

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

Choroba

Synonimy

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

606070

Kod ICD10

G71.0

Kod ICD11

8C75

*Źródło

orphanet