Visceral Manifestations of von Hippel — Lindau Disease: Value of Ultrasound and CT Imaging

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Abstract

Visceral manifestations of von Hippel - Lindau disease (VHLD) are generally asymptomatic and their early detection is of considerable help in the management. This communication documents the usefulness of imaging studies in detecting visceral manifestations in two cases of VHLD.

INTRODUCTION

von Hippel - Lindau disease (VHLD) is a rare autosomal dominant disorder characterized by CNS haemangioblastomas, retinal angiomas, renal cell carcinomas (RCC), phaeochromocytomas and visceral cysts.1 Visceral manifestations particularly phaeochromocytomas and RCC are associated with significant morbidity and mortality.2,3 These associated neoplasms are often overlooked since they are asymptomatic at initial assessment of patients and the usefulness of radiological screening in their detection is well documented.1,2 This report documents two cases of clinically unsuspected VHLD recognized during computed tomography (CT) evaluation of a renal mass in one case and ultrasound (US) screening of the abdomen in another patient with a cerebellar tumor. This study emphasizes the importance of CT in the demonstration of co-existing phaeochromocytomas and pancreatic tumors during pre-operative assessment in VHLD.

CASE REPORT 1

A 37 years male presented with painless hematuria and loss of weight for three months. He was a diabetic and hypertensive for five years. The resting pulse rate was 110/min and there was mild tremor of both hands. Blood pressure was 140/100 mm Hg. Clinical examination of the abdomen was noncontributory. Urine sediment revealed RBCs. US of the abdomen revealed a multicystic lesion in the pancreas, and a mixed echogenic mass in the mid and lower pole of the right kidney. Contrast enhanced CT (CECT) revealed a moderately enhancing mixed density mass in the mid and lower

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pole of the right kidney with multiple renal cysts in both kidneys. Multiple cystic lesions also replaced the pancreatic parenchyma. In addition an intensely enhancing mass was also noted in the left adrenal gland. Metabolic assessment showed elevated urinary metanephrines 585 mg/24h (Normal = 20 to 345 mg/24h) and normetanephrines - 2500 mg/24h (Normal = 30 - 440 mg/24h). Thyroid function studies and imaging study of the brain showed no abnormalities. Phenoxybenzamize and atenolol with unrestricted salt and fluid intake were initiated. After adequate duration of preoperative drug therapy a left adrenalectomy and a right nephrectomy was undertaken. The pancreas was found to be cystic at surgery and fluid was aspirated for analysis. Histopathology revealed a renal cell carcinoma - clear cell type, nuclear grade - II, a left adrenal phaeochromocytoma and a pancreatic serous cystadenoma.

**Case Report 2**

A 31 years old normotensive male patient presented with headache and occasional vomiting of one-year duration. His vital signs and clinical examination were unremarkable. CT and MRI of the brain revealed an enhancing space occupying lesion in the cerebellar parenchyma with compression of the fourth ventricle with moderate hydrocephalus. Multiple prominent vessels in the periphery of the lesion suggested the possibility of haemangioblastomas. Ultrasound examination of abdomen was performed as part of the pre-operative evaluation, to exclude any co-existing lesions. This revealed multiple complex cystic lesions in both kidneys and pancreas. CECT abdomen revealed multiple cystic lesions with solid components in both kidneys, few cysts in the pancreas and a small lesion in the left adrenal gland. Patient underwent preliminary VP shunt followed by stereotactic biopsy of the cerebellar lesion, which was found to be a haemangioblastoma on histology. The complex cystic masses of both kidneys were evaluated by fine needle aspiration cytology; no malignant cells were found. The patient was advised to come for periodical review.

**Discussion**

Common manifestations of VHLD include retinal angioma and hamangioblastoma of the CNS.1,2 Abdominal manifestations of VHLD are less common1 and include renal cell carcinoma, renal cysts, phaeochromocytoma, epididymal cysts, multiple cysts and serous cystadenoma of the pancreas. Rarely, endolymphatic sac tumors, liver and lung lesions have been reported. The abdominal lesions of VHLD tend to be asymptomatic (49%) at initial presentation and are generally diagnosed later than the cranial manifestation.1 Once the disease is suspected on the basis of a cranial or retinal lesion a vigorous effort should be made to detect possible abdominal lesions in affected individuals.

Phaeochromocytomas occur in about 15-35% of all individuals with VHLD.1-3 Patients with phaeochromocytoma as part of VHLD are less likely to have symptoms attributable to excess catecholamine release than those with sporadically occurring tumors. Although any person with a phaeochromocytoma is susceptible to life-threatening paroxysms, patients with phaeochromocytoma with VHLD are at particular risk, because they undergo frequent diagnostic and therapeutic procedures to evaluate and treat associated lesions.3 Hence it is mandatory to screen all patients with suspected VHLD for phaeochromocytoma before planning surgical or interventional procedures.1,3 In one of our patient under report, a clinically unsuspected phaeochromocytoma was detected during preoperative assessment of a renal tumour by CT, thereby preventing a potential catastrophe.

Pancreatic lesions of VHLD include cysts (accounts for 72%), serous cystadenomas, solid non-functional islet cell tumors and adenocarcinomas.1,5 CT is a consistent and reliable method in detecting pancreatic lesions as US failed to depict the pancreatic body and tail adequately due to the overlying bowel in one of our cases.

Multiple renal cystic lesions are the most common manifestation of VHLD (accounting for 75%)1 and a RCC in VHLD varies from 25% to 38%, accounting for 20% - 50% of deaths.2,5 These can be bilateral and multifocal, in 60% of cases.5 Usually detection of sporadic non-familial renal cancers occurs in the 6th decade.5 Early renal cell carcinoma complicating VHLD is difficult to detect by US2,4 and distinction between complicated / hemorrhage cyst and RCC is not always
easy in US. CECT depicts its enhancing solid component of RCC readily and CT is most sensitive in early detection.

Apart from lesions of the kidney, pancreas and adrenal gland, other lesions described in VHLD\textsuperscript{1,2} such as adenomas, cysts of the liver and spleen were not seen in our cases. Such lesions are all potentially detectable by ultrasound and CT.

It is generally agreed that a careful prospective follow up is necessary in all patients with VHLD and their asymptomatic family members.\textsuperscript{1,2,4} While inexpensiveness and absence of ionizing radiation make ultrasound a useful initial screening procedure, CECT is a more reliable imaging modality for small pancreatic and adrenal lesions of VHLD.

\textbf{REFERENCES}