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.