Genetic Testing and VHL

This document discusses how you can learn about your own genetic information. For more information on the genetics of VHL, please see “Genetics 101.”

The only way to confirm a clinical diagnosis of VHL is through genetic testing. The first step is to meet with a genetic counselor or other genetics provider to discuss your options for genetic testing. It is important to meet with them as this is a clinical test that could have medical and emotional implications for you and your family members.

Who are Genetic Counselors and How do I Find One?

Genetic counselors are healthcare professionals who have training in both medical genetics and counseling. They help guide and support people seeking more information about inherited diseases, genetic testing, and the impact of genetic test results on their health and their family. Genetic counselors meet with people with VHL, as well as people concerned about their family history of VHL. Ask your doctor if they are connected with a department of “cancer genetics,” or look online to locate a genetic counselor with whom you can meet in-person or by phone: http://www.nsgc.org/findageneticcounselor.

What Happens at a Genetic Counseling Visit?

- Despite living with continual uncertainty, many participants seemed to maintain a positive attitude. Several participants positively framed their VHL experiences while others exhibited perseverance. A genetic counselor asks questions about your medical history, including any cancer diagnoses and cancer screening.

- The genetic counselor takes a detailed family history and draws out a three-generation pedigree. In advance of a genetic counseling appointment, it is important to obtain the following information about your immediate and extended family:
  - Ethnicity and/or family origins
  - Ages and causes of death
  - Known genetic conditions
  - Genetic testing – bring a copy of any family member’s genetic test results to the visit
  - Health history including cancer diagnoses, tumor growth, vision loss, hearing loss, pregnancy history, mental health, and any other significant health problems
    - Age of onset, diagnosis, and treatment

- They do a risk assessment based on medical and family history information and may discuss options for genetic testing. Genetic testing is a personal decision and is always optional. The genetic counselor can discuss the potential benefits and limitations of genetic testing and answer questions that are bound to arise now or in the future.
• If you are interested, a genetic counselor can facilitate the genetic testing process, including working with you to choose a clinical testing laboratory, filling out paperwork, and coordinating a blood draw or saliva sample. They can communicate directly with the clinical testing laboratory and work with you on billing or insurance issues.

• If you decide to pursue genetic testing, a genetic counselor can play an important role in explaining your genetic test results. They can connect you with resources to start surveillance, if necessary, as well as help you to navigate sharing these results with family members.

I Think I Got Genetic Testing a Long Time Ago
Should I Get it Again

Genetic testing technologies have greatly improved over the past few years. People may wish to be re-tested using more modern methods of DNA analysis. These improved techniques are significantly more reliable. This could also allow for more accurate testing of any “at risk” family members. Talk to a genetic counselor to see if any updated tests are available for you.

What are the Possible Results of the Genetic Test, and What Does this Mean for my Health and My Family Members?

• Positive (or Pathogenic) – The lab identified a disease-causing change in the vhl gene, which confirms a diagnosis of VHL. The VHL Alliance has Active Surveillance Guidelines to help manage this increased risk for cancer and other non-cancerous tumors. Family members may also be at risk for VHL and are encouraged to meet with a genetic counselor to discuss the option of a targeted genetic testing for the same variant.

• Negative (or Benign) – The lab did not identify a disease-causing change in the vhl gene: it is unlikely that this individual has VHL.*

• Variant of Unknown Significance (VUS) – The lab identified a change in the vhl gene, but they do not have enough data to determine whether this change is disease-causing or not. Over time, as the lab accumulates more data, they will reclassify this result. Genetic counselors are helpful resources in determining how to best navigate this result.

Genetic counselors are important resources in navigating this kind of result. There is always a very small margin of error in genetic testing. Additionally, current technologies likely cannot detect every possible mutation in the vhl gene. Sometimes more comprehensive genetic testing may be available, such as multi-gene panel testing or exome sequencing. There are some cases where the vhl mutation only occurs in some of the cells, and this can be difficult to detect using a standard genetic test (see “mosaicism” for more information). Genetic counselors can work with you and your other healthcare providers to discuss the impact of this test result on your diagnosis and surveillance.

* I have a clinical diagnosis of VHL, but my genetic test results were negative. What do I do now?

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