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A Case Report on Kearns Sayre Syndrome: A Rare Oculomyoencephalopathy

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Abstract: ***A 22 year short-statured female presented with progressive drooping of both upper eyelids, gradually progressive restriction of eye movements in all gazes. No complaint of diplopia or diminution of vision, no diurnal variation; no history of prior ocular surgery or trauma. She complained of generalised weakness and mild hearing loss. BCVA was 6/6 OU. Chin & Brow elevation present, B/L mild ptosis with preserved Bell's phenomenon. B/L EOM movement limited in all gazes. Direct & consensual reflexes normal. B/L pigmentary retinopathy was present. Audiometry showed B/L sensorineural deafness. ECG showed AV conduction delay with LVH. Multidisciplinary management for ptosis correction, vision & hearing aids was given.***

Keywords: KSS, Ptosis, Hearing loss, conductive hearing loss, Sensorineural Hearing Loss, cardiac abnormalities, red ragged fibers, EOM restriction, skeletal muscle biopsy

1. Introduction

Kearns Sayre Syndrome is a rare mitochondrial cytopathy where there are deletions and/or duplications of mitochondrial DNA, which codes for proteins of the respiratory chain reaction. Its prevalence is 1 in 100000 individuals. It's a CLASSICAL TRIAD of – (a) Chronic Progressive External Ophthalmoplegia (b) Atypical retinal pigmentary degeneration(c) Onset occurring before 20 years of age.

2. Case Report

A 22 year old female presented with progressive drooping of upper lids in both the eyes. She had restriction of eye movements in both eyes, which was gradual in onset & progressive. She also complained of mild hearing loss on the left side. The ptosis was mild& progressive, first noticed by her in her teenage, present since 4 years. There was no association with jaw movement, no diurnal variation. Chin lift was remarkable while viewing at distance. There was no ocular redness, no pain, no photophobia, no diplopia or diminution of vision. She had generalized weakness. She had no difficulty in breathing, speech or swallowing. Her appetite, bladder and bowel habits were maintained.

She had a visual acuity of 6/12, N6 in right and 6/9, N6 in the left eye. Fatigability and Ice Pack tests were negative. MRD-1 was 3cm & 2 cm in right & left eye, respectively. MRD-2 was 7cm & 8cm in right & left eye respectively. LPS function was good. Direct and consensual pupillary reflex were normal. EOM movement in bilateral eyes was restricted in all gazes, insidious and painless. 15^0 exotropia was noted in right eye. Pigmentary retinopathy was found in both the eyes.

Investigations
The patient was subjected to a battery of investigations, as follows:

Routine Blood investigations showed Relative lymphocytosis & normocytic normochromic anaemia.

Audiology:
Audiometry showed moderate mixed hearing loss in left ear.

Neuropsychiatric evaluation of the patient was insignificant.

CSF Analysis:
CSF protein & LDH were raised

MRI Brain: Diffuse cerebral atrophy, prominent ventricular system noted.
Cardiology:
ECG revealed AV conduction delay with LVH &LAD.
2D ECHO- Normal study

Endocrinology- No endocrinopathy.

Skeletal muscle biopsy - Modified Gomori Trichrome staining of the skeletal muscles showed red ragged fibres, confirming the diagnosis.

Final Diagnosis

Kearns Sayre Syndrome

Management
A multidisciplinary team approach was mandated including the Department of Ophthalmology & Otorhinolaryngology, to provide treatment. The patient was managed conservatively with topical artificial tear substitutes and oral Co-enzyme Q_{10}. Her ptosis was surgically corrected done with Frontalis Sling operation. BCVA 6/6, N6 in both eyes was achieved with aid of corrective glasses. Her hearing deficit was corrected with the aid of cochlear implant. She was given Genetic counselling for KSS.

Follow– up
BCVA was maintained at 6/6, N_6 with the help of corrective glasses. Her ptosis had improved& EOM unrestricted. She was showing improving hearing levels with the aid. She was followed up regularly with cardiology consultations.

3. Discussion

The ocular course of Kearns Sayre Syndrome is relatively benign. The majority of KSS patients have near normal vision. Visual function is well preserved in such cases suggesting that the retinopathy minimally affects the

photoreceptor function. Ophthalmology, Audiology, Cardiology, Endocrinology, Neurology, and Neuro-Psychiatry consultations must be made in tandem for a diagnosis of KSS to be established.

There is no definitive treatment for this condition but generally symptomatic and palliative. Around 20% of deaths in KSS patients are attributed to cardiac causes. Management includes prophylactic cardiac pacing for advanced AV conduction defects, shown to improve survival. Ophthalmologic manifestations may be treated with surgery, usually done for cosmesis, but carriers a high risk of recurrence and possible ocular complications.

Dietary supplements (Coenzyme Q_{10}) to control the progressive effects of the disease have been tried. Coenzyme Q_{10} benefits by improving lactate metabolism, heart & skeletal muscle function, but doesn't improve the ocular manifestations. Cochlear implants should be used in patients with significant sensorineural deafness.

Long term follow-up for cardiomyopathies / endocrinopathies and proper genetic counselling is advised.KSS is a challenging diagnosis because of the low prevalence &under-documentation of cases. It is imperative to have a high index of suspicion for KSS when encountering cases of oculomusculoskeletal disorders in subjects below 20 years of age in view of high morbidity / mortality associated with KSS. Ophthalmologists play important role because they may be the physician of first contact in majority of such patients.

4. Declarations

Funding: Nil

Conflict of interest: None.

Ethical approval: The case study was conducted as per the guidelines of the Declaration of Helsinki and the institutional ethics committee.

5. Consent

Written informed consent was obtained from the patient for publication and/or presentation of her medical data, including this case report and accompanying images.

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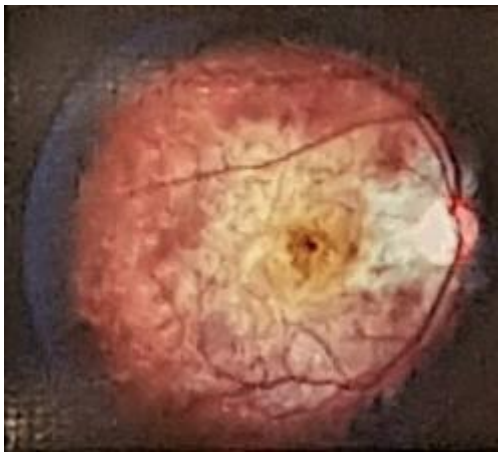

Figure: Pigmentary Retinopathy Right Eye

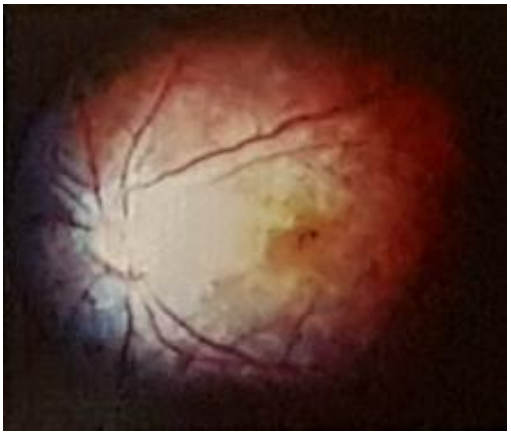

Figure: Pigmentary Retinopathy Left Eye

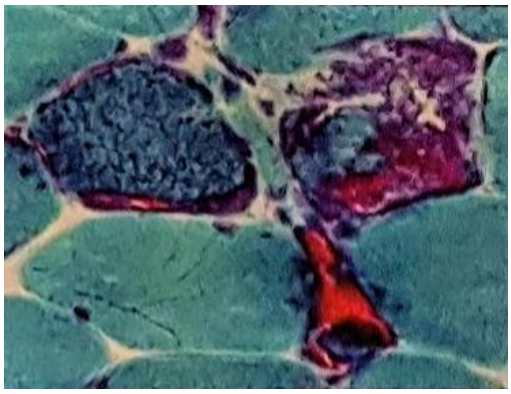

Figure: Red Ragged Fibres seen on Trichrome Gomori Staining of the skeletal muscles

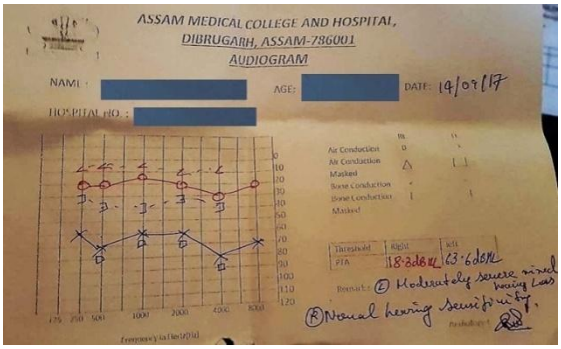

Figure: Audiometry showing Moderately Mixed Hearing loss on left side

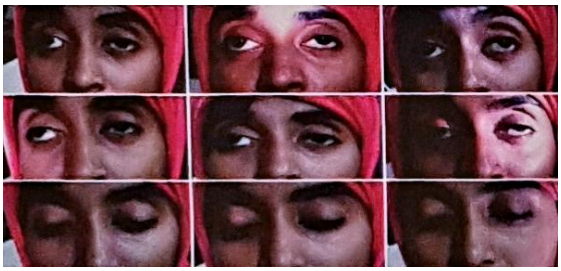

Figure: EOM restriction in all gazes bilaterally