## Opis choroby *

Definicja

A rare subtype of autosomal recessive limb-girdle muscular dystrophy disorder characterized by infantile to childhood-onset of slowly progressive, principally proximal, shoulder and/or pelvic-girdle muscular weakness that typically presents with positive Gowers' sign and is associated with elevated creatine kinase levels, hyporeflexia, joint and achilles tendon contractures, and muscle hypertrophy, usually of the thighs, calves and/or tongue. Other highly variable features include cerebellar, cardiac and ocular abnormalities.

Dane

**Klasyfikacja**
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2U
Autosomalna recesywna dystrofia obręczowo-kończynowa typu 2U
LGMD2U
ISPD-related LGMD R20
LGMD type 2U
LGMD2U
Limb-girdle muscular dystrophy type 2U

**Kod ORPHA**
352479

**Kod OMIM**
616052

**Kod ICD10**
G71.0

**Kod ICD11**
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*Źródło

orphanet