

SHC 21. EFFECT OF IRP 1 AND IRP 2 GENE POLYMORPHISMS ON BLOOD IRON LEVELS OF THALASSEMIA PATIENTS IN TURKISH POPULATION

Yasemin KARTAL, Zeliha KAYAALTI, Dilek Kaya-AKYÜZLÜ, Fezile ÖZDEMİR, Tülin SÖYLEMEZOĞLU

Department of Physiology, Hacettepe University, Faculty of Medicine, Turkey

Department of Forensic Chemistry and Forensic Toxicology, Ankara University, Institute of Forensic Sciences, Turkey

In thalassemia, repeated blood transfusions, cell destruction and increased absorption of iron from intestine lead to iron accumulation which causes toxicity. The aim of this study was to determine the effect of iron-regulatory protein 1 (IRP1) and iron-regulatory protein 2 (IRP2) gene polymorphism on blood iron levels in 100 thalassemia patients in Turkish population.

The study population comprised 100 genetically unrelated thalassemia patients (50 female and 50 male). Firstly, the genotypes of G>A-IRP1 (rs867469) polymorphism were determined by the Allele-Specific Oligonucleotide-Polymerase Chain Reaction method. The genotypes of G>A-IRP2 (rs17483548) were investigated by Polymerase Chain Reaction-Restriction Fragment Length Polymorphism method. Secondly, blood iron concentrations were measured by Flame Atomic Absorption Spectroscopy.

The genotype frequencies of IRP1 were found as 54% homozygote typical (GG), 34% heterozygote (GA) and 12% homozygote atypical (AA) and the genotype frequencies of IRP2 were found as 42% homozygote typical (GG), 46% heterozygote (GA) and 12% homozygote atypical (AA). Although individuals with the GG genotype of IRP1 gene polymorphism had higher blood iron level ($300.42 \pm 61.06 \mu\text{g/L}$) than those with GA ($284.31 \pm 48.23 \mu\text{g/L}$) and AA ($280.85 \pm 62.64 \mu\text{g/L}$) genotypes, according to the statistical analysis the results were not statistically significant ($p > 0.05$). Although individuals with the GG genotype of IRP2 gene polymorphism had also higher blood iron level ($298.35 \pm 42.30 \mu\text{g/L}$) than those with GA ($289.47 \pm 64.83 \mu\text{g/L}$) and AA ($284.44 \pm 73.69 \mu\text{g/L}$) genotypes, according to the statistical analysis the results were not statistically significant ($p > 0.05$). Further studies with larger study groups and different populations can elucidate the influence of this polymorphism on thalassemia

This study was supported by the Scientific and Technical Research Council of Turkey (TUBITAK), Project number 114S312.

* yagmuryaseminkartal@gmail.com

Corresponding Author: zkayaalti@gmail.com