Case 9555

Von Hippel-Lindau Disease: SNC, renal and pancreatic involvement
Published on 12.09.2011

DOI: 10.1594/EURORAD/CASE.9555
ISSN: 1563-4086
Section: Abdominal imaging
Area of Interest: Neuroradiology brain Abdomen
Procedure: Diagnostic procedure
Imaging Technique: MR
Imaging Technique: CT
Special Focus: Cysts Neoplasia Case Type: Clinical Cases
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Patient: 25 years, male

Clinical History:

Male patient, 25 years old, with history of intermittent episodes of headache and nausea. Two weeks before onset of severe headache associated with positional vertigo and nystagmus.

Imaging Findings:

The patient underwent a brain MRI that revealed a well-demarcated cystic lesion with a highly vascular mural nodule. Mural nodule has slightly low-medium signal intensity on T1-weighted images (Fig. 1 a), high signal intensity on T2-WI and avidly enhances after the administration of contrast material (Fig. 1 b).

These MR imaging characteristics are compatible with cerebellar haemangioblastoma. The most useful sequence for detection of these lesions is unenhanced and contrast-enhanced T1-WI.

Since this lesion manifests itself in a young person and there is a known association with Von Hippel Lindau (VHL) disease, abdominopelvic CT was carried out which showed multiple simple pancreatic cysts and renal cortical cysts as well as the presence of solid renal tumour (Fig. 2 and 3). CT is essential for the diagnosis of kidney and pancreatic involvement. Ultrasound is less sensitive than CT in detecting small lesions. MRI is important in patients with renal failure.

Discussion:

The VHL disease is a rare hereditary genetic disorder, with multisystemic manifestations. Inheritance is autosomal dominant, associated with inactivation of a tumour suppression gene located on chromosome 3p25.5 [1-3].

The spectrum of clinical manifestations of the disease is broad and includes retinal and SNC haemangioblastomas, renal and pancreatic cysts and tumours, pheochromocytomas, epididymal cystadenomas, endolymphatic sac tumours. The diagnostic criteria include: (1) more than one CNS haemangioblastoma, (2) one CNS haemangioblastoma and visceral manifestations and (3) any manifestation and a known family history of VHL disease [2-5].

The CNS haemangioblastomas associated with VHL disease occur mainly in the cerebellum, spinal cord and
medulla. Occur in younger patients, are often multiple and have a worse prognosis than sporadic. They are usually cystic with a solid mural component (55-60%) which is highly vascular. The pure solids are less frequent, have a worse prognosis and higher recurrence rate. Symptoms of cerebellar lesions include headache, vertigo, ataxia, vomiting, nystagmus, and ninth cranial nerve palsy. Ocular and CNS manifestations usually precede renal involvement [1, 4, 6].

Renal involvement manifests by a continuum of lesions from simple cysts to renal cell carcinoma (RCC) and is multicentric and bilateral in 75% of cases. The renal cysts occur in 59-63% and RCC in 24-45% of patients. Most cystic lesions remain cystic over time and often increase in size, some involute leaving scars. The number or size of cysts does not correlate with the malignant potential. The RCC associated with VHL disease occurs earlier, affects men and women with the same frequency and has a faster growth compared to sporadic RCC, which occurs later and predominantly in male patients. The RCC can arise in complex cystic precursor lesions. There is an increased incidence of tumours in the walls of cysts, so even the cysts with benign appearance should be monitored. Given the high risk of recurrence and uncommon metastatic involvement in tumours less than 7 cm the preferred method of treatment is conservative approach and nephron-sparing surgery. The RCC is responsible for the deaths of about 1/3 of these patients [2, 3-8].

The frequency of pancreatic involvement in VHL disease is 15–77%. Lesions include simple pancreatic cysts (91%), serous cystadenomas (12%), neuroendocrine tumors (7–12%) and combined lesions (11%). The most common are pancreatic cysts which are benign, distributed randomly throughout the parenchyma and vary in size. When most of the parenchyma is replaced by multiple cysts pancreatic insufficiency can occur. Pancreatic adenocarcinoma and hemangioblastoma have also been described in VHL disease [1,2,7,8].

Differential Diagnosis List: Von-Hippel-Lindau disease, Sporadic cerebellar haemangioblastoma, Sporadic renal cell carcinoma, Extensive cystic renal involvement by VHL disease may mimic autosomal dominant polycystic kidney disease

Final Diagnosis: Von-Hippel-Lindau disease

References:

manifestations. Radiology 174:815-820 (PMID: 2305064)
Figure 1

a

Description: Axial unenhanced T1-weighted MR image. Cystic well-demarcated lesion in the left cerebellar hemisphere with solid mural nodule with slightly low-medium signal intensity on unenhanced T1-WI. **Origin:** Department of Radiology, IPO Porto

b

Description: Axial enhanced T1-weighted MR image - the solid mural nodule is highly vascular and avidly enhances with gadolinium. **Origin:** Department of Radiology, IPO Porto
**Figure 2**

**Description:** Cortical solid nodule, deforming the kidney contour located in the upper right kidney, that enhances in arterial phase. **Origin:** Department of Radiology, IPO Porto
Description: Presence of several simple cystic lesions scattered throughout the pancreatic parenchyma.
Simple cortical cyst in the upper left kidney. **Origin:** Department of Radiology, IPO Porto