DISEASE:
Hereditary myopathy with lactic acidosis due to ISCU deficiency

NAME:	Hereditary myopathy with lactic acidosis due to ISCU deficiency
DESCRIPTION:	A rare disease characterised by myopathy with severe exercise intolerance and deficiencies of skeletal muscle succinate dehydrogenase and aconitase.
ORPHACODE:	43115
SYNONYMS:	Aconitase deficiency ISCU myopathy Iron-sulfur cluster deficiency myopathy Myopathy with exercise intolerance, Swedish type
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ISCU
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RELATED CONTENT

Related Genetic Tests

- Mitochondrial disorders (gene panel)
- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- iron-sulfur cluster assembly enzyme

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB
- mitochondrial disease, nuclear based (343 genes) - VUB