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IMAGE | LIVER

Hereditary Hemorrhagic Telangiectasia With Liver Vascular Malformation Presenting With High-Output Heart Failure

Gurshawn Singh, MD[1], Talal Adhami, MD[2], and Naim Alkhouri, MD[2]

[1]*Department of Internal Medicine, Cleveland Clinic, Cleveland, OH*
[2]*Department of Gastroenterology and Hepatology, Cleveland Clinic, Cleveland, OH*

Case Report

Hereditary hemorrhagic telangiectasia (HHT) is an autosomal dominant genetic disease manifested by formation of telangiectasias and visceral vascular malformations of organ systems, including the skin, lungs, gastrointestinal tract, brain, and liver. Hepatic involvement may lead to portal hypertension, high-output cardiac failure, and biliary strictures.[1] Diagnosis requires patients to have 3 of 4 clinical characteristics: epistaxis, telangiectasia on the skin, visceral lesions, and a first-degree relative with HHT.[2] Management involves controlling the source of bleeding by packing the nasopharynx, endoscopic therapies, or pulmonary or hepatic embolization.[3]

A 48-year-old female with HHT diagnosed at age 21 years presented with worsening shortness of breath and lower extremity edema over a 6-month period. She had heart failure with high cardiac output of 8.2 L/min. Prior chest and brain CT showed no AV malformations and she had no previous history of upper or lower gastrointestinal bleeds. Abdominal computed tomography showed markedly dilated portal vein, hepatic vein, and inferior vena cava with heterogeneous enlarged liver and innumerable hyperdense nodules (Figure 1). Vascular ultrasound of the liver showed high velocity and low resistive indices of the hepatic arteries with multiple hepatic intraparenchymal shunts. The patient developed oropharyngeal bleeding that rapidly progressed, leading to aspiration of blood, cardiac arrest, and death.

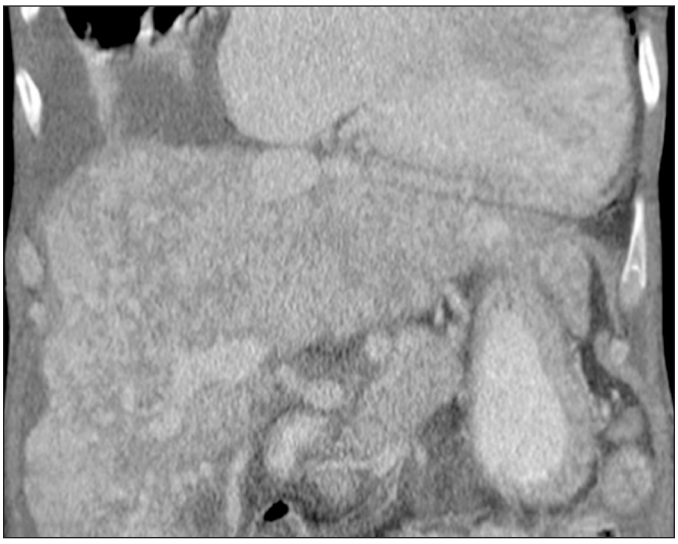

Figure 1. Abdominal CT showing markedly dilated portal vein, hepatic vein, and inferior vena cava with heterogeneous enlarged liver and innumerable hyperdense nodules.

Liver angioectasias in HHT can cause increased preload leading to high-output heart failure. Recognition of imaging findings is important for determining diagnosis and severity of disease in patients with suspected HHT. Per AASLD guidelines, hepatic artery embolization should be avoided, and the only definitive treatment in symptomatic patients with HHT is liver transplantation.[4]

Disclosures

Author contributions: G. Singh drafted the manuscript. T. Adhami edited the manuscript. N. Alkhouri drafted and edited the manuscript, and is the article guarantor.

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The patient is now deceased, so the authors made every effort to contact her next of kin for informed consent, but were unsuccessful. However, the authors feel that the patient information is sufficiently anonymous and that the patient would not object to this publication.

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Correspondence: Naim Alkhouri, Department of Gastroenterology and Hepatology, Digestive Disease Institute, Cleveland Clinic, 9500 Euclid Avenue, Cleveland, OH 44195 (alkhoun@ccf.org).

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