Images in Medicine

Hereditary haemorrhagic telangiectasia

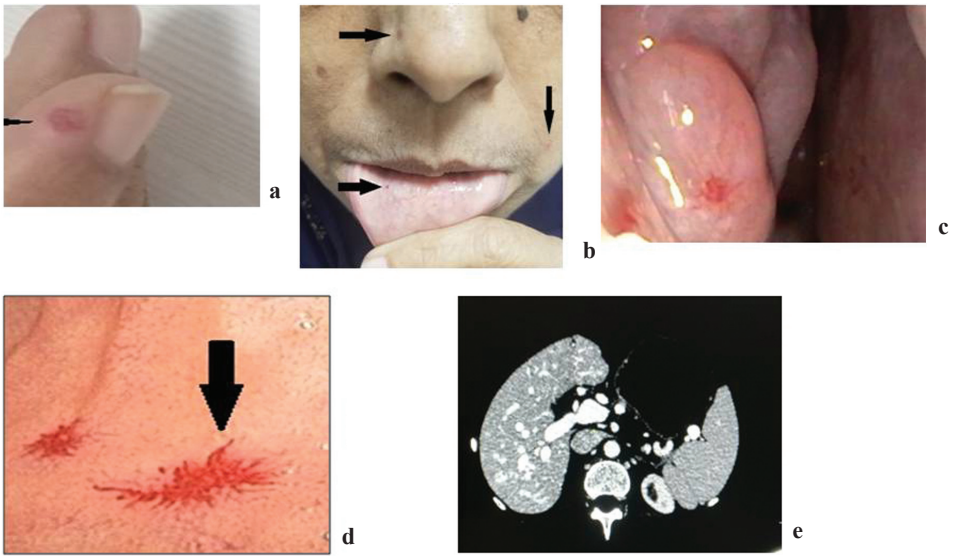

FIG 1. (a) Telangiectasia on the fingertip; (b) telangiectasia on buccal mucosa, cheek and nose; (c) telangiectasia in the nasal cavity; (d) duodenal telangiectasias; (e) computed tomography of liver showing venovenous malformations

Hereditary haemorrhagic telangiectasia (HHT), also known as Rendu–Osler–Weber disease, is an autosomal dominant disorder of the fibrovascular tissue.[1] It is characterized by the classical triad of mucocutaneous telangiectasia, arteriovenous malformations with recurrent epistaxis and haemorrhages.[2]

A 60-year-old female presented with a history of passage of intermittent black tarry stools in the preceding 10 years requiring multiple blood transfusions and intravenous iron preparations. There was a history of recurrent episodes of epistaxis since childhood requiring nasal packs. Her elder sibling and daughter had a similar history of recurrent epistaxis and transfusion-dependent anaemia. On examination, she was pale with punctuate tiny macules on the fingertips of both hands, tongue and lips (Fig. 1a and b). Nasal endoscopy showed multiple nasal angiodysplasias (Fig. 1c). Oesophagogastroduodenoscopy revealed multiple angiodysplasias in the stomach and the duodenum (Fig. 1d). Colonoscopy was normal. Capsule enteroscopy showed oozing of blood from angiodysplastic lesions in the jejunum. Rest of the small bowel was normal. Argon plasma coagulation was done for the bleeder using balloon enteroscopy. Computed tomography of the abdomen with an angiogram showed aneurysmal dilatation of the portal vein with multiple venovenous malformations (Fig. 1e). Two-dimensional echocardiogram showed moderate pulmonary arterial hypertension. Based on Curacao criteria,[3] the patient fulfilled the definitive criteria for the diagnosis of HHT, i.e. autosomal mode of inheritance, telangiectasia of the fingertips, spontaneous and recurrent epistaxis and gastrointestinal bleed. She was managed initially with lanreotide and later with thalidomide without much benefit. The patient is currently on symptom-based supportive care.

Conflicts of interest. Nil

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