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Case Report

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PULMONARY ARTERIOVENOUS MALFORMATION WITH HEMOPTYSIS IN OSLER RENDU WEBER DISEASE – SUCCESSFUL TREATMENT BY ENDOVASCULAR COILING

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ABSTRACT

Hemoptysis is a common presenting symptom in pulmonary practice and the differentials include chronic bronchitis in smokers, tuberculosis, carcinoma lung, coagulation abnormalities and arteriovenous malformations. Pulmonary arteriovenous malformations are uncommon but important cause of hemoptysis which require prompt recognition and appropriate management measures. We present the case of a 20 year old gentleman who presented with hemoptysis and epistaxis, who on further evaluation had features of Osler Rendu Weber disease and a left upper lobe pulmonary arteriovenous malformation. Since the hemoptysis persisted despite conservative measures, successful treatment of the vascular lesion could be achieved with endovascular coil occlusion of the affected vessel.

KEYWORDS: Hereditary hemorrhagic telangiectasia, pulmonary arteriovenous malformation, vascular coiling.

INTRODUCTION

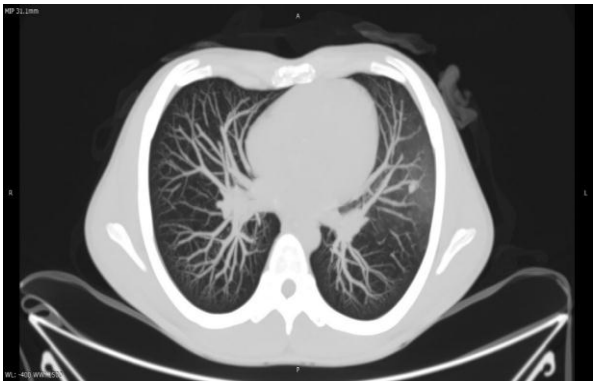
Hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu disease (OWRD), is an uncommon genetic disorder with autosomal dominant inheritance.[1] The disease is characterised by mucocutaneous telangiectasias and arteriovenous malformations. It is a systemic vascular dysplasia, hence any site may be affected although lesions involving nasopharynx, central nervous system (CNS), lung, liver, gastrointestinal (GI) tract and skin are often noted. Recurrent troublesome epistaxis is the most common presentation and may lead to severe anaemia necessitating transfusion. In 90% of patients the disease is manifested by fourth decade.[2] The diagnosis of HHT is made clinically on the basis of the Curaçao criteria, established in June 1999 by the Scientific Advisory Board of the HHT Foundation International.[3]

Pulmonary arteriovenous malformations (AVMs) are present in 15-33% of patients with the disease.[3] Most cases are symptomatic but the most dramatic presentation of PAVMs is with hemoptysis. Dyspnea and exercise intolerance can be presenting symptoms of pulmonary AVMs. Some lesions may cause enough right-to-left shunting to result in cyanosis, hypoxemia, and secondary polycythemia. Although minor hemoptysis can be managed with conservative measures, recurrent life threatening bleed from PAVMs may necessitate endovascular interventions or surgical resection of involved parenchyma. Surgical resection is immensely challenging in OWRD given the bilateral and often multiple nature of PAVMs.

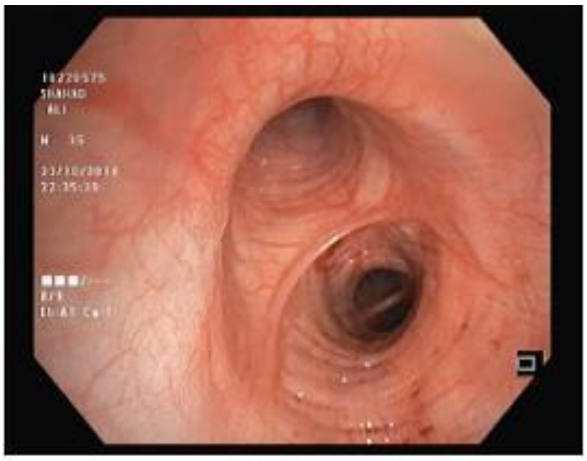
We describe the case of an 18 year old gentleman who presented with hemoptysis and epistaxis who, on evaluation, met with the diagnostic criteria for ORWD. He had a bleeding PAVM of the left upper lobe which was treated successfully with endovascular coiling.

CASE REPORT

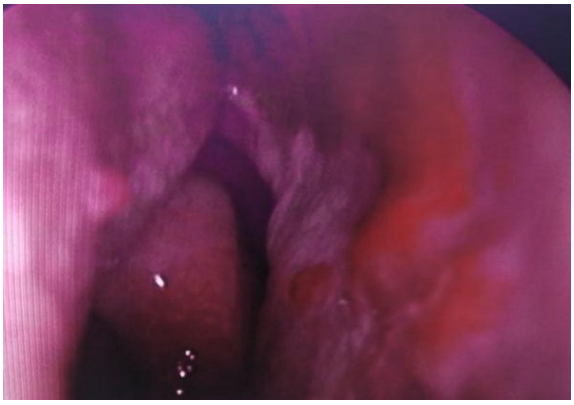
A 20 year old gentleman presented with 2 episodes of moderate hemoptysis, each bout amounting to approximately 100 ml for 3 days. His past history was remarkable for recurrent episodes of epistaxis from the age of 11 years which subsided with conservative measures, the last episode being a week back. He had no medical comorbidities. He denied usage of tobacco products, ethanol consumption or any high risk sexual behavior. Complete hemogram, blood sugars, renal functions, liver function tests and coagulation work up was normal. A chest radiograph was essentially normal. CT scan of the chest with contrast study (Fig 1) revealed a pulmonary arteriovenous malformation in the left upper lobe with surrounding ground glass opacities consistent with alveolar haemorrhage.



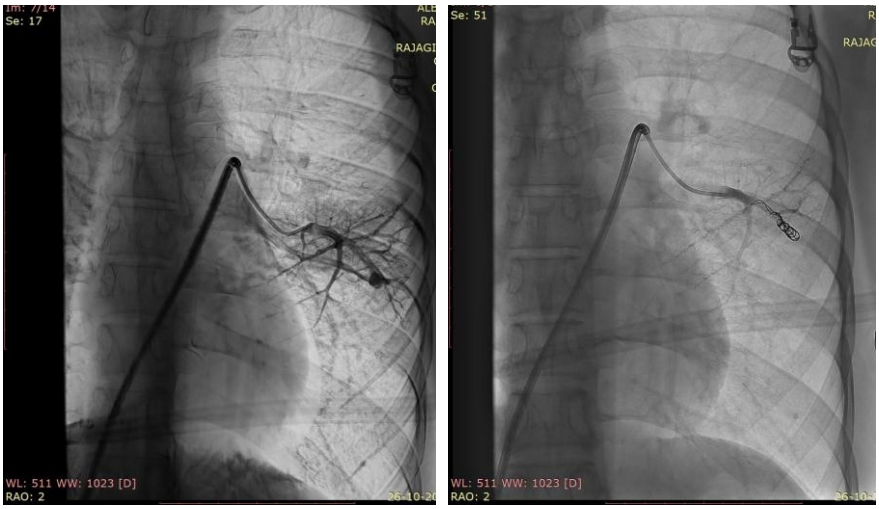
A bronchoscopy (Fig 2) revealed multiple telangiectatic vessels in the airway mucosa from the nasal cavity to bronchial segmental level.



The upper airway findings were re-examined with diagnostic nasal endoscopy (Fig 3) by the otolaryngologist. Given the clinical features, a diagnosis of Osler Rendu Weber Disease was arrived at. Interrogation of first degree family relatives did not reveal any clinically significant affection.



Since his hemoptysis did not subside with conservative measures, he was taken up for pulmonary angiography. Procedure was performed without any anaesthesia or sedation. Right femoral vein access was secured using 9F 11 cm long introducer sheath. 7Fr 63 cm Check-Flo Performer Introducer (Mullin's curve sheath) (Cook Medical, Bloomington IN) was introduced into pulmonary artery with tip into left pulmonary artery and diagnostic angiographic run was taken from left pulmonary artery and left descending pulmonary artery using 6F JR (Judkins Right) guiding catheter, which did not demonstrate the fistula. 5 F vertebral glide catheter was inserted through 7F sheath and pulmonary artery branch suspected to be supplying the lesion was cannulated based upon guidance from CT angiography. Super-selective run of this branch demonstrated small vascular sac measuring 7 mm, connected to the distal aspect of this pulmonary artery branch with adjacent early draining pulmonary vein branch suggestive of arterio-venous fistula (Figure 4). Microcatheter (Cantata, Cook Medical, Bloomington IN) was introduced through the vertebral glide catheter and tip positioned near the fistula site. Contrast injection from microcatheter showed filling of fistula sac and adjacent pulmonary vein branch.



Coil embolization of pulmonary vein branch supplying the fistula was done just proximal to fistula site using 6mmx20 mm interlock coil (Interlock-18 Fibered IDC Occlusion System, Boston Scientific, Marlborough, MA) and 4mmx4mm VortX-18 Fibered Platinum Coil (Boston Scientific, Marlborough, MA). Post procedure

angiography run showed complete occlusion of distal aspect of the pulmonary artery branch supplying the fistula with no filling of fistula (Figure 5). Thus the affected vessel was successfully blocked with vascular occlusion coil.

DISCUSSION

The earliest decription of a hereditary epistaxis was made by Sutton in 1864. In 1909, it was Hanes who named the disease as "hereditary hemorrhagic telangiectasias". But it is popularly mentioned as Osler-Weber-Rendu disease, after the noted contributions in describing the disease made by three eminent physicians.[4] OWRD is the first identified human disease caused by defects in a TGF-β superfamily receptor. The disease affects blood vessels throughout the body causing vascular dysplasia and results in a tendency for bleeding. HHT has been classified into the following four types (type 1 to 4). Endoglin gene (*ENG;* HHT type 1) and the ALK-1 gene (*ALK1;* HHT type 2) are the genes most often implicated in HHT.[5] Endoglin and ALK-1 are type III and type I TGF-β receptors, and both are exclusively expressed on vascular endothelial cells. In patients with Type 1 HHT, commonly Pulmonary and cerebral AVMs (CAVM's) are seen whereas in type 2, severe GI bleed and hepatic AVMs are noted.

The overall prevalence of this disease is 1 per 5000-10,000 population. The highest rates are seen in parts of the Dutch Antilles among the Afro-Caribbean population[6], where a prevalence of around 1 case per 200 persons was identified. Such epidemiological data are lacking in Indian subcontinent.

ORWD is equally common in either sex. It can remain clinically silent in the mildest cases. The diagnosis of this rare condition is made on the basis of the Curaçao criteria established in June, 1999. The typical triad of ORWD is made up of telangiectasia of the skin and mucous membranes, epistaxis, and a positive family history. The fourth criteria is visceral lesions (AVM) involving lungs, liver, brain, spine and gastrointestinal system. The diagnosis is confirmed by the presence of at least three of these manifestations.[3]

It is crucial to identify visceral involvement early to avoid complications. The risk of GI tract bleeding increases in patients older than 50 years. Epistaxis is the most common presenting symptom in ORWD, occurring in upto 90% of the patients. The mean age of onset of epistaxis is 12 years, and almost all exhibits this symptom by the age of 40 years.[7] Theoretically telangiectasias can occur anywhere on the skin or mucous membranes, but common affected sites are face, chest, and hands. They usually make their appearance by the end of third decade and increase in number with age. About 15 to 30% of the patients with HHT1 develop pulmonary AVMs and about 70% of pulmonary AVMs are due to HHT .[7] Hence, the disorder should be considered in any patient diagnosed with a pulmonary AVM. On the corollary, all patients with HHT should be screened for pulmonary AVM. The most sensitive test for screening of PAVM is contrast echocardiography although CT pulmonary angiography is the preferred investigation for localizing the lesion prior to surgical or endovascular therapy.

The treatment of clinical manifestations in ORWD is predominantly symptomatic. Epistaxis usually responds to conservative measures. Topical estrogen has been tried as nasal mucosal vasculature is hormone responsive. Laser coagulation of nasal telangiectasias, septal surgery, Youngs procedure, and angiographic embolization have been attempted, with varying results.

Isolated symptomatic PAVMs can be surgically resected. The interventional management of PAVM has been extensively reviewed previously.[8] Pulmonary AVMs can be successfully embolized with coils also. PAVM if left untreated, due to the right to left shunt dyspnea, hypoxemia, paradoxical emboli to the left side circulation, stroke and intracranial abscess can occur. Any AVM that is 3 mm or more in diameter is ideally treated as the risk of bleed is high. Also it is associated with a high risk of morbidity and mortality (0–55%).[9] Medical therapies under investigation include vascular endothelial growth factor (VEGF)[10] for the treatment of epistaxis and advanced liver disease and thalidomide for GI bleeding.

SUMMARY

The case reports summarises a young gentleman with hemoptysis as well as epistaxis who on evaluation had features of Osler Rendu Weber syndrome and a left upper lobe pulmonary arteriovenous malformation. Successful control of hemoptysis was achieved by endovascular coiling and obliteration of vascular neck. He remains under clinical follow up.

REFERENCES

1. Porteous ME, Burn J, Proctor SJ. Hereditary haemorrhagic telangiectasia: a clinical analysis. *J Med Genet*, 1992; 29(8): 527-30.
2. Plauchu H, de Chadarévian JP, Bideau A, Robert JM. Age-related clinical profile of hereditary hemorrhagic telangiectasia in an epidemiologically recruited population. *Am J Med Genet*, 1989; 32(3): 291-97.
3. Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJ, Kjeldsen AD, Plauchu H. Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). *Am J Med Genet*, 2000; 91(1): 66-67.
4. Cooper B. William Osler on Telangiectatic Syndromes. *Baylor University Medical Center Proceedings*, 1999; 12(4): 238-40.
5. Shovlin CL, Hughes JM, Scott J, Seidman CE, Seidman JG. Characterization of endoglin and identification of novel mutations in hereditary

hemorrhagic telangiectasia. *Am J Hum Genet*, 1997; 61(1): 68-79.
6. Westermann CJJ, Rosina AF, De Vries V, de Coteau PA. The prevalence and manifestations of hereditary hemorrhagic telangiectasia in the Afro-Caribbean population of the Netherlands Antilles: a family screening. *Am J Med Genet A*., 2003; 116A(4): 324-28.
7. Plauchu H, de Chadarévian JP, Bideau A, Robert JM. Age-related clinical profile of hereditary hemorrhagic telangiectasia in an epidemiologically recruited population. *Am J Med Genet*, 1989; 32(3): 291-97.
8. Meek ME, Meek JC, Beheshti MV. Management of Pulmonary Arteriovenous Malformations. *Semin Intervent Radiol*, 2011; 28(1): 24-31.
9. Chamarthy MR, Park H, Sutphin P, Kumar G, Lamus D, Saboo S, Anderson M, Kalva SP. Pulmonary arteriovenous malformations: endovascular therapy. *Cardiovascular Diagnosis and Therapy*, 2018; 8(3): 338-349-349.
10. Epperla N, Hocking W. Blessing for the Bleeder: Bevacizumab in Hereditary Hemorrhagic Telangiectasia. *Clin Med Res*., 2015; 13(1): 32-35.