Opis choroby *

Definicja

A rare congenital myopathy characterized by neonatal onset of severe muscle weakness with selective atrophy/hypotrophy or absence of type II myofibers. Patients present at birth with hypotonia and respiratory failure, as well as mild facial and severe axial and proximal upper and lower limb weakness with areflexia and mild contractures. Eye movements and cardiac function are normal.

Dane

Klasyfikacja
Choroba

Synonimy
Congenital myopathy with fast-twitch fiber atrophy
Congenital myopathy with reduced type II muscle fibers
Congenital myopathy with type 2 muscle fiber atrophy
Congenital myopathy with type II fiber atrophy
Congenital myopathy with fast-twitch fiber atrophy
Congenital myopathy with reduced type II muscle fibers
Congenital myopathy with type 2 muscle fiber atrophy
Congenital myopathy with type II fiber atrophy

Kod ORPHA
544602

Kod OMIM
618414

Kod ICD10
G71.2

Kod ICD11
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*Źródło

orphanet