Pulmonary Arteriovenous Malformation in Childhood: A Case Report

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Abstract:

The pulmonary arteriovenous malformation is an abnormal communication between pulmonary artery and pulmonary vein or between a bronchial artery and pulmonary vein. A 4 year old female child presented with dyspnea on exertion,easy fatigability, bluish discoloration of nail bed and weight loss since last 1 year. On examination, patient was undernourished with presence of central cyanosis, conjunctival plethora and grade 2 clubbing of 4 limbs. Systemic examination was normal. The hemogram showed polycythemia with rest of the parameters were within normal range. The chest X ray was suggestive of right lower lobe opacity and CECT Thorax suggestive of segmental opacity with air bronchogram in basal segment of right- lower lobe possibility of infective etiology. CT pulmonary angiogram was suggestive of large pulmonary arterio-venous malformation in right lower lobe which was plugged with endovascular plug and complete obliteration was achieved. There was dramatic improvement in dyspnea and cyanosis.

Keywords : Children, Endovascular Plug, Pulmonary Arteriovenous Malformation

Introduction:

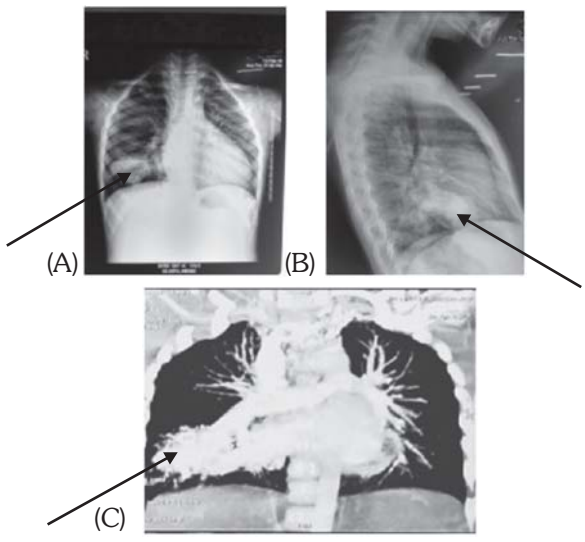
Pulmonary Arterio-venous malformation (PAVM) is rare anomaly with common presenting complaints like dyspnea on exertion, fatigue, nasal bleeding, bluish discoloration of lips & nail beds and hemoptysis. These arteriovenous malformations are communication between pulmonary artery and pulmonary vein or between a bronchial artery and the pulmonary vein. [1] Only 10% of the cases of pulmonary arteriovenous malformations are identified in infancy or childhood. It can causes severe morbidity & mortality by occurrence of complications like cerebral vascular accident, brain abscess, hemothorax, life-threatening hemoptysis if not treated early and adequately. [2]

Case Report:

A 4 year old female child presented to paediatric emergency room with complaints of dyspnea on exertion, easy fatiguability, bluish discoloration of nail beds and weight loss since last 1 year. She had no complaint of cough, fever, swelling over body, chest pain or squatting episode. Patient had history of 1 episode of generalised tonic clonic type of convulsion before 6 months. There was no past or family history of congenital heart disease, pulmonary disease, malignancy, epilepsy and tuberculosis.

On examination, patient was undernourished with presence of central cyanosis, conjunctival plethora and grade 2 clubbing of 4 limbs. On auscultation bilateral breath sounds were present without any crepitation or rhonchi; heart sounds were also normal without any audible murmur. Per Abdomen and nervous system examination was normal. The oxygen saturation of all four limbs was between 65-70 % with little improvement Oxygen supplementation. On the basis of above findings Cyanotic heart disease was suspected and child was admitted for further evaluation.

Figure 1 : (A & B) Chest X - ray showing right lower zone opacity (C) CT pulmonary angiogram showing large pulmonary arterio-venous malformation in right lower lobe.



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Basic investigation revealed, Hb-18 gm/dl (normal Hb11-15 gm/dl) with hematocrit of 55% (normal value 36-45%) while renal function test, liver function test were normal; chest X ray was suggestive of right lower zone opacity and 2D Echocardiography was normal. MRI brain with contrast and angiogram was normal which was done to rule out cerebral cause of seizure. On the basis of the above reports, S.erythropoietin level was done (30mIU/L) which was raised (normal 4-20 mIU/ml). Bone marrow biopsy showed normal functioning marrow. CECT Thorax was suggestive of segmental opacity with air bronchogram in basal segment of right lower lobe possibility of infective etiology. As patient had no complaint of fever and cough & on investigation total WBC count along with differential count was within normal range, thus infective aetiology was less likely. Then, we went for CT pulmonary angiogram which was suggestive of large pulmonary arterio-venous malformation in right lower lobe that was formed by branches of descending branch of right pulmonary artery and drained by right inferior pulmonary vein into left atrium. It was Idiopathic type of congenital pulmonary arteriovenous malformations as it was not associated with epistaxis or superficial telangiectases (Hereditary hemorrhagic telangiectia), no history of inheritance or secondary infection or cardiothoracic-vascular intervention, fewer physical findings and on investigation lesion was isolated and static in size.

The child was planned for embolization therapy; the descending branch of right pulmonary artery collateral was plugged with Amplatzer vascular plug –II size 16mm and venous drainage of collaterals communication was plugged with Amplatzer vascular plug –II size 22mm. The post-operative angiogram showed no flow across the fistula and child was put on low dose aspirin therapy. Following the above intervention, child's dyspnea and cyanosis improved gradually. On follow up at 3 months, child was asymptomatic with SpO2 of 92-95 % in all four limbs.

Discussion :

Pulmonary arteriovenous malformation (PAVM) was first described by T Churton in 1897. In a 1953 study from The Johns Hopkins Hospital, 3 cases of PAVM were detected in 15,000 consecutive autopsies. There is a recognized female predilection with Female to Male ratios ranging from 1.5 to 1.8:1. The estimated incidence is around 2-3 per 100,000. [3]

Anatomically, they are classified into focal, complex and diffuse. Focal type is commonest; has a single feeding segmental artery; it may have multiple subsegmental branches at single segmental level whereas complex types have multiple segmental feeding arteries (~20%). The diffuse type is rare (~5%) have combination of simple and complex AVMs within a diffuse lesion. [4]

Based on etiology, PAVM is of four types, of which most common is HHT [5] (~85%). HHT is an autosomal dominant disorder in which vascular malformations is commonly seen in the skin, nasopharynx, GI tract, lungs, and brain. HHT is a triad of cutaneous telangiectasia, recurrent epistaxis, and a family history of this disorder.[5] Idiopathic congenital pulmonary arteriovenous malformations (~15%) are likely to be single. They are less likely to become enlarged, and successfully treated with embolotherapy. Pulmonary arteriovenous malformations may also be acquired rarely secondary to chronic infections (~ <1%) such as schistosomiasis, actinomycosis, tuberculosis; liver cirrhosis and metastatic thyroid cancer. PAVM may be iatrogenic (~ <1%) due to cardiovascular intervention like Glenn or modified Fontan procedures for congenital cyanotic heart disease. [6]

Endoglin and AVRL-1 genes are implicated in angiogenesis in HHT responsible for pulmonary arteriovenous malformation.[7] In a PAVM, blood bypasses the normal oxygen-exchanging pulmonary capillary bed, returning desaturated blood to the pulmonary veins. whenthe shunting becomes significant, cyanosis results which leads to a compensatory rise in hematocrit and hemoglobin concentration. PAVM are more common in lower lobes which leads to orthodeoxia (accentuation of arterial hypoxemia in erect position, improved by assumption of recumbent position) and platypnea in advanced cases. Bleeding from nose and GI telangiectasias may reduce the hemoglobin in HHT leading to anemia and stroke from paradoxical embolization. Mortality is higher in bilateral, diffuse pulmonary arteriovenous malformations (up to 55%) than the focal variety due to availability of effective therapeutic interventions. [8, 9]

Approximately 70% of PAVM are associated with hereditary hemorrhagic telangiectia (HHT) and conversely, approximately 15 to 35% of HHT patients have PAVM. According to pathologic anatomy in 350 patients with PAVM, it was found that 75% had unilateral disease, 36% had multiple lesions, and half of those with multiple lesions had bilateral disease. [10]

In paediatric age group, few cases have been reported. The age of presentation of PAVM depends on size of lesion, diffuse or isolated, unilateral or bilateral. The

patient presents earlier if size of PAVM is large, diffused or bilaterally located. Most common presentations were dyspnea with or without exertion and cyanosis. Symptoms in early life may vary from being totally absent to severe with cyanosis, congestive heart failure, and even fulminant respiratory failure. [11, 12] The most common complaint in symptomatic patient is epistaxis mostly associated with HHT which was absent in this patient. The 2nd most common complaint is dyspnea. Superficial telangiectases attributable to HHT are the most common and frequently the only—physical finding in patients with PAVM which was absent in this patient. Murmurs or bruits over the site of the PAVM are heard in about 46% of patients. [13] The murmurs are most audible during inspiration, and usually increase on assuming positions which put the PAVM in dependent position.[14] Digital clubbing and cyanosis were seen in only 39 % and 34% of patients, respectively. [13] Despite being reported in textbooks as classic for PAVM, the triad of dyspnea, cyanosis, and clubbing was unequivocally present in only 10% of patients with PAVM in one study. [15]

Symptoms are often insidious, as the arteriovenous malformations slowly enlarge. Dyspnea, especially with exercise, may develop over many years. In severe cases, dyspnea in the upright position (platypnea) & visible cyanosis may be present. Hemoptysis may occur and less commonly seen symptoms are chest pain, cough, headache, tinnitus, dizziness, dysarthria, syncope, vertigo and diplopia which are all due to hypoxemia, polycythemia or paradoxical embolization through malformations (PAVM).

Definite therapy for pulmonary arteriovenous malformations (PAVM) involves therapeutic embolization or surgical resection in form of local excision, segmentectomy, lobectomy, or pneumonectomy. [16] Embolization therapy [17] is a form of treatment based on occluding the feeding arteries which includes Amplatzer vascular plugging except in secondary cause where underlying cause are treated such as due to liver failure which are treated with liver transplantation. Embolization therapy eliminates the need of surgical intervention and has less morbidity & mortality. [8, 18]

The patient was improved dramatically in terms of dyspnea, cyanosis and fatigability following embolization therapy and refrain from complications of PAVM giving better life to patient free from morbidity.

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