American Journal of
Biomedical Science & Research

www.biomedgrid.com
ISSN: 2642-1747

Case Representation

Not Your Usual Gastrointestinal Bleed: Hereditary Hemorrhagic Telangiectasia Unmasked

Huipeng Zhang[1], Xia Xu[2], Ahemala Duishanbai[3], Xin Long[4], Jing Zhang[5] and Yanbo Yu[6]*

[1]*Huipeng Zhang, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan 250012, Shandong, China*

[2]*Xia X, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan 250012, Shandong, China*

[3]*Ahemala Duishanbai, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan, China*

[4]*Xin Long, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan 250012, Shandong, China*

[5]*Jing Zhang, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan 250012, Shandong, China*

[6]*Yanbo Yu, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan 250012, Shandong, China*

***Corresponding author:** Yanbo Yu, Department of Gastroenterology, Qilu Hospital of Shandong University, Jinan 250012, Shandong Province, China.

To Cite This Article: *Yanbo Yu, Not Your Usual Gastrointestinal Bleed: Hereditary Hemorrhagic Telangiectasia Unmasked. Am J Biomed Sci & Res. 2025 29(1) AJBSR.MS.ID.003753,* **DOI:** 10.34297/AJBSR.2025.29.003753

Received: October 28, 2025; **Published:** November 04, 2025

Abstract

Hereditary hemorrhagic telangiectasia is a rare autosomal dominant genetic disorder. Gastrointestinal bleeding is a common but often overlooked complication of HHT and is frequently misdiagnosed as a more common disease. We report a case of a 55-year-old woman with persistent black stool and anemia. These symptoms were initially attributed to Helicobacter pylori-related ulcers. After the conventional treatment proved ineffective, subsequent imaging examinations revealed hepatic arteriovenous malformations, which prompted us to conduct genetic testing to confirm the diagnosis of HHT type 2. Then we began targeted anti-angiogenic therapy. After that, the bleeding stopped and the hemoglobin level rose. This case demonstrates that HHT can simply present as gastrointestinal bleeding, that is, it can disguise itself as refractory gastrointestinal bleeding, emphasizing the importance of a comprehensive assessment including endoscopy and genetic evaluation.

Keywords: Gastrointestinal bleeding, Hereditary hemorrhagic telangiectasia, Melena, Endoscopic

Abbreviations: AVMs: Arteriovenous Malformations; HHT: Hereditary Hemorrhagic Telangiectasia; TGF-β: Growth Factor-Beta; ALK1: ACVRL1; H. Pylori: Helicobacter pylori

Introduction

Hereditary Hemorrhagic Telangiectasia (HHT) is a rare autosomal dominant genetic disorder. It is characterized by recurrent bleeding, telangiectasia, and Arteriovenous Malformations (AVMs) [1]. The most common symptoms include epistaxis, telangiectasia on the skin and mucous membranes, and AVMs in the lungs, liver, and brain. However, clinical observations have shown that some patients may present with uncommon symptoms confined to a single system, such as gastrointestinal bleeding [2]. The lack of sensitivity in diagnosing HHT can delay the use of effective treatment methods. This case report describes such a situation. After standard treatments for gastrointestinal bleeding failed to produce satisfac-

tory results, we discovered the presence of intrahepatic AVMs on the patient's CT scan which drawn our attention to HHT.

Case Presentation

A 55-year-old woman was admitted with intermittent melena and upper abdominal pain for eight years. Three months before presentation, she underwent a gastroscopy revealing a duodenal ulcer and Helicobacter pylori (H. pylori) infection, whereas colonoscopy showed no obvious abnormalities. After receiving standard treatment for duodenal ulcers and eradication of H. pylori, the symptoms did not improve significantly. Laboratory tests showed hemoglobin at 85.0 g/L and contrast-enhanced computed tomography demonstrated cardiophrenic angle and intrahepatic AVMs (Figure 1), raising the possibility of HHT. Capsule endoscopy showed multiple capillary telangiectasias in the small intestine (Figure 2). Subsequently, to identify the etiology and confirm the diagnosis, she underwent genetic testing, which revealed a pathogenic c.558G>A nonsense mutation in the ENG gene [3]. Consequently, we initiated systemic anti-angiogenic therapy with thalidomide 50 mg once daily for her [4]. At the six-week follow-up, melena had virtually resolved and hemoglobin levels had increased to 102 g/L.

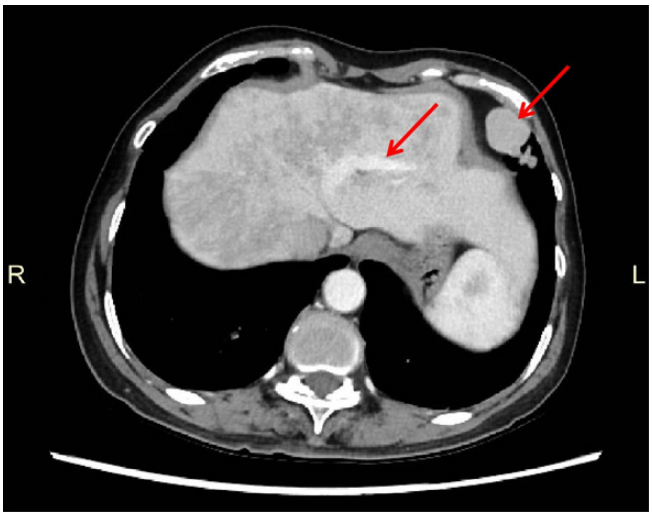

Figure 1: Contrast-enhanced Computed Tomography (CT) images: A soft tissue density nodule is noted in the left cardiophrenic angle, suggestive of an Arteriovenous Malformation (AVM). The hepatic parenchyma demonstrates heterogeneous enhancement during the arterial phase, with early opacification of the left hepatic vein. Overall, these findings are suspicious for Hereditary Hemorrhagic Telangiectasia (HHT), and further clinical evaluation is recommended to exclude this condition.

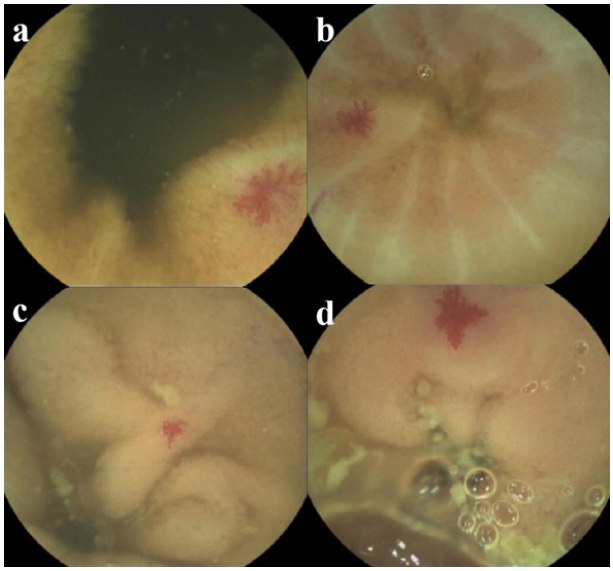

Figure 2: Capsule endoscopy findings: Multiple patchy areas of erythematous mucosa are observed in the jejunum and ileum, with prominent dilated vascular patterns on the surface.

Discussion

HHT, also known as Osler-Weber-Rendu syndrome, is an autosomal dominant vascular disorder characterized by aberrant blood vessel formation. The pathogenesis stems from loss-of-function mutations in genes involved in the transforming growth factor-beta (TGF-β) signaling pathway, predominantly ENG and ACVRL1 (ALK1) [5,6]. This genetic defect leads to the development of mucocutaneous telangiectasias and AVMs [7]. The clinical hallmarks include recurrent epistaxis, gastrointestinal bleeding, and complications from AVMs in the pulmonary, cerebral, and hepatic circulations, such as stroke, brain abscess, and high-output heart failure [5,6].

This case discusses the diagnostic challenges and management strategies of a 55-year-old female patient. The patient presented with long-term intermittent black stools and upper abdominal pain. At first, it seemed that we identified the diseases that could explain these symptoms. Because the previous gastroscopy showed duodenal ulcer and Helicobacter pylori infection. After that, we began the standard treatment for eradicating Helicobacter pylori. However, as the patient's symptoms persist, this reminds us that it is necessary to conduct further investigations beyond typical peptic ulcer diseases.

The evidence of intrahepatic arteriovenous malformations and laboratory anemia found through enhanced computed tomography has raised our suspicion of HHT [5]. Subsequent capsule endoscopy revealed multiple dilated capillaries in the small intestine, which also supported this suspicion. Ultimately, we confirmed this diagnosis through genetic testing. We found a pathogenic c.558G> A nonsense mutation in the ENG gene, and this mutation is usually associated with HHT [6].

This case demonstrates that gastrointestinal symptoms may sometimes mask more complex systemic vascular diseases, emphasizing the importance of considering HHT in the differential diagnosis of gastrointestinal bleeding. In this case, it is crucial to identify the ENG gene mutation as it establishes a direct connection with the pathophysiology of avm, providing clear evidence for potential genetic diseases [6]. Subsequently, we began to use thalidomide for systemic anti-angiogenic treatment, which led to significant clinical improvement. The reduction in black stools and the increase in hemoglobin levels during the subsequent 6-week follow-up confirmed this. This result highlights the effectiveness of targeted therapy in the treatment of HHT-related bleeding [8].

Conclusion

This case illustrates how Hereditary Hemorrhagic Telangiectasia (HHT) can masquerade as more common causes of gastrointestinal bleeding, particularly in cases where conventional treatments are ineffective. In this context, even in the absence of other characteristic clinical features, the presence of extra-intestinal arteriovenous malformations should raise clinical suspicion for HHT, and prompt consideration of a definitive diagnostic evaluation. A thorough endoscopic assessment, supplemented by genetic testing, is critical for ensuring timely diagnosis and appropriate management of this potentially treatable condition.

Acknowledgement

Not applicable.

Conflicts of Interest

The authors declare no conflicts of interest.

References

1. Govani FS, Shovlin CL (2009) Hereditary haemorrhagic telangiectasia: a clinical and scientific review. European journal of human genetics : EJHG 17(7): 860-871.
2. Samuel B Jackson, Nicholas P Villano, Jihane N Benhammou, Michael Lewis, Joseph R Pisegna, et al. (2017) Gastrointestinal Manifestations of Hereditary Hemorrhagic Telangiectasia (HHT): A Systematic Review of the Literature. Digestive diseases and sciences 62(10): 2623-2630.
3. Urszula Cymerman, Sonia Vera, Amna Karabegovic, Salma Abdalla, Michelle Letarte, et al. (2003) Characterization of 17 novel endoglin mutations associated with hereditary hemorrhagic telangiectasia. Human mutation 21(5): 482-492.
4. Kritharis A, Al Samkari H, Kuter DJ (2018) Hereditary hemorrhagic telangiectasia: diagnosis and management from the hematologist's perspective. Haematologica 103(9): 1433-1443.
5. Faughnan ME, Mager JJ, Hetts SW, Palda VA, Ratjen F, et al. (2021) Second International Guidelines for the Diagnosis and Management of Hereditary Hemorrhagic Telangiectasia. Annals of internal medicine 174(7): 1035-1036.
6. Bayrak Toydemir P, Mao R, Lewin S, McDonald J (2004) Hereditary hemorrhagic telangiectasia: an overview of diagnosis and management in the molecular era for clinicians. Genetics in medicine : official journal of the American College of Medical Genetics 6(4): 175-191.
7. Shovlin C L (2010) Hereditary haemorrhagic telangiectasia: pathophysiology, diagnosis and treatment. Blood reviews 24(6): 203-219.
8. Al Samkari H (2022) Systemic Antiangiogenic Therapies for Bleeding in Hereditary Hemorrhagic Telangiectasia: A Practical, Evidence-Based Guide for Clinicians. Seminars in thrombosis and hemostasis 48(5): 514-528.