

Pharmacogenetic Analysis of Inter-Ethnic Variability in the Uptake Transporter SLCO1B1 Gene in Colombian, Mozambican, and Portuguese Populations

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Abstract : There is no epidemiologic data on this gene polymorphism in several countries. Therefore, this study aimed to assess the genotype and allele frequencies of the gene variant in three countries. This study involved healthy individuals from Colombia, Mozambique, and Portugal. Genomic DNA was isolated from blood samples using the Qiamp DNA Extraction Kit (Qiagen). The isolated DNA was genotyped using Polymerase Chain Reaction (PCR) - Restriction Fragment Length Polymorphism. Microstat and GraphPad quick cal software were used for the Chi-square test and evaluation of Hardy-Weinberg equilibrium, respectively. A total of 181 individuals' blood sample was analyzed. Overall, TT (74.0%) genotype was the highest, and CC (7.8%) was the lowest. Country wise genotypic frequencies were Colombia 47(70.2%) TT, 12(17.9%) TC and 8(11.9%) CC; Mozambique 47(88.7%) TT, 5(9.4%) TC, and 1(1.9%) CC; and Portugal 40(65.6%) TT, 16(26.2%) TC, and 5(8.2%) CC. The reference (T) allele was highest among Mozambicans (93.4%) compared to Colombians (79.1%) and Portuguese (78.7%). Mozambicans showed statistically significant genotypic and allelic frequency differences compared to Colombians ($p < 0.01$) and Portuguese ($p < 0.01$). Overall and country-wise, the CC genotype was less frequent and relatively high for Colombians and Portuguese populations. This finding may imply statins risk-benefit variability associated with CC genotype among these populations that needs further understanding.

Keywords : c.521T>C, polymorphism, SLCO1B1, SNP, statins

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