

## Opis choroby \*

### Definicja

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a childhood onset of progressive shoulder and pelvic girdle muscle weakness and atrophy frequently associated with calf hypertrophy, diaphragmatic weakness, and/or variable cardiac abnormalities. Mild to moderate elevated serum creatine kinase levels and positive Gowers sign are reported.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal recessive limb-girdle muscular dystrophy type 2C  
Dystrofia obręczowo-kończynowa z powodu niedoboru gamma-sarkoglikanu  
Gamma-sarkoglikanopatia  
LGMD2C  
Gamma-sarcoglycan-related LGMD R5  
Gamma-sarcoglycanopathy  
LGMD due to gamma-sarcoglycan deficiency  
LGMD type 2C  
LGMD2C  
Limb-girdle muscular dystrophy due to gamma-sarcoglycan deficiency  
Limb-girdle muscular dystrophy type 2C

#### Kod ORPHA

353

#### Kod OMIM

253700

#### Kod ICD10

G71.0

#### Kod ICD11

8C70.41

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#### [\\*Źródło](#)

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