Genotype and allele frequencies of *MDR1* gene C1236T polymorphism in a Turkish population

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**ABSTRACT.** Human P-glycoprotein (P-gp) is encoded by the *MDR1* gene, which is located on chromosomal region 7q21 and consists of 28 exons. To date, over 50 single nucleotide polymorphisms (SNPs) have been reported for the *MDR1* gene. The effect of these polymorphisms on P-gp function or their clinical impact is in most cases unknown, but some of the SNPs are known to be of functional relevance and can also alter the pharmacokinetics of substrate drugs. The aim of the current study was to analyze for the first time an existing silent *MDR1* C1236T (Gly412Gly) polymorphism in a Turkish population. The genotype frequencies of C1236T SNP in a Turkish population were also compared with those in other populations. One hundred unrelated healthy subjects (48 females, 52 males) were included in this study and all them were of Turkish ethnicity. The genotyping of the C1236T SNP was performed by the polymerase chain reaction (PCR)-restriction fragment length polymorphism (RFLP) method. The frequencies of the wild-type C and mutant T alleles were 45.5 and 54.5%, respectively. The distribution of C1236T genotype frequencies in our study group was found to be similar to that in Czech, Polish, Portuguese, Russian, Malay, and Japanese populations and different from that in French, German, Chinese, and Indian populations. The distributions of CC, CT, and TT genotypes were 20.0, 51.0, and 29.0%, respectively. Our study provides a framework for future studies concerning the role of polymorphic variants of *MDR1* gene.
in the genesis of various diseases or in designing future pharmacogenetic and pharmacokinetic studies conducted with P-gp substrates in the Turkish population.

**Key words:** *MDRI*; C1236T; Polymorphism; Turkish population