Osler Rendu Weber Syndrome A Case Report

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Abstract: Osler-Weber-Rendu syndrome, also known as hereditary hemorrhagic telangiectasia, is a rare autosomal dominant disorder manifested by telangiectasias of the skin and mucous membranes and arteriovenous malformations of various organ systems. We present a case of Osler-Weber-Rendu syndrome with 4 affected members in her family.

Keyword: Hereditary hemorrhagic Telangiectasia, osler rendu weber syndrome, telangiectasia, Recurrent nose bleeds, Purpuric spots

Introduction: Osler-Weber-Rendu syndrome, or hereditary hemorrhagic telangiectasia (HHT), is a rare genetically determined autosomal dominant disorder identified by the triad of telangiectasia, recurrent epistaxis, and a positive family history for the disorder.[1] Presenting at any age, the disease has a wide spectrum of presentations, varying from asymptomatic to multiple organ involvement. The major cause of morbidity and mortality lies in the presence of multi organ arterio venous malformations (AVMs) and the associated hemorrhage. We present a case of HHT with typical mucocutaneous and pulmonary AVMs and association in 4 other members of the same family.

Case report
A 45-year-old postmenopausal lady born to nonconsanguineous parents presented with two episodes of Hemoptysis, multiple red raised lesions on the undersurface of the tongue, lower lip mucous membrane and around right hip region, the lesions are present for at least 25 years duration. She also had breathlessness on exertion and easy fatigability since 10 years. She had a history of recurrent spontaneous nose bleeds since early adulthood, and occasional bleeds from those on the tongue, she had no previous history of blood transfusions.

There was no history of bleeding gums, abdominal pain, bleeding rectum, headaches, seizures, visual disturbances, menorrhagia. On clinical examination patient was moderately built and nourished, pallor, cyanosis of central type, clubbing which is pan digital, and dermatologically

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nodular blanchable cherry red lesions 4 in number present over the right hip region.

Telangiectasia present on undersurface of the tongue, mucous membrane of lower lip. Abdomen examination revealed no organomegaly, examination of cardiovascular system revealed mild tachycardia, continuous bruit over right scapular region, and arterial saturation of 85%. Examination of upper respiratory system revealed telangiectasia over nasal mucosa in and around little’s area.

Basic blood investigation is normal except microcytic hypochromic picture, peripheral smear confirmed the finding. Liver and kidney function tests are within normal limits. ECG showed LAHB, LAD, and features suggestive of right ventricular hypertrophy.

X-Ray Chest showed mild Cardiomegaly, Echo is normal, Bubble contrast echo study showed appearance of bubble after fourth cycle in left side of heart chamber.
CT Pulmonary Angiogram

CT-Angiogram showed the Pulmonary AV Malformations. liver and brain showed no AVM, Fundus examination revealed no telangiectasia over retina.

Discussion
While Henri Rendu (1896), Sir William Osler (1901), and Frederick Parks Weber (1907) emphasized and published detailed observations of the syndrome which bears their names, it was Sutton (1864) who first described Osler-Weber-Rendu disease and Benjamin Guy Babington (1865) was the first to note its familial nature.

A number of variants of HHT have been described in literature. HHT type-1 and type-2 are due to defective endoglin (ENG) and activin like receptor kinase (ALK1) genes, respectively. Mutations of ENG are located on the long arm of chromosome 9 (9q33-34), whereas ALK1 mutations are on the long arm of chromosome 12 (12q13).[2,3] HHT type-3 involves mutations of the long arm of chromosome 5 (5q31.1-32) and type-4 maps to the short arm of chromosome 7 (7p14).[4,5] A HHT-juvenile polyposis overlap syndrome due to mutations of SMAD4 has also been described.[6] Patients with the HHT type-1 genotype have higher prevalence of pulmonary and cerebral AVMs, and more severe GI bleeding than in those with the HHT type-2 genotype. Conversely, the prevalence of hepatic AVMs is higher in patients with HHT type-2 than in those with HHT type-1.

[7] Although the precise mechanism remains poorly understood, bleeding tendency in HHT is attributed to localized vessel wall weakness. Bubble contrast echocardiography can be done as a screening procedure to identify the pulmonary AVM.

Diagnosis is based on the four components of the Curacao criteria established by the Scientific Advisory Board of the HHT Foundation International, Inc., viz. (1) epistaxis: spontaneous and recurrent; (2) telangiectasias: multiple, at characteristic sites, including lips, oral cavity, fingers, and nose; (3) presence of internal lesions: GI telangiectasia, pulmonary, hepatic, cerebral, and spinal AVMs; and (4) family history: first-degree relative with HHT according to these criteria. The diagnosis is considered definite if any three of the above mentioned criteria are present and possible if any two of the criteria are present. The diagnosis is unlikely if less than two criteria are present.[8] Our case was unique with the presence of all four criteria confirming the diagnosis.

The varied treatment modalities include estrogen, E-amino-caproic acid, cryotherapy, cautery, infrared coagulation, radiofrequency, pulse dye laser, Nd-YAG laser, and surgical ablation - all of which may be fraught with risks.[9-13]
Pulmonary and CNS AVMs and their hemorrhagic or embolic complications, viz. brain abscess and stroke are responsible for most of HHT’s 10% mortality rate. Lung lesions, once identified, are usually treated to prevent episodes of bleeding and more importantly embolism to the brain. This is particularly done in lesions with a feeding blood vessel of 3 mm or larger, as these are the most likely to cause long-term complications unless treated. The most effective current therapy is embolization with detachable metal coils. The procedure involves puncture of a large vein (usually under a general anesthetic), followed by advancing of a catheter through the right ventricle and into the pulmonary artery, after which radiocontrast is injected to visualize the AVMs (pulmonary angiography).

Once the lesion has been identified, coils are deployed that obstruct the blood flow and allow the lesion to regress. In experienced hands, the procedure tends to be very effective and with limited side effects, but lesions may recur and further attempts may be required. CTA scans are repeated to monitor for recurrence.[17][18][19][20] Surgical excision has now essentially been abandoned due to the success of embolotherapy.[19][20] It is crucial that a long-term follow-up for identification of potential complications is maintained and patients are counseled regarding the autosomal dominant nature of the condition.

Though cases have been reported with pulmonary AVM and mucocutaneous telangiectasias, our case is particularly unique as it had all the four criteria for diagnosis and that 4 members of the same family could be identified with this disease.

REFERENCES


