DISEASE:
Mitochondrial myopathy with reversible cytochrome C oxidase deficiency

NAME:	Mitochondrial myopathy with reversible cytochrome C oxidase deficiency
DESCRIPTION:	A rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a potentially life-threatening, severe myopathy manifesting in the neonatal to early infantile period, followed by marked, spontaneous improvement of muscular function by early childhood. Associated biochemical findings include lactic acidosis and a transient, marked decrease in respiratory chain activity.
ORPHACODE:	254864
SYNONYMS:	Benign COX deficiency Infantile reversible cytochrome C oxidase deficiency myopathy Mitochondrial myopathy with reversible COX deficiency Mitochondrial myopathy with reversible complex IV deficiency Reversible infantile cytochrome C oxidase deficiency Reversible infantile respiratory chain deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	TRMU MT-TE
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