A 26-Year-Old Female with New Onset Changes in her Vision

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Patient Presentation:

A 26-year-old female was referred to our institute for new onset changes in her vision. She stated that her vision turned red/orange for 45 minutes a few days ago. She complained of some occasional flashes and floaters stable over many years as well as difficulty seeing at night. She has a past history of myopic degeneration, endometriosis, and scoliosis. Per the patient, her grandfather, great grandfather, and 3 great uncles all went blind for reasons unknown. She denied any illicit drug use.

Examination and Testing:

Visual acuity was 20/50 and 20/30 in the right and left eyes respectively. Intraocular pressures were normal. Anterior segment examination had no significant findings. Posterior examination displayed normal macula and optic discs with bilateral arteriole attenuation. In the periphery of both eyes, bone spicule pigmentation was found in the inferonasal area of the right eye and the inferior region of the left eye. (Figure 1) OCT was normal architecture in both eyes. Humphry visual field 30-2 testing revealed nonspecific defects clustered superiorly in the left eye and superotemporally in the right eye. This correlated well with location of the pigmentary changes on the examination. (Figure 2) Electroretinogram fixed luminance flicker, which tests for the function of rods in the retina, revealed decreased signal strength in both eyes. (Figure 3)

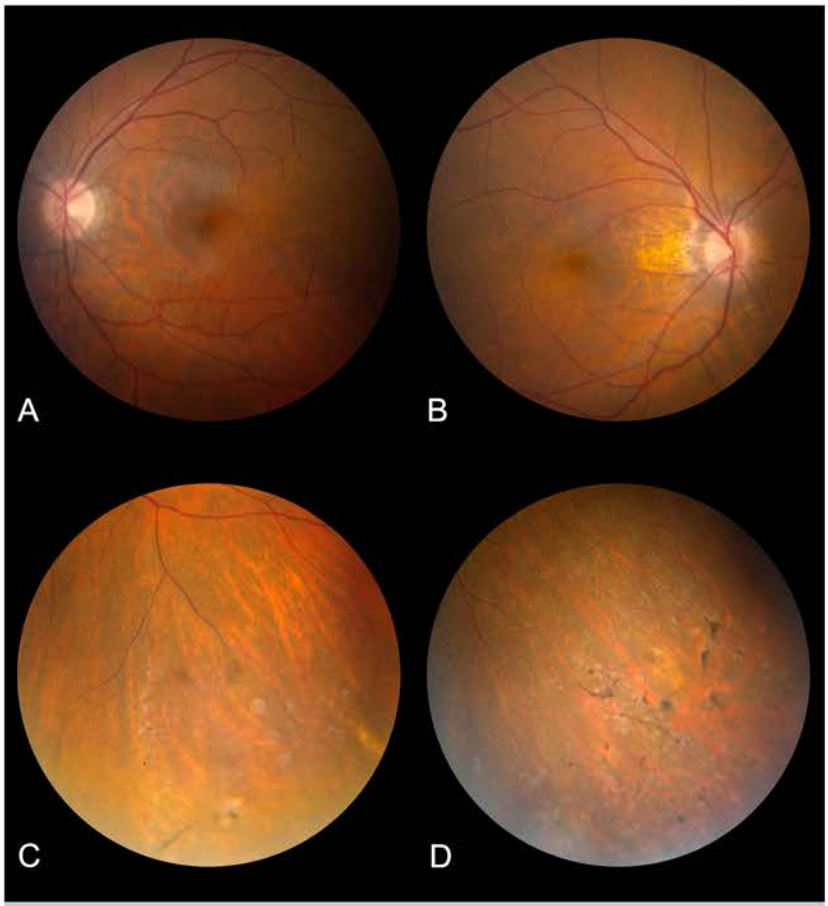

Figure 1 – The posterior pole demonstrated moderate arteriole attenuation in both left (A) and right (B) eyes. Peripheral examination revealed sectoral bone spicule RPE pigmentary changes in left (C) and right (D) eyes.

Differential Diagnosis:

The differential for peripheral pigmentary changes in a bone spicule pattern can encompass a wide variety of pathology. Retinitis pigmentosa is the highest on the differential in a patient with night vision changes and family history of vision loss. Vitamin A deficiency can also cause night vision problems and pigmentary changes. A history of congenital or remote syphilis and rubella can result in pigmentary retinopathy. Diffuse unilateral subacute neuroretinitis is a progressive parasitic disease affecting the outer retina and RPE that can also leave pigmentary changes. A history of trauma can cause changes in the retinal pigment epithelium (RPE) in

the form of scarring and hypertrophy, however a bilateral presentation would be unusual. Chronic CSR can cause guttering and RPE pigmentation in the inferior region, but a normal OCT and fundus examination would be unlikely.

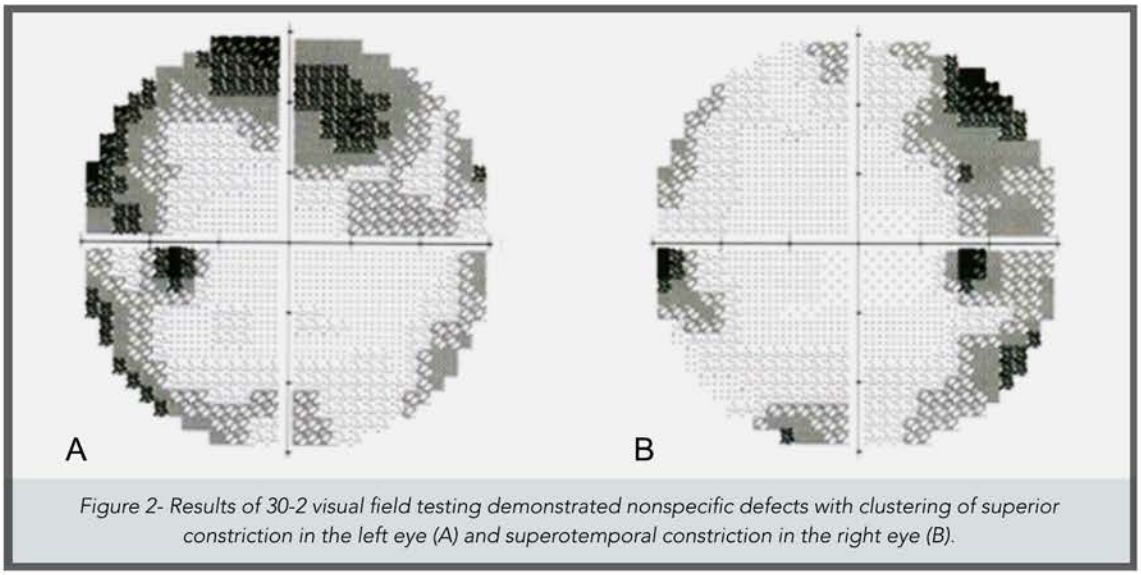

Figure 2- Results of 30-2 visual field testing demonstrated nonspecific defects with clustering of superior constriction in the left eye (A) and superotemporal constriction in the right eye (B).

Discussion:

Retina pigmentosa (RP) encompasses a large group of inherited disorders that result in slow progressive damage to the photoreceptors in the retina. It is estimated to affect approximately 1/4000 people worldwide. It typically presents in childhood with difficulty navigating in the dark, long periods of dark adaptation, visual field constriction, and photophobia. Classic examination findings are waxy pallor of the optic disc, severe vascular attenuation, bone spicule pigmentation of the peripheral RPE, and cystoid macular edema. It progresses slowly over decades and can eventually end in blindness. Genetically, there are over 90 known mutations which can cause an extremely large variance in the presentation and prognosis.

One unusual variant is known as sectoral RP, which is extremely rare. Its true incidence is not known due to most affected individuals only discovered on routine exam opportunistically. Classically it has exam findings of bilateral bone spicule hyperpigmentation patters along the inferotemporal arcade. Visual fields demonstrate superior scotomas correlating with the area of pigmentation. ERG demonstrates decreased rod greater than cone function. Generally, these patients are asymptomatic, but can present with various levels of night vision loss. This variant is generally a stationary to slowly progressive disease, with most patients retaining good central visual acuity even in late stages of the disease. Although controversial, some studies have shown some decrease in progression with the use of Vitamin A.

More recently, LUXTURNA™ (voretigene neparvovecrzyl) has become the first FDA-approved gene therapy

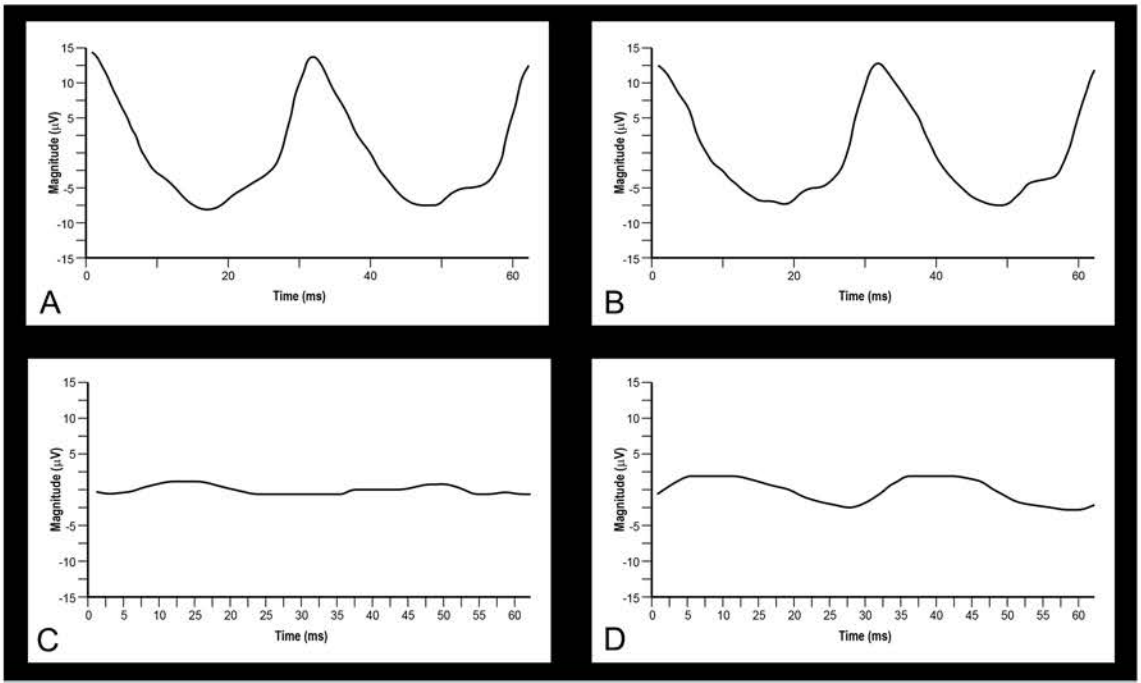

Figure 3 – ERG results of fixed luminance flicker testing in normal left (A) and right (B) eyes. Our patient demonstrated decrease signal strength in both left (C) and right (D) eyes.

for a genetic disease indicated for patients with a specific RPE65 gene mutation. Patients showed statistically significant improvement in visual acuity, contrast sensitivity, light perception, and visual field functions one year after receiving treatment. Although this gene therapy is currently limited to only a select subset of RP patients, this novel breakthrough is one step closer to finding a cure for numerous retinal degenerative diseases.

References:

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