

A Picture Paints a Thousand Words

Brian Christopher* and John Keohane

Department of Gastroenterology, Our Lady of Lourdes, Drogheda, Ireland

*Corresponding author: Brian Christopher, Dept of Gastroenterology, Our Lady of Lourdes, Drogheda, Ireland; E-mail: John.Keohane@hse.ie

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Description

A 56 year old woman was referred by her general practitioner with ongoing malaise. She had a history of recurrent epistaxis. On examination, there was extensive mucocutaneous telangiectasia (Figure 1). Her father and brother have a history of epistaxis and facial telangiectasia. Her presenting haemoglobin was 6.6 g/dl (normal, 13.0-18.0 g/dl), microcytic picture and ferritin of 3 ng/ml (normal, 22-274 ng/ml). She was transfused with packed red cells.



Figure 1(a): Mucocutaneous telangiectasias



Figure 1(b): Mucocutaneous telangiectasias

Push enteroscopy revealed multiple gastrointestinal telangiectasia. These were successfully coagulated with bipolar electrocoagulation (Gold Probe; Microvasive BostonScientific, Natick, MA) (Figure 2). Ileocolonoscopy was normal. She was discharged on oral iron. A diagnosis of hereditary hemorrhagic telangiectasia (HHT) [1-3] was made based on epistaxis history, family history and presence of mucocutaneous telangiectasias.



Figure 2: Small bowel angiodysplasia

HHT also known as Osler-Weber-Rendu syndrome is an autosomal dominant disorder. Patients may present with epistaxis or gastrointestinal bleeding with characteristic mucocutaneous telangiectasia on mucosal surfaces. Arteriovenous malformations (AVM's) can be present in lung (50%), hepatic (30%) and cerebral circulations (10%). Pulmonary AVM's may lead to paradoxical embolic stroke and brain abscess. Large hepatic AVM's may cause anaemia and cardiac failure. Cerebral AVMs are usually silent and multiple. Headaches and seizures may occur due to ischaemia or haemorrhage. The prevalence is between 1: 5000 and 1: 80003.

By age 16, 70% manifest signs rising to 90% at age 40. The international consensus diagnostic criteria (Curacao criteria) are based on findings below:-

- Spontaneous / recurrent epistaxis
- Multiple mucocutaneous telangiectasias
- Visceral involvement
- First degree relative with HHT

The diagnosis of HHT is definite when three or all of the above criteria are met. If two are met, the diagnosis is suspected. The treatment is individualised, supportive, and depending on the viscera affected. Mortality and morbidity is directly linked to the extent of visceral involvement.

References

1. Garcia-Tsao G, Korzenik JR, Young L, Henderson KJ, Jain D, et al. (2000) Liver disease in patients with hereditary hemorrhagic telangiectasia. *N Engl J Med* 343: 931-936.
2. Bharatha A, Faughnan ME, Kim H, Pourmohamad T, Krings T, et al. (2012) Brain arteriovenous malformation multiplicity predicts the diagnosis of hereditary hemorrhagic telangiectasia: quantitative assessment. *Stroke* 43: 72-78.
3. Govani FS, Shovlin CL (2009). Hereditary haemorrhagic telangiectasia: a clinical and scientific review. *European Journal of Human Genetics* 17 : 860-871.