Osler-Weber-Rendu Syndrome

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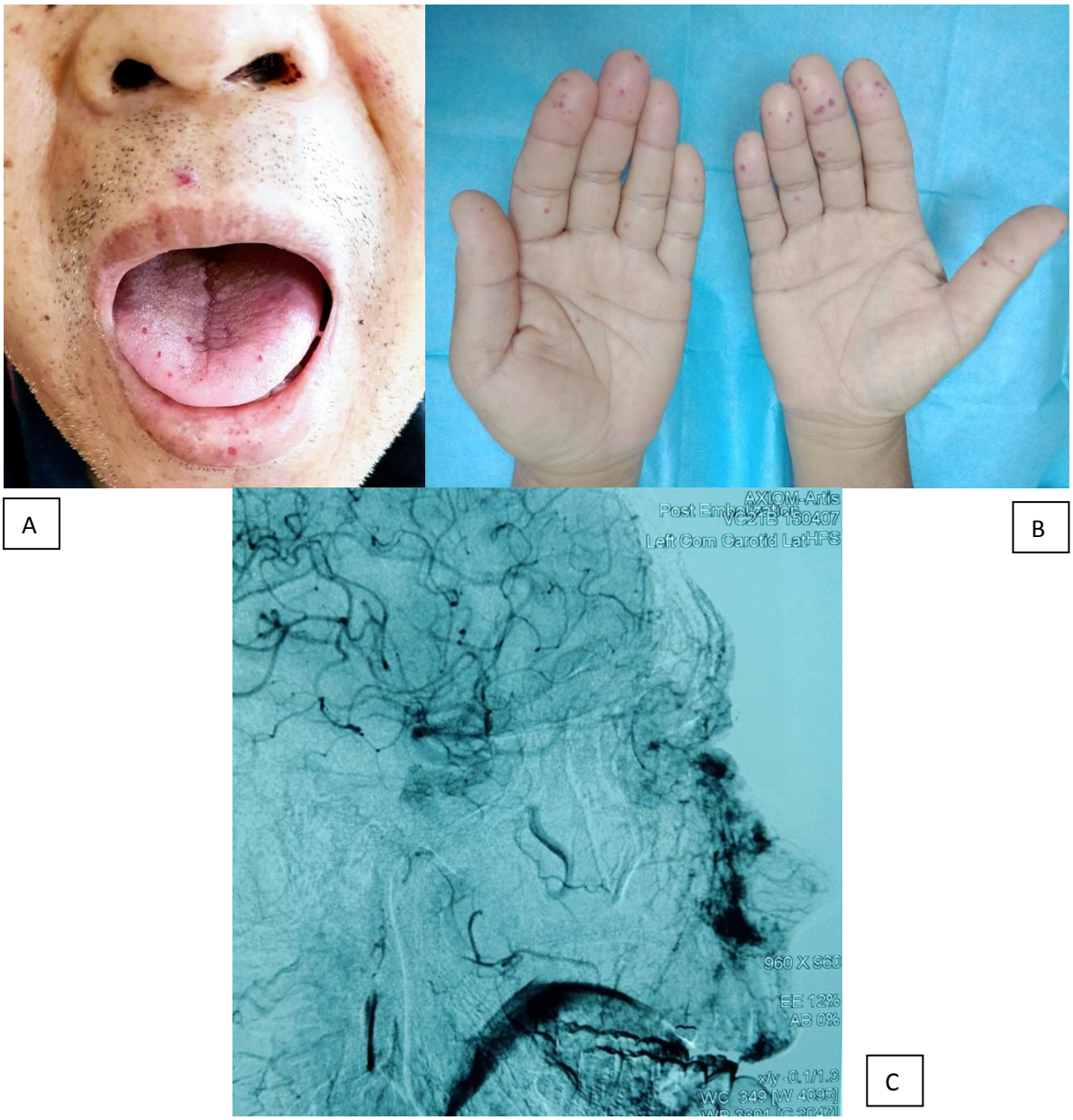


Figures: A) Telangiectasias on the tongue, lip and peri-oral region, B) palmer aspects of the hands and C) post embolisation image showing contrast blush in the nasal, upper lip and hard palate areas.

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 A), face and scalp, digits and the palmer aspect of the hands (Figure B). He also has numerous telangiectasia in the nasal passage. Patient recounted that bleeding can be severe; profuse epistaxis and bleeding from finger tips and scalp with spurting that can reach the ceiling. Upper gastrointestinal endoscopy showed several telangiectasia 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 cauterisation. Embolization of the left internal maxillary artery had to be done (Figure C) 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; a) spontaneous recurrent epistaxis, b) mucocutaneous telangiectasia, c) AVMs of visceral organs, and d) first degree relatives with similar condition. The diagnosis is definite if ≥3 criteria and possible if only 2 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 embolisation. (2) Treatment consist of iron replacement, transfusions and ablative therapies for the telangiectasia. (3) Thalidomide (anti-angiogenesis) can be tried but recently anti-vascular endothelial growth factor (anti-VEGF) antibody (Bevacizumab) has been shown to be beneficial. (4)

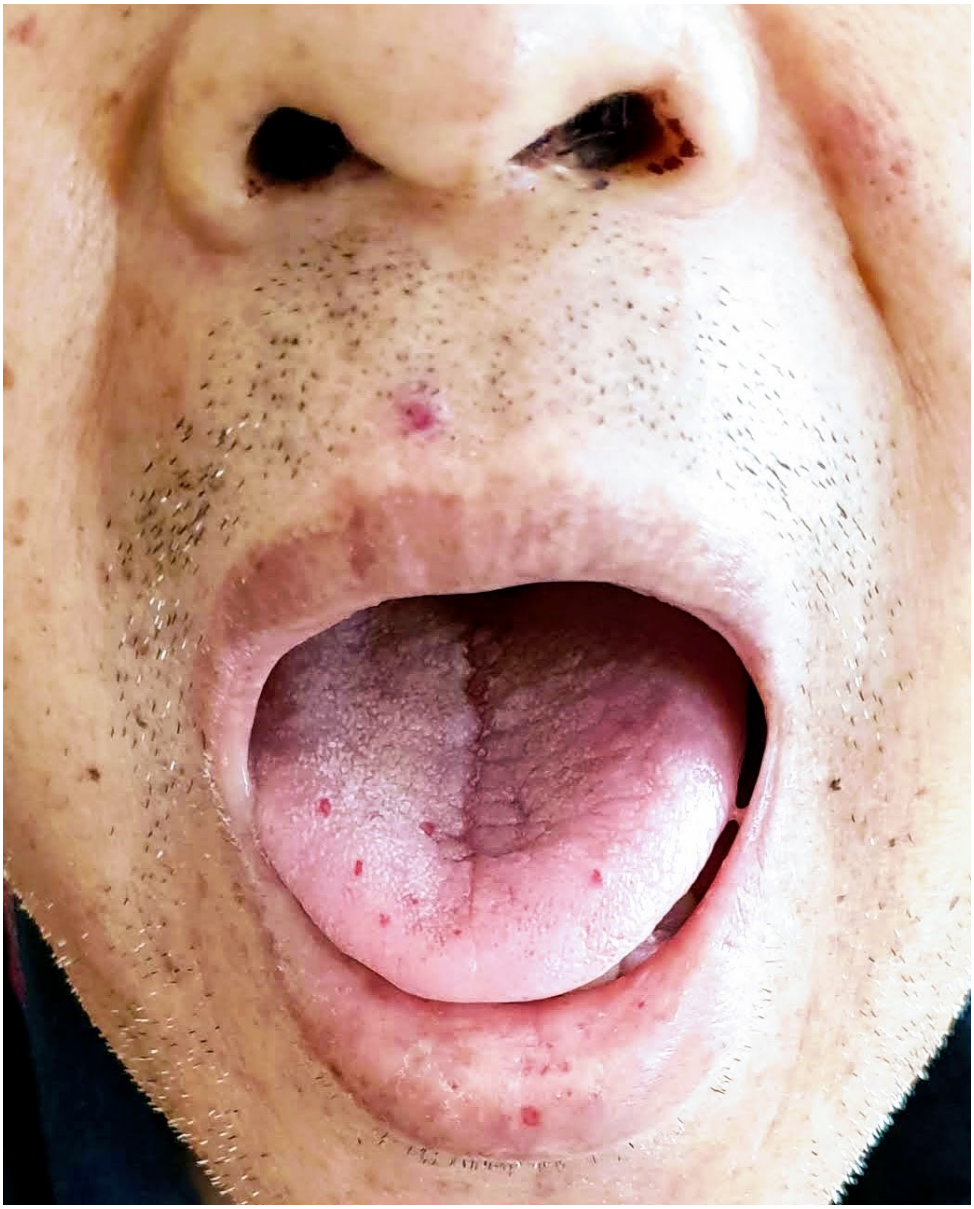
REFERENCES

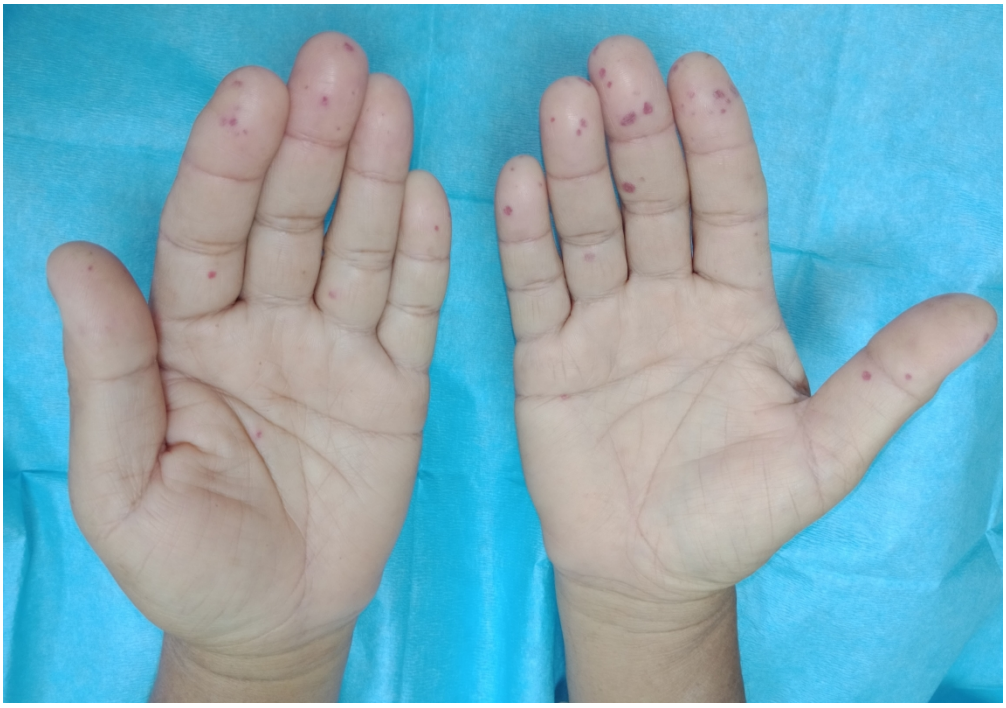
1: Shovlin CL, Guttmacher AE, Buscarini E, Faughnan ME, Hyland RH, Westermann CJ, et al. Diagnostic criteria for hereditary hemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). Am J Med Genet 2000; 91:66-7.

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.





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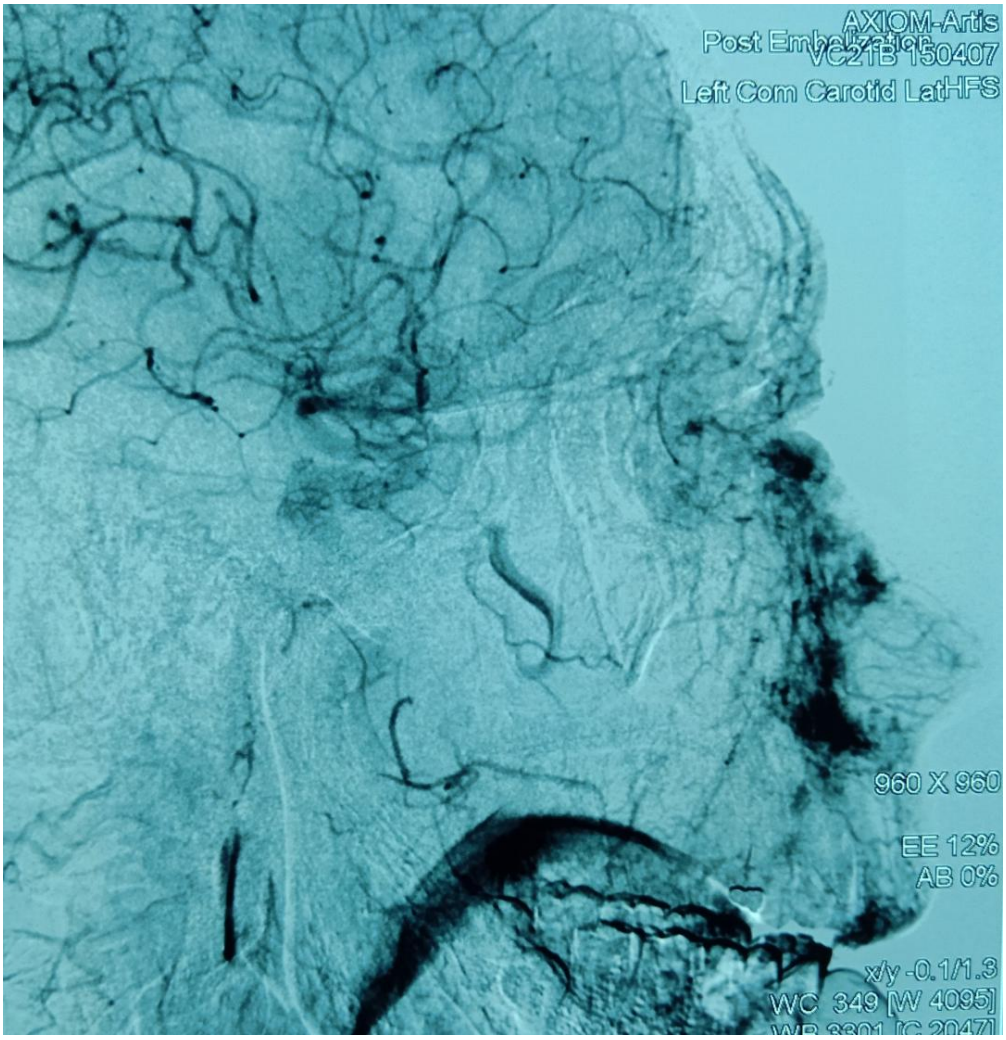

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