DISEASE:
Kearns-Sayre syndrome

NAME:	Kearns-Sayre syndrome
DESCRIPTION:	A rare inborn error of metabolism that is characterized by progressive external ophthalmoplegia (PEO), pigmentary retinitis and an onset before the age of 20 years. Common additional features include deafness, cerebellar ataxia and heart block.
ORPHACODE:	480
XREF(S):	Orphanet ICD-10 OMIM MeSH MedDRA
ANALYTE(S):	MT-ATP8 RRM2B MT-TL1
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RELATED CONTENT

Related Genetic Tests

- Leigh / NARP Syndrome
- Mitochondrial disorders (gene panel)
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- mitochondrially encoded ATP synthase membrane subunit 8
- mitochondrially encoded tRNA-Leu (UUA/G) 1
- ribonucleotide reductase regulatory TP53 inducible subunit M2B

Related Gene Panels

- Leigh syndrome (mtDNA / 37 genes) - VUB
- mitochondrial disease, nuclear based (343 genes) - VUB