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A Case Report on Osler -Weber Rendu Disease

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Abstract

Osler weber rendu disease, also known as hereditary hemorrhagic telengiectasia is a rare autosomal dominant genetic disorder that leads to abnormal blood vessel formation in skin, mucous membrane and in organs such as lungs, liver and brain. A 68 year old postmenopausal lady was admitted to our hospital following multiple episodes of hematemesis and malena for past 5 days. Upper GI endoscopy revealed multiple telengiectasia with active spurting, gastric polyp. Patient was treated with 1 unit of PRBC transfusion for anemia, Inj.Tranexamic acid 500 mg IV tid, Inj. Ondanseteron 4 mg IV for hematemesis. Tab. Thalidomide 100 mg hs have been given for treating hereditary hemorrhagic telengiectasia. The treatment is only palliative, with no consensus on the best treatment option. It is essential to promote control of the disease as long as possible.

Introduction

Osler weber rendu disease, also known as hereditary hemorrhagic telengiectasia is a rare autosomal dominant genetic disorder that leads to abnormal blood vessel formation in skin, mucous membrane and in organs such as lungs, liver and brain. In small blood vessels, these abnormalities are called telengiectasias, when they occur in larger vessels they are called arteriovenous malformations. Both are potential source of serious morbidity and mortality. Recent epidemiological studies revealed prevalence of 1:5000-8000 persons worldwide.HHT has a higher prevalence in certain population such as in afro Caribbean residents. [1] The name Osler weber render syndrome for the doctors who worked on researching this condition.

Case Report

A 68 year old postmenopausal lady was admitted to our hospital following multiple episodes of hematemesis and malena for past 5 days. The patient has co morbidities such as mild pulmonary arterial hypertension and asthma under treatment. She had multiple episodes of epistaxis, palpitations, abdominal pain. There was no history of bleeding gums and rectum, headache, seizure, visual disturbance or bluish discoloration of fingertips or nose. Family history with brother having telengiectasia was known.

On general examination patient was conscious, oriented, and anemic with pallor, abdominal tenderness in abdomen and absence of organomegaly. No abnormalities were detected on chest x-ray. The laboratory work-up at admission revealed the following: WBC, 2,200cells/mm3; platelet count, 250,000/mm3; hemoglobin, 6.9 g/dl and serum iron 34 ug/dl. Investigations including bleeding time, coagulation time, and prothrombin time (PT), activated partial thromboplastin time (aPTT), and stool occult blood reported to be normal. Endoscopy of the upper digestive tract was performed. Serum for antinuclear antibody (ANA), urine for hemoglobinuria and sickling test were found to be negative. Upper GI endoscopy revealed multiple small, sessile, numerous telengiectasia in the pharynx and oesophagus (figure: 1).Tiny polyps were evident at the fundus region, active spurting telengiectasia seen at proximal body (figure: 2), multiple telengiectasia and small polyps were found at antrum and duodenum (figure: 3 & figure: 4), active ooze noted at proximal body along the lesser curve.

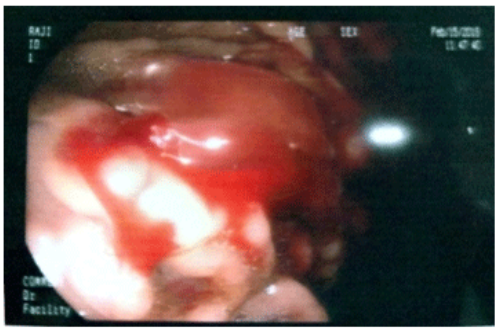


Figure 1: Tiny polyps were evident at the fundus region, active spurting telengiectasia seen at proximal body

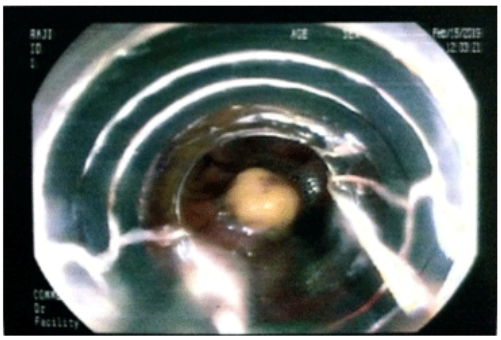
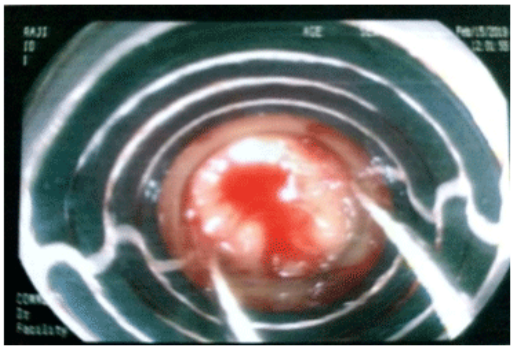


Figure 2: Multiple telengiectasia and small polyps were found at antrum and duodenum



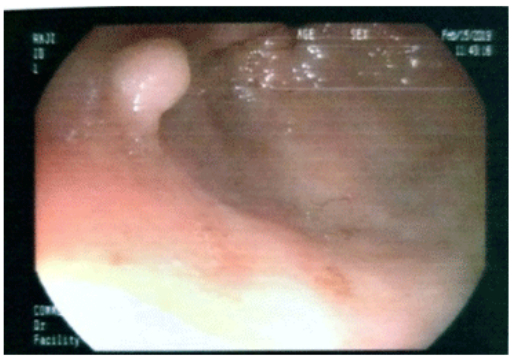


Figure 3&4: Active ooze noted at proximal body along the lesser curve

Patient was treated with 1 unit of PRBC transfusion for anemia, Inj.Tranexamic acid 500 mg IV tid, Inj. Ondanseteron 4 mg IV for hematemesis.Tab.Thalidomide 100 mg HS has been given for treating hereditary hemorrhagic telengiectasia. Hence the severity of nasal bleed and hematemesis were significantly reduced, haemoglobin level improved and there was no side effects reported by the patient.

Discussion

Diagnosis is based on the Curaçao criteria:

1) **Epistaxis** - spontaneous, recurrent nosebleeds
2) **Telangiectasias -** multiple, at characteristic sites (lips, oral cavity, fingers, nose)
3) **Visceral lesions -** such as gastrointestinal telangiectasias (with or without bleeding), pulmonary AVM, hepatic AVM, cerebral AVM, spinal AVM
4) **Family history -** a first-degree relative with HHT. Three criteria indicate a definite diagnosis of the disorder; two a possible or suspected case [1]. The primary and most common manifestation of HHT is usually epistaxis that begins during childhood or adolescence at a mean age of 12 years. [2] Telangiectasias do not usually appear until after puberty but may not occur until adulthood. They typically occur on the face, lips, tongue, palms, and fingers including the periungual area and the nail bed. Telangiectasias are dilated blood vessels that appear as thin spider web-like red and dark purple lesions that blanch with pressure. AVMs are abnormal connections between arteries and veins that bypass the capillary system. Patients with HHT have multiple AVMs throughout the body. However, the most important AVMs for which clinicians should screen are in the brain, lungs, GI tract, and liver. AVMs in the lung and brain can be asymptomatic [3].

The overall treatment of HHT is oriented towards the predominant clinical manifestation and its severity. The first step in epistaxis management should always be necessary patient counseling and use of preventive measures within the home to prevent the nasal mucosa from becoming dry. These include nasal humidification, use of over-the-counter saline sprays or ointments to keep the nasal mucosa moist and avoidance of nasal trauma as per Athena Kritharis, Hanny Al-Samkari et al, [4]. In a double blind randomised controlled trial, 50 µg ethinyloestradiol and 1 mg norethisterone resulted in a significant reduction in transfusion requirements in 10 patients with a mean transfusion requirement of 19.4 packed cells units per year. These data have been extrapolated in clinical practice to patients with lesser degrees of hemorrhage not necessarily associated with regular transfusion requirements. The use of higher dose conjugated oestrogens in the "hormone replacement" range of over 625 µg ethinyloestradiol equivalent, or prothrombotic agents such as tranexamic acid and aminocaproic acid are also widespread in management of HHT related bleeding as per M E Begbie, G M F Wallace, C L Shovlin et al [5]. Hormonal or antifibrinolytic therapy may be used as adjunct therapy to prevent ongoing bleeding. In patients that have liver AVMs, embolization is not recommended given the risk post-embolization necrosis and death. Surgical intervention should only be considered if they become symptomatic or develop complications. For those with significant hepatic AVM involvement, partial liver resection is a safe treatment option. If a patient's hemoglobin and hematocrit are low, an upper endoscopy should be completed if the anemia is disproportionate to epistaxis. Iron supplementation should be initiated, either oral or intravenous (IV) and endoscopic cauterization can be considered. It is recommended that acute epistaxis be managed with low-pressure, less-traumatic packing techniques. In some studies, a mild benefit was shown for the use of humidifiers to prevent chronic epistaxis [5].

Anti angiogenic factors can be useful in treating vascular malformations, such that thalidomide inhibits tumor necrosis factor-alpha, thereby acting as a potent anti angiogenic drug in case of this patient as well.

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