

**STUDY ON GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD)  
VARIANTS AND ITS ASSOCIATION WITH HAEMOGLOBINOPATHIES  
AMONG THE TRIBAL POPULATION OF MALARIA ENDEMIC INDO-  
BHUTAN BORDER DISTRICTS OF BTR, ASSAM, INDIA.**



A Thesis submitted to Bodoland University for the Degree of Doctor of Philosophy  
in Biotechnology in the Faculty of Science and Technology, 2023

**By**

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## DECLARATION

I do hereby declare that the research work embodied in this thesis entitled "**Study on Glucose-6-Phosphate Dehydrogenase (G6PD) variants and its association with haemoglobinopathies among the tribal population of malaria endemic Indo-Bhutan border districts of BTR, Assam, India**" has been carried out by me under direct guidance and supervision of Prof. (Dr.) Jatin Sarmah, Department of Biotechnology, Bodoland University, Kokrajhar, Assam, India.

The work is original and has not been submitted in part or in full for any degree or diploma to any university.

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## CERTIFICATE

This is to certify that the thesis entitled “**Study on Glucose-6-Phosphate Dehydrogenase (G6PD) variants and its association with haemoglobinopathies among the tribal population of malaria endemic Indo-Bhutan border districts of BTR, Assam, India**”, which has been submitted by Miss Noymi Basumatary for the award of the degree of Doctor of Philosophy under Bodoland University is a record of original research works carried out by her under my direct guidance and supervision. She has fulfilled all the requirements for submitting the thesis.

The results embodied in the thesis have not been submitted to any other university or institution for the award of any degree or diploma.

  
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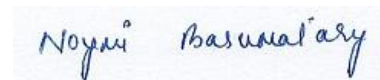
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## ABBREVIATIONS

|                   |  |
|-------------------|--|
| G6PD:             | Glucose-6-Phosphate Dehydrogenase                    |
| PPP:              | Pentose Phosphate Pathway                            |
| NADPH:            | Nicotinamide Adenine Dinucleotide Phosphate Hydrogen |
| ROS:              | Reactive Oxygen Species                              |
| GSH:              | Glutathione  |
| RBC:              | Red Blood Cell                                       |
| WHO:              | World Health Organization                            |
| <i>P. vivax</i> : | <i>Plasmodium vivax</i>                              |
| <i>P. ovale</i> : | <i>Plasmodium ovale</i>                              |
| Hb:               | Haemoglobin  |
| HbS:              | Haemoglobin S  |
| HbE :             | Haemoglobin E  |
| HbC:              | Haemoglobin C  |
| HbSS:             | Haemoglobin S homozygous                             |
| HbCC:             | Haemoglobin C homozygous                             |
| Hb AE:            | Haemoglobin E heterozygous                           |
| Glu:              | Glutamine  |
| Lys:              | Lysine   |
| Arg:              | Arginine   |
| Trp:              | Tryptophan   |
| His:              | Histidine  |
| Met:              | Methionine   |
| Thr:              | Threonine  |

|       |  |
|-------|--|
| Ala:  | Alanine                                      |
| Gly:  | Glycine                                      |
| Ile:  | Isoleucine                                   |
| Cys:  | Cystein                                      |
| Phe:  | Phenylalanine                                |
| Val:  | Valine                                       |
| Tyr:  | Tyrosine                                     |
| Ser:  | Serine                                       |
| Pro:  | Proline                                      |
| Asp:  | Aspartic acid                                |
| Asn:  | Asparagine                                   |
| NR:   | Not reported                                 |
| HbD:  | Haemoglobin D                                |
| NHM:  | National Helth Mission                       |
| HPFH: | Hereditary Persistence of Foetal Haemoglobin |
| HbQ:  | Haemoglobin Q                                |
| HbK:  | Haemoglobin K                                |
| NAC:  | N-acetyl-cysteine                            |
| LC:   | L-cysteine                                   |
| TCA:  | Tricarboxylic acid                           |
| ME1:  | Malic enzyme 1                               |
| IDH1: | Isocitrate dehydrogenase 1                   |
| CADD: | Computer-aided drug design                   |
| MD:   | Molecular dynamics                           |
| BTR:  | Bodoland Territorial Region                  |

|             |  |
|-------------|--|
| CBC:        | Complete Blood Count                       |
| WBC:        | White Blood Cell                           |
| MCV:        | Mean Corpuscular Volume                    |
| MCH:        | Mean Corpuscular Haemoglobin               |
| MCHC:       | Mean Corpuscular Haemoglobin Concentration |
| PCV:        | Packed Cell Volume                         |
| RDW:        | Red Cell Distribution Width                |
| SPSS:       | Statistical Package for Social Sciences    |
| Buffer AL:  | Lysis buffer                               |
| Buffer AW1: | Wash buffer 1                              |
| Buffer AW2: | Wash buffer 2                              |
| Buffer AE:  | Elution buffer                             |
| DF:         | Dilution factor                            |
| bp:         | Base pair                                  |
| Taq:        | <i>Thermus aquaticus</i>                   |
| PCR:        | Polymerase Chain Reaction                  |
| HaeIII:     | <i>Haemophilus aegyptus III</i>            |
| NlaIII:     | <i>Neisseria lactamica III</i>             |
| FokI:       | <i>Flavobacterium okeanokoites I</i>       |
| HindIII:    | <i>Haemophilus influenza III</i>           |
| MboII:      | <i>Moraxella bovis II</i>                  |
| BstUI:      | <i>Bacillus stearothermophilus I</i>       |
| BstXI:      | <i>Bacillus stearothermophilus X I</i>     |
| HhaI:       | <i>Haemophilus haemolyticus I</i>          |
| AflII:      | <i>Anabaena flosaquae II</i>               |



|                    |  |
|--------------------|--|
| NdeI:              | <i>Neisseria denitrificans I</i>                                       |
| RCSB PDB:          | Research Collaboratory for Structural Bioinformatics Protein Data Bank |
| ADT:               | AutoDock Tool  |
| MS:                | Molecular Surface  |
| ADMET:             | Absorption, Distribution, Metabolism, Excretion and Toxicity           |
| GROMACS:           | GRONingen Machine for Chemical Simulations                             |
| GROMOS:            | GRONingen Molecular Simulation   |
| SPCE:              | Simulation Program with Integrated Circuit Emphasis                    |
| NaCl:              | Sodium Chloride  |
| NTP:               | Number of particles, pressure and temperature                          |
| RMSD:              | Root Mean Square Deviation   |
| RMSF:              | Root Mean Square Fluctuation   |
| RG:                | Radius of Gyration   |
| SASA:              | Solvent Accessibility Surface Area                                     |
| HbA <sub>0</sub> : | Adult haemoglobin  |
| HbA <sub>2</sub> : | Smaller component of adult haemoglobin                                 |
| SD:                | Standard deviation   |
| TLC:               | Total leucocyte count  |
| LYM:               | Lymphocytes  |
| A <sub>260</sub> : | Absorbance at 260nm  |
| A <sub>280</sub> : | Absorbance at 280nm  |
| 3D:                | Three dimensional  |
| 2D:                | Two dimensional  |
| MW:                | Molecular weight   |
| TPSA:              | Topological polar surface area   |

|        |   |
|--------|---|
| HbA:   | Hydrogen bond acceptor                    |
| HbD:   | Hydrogen bond donor                       |
| LD50:  | Lethal dose                               |
| CNSHA: | Chronic non-spherocytic hemolytic anaemia |
| Ki:    | Inhibition constant                       |
| Km:    | Michaelis constant                        |

## STANDARD UNITS

|                   |                              |
|-------------------|------------------------------|
| km <sup>2</sup> : | Square kilometre             |
| mg:               | Milligram                    |
| kg:               | Kilogram                     |
| µl:               | Microlitre                   |
| U/g:              | Units per gram               |
| g/dL:             | Gram per decilitre           |
| ml:               | Millilitre                   |
| Million/Cumm:     | Million per cubic millimetre |
| pg:               | Picogram                     |
| Cells/Cumm :      | Cells per cubic millimeter   |
| fL:               | Femtolitre                   |
| Å:                | Angstrom                     |
| mM:               | Millimolar                   |
| nm:               | Nanomolar                    |
| µg/ml:            | Microgram per millilitre     |
| kcal/mol:         | Kilocalorie per mole         |
| Da:               | Dalton                       |
| nm:               | Nanometre                    |