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Rendu-Osler-Weber Disease: Typical Hepatic Findings

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1. Abstract

Hereditary Hemorrhagic Telangiectasia (HHT) or Rendu–Osler–Weber disease is a rare autosomal dominant multisystem disease characterized by angiodysplastic lesions involving the skin, lungs, gastrointestinal tract, brain and liver.

Liver involvement consists of vascular, parenchymal, and biliary lesions characterized by the presence of intrahepatic shunts, disseminated intraparenchymal telangiectases and other vascular lesions.

Multiphasic CT and MRI play a crucial role in the assessment of liver involvement in the context of Rendu-Osler disease. We present the case of a 34-old-male patient with a typical hepatic findings of Rendu-Osler-Weber disease.

2. Introduction

Hereditary Hemorrhagic Telangiectasia (HHT) or Rendu–Osler–Weber disease is a rare autosomal dominant disease characterized by angiodysplastic lesions [1]. It's a multisystem disease involving the skin, lungs, gastrointestinal tract, brain and liver.

Hepatic involvement has been considered uncommon. The presence of intrahepatic shunts, disseminated intraparenchymal telangiectases and other vascular lesions are the typical findings of hepatic involvement [2]. CT and MRI allow a detailed study of hepatic lesions, such as, telangiectasies, arteriovenous malformations, perfusion disorders, biliary lesions.

3. Case Report

A 34-year-old male patient with a history of recurrent epistaxis and a family history of epistaxis, presented for right hypochondria pain with anemic syndrome. The physical examination found an apyretic patient patient with tachycardia (HR: 100B/min). There was cutaneous-mucous pallor, telangiectasia of the tongue and digital hippocratism.

Laboratory datas found iron deficiency anemia with hemoglobin: 8 g/dl, a low mean corpuscular volume: 62fl and mean corpuscular hemoglobin concentration,: 28 pg, ferritin: 8 µg/dL

The CT hepatic angiography showed a heterogeneously enhanced hepatic parenchyma after contrast injection, realizing a mosaic appearance to the liver, with early opacification of the hepatic veins in favor of early venous return (Figure 1, a, b). There was a round lesion in segment VII of the liver, measuring 15mm, not enhanced, centered by an arterial branch in favour of a thrombosis aneurysm (Figure 2 c, d), with multiple intrahepatic shunts: arterioportal shunts, arterio-venous shunts and porto-venous shunts (Figure 3 e, f). Liver MRI showed a typical manifestation of Rendu Osler's disease. There was liver telangiectasia, regenerative nodules (Figure 4, g, h) and a thrombosis arterial aneurysm at segment VII. It was associated with intra hepatic ducts dilatation related to ischemic cholangitis, secondary to shunts with hypoperfusion of the bill duct wall (Figure 5, i, j).

The diagnosis of Rendu-Osler-Weber's disease was based on: Spontaneous and repeated epistaxis, family history of epistataxis, lingual telangiectasia, and hepatic arteriovenous malformations.

The patient received iron supplementation. Due to the persistence of the anemic syndrome, bi-monthly treatment with Bevacizumab was introduced for two months. The evolution was marked by clinical and biological improvement with normalization of hemoglobin rate.

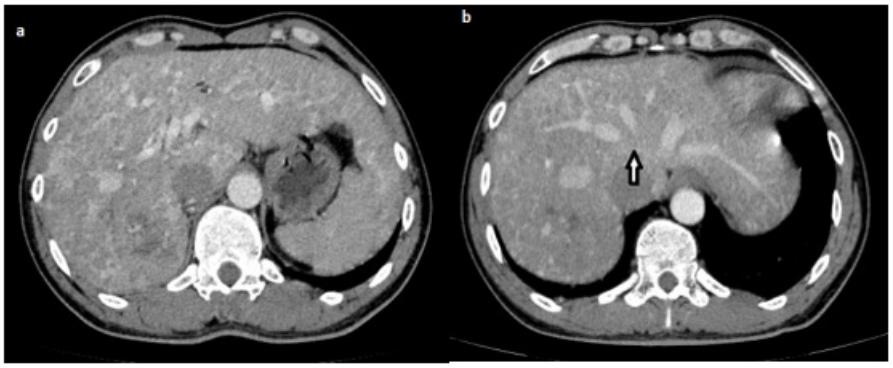

Figure 1: Hepatic CT angiography at the arterial phase showing a heterogeneously enhanced hepatic parenchyma realizing a mosaic appearance to the liver (a), with early opacification of the hepatic veins (b,arrow).

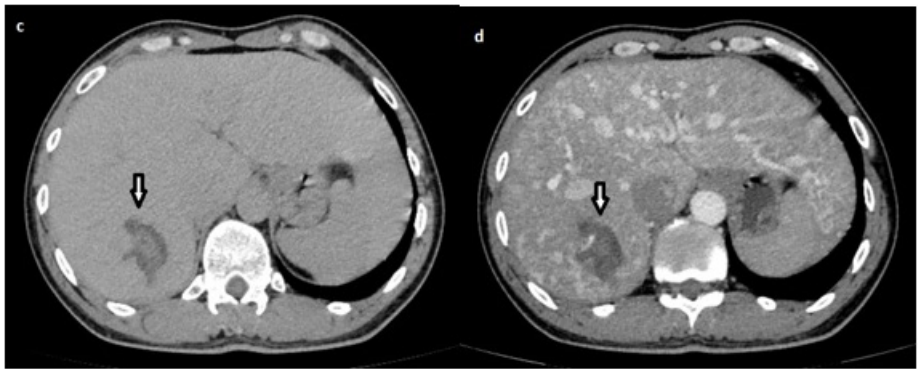

Figure 2: Hepatic CT scans before contrast enhacement (c) and after contrast enhancement at the arterial phase (d) showing a round lesion in segment VII not enhanced, centered by an arterial branch in favour of a thrombosis aneurysm (arrow).

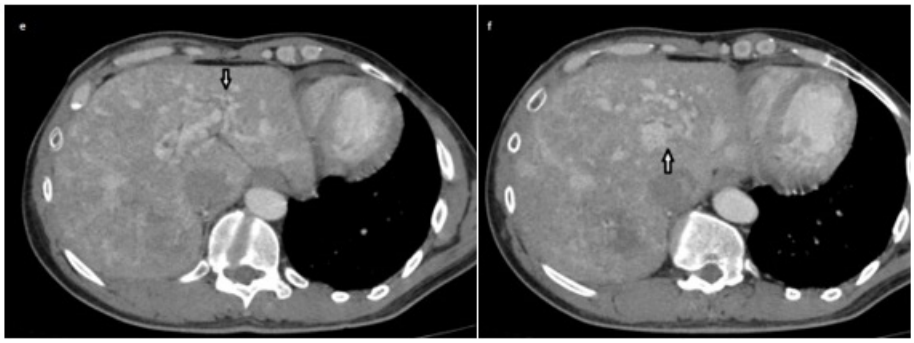

Figure 3: Hepatic CT angiography at the arterial phase showing multiple intrahepatic shunts: arterio-venous shunts (hepatic artery/ hepatic vein) (e, arrow), arterioportal shunts (hepatic artery/portal vein), and porto-venous shunts (portal vein/hepatic vein) (f, arrow).

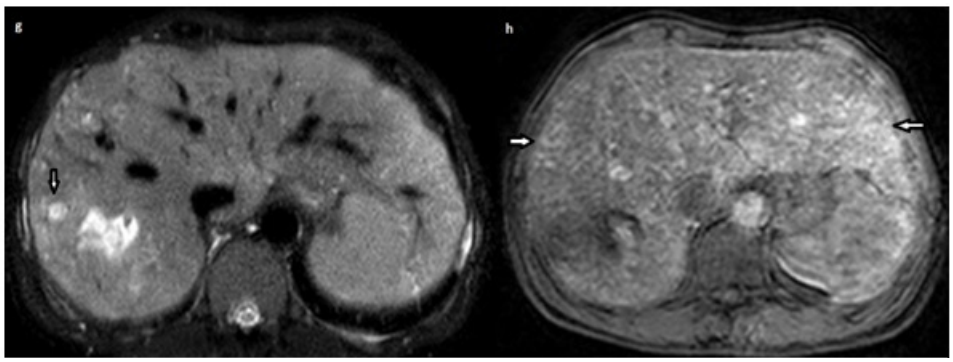

Figure 4: Hepatic MRI on T2 weighted sequence (g) and T1 weighted sequence after contrast injection at the arterial phase (h) showing regeneration nodules (g, arrow) with telangiectasia (h, arrow).

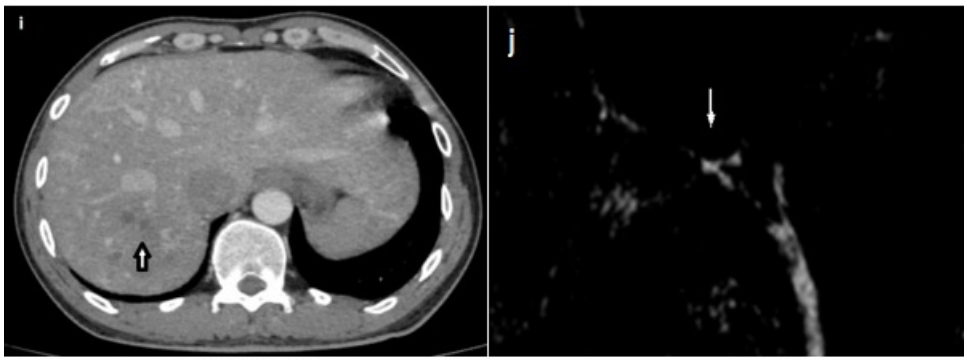

Figure 5: Hepatic CT angiography at the arterial phase (i) and cholangiopancreatography MRI (j) showing segmental dilatation of the intra hepatic bile ducts related to ischemic cholangitis (arrow)

4. Discussion

Hereditary Hemorrhagic Telangiectasia (HHT) also known as Rendu–Osler–Weber disease is an autosomal dominant vascular disease characterized by angiodysplastic lesions including telangiectases and arteriovenous malformations that involve many organs [1]. This disease occurs with an estimated frequency of 10 to 20 per 100,000 individuals [2].

Most common clinical manifestations include skin and mucosal telangiectasias and epistaxis, besides gastrointestinal, pulmonary and intracerebral bleeding. The onset of symptoms is around 10 years of age and with increasingly severe episodes. HHT is diagnosed on the basis of the presence of three among four Curacao criteria, as follows: skin and mucosal telangiectasias; recurrent spontaneous epistaxis; visceral arteriovenous-malformations and positive family history. HHT can be confirmed through identification of the involved mutations with molecular biology techniques (mutation of the endoglin gene, mutation of the activin gene) [3].

The organs most frequently involved are the skin, lungs, gastrointestinal tract, and brain; hepatic involvement, almost always defined by clinical criteria, has been considered uncommon, and its prevalence has ranged from 8% to 31% and consists of vascular, parenchymal, and biliary lesions [1]. The presence of intrahepatic shunts, disseminated intraparenchymal telangiectases and other vascular lesions are the typical findings of hepatic involvement. Patients with hepatic involvement can be asymptomatic, but congestive heart failure, portal hypertension, portosystemic encephalopathy, cholangitis, and atypical cirrhosis have been reported in the literature, so a correct diagnosis is important for the follow-up of symptomatic and asymptomatic patients [2].

Diagnostic imaging has a fundamental role in the identification of hepatic vascular alterations. Ultrasonography in association with color Doppler is the usual method for screening patients with Rendu-Osler disease and suspected liver involvement. It's able to demonstrate intraparenchymal shunts and other vascular malformations, and it offers qualitative and quantitative analyses of the arterial, venous, and portal flows. Helical CT, in particular the multidetector row hCT scanner allows a complete multiphasic study of the hepatic vascular system. MRI performed with dynamic and angiographic sequences (angio-MRI) provides similar information to that obtained with CT in the study of liver disorders [2].

Diffuse Hepatic Vascular Malformations are characteristic of HHT and range from tiny telangiectases to transient perfusion abnormalities and large confluent vascular masses. Liver telangiectasies are early manifestations of hepatic involvement in HHT and may progress to form more complex vascular malformations and shunts, with up to 21% of patients demonstrating an increase in size and complexity of hepatic VMs after long-term follow-up [2, 4]. Focal or diffuse telangiectases are the most frequently observed hepatic lesion. They appear like hypervascular rounded masses, measuring a few millimeters in size, but may reach 9 mm in diameter, with usually an asterisk shape. Coronal MIP images are useful in appreciating telangiectases, particularly when they are in close proximity to the large vertically oriented vessels, making them difficult to appreciate on transverse images [2, 4].

Large Confluent Vascular Masses defined as "large areas of multiple telangiectases that coalesce or large shunts that are directly visible." Any enhancing lesion with a diameter more than 10 mm is called a large confluent vascular mass. These larger vascular pools usually demonstrate early enhancement that is seen during both arterial phases, with enhancement persisting in the hepatic phase [2, 4].

Hepatic Shunts: Three pattern of intrahepatic shunts between the major vessels of the liver are possible: arteriosystemic (hepatic artery to hepatic vein), arterioportal (hepatic artery to portal vein), and portosystemic venous (portal vein to hepatic or systemic veins) [2, 4].

- **Arterio-Systemic Shunt:** They are rare and usually associated with hepatic neoplasms. They consist of abnormal connections between hepatic arteries and hepatic veins. CT findings consist in the early enhancement of one or more hepatic veins during the early arterial phase, and are usually seen with coexistent telangiectases or large vascular masses. The early filled hepatic vein involved by the shunt may be enlarged. Direct arteriovenous fistulas can also be seen in isolation or in association with focal hepatic vascular masses. A high output cardiac failure secondary to intrahepatic

shunt left-to-right due to arteriosystemic venous shunts is a possible serious complication. The CT shows an early visualization of hepatic veins and the inferior vena cava. These findings are usually associated with other signs of passive hepatic congestion such as hepatomegaly with heterogeneous enhancement during the arterial phases [2, 4].

- **Arterioportal Shunts:** Consist of abnormal anastomoses between the hepatic arteries and the portal veins. The multiphasic CT features of arterioportal shunting are indirect and include the early and prolonged enhancements of the portal vessels during the early arterial phase. These types of shunts are typically subcapsular or peripheral and may be associated with areas of perfusion disorder which are sometimes the only manifestation of the intraparenchymal arterioportal shunts [2, 4].
- **Porto-Systemic Shunts (portal vein to hepatic vein):** shunts are rarely seen in HHT. These shunts are seen in the hepatic phase, with a dilated portal vein branch (during the portal venous phase) communicating with the large hepatic vein, usually through a focal hepatic mass. Although vascular shunting is a prominent feature of HHT, other pathologic conditions such as neoplasms and traumatic or iatrogenic intrahepatic arteriovenous fistulas can cause intrahepatic vascular shunts [4].

Hepatic Perfusion Disorders: In contrast to the cirrhosis, the perfusion abnormalities in HHT are frequently more diffuse and inhomogeneous or ill-defined which can be related to the more widespread underlying vascular lesions. Hepatic perfusion abnormalities are identified as an inhomogeneous attenuating pattern within the liver parenchyma. They are better seen during the early arterial and late arterial phases, almost always disappearing in the hepatic phase as the hepatic parenchyma becomes homogeneous [2, 4].

Focal perfusion defects similar to transient attenuation defects of hepatic attenuation ("THAD" lesions) can be seen alone with a peripheral location, triangular shape and straight margins. Such defects may be an indirect sign of other abnormalities such as arterioportal shunt, tiny telangiectasia or serous venous drainage [4].

Other lesions: HHT patients rarely develop cirrhosis secondary to extensive necrotizing cholangitis. In the magnetic resonance cholangiopancreatography sequences, ischemic cholangitis was defined as irregular biliary ducts with narrowing and upstream dilatation in the peripheral intrahepatic-biliary tracts, with a diffuse or segmental distribution or a "pruned tree" appearance. The obstruction and dilatation of the bile ducts due to compression of enlarged vascular structures have also been studied [5].

5. Conclusion

Rendu–Osler–Weber disease is a rare multisystem disease characterized by angiodysplastic lesions involving the skin, lungs, gastrointestinal tract, brain and liver. Liver involvement consists of vascular, parenchymal, and biliary lesions. Imaging plays an important role in the assessment of liver involvement, which is usually very suggestive of the disease.

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