



Von Hippel-Lindau Disease at a Glance

What is VHL?

VHL, or von Hippel-Lindau disease, is a genetic disorder characterized by tumors in up to ten areas of the body. The tumors can be benign or cancerous, and appear and grow unpredictably throughout a patient's life.

Von Hippel-Lindau, is named after two physicians. Eugen von Hippel was a German ophthalmologist who first described tumors in the eyes, called hemangioblastomas, in 1893-1911. Arvid Lindau, a Swedish pathologist, was the first to describe the tumors in the brain and spine in 1926. The *VHL* gene was identified in 1993.

Understanding VHL is a key to finding a cure for cancer. This was underscored when the 2019 Nobel Prize in Medicine was awarded to Dr. William Kaelin, Jr., Sir Peter J. Ratcliffe, and Dr. Gregg L. Semenza. These researchers studied how the *VHL* tumor suppressor gene perceives oxygen levels - a mutated *VHL* gene recognizes a lack of oxygen, leading to a chain reaction which ultimately causes tumor growth.

Some key facts about VHL...

- The incidence of VHL is approximately 1 in 36,000, the same occurrence as glioblastoma, which took the lives of Senators John McCain and Ted Kennedy.
- Because of its rarity, VHL is often undiagnosed or misdiagnosed.
- Children of parents with VHL have a 50% chance of inheriting VHL.
- 20% of VHL cases are first in family due to a spontaneous mutation. These are referred to as *de novo* cases.
- Tumors can develop in up to ten different parts of the human body: brain, spine, retina, kidney, pancreas, adrenal gland, inner ear, reproductive tract, liver, and lungs.
- Malignant tumors, generally found in the kidney and pancreas of VHL patients, can metastasize or spread.
- Benign tumors, although they do not spread, can cause life-changing or life-threatening problems for patients due to their locations and/or size.
- Patients must follow a strict regimen of active surveillance to identify tumors. This regimen includes MRIs, ultrasounds, blood samples, and more. Lack of timely intervention can lead to high rates of medical disability and mortality.
- Eight drugs currently in use to treat breast and kidney cancer are direct results of VHL research.

For more information, see: vhl.org – the web site of the VHL Alliance

References

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