An extremely rare case of a pediatric peripheral primitive neuroectodermal tumour: Orbital primitive neuroectodermal tumour

Çok nadir bir pediatrik periferik primitif nöroektodermal tümör vakası: Orbital primitif nöroektodermal tümör

Hacer BAL, Cennet ŞAHİN, Erkin ARIBAL

Abstract

Primitive neuroectodermal tumours (PNETs) are a group of malignant soft tissue tumours of neuroepithelial origin that arise from primitive neural crest cells. Most of the PNETs occur in the central nervous system (CNS). If the origin is outside the CNS it is called peripheral primitive neuroectodermal tumour (pPNET). Histopathologically, PNETs consist of small round cells with a hyperchromatic nucleus, high nuclear-cytoplasmic ratio and varying degrees of neural differentiation detectable by immunohistochemical or ultrastructural techniques. pPNETs occur predominantly in children and young adults and show no gender difference. Occurrences of pPNETs in the orbit are infrequent and to the best of our knowledge only eighteen cases have been reported in the literature up to now. In this study, we present clinic, radiologic and histopathologic features of an orbital mass in an 8-year-old boy, which was diagnosed as a primary orbital pPNET confirmed by immunohistochemistry.

Key words: Pediatric, Peripheral primitive neuroectodermal tumour, Orbita

Table I. The sarcomas relevant to the differential diagnosis of primary pPNET

<table>
<thead>
<tr>
<th>PNET</th>
<th>HIC-2 gene (+), NSE (+), synaptophysin (+), glial fibrillary acidic protein (GFAP) (+), MIC2 gene (+)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ewing’s Sarcoma</td>
<td>PGP9.5 antibody (+), MIC2 gene (+)</td>
</tr>
<tr>
<td>Rhabdomyosarcoma</td>
<td>Actin (+), Vimentin (+), Desmin (+), S-100 (-)</td>
</tr>
<tr>
<td>Lymphoma</td>
<td>LCA (+), CD45 (+), CD20 (+), CD3 (+)</td>
</tr>
<tr>
<td>Neuroblastoma</td>
<td>PGP9.5 antibody (+), MIC2 gene (-)</td>
</tr>
<tr>
<td>Osteogenic Sarcoma</td>
<td>Osteoid (+)</td>
</tr>
</tbody>
</table>

Case Report

An 8-year-old boy was brought to our hospital with complaints of progressive protrusion of the left eye ball for two months and visual lost for one week. On external ocular examination there was a localized, tender, firm, fixed, non-pulsating globular mass in the superolateral orbit. There was an inferior and medial deviation of the left eye, restriction of ocular movements superolaterally and total visual loss.
He had minimal ptosis and he had no ocular pain. There was no history of injury, fever, chronic cough or other systemic symptoms. The patient had magnetic resonance imaging (MRI) and computed tomography (CT) with a pre-diagnosis of an ocular mass. On non-contrast cranial CT images there was a 36x20 mm space-occupying lesion in the left lateral orbital region, causing inferomedial and anterior deviation of the left eye (Figure 1). Also there were calcifications on posterolateral wall of the eye. On contrast enhanced cranial MRIs, an extension of the lesion along the left optic nerve, optic chiasm and optic tract was seen clearly (Figure 2). There was also a persistent primary hyperplastic vitrea on the left eye. A neoplastic process was suggested.

Extensive systemic investigations were done subsequently to rule out any other foci of tumour; these included a complete hemogram, CT of thorax-abdomen-pelvis, spinal MRI and bone marrow biopsy. These were all normal and therefore a diagnosis of a primary orbital malignancy was made. The patient had a left craniotomy, excision of the orbital rim and the mass, a duroplasty three days after the diagnosis. Histopathologic examination of the tumour revealed a diffuse collection of small malignant round to oval cells with high mitotic activity. Immunohistochemically, the majority of the tumour cells were positive for CD99 (MIC2 gene), neuron specific enolase (NSE) and the synaptophysin gene. Periodic acid-Schiff (PAS) reaction which rules out Ewing’s sarcoma was negative. Based on the above findings the patient was diagnosed as having pPNET of the left orbit. Cytogenetic studies showed the characteristic t(11;22) chromosomal translocation that confirmed the pPNET diagnosis. Following the operation, the patient had chemotherapy and radiotherapy treatments. Cranial CT and MRI were performed during postoperative 24 hours (Figure 3), and control images were obtained two months later, showing no recurrence and a total resection of the mass (Figure 4).

**Discussion**

PNET cells are derived from neuroectoderm which appears primitive. There are two types based on the tumour location: (1) PNETs in the CNS; (2) pPNETs outside the nervous system [1-3]. Primary orbital location which is a type of pPNET, as we present in our case, is extremely rare [1,3]. pPNET occurs predominantly in children and young adults and show no gender difference [1,6,7]. Imaging and histopathology, though supportive, do not confirm the diagnosis. Immunohistochemical and cytogenetic studies help in the confirmation of diagnosis. Microscopically, primary pPNETs are cellular tumours with small round cells that have hyperchromatic nuclei, and a high nuclear–cytoplasmic ratio. pPNETs differ in their degree of neuroectodermal differentiation. They are classified under Ewing’s family of tumours (extraosseous type), with which they share histopathological and cytogenetic similarity [1,3,8]. Immunohistochemical and cytogenetic
The treatment for pPNET usually involves a combination of surgery and chemotherapy with or without radiotherapy [3,6,11]. The modality of treatment depends on the type, site and size of tumour, extent of metastasis as well as the age and general health status of the patient. Surgery has been the initial treatment for orbital pPNET in most cases and has been the initial therapeutic approach used in our patient, although some of the published cases have been treated with chemotherapy and radiotherapy without surgery, with good results [6]. Our case was managed with successful resection of the mass, chemotherapy and external beam radiotherapy. There was no recurrence on the control investigation performed two months after the operation.

Conclusion

When a primary hypercellular small round cell tumour of the orbit is encountered, the recently recognized rare pPNET of the orbit should be considered in the differential diagnosis although the orbit is an extremely rare site for such a tumour. Accurate diagnosis of these tumours is of paramount importance. Immunocytoology helps to confirm the diagnosis. Management should be aggressive using multimodality treatment approaches given at the appropriate time. These patients should be followed up for life to rule out recurrence, metastasis and treatment-related malignancies.

References