

## Opis choroby \*

### Definicja

A rare, genetic, non-dystrophic myopathy disease characterized by childhood-onset severe external ophthalmoplegia, typically without ptosis, associated with mild, very slowly progressive muscular weakness and atrophy, involving the facial, neck flexor and limb (upper > lower, proximal > distal) muscles. Muscle biopsy shows type 1 fiber uniformity, absent, or abnormally small, type 2A fibers, increased variability of fiber size, internalized nuclei and/or fatty infiltration.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

363677

#### Kod OMIM

605637

#### Kod ICD10

G71.2

#### Kod ICD11

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### \*Źródło

orphanet