

## Osler weber rendu syndrome: A rare case report

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### Abstract

**Introduction:** Hereditary hemorrhagic telangiectasia (HHT) also known as Osler-Weber-Rendu syndrome is a rare fibrovascular dysplasia that makes vascular walls vulnerable to trauma, rupture causing skin and mucosal bleeding. It is an autosomal dominant inheritance characterized by recurrent epistaxis and telangiectasia on the face, hands and oral cavity; visceral arteriovenous malformations and positive family history. We report a case of Osler Weber Rendu syndrome in a 50 year old male.

**Keywords:** Osler weber rendu, Epistaxis, Telangiectasia.

### Introduction

Osler weber rendu syndrome also known as hereditary hemorrhagic telangiectasia, is characterized by small tuft of dilated capillaries scattered over the skin and mucous membranes. It is an autosomal dominant disorder. The clinical triad of hereditary hemorrhagic telangiectasia consists of recurrent epistaxis, telangiectasias and positive family history. Recurrent epistaxis is the most frequent symptom which is usually of childhood onset. Telangiectasia presents as punctate, linear or spider like appearance involving skin, tongue, ears, palms, soles, finger tips, nail beds, oral and nasal mucous membranes. The mucosal lesions usually blanch on pressure and they are more vulnerable to spontaneous rupture and leads to bleeding. These are often associated with Arterio venous malformations affecting lung, liver and/ or brain.

These patients have an abnormal plasma concentrations of transforming growth factor-beta (TGF- $\beta$ ) and vascular endothelial growth factor (VEGF) secondary to mutations in ENG, ACVRL1 and MADH4. Iron deficiency and associated anemia are frequent complications of the disease due to recurrent epistaxis and/or gastrointestinal bleeding.

### Case Report

A 50 yr old male presented with telangiectasias on the tongue for past 6 months. He has been suffering from frequent episodes of mild to severe nasal bleeding since childhood. Blood transfusions were given twice for correction of anaemia. No history of malena. History of similar complaints in the family members (mother, daughter and grandmother).

### Clinical examination

Multiple punctate, branched, linear telangiectasias on the dorsal and ventral surface of the tongue was seen (Fig. 1). Pallor present. Examination of hair, nail, skin and genitals were normal.



**Fig. 1: Telangiectasias on the dorsal and ventral surface of tongue**

### Investigations

CBC - 6.6 gm/dl, Renal function test, Liver function test, Urine analysis - WNL, HBsAg - normal, VCTC- non reactive, Peripheral smear - microcytic hypochromic and normocytic normochromic anaemia, BT, CT, PT, APTT, INR- within normal limits, ECG, ECHO, X ray chest - NAD, upper GI endoscopy - normal study, USG ABDOMEN - hepatomegaly and Hepatic Doppler - Normal study.

### Management

The patient was treated with anterior nasal packing and 3 units of packed red blood cells, and he underwent septal dermoplasty for recurrent epistaxis.

### Differential diagnosis

Spider angiomas, Ataxia telangiectasia, Rothmund syndrome, Bloom syndrome. Patient advised follow up for GI scopy and USG abdomen.

### Discussion

Henri Rendu (1896), Sir William Osler (1901), and Frederick Parks Weber (1907) were emphasized and published this syndrome which bears their names. In 1864 Sutton, first described Osler-Weber-Rendu disease and in 1865, Benjamin Guy Babington was the first one to note its familial nature.

Hereditary haemorrhagic telangiectasia is an autosomal dominant disorder with incidence of 1:5000 to 1:8000.

Males and females are equally affected. Mutation of genes ACVRL1, ENG and SMAD4 causes HHT. HHT 1 associated with ENG gene mutation. HHT 2 associated with ACVRL1 gene mutation. Mutation in endoglin (ENG) and ACVRL1 gene together accounts for 85% of cases. HHT 1 patients are increased risk of pulmonary Arterio venous malformations whereas HHT 2 patients tend to have milder phenotype, late age of onset but increased liver manifestations. Patients with mutation of SMAD 4 have HHT with Juvenile polyposis.

Diagnosis based on the curacao criteria includes

1. Epistaxis which is spontaneous and recurrent.
2. Telangiectasias over the lips, oral cavity, fingers and nose.
3. Visceral lesions like GI bleeding, Arterio venous malformations of pulmonary, liver, cerebral.
4. First degree relative positive family history.

**Definite diagnosis:** If 3 criteria are present, Possible diagnosis: 2 criteria, Unlikely diagnosis: 1 criterion.

As our patient had three features like Recurrent epistaxis, telangiectasia of tongue and family history, he fulfilled the criteria.

Frequent nasal bleeding and melena due to telangiectasia in the nose and GIT. Epistaxis is the most frequent and persistent sign. Worsening of epistaxis may enhance high-output cardiac failure from arterio venous malformations. GI bleeding is the presenting sign in upto 25% of patients; however, 40–50% develop GI bleeding sometime even during the course of their disease.

The spleen may be enlarged. Pulmonary and CNS Arterio venous malformations may appear later in life. Liver failure can result from diffuse intrahepatic shunting— hepatic artery to vein, bypassing the liver parenchyma. Retinal arterio venous aneurysms occur only rarely. Other sites of bleeding may be the kidney, spleen, bladder, liver, meninges, and brain.

The risk of cerebral hemorrhage from cerebral arterio venous malformations, cerebral abscesses, and pulmonary hemorrhage from pulmonary arterio venous malformations is probably high enough that asymptomatic patients should be screened for the presence of cerebral and pulmonary Arterio venous malformations. Pregnancy can also exacerbate HHT.

The telangiectasia tend to increase in number during middle age. First appearance of telangiectasia on the under surface of tongue and floor of the mouth is around puberty.

Pulmonary arterio venous fistula or intracranial arterio venous fistula and bleeding are the cause of death in these patients.

The treatment is symptomatic and multidisciplinary approach. Regular follow up of oximetry and blood count for anaemia should be done. Bleeding episodes are treated supportively with iron supplementation and RBC transfusions. Our patient had given multiple blood transfusions during bleeding episodes and advised for regular follow up. The tendency of epistaxis has been reduced by estrogen therapy.

Dermoplasty of bleeding nasal septum performed by replacement of mucous membrane with skin from the thigh or buttock Topical tranexamic acid has been used to control epistaxis. Interventional radiology with selective embolization can treat pulmonary and CNS arterio venous malformations, avoiding invasive surgeries. liver transplantation required in patients presenting with liver failure.

Blocking vascular endothelial growth factor (VEGF) with thalidomide or lenalidomide can reduce GI bleeding. Bevacizumab mono clonal inhibitor of VEGF given intravenous shows some promising results in severely ill patients. it reduces size and flow of hepatic AVMs, reversing liver and heart failure, reduces the transfusion need.

## Conclusion

This case is reported for its rarity. For any case presenting with oral telangiectasias, this syndrome has to be kept in mind.

**Conflicts of Interest:** None.

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