

Hereditary Hemorrhagic Telangiectasia



PLAY PICMONIC

Pathophysiology

Autosomal Dominant

Domino

Hereditary hemorrhagic telangiectasia is inherited in an autosomal dominant modality. This means one copy of the abnormal gene must be present in order for the disease to develop.

Mutation in TGF-beta

Mutant Transformer-giant-flagger-Beta-fish

Hereditary hemorrhagic telangiectasia involves the mutation of various genes that code for TGF-beta (e.g., ALK-1). These mutations then lead to structural defects in the vessel walls, which contribute to venous pooling and the development of small or large arteriovenous shunts.

Clinical Features

Nosebleed (Epistaxis)

Nose-bleeding

The clinical features of hereditary hemorrhagic telangiectasia can involve recurring epistaxis, telangiectasia of the mucosa and skin, GI bleeding, hematuria, and the formation of AVMs.

Telangiectasias

Tarantula-taser

The clinical features of hereditary hemorrhagic telangiectasia can involve recurring epistaxis, telangiectasia of the mucosa and skin, GI bleeding, hematuria, and the formation of AVMs.

GI Bleeding

GI-guy Bleeding

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Hematuria

Red-urinal

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AVM Formation

A Tangle of Blood Vessels

The clinical features of hereditary hemorrhagic telangiectasia can involve recurring epistaxis, telangiectasia of the mucosa and skin, GI bleeding, hematuria, and the formation of AVMs. The formation of AVMs (e.g., liver, lung, brain) results in an abnormal connection of a high-flow nature between an artery and a vein.

Diagnosis

Curaçao Criteria

Blue Curaçao

The diagnosis of hereditary hemorrhagic telangiectasia involves the Curaçao criteria. This criterion encompasses nosebleeds (epistaxis), telangiectasias, any signs of visceral involvement, and/or a family history.

Genetic Testing

Genes and Test-tubes

Hereditary hemorrhagic telangiectasia can be diagnosed either through specific criteria or through genetic testing.

Complication

High Output Heart Failure

High Output Dead Heart

A complication of hereditary hemorrhagic telangiectasia involves high-output cardiac failure. Other potential complications include anemia due to chronic GI bleeding or hematuria and a brain abscess or stroke due to AV shunt development bypassing normal lung functioning, thereby allowing emboli to enter arterial circulation.