INTRODUCTION: Von Hippel-Lindau disease (VHL) is an autosomal dominant multisystemic cancer syndrome due to a mutation of the VHL tumor suppressor gene on chromosome 3, region p25-26, with an incidence of 1/36,000 in newborns. Patients are at risk of developing cerebellar, spinal and retinal hemangioblastoma, renal cell carcinoma, pheochromocytoma, pancreatic neuroendocrine tumors, pancreatic and renal cysts, and epididymal cystadenoma. The most common causes of death from VHL are metastases from renal cell carcinoma and neurological complications from cerebellar hemangioblastomas. Molecular analysis of the VHL gene is clinically available and indicated in patients with known or suspected VHL. CASE REPORT: A 19-year-old woman was surgically treated for cerebellar hemangioblastoma in 1998 and for renal cell carcinoma of the right side in 2002. Familial VHL was subsequently diagnosed as the patient’s mother was found to be affected with bilateral polycystic kidney disease with chronic renal failure as well as hemangioblastoma of the retina and medulla oblongata. The mother underwent surgery for bilateral renal cell carcinoma in 2003. CONCLUSION: The multitude of VHL-associated tumors and intra-familial variability in clinical expressivity render early diagnosis of VHL difficult. We therefore shortly illustrate the spectrum of clinical phenotypes and the VHL screening and surveillance guidelines.