Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy characterized by proximal muscle weakness presenting in early childhood (with occasional falls and difficulties in climbing stairs) and a progressive course resulting in loss of ambulation in early adulthood. Muscle atrophy and multiple contractures have also been reported in rare cases.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2Q
Autosomalna recesywna dystrofia obręczowo-kończynowa z powodu niedoboru plektyny
LGMD2Q
LGMD type 2Q
LGMD2Q
Limb-girdle muscular dystrophy type 2Q
Plectin-related LGMD R17

Kod ORPHA
254361

Kod OMIM
613723

Kod ICD10
G71.0

Kod ICD11
-

---

*Źródło

orphanet