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