Opis choroby *

Definicja

A rare genetic neuromuscular disease characterized by early onset of proximal or generalized muscle weakness, external ophthalmoplegia with or without ptosis, and joint contractures. Hypotonia, neonatal respiratory distress necessitating ventilation, and severe dysphagia have also been reported. The disease is of variable severity and non- or slowly progressive. Patients typically remain ambulatory. Muscle biopsy may show predominance of type 1 fibers, marked variability in fiber size, increased internal nuclei, and proliferation of perimysial and endomysial connective tissue.

Dane

Klasyfikacja
Choroba

Synonimy
HIBM3
Dziedziczna miopatia z ciałkami wtrętowymi typu 3
HIBM3
IBM3
Miopatia z ciałkami wtrętowymi typu 3
Hereditary inclusion body myopathy type 3
IBM3
Inclusion body myopathy type 3

Kod ORPHA
79091

Kod OMIM
605637

Kod ICD10
G71.8

Kod ICD11
4A41.21

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*Źródło

orphanet