



## **VHL gene: What You Need to Know**

### **What does it mean to test positive for a VHL gene mutation?**

Mutations in the *VHL* gene cause a condition known as von Hippel-Lindau (VHL) disease.

### **Do I have an increased risk for cancer if I have a VHL gene mutation?**

If you have a *VHL* gene mutation, you have a greater risk of developing 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 invade nearby healthy tissue and organs. Cancerous cells have the potential to spread, or metastasize, to other sites of the body. Benign and malignant tumors are often treated in different ways.

### **What type of benign tumors am I more likely to get if I have a VHL gene mutation?**

- 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 in men and near the fallopian tubes in women.
- 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 cancer am I more likely to get if I have a VHL gene mutation?**

Individuals with VHL have an increased risk for developing malignant cancers of the kidney and pancreas.

### **Is there anything else that I will be at risk for if I have a VHL gene mutation?**

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

### **What is the chance that my family members will have a VHL mutation if I test positive?**

There is a 50% chance that a person with a mutation will pass it on to each of his/her children. In most cases, brothers and sisters of a person with a mutation have a 50% chance to have the mutation. Additionally, other family members are at risk to have the mutation.