Case Report
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Abstract
A 52 year-old African American male has experienced a gradual decrease in visual acuity from 2008 to present. Ocular exams are unremarkable aside from the decrease in visual acuity. Grossly abnormal ERG results confirm progressive cone-rod dystrophy.

I. Case History
- Patient demographics: 52 year-old African American male
- Chief complaint: Vision continues to slowly decline OU. Patient reports increased difficulty reading the Bible and other small print.
- Ocular history: Myasthenia gravis x 25 years s/p thymectomy 1985. binding antibody (+), blocking/modulating antibody (-). Presented with ocular symptoms, but has become systemic. Mestinon was tried for his ocular symptoms, but it didn’t help. The patient has been on prednisone since. Doing well on prednisone 10mg QOD x many years. Diplopia/ptosis well controlled at this point. Further tapering (below 5mg/day or 10mg QOD) has been unsuccessful. Patient is follow by Neurology at Marion VA.
- Medical history: Hypertension, elevated cholesterol, and pulmonary problems
- Medications: 1) Colestipol HCl 1GM TAB to lower cholesterol
  2) Diltiazem 180MG SA CAP for heart and blood pressure
  3) Fenofibrate 145MG TAB to lower cholesterol and triglycerides
  4) Hydrochlorothiazide 25MG TAB for fluid and blood pressure
  5) Hydrocodone 5/Acetaminophen 500MG TAB PRN for pain
  6) Lisinopril 20MG TAB to lower blood pressure
  7) Prednisone 10MG TAB for myasthenia gravis
- Allergies: Niacin, Zocor, Pravastatin, Crestor, Zetia 10MG TAB, Lopid 600MG TAB

II. Pertinent findings
- Clinical: Progressively declining BCVA OU
08/2003 20/20 OD, 20/20 OS  
11/2008 20/20 OD, 20/20 OS  
04/2009 20/25 OD, 20/30 NIPH OS  
06/2010 20/40 OD, 20/50 OS  
09/2010 20/30 OD, 20/50-2  
12/2010 20/30 OD, 20/60-2

Entrance testing, slit lamp, intraocular pressure, dilated fundus examination all within normal limits OU; no ocular manifestations of myasthenia gravis.

09/2010 30-2 threshold HVF: Reliable, generalized reduction in sensitivity OU without specific defects

-Physical: Unremarkable
-Radiology studies: 12/22/2010 CT scan, results normal
-Neuro-ophthalmology consult for progressively declining BCVA OU of unknown etiology:

01/2011 20/50 OD, 20/50 OS  
05/2011 20/50 OD, 20/80 OS  

Patient reports of increased photophobia over time and increased glare  
No nyctalopia or sudden changes  
Ishihara: control plate only OU. Patient not known to be dyschromatopic in the past.

Dilated fundus examination: faint macular RPE changes OU and slightly dull FLR bilaterally  
OCT: normal appearing foveas

Corneal Topography: ~0.5D cyl OU without significant irregularity

Pattern ERG results: Pattern ERG was performed using standardized ISCEV protocol. Responses were obtained using 1.0 and 0.75 degree checkerboard stimulus, with 4 Hz reversal rate. P50 peaks were severely reduced for both eyes, for both stimulus sizes. P50 latencies were delayed OU.

Retinal Angiography: Fluorescein retinal angiography showed normal choroidal and retinal vascular filling. No macular leakage was noted in either eye. No vascular sheathing was noted.

05/2011 HVF: generalized depression OU without specific defects
-Laboratory studies: CAR-Ab, ANA, Vit A/B12/B1/B6/folate, and SPEP ordered by neuro-ophthalmologist, still awaiting results
-Low vision evaluation:
Goal: Read small print more easily. Patient also reports too much light bothers his eyes.

05/2011 20/50+2 OD, 20/100-1 OS

Devices discussed/demonstrated:
Large print – patient uses a large print Bible
Lighting - recommended patient place floor lamp so it shines onto target over his right shoulder
Habitual correction with Add +2.25 – BVA 20/40 letters, slow
Coil 3x LED HHM - BVA 20/25 letters .8M fluid reading, patient reports LED light helps him see better
Eschenbach 3x LED SM - BVA .8M fluid reading .5 reading slow, patient likes the larger field of view. Prefers battery handle over cord.
Cocoons - to fit over his glasses, patient thinks having greater protection above and on the side of his glasses will make him more comfortable outside.
Patient also interested in transition lenses. Educated patient lenses don't get as dark in car.

III. Differential diagnosis
- Primary/leading: Severe cone-rod dystrophy; myasthenia gravis doing well on prednisone
- Others: Retrobulbar lesion, keratoconus, age-related macular degeneration, Stargardt disease, chloroquine/hydroxychloroquine maculopathy, congenital color blindness, retinitis pigmentosa, optic neuropathy or atrophy secondary to AION or NAION

IV. Diagnosis and discussion
- Elaborate on the condition: Cone-rod dystrophy is a genetic condition causing cone dysfunction with associated, but less severe, rod dysfunction.(1,2) This dystrophy is most often inherited sporadically or in an autosomal dominant pattern; and less often as autosomal recessive and X-linked forms.(1,3) Symptoms include bilateral slowly progressing decreased vision, photophobia, color vision difficulty, and worse vision in day than night.(1,3,4) Cone-rod dystrophy can be distinguished from other dystrophies by a reduction in visual acuity later in life with progression of the symptoms.(4,5) Ocular signs of advanced disease may include pendular nystagmus, a Bull’s eye macular appearance, a Stargardt’s – like macular lesion, attenuated vessels, optic atrophy beginning as temporal pallor, deutan-tritan color defects, decreased ERG single and flicker responses, dark adaptation abnormalities of cones, and EOG can be affected in severe cases.(1,3,4) Recognizably, ERG measurements assist in the diagnosis of this hereditary disease.
Spectral sensitivity measurements reveal reduced function of all three cones in cone-rod dystrophy and a single cone mechanism in selective cone dystrophy. Moreover, in cone-rod dystrophy the ERG reveals a reduction in the amplitudes of the photopic system and often mild involvement of the scotopic part. The yearly rate of change rod and cone ERG thresholds are comparable in patients with cone-rod dystrophy.

-Expound on unique features: This patient has no known family history of cone-rod dystrophy. The only indication of pathology was the presence of reduced BCVA in an individual with otherwise unremarkable ocular exam results. Further testing revealed generalized reduction in sensitivity on HVF, color vision defects, and reduced ERG flicker responses for rods and cones. The patient’s full field ERG demonstrated severely reduced and nearly obliterated photopic and 30 Hz flicker responses. Rod specific responses were reduced to ~10% normal OU. Maximal flash dark adapted responses were reduced to ~10% normal OD, ~20% normal OS. Pattern ERG showed marked attenuation of P50 peak for both eyes. A small, discernible waveform was obtained for both eyes, and these responses were reproducible. These results would indicate widespread cone and rod dysfunction OU, with more prominent cone involvement. The abnormal PERG suggests involvement of the central macular cones as well. There is no evidence to suggest chronic prednisone use is a causative agent here.

V. Treatment, management

-Treatment and response to treatment: No cure is available. Treatments include genetic counseling, low vision rehabilitation, and follow-up ERG in 1-2 years to monitor progression. This patient responded well during his low vision evaluation. He was motivated to try several low vision devices and thinks they will help him in his daily activities. A 3x HHM, 3x SM, cocoons, and spectacles with transition lenses were ordered and mailed to patient’s home. Yearly comprehensive exams are recommended to monitor visual acuity, ocular health, and low vision devices.

-Bibliography, literature review encouraged


**VI. Conclusion**

-Clinical pearls: In early stages of cone-rod dystrophy, the fundus may have a normal appearance, despite decreasing visual acuity. Fundus findings may not be present until more advanced stages of the condition. Reduced ERG responses help quantify the level of rod and cone involvement in cone-rod dystrophy.