Short communication

Retinitis pigmentosa sine pigmenti. Debut with macular oedema

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A R T I C L E   I N F O

Article history:
Received 4 September 2012
Accepted 17 September 2013
Available online 26 September 2014

Keywords:
Retinitis pigmentosa sine pigmenti
Macular edema
Acetazolamide
Optical coherence tomography
Night blindness

A B S T R A C T

Case report: A 25-year-old woman, with metamorphopsia in her left eye of one year onset. The examination revealed a bilateral cystoid macular edema (CME) and vascular attenuation. We describe the diagnostic tests as well as differential diagnosis and treatment response with carbonic anhydrase inhibitors.

Discussion: The retinitis pigmentosa sine pigment is a subtype of atypical retinitis pigmentosa characterized by the absence of pigment deposits. The night blindness is milder, and perimetric and electoretinographic impairment is lower. CME is an important cause of central vision loss, and responds to anhydrase carbonic inhibitors.

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R E S U M E N

Retinosist pigmentaria sine pigmento. Inicio con edema macular

Caso clínico: Mujer de 25 años que presenta metamorfopsia y disminución de agudeza visual en ojo izquierdo de un año de evolución. A la exploración se aprecia un edema macular quístico (EMQ) y atenuación vascular bilateral. Se describen las pruebas diagnósticas, así como el diagnóstico diferencial y la respuesta al tratamiento con inhibidores de anhidrasa carbónica.

Discusión: La retinosist pigmentaria sine pigmento es un subtipo de retinosist pigmentaria atípica, caracterizada por la ausencia de depósitos pigmentarios. La hemeralopía es más leve y la afectación campimétrica y electoretinográfica es menor. El EMQ constituye una causa importante de pérdida de visión central, y responde al tratamiento con inhibidores de anhidrasa carbónica.

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Introduction

Retinitis pigmentosa (RP) is the most frequent retinal dystrophy and the fourth cause of blindness in the world.\textsuperscript{1} It comprises a heterogeneous range of degenerative retinal diseases characterized by progressive loss of photoreceptors and retina pigment epithelium (RPE) involvement, which in many cases leads to blindness. RP comprises shared clinical expressions including hemeralopia, concentric and visual field reduction, retinal pigment deposits and diminished electroretinogram. Typically, central vision is not involved up to later stages of the disease except with some associated complications such as cystic macular edema (CME) or cataract.\textsuperscript{2}

RP sine pigmento is a subtype of atypical RP, characterized by the absence of pigment changes. The management of the disease must involve the exclusion of associated systemic diseases as well as social and genetic counseling.

Clinic case

Female, 25, of Romanian origin, exhibiting metamorphopsia and slight visual acuity (VA) reduction in the left eye (LE) with one year evolution. No relevant history. Baseline VA was of 0.8 in the right eye (RE) and 0.9 in LE, exhibiting non-tributary correction slight hypermetropia (+0.50 in both eyes). Anterior segment biomicroscopy and intraocular pressure were normal, while funduscopy revealed a grayish reflection, CME.

Fig. 1 – Baseline RE and LE fundus.

Fig. 2 – Baseline patient LE OCT.
Fig. 3 – RE (top) and LE (bottom) angiograph.

Fig. 4 – LE OCT after one year evolution.
Fig. 5 – Patient visual field 30:2. Humphrey-type campimetry (Zeiss) with standard SITA strategy.

and slight bilateral vascular attenuation in both eyes, without other relevant findings (Fig. 1). Optic coherence tomography (OCT) confirmed CME together with subfoveal intraretinal cysts in RE (Fig. 2). Angiograph verified CME together with grayish dotted RPE (Fig. 3). Autofluorescence was not performed because said test was not carried out during the evolution of the patient.

The patient did not refer dyschromatopsia but reported difficulties adapting to darkness for many years although she did not consider this important.

Campimetry, laboratory study with serology and antibodies as well as chest and sacroiliac x-rays were requested considering an inflammatory or infectious process, together with electrophysiological tests due to suspected retinal
dystrophy. Treatment was initiated with oral acetazolamide 250 mg every 8 h. Patient response was favorable with CME reduction, metamorphopsia and VA improvement (1.0 in both eyes). Accordingly, dosage was progressively reduced without the patient experiencing relapses, although asymptomatic bilateral subfoveal cysts persisted (Fig. 4).

Campimetry revealed concentric visual field reduction vis-à-vis the central area in both eyes (Fig. 5). Electrooculogram exhibited wide response to attenuated lights while the electroretinogram showed highly attenuated retinal responses in photopic as well as scotopic conditions. Visual evoked potentials and 100 Hue Farnsworth-Munsell test were normal. A differential diagnostic was considered between rod-cone dystrophy and RP sine pigmento, finally choosing the latter option due to clinical characteristics and the results of supplementary tests.

The patient has 4 brothers and it was possible to carry out an ophthalmological examination to only one, with entirely normal results (Fig. 6).

**Discussion**

Diagnosing retinosi sine pigmento on the basis of purely ophthalmoscopic findings is obviously more difficult than typical retinosis with pigmented deposits. Changes in RPE can be very subtle. In the case reported herein, arteriole attenuation was marked but without exhibiting additional semiology.

In this disease the amount of pigment is variable. For this reason, many authors propose that RP sine pigmento could be an extreme pattern of RP which could evolve to the typical form. However, at this point in time it is a specific subtype of atypical RP. In these patients, hemeralopia is less acute and in general they exhibit lower campimetry and electroretinographic involvement, in contrast with this case.  

CME is associated with RP in a given percentage of patients, compromising central vision. Sandberg et al. reported an incidence of CME diagnosed with OCT of 28% in a cohort of over 300 patients with classic RP, mainly in the dominant type. In addition, they demonstrated that in these patients VA was linked to foveal retinal thickness as well as to the presence of macular cysts. In the present case, the patient exhibited discrete visual involvement in LE, but RE macular cysts did not affect VA.

It has been demonstrated that treatment of CME associated to RP responds to topical and systemic treatment with anhydrase carbonic inhibitors (ACI). However, in some patients anatomic improvement is not accompanied by VA improvements due to likely loss of photoreceptors prior to the edema or as part of it.

By way of conclusion, the case reported herein is an atypical case of RP sine pigmento diagnosed due to CME together with subtle changes in the RPE and vasculature in a patient whose only symptom for many years was hemeralopia. The patient exhibited positive evolution to treatment with systemic ACI both in central retinal thickness reduction demonstrated by OCT and in visual recovery.

**Conflict of interests**

No conflict of interests has been declared by the authors.

**References**

