Hereditary hemorrhagic telangiectasia

Genetics

-ACVRL1 (25-57%), ENG (39-59%), GDF2, SMAD4 (1-2%)

-AD; mostly inherited

Clinical findings:

-Epistaxis (nosebleeds), spontaneous and recurrent (95%)

-Mucocutaneous telangiectasias (small blanchable red spots at characteristic sites, including lips, oral cavity, fingers, and nose) (80%)

-Arteriovenous visceral malformation (arteriovenous malformation lacks capillaries and consists of direct connections between arteries and veins)

-Hemorrhage is often the presenting symptom of cerebral AVM; exercise intolerance

Etiology

-Overall incidence in North America is estimated at 1:10,000

-Elevated risk for DVT

Pathogenesis

-Haploinsufficiency

Genetic testing/diagnosis

-Diagnosis: >3 of the following clinical features: epistaxis (nose-bleeds), mucocutaneous telangiectases, visceral AVMs, and/or a family history of HHT

-Serial single-gene testing or gene panel

Others

-Clinical: contrast echo for pulmonary AVM, head MRI for cerebral AVM, US for hepatic AVM

-Liver transplant if hepatic AVM is causing heart failure