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Ophthalmology

Primary Orbital Peripheral Neuroectodermal Tumour – A Rare Presentation.

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ABSTRACT Primitive neuroectodermal tumor(PNET) is a small round cell malignant tumor of neuroectodermal origin. Most of the PNET's occur in the central nervous system. Those occurring outside are called peripheral PNET(pPNET). We report a case of an adult male presenting with eccentric proptosis with extraconal and intraconal mass, diagnosed as malignant round cell tumor on histopathology and pPNET on immunohistochemistry.

KEYWORDS : primary neuroectodermal tumour, histopathology, immunohistochemistry.

INTRODUCTION:

Primitive neuroectodermal tumor (PNET) includes a wide array of lesions with varying degrees of differentiation affecting both the central and peripheral nervous system. It is seen in young adults and adolescents most commonly with no sex predilection. Primary orbital pPNET is rare with only 9 cases reported in literature. It shows characteristic small round cell tumor with rosette or pseudorosette and in some cases there are findings on neurosecretory granules. This report presents the clinical, radiological and histopathological findings of a left orbital mass in an adult male.

Case Report:

A 54 year old male patient came to our OPD with chief complaints of progressive, painless and progressive forward protrusion of the left eye since 5-6 months. He also had gradual, progressive diminution of vision in left eye since 2 months; without any diplopia.

Patient gives history of pulmonary tuberculosis 6 years back, for which treatment has been taken as per schedule.

There were no systemic symptoms including loss of appetite, loss of weight , fever or headache. No significant medical, surgical or family history was elicited.

On Examination :

His uncorrected vision in the right eye was 6/24 improving to 6/9 with spectacles, while in the left eye was Fc(3m) improving to 6/18 with spectacles. Intraocular pressure was 14.6 mm Hg in both eyes. There was a prominent cystic, painless, non tender and movable swelling of size about 1.5 X 1.5 cms present about 1 cms lateral to lateral canthus of left eye. Diffuse multiple painless, non tender swellings present extending from the swelling upwards between the eyebrow and eyeball. Eccentric proptosis of the eyeball was noted with downward displacement of eyeball. Pulsations & transillumination were absent, with no postural variations. Hertel exophthalmometry was 12 mm on the right side and 23 mm on the left side.

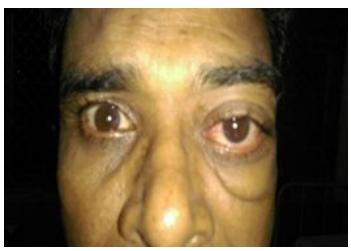

Fig 1 : front view showing eccentric proptosis

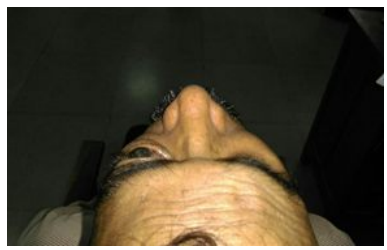

Fig 2: Nafziger' s test showing proptosis

Anterior and posterior segment examination was normal in both eyes. Systemic examination revealed no abnormality. B scan ultrasonography gave findings suggestive of retrobulbar space occupying lesion. A USG guided FNAC from the left retrobulbar mass was done which showed reactive lymphoid population comprising of mature & immature lymphoid cells, histiocytes, neutrophils and plasma cells.

An MRI of the brain and orbit , with contrast, showed an altered signal intensity lesion in the intraconal & extraconal compartment of the left orbit involving the left lacrimal gland. The lesion was encasing and medially displacing the lateral rectus and the optic nerve with anterior displacement of the globe. Preseptal extension into the upper eyelid was noted.

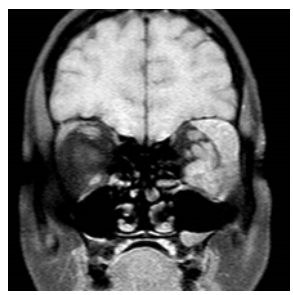

Fig 3:MRI (coronal section)

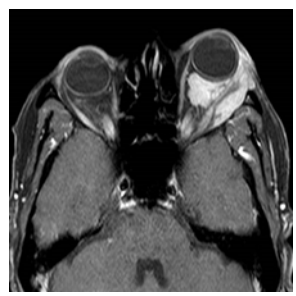

Fig 4: MRI (axial section)

In view of the MRI and cytologic findings, a probable diagnosis of Mickulicz disease was considered.

However, due to increase in the proptosis with signs of compressive optic neuropathy and for confirmatory diagnosis, patient was subjected to left frontal craniotomy with lateral orbital decompression on 24 september 2015. The mass was sent for histopathological examination.

Paraffin sections were stained with Hematoxylin-Eosin and Periodic Acid Schiff stains. Histopathological study revealed tumour tissue comprising of small round cells arranged in diffuse sheets & rosette pattern at places with intervening vascular channels showing hyalinisation. Cells had a centrally placed round hyperchromatic nuclei with scanty cytoplasm, with significant mitotic activity. Immunohistochemistry was done for HIC-2 gene (CD99), neuron specific enolase(NSE), leucocyte common antigen (LCA)CD45, S-100 protein, desmin and MIC-2. It revealed primitive neuroectodermal tumor with the cells positive for CD-99 , NSE and FLI-1(nuclear marker of endothelial differentiation) markers. Thus, a diagnosis of malignant round cell tumor was made.

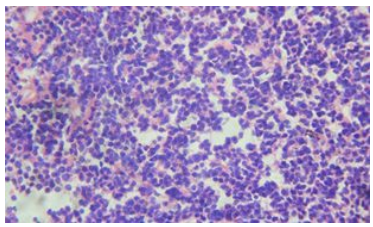

Fig 5 : H&E section

Postoperatively, the left upper eyelid swelling reduced significantly. The patient was started on chemotherapy with vincristine, doxorubicin and cyclophosphamide. So far the patient has received 3 cycles of chemotherapy, out of the 6 scheduled. So far, the patient has been well with subsidence of the proptosis and no sign of recurrence or metastasis.

Discussion:

PNET is a malignancy common in adolescents and young adults with no gender bias. They are classified under Ewing's family of tumors with which it shares its histopathological and cytogenetic similarity.[1,2,3] All are characterised by translocations involving the EWS gene at 22q12, usually t(11;22)(q24;12).[1,2]

The differential diagnosis of pPNET of orbit include other small blue round cell tumors including rhabdomyosarcoma, lymphoma, Ewing's sarcoma, neuroblastoma and hemangioblastoma. Immunohistochemistry helps in differentiationg this from other tumors. [6,7] Specific marker for primary peripheral PNET is HIC-2 gene (CD99),NSE and GFAP.[6,7]The first two were positive in our case, thus confirming the diagnosis.

Microscopically, primary peripheral PNET is a cellular tumor with characteristic small round cells with hyperchromatic nuclei, and a high nuclear/cytoplasmic ratio.[6,9] There are varying degrees of neuronal differentiation, NSE expressivity, Homer Wright rosette formation and neurofilament protein expression. Our case was NSE positive with presence of rosettes.[4,6,7]

Out of the nine previous reported cases of isolated orbital PNET, age group varied from less than one year to 13 years of age with two cases reported in adults (28 and 52yrs) with a predilection for the lateral orbit.[8] In our case, tumor was initially present in the orbit laterally encasing the lacrimal gland with extraconal and intraconal involvement, with no bony involvement. Management varies with different cases, chemotherapy and radiotherapy being the options.[2,3,4] Our patient was subjected to lateral orbital decompression and has received 3 cycles of chemotherapy till now, with no evidence of recurrence or metastasis as yet.

Primary peripheral PNET of the orbit is a rare disease which poses a diagnostic challenge. Imaging and histopathology help in supporting the diagnosis, but immunohistochemistry is confirmative. Management should be planned according to the individual need and should comprise of multimodality treatment approach. Follow up should be long term to rule out recurrence, metastasis and treatment related ocular and extraocular complications.

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