DISEASE:
DNA2-related mitochondrial DNA deletion syndrome

NAME:	DNA2-related mitochondrial DNA deletion syndrome
DESCRIPTION:	A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by either late-onset myopathy with progressive external ophthalmoplegia and muscular weakness (predominantly limb-girdle) or early-onset myopathy presenting with decreased fetal movements, congenital ptosis, progressive external ophthalmoplegia, hypotonia and, variably, joint contractures. Reduced content and multiple deletions of mitochondrial DNA is observed in muscle biopsy.
ORPHACODE:	352470
SYNONYMS:	Mitochondrial DNA deletion syndrome with limb-girdle weakness Mitochondrial DNA deletion syndrome with progressive myopathy mtDNA deletion syndrome with limb-girdle weakness mtDNA deletion syndrome with progressive myopathy
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	DNA2
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