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

orphanet