

## Opis choroby \*

### Definicja

A rare congenital myopathy characterized by neonatal onset of severe muscle weakness with selective atrophy/hypotrophy or absence of type II myofibers. Patients present at birth with hypotonia and respiratory failure, as well as mild facial and severe axial and proximal upper and lower limb weakness with areflexia and mild contractures. Eye movements and cardiac function are normal.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Congenital myopathy with fast-twitch fiber atrophy  
Congenital myopathy with reduced type II muscle fibers  
Congenital myopathy with type 2 muscle fiber atrophy  
Congenital myopathy with type II fiber atrophy  
Congenital myopathy with fast-twitch fiber atrophy  
Congenital myopathy with reduced type II muscle fibers  
Congenital myopathy with type 2 muscle fiber atrophy  
Congenital myopathy with type II fiber atrophy

#### Kod ORPHA

544602

#### Kod OMIM

618414

#### Kod ICD10

G71.2

#### Kod ICD11

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