

Signs and symptoms associated with VHL disease include headaches, problems with balance and walking, dizziness, weakness of the limbs, vision problems, and high blood pressure. Conditions associated with VHL disease include angiomatosis, hemangioblastomas, pheochromocytoma, renal cell carcinoma, pancreatic cysts (pancreatic serous cystadenoma), endolymphatic sac tumor, and bilateral papillary cystadenomas of the epididymis (men) or broad ligament of the uterus (women).

Angiomas occur in 37.2% of patients presenting with VHL disease and usually occur in the retina. As a result, loss of vision is very common. However, other organs can be affected: strokes, heart attacks, and cardiovascular disease are common additional symptoms.

Approximately 40% of VHL disease presents with CNS hemangioblastomas and they are present in around 60-80%. Spinal cord hemangioblastomas are found in 13-59% of VHL disease and are specific because 80% are found in VHL disease.

Although all of these tumours are common in VHL disease, around half of cases present with only one tumour type.

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