DISEASE:
Autosomal recessive progressive external ophthalmoplegia

NAME:	Autosomal recessive progressive external ophthalmoplegia
DESCRIPTION:	A rare genetic, neuro-ophthalmological disease characterized by progressive weakness of the external eye muscles, resulting in bilateral ptosis and diffuse, symmetric ophthalmoparesis. Additional signs may include generalized skeletal muscle weakness, muscle atrophy, sensory axonal neuropathy, ataxia, cardiomyopathy, and psychiatric symptoms. It is usually more severe than autosomal dominant form.
ORPHACODE:	254886
SYNONYMS:	arPEO
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	POLG TK2
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RELATED CONTENT

Related Genetic Tests

- Charcot-Marie-Tooth (other than type 1A) (gene panel, IPN panel)
- Mitochondrial disorders (gene panel)
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- DNA polymerase gamma, catalytic subunit
- thymidine kinase 2

Related Gene Panels

- Inherited Peripheral Neuropathies gene panel (139 genes) - KUL
- mitochondrial disease, nuclear based (343 genes) - VUB
- mitochondrial disorders, mitochondrial DNA based / mtDNA resequencing - VUB