

## 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

---

#### \*Źródło

orphanet