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

orphanet