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