THE USE OF OPTICAL COHERENCE TOMOGRAPHY IN HEREDITARY OPTIC NEUROPATHIES: DESCRIPTION OF A FAMILY

UTILIZACIÓN DE LA TOMOGRAFÍA ÓPTICA DE COHERENCIA EN NEUROPATÍAS ÓPTICAS HEREDITARIAS. DESCRIPCIÓN DE UNA FAMILIA

SÁNCHEZ-TOCINO H¹, DE-ANDRÉS-SANTOS A¹, PASCUAL-GONZÁLEZ P², SANCHIDRIÁN-MAYO M²

ABSTRACT

Case report: This report describes two siblings, a woman aged 44 years and her brother aged 29 years, who both complained of visual loss in both eyes. The woman had bilateral optic nerve (ON) temporal pallor and severe reduction of ON fibre layer thickness in this area. Both she and her brother had cecocentral defects in perimetry and color vision deficiency with a marked tritanopia deficit.

Discussion: ON pallor, limited to the temporal region, is the basic criterion for the diagnosis of dominant optic atrophy. Optical coherence tomography can be a useful technique in defining the diagnosis and improving the follow-up of patients with this pathology (Arch Soc Esp Oftalmol 2008; 83: 57-62).

Key words: Optical coherence tomography, dominant optic atrophy, hereditary optic atrophy, temporal disk pallor, Kjers’s disease.

RESUMEN

Caso clínico: Se presenta una mujer de 44 años y su hermano de 29 años con mala agudeza visual en ambos ojos. Presentan ambos una palidez temporal en el nervio óptico (NO) bilateralmente, con una severa disminución de fibras del NO en esta área, déficit centrocecal en el campo visual y una alteración de los colores en forma de tritanopia.

Discusión: La palidez del NO que afecta a la región temporal es un criterio básico en el diagnóstico de atrofia óptica dominante. La Tomografía Óptica de coherencia se presenta como una técnica útil en el diagnóstico y posterior seguimiento de esta patología.

Palabras claves: Tomografía Óptica de coherencia, atrofia óptica dominante, atrofia óptica hereditaria, palidez temporal del disco, enfermedad de Kjer’s.
CASE REPORT

A forty four-year-old woman who referred visual acuity reduction dating one year back. The VA with the best correction was of 0.2 in both eyes (BE). Both pupils were bradichoric. The anterior segment and intra-ocular pressure (IOP) were normal. The eye fundus showed an important pallor, close to atrophy, in the temporal sector of BE but greater in the right eye (RE) (fig. 1). The angiofluoresceinography (AFG) did not evidence tincture or microvascular alterations or peri-papillary telangiectasiae (fig. 1).

The Optic Coherence Tomography (OCT) study of the nervous fiber layer thickness (NFL) automatically calculated and evaluated the parapapillary thickness parameters in the vertical and horizontal axis along a circular scan. For the measurements, the «Fast RNFL Thickness (3.4)» was utilized under pharmacological midriasis. The measurements were analyzed by means of the «RFNL Thickness Average Analysis» protocol which is able to quantify the total average thickness in microns (μ) in for peri-papillary sectors and in nine o’clock angles; and «RNFL Thickness Serial Analysis Report» was utilized for studying the differences in the thickness of different explorations.

The OCT marked an important reduction of the mean NFL, mainly at the level of a temporal region and the inferior quadrant in the RE and the entire papilla in the LE (fig. 2). The colour test showed an important dyschromatopsia in BE without a clear pattern, but with a greater alteration in the blue-yellow axis. The visual field showed a central defect which was centrocecal in BE (fig. 3). The visual evoked potentials (VEP) were of low ampli-
The patient was a heavy smoker (approximately 40 cigarettes per day) and referred a poor diet. She was advised to quit smoking and to take a vitamin complex. The analysis carried out with levels of vitamin B12 and folic acid gave normal results. The magnetic nuclear resonance study did not show signs of demyelinization.

Even though after six months the vision had improved to 0.3 in BE, subsequent controls carried out in two years of follow up did not evidence any changes in VA, visual field or OCT.

One year ago the patient’s 29 year-old brother attended the practice after being referred to our practice due to a reduction of VA. He mentioned he had a sister in treatment for «something» related to the eyesight.

The VA was of 0.3 in BE, in binocular 0.4. The eye fundus revealed a temporal papillary pallor in BE without telangiectasiae. The OCT showed an
important reduction of the mean NFL thickness, mainly at the expense of the temporal and inferior sectors (fig. 4). The visual field study revealed central and centrocecal defects in BE (fig. 5). The patient exhibited dyschromatopsia and tritanopia (fig. 6). The VEP type pattern shows a reduction of amplitude with slightly extended latency. At present, the VA has gone down to 0.2 in BE, 0.3 binocular. No changes were found in the OCT or the visual field. As the patient is a smoker (approx. 20 cigarettes per day), he was advised to quit smoking.

Both patients attend six monthly checkups and an application has been filed for carrying out a genetic study.

**DISCUSSION**

Almost all hereditary optic neuropathies are bilateral, approximately symmetrical and with loss of central vision. The Kjer dominant optic atrophy type (ARE) is the most frequent hereditary primary optic neuropathy. Its prevalence is estimated in 1:50,000 (1). The loss of VA usually occurs in the first or second decade of life. It is reduced in symmetrical manner and can be found in levels of 20/200. It has been found that in some families VA is worse in older members, suggesting that VA reduces with age (2). In the instant case, the female referred a progressive loss of visual acuity. She had consulted on several occasions due to finding difficulties in short range of vision and reading which did not improve with eyeglasses. The male was referred to our practice by his general practitioner due to loss of vision which was discovered in an ophthalmological checkup carried out by his employer’s medical facility. Even though the sister was seventeen years older and her visual acuity, colour alteration and visual field were more altered, there was no important variation between both patients.
patients, which leads us to think that the disease progresses very slowly and remains without changes for a long time or that the brother is due to exhibit a more progressive condition. The most frequent chromatic alteration is tritanopia, although general dyschromatopsiae have been described. Typically, the visual field exhibits paracentral or centrocecal scotoma and these patterns were present in both patients. The optic atrophy can be subtle, more frequently temporal but sometimes diffuse, covering the entire optic nerve (ON). The VEP showed a reduction in wavelength amplitude. The above cases exhibited a wavelength reduction in VEP together with a slight increase of latencies in both cases. This finding has already been described by other authors (2). The OCT images revealed an important reduction of the mean NFL thickness, mainly at the expense of the temporal sector. The literary references we have consulted included very few articles or similar case reports describing the characteristics of these conditions in OCT studies. The findings by means of this technique are described as being equivalent to those found in metabolic or toxic optic neuropathy and different to those found in ischaemia or inflammation conditions. The latter appear to be more frequent in the age range of the instant cases (4).

Leber’s optic neuropathy is related to an alteration in mitochondrial DNA. The prevalence in males is of 80-90%. Visual loss is typical between 15-35 years of age (1). During the acute phase hyperaemia appears in the head of the ON, blurry margins, vessel tortuosity and dilatation, peri-papillary telangiectasiae and absence of disc exudation in FAG. The fact that these findings are more frequent in males help bus to differentiate it from ARE.

Barboni described 38 patients with Leber optic neuropathy and found in the OCT study a greater NFL increase in the superior, inferior and nasal sectors in the early stages and a diffuse reduction in all the quadrants in atrophy stages, even though the patient recovered visual acuity after the acute phase had a greater NFL thickness in the superior, inferior and nasal quadrant in comparison to those who did not recover VA, where these differences were not found in the temporal quadrant and considerably lower thicknesses in all patients (5).

It is also necessary to make a differential diagnostic with retinal degenerations and dystrophies. To this end, we must focus on the attenuation of vessels and electoretinogram alterations as well as on other systemic pathologies and neurological syndromes which can exhibit primary development with optic neuropathies (1).

OCT can be very useful in the diagnostic of optic neuropathies based on nutritional, toxic or hereditary causes where a reduction of the maculopapillary fibre layer can typically be identified (5). OCT allows the observer to identify NFL losses in the
temporal sectors. In the case of optic neuropathies, this loss is improved with vitamin supplements and by quitting smoking or drinking. The NFL thickness reductions in ischaemic or inflammatory optic neuropathies can develop with several patterns, either diffuse and affecting the entire disc in inflammatory forms, while in non-artheritic ischaemic forms exhibiting a loss of altitudinal field and affecting more of the upper NFL. Bilateral and symmetrical involvement is more infrequent in this type of neuropathies.

OCT is a simple, non-invasive test which could become a routine examination in the daily neuroophthalmology practice for all types of neuropathies. OCT is demonstrating its advantages not only in diagnostic but more importantly in follow-up (5).

REFERENCES