Von Hippel Lindau syndrome

Aflac Cancer & Blood Disorders Center
CHILDREN'S HEALTHCARE OF ATLANTA

Von Hippel Lindau syndrome is a genetic condition in which individuals are born with a predisposition to develop a range of benign and malignant tumors. These include a certain kind of kidney tumor called renal cell carcinoma, as well as tumors of the liver, pancreas and adrenal gland (pheochromocytomas). Individuals with Von Hippel Lindau syndrome are also at an increased risk to develop cerebellar hemangioblastomas and retinal angiomas. Additionally, there is an increased risk to develop endolymphatic sac tumors that may cause hearing loss.

Cause

Von Hippel Lindau syndrome is caused by genetic changes in the VHL gene, which acts as a tumor suppressor. A tumor suppressor gene, when working properly, encodes proteins that prevent the growth and development of tumors in the human body.

Diagnosis

Von Hippel Lindau syndrome can be diagnosed through genetic testing that is facilitated by a genetic counselor or genetics services provider.

Surveillance

Guidelines exist for surveillance of children and adults with Von Hippel Lindau

References

ncbi.nlm.nih.gov/books/nbk1463/