Mass General Brigham
Mass General Cancer Center

VHL gene: what you need to know

What does it mean to have a *VHL* pathogenic variant?

Having a *VHL* pathogenic variant, also known as a mutation, means a person has a version of the *VHL* gene that does not work properly. This causes a condition known as **von Hippel-Lindau (VHL) disease**.

Do I have an increased risk for cancer if I have VHL?

If you have a VHL, you have a greater risk of developing certain types of benign tumors, as well as certain types of malignant tumors. A person with VHL has nearly 100% chance of developing one or more VHL tumors in their lifetime.

Benign tumors are not cancerous and do not spread, whereas malignant tumors are cancerous and can spread (metastasize) to nearby healthy tissue and organs. Malignant tumors have the potential to spread to more distant sites of the body. Benign and malignant tumors are often treated in different ways.

What type of benign tumors am I more likely to develop if I have VHL?

- Benign tumors on the retina (back of the eye), brain, or spine called hemangioblastomas.
- Benign tumors located in the inner ear called endolymphatic sac tumors.
- Benign tumors in the scrotum or near the fallopian tubes.
- A specific type of adrenal gland tumor seen in VHL, called a pheochromocytoma, is usually benign. Pheochromocytomas are rarely malignant in individuals with VHL (~3%).

What type of malignant tumors am I more likely to develop if I have VHL?

- Malignant tumors of the kidney called renal cell carcinoma (kidney cancer).
- Pancreatic neuroendocrine tumors, which can be malignant.

Is there anything else that I will be at risk for if I have VHL?

People with VHL can develop benign fluid-filled sacs called cysts in the kidneys and pancreas.

If I have VHL, what is the chance that my family members will have VHL?

There is a 50% chance that a person with a pathogenic variant in *VHL* will pass it on to each of their children. In most cases, siblings of a person with a pathogenic variant have a 50% chance to have the pathogenic variant. Additionally, other family members are at risk to have the pathogenic variant.