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Pigmented Paravenous Chorioretinal Atrophy

A healthy 38 year-old female of Indian descent presented with a complaint of decreased night vision in both of her eyes over the last three years. She reports a surgical history of “lazy” eye repair at 4 years old in India. Her family history is significant for retinitis pigmentosa in her mother and a “retinochoroiditis” in her sister. She has no prescribed medications and her review of systems was negative.

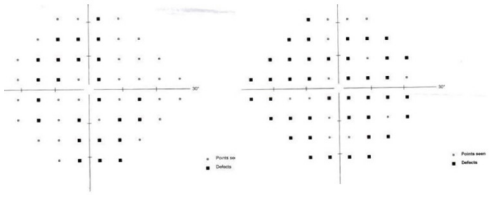

Figure 1: The right eye demonstrates generalized constriction on a 24-2 visual field. The left eye demonstrates geographic scotomas that do not respect the vertical midline on a 24-2 visual field.

On initial presentation, her vision in the right eye was 20/40 and the left eye was 20/20. She had diminished color vision in both eyes being able to identify 6 out of 10 color plates correctly in the right eye and 5 out of 10 color plates correctly in her left eye. An alternating exotropia was present. Her anterior segment exam was unremarkable and pupils were normal without an afferent pupillary defect. Her visual field tests demonstrated generalized depression in her right eye and the left eye had focal areas of depression both superior and inferior that did not respect the vertical midline (Figure 1).

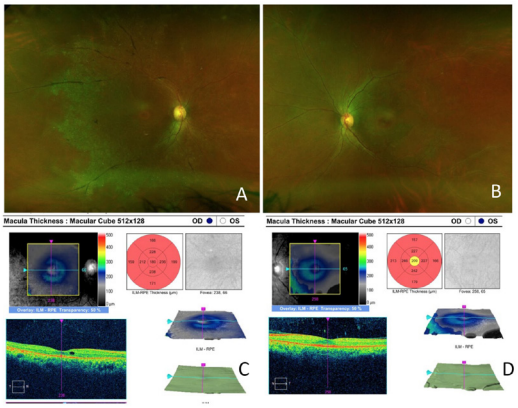

Figure 2: A & B) Fundus photographs demonstrating rpe changes in the macula in both eyes. Bone spicules with retinochoroidal atrophy were also noted along the arcades in both eyes. C & D) OCT demonstrating a small cyst in the right eye along with overt thinning in the

On dilated examination, her optic nerves appeared healthy and her vessels were slightly attenuated. There were subtle macular retinal pigment epithelial (RPE) changes in both eyes. Bone spicules and whitish chorioretinal atrophy were noted along the arcades in the right eye more dominantly than the left. Optical coherence tomography (OCT) of the macula demonstrated overt thinning of the rpe and outer retina in both eyes, in addition a small cyst was noted in the right eye (Figure 2). Fundus autofluorescence demonstrated a dark area of hypoautofluorescence with a rim of hyperautofluorescence (Figure 3). Fluorescein angiography demonstrated areas of hyperfluorescence secondary to window defects overlying areas of atrophy and hypofluorescence in areas blocked by the pigment clumps and bone spicules (Figure 4).

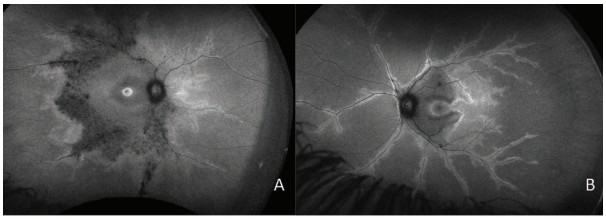

Figure 3: A & B) Fundus autofluorescence demonstrating hypoautofluorescence with a rim of hyperautofluorescence at the transition zone.

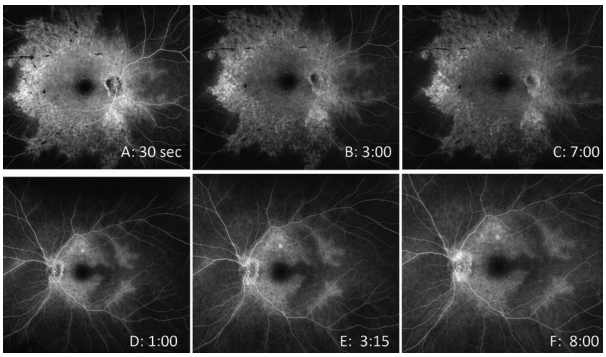

Figure 4: A –F): Fluorescein angiography demonstrating hyperfluorescence overlying areas of rpe atrophy. There is also hypofluorescence where the fluorescence is blocked by pigment clumping. No significant leakage was noted in either eye.

Bone spicules represent pigment migration into the interstitial spaces of the retina from the breakdown of RPE cells. The pigment accumulation tends to be greater surrounding retinal vessels therefore patients tend to develop spicule-shaped deposits or pigmentary cuffing around blood vessels[1]. Although the classic association of “bone spicules” and “pigmentary retinopathy” is with retinitis pigmentosa there are actually numerous conditions that can be associated with these findings. The differential diagnoses for pigmentary retinopathy includes retinitis pigmentosa, pigmented paravenous retinochoroidal atrophy, hydroxychloroquine retinopathy, autoimmune retinopathy, and numerous inflammatory and infectious conditions including serpiginous choroidopathy, sarcoidosis, syphilis, cytomegalovirus, and tuberculous[2-5].

Given the predominance of bone spicules and RPE atrophy primarily along the retinal veins and peripapillary region, pigmented paravenous chorioretinal atrophy (PPCRA) was highest on our differential. However, labwork for tuberculosis, rheumatoid arthritis, lupus, syphilis and HLA-B27 were performed and negative. Baseline exams were performed on her affected mother and sister (Figure 5). Interestingly, her mother’s fundus exam was characteristic for retinitis pigmentosa while her sister’s examination was characteristic for PPCRA. Given the strong family history of pigmentary retinopathy genetic testing was performed on the patient, her mother, and her sister. Interestingly, the only ocular mutation that was found in all family members was GRM6 which was noted to be of uncertain significance.

Pigmented paravenous chorioretinal atrophy (PPCRA) is a rare form of bilateral chorioretinal atrophy characterized by pigment clumps and RPE atrophy distributed along the retinal veins and peripapillary region. It was initially described in 1937 and thought to be of infectious etiology, first noted in a patient with a history of tuberculous and then noted in a second patient with a history of syphilis[3]. However as more cases have been reported multiple causal relationships including genetic, congenital, degenerative, inflammatory, and infectious etiologies have been theorized[2-5].

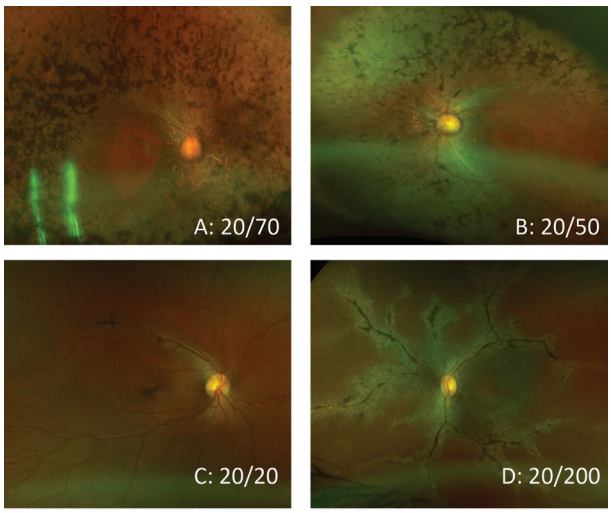

Figure 5: A & B): Fundus photographs of the patient's 64 year old mother demonstrating attenuated vessels and diffuse bone spicule formation in the periphery. C & D): Fundus photographs of the patient's 34 year old sister demonstrating atrophy and pigment clumping primarily along the arcades in both eyes. There is also atrophy noted in the macula of the left eye accounting for the limited vision.

Patients affected by PPCRA are usually asymptomatic however they may develop a decrease in vision, nyctalopia, and visual field loss depending on the extent and location of the atrophy. Approximately 26% of patient have macular involvement with severe disruption of the outer retina and RPE which is the main cause for vision loss[2]. Variable visual field presentations can be noted with PPCRA including geographic scotomas, peripheral constriction, and an enlarged blind spot[3]. The majority of eyes have a symmetrical presentation which tends to be non-progressive or slowly progressive over time[2,4].

Unfortunately, at this time there is no treatment for PPCRA. Annual examinations with visual field testing, fundus autofluorescence, and OCT may be useful to help monitor for slow progression. The management of pigmentary retinal disorders can be challenging given the broad number of inflammatory, infectious, and inherited disorders. At NJ Retina we offer a comprehensive approach beginning with a thorough history, careful examination, and advance imagining techniques combined with experience in order to best evaluate these patients. We fortunately also have access to a genetic testing panel for our patients suspected of having an inherited retinal disorder. As these patients are at risk for vision loss; our empathy, experience, and advanced tools are crucial in providing optimal care and support.

References:

1. *Vela, José I., et al. "Progression of retinal pigmentation mimicking unilateral retinitis pigmentosa after bilateral pars planitis: a case report." BMC ophthalmology 18.1 (2018): 1-3.*
2. *Lee, Eun Kyoung, et al. "Pigmented Paravenous Chorioretinal Atrophy: Clinical Spectrum and Multimodal Imaging Characteristics." American Journal of Ophthalmology 224 (2021): 120-132.*
3. *Huang, Hou-Bin, and Yi-Xin Zhang. "Pigmented paravenous retinochoroidal atrophy." Experimental and therapeutic medicine 7.6 (2014): 1439-1445.*
4. *Murray, A. T., and G. R. Kirkby. "Pigmented paravenous retinochoroidal atrophy: a literature review supported by a unique case and insight." Eye 14.5 (2000): 711-716.*
5. *Kükner, A. Şahap, et al. "Pigmented Paravenous Retinochoroidal Atrophy." Ophthalmologica 217.6 (2003): 436-440.*

At the **forefront** of **clinical research**

NJRetina continuously conducts clinical trials at key locations. Our clinical research coordinators will be happy to discuss the inclusion/exclusion criteria or any other aspect of these studies with you or your patients. If you have any questions, please feel free to contact:

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Enrolling Studies:

Dry AMD
Teaneck and Toms River
Gallego: A Phase II, Multicenter, Randomized, Single-masked, Sham-controlled Study to Assess Safety, Tolerability, and Efficacy of Intravitreal Injections of FHTR2163 in Patients with Geographic Atrophy Secondary to Age-related Macular Degeneration (Gallego)

Diabetic Macular Edema (DME)
Teaneck
Gleam: A Prospective, Randomized, Double-masked, Active Comparator-Controlled, Multi-center, Two-arm, Phase 3 Study to Evaluate the Efficacy and Safety of Intravitreal KSI-301 Compared with Intravitreal Aflibercept in Participants with Visual Impairment Secondary to Treatment-naive Diabetic Macular Edema.

Diabetic Retinopathy
Teaneck
Pavilion: A Phase III, Multicenter, Randomized Study of the Efficacy, Safety, and Pharmacokinetics of the Port Delivery System with Ranibizumab in Patients with Diabetic Retinopathy

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Altitude: A Phase 2, Randomized, Dose-escalation, Observation-controlled Study to Evaluate the Efficacy, Safety, and Tolerability of RGX-314 Gene Therapy Delivered Via One or Two Suprachoroidal Space (SCS) Injections in Participants with Diabetic Retinopathy (DR) without Center Involved-diabetic Macular Edema (CI-DME) (ALTITUDE)

Retinal Vein Occlusion
Toms River
Balaton: A Phase 3, Multicenter, Randomized, Double-masked, Active Comparator-controlled Study to Evaluate the Efficacy and Safety of Faricimab in Patients with Macular Edema Secondary to Branch Retinal Vein Occlusion

Teaneck
Beacon: A Prospective, Randomized, Double-masked, Active Comparator-controlled, Multi-center, Two-arm, Phase 3 Study to Evaluate the Efficacy and Safety of Intravitreal KSI-301 Compared with Intravitreal Aflibercept in Participants with Visual Impairment Due to Treatment-naïve Macular Edema Secondary to Retinal Vein Occlusion (RVO)

Wet AMD
Edison
Opthea Coast: A Phase 3, Multicenter, Double-masked, Randomized Study to Evaluate the Efficacy and Safety of Intravitreal OPT-302 in Combination with Aflibercept, Compared with Aflibercept Alone, in Participants with Neovascular Age-related Macular Degeneration (nAMD)

Toms River
Opthea Shore: A Phase 3, Multicenter, Double-masked, Randomized Study to Evaluate the Efficacy and Safety of Intravitreal OPT-302 in Combination with Ranibizumab, Compared with Ranibizumab Alone, in Participants with Neovascular Age-related Macular Degeneration (nAMD)

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