ENT MANIFESTATIONS IN HEREDITARY HEMORRHAGIC TELANGIECTASIA: A CASE REPORT
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ABSTRACT: Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant disorder, which affects various internal organs and has a tendency for bleeding. It has a classic triad of mucocutaneous telangiectasias, recurrent hemorrhages and positive familial history of first degree relative. Here we report a clinically proven case of Osler Weber Rendu syndrome in a female patient.

KEYWORDS: Arteriovenous malformation, Epistaxis, Hereditary hemorrhagic telangiectasia, Osler Weber Rendu syndrome.

INTRODUCTION: Hereditary hemorrhagic telangiectasia (HHT), also known as Osler Weber Rendu disease, is a rare systemic fibrovascular dysplasia which bears, as basic defect, an alteration in the elastic and muscle layers of vessel walls, making them more vulnerable to spontaneous ruptures and injuries.¹,²

The disease is autosomal dominant, although in about 20% of the cases, there is no family history.³ The incidence in the general population is of 12/100,000 and has a homogeneous race and gender distribution.³

CASE REPORT: A 48 year old woman came with complaints of recurrent episodes of epistaxis and Bleeding from oral cavity since 15 years with worsening of the clinical Picture in the 6 months, minimum of two to three episodes in a month and so patient underwent endoscopic nasal cautery. Patient had a family history (mother) of similar complaints. Patient did not give any history of having cough, nausea, vomiting, abdominal pain, fever, sweats or chills, and dysphagia. Patient had good appetite, moderate sleep and no weight loss.

ON EXAMINATION: On ENT Evaluation.
NOSE: Little’s area in right nasal cavity (Figure 1) and left nasal cavity (Figure 2) showed telangiectatic spots and fibrotic bands seen as the result of cautery.
ORAL CAVITY: Telangiectatic spots seen on the anterior 2/3 rds of tongue (Figure 3).
CASE REPORT

Patient underwent Portal venous doppler, which revealed Chronic parenchymal liver disease with portal cavernoma. Imaging of the abdomen showed features suggestive of Portal cavernoma, Cirrhosis of liver and Intra hepatic and Extra hepatic porto shunt system and cholelithiasis. With these findings, a diagnosis of HHT was made. And patient was managed conservatively with topical hemostytic agents and nasal packing and supportive treatment was given with vit k injections and systemic hemostytic agents.

**DISCUSSION:** HHT, first recognized in 1896,[4] is an inherited disorder and there is no age cutoff.[4]

Curacao criteria are used for the clinical diagnosis of HHT, consisting of recurrent epistaxis, mucocutaneous telangiectasias, visceral AVM and an affected first degree relative.[5] Diagnosis is definite if three criteria are met; It is suspected or possible with two, and is unlikely if less than two criteria are met. Our case had a certain clinical diagnosis as she met three out of the four criteria for HHT: Recurrent epistaxis, telangiectasias of tongue, GI and hepatic AVMs, whereas familial history could not be confirmed as relatives were not available for direct medical examination.

Otorhinolaryngological manifestations are the most frequent. Recurrent epistaxis is the main complaint seen in more than 90% cases.[6]

Blood vessels from other regions may also be involved, especially those from the skin, lips and mouth in 80% of cases, Pulmonary Arteriovenous Malformations (AVM) in about 40% of cases, Hepatic AVMs in at least 30% of cases, gastrointestinal tract in about 15% of cases and brain (Cerebral AVMs in 10% and Spinal AVMs in 1% cases).[6]

HHT can also be confirmed by genetic analysis.[7] Some patients with a clinical diagnosis of probable or unlikely HHT do not show any symptoms. In such cases of absence of typical symptoms of HHT, only genetic analysis provides an accurate diagnosis.[8]

Pathogenesis in patients with HHT is yet to be elucidated.[8]

However, a germ line mutation in at least two distinct genes, endoglin and ALK1/ACVRL1, are found in the majority of HHT patients.[8] Mutations in endoglin or ALK1/ACVRL1 genes account for HHT type 1 and HHT type 2, respectively.[8] The precise mechanisms underlying HHT related vascular abnormalities are still uncertain, although recent studies suggest that endoglin or ALK1/ACVRL1 encode for proteins involved in angiogenesis, vascular remodeling and/or endothelial arteriovenous identity.[8]

Treatment is directed towards the presenting clinical manifestations, managing complications and giving supportive care.[9] Anemia from recurrent nasal or GI bleed is treated by oral/parenteral iron, combined estrogen progesterone preparations and blood transfusion in severe cases.[9] Argon plasma coagulation may be useful when the telangiectatic lesions in the gastrointestinal tract are localized.[9]

Management for epistaxis needs establishing the bleeding site, stopping the bleeding and treating the underlying cause.[9] Treatment of epistaxis involves the use of ointments (to reduce drying of nasal mucosa), nasal sprays, creams and hemostatic agents.[9]

Telangiectasias have been successfully treated with estrogens,[10] aminocaproic acid,[11] endoscopic thermal ablation,[12] and thalidomide.[13] Bevacizumab has been used for HHT and severe liver involvement.[14]
HHT is not that rare but remains under reported. Most patients have a normal life expectancy but about 10% die of complications viz. hemorrhage, or stroke. The first degree relatives of HHT patients should be screened for the condition. Many patients don't require treatment other than oral iron supplements, where as some may require transfusions and nasal packing.

Surgery has limited use but may be useful in emergency viz. septal dermatoplasty and septal closure in which it has been found to be effective. Despite numerous case reports, understanding of the disease is not fully appreciated by clinicians, who often fail to recognize the disorder until severe manifestations occur. A recent study estimated a diagnostic delay in HHT showing that patients receive a definite diagnosis only after nearly 3 decades from disease onset.

A higher degree of suspicion and awareness is needed to identify this disease early, to reduce morbidity, and to improve outcomes and quality of life.

INFORMED CONSENT: Written informed consent was obtained from patient who participated in this.

CONCLUSION: Hereditary Hemorrhagic Telangiectasia is a multisystemic disease, first manifested by repetition epistaxis. Thus, it is fundamentally important that the otorhinolaryngologist be up to date in relation to the disease's etiopathogenesis and treatment options, so as to perform correct diagnosis as well as to prevent systemic complications of this disease.

A higher degree of suspicion and awareness is needed to identify this disease early, to reduce morbidity, and to improve outcomes and quality of life. Hence this case presentation.

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