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Case Report

Lung Agenesis in an Adult Patient With Congenital Heart Disease

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ABSTRACT

Pulmonary agenesis is a very rare congenital disorder with a chromosome recessive inheritance. Half of the cases with pulmonary agenesis suffer from other congenital disorders such as cardiovascular, skeletal, and gastrointestinal defects. Clinical manifestations vary from respiratory problems to recurrent chest infections, which occur in different stages of life including infancy, childhood, and adolescence. The mortality rate of pulmonary agenesis is about 50% among neonates, especially if it is associated with other disorders like cardiac anomalies. In this paper, we report a case of left-sided lung agenesis in a 32-year-old woman. ***(Iranian Heart Journal 2021; 22(4): 135-139)***

KEYWORDS: Congenital heart defect, Pulmonary agenesis, Respiratory infections

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Pulmonary agenesis is a very rare congenital disorder with a chromosome recessive inheritance. [1] Only 200 cases of this disorder have been reported in the literature. [2] Pulmonary angiogenesis might happen unilaterally or bilaterally, and patients with bilateral agenesis do not survive. [3]

The prevalence of bilateral and unilateral types of pulmonary agenesis in every 1000 live births is 0.5 and 1, respectively. The condition is more common among male babies, and it usually presents itself with left-sided lung involvement in most patients. Moreover, half of the cases suffer from other congenital disorders such as cardiovascular, skeletal, and gastrointestinal defects. The clinical manifestations of this defect vary from respiratory problems to recurrent chest infections, which occur in different stages of life including infancy, childhood, and adolescence. [4] We herein present the clinical manifestations and paraclinical data of a case of left-sided lung agenesis.

Case Report

A 32-year-old woman was referred to our clinic with the general appearance of central cyanosis and clubbing. The patient's history indicated cyanosis at birth, which had progressively exacerbated. Left-sided tracheal deviation, left hemithorax flattening, and left-sided chest movements were evident in the physical examination of the respiratory system. The left chest percussion note was dull, with no breath audible over the left hemithorax; additionally, there was a left-deviated point

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of maximal impulse. However, no abnormality was found in the right hemithorax examination. The oxygen saturation level was 71% and 84% for the upper and lower extremities, respectively.

A homogenous opaqueness was discovered in the chest X-ray at the middle and lower areas of the right lung, which shifted the heart and the trachea to the left (Fig. 1). A chest computed tomography indicated that the heart was placed in the left hemithorax with no left main pulmonary artery visible, which might have been agentic. In the chest computed tomography, a structure resembling bronchocele was evident in the posteromedial region of the hemithorax, which was an extension of the left main bronchus. Another small adjunct bronchus was observed on the left-hand side (Fig. 2). Transthoracic echocardiography showed severe right ventricular enlargement, dysfunction, and hypertrophy, as well as a large perimembranous ventricular septal defect, a large patent ductus arteriosus, and severe tricuspid regurgitation (the tricuspid regurgitation gradient ≈120 mm Hg) (Fig. 3). For the exploration of further associated congenital anomalies, abdominal sonography was performed, and it indicated no anomalies. We decided to manage her condition medically by using diuretics and bosentan and asked her to have regular follow-ups. We discussed the patient's condition and its probable complications in detail with herself and her companions. We also prescribed her Influenza and pneumococcal vaccines.

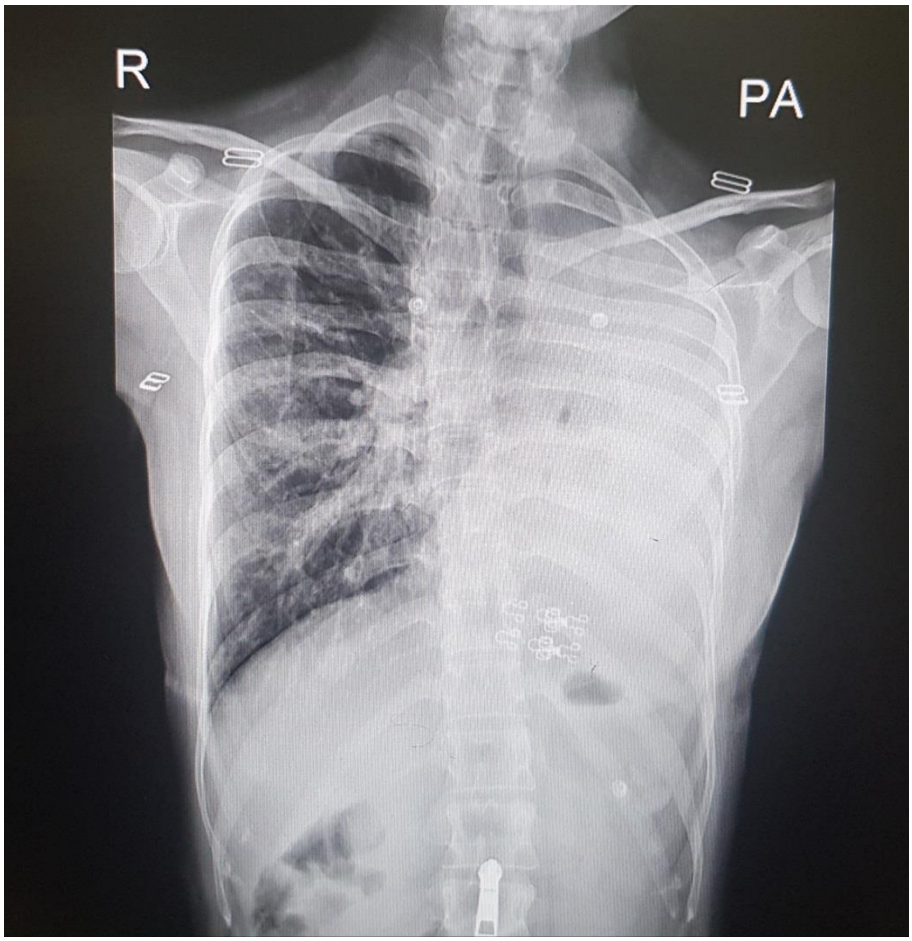

Figure 1. The chest X-ray in the posteroanterior view shows a white lung and the shifting of the heart to the left.

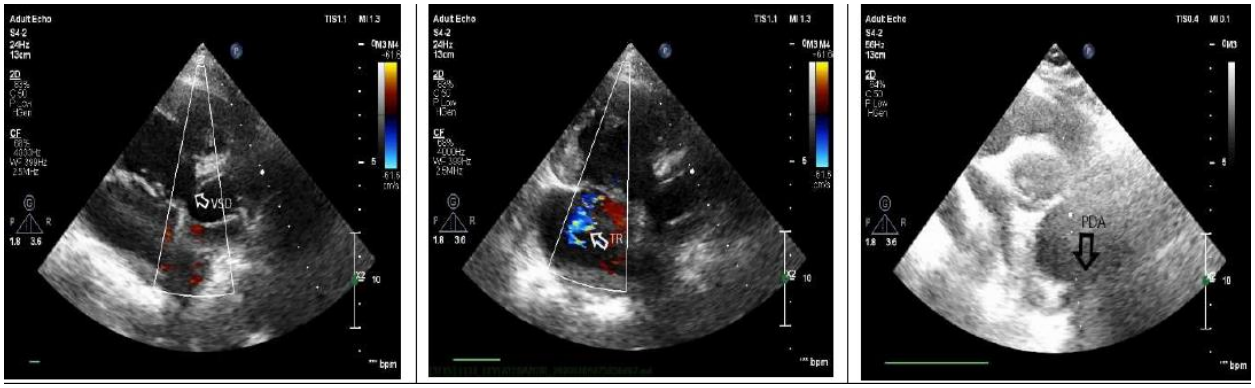

Figure 2. Transthoracic echocardiography shows a ventricular septal defect, a patent ductus arteriosus, and severe tricuspid regurgitation.

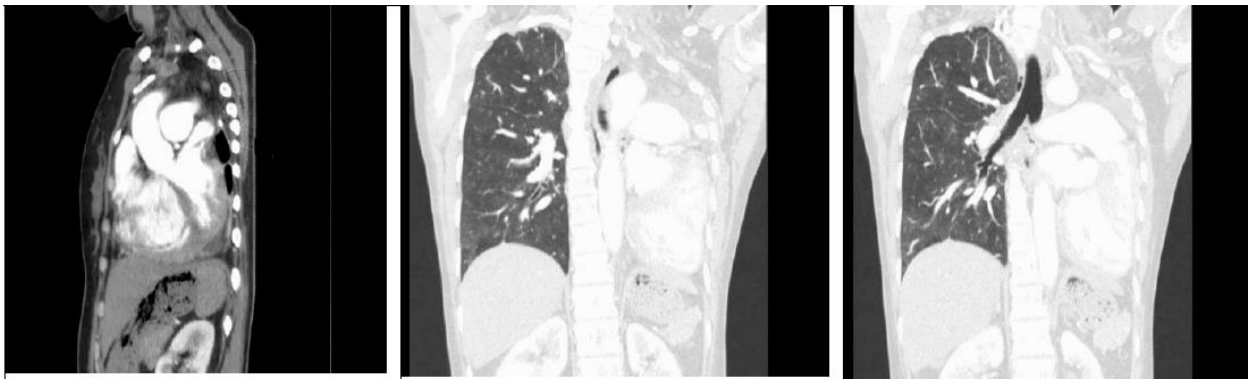

Figure 3. Cardiac computed tomography angiography shows the shifting of the heart to the left and the absence of the right bronchus.

DISCUSSION

Lung agenesis is a very rare congenital developmental problem in which 1 or both of the lungs do not evolve. [1] Although the first case of this anomaly was reported in 1673 by Depozze, who noticed unilateral pulmonary agenesis in a female autopsy, it was not until 1923 when Muhammed reported left-sided pulmonary agenesis in a forensic medicine autopsy. [3]

Most patients with pulmonary agenesis experience congenital anomalies such as genitourinary, cardiovascular, and skeletal defects. [5] We noticed a cardiac anomaly in the case presented herein. The mortality rate of similar cases with unilateral pulmonary agenesis is 50%, which could be higher if the agenesis is for the right lung or associated with other disorders like cardiac anomalies. This is justified by tracheal pulmonary agenesis as a result of a greater mediastinal shift. [6]

The first classification of pulmonary agenesis was presented by Schneider and Schawatbe, and then it was modified by Boyden into 3 variations according to the developmental stage of the lung bud [7]:

Type I is agenesis associated with the complete absence of lung parenchyma, bronchus, and blood supply to the affected lung side. Type II is aplasia in which the lung parenchyma has not been developed completely and some undeveloped bronchus is evident. Type III is hypoplasia in which variable amounts of the lung parenchyma, bronchial tree, and vasculature are present. Based on this classification, our patient was

classified as type II because of the presence of a rudimentary bronchus on the affected side.

Although the exact underlying cause of the disease is not known yet, several hypotheses including environmental agents, shortages of vitamin A, consanguinity, medicines, and intrauterine infections have been raised. The disease is commonly diagnosed in childhood because of frequent pulmonary infections or during other evaluations. Nonetheless, cases have been reported as early as adolescence, and the oldest person ever reported with unilateral agenesis was 72 years old. [4] Currently, several methods including computed tomography, pulmonary angiography, and bronchography exist for the diagnosis and evaluation of lung agenesis; however, contrast-enhanced computed tomography forms the standard investigation for the diagnosis, and the other methods are rarely required. [8] Bronchoscopy for the direct visualization of the bronchus anatomy may be done, but it is not necessary. [9] Lifelong prognoses are influenced by the agenesis sidedness, the status of the residual lung tissue, and other associated anomalies, seen in up to 50% of cases. [7] Congenital diseases such as patent ductus arteriosus, ventricular septal defect, atrial septal defect, narrowed trachea, and tracheoesophageal fistula might be associated. [10] For the case presented here, we drew upon echocardiography and abdominal ultrasonography. [11]

Treatment is based on associated anomalies and patient conditions. Asymptomatic patients require no regular medical treatment, but chest infections must be treated aggressively. [11] Some types of surgical corrections have been attempted in specific patients, including mediastinal deviation and tracheal abnormality, which may lead to respiratory distress. Additionally, such other strategies as inflatable prosthesis placement, aortopexy, and diaphragmatic translocation have been reported. [12]

CONCLUSIONS

Lung agenesis is a very rare congenital disease of the lung that is common on the left side and in males. However, in this paper, we reported left-sided lung agenesis in a female patient in association with different cardiac defects, which makes it even more uncommon. Early and correct recognition of the disease is crucial to diminish fibrosis in patients with unilateral lung agenesis, which can occur as a result of recurrent chest infections. The base of treatment consists of observation and aggressive and early management of chest infections and surgical correction in particular conditions.

Declarations

Ethical approval was waived given the retrospective observational design of the study. We obtained the patient's consent to anonymously publish her case.

-**Availability of Data and Material**: Not applicable

-**Conflict of Interest:** None declared

-**Funding**: None

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