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

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*Źródło

orphanet