

**DISEASE:**  
**X-linked centronuclear myopathy**

|                     |                                                                                                                                                                                            |
|---------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>NAME:</b>        | X-linked centronuclear myopathy                                                                                                                                                            |
| <b>DESCRIPTION:</b> | A rare X-linked congenital myopathy characterized by numerous centrally placed nuclei on muscle biopsy and that presents at birth with marked weakness, hypotonia and respiratory failure. |
| <b>ORPHACODE:</b>   | 596                                                                                                                                                                                        |
| <b>SYNONYMS:</b>    | X-linked myotubular myopathy<br>XLCNM<br>XLMTM                                                                                                                                             |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>ICD-10</u><br><u>MeSH</u><br><u>OMIM</u>                                                                                                                             |
| <b>ANALYTE(S):</b>  | <u>MTM1</u>                                                                                                                                                                                |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                        |
| <b>CHANGED:</b>     | 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 artrogryposis (gene panel)

### Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

### Related Analytes

- myotubularin 1

### Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB

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