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Hereditary hemorrhagic telangiectasia: A rare genetic disorder causing recurrent anaemia

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Abstract

Gastrointestinal (GI) vascular lesions are one of the most common causes of occult bleeding. The term angiodysplasia and telangiectasias are used interchangeably but angiodysplasias are confined to the GI tract and vascular telangiectasias are a part of systemic diseases like systemic sclerosis or genetic disorder like Hereditary Hemorrhagic Telangiectasia (HHT). Here we present a case of a middle-aged lady who presented to us with a history of easy fatigability and unexplained iron deficiency anemia. She had a history of recurrent epistaxis.

On examination, the patient had lower palpebral telangiectasia. On investigation, gastrointestinal endoscopy revealed multipletelangiectasias throughout the entire GI tract. Based on the history, clinical examination and endoscopy she was diagnosed to have HHT. She was successfully managed with thalidomide. High clinical suspicion is required to diagnose these patients. A thorough physical examination may reveal mucocutaneous telangiectasias which can help to clinch the diagnosis.

Keywords: Occult gastrointestinal bleed; Hereditary hemorrhagic telangiectasia; Arterio-portal shunt; Thalidomide.

Abbreviation: EGD: Esophagogastroduodenoscopy; GI: Gastrointestinal; HHT: Hereditary Hemorrhagic Telangiectasia.

Description

A female in her 40s presented with a history of gradually progressive easy fatigability for 12 years. On the subsequent inquest, the patient revealed a history of occasional spontaneous nose bleeds from 20 years of age but she did not have a history of recent significant nose bleeds. Detailed family history revealed that her father had a history of recurrent epistaxis. On examination, the patient had pallor, and the lower palpebral conjunctiva showed the presence of tortuous and dilated vessels suggestive of telangiectasia (Figure 1A). An Esophago Gastro Duodenoscopy (EGD) and colonoscopy also showed the presence of telangiectasias (Figure 1B, C, D and E). Based on the presence of recurrent epistaxis and mucosal telangiectasia in a patient with a family history of epistaxis, according to Curacao criteria, the patient was diagnosed with Hereditary Hemorrhagic Telangiectasia (HHT). Her triple-phase computed tomography and angiography of the abdomen revealed abnormal communication of the portal vein and hepatic artery with volume redistribution. However, there was no evidence of cirrhosis, ascites or portal hypertension. The patient was started on the tablet Thalidomide 100 mg once a day. On follow-up, her hemoglobin improved, with no further episodes of bleeding from any site. HHT is an autosomal dominant hereditary disorder with incomplete penetrance. Around 80% of patients with HHT present with recurrent epistaxis, whereas skin lesions appear late in the disease course [1]. Stomach and small bowel involvement are found in 64 to 91% of patients which can be picked up on

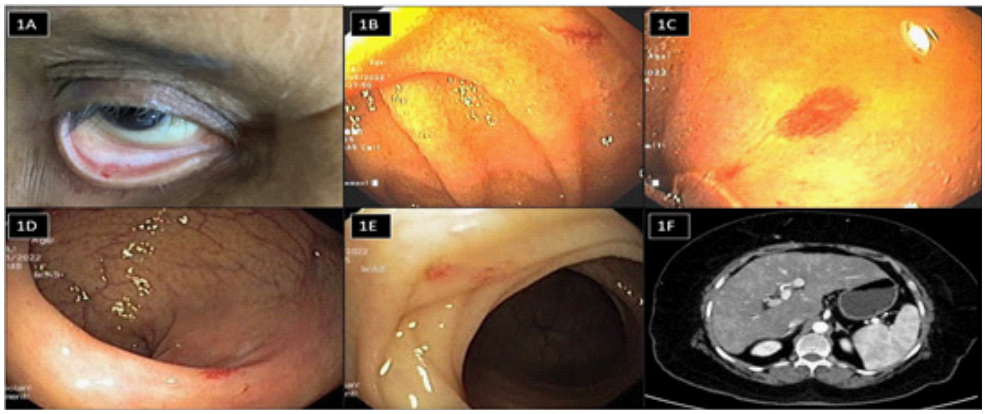

Figure 1: (A): The lower palpebral conjunctiva showing the presence of telangiectasia; **(B,C,D,E):** Esophagogastroduodenoscopy and colonoscopy images showing the presence of telangiectasias; **(F):** A computed tomography scan showing a suspected communication of the portal vein and hepaticartery with out any other evidence of vascular malformations.

endoscopy [2]. The liver can also be involved in HHT. Abnormal vascular connections in the liver are seen in 70% of the patients, and mostly they are asymptomatic [3]. Conservative management options include family screening, iron replacement therapy for anemia, genetic counselling, and anti-angiogenic drugs [4].

Declarations

Conflict of interest: None of the other authors has any conflict of interest to declare concerning this paper.

Informed consent: Consent was taken from the patient for the publication of this report and the images.

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