UNILATERAL VITELLIFORM MACULAR INJURY

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PURPOSE

To report a case of an unilateral vitelliform macular injury dystrophy in an elderly patient.

INTRODUCTION

Vitelliform Macular Dystrophy, also known as Best's Disease, is a rare, bilateral disease of autosomal dominant inheritance, with variable penetrance and expressivity.[1]

Individuals affected by Best's disease develop a yellow "egg yolk" (vitelliform) macular lesion usually in childhood (there is also adult disease), which eventually breaks down, leaving a geographical atrophic appearance.[5] At the end of the disease, atrophy may be difficult to distinguish from other types of degeneration or macular dystrophies. The disease can be identified at various clinical stages, from a pre-vitelliform stage, when there are no substantial eye changes, to choroidal vascularization, which is generally associated with a poor prognosis.[2]

Most patients maintain good visual acuity (VA) throughout the disease.[2]

The evolution of this pathology may be initially characterized by a normal fovea, but with altered electrooculogram (EOG) (pre-vitelliform stage). Then the vitelliform stage, characterized by subretinal lesions. Lipofuscin deposits accumulate in the periphery surrounding the lesion, causing the separation of the photoreceptors of the RPE, giving rise to the vitelirruptive stage. Then healing occurs and choroidal neovascularization with irreversible central vision loss can occur.[3]

Currently, the basis for the diagnosis of Vitelliform Macular Dystrophy is genetic testing. It consists of a mutation in the VMD2 gene, located on chromosome 11q13, which is responsible for encoding the bestrophin protein.

The aim of this paper is to present a case of unilateral vitelliform dystrophy in an elderly patient.

RESULTS

A 89-year-old white female from Salvador came to the service complaining of nonspecific eye discomfort. There was a previous pathological history of esophageal cancer, with chemotherapy completed four months ago. Ocular history reported phacoemulsification with intraocular lens (IOL) implantation for 03 years in both eyes (AO). No relevant data on family history.

At the ophthalmological examination, she presented AV with the best correction equal to 20/25 AO. On biomicroscopic examination in AO, she presented anterior blepharitis and pseudophakia, with topical IOL, without evidence of posterior capsule opacity. Applanation tonometry with value of 15mmHg in AO.

The fundus examination showed a yellowish-white lesion in the macular region in the right eye (OD) (Figure 1). In the left eye (EO), there were no significant findings.

Retinal multimodal evaluation was performed in OA, without significant findings in the OE. On autofluorescence examination, in OD the lesion was short-wave hyperfluorescent (Figure 2). Optical coherence tomography (OCT) of the macula (Figure 3) demonstrated subretinal median reflectivity content causing cupuliform elevation of the nasal parafoveal neurosensory retina, with signs of degeneration of the outermost retinal layers overlying the lesion, and fusiform alteration at the level of the RPE causing posterior optical shading.

The "en-face" OCT image at the level of the external limiting membrane presents a lesion with an internal content of medium reflectivity and high reflectivity contours (Figure 4).

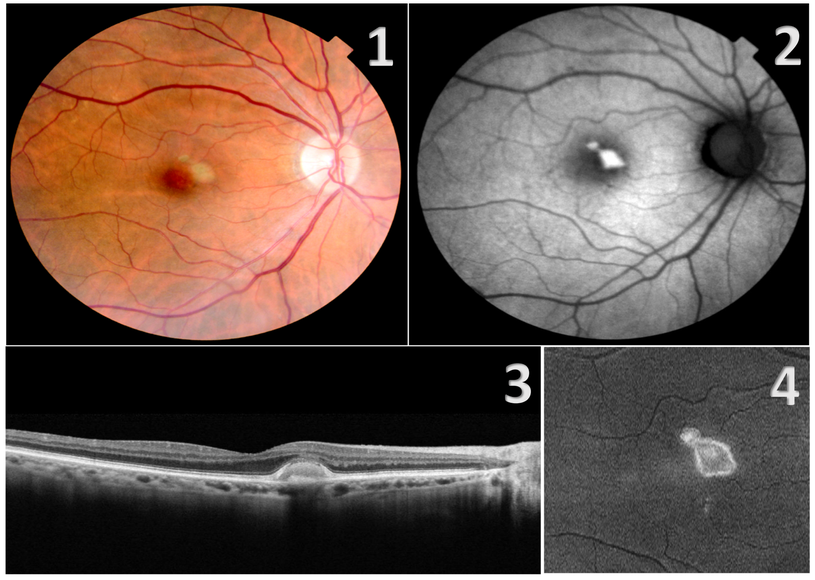
DISCUSSION

We present a case of a 89-year-old female patient with preserved visual acuity, presenting a yellowish-white lesion in the OD macula region visualized at fundus. OCT showed a medium subretinal reflectivity content causing cupuliform elevation of the sensorineural retina. In the contralateral eye there were no significant findings.

Pathophysiology is the coding of a defective protein, bestrofin1. Bestrofin1 is involved in the transport of Ca2 + and Cl- channels in the RPR basolateral plasma membrane. This change leads to a buildup of toxic substances between the photoreceptors and the RPE and, consequently, RPE atrophy and visual impairment. It can range from asymptomatic (subclinical) forms to severe complications such as choroidal neovascularization and disciform scarring.

In this case, the yellowish-white color shown shows the accumulation of lipofucsin in the RPE and in the subretinal region.

Molecular genetic studies are more sensitive in detecting carriers than electrophysiological tests. [5] [6] Differential diagnosis should be made with other retinal dystrophies and maculopathies in general.



Caption: 1) Color retinography showing macular vitelliform lesion. 2) Autofluorescence showing hyperautofluorescent character of the lesion. 3) SD-OCT presenting subretinal medium reflectivity content deposit causing cupuliform elevation of the sensorineural retina, with signs of degeneration of the outermost retinal layers overlying the lesion, and fusiform alteration at the level of the RPE, causing posterior optical shading. 4) Facing OCT at the level of the external limiting membrane presents a lesion with internal content of medium reflectivity and contours of high reflectivity.

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