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Von Hippel-Lindau: Current Evidence in Diagnosis, Treatment, and Nursing Implications

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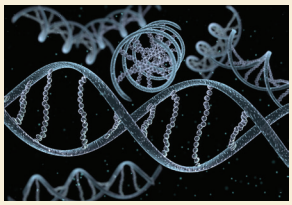

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Background: Von Hippel-Lindau (VHL) is a rare autosomal dominant hereditary disorder that predisposes individuals to benign and malignant tumors in the brain, eyes, kidneys, pancreas, genital tract, or other body parts. The VHL gene, which is located on the short arm of chromosome 3, prevents cells from dividing too rapidly. Mutations in the VHL gene result in uncontrollable cell growth and tumor formation.

Objectives: The purpose of this article is to summarize the current research literature describing diagnosis, treatment, and nursing implications of VHL.

Methods: Three electronic databases, relevant journals, and relevant websites were searched.

Findings: The majority of patients affected with VHL have an affected parent, but a small percentage develop VHL from a new mutation that takes place in a single egg or sperm during conception or from a post-conception mutation. Genetic testing, either through sequence analysis, Southern blot analysis, or quantitative polymerase chain reaction, is considered standard in evaluating patients suspected of having VHL. A diagnosis of VHL can be made by identifying one VHL tumor for a patient who has a confirmed family history of VHL. The presence of at least two tumors is required to make a diagnosis of VHL in a patient without a positive family history. The nursing role includes providing resources on VHL genetic counseling, genetic testing, and palliative care.

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Von Hippel-Lindau (VHL) is an autosomal dominant hereditary disorder in which a single mutated gene is transmitted to offspring from an affected parent (National Cancer Institute [NCI], 2015). Mutations in the VHL gene predispose individuals to various types of benign and malignant neoplasms in different organs (NCI, 2015). For instance, central nervous system tumors are named hemangioblastomas. Other types of VHL tumors may develop in the kidneys, the pancreas, or other organs (NCI, 2015).

Diagnosis, treatment, and evaluation of patients with VHL require a multidisciplinary approach. Specialists and healthcare personnel who are involved in treating patients with VHL may include oncology surgeons, neurosurgeons, general surgeons, ophthalmologists, endocrinologists, neurologists, genetic counselors, nurse practitioners, and nurses (NCI, 2015). VHL is likely to be a burden for patients, families, nurses, and healthcare personnel who are not familiar with its complexity. Therefore, the multiple roles that nurses play become critical as they embrace the need to act as a liaison, educator, and coordinator in managing VHL. Because nurses are on the front line of patient care, they must have a basic understanding of VHL and current evidence regarding its diagnosis and treatment.

The term *von Hippel-Lindau* was first used in 1936 (Maher, Neumann, & Richard, 2011). VHL is named for two famous European physicians, Eugen von Hippel and Arvid Lindau. The German ophthalmologist Eugen von Hippel first described angiomas in the eye in 1904. Arvid Lindau, a Swedish pathologist, described the angiomas of the cerebellum, kidney, and spine in 1926 (Molino, Sepe, Anastasio, & De Santo, 2006). However, the term VHL was not in common use until the 1970s (Maher et al., 2011).