Cleveland Clinic
Genomic Medicine Institute

Von Hippel Lindau Syndrome (*VHL* gene)

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VHL is a condition characterized by abnormal growth of blood vessels in various parts of the body. The most common symptoms include hemangioblastomas (tumors of blood vessels) of the brain, spinal cord, and retina, as well as kidney cysts and kidney cancer. Some individuals with VHL develop pheochromocytomas, tumors in the inner ear, pancreatic cysts, and pancreatic tumors.

The *VHL* gene is passed on in families by autosomal dominant transmission, whereby each child of a carrier has a 50% (1 in 2) chance of inheriting the abnormal copy from the carrier parent. In some instances, an individual may be the first in their family to have a *VHL* mutation. This is referred to as a *de novo* mutation. Twenty percent of individuals with *VHL* have a *de novo* mutation.

The Center for Personalized Genetic Healthcare