DISEASE:
GNE myopathy

NAME:	GNE myopathy
DESCRIPTION:	GNE myopathy is a rare autosomal recessive distal myopathy characterized by early adult-onset, slowly to moderately progressive distal muscle weakness that preferentially affects the tibialis anterior muscle and that usually spares the quadriceps femoris. Muscle biopsy reveals presence of rimmed vacuoles.
ORPHACODE:	602
SYNONYMS:	DMRV Distal myopathy with rimmed vacuoles Distal myopathy, Nonaka type HIBM2 Hereditary inclusion body myopathy type 2 IBM2 Inclusion body myopathy type 2 Nonaka myopathy Quadriceps-sparing myopathy

XREF(S):	Orphanet OMIM MeSH ICD-10 OMIM
ANALYTE(S):	GNE
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RELATED CONTENT

Related Genetic Tests

- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB