Pathophysiology, diagnosis and management of hereditary hemorrhagic telangiectasia (HHT)

Physiological signaling pathway

- TGFβ
- TGFβRII
- ALK5
- BMP9
- Endoglin
- SMAD 2/3
- SMAD 1/5/8
- Complex SMAD4
- Endothelial cell proliferation
- Vascular smooth muscle cell migration
- VEGF production

Pathophysiological signaling pathway in HHT

- TGFβ
- TGFβRII
- ALK5
- ALK1
- BMP9
- Mutations
- SMAD 2/3
- SMAD 1/5/8
- Complex SMAD4
- VEGF production
- Endothelial cell proliferation
- Vascular smooth muscle cell migration
- Arteriovenous malformations
- Talangiectasia
- Manifestations of HHT

CLINICAL MANIFESTATIONS
- Mucocutaneous telangiectasias
- Arteriovenous malformations
- Epistaxis
- GI bleeding
- Iron deficiency
- Iron deficiency anemia
- Ischemic and hemorrhagic stroke
- Brain abscess
- Heart failure
- Liver failure

DIAGNOSTIC CRITERIA (Curaçao criteria)
- Spontaneous and recurrent epistaxis
- Telangiectasias at characteristic sites
- Visceral arteriovenous malformations or telangiectasias
- A first degree relative with HHT

Kritharis et al., Haematologica, 2018