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Case Study

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INTRAMEDULLARY SPINAL GANGLIOGLIOMA WITH ANAPLASIA AND *KIAA1549-BRAF* FUSION, AN UNCOMMON ENTITY: A CASE REPORT

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ABSTRACT

Ganglioglioma is a rare, slow-growing primary central nervous system (CNS) tumor which most frequently occurs in the temporal lobes, typically resulting in seizures. Ganglioglioma of the spinal cord is exceedingly rare, only approximately 178 cases of spinal cord ganglioglioma have been reported, mostly in children or young adults. The gold Standard of treatment is gross total resection; however, the optimal treatment in the setting of subtotal resection has not been clearly established. We report an unusual case of an intramedullary cervical spinal cord ganglioglioma with anaplasia and KIAA1549-BRAF fusion in a 4-year-old boy who underwent subtotal resection followed by adjuvant radiotherapy.

KEYWORDS: Ganglioglioma with anaplasia, KIAA1549-BRAF fusion.

INTRODUCTION

Ganglioglioma was first introduced as a CNS neoplasm by Ewing in 1928. Ganglioglioma of the spinal cord is exceedingly rare, only approximately 178 cases of spinal cord ganglioglioma have been reported.[1] Only 1.1% of all spinal tumors show this pathology. Gangliogliomas most commonly occur in the temporal lobe and cerebellum. In the spinal cord there is a predilection for the cervical region. Children are more commonly affected than adults. No gender predominance is seen for spinal tumors. The most common symptoms for spinal tumors are back pain and limb weakness. Spinal lesions are found earlier than cerebral lesions.[2]

CASE REPORT

A 4-year-old male child reported to our hospital with history of torticollis and left-hand weakness of 2-month duration. There were no other deficits noted. MRI spine revealed an intramedullary tumor extending from C3 cervical segment to C7 cervical segment, with surrounding syrinx. All possible risks and benefits of surgery were discussed with family and once agreed, patient underwent C1 to T1 laminoplasty and decompression of the intramedullary tumor under neuromonitoring. The postoperative period was uneventful. Histopathology revealed the lesion to be a Ganglioglioma with anaplasia and molecular testing confirmed the presence of KIAA1549-BRAF fusion. As total resection of tumor was not done, the child was referred to radiation oncology, and he was irradiated. Child is currently under follow up in OPD and is doing fine, with mild left sided weakness and walking with a limp.

DISCUSSION

Spinal ganglioglioma are rare and literature only provides 22 cases of ganglioglioma with anaplasia in the spine, with thoracic spine most frequently involved area (14/22), followed by the cervical (6/22) and lumbar (5/22) spine.[3]

Radiological characteristics are not pathognomonic. MRI is the diagnostic modality of choice. Ganglioglioma might be hyperintense, isointense, hypointense or heterogeneous on T1; it is mostly hyperintense on T2 and mostly enhances heterogeneously with contrast.[4]

Ganglioglioma with anaplasia ("anaplastic ganglioglioma") is a glioneuronal tumor composed of admixture of neoplastic ganglion and glial cells, with increased mitotic activity, high Ki67 proliferation index, necrosis and microvascular proliferation. As these are rare, further studies are needed to establish their diagnostic criteria. Most common genetic aberrations in ganglioglioma is the BRAF p.V600E mutation, occurring in 10-60%, mostly cortical tumors and less in spinal tumors. BRAF gene fusions are seen in gangliogliomas lacking BRAF mutations, most commonly with KIAA1549 as fusion partner, seen especially in spinal tumors.[5]

Total surgical resection remains the standard treatment. Preoperative radiological presumption of ganglioglioma is of clinical importance. Usually, more than eight segments of the spinal cord are involved, and its length is two times that of astrocytoma and ependymoma.[4] Holospinal involvement, scoliosis and bone remodeling, mixed intensity on T1, significant cysts or syrinx, absence of oedema and enhancement within the tumor center, hemosiderin or calcification and young age help in diagnosis.[4] The higher intensity of fluid content of cysts, irregular margins with soft tissue compartment and gadolinium enhancement help in differentiating them from syrinx.

Total surgical resection is the treatment of choice for ganglioglioma. Early surgical intervention follows two main advantages: first, early surgery leads to better functional status, and second, easier resection due to smaller size. Some argue that complete resection might not be feasible because no well-defined plane of cleavage can be identified. It is believed that spinal lesions carry a high morbidity rate even with microsurgical techniques. Surgical outcomes may include recovery, no change in neurological status, or progression of neurological deficit.[4] The recurrence is more common in spinal gangliogliomas than in their cerebral counterparts.[4]

Close follow-up is necessary. It is advised that patients be seen every 6 months for 2 years postoperatively, and annually thereafter.[4]

Radiotherapy is usually not suggested. The reasons to not do radiotherapy include the risk of differentiation of the astrocytic component into glioblastoma, the probability of irradiation necrosis, and increase neurological compromise.

Radiotherapy is considered for cases in which the radical resection of tumor is not feasible, when progression is evident and for malignant histopathology.[6] The role of chemotherapy is still under debate.[7] For recurrent gangliogliomas, a second surgery should be considered, however radiotherapy appears to be a safe and effective treatment option.[6]

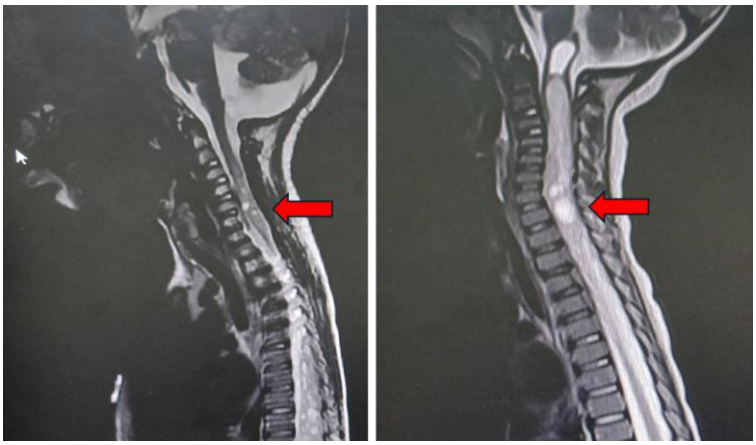

Figure 1: MRI. Intramedullary cervical spinal ganglioglioma extending from C3 cervical segment to C7 cervical segment, with surrounding syrinx.

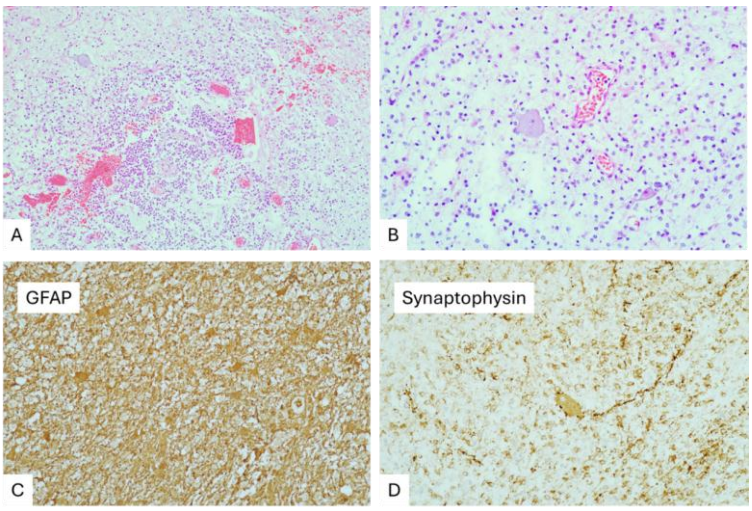

Figure 2: Ganglioglioma with anaplasia ("anaplastic ganglioglioma"). A-B. Glioneuronal tumor composed of admixture of neoplastic ganglion and glial cells (H&E). C. GFAP immunostain highlighting the neoplastic glial component. D. Synaptophysin immunostain highlighting the neoplastic ganglion cell.

CONCLUSION

Ganglioglioma must be considered in the differential diagnosis of tumors affecting the spinal cord. In cases of suspected spinal ganglioglioma showing no sharp delineation from the surrounding tissue, a subtotal tumor removal should be considered to prevent severe neurological deficits.

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