Kearns-Sayre syndrome (KSS): A Rare Case

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ABSTRACT
We describe a case report of 27 year old male with progressive drooping of both upper eyelids for eight years, associated with inability to move eyes towards a particular direction. After thorough clinical evaluation, ocular investigations, various blood tests and cardiology evaluation, a diagnosis of Kearns-Sayre syndrome (KSS) was made. KSS is a rare mitochondrial myopathy, seen in first two decades of life involving ocular, cardiac, neurological and renal systems.

KEY WORDS: mitochondrial, myopathy, retinopathy.

INTRODUCTION
Kearns-Sayre syndrome (KSS) is a mitochondrial disease with features of chronic progressive ophthalmoplegia, pigmentary retinopathy and an age of onset younger than 20 years. It is seen to occur in the first two decades of life, affecting an active age group and with associated life threatening complications. Other systems that might be involved are neurological, endocrine and renal systems. Most cases have no positive family history and appear to be due to spontaneous mitochondrial mutations.

CASE REPORT
A 27 year old male from Kathmandu presented with complaints of progressive drooping of both upper lids and inability to move eyes towards a particular direction noticed 8 years ago. Drooping had progressively increased to an extent where he had to lift his upper lids with his fingers to see well. He also had increasing diminution of vision during early evenings for past 10 years with history of tripping and falling in dim lit places. Patient also complained of inability to turn his eyes while viewing an object and had to turn his face towards what he wants to view. He also complained of generalized tiredness. Patient also had two episodes of loss of consciousness 8 years back. He was diagnosed as a case of complete heart block and underwent pacemaker implant. None of his three elder brothers or family member has similar systemic or ocular complaint. He studied until grade 12, and did not pursue higher studies. He had no complaints of decreased hearing and difficulty in movements.

On examination, the patient had a best corrected visual acuity of 6/12 in both eyes with −4.00 DS OU, brow lift, chin elevation, restriction of extraocular movements in all directions of gaze, palpebral fissure height in right eye was 3mm and in left eye was 4mm. Patient had poor LPS (levator Palpebral Superioris) function of 4 mm and poor Bells phenomenon. Remaining anterior segment findings were not significant. Posterior segment findings showed normal disc, normal blood vessels and pigmentedary changes in the retina. He was evaluated in the oculoplasty, neuro-ophthalmology, retina and orthoptics department in Nepal Eye Hospital.

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Investigations done included visual field analysis by Goldmann perimetry. Blood tests included RBS – 94 mg/dl, normal thyroid function tests, normal sodium and potassium.

Patient was referred to cardiac, neurology and endocrine departments in Bir Hospital and TUTH. He had a normal ECG, echocardiography, and pacemaker was noted.

Patient was advised bilateral frontalis sling for severe ptosis, which patient declined. He was advised 6 monthly follow ups for refraction, progression of ptosis, pigmentary retinopathy and visual field status. Follow up with a cardiologist was also advised.

Other tests which we would have liked to have done but were not done due to unavailability or patients reluctance were CSF analysis of protein level and lactate level, muscle biopsy and PCR determination of mtDNA mutations or deletions.

He was prescribed Capsule coenzyme q10 150mg once daily by Neurology department.

**DISCUSSION**

Kearns-Sayre syndrome, a rare mitochondrial myopathy was first described in 1958. Remes et al estimated a prevalence of 1.6 cases per 100,000 population in a Finland.\(^1\) The high concentration of mitochondria in extraocular muscles than in other skeletal muscles may account for the predominance of ocular findings.

The mean age of KSS presentation is 17 years, but the mean age at diagnosis is usually at 26 years with 69% male preponderance.\(^2\)

This patient was clinically diagnosed as a case of Kearns-Sayre as the obligate triad of onset before age 20, extraocular muscle weakness (ptosis and ophthalmoparesis), and pigmentary retinopathy, plus one of the following: ataxia, cerebrospinal fluid protein level greater than 100 mg/dL, or cardiac conduction block was fulfilled.\(^3\)

Mitochondrial DNA, is transmitted exclusively by the mother via the ovum. Mitochondrial DNA replicates more frequently resulting in considerable phenotypic variability. It codes for oxidative phosphorylation proteins. Among these are subunits of cytochromes band c, and adenosine triphosphate (ATP) synthase.\(^4\)

Electron microscopic picture of mitochondria is
variable. The picture of “ragged red fibres” from stained muscle biopsies is non-specific and not always present in all cases.

KSS has been distinguished from CPEO (chronic progressive external ophthalmoplegia), by the fact that patients with Kearns–Sayre syndrome have identical mitochondrial DNA deletions in other tissues, whereas patients with isolated CPEO who have had other tissue tested have the deletion only in muscle tissue.

Ptosis is usually the first ocular presenting symptom. This is followed by chronic progressive ophthalmoplegia, pigmentary retinopathy, incomitant strabismus and weakening of facial muscles.

Neurological signs such nonspecific weakness, ataxia, and hearing loss are seen. Endocrinological disturbances include growth retardation, hypothalamic-pituitary adrenal insufficiency, diabetes mellitus, thyroid abnormalities, hypoparathyroidism and hyperaldosteronism. Cardiac anomalies, such as conduction disturbances and arrhythmia may occur at any time in KSS patients. Such disturbances can lead to sudden death. Patient might also present with renal tubular acidosis.

Ubiquinone, or coenzyme Q10 (CoQ) deficiency has been noticed in few patients with KSS. This enzyme is involved in normal mitochondrial respiration. Treatment has shown improved systemic function but no known effect on ophthalmoplegia, ptosis, or retinopathy.

Our patient claims he has had decreased tiredness after taking ubiquinone supplements, however no improvement in ocular movements was noted after one month of use. No side effects were noted.

REFERENCES