Open Peer Review on Qeios

Von Hippel-Lindau Syndrome

National Cancer Institute

Source

National Cancer Institute. Von Hippel-Lindau Syndrome. NCI Thesaurus. Code C3105.

An inherited familial cancer syndrome which is characterized by development of capillary hemangioblastomas of the central nervous system and retina; clear cell renal carcinoma; pheochromocytoma; pancreatic tumors; and inner ear tumors. The syndrome is associated with germline mutations of the VHL tumor suppressor gene, located on chromosome 3p25-26. Symptoms of VHL syndrome may not be apparent until the third decade of life. CNS hemangioblastoma is the most common cause of death, followed by clear cell renal cell carcinoma. --2004