DISEASE:
Barth syndrome

NAME:	Barth syndrome
DESCRIPTION:	Barth syndrome (BTHS) is an inborn error of phospholipid metabolism characterized by dilated cardiomyopathy (DCM), skeletal myopathy, neutropenia, growth delay and organic aciduria.
ORPHACODE:	111
SYNONYMS:	3-methylglutaconic aciduria type 2 BTHS Cardioskeletal myopathy with neutropenia and abnormal mitochondria Cardioskeletal myopathy-neutropenia syndrome MGA2 X-linked cardioskeletal myopathy and neutropenia
XREF(S):	Orphanet MeSH ICD-10 OMIM
ANALYTE(S):	TAFAZZIN
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Cardiomyopathy, hereditary (gene panel)
- Cardiopathies, hereditary (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- tafazzin, phospholipid-lysophospholipid transacylase

Related Gene Panels

- Cardiomyopathy, hereditary (208 genes) - VUB
- Cardiopathies, hereditary (102 genes) - KUL