Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy, that can present from birth to early childhood, characterized by hypotonia, microcephaly, mild proximal muscle weakness (leading to delayed walking and difficulty climbing stairs), mild intellectual disability and epilepsy. Additional manifestations reported in some patients include cataracts, nystagmus, cardiomyopathy, and respiratory insufficiency.

Dane

Klasyfikacja
Choroba

Synonimy
Autosomal recessive limb-girdle muscular dystrophy type 2T
LGMD2T
GMPPB-related LGMD R19
LGMD type 2T
LGMD2T
Limb-girdle muscular dystrophy type 2T

Kod ORPHA
363623

Kod OMIM
615352

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet