

Dyspnea in a patient with Hereditary Hemorrhagic Telangiectasia

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Citation

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Abstract

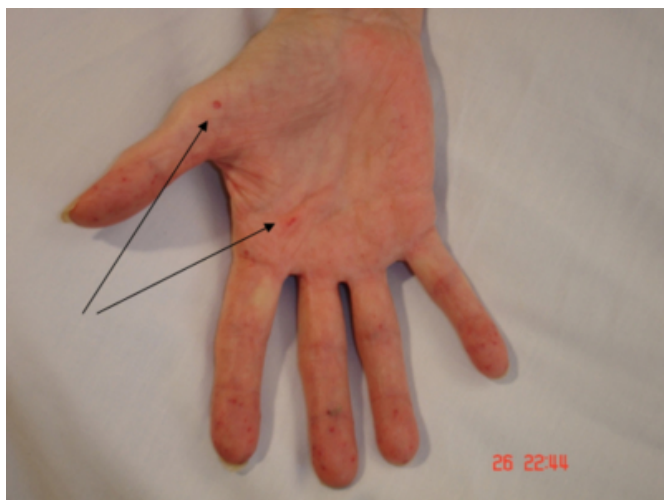
Pulmonary arteriovenous malformation's (PAVMs) are rare pulmonary vascular anomalies, occurring at increased frequency in patients with hereditary hemorrhagic telangiectasia (HHT). PAVMs, in which left pulmonary artery communicates directly with a pulmonary vein, usually present with cyanosis. We discuss a case of an abnormal communication between pulmonary artery and left pulmonary vein presenting as congestive heart failure.

CASE HISTORY

A 77 year old female with history of hereditary hemorrhagic telangiectasia and recurrent epistaxis presented to the Emergency Department with complaints of fatigue, shortness of breath, cough and lower extremity edema of several days duration. Physical examination revealed a temperature of 98 °F , pulse of 100 beats per minute, a respiratory rate of 20 per minute; pulse oximetry revealed a saturation of 90% on room air. Physical examination showed multiple telangiectases of the lips, tongue, palate and fingers consistent with her history of hereditary hemorrhagic telangiectases (Figure 1).

Figure 1

Figure 1: Multiple telangiectasia of the hands (arrows)

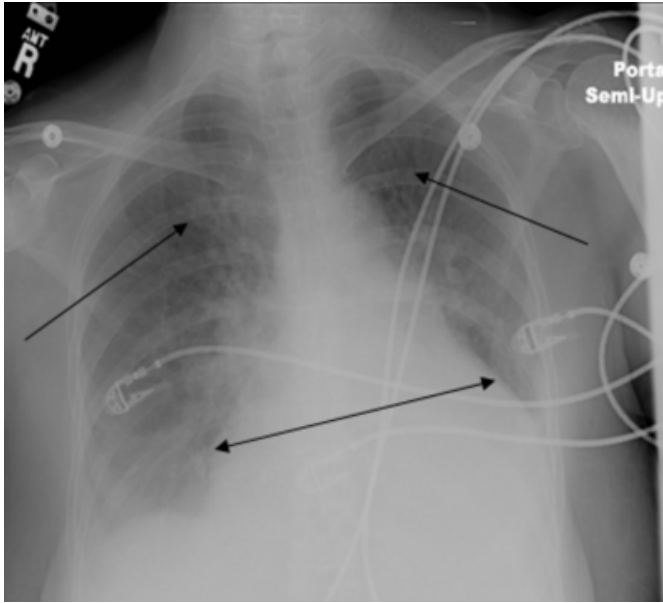


She had an elevated jugular venous pressure and lung examination revealed bibasilar crackles and diminished

breath sounds in the left base. Cardiac examination was significant for an S3 gallop and a loud left hemi thorax bruit was audible which increased with inspiration and the Muller maneuver (forced inspiration with a closed glottis after full expiration), and decreased with expiration and the Valsalva maneuver confirming a pulmonary bruit. Abdominal examination revealed no hepatosplenomegaly and lower extremity exam showed 2 + pitting edema. Her laboratory workup revealed a cardiac BNP (brain natriuretic peptide) of 750 pg/mL. An arterial blood gas revealed partial pressure of oxygen (Pao₂) at 80 mmHg and an oxygen saturation of (SaO₂) of 90% on room air. Chest radiograph was consistent with bilateral vascular congestion and left sided pleural effusion (Figure 2).

Figure 2

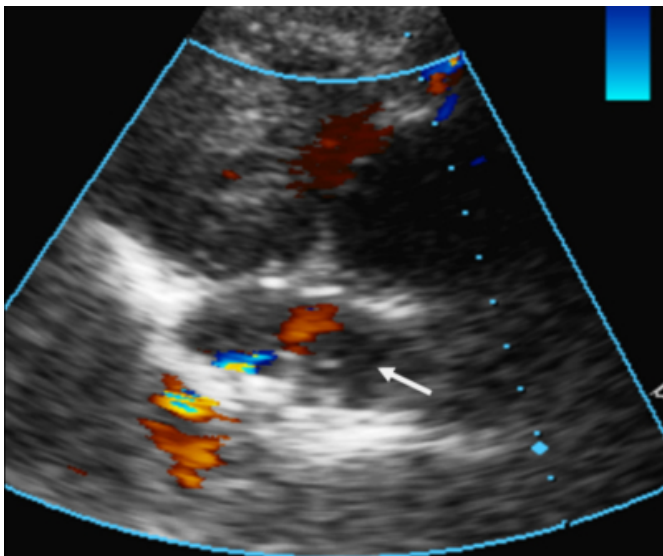
Figure 2: Chest X-Ray showing bilateral vascular congestion and cardiomegaly (arrows)



A clinical diagnosis of congestive heart failure was entertained and she was managed with intravenous diuretics and oxygen. As part of usual cardiac work-up she had a 2D-echocardiogram which showed a dilated varix between left pulmonary artery and vein (Fig 3). The echo also revealed a hyperdynamic left ventricle with an ejection fraction of 70-75%.

Figure 3

Figure 3: 2D echo showing the arteriovenous malformation with blood flow (arrow)



An ultrasound of the liver did not show any AV malformations and a CT scan of chest, revealed a large AV

malformation approximately 2.5 x 3.5 x 9.4 cm in the left lower lobe of the lung, draining into a markedly dilated inferior pulmonary vein. CT scan of abdomen and pelvis and brain, however did not show any AV malformations. Magnetic resonance angiography (MRA) of her chest revealed cardiomegaly with enlarged right atrium and a large coronary sinus with a large inferior pulmonary vein varix on the left primarily supplied by a prominent inferior pulmonary artery (Figure 4). A cineangiogram revealed filling of the pulmonary vein varix by the inferior pulmonary artery (Figure 5).

Figure 4

Figure 4: MRA of the chest showing the left pulmonary vein varix supplied by the pulmonary artery (arrow)

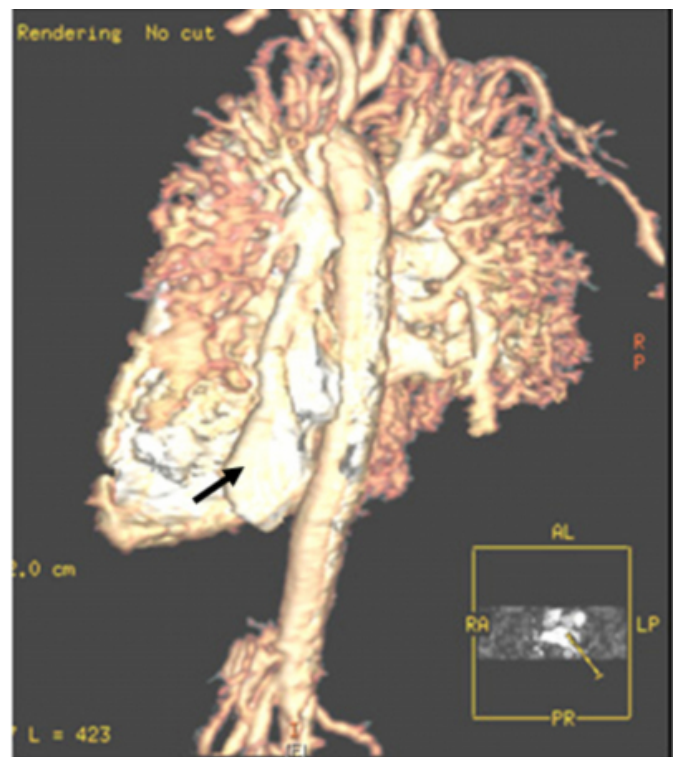
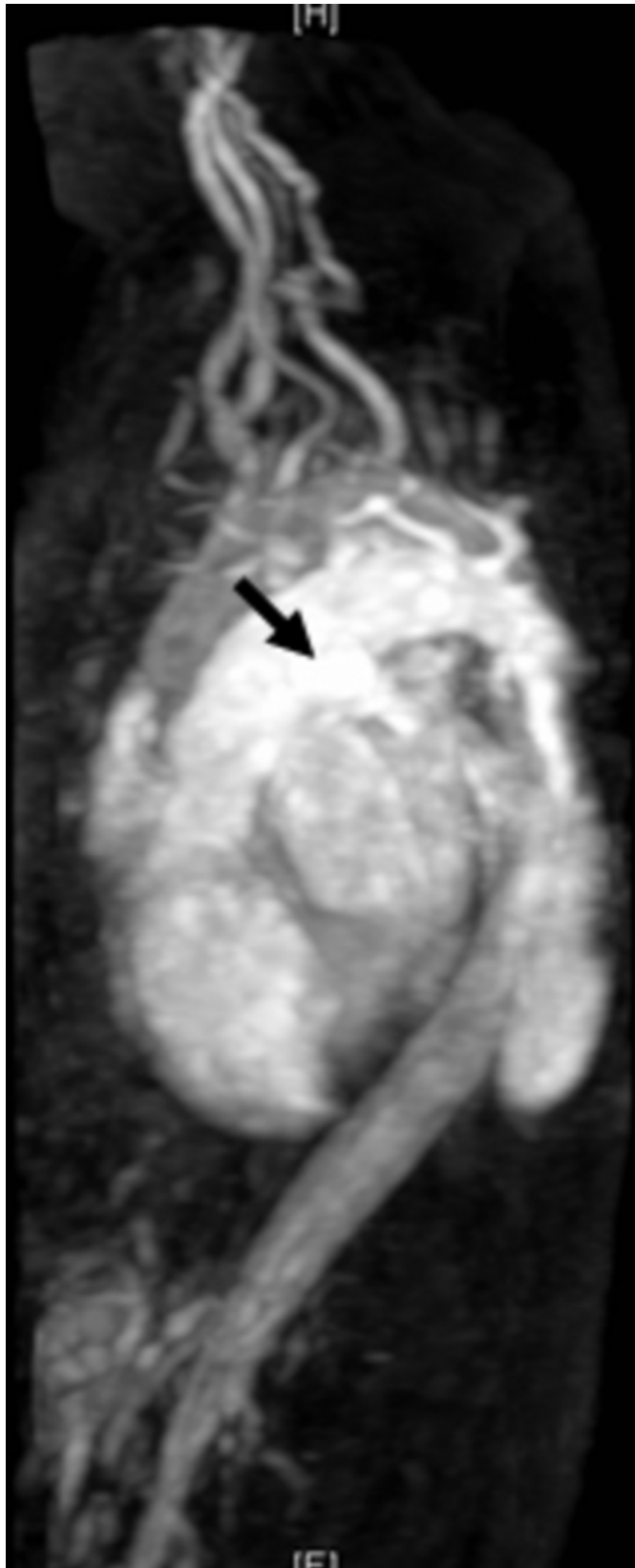


Figure 5

Figure 5: C T angiogram demonstrating the filling of the varix from the pulmonary artery (arrows)



The patient improved on intravenous diuretics and was

offered a transcatheter coil embolization as a treatment option for her PAVMs. However, the patient refused this treatment option stating that she needed time to consider it and was discharged without complications on day 3 of her admission.

DISCUSSION

Pulmonary arteriovenous malformations (PAVMs) consist of direct connections between a branch of a pulmonary artery and a pulmonary vein through a thin-walled aneurysm.

PAVMs are commonly congenital in nature. Since their first description at autopsy in 1897¹, these abnormal communications have been given various names including pulmonary arteriovenous fistulae, pulmonary arteriovenous aneurysms, hemangiomas of the lung, cavernous angiomas of the lung, pulmonary telangiectases, and pulmonary arteriovenous malformations².

PAVMs occur twice as often in females as in males. Approximately 70% of all PAVMs occur in association with Hereditary Hemorrhagic Telangiectasia (HHT) and approximately 15%-35% patients with HHT have PAVMs^{3,4,5,6,7} which range from diffuse telangiectases to large complex structures consisting of a bulbous aneurysmal sac between dilated feeding arteries and draining veins. The HHT subgroup with endoglin mutations has a higher risk (40%) than HHT patients with ALK1 mutations^{5,6,7,8}. Most patients are asymptomatic and account for 13% to 55% of patients in different series^{4,5,6,7,8,9,10,11}. The classic triad of dyspnea, cyanosis, and clubbing is present in only 10% of patients with PAVMs⁴. The direct communication between the pulmonary and systemic circulation bypasses the capillary bed and this right-to-left-shunt causes hypoxemia and the absence of a filtering capillary bed allows embolism that can reach the systemic arteries inducing clinical sequelae, especially in the cerebral circulation with brain abscesses and stroke. These processes account for clinical features such as dyspnea, fatigue, hemoptysis, cyanosis and polycythemia¹². The most common presenting symptom is dyspnea on exertion (31% to 67% of patients), and severity of dyspnea is related to the degree of hypoxemia and magnitude of the right-to-left shunt. Majority of the patients with PAVMs tolerate hypoxemia well and are relatively asymptomatic unless the arterial oxygen pressure is less than 60 mm Hg. This reflects the fact that pulmonary arteriovenous dilatation is the hallmark of lung involvement in HHT with PAVMs, leading to decreased pulmonary vascular resistance and increased cardiac output, with

normal to low pulmonary arterial pressure and ability to generate supranormal cardiac outputs. Cyanosis resulting from the right-to-left shunt is usually not evident until adolescence or adulthood. Epistaxis, melena, and neurologic symptoms (headache, vertigo, numbness, paresthesias, syncope, or confusion) should alert the clinician to the possibility of coexisting HHT as in our case. The most common physical exam findings are cyanosis, clubbing, and pulmonary vascular bruit. Classically, the pulmonary bruit is increased by inspiration and the Muller maneuver (forced inspiration with a closed glottis after full expiration). This is caused by an increase in the pulmonary blood flow. The bruit is decreased by expiration and the Valsalva maneuver, by decreasing venous return to the lung^{13,14}.

Congestive heart failure is an unusual cause of dyspnea and therefore of clinical presentation in patients with PAVMs. Patients of PAVMs who develop congestive heart failure usually have arteriovenous malformations (AVMs) in other vascular beds as occurs typically in patients with HHT where AV malformations can occur in the gastrointestinal tract, liver and the central nervous system. HHT occurs in 0.01% of the population. Affected individuals usually present with recurrent hemorrhage from AVMs in mucocutaneous epithelium. These malformations result in a high output heart failure. High-output heart failure is defined as symptoms of heart failure (dyspnea at rest or with varying degrees of exertion, orthopnea, paroxysmal nocturnal dyspnea, and edema, either pulmonary or peripheral) in the presence of an above normal cardiac index ($> 3.0 \text{ L/min/m}^2$). A number of conditions viz; HHT (Osler-Weber-Rendu disease), arteriovenous fistula, chronic anemia, Paget's disease, hyperthyroidism, beriberi, and polycythemia vera may lead to a reflective increase in cardiac output, which can be associated with heart failure. However, the elevated cardiac output itself does not lead to symptoms of congestive heart failure. Over time, because of the increase in blood volume, the right atrial pressure, pulmonary artery pressure, and left ventricular end-diastolic pressure gradually increase until the myocardium decompensates^{12,13,14,15}. Such conditions were not apparent in this case. The patient had no signs and symptoms to suggest Paget's disease. Her Alkaline phosphatase was normal i.e. 203 U/L (range 54-217U/L) and there was no bone pain or history of fractures. She was not polycythemic, her hematocrit being 34%. Again the patient did not have any history of chronic anemia though from time to time she would get a minor episode of epistaxis but all along she had maintained hemoglobin in the range of 10.2 g/dL to 11.3 g/dL. The patient did not betray any clinical

features of hyperthyroidism and her TSH was normal i.e. 1.975 IU/mL (range 0.350-5.5 IU/mL). Again no evidence of liver and kidney disease was found and there was nothing to suggest a diagnosis of carcinoid syndrome, a rare cause of high output heart failure.

Our patient however had no evidence of AVMs at any other site except the pulmonary vascular bed. An extensive imaging workup failed to reveal any arteriovenous malformations elsewhere. This is a unique case of isolated PAVMs presenting with heart failure.

CONCLUSION

We conclude that patients with hereditary hemorrhagic telangiectasia can develop high output heart failure with isolated PAVMs and in the absence of arteriovenous malformations elsewhere.

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