

## Diffuse Pulmonary Arteriovenous Malformation in Children: Essential Value of Contrast Echocardiography in Diagnosis

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### Abstract

Pulmonary arteriovenous malformations (PAVM) are abnormal communications between pulmonary arteries and pulmonary veins, and most commonly congenital in nature. Although rare, it is an important consideration in cyanotic patients of unknown cause. We report 3 cases with diffuse PAVM in children with different clinical manifestations and initial diagnosis was made by transthoracic contrast echocardiogram.

Transthoracic contrast echocardiography (TTCE) is valuable as initial diagnostic tools for diffuse PAVM. Pulmonary angiography should be reserved for therapeutic purposes for PAVM rather than diagnostic.

**Key Words:** Children, Pulmonary arteriovenous malformation, Transthoracic contrast echocardiography.

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## 1- INTRODUCTION

Pulmonary arteriovenous malformations (PAVM) are abnormal pulmonary blood vessels in which there is a direct connection between arterial and venous vessels without intervening capillaries. As a result of this abnormality, PAVM can present with a wide spectrum of clinical manifestations. These include hypoxemia, life threatening haemorrhage, hemothorax, right to left shunting leading to paradoxical embolisation, manifested as stroke and brain abscess (1, 2).

Approximately 85% of PAVM are simple. Diffuse PAVM are of smaller percentage, in which there is disseminated involvement of multiple pulmonary segments (3, 4). The purpose of this case series was to describe the variable clinical presentation and essential value of transthoracic contrast echocardiography (TTCE) as an initial diagnostic test in children with diffuse PAVM.

## 2- CASE SERIES REPORT

### 2-1. Case. 1

A one-year-old boy with history of recurrent respiratory infections since 1-month-old and incidental findings of dextrocardia and situs inversus in screening echocardiography. He presented at 10-month-old with 2 weeks history of fever and cough with associated respiratory distress requiring intubation and ventilator support.

He was positive for pulmonary tuberculosis and treatment was started. His oxygen saturations remain in the low 80s despite of his clinical improvement. He was cyanosed and clubbed. A hyperoxia test showed the Partial Pressure of Oxygen (PaO<sub>2</sub>) of 62.2mmHg with 100% oxygen and echocardiography showed dextrocardia, normal cardiac structure with positive contrast echocardiography which raised the suspicion of PAVM (**Figure.1**). A high resolution computed tomography

(HRCT) of the thorax showed no obvious intra-pulmonary shunting or bronchiectatic changes. Subsequent pulmonary angiography confirmed diffuse PAVM with no evidence of pulmonary hypertension.

### 2-2. Case. 2

A eleven-year-old girl with history of right cerebral abscess undergone a burr hole and aspiration. Three years later she had recurrent abscess at left temporoparietal region. She had a surgical drainage and 6 weeks of intravenous antibiotics.

She had central cyanosis, clubbing, oxygen saturation 85 to 88% and upper motor neuron lesion signs. Chest X-ray, High Resolution CT of the Chest (HRCT) thorax was normal and echocardiography showed a normal cardiac structure. Transthoracic contrast echocardiography (TTCE) is suggestive of PAVM. Pulmonary angiography confirmed diffuse PAVM.

### 2-3. Case. 3

A four-year-old girl with history of recurrent epistaxis and increasing cyanosis since two years old. She also had lethargy and reduced effort tolerance. On examination, he had soft dysmorphic features, small for age and no mucocutaneous telangiectasia. Her heart rate was 120/min, blood pressure 87/60mmHg. She had orthodeoxia with her oxygen saturation 84% during lying, and 74% on standing.

Both respiratory and cardiovascular examination were unremarkable. Hemoglobin level was 148g/L and no thrombocytopenia. A TTCE suggestive of PAVM. HRCT thorax was reported as normal. Diffuse PAVM was confirmed with pulmonary angiography (**Figures 2a, 2b**) and no evidence of pulmonary hypertension with a mean Pulmonary Artery (PA) pressure 13mmHg.



**Fig.1:** Positive transthoracic contrast echocardiography in PAVM.



**Fig. 2(a):** Diffuse PAVM right lung.



**Fig. 2(b):** Diffuse PAVM left lung.

### 3- DISCUSSION

PAVMs are rarely diffuse. Faughnan et al., defined diffuse PAVM as every segment of one or more lobes diffusely involved by small PAVM (4). Though small, it is an important subset of PAVM as it caused significant morbidity and mortality. In our series, suspicion of PAVM was thought from symptoms of cyanosis in 2 patients and complication of cerebral abscess in 1 patient. All three patients have no respiratory and cardiovascular causes to explain the underlying hypoxemia. High suspicion of PAVMs in the patients was confirmed by a positive agitated saline contrast transthoracic echocardiography. In all patients baseline HRCT was normal.

All three patients do not met diagnosis of hereditary hemorrhagic telangiectasia (HHT) using the International Clinical Diagnostic (Curacao) Criteria (5). Only patient in case 3 had orthodeoxia which is known to be more common in HHT related PAVM. However it is possible that patient in our series has unrecognized HHT as the clinical expression of HHT is age related (6). Approximately 80 to 90% of patients presenting with PAVMs eventually had HHT (7, 8).

Presenting symptoms in all the three patients which includes hypoxemia and neurological symptoms were correlated with diffuse PAVM. Other most frequently symptoms in diffuse PAVM patients were hemoptysis (4, 9). TTCE is a rapid, simple and minimally invasive examination used as an initial PAVMs screening specifically looking for intrapulmonary shunting. After intravenous injection of agitated saline solution, echoic microbubbles can be easily depicted in the left cardiac cavities after three to five cardiac beats, thus suggesting the presence of a right-to-left pulmonary shunt (whereas microbubbles that are immediately visible after injection may suggest an intracardiac shunt). Reported diagnostic sensitivity of TTCE is

high (up to 97%), and the negative predictive value is 99% in published series (10). In PAVM typically the microbubbles arising from the pulmonary veins before appearing in the left atrium. The optimal echocardiography view is either from a standard apical four-chamber or subcostal coronal view. Diagnostic testing to identify intrapulmonary shunt, other than TTCE, are Chest CT scan and pulmonary angiography. Chest CT scans as an additional test in characterizing PAVMs in patients with positive contrast echocardiography may miss a diffuse PAVMs. We thought this could be due to microscopic PAVM and the diffuse involvement. HRCT in all three patients were reported as normal, and no intrapulmonary shunting was seen in both arterial and venous phase.

Pulmonary angiography, which was initially done for therapeutic purposes, confirmed the diffuse PAVM in all patients. There were no hepatic shunt and no pulmonary hypertension. Due to the diffuse PAVM, management was very challenging. No transcatheter embolotherapy was offered to all patients. In pediatric patients, transthoracic contrast echocardiography is a safe test with no exposure to ionizing radiation. In developing countries with limited resources and financial issues, a simple contrast echocardiography for diagnosis and reserving a pulmonary angiogram for characterization and intervention are probably a more cost effective way to screen and treat children with PAVMs.

Therefore a screening policy for PAVMs based on TTCE would decrease biological risk due to ionizing radiations. An economic benefit can also be considered for the use of TTCE as first line examination for PAVM screening wherever TTCE is cheaper than CT (11). Unlike most patients with focal PAVM who usually successfully treated by transcatheter embolotherapy (TCE),

management of patients with diffuse PAVM are more difficult and approach to treatment, are ill defined.

#### 4- CONCLUSION

PAVM is an important diagnosis to consider in persistent hypoxemia in the absence of respiratory and cardiac disease. A normal HRCT thorax may not rule out diffuse PAVM. Hence, transthoracic contrast echocardiography is an extremely safe, affordable, sensitive and non-invasive test for the detection of PAVM. Pulmonary angiogram should be reserved for characterization of PAVM and treatment.

**5- CONFLICT OF INTEREST:** None.

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