Chronic Progressive External Ophthalmoplegia (CPEO) Secondary to Kearns-Sayre Syndrome (KSS) in a 10-year old Black Female

Abstract (33 words)

Background: Kearns-Sayre syndrome is one cause of CPEO that is difficult to diagnose based on its wide array of clinical manifestations. We report a rare case of a patient currently diagnosed with KSS.

Case Report Outline

I. Case History

- 9 YOBF presents on 10/13/2009 for strabismus consult from local optometrist
- Chief complaint of blurry vision in both eyes that affects distance vision with gradual onset; occurs intermittently and frequently
- Ocular history: patient failed school vision screening in 2007; glaucoma suspect OU secondary to large physiological cupping; compound myopic astigmatism OU first diagnosed in 2007 for which the patient wears glasses
- Medical history: torticollis, asthma, seasonal allergies, no known drug allergies
- Medications: Advair discus 100-50mcg PRN for symptoms, Singulair 4mg PRN for symptoms, Claritin 10mg PRN for allergies
- Pediatric/Pregnant history: normal term pregnancy and normal development

- On 05/10/10 patient returns for evaluation of droopy lids OS>OD

- 07/07/10 patient complains of itching eyes OU for two weeks with gradual onset; symptoms are constant and patient reports Visine helps upon instillation

II. Pertinent Findings

10/13/2009

Clinical:
- VAcc: 20/30-1 OD, 20/30-2 OS
- Current glasses: OD: -5.00 +1.00 x169 OS: -4.25 +0.50 x027
- Pupils: PERRL(-)APD OU
- Conf. fields: FTFC OU
- EOMs: restriction on adduction in upgaze OD
  Good alignment in primary and downgaze OU
- SLE, IOPs and DFE: unremarkable ocular health

Physical:
- left head tilt and face turn

05/10/2010

- VAcc: 20/70 OD, 20/60 OS
- Lids: bilateral ptosis OS>OD
- EOMs: dramatically reduced OU (ophthalmoplegia) with mainly central fixation
  OD>OS
-Systemically asymptomatic

07/07/10
- SLE: palp conj hyperemia 1+ OU, papillae 1+ OU
- Lids: bilateral ptosis OS>OD
- EOMs: ophthalmoplegia OU

07/16/10
Laboratory studies:
- ECG revealed minor PR wave abnormality, although no heart conduction deficits
- Ice pack test showed no improvement of upper lid ptosis OU

09/??/10
Laboratory studies:
- Histological slides of extraocular muscles to be prepared for evaluation

III. Differential diagnosis
- Brown’s tendon sheath syndrome
- Chronic Progressive External Ophthalmoplegia (CPEO)
  - specifically Kearnes-Sayre syndrome
- Myasthenia Gravis (MG)
- INO/BINO

IV. Diagnosis and discussion
- Kearnes-Sayre syndrome (KSS): an inherited mitochondrial myopathy affecting patients in 1st or 2nd decades of life
- Ocular manifestation: slowly progressive, symmetrical ophthalmoplegia (CPEO) with bilateral ptosis and no diplopia; pigmentary retinopathy
- Unique features: cardiac conduction abnormalities (heart block); ragged red fibers on extraocular muscle histological slides with mitochondrial proliferation; deafness and increased CSF protein may also be evident; fatigue and proximal muscle weakness are sometimes evident

V. Treatment, management
10/13/09
- Patient initially diagnosed with Brown’s tendon sheath syndrome OD; superior oblique tendon sheath graft surgery discussed with patient and mother but not recommended at that time

05/10/10
- Patient was re-diagnosed with having CPEO vs. MG vs. other cause for ophthalmoplegia
- Consulted with retinal ophthalmologist at practice who reported unremarkable peripheral fundus and no papilledema OU
- Patient referred to Willmer Eye Institute (WEI) this visit and instructed parents to go to Johns Hopkins ER for neuro-ophthalmology consult
07/07/10
- WEI believes KSS to be cause of patient’s ophthalmoplegia; they ordered ECG testing and MG work-up
- Patient treated for chronic allergic conjunctivitis OS>OD and given prescription for Pataday igt OU qam; instructed to continue follow-up care with Johns Hopkins University

09/01/10
- Histology slides of extraocular muscles to be prepared this month to evaluate for ragged red fibers and mitochondrial proliferation

Bibliography to come with final poster/paper.

VI. Conclusion
Kearns-Sayre syndrome-related CPEO is a rare diagnosis that can be easily overlooked. A comprehensive ocular evaluation with dilated fundus exam and further laboratory testing (including ECG and histological analysis) are required to definitively diagnose this condition. The optometrist’s role when assessing ophthalmoplegia including necessary neuro-ophthalmology referral is crucial to the patient’s treatment and management.

Clinical Pearls:
- saccades are slow in CPEO vs. quick saccades over a short distance in ocular MG

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