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Case Study

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SPINAL HEMANGIOBLASTOMA AN UNCOMMON ENTITY: A CASE REPORT

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ABSTRACT

Spinal hemangioblastomas (HBs) are a rare, especially in the paediatric population. Spinal cord hemangioblastoma is an uncommon vascular neoplasm with a benign nature and is associated with Von Hippel-Lindau (VHL) disease in 20-30% of patients. Total removal of these tumors without significant neurological deficit remains a great challenge.

KEYWORDS: Spinal hemangioblastomas (HBs).

INTRODUCTION

Hemangioblastoma is a highly vascular tumors which can be found throughout the central nervous system. In the spinal cord, hemangioblastomas account for approximately 2% of the primary spinal cord tumors. They are classified as benign neoplasms (WHO Grade 1), but they can cause significant morbidity and mortality through mass effect. 70-80% of hemangioblastomas occurring in the spinal cord are usually sporadic isolated lesions and the other 20% may be associated with a rare genetic disorder known as Von Hippel-Lindau (VHL) disease.[1]

CASE REPORT

A 52-year-old female presented to our hospital with low backache with right sided radiation. On examination no gross neurological deficit was identified. Patient then underwent MRI lumbosacral spine which revealed an intradural mass lesion at level of L2/3, seen best on T1 fat saturated sequence with neural compression (Figure 1). Patient was explained all the possible risks and benefits of surgery, and once she agreed, she underwent a L2/3/4 laminectomy and excision of the intradural lesion in piecemeal fashion. The lesion was very vascular on gross examination. Postoperative period was uneventful, and patient was discharged home. She is now on regular OPD follow up. Histopathology revealed lesion to be a hemangioblastoma (Figure 2). Patients further clinical and radiological examination did not reveal any other lesion to qualify her for a Von Hippel Lindau disease diagnosis.

DISCUSSION

Hemangioblastoma most commonly occur as sporadic lesions, however Von Hippel-Lindau (VHL) disease denotes an important subset of patients, who present with hemangioblastomas throughout the central nervous system (CNS). Multiple hemangioblastomas in the retina, cerebellum, brain stem and spinal cord are usually associated with VHL disease. The basis of familial inheritance of VHL disease is a germline mutation in the VHL tumor suppressor gene, which is located on the short arm of chromosome 3p25.[2] Magnetic resonance (MR) imaging is valuable in the diagnosis of hemangioblastomas of the spinal cord. On T1-weighted MR imaging with contrast enhancement, the tumors appear as bright enhancing lesions and are usually located on the dorsal surface of the spinal cord. On T2-weighted image, may highlight associated oedema and syrinx. Spinal cord hemangioblastomas usually arise in the region of the dorsal roots or at the dorsal root entry zone.[3]

In both sporadic and VHL associated hemangioblastomas, allelic loss or mutations of the VHL gene are found in the stromal cells. VHL is a tumor suppressor gene that negatively regulates the hypoxia inducible factor (HIF1 Loss or mutation of VHL gene causes accumulation of HIF1 in the stromal cells, triggering increased transcription HIF1 regulated genes (such as VEGF, erythropoietin) leading to increase the activities of hematopoietic stem cells, endothelial cells and myeloid cells. These play an important role in the development of VHL disease associated neoplasms.[4]

The histological hallmark feature of hemangioblastomas is their biphasic tissue composition comprising neoplastic stromal cells and non-neoplastic dense capillary networks. The neoplastic stromal cells are large and polygonal in shape and have clear vacuolated cytoplasm (full of lipid vacuoles), with round or oval nuclei and prominent nucleolus. The neoplasm's highly vascular nature is due to the dense capillary network. Immunohistochemical stains help in to differentiating hemangioblastomas from other central nervous system (CNS) tumors. The neoplastic stromal are usually immunopositive for alpha-inhibin, S100 protein, D2-40 and vimentin.[5]

A comparison study of recurrence and surgical outcome of spinal hemangioblastoma in sporadic and Von Hippel Lindau disease, done in 2023, revealed that a gross total resection may sufficiently decrease tumor recurrence, especially in patients with VHL associated hemangioblastoma. This study also revealed that in sporadic hemangioblastoma, postoperatively there is usually good functional improvement, and the long-term functional prognosis is favorable.[6]

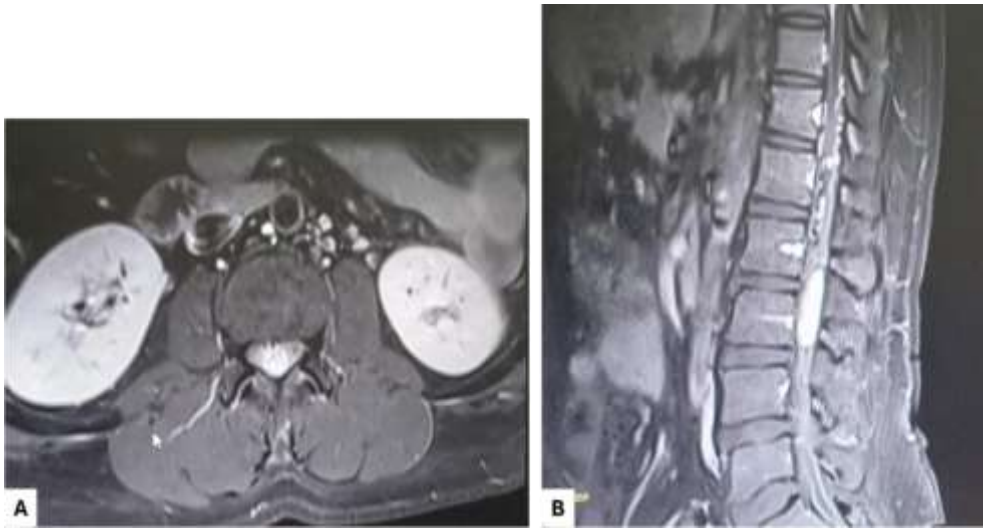

Figure 1: Axial and sagittal MRI Lumbar spine revealing the hemangioblastoma.

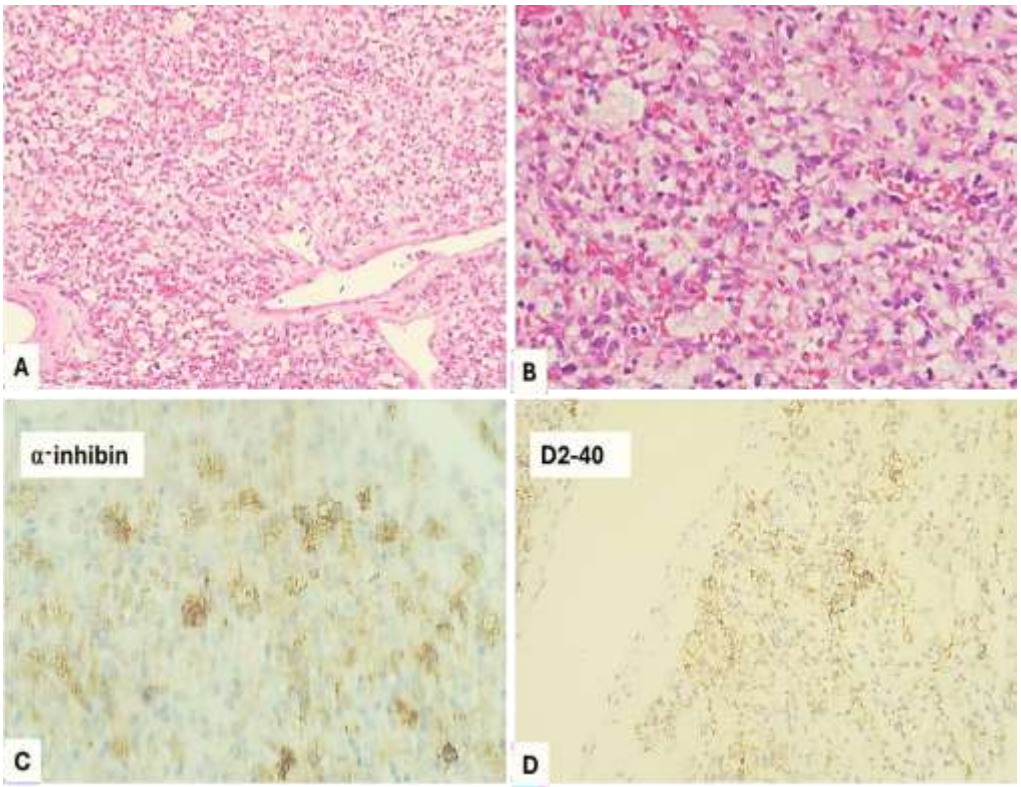

Figure 2: Histological picture of the spinal hemangioblastoma with neoplastic foamy stromal cells and dense non-neoplastic capillary network (A, B). Neoplasm is immunopositive for inhibin and D2-40 (C, D).

CONCLUSION

VHL-associated spinal hemangioblastomas differ from sporadic tumors in terms of age, presenting symptoms, multifocality, and rate of recurrence. Recurrences and new tumorigenesis can occur, especially in VHL patients, indicating the need for life-long follow up for VHL patients.

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