Opis choroby *

Definicja

A subtype of autosomal recessive limb girdle muscular dystrophy characterized by a variable age of onset of progressive, typically symmetrical and selective weakness and atrophy of proximal shoulder- and pelvic-girdle muscles (gluteus maximus, thigh adductors, and muscles of the posterior compartment of the limbs are most commonly affected) without cardiac or facial involvement. Clinical manifestations include exercise intolerance, a waddling gait, scapular winging and calf pseudo-hypertrophy.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2A
Dystrofia obręczowo-kończynowa z powodu niedoboru kalpainy
Kalpainopatia pierwotna
LGMD2A
Calpain-3-related LGMD R1
LGMD type 2A
LGMD2A
Limb-girdle muscular dystrophy due to calpain deficiency
Limb-girdle muscular dystrophy type 2A
Primary calpainopathy

Kod ORPHA
267

Kod OMIM
618129, 253600

Kod ICD10
G71.0

Kod ICD11
8C70.41

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*Źródło

orphanet