Von Hippel Lindau disease (ECR 2012 Case of the Day)
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Section: Abdominal imaging
Area of Interest: Abdomen
Procedure: Complications
Imaging Technique: CT
Special Focus: Cysts Neoplasia Case Type: Clinical Cases
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Patient: 48 years, male

Clinical History:

This 48-year-old male patient presented to hospital with a two-month history of malaise, nausea and weight loss. His past medical history was unremarkable apart from a neurosurgical procedure in childhood.

Imaging Findings:

Fig. 1a: Axial image from a contrast-enhanced abdominal CT at the level of the pancreas showing cystic transformation of the body and tail of the pancreas.
Fig. 1b: Axial image from a contrast-enhanced CT of the abdomen at the level of the portal vein showing similar cystic changes in the head and neck of the pancreas.
Fig. 1c: Further axial image from a contrast-enhanced CT of the abdomen shows cystic change involving the uncinate process of the pancreas.

Discussion:

Von Hippel Lindau (VHL) disease is a rare, autosomal dominantly inherited multisystem disorder characterized by development of a variety of benign and malignant tumours. The gene for VHL has a high penetrance but variable phenotypic expression [1].

Clinical perspective: About 40 different lesions in 14 different organs have been described including retinal and central nervous system haemangioblastomas, endolymphatic sac tumours, renal cysts and tumours, pancreatic cysts and tumours, pheochromocytomas, and epididymal cystadenomas [1]. Common causes of death in VHL patients are renal cell carcinoma and neurologic complications from cerebellar haemangioblastomas [1]. VHL may present at any time from infancy to the seventh decade and the median life expectancy is 49 years [2, 3].

Imaging perspective: The various manifestations can be demonstrated with different imaging modalities such as ultrasonography, computed tomography, magnetic resonance imaging, and nuclear medicine. Pancreatic cysts are rare in healthy persons and the presence of a single pancreatic cyst in a patient with a family history of VHL is highly suggestive of the disease. This may be the only abdominal manifestation of the disease preceding others by several years [4]. Pancreatic involvement in VHL disease includes simple pancreatic cysts (50%–91%), serous microcystic cystadenomas (12%), and rarely adenocarcinomas. Pancreatic neuroendocrine tumours also occur [5].

The renal lesions vary from simple cysts to hyperplastic cysts, cysts containing clear cell carcinoma and solid...
tumours. Renal cell cancers associated with VHL are either multicentric and bilateral solid hypervascular masses or complex cystic masses with mural nodules and thick septa. CT is more sensitive than US for detection of small lesions and solid tumours enhance briskly after contrast administration. Nevertheless, US may be preferable for surveillance purposes to reduce the amount of radiation exposure to the patient to as low as reasonably possible. MR imaging is especially useful in young patients and those with renal failure who still require screening. The best sequences are fast T2-weighted images or contrast-enhanced T1-weighted images with fat suppression.

Outcome: The patient underwent nephron-sparing surgery for the small left renal mass which proved to be a renal cell carcinoma.

Take home message: Although genetic testing is available, the manifestations of VHL are protein; therefore, imaging plays a key role in identification of abnormalities and subsequent follow-up of lesions (1). Screening is important because the lesions in VHL disease are treatable; thus, early detection allows use of more conservative therapy and may enhance the patient’s length and quality of life [2, 3].

**Differential Diagnosis List:** Von Hippel Lindau disease – pancreatic, renal and cerebellar manifestations., Mucinous cystic pancreatic tumour, Von Hippel Lindau disease, Serous cystadenoma of the pancreas, AD polycystic kidney disease, Intraductal papillary mucinous tumour of the pancreas

**Final Diagnosis:** Von Hippel Lindau disease – pancreatic, renal and cerebellar manifestations.

**References:**


Figure 1

**a**

Description: Axial image from a contrast enhanced abdominal CT at the level of the pancreas showing cystic transformation of the body and tail of the pancreas. **Origin:** Fenlon H, Department of Radiology, Mater Misericordiae University Hospital, Dublin, Ireland.

**b**

Description: Axial image from a contrast enhanced CT of the abdomen at the level of the portal vein showing similar cystic changes in the head and neck of the pancreas. **Origin:** Fenlon H, Department of Radiology, Mater Misericordiae University Hospital, Dublin, Ireland.
Description: Further axial image from a contrast enhanced CT of the abdomen shows cystic change involving the uncinate process of the pancreas. Origin: Fenlon H, Department of Radiology, Mater Misericordiae University Hospital, Dublin, Ireland.