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Harold C. Simmons Cancer Center

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What you should know about Von-Hippel Lindau (VHL) syndrome

VHL syndrome is a rare condition that is caused by mutations in the *VHL* gene. Individuals with VHL are at increased risk to develop multiple tumors all over the body. Many of the tumors associated with this disease are vascular (formed from blood vessels). Most of the tumors in individuals with VHL are not cancerous, but they can cause health problems.

The risk for tumors associated with VHL

VHL is associated with the following risks for cancer:

- 13-72% risk for benign tumors of the brain and spine (hemangioblastomas), which can lead to headaches, vomiting, dizziness, and sometimes difficulty walking
- 25-60% risk for kidney cancer
- 10-20% risk for benign tumors in the adrenal gland (called pheochromocytomas), which can cause high blood pressure
- 17-56% risk for pancreatic tumors, which are usually not cancerous
- 25-60% risk for retinal hemangioblastomas, which are non-cancerous tumors in the eye, which often do not have symptoms. Retinal hemangioblastomas can occasionally cause vision loss
- 11-16% risk for tumors in the ear, which usually are benign, but can be associated with hearing loss
- 25-60% risk for benign cystadenomas of epididymis in males

The risks to family members

VHL is inherited in an autosomal dominant fashion. This means that the children, brothers, sisters, and parents of an individual with VHL have a 50% (1 in 2) chance of having the mutation. Approximately 20% (1 in 5) of individuals with VHL do not have a family history of VHL, and thus have a new mutation.

Managing the risk

The following screening practices are recommended for individuals with VHL:

- Yearly ophthalmologic examination, beginning before age 5
- Yearly blood pressure monitoring and measurement of urinary metabolites beginning at age 5
- Yearly abdominal ultrasounds beginning at age 16, followed by CT or MRI to evaluate any areas of concern
- Audiologic evaluation if hearing deficits are suspected