DISEASE:
Alpha-B crystallin-related late-onset myopathy

NAME:	Alpha-B crystallin-related late-onset myopathy
DESCRIPTION:	A rare, genetic, alpha-crystallinopathy disease characterized by adult-onset myofibrillar myopathy, variably associated with cardiomyopathy and/or posterior pole cataracts. Patients typically present progressive proximal and distal muscle weakness and wasting of lower and upper limbs, often with velopharyngeal involvement including dysphagia, dysphonia and ventilatory insufficiency. Electromyography shows myopathic features and muscle biopsy reveals myofibrillar myopathy changes.
ORPHACODE:	399058
SYNONYMS:	Alpha-B crystallin-related late-onset distal myopathy Late-onset distal crystallinopathy
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CRYAB
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- Centrum Medische Genetica - UZ Brussel VUB

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