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Hereditary Haemorrhagic Telangiectasia

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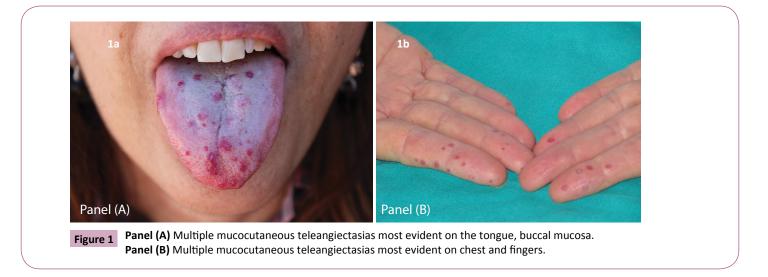
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Clinical Image

Multiple mucocutaneous teleangiectasias is most evident on the tongue (Figure 1, Panel A), buccal mucosa, chest and fingers (Figure 1, Panel B) in fatigue and iron-deficiency anemia, due to recurrent spontaneous epistaxis and a recent episode of gastrointestinal bleeding.

The clinical picture and the family history suggest the diagnosis of Hereditary Haemorragic Teleangiectasia (HHT). A missense mutation in the gene coding for the Activin receptor-like kinase ALK-1 was detected and confirmed the diagnosis of HHT. Treatment with laser photocoagulation of nasal teleangiectasias and iron infusions was found to be relevant with improvement of the symptoms.