Case Report

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Congenital unilateral absence of right pulmonary artery with VSD and wide aortopulmonary window in an adult patient: a case report

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Introduction and background: Unilateral absence of pulmonary artery (UAPA) is a very rare condition, with an estimated prevalence of 1 in 200,000 population, which is commonly associated with various cardiovascular anomalies or can occur in an isolated manner. Isolated cases survive to adulthood and remain asymptomatic, but they may frequently experience hemoptysis, repeated infections, or symptoms like dyspnea and chest pain. Due to the rarity of the disorder and its ambiguous appearance, diagnosis can be very challenging.

Case presentation: We present a case of a 28-year-old male who visited our center with the diagnosis of ventricular septal defect with Eisenmenger syndrome elsewhere for further evaluation and was found to have right-sided UAPA with ipsilateral pulmonary hypoplasia and some associated cardiac anomalies.

Clinical discussion: Discussions are held regarding typical chest radiograph findings, diagnostic methods, and possible therapies.

Conclusion: Physicians should be aware of UAPA, which might go undiagnosed for several years despite regular medical care and can show up later in life, causing chronic respiratory symptoms along with Eisenmenger syndrome and ventricular septal defect like in our case.

Keywords: aortopulmonary window, pulmonary artery proximal interruption, unilateral absence of pulmonary artery, ventricular septal defect

Introduction

Unilateral absence of pulmonary artery (UAPA) is a rare congenital anomaly in which one main branch of the pulmonary artery is completely absent, with an estimated prevalence of 1 in 200,000[1]. UAPA artery can present as an isolated anomaly or may be associated with other congenital heart defects such as tetralogy of Fallot, atrial septal defect, ventricular septal defect (VSD), truncus arteriosus, and others[2]. In the majority of cases, patients are diagnosed in infancy or childhood. Only some cases are asymptomatic, and thus remain undiagnosed till adulthood[2].

In this paper, we report a 28-year-old male with an absence of a right pulmonary artery (RPA) and wide aortopulmonary window associated with VSD. This case report has been written as per Surgical CAse REport (SCARE) 2020 criteria[3].

HIGHLIGHTS

- Unilateral absence of the pulmonary artery (UAPA) is a rare congenital anomaly with a prevalence of 1 in 200,000.
- UAPA is more accurately described by the terms 'pulmonary artery proximal interruption,' 'nonconfluent pulmonary artery,' or 'ductal origin of the distal pulmonary artery.'
- Computed tomography and MRI are definitive diagnostic techniques of UAPA with accurate anatomical depiction.
- Therapeutic approach for UAPA is based on the symptoms of the patient, pulmonary artery anatomy associated with cardiovascular anomalies, aortopulmonary collateral, and pulmonary artery hypertension.

Case presentation

A 28-year-old male visited the outpatient clinic with shortness of breath for 1 day associated with generalized weakness. Shortness of breath developed on normal physical activity and on walking uphill (NYHA grade II). He denied chest pain, palpitation, cough, syncope, lower extremity swelling, orthopnea, and paroxysmal nocturnal dyspnea. There was a history of multiple hospitalizations in the past for recurrent lower respiratory tract infections, and he was diagnosed in one of the other centers as VSD with Eisenmenger syndrome after which the patient visited our cardiac center.

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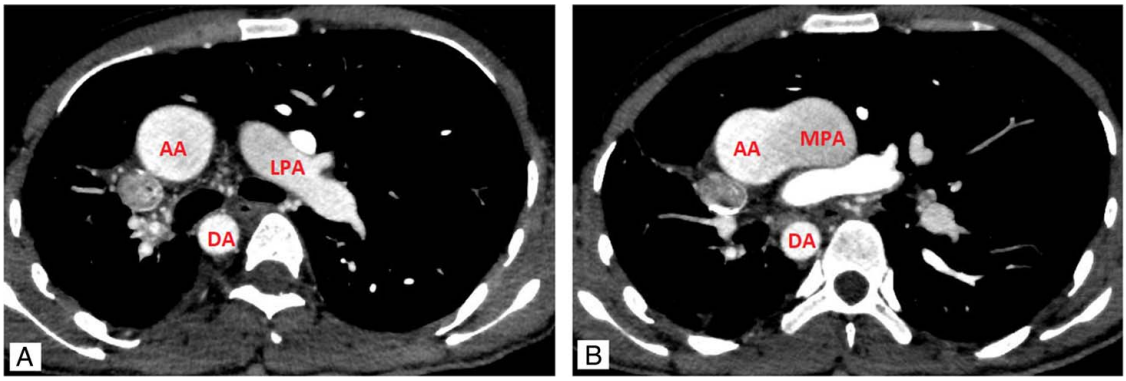

Figure 1. Axial postcontrast computed tomography image (A) shows an absence of the right pulmonary artery when the left pulmonary artery (LPA) is visible at the same level supplying the left lung; however, lobar pulmonary arteries are still visible in right lung secondary to supply of blood via major aortopulmonary collateral arteries. Image (B) shows wide communication between the ascending aorta (AA) and the main pulmonary trunk (MPA). DA, descending aorta.

Physical examination revealed an averagely built male, his pulse rate was 74 beats/min, respiratory rate was 20 breaths/min, and blood pressure was 130/80 mmHg with no jugular venous distention. The chest was more or less symmetrical with decreased breath sound on the right side. Clubbing and cyanosis were present, and the rest of the physical examination was unremarkable. Hemoglobin was 22 g/dl with other hematologic and biochemical profiles within normal ranges.

Electrocardiography showed sinus rhythm with right axis deviation and P-pulmonale. A chest radiograph revealed the decreased volume of the right lung and hyperinflation of the left lung with a right shift of the mediastinum. An echocardiogram showed perimembranous VSD 1.2 cm with bidirectional shunt, severe pulmonary artery hypertension (PAH) with a pulmonary regurgitation peak gradient of 91 mmHg, mild right ventricular hypertrophy, and normal ejection fraction.

Pulmonary computed tomography (CT) angiogram revealed the absence of the RPA (Fig. 3B), right-sided aortic arch, and small hypoplastic right lung with the displacement of the heart and mediastinum to the right (Fig. 2B) along with wide aortopulmonary window (Fig. 3B).

Contract-enhanced computed tomography of the chest revealed an absent RPA with multiple medium-sized major aortopulmonary collateral arteries MAPCAs in the right hilar region supplying lobar/segmental pulmonary arteries of the right lung (Fig. 1A). There was a decreased volume of the right lung compensated by hyperinflation of the left lung with retrosternal herniation toward the right side (Fig. 2A). A few focal fibrotic changes were noted in the right lung with fissure thickenings (Fig. 2A). There was an abnormal wide communication between the proximal ascending aorta and the main pulmonary artery; communication measuring 30 mm in diameter (Figs 1B, 3A).

Discussion

Congenital absence of pulmonary artery was first reported in 1868 by Fraentzel[4]. Unilateral absence of the pulmonary artery is twice as common on the right side[2]. Due to the lack of an adequate number of these cases, the prevalence of left-sided versus right-sided UAPA shows discrepancies that vary from study to study [2,5]. The involution of the proximal sixth aortic arch and the persistence of the connection of the intrapulmonary pulmonary artery to the distal sixth aortic arch causes the absence of the pulmonary artery[2]. This anomaly is more accurately described by the terms 'pulmonary artery proximal interruption,', 'nonconfluent pulmonary artery,' or 'ductal origin of the distal pulmonary artery'[6].

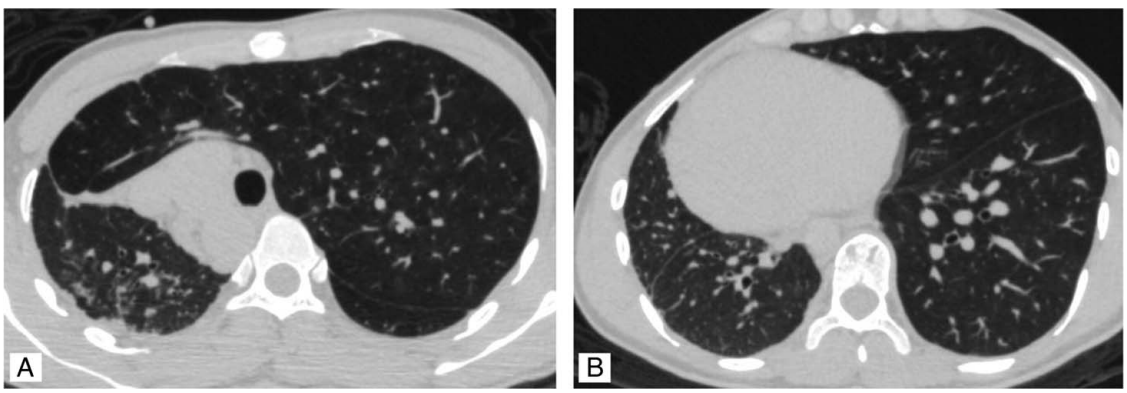

Figure 2. Axial computed tomography images in the lung window show the decreased volume of the right lung (i.e. hypoplastic lung) with an ipsilateral shift of mediastinum (B) and compensatory hypertrophy of the left lung with retrosternal herniation (A). Few focal fibrotic changes are noted in the right upper lobe, predominantly toward the posterior pleura.

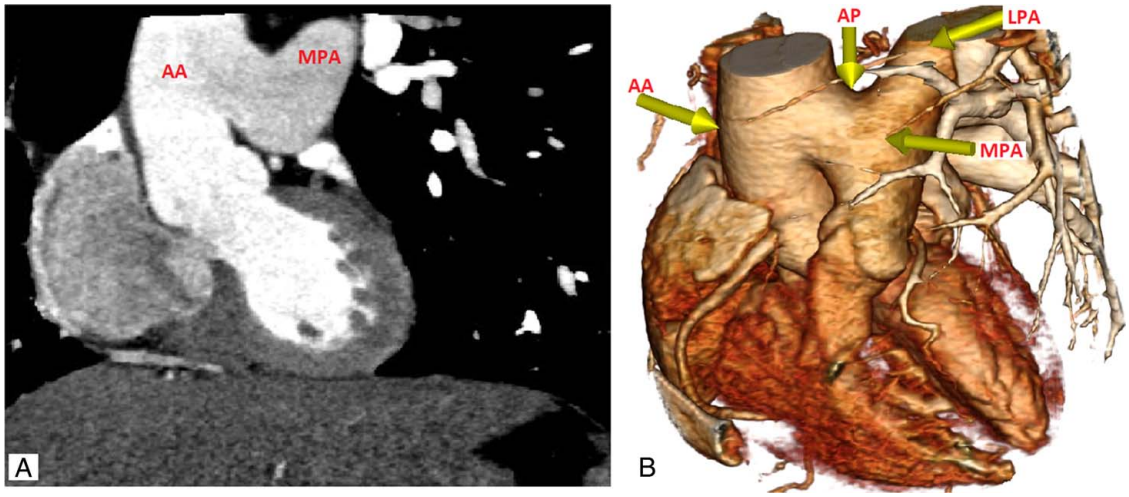

Figure 3. Coronal postcontrast computed tomography image (A) of the heart and three-dimensional reconstructed image of the heart and great vessels (B) show wide communication between the ascending aorta (AA) and main pulmonary trunk (MPA) with image (B) also demonstrating the absence of the right pulmonary artery; however, left pulmonary artery (LPA) is visualized. AP, aortopulmonary communication.

Although right-sided UAPA is common, the left-sided seems to be more frequently associated with congenital cardiovascular anomalies such as tetralogy of Fallot, VSD, atrial septal defect, coarctation of the aorta, right aortic arch, truncus arteriosus, patent ductus arteriosus, aortopulmonary window, and pulmonary atresia. Patients with isolated right UAPA survive into adulthood with minimal symptoms, which makes it difficult to diagnose at an early stage[7]. In this patient, right-sided UAPA is associated with congenital cardiovascular anomalies, which were diagnosed in his adulthood.

Symptomatic presentation of UAPA shows two different patterns; during infancy, it presents with congestive cardiac failure and PAH, whereas in adulthood, it remains asymptomatic or presents with exercise intolerance and hemoptysis[8,9]. Incident of PAH is quite high with UAPA irrespective of the associated defect[7]. Other common symptoms are recurrent pulmonary infections, decreased exercise tolerance, and mild dyspnea during exertion[7]. This patient has a history of recurrent lower respiratory tract infections, dyspnea on exertion, and is under treatment for severe PAH.

Diagnosis of UAPA is a difficult and challenging task, especially when the patient is previously misdiagnosed. The diagnosis of UAPA is based on taking a complete medical history, physical examination, and imaging examinations. The chest radiograph often shows a reduction in the size of the affected hemithorax, compensatory hyperinflation of the contralateral hemithorax, elevation of the ipsilateral hemidiaphragm, an absent ipsilateral and enlarged contralateral pulmonary artery shadow, and ipsilateral mediastinum shift[10]. Most of these findings were present in this patient and were previously interpreted as a result of the respiratory tract infection.

Echocardiography is a useful tool to establish the diagnosis, exclude cardiac and vessel abnormalities, and evaluate the presence or development of PAH. CT and MRI are definitive diagnostic techniques of UAPA with accurate anatomical depiction. Both techniques are not just used for diagnostic purposes but also to note other findings such as variable collateral circulation such as major aortopulmonary collateral arteries, mosaic parenchymal changes, and bronchiectasis secondary to recurrent infections. Although angiography is the gold standard for the diagnosis of pulmonary artery agenesis, it is now rarely used due to the emergence of CT and MRI and is only taken into consideration for individuals who require surgical intervention[1,2,11].

The therapeutic approach for UAPA is based on the symptoms of the patient, pulmonary artery anatomy associated with cardiovascular anomalies, aortopulmonary collateral, and PAH. No treatment is required in patients without any evidence of cardiopulmonary dysfunction. They should be closely monitored and followed up regularly[2,9]. Other treatment options, including revascularization of the distal affected pulmonary artery and lobectomy, or selective embolization of systemic arterial supply, are considered in symptomatic patients[2,7]. Medical management includes specific management of coexisting PAH, including endothelin receptor antagonists like bosentan or prostaglandin antagonists and other supportive therapy like diuretics[12].

Conclusions

UAPA is a rare congenital anomaly, and the diagnosis in an adult makes this case unique and interesting. Physicians should be aware of UAPA, which might go undiagnosed for several years despite regular medical care and can show up later in life, causing chronic respiratory symptoms along with Eisenmenger syndrome and VSD like in our case.

Ethical approval

This is a case report; therefore, it did not require ethical approval from the ethics committee.

Patient consent

Written informed consent was obtained from the patient for the publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

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Author contribution

All authors were involved in writing the paper, the collection of data, the revision of it critically for important intellectual content, review, and editing.

Conflicts of interest disclosure

All the authors declare that they have no conflicts of interest.

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Guarantor

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