DISEASE:
Vocal cord and pharyngeal distal myopathy

NAME:	Vocal cord and pharyngeal distal myopathy
DESCRIPTION:	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.
ORPHACODE:	600
SYNONYMS:	Distal myopathy with vocal cord weakness MATR3-related distal myopathy VCPDM
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	MATR3
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Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

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