

## Family Testing for von Hippel-Lindau Disease: Information for Patients and their Families

If a genetic variant (mutation) that is associated with von Hippel-Lindau Disease has been detected in you or in one of your blood relatives, please read this document and share it with other members of your family (you can download copies from <http://www.correlagen.com/fields/other/other.jsp>). This document explains how family testing for von Hippel Lindau Disease can benefit your extended family.

### Why is family testing for von Hippel-Lindau Disease important?

Von Hippel-Lindau Disease (VHL) is a dominantly inherited disease. That means that the parents, children, and siblings of a patient with VHL are at a 50% risk of also being affected. In other words, on average half of all siblings and children (and one parent) of a patient with VHL are also affected with VHL.

### How can genetic testing be used for family testing?

VHL can be caused by a single mutation in the gene *VHL*. Detecting a disease-causing mutation in the gene *VHL* therefore allows diagnosis of VHL. This type of diagnostic test is referred to as “genetic testing.” Disease-causing mutations can occur in any part of the gene *VHL*, but the same mutation – called the “familial mutation” – typically accounts for all of disease within one extended family. Therefore, the entire gene region is screened in the first family member who undergoes genetic testing. This first-to-be-tested family member is commonly called the “index patient” or the “proband” for a family. In blood-relatives of the index patient, only gene regions known to contain the “familial mutation” have to be tested. This limited genetic testing is much more straightforward and much less costly than the initial full-gene testing.

### Why is genetic testing better than other diagnostic methods for family testing?

Genetic testing is often a much better tool than diagnosis based on symptoms to identify affected and unaffected members of a family with a dominantly inherited disease. For example, a family member without any symptoms of the disease may be unaffected, but he or she may also be presymptomatic – that is, he or she may simply not have developed symptoms of the disease yet. Sometimes, symptoms may be present, but it may be unclear if they are due to the genetic disease or due to unrelated reasons. Genetic testing can determine if a family member is unaffected, presymptomatic, or affected by screening for the presence of the familial mutation. If the familial mutation is not present, the family member is unaffected. If the familial mutation is present, the family member is presymptomatic or affected. Unaffected family members no longer have to worry about developing the disease or passing it on to their children. Presymptomatic or affected family members know that they have to be vigilant for symptoms of the disease and seek treatment as soon as symptoms appear. If treatment is started early, the outcome for the patient is often much better.

### Where can I get genetic testing?

Genetic testing for VHL should be ordered by a physician. That means you should make an appointment with your doctor to discuss genetic testing for VHL. Since genetic testing is a relatively new diagnostic method that many physicians may not use routinely, we recommend that you take this flyer with you when you see your doctor. Before you see your doctor, you may also want to visit [www.correlagen.com](http://www.correlagen.com) for more information on genetic testing in general, for more information on VHL and the benefits of genetic testing for VHL, and for links to other websites with information on VHL and genetic testing.

### Is genetic testing a complicated procedure?

Fortunately, getting a genetic test is not complicated at all. Genetic testing is typically performed on a small blood sample or a cheek swab. For a cheek swab, a cotton-tipped applicator is rubbed against the inside of the cheek – an easy and painless procedure. Please remember to tell your doctor exactly what the familial mutation is in your family, so that he or she can specify it when ordering the genetic testing. If you don't know what the mutation is, please ask the index patient in your family.

**What is von Hippel-Lindau Disease?**

Von Hippel-Lindau Disease (VHL) belongs to the familial cancer syndromes, which are a group of diseases characterized by a very high lifetime risk of developing certain types of tumors or cancers. Tumors characteristic for VHL can occur in the eyes, the adrenals, the kidneys, and many other tissues. Once VHL has been diagnosed, annual tumor screening can help to detect tumors before they become symptomatic and allow early treatment. Early diagnosis of VHL has been shown to increase survival of patients with VHL-associated kidney cancer, one of the leading causes of death from VHL. Similarly, timely treatment of VHL-associated tumors of the eye can prevent vision loss and blindness. The key to treatment success is to start therapy early. That's why family testing for VHL, allowing identification of presymptomatic family members, is so important.

Please visit <http://www.correlagen.com/fields/other/other.jsp> for more information on VHL.

