Opis choroby *

Definicja

Congenital myopathy with internal nuclei and atypical cores is a rare genetic skeletal muscle disease characterized by neonatal hypotonia, distal more than proximal muscle weakness, progressive exercise intolerance with prominent myalgias, and mild-to-moderate overall motor impairment with preserved ambulation. Face, extraocular, cardiac, and respiratory muscles are unaffected. Mild cognitive impairment is also noted in most patients.

Dane

Klasyfikacja
Choroba

Synonimy
CNM4
CNM4
Miopatia z ośrodkowo ułożonymi jądrami typu 4
Centronuclear myopathy type 4

Kod ORPHA
319160

Kod OMIM
614807

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet