

## Opis choroby \*

### Definicja

A subtype of autosomal recessive limb girdle muscular dystrophy characterized by a variable age of onset of progressive, typically symmetrical and selective weakness and atrophy of proximal shoulder- and pelvic-girdle muscles (gluteus maximus, thigh adductors, and muscles of the posterior compartment of the limbs are most commonly affected) without cardiac or facial involvement. Clinical manifestations include exercise intolerance, a waddling gait, scapular winging and calf pseudo-hypertrophy.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal recessive limb-girdle muscular dystrophy type 2A  
Dystrofia obręczowo-kończynowa z powodu niedoboru kalpajny  
Kalpainopatia pierwotna  
LGMD2A  
Calpain-3-related LGMD R1  
LGMD type 2A  
LGMD2A  
Limb-girdle muscular dystrophy due to calpain deficiency  
Limb-girdle muscular dystrophy type 2A  
Primary calpainopathy

#### Kod ORPHA

267

#### Kod OMIM

618129

#### Kod ICD10

G71.0

#### Kod ICD11

8C70.41

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#### [\\*Źródło](#)

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