

## Opis choroby \*

### Definicja

A form of limb-girdle muscular dystrophy characterized by slowly-progressive, mainly proximal, muscle weakness presenting in early childhood (with difficulties walking and climbing stairs) and mild to severe intellectual disability. Additional manifestations reported include microcephaly, mild increase in thigh or calf muscles, and contractures of the ankles.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Alpha-dystroglycan-related LGMD R16

LGMD2P

Autosomal recessive limb-girdle muscular dystrophy type 2P

LGMD type 2P

LGMD2P

Limb-girdle muscular dystrophy type 2P

#### Kod ORPHA

280333

#### Kod OMIM

613818

#### Kod ICD10

G71.0

#### Kod ICD11

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orphanet