



Hereditary Haemorrhagic Telangiectasia

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Received: Sep 10, 2026

Accepted: Sep 22, 2026

Published Online: Sep 29 2026

Journal: Journal of Clinical Images

Publisher: MedDocs Publishers LLC

Online edition: <http://meddocsonline.org/>

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Abstract

A 52-year-old woman presented with fatigue and iron-deficiency anemia (hemoglobin 8,1 g/dl, mean corpuscular volume 61 μm^3 , iron 13 $\mu\text{g}/\text{dl}$, ferritin 2 ng/ml) due to recurrent spontaneous epistaxis and a recent episode of gastrointestinal bleeding. Physical examination revealed multiple mucocutaneous telangiectasias most evident on the tongue (Panel A), buccal mucosa, chest and fingers (Panel B); a CT scan of the chest and abdomen revealed multiple arteriovenous malformations in the liver. The patient's mother and the brother had similar clinical manifestations. Based on the clinical picture and the family history, Hereditary Haemorrhagic Telangiectasia (HHT) was diagnosed according to Curaçao international diagnostic criteria [1,2]. A molecular genetic test revealed a missense mutation in the gene encoding activin receptor-like kinase ALK-1 in the patient and the family, confirming the diagnosis of HHT (Osler-Weber-Rendu syndrome). The patient was treated with laser photocoagulation of nasal telangiectasias and iron infusions, with relevant improvement of the symptoms.

Although HHT is best managed in specialist centres with a multidisciplinary approach, it requires local coordinate care for the timely treatment of life-threatening clinical manifestations.



Figure 1



Figure 2



References

1. ME Begbie, GMF Wallace, CL Shovlin. Hereditary haemorrhagic telangiectasia (Osler-Weber-Rendu syndrome): a view from the 21st century. *Postgrad Med J.* 2003; 79: 18-24.
2. CL Shovlin, AE Guttmacher, E Buscarini, et al. Diagnostic criteria for hereditary haemorrhagic telangiectasia (Rendu-Osler-Weber syndrome). *Am J Med Genet.* 2000; 91: 66-7.