CASE REPORT

Osler–Weber–Rendu syndrome

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Learning points for clinicians
Gastrointestinal bleeding occurs in about one-third of patients with Hereditary Hemorrhagic Telangiectasia, however, it contributes less frequently to iron deficiency anemia in patient than unrecognized nasal bleeding. Early recognition with localized management is important.

Introduction
Osler–Weber–Rendu syndrome, also known as Hereditary Hemorrhagic Telangiectasia (HHT), is an autosomal dominant disorder. Telangiectasias and Arterio-Venous Malformations (AVMs) are vascular lesions present in HHT, most commonly causing epistaxis and gastrointestinal bleeding.¹ While epistaxis presents as early as childhood, the gastrointestinal manifestations of HHT develop with increasing age.²

Case presentation
A 62-year-old Puerto Rican male presented to emergency room because of generalized weakness fatigue, pallor. Patient reported melena and occasional hematochezia. Vitals were within normal limits. Systemic examination was significant for telangiectasias on oral mucosa and tongue. Patients stool was found frankly positive for fecal occult blood test.

Complete blood count showed hemoglobin of 3.8 g/dl and hematocrit of 13.2%. Reticulocytes were high. Red cell indices showed Microcytic Hypochromic anemia. Iron studies showed anemia of iron deficiency from chronic blood loss. His colonoscopy showed up to 36 colonic AVMs. Esophagogastroduodenoscopy showed multiple AVMs (Figure 1A and B) which were treated with argon plasma coagulation. Patient received 6 units of packed RBCs. Ferrous sulfate and pantoprazole were initiated. Patient improved symptomatically and hemoglobin increased to 8.1 g/dl by discharge.

Discussion
Majority of patient with HHT experience epistaxis, mucocutaneous telangiectasia and a tendency to develop gastrointestinal bleeding leading to iron deficiency anemia.¹

Gastrointestinal bleeding is present in about one-third of the patients with HHT and the approach to which focuses on localizing the source of the bleed. Although vascular lesions in HHT can be present throughout the gastrointestinal tract, AVMs are most commonly found in the stomach and duodenum.³

Patients that have a long standing history of recurrent epistaxis, gastrointestinal bleeding and symptomatic iron deficiency should be investigated for HHT.

International consensus diagnostic criteria have been developed based on the presence of the following four findings:
1. Spontaneous and recurrent epistaxis.
2. Multiple mucocutaneous telangiectasia.
3. Visceral involvement with AVM.
4. Family history of HHT.

Based on this criteria, patients can be classified as ‘definite’, ‘suspected’ and ‘unlikely’ when three or four, two, or zero to one of the above mentioned criteria is present.⁴

Management of HHT is primarily aimed at treating the resultant iron deficiency anemia and preventing complications of vascular lesions. Localized treatment options are used for localized source of bleeding with symptomatic HHT. This usually allows the patients to avoid systemic therapy with side effects.

Conflict of interest: None declared.

Submitted: 23 May 2016; Revised (in revised form): 2 June 2016

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References


Figure 1. (A) Stomach—arteriovenous malformations in antrum with antral bleeding. (B) Stomach—post APC of antral AVMs.