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TECTAL MESENCHYMAL TUMOUR OF UNCERTAIN DIFFERENTIATION - A RARE CASE REPORT

Dr Venkatesan Sanjeevi

Dr. Chandan Y S

ABSTRACT Tectal mesenchymal tumour of uncertain differentiation is very rare tumour in tectal plate region. We report a 48 years old male presented with complaints of double vision, hard of hearing and drooping of eyelids. On examination patient had partial ptosis of both eyelids, downbeat nystagmus, convergence retraction nystagmus, upward gaze palsy and hard of hearing on both side. MRI of brain with contrast showed Heterogeneously enhancing lesion with poorly defined margins occupying the central portion and left side of the tectum of the midbrain. Patient underwent midline sub occipital craniotomy, supra cerebellar-infratentorial approach and complete excision of tumour. Histopathological examination and correlating with immunohistochemistry patchy EMA positive, Vimentin , SMA, BCL2, FLI-1 and patchy TLE1 positive-suggestive of Tectal Intracranial Mesenchymal tumour of Uncertain differentiation. Further Molecular study- soft tissue sarcoma panel including CIC fusion, SS18-SSX1 and SS18-SSX2 fusion by Next Generation Sequencing (NGS) is advised to study in detail about the intracranial mesenchymal tumour of uncertain differentiation.

KEYWORDS : Tectal mesenchymal tumour, Diplopia, Nystagmus, Immunohistochemistry

INTRODUCTION

Tectal plate is a rare location for a tumour. Different types of pathology arising in that location including tumours, vascular lesions, inflammatory and infectious processes. Among the tumours found in tectal plate, the most common is Astrocytoma,(1) but the other types have been described, such as oligodendroglioma, ependymoma, ganglioglioma, medulloblastoma, primitive neuroectodermal tumours, metastasis, lipoma, melanoma, dysembryoplastic neuroepithelial tumour (DNET), cavernoma, abscess and periaqueductal gliosis. Most of the patients presents with features of Parinaud syndrome includes paralysis of upward gaze, Pseudo-Argyll Robertson pupils, Convergence-retraction nystagmus and Eyelid retraction (Collier's sign) due to damage to the tectal plate or posterior commissure of midbrain.(2) In some patient may develop non communicating or obstructive hydrocephalus due to narrowing or obstruction of cerebral aqueduct.(3)(4) Along with clinical examination Radiological imaging plays a key role in diagnosis and helps in plan of surgical approaches. Either biopsy, partial or complete excision of the lesion based on the different pathology. Histopathological examination and immunohistochemistry plays a vital role in differentiating the various tumours, in addition to that advanced Molecular study- Next generation sequencing (NGS) helps in diagnosing tumours of uncertain differentiation. The prognosis is based on the underlying pathology, in some patients may require adjuvant chemotherapy, radiotherapy or both.

Case Presentation

A 48 years old male presented with complaints of double vision, hard of hearing and drooping of eyelids for 3 months duration, gradually progressing in nature. On examination patient had partial ptosis of both eyelids, downbeat nystagmus, convergence retraction nystagmus, upward gaze palsy and hard of hearing on both side. MRI of brain with contrast, multivoxel spectroscopy and tractography was done, which showed T2/FLAIR hyper intense lesion with ill-defined margins occupying the central portion and left side of the tectum of the midbrain. Lesion appears hypo intense on the T1-weighted images. No diffusion restriction. Heterogeneous enhancement of the lesion is observed with intense enhancement noted in the central portion of the lesion. The lesion encircles the aqueduct. Ill-defined non-enhancing T2 flair hyper intensity noted in the superior cerebellar peduncle on the left side possibly due to oedema. The cranial margin of the lesion is ill-defined and appears to extend to the mesencephalon-diencephalon junction. Multivoxel spectroscopy shows elevated choline and decreased NAA with abnormal Cho/NAA and Cho/Cr ratios in the lesion . Diffusion tensor imaging and tractography done.(Figure 1) The superior and inferior cerebellar peduncles appear symmetric on both sides with symmetric fractional anisotropy values- reported as Heterogeneously enhancing lesion with poorly defined margins occupying the central portion and left side of the tectum of the midbrain, lesion demonstrates elevated choline peak with decreased NAA on the multi voxel spectroscopy -findings compatible with tectal glioma. Patient underwent midline sub occipital craniotomy, supra cerebellar-infratentorial approach and complete excision of tumour under Intraoperative neuromonitoring (IONM). Post op images showed near total excision.(Figure 2)

Histopathological examination was done showed multiple fragments of tissue few showing tumour cells and few showing normal brain parenchyma with reactive gliosis. Tumour cells are arranged in intersecting fascicles at places and sheets at places. Cells have epithelioid morphology at places with round nucleus and mild nuclear atypia with clear cytoplasm. Cells have spindled morphology at places. Intervening spaces have many hyalinized thin blood vessels and hyalinized collagen fibres. Mitotic activity is low 2 - 3 / 10hpf. No microvascular proliferation/necrosis seen.(Figure-3) Correlating with immunohistochemistry patchy EMA positive, Vimentin , SMA, BCL2, FLI-1 and patchy TLE1 positive-suggestive of Tectal Intracranial Mesenchymal tumour of Uncertain differentiation.(Table-1)(Figure 4) Advised Molecular study - soft tissue sarcoma panel including CIC fusion, SS18 - SSX1 and SS18 - SSX2 fusion by Next Generation Sequencing. (NGS).

DISCUSSION

Intracranial mesenchymal tumours are a rare type of neoplasm (0.3% of all soft tissue tumours). Mesenchymal tumours are most frequently localized in the subcutaneous tissue (typically in the limbs and hands) of young adults and have rarely been diagnosed in the central nervous system. The commonest location for the mesenchymal tumours are extra axial cerebral convexities, falx, lateral ventricles, tentorium, cerebellopontine angle and spinal cord. In fifth edition of the WHO Classification of Tumours of the Central Nervous System (CNS), published in 2021 classifies Mesenchymal soft tissue tumours of Uncertain differentiation into Intracranial mesenchymal tumour, (FET-CREB fusion-

positive), CIC-rearranged sarcoma, Primary intracranial sarcoma, (DICER1-mutant) and Ewing sarcoma. Tectal plate is a rare location for a tumour. Mesenchymal tumour in tectal plate region is very rare, not reported in literature so far. Among tumours found in the tectal plate, the most common is the astrocytoma, but other types have been described, such as oligodendroglioma, ependymoma, ganglioglioma, medulloblastoma, primitive neuroectodermal tumours, metastasis, as well as lipoma, melanoma, dysembryoplastic neuroepithelial tumour, cavernomas, abscess, and periaqueductal gliosis.(5) Tectal tumours may extrude from the tectum into the lumen of the cerebral aqueduct and subsequently protrude to the third ventricle, pushing away the posterior commissure and enlarging the orifice. Tectal distortion or thickening caused by a localized mass, leading to aqueductal compression and hydrocephalus.

Parinaud syndrome features includes paralysis of upward gaze, Pseudo-Argyll Robertson pupils, Convergence-retraction nystagmus and Eyelid retraction (Collier's sign) with or without non communicating hydrocephalus features will be typical presentation in case of tectal plate tumours. The radiologic investigation of these tumours have been up graded in the CT scan era with better visualization of the tectal region; but the majority of lesions continue to appear as noncommunicating hydrocephalus alone, although calcification or a hypo dense lesion on the tectal plate can be observed. MRI of these tumours reveals tectal distortion or thickening caused by a localized mass, leading to aqueductal compression and hydrocephalus; characteristic T1 hypo intensity and T2 hyper intensity with strong enhancement after gadolinium administration. MRI is an accurate and non-invasive method of diagnosis that can be indicated in all cases of late onset hydrocephalus and aqueductal obstruction, especially in adults. Even when present, contrast enhancement after gadolinium injection was an independent factor of tumour grading. In our case the lesion appears hypo intense on the T1-weighted images, hyper intense on T2/FLAIR - weighted images with ill-defined margins occupying the central portion and left side of the tectum of the midbrain, No evidence of haemorrhage /calcification within the lesion. Heterogeneous enhancement of the lesion is observed with intense enhancement noted in the central portion of the lesion.

The lesion encircles the aqueduct. Ill-defined non-enhancing T2 flair hyper intensity noted in the superior cerebellar peduncle on the left side possibly due to oedema. Multivoxel spectroscopy shows elevated choline and decreased NAA with abnormal Cho/NAA and Cho/Cr ratios in the lesion.(6) Surgery is the gold standard treatment; adjuvant radiation therapy and chemotherapy with sarcoma-based regimens have been used in rare cases when complete surgical excision was not recommended. In terms of prognosis, these tumours show a tendency for local relapse. Endoscopic third ventriculostomy (ETV) has been used in patients with tectal gliomas,(7)(8) both as an initial procedure and after shunt failures. Histopathological examination was done showed multiple fragments of tissue few showing tumour cells and few showing normal brain parenchyma with reactive gliosis. Tumour cells are arranged in intersecting fascicles at places and sheets at places. Cells have epithelioid morphology at places with round nucleus and mild nuclear atypia with clear cytoplasm. Cells have spindled morphology at places. Intervening spaces have many hyalinized thin blood vessels and hyalinized collagen fibres. Mitotic activity is low 2 - 3 / 10hpf. No micro vascular proliferation / necrosis seen. Correlating with immunohistochemistry patchy EMA positive, Vimentin , SMA, BCL2, FLI-1 and patchy TLE1 positive-suggestive of Tectal Intracranial Mesenchymal tumour of Uncertain differentiation. Advised Molecular study - soft tissue sarcoma panel including CIC fusion, SS18 - SSX1 and SS18 - SSX2 fusion by Next Generation Sequencing. (NGS) (9)(10)(11)

CONCLUSION

In conclusion, our case highlights the necessity and mandatory molecular studies workup for these rare diseases, which are crucial for refining personalized therapies and exploring novel treatment options.

Preoperative Magnetic Resonance Images

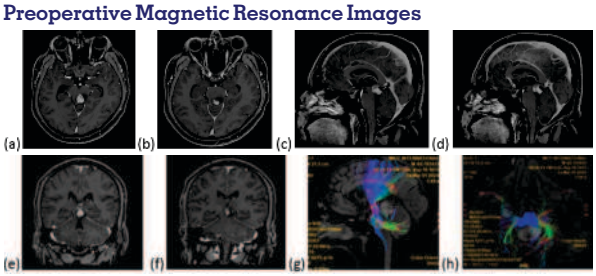

Figure 1:

Postoperative Magnetic resonance Images

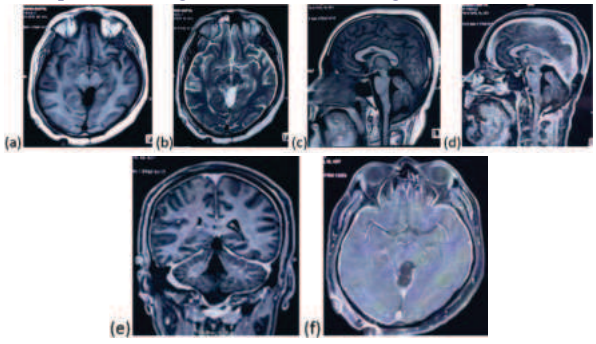

Figure 2:

Histopathological examination (H&E section-200x magnification)

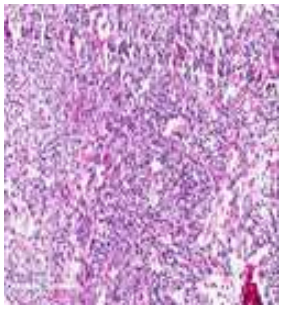

Figure 3:

Immunohistochemistry

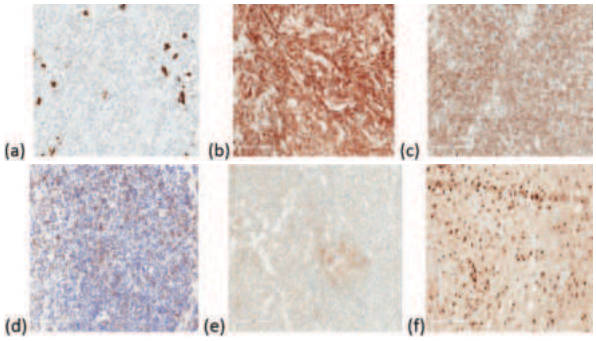

Figure 4:

Preopertive Magnetic resonance images

Figure 1: (a)(b). Axial section T1-weighted contrast, (c)(d). Sagittal T1-weighted contrast, (e)(f). Coronal T1-weighted contrast showing heterogeneously enhancing lesion with poorly defined margins occupying the central portion and left

side of the tectum of the midbrain. The cranial margin of the lesion is ill-defined and appears to extend to the mesencephalon-diencephalon junction. (g)(h). Diffusion tensor imaging and tractography showing Corticospinal tracts are well visualised with symmetric fractional anisotropy values on both sides. Corticospinal tracts are anterior to the lesion in the tectum of the midbrain. There is no distortion/destruction of the tracts. The superior and inferior cerebellar peduncles appear symmetric on both sides with symmetric fractional anisotropy values.

Postoperative Magnetic resonance images

Figure 2: (a). Axial section T1-weighted, (b). Axial section T2-weighted, (c). Sagittal T1-weighted, (d). Sagittal T1-contrast, (e). Coronal T1-contrast and (f). Axial T1-contrast images showing complete excision of Tectal tumour without any tumour residue with post operative changes.

Histopathological examination (H&E section-200x magnification)

Figure 3: Spindled tumour cells with bland morphology arranged in fascicles

Immunohistochemistry

Figure 4:

(a). GFAP negative in the tumour cells
(b). SMA positive in tumour cells
(c). FLI-1 nuclear positive in tumour cells
(d). TLE-1 nuclear positive in tumour cells
(e). EMA Patchy focal positive in tumour cells
(f). Ki67 highlighted the proliferation index

Table 1: Immunohistochemistry (IHC) Markers And Results

S.No	IHC markers	Result
1.	GFAP - Clone : EP13	Negative
2.	P53 - Clone : BP53-12	Scattered positive (wild type)
3.	IDH-1-Clone : R132H(H09)	Negative
4.	ATRX - Clone -D-S	Retained expression
5.	Ki67 - Clone - MIB-1	30%
6.	Olig.2 - Clone - EP112	Negative
7.	S100 - Clone - EP32	Negative
8.	EMA - Clone - E29	Positive in scattered tumour cells
9.	PR - Clone - 1E2	Negative
10.	Synaptophysin - Clone - SP11	Negative
11.	NeuN - Clone - PFOX3	Negative
12.	CD34 - Clone - QBEnd10	Negative
13.	Inhibin - A - Clone - R1	Negative
14.	CAIX - Clone - H11	Negative
15.	Vimentin - Clone - V9	Positive
16.	SMA - Clone - IA4	Positive
17.	INI -1- Clone - 25	Retained expression
18.	TLE -1- Clone - IFS	Patchily positive
19.	CD99 - Clone - EP8	Negative
20.	FLI -1- Clone - G148-222	Positive
21.	NKX2.2 - Clone - NK2/294	Negative
22.	BCL2 - Clone - 124	Positive

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