OCCULT MACULAR DYSTROPHY: A CASE REPORT.

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PURPOSE

The aim is to report a case of Occult Macular Dystrophy in a female patient without previously ocular diseases.

METHODS

Review of the patient's medical record.

CASE REPORT

A 53-year-old woman presented with a 3-month history of bilateral visual loss, predominantly in the right eye. The patient's best corrected visual acuity was 20/400 in OD and 20/200 in OS. Biomicroscopy showed no changes. IOP was 12 mmHG in both eyes. Fundoscopy showed no changes consistent with low vision (Figures 1 and 2).

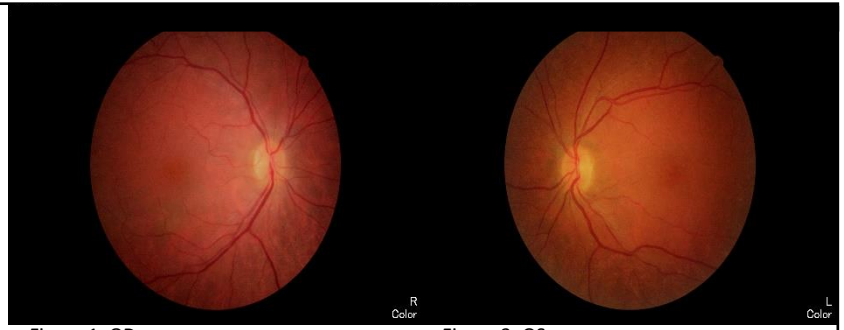
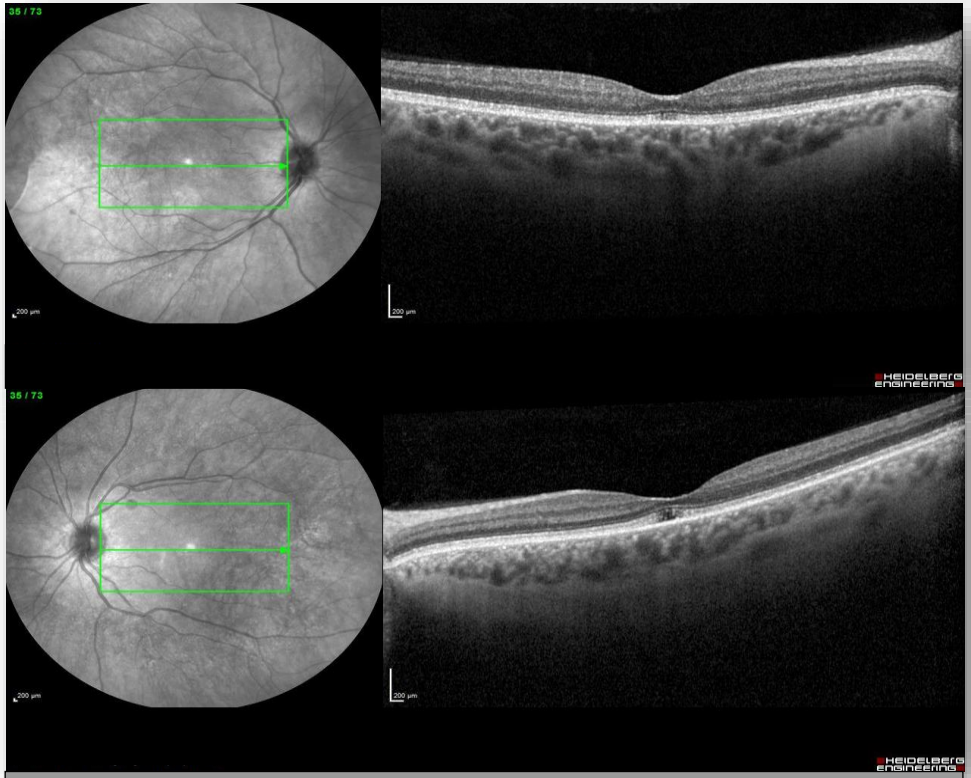


Figure 1: OD Figure 2: OS

Based on the tomographic findings (Figures 3 and 4) with cavitation in the outer retinal layers, the multifocal ERG (Figure 5) with absence of foveal peak and significant decrease in amplitude, and excluding other hypotheses such as Mactel, tamoxifen use, foveolitis, and the negative results of the infectious and inflammatory screening tests, we concluded that the main diagnostic hypothesis was occult macular dystrophy (OMD).



Figures 3 and 4: OCT shows cavitations in the outer layers of the retina in both eyes, with no changes in the other layers.

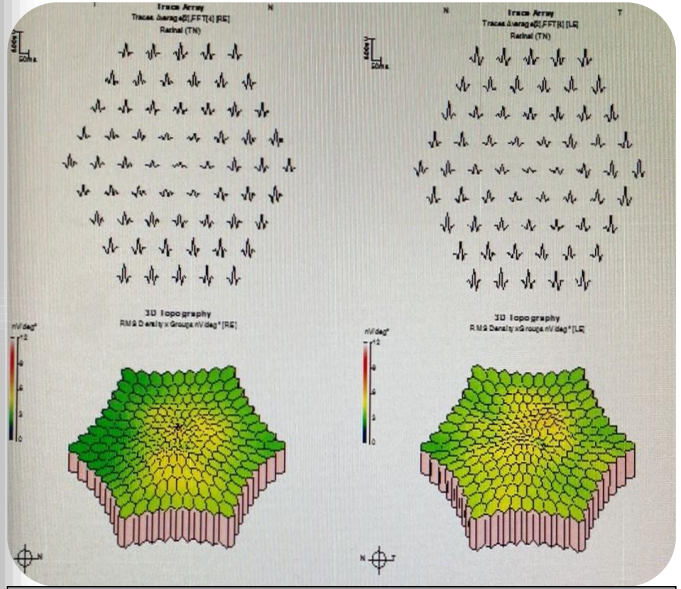


Figure 5: with an absence of the foveal peak and a significant decrease in amplitude

DISCUSSION

Occult macular dystrophy (OMD) is a condition that causes progressive vision loss[1,2] due to the loss of foveal cones[1]. It is associated with abnormal focal macular electroretinogram (ERG) but appears normal in fundoscopy, fluorescein angiography, and full-field ERG[2,4].

Symptoms can appear at any age, ranging from 6 to 81 years, and include loss of visual acuity and color vision[3]. The disease is often diagnosed late due to the normality of fundoscopy[5], which remains unchanged even in advanced stages. The fundus' good appearance is likely due to the retinal pigment epithelium's good function[6].

To make an early diagnosis, suspected cases should be evaluated in the following order: Optical Coherence Tomography (OCT) [6,7], followed by Fluorescein Angiography, ERG full-field, and multifocal.

Currently, there is no effective treatment for this idiopathic form of macular degeneration [1,7].

REFERENCES

1. Davidson AE, Sergouniotis PI, Mackay DS, Wright GA, Waseem NH,Michaelides M, et al. RP1L1 variants are associated with a spectrum of inherited retinal diseases including retinitis pigmentosa and occultmacular dystrophy. Hum Mutat. 2013;34(3):506–14.
2. Fu Y, Chen KJ, Lai CC, Wu WC, Wang NK. Clinical features in a case of occult macular dystrophy with rp1l1 mutation. Retin Cases BriefRep. 2019;13(2):158-161.
3. 4. Fujinami K, Tsunoda K, Hanazono G, Shinoda K, Ohde H, MiyakeY. Fundus autofluorescence in autosomal dominant occult maculardystrophy. Arch Ophthalmol. 2011;129(5):597–602.
4. Hanazono G, Ohde H, Shinoda K, Tsunoda K, Tsubota K, MiyakeY. Pattern-reversal visual-evoked potential in patients with occultmacular dystrophy. Clin Ophthalmol. 2010; 4:1515–20.
5. Kitaguchi Y, Kusaka S, Yamaguchi T, Mihashi T, Fujikado T. Detectionof photoreceptor disruption by adaptive optics fundus imaging and Fourier-domain optical coherence tomography in eyes with occult macular dystrophy. Clin Ophthalmol. 2011;5:345–51.
6. Viana KI, Messias A, Siqueira RC, Rodrigues MW, Jorge R. Structure-functional correlation using adaptive optics, OCT, and microperimetry in a case of occult macular dystrophy. Arq Bras Oftalmol.2017;80(2):118–21.
7. Ahn SJ, Cho SI, Ahn J, Park SS, Park KH, Woo SJ. Clinical and genetic characteristics of Korean occult macular dystrophy patients. InvestOphthalmol Vis Sci. 2013;54(7):4856–63.