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.