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Pathology

Spinal Hemangioblastoma – A Rare Entity

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Abstract | **Case Report**

Spinal Hemangioblastomas are rare benign vascular tumors of the central nervous system presenting in adults and are seen in association with VHL gene mutation. MRI is the preliminary diagnostic modality whereas histopathological diagnosis after complete surgical resection of tumor remains a confirmatory modality. Here we present an extremely rare case of a 2 year old female child with complaints of lower limb weakness who was diagnosed histopathologically with spinal hemangioblastoma.

Keywords: Spinal Hemangioblastoma, CNS Tumors, Benign Vascular Tumor.

INTRODUCTION

Hemangioblastomas are benign, highly vascularized tumors of the central nervous system [1]. Spinal hemangioblastoma accounts for 2–10% of intramedullary spinal cord tumors [2]. An estimated 10–40 per-cent are associated with a germline mutation of the Von-Hippel Lindau (VHL) tumor suppressor gene. These tumors can contain endothelial cells, pericytes, and stromal cells [1].

MRI has increased the diagnostic accuracy, and patients with spinal hemangioblastoma often have rapid progression of symptoms [2]. Slow progression is associated with delayed symptoms, however, for the symptomatic patient, surgical resection is the preferred option, with accompanying pathological analysis [1].

CASE REPORT

We present a case of a 2 year old female child who presented to the OPD at KIMSDU, Karad with complaints of weakness in bilateral lower limbs which was associated with bowel and bladder disturbances. There was no history of any loss of consciousness. No other past medical and surgical illnesses were present. Her vitals were stable and general examination revealed no abnormalities. CNS examination revealed loss of power in lower limbs. Laboratory findings were within normal limits. The MRI report of her dorso-lumbar spine showed a well-defined oblong intramedullary lesion involving the conus medullaris extending from D12 to L2 causing focal expansion of canal with areas of hemorrhages within. Minimally invasive spinal surgery was conducted and specimens were sent to the department of histopathology, KIMSDU, Karad.

GROSS EXAMINATION

We received six, tiny, grey white to grey brown, soft, friable tissue bits with foci of hemorrhages. The largest tissue bit measured 0.8 x 0.6 x 0.4 cm and the smallest tissue bit was tiny. Tissue was fixed in formalin and submitted entirely for processing.

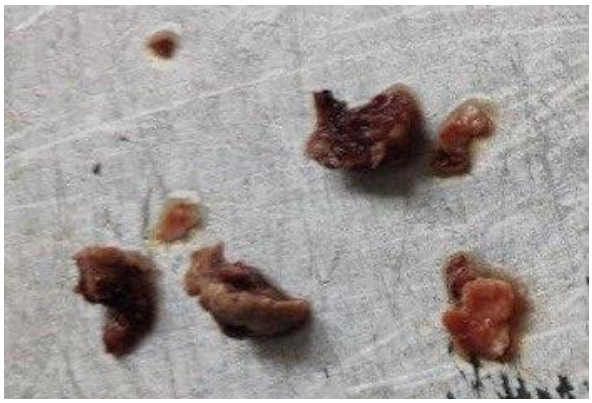

Figure 1: Gross examination of spinal hemangioblastoma biopsy

Figure 2: Tissue section mount of hemangiblastoma biopsy

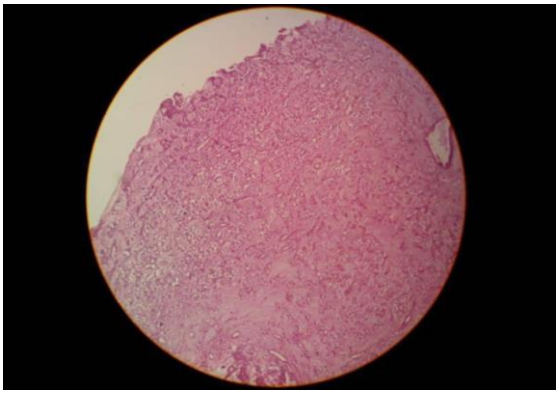

Figure 3: Scanner view of spinal hemangioblastoma

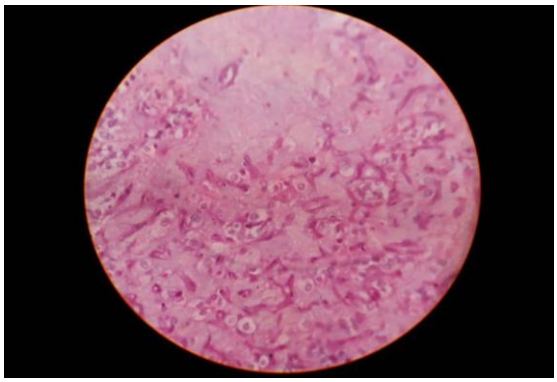

Figure 4: Low power view of spinal hemangioblastoma

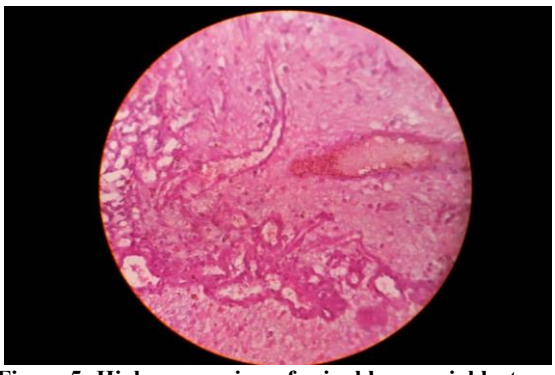

Figure 5: High power view of spinal hemangioblastoma

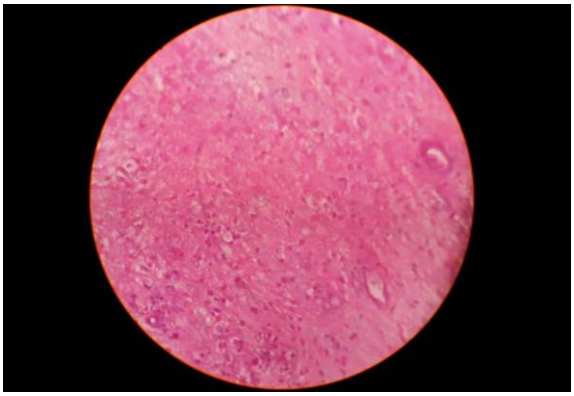

Figure 6: Areas of necrosis with inflammatory cell infiltration

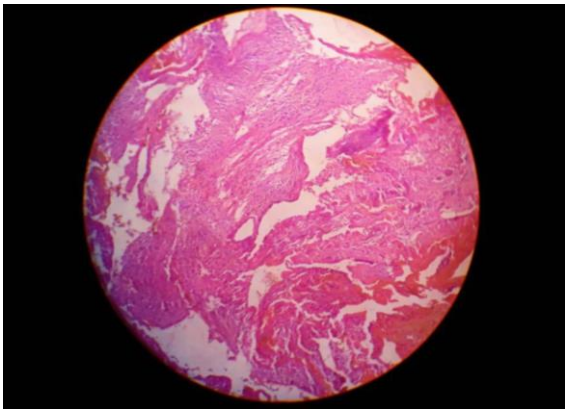

Figure 7: Areas of hemorrhage

MICROSCOPY

Microscopy revealed benign neoplastic stromal cells arranged compactly amongst numerous blood vessels. Individual cells showed round to polygonal morphology with mild nuclear pleomorphism and clear to pale foamy cytoplasm. Rare mitotic figures were noted. The vessel walls were demarcated with tumor cells. Areas of necrosis with inflammatory infiltrate as well foci of hemorrhages were noted.

DISCUSSION

Hemangioblastomas are rare neoplasms, and classified as World Health Organization (WHO) grade I benign tumors [3]. Hemangioblastomas are highly vascular tumors of the central nervous system. They can occur anywhere in the central nervous system such as the brain, the optic nerve, retina and spinal cord [1]. Spinal hemangioblastoma is a rare disease that accounts for 2–15% of intramedullary spinal cord tumors. Spinal hemangioblastomas often occur as intramedullary tumors that progress from under the pia mater to the intramedullary region. Thus, intramedullary tumor location is relatively frequent. However, in rare locations, tumors that have progressed from intramedullary to extramedullary region, intradural-extramedullary tumors derived from nerve roots, and few extramedullary tumors have also been mentioned [3].

Many of these cases of hemangioblastomas are sporadic, but around 10–40 percent are associated with the germline mutation of the Von-Hippel Lindau (VHL) tumor suppressor gene [1]. Von-Hippel Lindau disease is a rare, autosomal-dominant condition characterized by retinal capillary hemangiomas, pancreas and kidney cysts, or benign renal tumors with an increased risk for malignant transformation into carcinomas. The VHL/Von-Hippel Lindau gene, a tumor suppressor gene mutation, is located in chromosome 3p25-26, which is detectable in some sporadic hemangioblastomas. Approximately 20% to 45% of patients also suffer from VHL disease, which causes significant neurological decline and death, and the natural history and optimal management for which are not understood. They frequently occur in young adults of age 30 to 40 years, with some patients having a positive family history. Occurrence in pediatric group is extremely rare. The early and most common signs and symptoms include pain and sensory dysfunction. Other signs include myelopathy, hyperreflexia, scoliosis, weakness, and incontinence [3].

The increased use of MRI has simplified preoperative diagnostic imaging of intramedullary hemangioblastomas [2]. These tumors can contain endothelial cells, pericytes, and stromal cells [1]. They are all considered to be embryologically arrested mesodermal hemangioblasts composed of stromal and endothelial cells and mast cells [3]. The benign tumor cells are arranged compactly within the vascular component of blood cells. Neoplastic cells are stromal cells with round to polygonal morphology exhibiting mild nuclear atypia and clear, pale, foamy cytoplasm. Occasional hyaline cytoplasm can also be seen. Mitotic figures are rare or absent. Abundant areas of hemorrhages are noted. Tumor necrosis and inflammatory cell infiltrate are also noted.

Surgery follows a two-fold rationale. Firstly, patients who have primary spinal cord tumors and undergo complete surgical resection of the tumor have acceptable rates of neurological improvements. Secondly, surgery allows for the direct acquisition of tissue to confirm the histopathological diagnosis of hemangioblastoma [1]. A histopathologist thus plays a vital role to determine the further consequences of the disease.

In sporadic cases, a good prognosis may be expected after successful surgery, as systemic comorbidities do not exist. Patients with VHL disease have comparatively worse outcomes after surgery due to the systemic nature of the disease. With a 6% to 20% recurrence rate in the literature, longer and continued follow-up is needed in all of these series to determine the true recurrence rate of sporadic Spinal Hemangioblastomas and long-term outcomes.

CONCLUSION

Spinal hemangioblastomas are rare, benign, vascular tumors of the Central Nervous System classified as WHO Grade I tumors. They are either sporadic, showing slow progression of disease with relatively less or no symptoms, or are associated with VHL gene mutation associated with the Von-Hippel Lindau disease, that show rapid progression and have poorer outcomes. Surgical resection remains treatment of choice and histopathological diagnosis to confirm as benign neoplastic is vital to rule out malignancy or a non-neoplastic etiology.

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