Case Report

Osler-Weber-Rendu disease: A rare cause of recurrent hemoptysis

Amir M Khoja, Rahul K Jalan, Dheeraj L Jain, Omkar V Kajale

Department of Chest Medicine and Thoracic Endoscopy, Ruby Hall Clinic, Pune, Maharashtra, India

ABSTRACT

Osler-Weber-Rendu disease, also known as hereditary hemorrhagic telangiectasia, is a rare autosomal dominant condition causing systemic fibrovascular dysplasia. It has an incidence of 1-2/100,000. Phenotypic variation is extreme ranging from asymptomatic to severely symptomatic, from cases with no or few mucocutaneous lesions to those with diffuse cutaneous telangiectasia. We discuss a case of Osler-Weber-Rendu disease causing diffuse cutaneous telangiectasia and hemoptysis. The patient presented with complaints of hemoptysis and was extensively examined and investigated before being diagnosed with Osler-Weber-Rendu disease. We successfully managed the patient's hemoptysis by bronchial artery embolization. This case emphasizes the need for careful examination and investigation and to consider such rare diseases when all the common causes of hemoptysis are ruled out. An early and proper diagnosis will lead to more effective management of such a rare disease with few treatment options available.

KEY WORDS: Bronchial artery embolization, hemoptysis, Osler-Weber-Rendu disease, telangiectasia

Address for correspondence: Dr. Amir M Khoja, Dr. Khoja Clinic, NIRMAL, Next to Hotel Madhuban, Dhole Patil Road, Pune - 411 001, Maharashtra, India. E-mail: amirmkhoja@gmail.com

INTRODUCTION

Osler-Weber-Rendu disease or hereditary hemorrhagic telangiectasia 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.[1] 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.[2] The mucosal lesions usually blanch on pressure and are highly vulnerable to spontaneous rupture and cause bleeding. The diagnosis of this rare condition is made on the basis of the Curaçao criteria established in June, 1999 that includes the following factors: epistaxis, telangiectasia, visceral lesions, and family history. The diagnosis is confirmed by the presence of at least three of these manifestations. Epistaxis from nasal mucosal telangiectasia being the most common presentation of this disease that is seen in 32-85%[3] of patients. The respiratory symptom most commonly present at the time of diagnosis is dyspnea and hemoptysis is a less common symptom.[2] Hemoptysis ranges in frequency from 4 to 18%.[3] This case presented with hemoptysis that is a rather uncommon manifestation of the disease causing a diagnostic dilemma.

CASE REPORT

A 19-year-old nonsmoker female patient presented with complaints of hemoptysis with multiple episodes each of 100-200 mL and breathlessness at rest since 1 day. She had a history of epistaxis 2 years ago, complete details of which were not available. There was no history of hemoptysis prior to this episode neither was there any history of pulmonary tuberculosis, exposure to toxic fumes, or travel. The patient's mother had a

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history of severe unexplained menorrhagia. Her family history was not significant. On examination, the condition of the patient was found to be vitally stable. There was no evidence of cyanosis, clubbing, lymphadenopathy, pedal edema, and icterus. Telangiectatic lesions were seen in her lower lip. The respiratory system examination of the patient revealed a decrease of air entry into the right infrascapular region with crepitations heard in the right infrascapular region. Rest of the results of the system examination were normal.

Hematologic investigations showed a hemoglobin level of 10 g/dL, total leucocyte count of 12,000 cells/cm^3, and platelet count of 2.9 × 10^3/L. Her coagulation profile was normal. X-ray of her chest showed an inhomogeneous opacity in the right lower zone with blunting of the costophrenic angle. High-resolution computed tomography (HRCT) of her thorax with CT pulmonary angiography revealed multiple patchy areas of consolidation, air space opacities, and ground glass opacities in both the lungs. Cardiomegaly with mild right atrial and right ventricular dilatation were also diagnosed. Pulmonary arteries and veins looked normal with no e/o arteriovenous malformation or embolus. Two-dimensional echocardiogram (2D Echo) was suggestive of left ventricular ejection fraction (LVEF) in 65% with mild right atrial and ventricular dilatation; no other cardiac abnormality was detected.

Bronchoscopy under awake sedation was done that showed telangiectasia in the Little's area [Figure 1], vocal cords [Figure 2], trachea [Figure 3], and all over the bronchial mucosa. Multiple pin-point areas of bleeding were seen near the telangiectatic vessels [Figure 4]. Multiple blood clots in the right middle and lower lobe bronchus with areas of active bleeding were seen. All the clots were removed and hemostasis was achieved by argon plasma coagulation of the telangiectatic vessels. Abdominal ultrasonography (USG) with hepatic Doppler was done that showed no arteriovenous malformation. Hematological opinion was taken and taking all the findings into consideration a diagnosis of Osler-Weber-Rendu disease was done. The patient was started on thalidomide (100 mg) taken orally once a day and was under observation. She had no further episode of hemoptysis, hence, was discharged.

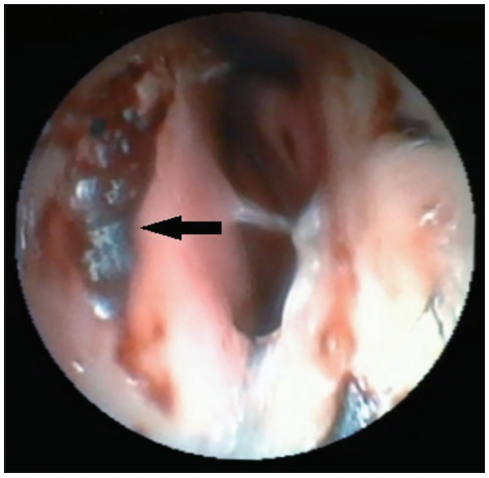

Figure 1: Telangiectatic lesion in the Little's area

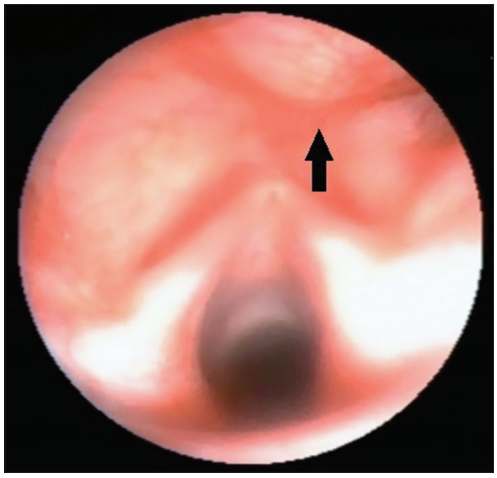

Figure 2: Telangiectatic lesion over vocal cords

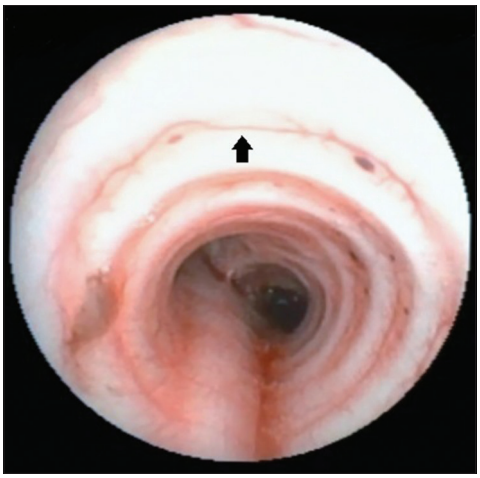

Figure 3: Telangiectatic lesion in the trachea

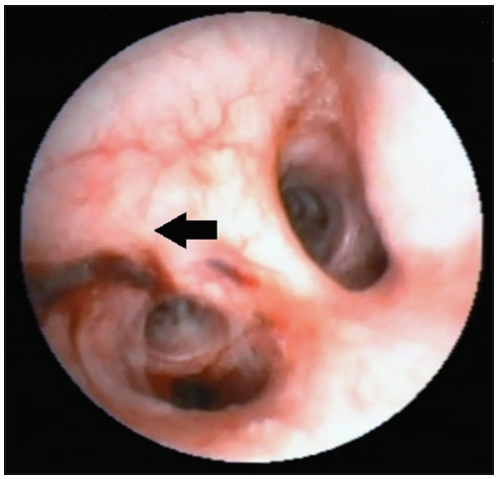

Figure 4: Telangiectatic lesion in the left main bronchus

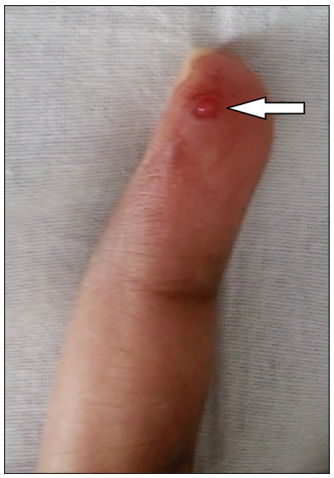

Figure 5: Telangiectatic lesion on the middle finger of the left hand

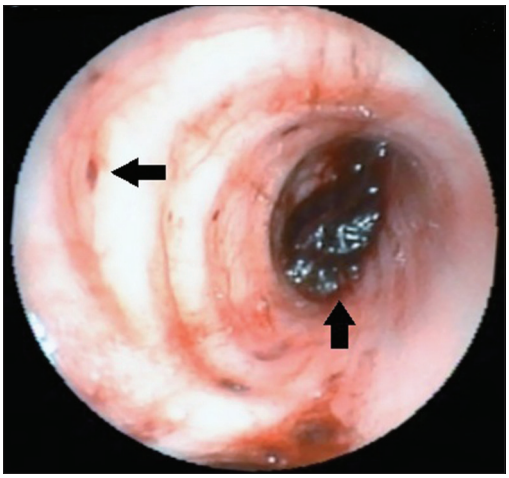

Figure 6: Blood clot in the right middle lobe (RML)

Fifteen days later, the patient returned with complaints of hemoptysis, with four episodes each of 20-30 mL in the last 4 days. Telangiectatic lesions were present on the left middle finger [Figure 5] with active bleeding and were not responding to the treatment. X-ray of her chest showed collapsed right middle lobe with tracheal and mediastinal shift. Bronchoscopy of the patient revealed telangiectatic lesions all over the bronchial mucosa and clots occluding the right middle lobe [Figure 6]. Although clots were removed, active bleeding was seen through the medial segment of the middle lobe. *N*-butyl cyanoacrylate was instilled in the right middle lobe and argon plasma coagulation of all the telangiectatic lesions was done. Hemostasis was achieved.

Next day, the patient complained of epistaxis from the left nostril and massive hemoptysis with clots. The epistaxis was controlled by nasal packing. Bronchial artery angiogram was done that showed blush and leakage from the right middle lobe artery. Embolization of the right middle lobe artery was done with gel foam and hemostasis was achieved. The patient was discharged after an observation period of 4 days.

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- β, which will mediate vascular remodeling by affecting the extracellular matrix production.[4,5] Sixty percent of the patients with pulmonary arteriovenous malformations have hemorrhagic hereditary telangiectasia; however, only 15-33% of people with this disease have pulmonary arteriovenous malformations.[6] Patients diagnosed with this disease are mostly in their thirties or forties with the mean age being 38-40 years[3] However, occasionally the condition is detected in infancy.[2] Advanced stages of this disease are associated with the development of arteriovenous malformations in the lungs, liver, central nervous system (CNS), and gastrointestinal tract.[7] The most common complication is hemorrhage, either in the bronchial tree producing hemoptysis or rarely into the pleural cavity producing hemothorax.[2]

Management of this disease is aimed at relieving the symptoms, management of complications, and preventing the progression of the disease. Management of pulmonary complications would require embolization with gel foam or coil. Angiogenesis-inhibiting substances like Thalidomide and Bevacizumab have been shown to be effective in treating patients with advanced diseases. An important aspect in the treatment of such patients is to keep the systolic blood pressure around 100 mmHg to prevent recurrences.

Our case is unique as it probably represents a sporadic mutation leading to Osler-Weber-Rendu disease that leads to the patient's symptoms. The age of the patient at the onset of symptoms was 19 years, which is quite less than the mean age reported in many large case series. After embolization, the patient was advised to continue thalidomide intake. In the last 6-month follow-up, the patient did not have recurrence of any of her symptoms.

In a country like India where hemoptysis is most commonly associated with tuberculosis, it is important to study the symptoms of such rare diseases before initiating the treatment. As only few treatment options are available for such diseases, the rates of morbidity and mortality are significantly high.

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Conflicts of interest
There are no conflicts of interest.

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