CASE REPORT
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Hereditary hemorrhagic telangiectasia

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Abstract

Hereditary Hemorrhagic Telangiectasia (HHT) or Osler-Weber-Rendu disease is an autosomal dominant genetic disorder. HHT frequently presents with gastrointestinal bleeding with diagnostic and therapeutic challenge. Hepatic involvement in this disease is increasingly recognized and poses another therapeutic challenge. With advances in genetic screening and diagnostic procedures, and the increasing awareness of the condition by physicians and patients, this disease is now diagnosed more often. This case report reviews the available literature on various manifestations of HHT, and the various diagnostic and therapeutic modalities available for its management.

Keywords: Gastrointestinal; hepatic; hereditary; Osler-Rendu-Weber disease; telangiectasia

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Introduction

Hereditary hemorrhagic Telangiectasia (HHT) is an autosomal dominant disorder of the blood vessels, which is reported to affect one in 5,000 people. The disorder is also sometimes referred to as Osler-Weber-Rendu (OWR) disease.

It typically manifests as mucocutaneous or visceral angiodysplastic lesions (telangiectasias and arteriovenous malformations (AVM)), which most frequently involves skin, liver, lung, gastrointestinal tract, and brain. A recent study has shown that up to 74% of patients of HHT have liver involvement. [1]

We report a case of HHT with predominant liver involvement and present a brief review of literature.

Case Report

A 63-year-old male was presented with generalized weakness of 4 months duration and malena for 3 weeks. He also has past history of recurrent epistaxis. The routine blood test showed severe anemia and hemoglobin of 2.68 gm% with normal coagulation profile. Per abdomen examination showed hepatosplenomegaly. Sonography of the abdomen showed enlarged liver with normal echotexture. No focal lesion was seen. The hepatic artery was enlarged and tortuous and the hepatic veins were very prominent. Multi Detector Computed Tomography (MDCT) of the abdomen showed multiple small ill-defined enhancing lesions scattered throughout the liver [Figure 1]a. Enlarged hepatic veins were seen opacifying in the arterial phase indicating arterio-hepatic venous shunting [Figure 1]b.The left portal vein was also opacified in the arterial phase indicating arterio-portal venous shunting [Figure 1]a. The portal vein was of normal caliber. Mild focal Intra hepatic biliary radical dilatation was present in the left lobe. No venous collaterals were seen in the abdomen. The spleen was normal. No other abnormality was seen in the abdomen.

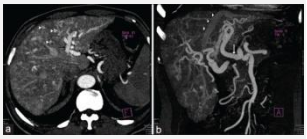

Figure 1: (a) Dynamic contrast enhanced MDCT scan of liver arterial phase. Axial MIP reconstructed image shows dilated hepatic artery (black arrowhead) and early filling of left portal vein (black arrow), suggesting presence of arterio-portal venous shunt. Multiple telangiectasias (white arrowhead), (b) Dynamic contrast enhanced MDCT scan of liver arterial phase. Coronal MIP reconstructed image shows dilated hepatic artery (arrow) and early filling of hepatic veins (arrowheads), indicating the presence of arterio-hepatic venous shunt

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Screening of the chest was done by covering the chest while doing computed tomography (CT) abdomen and was normal. The screening for the intracranial AVM was debated, as the patient was asymptomatic and high risk of diagnostic and treatment modalities.

As upper and lower gastrointestinal (GI) scopy was negative, capsule endoscopy done subsequently showed multiple telangiectasias in antrum of the stomach, duodenum, and jejunum. Active bleeding was seen in the mid ileum.

As the patient continued to have malena, diagnostic catheter angiogram was planned. After obtaining informed consent, patient was brought to the angiography suit. The procedure was done on single plane digital subtraction angiography (DSA) unit (Polystar Siemens Germany). Using standard technique, procedure was performed with 5 F right femoral access. After placement of the catheter, 3000 IU heparin was given through the sheath for thromboembolic prophylaxis. The celiac artery and superior mesenteric artery (SMA) selective injections done by using 4 F diagnostic Simmons I catheter, which showed tortuous dilated common hepatic artery and replaced right hepatic artery arising from SMA [Figure 2]. The hepatic vasculature showed abnormal tortuous arterial branches and intense parenchymal blush followed by early opacification of the hepatic veins. SMA injection showed an arteriovenous (AV) fistula in the mid ileum, which appeared to be the cause for malena [Figure 3]a and b. In view of multiple arteriovenous malformations (AVMs), a radiological diagnosis of HHT was made.

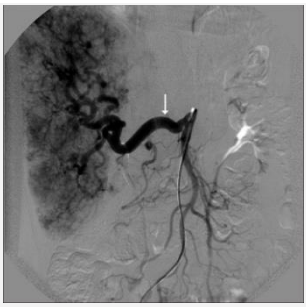

Figure 2: DSA: Selective superior mesenteric artery angiogram shows dilated replaced right hepatic artery arising from SMA (arrow). Intrahepatic RHA show diffuse telangiectasias

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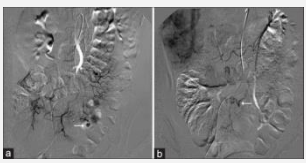

Figure 3: (a) DSA: Selective SMA angiogram in arterial phase showing ileal AV fistula (arrow), (b) DSA: Selective SMA angiogram in arterial phase showing early draining vein (arrow)

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As patient was having malena, coil embolization of the mid ileal AVF was planned in same sitting. The 2.7 F microcatheter (prograte, terumo corp.) was navigated coaxially through the diagnostic catheter into the culprit vessel and was embolized with microcoils (Cook medical) [Figure 4]a and b. The patient was stable post procedure for a week's time. One-week post embolization, the patient continued to have malena. Repeat catheter angiogram done showed persistence of AVF, which was being fed, by an adjacent branch of SMA. As the feeders were very small, tortuous collaterals form adjacent vessels, and the main access vessel was already occluded by previous coil embolization, we doubted feasibility of endovascular embolization after discussing with our gastrointestinal surgeon and physician. The affected bowel was resected surgically. Surgical pathology of resected bowel and intra-operative liver biopsy showed multiple AVMs and cirrhosis of liver.

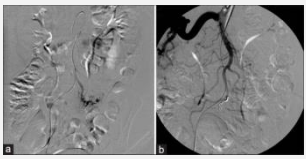

Figure 4: (a) DSA super selective angiogram through microcatheter showing AVF with early draining vein, (b) DSA super selective angiogram through microcatheter showing feeding vessel embolization with microcoils

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Discussion

HHT is an autosomal dominant inherited vascular disease with high penetrance but variable symptomatology. Two subtypes have been defined with mutations in chromosome 9q34 in type 1 and mutations in chromosome 12q31 in type 2. Furthermore, the SMAD4 gene has been shown involved in a very rare HHT type combined with juvenile polyposis. [2],[3]

Genetic testing for HHT has become available only recently in a few countries. It can confirm the clinical diagnosis and predict the spectrum of disease a person is likely to have.

HHT usually presents after the third decade with peak incidence being in 6 th decade. [4]

The manifestations of HHT are localized to the capillaries and are caused by a defect in normal growth and repair of endothelial cells. [5]

It is recognized that the manifestations of HHT are not seen generally at birth, but develop with increasing age such that nose bleeds are usually the earliest sign of disease, often occurring in childhood, pulmonary AVM's becoming apparent from puberty, with mucocutaneous and GI telangiectasia developing progressively with age. [6]

The established clinical diagnostic criteria for HHT are: [7]

1. Nosebleeds (epistaxis), which are spontaneous and recurrent (may be mild or severe)
2. Telangiectases on the skin or mucous membranes (mucocutaneous). Telangiectases are small red spots that blanch under pressure, located at characteristic sites, including the lips, oral cavity, fingers, and nose
3. Visceral AVMs, consisting of direct connections between arteries and veins. They may be located in the lungs, brain, liver, spinal cord, or GI tract
4. A first-degree relative (brother, sister, parent or child) with HHT, based on these diagnostic criteria.

A diagnosis of HHT is considered definite when three or more of these features are present, possible or suspected when two findings are present, and unlikely with fewer than two findings.

In our case, first three features were present. Family history of HHT was not present.

Virtually all small vessels (arterioles, capillaries, and venules) may be dilated and intercommunicate extensively. 60-70% of patients show telangiectases of the lips, tongue and finger tips. Epistaxis and GI bleeding occur despite normal hemostatic factors. [8]

The presence of hepatic telangiectasia and dilation of the common hepatic artery as signs of HHT liver disease have been described in many other studies. [1],[7],[9] These two findings are pathognomonic for HHT liver involvement, particularly in the presence of a compatible clinical history. Recognition of the condition is important since inadvertent biopsy can result in uncontrolled bleeding.

On CT, round strongly enhancing lesions with a diameter of less than 10 mm and a predominantly peripheral arrangement are considered parenchymal hepatic telangiectasias and enhanced lesions with a diameter of more than 10 mm are considered large confluent vascular masses. [1]

Most of the patients with hepatic involvement are asymptomatic. [9] Increased blood flow through AV fistulas, may cause hepatomegaly, pain in the right upper quadrant of the abdomen, or a pulsatile liver. Left to right shunting through liver fistulas may lead to high output cardiac failure. [1] Hepatic artery to portal vein shunts may lead to portal hypertension, with bleeding GI varices and encephalopathy. Fibrovascular histological changes may occur in liver, which lead to cirrhosis and its complications (portal hypertension, and tumor). Rarely, biliary strictures and dilatation are seen because of biliary ischemia due to shunting of blood away from peribiliary plexus, by arteriovenous fistulae (AVF). [10],[11] Anatomic variation of hepatic artery supply is also associated with HHT and is noted in our case. [1]

Liver involvement in HHT needs treatment only if the patient shows signs of liver and heart failure because of liver AVMs. There are no definite criteria for the embolization of these AVMs and decision should be taken on case-by-case basis. [12]

Nosebleeds are usually treated conservatively with humidification. Laser therapy or septal dermoplasty are considered if conservative treatment fails. Embolization is usually effective for only 6-8 weeks and used only on emergency basis as a temporary measure.

Skin telangiectasias/AVMs are treated, if symptomatic with laser therapy.

Lung and brain AVMs should be treated before they cause symptoms. Embolization is the method of choice for their management. Treatment of brain AVMs depends on location, size, and structure involved. Surgery, embolization, or stereotactic surgery may be needed separately or in combination for their treatment.

GI hemorrhage probably contributes less frequently to iron deficiency than under-recognized nose bleeds: In many patients, improvement in nasal hemorrhage is able to reduce iron and transfusion requirements significantly. However, recurrent gastrointestinal tract hemorrhage is a frequent issue affecting 13-33% of HHT affected individuals, particularly in later years. [6],[13] It often presents as an iron deficiency anemia but occasionally as an acute GI hemorrhage. The onset is usually from the 5 th or 6 th decade. Telangiectasia occurs throughout the gastrointestinal tract, and is more common in the stomach or duodenum, than in the colon. They are visualized by endoscopy and are similar in size and appearance to mucocutaneous telangiectases but may be surrounded by an anemic halo. Less commonly AVMs and aneurysms may occur, depicted by GI angiography. Most patients are satisfactorily managed conservatively with oral iron therapy and, if necessary, blood transfusions. Repeated laser therapy may also be used to control bleeding in the short term, though results are not as good as in the non-HHT population. [6] Embolization has limited

success due to recurrent disease, but may be useful for emergency control of hemorrhage from discrete lesions, as may surgery. [6] Antihemorrhagic medical treatments have been sought. The only therapy supported by evidence from randomized controlled trials is the use of female hormones (50 μg ethinyloestradiol and 1 mg norethisterone) in heavily transfusion dependent patients. [6]

In our case, there was single major feeder to AVF and was embolized with the steel microcoils. The use of glue and Polyvinyl alcohol (PVA) particles was deferred as we thought; these may reach till the precapillary venules and or arterioles increasing chance of bowel gangrene post procedure. [14] We know the coli embolization of feeder artery can provide relief for acute symptoms, but it is ineffective for long term management as collaterals develop. [12] Same was the reason for recurrence of hemorrhage in our case. To our knowledge, no uniform method for treating this subgroup of patients can be found in the literature.

We now believe that the issue of failure of the embolization procedure must be part of the consent. Although all of the treatments in current practice allow some relief to patients with HTT with GI hemorrhage, no treatment allows for a definitive cure in most patients. [12]

Retrospectively, in our view, use of larger PVA particles for embolization would have been better choice to reduce perfusion pressure in the AVF and to get better results and short-term benefit rather than proximal arterial coil embolization.

Even though our patient with GI hemorrhage required subsequent surgical therapy, we feel that endovascular therapy still has a role in HHT-related gastrointestinal hemorrhage. It may also be done in cases of acute intractable GI hemorrhage, for instance to create the opportunity for surgical intervention. Further evaluations are needed to evaluate the role of endovascular embolization for GI hemorrhage in HTT.

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