DISEASE:
Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy

NAME:	Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy
DESCRIPTION:	A rare mitochondrial disease characterized by adult onset of progressive external ophthalmoplegia, exercise intolerance, muscle weakness, manifestations of spinocerebellar ataxia (e.g. impaired gait, dysarthria) and mild motor peripheral neuropathy. Respiratory insufficiency has been reported in some cases.
ORPHACODE:	329336
SYNONYMS:	Adult-onset CPEO with mitochondrial myopathy
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	RRM2B RNASEH1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Chronic progressive external ophthalmoplegia (CPEO) (Full sequencing of mtDNA genome)
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- ribonuclease H1
- ribonucleotide reductase regulatory TP53 inducible subunit M2B

Related Gene Panels

- mitochondrial disorders, mitochondrial DNA based / mtDNA resequencing - VUB