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Osler-Weber-Rendu Syndrome: A Case Report on a Rare Vascular Disease Presented with Upper Gastrointestinal Bleeding and Anemia

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ABSTRACT

Osler Weber Rendu Syndrome, also known as Hereditary hemorrhagic telangiectasia, is an autosomal dominant disorder characterized by abnormal blood vessel formation-telangiectasia and arteriovenous malformations. The most common clinical manifestation is spontaneous and recurrent nosebleeds, telangiectasia on lips, tongue, buccal, and gastrointestinal mucosa [1].

Until today, only few cases have been reported from Nepal. Here we report case of a 50-year-old female from Damak, who presented with a history of multiple foci of telangiectasia over the tongue, fatigue, anemia and black discoloration of stool. The clinical diagnosis was confirmed by Curacao Criteria characterized by epistaxis, Mucocutaneous Telangiectasia, visceral AVMs, and Family history. These are few cases report of Osler Weber Rendu Syndrome from Nepal and few documented cases who presented with typical telangiectasia on lips and anemia secondary to UGI bleed - malena. We suspect that Osler Weber Rendu Syndrome might not have been considered in the differential diagnosis of Anemia in patient presenting with telangiectasia and UGI bleeding in Nepal and suggest that it is to be kept as a differential in the given scenario.

INTRODUCTION

Osler-Weber-Rendu syndrome, also known as hereditary hemorrhagic telangiectasia (HHT), is a rare autosomal dominant genetic disorder with a prevalence of 1:5000 to 1:8000. HHT gene pathogenic variants lead to the development of aberrant vascular structures that range from dilated micro-vessels to large arteriovenous malformations (AVMs) [2]. The eponym recognizes the 19th century physicians William Osler, Henri Jules Louis Marie Rendu, and Frederick Parkes Weber, who each independently described the disease. Abnormal angiogenesis occurs in the skin, mucous membranes, as well as in the visceral organs such as the lungs, liver, and brain. The syndrome is characterized by epistaxis along with a mucocutaneos manifestation and often in association with the clinical spectrum of various malformations of the liver, lungs, and gastrointestinal tract, which may be asymptomatic or exhibit a wide range of clinical symptoms [3].

There are three main types of HHTs, which include Type 1, Type 2, and HHT-Juvenile polyposis overlap syndrome caused by mutations in ENG (endoglin), ACVRL1 (activin receptor-like kinase 1), and SMAD4 (transcription factor), respectively. These mutations affect TGFβ (transforming growth factor beta) pathway, disrupting the key balance between angiogenesis inducers and inhibitors in favor of angiogenesis and consequent development of multiple telangiectasias and AVMs in various body regions. Recurrent nosebleeds are common and occur in approximately 90% of affected individuals, while other symptoms include: gastrointestinal bleeding causing malena and severe symptomatic microcytic anemia [4]. Although there is a wide array of manifestations ranging from asymptomatic to life-threatening, most of the time, the disease presents with a history of spontaneous and recurrent epistaxis, symptomatic anemia-fatigue, palpitations, sob and chronic GI bleeding.

"Curacao criteria" is used to clinically diagnose the patients as HHT, which includes recurrent spontaneous nosebleeds, mucocutaneous telangiectasias, visceral involvement, and first-degree family history of HHT. If ≥ 3 criteria are met, the diagnosis is definitive.

Treatment of the disease is multimodal, primarily focusing on limiting the bleeding from various sites like the nose and gastrointestinal tract, and correcting the anemia with iron supplementation and blood transfusion when necessary as well as anti-VEGF agents like bevacizumab, pazopanib, and thalidomide, decreasing the frequency of bleeding, need for transfusion, and improving the overall quality of life.

Case Report

A 50 years-old female, resident of Damak, Terai region of Nepal, presented on July 2023 with a history of multiple episodes of spontaneous nasal bleeding (epistaxis), red color multiple small vessel visible in tongue, fatigue, palpitation and black discoloration of stool. Her lab investigations revealed positive fecal occult blood and very-low Hemoglobin (Hb) of 5.7 g/dl requiring multiple transfusions. Two months later, she presented again with similar symptoms, for which an extensive work-up was done to find out the possible cause of iron deficiency anemia. However, all work-ups, including autoimmune profile, stool microscopy, and bone marrow biopsy, were inconclusive with normal vitamin B12 and folate levels.

Esophagogastroduodenoscopy (EGD) was performed as per upper gastrointestinal endoscopy findings, positive fecal occult blood test,

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and normal colonoscopy, she was diagnosed as antral gastritis and was given blood transfusions, iron supplements, and eradication therapy for *Helicobacter pylori*. In coming years, the patient had similar symptoms requiring multiple hospitalizations. In 2024, She presented with nasal epistaxis and multiple telangiectasia of lips and tongue and iron deficiency anemia and thrombocytopenia which was treated with steroids. She was refractory to steroids. The patient was further investigated with EGD showing multiple Arteriovenous (AV) malformations in the gastroesophageal junction, gastric fundus, and pylorus, and the second part of the duodenum with active bleeding and ulcerations. In addition, a colonoscopy revealed AV malformations in the rectosigmoid region. After carefully reviewing the patient's medical history and family history and the features of endoscopy, a diagnosis of Osler Weber Rendu syndrome was made. The patient subsequently started on bevacizumab and showed dramatic improvement. A workup for complications associated with Osler Weber Rendu syndrome, a CT angiogram of the brain and chest was done as well as virology and cytogenetics were normal.

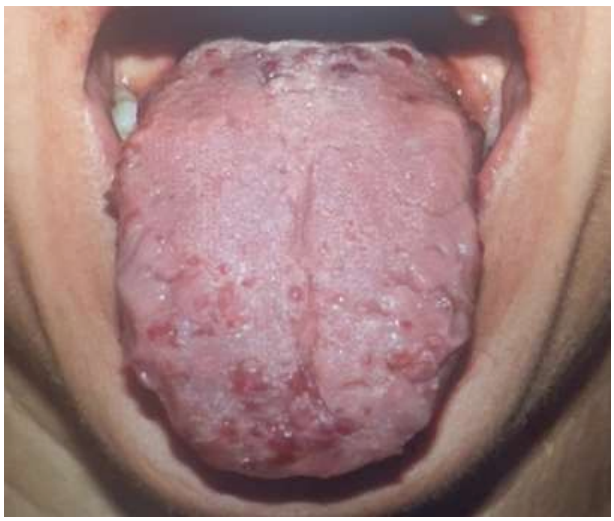

Figure 1: Telangiectasia of the Tongue.

Discussion

Osler-Weber-Rendu syndrome, also known as Hereditary Hemorrhagic Telangiectasia (HHT), is a rare autosomal dominant genetic disorder with a prevalence of 1:5000to 1:8000. However, few cases are reported from Asia and a handful of cases from neighboring country India. The syndrome is characterized by epistaxis along with a muco-cutaneos manifestation and often in association with the clinical spectrum of various malformations of the liver, lungs, and gastrointestinal tract, which may be asymptomatic or exhibit a wide range of clinical symptoms.

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Conclusion

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Conflict of Interest: None

Ethical Consideration: Not Required

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Declaration: Patient approve the article for publication

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