Implication of HMOX1 and CCR5 Genotypes on Clinical Phenotype of Egyptian Patients with Sickle Cell Anemia

Abstract:

Background: Sickle cell disease (SCD) is a relatively common inherited hemolytic anemia among individuals of African descent. Genetic factors might clarify clinical diversity of the disease and variations in treatment response. Some researchers investigated hemeoxygenase1 (HMOX1) or chemokine receptor 5 (CCR5Δ32) genotypes among SCD patients and their correlation with fetal hemoglobin (HbF) and disease severity. However, there is no such records among Arab nations. ***Aim:*** We aimed to estimate the prevalence of the HMOX1-413 A>T (rs2071746) and CCR5Δ32 (rs333) polymorphisms, and to assess their effect on SCD phenotype and HbF level among Egyptian patients. ***Material and Methods:*** Polymerase chain reaction assay was used to determine these polymorphisms among 100 SCD patients and 100 healthy controls. ***Results:*** Though not statistically significant, the frequency of individual carrying HMOX-1 polymorphic; AT and TT genotypes in both patient and control groups were (92% and 85% respectively). Regarding CCR5Δ32 polymorphisms, all SCD patients harbored the wild genotype (100%), while the heteromutant genotype was encountered in 2% of our controls. Patients harboring mutant HMOX-1 had less frequent vasoocclusive crisis (VOC)/ lifetime, less VOC in the last year, less incidence of stroke, less frequency of hospitalization, and responded more frequently to hydroxyurea with statistically significant differences (p=0.028, 0.007, 0.046, 0.007, and 0.011 respectively). No significant associations with HbF level or other hematologic parameters were encountered among our cohort. ***Conclusion:*** Our study results suggest a protective effect of mutant HMOX-1 genotypes in ameliorating phenotypic severity of disease. HMOX1-413 A>T (rs2071746) polymorphisms might prove to be a prognostic marker among Egyptian SCD, but not CCR5Δ32 (rs333) polymorphisms.

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