

Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy characterized by an onset in childhood or adolescence of rapidly progressive proximal limb muscle weakness (particularly affecting the neck, hip girdle, and shoulder abductors), hypertrophy in the calves and quadriceps, ankle contractures, and myopia.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive limb-girdle muscular dystrophy type 20
LGMD20
LGMD type 20
LGMD20
Limb-girdle muscular dystrophy type 20
POMGNT1-related LGMD R15

Kod ORPHA

206564

Kod OMIM

613157

Kod ICD10

G71.0

Kod ICD11

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*Źródło

orphanet