This 63-year-old man is being investigated for a transfusion dependent anaemia.

1. What is the clinical sign demonstrated?
ANSWER

This is telangiectasia of the tongue and the palm. Due to hereditary haemorrhagic telangiectasia.

DISCUSSION

Hereditary haemorrhagic telangiectasia (HHT) or Osler-Rendu-Weber disease, is inherited as an autosomal dominant trait with varying degree of penetrance. Despite being a hereditary condition, HHT may not manifest clinically until the fourth or later decade of life. (2)

HHT is characterized by,

- mucocutaneous telangiectases
- arterio-venous malformations
- and be a possible cause of serious morbidity and mortality (1).

Spontaneous, recurrent, nose bleeds from telangiectasia of the nasal mucosa are the most common clinical manifestations of HHT (3). Arterio-venous malformations are known to occur in pulmonary, cerebral and hepatic circulation (3).

DIAGNOSIS

The diagnosis of HHT is made clinically on the basis of positive 3 out of the 4 Curaçao criteria (4).

- recurrent spontaneous epistaxis
- multiple mucocutaneous telangiectasias
- oral cavity, lips, fingers and nose
- arterio-venous malformations (AVMs) in lung, liver, brain, spinal cord, GI tract
- first degree relative with HHT

Genetic testing can be used to confirm a clinical diagnosis or to establish a diagnosis in suspected or asymptomatic patients. Majority of HHT cases are due to mutations of

- ACVRL1 (ALK1)
- ENG and
- SMAD4 genes (5)
TREATMENT

Treatment is mainly supportive.

- Hemostatic agents - tranexamic acid
- Frequent blood transfusions
- Iron supplementation.
- In visceral AVMs
  - Surgical resection
  - Embolisation
  - Endoscopic ablation
  - Transplantation - in large hepatic AVMs

Anti-oestrogen medications - Tamoxifen has been used with good results.

VEGF inhibitor (angiogenesis inhibitor) Bevacizumab (Avastin) had shown good response initially, the results were not positive in follow up trials. (6)

REFERENCES