

## A Case Report of Osler Weber Rendu Syndrome

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**Abstract:** Hereditary hemorrhagic telangiectasia (HHT) or Osler-Weber-Rendu disease is a rare fibrovascular dysplasia that makes vascular walls vulnerable to trauma and rupture causing skin and mucosal bleeding. It is of autosomal dominant inheritance characterized by recurrent epistaxis and telangiectasia on the face, hands and oral cavity; visceral arteriovenous malformations (AVMs) and positive family history. Epistaxis is often the foremost manifestation. It is associated with AVMs in several organs. There are possible hematologic, neurologic, pulmonary, dermatologic and gastrointestinal complications. Treatment is supportive and helps prevent complications. We report herein a patient with this syndrome who came to Pediatric Intensive Care Unit at our hospital.

**Keywords:** hemorrhagic telangiectasia, arteriovenous malformations, epistaxi

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### I. Introduction

Hereditary hemorrhagic telangiectasia (HHT) or Osler-Weber-Rendu disease is a rare fibrovascular dysplasia that makes vascular walls vulnerable to trauma and rupture causing skin and mucosal bleeding. It is of autosomal dominant inheritance characterized by recurrent epistaxis and telangiectasia on the face, hands and oral cavity; visceral arteriovenous malformations (AVMs) and positive family history. Epistaxis is often the foremost manifestation. It is associated with AVMs in several organs. There are possible hematologic, neurologic, pulmonary, dermatologic and gastrointestinal complications. Treatment is supportive and helps prevent complications. We report herein a patient with this syndrome who came to Pediatric Intensive Care Unit at our hospital

### II. Case Report

A CASE of 3 year old female, first child born of non-consanguineous marriage, presented with cyanosis and respiratory distress. Patient had convulsions and epistaxis 4 days back.

Family history-Father and paternal uncle expired because of unknown bleeding disorder.

#### On Examination:

General Examination- cyanosis and clubbing was present. The patient was anemic with pallor. There were multiple telangiectasias in the typical locations like hands, legs and tongue.

#### On investigation:

Hb-14.1, WBC-11900, Platelet-2.75 lac, HCT 35.3.

SPO<sub>2</sub>- 80 percent

ESR-30

Urine-normal

BUN 12 Creatinine-0.7

CSF-Normal.

BT, CT, PT, PTT were Normal.

SGPT, SGOT-Normal.

Chest X-ray- Normal

**2D ECHO** – Essentially normal heart with No abnormality, good biventricular function. High index of suspicion of Pulmonary AV malformation In view of Normal 2D Echo she was investigated further to determine cause of cyanosis and clubbing.

**CT Scan Brain-** Ill-defined area of hemorrhage measuring 2.7\*2.4 cm in right parieto-occipital region extending into ipsilateral lateral ventricle. Hypertrophied right middle cerebral artery, and right posterior cerebral artery. A dilated straight sinus suggested drainage of AVMs.

CT Chest revealed bulky lobulated homogeneity within right hemithorax s/o presence of AVMs. ALSO showed Heterogeneity of LIVER enhancement s/o intrahepatic AVMs.

Cerebral Angiography showed multiple cerebral and cerebellar AVMs, Pial AVFs, Right frontal telangiectasias and Right sigmoid sinus diverticulum.

Pulmonary Angiography- Two large pulmonary AVMs one in the Right upper lobe and other in Right lower lobe.

### III. Figures



**Fig 1** And **2** Showing Multiple Telangiectasia

**Fig 3** Of Cerebral Angiography Showing Multiple Avms

**Fig 4** Showing Normal Xray ; **Fig 5** Of Pulmonary Angiography Showing Two Large Pulmonary Avms

### IV. Discussion

While Henri Rendu (1896), Sir William Osler (1901) and Frederick Parkes 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[1]. A number of variants of hereditary hemorrhagic telangiectasia (HHT) have been described in literature. HHT type 1 (HHT1) and HHT type 2 (HHT2) are due to defective endoglin (ENG) and activin receptor-like kinase 1 (ALK1) genes, respectively. Mutations of ENG are located on the long arm of chromosomes 9 (9q33-34), whereas ALK1 mutations are on the long arm of chromosome 12 (12q13). HHT type 3 (HHT3) involves mutations of chromosome 5 (5q31p-32) and HHT type 4 (HHT4) maps on short arm of chromosome 7 (7p14)[2]. An HHT-juvenile polyposis overlap syndrome due to mutations of SMAD4 has also been described.[3,5]

Patients with HHT1 genotype have higher prevalence of pulmonary and cerebral arteriovenous malformations (AVMs) and more severe GI bleeding than in those with HHT2 genotype. Conversely, the prevalence of hepatic AVMs is higher in patients with HHT2[4,7]. Although the precise mechanism remains poorly understood, bleeding tendency in HHT is attributed to localized vessel wall weakness.

Diagnosis depends on the presence of four components known as the Curacao criteria established in June 1999 by the Scientific Advisory Board of the HHT Foundation International, Inc. These four criteria are: Â Epistaxis: Spontaneous and recurrent Â Telangiectasias: Multiple, at characteristic sites including lips, oral cavity, fingers and nose Â Presence of internal lesions: GI telangiectasia, pulmonary, hepatic, cerebral and spinal AVMs[6,8]

**Â Family history:** First-degree relatives with HHT according to these criteria. The diagnosis is unlikely if less than two criteria are present. Our case fulfilled all four criteria. His symptomatology was attributed to anemia resulting from frequent bleeding from the vascular lesions. The varied treatment modalities include estrogen, L-amino- caproic acid, cryotherapy, cautery, infrared coagulation, radiofrequency, pulse dye laser, Nd-YAG laser and surgical ablation, all of which may be fraught with risk. Asymptomatic pulmonary and central nervous system AVMs and their hemorrhagic or embolic complications viz. brain abscess and stroke are responsible for most of 10% mortality rate associated with HHT. 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.[9,10,11,12]

## V. Conclusion

Though cases have been reported with digital and mucocutaneous telangiectasia, our case is particularly unique as it had all the four criteria (curacao criteria) for diagnosis and also two other members of the same family could be suspected with the disease.

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