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LUNG UNDERDEVELOPMENT: CASE REPORTS AND A LITERATURE REVIEW

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ABSTRACT

Lung underdevelopment is a rare congenital anomaly with variable clinical significance and presenting symptoms. It usually manifests during childhood. We present two cases of developmental lung anomaly subtypes and discuss clinical presentation and outcomes in such patient populations.

KEYWORDS

Lung agenesis, hypoplasia, TB infection, PFT, Pulmonary hypertension, congenital malformation

LEARNING POINTS

- Pulmonary underdevelopment is a challenging diagnosis and should be considered in patients with unilateral opacification on chest radiograph.
- Childhood developmental history is critical for diagnosis as delayed, or misdiagnoses are common. Definitive diagnosis can be made by computed tomography scan.
- Management is watchful waiting with close monitoring, with long term prognosis remaining unclear.

CASE PRESENTATION

The two patients shared a similar background prior to presentation. Both emigrated from Venezuela with documented positive purified protein derivative (PPD) tests during childhood.

Case one

A 39-year-old Venezuelan female presented to our outpatient clinic with a complaint of dyspnea on exertion. She described her symptoms as shortness of breath with a hastened pace on ground level or walking uphill. She denied any coincidental cough, fever, chills, or other constitutional symptoms. She was noted to have a positive PPD test back in 2000 as part of her immigration process but was never treated for tuberculosis (TB).

Physical examination findings

On examination at the time of presentation, blood pressure was 97/62 mmHg, heart rate was 84/min, respiratory rate was 18/min, and temperature was 37.1 °C. The patient's oxygen saturation was 97% on room air. Physical examination

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revealed depressed lung sounds over the left lower lung field. The remaining lung areas were clear to auscultation with normal chest expansion, cardiac sounds could be easily heard from the back, the abdomen was soft, and on-tender, and no lymph node could be palpated.

Diagnostic studies

A chest X-ray showed opacification of the left lung marking with mediastinal shift *(Fig. 1)*. Subsequently, a computed tomography (CT) scan of the chest showed complete agenesis of the left lung, anterior herniation of the right lung with left mediastinal shift and absence of corresponding left main pulmonary artery *(Fig. 2)*. Pulmonary function testing (PFT) showed evidence of moderately severe obstruction with a forced expiratory volume in one second/forced vital capacity (FEV1/FVC) ratio of 62% (if using moderately severe obstruction as a descriptor, consider noting the FEV1 alone), decreased total lung capacity (TLC), and vital capacity (VC) with normal diffusing capacity for carbon monoxide (DLCO) *(Fig. 3)*.

Final diagnosis

Pulmonary agenesis.

Case two

A 53-year-old Venezuelan male with a past medical history of Crohn's disease presented to the clinic with a 1-year history of progressive exertional dyspnea, with symptoms primarily described as limited walking on ground level, requiring frequent stops in order to catch his breath. He denied cough, fevers, chills, or hemoptysis. He was on azathioprine and mesalamine for otherwise well-controlled Crohn's disease. Notably, he also tested positive for a PPD skin test as a child; it was unclear if he was ever treated for TB. He was diagnosed with asthma by his primary care provider.

Physical examination findings

On examination at the time of presentation, blood pressure was 135/81 mmHg, heart rate was 83/min, respiratory rate was 16/min, and temperature was 36.8 °C. The patient's oxygen saturation was 97% on room air. Physical examination revealed bronchial breath sounds over the left lung fields. The remaining areas were clear to auscultation. Cardiac sounds revealed no murmurs or extra heart sounds. The abdomen was soft and on-tender, and no lymph node could be palpated.

Diagnostic Studies

A CT scan of the chest was initially interpreted as showing left pneumonectomy but, upon further inspection, revealed fibrosis and underdevelopment of the left lung, most likely related to his previous TB infection, with posterior herniation of the right lung *(Fig. 4)*. Pulmonary function testing showed evidence of moderate-severe obstruction with FEV1/FVC ratio of 68%, normal TLC and VC, and a normal DLCO *(Fig. 5)*.

Final Diagnosis

Pulmonary hypoplasia due to previous TB infection.

DISCUSSION

Developmental lung anomalies (DLA) are rare congenital

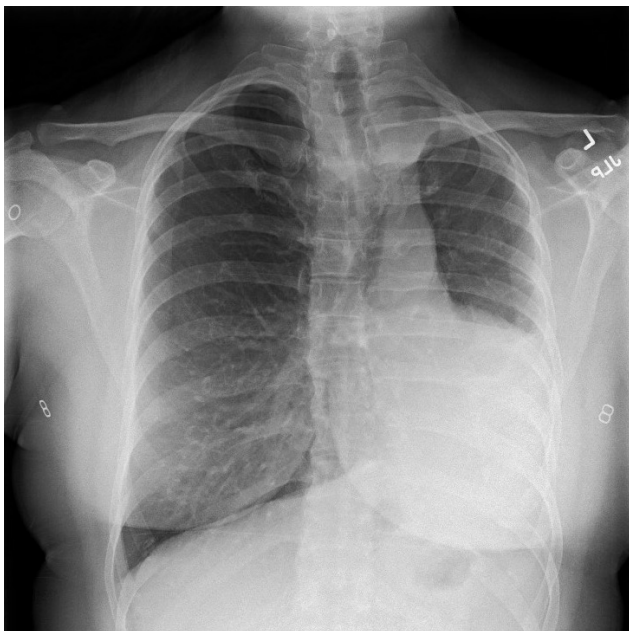

Figure 1. Chest X-ray showing volume loss and left sided tracheal deviation.

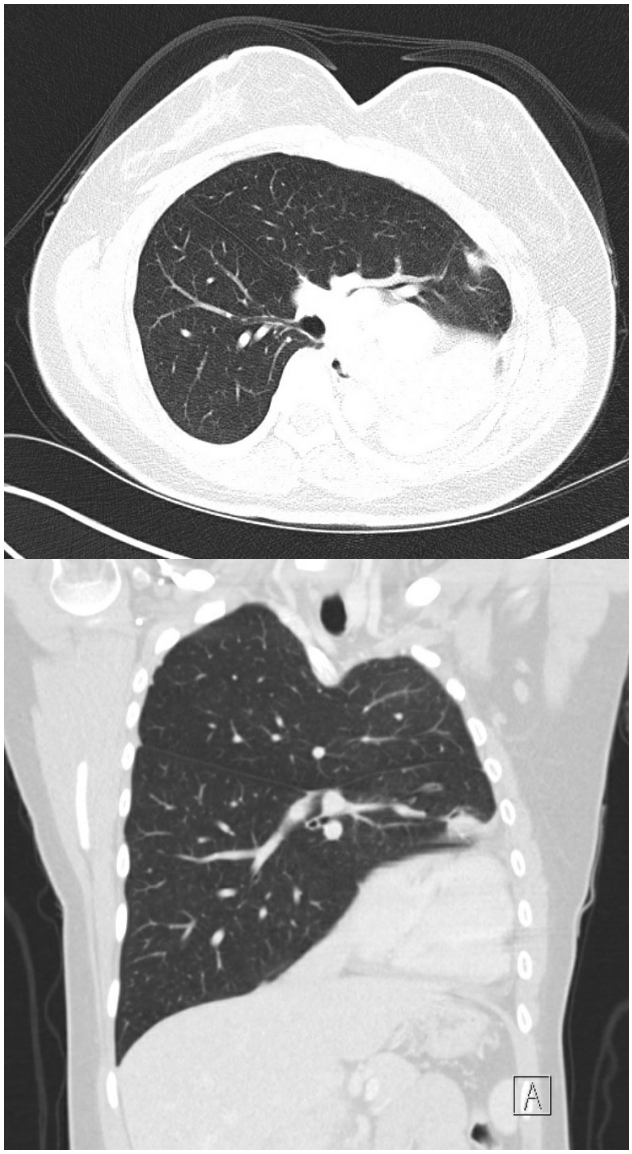

Figure 2. Computed tomography scan: axial and coronal views showing agenesis of the left lung with mediastinal shift.

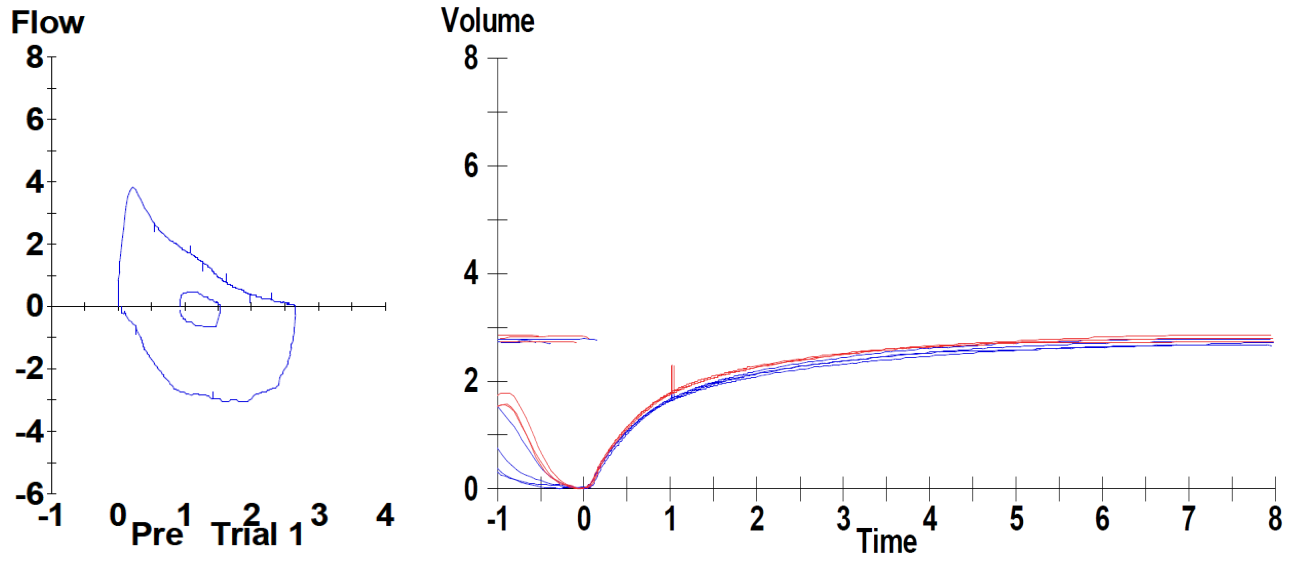

Figure 3. Flow/volume loop and volume/time curve.

malformations that vary in clinical and radiological presentation, with a reported incidence of 35 cases per 100,000. Such cases are usually diagnosed in early childhood. They can be divided into three categories based on the affected component: bronchopulmonary, vascular anomalies, and combined lung/vascular anomalies[1].

Pulmonary underdevelopment is a subgroup of congenital defects within the vascular anomaly group, as there is usually associated variable development of the pulmonary artery[1]. Although pulmonary underdevelopment may be an isolated phenomenon, nearly 50% of reported cases are linked to other congenital defects, including cardiovascular or genitourinary[2].

The left side is most commonly affected, with no clear gender or ethnic predisposition. This can be further categorized into three types: 1) type 1, or agenesis with complete absence of the lung parenchyma, vasculature, and bronchial tree; 2) type 2, or aplasia with rudimentary bronchi and complete absence of lung parenchyma; 3) type 3, or hypoplasia with variable amounts of lung parenchyma, bronchial tree, and supporting vasculature[1,3,4].

As the primary etiology remains unknown, secondary causes of pulmonary hypoplasia should be investigated, including diaphragmatic hernia, lung fibrosis secondary to antecedent childhood infection, chest wall deformities, and pleural effusions[4]. Thorough history-taking, physical examination, and radiographical investigations remain the gold standard in assessing such clinical patterns.

Clinical presentation of pulmonary underdevelopment depends on its type and simultaneously organogenesis[3]. It can vary from asymptomatic cases incidentally noted on chest imaging to presentations with a wide range of respiratory symptoms, including cough, dyspnea, and recurrent chest infections. Differential diagnoses on the basis of imaging may include complete atelectasis of ipsilateral lung secondary to mucus or endobronchial obstruction, previous thoracotomy with lobectomy, or pneumonectomy with compensatory hyperinflation of the other lung[1].

Prognosis usually depends on the presence of coexisting congenital anomalies. Management primarily focuses on symptomatic relief and control of recurrent infections of the underdeveloped lung parenchyma with limited data on life expectancy[1].

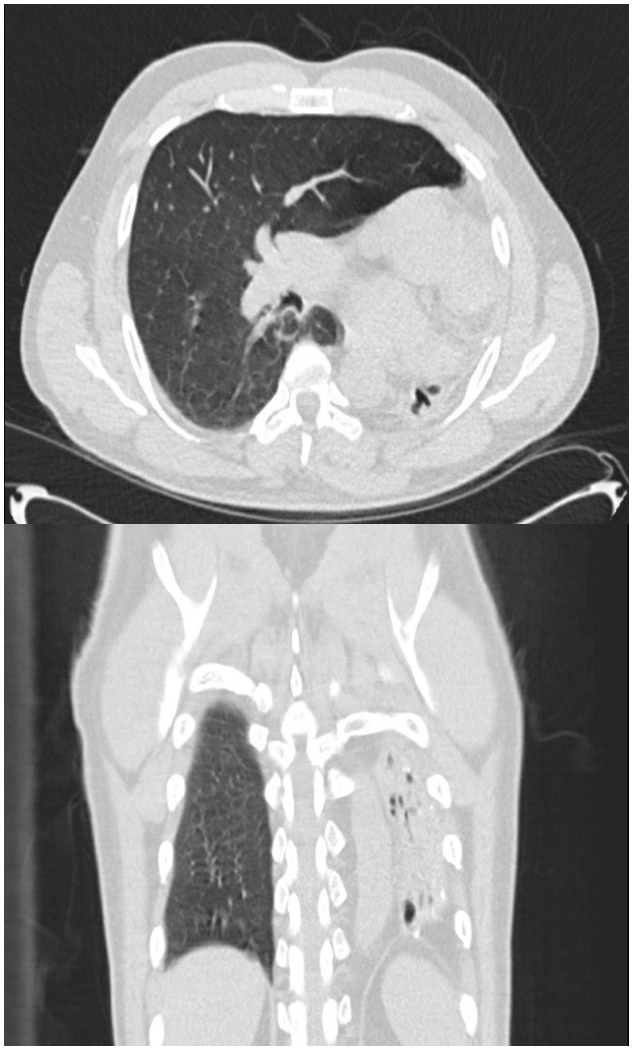

Figure 4. Computed tomography scan: axial and coronal views showing hyperplasic left lung with mediastinal shift.

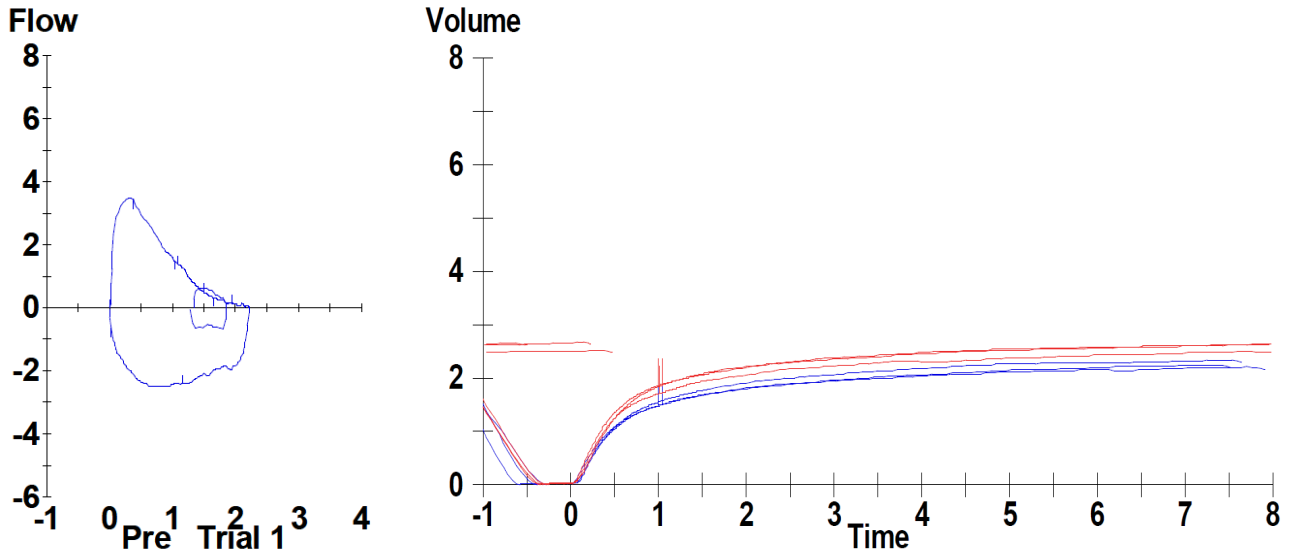

Figure 5. Flow/volume loop and volume/time curve.

Radiographic discussion

Pulmonary underdevelopment has varied radiographic manifestations depending on the affected side and contralateral side compensatory hyperinflation. Chest X-ray shows unilateral opaque hemithorax with volume loss physiology and mediastinal shift toward the ipsilateral direction. Those findings are nonspecific and can be mistaken for lung atelectasis or even pneumonectomy, as highlighted in our second case. CT scan remains the principal investigation for diagnosis, offering a better understanding of thoracic anatomy, bronchial development, hyperinflation, and herniation of the contralateral lung to the affected side and the possible presence of associated cardiovascular anomalies[1,4]. Magnetic resonance imaging (MRI), pulmonary artery angiography, and direct airway visualization by bronchoscopy should be considered in individual cases[4].

The long-term effect of pulmonary underdevelopment on PFT remains poorly studied given disease rarity, as most published reports describe post-lung cancer or chronic obstructive pulmonary disease lobectomy or pneumonectomy effect on PFT[5,6]. Interestingly, both of our patients had evidence of airway obstruction with no bronchodilator response. Hypothetically, in patients with lung underdevelopment, it can be due to compressed airways leading to the affected lung and compensatory hyperinflation of the contralateral lungs seen in our second case.

Limited data suggest thatiè after pediatric pneumonectomy in patients under the age of five pulmonary function tests can be normal due to neoalveolazation[5]. The development of pulmonary hypertension remains a concern as loss of the unilateral pulmonary vascular bed leads to cardiac physiology changes affecting the right ventricle and, ultimately, the tricuspid valve[5].

Clinical course

Both patients were given selective short-acting beta-agonist inhalers to be used as needed for their dyspnea, with a plan to follow closely in the outpatient clinic and annual PFT.

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