

## 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

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### \*Źródło

orphanet