

MITO PROFILE

KEARNS-SAYRE SYNDROME INFORMATION

PROVIDED BY THE UNITED MITOCHONDRIAL DISEASE FOUNDATION

KEARNS-SAYRE SYNDROME (KSS)

KSS is a slowly progressive multi-system mitochondrial disease that often begins with drooping of the eyelids (ptosis). Other eye muscles eventually become involved, resulting in paralysis of eye movement. Degeneration of the retina usually causes difficulty seeing in dimly lit environments.

KSS is characterized by three main features:

- typical onset before age 20 although may occur in infancy or adulthood
- paralysis of specific eye muscles (called chronic progressive external ophthalmoplegia – CPEO)
- degeneration of the retina causing abnormal accumulation of pigmented (colored) material (pigmentary retinopathy).

In addition, one or more of the following conditions is present:

- block of electrical signals in the heart (cardiac conduction defects)
- elevated cerebrospinal fluid protein
- incoordination of movements (ataxia).

Patients with KSS may also have such problems as deafness, dementia, kidney dysfunction, and muscle weakness. Endocrine abnormalities including growth retardation, short stature, or diabetes may also be evident.

KSS is a rare disorder. One online eMedicine article found only 226 cases worldwide in the literature by 1992 (*Kearns-Sayre Syndrome* by Ewa Posner, MD). It is usually caused by a single large deletion (loss) of genetic material within the DNA of the mitochondria (mtDNA), rather than in the DNA of the cell nucleus. These deletions, of which there are over 150 species, typically arise spontaneously. Less frequently, the mutation is transmitted by the mother.

As with all mitochondrial diseases, there is no cure for KSS. Treatments are based on the types of symptoms and organs involved, and may include: Coenzyme Q10, insulin for diabetes, cardiac drugs, and a cardiac pacemaker which may be life-saving. Surgical intervention for drooping eyelids may be considered but should be undertaken by specialists in ophthalmic surgical centers.

KSS is slowly progressive and the prognosis varies depending on severity. Death is common in the third or fourth decade and may be due to organ system failures.

RESOURCES FOR KSS INFORMATION

Kearns-Sayre Syndrome, online eMedicine and OMIM articles by same title Available through UMDF website www.umdf.org, then *For patients and families*, then *disease descriptions*. Click on *KSS* and follow the links for other resources, which are the eMedicine and OMIM articles

Kearns-Sayre Syndrome – 3 Case Reports and Review of Clinical Features by Seon Bae Park, Kyoung tak Ma, Koung Hun Kook, and Sang Yeul Lee
<http://www.eymj.org/2004/pdf/08727.pdf>

Kearns-Sayre Syndrome short description from NORD. Full text may be ordered from site.
http://www.rarediseases.org/search/rdbdetail_abstract.html?disname=Kearns%20Sayre%20Syndrome

NINDS Kearns-Sayre Syndrome Information Page
http://www.ninds.nih.gov/disorders/kearns_sayre/kearns_sayre.htm

Kearns-Sayre Syndrome
http://malattierare.pediatria.unipd.it/pubblicaMR/mr_dx_ing.asp?mr=196

Mitochondrial DNA Deletion Syndromes
<http://www.geneclinics.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=4bDo8wuPEJdqm&gry=&fcn=y&fw=Tex-&filename=/profiles/kss/index.html>



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