QJM: An International Journal of Medicine, 2020, 1–2

doi: 10.1093/qjmed/hcaa094
Advance Access Publication Date: 14 March 2020
Clinical picture

CLINICAL PICTURE

Osler–Weber–Rendu syndrome

A 57-year-old man with diabetes and chronic hepatitis B presented with epistaxis and symptomatic anaemia. His past medical history was relevant for recurrent presentations with epistaxis resulting in chronic iron deficiency anaemia requiring transfusions. Physical examination reviewed multiple telangiectasias affecting the tongue, lips (Figure 1A), face and scalp, digits and the palmer aspect of the hands (Figure 1B). He also has numerous telangiectasias in the nasal passage. Patient recounted that bleeding can be severe; profuse epistaxis and bleeding from fingertips and scalp with spurting that can reach the ceiling. Upper gastrointestinal endoscopy showed several telangiectasias affecting the duodenum. Computed tomography scan showed multiple small arteriovenous malformations (AVMs) affecting the liver. Brain and chest scans were normal. Each episode of epistaxis was treated with nasal packing and cauterization. Embolization of the left internal maxillary artery had to be done (Figure 1C) in a previous admission. There was no family history of similar condition.

Osler-Weber-Rendu Syndrome (Hereditary Haemorrhagic Telangiectasia) named after Sir William Osler (Canadian physician), Frederick Parkes Weber (English dermatologist) and Henri Jules Louis Marie Rendu (French physician) who independently described the condition in the late 19th and early 20th centuries is a rare autosomal dominant condition (mutation endoglin [HHT1] or ACVRLK1 [HHT2] genes) characterized by presence of multiple AVMs and recurrent epistaxis.[1] Clinical diagnosis is based on the Curacao criteria: (i) spontaneous recurrent epistaxis, (ii) mucocutaneous telangiectasia, (iii) AVMs of visceral organs and (iv) first degree relatives with similar condition. The diagnosis is definite if ≥3 criteria and possible if only two criteria are met. The condition only becomes apparent in adulthood. Recurrent epistaxis and chronic iron deficiency anaemia are common presentations. Large AVMs can be associated with high-output cardiac failure due to shunting and stroke due to paradoxical embolization.[2] Treatment consists of iron replacement, transfusions and ablative therapies for the telangiectasia.[3] Thalidomide (anti-angiogenesis) can be tried but recently anti-vascular endothelial growth factor antibody (Bevacizumab) has been shown to be beneficial.[4]

Photographs and text from: B.I. Mani, A.R. Rubel, W.A. Chauhdary, A. Bashir, Z.N. Soe, N. Javed, S.M.A. Sharif, M.T. Hla Aye and V.H. Chong, Department of Medicine, PMMPHAMB Hospital, Jalan Sungai Basong, Tutong, Brunei Darussalam. email: babuivan@gmail.com

Consent for publication

Consent has been obtained from patient for publication.

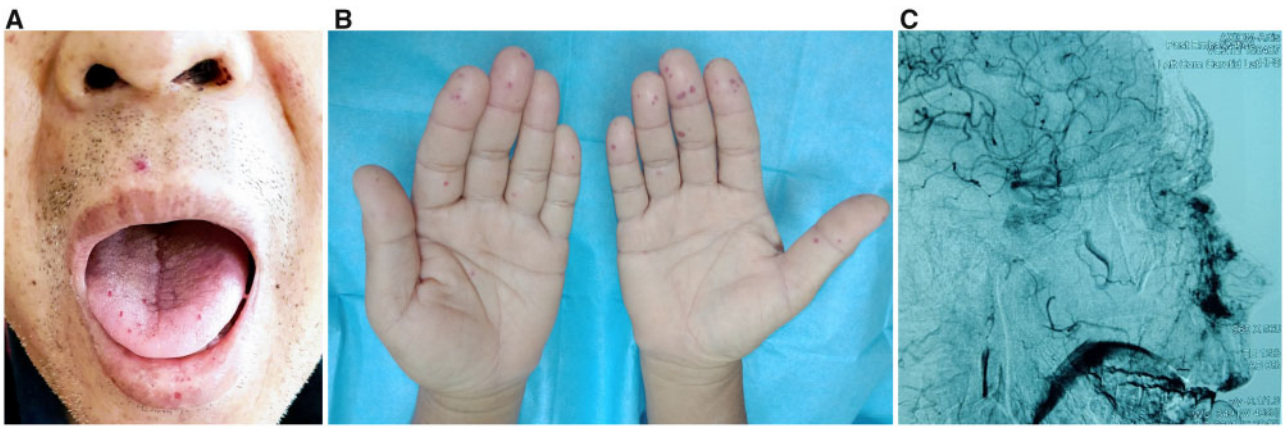

Figure 1. (**A**) Telangiectasias on the tongue, lip and peri-oral region, (**B**) palmer aspects of the hands and (**C**) post-embolization image showing contrast blush in the nasal, upper lip and hard palate areas.

Conflict of interest: None declared.

References

1. Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJ, *et al*. Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). *Am J Med Genet* 2000; **91**:66–7.
2. Moussouttas M, Fayad P, Rosenblatt M, Hashimoto M, Pollak J, Henderson K, *et al*. Pulmonary arteriovenous malformations: cerebral ischemia and neurologic manifestations. *Neurology* 2000; **55**:959–64.
3. Geisthoff UW, Nguyen HL, Röth A, Seyfert U. How to manage patients with hereditary haemorrhagic telangiectasia. *Br J Haematol* 2015; **171**:443–52.
4. Bose P, Holter JL, Selby GB. Bevacizumab in hereditary hemorrhagic telangiectasia. *N Engl J Med* 2009; **360**:2143–4.