Opis choroby *

Definicja

A rare autosomal recessive congenital myopathy characterized by numerous centrally placed nuclei on muscle biopsy and clinical features of a congenital myopathy including facial weakness, ocular abnormalities (ptosis and external ophthalmoplegia) and predominant proximal muscle weakness of variable severity with possible distal involvement.

Dane

Klasyfikacja
Choroba

Synonimy
AR-CNM
AR-CNM

Kod ORPHA
169186

Kod OMIM
615959

Kod ICD10
G71.2

Kod ICD11
8C72.01

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*Źródło

orphanet