Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy characterized by an infantile onset of hypotonia, axial and proximal lower limb weakness (with severe weakness noted after febrile illnesses), cardiomyopathy and normal or reduced intelligence. Hypertrophy of calves, thighs, and triceps have also been reported in some cases.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive LGMD type 2M
LGMD2M
Autosomal recessive limb-girdle muscular dystrophy type 2M
Fukutin-related LGMD R13
LGMD type 2M
LGMD2M

Kod ORPHA
206554

Kod OMIM
611588

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet