Familial Exudative Vitreoretinopathy: Case Report

RETINA CURITIBA
CENTRO DE TRATAMENTO ESPECIALIZADO

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PURPOSE

Report the case of a patient with a diagnosis suggestive of familial exudative vitreoretinopathy.

METHODS

Case report through analysis of patient records monitored in the service of Retina Curitiba / Hospital da Visão de Curitiba for 6 years.

RESULTS

LSR, female, 14 years old, without comorbidities, born at term of cesarean delivery without complications. In 2013, at the age of 8 years old, she was referred to the ophthalmology service due to difficulties in school performance. Best corrected visual acuity (BCVA) was 20/40 in the right eye and 20/400 in the left eye (LE), slit-lamp biomicroscopy presented without changes and binocular indirect ophthalmoscopy showed vitreous opacities and temporally dragged macula and retinal vessels in both eyes. The findings raised the hypothesis of familial exudative vitreoretinopathy (FEVR) and retinopathy of prematurity (ROP), with ROP subsequently excluded due to clinical evolution and absence of perinatal changes. She was followed up for 6 years, evolving with 10 diopters exotropy in LE and maintained BCVA.

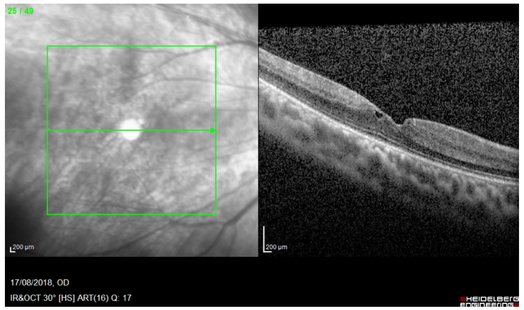

Figure 1: Right eye macular OCT

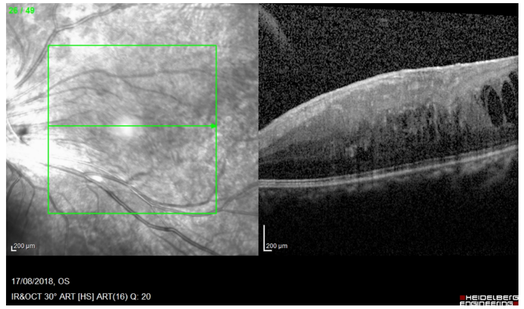

Figure 2: Left eye macular OCT showing important retinal distortion

DISCUSSION

FEVR is a rare inherited disease, usually with an autosomal dominant pattern, characterized by failure of peripheral retinal vascularization. Vascular changes are similar to those found in ROP, which is an important differential diagnosis to be excluded in the absence of perinatal disorders. FEVR generally presents asymmetrical severity of avascular peripheral retina and varied penetrance within the same family, with changes in vascularization present since birth, with late progression or stabilizing in early adulthood. Through clinical and complementary exams, such as indirect binocular ophthalmoscopy and angiofluoresceinography, it is possible to identify features suggestive of the condition, such as peripheral avascular changes, dragged vessels – usually temporally - and macular ectopia. Treatment is performed through serial examinations and management of retinal changes.

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