

We Need You!

The only way we can learn more about VHL is to gather information from lots of affected families and merge that with the clinical experience of physicians and the laboratory research of scientists. Your research, your ideas, will help us all.

Come and learn from what has already been compiled, and add your own questions and ideas to help us all live better with VHL.

We Need All Our Friends!

We need the moral support and financial participation of all our friends and relations, to find better ways to diagnose and manage VHL, and ultimately to find a cure. Please help.

CARING . . .

An international network of family support groups.

SHARING . . .

In person, on the phone, in online discussion groups in five languages, and through the *VHL Family Forum*

LEARNING . . .

from each other and from our physicians and medical teams

EDUCATING . . .

ourselves, the medical community, and the general public.

CLINICAL CARE CENTERS

Call for a referral to an institution participating in the international VHLFA information network

LOCAL FAMILY SUPPORT CHAPTERS

Call for the contact person in your area, or to start a new group.

Publications

The VHL Handbook and Kids' Handbook
Your Family Health Tree
VHL Family Forum, 3-4 issues a year
Multiple languages available

The VHL Family Alliance is a member of:

Family Voices
Genetic Alliance
National Coalition for Cancer Survivorship
National Organization for Rare Disorders



Dedicated to improving diagnosis, treatment, and quality of life for individuals and families affected by von Hippel-Lindau syndrome.

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IMAGING AND VHL



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Imaging and VHL

von Hippel-Lindau (VHL) is caused by one tiny little misspelling in one gene. Everybody on the planet has two copies of the VHL gene—every human, every dog and cat, every earthworm, every fruit fly, every sea urchin has two copies of the VHL gene.

People who have the medical condition called VHL, have one copy of the VHL gene that works just fine, and a tiny flaw in the second copy that causes it not to work right.

Because of this little flaw, people with VHL are at increased risk of tumors of the retina, brain, inner ear, spinal cord, kidneys, pancreas, and adrenal glands.

People ask “what are the symptoms?”—Well, it depends which issues come up first. But in most cases, if you wait for symptoms to arise, the problem is already pretty big and may be hard to fix.

The best approach to maintaining health in VHL is EARLY DIAGNOSIS—usually through surveillance imaging. By the time there are symptoms, a lot of opportunities are already lost.

- X-ray will not see VHL tumors or other soft structures at all.
- Ultrasound generally does not provide a picture of sufficient clarity to see small tumors developing.
- CT provides good image quality, but we are mindful of the accumulation of radiation. For people who need scans every 1-2 years, or more often when monitoring the growth of a tumor, it is important to minimize the use of radiation. There are also concerns about contrast medium, as it can be harmful to the kidneys, one of the organs affected by VHL.



- MRI can provide good image quality without radiation. Protocols have been developed which minimize the risks of harmful side effects, especially for people with less than perfect kidney function. We are hoping to see the cost of MRI's come down as the machines become less expensive and more available.
- PET scanning plays a key role in finding and treating neuroendocrine tumors.

Finding Issues Early

RETINA

Seeing problems in the retina is easier than seeing problems in the kidney or other internal organs. The retinal specialist can look straight through the dilated pupil and see the retina. Looking before symptoms emerge allows treatment of very small lesions, long before they threaten vision.

BRAIN/SPINAL CORD and INNER EAR

Waiting for symptoms in the brain or spinal cord increases the risk of serious deficits and disability. Checking the brain and spinal cord at least every two years with surveillance MRI allows us to see problems developing, and to make a plan for how to manage tumors. If we see a cyst or syrinx developing, we can make sure we have the right surgical team assembled and can watch for indications that it is time to intervene and remove the tumor before symptoms occur.

The tumors of the inner ear, or endolymphatic sac, are so very small that they will not show up on a regular MRI of the brain. The most efficient surveillance MRI that will cover both the most critical areas of the brain and spine as well as the inner ear is an “MRI with contrast of brain and cervical spine, with thin cuts through posterior fossa, and attention to inner ear/petrous temporal bone to rule out both ELST and hemangioblastomas of neuraxis”.

KIDNEY, PANCREAS

Waiting for symptoms in these organ systems is a very bad idea. VHL tumors in the kidney and pancreas can grow into metastatic cancer. It goes without saying



that it is best to avoid metastatic kidney cancer or pancreatic cancer. If we find tumors at earlier stages, it is possible to monitor tumor growth, differentiate cancer tumors from benign tumors, and intervene before the tumor gains the ability to metastasize. We

maintain a delicate balance between keeping the organ healthy and avoiding metastatic cancer.

PHEOCHOMOCYTOMA/PARAGANGLIOMA

Imaging may also find a tumor on or near the adrenal glands. When symptoms occur, blood and urine tests can confirm that a pheo or para is present. The next question is: where is it? These neuroendocrine tumors can occur anywhere along the sympathetic nervous system, roughly along a line from your earlobe to your groin, on either side of the body. Nuclear medicine tests can be used to find them. A tracer (MIBG or F-DOPA) is injected into the patient's bloodstream, seeks the cell type, and clings to the cells. The nuclear medicine scan then takes a picture and the tracer lights up like a light-bulb, showing the surgeon where the pheo/para is located.

SCANNING IS ESSENTIAL

Scanning gives us the advance warning that we need to gather the right health care team, make a careful plan, and ensure that each intervention is as gentle as possible and is carefully targeted to correct the problem with minimal damage to healthy tissues.

Advances over the last twenty years are primarily due to EARLY DIAGNOSIS and BETTER TREATMENT. Thanks in large part to medical imaging, people with VHL are living longer, healthier lives than ever before in history.