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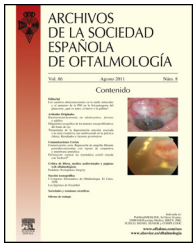

Short communication

Debut of Leber's hereditary optic neuropathy. Macular segmentation analysis using optical coherence tomography☆

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ABSTRACT

Case report: Two clinical cases are presented of two family relatives newly diagnosed with Leber hereditary optic neuropathy (LHON) and G11778A mutation analysis by optical coherence tomography (Cirrus HD-OCT, Carl Zeiss Meditec, Dublin, California, USA) layer peripapillary fibers retina (RNFL) and ganglion cell and internal plexiform layers (GCL/IPL) using macular segmentation.

Discussion: The analysis of the macula by OCT segmentation (version 6.0 Cirrus OCT) allows the GCL/IPL to be evaluated without the interindividual variability of peripapillary RNFL distribution or the presence of edema of the optic disk. When an analysis of the peripapillary RNFL, it does not provide information on this neuronal damage, which itself is evidence in the study of GCL/IPL.

Debut de neuropatía óptica hereditaria de Leber. Análisis mediante segmentación macular con tomografía de coherencia óptica

Palabras clave:
Neuropatía óptica
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RESUMEN

Caso clínico: Se presentan los casos clínicos de 2 familiares directos diagnosticados recientemente de neuropatía óptica hereditaria de Leber (NOHL) mutación G11778A así como el

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Tomografía coherencia óptica
Segmentación macular

análisis mediante tomografía de coherencia óptica (TCO) (Cirrus HD-OCT, Carl Zeiss Meditec, Dublin, California, EE. UU.) de la capa de fibras de la retina peripapilar (CFNR) y de la capa de células ganglionares y la plexiforme interna de la retina (CCG/PI) mediante segmentación macular.

Discusión: El análisis de la mácula mediante segmentación con TCO (versión 6.0 de OCT-Cirrus) nos permite evaluar la CCG/PI sin la variabilidad interindividual de la CFNR peripapilar o por la presencia de edema en el disco óptico. En los casos que presentamos el análisis de la CFNR peripapilar no aporta información sobre el daño neuronal presente, que sí se evidencia en el estudio de la capa de CCG/PI.

Introduction

Leber hereditary optic neuropathy (LHON) is a neurodegenerative disease of mitochondrial maternal inheritance which involves the optic nerve, producing a sudden loss of visual acuity (VA) in young carrier adults which, in some cases, can partially improve[1] (60% in mutations 3460 and 14,484 and 5% in 11,778). The prevalence of LHON is of one patient between 15,000 and 50,000 inhabitants, with onset usually between the age of 15 and 30, with sequential mono-or binocular involvement and severe irreversible impairment in the majority of cases. Over 90% of the mutations which account for LHON occur in positions 11,778, 3460 and 14,484.[1–6]

LHON ophthalmoscopy reveals papillary telangiectasiae, disk edema and absence of leak in angiography, although in many cases it could exhibit a normal appearance which subsequently evolves toward optic disk atrophy. Differential diagnostic should consider other optic neuropathies such as dominant autosomic optic atrophy or the Wolfram syndrome.[2,3,5]

Two cases of simultaneous LHON debut in 2 direct relatives with mutation G11778A are presented, together with optic coherence tomography analysis (OCT) (Cirrus HD-OCT, Carl Zeiss Meditec, Dublin, California, USA) of the peripapillary retina nerve fiber layer (RNFL) and ganglion cell layer/retinal internal plexiform (GCL/IPL) by means of macular segmentation.

Case reports

Case 1: Male, 20, who referred sudden vision reduction in the right eye (RE) with 2 weeks evolution, followed 5 days later by the left eye (LE). The patient did not exhibit any of the residual or ocular symptoms, relevant family history or any drug allergy. He visited the emergency department, where the initial examination produced VA of finger counting in both eyes (BE), normal anterior pole, intraocular pressure of 12 mm Hg in BE and ocular fundus (OF) with slightly engorged vessels and papillary paleness in BE (Fig. 1). Refraction analysis under cycloplegia produced +0.50 in BE. The patient was referred to the Neuro-ophthalmology unit where he was diagnosed with LHON, mutation G11778A. OCT analysis of the peripapillary retina nerve fiber layer (Fig. 2 left) exhibited slight edema in the nasal and superior quadrants in RE and slight temporal atrophy in LE edema in nasal and superior quadrants in RE and temporal atrophy in LE. Macular segmentation exhibited generalized GCL/IPL atrophy in BE (Fig. 2 right).

Case 2: Female, 38, cousin of the previous patient, who referred sudden loss of vision in LE with one week evolution without any other relevant visual or ocular symptom. Upon exploration, the patient exhibited VA of 1.0 in RE and finger counting in LE, normal anterior pole, intraocular pressure of

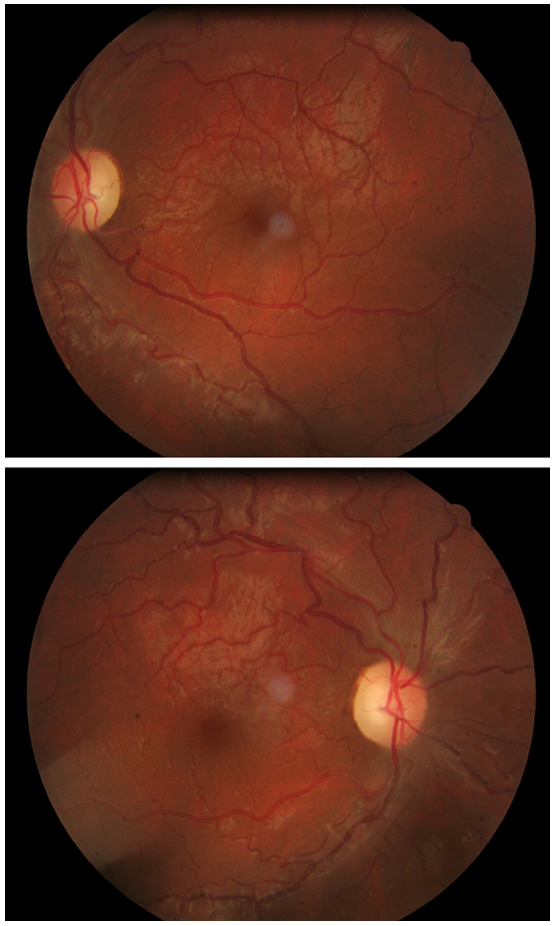

Fig. 1 – Case 1: ocular fundus with slight engorgement and vascular tortuosity and papillary paleness with temporal bilateral predominance.