Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy characterized by proximal weakness (manifesting as slowness in running) presenting in infancy, along with calf hypertrophy, mild lordosis, scapular winging and normal intelligence (or mild intellectual disability).

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2N
LGMD2N
LGMD type 2N
LGMD2N
Limb-girdle muscular dystrophy type 2N
POMT2-related LGMD R14

Kod ORPHA
206559

Kod OMIM
613158

Kod ICD10
G71.0

Kod ICD11
-

---

*Źródło

orphanet