A 16-Year-Old Caucasian Male Referred for Retinitis Pigmentosa Evaluation

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Introduction:

A 16-year-old Caucasian male is initially referred for second opinion in evaluation for retinitis pigmentosa. He presents with complaints of blurred vision, reportedly worsening over the prior six months. Review of systems was negative, and his past ocular history is notable for a history of patching as a child in the treatment of amblyopia. Family history is unremarkable for any ophthalmologic pathology. His birth history was notable for a peripartum infection in his mother who was treated with antibiotics, however, our patient was without concern for infection at the time. He has no other medical comorbidities.

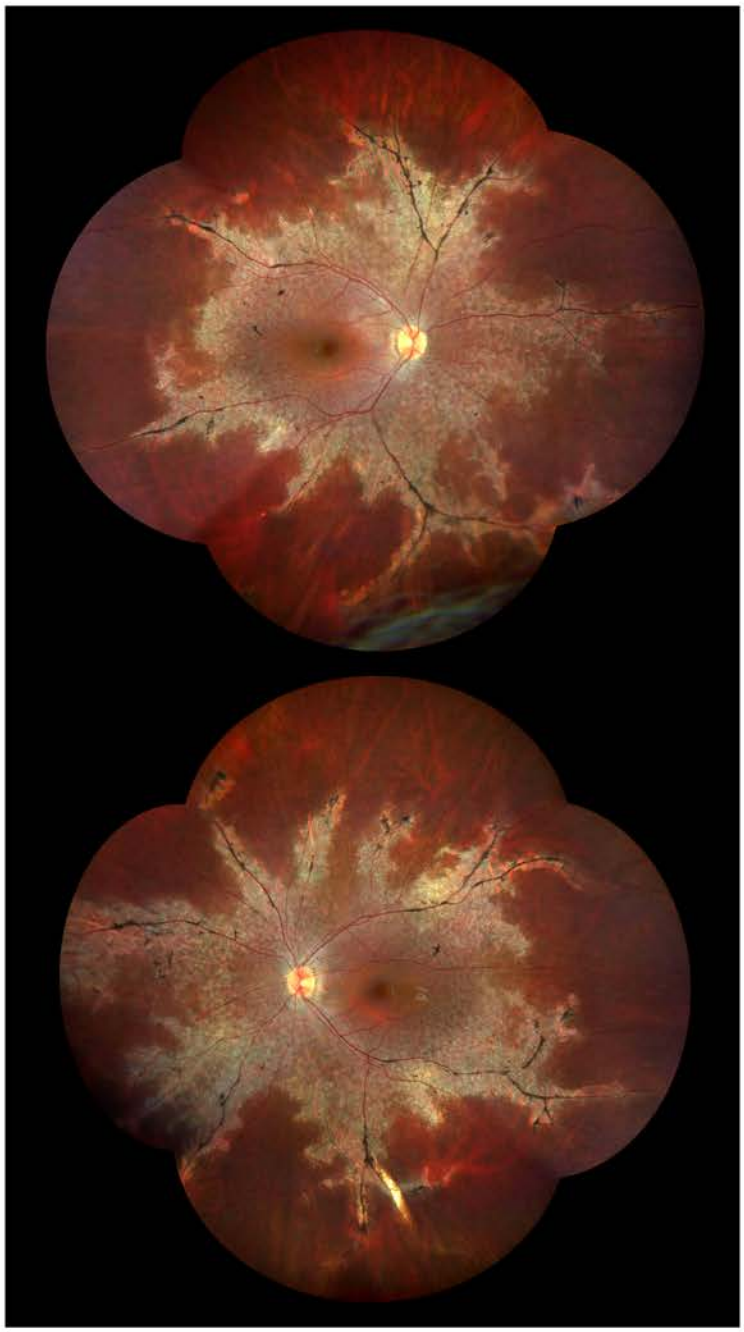

Figure 1: Fundus photos show significant parafoveal yellowing with paravenous pigmentation and chorioretinal scarring bilaterally.

Examination and Work-Up:

At presentation, visual acuity was 20/25 in the right eye and 20/20 in the left eye with intraocular pressures of 14 and 13, respectively. The anterior segment exam was unremarkable and specifically without cataract or anterior chamber inflammation. The anterior vitreous was notable for trace pigmented cells in both eyes. Optic nerves were pink and sharp. There was notable yellowing of the parafoveal areas with paravenous pigmentation and chorioretinal scarring extending into the periphery bilaterally. (Figure 1) Optical coherence tomography of the macula was notable for parafoveal outer retinal thinning. (Figure 2) Fundus autofluorescence was significant for a parafoveal ring of hyper-autofluorescence and peripheral hypo-autofluorescence.

(Figure 3) The patient presented with genetic testing completed using the Blueprint Genetics (Seattle, WA) panel with no pathologic variants detected. Our patient was sent for an electroretinogram (ERG) and focused lab work–up — complete blood count, angiotensin converting enzyme, syphilis testing, and QuantiFERON gold. Labs were within normal limits or negative. The ERG demonstrated mildly reduced cone function. A Goldman Visual Field demonstrated generalized constriction with paracentral scotomas greater in the right eye than the left.

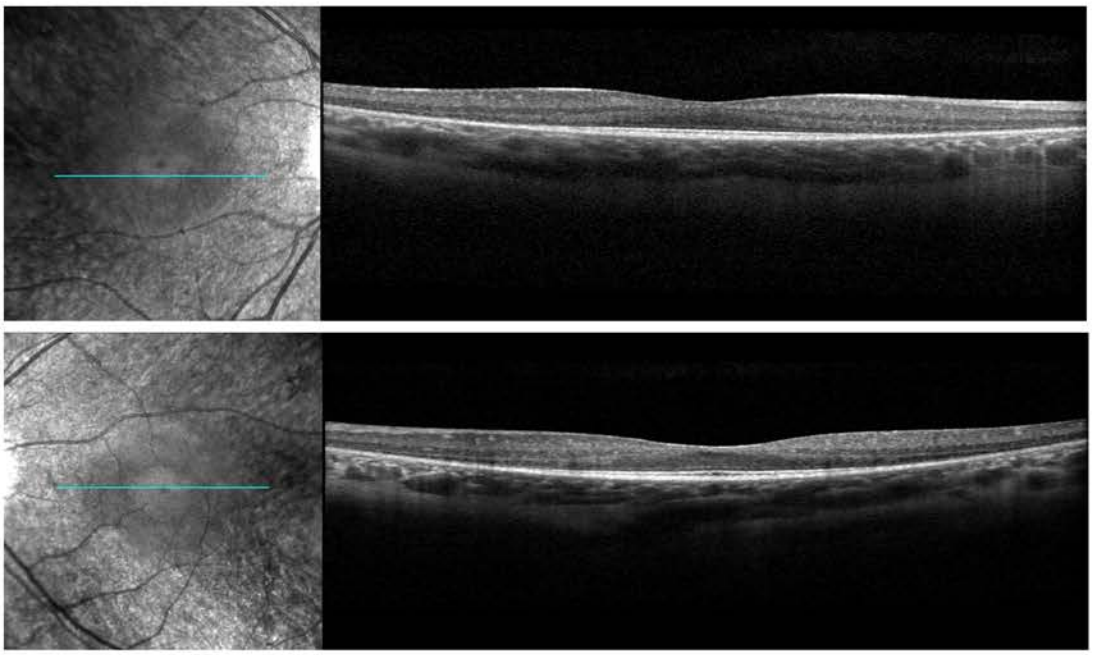

Figure 2: Optical coherence tomography of the macula shows parafoveal outer retinal thinning bilaterally.

Discussion:

Our patient presented with reported vision changes over the last six months and a previous diagnosis of retinitis pigmentosa. Examination findings were most notable for parafoveal atrophy with paravenous pigmentation and chorioretinal atrophy extending into the periphery. Visual function was centrally intact though notable visual field defects were detected. Given the characteristic fundus findings, a diagnosis of pigmented paravenous retinochoroidal atrophy (PPRCA) was given.

First described in 1937 by Hewitson-Brown, PPRCA is a poorly understood disorder. Its etiology is presumed to be an inflammatory insult leading to delayed retinal development. Various more specific infectious and inflammatory causes have been postulated as being associated including sarcoidosis, syphilis, tuberculosis, and others. PPRCA is believed to occur sporadically, however, there have been genetic associations cited.[1] Genes implicated include CRB1, CRK, and HK1, all of which have also been implicated in retinitis pigmentosa. [2,3,4]

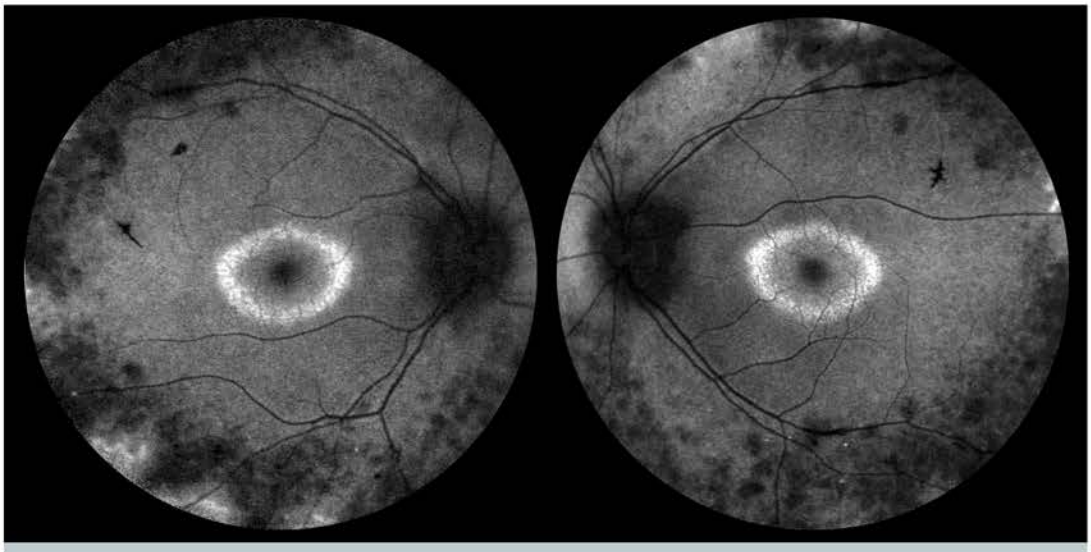

Figure 3: Fundus autofluorescence demonstrates a parafoveal ring of hyper-autofluorescence and patchy peripheral hypo-autofluorescence bilaterally.

Patients typically complain of very mild visual symptoms and uncommonly nyctalopia with visual field deficits varying depending on the extent of retinal involvement. ERGs also vary and may show abnormal rod response, cone response, or combined responses. [1]

On fundoscopic exam, PPRCA is characterized by paravenous pigment clumping with surrounding radial zones of retinochoroidal atrophy. The minority of cases affect the macula but can take on a range of presentations when they do and include macular edema, epiretinal membranes, and atrophy. Fortunately, PPRCA is typically slowly progressive or non-progressive.[1]

References:

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3. Oh JK, Nuzbrokh Y, Lee W, et al. A mutation in CRX causing pigmented paravenous retinochoroidal atrophy. European Journal of Ophthalmology. 2022;32(1):NP235-NP239. doi:10.1177/1120672120957599.

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