Von Hippel-Lindau Disease Presenting With Cranial and Spinal Hemangioblastomas: MRI Findings

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Abstract- Hemangioblastoma (HB) is one of the most common primary neoplasms of the posterior fossa in adults. Although single tumors can be sporadic, multiple tumors are almost always associated with von Hippel-Lindau (VHL) disease. In our case report, we are describing a 39-year-old woman with complaints of severe headache, pain, and numbness in both arms. She had a history of VHL disease. We performed contrast-enhanced cranial and cervical spine magnetic resonance imaging (MRI) examinations. Given the patient's history of VHL disease and characteristic MR imaging features of lesions, cranial and cervical HBs were considered for diagnosis. Surgical excision is the main treatment of these tumors and follow-up optimal imaging of these patients is crucial. In the screening of VHL patients, whole spinal axis imaging in conjunction with routine MRI studies must always be performed to rule out spinal HBs.
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Keywords: Hemangioblastoma; Magnetic resonance imaging; Von hippel-lindau

Introduction

von Hippel-Lindau (VHL) disease is a rarely seen familial syndrome. It is characterized by retinal angiomas, multiple central nervous systems (CNS) hemangioblastomas, and also cysts and tumors of the various abdominal organs. Hemangioblastoma (HB) is a rarely seen vascular tumor of the CNS. Their peak age is between 20 and 50 years of age. Magnetic resonance imaging (MRI) is the preferred modality in diagnosing the CNS manifestations of VHL disease due to its proven multiplanar capability in brain and spine imaging (1).

Case Report

A 39-year-old woman applied to the hospital with complaints of severe headache, pain, and numbness in both arms. She had a history of VHL disease and following her neurological examination, cranial and cervical spine MRI examinations were carried out. Before examinations, informed consent was obtained from the patient. The cranial and cervical spine MRI examinations were performed using a 1.5-tesla magnet (Avanto-SQ Engine; Siemens, Erlangen, Germany). For cranial examination, axial and sagittal T1 weighted, axial and coronal fast spin echo (FSE) T2 weighted, axial and coronal FSE IR (FLAIR), axial SWI (susceptibility weighted image), and for cervical spinal examination, axial and sagittal T1 and FSE T2 weighted images were obtained. Afterward, following intravenous gadolinium diethylenetriaminepentaacetic acid (DTPA) administration (0.1 mmol/kg) axial, sagittal, and coronal post-contrast T1 weighted images were also obtained for both examinations. On the axial T1 weighted image, a hypointense mass was found in the left cerebellar region. The axial FSE T2 weighted image showed a 55x39 mm fairly hyperintense cystic mass lesion in the left cerebellar hemisphere extending to and compressing the fourth ventricle (Figure 1a). On the post-contrast coronal T1 weighted image, a 5 mm mural nodule showing contrast enhancement at the left lateral portion of the mass was present (Figure 1b). The cervical spinal examination yielded a cystic mass lesion extending from the level of the foramen magnum to the whole cervical spinal cord which exhibited heterogeneous hyperintense

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signal intensity on FSE T2 weighted sagittal images (Figure 1c). Following contrast administration; multiple, subcentimeter, contrast-enhancing nodular lesions were detected in the proximal cervical spinal cord (Figure 1d). Given the patient's history of VHL disease and characteristic MR imaging features of lesions, cranial and cervical HBs were considered for diagnosis and the following operation confirmed our diagnosis.

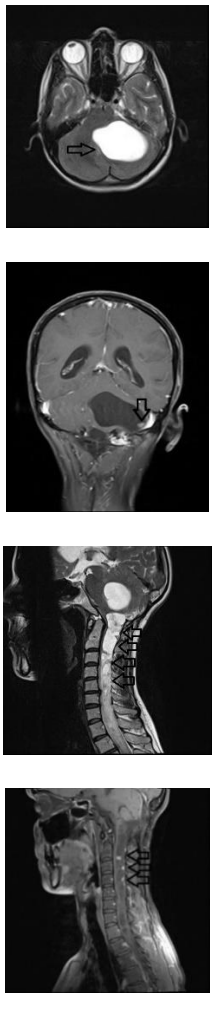

Figure 1. a: Axial FSE T2 weighted image, a hyperintense cystic mass lesion measuring 55x39 mm in diameter in the left cerebellar hemisphere is present. The mass extends to the vermis and compresses the fourth ventricle. b: Coronal post-contrast T1 weighted image, a 5 mm mural nodule in the lateral part of the cystic mass showing contrast enhancement. c: Cervical spine FSE T2 weighted sagittal image, a heterogenous hyperintense signal intensity cystic mass lesion extending along the whole cervical spinal cord. An incidental central disc protrusion is also present at the C5- C6 level. d: Cervical spine post-contrast T1 weighted sagittal image; multiple, contrast-enhancing, millimetric nodular lesions are visible in the proximal cervical spinal cord

Discussion

VHL disease can be diagnosed if the patient has a- multiple retinal and CNS HBs and or b- one HB together with a cyst in the liver, pancreas, kidney, or epididymis or a renal cell carcinoma or phaeochromocytoma. A family history of VHL disease with at least one HB, renal cell carcinoma or phaeochromocytoma, or visceral cyst is also sufficient to establish the diagnosis (1). HB is a vascular tumor of the CNS, usually occurring in the brain, brain stem, spinal cord, and retina, and most often found in the cerebellum (2).

It is one of the most common primary neoplasms of the posterior fossa in adults. Although single tumors can be sporadic, multiple tumors are nearly always seen with VHL disease (3). Sporadic tumors usually appear in the 5th and 6th decades of life, whereas VHL-associated ones occur at earlier periods, namely in the 3rd or 4th decades (2,4).

Cerebellar HBs are classified into four types based on their histological and radiological features (3). Type 1 is a simple cyst without accompanying a macroscopic nodule (5%). Type 2 is a cystic lesion with an accompanying mural nodule (60%). Type 3 is solid type tumors (26%). Type 4 is solid type tumors containing small internal cysts (%9).

HBs are capillary-rich and benign neoplasms of the CNS. But there is no systemic chemotherapy treatment protocol available for these patients except for surgery (5). Surgical excision is regarded as the treatment of choice for these tumors, but accurate timing and requirement should be carefully scrutinized (3). Follow-up optimal imaging timing of these patients should also be well established (6).

We can thus conclude that, in the evaluation of VHL patients, whole spinal axis imaging in conjunction with routine MR studies must always be performed in order not to overlook possible accompanying spine hemangioblastomas.

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