

Kardiomiopatia - hipotonia - kwasica mleczanowa

Kod Orpha: 91130 Kod OMIM: 610773

Opis choroby *

Definicja

Cardiomyopathy-hypotonia-lactic acidosis syndrome is characterised by hypertrophic cardiomyopathy, muscular hypotonia and the presence of lactic acidosis at birth. It has been described in two sisters (both of whom died within the first year of life) from a nonconsanguineous Turkish family. The syndrome is caused by a homozygous point mutation in the exon 3A of the *SLC25A3* gene encoding a mitochondrial membrane transporter.

Dane

Klasyfikacja

Choroba

Kod ORPHA

91130

Kod OMIM

610773

Kod ICD10

G71.3

Kod ICD11

5C53.30

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.