Opis choroby *

Definicja

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by an onset in late adolescence or early adulthood of slowly progressive, proximal weakness and atrophy of shoulder and pelvic girdle muscles. Cardiac and respiratory muscles are not involved. Hypertrophy of the calf muscles and highly elevated serum creatine kinase levels are frequently observed.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2B
Dystrofia obręczowo-kończynowa z powodu niedoboru dysferliny
LGMD2B
Dysferlin-related LGMD R2
LGMD due to dysferlin deficiency
LGMD type 2B
LGMD2B
Limb-girdle muscular dystrophy due to dysferlin deficiency
Limb-girdle muscular dystrophy type 2B

Kod ORPHA
268

Kod OMIM
253601

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet