Autosomalna recesywna dystrofia obręczowo-kończynowa typu 2L
Kod Orpha: 206549 Kod OMIM: 611307

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
-

---

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.