

## Opis choroby \*

### Definicja

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by childhood onset of progressive proximal weakness of the shoulder and pelvic girdle muscles, resulting in difficulty walking, scapular winging, calf hypertrophy and contractures of the Achilles tendon, which lead to a tiptoe gait pattern. Cardiac and respiratory involvement is rare.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Alpha-sarcoglycan-related LGMD R3  
Alfa-sarkoglikanopatia  
Dystrofia obręczowo-kończynowa z powodu niedoboru alfa-sarkoglikanu  
LGMD2D  
Alpha-sarcoglycanopathy  
Autosomal recessive limb-girdle muscular dystrophy type 2D  
LGMD due to alpha-sarcoglycan deficiency  
LGMD type 2D  
LGMD2D  
Limb-girdle muscular dystrophy due to alpha-sarcoglycan deficiency  
Limb-girdle muscular dystrophy type 2D

#### Kod ORPHA

62

#### Kod OMIM

608099

#### Kod ICD10

G71.0

#### Kod ICD11

8C70.41

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#### \*Źródło

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