

Opis choroby *

Definicja

Limb-girdle muscular dystrophy due to POMK deficiency is a form of limb-girdle muscular dystrophy presenting in infancy with muscle weakness and delayed motor development (eventually learning to walk at 18 months of age) followed by progressive proximal weakness, pseudohypertrophy of calf muscles, mild facial weakness, and borderline intelligence.

Dane

Klasyfikacja

Choroba

Synonimy

LGMD due to POMK deficiency

LGMD spowodowana niedoborem POMK

Kod ORPHA

445110

Kod OMIM

616094

Kod ICD10

G71.0

Kod ICD11

8C70.41

*Źródło

orphanet