

Opis choroby *

Definicja

A rare genetic, congenital, non-dystrophic myopathy characterized by neonatal or infantile-onset hypotonia and mild to severe generalized muscle weakness.

Dane

Klasyfikacja

Choroba

Synonimy

CFTDM

CFTDM

Kod ORPHA

2020

Kod OMIM

617760

Kod ICD10

G71.2

Kod ICD11

8C72.1

*Źródło

orphanet