Opis choroby *

Definicja

A mild subtype of autosomal recessive limb girdle muscular dystrophy characterized by slowly progressive proximal muscle weakness and wasting of the pelvic and shoulder girdles with onset that usually occurs during the second or third decade of life. Clinical presentation is variable and can include calf psuedohypertrophy, joint contractures, scapular winging, muscle cramping and/or facial and respiratory muscle involvement.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2H
Dystrofia obręczowo-kończynowa z powodu niedoboru TRIM32
LGMD2H
LGMD due to TRIM32 deficiency
LGMD type 2H
LGMD2H
Limb-girdle muscular dystrophy due to TRIM32 deficiency
Limb-girdle muscular dystrophy type 2H
Sarcotubular myopathy
TRIM32-related LGMD R8

Kod ORPHA
1878

Kod OMIM
254110

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet