DISEASE:
Autosomal dominant progressive external ophthalmoplegia

NAME:	Autosomal dominant 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 skeletal muscle weakness, cataracts, hearing loss, sensory axonal neuropathy, ataxia, parkinsonism, cardiomyopathy, hypogonadism and depression. It is usually less severe than autosomal recessive form.
ORPHACODE:	254892
SYNONYMS:	adPEO
XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	POLG POLG2 SLC25A4 TWNK RRM2B
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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)
- Progressive external ophthalmoplegia

Related Laboratories

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

Related Analytes

- DNA polymerase gamma, catalytic subunit
- DNA polymerase gamma 2, accessory subunit
- ribonucleotide reductase regulatory TP53 inducible subunit M2B
- solute carrier family 25 member 4
- twinkle mtDNA helicase

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

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