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Case Report

Paediatrics Section

Pulmonary Arteriovenous Malformations in a 18-months-old Child: A Case Report

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ABSTRACT

Pulmonary Arteriovenous Malformation (PAVM) is a cardiovascular anomaly in which an anatomic communication between the pulmonary artery and pulmonary vein is formed, resulting in an additional cardiac shunt from right to left side. PAVM is a rare disease, reported more in females, but in newborns comparatively higher in males. Cases can vary from asymptomatic to symptomatic such as having dyspnoea, hypoxia, cyanosis, neurological symptoms. They are mostly congenital having association with Hereditary Haemorrhagic Telangiectasia (HHT). Herby, author present a case of 18-months-old female admitted with peripheral cyanosis in the Paediatric Department. Her saturation level was 76%, after admission she received three doses of salbutamol nebulisation and was kept on 3 litre oxygen until her saturation increased to around 95%. On auscultation, a murmur of grade 2/6 heard in the left axilla with clear lung fields. Chest X-ray detected a prominent lobulated opacity in the left sided lung adjacent to the heart border. An echocardiogram was done which turned out normal. Thoracic Computed Tomography (CT) scan with contrast showed a vascular malformation in the apical region of the left lower lobe. Pulmonary angiography showed multiple vascular lesions in the peripheries which was consistent with arteriovenous malformation. The child was diagnosed with PAVM. A transcatheter embolisation and coil closure of AVM was performed and postprocedure the child was stable.

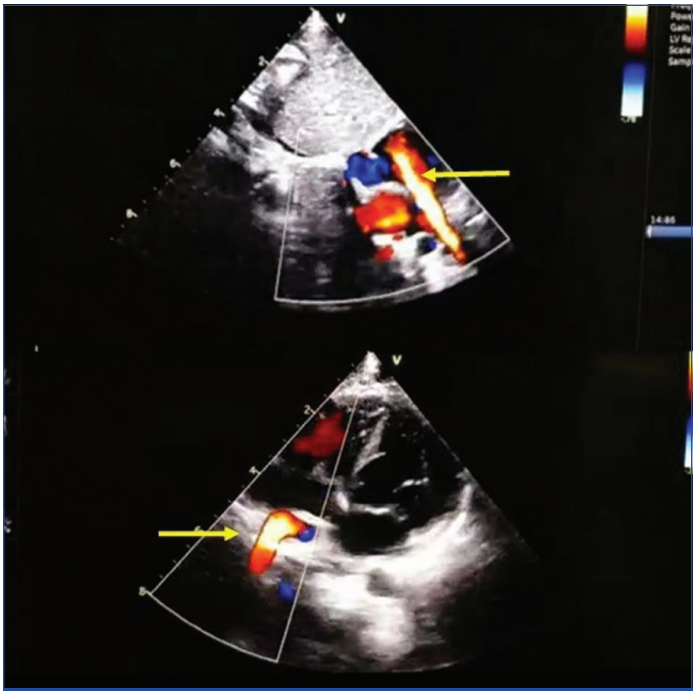
Keywords: Cardiovascular anomaly, Hereditary haemorrhagic telangiectasia, Pulmonary angiography, Peripheral cyanosis

CASE REPORT

An 18-months-old female child was presented to the Paediatric Emergency Unit with complaint of peripheral cyanosis since two to three days. She was delivered by elective caesarean section at 38 weeks' term and she was absolutely in normal condition at discharge. Her delivery was uneventful and she was on exclusive breastfeeding. She was treated for bronchopneumonia at seven months with intravenous (i.v.) azithromycin 5 mg/kg once daily for three days, i.v. methylprednisolone 1 mg/kg/day, and supplemental oxygen and was discharged after seven days. She had frequent visits to the emergency for recurrent chest infection which was treated as bronchiolitis. There was no significant family history.

At admission the pulse oximetry showed a saturation of 76% on room air, heart rate of 135 beats/min, a respiratory rate of 40 breaths/min and blood pressure of 108/61 mmHg. The clinical examination revealed an active well perfused child, afebrile, not in distress. On auscultation, there was continuous murmur 2/6 heard in the left axilla with clear lung fields. Haemoglobin level was 16.6 gm/dL with haematocrit of 47.2%. Capillary blood gas analysis and other laboratory findings were normal. Chest X-ray detected a prominent lobulated opacity in the left sided lung adjacent to the heart border. Echocardiogram was normal [Table/Fig-1]. A contrast thoracic Computed Tomography (CT) scan showed a vascular malformation in the apical region of the left lower lobe [Table/Fig-2]. Pulmonary angiography showed multiple vascular lesions in the peripheries which was consistent with arteriovenous malformation [Table/Fig-3,4]. She was diagnosed with Pulmonary Arteriovenous Malformation (PAVM). A transcatheter embolisation and coil closure of the AVM was performed at another facility (Al Qassimi Hospital, UAE), the very next day following hospitalisation.

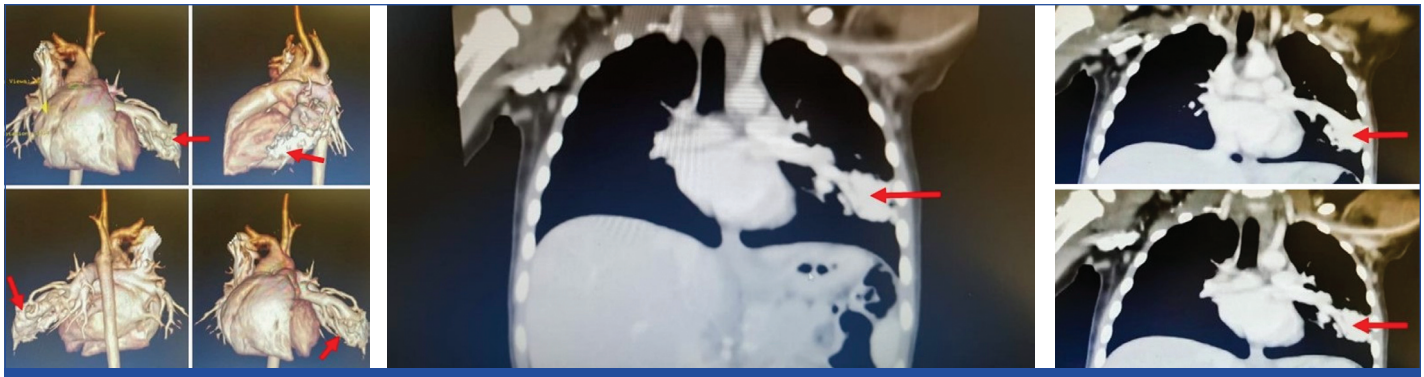
Preoperative evaluation was within normal limits. Under general anesthesia, a 6F catheter was used to access the right femoral vein, followed by a 4F catheter to reach the right femoral artery. The Refractory Right Ventricle (RRV) pressure was 18 mmHg, the End Diastolic Pressure (EDP) was 8 mmHg, and the pulmonary artery pressure was 20/13 mmHg.



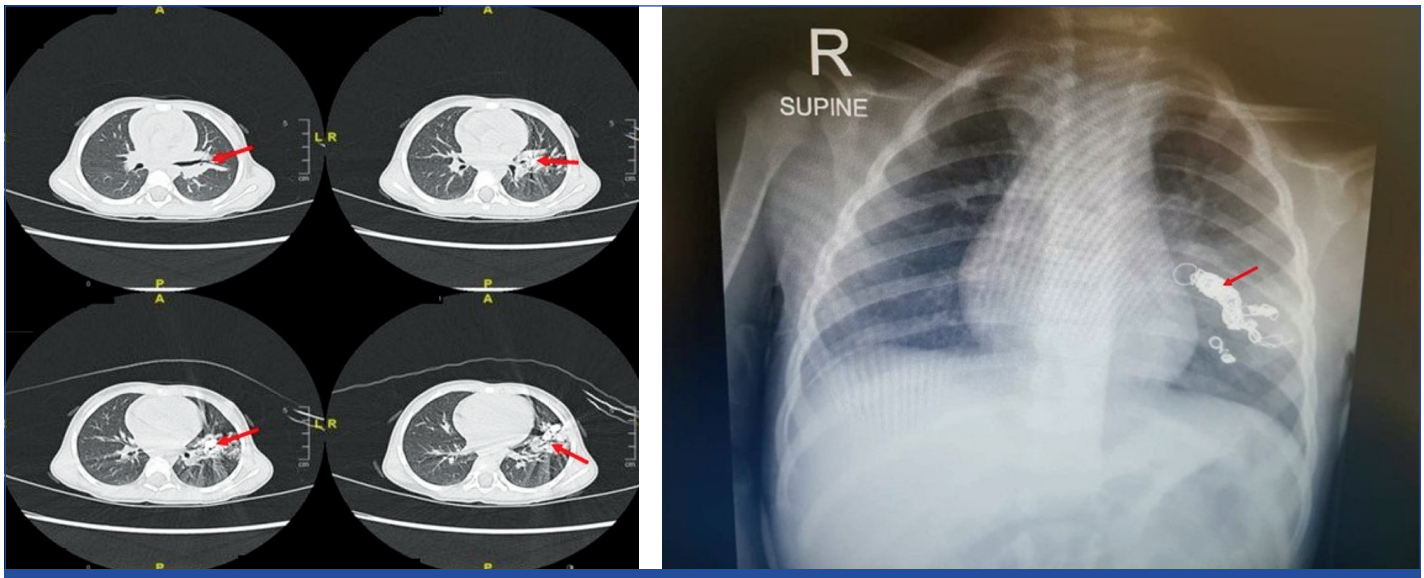
[Table/Fig-1]: Echocardiogram showing normal intracardiac structure and function.

Left pulmonary angiogram showed a massive malformation connecting the left lower lobar artery with left upper pulmonary vein to Left Atrium (LA). The left upper vein dilated due to a large venous return [Table/Fig-5]. There were multiple feeding arteries to the pulmonary vein. Two small feeding arteries were closed using a 6 mm by 20 cm interlock coil, followed by a 4 mm by 8 cm coil. Then a Helical coil of 8 mm by 30 cm was used. Three interlock coils- one 12 mm by 30 cm, 6 mm by 20 cm, and 5 mm by 5 mm were used to seal the major feeding artery with various branches. The AVM was removed through an 8F long sheath.

There was an estimated blood loss of 25 cc. A total of 2000 units of heparin were utilised. There was no major complication. The saturation



[Table/Fig-2]: Volume Rendering Computed Tomography showing consistent with vascular malformation in the apical region of the left lower lobe and dilated pulmonary vein draining to the left atrium- Pulmonary arteriovenous malformation (red arrow). **[Table/Fig-3]:** Pulmonary CT angiography- shows a vascular malformation (red arrow) in the peripheries which was consistent with pulmonary arteriovenous malformation. **[Table/Fig-4]:** Pulmonary CT angiography showing intense contrast involvement consistent with a PAVM and draining vein (red arrow). (Images from left to right)



[Table/Fig-5]: Computed tomography angiography chest without contrast. The patient status was postoperative intervention for pulmonary AVM. It shows (red arrow) surgical clips with resultant metallic artifacts noted at the operative site. Residual serpiginous vascular channels are still noted at the operative bed. Both lungs are clear from active parenchymal lesions. **[Table/Fig-6]:** Follow-up anterio-posterior chest X-ray following the application of interlock coils. Both lungs are clear. (Images from left to right)

increased from 76% to 95%. Postoperative the child was stable. She was discharged after three days and was followed-up every week for two months, then every three months and no complications were observed during the follow-up period [Table/Fig-6].

DISCUSSION

Pulmonary arteriovenous malformations is a rare disease in the United Arab Emirates. The incidence of PAVM is about 1:50,000 cases. Its common in women but a male predominance is seen in newborns [1]. It was first discovered during an autopsy in 1897 and first diagnosed during life in 1939 [2].

The pathogenesis of PAVM is still unknown. Around 70% of cases of PAVM have a strong association with Hereditary Haemorrhagic Telangiectasia (HHT) but the case presented in this report did not have any such association [3]. PAVMs can be the simple type (80% of the cases) or complex type (20%). PAVMs are classified into five different groups:

Group 1: consists of multiple small arteriovenous fistulas without aneurysm.

Group 2 and 3: consists of large arteriovenous aneurysm.

Group 4: consists of large venous aneurysm with systemic artery communication and without fistula subsequently.

Group 5: has anomalous venous drainage with fistulas.

PAVM are usually in the lower and subpleural lung zones [4].

Pulmonary arteriovenous malformations include clinical manifestation and radiological features that are similar to a variety of different diseases. When evaluating a patient with PAVM, keep the following differentials in mind: Pulmonary infarct, pulmonary artery aneurysm, pulmonary contusion, pulmonary hamartoma, pulmonary varices, pulmonary bronchocele, primary lung malignancy, carcinoid tumor, calcified or infectious granuloma [4-6].

Studies have demonstrated that 80% of HHT families have a disease-causing mutation in two genes- the Activin Receptor-like kinase gene (ACVRL1) found on chromosome 12 which, codes activin receptor-like kinase 1 protein or the Endoglin Gene (ENG) found on chromosome 9 that codes endoglin protein. Researches show mutations of the gene represented as Mother Against Decapentaplegic Homologue 4 (MADH4), coding for SMAD4 protein, was found in 1% to 3% of HHT patients with an uncommon combination of mixed familial Juvenile Polyposis (JP) and HHT [7].

Cases range from asymptomatic to more serious with congestive heart failure, cyanosis and respiratory failure. Symptoms correlates with the size of the lesion [6]. Epistaxis is common due to the bleeding from mucosal telangiectasia [8]. Dyspnoea can be seen in patients with giant or multiple PAVM, and in patients having cyanosis and clubbing. Some have platypnea, indicating reduced blood flow in supine position [8]. Approximately 20% of patients have bleeding in the gastrointestinal tract [9]. Some uncommon symptoms like headache, chest pain, cough, vertigo, dysarthria can be seen, correlated with conditions like polycythemia, hypoxemia, and stroke [10].

Most common physical finding in PAVM is superficial telangiectasia associated with HHT. These are ruby colored papule, slightly rounded, 1-3 mm, sharply demarcated with few dendritic projections from their surrounding skin lesions extensively distributed on the face, mouth,

chest, upper extremities [10]. Murmurs or bruits, which is best heard during inspiration, are present in 46% of patients whereas cyanosis and clubbing is seen in 34%. The PAVM triad of dyspnoea, cyanosis, and clubbing is seen in 10% of patients [11].

PAVM can be diagnosed using chest X-ray, CT, contrast echocardiography, pulmonary angiography, radionuclide perfusion lung scanning and magnetic resonance imaging. Chest X-ray can detect and screen for PAVMs, but it does not detect small lesions. PAVM appears as rounded, well-defined nodular lesions of varying sizes, from 1 to 2 cm in diameter with branching afferents feeding and dilated efferent vessels. Complex PAVMs involves entire lung segments, appears as an area of generalised increased opacity, marked vascular prominence, and the absence of discrete lung nodules [7]. A non contrast CT shows a homogeneous, well-circumscribed, non calcified nodule or as a serpiginous mass connected with blood vessels [8].

Intravenous contrast injection reveals enhancement of the feeding artery, the aneurysm, and the draining vein on early-phase sequences, which is necessary to diagnose PAVM [7]. Multiple Regression Analysis (MRA) analyses the pulmonary arterial and venous enhancement kinetics to assess PAVM patency.

Transthoracic Contrast Echocardiography (TTCE) is a screening test for PAVMs. Intrapulmonary right-to-left shunt can be detected using TTCE with agitated saline contrast. The echocardiographic contrast when injected into a peripheral vein appears as a white cloud of echoes against a black anechoic background. It is normal for these micro-cavitation's to be filtered out by the pulmonary capillary bed, their occurrence in the heart's left side indicates intracardiac Right-to-Left Shunting (RLS), while a delayed emergence is indicative of intrapulmonary RLS [10]. Appearance of bubbles in the left atrium after injection of agitated saline solution is a positive Echocardiogram (Echo) [11].

Radionuclide perfusion lung is not used routinely because it does not provide much detail about the anatomical features and a positive result does not confirm the diagnosis of PAVM but a negative result definitely excludes the diagnosis [11]. Risk of morbidity in untreated patients is 50% and 3% in treated ones. Treatment is recommended for all patients with lesions less than 2 cm in diameter on chest radiography regardless of the symptoms [11]. The goal is to prevent neurological complications, hypoxia and high-output heart failure [11]. The mainstay of treatment is percutaneous embolisation first done in 1978 [11]. Metallic coils and balloons are used for embolisation with a success rate of >95%, although recanalization can occur [12].

CONCLUSION(S)

Pulmonary arteriovenous malformations are rare with a wide spectrum of symptoms. For a final diagnosis, a thorough history taking and a high level of suspicion are critical. A chest X-ray generally suggests PAVM, and CT scan or pulmonary angiography is usually required to confirm the diagnosis. Embolotherapy is safe and effective standard treatment for PAVMs. Patients should be followed-up on a routine basis.

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