Opis choroby *

Definicja

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a childhood onset of progressive shoulder and pelvic girdle muscle weakness and atrophy frequently associated with calf hypertrophy, diaphragmatic weakness, and/or variable cardiac abnormalities. Mild to moderate elevated serum creatine kinase levels and positive Gowers sign are reported.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2C
Dystrofia obręczowo-kończynowa z powodu niedoboru gamma-sarkoglikanu
Gamma-sarkoglikanopatia
LGMD2C
Gamma-sarcoglycan-related LGMD R5
Gamma-sarcoglycanopathy
LGMD due to gamma-sarcoglycan deficiency
LGMD type 2C
LGMD2C
Limb-girdle muscular dystrophy due to gamma-sarcoglycan deficiency
Limb-girdle muscular dystrophy type 2C

Kod ORPHA
353

Kod OMIM
253700

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet