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

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*Źródło

orphanet