

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

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Kod ORPHA

169186

Kod OMIM

615959

Kod ICD10

G71.2

Kod ICD11

8C72.01

*Źródło

orphanet