

An exceptional cause of iron deficiency anemia: Osler-Weber-Rendu Syndrome

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Abstract

Osler-Weber-Rendu syndrome, as known as hereditary hemorrhagic telangiectasia, is a kind of vascular dysplasia that involve skin, mucosal surfaces and internal organs. It has an autosomal dominant inheritance pattern. Incidence of the disease is 1-2/100000. Here we report a case of iron deficiency anemia after repeated multiple attacks of nosebleed whom diagnosed with Osler- Weber-Rendu syndrome.

Keywords

Osler-Weber-Rendu syndrome; iron deficiency anemia; vascular dysplasia

Introduction

Osler-Weber-Rendu (OWR) syndrome, as known as hereditary hemorrhagic telangiectasia, is a kind of vascular dysplasia that involve skin, mucosal surfaces and internal organs [1]. It's incidence in general population is 1-2/100000 [2]. Diagnostic criteria of OWR syndrome are as follows: (a) spontaneous and recurrent nosebleed, (b) more than one telangiectasia in characteristic localizations (lips, oral cavity, nasal cavity, and fingers), (c) visceral lesions (gastrointestinal telangiectasia, pulmonary, hepatic, spinal and cerebral arteriovenous malformations (AVM)), and (d) history of OWR syndrome in a first degree relative³. Diagnosis is certain if three of four criteria met, however, diagnosis is likely if two criterion present and not likely if less than 2 criterion present [3].

Epistaxis due to the rupture of telangiectasia in nasal cavity is the most common and the earliest symptom of OWR syndrome [4]. It emerge in about 95% of the patients with a frequency of about 18 times a month [4]. Median age for the beginning of epistaxis is 12 years. Epistaxis may be as severe as requiring treatment for iron deficiency anemia, however, it may also be mild in some of the cases [5]. Especially in those subjects with mild symptoms, diagnosis can be missed.

We present here a case with iron deficiency anemia that diagnosed with OWR syndrome after clinical evaluation.

Case Report

A fifty one year old man admitted to internal medicine clinic of our institution with fatigue and weakness. He had history of frequent epistaxis episodes since childhood. However he has not been

diagnosed with OWR syndrome due to the bleeding was mild most of the times. His mother had a diagnosis of OWR syndrome a few years ago. Vital signs of the patient was normal on admission. Besides he was looking pale, multiple telangiectases seen on lips and tongue mucosa on physical examination (*Photos 1 and 2*).

His hemoglobin level was 10.9 g/dl, hematocrit was 32% mean corpuscular volume was 78 fL in hemogram test. Other blood tests were as follows: serum iron: 26 ug/dl (reference range: 31-144 ug/dl), iron binding capacity: 326 (reference range: 69-240ug/dl), and ferritin: 6,7 ng/ml (reference range: 21-274). Other biochemical tests were normal. Diagnosis of iron deficiency anemia was established and underlying cause researched. Urine analysis was negative for blood.

Abdominal sonography and thoracic angiography were normal. Upper gastrointestinal endoscopy revealed multiple telangiectasia on oropharynx, millimetric angiodysplastic lesions in corpus and antrum of the stomach and in duodenum. A colonoscopy revealed 2 millimetric angiodysplasias in sigmoid colon. Diffusion MR imaging of the brain revealed 11x8 mm telangiectasia adjacent to the right insular cortex (*Figures 1, 2 and 3*).

Iron replacement initiated for anemia. He was referred to a more experienced neurosurgery center after neurosurgery consultation for treatment of cerebral telangiectasia. Patient was discharged with a suggestion of revisit of our clinic after 2 months.

Discussion

Osler- Weber-Rendu syndrome is a kind of vascular dysplasia that involve skin, mucosal surfaces and internal organs. Earliest and most common symptom of the syndrome is epistaxis [4]. Although chronic iron deficiency anemia may develop cause of epistaxis, it may also be mild and may not require iron replacement in some cases [5]. Present case had an iron deficiency anemia at the time of admission.

Some of the cases with OWR syndrome define exacerbation of the epistaxis episodes, however, severity of the episodes may not be changed and even may reduce by time. Lesions may occur in central nervous system in infancy or in childhood, however, may delay till adult age, as seen in present case [6]. Despite pulmonary AVM increase the risk of neurologic symptoms, headache and seizures may occur in any time when intracranial bleeding develops [7]. There was a cerebral AVM in present case, but, luckily, he had no past history of intracranial bleeding.

Some authors may suggest further investigation of central nervous system for AVMs by angiography due to MR imaging might underestimate the number of these lesions [7,8]. Therefore, we performed a diffusion MR instead of conventional MR imaging.

Genetic testing is suggested for patients with suspected OWR syndrome. However, due to financial limitations of the patient, he was not consulted to genetic department.

Telangiectases of skin, nose, liver and gastrointestinal system were treated when bleeds, however, pulmonary and cerebral lesions should be treated even in patients without symptoms [9]. Therefore we referred the patient to a more sophisticated neurosurgery center after consultation to neurosurgery department of our institution.



Photos



Figures

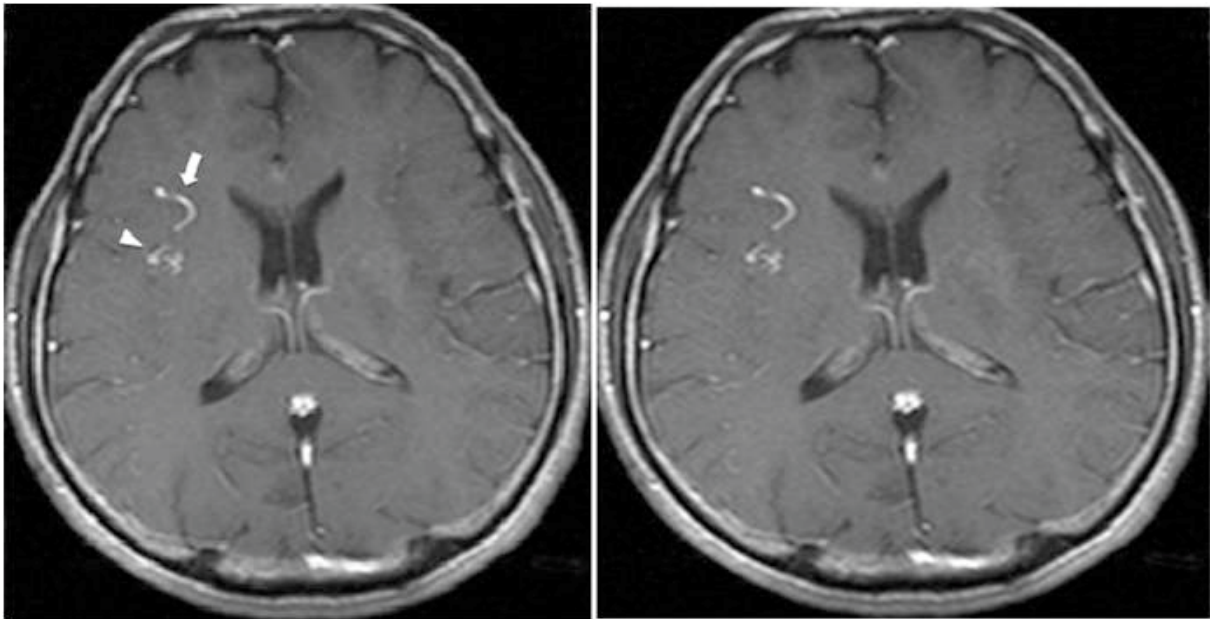


Figure 1: Axial contrast enhanced T1 weighted MR image. A bundle of vessels forming a telangiectasia shows contrast enhancement in right insula (arrowhead). Anterior to the telangiectasia, a feeding vessel, which is filled with contrast medium, can also be seen (white arrow).

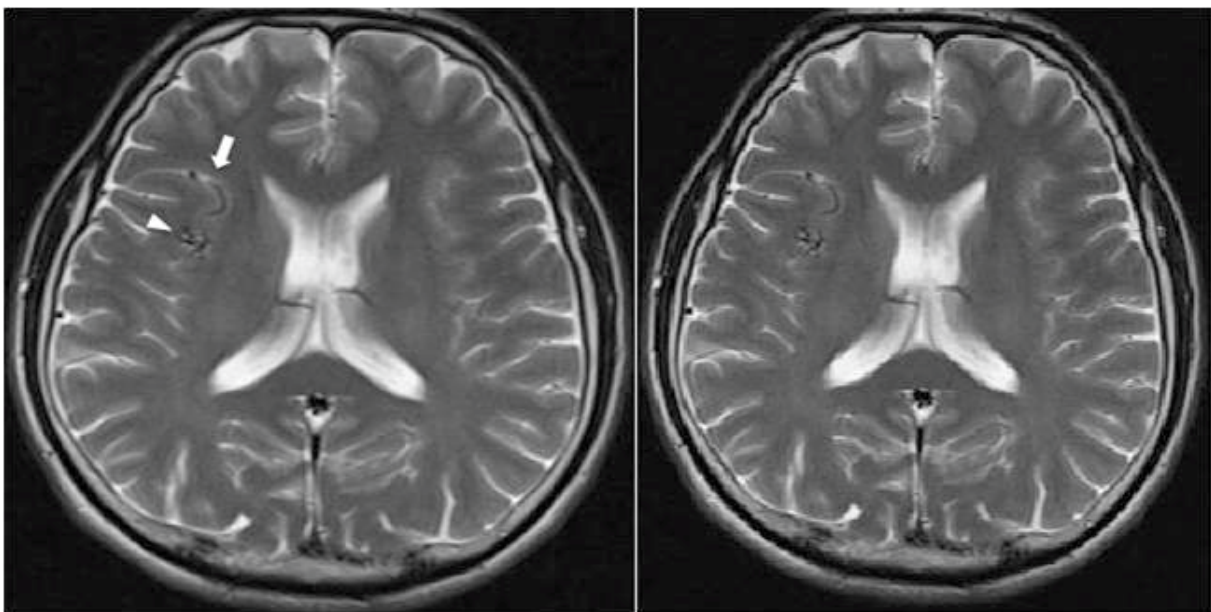


Figure 2: Axial T2 weighted MR image. Flow voids of the vessels in telangiectasia can be seen in right insula (arrowhead). A feeding vessel can be seen anterior to the telangiectasia (white arrow).



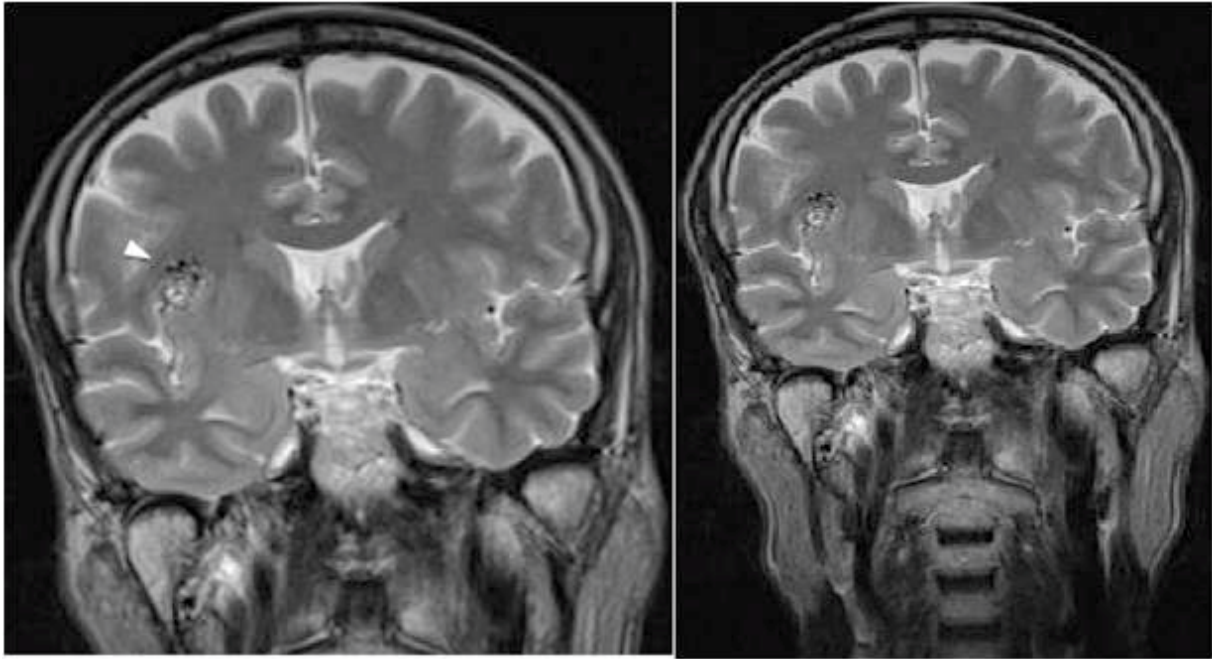


Figure 3: Coronal T2 weighted MR image. Flow voids in blood vessels of telangiectasia can be seen in superior part of right insula (arrowhead).

Conclusion

In conclusion, OWR syndrome should be kept in mind in patients with iron deficiency anemia who have a medical history of multiple bleeding episodes. Screening is strongly advised for the family members.

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