

Opis choroby *

Definicja

A form of limb-girdle muscular dystrophy characterized by childhood-onset of progressive proximal muscle weakness (leading to reduced ambulation) with myalgia and fatigue, in addition to infantile hyperkinetic movements, truncal ataxia, and intellectual disability. Additional manifestations include scoliosis, hip dysplasia, and less commonly, ocular features (e.g. myopia, cataract) and seizures.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal recessive limb-girdle muscular dystrophy type 2S
LGMD2S
LGMD type 2S
LGMD2S
Limb-girdle muscular dystrophy type 2S
TRAPPC11-related LGMD R18

Kod ORPHA

369840

Kod OMIM

615356

Kod ICD10

G71.0

Kod ICD11

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[*Źródło](#)

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