OPTICAL COHERENCE TOMOGRAPHY IN VITELLIFORM MACULAR DYSTROPHY TYPE 2

TOMOGRAFÍA DE COHERENCIA ÓPTICA EN LA DISTROFIA MACULAR VITELIFORME TIPO 2

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ABSTRACT

Case report: We report the case of a 10-year-old boy with a progressive bilateral vitelliform macular dystrophy, and his father with terminal stage disease in both eyes; we studied the development and progression of this condition with optical coherence tomography.

Discussion: Optical coherence tomography is a useful noninvasive tool that complements other diagnostic modalities and improves the follow up assessment. It provides additional information on the morphology of the lesion as well as identifying secondary changes in the adjacent retina. It also demonstrates the location of any yellowish material under the sensory retina (Arch Soc Esp Oftalmol 2008; 83: 501-504).

Key words: Vitelliform macular dystrophy, Vitelline dystrophy, Vitelliruptive degeneration, Best's disease, Optical coherence tomography.

INTRODUCTION

Best’s disease or vitelliform dystrophy type 2 (VMD 2) is a dystrophy of the retinal pigment epithelium (RPE) comprised within hereditary macular dystrophies (autosomic dominant). The gene involved in Best’s disease is in chromosome 11q12.3 which codes for bestrofine (1). A variable deposit of yellowish material appears, frequently attributed to the lipofuscin deposit in the retinal pigmentary epithelium (1).

RESUMEN

Caso clínico: Presentamos el caso de un niño de 10 años con una distrofia macular viteliforme bilateral, en progresión, y el padre con estadios finales en ambos ojos; se estudia la evolución con la tomografía óptica de coherencia.

Discusión: La tomografía óptica de coherencia es una herramienta útil y no invasiva, que permite complementar el diagnóstico y el seguimiento de los pacientes, aportando datos morfológicos de la lesión así como cambios secundarios en la retina adyacente. Demuestra la localización del material amarillento depositado bajo la retina sensorial.

Palabras clave: Distrofia macular viteliforme, Distrofia Vitelina, Degeneración Viteloeruptiva, Enfermedad de Best, Tomografía de coherencia óptica.
The evolution of vitelliform dystrophy can be observed in children from 3 to 15 years of age, with the following stages: normal fovea (with alteration of the electrooculogram—EOG—) or pre-vitelliform stage, vitelliform stage, pseudohypopion stage, beaten yolk stage and atrophic-cicatricial stage.

**CASE REPORTS**

**Case 1**

A 10 year-old child which is diagnosed with bilateral vitelliform maculopathy. The patient has the familial history of his father with Best disease (autosomic dominant) in final stage.

The initial visual acuity (VA) of the patient with maximum correction in the right eye (RE) is of 0.7 and 0.6 in the left eye (LE).

The funduscopic exploration of the RE (fig. 1 A) showed a yellowish and rounded macular lesion, forming an inferior level (vitelliform injury in pseudohypopion stage). The LE (fig. 2 A) exhibited a subfoveal yellowish lesion in vitelliform stage. Fluorescein angiography (FA) was made to complete the study of the injury (figs. 1 B and 2 B).

A RE Optical Coherence Tomography (OCT) — macular thickness protocol— revealed subretinal hyper reflectiveness in the lower area of the injury with attenuation of light, corresponding to the deposited material. The superior area of the lesion exhibited hypo reflectiveness corresponding to the subretinal liquid, which separates the RPE and neuroepithelium (fig. 1 D). The left eye (fig. 2 D) revealed an over-elevation of the retina with increases subretinal reflectiveness (deposited material and RPE) and attenuation of underlying tissue. There is a loss of definition of the other two hyper reflective bands observed in normal conditions at this level.

After 8 months (fig. 3), the patient exhibited a severe loss of VA, finger counting at two meters with maximum correction in both eyes. The eye fundus exploration revealed a cicatricial stage in both eyes. OCT showed retinal atrophy associated to residual subretinal fibrosis, shown as a reduction of the retinal thickness and subretinal hyper reflectiveness.

**Case 2**

The father of the above patient, aged 46, referred poor eyesight since childhood. His visual acuity with maximum correction is of 0.1 in RE and 0.175 in LE. The eye fundus exploration showed a cicatricial lesion in both eyes with associated RPE hyperplasia. The RE OCT revealed a severe atrophy at the foveal level and the existence of serous subretinal liquid in the LE, as well as small hyper reflective grains in the external profile of the neurosensory retina, similar to the image observed in chronic central serous chorioretinopathy (fig. 4).

![Fig. 1: Right eye: A) Retinography showing a macular lesion in pseudohypopion stage. B) Aneritra retinography. C) FA: inferior hypofluorescence due to screen effect. D) OCT: subretinal hyper reflectiveness, corresponding to the material deposit and hypo-reflectiveness corresponding to the subretinal liquid.](image1)

![Fig. 2: Left eye: A) Retinography: yellowish rounded lesion at subfoveal level (vitelliform stage); B) Aneritra retinography; C) FA showing blockage of fluorescence which produces the injury; D) OCT: elevation of the retina with increased subretinal reflectiveness and attenuation of underlying tissue.](image2)
DISCUSSION

Notwithstanding the striking characteristics of the injury, VMD type 2 maintains a good initial visual acuity. Frequently, the deterioration of vision is secondary to the underlying retinal atrophy or, less frequently, to hemorrhage, scarring or choroidal neovascular membrane.

This disease produces an accumulation of a yellowish material in the centre of the macula, in a circumscribed and cup-shaped configuration in the vitelliform stage, but there is a variable quantity of subretinal liquid present which increases in time (1). This aggregate material tends to settle, forming a level between itself and the clear subretinal liquid. The usual evolution is towards its disappearance, beginning from the central part of the macular injury (1), with the deposits remaining at the margins thereof.

The characteristics of vitelliform dystrophy can be assessed with OCT but there is some debate about the localization of the deposited material (4). Some histopathological studies have described that this material can be under the RPE or the retina (5). The Stratus ® OCT model shows that the material deposit is located between the two high reflectiveness layers corresponding to the retinal pigmented epithelium and to the union of external and internal segments of photoreceptors (4,5).

The separation of both due to the deposits is particularly visible in the margins (4). In our case, if we observe the RE image (pseudohypopyon stage), the upper area of the injury shows hyper reflectiveness in the margin corresponding to the presence of subretinal liquid. In more advanced cases it is possible to follow up on the injury because the retinal thickness above the injury is correlated to the visual acuity.

The usefulness of Optical Coherence Tomography is demonstrated as a supplementary tool which can provide morphological data about the injury, the localization of the material deposits and the stage thereof, apart from showing the appearance of secondary changes in the overlying retina.

REFERENCES