

**GENETIC TEST:**

**Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4**

|                                   |                                                                                                         |
|-----------------------------------|---------------------------------------------------------------------------------------------------------|
| <b>FULL NAME:</b>                 | Spinal muscular atrophy (SMA) type 1 (Werdnig-Hoffmann), type 2, type 3 (Kugelberg-Welander) and type 4 |
| <b>TEST TYPE:</b>                 | Clinical                                                                                                |
| <b>TEST SPECIALTY:</b>            | Molecular Genetics                                                                                      |
| <b>TEST PURPOSE:</b>              | Carrier diagnosis,<br>Post-natal Diagnosis,<br>Prenatal diagnosis                                       |
| <b>SPECIMEN:</b>                  | Peripheral (whole) blood on EDTA,<br>Chorionic villi,<br>Amniotic fluid                                 |
| <b>METHOD CATEGORY:</b>           | Deletion/duplication analysis                                                                           |
| <b>METHOD TECHNIQUE:</b>          | MLPA based techniques                                                                                   |
| <b>RIZIV CODE:</b>                | 565456-565460                                                                                           |
| <b>ACCREDITATION (ISO 15189):</b> | 2022-12-22 / 2027-12-21                                                                                 |

|                                   |                                                                                                                                                                                         |
|-----------------------------------|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>EQA:</b>                       | <ul style="list-style-type: none"> <li>• Spinal Muscular Atrophy,</li> <li>• Spinal Muscular Atrophy,</li> <li>• Spinal Muscular Atrophy,</li> <li>• Spinal Muscular Atrophy</li> </ul> |
| <b>TURNAROUND TIME (MAXIMUM):</b> | 2 months                                                                                                                                                                                |
| <b>CREATED:</b>                   | 23 Jul 2019 - 12:08                                                                                                                                                                     |
| <b>CHANGED:</b>                   | 25 Jan 2023 - 09:17                                                                                                                                                                     |

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## RELATED CONTENT

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### Related Diseases

- [Proximal spinal muscular atrophy type 1](#)
- [Proximal spinal muscular atrophy type 2](#)
- [Proximal spinal muscular atrophy type 3](#)
- [Proximal spinal muscular atrophy type 4](#)

### Related Laboratories

- [Centre de Génétique Médicale UCL](#)

### Related Analytes

- [survival of motor neuron 1, telomeric](#)
- [survival of motor neuron 2, centromeric](#)

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