

DISEASE:
Combined oxidative phosphorylation defect type 7

NAME:	Combined oxidative phosphorylation defect type 7
DESCRIPTION:	Combined oxidative phosphorylation defect type 7 is a rare mitochondrial disease due to a defect in mitochondrial protein synthesis characterized by a variable phenotype that includes onset in infancy or early childhood of failure to thrive and psychomotor regression (after initial normal development), as well as ocular manifestations (such as ptosis, nystagmus, optic atrophy, ophthalmoplegia and reduced vision). Additional manifestations include bulbar paresis with facial weakness, hypotonia, difficulty chewing, dysphagia, mild dysarthria, ataxia, global muscle atrophy, and areflexia. It has a relatively slow disease progression with patients often living into the third decade of life.
ORPHACODE:	254930
SYNONYMS:	COXPD7 Severe C12ORF65-related COXPD Severe C12ORF65-related combined oxidative phosphorylation defect
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	MTRFR
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14
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