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CASE REPORT

A Rare Case of a Large Aortopulmonary Window with Anomalous Right Pulmonary Artery From Ascending Aorta in a Young Adult

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Abstract

Background: The aortopulmonary window, a rare congenital condition, is an abnormal communication between the ascending aorta and the major pulmonary artery. This anomaly often necessitates early surgical intervention because its prognosis is unfavorable. Cases of adult survival without treatment are exceedingly rare. This condition is typically fatal in childhood if left untreated. In rare instances, it may manifest in adults, presenting symptoms similar to those of pulmonary hypertension. These patients exhibit clinical presentations that are indistinguishable from those of more common conditions characterized by a left-to-right shunt. Through careful transthoracic echocardiography, it is possible to identify the defect in the aortopulmonary septum.
Results: We present a unique case of a 23-year-old adult diagnosed with a large, unrepaired aortopulmonary window, illustrating the challenges in diagnosing and managing such rare adult presentations.

Keywords: Aortopulmonary window, Adult congenital heart disease, Echocardiography, Rare cardiac anomalies

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Introduction

The aortopulmonary (AP) window, or aortopulmonary septal defect, is an uncommon congenital heart condition, constituting only 0.2–0.6% of all congenital heart diseases [1]. This defect involves an atypical connection between the ascending aorta and pulmonary trunk caused by abnormal embryonic development of the aortopulmonary trunk. Most patients with this condition experience congestive heart failure in infancy due to a left-to-right shunt. Without treatment, survival past infancy and early childhood is rare [2]. However, in patients with a minor defect, symptoms of pulmonary arterial hypertension (PAH) may emerge in adulthood. A notable case involves an adult with a large AP window. The prognosis for untreated cases, especially those with a large AP window, is poor, with a high mortality rate within the first year [3,4]. Rarely do some patients reach adulthood. A reported case involves a 23-year-old man with a large AP window, an anomalous origin of the right pulmonary artery from the aorta (AORPA), and Eisenmenger syndrome, who had not undergone surgery.

Case Report

A male patient, age 23, arrived at our hospital's outpatient department complaining of dyspnea (NYHA class II) for the previous year, which included chest heaviness, chest pain, and shortness of breath. There was no history of paroxysmal nocturnal dyspnea or orthopnea. Additionally, the patient had a history of frequent palpitations during periods of activity dating back five years, during which time he did not seek medical attention. There was evidence of a history of recurrent chest infections in childhood. Abdominal fullness, cyanosis, or pedal edema were not present in the past.

The patient had a systemic examination and showed 149/90 mmHg of blood pressure, 89 beats per minute of pulse, and 19 breaths per minute of respiration. In the indoor air, his saturation level of oxygen was 95%. A detectable second heart sound (P2) and a notable grade 3 parasternal heave were discovered during a cardiovascular test. Auscultation revealed a discernibly louder second heart sound. Furthermore, over the left parasternal area, notably in the second and third intercostal spaces, a grade 3/6 systolic murmur was detected. Aortic and tricuspid valve failure was indicated by the presence of an additional pan systolic murmur over the lower left parasternal region, which become more intense during inspiration.

The findings of the laboratory tests were normal for complete blood count, B-type natriuretic peptide levels, liver and renal function, and urinalysis. The ECG showed enlarged atria and a normal sinus rhythm. The left ventricular ejection percentage was determined to be 59%, and the left ventricular end-diastolic diameter was within normal ranges. Cardiomegaly was discovered by chest radiography. In the parasternal long-axis view, echocardiography also revealed a significant void measuring roughly 22 mm between the ascending aorta and the main pulmonary artery bifurcation, along with evidence of bidirectional shunting on colour Doppler flow imaging. a right pulmonary artery that rises prominently from the ascending aorta. One could observe the main

pulmonary artery giving rise to the left pulmonary artery.

The results of the echocardiography revealed congenital cardiac disease with an AP window and an abnormal origin of the ascending aorta of the right pulmonary artery. A thickened tri-leaflet aortic valve, severe aortic stenosis, peak/mean 160/100 mmHg, mild sub-valvular and valvular pulmonary stenosis, and dilated branch pulmonary arteries were among the other findings. The aorta-pulmonary artery connection was further established by contrast-enhanced CT images of the lungs. The ascending aorta, which seems dilated (44mm), is the source of the right pulmonary artery, whereas the pulmonary trunk continues as the left main pulmonary artery. Surgery was ruled out in light of the final diagnosis, which included an AP window along with an aberrant origin of the right pulmonary artery (AORPA) from the ascending aorta (Figure 1), severe pulmonary arterial hypertension, and Eisenmenger syndrome. The patient was advised to take medication to help lower the pulmonary arterial pressure.

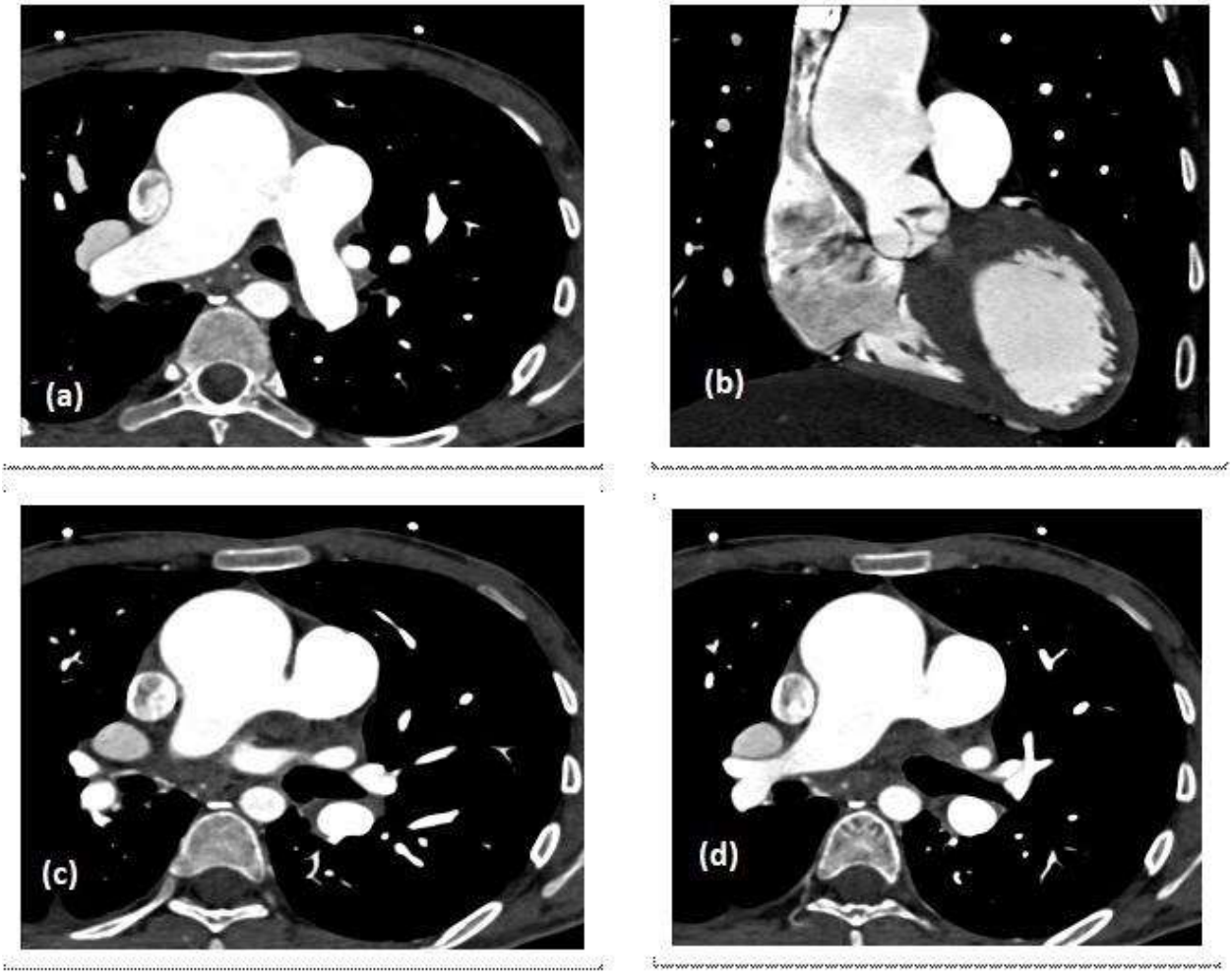

Figure 1. Contrast-enhanced computed tomography images showing a large aortopulmonary window in axial(a) and coronal sections. (b) Right pulmonary artery arising from the ascending aorta. (c) Left pulmonary artery continuation of the main pulmonary artery.

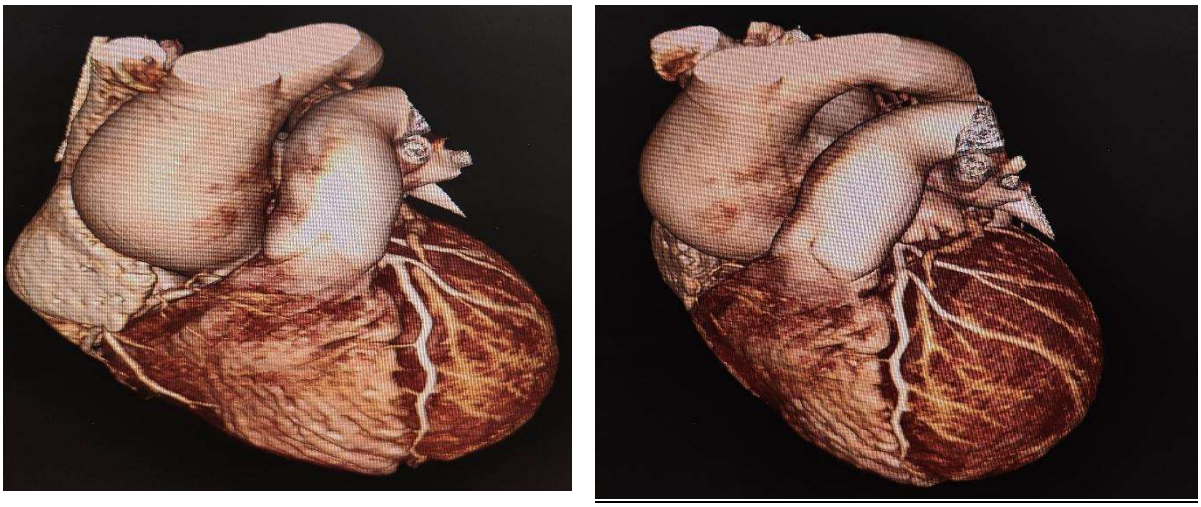

Figure 2. 3D VR image showing aortopulmonary window between ascending aorta and pulmonary artery. Note Anomalous right pulmonary artery arising from ascending aorta.

Discussion

The ascending aorta and the pulmonary artery are directly connected in the aortopulmonary septal defect (APSD), an uncommon congenital cardiac disease that accounts for 0.2%–0.6% of congenital cardiac malformations. This is caused by inadequate embryonic separation of these arteries [1]. It can happen on its own or in conjunction with other cardiac conditions such patent ductus arteriosus or ventricular septal defect. Problem size influences the clinical presentation, which can range from early childhood congestive heart failure to long-term adult survival with Eisenmenger syndrome consequences, especially if the problem is left untreated [3].

One of the most essential diagnostic methods for determining APSD is echocardiography. Surgery is the recommended course of action, ideally performed in early childhood. The most popular technique is transaortic patch closure, while minor lesions with distinct edges may benefit from a transcatheter approach. Pulmonary vascular disease (PVD) can be avoided with urgent surgery, which also has good short- and long-term results. On the other hand, further congenital abnormalities may complicate surgery and have an impact on the outcome [7].

Untreated APSD in adult patients is rare, with survival into adulthood without developing irreversible PVD being exceptionally rare. In such cases, medical management focuses on symptom relief, especially for pulmonary hypertension, using medications such as endothelin receptor antagonists and phosphodiesterase inhibitors, and for heart failure. The management of these patients is challenging, especially when irreversible PVD and Eisenmenger's syndrome are present, which eliminates surgical options [4].

The prognosis of APSD varies; early detection leads to better outcomes. Delayed diagnosis can result in severe complications, including irreversible PVD and Eisenmenger syndrome, which significantly impact patient prognosis [5,8]. Therefore, early detection and management of APSD are essential for effective treatment.

Conclusion

This case report underscores the uncommon occurrence of an aortopulmonary septal defect coexisting with an anomalous origin of the right pulmonary artery from the aorta in a young adult. It emphasizes the indispensable role of advanced imaging studies, particularly computed tomography angiography, as critical techniques for unveiling detailed cardiac structures essential for appropriate surgical planning [2]. The report brings to light adult presentations of congenital heart disease, which have been less prominent in medical literature, and advocates for a multidisciplinary forum for discussing the diagnosis and management of such patients.

Statements and Declarations

Conflicts of interest

The authors declares that they do not have conflict of interest.

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