Case report

Osler–Weber–Rendu disease: report of 3 cases in a family

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Introduction

Osler–Weber–Rendu disease, also referred to as hereditary haemorrhagic telangiectasia, is a genetic disease, an autosomal dominant disorder characterized by telangiectasias and aneurysms with primary involvement of the mucosa. The reported prevalence of this disease is 1–2 cases per 100 000 population [*1*]. The disease most commonly occurs in white patients, but has also been described in patients of Asian, African and Arab descent, and occurs with equal frequency and severity in both sexes. The syndrome most often presents by the third decade of life, but may be clinically silent [*1,2*].

History of epistaxis is noted in 50%–90% of affected patients. Telangiectasias are most commonly seen on nail beds, palms, lips, tongue, ears, face and chest skin. Similar findings on other skin and mucosal surfaces may be seen in this syndrome [*3–5*].

We describe 3 males from a single family affected with Osler–Weber–Rendu disease. The proband case had been admitted to a general hospital with primary presentation of severe chronic anaemia and with a history of blood transfusion and diagnosis of peptic ulcer. Diagnosis of this syndrome was considered when the patient was referred for dermatologic consultation because there were some telangiectatic vasculae on his earlobe.

Case reports

Case 1

A 63-year-old Iranian man who complained of fatigue, pale appearance, weakness and asthenia was the proband case. He had been hospitalized in the internal medicine ward of Afzalipoor Hospital in Kerman for further evaluation and to determine the cause of the anaemia. In his past history he mentioned many episodes of epistaxis. He had a history of 7 blood transfusions, all of them because of severe anaemia. All attacks of fatigue had appeared after episodes of haematemesis or strong melaena. Comprehensive clinical observation and physical examination of the patient were performed as well as dermatological consultation. Many mat-like telangiectasias were found. Erythematous patches were apparent on the nose tip and also on the nose wing surface. Similar eruptions were seen on lip vermilions and tongue tip, all of which suggested a diagnosis of Osler–Weber–Rendu disease. Endoscopic examination and rectosigmoidoscopy were performed, and showed many telangiectatic malformations that were distributed on the mucosal surface.

Typical telangiectasis on both lips and on the tongue tip, along with gastrointestinal telangiectasia are all supportive of a diagnosis of Osler–Weber–Rendu disease.

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Received: 28/04/05; accepted: 25/09/05

The patient had no history of any other disorders such as heart or renal failure or pulmonary disease. Laboratory examination showed severe anaemia (haemoglobin = 7 g/dL) with slight abnormalities in liver function tests. No serologic evidence was found of viral hepatitis B or C.

Case 2

This patient was a 61-year-old man, the brother of the patient in Case 1, who had a similar history, i.e. paleness of face, faintness and gastrointestinal bleeding. On physical examination he had many telangiectatic lesions on the nasal alae, lower lip, tongue, ears and in both conjunctivas. He had had 2 episodes of severe anaemia in the past and had to have blood transfusions due to a decrease in serum haemoglobin level to 9 g/dL.

Case 3

This was a 28-year-old male, the first son of the patient in Case 2, with a similar history, faintness, gastrointestinal bleeding and feebleness. On physical examination we found he had a few telangiectasias on his lip and nasal mucosae. He also complained of recurrent epistaxis and had a history (1 episode) of decrease in serum haemoglobin level (10.5 mg/dL), and had been given a blood transfusion.

Discussion

Osler–Rendu–Weber disease has multiple manifestations. Severe anaemia due to chronic gastrointestinal bleeding in our 3 cases, who were male Caucasian race, was the first presentation of this syndrome. All had a history of hospital admission for gastrointestinal bleeding due to subclinical peptic ulcer and had been given blood transfusions. Diagnosis was finally determined through dermatological consultation.

The clinical manifestations of this condition are caused by the development of abnormal vasculature, including telangiectases [*1,2*]. Clinically, nasal telangiectasia is more likely to bleed than cutaneous telangiectasia. This may be because of stronger tissue supporting telangiectases in the skin compared with the relatively weak tissue in mucous membranes [*2–4*]. The cause may be loss of the muscular layer and greater disturbance of the elastic lamina of vessel walls in this area, and this may give rise to aneurysms in multiple organ systems [*5,6*].

Most commonly, telangiectases involve the mucous membranes, the skin, the conjunctiva, the retina and the gastrointestinal tract. Arterial venous malformations may found in the lungs, brain and liver [*3,4*]. Patients are at risk of haemorrhage from multiple sites (especially the nasal mucosa), pulmonary haemorrhage, high-output cardiac failure, ischaemic stroke, migraine and paradoxical emboli, but fewer than 10% of patients die of complications of the disease [*3,4*]. Recurrent epistaxis is observed in up to 90% of patients; in half of these, epistaxis becomes more diffuse and serious with age, and blood transfusions are required in 10%–30% of cases.

None of our patients had complained of pulmonary haemorrhage in the past. Although patients with pulmonary vascular malformation and telangiectasis of the gastrointestinal tract are at risk of life-threatening internal haemorrhage, pulmonary vascular malformations are present in only 15%–20% of patients, and in half of these are usually asymptomatic [*5–7*], as in the cases reported here.

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