Opis choroby *

Definicja

A subtype of autosomal recessive limb girdle muscular dystrophy characterized by a childhood to adolescent onset of progressive pelvic- and shoulder-girdle muscle weakness, particularly affecting the pelvic girdle (adductors and flexors of hip). Usually the knees are the earliest and most affected muscles. In advanced stages, involvement of the shoulder girdle (resulting in scapular winging) and the distal muscle groups are observed. Calf hypertrophy, cardiomyopathy, respiratory impairment, tendon contractures, scoliosis, and exercise-induced myoglobinuria may be observed.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2E
Beta-sarkoglikanopatia
Dystrofia obręczowo-kończynowa z powodu niedoboru beta-sarkoglikanu
LGMD2E
Beta-sarcoglycan-related LGMD R4
Beta-sarcoglycanopathy
LGMD due to beta-sarcoglycan deficiency
LGMD type 2E
LGMD2E
Limb-girdle muscular dystrophy due to beta-sarcoglycan deficiency
Limb-girdle muscular dystrophy type 2E

Kod ORPHA
119

Kod OMIM
604286

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet