DIAGNOSIS AND TREATMENT OF GYRATE ATROPHY: A CASE REPORT.

Allan Gomes da Silva[1]; Philipe Franco do Amaral Tafner [1]; Deborah dos Santos Musich[1]; Fabiana Jallad Sallum[1]; Gabriel Castilho Sandoval Barbosa[1]; Julia de Ávila Fowler[1]; Priscila Nascimento Hess [2], Julio Zaki Abucham Neto[3].

1 Resident in Ophthalmology of Faculdade de Medicina do ABC (FMABC). 2 Retina and Vitreous fellow of Faculdade de Medicina do ABC (FMABC). 3 Chief of Retina and Vitreous sector of Faculdade de Medicina do ABC (FMABC).

PURPOSE

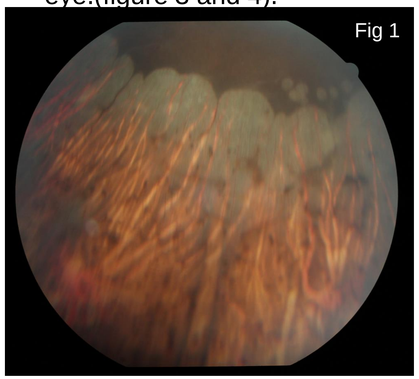
To describe and discuss the management of a young female with gyrate atrophy and yours complications.

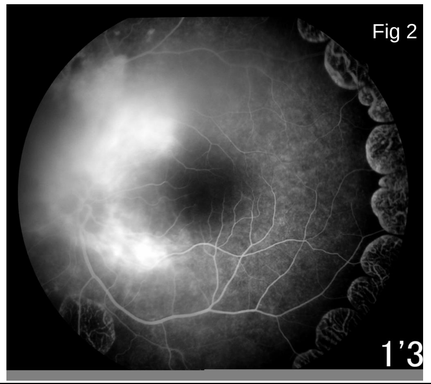
INTRODUCTION

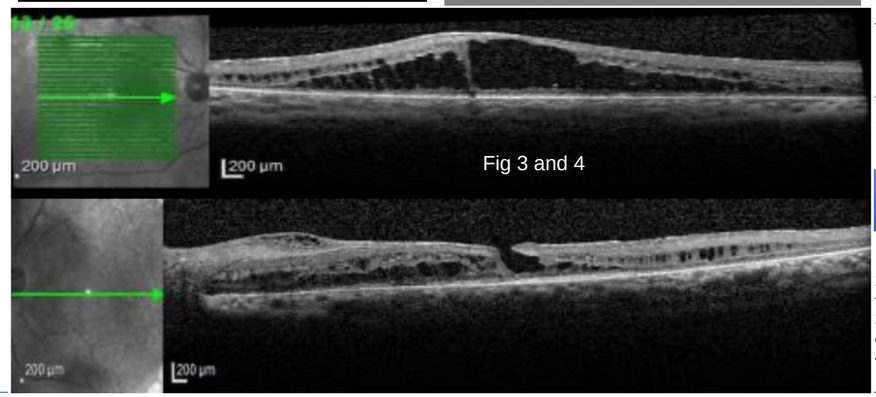
Gyrate Atrophy is a rare autosomal recessive genetic disorder of the retina. It`s characterized by early onset in childhood, with progressive chorioretinal degeneration. Fundus exam reveal characteristic circular lesions with well-defined borders that initiate at the peripheral fundus and progress centripetally toward the posterior pole.[1-4] Patients usually presents early axial myopia, cataract (posterior subcapsular cataract, most frequently) and nyctalopia in the first decade of life.[1;2] Continuous visual loss is correlated to coalescence of the atrophic lesions as the disease progresses. Advanced stage occurs with macular involvement, generally because cystoid macular edema, epiretinal membrane and choroid neovascularization.[2;3] Biochemically defined by a lack of the ornithine-delta-aminotransferase enzyme, which course with high systemic ornithine levels.[3] There are only a few papers published about this rare genetic disorder.

RESULTS

A 20-year-old woman presented with progressive visual loss since childhood. Reported as family history, parents with consanguinity. On examination, Visual acuity with best correction of 20/250 OD and 20/80 OS, was measured. Biomicroscopy highlighted posterior pole cataract in both eyes and Iris atrophy OD. Fundus exam reveals abnormal reflectivity of the macular surface, thinning vessels, ghost vessels and sharply demarcated circular areas of chorioretinal atrophy with hyperpigmented margins in the mid to distal periphery in both eyes. On the left eye a macular hole was hypothesized.(figure 1 and 2). Optical Coherence Tomography demonstrated epiretinal membrane in both eyes and revealed a macular hole on the left eye.(figure 3 and 4).







The diagnosis hypothesis of Gyrate Atrophy was suggested and treatment with dorzolamide hydrochloride was initiated. Patient was reevaluated after a month with no response to treatment, presenting visual acuity of 20/250 in both eyes and no other changes on the exam. Intra ocular Anti-VEGF was prescribed.

DISCUSSION

Ornithine-delta-aminotransferase enzyme (OAT) is expressed among several tissues. At the retina, high levels of ornithine and its metabolites due to the lack of OAT leads to degeneration of photoreceptors and chorioretinal atrophy. Published reviews demonstrate high levels of systemic ornithine 10 to 20 times higher than patients with regular OAT, especially in Aqueous Humor, cerebrospinal fluid and urine. However, no relation with ornithine levels and progression of the disease was found. Gyrate Atrophy is hard to make early diagnosis due to the absence of new-born screening exams being able to detect the disease. Moreover, there are only a few or none abnormalities at the first weeks of life. The disease progresses as patient grows and realize the first symptoms, the ophthalmologic exam allied to fluorescein angiography and OCT are helpful to diagnose and manage the disease. Although, early diagnosis is not easy to achieve, it is very important to stop progression, as some patients can respond to supplementation of pyridoxine (B6) and diet restriction of arginine.

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