

Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy characterized by the onset of slowly progressive proximal muscle weakness during childhood (with fatigue and difficulty running and climbing stairs) and developmental delay. Mild intellectual deficit and microcephaly, without any obvious structural brain abnormality, are found in all patients. Mild pseudohypertrophy and joint contractures of the ankles have also been reported.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive limb-girdle muscular dystrophy type 2K

Dystrofia obręczowo-kończynowa-
niepełnosprawność intelektualna

LGMD2K

LGMD type 2K

LGMD2K

Limb-girdle muscular dystrophy type 2K

Limb-girdle muscular dystrophy-intellectual
disability syndrome

POMT1-related LGMD R11

Kod ORPHA

86812

Kod OMIM

609308

Kod ICD10

G71.0

Kod ICD11

8C70.41

*Źródło

orphanet