

Von Hippel-Lindau Syndrome

Von Hippel-Lindau syndrome (VHL) is a rare genetic disorder. Having VHL increases the risk of developing several types of cysts and tumors throughout one's life. Many of the tumors that happen in VHL are benign (non-cancerous) but some can be malignant (cancerous). VHL is hereditary, meaning that it can be passed to an individual from their parents. Each child of a parent with VHL has a 50%, or 1 in 2 chance of inheriting the VHL gene mutation.

VHL can affect several organs in the body. It affects people differently, even between members of the same family. However if you have VHL, you will almost certainly develop at least one type of tumor. Most persons with VHL will develop multiple tumors. VHL most commonly causes cysts or tumors in these areas:

- Retinae (inner part of the eye)
- Brain
- Spinal cord
- Kidney
- Endocrine pancreas
- Inner ear
- Adrenal gland

Causes of VHL

VHL is caused by a genetic change, called a mutation, in the *VHL* gene. Genes are the set of instructions that tell all of the cells in our bodies what to do. We have two copies of our genes. Our mother gives us one copy of genes and our father gives us one copy. Sometimes, a change or mutation happens in a gene that causes the gene to stop working properly. When it is working properly, the *VHL* gene controls cell growth and helps prevent tumors from forming. In the case of VHL, a mutation in the *VHL* gene causes the gene to stop working properly. This genetic change can be passed to sons and daughters from their mother or father.

Symptoms of VHL

Symptoms of VHL can arise at any age and are dependent on the type of tumor someone has. Some tumors may cause problems such as:

- Vision loss or impairment
- Headaches and/or changes in balance or strength
- Changes in blood pressure (hypertension)

Other tumors may not cause obvious symptoms. The age when symptoms first appear and the type of symptoms may be very different among individuals and their relatives. It is very important for people with VHL to talk with their health care providers about any changes to their health, as these could be early signs of a tumor. The goal of regular screening and check-ups for people with VHL is to find tumors early before they cause many medical problems.

Early Detection, Testing and Screening for VHL

Early detection can prevent, delay or aid in treating serious symptoms caused by VHL. Early detection measures include genetic testing and annual screening exams.

Genetic Testing for VHL

Genetic testing, which is often done with a blood sample, is very important to help diagnose VHL. If a mutation in the *VHL* gene is found, then the VHL diagnosis can be confirmed. Next, other family members may have the same genetic test to learn whether or not they carry the same gene mutation and have VHL. This helps identify who in the family needs annual screening for VHL and who does not.

Annual Screening

Annual screening tests are recommended for persons with VHL. These tests may include physical exams, blood tests and imaging. How often these tests are performed will be different depending on each patient's age, family history, sex and personal medical history. It may also differ depending on the type of *VHL* gene mutation someone has. A team of specialists work with families to develop a regular screening plan and to review the results.

Careful screening can result in the earlier detection of the tumors and cysts that happen in VHL. This can help with earlier and more effective treatment while minimizing the medical issues associated with the tumors and cysts.

Treatment

Currently, there is no cure for VHL and the tumors cannot be prevented. However, early diagnosis and treatment of symptoms can greatly improve quality of life. The type of treatment your medical team recommends will depend on many factors. The recommended treatment for the same type of tumor could be different between family members. Currently, a combination of surgery, medicine, radiation treatments and/or laser therapies may be used to treat tumors.

Your care team may include:

- Ophthalmologists (eye doctors)
- Neurosurgeons (doctors who specialize in brain and spine surgery)
- Oncologists (cancer doctors)
- Urologists (doctors who specialize in care of the kidneys and urinary tract)
- Endocrinologists (doctors who specialize in the care of the glands in the endocrine system)

Children with VHL

Children with VHL have special requirements that should be addressed by a pediatrician familiar with VHL. The pediatrician is an important team member who works with other specialists to test, screen and treat children living with VHL.

The VHL Clinical Center at MD Anderson Cancer Center

The VHL Clinical Center's mission is to provide patients compassionate care, delivered by expert doctors and counselors. Patients receive a plan of care developed to meet their specific needs. Afterwards, patients have screening, treatment and follow-up visits needed for short-term and long-term care.

The VHL Clinical Center is uniquely qualified to care for patients living with VHL. The health care team includes experts in many different cancer specialties who are trained to care for and treat patients (both adults and children) living with VHL. For more information about the VHL Clinical Center, contact a genetic counselor at 713-745-7391.