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VON-HIPPEL LINDAU'S DISEASE: A CASE STUDY

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This article is dedicated to the loving memory of my teacher Oleg Mihailovich Korolenko.

Introduction: Von Hippel-Lindau disease is an autosomal dominant neoplasia syndrome that results from a germline mutation in the VHL gene. Germline mutations in the VHL gene lead to the development of several benign or malignant tumours, and cysts in many organ systems. Affected individuals might develop CNS lesions including cerebellar, spinal cord, brainstem, nerve root, and supratentorial haemangioblastomas, as well as retinal haemangioblastomas and endolymphatic sac tumours. Visceral features of the disorder include renal cysts and carcinomas, phaeochromocytomas, pancreatic cysts and neuroendocrine tumours, as well as epididymal and broad ligament cystadenomas.

Aim: To properly diagnose and prevent complications that may occur in patients.

Materials and methods: Computer tomography scan of the head, Anamnestic data, Clinical objective examination data.

Results: Patient X, male, 50 years old presented to the neurological department of KNMU with painless decrease of vision in the right eye. The intraocular pressure was within normal limits. The results of bilateral biomicroscopic examination of anterior segments were unremarkable. Right eye fundus examination revealed lesions in the superior retina with dilated vessels and tortuous draining vein which is a characteristic for Retinal hemangioblastoma. Systemic studies, including magnetic resonance imaging of the brain revealed bilateral cerebellar tumors. His family history was without any hereditary diseases. On the basis of clinical and investigational findings, he was diagnosed with Von-Hippel Lindau's disease. Diagnosis of von Hippel-Lindau disease is often based on clinical criteria. Patients with a family history, and a CNS haemangioblastoma (including retinal haemangio-blastomas), phaeochromocytoma, or clear cell renal carcinoma are diagnosed with the disease. Those with no relevant family history must have two or more CNS haemangioblastomas, or one CNS haemangioblastoma and a visceral tumour (with the exception of epididymal and renal cysts, which are frequent in the general population) to meet the diagnostic criteria.

Conclusion: Specific correlations of genotype and phenotype have emerged in affected families. Several familial phenotypes of von Hippel-Lindau disease are now recognised, providing useful information to screen and counsel affected individuals. Type 1 families have a greatly reduced risk of phaeochromocytomas, but can develop all the other tumour types generally associated with the disease. Type 2 families have phaeo-chromocytomas, but have either a low-risk (type 2A) or high-risk (type 2B) for renal cell carcinomas. Type 2C families have phaeochromocytomas only, with no other neoplastic findings of VHL. The new insights into the underlying mechanisms of tumour formation, greater knowledge of the natural history of the various lesions associated with von Hippel-Lindau disease, and more precise diagnostic studies (laboratory and imaging) should lead to an improved quality of life and extend the life expectancy of affected individuals. The diverse multisystem effects of this disease need careful, selective, and coordinated planning to determine the treatment of individual lesions that will provide the best long-term management of these patients.