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