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PORTAL HYPERTENSION IN OSLER-WEBER-RENDU SYNDROME: A CASE REPORT

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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 patient with Osler-Weber-Rendu syndrome with portal hypertension without evidence of liver cirrhosis.

KEYWORDS : Hereditary haemorrhagic telangiectasia, Osler-Weber-Rendu syndrome, Ultrasound, CT scan

INTRODUCTION

Hereditary haemorrhagic telangiectasia (HHT, Osler-Weber-Rendu syndrome) has been subjected to under-reporting for many years. Recent careful epidemiological studies in France, Denmark, and Japan, however, revealed an incidence of one in 5–8000.[1-3]. HHT is an autosomal dominant condition. Despite the fact that nearly 20% of the cases do not have a family history; they could represent sporadic mutations.[4]. The rare case of systemic fibromuscular dysplasia is characterized by the presence of small, red, telangiectatic lesions of 2-3 mm diameter that are found on the skin and mucosal surfaces, particularly the face, lips, mouth, and nasopharynx.[5]. Epistaxis from nasal mucosal telangiectasia being the most common presentation of this disease that is seen in 32-85% of patients.[6]. The respiratory symptom most commonly present at the time of diagnosis is dyspnea and hemoptysis are a less common symptom.[5]. Hemoptysis ranges in frequency from 4 to 18%.[6].

Case report:

A 50-year-old male known case of Osler-Weber-Rendu Syndrome with history of recurrent admissions to hospital for blood transfusion due to recurrent epistaxis presented with complaints of shortness of breath (SOB), abdominal distension, swelling of feet, and easy fatiguability since last 15 days. SOB was associated with orthopnea and dyspnea in left recumbent position.

On physical examination, the patient had tachycardia, pallor, raised jugular venous pressure, and pedal edema. Oral telangiectasias were also present.

Respiratory system examination revealed shifting of trachea to left side and decreased chest movement on the right side. Breath sounds were absent in the right infra-axillary area and diminished in right infrascapular area.

Cardiovascular examination revealed tachycardia along with pan systolic murmur in tricuspid area.

Abdominal examination showed soft and distended abdomen with flanks full with shifting dullness.

On laboratory investigations, the patient was reported to be severely anemic (Hb 6.5 g/dl), leucopenia (TLC 3.7 X 10^9/L) with hypoalbuminemia (Albumin 3.3 g/dl). Peripheral blood film showed microcytic hypochromic anemia and leucopenia. Serum iron examination was suggestive of iron deficiency (Fe 20 μg/dl; total iron binding capacity 471 μg/dl; transferrin saturation 4.89%; serum ferritin 7.49 ng/ml).

Electrocardiography was suggestive of left ventricular hypertrophy and chest X-ray showed right sided pleural effusion which was tapped and found to be transudative (pleural fluid LDH/serum LDH=0.51; pleural fluid protein/serum protein=0.42). Ascitic fluid was tapped and was found to have high serum ascitic albumin gradient and low protein (Table 1).

On further evaluation, ultrasound abdomen showed hepatomegaly with coarse echotexture and nodular outline with splenomegaly. ultrasound color Doppler showed multiple intrahepatic tortuous dilated vessels in both lobes of liver (Figure 2a) with portal vein (PV) diameter of 15 mm and velocity of 34.89 cm/sec (Figure 2b).

Triple phased CT scan of abdomen showed dilated celiac artery, left gastric, common hepatic and its branches, splenic and superior mesenteric artery with tortuous vessels arising from left gastric artery. PV hypertension with dilated PV with splenomegaly with evidence of tortuous dilated collaterals was seen at porta hepatis, splenic hilum and perigastric region. Hepatomegaly without evidence of cirrhosis; however, features of transient hepatic attenuation difference were seen in liver parenchyma. Ascites, right pleural effusion and feature suggestive of congestive cardiac failure with early filling of inferior vena cava were also present.

Upper gastrointestinal endoscopy revealed multiple telangiectasias along with 3 active bleeding sites in stomach and telangiectasias in 2nd part of duodenum (Figure 3).

DISCUSSION

Osler-Weber-Rendu disease is a hereditary disease that can present with sporadic mutation and different phenotype variations. Many genes have been implicated with the disease pathogenesis; however, the most important one so far is located on 9q that codes for a surface glycoprotein endoglin, a surface receptor for TGF-□, which will mediate vascular remodeling by affecting the extracellular matrix production.[7,8] Sixty percent of patients with pulmonary arteriovenous malformations have hemorrhagic hereditary telangiectasia; however, only 15-33% of people with this disease have pulmonary arteriovenous malformations.[9] Patients diagnosed with this disease are mostly in their thirties or forties with the mean age being 38-40 years.[6] However occasionally the condition is detected in infancy.[5]

The diagnosis of OWRD or HHT is based on Curacao clinical criteria followed as[10]: epistaxis; telangiectasias—characteristic sites include lips, oral cavity, fingers and nose; visceral lesions including gastrointestinal, hepatic, PAVMs, cerebral AVMs (CAVMs) or spinal AVMs; family history with a first-degree relative with HHT. Presence of all three criteria makes a definite diagnosis and two are needed for a possible diagnosis.

HHT is a common cause of hepatic involvement in the form of focal nodular hyperplasia and hepatic AVMS, more commonly reported with activin receptor-like kinase-1 mutation with a prevalence of up to 84%. However, portal hypertension is uncommon with presence in less than 10% patients.[11,12] Portal hypertension and hepatic encephalopathy more commonly appear to result from increased sinusoidal blood flow, leading to enhanced deposition of fibrous tissue and pseudo cirrhosis of the liver.[13,14]

Prognosis

Most HHT patients who have adequate access to healthcare will have normal life expectancies. There is a bimodal distribution of mortality, with peaks at age 50 and then from 60 to 79. Most of the mortality of HHT is the result of complications of AVMs, particularly in the brain, lungs, and GI system.[15]

Table 1: Ascitic fluid analysis

	Value
Appearance	Straw and clear
Total protein	2.1 g/dl
Albumin	1.0 g/dl
Glucose	90 mg/dl
WBCs	5.9 X10^3/L (N10%, L90%)
SAAG	2.3

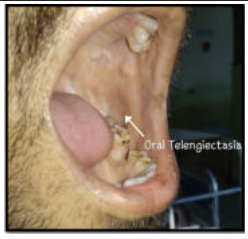


Figure 1: Oral telangiectasia

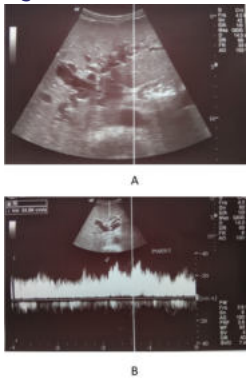
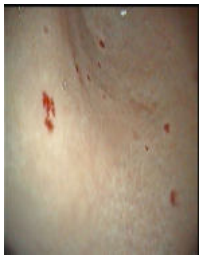


Figure 2: Ultrasound Color Doppler A) Tortuous dilated vessels B) portal vein velocity



CONCLUSION

Osler-Weber-Rendu syndrome is a rare cause of vascular dysplasia involving many organs and systems. A case of portal hypertension in a patient with HHT was presented.

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