

# HEREDITARY HEMORRHAGIC TELANGIECTASIA: A RARE CAUSE OF HEPATIC ENCEPHALOPATHY DUE TO PORTOSYSTEMIC SHUNT

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## ABSTRACT

Hepatic encephalopathy is a common complication of end-stage liver disease. It happens in the presence of significant hepatic dysfunction and the diversion of the portal blood to the systemic circulation (porto-systemic shunts). Hereditary Hemorrhagic Telangiectasia (HHT) or Osler-Weber-Rendu disease, can also involve the liver along with the skin, mucous membranes, lung, brain, and gastrointestinal tract. The prevalence of hepatic involvement in HHT ranges between 41% and 78%. Although

most patients with HHT are asymptomatic, some may have symptoms due to high-output heart failure, portal hypertension and biliary disease. Less commonly, patients may also develop porto-systemic encephalopathy. In this case report, we present a woman aged 63 years, who was diagnosed as having hepatic encephalopathy due to a portosystemic shunt.

**Keywords:** Portosystemic shunt, hepatic encephalopathy, hereditary hemorrhagic telangiectasia *Nobel Med 2015; 11(3): 74-76*

## HEREDİTER HEMORAJİK TELENJEKTAZİ: PORTOSİSTEMİK ŞANTA BAĞLI NADİR BİR HEPATİK ENSEFALOPATİ NEDENİ

### ÖZET

Hepatik ensefalopati son dönem karaciğer hastalığının sık bir komplikasyonudur ve karaciğer disfonksiyonu ve/veya portal kanın sistemik dolaşıma portosistemik şantlar ile karışımı sonucu gelişmektedir. Hereditör hemorajik telenjektazi (HHT) veya Osler-Weber-Rendu olarak da bilinen klinik tabloda deri, mukozal membranlar, akciğer, beyin ve gastrointestinal sistem ile birlikte karaciğer tutulumu da görülebilmektedir.

Karaciğer tutulum oranı %41-78 arasında değişmektedir ve hastalarının çoğu asemptomatik olmasına rağmen bu hastalarda portal hipertansiyon, biliyer hastalıklar ve yüksek debili kalp yetersizliği de gelişebilmektedir. Bu hastalarda nadiren portosistemik şantlara bağlı ensefalopati gelişebilmektedir. Bu olgu sunumunda HHT tanısı ile takipli ve portosistemik şanta bağlı hepatik ensefalopati gelişmiş 63 yaşında bir kadın hastayı paylaşmaktayız.

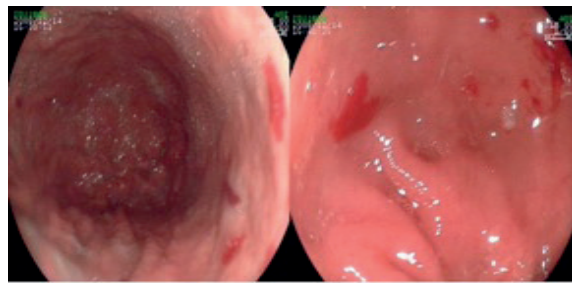
**Anahtar kelimeler:** Portosistemik şant, hepatik ensefalopati, Hereditör hemorajik telenjektazi *Nobel Med 2015; 11(3): 74-76*

## INTRODUCTION

Hepatic encephalopathy (HE) is a common and potentially devastating complication of liver disease.<sup>1</sup> Hepatic encephalopathy, which is a neuropsychiatric syndrome, is caused by two main mechanisms: the presence of important hepatic dysfunction and the diversion of the portal blood to the systemic circulation, without purification of toxic intestinal substances by the liver (porto-systemic shunts).<sup>2</sup> Hereditary hemorrhagic telangiectasia (HHT) or Osler-Weber-Rendu disease is an autosomal dominant genetic disease that is characterized by widespread telangiectases, which can involve the skin, mucous membranes, lung, brain, gastrointestinal tract and/or liver. The prevalence of hepatic involvement in HHT ranges between 41% and 78%.<sup>3</sup> Liver involvement in HHT is characterized by widespread diffuse liver vascular malformations that give rise to three types of shunting: arteriovenous (hepatic artery to hepatic vein), arterioportal (hepatic artery to portal vein), and portovenous (portal vein to hepatic vein).<sup>4</sup> However, most patients are asymptomatic; patients with HHT may have symptoms due to high-output heart failure, portal hypertension and biliary disease. Less commonly, patients may also develop porto-systemic encephalopathy, a condition that has been reported in 3 cases, two of which had or later developed symptoms of heart failure. In this case report, we present a patient with HHT complicated with encephalopathy.

## CASE

A woman, aged 63 years, was admitted with acute onset of melena and altered consciousness. In her past medical history she was diagnosed as having HHT and recurrent gastrointestinal bleeding attacks. In addition, she was diagnosed as having primary central nervous system (CNS) lymphoma and received 3 cycles of chemoradiotherapy 3 years ago. She had been in remission for 2 years. In her physical examination, we found that patient had slow movements, disorientation in time and space, paleness, telangiectases on her face and anterior of her body. In her cardiovascular examination, there was a 1/6 systolic murmur in the aortic area, blood pressure 110/70 mmHg, there was no orthostatic hypotension, and the pulse rate was 98/minute. Melena was diagnosed during her digital rectal examination. The laboratory analysis results were as follows: Hb: 8 gr/dl, MCV: 65 fl, transferrin saturation <10%, ferritin: 5 mg/dl, platelet: 500,000/ul, ammonia: 75 mg/dl (N<60) and coagulation tests, albumin, AST/ALT/ALP/GGT/bilirubin were normal. Cranial magnetic resonance imaging (MRI) was performed because the patient had altered consciousness and CNS lymphoma



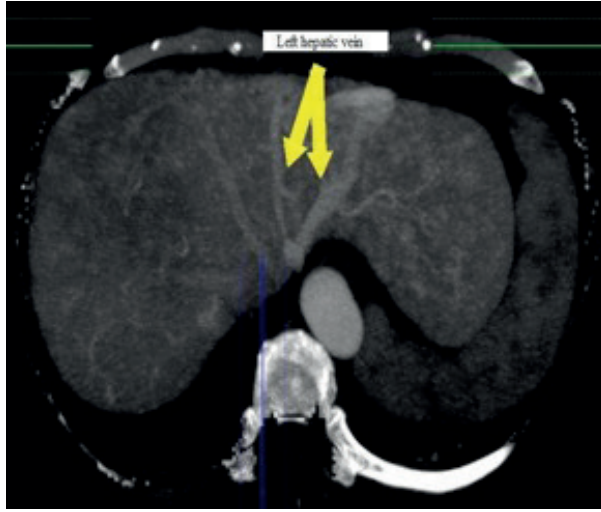
**Figure 1.** Gastric telangiectasia of patient in the gastroscopy

on her medical history. The results of the cranial imaging showed there were some changes compatible with age, but there were also secondary changes in the right parietal lobe due to biopsy at the time of diagnosis for CNS lymphoma. Referring to these cranial imaging findings, primer CNS lymphoma recurrence was excluded. Hepatic dysfunction was considered to explain the patient's altered consciousness because she had elevated ammonia levels. However, laboratory tests such as abnormal albumin, coagulation test, bilirubin and AST/ALT/ALP/GGT showed no signs of hepatic dysfunction. A gastroscopy was performed to evaluate the etiology of the upper gastrointestinal bleeding. The procedure revealed that there were no esophageal varices and that the origin of the hemorrhage was found to be a gastric telangiectasia (Figure 1). Even though the patient had high ammonia levels and altered consciousness, there were no laboratory findings that supported hepatic dysfunction. Therefore, we considered a portosystemic shunt and hepatic encephalopathy due to the liver involvement of HHT. Abdominal computed tomography angiography was the performed and a portohepatic shunt was found (Figure 2, 3). As a result, it was considered that the upper gastrointestinal bleeding had induced the hepatic encephalopathy due to the portohepatic shunt. The altered consciousness of the patient resolved with a low-protein diet, branched-chain amino acids, and lactulose.

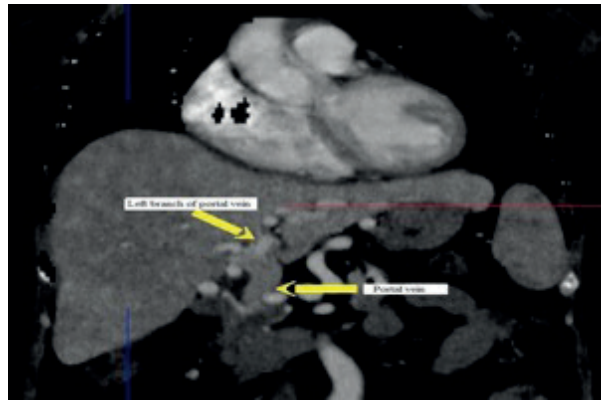
## DISCUSSION

Hereditary hemorrhagic telangiectasia is a vascular disease with hereditary autosomal-dominant transmission that has a frequency of 10 to 20 per 100,000 individuals.<sup>5</sup> The most frequently involved organs are the skin, lungs, gastrointestinal tract, and brain; hepatic involvement has been considered uncommon. Intrahepatic shunts, disseminated intraparenchymal telangiectases, and other vascular lesions are the typical findings of hepatic involvement.<sup>6</sup> Although patients with liver involvement are generally asymptomatic, congestive heart failure, portal hypertension, portosystemic encephalopathy, cholangitis, and atypical cirrhosis have been reported.<sup>7,8</sup> When the liver is involved in HHT, desaturated blood

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**Figure 2.** When there is no contrast in inferior vena cava in early arterial phase, contrast is present in left hepatic vein. This image supports systemic arterial shunt.



**Figure 3.** When there is no contrast in portal vein in early arterial phase, contrast is present in left branch of portal vein. This image supports systemic arterial shunt.

in the hepatic artery is shunted through the fistula into the portal or hepatic vein, bypassing the sinusoids.<sup>9</sup> The clinical manifestations depend on the magnitude of the shunt and comorbid conditions. HHT is a very rare etiology of hepatic encephalopathy. There are only 3 case reports in the literature about HHT with encephalopathy. Fukunaga et al demonstrated hepatic encephalopathy due to a portosystemic shunt in a patient with HHT in their case report, similar to our case.<sup>10</sup> Our patient was not considered as to have cirrhosis because of the synthesis function of the liver was normal and the imaging was not compatible with cirrhosis. In addition, the cranial imaging and other laboratory analyses failed to produce any finding that could explain the patient's mental status. Depending on the underlying disease, hepatic encephalopathy is subdivided into: type A, which result from acute liver failure; type B, predominantly from a portosystemic bypass or shunting; and type C, from cirrhosis.<sup>11</sup> The study of Ozturk et al showed that the most common factor that induced hepatic encephalopathy was spontaneous ascites infection. This case report also showed that gastrointestinal system bleeding was an important factor that could cause hepatic encephalopathy attacks.<sup>12</sup>

In conclusion, in a patient with known HHT and portosystemic shunt, after excluding alternative diagnoses, the patient's consciousness disturbance was attributed to hepatic encephalopathy induced by an upper gastrointestinal bleed.

\* The authors declare that there are no conflicts of interest.

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