International Clinical and Medical Case Reports Journal
Case Report (ISSN: 2832-5788)

Uncommon Cause of Hypoxaemia: A Case Report of Pulmonary Arteriovenous Fistula

Ponaswath K[1*], Sukanya N[2] , Kiruthika J[3] , Kalpana Ramanathan[4] , Chandrasekar S[5]

[1]Post Graduate of General Medicine, Department of General Medicine, Government Stanley Medical College and Hospital, Chennai, India

[2]Senior Assistant Professor of General Medicine, Department of General Medicine, Government Stanley Medical College and Hospital, Chennai, India

[3]Assistant Professor of General Medicine, Department of General Medicine, Government Stanley Medical College and Hospital, Chennai, India

[4]Professor, Department of General Medicine, Government Stanley Medical College and Hospital, Chennai, India

[5]Professor and Head, Department of General Medicine, Government Stanley Medical College and Hospital, Chennai, India

Citation: *Ponaswath K, Sukanya N , Kiruthika J , Kalpana Ramanathan, Chandrasekar S. Uncommon Cause of Hypoxaemia: A Case Report of Pulmonary Arteriovenous Fistula. Int Clinc Med Case Rep Jour. 2025;4(4):1-7.*

Received Date: 05 April, 2025**; Accepted Date:** 08 April, 2025**; Published Date:** 10 April, 2025

[*]**Corresponding author:** Ponaswath K, Post Graduate of General Medicine, Department of General Medicine, Government Stanley Medical College and Hospital, Chennai, India

ABSTRACT

Pulmonary arteriovenous malformation (PAVM) is a rare vascular anomaly characterised by an abnormal right-to-left shunt, leading to impaired gas exchange and disruption of pulmonary filtration. Although uncommon in the general population, PAVMs are a crucial differential diagnosis in patients presenting with hypoxaemia, pulmonary nodules, or haemoptysis. Here, we report a rare case of a 38-year-old female who presented with complaints of headache and was subsequently diagnosed with a pulmonary arteriovenous fistula.

Keywords: Osler-Weber-Rendu syndrome; Central cyanosis; Pandigital clubbing; Paradoxical embolism; Pulmonary arteriovenous fistula

INTRODUCTION

Pulmonary arteriovenous malformations (PAVMs), also known as pulmonary arteriovenous fistulae, are abnormal direct connections between the pulmonary artery and vein, bypassing the normal capillary network and resulting in a right-to-left shunt. First described by Churton in 1897, PAVMs were later recognised as a key manifestation of hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu syndrome.While many individuals with PAVMs remain asymptomatic, undiagnosed cases carry a risk of severe complications, including

ischaemic stroke, myocardial infarction, cerebral abscess, massive haemoptysis, and haemothorax.[1] Pulmonary arteriovenous malformations (PAVMs) are rare in the general population. This was demonstrated in a study of 15,000 consecutive autopsies, where only three cases were identified. However, the true prevalence may be higher than autopsy data.[1] A large-scale study analysing over 21,000 screening computed tomography (CT) scans, which can detect smaller PAVMs, estimated a prevalence of approximately 1 in 2,600 individuals.[1] PAVMs are 1.5 to 1.8 times more common in females than in males and are predominantly congenital. In the general population, PAVMs are typically solitary (90%) and unilateral (97%), with a strong predilection for the lower lobes, where at least two-thirds are found.[1] Approximately 85% of pulmonary arteriovenous malformations (PAVMs) are classified as simple, meaning they receive blood supply from one or more arteries within a single pulmonary segment. In contrast, complex PAVMs, which account for 5%-10% of cases, have multiple arterial feeders from different pulmonary segments.[2]

CASE PRESENTATION

A 34-year-old female presented with complaints of headache and neck pain for two days. She had no history of fever. Three years ago she had undergone burr hole evacuation for a left occipital pyogenic brain abscess, and 2 months later she developed right-sided hemiparesis due to a left middle cerebral artery infarct. One year later, she had a severe headache and vomiting and was diagnosed with cortical vein thrombosis and was started on oral anticoagulants. In view of a previous history of arterial and venous infarcts, the patient was advised to have further evaluation but was lost to follow-up.

On examination, the patient was drowsy and was partially obeying oral commands. She had pandigital clubbing of grade III and central cyanosis. She was tachypneic and had persistent hypoxaemia with room air saturation of 78%. Neurological examination revealed increased tone and reduced power in the right upper and lower limbs, consistent with her previous stroke, while cranial nerve function, sensory examination, and higher mental functions were normal. Other systemic examinations were unremarkable. Notably, her mother and maternal uncle also exhibited pandigital clubbing, though there was no family history of thrombotic episodes.

Laboratory investigations showed increased haemoglobin (18 g/dl), a haematocrit of 55%, and an elevated PT-INR with otherwise lab normal parameters. In view of her persistent headache, a CT brain was performed, which revealed a subdural haemorrhage in the right front temporoparietal region along with chronic infarcts in the left middle cerebral artery territory. The findings were suggestive of an acitrom induced intracranial haemorrhage, for which she was managed with a fresh frozen plasma transfusion and IV Vitamin K. Her sensorium improved after 5 days. Given her low SpO2, cyanosis, and pandigital clubbing, cyanotic heart disease was suspected, and an

echocardiogram was performed, which was normal. Arterial blood gas (ABG) analysis revealed a pH of 7.42, a PaO2 of 56 mmHg, and a PaCO2 of 40 mmHg, indicating chronic hypoxaemia. Serum erythropoietin level was done in view of polycythaemia, hypoxaemia, and recurrent thrombosis and was found to be elevated, suggesting secondary polycythaemia.

On the tenth day of hospitalization, the patient developed pain, swelling, erythema, and calf tenderness in the right lower limb. A bilateral lower limb venous Doppler ultrasound detected an echogenic thrombus extending from the posterior tibial vein to the common femoral vein in the right lower limb, confirming deep vein thrombosis (DVT). She was started on anticoagulation with Fondaparinux and later bridged with Acitrom due to her high thromboembolic risk. Further evaluation for an underlying hypercoagulable state was initiated.(Table 1)

Table 1: Investigations for hypercoagulable state

D-Dimer	700ng/ml (<500ng/ml)
Serum homocysteine	12 µmol/L (5-15 µmol/ml)
APLA profile	Negative
Serum erythropoietin	**35** mIU/ml (4-26 mIU/ml)
APLA	Negative

A repeat echocardiogram was performed to rule out an intracardiac shunt, which was normal. Further transthoracic saline bubble contrast echocardiography (TTCE) was done. This revealed saline microbubbles entering the left heart chambers after three cardiac cycles, suggesting a PAVM. To confirm the diagnosis, CT pulmonary angiography, the gold standard for detecting PAVMs, was performed. The scan showed multiple small subpleural abnormal communications between terminal branches of the pulmonary artery and pulmonary vein in the left anterior basal segments, left lower lobe, and lingula, with diffusely dilated and prominent pulmonary arterial and venous branches extending to the pleural surface.(Figure 1) This established the final diagnosis of a pulmonary arteriovenous malformation with secondary polycythaemia, which explained the patient's prior brain abscess and CVA as a result of paradoxical embolism. The patient was advised to undergo digital subtraction angiography (DSA) and Embolotherapy for the AV malformation. Given the suspicion of hereditary hemorrhagic telangiectasia (HHT), a detailed genetic workup was planned.

However, due to financial constraints, she declined further treatment. She was discharged on oral anticoagulation and is currently under regular follow-up.

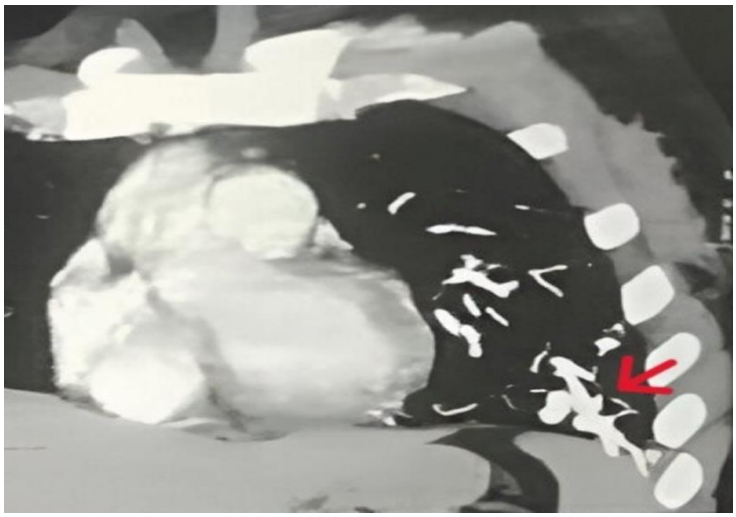

Figure 1: CT Pulmonary Angiography showing small subpleural abnormal communications between terminal branches of the pulmonary artery and pulmonary vein (red arrow).

DISCUSSION

Approximately 70% of pulmonary arteriovenous malformations (PAVMs) are associated with hereditary hemorrhagic telangiectasia (HHT). Conversely, 15% to 35% of individuals with HHT have PAVMs. Genetic mutations in endoglin (HHT1) and activin receptor-like kinase 1 (HHT2) disrupt TGF-β signalling, leading to impaired vascular remodelling, which is believed to contribute to PAVM formation. Other proposed mechanisms include defects in arterial loops, incomplete resorption of vascular septae, or abnormal capillary development during foetal growth.[3] HHT is an autosomal dominant disorder characterised by arteriovenous malformations (AVMs) affecting multiple but specific organs and tissues. Vascular lesions typically include small telangiectasias of the skin, nasal mucosa, and gastrointestinal tract, as well as larger AVMs most commonly involving the central nervous system, lungs, and liver(Table 2). Once a pathogenic gene mutation is identified in a clinically affected family member, genetic testing is recommended for at risk relatives to facilitate early detection and management.[4]

Table 2: Curacao criteria

Epistaxis	>1 episode of spontaneous bleed (nocturnal bleeding is particularly suspicious)
Multiple telangiectasias	Common sites include the lips, oral cavity, fingers and nose
Visceral lesions	Common sites include pulmonary AVM,hepatic AVM,cerebral AVM,Spinal AVM, and GI telangiectasias
Family history	A first degree relative with an HHT diagnosis made by these criteria

A diagnosis of HHT is definite if three or more criteria are met, possible/suspected if two criteria

are met, and unlikely if less than two criteria are met.[5]

Most patients with pulmonary arteriovenous fistulae (PAVFs) are asymptomatic and are often diagnosed incidentally during routine imaging. However, the classic clinical triad of exertional dyspnoea, cyanosis, and digital clubbing can prompt further investigation, leading to early detection. Symptoms typically manifest in the third decade of life, though they may appear much earlier. In patients with hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu syndrome Weber disease, PAVFs may present with a range of symptoms beyond respiratory complaints. Common manifestations include recurrent epistaxis, haematuria, melena, haemoptysis, or neurological symptoms such as headache, vertigo, numbness, paresis, syncope, confusion, and dysphagia, reflecting potential cerebral involvement.[6] The diagnosis of pulmonary arteriovenous malformations (PAVMs) can be made using various imaging and functional modalities, including pulmonary function tests (PFTs), chest radiography (CXR), transthoracic contrast echocardiography (TTCE), radionuclide lung perfusion scanning, computed tomography (CT), and three- dimensional magnetic resonance (MR) angiography. Chest radiography is often the initial diagnostic tool, with 98% of patients showing abnormalities, typically a well-demarcated lobulated mass, most commonly in the lower lobes.[4] Oxygenation is frequently impaired in PAVM patients, as demonstrated in a review of 349 cases, where 32% had an arterial oxygen saturation (SaO_2) below 76%,23% between 76-85%, 26% between 86-90%, 14% between 91-95%, and only 6% above 96%.[3] TTCE is a highly sensitive , minimally invasive screening tool for PAVMs,with a diagnostic sensitivity of up to 97% and a negative predictive value of 99%. It is particularly useful in evaluating right-to-left shunts, whether cardiac or intrapulmonary.
However, its utility is sometimes limited by cost, availability, and the detection of clinically insignificant shunts. CT pulmonary angiography remains the gold standard for PAVM diagnosis, offering rapid scanning, high- resolution imaging, lesion characterisation, treatment planning, and follow-up assessment. Three- dimensional contrast MR angiography is occasionally used for detailed visualisation of thoracic vascular structures.[4]

Treatment options for PAVMs include percutaneous image-guided embolisation, surgical resection, and hormonal therapy. Embolisation is the preferred and most effective treatment, involving occlusion of the feeding artery using coils or balloons. It is recommended for symptomatic PAVMs or lesions larger than 3 mm, particularly if associated with symptoms. Follow-up imaging is essential to confirm successful closure. Surgical excision is considered for patients in whom embolisation fails or those with life-threatening pulmonary haemorrhage due to PAVM rupture. In diffuse cases, lung transplantation may be required. In patients with PAVMs associated with symptomatic hereditary hemorrhagic telangiectasia (HHT), hormonal therapy with systemic oestrogen-progesterone has been utilised. Additionally, anticoagulation therapy is recommended for all patients with PAVMs to reduce thromboembolic complications.[1] Pulmonary

arteriovenous malformations (PAVMs) generally do not resolve spontaneously and tend to remain stable, although approximately 25% may enlarge by 0.2-0.3 mm per year. Their natural history carries significant morbidity and mortality, with untreated cases having a mortality rate as high as 50% compared to just 3% with treatment, largely due to complications such as stroke and brain abscess. The risk of rupture increases during the second and third trimesters of pregnancy, highlighting the need for vigilant management during this period. Prophylactic antibiotics are recommended for patients with PAVMs undergoing surgical or dental procedures to reduce the risk of septic emboli. Given the potential for growth over time, even in treated cases, long-term follow-up is essential, typically every five years, to monitor for the enlargement of small PAVMs that might eventually pose a risk for paradoxical embolisation and stroke.[1]

Furthermore, literature indicates that recanalisation or the development of collateral circulation occurs in up to 20% of patients after embolisation, often due to previously unrecognised feeding arteries. These factors underscore the importance of continuous monitoring and comprehensive long-term management to optimise patient prognosis and outcomes.[7]

CONCLUSIONS

Pulmonary arteriovenous malformation should be considered in patients with unexplained dyspnoea or hypoxaemia, as well as in those with a history of stroke or brain abscess. Genetic testing is advised, particularly in families with hereditary hemorrhagic telangiectasia, to identify at-risk individuals and allow for proactive screening. Without treatment, PAVMs can result in serious complications, including stroke, brain abscess, and life-threatening pulmonary haemorrhage. Treatment decisions are guided by the patient's symptoms, the size of the feeding artery, and individual tolerance, with options ranging from embolisation to surgical intervention. Ultimately, a patient-centred approach that emphasises collaboration and coordinated care is essential in managing this rare vascular condition.

REFERENCES

1. Danyalian, A., Sankari, A., & Hernandez, F. (2024). Pulmonary arteriovenous malformation. In StatPearls [Internet]. StatPearls Publishing.
2. Pollak JS, Saluja S, Thabet A, Henderson KJ, Denbow N, White Jr RI: Clinical and anatomic outcomes after embolotherapy of pulmonary arteriovenous malformations. J Vasc Interv Radiol . 2006 Jan;17(1):35-44; quiz 45.
3. Gossage JR, Kanj G. Pulmonary arteriovenous malformations: a state of the art review . American journal of respiratory and critical care medicine. 1998;1:643-61.
4. Kaufman CS, McDonald J, Balch H, Whitehead K: Pulmonary arteriovenous malformations: what the interventional radiologist should know. Semin Intervent Radiol. 2022;39(3):261–270.

5. Meek ME, Meek JC, Beheshti MV: Management of pulmonary arteriovenous malformations. InSeminars in interventional radiology. 2011;1:024-031.
6. Prager RL, Laws KH, Bender Jr HW: Arteriovenous fistula of the lung. The . Annals of thoracic surgery. 1983;1:231-9.
7. Li T, Hao P, Lv X, Tian H: Treatment of a rare recurrent pulmonary arteriovenous malformation after multiple embolization by robotic-assisted thoracoscopic surgery. J Surg Case Rep . 2022 Feb 2;2022(2):rjac006.