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 Synonimy

Choroba 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 Kod ICD10 611588 G71.0

Kod ICD11

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*Źródło

orphanet