

DISEASE:
Distal myopathy with anterior tibial onset

NAME:	Distal myopathy with anterior tibial onset
DESCRIPTION:	A rare, genetic neuromuscular disease characterized by a progressive muscle weakness starting in the anterior tibial muscles, later involving lower and upper limb muscles, associated with an increased serum creatine kinase levels and absence of dysferlin on muscle biopsy. Patients become wheelchair dependent.
ORPHACODE:	178400
SYNONYMS:	Distal anterior compartment myopathy
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>DYSF</u>
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- Centrum Medische Genetica - UZ Brussel VUB

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- dysferlin

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Source URL: <http://gentest.healthdata.be/disease/371>