Microarray Technique for Studying Genetic Diversity in Saudi Sickle Cell Patients

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ABSTRACT

Introduction: Sickle cell anemia is one of the most common heritable hematologic diseases affecting humans. Approximately 3 million individuals had genetic blood diseases in Saudi Arabia, of whom 30% lived in Dammam Region. The aim of this study was to conduct complete gene survey studies using microarray technology.

Material and Methods: Blood samples from 90 unrelated sickle cell disease patients were obtained from the KKUH, Riyadh between from January 2017 and to June 2020. In this study, linkage disequilibrium has been determined between single nucleotides polymorphism loci in the same region of beta globin gene to identify which of them had a role for the unique variable appearance of the disease affect. To achieve such goal, the Haploview program was used.

Results: The obtained results revealed the region from 5246694 to 5251625 which contains 9323 bases, showing three single-nucleotide polymorphism (SNPs) in the beta globin gene region in chromosome number 11, besides the haplotypes that were appeared in the samples under investigation. This study also showed a significant correlation between SNP2 - SNP3 and between SNP 1 - SNP3, and a negative correlation between SNP1 - SNP2.

Conclusion: This study has used genome-wide association study (GWAS) in understanding the genetic diversity that explains the phenotypic shape of sickle cell disease (SCD) patients in Saudi Arabia. It is therefore important to conduct further studies at a large level in Saudi Arabia to confirm these important results, which will increase current understanding of the SCD's nature.

Keywords: Genetic origin, Mutation, Sickle cell disease, Single nucleotide, Single phenotype

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