Condition: Hereditary hemorrhagic telangiectasia (Osler-Rendu-Weber syndrome)

Inheritance:

Autosomal dominant.

Genetic etiology:

Mutation either in *ACVRL1* gene encoding serine/threonine-protein kinase receptor R3, *ENG* encoding endoglin, or SMAD4. Most mutations result in loss of function alleles.

Frequency:

Approximately 1/10,000.

Clinical features:

Affected individuals develop multiple arteriovenous malformations on skin, mucous membranes, and in deeper tissues. Clinically this may present with nose bleeds or GI bleeding. Telangiectasias are visible on the skin and mucous membranes. AV fistulas can occur in the lung and lead hypoxemia, or to stroke or brain abscess.

Management:

Monitoring for major complications, especially pulmonary AVMs; supportive care.

Genetic counseling:

Transmitted as dominant traits; genetic testing is available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.