Atypical Retinal Dystrophy - Case Report

RETINA CURITIBA
CENTRO DE TRATAMENTO ESPECIALIZADO

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PURPOSE

Report the case of a patient with an atypical retinal dystrophy under investigation

METHODS

Case report through analysis of patient records monitored in the service of Retina Curitiba / Hospital da Visão de Curitiba

RESULTS

TMS, female, 66 years old, referred for ophthalmological evaluation due to mild visual loss. The patient presents a history of idiopathic juvenile artrithis, hearing loss since childhood and amaurotic mother without a definite cause. On ophthalmological examination, best corrected visual acuity (BCVA) was 20/40 in both eyes, slit-lamp biomicroscopy presented without relevant findings and intraocular pressure was 14 mmHg bilaterally. Indirect binocular ophthalmoscopy and angiofluoresceinography showed atrophy of EPR in the posterior pole, pallor of the optic disc, bone spicules in the periphery and vascular thinning.

The patient underwent complementary exams wich indicated: Optical coherence tomography - thinning of the outer layer of the retina; Electroretinography - symmetrical alteration in both eyes with increased response latency and wave amplitude close to the lower limit, compatible with retinal dystrophy affecting photopic and scotopic response; Visual field - with intense decrease in total retinal sensitivity; Audiometry - showed sensorineural hearing loss from 2 kHz bilateral; Impendanciometry / Immitanciometry - presented normal type A curve tympanograms and absent acoustic reflexes in both sides.

Figure 1: Right eye macular OCT

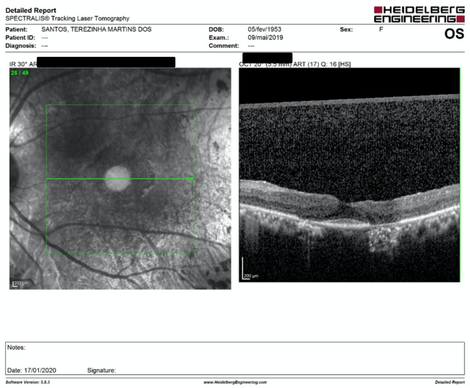

Figure 2: Left eye macular OCT

DISCUSSION

Main diagnosis suggested is Retinitis Pigmentosa (RP), the most common form of hereditary retinal dystrophies. Symptoms can appear only in adulthood, which include: nictalopia, decreased visual field, photopsia, headache and low visual acuity. In fundscopy, we find the classic RP triad: pale optic nerve, vascular attenuation and bone spicules. In the optical coherence tomography exam, we can find edema, fibrosis or atrophy of the retinal layers in the macula, which differs from the atrophy aspect of the external retinal ring layers as described in this case. In general, the electrophysiological aspect in RP can be very variable, from normal to undetectable responses depending on the severity of the case.

In dystrophy of cones and rods we find a greater central involvement with loss of color vision in earlier stages.

We must still take into account the causes of pigmentary pseudoretinosis such as rubella retinosis, syphilis, herpes, toxoplasmosis, paraneoplastic syndromes and drug toxicity.

It is important to remember that Hidden Acute External Zonal Retinopathy (AZOOR), which is still poorly elucidated, is a focal degenerative disease of photoreceptors and should be suspected in case of discrepancy between functional signs and the absence of abnormalities on fundoscopy. Among the diagnostic criteria, there is the sudden appearance and rapid worsening of central and paracentral scotomes, absence of white spots, atrophy of the epithelium of the retinal pigment epithelium (in 43% of patients), and loss of the ellipsoid zone in the areas corresponding to the scotomas.

Finally, determining the genetic abnormality of retinal dystrophy can help the diagnosis, predict the prognosis and family risks and suggest the best treatment. Thus, a complete patient assessment is essential for early diagnosis and therapeutic intervention in order to slow down the clinical manifestations of the disease, prolonging the patient's quality of life.

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