

Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy most often characterized by an adult onset (but ranging from 11 to 51 years) of mainly proximal lower limb weakness, with difficulties standing on tiptoes being one of the initial signs. Proximal upper limb and distal lower limb weakness is also common, as well as atrophy of the quadriceps (most commonly), biceps brachii, and lower leg muscles. Calf hypertrophy has also been reported in some cases. LGMD2L progresses slowly, with most patients remaining ambulatory until late adulthood.

Dane

Klasyfikacja

Choroba

Synonimy

Anoctamin-5-related LGMD R12

LGMD2L

Autosomal recessive limb-girdle muscular dystrophy type 2L

LGMD type 2L

LGMD2L

Limb-girdle muscular dystrophy type 2L

Kod ORPHA

206549

Kod OMIM

611307

Kod ICD10

G71.0

Kod ICD11

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*Źródło

orphanet