CLINICAL IMAGE

Hereditary hemorrhagic telangiectasias as a rare cause of recurrent gastrointestinal bleeding

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A 52-year-old man was referred to our department because of gastrointestinal bleeding presenting as tarry stools, severe to moderate anemia, and fatigue for over 6 months. His past medical history included recurrent epistaxis and 2 syncopal episodes over the previous 8 years. His father died in his forties of heart attack, and the remaining family history was unremarkable. Several months before admission, the patient underwent endoscopic evaluation in another hospital, showing multiple gastric and duodenal vascular lesions and small hyperplastic colon polyps removed during colonoscopy. At that time, the treatment was confined to blood transfusions and oral iron supplementation. On admission, a physical examination showed red macular telangiectasias on the labial mucosa, tongue (FIGURE 1A), and trunk. Severe normocytic anemia with a hemoglobin level of 7.7 g/dl was found. On the basis of clinical picture, he was diagnosed with hereditary hemorrhagic telangiectasia (HHT) based on the Curaçao criteria.[1] No arteriovenous malformations of the brain, lungs, and liver were identified on magnetic resonance imaging or high-resolution computed tomography. Coronary angiography showed normal coronary vessels. Upper gastrointestinal endoscopy revealed multiple telangiectasias of 2 to 6 mm in diameter in the duodenum and stomach (FIGURE 1B) and single telangiectasias in the esophagus. Capsule endoscopy was performed showing numerous flat telangiectasias distributed mainly in the jejunum and duodenum (FIGURE 1C). Subsequently, the patient underwent 4 push enteroscopy procedures with argon plasma coagulation of the dominant lesions. Since the procedures had a moderate effect, the patient was additionally put on oral estrogen-progesteron hormonal therapy. During the follow-up, the clinical symptoms resolved and control levels of hemoglobin ranged from 11.5 to 12 g/dl.

HHT, also known as Osler–Weber–Rendu disease, is a dominantly inherited genetic disorder of blood vessel development, characterized by epistaxis, mucocutaneous telangiectasias, and visceral arteriovenous malformations (AVMs). Mutations in at least 5 genes are associated with HHT;

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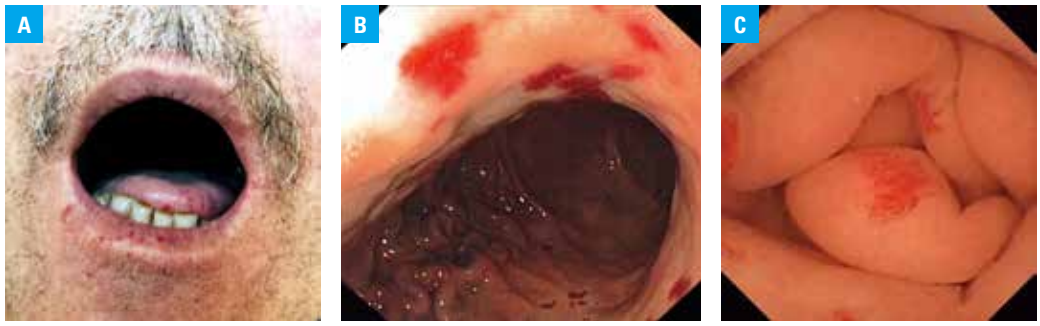

FIGURE 1 A – lip and tongue telangiectatic lesions; B – endoscopic image of telangiectasias in the stomach; C – capsule endoscopy image showing multiple telangiectasias in the jejunum

however, 2 of them (*ENG* and *ACVRL1/ALK1*) are responsible for most of the cases.[2,3] The diagnosis of HHT is based mainly on the clinical picture, although genetic testing may be required in doubtful cases. The clinical Curaçao criteria[1] include epistaxis, telangiectases, visceral lesions (organ AVMs and gastrointestinal telangiectasia), and family history. Our patient fulfilled 3 criteria, which corresponds to a definite diagnosis.

The major clinical presentation of HHT is epistaxis, which occurs in 90% of patients, usually at a younger age. In the further course of the disease, AVMs may develop in pulmonary, hepatic, and cerebral vasculature, causing a broad spectrum of complications including pulmonary arterial hypertension, pulmonary hemorrhage, and cerebral abscesses.[3] Moreover, gastrointestinal telangiectasias are a common source of hemorrhage, which may contribute to iron deficiency anemia. In severe cases of gastrointestinal bleeding, endoscopic or surgical intervention is necessary; however, mild bleeding can be controlled pharmacologically. Systemic estrogen-progesterone therapy has been proved to reduce bleeding in patients with HHT.[4,5] Other medications used in these patients include aminocaproic acid, immunomodulators such as interferon α or thalidomide, and monoclonal antibodies such as bevacizumab.[2,3]

Taken together, we presented a rare case of a patient with gastrointestinal bleeding due to HHT, in whom an appropriate diagnosis and treatment resulted in a good clinical outcome.

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