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## Short communication

# Optical coherence tomography in the diagnosis of achromatopsia<sup>☆,☆☆</sup>

C. Burgueño-Montañés\*, M. Colunga-Cueva

Hospital Universitario Central de Asturias, Oviedo, Spain

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### ABSTRACT

**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

### RESUMEN

**Caso clínico:** Varón de 55 años con nictalopía, 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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#### Palabras clave:

Acromatopsia

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\* Corresponding author.

E-mail address: [casamata@gmail.com](mailto:casamata@gmail.com) (C. Burgueño-Montañés).

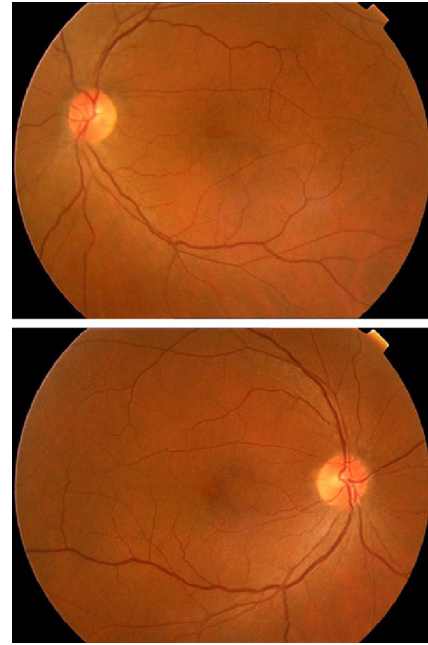
**Introduction**

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.<sup>1,2</sup>

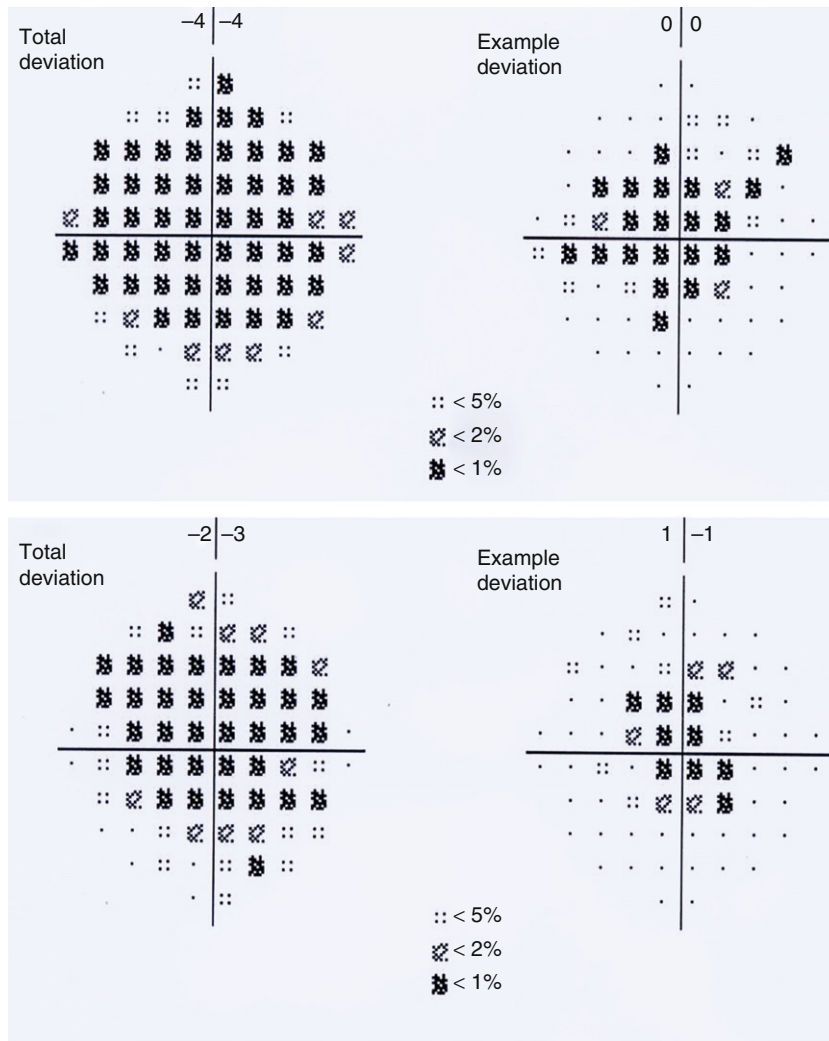
**Clinic case**

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.**

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