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Management of von Hippel-Lindau Disease: An Interdisciplinary Review

Supplemental Material

Case Report

A 56 year old female was initially treated for a retinal disruption with laser coagulation. Four years later she complained of flank pain during a routine gynaecological control. Elevated blood pressure was measured and multiple renal cysts were detected on ultrasound.

This constellation led to the diagnosis of polycystic kidney disease (PKD) with the ophthalmologic diagnosis not being taken into consideration. Follow-up ultrasound after four years and optimal antihypertensive treatment were recommended.

At the same time her 26 year old daughter underwent resection of a cystic cerebellar haemangioblastoma (HB). It was classified as an isolated lesion. Knowing about the hereditary nature of her mother's PKD, the patient requested an ultrasound of the kidneys. This examination revealed renal cysts, but no further diagnostic workup was undertaken.

About four years later, the daughter presented for abdominal cramps to the emergency department. The symptoms were caused by constipation. However, atypical renal cysts were again seen on abdominal ultrasound and further imaging was performed. On abdominal computed tomography (CT) one renal lesion was considered suspicious for malignancy and resection was recommended. More importantly, by reviewing the patient's history (cerebellar HB and renal cysts) and being aware of the positive family history of retinal lesions and polycystic kidneys, the treating urologist was the first physician raising the differential diagnosis of VHL disease. A nephron-sparing tumourectomy was performed. Histology revealed a clear cell renal cell cancer (ccRCC).

After genetic counselling, molecular genetic analysis confirmed the diagnosis of VHL disease in the mother and the daughter. Since then, both are managed within the St. Gallen VHL Multidisciplinary Group. In the meantime, the daughter has undergone several renal tumour resections due to multiple bilateral ccRCCs. If the urologist had not made the association between the anamnestic clues and would have performed standard nephrectomy rather than a tumourectomy, the daughter might be on haemodialysis since the age of 35.