Cerebellar Hemangioblastoma- A Case Report

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ABSTRACT

Cerebellar hemangioblastomas are rare neoplasms of cerebellum with uncertain histogenesis classified under WHO grade I category. Most of these lesions are sporadic and few of them though rare, are associated with von Hippel-Lindau disease (VHL). We report a case of cerebellar hemangioblastoma in a 31 years old male who presented with headache and vomiting. MRI showed a cystic lesion in the cerebellum. Intraoperative squash cytology was suggestive of a vascular neoplasm. Histopathological examination of the tumor was done and a diagnosis of cerebellar hemangioblastoma was given. Immunohistochemistry using NSE and CD31 revealed positivity and confirmed the diagnosis. The clinical radiological and histopathological and immunohistochemical findings were discussed.

KEYWORDS: Cerebellar Hemangioblastoma, immunohistochemistry, Von Hippel-Lindau disease.

INTRODUCTION

Hemangioblastomas are uncommon, slow growing tumors of the central nervous system of adults, which most commonly occur in the cerebellum, brain stem or spinal cord. Hemangioblastomas are benign neoplasms of the central nervous system that account for 1-2% of primary brain tumors.[1]These tumors arise from excess growth of blood vessels in the brain. They account for approximately 4% of all spinal cord tumors and 7-10% of tumors arising in the posterior cranial fossa in adults. The tumors have a predilection for the posterior cranial fossa, commonly arising in the cerebellum.[2,3] They are seldom seen in the supratentorial compartment of the brain.[4]These tumors are more common in men.[5]

Approximately 20% of the hemangioblastomas occur in association with VHL syndrome while majority (80%) arise sporadically. Whether hemangioblastomas occur sporadically or as part of VHL syndrome, they are identical in clinical presentation, grossly and microscopically.

CASE REPORT

A 31 yrs old male presented with headache and vomitings since 3 months, followed by speech difficulty. There was no history of seizures. Patient had no relevant past or personal history or other co morbidities. MRI revealed a large well circumscribed cystic lesion in the right cerebellar hemisphere and vermis with a nodular densely enhancing component along its posterior wall with mass effect and cerebellar tonsillar herniation.

Intraoperative squash cytology was suggestive of a vascular neoplasm of cerebellum. The excised tumor was sent for histopathological examination. The tumor consisted of multiple gray brown soft tissue bits approximately measuring 3cc in volume. The entire tissue was submitted for processing.

Histopathological examination revealed a highly vascular tumor with stromal cells arranged in nests. Individual tumor cells were large with abundant vacuolated cytoplasm and centrally placed nuclei along with vascular channels of various caliber which were lined by endothelial cells. Areas of hemorrhage were noted. A diagnosis of cerebellar hemangioblastoma was given. Immunohistochemistry was done which confirmed the diagnosis. Stromal cells showed positivity for Neuron Specific Enolase and lacked the endothelial markers like CD31 which highlighted the capillary endothelial cells.
DISCUSSION

Hemangioblastomas are benign vascular tumors of the central nervous system composed of neoplastic stromal cells and blood vessels. According to WHO Classification of CNS Tumors- 2012, Cerebellar hemangioblastomas are categorized under grade 1 tumors of borderline or uncertain behavior that occur most commonly in the posterior cranial fossa. These tumors were also categorized as “other neoplasms related to meninges.”[6] These tumors account for 1-2% of primary intracranial tumors.

Although approximately 80% of hemangioblastomas appear to be sporadic, some of these may represent occult cases of VHL disease that can be detected if the patients are appropriately screened for germ line VHL mutations. In VHL syndrome the pVHL protein is dysfunctional due to mutation and/or gene splicing. In normal circumstances Pvh1 is involved in the inhibition of Hypoxia Inducible Factor 1α by ubiquitin mediated proteosomal degradation. In these dysfunctional cells pVHL cannot degrade HIF-1α causing it to accumulate. HIF-1α causes the production of Vascular endothelial growth factor, Platelet derived growth factor-B, Erythropoietin and Transforming growth factor-α, which act to stimulate growth of cells within the tumor.[7]

Hemangioblastomas may be purely cystic (5%), purely solid (26%), cyst with a mural nodule (60%) or solid tumor with internal cysts (9%).[8] The cyst fluid contains amino acids, nitrogen, mucoprotein and alkaline phosphatase similar to that of blood, suggesting that the cyst fluid arises by diffusion from the vascular component of the mural nodule.[9] Symptoms of cerebellar hemangioblastomas depend on tumor size and location.[10] Serial MRI studies of patients with hemangioblastomas have shown that cyst formation can arise from solid tumor.[11] The combined tumor and cyst growth rates and sizes are significant
predictors of symptom development for hemangioblastomas in the cerebellum.

In 1931, Lindau[12] hypothesized that these tumors may be derived from a congenital anlage and that the histological picture revealed an embryological type of tumor cells. Stein et al suggested an angiomesenchymal origin of hemangioblastomas.[13]

The histogenesis of cerebellar hemangioblastomas is uncertain. Tissue microdissection combined with deletion analysis of the VHL gene locus, have identified that the stromal and not the vascular cells are neoplastic.[14] Neural cell adhesion molecule NCAM/CD56 is consistently immunoreactive. Factor XIIIa has been reported to be expressed by stromal cells while other studies found it exclusively expressed by the reactive vascular component. Suggested origins of the tumor include glial cells, endothelial cells, arachnoid cells, embryonic choroid plexus, neuroendocrine cells etc.

The prognosis of hemangioblastomas is excellent if surgical resection can be achieved, which is often possible. Permanent neurological deficits are rare and can be avoided when they are diagnosed and treated early. Although the imaging findings of hemangioblastomas are well described, use of appropriate immunohistochemical stains is required to establish the correct diagnosis. Stromal cells of hemangioblastomas variably express neuron-specific enolase, vimentin and S-100 protein and do not react with endothelial cell markers like CD31 and usually do not express Glial Fibrillary Acidic Protein.

CONCLUSION

Cerebellar hemangioblastomas are rare benign lesions of CNS. Majority occur as sporadic cases without association with VHL syndrome. Many of these tumors are solid and show a cystic component. Radiological histopathological and immunohistochemical studies clinch the diagnosis.

REFERENCES


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