Opis choroby *

Definicja

A subtype of autosomal dominant limb-girdle muscular dystrophy characterized by an adult-onset of slowly progressive, proximal pelvic girdle weakness, with none, or only minimal, shoulder girdle involvement, and absence of cardiac and respiratory symptoms. Mild to moderate elevated creatine kinase serum levels and gait abnormalities are frequently observed.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal dominant limb-girdle muscular dystrophy type 1D
LGMD1D
DNAJB6-related LGMD D1
LGMD type 1D
LGMD1D
Limb-girdle muscular dystrophy type 1D

Kod ORPHA
34516

Kod OMIM
603511

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet