Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy that usually has a childhood onset (but can range from the first to third decade of life) of severe progressive proximal weakness, eventually involving the distal muscles. Some patients may remain ambulatory but most are wheelchair dependant 20 years after onset.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2J
LGMD2J
LGMD type 2J
LGMD2J
Limb-girdle muscular dystrophy type 2J
Titin-related LGMD R10

Kod ORPHA
140922

Kod OMIM
608807

Kod ICD10
G71.0

Kod ICD11
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*Źródło

orphanet