Implications of Undiagnosed Kearn-Sayre Syndrome in Chronic Progressive External Ophthalmoplegia Patients
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Abstract

Kearn-Sayre Syndrome is a rare mitochondrial disorder with ocular manifestations of ophthalmoplegia, ptosis, and pigmentary retinopathy. Due to associated cardiac complications and risk of mortality, proper diagnosis and referral is critical.

I. Case History

1. Patient demographics
   - 46 year old Hispanic male
   - Presented with longstanding diplopia at distance and near, and headaches when reading

2. Ocular history
   - Chronic progressive external ophthalmoplegia (CPEO) OU s/p strabismus repair 1998
   - Ptosis OU s/p repair OS 1998

3. Medical history
   - Depression/Chronic PTSD
   - Chronic pain syndrome
   - Vitamin D deficiency
   - Syncope/lightheadedness/palpitations
   - Dyspnea
   - Ankle and foot edema
   - Night sweats/heat and cold intolerance
   - Tinnitus

4. Medications
   - Benzoyl peroxide, isotretinoin, trazodone, naproxen, artificial tears, lubricating ointment, cholecalciferol, imdur

II. Pertinent findings

1. Clinical
   - BCVA 20/30-2 OD/OS
   - Minimal adduction, significant restriction in abduction, elevation, and depression OD/OS, restriction OS > OD
   - Near Point Convergence: receded at 35cm
   - Cover Test: small ET at distance, moderate XT at near

2. Physical
   - Lagophthalmos OU, pannus inferonasal OU, exposure keratopathy OU
   - Ptosis OD>OS
   - Macular RPE changes OU
   - Enlarged C/D OU, PPA OU

3. Laboratory studies
   - Blood work: elevated creatine phosphokinase (791, 2577, 373), elevated aldolase (12.4), negative ANA/RH factor, negative Lyme screen
   - Cardiac catheterization, coronary angiography, femoral angiography – normal coronaries, transient vasospasm noted during case, normal left ventricular end diastolic pressure
   - EKG: Bundle branch block/interventricular conduction delay was present. Two supraventricular tachycardia runs occurred. Ventricular bigeminy was present. Small area of inferolateral
ischemia was noted.

4. Radiology studies
- MR: abnormal increased FLAIR and T2 signal along corticospinal tracts bilaterally. Suggestive of ALS. Suggest MR spectroscopy and DTA. No change from 2015.
- EMG/NCS: Abnormal EMG/NCS right upper and right lower extremities.

III. Differential diagnosis
1. Primary/leading
- Kearn-Sayre Syndrome and Chronic Progressive External Ophthalmoplegia
2. Others
- Amyotrophic lateral sclerosis
- Myasthenia Gravis
- Sarcoidosis
- Thyroid Eye Disease (Grave’s Disease)
- Botulism
- Oculopharyngeal dystrophy
- MELAS (Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-like episodes) Syndrome

IV. Diagnosis and discussion
Kearn-Sayre Syndrome is a rare mitochondrial encephalomyopathy secondary to deletions in mtDNA without any racial predilection. Patients present with muscle weakness, dysfunction of the central nervous system, endocrinopathies, cardiac abnormalities, or ocular manifestations. Muscle weakness can often present as ptosis, dysphagia, and ophthalmoplegia. CNS manifestations are deafness, ataxia, and dementia. The greatest significance of management are secondary to cardiovascular complications such as atrioventricular block, his-ventricular interval prolongation, dilated cardiomyopathy, and Stokes Adam syncope – with sudden death being reported in 20% of patients.

Kearn-Sayre Syndrome is characterized by a triad: age of onset <20 years; chronic progressive external ophthalmoplegia; and pigmentary degeneration of the retina. The true incidence of the syndrome is unknown. Diagnosis of Kearn-Sayre Syndrome can be made based on clinical impression and lab testing, or with genetic testing.

V. Treatment, management
In office management of diplopia is treated with prism spectacles separated for distance and near to give the patient clear, single vision. Artificial tears and lubricating ointment at night can be used in combination with a moisture chamber and a lid crutch for palliative treatment of exposure keratopathy secondary to lagophthalmos and ptosis. While ptosis repair is an option, many oculoplastic surgeons are hesitant to perform surgery as the ptosis will continue to progress. In these cases, a reversible silicon sling is preferred.

If a chronic progressive external ophthalmoplegia patient is suspected to have Kearn-Sayre Syndrome, the proper referrals to cardiology, endocrinology, neurology, audiology, and ophthalmology must be made. Genetic counseling may be necessary. Recent studies show CoQ10 to hold potential as a treatment option for Kearn-Sayre Syndrome.

1. References
VI. Conclusion

Kearn-Sayre Syndrome patients often present to optometrists with complaints of diplopia and dry eyes. Management of symptoms can significantly improve quality of life, but the proper referrals will ultimately save lives.