Case Report

A silent anomaly: congenital unilateral absence of pulmonary artery incidentally discovered in adulthood, presenting as pulmonary hypertension and chronic respiratory failure—case report

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Background: Unilateral absence of the pulmonary artery (UAPA) is a rare congenital anomaly that results from the failure of the sixth aortic arch to connect with the pulmonary trunk during embryogenesis. Left-sided UAPA is often associated with congenital heart diseases, whereas right-sided UAPA is typically isolated and asymptomatic. There is limited literature available on individuals diagnosed with UAPA in mid-adulthood. Therefore, this case report seeks to contribute to the existing knowledge, aiming to enhance understanding of UAPA in this age group and its associated management goals.

Case Description: Case 1 involves a 44-year-old man with a history of heavy smoking and chronic obstructive pulmonary disease (COPD) as well as noncompliance with medication who presented with hypoxia, hemoptysis, and worsening dyspnea. Further examinations revealed severe pulmonary arterial hypertension (PAH) due to congenital absence of the right main pulmonary artery (PA) and extensive systemic arterial collateralization. Treatment included bronchodilators, supplemental oxygen, sildenafil, ambrisentan, and spironolactone. However, he ultimately succumbed to right ventricular failure. A previous computed tomography (CT) angiogram had shown absence of the right PA, indicating a congenital condition, but the patient was lost to follow-up after being referred for a lung transplant. Case 2 describes a 46-year-old woman of Ghanaian descent with uncontrolled asthma and a significant history of dyspnea and respiratory distress linked to an aspirin allergy. Investigations revealed an absent right PA leading to a hypoplastic right lung with chronic changes, and severe PAH. Treatment with corticosteroids and nebulization led to improvement, but a right heart catheterization confirmed elevated pulmonary pressures and a left-to-right shunt due to collateral circulation. An aortogram showed a large fistulous connection to the hypoplastic lung. A complex intervention involving the placement of coils in the collateral vessels achieved a successful outcome.

Conclusions: UAPA in adulthood is a diagnostic and therapeutic challenge due to its rarity, necessitating careful evaluation and individualized treatment approaches.

Keywords: Unilateral absence of the pulmonary artery (UAPA); pulmonary hypertension (PH); case report

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Introduction

Unilateral absence of the pulmonary artery (UAPA) is a rare congenital anomaly resulting from the failure of the sixth aortic arch to connect with the pulmonary trunk during embryogenesis (1,2). Left-sided UAPA is more commonly associated with congenital cardiovascular malformations making it easy to diagnose and treat early in life, whereas right-sided UAPA is often asymptomatic and incidentally discovered on chest radiographs (3,4). Although UAPA is typically diagnosed in adolescence (average age: 14 years), we encountered atypical presentations at our institute leading to diagnoses in mid-adulthood, prompted by symptoms of pulmonary hypertension (PH) and respiratory failure.

The scarcity of literature regarding isolated UAPA presents challenges in diagnostic and therapeutic planning (5). Therefore, our goal is to add to the existing knowledge base, develop a deeper understanding of its clinical presentation, management, and the ability to predict outcomes. The cases presented originate from a single-center, retrospective design and are non-consecutive. We present this article in accordance with the CARE reporting checklist (available at https://shc.amegroups.com/article/view/10.21037/shc-23-48/rc).

Highlight box

Key findings

- Unilateral absence of the pulmonary artery (UAPA) is a rare congenital anomaly usually identified during adolescence. However, atypical presentations prompted by the development of pulmonary hypertension (PH) and respiratory failure may lead to diagnosis in mid-adulthood with worsening clinical condition.

What is known and what is new?

- Due to slow progression of the disease, individuals with UAPA frequently experience symptoms for a prolonged time without fully grasping the gravity of their situation, ultimately resulting in adverse consequences.
- Managing UAPA poses challenges due to the absence of a universally accepted treatment protocol. Treatment is typically tailored to the individual's clinical presentation. In cases of PH, pharmacotherapy with vasodilators is commonly pursued, whereas surgical options like selective embolization are reserved for individuals experiencing massive hemoptysis. Notably, lung transplant may emerge as a definitive treatment option.

What is the implication, and what should change now?

- The gradual advancement of UAPA and its atypical presentations lead to patients experiencing symptoms for an extended duration without recognizing the gravity of their condition. Clinicians must maintain a high index of suspicion and conduct thorough scrutiny, employing both general and specialized investigations, when determining the underlying cause.

Case presentation

Case 1

A 44-year-old man was brought to the emergency department with hypoxia, hemoptysis, and progressive dyspnea. He complained of nonproductive cough, intermittent wheezing, and progressive dyspnea with minimal activity over the previous year. He was a heavy smoker and diagnosed with chronic obstructive pulmonary disease (COPD), requiring ongoing home oxygen treatment. His medical history was noteworthy for poorly controlled COPD exacerbations attributed to noncompliance with prescribed medications, leading to recurrent emergency room visits. Further investigation revealed an extensive history of substance abuse and nonadherence to oxygen therapy. Physical examination revealed mild lower extremity edema and bilateral expiratory wheezing. The patient was initially stabilized on fluids, his hemoptysis resolved spontaneously without any further episodes or need for embolization. Subsequent imaging, with a contrast-enhanced computed tomography (CT), did not reveal any active bleeding sites. On further workup, chest X-ray demonstrated a small-volume right hemithorax with increased reticular opacities, mild rightward shift of the cardiomediastinal silhouette, an empty right pulmonary bay (hilum), and compensatory hypertrophy of the left lung (*Figure 1*). These findings were further characterized on contrast-enhanced CT, revealing a markedly dilated main pulmonary arterial trunk measuring up to 4.2 cm in transverse diameter consistent with severe pulmonary arterial hypertension (PAH), an absent right main pulmonary artery (PA), and extensive systemic arterial collateralization in the hypoplastic right lung, most compatible with congenital proximal interruption of the PA. Moderate emphysema and compensatory hyperinflation of the left lung was also observed (*Figure 2*). Electrocardiogram showed a normal sinus rhythm with right axis deviation, incomplete right bundle branch block, and right ventricular hypertrophy. A limited echocardiogram study revealed a dilated right ventricle with reduced function. Once stable, the patient underwent a right heart catheterization, revealing a mean PA pressure of 34 mmHg, a right atrial (RA) pressure of

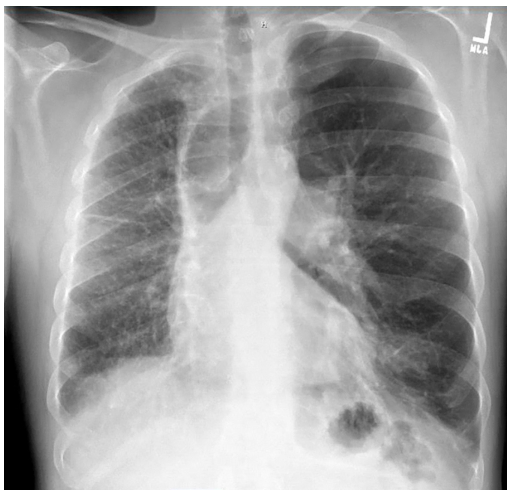

Figure 1 Frontal chest radiograph shows decreased right lung volume with increased reticular opacities, rightward cardiomediastinal shift, and an empty right hilum. Compensatory hyperinflation of the left lung and enlarged main pulmonary artery is also present.

7 mmHg, and a PA wedge pressure of 10 mmHg. The patient was continued on bronchodilators and supplemental oxygen therapy. Sildenafil and ambrisentan for PAH were initiated and spironolactone 20 mg, 3 times a day, was administered to help with his volume status. Despite initial stabilization, the patient succumbed to ongoing right ventricular failure and dysrhythmias leading to cardiac arrest.

Interestingly, a CT angiogram done a year previously revealed absence of the right PA, with perfusion to the right lung being supplemented through collateral vessels originating from the bronchial artery, the costal artery, or the thoracic aorta. The reduced volume of the right lung suggested the possibility of a congenitally small lung; however, the cardiac contour was normal without evidence of congenital cardiac defects. Additionally, hypertrophy of the left PA was observed, along with clear visualization of the aorta and the left PA, including its segmental branches. At that time, the patient was referred to a lung transplant center but was lost to follow-up.

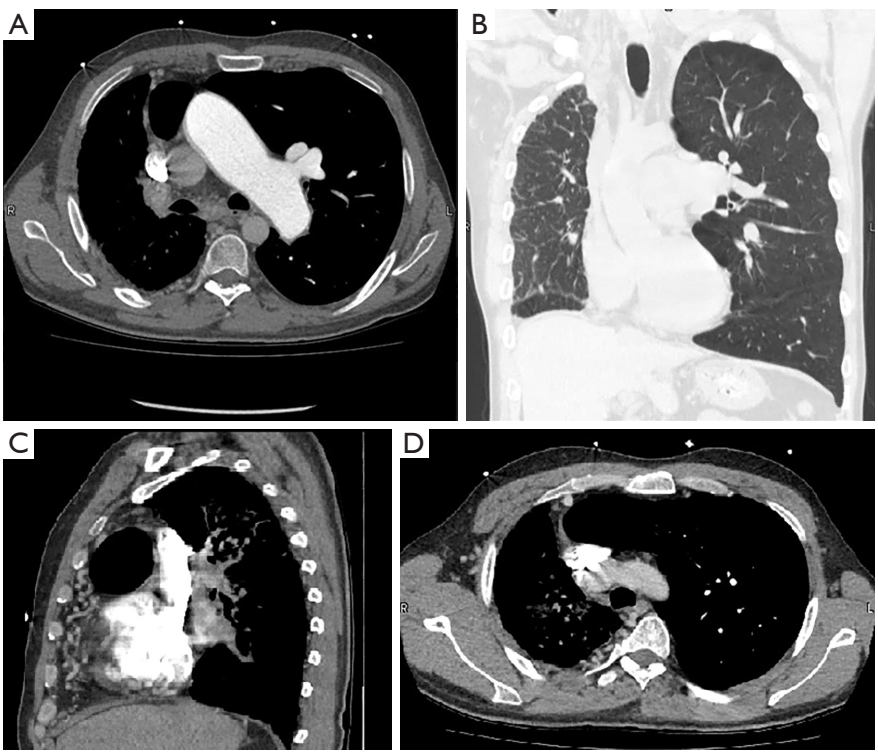

Figure 2 CT images. (A) Axial CT image with intravenous contrast (mediastinal window) shows complete absence of the right main pulmonary artery and a dilated main and left pulmonary artery. (B) Coronal CT image (lung window) shows hypoplasia of the right lung with elevated right hemidiaphragm and contralateral hyperinflation of the left lung. Sagittal (C) and axial (D) CT images of the chest with intravenous contrast (mediastinal window) show extensive systemic to pulmonary artery supply to the right lung, including bronchial, intercostal, and internal thoracic collaterals. CT, computed tomography.

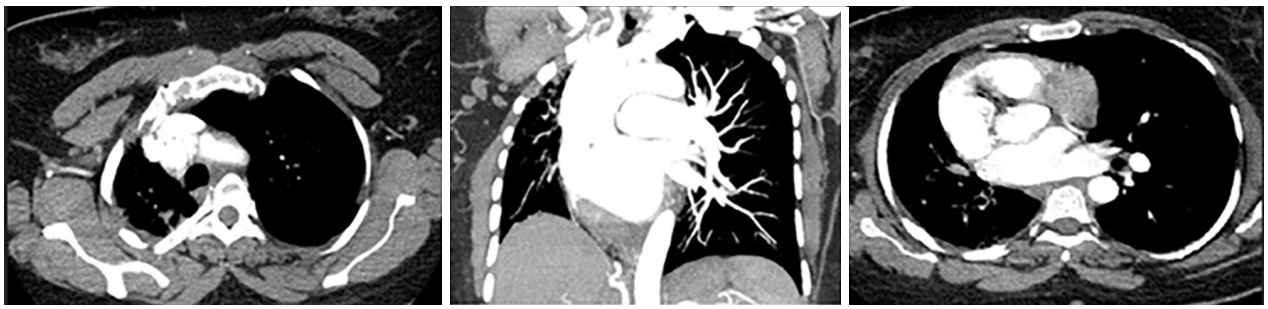

Figure 3 Chest CT in different views revealed the absence of the right pulmonary artery, resulting in hypoplasia of the right lung. The right lung is supplied by a hypertrophied bronchial artery branching off of the aorta. Chronic interstitial changes, scarring and bronchiectasis are seen in the hypoplastic right lung. CT, computed tomography.

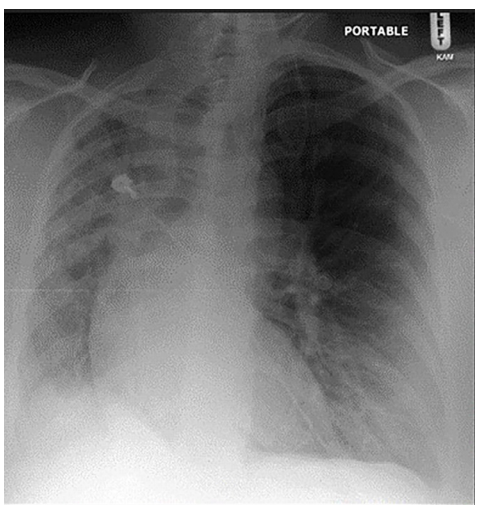

Figure 4 Chest X-ray showing diminished lung volume on the right with an ipsilateral shift of the heart and mediastinum.

Case 2

A 46-year-old woman of Ghanaian descent with a history of uncontrolled asthma presented to the emergency department with symptoms of asthma exacerbation that had been present for 2 days. Since being diagnosed with asthma at the age of 31, she has had regular visits to the emergency department for upper respiratory infections. She also reported a considerable history of dyspnea with exertion, particularly during daily activities, and previous respiratory distress due to an allergic response to aspirin. Furthermore, her family history revealed several incidences of nasal polyps among her relatives.

A chest X-ray revealed reduced lung volume on the right side as well as ipsilateral heart and mediastinum displacement. Chest CT and chest X-rays revealed absence of the right PA (*Figures 3,4*), resulting in a hypoplastic right lung fed by a hypertrophied bronchial artery branching out from the aorta. Furthermore, the underdeveloped right lung revealed chronic interstitial changes, scarring, and bronchiectasis. A ventilation-perfusion scan revealed a lack of blood flow to the right lung as well as perfusion abnormalities in the base of both lung (*Figures 4,5*). Echocardiogram revealed a PA pressure of 77 mmHg with a right ventricular systolic pressure of 76.7 mmHg. In addition, moderate tricuspid regurgitation and mild right ventricular dilatation along with moderate right ventricular hypokinesis was noted.

The patient was admitted to the hospital and treated with oral corticosteroids and a nebulizer and exhibited remarkable clinical improvement within 3 days. An outpatient right heart catheterization to evaluate the possibility of PH revealed an elevated mean RA pressure of 10 mmHg, a right ventricular pressure of 45/10 mmHg, and an end-diastolic pressure of 12 mmHg. The left PA pressure was 46/24/30 mmHg, and the pulmonary capillary wedge pressure was 10 mmHg, with an approximate transpulmonary gradient of 20 mmHg. Cardiac output, calculated using the FICK equation, was 4.56 L per minute, with a cardiac index of 3.55 L per minute/m^2. The calculated pulmonary vascular resistance was 4.38 mmHg × min/L or 350 per cm^3.

Oxygen saturation measurements demonstrated a rise in saturation from the superior vena cava to the right atrium, indicating the presence of a left-to-right shunt attributed to collateral circulation. The partial pressure of oxygen in the aorta climbed to 484 mmHg after a nitric oxide challenge (20 ppm) and continued high-flow oxygenation, while the left PA pressure increased to 52 mmHg. After a 5-minute period, the aortic partial pressure increased to 514 mmHg,

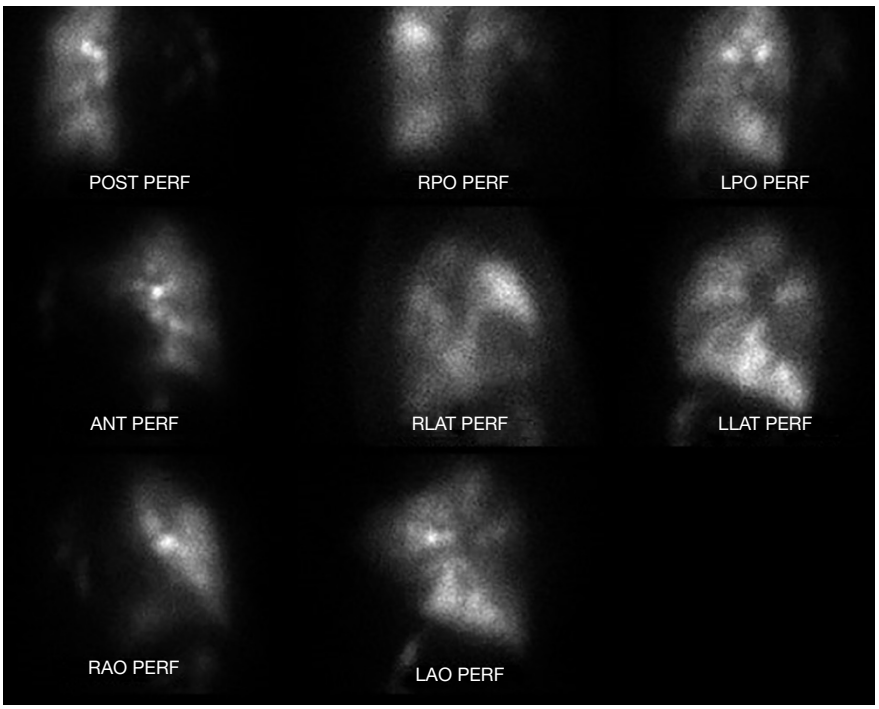

Figure 5 Ventilation perfusion scan revealed absent perfusion in the right lung with perfusion defects in both lung bases. POST, after; PERF, perfusion; RPO PERF, right posterior oblique perfusion; LPO PERF, left posterior oblique perfusion; ANT PERF, anterior perfusion; RLAT PERF, right lateral perfusion; LLAT PERF, left lateral perfusion; RAO PERF, right anterior oblique perfusion; LAO PERF, left anterior oblique perfusion.

indicating a good response to the vasodilatory effects of nitric oxide.

An aortogram revealed a large trifurcating fistulous connection originating from the proximal descending aorta and extending to the underdeveloped right lung, with indications of sequestration observed as contrast material entered the heart chambers during the contrast injection phase. Consequently, the patient underwent a complex structural intervention, involving the placement of seven coils: two in the superior, three in the middle, and two in the inferior collateral vessels, resulting in an excellent technical outcome.

Ethical consideration

All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patients for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

Discussion

UAPA is an extremely rare congenital malformation that affects 1 in 200,000 adults. In most cases, the right PA, opposite to the aortic arch, is affected due to altered development of the sixth aortic arch segment that fails to connect with the pulmonary trunk (4). Subsequently, extensive systemic collateral branches develop to supply the ipsilateral peripheral pulmonary arteries, which can often lead to hemoptysis, as seen in the patient presented in case 1. UAPA rarely occurs in isolation, as it did in this case. It is more frequently seen in conjunction with congenital cardiac defects such as tetralogy of Fallot or septal defects (1-3).

Dyspnea, exercise intolerance, recurrent chest infections, PH, hemoptysis, and chest pain are all nonspecific clinical manifestations of UAPA. Therefore, a thorough evaluation is necessary to diagnose this condition as it is easy to miss. A chest radiograph will classically show ipsilateral volume loss

with diaphragmatic elevation and shift of the mediastinum toward the affected side as well as hyperaeration of the contralateral lung (2,3,5). Fine non-branching linear opacities can be seen at the lung periphery indicating enlarged intercostal and transpleural pulmonary collaterals. Pulmonary angiography is the gold standard for diagnosing UAPA, however, CT, magnetic resonance imaging (MRI), and echocardiogram are more often used to confirm the diagnosis (6).

Isolated UAPA with PH has rarely been documented, and risk factors for the development of PH are unknown. In most cases, PH is mild, unlike our patient in case 1 for which right heart catheterization findings confirmed the diagnosis of severe PH. According to one theory, PH develops as a result of pulmonary arterial vascular remodeling, which occurs in response to increased blood flow through the single patent PA leading to endothelial intimal injury, intimal hyperplasia, medial hypertrophy, and ultimately collagenous replacement of the intima in advanced disease. Although most cases of PH have been described as mild, PH in the patient described in case 1 may have been exacerbated by his past noncompliance with O_2 therapy, multiple COPD exacerbations, and substance abuse in addition to his UAPA (6,7).

Since a universally accepted form of therapy does not exist, UAPA continues to pose therapeutic challenges. Serial echocardiography is used to monitor asymptomatic patients for the emergence of PH. Adult patients receive therapy that is specific to their clinical presentation. Due to extensive fibrosis of the intrapulmonary arteries, surgical options are often not viable (8). Therefore, patients with PH have largely pursued pharmacotherapy with vasodilators like endothelin receptor antagonists, calcium channel blockers, and intravenous prostacyclins with improvements seen in their dyspnea and PH. Massive hemoptysis may be treated with selective embolization of the systemic collateral circulation, and pneumonectomy may be preferred in those with recurrent hemoptysis, with the exception of high risk patients due to the technical difficulties with embolization brought on by the extensive collateral circulation present in UAPA (8-10). Despite a favorable response to medical therapy observed in our patient in case 2, it was deemed necessary for her to undergo a complex structural intervention to attain the optimal outcome. Conversely, our patient in case 1 exhibited resistance to all medical interventions, leading to the determination that lung transplantation remained the most suitable course of treatment.

Conclusions

To summarize, given the slow progression of their disease, patients with UAPA often experience symptoms for an extended period without recognizing the seriousness of their condition which eventually leads to detrimental effects. This case highlights the diagnostic and therapeutic challenges that patients with rare causes of PH may experience as well as the need for a high index of suspicion and meticulous scrutiny of both general and specialized investigations when establishing the etiology.

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Footnote

Reporting Checklist: The authors have completed the CARE reporting checklist. Available at https://shc.amegroups.com/article/view/10.21037/shc-23-48/rc

Peer Review File: Available at https://shc.amegroups.com/article/view/10.21037/shc-23-48/prf

Conflicts of Interest: All authors have completed the ICMJE uniform disclosure form (available at https://shc.amegroups.com/article/view/10.21037/shc-23-48/coif). The authors have no conflicts of interest to declare.

Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patients for publication of this case report and accompanying images. A copy of the written consent is available for review by the editorial office of this journal.

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