## Opis choroby \*

## Definicja

Vocal cord and pharyngeal distal myopathy (VCPDM) is a rare autosomal dominant distal myopathy characterized by adult onset of muscle weakness in the feet and hands (slowly progressing to involve proximal limb muscles) combined with vocal or swallowing dysfunction and frequent respiratory muscle involvement in later stages. Normal to mildly elevated creatine kinase (CK) serum levels and rimmed-vacuolated dystrophic muscle fiber changes are associated laboratory and pathologic findings.

Dane

**Klasyfikacja** Synonimy

Choroba Distal myopathy with vocal cord weakness

MATR3-related distal myopathy

**VCPDM** 

Distal myopathy with vocal cord weakness

MATR3-related distal myopathy

**VCPDM** 

**Kod ORPHA** 

600

**Kod OMIM** 

**Kod ICD10** 

606070

G71.0

**Kod ICD11** 

8C75

## \*Źródło

orphanet