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CASE REPORT

Suprasellar Desmoplastic Infantile Astrocytoma: A Rare Case Report

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Abstract

Desmoplastic infantile astrocytoma (DIA) is a rare primary brain tumour representing 0.4% of all brain tumours and 1.25% of intracranial tumors in children. Here we present a case of 1-year-old child with complaints of irritability for 3 months and recurrent vomiting on food intake for 2 months. MRI showed a large suprasellar mass showing T1-hypointensity, T2/FLAIR-hyperintensity and intense enhancement post-contrast administration. The patient underwent frontal craniotomy and excision of the space occupying lesion. Histological examination revealed DIA, immunopositive for Glial Fibrillary Acidic Protein(GFAP). Here we present this case due to its rarity.

Keywords

Desmoplastic infantile astrocytoma (DIA), Suprasellar mass, Glial Fibrillary Acidic Protein (GFAP)

Introduction

Desmoplastic Infantile Astrocytoma (DIA) is a rare biphasic tumor representing 0.4 % of all brain tumors, 1.25% of intracranial tumors in children.[1,2] Desmoplastic infantile astrocytoma (DIA) is a rare primary brain tumor seen commonly in children before the age of 24 months.[3] It is a benign neoplasm often occurring due to *BRAF* V600E mutation.[3] They typically arise from the cerebral hemispheres, but apresentation in the suprasellar location is very rare.[4] It has a glioneuronal counterpart called the desmoplastic infantile ganglioglioma (DIG), both of which are considered as CNS Grade 1 lesions.[3,5,6] Radiologically they are often large solid and cystic tumors which are located superficially.[3] It usually has a favorable prognosis, post neurosurgical excision of the space occupying lesion in the brain.[3]This case report on desmoplastic infantile astrocytoma is a rare central nervous tumor of infancy. The unusual presentation at the suprasellar region is considered very rare and it discusses the importance of including this entity in the differential diagnosis of paediatric central nervous system malignancies.

Case Report

A 1-year-old child had come to the out patient department with complaints of having irritabilitysince 3 months and recurrent vomiting on food intake for 2 months. MRI brain was done which showed a large suprasellar mass showing predominant T1 hypointensity, T2/ FLAIR hyperintensity and intense enhancement post contrast administration. The patient underwent frontal craniotomy and excision of the space occupying lesion.We at the pathology department, had received the tumour tissue in formalin solution.Macroscopically, there were multiple grey white soft tissue fragments on aggregate measuring 0.8x0.6 cm which were all embedded and processed. Histopathological examination revealed, a neoplasm composed of predominantly spindled cells with

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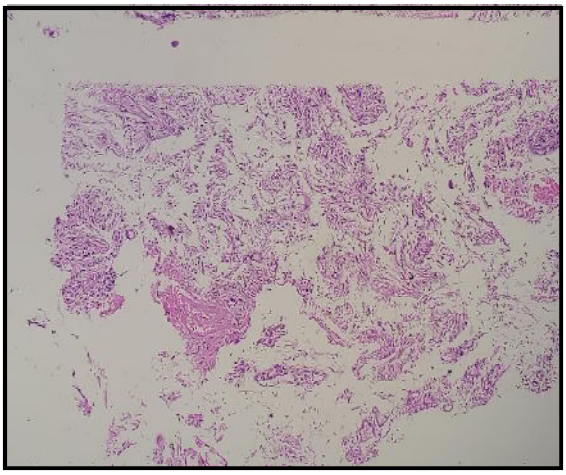

Fig 1 shows Histopathological examination-Low power view of DIA

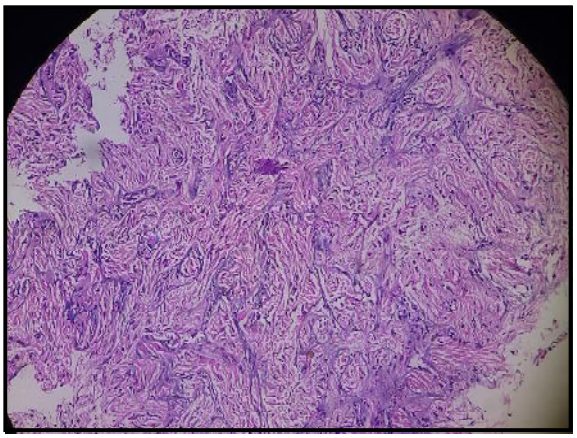

Fig 2 shows Histopathological examination-High power view of DIA

scant eosinophilic cytoplasm and uniform round to oval nuclei in a background of abundant fibrillary material and few intermixed capillary sized blood vessels. No mitosis, nuclear pleomorphism and necrosis was noted. It was further found to be immunopositive for Glial Fibrillary Acidic Protein (GFAP).

Discussion

Desmoplastic Infantile Astrocytoma (DIA) is a rare benign tumor of the central nervous system characterized by the presence of neuroglial cells in a desmoplastic stroma.[7] It is a biphasic tumor, with - a desmoplastic leptomeningeal component, and a neuroepithelial component.[8] They represent 0.4 % of all brain tumors and 1.25% of intracranial tumors in children and 1.3-15.8% of the brain tumors in infants.[8] Patient commonly present with enlarging head circumference with tense

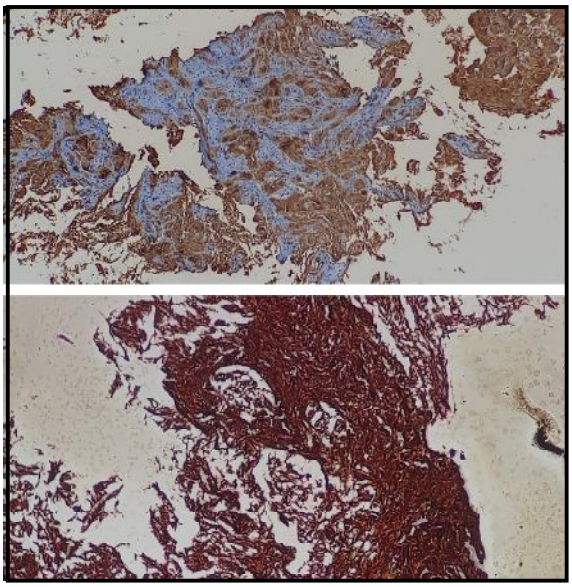

Fig 3- A and B shows Immunohistochemistry-Immunopositive for Glial Fibrillary Acidic Protein (GFAP)

bulging fontanelles.[4] They typically arise from superficial cerebral hemispheres and leptomeninges and were previously called 'superficial cerebral astrocytomas attached to dura' and 'desmoplastic cerebral astrocytoma of infancy'.[4,7] Their presentation in the suprasellar location is considered very rare.[4] It has a glioneuronal counterpart called the desmoplastic infantile ganglioglioma (DIG), both of which are classified as Grade 1 lesions of the central nervous system.[5] DIG differs from DIA, by the presence of a neuronal component in DIG.[4]It is thought that these tumors originate from immature cells that are capable of divergent differentiation along the astrocytic and neuronal lines, and are also capable of causing a desmoplastic reaction in the leptomeninges adjacent to it.[4] Male children are commonly affected, however, in our case, patient was a female child.[5] DIAS are IDH- and histone H3-wildtype tumours with activation of the MAPK signalling pathway, via mutation or fusion involving BRAF or RAF1 mutations and lack the CDKN2A and/or CDKN2B homozygous deletion.[8]

Radiologically they are large superficially positioned supratentorial lesions and are seen as brightly contrast-enhancing masses with a subjacent, uni- or multiloculated cyst.[6] Histological examination reveals collagen- and reticulin-rich desmoplastic regions populated by plump spindled cells in loose fascicular or storiform array.[6] In DIA, the neuroepithelial component comprises an

neoplastic astrocytic population.[6] DIG shows a neoplastic ganglion cell component in addition.[7] These tumors characteristically show a poorly differentiated neuroepithelial component, composed of small embryonal cells seen within a reticulin-free fibrillar matrix.[6] Multicystic or multinodular cortical component without desmoplasia may be seen. Mitosis is generally absent.[6] Calcifications are common but perivascular mononuclear inflammatory infiltrates, necrosis, glomeruloid microvascular proliferation and xanthomatous cells are absent.[8] Immunopositivity for GFAP indicates glial astrocytic elements.[8]

Their differential diagnosis includes ganglioglioma, pleomorphic xanthoastrocytoma and infant type hemispheric glioma.[8]

Desmoplastic Infantile Astrocytoma has excellent prognosis with surgical resection being the recommended therapeutic option as most tumors are rendered stable or regrow slowly.[3] They can undergo spontaneous regression as well as malignant transformation and hence it requires long term follow up.[9] Suprasellar, multifocal lesions and leptomeningeal spread are associated with poor prognosis.[4,5,10] It is also imperative to include this in the differential diagnosis of CNS tumors in infants.

Conclusion

Desmoplastic Infantile Astrocytoma has excellent prognosis with surgical resection being the recommended therapeutic option. They can undergo spontaneous regression as well as malignant transformation and hence it requires long term follow up. Suprasellar, multifocal lesions and leptomeningeal spread are associated with poor prognosis. Hence it is important to include it in the differential diagnosis of CNS tumors in infants.

The authors certify that they have obtained all appropriate patient consent forms. The presented patient had given their written informed consent, for publication of data and images of this patient.

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