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Genetic Polymorphism of Cytochrome P450 2D6*4 and 2D6*5 in an Adult Population Sample from Costa Rica

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ABSTRACT Currently, there is a defined group of drugs, previously established by the Food and Drug Administration (FDA), that requires pharmacogenomic tests, due to possible polymorphisms in the cytochromes that metabolizes them, leading to a potential modification of their pharmacokinetical and/or pharmacodynamical properties, and therefore their dosage. The main objective of this study was to determine the frequency of two single-nucleotide polymorphism (SNP) for CYP2D6 cytochrome, related to a poor metabolizer phenotype: CYP2D6*4 and CYP2D6*5 in a group of adults from Costa Rica. CYP2D6*4 allele was determined by PCR RFLP methodology and CYP2D6*5 allele by multiplex PCR in 389 adult blood samples from Costa Rica. The allelic frequency determined for CYP2D6*4 was of 12.2 percent, and of 2.8 percent for CYP2D6*5. There were 23 subjects with a homozygote polymorphic genotype (4*/4*) and 3 with a double heterozygote mutation (wt/*4 and wt/*5); that corresponds to a 6.7 percent of the sample. The relevance of this finding is due to the fact that a slower metabolism for selective substrates has been demonstrated in subjects from Costa Rican origin, due to SNP in alleles for CYP2D6; nevertheless, other CYP2D6 polymorphisms implied in this phenotype should be analyzed, in order to give a definitive characterization.