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CASE REPORT

The Missing Lung: A 33-Year-Old's Battle with Congenital Agenesis and Cor Pulmonale

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Abstract

Unilateral lung agenesis is a rare congenital disorder, often diagnosed in childhood due to recurrent infections. We present a 33-year-old male with left lung agenesis, manifesting as progressive dyspnea, pedal edema, and severe pulmonary hypertension (PH). Clinical examination revealed absent left lung sounds, mediastinal shift, and signs of right heart failure. Imaging confirmed left lung agenesis, while echocardiography demonstrated severe PH (PASP 110 mmHg). Management included oxygen therapy, diuretics, and antibiotics for superimposed infection. This case underscores the importance of early recognition and multidisciplinary management in congenital lung anomalies to prevent PH and cor pulmonale.

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Case Presentation

A 33-year-old male, with no history of consanguinity or familial congenital disorders, presented with progressive exertional dyspnea (NYHA class III) for seven months and bilateral pedal edema for two months. He reported recurrent chest infections since childhood but had no prior cardiopulmonary evaluation. On admission, he was dyspneic (SpO, 90% on room air), tachycardic (HR 104 bpm), and hypotensive (BP 90/60 mmHg). Physical examination revealed elevated jugular venous pressure (JVP), a loud P, , palpable P, , and left parasternal heave. Respiratory findings included left-sided chest flattening, absent breath sounds, and dull percussion on the left hemithorax, with tracheal and mediastinal shift to the left.

Investigations confirmed the diagnosis. Chest X-ray showed complete left hemithorax opacification with rib crowding and compensatory right lung hyperinflation. CT chest revealed left lung agenesis, mediastinal shift, and right lower lobe ground-glass opacities with bronchiectasis. Fiberoptic bronchoscopy confirmed the absence of the left bronchial tree. ECG demonstrated P pulmonale and right ventricular hypertrophy (RVH) with strain, while echocardiography estimated a pulmonary artery systolic pressure (PASP) of 110 mmHg with preserved left ventricular function (LVEF 62%). Sputum culture grew *Klebsiella pneumoniae*, sensitive to ceftriaxone.

The patient was initiated on long-term oxygen therapy (LTOT), intravenous antibiotics (empirical

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followed by culture-directed), diuretics, and pulmonary vasodilators (Ambrisentan 5 mg/day) for severe PH. After 3 months of follow-up, his symptoms showed modest improvement: SpO, 94% on 2 L/min LTOT, reduced JVP elevation, and mild resolution of bipedal edema. Repeat echocardiography demonstrated a decline in PASP to 87 mmHg, though right heart strain persisted. He was advised to continue current therapy with close cardiopulmonary monitoring.

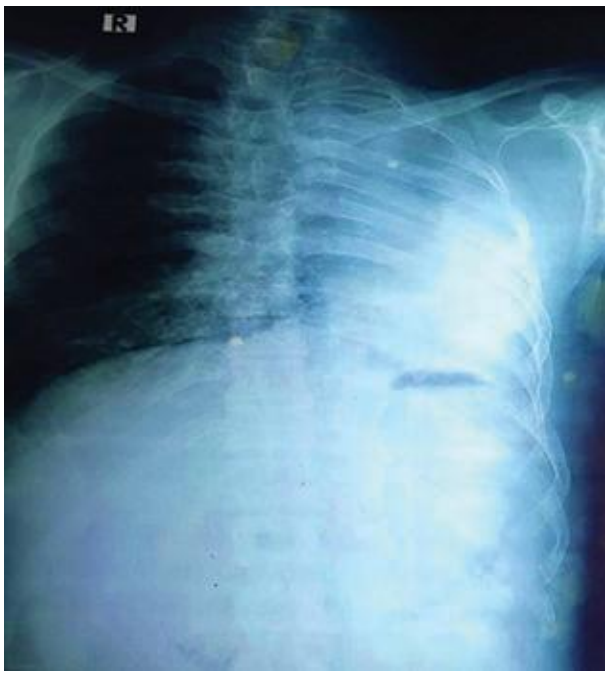
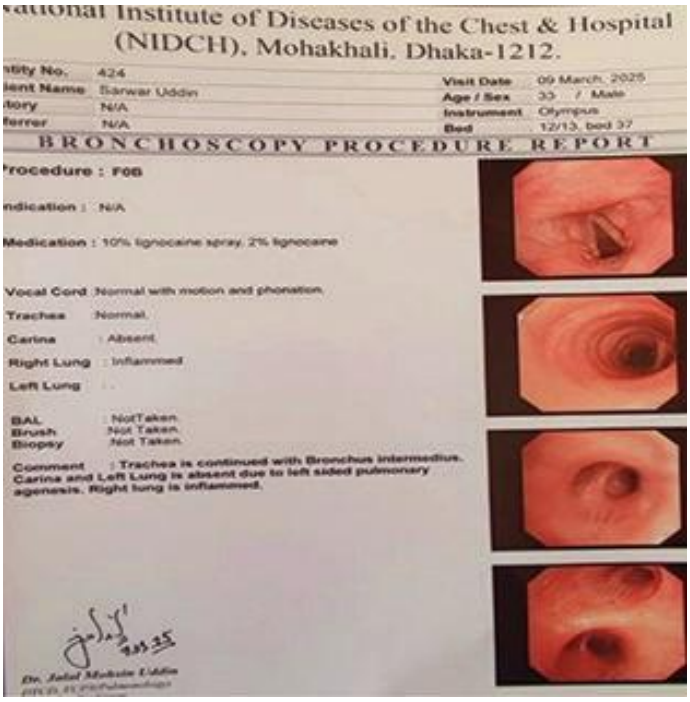


Figure 1: *Chest X-ray demonstrating left hemithorax opacification, rib crowding, and right lung hyperinflation.*

Figure 2: *CT scan revealing left lung agenesis, mediastinal shift, and right lower lobe Ground glass opacity."*


National Institute of Diseases of the Chest & Hospital
(NIDCH), Mohakhali, Dhaka-1212.

Visit Date	09 March, 2025		
Age / Sex	33 / Male		
Instrument	Olympus		
Bed	12/13, bed 37		

BRONCHOSCOPY PROCEDURE REPORT

Procedure : FOB

Indication : N/A

Medication : 10% lignocaine spray, 2% lignocaine

Vocal Cord : Normal with motion and phonation.

Trachea : Normal.

Carina : Absent.

Right Lung : Inflammed

Left Lung :

BAL : NotTaken.
Brush : Not Taken.
Biopsy : Not Taken.

Comment : Trachea is continued with Bronchus intermedius. Carina and Left Lung is absent due to left sided pulmonary agenesis. Right lung is inflammed.

Dr. Jalal Mahsin Uddin

Figure 3: *Bronchoscopic view confirming left bronchial agenesis.*

Discussion

Unilateral lung agenesis is an exceedingly rare congenital anomaly resulting from the complete absence of lung parenchyma, bronchi, and vasculature on one side. Left-sided agenesis, as seen in this case, is slightly more common than right-sided agenesis and is often associated with a better prognosis due to less severe mediastinal shift and cardiac compression.[1] However, the remaining lung undergoes compensatory hyperinflation, increasing susceptibility to recurrent infections, bronchiectasis, and progressive respiratory insufficiency.[2] In our patient, chronic hypoxia due to reduced pulmonary vascular bed and recurrent infections likely contributed to the development of severe pulmonary hypertension (PH), a well-documented complication in such cases.[3]

The pathophysiology of PH in lung agenesis involves multiple mechanisms. The absence of pulmonary vasculature in the affected side increases blood flow to the contralateral lung, leading to endothelial dysfunction, vascular remodeling, and elevated pulmonary vascular resistance.t Chronic hypoxia further exacerbates PH by inducing vasoconstriction and vascular smooth muscle proliferation.u Over time, this results in right ventricular hypertrophy (RVH), right heart failure, and cor-pulmonale, as evidenced by our patient's echocardiographic findings (PASP 110 mmHg) and clinical signs of right-sided congestion (elevated JVP, pedal edema).v

Diagnosis of lung agenesis requires a high index of suspicion, particularly in adults with a history of recurrent respiratory infections and unexplained dyspnea. Imaging plays a crucial role, with chest X-ray revealing mediastinal shift and hemithorax opacification, while CT provides definitive confirmation of absent lung tissue.w Fiberoptic bronchoscopy is useful to rule out bronchial atresia or other obstructive pathologies.x In our case, echocardiography was pivotal in assessing PH severity and right heart function, though right heart catheterization remains the gold standard for PH diagnosis.y

Management of lung agenesis with PH is primarily supportive. Long-term oxygen therapy (LTOT) is essential to alleviate hypoxia and reduce pulmonary vasoconstriction.[1]p Diuretics are used for volume overload, while pulmonary vasodilators (e.g., phosphodiesterase-5 inhibitors, endothelin receptor antagonists) may be considered in selective cases, though evidence in congenital lung anomalies is limited.[11] Infection prevention through vaccinations and prompt antibiotic therapy for respiratory infections is critical to prevent further lung damage. In advanced cases, lung transplantation may be the only definitive treatment, though comorbidities such as severe PH and right heart failure pose significant challenges.[12]

This case highlights the delayed presentation of lung agenesis and underscores the importance of early recognition and multidisciplinary management. Regular follow-up with cardiopulmonary assessment, PH monitoring, and preventive care are vital to improving long-term outcomes in such patients.

Conclusion

Unilateral lung agenesis, though rare, can lead to severe complications such as pulmonary hypertension and cor pulmonale in adulthood. A high clinical suspicion, combined with imaging and hemodynamic assessment, is essential for diagnosis. Multidisciplinary management, including oxygen therapy, diuretics, and infection control, remains the cornerstone of treatment. Early intervention may mitigate disease progression and improve quality of life.

Consent

Verbal consent has been taken from the patient regarding publication of this report.

Conflict of Interest

No conflict of interest was declared by the authors.

References

1. Boyden EA. Developmental anomalies of the lungs. *Am J Surg.* 1955;89(1):79-89.
2. Mardini MK, Nyhan WL. Agenesis of the lung. *Chest.* 1985;87(4):522-527.
3. Galiè N, Humbert M, Vachiery **JL,** et al. 2015 ESC/ERS Guidelines for the diagnosis and treatment of pulmonary hypertension. *Eur Heart J.* 2016;37(1):67-119.
4. Haworth SG. Pulmonary hypertension in childhood. *Eur Respir J.* 1993;6(7):1037-1043.

5. Stenmark KR, Fagan KA, Frid MG. Hypoxia-induced pulmonary vascular remodeling. *Circ Res.* 2006;99(7):675-691.

6. McLaughlin VV, Archer SL, Badesch DB, et al. ACCF/AHA 2009 expert consensus document on pulmonary hypertension. ***J** Am Coll Cardiol.* 2009;53(17):1573-1619.

7. Biyyam DR, Chapman T, Ferguson MR, et al. Congenital lung abnormalities: embryologic features, prenatal diagnosis, and postnatal radiologic-pathologic correlation. *Radiographics.* 2010;30(6):1721-1738.

8. Berrocal T, Madrid C, Novo S, et al. Congenital anomalies of the tracheobronchial tree, lung, and mediastinum: embryology, radiology, and pathology. *Radiographics.* 2004;24(1):e17.

9. Hoeper MM, Bogaard HJ, Condliffe R, et al. Definitions and diagnosis of pulmonary hypertension. ***J** Am Coll Cardiol.* 2013;62(25 Suppl):D42-D50.

10. Weitzenblum E, Chaouat A. Hypoxic pulmonary hypertension in man: what minimum daily duration of hypoxaemia is required? *Eur Respir **J.*** 2001;18(2):251-253.

11. Humbert M, Sitbon O, Chaouat A, et al. Pulmonary arterial hypertension in France. *Am J Respir Crit Care Med.* 2006;173(9):1023-1030.

12. Yusen RD, Edwards LB, Kucheryavaya AY, et al. The Registry of the International Society for Heart and Lung Transplantation: thirty-second official adult lung and heart-lung transplantation report—2015. ***J** Heart Lung Transplant.* 2015;34(10):1264-1277.