Case Report

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A presentation of hemiplegia and recurrent melena in Osler Weber Rendu syndrome

Ayana S Kumar[1], Akshaya Suresh[1], Rakhi Krishna[1], Anoop Koshy[1], Roshni P R[*2]

[1]Department of Pharmacy Practice, Amrita School of Pharmacy, Amrita Vishwa Vidyapeetham, Kochi - 682041, Kerala, India

[2]Department of Gastroenterology, Amrita Institute of Medical Sciences and Research Center, Amrita Vishwa Vidyapeetham, Kochi - 682041, Kerala, India

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Abstract

Osler Weber Rendu syndrome, synonymously Hereditary hemorrhagic telangiectasia, is a rare systemic disease with multiple-organ involvement. This genetic disorder inherits in an autosomal dominant fashion, with the incidence found closer to 1 in every 5,000 people. The condition can be diagnosed at any age and has an equal chance of affecting both males and females. A 74-year-old male, having a history of left hemiplegia, was admitted with a recent history of recurrent melena. An upper enteroscopy revealed bleeding telangiectasia in the proximal jejunum managed with hemoclips. The possibility of Osler Weber Rendu syndrome was considered as he developed bleeding recurrently. According to Curacao Criteria, a minimum of two out of four symptoms should be present to ratify the disease, and the presence of three symptoms makes a definite diagnosis. Since there is no cure for the syndrome to date, therapy's mainstay entails meticulous supportive care (focused on managing the manifestations). The disease is a type of arteriovenous malformation associated with the mutation in either endoglin (ENG) or activin A receptor-like kinase-1 (ACVRL1) genes. He was given an intraarticular injection of methotrexate in the left wrist hand because of small joint polyarthritis. The patient was symptomatically and clinically better on discharge. Novel treatment strategy includes bevacizumab and thalidomide though their use has not been clinically approved.

*Corresponding Author

Name: Roshni P R
Phone: +91-9048540788
Email: roshnipr@aims.amrita.edu

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INTRODUCTION

Osler Weber Rendu syndrome, synonymously Hereditary Hemorrhagic Telangiectasia (HHT), is a fibrovascular tissue disorder that follows an autosomal dominant fashion, hits 1 in 5000 to 1 in 10,000 worldwide (Vukomanović *et al.*, 2014). Two out of four symptoms should at least present to confirm the disease, and the presence of three symptoms make a definite diagnosis according to Curacao Criteria, which covers spontaneous and persistent epistaxis, cutaneous and mucosal telangiectasia on the fingers, face, ear, and oral cavity, Arteriovenous Malformations (AVM) in the lung, brain, and liver, and family history (Juares *et al.*, 2008). Like the brain's vascular deformity, a range of CNS

complications prevails in these patients, (Brinjikji *et al.*, 2017) resulting in migraine and hemorrhage (subarachnoid or intraparenchymal). There is advancing telangiectasia on the lips, tongue, face, and hands (Agnollitto *et al.*, 2013).

Case history

A 74-year-old male, a known case of type 2 diabetes mellitus with a history of left hemiplegia, was admitted with a recent history of recurrent melena. CT scan abdomen done showed possible small bowel vascular malformation. An upper enteroscopy (Figure 1) was done; it revealed bleeding telangiectasia in the proximal jejunum. Hemoclips were applied, and bleeding was controlled. After three months, he was admitted again, then with nasal bleed. In the background of multiple site telangiectatic bleeds in the recent past (Figure 2), the possibility of Osler Weber Rendu syndrome was considered. ENT was consulted, and the patient's nasal telangiectasia was cauterized. He was given an intraarticular injection of methotrexate in the left wrist hand because of small joint polyarthritis. At the time of discharge, the patient was advised on methotrexate 10mg weekly once and folvite 5mg twice per week.

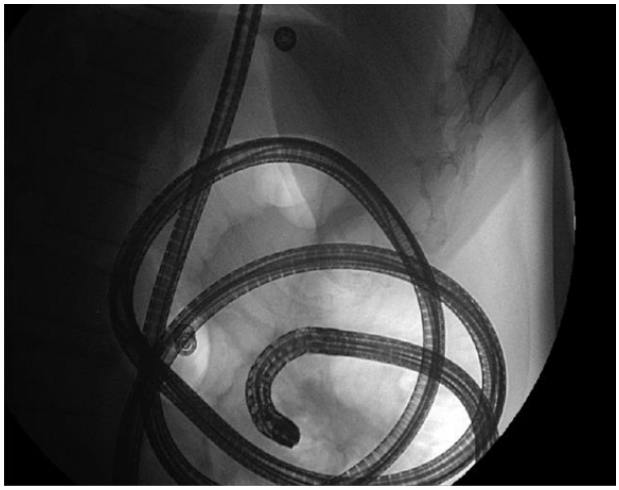

Figure 1: Upper enteroscopy showed bleeding Jejunal Dieulafoy Lesion.

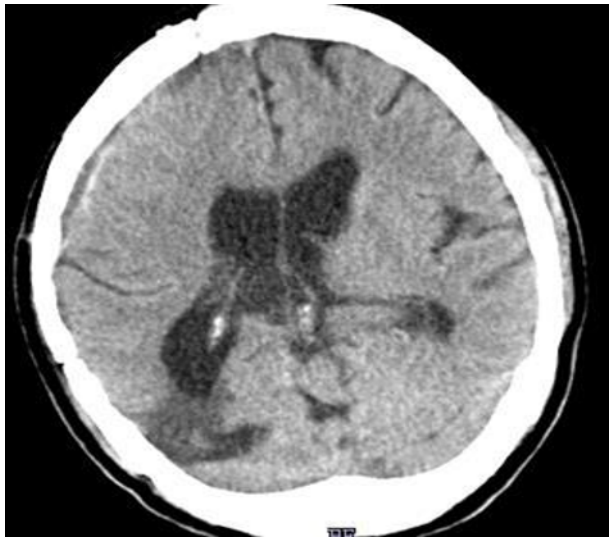

Figure 2: Hypodense area noted in the right parieto occipital region - likely to represent old infarct/sequelae to old intracranial bleed.

DISCUSSION

Osler Weber Rendu syndrome is a systemic disease with multiple organ involvement associated with the mutation in ENG or ACVRL1 genes. They begin to show the symptoms of hepatic, gastrointestinal, and cerebral damage. Pulmonary arteriovenous malformations form the majority of complications in the patients (Botella *et al.*, 2015). HHT patients may have severe cerebral deformity attributed to arteriovenous malformations. The patients complain of auditory and visual disturbances, headache, syncope, vertigo (Guttmacher *et al.*, 1995). About 90% of the patients show epistaxis, which is the spontaneous bleeding from the nasal mucosa due to the mucosal telangiectasia. The disease usually goes unsuspected in patients, and anemia is usually managed by iron supplements and blood transfusions in severe cases. The condition may be self-limiting, but the need for medical intervention ensues (Jolobe, 2017). Based on the severity of lesions and the site of location, various treatment modalities for HHT can be chosen like pharmacological, surgical, or both. The treatment's principal aim should be in reducing the vascular lesions and the complications associated with it. A combination of estrogen and progesterone helps increase the vascular wall thickness, but they being hormonal therapy, their use is often limited. Aminocaproic acid and tranexamic acid (anti-fibrinolytic), to an extent, reduce the risk of bleeding in these patients (Sadick *et al.*, 2006). A novel treatment approach entails bevacizumab and thalidomide though their use has not been clinically approved. Bevacizumab (5mg/kg every 14 days for total 6 IV injections) reduces the bleeding and hepatic complications, as the drug is an anti VGEF monoclonal antibody ending in an absolute reduction of angiogenesis (Maestraggi *et al.*, 2015; Kaliyadan, 2008).

CONCLUSION

Only in patients with severe and recurrent melena, the focus of care is managing anemia and suggesting endoscopy. Since the disorder advances gradually and often goes unrecognized or misdiagnosed, it worsens the condition, leading to the delayed

availing of treatment. Here we emphasize that gastroenterologists and professionals from other disciplines are pivotal in managing acute and chronic manifestations of HHT. Therefore, it seems crucial to record all cases found for them to get more conventional with the condition. Understanding and getting familiar with each case noted would help the professionals comprehend and equip them to be diagnostically conclusive about the disease at an early stage, by which half the battle is done.

Moreover, a thorough knowledge would further permit the physicians to detect any deviation or contrast in clinical manifestations from routine. This detection skill, which is only acquired by the meticulous observation, seems imperative because the primary therapy for HHT is organ dependent supportive care. Therefore, it is quintessential to study and analyze every case, which ultimately lends them with an idea of what treatment plan would work best for a particular individual, thereby shielding the sufferers from a break down with added complications. Hence, having a customized plan could serve the purpose of further enhancing patients' quality of life.

Also, all patients initially and predominantly reported with gastrointestinal manifestations of HHT must be strictly screened, as the patient may suffer from congestive heart failure, bleeding in the gastrointestinal tract, and portal hypertension. Furthermore, by reporting this case, we tend to update & take notice of the occurrence on one more case of a rare syndrome, highlighting the demand for gene therapies to be in action so that a standard therapy set in soon.

Conflict of interest

The authors declare that they have no conflict of interest for this study.

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