Von Hippel-Lindau Disease

Von Hippel-Lindau disease (VHL) is a rare genetic condition characterized by multiple hemangioblastomas and cysts in the body. This disease is caused by a mutation in the von Hippel-Lindau gene on chromosome 3. This gene is a tumor suppressor gene that codes for a protein complex that is involved in the ubiquitination and degradation of the transcription factor HIF. The HIF transcription factor plays an important role in the regulation of angiogenesis. Activation of HIF, caused by a mutation in the von Hippel-Lindau gene, results in enhanced angiogenesis leading to multiple hemangioblastomas in the body. Classic locations for these vascular tumors include the medulla, retina, and the cerebellum. Patients also frequently develop cysts in the kidney, liver, and pancreas. Individuals with this mutation are more likely to develop bilateral renal cell carcinomas and have increased the incidence of pheochromocytomas.



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Pathophysiology

Chromosome 3
Chromosome (3) Tree
This disease is caused by a mutation in the VHL gene on chromosome 3.

Presentation

Hemangioblastomas
He-man-angel-blast
Hemangioblastoma is a benign vascular tumor that occurs most commonly in the CNS and retina.

Medulla, Retina, Cerebellum
Medusa Red-tin Silver-cerebellum-bell
Von Hippel-Lindau disease is characterized by multiple hemangioblastomas that are classically found in the medulla, retina, and cerebellum. The involvement of these structures can result in headaches, ataxia, weakness, and vision problems.

Cysts
Cysts
A cyst is an enclosed sac that has a distinct membrane and is separated from the nearby tissue. Von Hippel-Lindau disease often causes multiple cysts throughout the body.

Kidney, Liver, Pancreas
Kidney, Liver, Pancreas
Von Hippel-Lindau disease often causes multiple cysts throughout the body, including the kidney, liver, and pancreas.

Develop Bilateral Renal Carcinomas
Bi-ladders Kidney Car-gnome
Patients with von Hippel-Lindau disease are at an increased risk of developing bilateral renal cell carcinomas. The classic triad of symptoms is hematuria, flank pain, and an abdominal mass, although this occurs in 10-15% of cases.

Pheochromocytomas
Phiat-chrome
Patients with von Hippel-Lindau disease are at increased risk of developing pheochromocytomas. This development is a neuroendocrine tumor of the chromaffin cells, which secrete catecholamines. This tumor is of neural crest cell origin and is often found in the adrenal medulla. It often presents with the classic triad of intermittent palpitations, headache, and diaphoresis.

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