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Gastrointestinal

# Congestive cirrhosis in Osler-Weber-Rendu syndrome: A rare case report

*Patrícia Leitão PhD*[a,*], *André Carvalho MD*[a], *Conceição Guerra MD*[a], *José Gonçalves MD*[a], *Isabel Ramos PhD*[a,b]

[a] *Radiology Department, Centro Hospitalar São João, Porto, Portugal*
[b] *Centro Hospitalar São João, Alameda Prof. Hernâni Monteiro, Porto, 4200-319, Portugal*

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ABSTRACT

Hereditary hemorrhagic telangiectasia or Osler-Weber-Rendu syndrome is a rare autosomal dominant vascular disorder characterized by epistaxis, mucocutaneous telangiectasias, and arteriovenous malformations affecting various organs and systems. The liver is a commonly involved organ (74% of patients with hereditary hemorrhagic telangiectasia), although symptomatic liver disease is quite infrequent. In symptomatic cases, clinical manifestations relate most commonly to the predominant type of vascular shunting present (arteriovenous, arterioportal, or portovenous). Clinically, liver disease can manifest as a high-output cardiac failure, portal hypertension, or biliary disease. Imaging plays an important role in diagnosis, characterization, and follow-up of liver involvement, with ultrasound, computed tomography, magnetic resonance imaging, and angiography being useful in this context. We present a case of congestive cirrhosis with florid liver findings in a patient with Osler-Weber-Rendu syndrome. Imaging findings that clinched the diagnosis are reviewed. A brief literature review is also provided.



## Introduction

Osler-Weber-Rendu syndrome is a rare hereditary disorder with varied manifestations. The pathophysiology of this syndrome is related to weakness of the vessel walls with resulting dilation of the vascular lumen and development of vascular shunts [1,2]. The most common clinical presentation is recurrent epistaxis and telangiectasias of the face, the lips, the limbs, and the trunk. Gastrointestinal and central nervous system hemorrhage and liver arteriovenous malformations are also frequently encountered.

Ultrasound is a useful tool in the detection of liver vascular shunts, whereas computed tomography (CT) and angiography can help in the characterization of these lesions and its associated complications.

Arteriovenous shunts are the most common subtype of liver shunts (ie, from the hepatic artery to the hepatic veins) and are associated with high-output cardiac failure secondary to an excessive return of blood to the heart [3].

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* *Corresponding author.*
E-mail address: patleitao20@gmail.com (P. Leitão).

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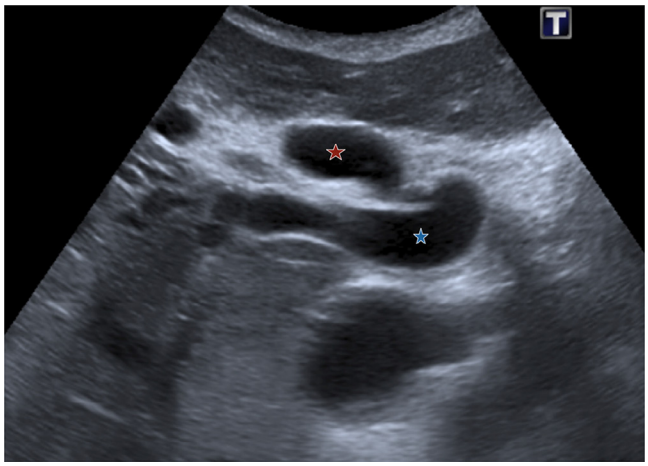

**Fig. 1 – Abdominal ultrasound image shows a very prominent common hepatic artery (red star) with a diameter at the extrahepatic level similar to that of the portal vein (blue star).**

## Clinical case

A 62-year-old woman with a known Osler-Weber-Rendu syndrome presented to the emergency department complaining of dyspnea, fatigue, increased abdominal girth, and peripheral edema. Physical examination revealed tachypnea, cold extremities, and bibasilar crackles on pulmonary auscultation. Signs and symptoms of a decompensated congestive heart failure prompted further evaluation with electrocardiogram and transthoracic echocardiogram. Right upper quadrant tenderness was also present at physical examination. An abdominal ultrasound was requested to evaluate for ascites or a possible acute hepatobiliary disease.

Cardiac tests showed signs consistent with severe right heart failure; abdominal ultrasound revealed moderate volume ascites and signs suggestive of cirrhosis, with liver enlargement, irregular contour, left lobe hypertrophy, and a diffusely heterogeneous ecotexture. The most relevant ultrasound findings were, however, a prominent common hepatic artery with a diameter at the extrahepatic level similar to that of the portal vein (Fig. 1); the presence of several tubular structures paralleling the portal vein at the hepatic hilum, representing dilated arterial branches; and markedly dilated and tortuous intrahepatic vessels suggestive of arterioportal shunts (Fig. 2).

A multiphasic contrast-enhanced CT scan of the abdomen was performed for further evaluation, revealing a dysmorphic liver, with mottled enhancement in the arterial and early portal venous phase (Fig. 3A and B). On delayed phase imaging, enhancement of the liver became more uniform, as seen in Figure 3C. The findings described on ultrasound were confirmed by noting marked tortuosity and an increased caliber of the common hepatic artery, multiple tortuous intrahepatic collateral circulation, and arteriovenous shunts (Fig. 3D). Hepatic vein engorgement, cardiomegaly with right-side predominance, ascites, and splenomegaly were also present. These findings were suggestive of liver involvement in the Osler-Weber-Rendu syndrome.

## Discussion

Hereditary hemorrhagic telangiectasia (HHT) or Osler-Weber-Rendu syndrome is a rare, autosomal dominant genetic disorder characterized by the development of angiodysplasias, as well as arteriovenous aneurysms and vascular shunts. HHT can affect multiple organic systems, with the most important being the lungs, the liver, and the brain. The diagnosis of HHT lies largely on a clinical basis, with epistaxis, telangiectasias, visceral lesions, and a positive family history being the 4 main clinical diagnostic criteria. For a definite diagnosis, at least 3 of the aforementioned criteria must be present, according to the Curacao criteria [4].

Although liver involvement is commonly present, symptomatic associated complications are rare. Because the liver has a dual blood supply and a dual venous drainage system, different types of vascular shunts can develop [5]. Clinical manifestations of liver involvement in HHT depend mostly on the predominant type and size of vascular shunt, and have been categorized into 3 distinct clinical patterns: (1) high-output cardiac failure, (2) portal hypertension, and (3) biliary disease. Cases of high-output cardiac failure are usually associated with arteriosystemic shunts (shunts between the common hepatic

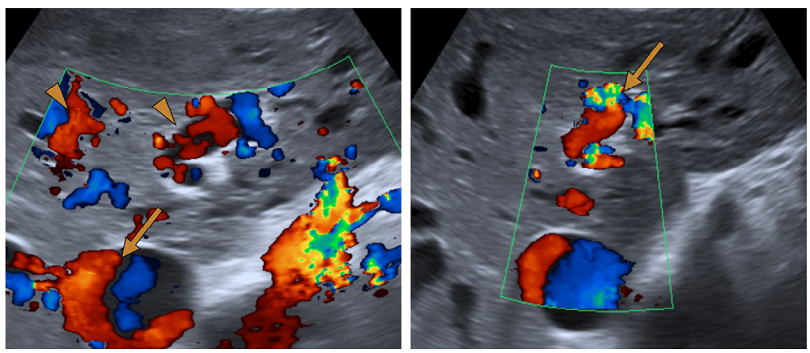

**Fig. 2 – Ultrasound images with color Doppler depicts several tubular structures paralleling the intrahepatic portal branches representing dilated arterial branches (arrowheads) as well as markedly dilated and tortuous intrahepatic vessels suggestive of arterioportal shunts (arrows).**