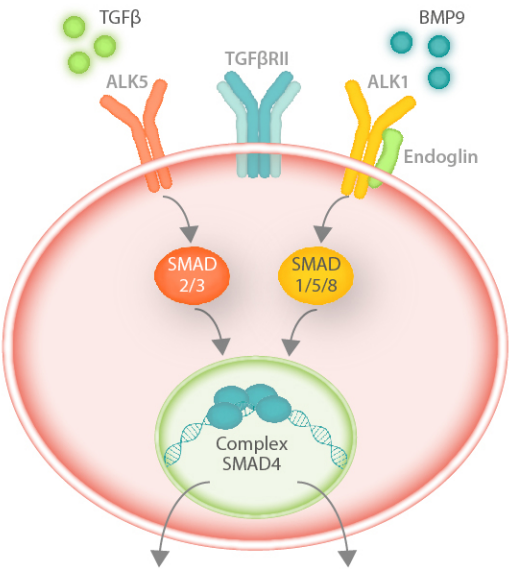
Pathophysiology, diagnosis and management of hereditary hemorrhagic telangiectasia (HHT)

Physiological signaling pathway



- ↓ Endothelial cell proliferation
- ↓ Vascular smooth muscle cell migration
- ↑ VEGF production

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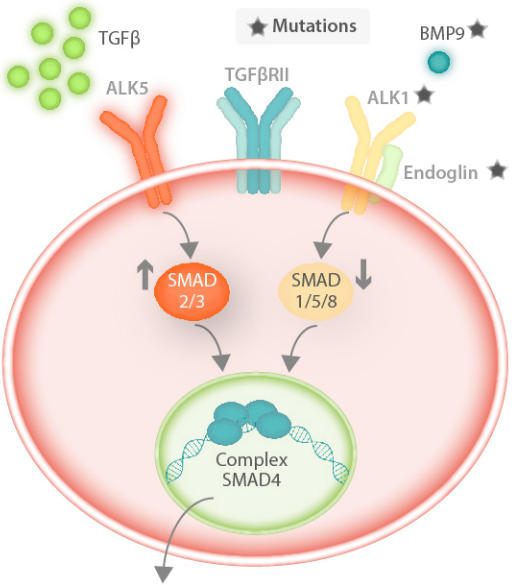
Pathophysiological signaling pathway in HHT



- ↑ VEGF production
- ↑ Endothelial cell proliferation

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- Arteriovenous malformations
- Talengiectasia
- 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