Opis choroby *

Definicja

A rare subtype of autosomal recessive limb-girdle muscular dystrophy characterized by atrioventricular block resulting in repeated syncope episodes, elevated creatine kinase serum levels and adult-onset of slowly progressive proximal limb skeletal muscle weakness and atrophy. Muscular dystrophic changes observed in muscle biopsy include diameter variability, increased central nuclei, and presence of necrotic and regenerating fibers.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy-cardiac arrhythmia syndrome
LGMD2X
Zespół autosomalnej recesywnej dystrofii obręczowo-kończynowej i arytmii serca
BVES-related LGMD
LGMD type 2X
LGMD2X
Limb-girdle muscular dystrophy 2X

Kod ORPHA
476084

Kod OMIM
616812

Kod ICD10
G71.0

Kod ICD11
-

---

*Źródło

orphanet