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Brief Report

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An unusual gastric submucosal prominence diagnosed as hereditary hemorrhagic telangiectasia

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Introduction

Hereditary hemorrhagic telangiectasia (HHT), also known as Osler-Weber-Rendu syndrome, is an autosomal dominant disease that can frequently be complicated by the presence of significant arteriovenous malformations (AVMs) in the brain, lungs, gastrointestinal tract, and liver [1]. The most prevalent hemorrhagic complications in patients with HHT include epistaxis, gastrointestinal hemorrhage, and anemia due to chronic blood loss [2]. This case involved an unusual gastric prominence that was initially diagnosed as a gastrointestinal stromal tumor (GIST); however, further investigations culminated in a diagnosis of HHT. The endoscopic appearance of this type of gastric prominence has not been previously reported in cases of HHT, providing a fresh perspective on the endoscopic manifestations of the disease.

Case report

A 64-year-old woman was admitted due to two episodes of hematemesis a month ago, each involving the expulsion of ~500 mL of blood. Initially, she was treated at a local hospital. The gastroscopy identified a submucosal prominence with superficial ulceration on its surface at the lesser curvature of the stomach, near the angular incisure (Figure 1A), tentatively suspected to be a GIST. Subsequently, she was referred to our hospital for further endoscopic intervention. On admission, her vital signs were stable, and no obvious abnormalities were found during the physical examination. The endoscopic ultrasound complemented by Doppler flow imaging revealed a pronounced blood flow signal within the prominence (Figure 1B and C). To further investigate the blood supply to the gastric submucosal prominence, the abdominal contrast-enhanced computed tomography (CT) examination and vascular interventional angiography were performed. The contrast-enhanced CT illustrated an augmented vessel sign at the lesser curvature of the stomach (Figure 1D). The arterial and portal venous phases of CT sections revealed tortuous hepatic arteries, aneurysmal dilatation of the proper hepatic artery, and early filling of hepatic veins, suggestive of a hepatic arteriovenous fistula (Figure 1E and F).

Given the findings from the CT images, HHT was suspected. Subsequently, a detailed physical examination was repeated, revealing multiple telangiectasias on the tongue, lips (Figure 1G), and fingertips (Figure 1H). Upon inquiry about epistaxis, the patient supplemented that she, her son, and her daughter all experienced intermittent nasal bleeding. Thus, the diagnostic criteria for HHT were fulfilled. The prominence noted may represent a mass composed of dilated and tortuously deformed microvasculature. To further elucidate the characteristics of the submucosal prominence, vascular interventional angiography was performed. The celiac trunk angiography revealed tortuous hepatic arteries and aneurysmal dilation, and faintly identified a cluster of tortuous small vessels at the lesser curvature of the stomach (Figure 1I and Supplementary Video 1). To verify the diagnosis and identify potential genetic mutations in the progeny, the patient and her children underwent whole exome sequencing. The result confirmed the diagnosis by identifying the variant NM_000020.2: c.1347delC (p.Thr450Profs*15) within the *ACVRL1* gene.

However, the patient declined the proposed treatment, citing the absence of subsequent gastrointestinal hemorrhage. Upon discharge, she was prescribed proton pump inhibitors and oral iron supplementation. The patient was monitored through telephone follow-ups. Over the next 6 months, she reported no episodes of gastrointestinal bleeding. It was advised that she undergo regular complete blood count monitoring. Should gastrointestinal bleeding recur, prompt intervention via endoscopy or angiography should be considered, and the antiangiogenic or antifibrinolytic agents are also recommended.

Discussion and conclusions

The diagnosis of HHT is established through the Curaçao criteria, which comprise four clinical features: recurrent epistaxis, mucocutaneous telangiectasias, visceral arteriovenous lesions, and a

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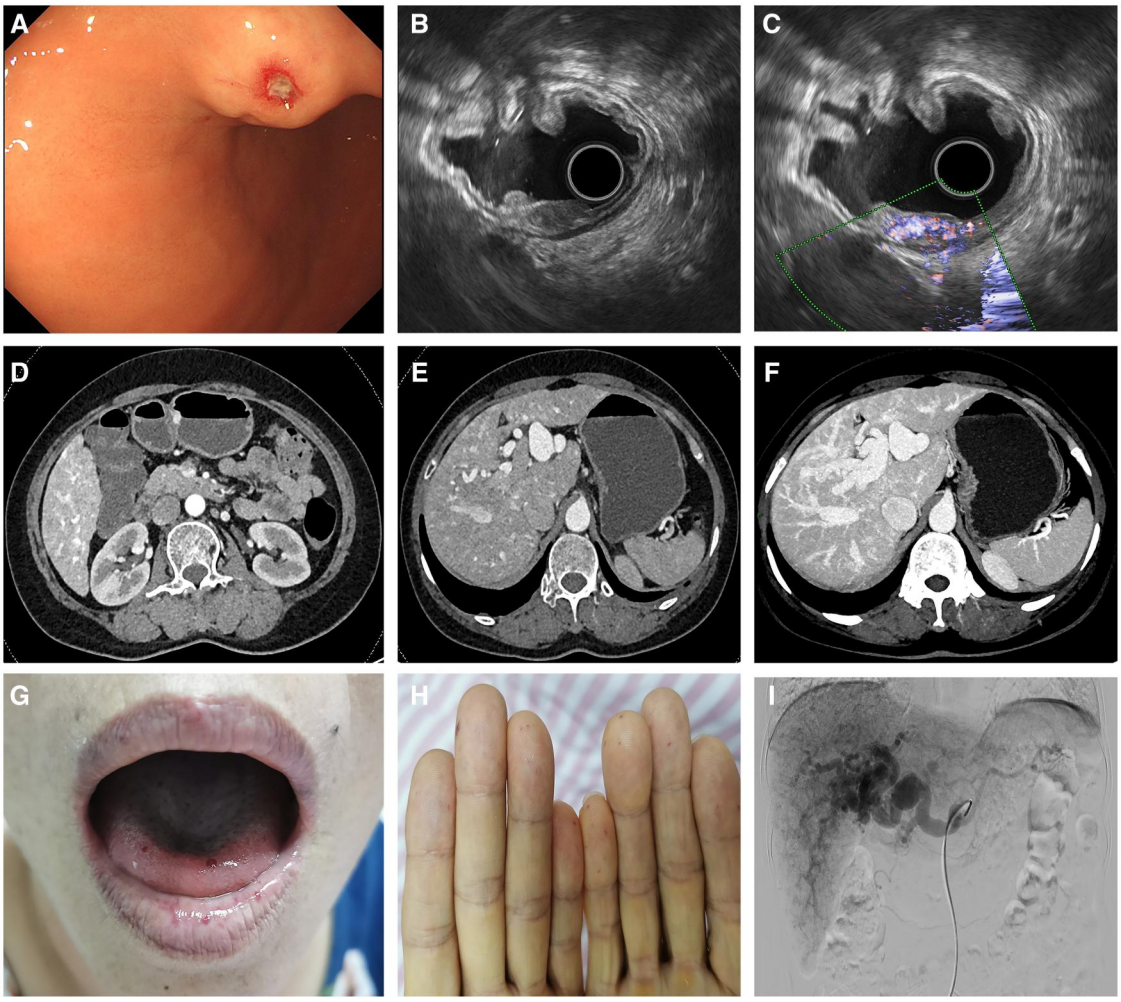

Figure 1. Clinical data of the case with hereditary hemorrhagic telangiectasia. (**A**–**C**) Endoscopy and endoscopic ultrasound showed a submucosal prominence at the lesser curvature of the stomach. (**D**–**F**) Contrast-enhanced computed tomography showed an enhanced vessel sign at the lesser curvature of the stomach, tortuous hepatic arteries, aneurysmal dilatation of the proper hepatic artery, along with precocious visualization of hepatic veins, indicating a hepatic arteriovenous fistula. (**G** and **H**) Multiple telangiectasias were found on the tongue, lips, and fingertips. (**I**) Celiac trunk angiography revealed tortuous hepatic arteries and aneurysmal dilatation of proper hepatic artery and specifically showed a cluster of tortuous small vessels at the lesser curvature of the stomach.

first-degree relative diagnosed with HHT. The clinical diagnosis is deemed definite with the presence of at least three features and considered possible or suspected when two of four criteria are met [3]. Gastrointestinal telangiectasias are thought to affect the majority of patients with HHT and are predominantly located in the stomach and proximal small bowel. Patients with HHT exhibit a high incidence of dilated and tortuous hepatic arteries, contributing to the specificity in the diagnosis of HHT [2, 4]. Over 80% of HHT patients possess identifiable gene mutations. Gene mutations described in HHT include *ENG*, *ACVRL1*, and *MADH4*, as well as other postulated loci [5]. HHT Type 1 (HHT1) results from mutations in the *ENG* gene, while HHT Type 2 (HHT2) is attributed to mutations in the *ACVRL1* gene. Liver AVMs may be more common in HHT2 than in HHT1 [6]. In this case, the patient was classified as HHT2 and exhibited vascular malformations in both the stomach and liver.

The management of HHT depends primarily on the specific complications manifested by the patients. Gastrointestinal telangiectasias can be treated through endoscopic techniques such as argon plasma coagulation [3]. Patients with recurrent bleeding, multiple AVMs, and small bowel AVMs may require additional pharmacological interventions [6]. The use of oral antifibrinolytics, intravenous bevacizumab, or other systemic antiangiogenic therapies could be considered by clinicians [3].

In this case, the endoscopic manifestation of HHT in the stomach differed markedly from previously reported cases, which typically presented as flaky mucosal erosion or shallow ulcers. Instead, a distinctive lesion characterized by a submucosal prominence with superficial ulceration was observed, an exceptionally rare phenomenon that remains undocumented. This case highlights that gastric prominences can occur not only in GISTs, leiomyomas, lipomas, neuroendocrine tumors, or ectopic pancreas, but can also be attributed to hemangiomas or vascular malformations. Assessing the vascularity of the submucosal prominence is crucial for determining the patient's subsequent treatment. The subtle telangiectasias on this patient's lips and fingertips were easily overlooked but could have facilitated an earlier diagnosis if recognized sooner. Consequently, it is imperative to perform a meticulous physical examination to detect vascular dilation on the skin and mucous membranes. Additionally,

this case emphasizes the importance of conducting endoscopic ultrasound with Doppler blood flow signals or contrast-enhanced CT prior to the resection of gastric prominences.

Supplementary data

Supplementary data is available at *Gastroenterology Report* online.

Authors' Contributions

L.Z. conceived the project and drafted the manuscript; H.L., J.N., and N.Y. helped to collect data; Y.L. revised the manuscript. All authors read and approved the final version of the manuscript.

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Conflicts of Interest

The authors declare that there are no conflicts of interest in this study.

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