

## Opis choroby \*

### Definicja

A rare, genetic, alpha-crystallinopathy disease characterized by adult-onset myofibrillar myopathy, variably associated with cardiomyopathy and/or posterior pole cataracts. Patients typically present progressive proximal and distal muscle weakness and wasting of lower and upper limbs, often with velopharyngeal involvement including dysphagia, dysphonia and ventilatory insufficiency. Electromyography shows myopathic features and muscle biopsy reveals myofibrillar myopathy changes.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Alpha-B crystallin-related late-onset distal myopathy

Dystlana krystalinopatia o późnym początku

Late-onset distal crystallinopathy

#### Kod ORPHA

399058

#### Kod OMIM

608810

#### Kod ICD10

G71.0

#### Kod ICD11

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#### \*Źródło

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