

Opis choroby *

Definicja

Lethal left ventricular non-compaction-seizures-hypotonia-cataract-developmental delay syndrome is rare, genetic, neurometabolic disease characterized by global developmental delay, severe hypotonia, seizures, cataracts, cardiomyopathy (including left or bi-ventricular hypertrophy, dilated cardiomyopathy) and left ventricular non-compaction, typically resulting in infantile or early-childhood death. Patients usually present metabolic lactic acidosis, failure to thrive, head lag, respiratory problems and decrease in respiratory chain complex activity. Highly variable cerebral abnormalities have been reported and include microcephaly, prominent extra-axial cerebrospinal fluid spaces, diffuse neuronal loss and cortical/white matter gliosis.

Dane

Klasyfikacja

Choroba

Kod ORPHA

478049

Kod OMIM

617228

Kod ICD10

E88.8

Kod ICD11

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*Źródło

orphanet