

## 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