Opis choroby *

Definicja

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by childhood onset of progressive proximal weakness of the shoulder and pelvic girdle muscles, resulting in difficulty walking, scapular winging, calf hypertrophy and contractures of the Achilles tendon, which lead to a tiptoe gait pattern. Cardiac and respiratory involvement is rare.

Dane

Klasyfikacja
Choroba

Synonimy
Alpha-sarcoglycan-related LGMD R3
Alfa-sarkoglikanopatia
Dystrofia obręczowo-kończynowa z powodu niedoboru alfa-sarkoglikanu
LGMD2D
Alpha-sarcoglycanopathy
Autosomal recessive limb-girdle muscular dystrophy type 2D
LGMD due to alpha-sarcoglycan deficiency
LGMD type 2D
LGMD2D
Limb-girdle muscular dystrophy due to alpha-sarcoglycan deficiency
Limb-girdle muscular dystrophy type 2D

Kod ORPHA
62

Kod OMIM
608099

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet