A 14-Year-Old Boy Referred for Abnormal Fundus Findings

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Introduction:

A 14-year-old boy was referred to the retina clinic for abnormal fundus findings and concern for retinitis pigmentosa. He was diagnosed with amblyopia in his left eye in early childhood. He reports gradually worsening vision in his right eye over several years. He had never had eye surgery, including strabismus surgery, and his parents had not noted any abnormal eye movements.

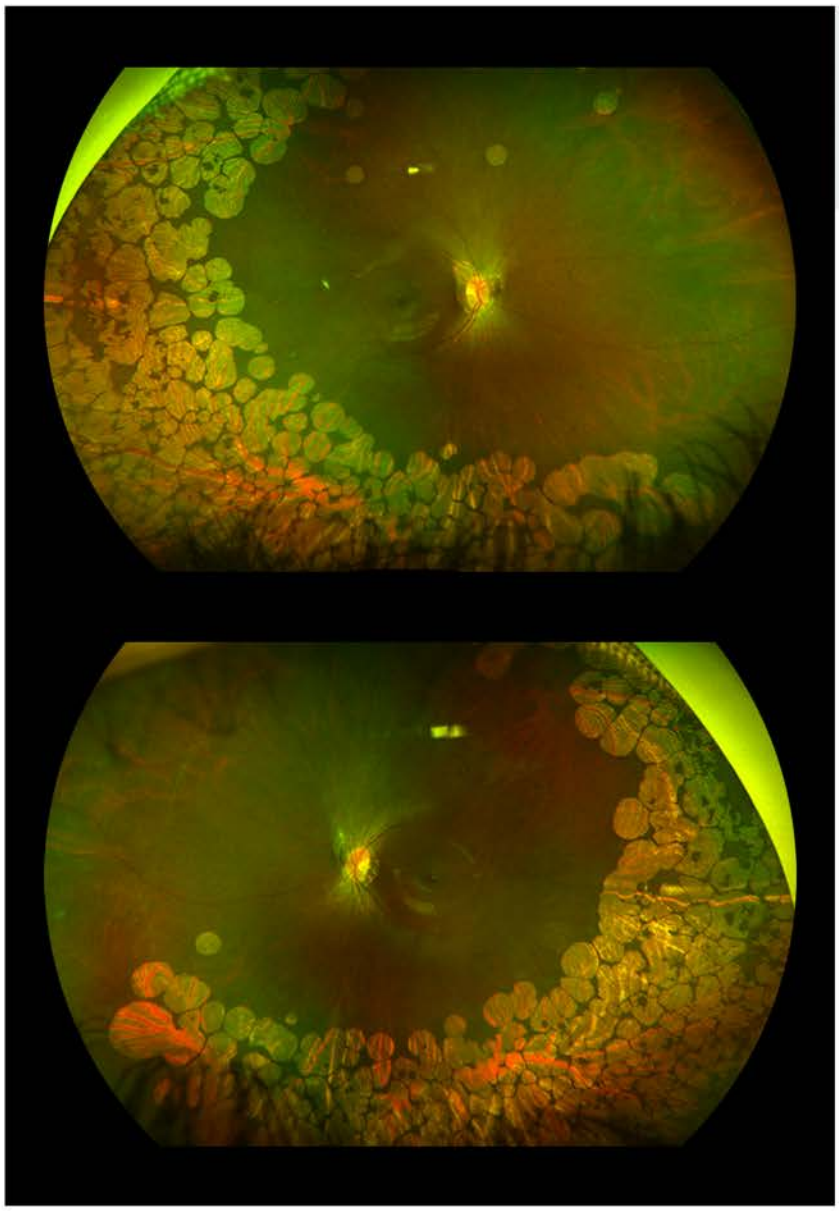

Figure 1. Bilateral widefield pseudo-color fundus images.

Exam:

Best corrected visual acuity was 20/50 in the right eye and 20/150 in the left eye. Pupillary responses were normal to light and accommodation in both eyes, and there was no relative afferent pupillary defect. Confrontational visual fields revealed mild generalized constriction in both eyes. On dilated funduscopic examination, both eyes demonstrated profound, nearly confluent, round patches of retinal pigment epithelial atrophy throughout the temporal and inferior periphery with some sparing of the nasal and superior periphery and complete sparing of the maculae, which were normal bilaterally. There was tilting of the optic discs with peripapillary atrophy in both eyes. (See Figure 1)

Diagnosis:

To aid in diagnosis, fundus autofluorescence was obtained, which demonstrated a hyper-autofluorescent ring in the macula centered on the fovea in both eyes as well as hypo-autofluorescence throughout the temporal and inferior periphery in both eyes corresponding to the areas of RPE atrophy (See Figure 2). Spectral domain optical coherence tomography also demonstrated macular edema in both eyes, greater on the right (see Figure 3). The patient was sent for an electroretinogram, for which all responses, including rod specific, maximal flash scotopic, photopic, and 30 Hz flicker, were non-recordable bilaterally, consistent with severe,

widespread cone and rod dysfunction. Plasma ornithine levels were ordered, but due to social factors, these have not been able to be obtained.

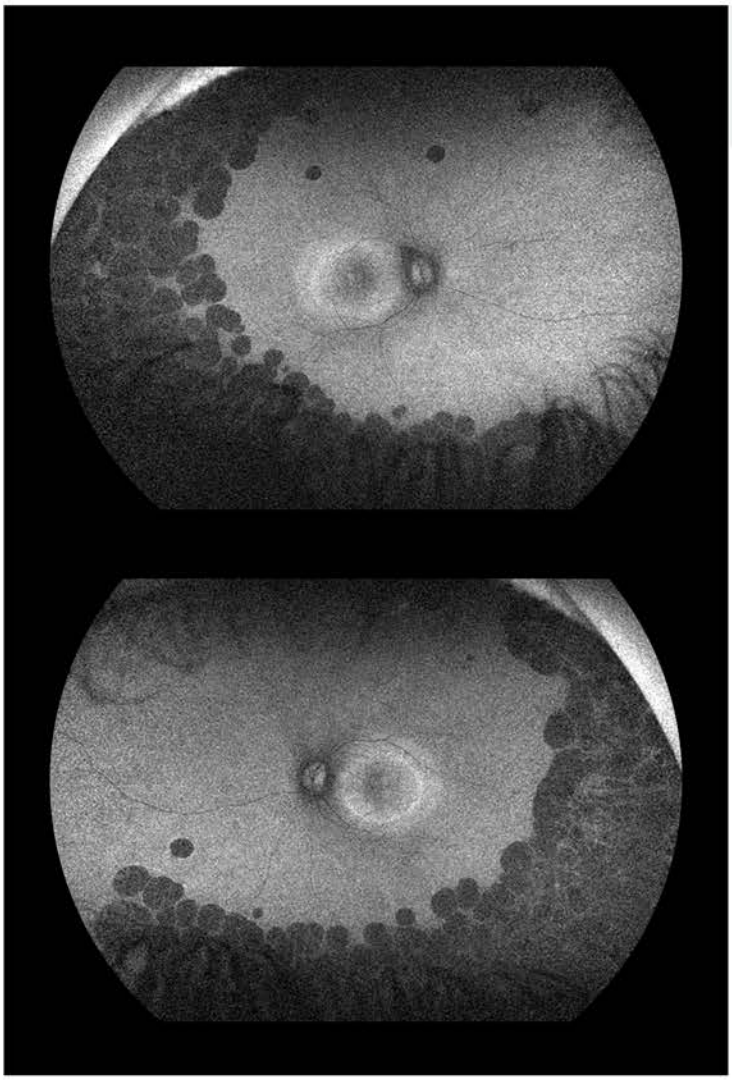

Figure 2: Fundus autofluorescence images of both eyes.

Discussion:

This represents a case of gyrate atrophy of the choroid and retina, more commonly shortened to gyrate atrophy. The differential diagnosis is limited due to the striking, near-pathognomonic pattern of symmetric peripheral retinal atrophy in a young patient but may include choroideremia, retinitis pigmentosa, myopic degeneration, cobblestone degeneration, multifocal choroiditis, late-onset retinal degeneration (LORD), and congenital stationary night blindness.

Gyrate atrophy is a retinal dystrophy that arises from a deficiency of ornithine aminotransferase, which is carried on the *OAT* gene on chromosome 10q26.[1-3] It has autosomal recessive inheritance and leads to a number of ocular findings, including early cataracts, myopia, progressive chorioretinal degeneration, and cystoid macular edema.[1] Patients frequently report nyctalopia and progressive loss of peripheral vision.[1] Due to the deficiency in ornithine aminotransferase, ornithine is not properly metabolized and reaches high concentrations in the serum. Reports cite a 10- to 20-fold increase in plasma ornithine concentrations.[1] The highest prevalence of gyrate atrophy in the world by current estimates is in Finland.[4]

Early in the course of gyrate atrophy, large areas of atrophy of the retinal pigment epithelium (RPE) and choriocapillaris develop in the far periphery.[2] In late stages, peripheral atrophic areas coalesce to form a scalloped border between the healthy and disease RPE, with a marked absence of outer retina, RPE, choriocapillaris, and medium-to-large choroidal vessels.[2] There is often hyperpigmentation of the remaining RPE. Systemic finding of gyrate atrophy may include mild cognitive impairment and language delay, although these are much less common features.

ERG testing in eyes with gyrate atrophy show diminished rod and cone responses, progressing to completely extinguished responses.[1]

There is no definitive treatment for gyrate atrophy, however treatments aimed at lowering plasma concentrations of ornithine have been demonstrated to slow the progression of chorioretinal lesions and, to a lesser extent, the progressive loss of visual function.[3,5] This is achieved by following an arginine-restricted and low-protein diet, as arginine is the precursor to ornithine in the urea cycle.[3] Arginine can be found in a variety of foods such as nuts, seeds, meat products, oats, cereals, corn, brown rice, dairy, chocolate, and seaweed. There is modest evidence to support dietary supplementation with vitamin B6, cofactor in the metabolism of ornithine by OAT, to attempt to increase residual enzyme activity.[6,7]

References:

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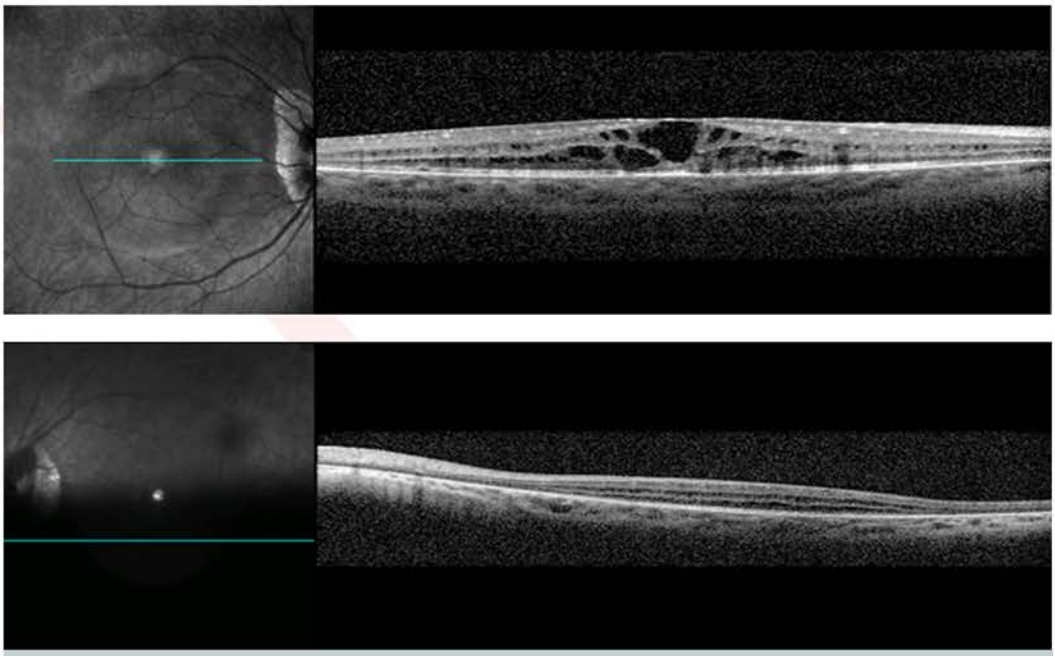

Figure 3. SD-OCT of the macula in both eyes, centered on the fovea in the right eye demonstrating macular edema, and inferior to the fovea in the left eye, demonstrating loss of outer retinal layers.

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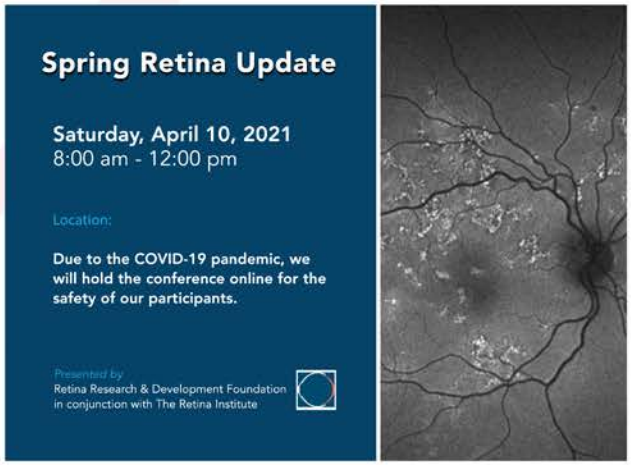

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