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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*Źródło

orphanet