Hereditary haemorrhagic telangiectasia

Pedro Magalhães-Costa, Cristina Chagas

DESCRIPTION
A 78-year-old Caucasian man was admitted for haematemesis and melena. At presentation, he was haemodynamically stable and laboratory showed an haemoglobin = 8.5 g/dL. On physical examination the only relevant findings were the presence of multiple, dot-sized (1–2 mm), small, red macular telangiectasias (figure 1) distributed in the lips, tongue and fingertips. Upper endoscopy revealed a gastric telangiectasia surrounded by an anaemic halo (figure 2), promptly managed with argon-plasma termocoagulation (35W; 1L). After 3 days

Figure 1 Multiple, dot-sized (1–2 mm), small, red macular telangiectasias, distributed in the lips, tongue and fingertips.

Figure 2 Upper endoscopy revealed a gastric telangiectasia surrounded by an anemic halo (A), promptly managed with argon-plasma termocoagulation (B).
in the ward and no evidence of gastrointestinal bleeding the patient was discharged. Retrospectively the patient referred multiple and spontaneous episodes of epistaxis in the past. Based on the Curacao diagnostic criteria he was diagnosed with hereditary haemorrhagic telangiectasia (HHT). HHT also known by the eponym Osler-Weber-Rendu disease is an autosomal dominant disorder characterised by the presence of multiple arteriovenous malformations in the skin, mucous membranes and internal organs (lung, liver and brain). Telangiectasias are typically observed in the lips, tongue, oral mucosa and fingertips in 75–90% of the affected individuals. Although 75% of these patients may have gastric and/or small-bowel telangiectasias on endoscopic examination, only one-third will eventually suffer from a gastrointestinal bleeding episode, usually after 40 years of age.

Contributors PM-C wrote the manuscript. PM-C and CC treated and followed the patient. CC critically revised the manuscript.

Competing interests None.

Patient consent Obtained.

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REFERENCES

Learning points

▸ Hereditary haemorrhagic telangiectasia is a rare cause of gastrointestinal bleeding.
▸ On the combination of epistaxis, gastrointestinal bleeding and mucocutaneous telangiectasias (lips, oral mucosa and fingertips), physician’s need to consider the diagnosis of hereditary haemorrhagic telangiectasia.
▸ Additional arteriovenous malformations—pulmonary, hepatic and cerebral—may be present and require further assessment, as well as family screening.