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Case report

OSLER-WEBER-RENDU SYNDROME: A RARE CAUSE OF UPPER GI BLEED

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ABSTRACT

The causes of upper gastrointestinal bleed are manifold with the most common being peptic ulcers and oesophageal varices. We present a rare cause of upper gastrointestinal bleeding due to Hereditary Hemorrhagic Telangiectasia presenting with hematemesis and malaena due to bleeding from telangectatic lesions in the stomach and duodenum. The patient also had typical mucosal and nail manifestations of the disease which aided in the diagnosis of the condition.

Keywords: Osler Weber Rendu, Hereditary Hemorrhagic telangectasia, Upper GI Bleed

INTRODUCTION

Hereditary Hemorrhagic Telangiectasia (HHT)[1,2] is a disorder characterized by multiple telangiectasias in the gastrointestinal tract, central nervous system and lungs. The criteria to be satisfied for the diagnosis of HHT[3] are epistaxis, mucocutaneous telangiectasias, and a positive family history. We report a typical case of HHT who presented to us with history of bleeding from oral and nasal cavity since several decades and recent onset of hematemesis and malaena. There was positive family history of epistaxis as well. Diagnosis was delayed as the patient did not seek tertiary level care for her complaints for several years and it was only the distressing complaint of blood stained vomiting which prompted her to seek an expert opinion.

CASE SUMMARY

A 65 year old female was brought to the Medicine Casualty with a history of hematemesis and melaena. She had recurrent epistaxis since 30 years of age, 2 episodes of bleeding from the tongue 20 years ago, recurrent episodes of melaena for the past 20 years and one episode of bleeding from the nails five years ago. Two of her siblings had epistaxis in the past. Her youngest son has epistaxis for past 2 years. On examination, she was pale, with no icterus, clubbing, or pedal oedema. Her vitals were stable. She had multiple telangiectasias in the nail bed (Fig. 1) and multiple telangiectasias in the oral cavity and tongue (Fig. 2). She had no hepatosplenomegaly or ascites. Her respiratory and central nervous system were within normal limits. Cardiovascular system revealed cardiomegaly. With the history of mucosal bleed. A positive family history, and multiple mucosal

telangiectasias, a diagnosis of Hereditary Hemorrhagic Telangiectasia (HHT) was made.

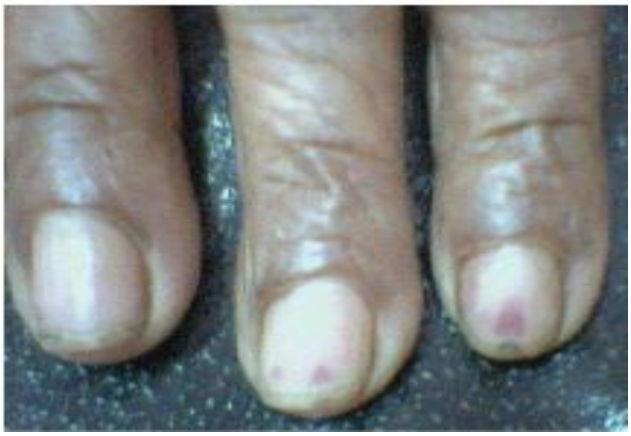

Fig. 1: Nail telangiectasias

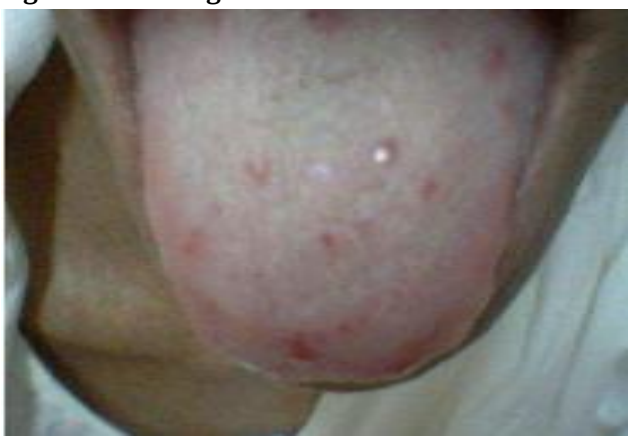

Fig. 2: Tongue And Palatte showing telangiectasias.

Investigations

Laboratory investigations revealed hemoglobin of 6.5g/dL , total count of 6600/mm^3 , differential count of 84 % neutrophils, 12% lymphocytes, and 4 % eosinophils , red cell count of 3.3 million/mm^3, haematocrit of 20%, mean corpuscular hemoglobin of 16.3pg, mean corpuscular volume of 62.5fl, a platelet count of 2.77 lakhs/mm^3, and erythrocyte sedimentation rate of 55 mm/hr. Peripheral smear confirmed microcytic hypochromic anemia. Her bleeding time and clotting time were 3mts and 6 mts respectively. Her prothrombin time was 16s (control 14s). Antinuclear antibody was negative. Her blood group was O positive. Chest X ray revealed cardiomegaly, with aortic knuckle calcification. ECG showed ST depression in the lateral leads. Echo-cardiogram showed good LV function, with no regional wall motion abnormality. Ultrasound showed fatty changes in her liver with prominent hepatic veins and no splenomegaly or ascites. Her portal vein size was 10.8mm. Endoscopy showed multiple hemorrhagic spots in fundus, body, and antrum of stomach (Fig. 3). She was managed with blood transfusion and supportive measures. She had no bleeding from other sites.

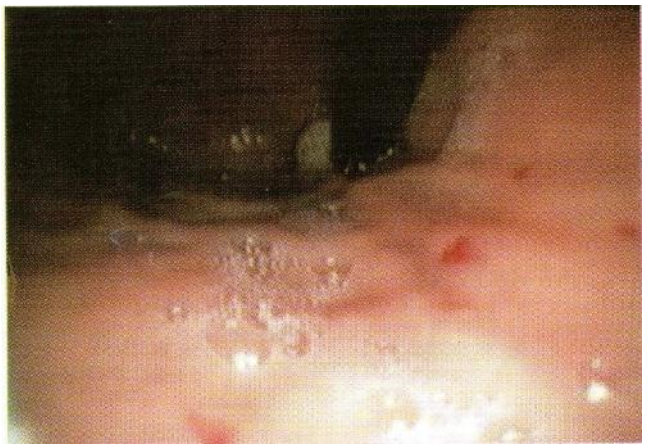

Fig. 3: Endoscopy showing multiple telangiectasias in the gastric fundus.

DISCUSSION

Heriditary Hemorrhagic Telangectasia or HHT was first described by Legg in 1876[4]. Later it was described separately by Rendu. Weber, and Osler[5]. 80 % of cases have a positive family history. There is increased frequency of HHT in blood group 'O' which happens to be the blood group of our patient as well. The criteria for diagnosis are epistaxis, mucocutaneous telangiectasia, and a positive family history. The earliest sign of this disease is epistaxis. Epistaxis and mucocutaneous lesions are constant features. Pulmonary involvement is seen in 30 % of cases, 70 % of which are in the lower lobes and 10 % seen bilaterally. The patient becomes symptomatic if the arteriovenous malformation (AVM) is more than 2 cm. The most common complaint being dyspnoea, followed by hemoptysis and bruit over the chest. Clubbing and cyanosis occurs in some cases due to right to left shunt in the lung. CT thorax and angiography can diagnose the lesions. In the gastrointestinal

tract, AVMs is seen most commonly in the stomach and small bowel. Hepatic involvement occurs in 10 % of cases which is characterized by high output cardiac failure, portal hypertension and biliary tract disease[6]. Upper GI endoscopy, angiography and Doppler aids in the diagnosis. Central nervous system manifestations include cerebral abscesses and embolism due to right to left shunt, and cerebral AVMs. Cerebral AVMs are seen in 10% of cases which are detected on MRI and MR Angiogram.

HHT is an autosomal dominant disease with two loci[7]. Depending upon the genetic mutation HHT is divided into two types - HHT 1 & HHT 2. The genetic locus of HHT 1 is an Endoglin gene on 9q3 and codes for TGF binding protein expressed on endothelial cells[7]. There is increased incidence of pulmonary AVMs in HHT 1. The genetic locus of HHT 2 is ALK 1 gene on chromosome 12 which encodes a member of TGF B receptor family[7]. The telangiectasias are angiodysplastic with insufficient smooth muscle contractile element and perivascular connective tissue weakness. There is increased circulating tissue plasminogen activator leading to the impaired thrombus formation. There is increased incidence of DIC, Factor IX deficiency and type 2 Von Willebrand's disease. Skin and digestive tract lesions can be managed by NdYAG laser ablation[2]. Pulmonary and central nervous system AVMs are tackled using interventional radiology techniques[3,8]. Gastrointestinal lesions can be treated by Epsilon Amino Caproic Acid[9], Estrogen and Progesterone[10], and endoscopic ablation. Liver AVMs are significantly harder to treat and liver transplantation may be the only curative therapy which can be offered[11]. Experimental treatments with anti-angiogenesis drugs have been evaluated in small clinical trials and have found have beneficial effects. Bevacizumab, an anti-VEGF antibody was found to reduce number and severity of episodes of epistaxis[12,13] and Thalidomide, another drug with anti-angiogenic properties was also found to be effective [14].

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