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Occult macular dystrophy

Distrofia macular oculta

Nabila Terra Demachki[1] https://orcid.org/0000-0003-1214-950X
Karolyna de Andrade Carvalho[2] https://orcid.org/0000-0001-5915-1055
Ricardo Gomes dos Reis[3] https://orcid.org/0000-0003-2680-2995

Abstract

We report a case of bilateral occult macular dystrophy in a 70-year-old woman with progressive low visual acuity, without justifiable fundoscopic or angiographic findings. Imaging tests were done to excluding expansive lesions and electrophysiological tests that suggested the diagnosis.

Keywords: *Macular degeneration; Electroretinogram; Electrophysiology; Visual acuity*

Resumo

Apresentamos um caso de distrofia macular oculta bilateral, em paciente de 70 anos com queixa de baixa acuidade visual progressiva, sem achados fundoscópicos ou angiográficos justificáveis. Foram realizados exames de imagem do sistema nervoso central que afastaram lesões expansivas e testes eletrofisiológicos que sugeriram diagnóstico.

Descritores: Degeneração macular; Eletrorretinograma; Eletrofisiologia; Acuidade visual

[1]Specialization Course in Ophthalmology, Universidade Federal do Rio de Janeiro, Rio de Janeiro, RJ, Brazil.
[2]Residency Program in Ophthalmology, Universidade Federal do Rio de Janeiro, Rio de Janeiro, RJ, Brazil.
[3]Ophthalmology Service, Universidade Federal do Rio de Janeiro, Rio de Janeiro, RJ, Brazil.

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Introduction

Occult macular dystrophy consists of progressive loss of sight [1, 2] due to the loss of foveal cones [1]with abnormal focal macular eletroretinogram (ERG) associated with fundoscopy, fluorescein angiography, and normal full- field ERG.[2-4]

It was initially described as an autosomal dominant disease of hereditary nature.[5]Among the mapped genes is RP1L1[1-3] encoding a photoreceptor of 2400 amino acids.[1]It is known that the existence of this mutation has great importance in the progression in the disease.[2]However, mutations of RP1L1 are not exclusive to occult macular dystrophy, and are also associated with phenotypes of retinitis pigmentosa.[1]

Two forms of occult macular dystrophy are currently described, being: hereditary and sporadic.[3]In sporadic form, a defect occurs in the junction of the external and internal segments of the photoreceptors.[4]

The onset of symptoms may be extensive (6-81 years), with loss of visual acuity and color vision.[3] The diagnosis of the disease is usually late because of the normality of fundoscopy,[3] since even in advanced stages it remains unchanged.[4] The good appearance of the eye fundus is probably due to the good function of the retinal pigment epithelium.[4, 6]

In order to define the diagnosis early and at a lower cost, suspicious cases should be evaluated following the order of Optical Coherence Tomography (OCT), followed by Fluorescein Angiography (FA), full field ERG, multifocal ERG, and later more advanced tests such as Computerized Tomography (CT) or Magnetic Resonance Imaging (MRI) of the skull.[3]The genetic test is the most accurate and difficult to access diagnostic test. Thus, the diagnosis depends on multimodal ophthalmic examination.[3]

To date, there is no effective treatment because it is an idiopathic form of macular degeneration.[3]

Case report

IRN, female, 70 years old, housewife from Rio de Janeiro sought care due to dissatisfaction with facectomy in with left eye (LE) and progressive low visual acuity (LVA) in both eyes. To the examination, she presented corrected visual acuity of 20/200 in the right eye (RE), and 20/400 in the LE, biomicroscopy of the anterior segment demonstrating nuclear cataract 2+/4+ in the RE, and topic intraocular lens in the LE, aplanation tonometry within normality in both the eyes (BE). Fundoscopy evidencing optic discs with regular contours and physiological excavation, preserved vascular arches, preserved foveal brightness, and inferior paving stone degeneration in BE (Figure 1).

In the case of LVA in both eyes, with non-specific fundoscopy, foveal atrophy in OCT, ERG with reduced amplitude, no abnormalities in imaging, PEV or AF examinations, a diagnosis of occult maculopathy was suggested. The genetic test was not carried out due to the difficulty in accessing the exam.

Discussion

Several studies have shown that the characteristic age of onset of occult macular dystrophy varies between 6 and 81 years, [3]and does not usually progress after 60 years of age.[7] In our report, the patient was diagnosed at age 70, corresponding to the age range mentioned.

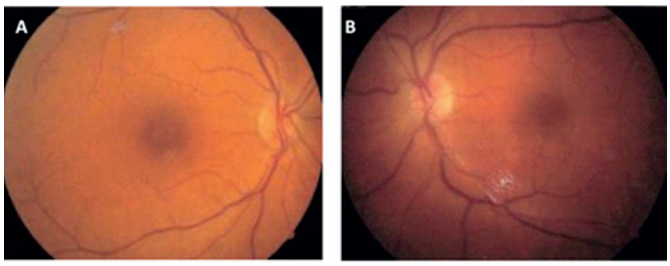

Figure 1: Retinography of the right eye (A) and left eye (B)

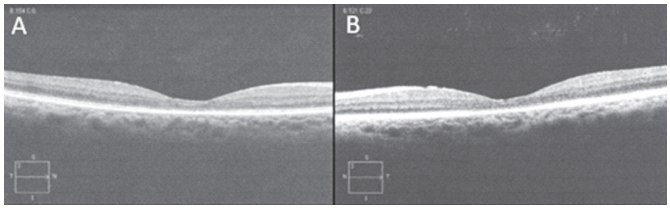

Figure 2: Optical Coherence Tomography of the right eye (A) and left eye (B)

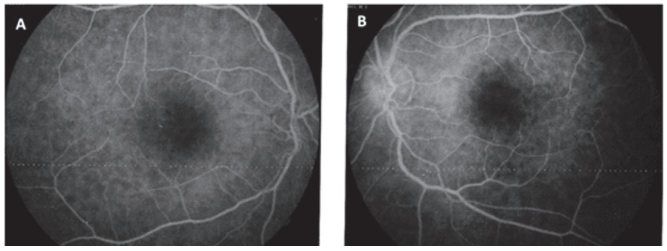

Figure 3: Fluorescein angiography of the right eye (A) and left eye (B)

In occult maculopathy there is progressive worsening of visual acuity with fundoscopy and FA generally normal, contributing little to the diagnosis. FA may show hyperfluorescence, especially in advanced disease,[4] with a circular signal around the fovea, which may mean lesion in the photoreceptors or retinal pigment epithelium.[7] In the case described, no change was observed in fundoscopy and FA.

OCT is essential in the diagnosis of occult macular dystrophy because it demonstrates the characteristic aspect of bilateral loss of the ellipsoid zone in the central region.[3- 5, 8] Based on the tomographic findings, it is known that occult dystrophy is related to photoreceptor deformities.[2] Thus, the pattern of visual loss is directly related to the degree of involvement of cones and rod cell photoreceptors.[1]

The patient in the case reported showed thinning of the foveal region in the OCT, a finding that corroborates the diagnosis of occult dystrophy. However, although characteristic, the loss of the ellipsoid zone is not pathognomonic of occult macular dystrophy, [5] and can still be found in exposure to alkyl nitrite and tamoxifen, and in solar retinopathy.[8]

Electrophysiological and genetic tests are important in differentiating occult macular dystrophy from dystrophy of cones and rods, and from achromatopsia. [8]

PEV in this condition may present with a reduction in amplitude or increase in latency, a non specific findin, but an additional finding in the differential diagnosis of amblyopia and optic nerve diseases such as retrobulbar optic neuritis.[5, 9] In the case presented, PEV was normal.

According to Fujinami et al. the multifocal ERG of occult macular dystrophy characteristically demonstrates alteration in

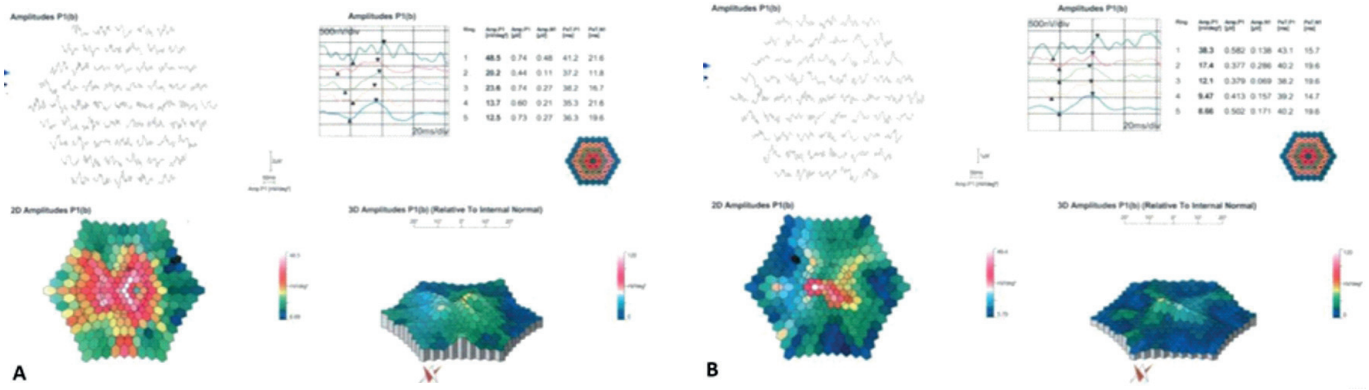

Figure 4: Multifocal eletroretinogram of the right eye (A) and left eye (B)

the 15th centrals.[4] Our patient presented normal full-field ERG in the RE and subnormal in the LE, and multifocal ERG with bilateral central reduction, similar to cases found in the literature. [7,10,11]

Microperimetry offers better ability to evaluate scotomas and fixation-related visual deficit in cases of occult macular dystrophy when compared to other perimetric techniques.[12] It usually shows a decrease in foveal sensitivity.[13]

We reported the case of a patient with occult macular dystrophy diagnosised with clinical examinations, electrophysiological tests, OCT and FA even in the absence of genetic tests. We emphasize the importance of this differential diagnosis in symptomatic cases with few or no fundoscopic finding.

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Corresponding author
Nabila Terra Demachki
Rua Sorocaba, 305 – apto 803
Botafogo – Rio de Janeiro/ RJ
CEP: 22271-110
E-mail: nabilademachki@gmail.com