RECURRENCE OF PERIPHERAL PRIMITIVE NEUROECTODERMAL TUMOR OF THE ORBIT WITH SYSTEMIC METASTASES

RECIDIVA DE TUMOR NEUROECTODÉRMICO PRIMITIVO PERIFÉRICO ORBITARIO CON METÁSTASIS SISTÉMICA

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ABSTRACT

Case report: A six-year-old boy presented with proptosis of the right eye. Imaging studies detected a mass in the medial wall of the right orbit. This mass was biopsied revealing a histopathologic diagnosis of primitive neuroectodermal tumor, so chemotherapy treatment was given. After seven years in remission he presented with a recurrence of the orbital tumor and was found to also have systemic metastases. Treatment with chemotherapy, radiotherapy and orbital exenteration was unsuccessful.

Discussion: The orbital occurrence of these tumors is extremely rare. Differentiation from other small round cell tumors requires immunohistochemical and ultrastructural techniques (Arch Soc Esp Oftalmol 2006; 81: 599-602).

Key words: Peripheral primitive neuroectodermal tumor, recurrence, orbit, proptosis, small round cell tumors.

RESUMEN

Caso clínico: Paciente varón de 6 años con proptosis del ojo derecho. Las pruebas de imagen detectaron una masa en la pared medial de la órbita derecha que fue biopsiada; el diagnóstico histopatológico fue de tumor neuroectodérmico primitivo. Él paciente recibió tratamiento quimioterápico. Tras 7 años en remisión el paciente presentó recidiva del tumor orbitario con metástasis sistémicas que fue tratada sin éxito con quimioterapia, radioterapia, y exenteración orbitaria.

Discusión: La localización orbitaria de estos tumores es extremadamente rara. El diagnóstico diferencial debe realizarse con otros tumores de células pequeñas redondeadas, siendo fundamentales las técnicas inmunohistoquímicas y/o ultraestructurales.

Palabras clave: Tumor neuroectodérmico primitivo periférico, recidiva, órbita, proptosis, tumor de células pequeñas redondeadas.

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INTRODUCTION

Primitive peripheral neuroectodermal tumors (PPNET) are soft tissue malign tumors which presumably originate in the neural crest and appear outside of the central and sympathetic nervous system. They account for 4-17% of soft tissue tumors in the pediatric stage.

The orbital location of these tumors is extremely unusual. To our knowledge, only ten cases have been published in literature (1-5), eight of them in the pediatric stage of life.

CASE REPORT

Male patient, 6 years old, without relevant personal or familial history, who attends the urgency service in January 1997 due to unilateral right exophthalmos which started 3 days before. Exploration revealed limitations in the supraduction of the right eye (RE) and reduced retropulsion and slightly painful exophthalmus in the same eye (RE exophthalmometry: 16 mm and left eye: 11 mm). The rest of the ophthalmological exploration gave normal results. Computerized axial tomography (CAT) and nuclear magnetic resonance (NMR) revealed an extraconal mass (28 mm x 18 mm x 15 mm) of compact appearance in the internal wall of the right orbit which displaced the eye in the inferior temporal direction and which captured contrast (figs. 1 and 2). No invasion of adjacent structures was evidenced.

Under general anesthesia, a transconjunctival orbitotomy was performed, partially resecting the tumor. The histopathological study revealed a malign tumor of small rounded cells with frequent mitosis patterns (fig.3); no rosettes or pseudo-rosettes were evident. The anatomic/pathological diagnosis was primitive neuroectodermal tumor.

A systemic assessment (cranial NMR, CAT of thorax, abdomen, pelvis and limbs, medula biopsy) yielded negative results, thus leading to the final diagnostic of PPNET.

The patient was referred to the oncology service where he received chemotherapy treatment as per

Fig. 1: MRN in T1, coronal section.

Fig. 2: NMR with contrast (Gadolinium) in T1, axial section (left) and sagittal section (right).
the protocol for malign mesenchimal tumors of the International Pediatric Oncology Society (vincristine, actinomicine, phosphamide, carboplatinum, epirubicine, ethoposide), leading to remission.

The patient’s evolution was positive in the assessments, while the systemic and orbitary image tests (echography and/or NMR) carried out at regular period were normal (fig. 4).

After seven years of remission, the patient visited the hospital in February 2004 due to RE proptosis. An orbitary NMR revealed a mass around the internal straight muscle which caused a lateral displacement of the orbitary content, without bone extension (fig. 5). A biopsy was made which was reported as a relapse of the neuroectodermal tumor since a proliferation of small rounded cells was detected, with abundant mitosis patterns and positive results for CD19 and sinaptophysine.

In spite of an aggressive chemotherapy treatment (carbo-platinum, topotecane, cyclophosphamid), local radiotherpay (doses of 5000 cGy), and craniofacial multidisciplinary exenteration, the tumor could not be controlled. Subsequent cerebrum extensions were identified as well as lung metastasis which caused the demise of the patient.

**DISCUSSION**

The peak age for prevalence of PPNET is the teenage years.

It must be taken into account that the orbitary location of this type of tumor is highly infrequent, when found in the orbit, it usually appears in the lateral walls (3).
The differential diagnostic of orbital PPNET must be made with other small rounded cell tumors including Ewing’s tumor, neuroblastoma metastasis, rhabdomyosarcome, lymphoma and osteogenic sarcome. The PPNET tumors were included within the extra-skeletal Ewing tumors, with which they share microscopic, genetic and molecular alterations, but they differ in that they exhibit varying degrees of neuronal differentiation. Many authors consider that PPNET’s and extra-skeletal Exing tumors represent extremes of the same group of tumors. Accordingly, immunohistochemical and/or ultrastructural techniques are essential for establishing the diagnostic between these two tumors and to differentiate them from other small rounded cell tumors.

At the immunohistochemical level, PPNETs are characterized by yielding positive results for vimentine, CD19, glycoprotein p30-p32, and neuronal markers (neurofilaments, sinaptofysine, chromogranine, specific neuronal enolase). At the structural level, in PPNETs we can find intermediate cytoplasmatic filaments, cytoplasmatic microtubules and secreting granules.

In the orbitary PPNETs we can find local bone involvement and extraorbital extensions (1,5), but systemic metastases are usual. There is only one published case of orbital PPNET with hepatic metastasis (4). To this date and knowledge after carrying out a vast network search, this is the third case described in literature of orbital recurrence after treatment (1,5) and the second case described with systemic metastasis (4). Of the ten patients whose cases were published, two died from the tumor (1) but we ignore the evolution to date of the remaining patients.

PPNETs are tumors which evolve rapidly with bad prognosis.

There is no consensus about the best therapeutic strategy. Some authors consider chemotherapy plus-minus radiotherapy as the first line of treatment (2).

REFERENCES