

## Opis choroby \*

### Definicja

A rare, genetic neuromuscular disease characterized by progressive external ocular, facial and pharyngeal muscle weakness, leading to variable degrees of ptosis, ophthalmoparesis, facial muscle atrophy, dysarthria and dysphagia, as well as distal muscle weakness and atrophy of lower and upper extremities. Respiratory muscle involvement is common, but sensorineural hearing loss, asymmetrical extremity weakness and severe proximal weakness are rare.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

OPDM

Miopatia oczno-gardłowa dystalna

OPDM

Oculopharyngeal distal myopathy

#### Kod ORPHA

98897

#### Kod OMIM

618940

#### Kod ICD10

G71.0

#### Kod ICD11

9C82.1

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#### \*Źródło

orphanet