

**OVERALL AIM: To understand the genomic diversities of the human Beta-globin gene (HBB) and defective Mannose Binding Lectin gene (MBL2 gene) to improve the diagnosis, management, and treatment of Sickle Cell Disease using genomic biomarkers and their related variants**

| Inputs                                                                                                                                                                                                                                                              | Activities                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           | Outputs                                                                                                                                                                                                                                                                                                                                                                                                                                                   | Outcomes                                                                                                                                                                                                                                                                                                              | Impact                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
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| <ul style="list-style-type: none"> <li>Professional expertise and services</li> <li>Study participants</li> <li>Genomic science technologies (Mass Array, PCR, Agarose Gel electrophoresis, bioinformatics platforms)</li> <li>Sample storage facilities</li> </ul> | <ul style="list-style-type: none"> <li>Recruit patients</li> <li>Collect, store, and transport blood samples</li> <li>Engage laboratories for genetic testing</li> <li>Conduct FBC, HPLC, and ELISA testing</li> <li>Extract DNA from patient samples</li> <li>Conduct haplotyping and genotyping on DNA samples</li> <li>Conduct bioinformatics analysis and interpretation of the results</li> <li>Identify the presence of Beta-globin gene haplotypes and MBL2 gene Single Nucleotide Polymorphisms (SNPs)</li> <li>Analyze demographic, clinical and laboratory data</li> </ul> | <ul style="list-style-type: none"> <li>Database with demographic and clinical patient data</li> <li>Collaborative working relationship with in-region laboratory</li> <li>Characterization of genetic heterogeneity and diversity in SCD patients</li> <li>Identification of genetic modifiers influencing susceptibility to bacteremia/infection in SCD patients</li> <li>Creation of a biobank for the collected samples for future research</li> </ul> | <ul style="list-style-type: none"> <li>Discovery of single-nucleotide polymorphisms (SNPs) and haplotypes in HBB gene and MBL2 gene linked to SCD outcomes</li> <li>Improved understanding of genomic disease correlation</li> <li>Availability of validated datasets for future research in pediatric SCD</li> </ul> | <p><b>Improved knowledge of SCD pathophysiology, diagnosis, management, and treatment</b></p> <ul style="list-style-type: none"> <li>Generation of benchmark data to strengthen GWAS in African populations</li> <li>Advanced clinical insights to explore genomic mediated care</li> </ul> <p><b>Improved quality of care for patients with SCD</b></p> <ul style="list-style-type: none"> <li>Improved precision and accuracy of diagnosis/prognosis of genetic diseases</li> <li>Improved risk assessment and clinical management of bacteremia in SCD through integration of genetic and laboratory parameters</li> <li>Improved resource utilization for diagnosis, treatment, and management of SCD</li> </ul> <p><b>Improved disease outcomes for patients with SCD</b></p> <p><b>Improved equity in global genomic research through inclusion of underrepresented populations</b></p> |