

## 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