

Opis choroby *

Definicja

Lethal infantile mitochondrial myopathy is a rare mitochondrial oxidative phosphorylation disorder characterized by progressive generalized hypotonia, progressive external ophthalmoplegia and severe lactic acidosis, which results in early fatality (days to months after birth). Patients may present with lethargy and areflexia and may associate additional features, such as cardiomyopathy, renal dysfunction, liver involvement and seizures.

Dane

Klasyfikacja

Choroba

Synonimy

LIMD

LIMD

LIMM

Śmiertelna niemowlęca choroba
mitochondrialna

LIMM

Lethal infantile mitochondrial disease

Kod ORPHA

254857

Kod OMIM

551000

Kod ICD10

G71.3

Kod ICD11

8C73.Y

[*Źródło](#)

orphanet