Opis choroby *

Definicja

A rare subtype of autosomal dominant limb-girdle muscular dystrophy characterized by slowly progressive proximal muscular weakness initially affecting the lower limbs (and later involving the upper limbs), hypotrophy of upper and lower limb-girdle muscles, hyporeflexia, calf hypertrophy, and increased serum creatine kinase. There is no involvement of oculo-facial-bulbar muscles and cardiac muscle.

Dane

Klasyfikacja
Choroba

Synonimy
LGMD1H
LGMD1H

Kod ORPHA
238755

Kod OMIM
613530

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet