

## Opis choroby \*

### Definicja

A mild subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a variable onset (ranging from infancy to adolescence) of progressive proximal upper and lower limb muscle weakness and atrophy. Mild scapular winging, calf hypertrophy, and lack of respiratory and cardiac involvement are also observed.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Autosomal recessive limb-girdle muscular dystrophy type 2G  
Dystrofia obręczowo-kończynowa z powodu niedoboru teletoniny  
LGMD2G  
LGMD due to telethonin deficiency  
LGMD type 2G  
LGMD2G  
Limb-girdle muscular dystrophy due to telethonin deficiency  
Limb-girdle muscular dystrophy type 2G  
Telethonin-related LGMD R7

#### Kod ORPHA

34514

#### Kod OMIM

601954

#### Kod ICD10

G71.0

#### Kod ICD11

8C70.41

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#### \*Źródło

orphanet