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 <i>SLC25A3</i>

Dane

Klasyfikacja Choroba

Kod ORPHA 91130

Kod OMIM 610773 Kod ICD10 G71.3

Kod ICD11 5C53.30

<u>*Źródło</u>

orphanet