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CASE REPORT

Unilateral pulmonary artery agenesis associated with pulmonary hypoplasia and pulmonary sequestration: a report of two pediatric cases

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Abstract

Isolated agenesis of the right or left pulmonary artery is a rare congenital anomaly resulting from malformation of the sixth aortic arch on the affected side during embryogenesis. Two pediatric cases of right pulmonary artery agenesis are reported: the first corresponds to an isolated case with associated pulmonary hypoplasia; the second also presents with pulmonary hypoplasia and is associated with pulmonary sequestration. Both cases were evaluated through imaging studies and followed up on an outpatient basis. Clinical presentation may range from asymptomatic cases to severe manifestations, such as pulmonary hypertension, hemoptysis, and congestive heart failure. Early suspicion and diagnosis are essential for appropriate management. This case report aims to contribute to the recognition of this rare condition and its possible anatomical associations.

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INTRODUCTION

The pulmonary artery develops from the sixth aortic arch. The proximal portion of this arch gives rise to the proximal segments of the right and left pulmonary arteries. On the right side, the distal portion loses its connection with the aortic arch, while on the left it maintains its connection as the ductus arteriosus[1,2].

Isolated agenesis of the pulmonary arteries, whether right or left, is a rare congenital abnormality resulting from a malformation of the sixth aortic arch during embryogenesis. During the embryonic and pseudoglandular periods, arterial development occurs parallel to the airways. As the fetus develops, the vessels increase in diameter and length, and in the postnatal phase, there is a marked branching, which corresponds to the proliferative alveolar development from early childhood until approximately eight years of age[3-6].

The prevalence of this condition is estimated at approximately one case for every 200,000 individuals. Some patients remain asymptomatic, while others may present with clinical manifestations such as hemoptysis, severe pulmonary hypertension, congestive heart failure, and cyanosis[1-4].

When agenesis occurs in the left pulmonary artery, it tends to be symptomatic and is often associated with other cardiac malformations. On the other hand, agenesis of the right pulmonary artery may be asymptomatic and arise as an incidental finding on imaging tests.

Two pediatric clinical cases of patients with unilateral agenesis of the right pulmonary arteries are reported, one isolated case and the other associated with ipsilateral pulmonary sequestration[3,5.]

CLINICAL CASE 1

A male patient, 4 years old at the time of his first consultation at the pediatric pulmonology outpatient clinic. He was born by cesarean section at 40 weeks of gestational age. The pregnancy was uneventful.

According to the report of the guardian, he was admitted to the neonatal intensive care unit for the diagnosis of pneumonia, requiring invasive mechanical ventilation. During hospitalization, there were two episodes of cardiorespiratory arrest and the need for phototherapy due to neonatal jaundice.

After hospital discharge, while still in preschool age, dyspnea associated with cyanosis and dry cough during great exertion were observed, with improvement after the use of bronchodilators. He has a history of four previous hospitalizations due to suspected recurrent pneumonia.

At 4 years of age, he had an episode of syncope, which prompted a new hospitalization. At the time, the chest X-ray showed a reduction in lung volume on the right, with ipsilateral mediastinal shift. Transthoracic echocardiogram revealed minimal tricuspid valve prolapse, with estimated pulmonary artery systolic pressure of 12 mmHg.

Carotid and vertebral Doppler ultrasonography showed increased systolic velocity in the common carotid arteries, but without significant structural changes. Serum immunoglobulins and sweat test results were within normal limits.

Bronchoscopy revealed hyperemia and diffuse edema of the mucosa with abundant mucoid secretion on the right, changes suggestive of active chronic bronchitis. Chest tomography showed volumetric reduction of the right lung, in addition to peripheral septal thickening in the right lower lobe, and retracted mediastinal structures on the right[7-10].

Chest angiotomography (Figure 1) revealed the following main findings: absence of the right branch of the pulmonary artery associated with moderate hypoplasia of the right lung with marked hypoperfusion; retraction of the mediastinal structures to the right; marked hypertrophy of the bronchial arteries; reduction in the caliber of the superior pulmonary veins; smooth thickening of the interlobular septa; peripheral pseudocysts in the right lung; and pulmonary sequestration at the base of the right lung.

The patient is currently 15 years old and undergoes regular follow-up in the pediatric pulmonology department, with optimized treatment for asthma using inhaled corticosteroids.

CLINICAL CASE 2

A male patient, 4 years old at the first consultation. Complete prenatal care, without complications. Obstetric morphological ultrasound without alterations

Patient pathological history includes full-term birth (38 weeks and 2 days, birth weight of 3,020 g), by cesarean section due to cephalopelvic disproportion. He presented good vitality at birth, without need for oxygen therapy.

At 24 hours of life, he underwent a heart test with altered results, and underwent an echocardiogram that showed patent foramen ovale (PFO) and patent ductus arteriosus (PDA). The test was repeated in the neonatal period, with a normal report.

In early childhood, he presented recurrent episodes of upper respiratory tract infections, all treated on an outpatient basis. There is a report of difficulty in gaining weight during the infancy phase, and a possible allergy to cow's milk protein (CMPA) was investigated. He had only one previous hospitalization, due to acute gastroenterocolitis.

During a routine consultation, a new echocardiogram was requested, which showed cardiac dextroposition. The chest X-ray showed mediastinal shift to the right, reduced right lung volume and increased anteroposterior diameter of the chest. Given these findings, the patient was referred for evaluation by a pediatric pulmonologist.

On inspection, the patient presented a slightly retracted chest to the right, mild dyspnea, and lung auscultation without adventitious sounds. A new chest X-ray confirmed reduced lung volume to the right, with ipsilateral mediastinal

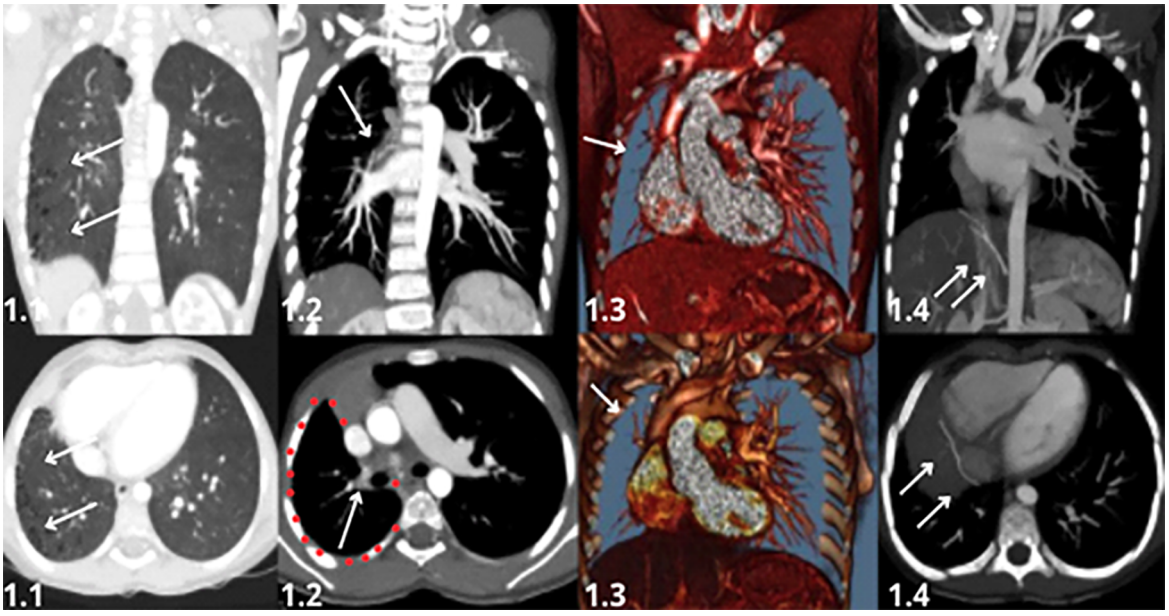


Figure 1. Chest angiotomography images with reconstruction with lung and mediastinal window, in the coronal, axial, *3D volume rendering, and Maximum Intensity Projection (MIP) planes.*
legend:
1.1. Peripheral pulmonary pseudocysts (arrows).
1.2. Hypertrophy of the bronchial arteries (arrows) and right pulmonary hypoplasia (dotted lines).
1.3. Absence of the right branch of the pulmonary artery (arrows).
1.4. Right pulmonary sequestration (arrows).

shift. Additional tests, including abdominal and urinary tract ultrasound, sweat test, complete blood count and biochemistry, showed no changes.

To clarify the diagnosis, a chest angiotomography (Figure 2) was requested, which showed: absence of the right branch of the pulmonary artery associated with hypoplasia and hypoperfusion of the right lung; retraction of the mediastinal structures to the right; collateral circulation by bronchial arteries; and reduction in the caliber of the pulmonary veins.

A more recent transthoracic echocardiogram showed mesocardia and mesoapex; agenesis of the right pulmonary artery; qualitatively normal biventricular dimension and systolic function; and slight tricuspid regurgitation, allowing the estimation of pulmonary artery systolic pressure at approximately 25 mmHg.

The preschooler is undergoing regular outpatient follow-up in the pediatric pulmonology service. Currently, he has shown good weight gain and remains asymptomatic.

DISCUSSION

The pulmonary artery develops from the sixth aortic arch. The proximal portion of the arch becomes the proximal segment of the right and left pulmonary arteries. On the right side, the distal portion loses its connection with the arch, while on the left it maintains its connection as the ductus arteriosus (figure 3).

During the embryonic and pseudoglandular periods, arterial development occurs in parallel with that of the airways. As the fetus matures, the vessels increase in diameter and length, and in the postnatal phase there is a marked increase in branching, which corresponds to the proliferative alveolar development of early infancy until approximately eight years of age. Unilateral pulmonary artery agenesis is a rare congenital anomaly that results from a malformation of the sixth aortic arch on the affected side during embryogenesis[3-6].

Pulmonary agenesis may occur as an isolated disorder or be associated with other congenital cardiovascular malformations, such as tetralogy of Fallot, atrial septal defect, coarctation of the aorta, right aortic arch, truncus arteriosus and pulmonary atresia[5], and may also be related to the presence of aortopulmonary collateral circulation. Collateral arteries of the affected lung usually arise from bronchial, intercostal, subdiaphragmatic, subclavian and coronary arteries[5]. Its estimated prevalence is one in every 200,000 individuals, and there does not appear to be a sex predilection[1-3,6-10].

The diagnosis is usually established in adolescence (median age of 14 years), but the affected patient may remain asymptomatic and receive a diagnosis later in adulthood, usually after an abnormal chest radiograph. The patients in this case report were diagnosed in the pediatric age group[1-3].

Two thirds of the patients described in the literature show alterations on the right side. The two patients presented demonstrated agenesis of the right pulmonary artery. Patients with this pathology have a normal pulmonary trunk and unilateral absence of a pulmonary artery branch[3].

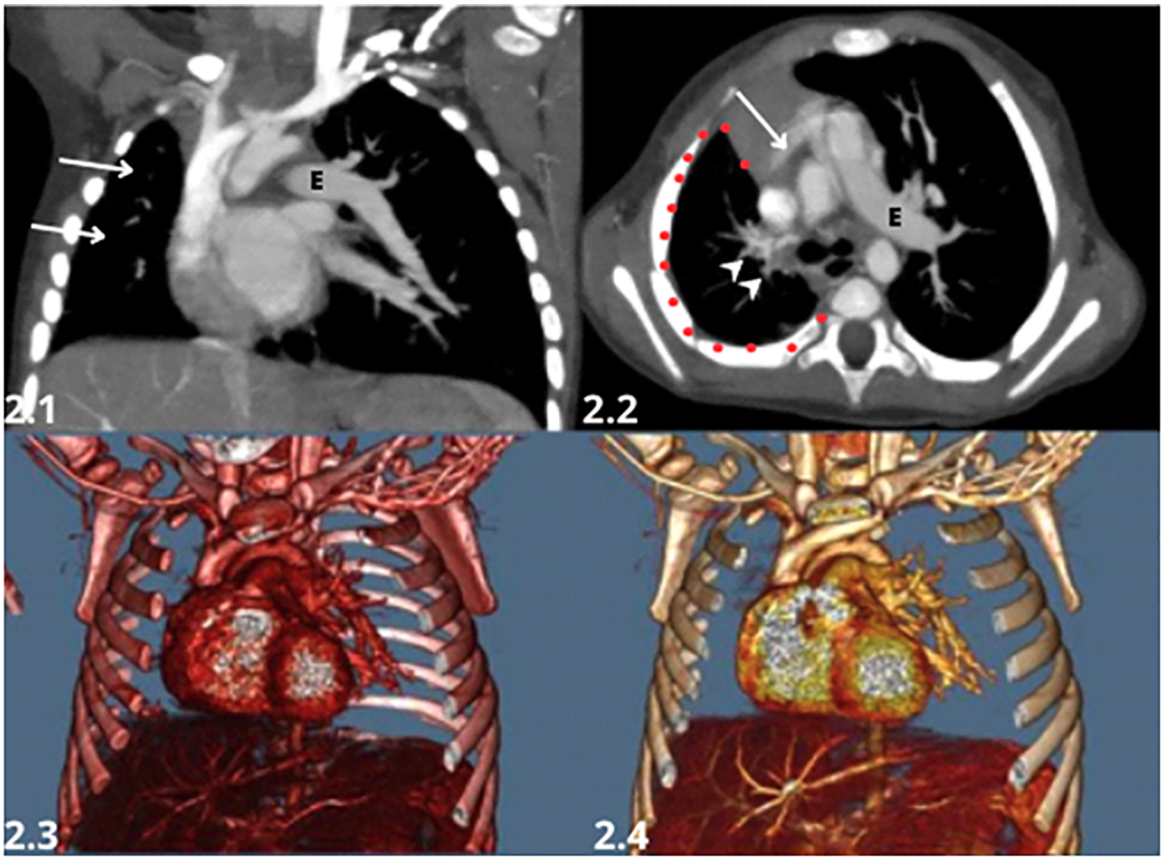


Figure 2. Chest angiotomography images with reconstruction with mediastinal window, in the axial, coronal and 3D volume rendering planes. Legend:
2.1 and 2.2 - Absence of the right pulmonary artery (arrows) and ipsilateral pulmonary hypoplasia (dotted), with collateral circulation by bronchial arteries (arrowheads). Left pulmonary artery (E).
2.3 and 2.4 - Hypodevelopment of the right hemithorax, with retraction of the mediastinal elements and deviation of the cardiac area.

The intrapulmonary vasculature and the distal portion of the pulmonary trunk may develop normally and receive vascularization via the bronchial vessels, resulting in a small, hypovascular lung on the affected side. The condition may be associated with other congenital cardiac anomalies, which was not observed in either of the two patients in this report.

Symptoms may include recurrent lung infections, dyspnea on exertion, chest pain, and hemoptysis[1,3-8]. The patient in Clinical Case 1 presented recurrent pneumonia since the neonatal period. Similarly, the patient in Clinical Case 2 presented recurrent upper respiratory tract infections and difficulty in gaining weight, in accordance with the findings described in the literature.

Pulmonary hypertension or pulmonary hemorrhage is observed in up to a quarter of patients and determines a worse prognosis, outcomes not observed in the two patients in the report[3].

The physical examination is nonspecific, and there may be a reduction in vesicular murmur in the affected hemithorax[1].

The chest radiograph typically shows volume asymmetry and changes in the transparency of the hemithorax, with reduced volume and hypertransparency on the affected side.

Echocardiography may reveal congenital cardiovascular defects or pulmonary hypertension. Chest angiotomography is an essential examination for characterizing vascular anatomy in unilateral pulmonary artery agenesis.

In Clinical Case 1, the echocardiogram revealed tricuspid valve prolapse, without clinical repercussions. The angiotomography demonstrated absence of the right branch of the pulmonary artery, associated with pulmonary hypoplasia and anatomical alterations in the pulmonary arteries and veins, findings frequently described in the literature in cases of unilateral pulmonary artery agenesis. However, the presence of pulmonary sequestration was also observed, a rare condition in this context, configuring an unusual finding in relation to what is usually reported[1,3-8].

In Clinical Case 2, the patient presented anatomical alterations in the neonatal period, with partial resolution over time. In the last echocardiogram, mesocardia, mesoapex and agenesis of the right pulmonary artery were observed. Angiotomography confirmed the absence of the right pulmonary artery, in addition to alterations in the ipsilateral pulmonary vasculature. These findings are consistent with data described in the literature, which report similar anatomical

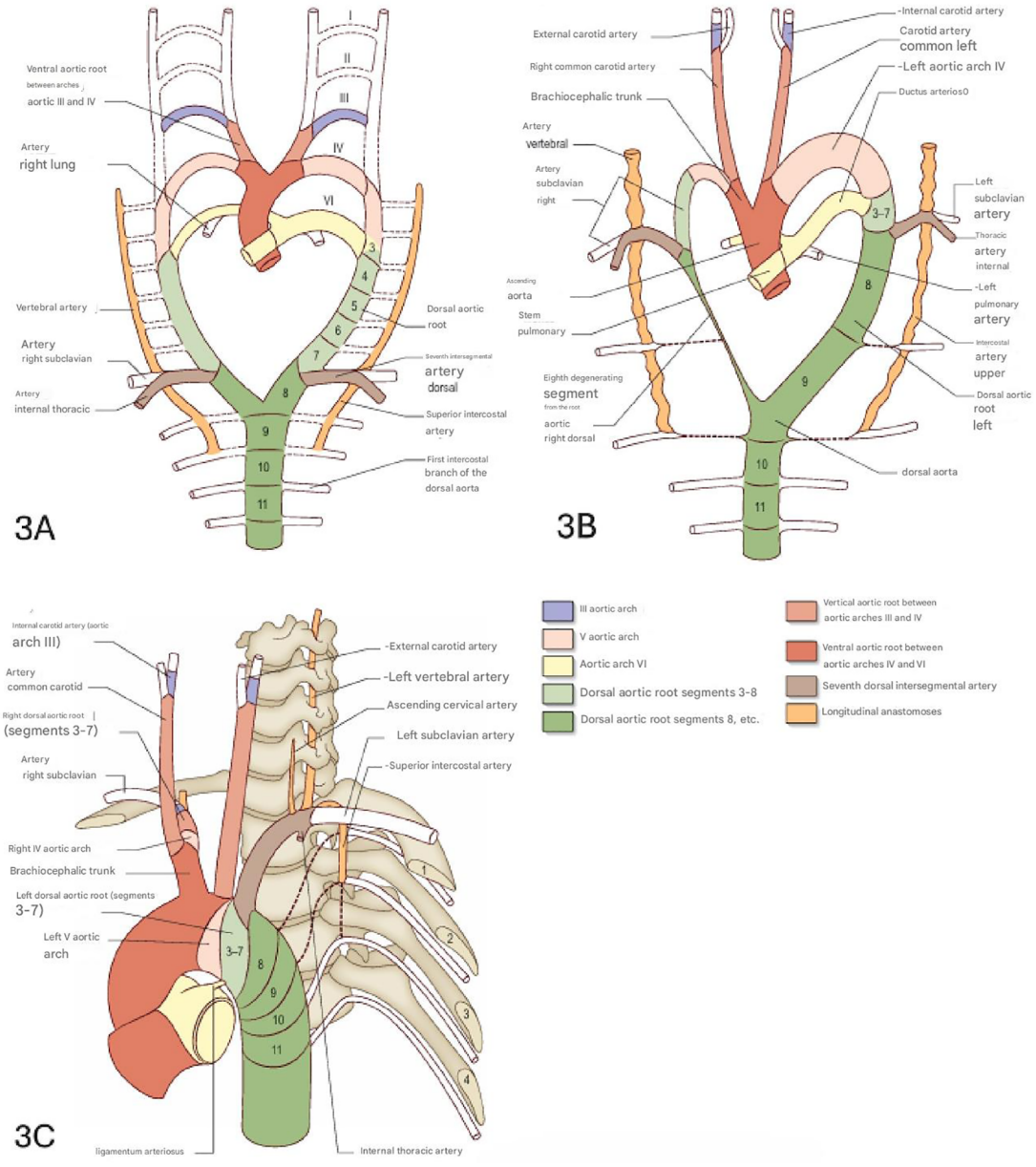


Figure 3. Arteries of the aortic arch and their derivatives (Reproduced from Gray's Anatomy 40th edition)[11]
Legend:
3A. Early development of the aortic arch, ascending aorta, bilateral aortic arches (I through VI), and bilateral dorsal aortic root and dorsal aorta.
3B. Later formation of the aortic arch: The I aortic arch regresses to form the maxillary artery. The II aortic arch regresses to form the stapedial artery. The III bilateral aortic arch becomes the internal carotid arteries, and the ventral aortic root between the III aortic arch and the IV aortic arch becomes the common carotid arteries. The IV right aortic arch and the proximal right dorsal aortic root become the right subclavian artery, and the right distal dorsal aortic root regresses. The IV left aortic arch and the proximal left dorsal aortic root form the transverse aortic arch between the left common carotid artery and the left subclavian artery, and the left distal dorsal aortic root forms the descending aorta. The VI proximal aortic arch forms the main and branch pulmonary arteries. The VI left distal aortic arch forms the conduit arteriosus, and the VI right distal aortic arch regresses.
3C. Final anatomy of the aortic arch.

variations in patients with unilateral agenesis of the pulmonary artery[1,3-8].

Bronchoscopy findings may include signs of chronic bronchitis, especially in patients with recurrent pulmonary infections, as well as vascular changes that may cause hemoptysis. Cases with completely normal bronchoscopy have also been described. The mortality rate reported in the literature is approximately 7%[1-3,9].

Pulmonary hemorrhage and pulmonary hypertension have a negative impact on long-term survival. Management of this condition may include surgical and pharmacological treatment. In the context of surgical treatment, revascularization and pneumonectomy may be considered in cases of pulmonary infections, recurrent hemoptysis, and pulmonary hypertension. Drug treatment for pulmonary hypertension is recommended for patients who cannot undergo surgical revascularization or in cases that do not show improvement after surgery[3-7]. Available options include endothelin receptor antagonists (e.g., bosentan), parenteral prostacyclins, vasodilators (e.g., sildenafil), and calcium channel blockers, which have variable responses[5].

In conclusion, congenital agenesis of the pulmonary arteries may be asymptomatic until adulthood or may be suspected by the presence of persistent imaging in the same location, suggesting recurrent pneumonia, and is a major diagnostic challenge due to the variations in clinical presentations. Its detection and characterization by echocardiogram or chest radiography are complex, therefore, chest CT angiography is an essential examination in the evaluation of the vascular anatomy of this pathology. It is important for the general pediatrician and the subspecialist (pulmonologist or pediatric cardiologist) to be aware of this spectrum of anomalies, whose incidental diagnosis is not uncommon[2].

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