# Wrodzona miopatia ze zredukowanymi włóknami mięśniowymi typu 2
# Kod Orpha: 544602 Kod OMIM: 618414

## 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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*Źródło

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.