

DISEASE:
Isolated complex III deficiency

NAME:	Isolated complex III deficiency
DESCRIPTION:	Isolated complex III deficiency is a rare, genetic, mitochondrial oxidative phosphorylation disorder characterized by a wide spectrum of clinical manifestations ranging from isolated myopathy or transient hepatopathy to severe multisystem disorder (that may include hypotonia, failure to thrive, psychomotor delay, cardiomyopathy, encephalopathy, renal tubulopathy, hearing impairment, lactic acidosis, hypoglycemia and other signs and symptoms).
ORPHACODE:	1460
SYNONYMS:	Isolated CoQ-cytochrome C reductase deficiency Isolated coenzyme Q-cytochrome C reductase deficiency Isolated mitochondrial respiratory chain complex III deficiency Isolated ubiquinone-cytochrome C reductase deficiency

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10
ANALYTE(S):	BCS1L MT-CYB UQCRQ UQCRB TTC19 UQCRC2 CYC1 LYRM7 UQCC2 UQCC3 UQCRFS1
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Related Analytes

- [BCS1 homolog, ubiquinol-cytochrome c reductase complex chaperone](#)
 - [cytochrome c1](#)
 - [LYR motif containing 7](#)
 - [mitochondrially encoded cytochrome b](#)
 - [tetratricopeptide repeat domain 19](#)
 - [ubiquinol-cytochrome c reductase complex assembly factor 2](#)
 - [ubiquinol-cytochrome c reductase complex assembly factor 3](#)
 - [ubiquinol-cytochrome c reductase binding protein](#)
 - [ubiquinol-cytochrome c reductase core protein 2](#)
 - [ubiquinol-cytochrome c reductase, Rieske iron-sulfur polypeptide 1](#)
 - [ubiquinol-cytochrome c reductase complex III subunit VII](#)
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