

Von Hippel Lindau Disease Presenting as Cervical Compressive Myelopathy

Sir,

A 20-year-old boy, product of non-consanguineous marriage, presented with insidious onset, progressive, sensori-motor quadriparesis (lower limbs followed by upper limbs) with bowel and bladder incontinence over 3 months. There was no history of radicular pain or constitutional symptoms. He was previously operated twice for recurrent pheochromocytoma (left suprarenal (2015) followed by right suprarenal (2018)) and was on replacement doses of hydrocortisone and fludrocortisone, with history of recent drug default. Family history was

significant for the demise of his father at the age of 45 years from the complications of a spinal tumor.

Examination revealed a systolic blood pressure of 80 mm of Hg. He was poorly nourished with pressure sores over the gluteal region and wasting of small muscles of hand. Power in the upper limbs was modified research council (MRC) grade 3/5 and grade 0/5 in lower limbs with paresthesia below C7. Reflexes were normal and plantars were not elicitable.

Magnetic resonance imaging (MRI) spine revealed an iso- to hyperintense intramedullary lesion with multiple intradural flow voids and homogenous contrast enhancement suggestive of a cervical hemangioblastoma [Figure 1]. MRI brain was within normal limits. Fundus and sonogram of the abdomen were normal.

The patient was clinically diagnosed to have vHL disease based on the Melmon and Rosen criteria,^[1] and advised surgery for his compressive cervical myelopathy. However, the patient refused for the same and was managed conservatively. Genetic study for vHL gene could not be done because of financial constraints. The patient's father also probably died of complications from a spinal hemangioblastoma, but no documents were available to ascertain the same.

Von Hippel Lindau (vHL) disease is an autosomal dominant, familial cancer syndrome with a prevalence of between 1:31,000 and 1:85,000.^[1-5] Its hallmarks are central nervous system (CNS) hemangioblastomas, retinal hemangiomas, and renal cell carcinomas (clear cell type).^[6] Cerebellum and spinal cord are the commonly involved sites in the CNS. Definitive diagnosis requires the demonstration of pathogenic mutations in the vHL gene on chromosome 3p25-26.^[7] However, clinical diagnosis can be made on the basis of the clinical diagnostic criteria proposed by Melmon and Rosen.^[8] Individuals with a positive family history of retinal or CNS hemangioblastoma require just one hemangioblastoma or a visceral lesion (phaeochromocytoma, pancreatic cyst/tumor, renal tumors, or papillary cystadenomas of epididymis) for diagnosing vHL. Individuals without a family history require either two or more hemangioblastomas or one hemangioblastoma and a visceral manifestation for vHL diagnosis.

Although intra- and inter-familial phenotypic variability is common among vHL patients, penetrance is age-related and almost complete by the seventh decade.^[2,3] The patients

may initially be asymptomatic for vHL manifestations but eventually have either a loss of function or malignant transformation of the affected organ.^[6] Therefore, annual ophthalmic, abdominal, and CNS examinations are imperative for diagnosing and treating these early, once a diagnosis of vHL has been made.^[9]

Spinal hemangioblastomas account for about 10% of spinal cord tumors.^[10] Majority (80%) are associated with vHL, especially in young patients. Pheochromocytomas occur in 7–18% of vHL patients.^[11] Spinal hemangioblastomas may be intramedullary, extramedullary, or both and are best imaged using contrast enhanced MRI sequences.^[12] These are histopathologically benign neoplasms with low oncogenic mortality, and neither invade nor metastasize.^[10] However, they cause significant morbidity because of their anatomical location leading to symptoms through pressure effect on adjacent structures or through hemorrhage. Gross total excision is the most common treatment employed. The other modalities include stereotactic radiosurgery and angioembolization.^[10] Surgery was offered to our patient, but refused.

Annual CNS examinations in addition to the recommended ophthalmic and abdominal examinations may facilitate early detection and improve outcomes in patients with vHL.^[9] It is also imperative to suspect a diagnosis of vHL in patients presenting with bilateral or recurrent pheochromocytomas and multiple renal cysts in order to make an early diagnosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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Figure 1: Sagittal (a) and axial (b) T2 -WIs show iso-hyperintense intramedullary lesion (arrows in a & c) on ventral aspect of spinal cord at C5-7 levels with multiple intradural flow-voids (arrowhead in a). The mass demonstrates homogeneous enhancement (arrows in b and d) in post-gadolinium sagittal (b) and axial (d) T1-WIs

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