Opis choroby *

Definicja

A rare subtype of autosomal dominant limb girdle muscular dystrophy characterized by an adult onset of proximal shoulder and hip girdle weakness (that later progresses to include distal weakness), nasal speech and dysarthria. Other frequent findings include tightened heel cords, reduced deep-tendon reflexes and elevated creatine kinase serum levels. Respiratory failure, as well as mild facial weakness and dysphagia, may also be observed.

Dane

Klasyfikacja
Choroba

Synonimy
LGMD1A
Dystrofia obręczowo-kończynowa z powodu niedoboru miotiliny
LGMD1A
Limb-girdle muscular dystrophy due to myotilin deficiency

Kod ORPHA
266

Kod OMIM
609200

Kod ICD10
G71.0

Kod ICD11
8C70.40

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*Źródło

orphanet