

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	Synonimy
Choroba	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	Kod OMIM	Kod ICD10
55596	609115	G71.0

Kod ICD11

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*Źródło

orphanet