

## **Case 10068**

### **Liver involvement in Hereditary Haemorrhagic Telangiectasia (HHT; Osler-Weber-Rendu syndrome)**

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**Section:** Abdominal Imaging

**Published:** 2012, Jul. 23

**Patient:** 42 year(s), female

## **Authors' Institution**

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## **Clinical History**

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42-year-old black woman with abnormal liver function and an ultrasound examination done outside our institution suggesting Caroli's disease due to dilated biliary tree and poorly defined nodular changes. She was also being followed in an ENT clinic for recurrent sporadic epistaxis.

## **Imaging Findings**

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CholangioMR was done because of suspicion of Caroli's disease, but failed to demonstrate evidence for this disease. The liver's parenchyma was diffusely heterogeneous and the celiac trunk and hepatic artery were dilated.

Doppler ultrasound demonstrated dilated celiac trunk and hepatic artery with a high peak velocity

and low resistance pattern. The liver's parenchyma was also notably heterogeneous with exuberance of its arterial branches.

CT showed hepatomegaly with dilated and tortuous celiac trunk, extra-parenchymal hepatic artery and its intra-parenchymal branches, dilated IVC and enhancement of the hepatic veins in the early arterial phase. Multiple intrahepatic hyperenhancing nodular changes are seen, ranging from a few millimetres in size to several centimetres, some suggestive of FNH.

## Discussion

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Hereditary Haemorrhagic Telangiectasia (HHT; Osler-Weber-Rendu syndrome) is an uncommon (1-2:10, 000) autosomal dominant disease characterised by widespread mucocutaneous and visceral angiodysplastic lesions. Liver involvement affects 8-78% of cases, but only 8% of these are symptomatic.<sup>(1-6)</sup>

Intrahepatic shunts can occur from the hepatic artery to the hepatic vein (arteriosystemic), the hepatic artery to the portal vein (arteriportal) or the portal vein to the hepatic vein (portosystemic). Arteriportal shunts are the most common and typically sub-capsular or peripheral. Arteriosystemic shunts are rare and often associated with arteriportal shunts. Both result in increased hepatic artery blood flow and dilation. Portosystemic shunts are rare diverting the portal flow to the hepatic veins thus rendering the liver susceptible to infarction.<sup>(3-5)</sup>

Three clinical sub-types occur<sup>(1)</sup>. The predominance of shunts to the hepatic veins eventually results in high-output heart failure, while shunts to the portal vein result in portal hypertension with splenomegaly and gastroesophageal varices, regenerative changes and fibrosis. Biliary ischaemia due to arteriovenous shunts causes biliary strictures, dilation, obstruction or sepsis.

Extra-hepatic shunts, hepatic portosystemic encephalopathy, mesenteric angina, cirrhosis and liver failure may also occur.<sup>(2, 3, 5, 6)</sup>

Usually widespread telangiectasias suggest the diagnosis and imaging is intended to stage the disease. In our patient, the liver involvement was the basis of the diagnosis and only then were the epistaxis and subtle skin telangiectasias (in dark skin) interpreted in light of HHT.

Colour Doppler ultrasound evaluates arterial, venous and portal flows showing intraparenchymal shunts and vascular malformations.<sup>(3, 7)</sup> A dilated common hepatic artery (>7mm) and intrahepatic hypervascularisation are highly sensitive and specific diagnostic signs, showing a high velocity and low resistance flow.<sup>(4, 6)</sup> Arterialised portal or hepatic veins can sometimes be shown.<sup>(4, 6, 8)</sup>

Multi-phase CECT studies are specially useful, showing dilated and tortuous vessels, early filling of the portal and hepatic veins and hyperenhancing parenchymal nodules which become isoenhancing in the portal phase. These nodules are vascular lesions (telangiectasias and confluent vascular masses).<sup>(2-4)</sup>

MRI and MRA demonstrate hepatic vascular malformations and the extent of parenchymal fibrosis and cirrhosis (T2-weighted sequences), fatty infiltration (chemical shift sequences) and telangiectasias (dynamic GE T1-weighted sequences).<sup>(7)</sup>

Angiography is now limited to pre-transplant work-up and hemodynamic measurements.<sup>(3)</sup>

Biopsy procedures are discouraged because of the bleeding risk.<sup>(3, 5)</sup>

The prognosis is usually good with medical treatment.<sup>(2)</sup>

Hepatic artery embolisation or ligation may be useful but are associated with hepatobiliary necrosis.<sup>(1, 3, 5)</sup> Liver transplant is the only definitive curative option but excessive intra-operative bleeding and recurrence have been reported.<sup>(1, 5, 6)</sup>

## Final Diagnosis

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Hereditary Haemorrhagic Telangiectasia (Osler-Weber-Rendu Syndrome)

## Differential Diagnosis List

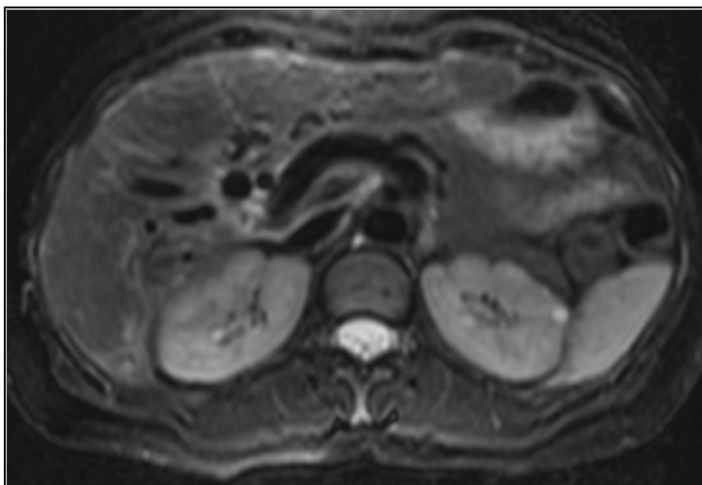
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Hepatic intraparenchymal perfusion changes, Isolated arteriosystemic shunts, CHC, Cirrhosis, Isolated vascular malformations

## Figures

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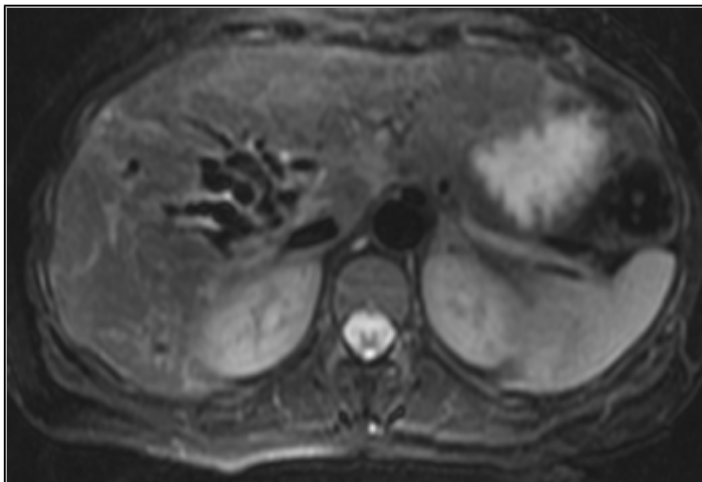
**Figure 1 CholangioMR**



CholangioMR done for the suspicion of Carolli's disease showed dilated celiac trunk and hepatic artery

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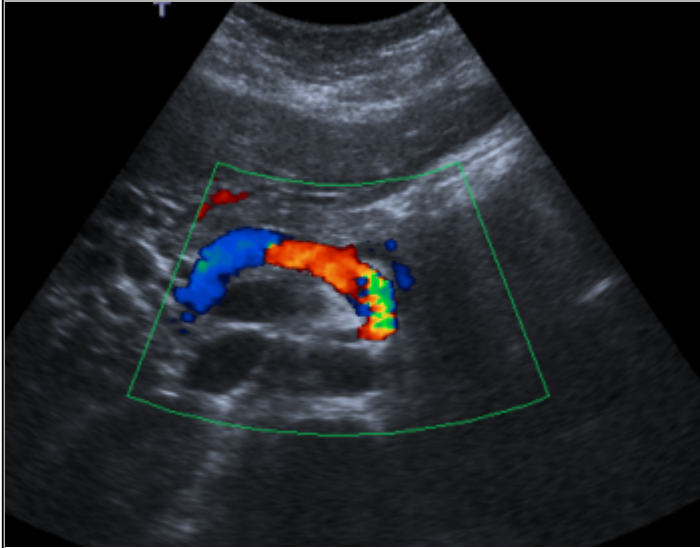
Area of Interest: Biliary Tract / Gallbladder; Liver;  
Imaging Technique: MR;  
Procedure: Diagnostic procedure;  
Special Focus: Arteriovenous malformations; Congenital;



CholangioMR done for the suspicion of Carolli's disease showed dilated celiac trunk and hepatic artery

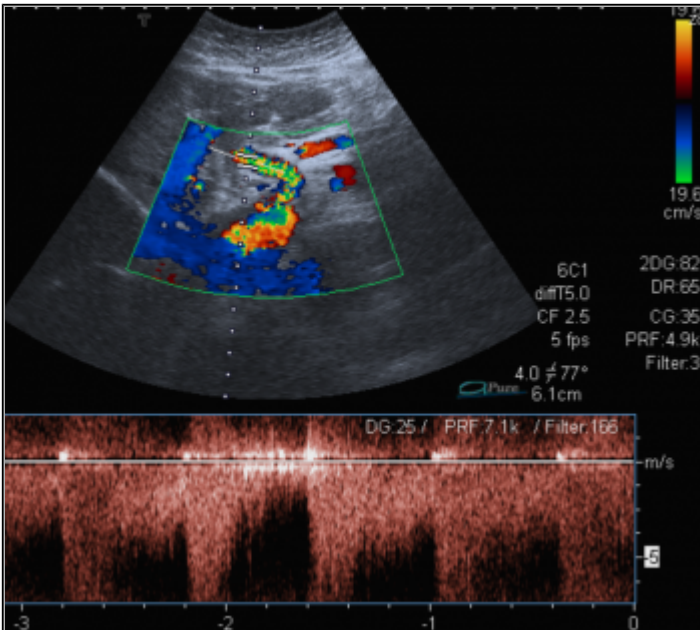
Area of Interest: Liver;  
Imaging Technique: MR;  
Procedure: Diagnostic procedure;  
Special Focus: Arteriovenous malformations; Congenital;

**Figure 2 Doppler US 1**



Marked dilation of the celiac trunk

Area of Interest: Abdomen; Interventional vascular; Liver;  
Imaging Technique: Ultrasound-Colour Doppler; Ultrasound-Spectral Doppler;  
Procedure: Diagnostic procedure;  
Special Focus: Genetic defects; Trauma;

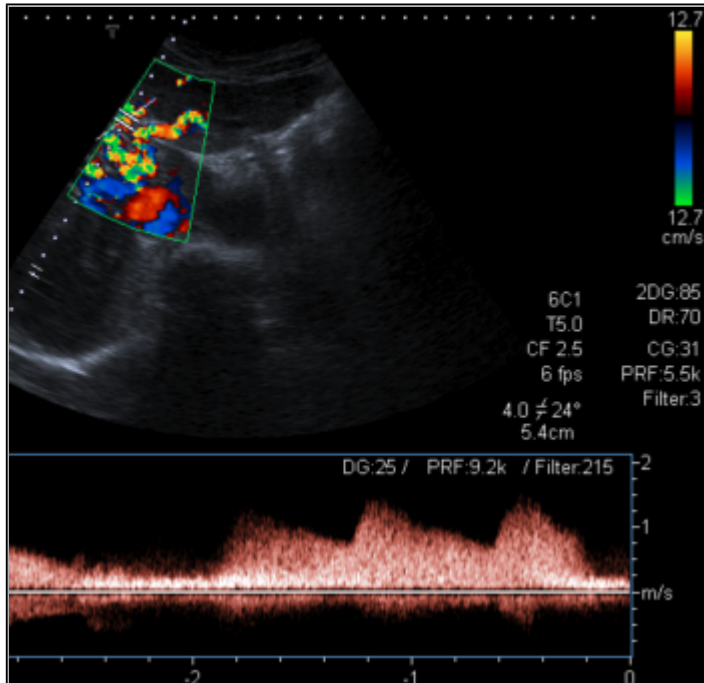


Hyperdynamic hepatic artery with peak-systolic flows higher than 5m/s and dilation of the celiac trunk and hepatic artery

Area of Interest: Liver;

Imaging Technique: Ultrasound-Colour Doppler;  
Procedure: Diagnostic procedure;  
Special Focus: Congenital;

**Figure 3 Doppler ultrasound 3**

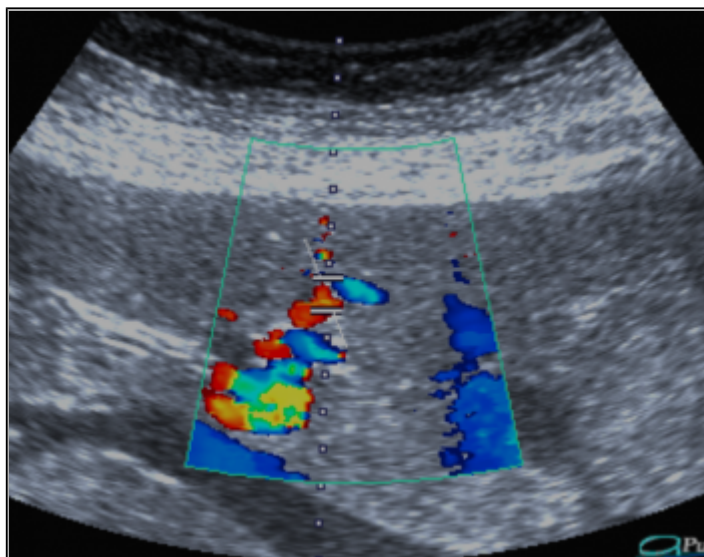


Exuberant high-flow intra-hepatic arteries

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Area of Interest: Liver;  
Imaging Technique: Ultrasound-Colour Doppler;  
Procedure: Diagnostic procedure;  
Special Focus: Congenital;

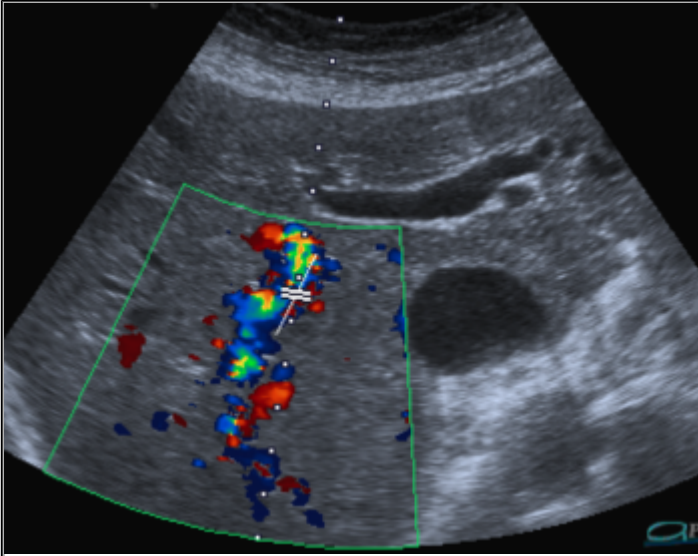
**Figure 4 Doppler ultrasound 4**



Arteriovenous shunts

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Area of Interest: Liver; Vascular;  
Imaging Technique: Ultrasound-Colour Doppler;  
Procedure: Diagnostic procedure;  
Special Focus: Congenital;

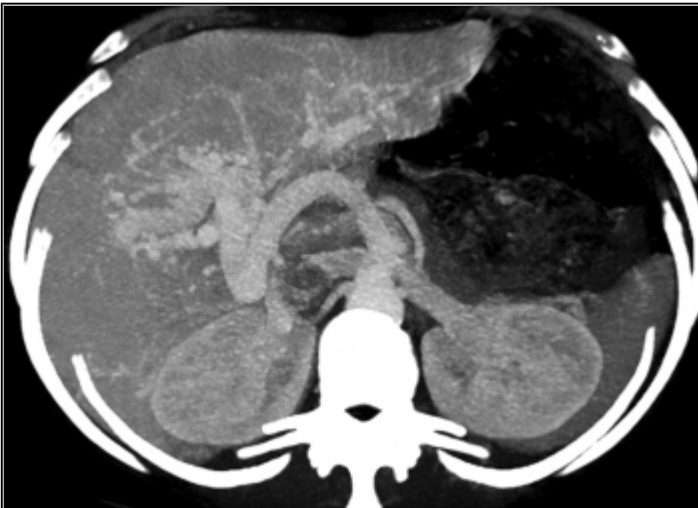


Arteriovenous shunts

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Area of Interest: Liver;  
Imaging Technique: Ultrasound-Colour Doppler;  
Procedure: Diagnostic procedure;  
Special Focus: Congenital;

**Figure 5 CT 3D reconstruction**



3D MIP Showing exuberant vascularisation of the liver with a dilated and turtuous hepatic artery

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Area of Interest: Liver;  
Imaging Technique: CT-Angiography;  
Procedure: Computer Applications-3D;  
Special Focus: Arteriovenous malformations; Congenital;

**Figure 6 CT**



Arterial phase CT shows a heterogenous liver with multiple hyperenhancing liver nodules, posing a differential diagnosis with HCC in cirrhosis

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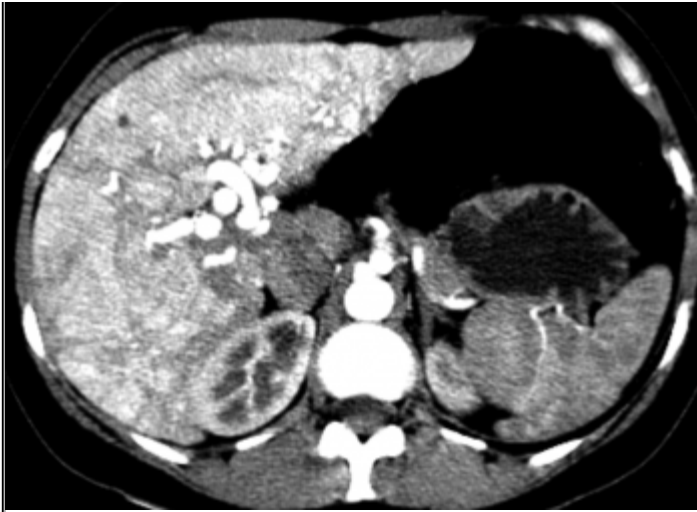
Area of Interest: Liver;  
Imaging Technique: CT-Angiography;  
Procedure: Diagnostic procedure;  
Special Focus: Arteriovenous malformations; Congenital;



Arterial phase CT shows a heterogenous liver with multiple hyperenhancing liver nodules, posing a differential diagnosis with multifocal HCC in cirrhosis

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Area of Interest: Liver;  
Imaging Technique: CT-Angiography;  
Procedure: Diagnostic procedure;  
Special Focus: Arteriovenous malformations; Congenital;



Arterial phase CT shows a heterogenous liver with multiple hyperenhancing liver nodules, posing a differential diagnosis with HCC in cirrhosis

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Area of Interest: Liver;  
Imaging Technique: CT-Angiography;  
Procedure: Diagnostic procedure;  
Special Focus: Arteriovenous malformations; Congenital;

**Figure 7 CT hepatic veins**



Early enhancement of the hepatic veins, before portal vein opacification is due to arterio-systemic shunts.

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Area of Interest: Liver;  
Imaging Technique: CT-Angiography;  
Procedure: Diagnostic procedure;  
Special Focus: Arteriovenous malformations; Congenital;

**Telangiectasia, Hereditary Hemorrhagic** [C14.907.823.780]

An autosomal dominant vascular anomaly characterized by the presence of multiple small telangiectases of the skin, mucous membranes, gastrointestinal tract, and other organs, associated with recurrent episodes of bleeding from affected sites and gross or occult melena. (Dorland, 27th ed)

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**Citation**

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URL: <http://www.eurorad.org/case.php?id=10068>