

## Von Hippel-Lindau Syndrome

Von Hippel-Lindau Disease (VHL) is a rare disorder that is caused by changes (mutations) in the VHL gene. 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 percent (%), or 1 in 2, chance of inheriting the VHL gene mutation.

VHL can affect several organs of the body and affects every patient differently. VHL can cause cysts or tumors of the:

- Retina (inner part of the eye)
- Brain
- Spinal cord
- Kidney
- Pancreas
- Inner ear

Patients may also have pheochromocytomas (usually benign tumors of the adrenal gland).

### Symptoms

Symptoms can arise at any age. The age when symptoms first appear and the type of symptoms may be very different among individuals and families. VHL can cause problems with vision at any age, even in children under the age of five. Other symptoms of VHL may include headaches and changes in balance and strength.

Patients may also experience changes in blood pressure and heat flashes if a pheochromocytoma is present.

### 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

Genetic testing of a blood sample can help diagnose VHL and can help determine if other family members have VHL.

#### Annual Screening

Annual screening tests are recommended for individuals with VHL. These tests may include physical exams, blood tests and scans. How often these tests are taken may be different depending on each patient's age, family history and medical history. A team of specialists work

with families to develop a screening plan and review the results.

Careful screening can result in the earlier detection of the tumors and cysts that can occur with VHL, allowing earlier and more effective treatment.

## **Treatment for VHL**

Currently, there is no cure for VHL. However, early diagnosis and treatment of the symptoms caused by VHL can greatly improve a patient's quality of life.

### **Retina**

Ophthalmologists (doctors specializing in treating the eyes) use laser therapy and other treatments to decrease the chance of vision loss caused by VHL.

### **Brain and Spinal Cord**

Neurosurgeons (doctors specializing in brain and spinal cord surgery) and radiation treatment specialists care for and treat brain and spinal cord tumors. Careful planning and treatment can prevent or minimize the loss of coordination or mobility.

### **Kidney**

Oncologists and urologists (doctors who care for organs of the urinary tract) treat kidney tumors. Treatment for kidney tumors is designed to keep as much kidney function as possible. One form of treatment is minimally invasive surgery.

### **Pancreas and Adrenal Gland**

Surgical and medical endocrinology experts (doctors who treat glands and hormones in the body) work together to manage and treat patients with pancreatic and adrenal symptoms. The health care team may prescribe medicine for the patient to take before and after surgery.

## **Children with VHL**

Children with VHL have special requirements that should be addressed by a pediatrician familiar with inherited disorders. The pediatrician plays an important role in the team of specialists who work together to test, screen and treat children living with VHL.

## **The VHL Clinical Center at M. D. Anderson Cancer Center**

The VHL Clinical Center's mission is to provide patients compassionate care, delivered by expert physicians 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 living with VHL. For more information about the VHL Clinical Center, contact a genetic counselor at 713-745-7391.