

## Opis choroby \*

### Definicja

A rare, genetic, skeletal muscle disease characterized by an early-onset hypotonia, muscle weakness, global developmental delay with intellectual disability, and cardiomyopathy. Congenital structural heart defects and ichthyosiform cutaneous lesions have also been associated. Muscle biopsy shows characteristic enlarged mitochondria located at the periphery of muscle fibers.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Congenital megaconial myopathy  
Wrodzona dystrofia mięśni typu megaconial  
Wrodzona dystrofia mięśniowa z zaburzeniami struktury mitochondriów  
Wrodzona miopatia typu megaconial  
Congenital muscular dystrophy due to phosphatidylcholine biosynthesis defect  
Congenital muscular dystrophy with mitochondrial structural abnormalities

#### Kod ORPHA

280671

#### Kod OMIM

602541

#### Kod ICD10

G71.2

#### Kod ICD11

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#### \*Źródło

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