DISEASE:
Isolated succinate-CoQ reductase deficiency

NAME:	Isolated succinate-CoQ reductase deficiency
DESCRIPTION:	A rare, mitochondrial oxidative phosphorylation disorder characterized by a highly variable phenotype. The severe, multisystemic disease involves brain, heart, muscles, liver, kidneys, and eyes and results in death in infancy. Mildly affected individuals have only isolated cardiac or muscle involvement in the adulthood. Histochemical and biochemical analysis reveals a global reduction of succinate dehydrogenase activity.
ORPHACODE:	3208
SYNONYMS:	Isolated mitochondrial respiratory chain complex II deficiency Isolated succinate dehydrogenase deficiency Isolated succinate-coenzyme Q reductase deficiency Isolated succinate-ubiquinone reductase deficiency
XREF(S):	Orphanet OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	SDHA SDHB SDHD SDHAF1
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RELATED CONTENT

Related Genetic Tests

- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Mitochondrial complex II deficiency
- Mitochondrial disorders (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- succinate dehydrogenase complex flavoprotein subunit A
- succinate dehydrogenase complex assembly factor 1
- succinate dehydrogenase complex iron sulfur subunit B
- succinate dehydrogenase complex subunit D

Related Gene Panels

- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG
- mitochondrial disease, nuclear based (343 genes) - VUB