Opis choroby *

Definicja

A rare genetic neuromuscular disease characterized by adult onset of slowly progressive distal and/or proximal muscle weakness in the upper and lower extremities, and early involvement of respiratory muscles leading to respiratory failure. Additional features are neck flexor weakness, foot extensor weakness, and, in rare cases, mildly impaired cardiac function. Muscle biopsy shows eosinophilic myofibrillar inclusions referred to as cytoplasmic bodies, as well as fiber size variation, increased internal nuclei and connective tissue, fiber splitting, and rimmed vacuoles.

Dane

Klasyfikacja
Choroba

Synonimy
Edström Myopathy
ADMERF
Dziedziczna miopatia z ciałkami wtrętowymi z wczesną niewydolnością oddechową
HIBM-ERF
Miopatia Edströma
Miopatia miofibrylarna z wczesną niewydolnością oddechową
HIBM-ERF
HMERF
Hereditary inclusion body myopathy with early respiratory failure
MFM-titinopathy
Myofibrillar myopathy with early respiratory failure
Myofibrillar myopathy-titinopathy

Kod ORPHA
178464

Kod OMIM
603689

Kod ICD10
G71.0

Kod ICD11
8C70.Y

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*Źródło

orphanet