Opis choroby *

Definicja

A subtype of autosomal recessive limb-girdle muscular dystrophy characterized by a variable age of onset of progressive weakness and wasting of the proximal skeletal muscles of the shoulder and pelvic girdles, frequently associated with progressive respiratory muscle impairment and cardiomyopathy. Calf hypertrophy, muscle cramps and elevated serum creatine kinase levels are also observed. Neuropsychomotor development is usually normal.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2F
Delta-sarkoglikanopatia
Dystrofia obręczowo-kończynowa z powodu niedoboru delta-sarkoglikanu
LGMD2F
Delta-sarcoglycan-related LGMD R6
Delta-sarcoglycanopathy
LGMD due to delta-sarcoglycan deficiency
LGMD type 2F
LGMD2F
Limb-girdle muscular dystrophy due to delta-sarcoglycan deficiency
Limb-girdle muscular dystrophy type 2F

Kod ORPHA
219

Kod OMIM
601287

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet