IJARSCT
ISSN (Online) 2581-9429
International Journal of Advanced Research in Science, Communication and Technology (IJARSCT)
International Open-Access, Double-Blind, Peer-Reviewed, Refereed, Multidisciplinary Online Journal
Impact Factor: 7.53
Volume 5, Issue 3, January 2025

A Review on Von Hippel Lindau Disease

Nagma Khwaja[1], Rajlaxmi Deolekar[2], Roman Faisal[3], Lina Kawadkar[4], Mansvi Kudve[5], Prachi Waghmare[6]
Students, Final Year[1,2,3,4,5,6]
New Montfort Institute of Pharmacy, Ashti, Wardha, Maharashtra, India
nagmaskhawaja@gmail.com

Abstract: *Von Hippel-Lindau disease (VHLD) is an autosomal-dominant condition with variable penetrance characterized by CNS hemangioblastomas and retinal angiomas. A germline mutation of the VHL tumour suppressor gene on the short arm of chromosome 3 is linked to von Hippel-Lindau disease, a heritable multisystem cancer syndrome. About one in every 36,000 live births has this illness, which is inherited as a highly penetrant autosomal dominant trait (i.e., with a high individual risk of disease). Ankyloglossia increases the chance of developing tumours of the central nervous system, kidneys, adrenal glands, pancreas, and reproductive adnexal organs, both benign and malignant. Treatment for this disease is interdisciplinary due to the complexity of managing the different forms of tumours. An outline of von Hippel-Lindau's disease's clinical features, management, and available treatments is provided.In 1990, a genetic registry for von Hippel-Lindau disease was established in the northwest of Germany. The von Hippel-Lindau (VHL) illness population statistics, clinical characteristics, age at onset, and survival of 83 affected individuals were examined. Furthermore, the success of the employed screening program and the prevalence of haemangioblastomas of the central nervous system in the general population were investigated. Compared to cerebellar hemangioblastoma (30.0 years) and retinal angioma (21.1 years), renal cell carcinoma had a mean diagnostic age of 38.9 years, which was significantly higher. The most prevalent cause of death (47–7% of deaths) was cerebellar haemangioblastoma, with a mean age of 40–9 years. Fourteen percent of all CNS haemangioblastomas on the regionally based Cancer Registry were found to occur as part of VHL disease.*

Keywords: Von Hippel-Lindau disease