DISEASE:
Childhood-onset nemaline myopathy

NAME:	Childhood-onset nemaline myopathy
DESCRIPTION:	Childhood onset nemaline myopathy, or mild nemaline myopathy is a type of nemaline myopathy (NM; see this terms) characterized by distal muscle weakness, and sometimes slowness of muscle contraction.
ORPHACODE:	171439
SYNONYMS:	Mild nemaline myopathy
XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10

ANALYTE(S):	TPM2 MYPN ACTA1 TPM3 NEB KBTBD13 KLHL41
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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

- actin alpha 1, skeletal muscle
- kelch repeat and BTB domain containing 13
- kelch like family member 41
- myopalladin
- nebulin
- tropomyosin 2
- tropomyosin 3

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB