

**GENETIC TEST:**  
**Congenital myotonia (Becker-Thomsen disease) (CLCN1 gene)**

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| <b>FULL NAME:</b>                 | Congenital myotonia (Becker-Thomsen disease) (CLCN1 gene)                                                                          |
| <b>TEST TYPE:</b>                 | Clinical                                                                                                                           |
| <b>TEST SPECIALTY:</b>            | Molecular Genetics                                                                                                                 |
| <b>TEST PURPOSE:</b>              | Carrier diagnosis,<br>Mutation confirmation,<br>Post-natal Diagnosis,<br>Pre-implantation genetic diagnosis,<br>Prenatal diagnosis |
| <b>SPECIMEN:</b>                  | Peripheral (whole) blood on EDTA,<br>Amniotic fluid,<br>Chorionic villi,<br>Cell culture                                           |
| <b>METHOD CATEGORY:</b>           | Sequence analysis: entire coding region<br>Mutation screening and sequence analysis of selected exons                              |
| <b>METHOD TECHNIQUE:</b>          | Bi-directional Sanger Sequence analysis                                                                                            |
| <b>RIZIV CODE:</b>                | 565471-565482                                                                                                                      |
| <b>ACCREDITATION (ISO 15189):</b> | 2021-10-07 / 2026-06-14                                                                                                            |

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| <b>EQA:</b>                       | <ul style="list-style-type: none"> <li>• DNA Sequencing - Sanger,</li> <li>• DNA Sequencing - Sanger,</li> <li>• DNA Sequencing - Sanger,</li> <li>• DNA Sequencing - Sanger,</li> <li>• DNA Sequencing - Sanger,</li> <li>• DNA Sequencing - Sanger,</li> <li>• DNA Sequencing - Sanger ,</li> <li>• DNA Sequencing - Sanger</li> </ul> |
| <b>TURNAROUND TIME (MAXIMUM):</b> | 3 months (10 working days for prenatal diagnosis)                                                                                                                                                                                                                                                                                        |
| <b>CREATED:</b>                   | 23 Aug 2019 - 21:40                                                                                                                                                                                                                                                                                                                      |
| <b>CHANGED:</b>                   | 01 Mar 2023 - 16:33                                                                                                                                                                                                                                                                                                                      |
| <b>URL:</b>                       | <a href="https://laboguide.uzbrussel.be/laboguide#Analyses:Becker-Thomsen%20myotonie&amp;&amp;&amp;...">https://laboguide.uzbrussel.be/laboguide#Analyses:Becker-Thomsen%20myotonie&amp;&amp;&amp;...</a>                                                                                                                                |

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