

Von Hippel-Lindau Syndrome

Von Hippel-Lindau syndrome (VHL) is a rare genetic condition that leads to an increased risk of developing certain types of cysts and tumors. The tumors can be benign (non-cancerous) or malignant (cancerous). Many of these tumors may begin developing in childhood or early adulthood.

VHL most often causes cysts or tumors in these areas:

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

VHL is caused by a change (mutation) in the VHL gene. A gene change causes the gene to stop working normally. The VHL gene normally works to protect the body from forming tumors. Mutations in the VHL gene cause the increased risk of developing tumors.

Genetic conditions can be passed down in families. Parents can pass VHL to their children. Each child of a parent with VHL has a 1 in 2 chance of having the VHL gene mutation.

Symptoms

VHL can affect several organs in the body. It affects people differently. Even members of the same family may have different symptoms. Most people with VHL will develop at least 1 type of tumor. Many people will develop multiple tumors.

Symptoms of VHL can start at any age. Symptoms depend on the type of tumor that develops. Some tumors may cause problems such as:

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

Some tumors may not cause clear symptoms. Changes in health could be early signs of a tumor. Tell your doctor about any changes in your health right away. Tumors are often easier to treat when they are found early.

Genetic Testing

Genetic testing will test your blood or saliva to look for mutations in the VHL gene. Genetic testing helps to diagnose VHL.

VHL gene mutations are usually passed down in families. Family members may benefit from genetic testing when a person in the family has VHL. Genetic testing can help to find out if other family members have VHL. This can help doctors decide which family members should consider additional screenings and check-ups.

Early Detection and Screening

Screening helps find tumors and cysts as early as possible. Early detection can prevent, delay or help in treating serious symptoms caused by VHL. Regular screening tests and check-ups are key to early detection.

Screening tests may include physical exams, blood tests and imaging. Your care team will help you develop a screening plan. Your screening plan will be tailored to your specific needs. Factors that may affect your screening plan include your age, family history, sex, personal medical history, and type of VHL gene mutation.

A team of specialists will help your family to develop a regular screening plan.

Treatment

Early diagnosis and treatment of symptoms can greatly improve your quality of life. The type of treatment your care team recommends will depend on many factors. It is possible that doctors may recommend different treatments for family members with the same type of tumor.

Current treatments for tumors include a combination of surgery, medicine, radiation treatments or laser therapies. A new medicine is available for people with VHL with specific tumors, such as kidney cancer or a pancreatic neuroendocrine tumor. You and your care team will discuss the best treatment options for you.

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 need specific medical care and should see a pediatrician (doctor who specializes in the care of children) who is familiar with VHL. The pediatrician will work with

other specialists to test, screen and treat the child with VHL.

The VHL Clinical Center at MD Anderson Cancer Center

At the VHL Clinical Center, expert doctors and genetic counselors provide compassionate and specialized care. The care team includes experts in many different cancer areas that are uniquely trained to care for adults and children with VHL.

Patients receive care plans that meet their specific health needs. The care team supports patients through screening, treatment and short-term and long-term care follow-up visits.

For more information about the VHL Clinical Center, contact a genetic counselor at 713-745-7391.

Resources

VHL Alliance

www.vhl.org/

VHL Alliance works to improve the quality of life and health outcomes for VHL patients, families, and caregivers with education, treatment options and advancements in medical research.