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Familial Lung Agenesis

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Abstract

Pulmonary agenesis (PA) is a very rare developmental anomaly of the lung. PA involving different members of a family is exceptional. Here, we report two cases of familial left pulmonary agenesis occurred in mother and daughter. Neither of them has other known malformations.

Keywords: Pulmonary agenesis; Lung; Congenital disease; Familial disease

Introduction

Pulmonary agenesis (PA) is a very rare congenital anomaly [1-5] of lung development defined as a complete absence of lung tissues, bronchi, and pulmonary vessels [3,6,7]. It may be uni- or bilateral [1,8] and may be associated with anomalies in other systems. Bilateral PA is not viable. The right-sided form carries the poorest prognosis due to the severity of co-existent congenital defects, mainly cardiovascular malformations [1-5,7,9]. Unilateral PA may be present to varying degrees of severity. Diagnosis in adulthood is rare [2-6,9].

Case Presentation

A 35-year-old woman with no medical history of interest was incidentally diagnosed with left pulmonary aplasia or agenesis by chest-X-ray at the age of 1 year. Chest CT scan performed when she was an adult showed absence of the left main pulmonary artery, the left pulmonary veins and ipsilateral bronchus remnant. Consequently, she was finally diagnosed with left pulmonary agenesis (PA) **(Figures 1, 2 and 3).**

Her daughter was diagnosed with left pulmonary aplasia or agenesis by prenatal ultrasound. Chest CT performed later showed similar findings as those found in her mother's CT and she was finally diagnosed with left PA as well **(Figures 4 and 5).** Neither of them has other known malformations or pathology in the right lung.

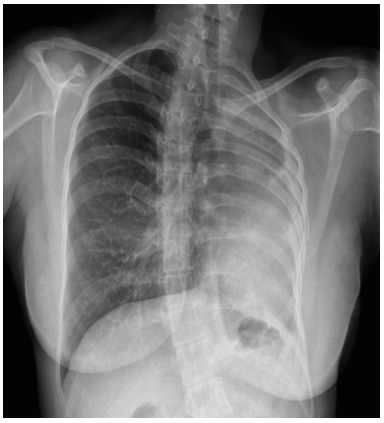

Figure 1: PA Chest X-Ray of the mother made at age of 35 shows a diffuse opacity in the left hemithorax, mediastinum structures deviated to the left side with compensatory hyperinflation of the right lung and decreased space between the left ribs.

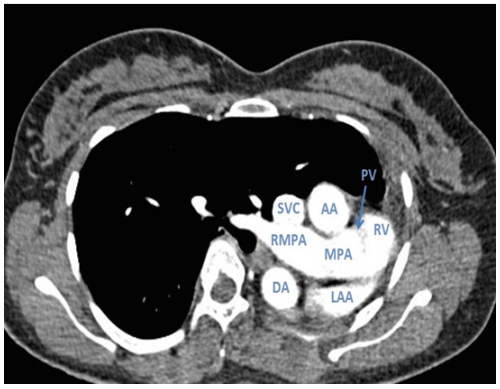

Figure 2: Chest CT scan at the level of the distal trachea confirms the absence of lung tissues at the left side and mediastinal ipsilateral shift. There is no left bronchus remnant. Also, there is an ipsilateral absence of the pulmonary artery and veins. [SVC: Superior Vena Cava; AA: Ascending Aorta; DA: Descending Aorta; MPA: Main Pulmonary Artery, RPA: Right Pulmonary Artery; LAA: Left Pulmonary Artery, PV: Pulmonary Valve; RV: Right Ventricle].

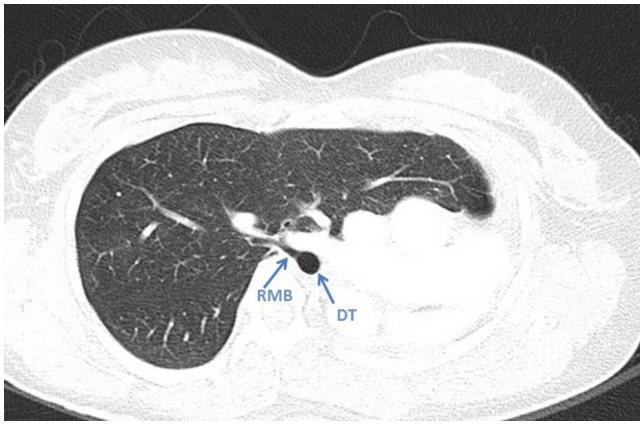

Figure 3: Chest CT scan at the same level, lung windows settings, shows hyperinflated right lung and herniation across the midline. [RMB: Right Main Bronchus; DT: Distal Trachea].

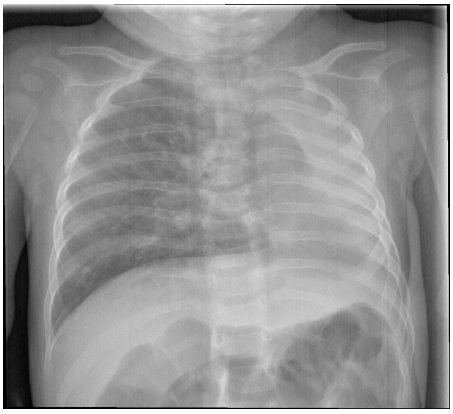

Figure 4: PA Chest X- Ray of the daughter made at age of 5 months shows similar findings as those of her mother with a diffuse opacity in the left hemithorax, leftward deviation of mediastinal structures and compensatory right lung hyperinflation.

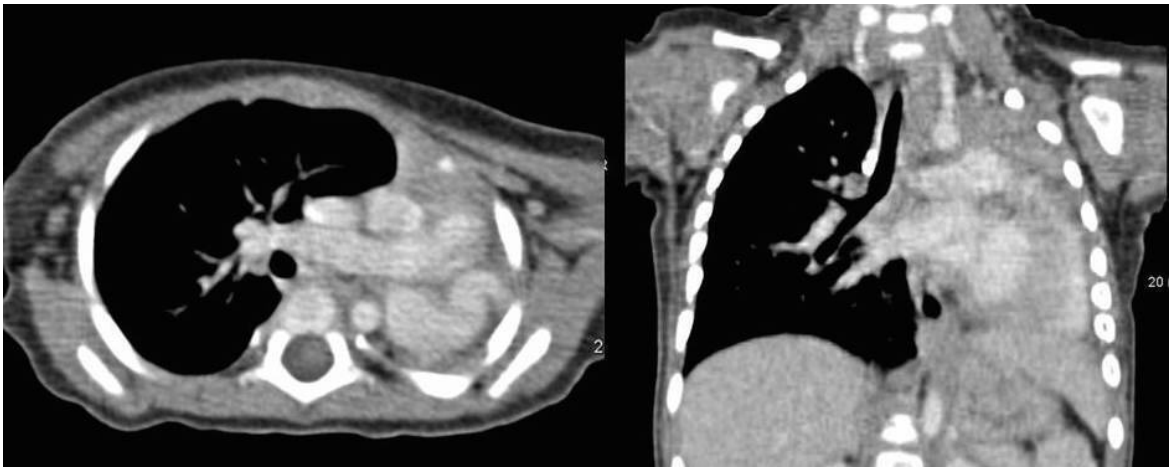

Figure 5: Chest CT of the daughter. Axial and coronal reconstructions show similar findings as those of her mother's scan: absence of left lung tissues and mediastinal ipsilateral shift. Also, there is an ipsilateral absence of the pulmonary artery and veins. There is no left bronchus remnant.

Discussion

PA is a rare congenital malformation [1-5] of lung development defined as a complete absence of lung tissues, bronchi, and pulmonary vessels [3,6,7]. It may be uni- or bilateral [2,3] being the latest not viable [1,8]. Familial occurrence is exceptional. In unilateral cases, left-side agenesis is more frequent (70%) [1-3,5,9,10]. The right-sided form carries the poorest prognosis due to the severity of co-existent anomalies, mainly cardiovascular [1-5,7,9].

Up to 50% of patients have associated congenital anomalies [5,6] mainly affecting skeletal, genitourinary, gastrointestinal and cardiovascular systems [1,4,5,7]. There are no differences between males and females [3].

Originally Schneider classified agenesis into three groups which was later on modified by Boyden: Type I (Agenesis): Complete absence of lung and bronchus and absence of blood vessels to the affected side; type II (Aplasia): Rudimentary bronchus with complete absence of lung tissue and type III (Hypoplasia): Presence of variable amounts of lung parenchyma, bronchial tree and supporting vasculature [1-5,7,10,11].

The respiratory system develops from the ventral wall of the foregut during gestation at 3-4 weeks (embryonic stage) [10,11] and completes a significant portion of its development immediately before, and after, birth [1,7,11].

Pulmonary arteries develop from the cono-truncal septation of the primitive heart and the sixth aortic arches and accompany the bronchial tree as it branches [7]. Agenesis of a

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pulmonary artery (right or left) affects growth of the ipsilateral lung parenchyma **(Figure 6).**

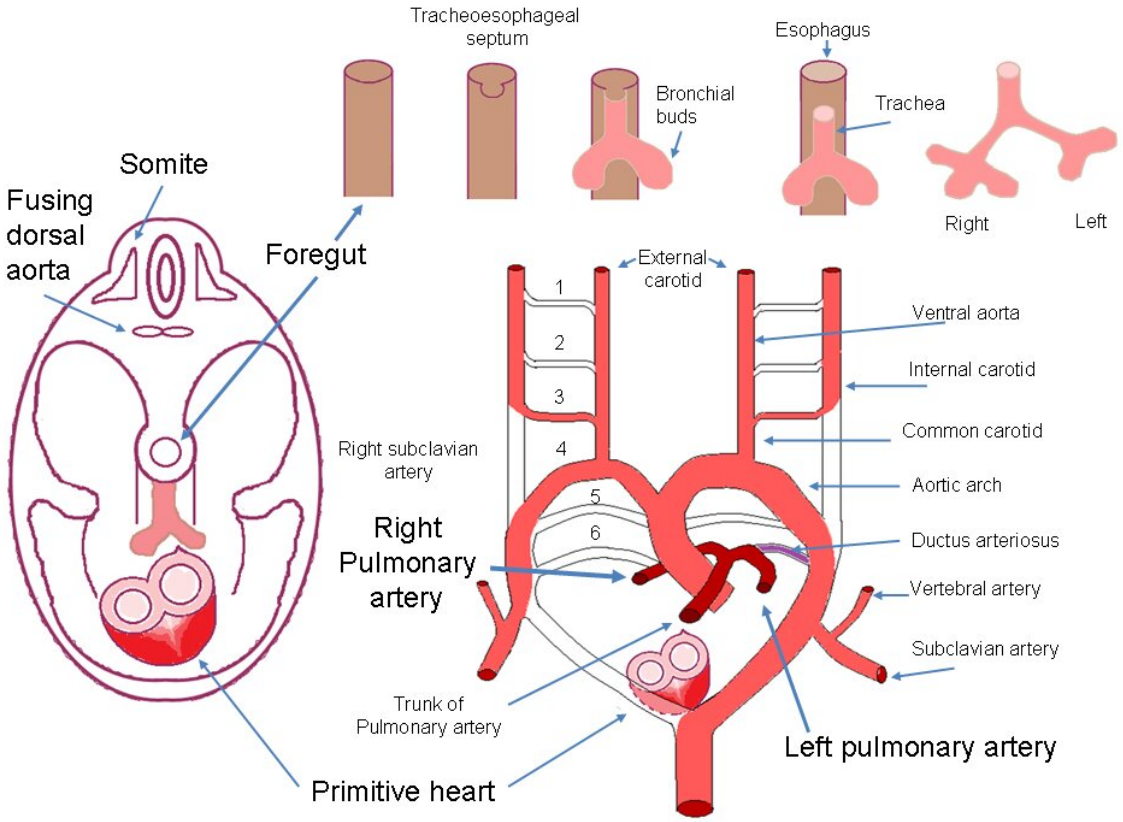

Figure 6: Diagrams of the development of the lower respiratory system and pulmonary arteries.

PA occurs during the embryogenic period (4 weeks of gestation), when the primitive lung is being formed. Although etiology is still unknown it is supposed to be multifactorial [1-3] and there are some hypotheses: abnormal blood flow in the dorsal aortic arch during the 4th week of gestation [11], vitamin A or folic acid deficiency, use of salicylates, intrauterine infections, drugs, parental consanguinity, genetic anomalies and environmental factors [1-4,6,8-10].

In our cases a genetic influence seems to be the most probably origin of the pulmonary agenesis. Familial anomalies of lung development have been more frequently published in lung hypoplasia [12-15] however, a case of lung agenesis was reported in identical twins [16] and another publication showed the genetic study of a case of PA with a chromosome abnormality [17].

PA is usually diagnosed in neonatal period in those patients with respiratory problems such as cyanosis, dyspnea, feeding difficulties [9] or recurrent infection [1-3,8]. Although less frequently, in asymptomatic cases, it can be diagnosed in adulthood [2-6,9].

Asymmetrical chest wall movements and decreased respiratory sound in pulmonary auscultation are seen on physical examination [1].

Diagnosis can be suspected in chest X-ray as a diffuse opacity in the affected side [1,4] with compensatory hyperinflation of the contralateral lung, mediastinal structures deviated to the affected side [3,6,8,11], decreased space between ribs and hemidiaphragm elevation [6,7].

Contrast enhanced CT is currently the standard imaging technique for diagnosis of lung agenesis. Pulmonary angiography is rarely required. Bronchoscopy for visualization of rudimentary or absent bronchus may be done but is not essential [2,11].

In prenatal cases PA can be as a hyperechoic hemithorax in prenatal ultrasound but fetal MRI is mandatory for confirmation [1]. Pulmonary aplasia, total atelectasia, diaphragmatic hernia, fibrothorax, total pneumectomy, and pleural effusion should be considered in the differential diagnosis [5,6].

No treatment is required in asymptomatic cases [2,4,10,18]. Surgery is needed depending on associated anomalies [1]. Survival prognosis depends on associated anomalies, affected side and functional capacity of the remaining lung [1-3,5,8,18].

Conclusion

Pulmonary agenesis is a rare congenital disease usually diagnosed in the childhood. During adulthood may mimic lung collapse and other underlying pathologies should be discarded. Its clinical presentation varies depending on other associated anomalies. Symptomatic treatment is necessary with concomitant chest infections. Familial PA is an extremely rare condition.

Authors Contributions

All authors have contributed equally to the design and preparation of this manuscript and as reviewers of its final content.

Disclosures

We declare that none of the authors have any conflicts of interest.

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None.

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