Opis choroby *

Definicja

A rare, mild subtype of autosomal dominant limb-girdle muscular dystrophy characterized by a typically adult onset of mild, progressive, proximal weakness of pelvic and shoulder girdle muscles and progressive, permanent finger and toes flexion limitation without flexion contractures. Normal to highly elevated creatine kinase serum levels are observed.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant limb-girdle muscular dystrophy type 1G
LGMD1G
HNRNPDL-related LGMD D3
LGMD type 1G
LGMD1G
Limb-girdle muscular dystrophy type 1G

Kod ORPHA
55596

Kod OMIM
609115

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet