Short communication

Optical coherence tomography in the diagnosis of achromatopsia☆,☆☆

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A B S T R A C T

Case report: The case of a fifty-five year-old male with nyctalopia, photophobia, poor color vision and nystagmus is presented. The initial suspected diagnoses were achromatopsia and blue-cone monochromatism, since both are clinically indistinguishable. Optical coherence tomography (OCT) showed the characteristic foveal reflectivity pattern of achromatopsia. This diagnosis was subsequently confirmed by genetic study.

Discussion: OCT is a non-invasive diagnostic imaging method that allows tissue morphology to be observed with high resolution. Its use might be of great help to distinguish clinically similar diseases.

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Tomografía de coherencia óptica en el diagnóstico de la acromatopsia

R E S U M E N

Caso clínico: Varón de 55 años con nictalopia, fotofobia, mala visión de los colores y nistagmo. Nos planteamos el diagnóstico diferencial entre la acromatopsia y el monocromatismo de conos azules, puesto que ambos son clínicamente indistinguibles. En la tomografía de coherencia óptica (OCT) nos encontramos un patrón de reflectividad foveal característico de la acromatopsia, diagnóstico que posteriormente confirmamos con el estudio genético.

Discusión: La OCT es un método de diagnóstico por imagen, no invasivo, que permite la visualización de los tejidos con alta resolución. Su aportación en enfermedades clínicamente similares es fundamental porque nos ayuda a hacer el diagnóstico.

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Introduction

Optic coherence tomography (OCT) is a noninvasive imaging and diagnostic method which allows observing tissue in vivo. The acquired images can be analyzed qualitatively by recognizing various morphological patterns as well as quantitatively on the basis of the thickening and distance between reflective layers. OCT has been very useful for studying macular pathologies, both for diagnostic as well as follow-up. 1,2

Clinic case

Male, 55, with nyctalopia, photophobia, poor color vision and nystagmus since birth. Sister with same symptoms.

Ophthalmological examination gave a visual acuity (VA) of 1/8 with correction—no gain with stenopeic in both eyes. Pendulum nystagmus. Anterior segment was normal and ocular fundus papilla and macula exhibited normal appearance but without foveal reflex (Fig. 1). Ocular pressure of 14 mmHg recorded in both eyes. The observed central visual field was 10-2: bilateral relative central scotoma (Fig. 2).

Fig. 1 – Normal ocular fundus.

Fig. 2 – Central visual field 10-2 (top, RE; bottom, LE): relative central scotoma in both eyes.
Color vision study: patient identified some numbers in the Ishihara test, indicating residual discrimination of color shades. In test 15 Hue Désaturé de Lanthony patient exhibited lines which did not follow any axis, while in the Farnsworth test type «100 Hue» deep chromatic amblyopia was identified (Fig. 3).

Neurophysiological examination: EOG with normal Arden index. PEV with pattern abnormal. ERG with amplitude decremented stimulus pattern. ERG with Ganzfeld stimulus, normal scotopic and abnormal photopic response, with highly reduced Flicker response.

The condition suggested an incomplete or atypical achromatopsia (AC), although differential diagnostic with blue cone monochromatism (BCM) must be performed because clinically they cannot be differentiated.

Macular study with Cirrus® HD OCT (Carl Zeiss Meditec Inc.) and spectral domain technology revealed normal foveal and diminished parafoveal thickness, interruption of the hyper-reflective thin line representing the union between the internal and external photoreceptor segments (Fig. 4). Foveal reflectiveness pattern characteristic of AC, therefore diagnostic was labeled as incomplete or atypical AC, confirmed 10 months later by molecular genetic study.

**Discussion**

Congenital cone system dysfunction, characterized by abnormal color vision and poor VA, includes complete and incomplete congenital AC and BCM.9

AC is a rare retinal disease of recessive autosomic inheritance. Generally, subjects exhibit since birth and in stationary manner very poor VA, photophobia, relative central scotoma, pendulum nystagmus which diminishes with time and various degrees of loss in color vision. Biomicroscopy was normal but reduced foveal reflex or abnormalities in the central or peripheral RPE can be found, or even bull’s eye maculopathy. Two conditions are distinguished: complete AC (typical or rod monochromatism) and incomplete or atypical AC.

Complete AC involves total absence of cone function, VA in the range of 0.1 and imperceptible color vision with all colors being equivalent to shades of gray. Three genes are involved: CNGA3 (25%), CNGB3 (50%) and GNAT2 (2%).1,2 Some of the individuals affected with CNGB3 mutation preserve a residual cone function in ERG up to middle-age which is progressively lost, to the point that responses to rod function deteriorate.4,5 In incomplete AC, some individuals can
Table 1 – Thickness. Hyper-Reflective Layers.

<table>
<thead>
<tr>
<th>Thickness</th>
<th>Hyper-reflective layers</th>
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<tr>
<td></td>
<td>P2union of segments</td>
</tr>
<tr>
<td>Foveal</td>
<td>Normal</td>
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<tr>
<td>Parafoveal</td>
<td>↓</td>
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Fig. 4 – Cirrus® HD OCT (top RE; bottom LE): normal foveal thickness and parafoveal diminished, with interruption of hyper-reflective layer P2 (union between photoreceptor internal and external segments).

preserve very slight albeit abnormal color vision, slightly better VA (0.1–0.25), CNGA3 mutations have also been identified and some long and/or middle wave length cones are probably present.

BCM is a recessive inheritance disease linked to X. Only the blue cones (short wavelength) encoded by genes located in chromosome 7 remain functional. BCM is generally stationary although some progressive cases have been described in elderly patients. Mutations have been identified in opsin OPN1LW, OPN1MW red and green.1,3

Barthelmonth et al.1 in 2006 and Varsányi et al.2 in 2007 analyzed macular thickness and morphology with Stratus OCT in patients with AC, BCM and normal controls. OCT revealed significant structural alterations in the macula of achromats and establish different foveal reflectiveness patterns allowing for a simple, reliable and clear differentiation between these 2 diseases (Table 1).

In AC, the hyper-reflective layer which represents the union of internal and external photoreceptor segments (P2) is not present. This photoreceptor region is rich in mitochondria and foveal thickness does not vary vis-à-vis normal eyes.

The contribution of OCT is crucial precisely in clinically similar diseases such as AC and BCM as it facilitates differentiation between both and accordingly enables adequate diagnostics.

Conflict of interest

No conflict of interest has been declared by the authors.

REFERENCES