the primitive lung bud fails to develop properly, often attributed to abnormal flow of blood in the 'dorsal aortic arch' during the gestational 4th week of life.[2] Onset and presentation of pulmonary agenesis exhibit considerable variability. Approximately half of all cases coincide with other congenital anomalies, notably including tracheoesophageal fistula (TEF) and VACTERL association. [4] Notably, right-sided agenesis presents a graver prognosis owing to its frequent associations with congenital cardiac abnormalities.[3] However, when isolated and devoid of other congenital abnormalities, unilateral pulmonary agenesis may manifest with milder symptoms and lead to prolonged survival rates. [1] This narrative is exemplified by a recent case involving 23-year-old females diagnosed with right lung agenesis, whose initial presentation was confused with a case of treated pulmonary tuberculosis at our center.

Case Report

A 23-year-old female patient was referred from the peripheral district hospital with a diagnosis of right fibrocavitatory disease and pulmonary tuberculosis with a complaint of shortness of breath on exertion. She had similar episodes in the past as well. She gave a history of antitubercular drug(ATT) for 6 months 2 years back, but her sputum was negative and the diagnosis was based on a chest radiograph. She had no history of any addiction to nicotine or tobacco. On general examination, she was undernourished. There was pallor but no other significant findings on general examination. On systemic examination, findings were suggestive of right-side mediastinal shift, which was confirmed on a new chest radiograph, which showed right-side homogeneous opacity with marked mediastinal shift and left lung hyperinflation (Figure 1).

Her oxygen saturation (spO2) was 87% on room air at rest with no distress and her blood pressure was 100/80 mmHg. She was admitted for further evaluation and CECT chest and bronchoscopy were planned. Her 2D echo is suggestive of a right side, along with moderate pulmonary artery hypertension. Her blood investigations, including blood counts and blood biochemistry, were within normal range. The

Address for correspondence: Ramakant Dixit,
Department of Respiratory Medicine, JLN Medical College, Ajmer, Rajasthan, India.
E-mail: dr.ramakantdixit@gmail.com

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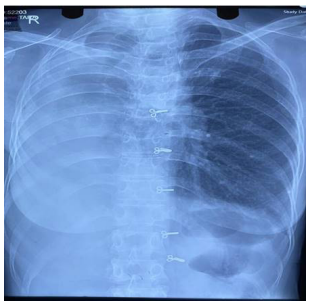


Figure 1: Chest radiograph showing tracheal and cardiac shifted towards right side along with homogeneous opacity in all zones right side.

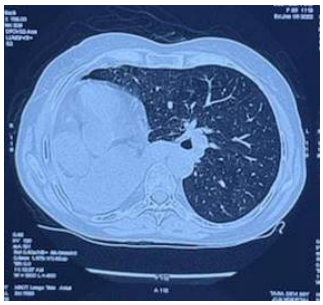
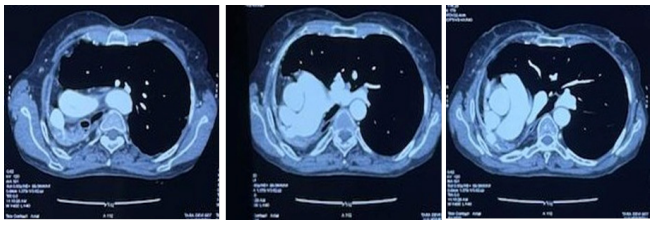


Figure 2: the lung window of the CT scan showing hyperinflation with left lung herniating towards right.



3 (A) **3 (B)** **3 (C)**

Figure 3 A, B, C: CECT chest film showing hyper inflated left lung herniating on right side. The mediastinal structure can be noted are completely shifted towards right side . In the serial cuts it can be appreciated that the trachea after bifurcation is continuing with left bronchus while the right end is ending abruptly after a short distance.

CT scan showed a left hyperinflated lung herniating towards the right side, with all mediastinal structures shifted to the right side with no lung parenchymal tissue on the right side. The right bronchus ended blindly (Figures 2, 3 A, B, C) and the computed tomography pulmonary angiography (CTPA) evidenced an absent right pulmonary trunk. Hence, the diagnosis of right lung agenesis was made. The Bronchoscopy abronchoscopyright side bronchus closed, thus confirming right lung agenesis. The patient was managed with oxygen, bronchodilator and supportive treatment and discharged after 1 week with advice of vaccination.

Discussion

Pulmonary agenesis represents a rare developmental anomaly marked by the complete absence of bronchi, pulmonary vessels, and lung tissues, with an incidence of approximately 34 cases per million live births.[1] In contrast, pulmonary hypoplasia involves a reduction in the size/number of vessels and alveoli while maintaining the gross morphology of the lung. It may manifest as localized involvement of a single lobe, affect the whole lung, or exceptionally occur bilaterally, though bilateral aplasia is nonviable. Unilateral agenesis may present with varying degrees of severity, with the left lung being more commonly affected as compared to the right, and a higher prevalence in males than females.[4]

Embryologically, pulmonary agenesis arises from a failure in the respiratory system's development from the foregut. This can result in bilateral pulmonary agenesis if there's an arrest in the growth at the stage of primitive lung bud, whereas unilateral lung agenesis occurs due to developmental issues at a later stage. Lobar agenesis arises from arrest in the development of one side of an older embryo, while pulmonary hypoplasia can occur from a failure of alveolar differentiation during the final trimester of pregnancy.[5,6]

Schneider initially classified pulmonary agenesis in 1912, with subsequent modifications by Boyden.[7,8] This classification distinguishes between types based on the extent of lung and bronchus absence, ranging from complete agenesis to rudimentary bronchus with no lung tissue. Type 1 is complete agenesis, where pulmonary parenchyma, including bronchus and vasculature, are completely absent. Type 2 is agenesis, where pulmonary parenchyma is completely lacking, but rudimentary bronchus is present on the affected side. Type 3 is hypoplasia, where a variable amount of pulmonary lung parenchyma with a decrease in the number of airways and alveolar tissue is seen. The case presented here fits into type 2, i.e., aplasia of a lung on the right side.

The etiology of pulmonary agenesis is multifactorial, involving genetic, teratogenic, and mechanical factors. It is often sporadic, with possible associations with chromosomal aberrations, consanguinity, intrauterine infections, vitamin A deficiency, and environmental factors.[4]

Clinical presentations vary widely, from neonatal respiratory distress to later-life symptoms and recurrent infections. Diagnosis involves imaging modalities like ultrasound, chest X-ray, and CT scans, along with bronchoscopy and, bronchography, and pulmonary angiography, in specific cases. Differential diagnoses of this condition include Scimitar syndrome, congenital malformations of the pulmonary airway, congenital lobar emphysema, Macleod syndrome, bronchopulmonary sequestration, etc., apart from destroyed lung.[9] The condition may be wrongly diagnosed as destroyed lung syndrome due to tuberculosis and the patient may receive antitubercular treatment without actual indication, as occurred in this case. However this can be differentiated by the fact that the chest wall symmetry is maintained on the affected side in hypoplastic lung disease.

Management strategies depend on the patient's symptoms and associated anomalies, ranging from

conservative approaches to surgical interventions.[6] Prognosis is influenced by the severity of co-morbid conditions and the degree of involvement of the remaining lung tissue in the disease process.[1,4] Right-sided diseases have more mortality as compared to left sided. Also, in case of isolated pulmonary involvement, a patient may survive upto adulthood as compared to those having pulmonary disease associated with other congenital anomalies.

In conclusion, diagnosing and managing pulmonary agenesis, especially in cases without additional congenital anomalies, presents unique challenges requiring thorough clinical evaluation and multidisciplinary care.

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