JBR–BTR, 2011, 94: 140-141.

CENTRAL NERVOUS SYSTEM LESIONS IN VON HIPPEL-LINDAU SYNDROME

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Key-word: von Hippel-Lindau disease

Background: A 48-year-old man presented with persisting tinnitus and progressive worsening occipital headaches, neckpain, ataxia and paresthesia of the scalp. There was no recent trauma in his recent history.

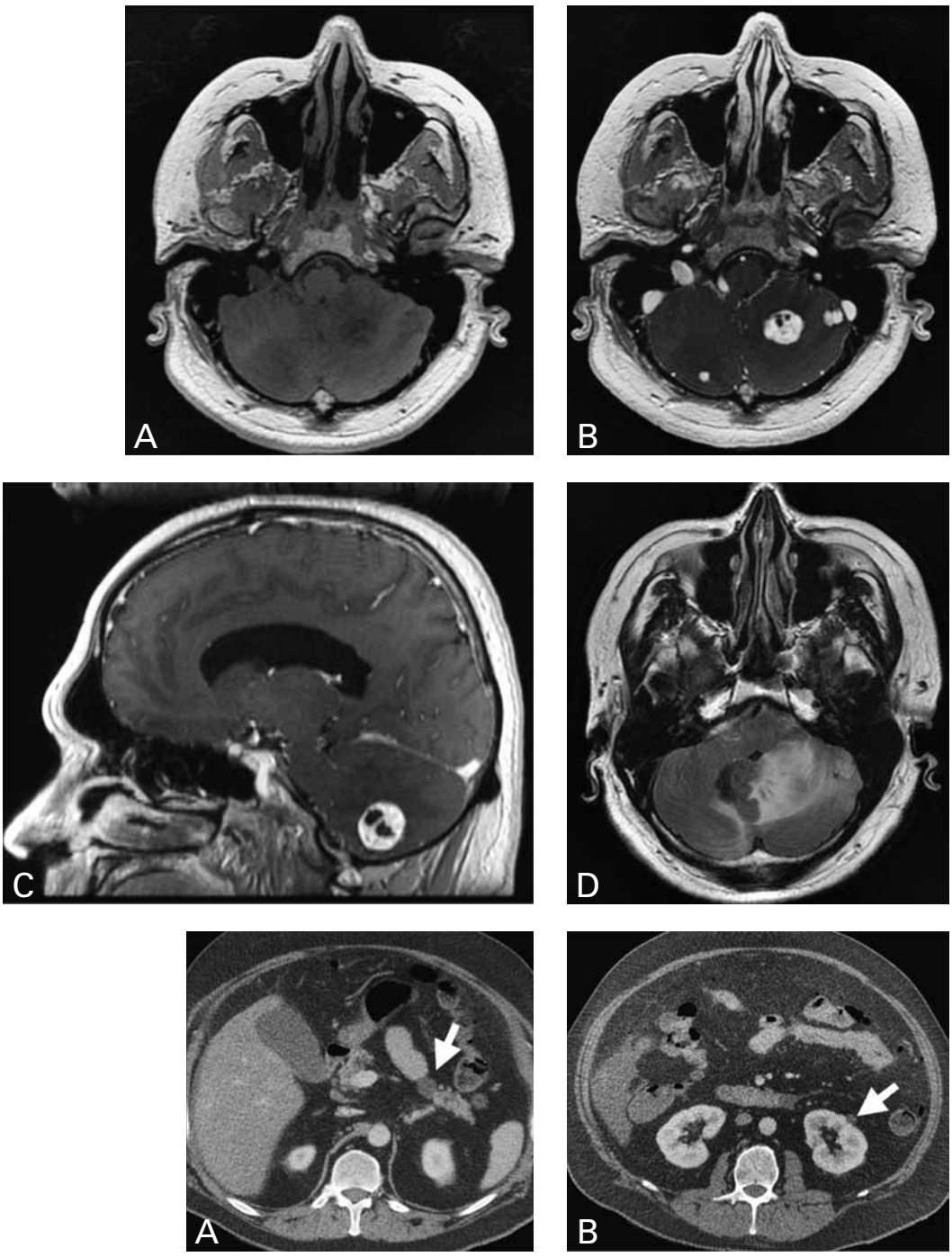

Fig.	1A	1B
	1C	1D
	2A	2B

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Work-up

MRI of the brain (Fig. 1) includes axial (A) and axial Gd-enhanced (B) T1 images (3D FSPGR). Depiction of three out of the four heterogeneously enhancing cerebellar masses with inlying cystic components in both cerebellar hemispheres. There is a definite mass effect. Sagittal Gd-enhanced T1 image (3D FSPGR) (C) demonstrates the craniocaudal extension of the largest cerebellar cystic lesion, situated in the left cerebellar hemisphere. On axial T2 Flair image (D) the parenchymal lesions are heterogeneously hyperintense with perilesional edema and compression of the left cerebellar peduncle.

CT scan of the abdomen (Fig. 2) shows on axial contrast-enhanced views a single cyst in the body of the pancreas and the left kidney.

Radiological diagnosis

The cystic aspect of the cerebellar lesions with mural enhancing nodules as well as the presence of enlarged blood vessels in the tumor region are highly suggestive for hemangioblastoma. The patient had a genetically confirmed family history of Von Hippel-Lindau disease (hemangioblastosis) with his father and three of his six sisters presenting hemangioblastosis. The cystic lesions found in the pancreas and the left kidney are also consistent with the diagnosis of *Von Hippel-Lindau disease*.

Von Hippel-Lindau is an autosomal dominant disease with the disorder matched to the short arm of chromosome 3. The disease is a multisystem disorder with phenotypic variability.

Manifestations can be clustered. In central nervous system hemangioblastomas and retinal angiomas, the tumors originate from the blood vessels and usually become symptomatic in the 3rd or 4th decade of life. Symptoms are variable and consist of headache, vomiting, confusion, blurred vision, neck pain and ataxia. At CT scan hemangioblastomas are cystic in 75% of cases and solid or mixed in the remaining cases. There is typically an intense tumor blush, characterized by the presence of an enhancing nodule. As a general rule hemangioblastomas do not exhibit calcifi cation, which is a useful sign in aiding distinction from other tumors, such as cystic astrocytoma. At MRI, the masses are often cystic in appearance with prolongation of both T1 and T2 relaxation times.

The solid component of the tumor is usually peripheral in location and shows avid enhancement follwing Gd-contrast administration. Because of large feeding and draining vessels small tubular areas of flow void may sometimes be seen within the nodule. On both CT and MRI, enhancement following administration of contrast medium is essential for the accurate detection of these lesions. Both CT angiography and conventional angiography are useful adjuncts in the detection and diagnosis of hemangioblastomas.

In renal lesions, renal cysts are frequently found (50-70%). They rarely produce clinical symptoms. In up to 70% of patients renal cell carcinoma occurs.

In case of pancreatic lesions, simple cysts are the most common finding and rarely of functional significance.

Pheochromocytomas occur in 7%-20% of patients.

In other organs, simple cysts can be found in many other parts of the human body, for example cyst adenomas of the epididymis and endolymphatic tumors of the inner ear.

Criteria to establish the diagnosis are the demonstration of at least two hemangioblastomas of the CNS, one retinal hemangioma of the CNS associated with a visceral manifestation of the disease, or known family history and at least one manifestation of the disease.

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