

## 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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