Opis choroby *

Definicja

A rare, autosomal dominant congenital myopathy characterized by numerous centrally placed nuclei on muscle biopsy and clinical features of a congenital myopathy (hypotonia, distal/proximal muscle weakness, rib cage deformities (sometimes associated with respiratory insufficiency), ptosis, ophthalmoparesis and weakness of the muscles of facial expression with dysmorphic facial features.

Dane

Klasyfikacja
Choroba

Synonimy
AD-CNM
AD-CNM

Kod ORPHA
169189

Kod OMIM
160150

Kod ICD10
G71.2

Kod ICD11
8C72.01

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*Źródło

orphanet