Case Report

Kearns–Sayre Syndrome


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Introduction

Kearns-Sayre syndrome is a rare neuromuscular disorder first described by Thomas P. Kearns and George P. Sayre in 1958. Incidence is 3/100000 live births and is a rare genetic disorder caused by mitochondrial myopathy due to mutations in mitochondrial DNA and typically develops before the age of twenty. Clinical triad of Kearns-Sayre syndrome are Chronic Progressive External Ophthalmoplegia (CPEO), salt and pepper like Pigmentary Retinopathy and Cardiac blocks. KSS prognosis is related to the number of tissues affected and the severity of the alterations. In this article we report a patient who presented with clinical features suggestive of Kearns-Sayre syndrome.

Case Report

19 year old male came to ophthalmology outpatient department with complaints of drooping of eye lids and difficulty in moving both eyes for the past one year, six episodes of syncope in past five months duration. There was no history of diplopia, ataxia and deafness. On examination both eyes vision was 6/60 with pin hole improvement to 6/24. Both eyes lids showed severe ptosis with poor Levator Palpebrae Superioris and restricted extraocular movements suggestive of Chronic Progressive External Ophthalmoplegia (CPEO), salt and pepper like Pigmentary Retinopathy and Cardiac blocks.

Discussion

Kearns-Sayre syndrome, a rare neuromuscular disorder was first described by Thomas P. Kearns and George P. Sayre in 1958. Incidence is 1-3/100000 live births1,2. The Kearns-Sayre syndrome is a genetic disorder caused by mitochondrial myopathy due to mutations in mitochondrial DNA involved in oxidative phosphorylation for energy production.
Mitochondrial DNA contains many genes for normal function but deletion removes 4,997 nucleotides, which includes twelve mitochondrial genes in Kearns–Sayre syndrome. Deletions of mitochondrial DNA result in impairment of oxidative phosphorylation and a decrease in cellular energy production. Regardless of which genes are deleted, all steps of oxidative phosphorylation are affected. Tissues with high energy demand such as muscle and nervous system are particularly vulnerable to mitochondrial dysfunction, a consequence of deletions, rearrangements or other mutations in mitochondrial DNA. Triad of Kearns-Sayre syndrome are Chronic Progressive External Ophthalmoplegia (CPEO), salt and pepper like Pigmentary Retinopathy and Cardiac blocks. Ptosis is the first sign in Kearns-Sayre syndrome. Other systemic involvements are deafness, ataxia, syncope, renal failure, seizures, dementia, short stature, hypocalcemia and diabetes. The cardiac manifestations of Kearns-Sayre Syndrome are the most important aspects of the disease for determining the prognosis. Manifestations of cardiac disease occur in 57% of patients with Kearns-Sayre Syndrome, including syncopal attacks, heart failure and cardiac arrest. Kearns-Sayre Syndrome is diagnosed by extra ocular muscle biopsy which shows the ragged-red cells (red fibers torn) due to intramuscular accumulation of abnormal mitochondria and it is specific for the diagnosis of mitochondrial myopathies. Increased amount of protein (>1g/l) in cerebrospinal fluids in CSF analysis is also specific for Kearns-Sayre syndrome. There is no specific treatment for Kearns-Sayre syndrome. Co enzyme Q10 was tried in certain myopathy cases. Cardiac blocks are managed with cardiac pacemakers and ptosis corrected by crutch glasses (Fig 5).

Conclusions

Awareness of the nature of components of the syndrome led us to early recognition of the systemic complications and plan appropriate referral and management. KSS prognosis is related to the number of tissues affected and the severity of the alterations. The disturbances in the cardiac conduction system are responsible for high morbidity and mortality of the disease.

References

1) Kearns TP, Sayre GP. Retinitis pigmentosa, external ophthalmoplegia and complete heart block: unusual syndrome with histologic study in one of two cases. Arch Ophthalmol 1958; 60:280-289.


4) Zeviani M, Moraes CT, Di Mauro S. Deletions of mitochondrial DNA in aernes-sayresyndrome. Neurology 1998; 1525-32


Diabetes is after women's heart!

In one of the largest studies of its kind published in Diabetologia (Diabetologia, May 2014 DOI: 10.1007/s00125-014-3260-6), a metaanalysis & systematic review was done on the data obtained from more than 850,000 subjects over a period of 50 years with particular reference to relationship between diabetes and heart disease. It was found that diabetic women have three times greater risk of developing coronary heart disease (CHD) than their non-diabetic counterparts; in diabetic men, the risk of CHD is two-times higher than in non-diabetic men. Taken overall, women have 44% increased risk of CHD compared to diabetic men. If these findings are confirmed, screening women for pre-diabetes and a more stringent follow-up of diabetic women is necessary to prevent CHD in them.

- Dr. K. Ramesh Rao