Kearns-Sayre Syndrome

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Introduction

Kearns-Sayre syndrome (KSS) is specified by pigmentary retinopathy and ophthalmoparesis which should begin before 20 years of age. There are also some other clinical features which are common; some of these freatures are the following: cerebellar ataxia, increased cerebrospinal fluid (CSF) protein, conduction block in heart, and proximal myopathy. Many of the patients also have short stature and some of them have multiple endocrinopathies such as diabetes mellitus, hypoparathyroidism, and Addison disease

Genetics

The disorder is caused by mutation in the genome which is located in the mitochondria. It is a 16 kilo-base-pair circular double-stranded DNA, several copies of which is found within the mitochondrial matrix. Disease occurs only when the proportion of mutated to wild-type mtDNA exceeds a tissue-specific limit.

Epidemiology

Kearns-Sayre syndrome is a rare disorder. Remarkable heterogeneity and different modes of inheritance have been reported. There is not total agreement on the rate of prevalence, but it is estimated to be about 1 to 1.5 per 100000 adult population.

Clinical Presentation

The following are the most prominent signs noted in patients with Kearns-Sayre syndrome (KSS):

Muscle weakness, CNS dysfunction (cerebellar ataxia, Dementia, night blindness, external ophthalmoplegia), cardiac disease (syncope, palpitations), symptoms of endocrine dysfunction.

Laboratory Studies

The following studies are indicated in Kearns-Sayre syndrome (KSS):

Urine measurements of pH, protein, glucose, and amino acid levels, serum creatine kinase level, blood lactate and pyruvate (usually increased). Cerebrospinal fluid (CSF) protein levels are very frequently elevated, and CSF lactate levels are elevated even if blood lactate levels are within normal limits. In young children, single large-scale deletions may be detectable in blood. Diagnosis may alternatively be established by muscle biopsy with histochemistry and mtDNA analysis for major rearrangements. Screening is recommended to rule out the endocrinologic abnormalities which frequently occur in these patients. Serum electrolytes, glucose, calcium, magnesium, and plasma cortisol should be assayed, as well as thyroid function.

Medical Care and Consultations

No disease-modifying therapy is available for Kearns-Sayre syndrome (KSS). Management is mainly supportive and entails close watching for early detection of probable associated problems. Supplementation with coenzyme Q10 and folinic acid may be indicated. Exercise may help patients with myopathy. . All patients with Kearns-Sayre syndrome need periodical visits by an ophthalmologist. Consult with a cardiologist regarding pacemaker insertion for heart block. Additional consultations (such as endocrinologist, neurologist, psychiatrist, and neuropsychologist) may be needed, based on the status of the patient and the presence of complications. Genetic counseling is also indicated.

Prognosis

Kearns-Sayre syndrome is a progressive disorder, and the prognosis for patients with the condition is poor. Death frequently occurs in the third or fourth decade of life. Like many other disorders with mtDNA deletion, women who have Kearns-Sayre syndrome have a higher risk of having children who are clinically affected. At present, this risk is estimated to be about 4%.

Key words: Kearns-Sayre, Mitochondrial, Ataxia, Retinopathy