

Pulmonary arteriovenous malformations: A case of missed diagnosis in a neonate

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SUMMARY

Pulmonary arteriovenous malformation (PAVM) is a congenital vascular abnormality that can cause persistent cyanosis in children. PAVMs can go undetected till adulthood; however, there have been several neonatal cases reported over the years. This case report describes a classical manifestation of a child with isolated PAVM whose diagnosis was likely missed during the neonatal period. A high level of clinical awareness of this condition is crucial as early diagnosis and appropriate treatment can prevent life-threatening complications and mortality. Diagnosis was confirmed by computed tomography pulmonary angiography and percutaneous transcatheter embolotherapy resulted in complete resolution of symptoms.

INTRODUCTION

Pulmonary arteriovenous malformations (PAVMs) are structurally abnormal vessels that provide communication between the branches of pulmonary arteries and pulmonary veins.¹ PAVM creates an intrapulmonary right-to-left shunt and allows the systemic venous blood to bypass the normal pulmonary capillary bed, which is crucial for gas exchange and filtration process.¹ Majority of cases presenting with PAVM eventually manifest with hereditary haemorrhagic telangiectasia; however, in a small group of patients, it appears to be sporadic.² PAVM is most commonly congenital in origin, but the condition often remains silent until adulthood, suggesting that the gradual enlargement of the PAVM occurs with increasing age.³ Although PAVM is a rare clinical problem in neonate, it can be potentially fatal. Therefore, a thorough assessment is essential for early diagnosis.⁴

CASE REPORT

A three-year-old girl presented with a one-month history of worsening cyanosis and deteriorating effort tolerance. She was born prematurely at 33 weeks of gestation at a district hospital and reported to have respiratory distress at birth requiring continuous positive airway pressure (CPAP) support. Chest radiographs revealed a homogenous opacity over the right upper and middle lobes (Figure 1), and she was

treated for congenital pneumonia with intravenous antibiotics. She responded well and successfully weaned off respiratory support by day 8 of life. As clinical improvement pursued, no repeat chest radiograph was done. She was referred to our centre for further evaluation of systolic murmur at 1 month of age, and echocardiography showed a patent ductus arteriosus with left pulmonary artery stenosis. However, no chest radiography was performed. She subsequently defaulted follow-up, and parents claimed that she was well with no history of fever, cough, recurrent respiratory infections, or cyanosis. There was also no history of epistaxis, haemoptysis, or skin telangiectasia. Parents denied any family history of cardiac condition or hereditary haemorrhagic telangiectasia.

Physical examination revealed that the child was cyanosed with digital clubbing and was in respiratory distress. Transcutaneous oxygen saturation was 69% before dropping to 55% when she raised from supine to upright position despite being put on supplemental oxygen 3 L/min via nasal prong. Her mucous membrane was normal. Cardiovascular examination was unremarkable; however, auscultation of the lungs revealed a bruit over the right middle zone that was more prominent at sitting position. She had polycythaemia with haemoglobin level of 15.7 g/dL. The chest radiograph showed a homogenous opacity over the right upper and middle lobes of the lungs (Figure 2). A bedside echocardiogram demonstrated a structurally normal heart with no evidence of pulmonary hypertension. However, the transthoracic contrast echocardiography (TTCE) was highly suggestive of PAVM. The diagnosis was confirmed by computed tomography pulmonary angiography (CTPA), which showed a multiple complex PAVM, predominantly in the anterior segment of the right upper, middle, and lower lobes.

The PAVMs were successfully embolised via a percutaneous transcatheter. Post-embolotherapy, no significant residual flow was seen through the PAVMs. Percutaneous pulse oximetry saturation on room air increased immediately from 70% to 100%. Her postoperative course was uneventful, and she was discharged after four days. Her exercise tolerance improved, and she remained asymptomatic with good weight gain during the three-month follow-up.

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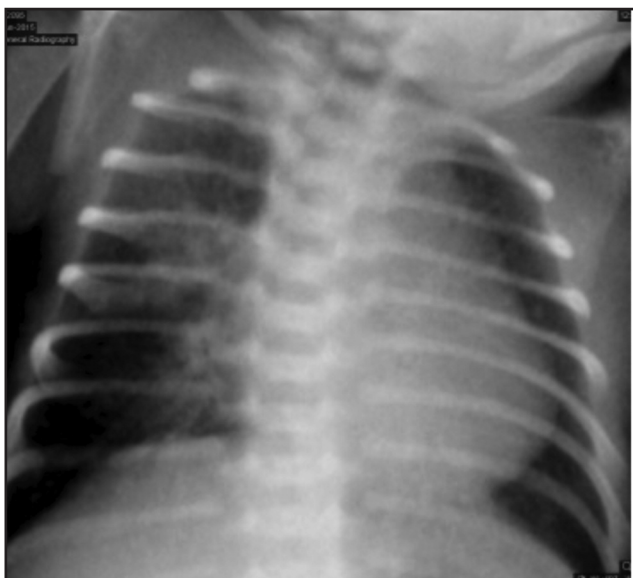


Fig. 1: Chest radiograph at birth showed a homogenous opacity over right upper and middle lobes.

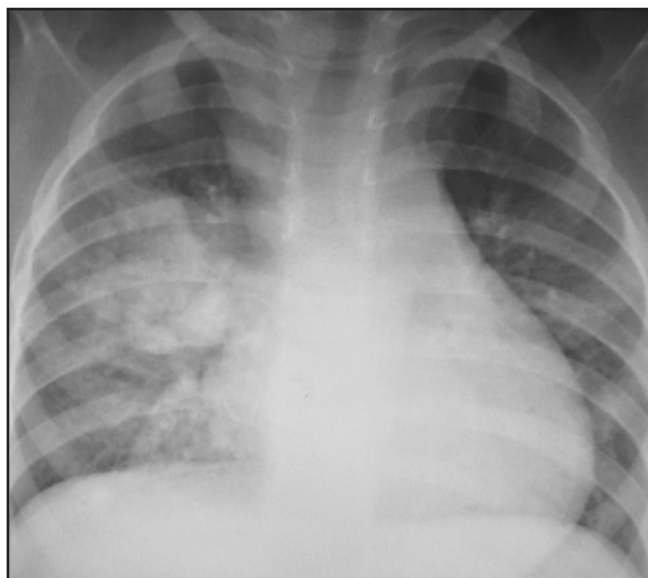


Fig. 2: Chest radiograph taken at three years of age showing similar opacities.

DISCUSSION

Over the past decades, PAVM has evolved from a rare curiosity to not an uncommon anomaly with an estimated prevalence of 1 in 2600 individuals.⁵ Numerous reviews and reports have documented the classical presentation and natural history of PAVM in adults and children; however, manifestation in the neonatal period is rare.⁴ Although majority of PAVM is congenital in origin, in most cases, it remains silent until adulthood with only 10% of cases detected among children.⁶

Our patient presented with the classic triad of dyspnoea, cyanosis, and clubbing accompanied by pulmonary bruit and orthodeoxia, the hallmark of PAVM. The latter is the result of worsening ventilation/perfusion mismatch due to gravitationally induced redistribution of blood flow to the lung bases where the vascular malformations are located when the patient is in an upright position.⁵ Our suspicion was supported by the classical appearance of a sharp, well-defined nodule or mass of uniform density on the chest radiograph as well as the appearance of microbubbles seen in the left side of the heart through transthoracic contrast echocardiography following the injection of agitated saline. The latter technique has high sensitivity (100%) but lower specificity (67%–91%).³ PAVM in our case was diagnosed via the gold standard imaging study that clearly delineates the exact anatomy of the anomaly. With CTPA, PAVM can be accurately predicted in 95% of cases, thus making it a necessary investigation for therapeutic planning.³

Interestingly, a retrospective review of our patient's chest radiograph suggests that she may have had the abnormality since neonatal period; however, the diagnosis was missed as she showed significant clinical improvement with supportive therapy and no documented cyanosis. Thus, no repeat imaging study performed to look for resolution of the radiological evidence. Furthermore, PAVM is not routinely

appreciated during standard echocardiogram and may be deceptive.⁴ Missed diagnosis of PAVM in the neonatal period is not uncommon.^{4,6} The chest radiograph findings were often misinterpreted as pneumonia or pulmonary hypertension. Fortunately, our patient did not exhibit a more devastating or life-threatening manifestation such as stroke, cerebral abscess, haemoptysis, or haemothorax.^{4,7} These are related to either loss of filtration function of the lung, leading to paradoxical systemic embolisation, or rupture of the abnormal vascular structure.

Our patient was successfully treated with percutaneous transcatheter embolotherapy. The main aim of the treatment is early occlusion of the fistula to alleviate hypoxia and to prevent any serious thromboembolic complications owing to paradoxical embolisation. Although it is highly effective and safe, embolotherapy may be potentially complicated by haemorrhage, arrhythmias, air embolism, or migration of the embolic agent.⁷ In 5%–15% of cases, recanalisation after re-embolisation is warranted. Surgical intervention is reserved for cases with serious bleeding, lesions not amendable to embolotherapy, or failure of embolotherapy.⁷

CONCLUSION

Although PAVMs are rarely manifested in the neonatal period, the unspecific chest radiographs findings should raise suspicion. A high level of awareness is crucial as early diagnosis and appropriate treatment can prevent life-threatening complications and avert death.

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