UVEITIS MASQUERADE SYNDROME PRESENTING AS A DIFFUSE RETINOBLASTOMA

SÍNDROME MASCARADA POR RETINOBLASTOMA DIFUSO

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ABSTRACT

Clinic report: A 3-year-old boy presented with an intermediate uveitis. Complete ophthalmic exam, ocular ultrasonography, magnetic resonance imaging and computerized tomography of the orbit were inconclusive. Determination of the aqueous humor/serum rate of Lactate dehydrogenase (LDH) was the key for the diagnosis of a diffuse retinoblastoma.

Discussion: A masquerade syndrome is the initial presentation in 1-3% of retinoblastomas. Aqueous humor puncture is contraindicated in patients with retinoblastoma but it might be the only way to achieve a correct diagnosis in these difficult and very unusual cases: enzymatic assays such as LDH offer a good sensitivity and specificity for the diagnosis of these patients (Arch Soc Esp Oftalmol 2009; 84: 477-480).

Key words: Intermediate uveitis, LDH, masquerade, retinoblastoma.

RESUMEN

Caso clínico: Paciente de tres años con sospecha de uveítis intermedia. La exploración oftalmológica, ecografía ocular, resonancia nuclear magnética y tomografía computerizada orbitaria no fueron concluyentes. La determinación de la tasa de lactato deshidrogenasa (LDH) en humor acuoso/suero fue la clave para llegar al diagnóstico de un retinoblastoma difuso.

Discusión: El síndrome mascarada es la forma de debut en un 1-3% de los retinoblastomas. La punción de cámara anterior está contraindicada en pacientes con retinoblastoma pero podría ser la única forma de llegar a un diagnóstico correcto en estos casos difíciles y poco frecuentes. Las pruebas enzimáticas como la LDH nos ofrecen una buena sensibilidad y especificidad para el diagnóstico de estos pacientes.

Palabras clave: Uveítis intermedia, LDH, mascarada, retinoblastoma.
INTRODUCTION

Diffuse retinoblastomas are infrequent and difficult to diagnose. These tumors can express as an intra-ocular inflammatory reaction, causing uveitis with presence of tumor cell groups simulating hypopion. The usual treatment consists in enucleation of the affected eye. The severity of the diagnostic has led to the development of different confirmation tests before enucleation. The determination of LDH in the aqueous humor can be useful in the management of these cases.

CLINICAL CASE

A three-year-old child was referred to our service with suspected left eye anterior uveitis resistant to topical treatment. The patient did not exhibit relevant personal or familial history. The slit lamp exploration showed a marked cellularity in the anterior chamber with a 1 mm hypopion. He also exhibited a vitreous cellular tyndall as well as a white lesion of about 2 mm diameter in the pars plana (fig. 1). Contralateral eye was normal.

An ocular echography was performed which it did not detect intralesional calcium (fig. 2). Nuclear magnetic resonance showed the superior temporal lesion whereas the computer tomography detected a very small intra-lesion calcification (fig. 3). It was decided to determine Lactate dehydrogenase (LDH), which exhibited a rate of aqueous humor/serum of 4. No eosinophiles or anti-toxocara antibodies were found in the aqueous humor. Left eye retinoblastoma was diagnosed and enucleation was decided with a propheses implant sutured to the straight muscles. The anatomic-pathological study confirmed the presence of a diffuse retinoblastoma with partial invasion of the iris, the ciliar body and the choroids (figs. 4-6). In addition, the extension study was completed with bone marrow aspiration and a lumbar punction, which gave negative results.

The case was classified as of middle histopathological risk due to the anterior chamber and sclera invasion without involvement of the postlaminar optic nerve. Six chemotherapy cycles were established with vincristine (1,5 mg/m 2 ) etoposid (150

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Fig. 1: Left ocular fundus image showing the vitreous tyndall together with a 2 mm diameter white lesion.

Fig. 2: ocular echography showing a heterogeneous intra-ocular mass without evidence of calcification.

Fig. 3: orbitary computerized tomography showing a nasal intra-ocular lesion with discrete calcification.
mg/m^2) and carboplatin (560 mg/m^2) (1). The treatment was completed without complications.

The genetic study of mutations in the RB gene was negative. Four years after the treatment of the patient remains healthy. The right eye is normal and he uses an external ocular prosthesis which is well tolerated.

**DISCUSSION**

The classic signs of retinoblastoma are leukocoria and strabismus. Presentation as ocular inflammation can occur in 1-3% of cases, generally associated to a relatively rare variant of diffuse infiltrating retinoblastoma. This generally unilateral variant expresses in older infants around four years old, in contrast with other forms of retinoblastoma with a mean age at diagnosis of 18 months (5).

Histologically, these tumors invade the retina in a diffuse manner without forming a tumor mass. The clinical signs are usually tyndall, pseudohypopion, conjunctival chemosis, hemovitreous and vitritis which constitute an obstacle for ocular fundus exploration. In these cases, the initial diagnostic is usually uveitis. Calcifications may not be present or can be detected by means of magnetic resonance, computerized tomography or echography, which could detect diffuse retinal thickening (2,5). In these infrequent cases an aqueous humor aspiration could be useful. The medical literature does not include large series assessing this procedure. We only have descriptions of isolated clinical cases. The procedure is carried out through a clear cornea to avoid the dissemination of tumor cells and must be reserved for cases in which a diagnostic cannot be reached by non-invasive techniques. The psychological study is diagnostic if tumor cells are detected. However, the small number of said cells and the difficulty of the process frequently produces false negatives (3).

Enzymatic determinations in aqueous humor are a good alternative in these cases. LDH is a glycolytic enzyme which appears at high concentrations in cells with a high metabolic rate. Normally its concentration in serum and aqueous humor is low and...
the LDH ratio between aqueous humor and serum is below 1.0 in patients who do not have retinoblastoma. High LDH levels suggest retinoblastoma but are not pathognomic. High LDH levels have been described in patients with Coats’ disease, traumatic hyphema and other non-tumoral pathologies (4). Other enzymes which participate in the glycolysis cycle, such as phosphoglucose isomerase (PGI) and neuronal-specific enolase (NSE) could also be useful, although there is limited usage experience. Recently it has been proposed to quantify the expression in aqueous humor of GD2 syntase, a marker of neural crest-derived cells, for diagnosing retinoblastoma. As with LDH, the PGI levels are detected by automatic spectrophotometry, a standard technique in all labs. NSE values require commercially available radioimmunometric or enzymoimmunoanalysis. For detecting GD2 synthase much more complex PCR techniques are utilized. All these enzymes are highly sensitive (nearly 100%), but even so none is free from false positives (4,5).

If a child exhibits uveitis, hyphema, hypopion, hemovitreous or retina detachment, we should discard the possibility of retinoblastoma obtaining an assessment by an expert ophthalmologist, as well as ocular echography and/or computerized tomography. The use of puncture-biopsy and enzymatic studies in aqueous humor can be useful but should be reserved only for doubtful cases due to the risk of tumor dissemination (5).

REFERENCES