Title: Ocular Manifestations of Spinocerebellar Ataxia 3

Abstract: Spinocerebellar ataxia (SCA) is a group of progressive, neuro-degenerative genetic disease, with no cure which can have multiple ocular manifestations. This is a case report of 33-year-old male with SCA3, who is noting ocular symptoms.

I. Case History

A. Demographics: 33 year old African American male

B. Chief Complaint: Intermittent horizontal diplopia, occurring for several months, increasing in frequency without correction

C. History

1. Ocular
   a. Refractive error
   b. Dry eye syndrome

2. Medical
   a. Spinocerebellar Ataxia type 3
      i. Onset of symptoms 8 years ago
      ii. Mother passed away from condition
   b. Vitamin D Deficiency
   c. Oropharyngeal dysphagia
   d. Stuttering
   e. Obstructive sleep apnea
   f. History of falls

D. Medications: Cholecalciferol 400 unit (Discontinued)

II. Clinical Findings

A. BCVA: OD: 20/20 OS:20/25

B. EOMs: FROM (-) pain/diplopia OD/OS, gaze-evoked nystagmus

C. Pupils: ERRL (-) APD

D. CVF: FTFC OD/OS

E. Saccades: Normal OD/OS

F. Maddox Rod: 4 prism diopter base in horizontal, isophoric vertical

G. Cover Test: 8 prism diopeters exophoria at distance and near
H. Near Point of Convergence: break at 1 inch, recovery at 6, (+) diplopia

I. Color Vision(Ishihara) 14/14 OD, 14/14 OS

J. IOP 12/12 GAT @10:54am

K. Slit lamp examination: unremarkable

L. Dilated fundus examination: unremarkable

M. Humphrey VF 30-2: OD: Scattered points of sup loss, (-) blind spot, reliable

OS: Scattered points of sup loss, reliable

N. Cirrus RNFL: OD/OS: normal thickness throughout

O. Cirrus Macula OCT: normal foveal contour/thickness, with patchy loss in ganglion cell complex

III. Differential Diagnosis

A. Traumatic Brain Injury

B. Multiple Sclerosis

C. Decompensating phoria likely secondary to Spinocerebellar Ataxia 3

IV. Diagnosis and Discussion

A. Diagnosis

1. Decompensating phoria secondary to Spinocerebellar Ataxia 3

B. Discussion

1. Diagnosis: Conclusive diagnosis of SCA 3 is through genetic testing for abnormal triglyceride repeats on the ATXN3 gene. Genetic testing should be considered if there is family history for the disease. Testing should also be considered if gait abnormalities, clumsiness, vision problems and diplopia.

2. Genetics: Autosomal Dominant, 50% chance of passing onto progeny. CAG triglyceride repeat on ATXN3 gene.

3. Prognosis: Condition progresses from onset until death. Disease is more severe the earlier the disease onset occurs. Onset occurs between second and the fifth decades, mean onset age is 37. Progression exhibits anticipation, in that the disease tends to onset sooner and more severely as generations move on.

4. Condition leads to ataxic gait, hyperreflexia, dysarthria, nystagmus, vocal cord paralysis, vestibular dysfunction, sleep disturbance- REM behavior disorder, slow saccadic eye movement. Pt currently requires a walker to move and is experiencing slight slurring of speech.

5. Life expectancy: mid-thirties to normal life span
6. Ocular findings: nystagmus, saccadic deficiencies that can result in up-gaze restriction, ophthalmoplegia, disconjugate eye movements that can lead to diplopia

V. Treatment/Management

A. Treatment: palliative

1. No cure

2. Manage signs/symptoms as it progresses: use cane/walker/wheelchair as walking becomes more difficult, prism or VT for diplopia

3. For this patient, a new spectacle prescription was given, OD: -0.25-1.00x105 OS: -0.25-0.50x045, and patient is to follow up 3 months after receiving glasses for a vision check. If diplopia persists or worsens, we will consider vision therapy and/or prism correction.

VI. Conclusion

A. As there is no cure, treatment is only palliative. Symptoms are to be managed directly. Additional eye tests that are problem indicated are ocular motilities, saccades, cover test, and ganglion cell complex testing. As the various Spinocerebellar Ataxias make up one of the most common kinds of neuro-degenerative diseases, it is important to know what testing is clinically relevant and what symptoms are pathognomonic of these conditions.

References:


