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