

Opis choroby *

Definicja

A rare, genetic, congenital myopathy disorder characterized by variable degrees of muscular weakness, frequently associated with severe nemaline myopathy-like disease (including neonatal hypotonia, lack of spontaneous movements, feeding and swallowing difficulties, frequent respiratory infections, respiratory insufficiency, early death), and histopathologic findings of large, densely packed, subsarcolemmal accumulations of thin, actin-immunopositive filaments (with or without intranuclear nemaline rods) on muscle biopsy.

Dane

Klasyfikacja

Choroba

Synonimy

Actin myopathy

Miopatia aktynowa

Kod ORPHA

98904

Kod OMIM

161800

Kod ICD10

G71.2

Kod ICD11

8C72.0Y

*Źródło

orphanet