Condition: Von Hippel-Lindau syndrome

Inheritance:
Autosomal Dominant**.**

Genetic etiology:
Mutation in *VHL* tumor suppressor gene.

Frequency:
Approximately 1/36,000.

Clinical features:
Individuals with von Hippel-Lindau syndrome develop hemangioblastomas of the brain, spinal cord, and retina. They may also develop renal cysts with risk of renal cell carcinoma, as well as cysts of the pancreas and epididymis, pheochromocytoma, and endolymphatic sac tumors (which cause deafness).

Management:
Screening for associated tumors and symptomatic treatment.

Genetic counseling:
Affected individuals at 50% risk of transmitting a gene mutation; both familial and de novo cases occur. Genetic testing is available.

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