

**GENETIC TEST:****Beta-globin hemoglobinopathies, Hemoglobinopathy C (HbC) (HBB hot spot mutation - p.Glu6Lys)**

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| <b>FULL NAME:</b>        | Beta-globin hemoglobinopathies, Hemoglobinopathy C (HbC) (HBB hot spot mutation - p.Glu6Lys)                                       |
| <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>  | Targeted variant analysis                                                                                                          |
| <b>METHOD TECHNIQUE:</b> | Bi-directional Sanger Sequence analysis                                                                                            |
| <b>RIZIV CODE:</b>       | 565390-565401                                                                                                                      |

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| <b>ACCREDITATION (ISO 15189):</b> | 2021-10-07 / 2026-06-14                                                                                                                                                                                                                                                                              |
| <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> </ul> |
| <b>TURNAROUND TIME (MAXIMUM):</b> | 2 months (10 working days for prenatal diagnosis)                                                                                                                                                                                                                                                    |
| <b>CREATED:</b>                   | 23 Aug 2019 - 16:28                                                                                                                                                                                                                                                                                  |
| <b>CHANGED:</b>                   | 01 Mar 2023 - 16:28                                                                                                                                                                                                                                                                                  |
| <b>URL:</b>                       | <a href="https://laboguide.uzbrussel.be/laboguide#Analyses:Hemoglobinopathie%20S&amp;&amp;&amp;&amp;975...">https://laboguide.uzbrussel.be/laboguide#Analyses:Hemoglobinopathie%20S&amp;&amp;&amp;&amp;975...</a>                                                                                    |

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