Chronic Progressive External Ophthalmoplegia Secondary to Kearns Sayre Syndrome

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INTRODUCTION

Kearns Sayre Syndrome (KSS) is a mitochondrial myopathy with ocular signs of chronic progressive external ophthalmoplegia, ptosis, pigmentary retinopathy, and strabismus. Patients with Kearns Sayre Syndrome are at an elevated risk for cardiac conduction abnormalities which can be as severe at atrioventricular (AV) block. Cardiac abnormalities occur in 50% of patients with KSS and is the leading cause of death in this population. Ocular findings in KSS can precede cardiac abnormalities. This case covers the signs and symptoms of Kearns Sayre Syndrome, systemic abnormalities and treatment and management options.

CASE PRESENTATION

A 43-year-old African American female presents to clinic with the complaint of slow, progressive loss of eye movement OU over the past 5 years and ptosis OU. She has a past ocular history pertinent for normal tension glaucoma OU and a longstanding exotropia OS. Her past medical history is pertinent for type 2 diabetes and an unknown mitochondrial myopathy.

TABLE 1

	OD	OS
VAcc	20/30 PH 20/25	20/40-2 PHNI
CVF	FTFC	FTFC
EOM	no movement in any field of gaze	no movement in any field of gaze
Pupils	ERRLA, (-)RAPD	ERRLA, (-)RAPD

DIAGNOSIS

The diagnosis for our patient is Kearns Sayre Syndrome. An electroretinogram (ERG) was performed on our patient's right eye to rule out other pigmentary dystrophies. The ERG was within normal limits. KSS is caused by mitochondrial DNA deletions or mutations. These abnormalities can be sporadic, autosomal dominant, or autosomal recessive. The extraocular muscles have an increased metabolic demand and therefore have an increased number of mitochondria causing extraocular muscle abnormalities such as chronic progressive external ophthalmoplegia, ptosis, and strabismus. There have also been reports of optic atrophy and corneal clouding. Systemic manifestations of KSS include: cardiac abnormalities, sensorineural hearing loss, ataxia, non-ocular muscle weakness, neuropathy, impaired intellectual function, and endocrine disorders. This disorder is diagnosed by clinical findings including the typical triad of progressive external ophthalmoplegia, pigmentary retinopathy, and onset before the age of 20 with one of the following additional signs: cardiac block, cerebellar symptoms, or elevated cerebrospinal fluid protein levels above 100 mg/dl. KSS can also be diagnosed with genetic testing and muscle biopsies. Skeletal muscle biopsies are preferred as the ragged red patten are clearly seen.

TABLE 2

	OD	OS
Adnexa	Normal	Normal
Lids/Lashes	Ptosis OD<OS	Ptosis OD<OS MRD1: -0.5mm MRD2: 2mm
Conjunctiva	White and quiet	White and quiet
Sclera	White and quiet	White and quiet
Cornea	Normal endothelium, epithelium, stroma, and tear film	Normal endothelium, epithelium, stroma, and tear film
Angles	3-4+ nasal and temporal	3-4+ nasal and temporal
Anterior Chamber	Deep and quiet	Deep and quiet
Iris	Normal	Normal
Lens	Clear lens capsule, cortex, and nucleus	Clear lens capsule, cortex, and nucleus

FIGURE 1

FIGURE 2

FIGURE 3

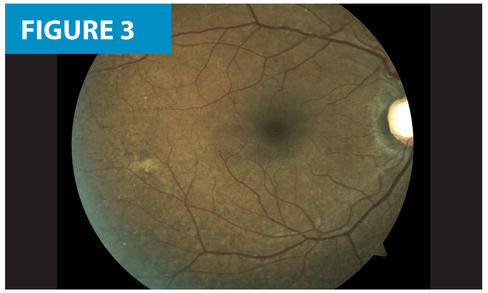


FIGURE 4

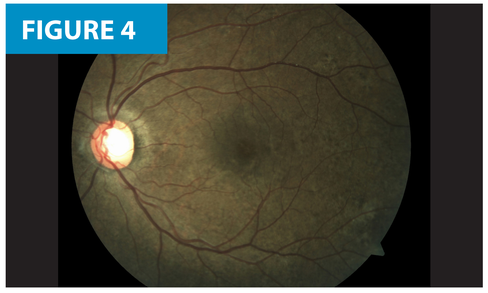


FIGURE 5

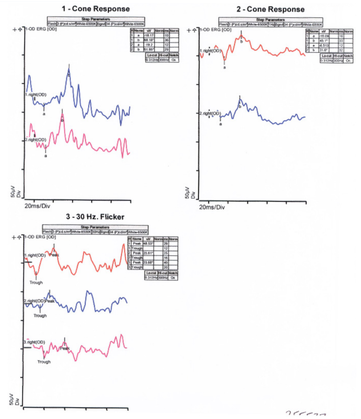
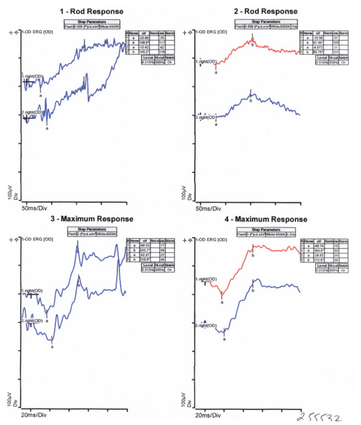


FIGURE 6



DISCUSSION

Ocular manifestations of KSS are managed primally with supportive therapy. Ptosis and strabismus surgery can be performed but the symptoms may progress after surgery. Cardiac abnormalities, typically conduction deficits, are present in 50% of patients with KSS and cause sudden death in 20% of patients. Syncope may be the first sign of cardiac involvement. Patients with KSS should be under the care of a cardiologist and receive electrocardiogram testing annually. Pacemakers may be placed at the first sign of cardiac involvement. 87% of patients with KSS had ptosis and ophthalmoplegia prior to cardiac abnormalities.

CONCLUSION

Ocular manifestations can be the presenting signs of KSS. If KSS is suspected, refer the patient to a cardiologist for regular monitoring as well as to a primary care physician to consider a muscle biopsy to confirm the diagnosis. As optometrists, we can monitor the patient for progression and treat patient symptoms with supportive therapy.

REFERENCES

Available upon request

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