

## 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

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#### [\\*Źródło](#)

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