

Osler-Weber-Rendu syndrome

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DESCRIPTION

A 57-year-old woman presented to the emergency department reporting of fresh rectal bleeding since 3 days ago. She had a history of spontaneous recurrent epistaxis since childhood but had never been fully evaluated. Her family history was also notable for a son with recurrent spontaneous epistaxis. Physical examination revealed telangiectasia of the tongue (figure 1) and fingertips (figure 2). Laboratory findings were compatible with severe iron deficiency anaemia. Lower gastrointestinal endoscopy showed arteriovenous malformation and telangiectasia in the colon (figures 3 and 4). With a history of spontaneous and recurrent epistaxis, multiple mucocutaneous telangiectasias, positive family history and gastrointestinal arteriovenous malformation and telangiectasia a clinical diagnosis of hereditary haemorrhagic telangiectasia (Osler-Weber-Rendu syndrome) was made. Radiological evaluation revealed no vascular malformation in other viscera.

Hereditary haemorrhagic telangiectasia (HHT) is an uncommon autosomal dominant disease that occurs on mucocutaneous surfaces (ie, nose, gastrointestinal tract and skin), lung, liver and brain.¹ The Curaçao criteria was developed for the



Figure 1 The telangiectasia of the tongue.



Figure 2 The telangiectasia of the fingertips.



Figure 3 The arteriovenous malformation and telangiectasia in the colon.

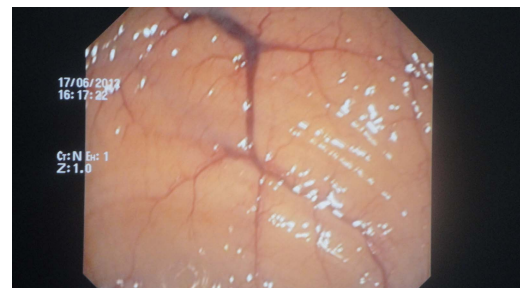


Figure 4 The arteriovenous malformation and telangiectasia in the colon.

diagnosis of HHT that contains epistaxis, family history, telangiectasias and visceral lesions.²

We were showed key recommendations with comments according to the Olitsky study for treatment of patient with HHT³ (Appendix).

Contributors The case was managed by GA and MR and read and approved the final manuscript. MR followed up the patient. MR and GA have.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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