Opis choroby *

Definicja

A rare autosomal recessive limb-girdle muscular dystrophy characterized by adult onset of progressive muscle weakness and atrophy in the proximal upper and lower limbs, leading to scapular winging and loss of independent ambulation. Respiratory function may become impaired in the course of the disease. Fatty degeneration of internal regions of thigh muscles sparing external areas has been reported, as well as a reduction of alpha-dystroglycan in muscle biopsies.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2Z
LGMD2Z
LGMD type 2Z
LGMD2Z
Limb-girdle muscular dystrophy type 2Z
POGLUT1-related LGMD R21

Kod ORPHA
480682

Kod OMIM
617232

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet