Case Report of a Young Male with Cone Dystrophy

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Abstract:
Cone Rod Dystrophies are estimated at 1 in 40,000. This case report outlines a 40 year old white male complaining of photophobia and painless, progressive, vision loss. Extensive testing supports a diagnosis of Cone Dystrophy.

I. Case History

40 year old white male complains of intense light sensitivity and trouble reading at both distance and near for the last year. Patient has never worn glasses before. He was in the U.S. Army and U.S. Navy, veteran of the first Gulf War and the war in Iraq. He sustained a head injury when parachuting out of a helicopter in Iraq in 2002.

Ocular History: none known

Medical History:

- Hyperlipidemia
- Hypertension
- Chronic Pain Syndrome
- Posttraumatic Stress Disorder
- Disloc/separ, A-C joint
- Depressive Disorder
- Atypical Chest Pain
- Subjective tinnitus
- Sensorineural hearing loss
- Insomnia
- Anxiety
- Reflex Sympathetic Dystrophy of the Lower Limb
- Chronic Migraine without Aura
Allergic rhinitis

Medications:

- Acetaminphen
- Alprostadil
- Alprazolam
- Cetirizine HCl
- Hydrochlorothizide/Lisinopril
- Minocycline
- Naltrexone
- Prazosin
- Simvastatin
- Tamsulosin
- Topiramate
- Venlafaxine HCl
- Clonidine

II. Pertinent findings

Clinical

BCVA 04/12: 20/40 OD and OS
BCVA 11/12: 20/100 OD and OS
BCVA 07/13: 20/200 OD and OS
BCVA 07/14: 20/400 OD and OS

Color Vision May 2012: 11/11 Ishihara OD and OS

- Nov 2012: normal color OD, mild tritan OS  Farnsworth D15
- Aug 2013: normal color OU  Farnsworth D15

All other entrance tests normal OU at all visits

Ophthalmoscopy: normal OU at all visits

Physical

Wheel chair bound
Others

ERG- Nov 2012: reduced full-field ERGs OU with predominant loss of cone function and reduced rod function OU

Aug 2013: normal rod ERG responses, reduced 30 Hz cone ERG responses

OCT- normal central macula thickness OU at all exams

Goldman perimetry- May 2012: moderately constricted field OS>OD

Nov 2012: severely constricted field OS>OD

August 2013: severely constricted fields OS=OD

III. Differential diagnosis

Hysterical amblyopia OU vs rod-cone dystrophy vs cone dystrophy

IV. Diagnosis and discussion

Diagnosis- Progressive Cone Dystrophy

Cone dystrophy is a bilateral disease that can be stationary or progressive, occurring spontaneously or inherited autosomal dominantly. Stationary cone dystrophy tends to remain stable and is usually present at birth, while progressive cone dystrophy tends to occur later in life. Fundus exam is usually normal in the early stages of the disease, as in our patient, with outer nuclear layer loss and RPE changes occurring in later stages. The exact pathogenesis is unknown at this time.

• Expound on unique features

Decreased vision not attributed to refractive error, extreme light sensitivity, normal or granular appearing macula on ophthalmoscopy, decreased cone function on ERG, visual field constriction, and reduced color vision. FA may reveal areas of hyperfluorescence in the macula region, indicating that the RPE has lost some of its integrity, allowing the underlying fluorescence from the choroid to be more visible.

V. Treatment, management

Patient was evaluated by neuro-ophthalmology, and referred to Low Vision. Patient currently uses darkly tinted lenses and handheld magnifiers for near work, and was advised to have DNA testing of ABCA4 gene at Carver Lab in Iowa City, IA.

• Treatment and response to treatment

No treatment currently available, vision has remained stable at 20/400 OD and OS

• Refer to research where appropriate
Some animal studies have shown that antioxidant vitamins can slow further vision loss. Research is ongoing.

• Bibliography, literature review encouraged


VI. Conclusion

Optometrists should be aware of the importance of ERG in the diagnosis of Cone Dystrophy. Use of ERG is critical in the diagnosis of unexplained vision loss in healthy young patients, and helpful in differentiating Cone Dystrophy from Rod-Cone Dystrophy. While fundus changes can be visible later in the disease process, early on the retina may appear normal. Timely and accurate diagnosis is helpful for patients and their families dealing with their condition.