DISEASE:
Mitochondrial DNA depletion syndrome, myopathic form

NAME:	Mitochondrial DNA depletion syndrome, myopathic form
DESCRIPTION:	A form of mitochondrial DNA depletion syndrome that displays a broad phenotypic spectrum but that is most often characterized by hypotonia, proximal muscle weakness, facial and bulbar weakness and failure to thrive.
ORPHACODE:	254875
SYNONYMS:	mtDNA depletion syndrome, myopathic form
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	TK2
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Mitochondrial disorders (gene panel)
- Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)
- mtDNA depletion syndrome

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- thymidine kinase 2

Related Gene Panels

- mitochondrial disease, nuclear based (343 genes) - VUB
- mtDNA depletion syndrome (6 genes) - VUB

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