

**DISEASE:**  
**Typical nemaline myopathy**

<b>NAME:</b>	Typical nemaline myopathy
<b>DESCRIPTION:</b>	Typical nemaline myopathy is a moderate neonatal form of nemaline myopathy (NM; see this term) characterized by facial and skeletal muscle weakness and mild respiratory involvement.
<b>ORPHACODE:</b>	171436
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">OMIM</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	<a href="#">ACTA1</a> <a href="#">CFL2</a> <a href="#">TPM2</a> <a href="#">NEB</a> <a href="#">KLHL41</a> <a href="#">LMOD3</a>
<b>CREATED:</b>	13 May 2019 - 01:02

**CHANGED:**

22 Jun 2023 - 16:14

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## RELATED CONTENT

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### Related Genetic Tests

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

### Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

### Related Analytes

- actin alpha 1, skeletal muscle
- cofilin 2
- kelch like family member 41
- leiomodin 3
- nebulin
- tropomyosin 2

### Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB