Abstract

CYP2D6 differences have already been demonstrated within Latin American populations by the CEIBA.FP Consortium of the Ibero-American Network of Pharmacogenetics (RIBEF, as per the acronym in Spanish). However, within the population of Costa Rica, no research has been conducted until now, even though this population has a trihybrid component ancestry that represents an interesting condition. Thus, the present study was aimed to determine the frequency of Ultra-rapid Metabolizers (UMs) and Poor Metabolizers (PMs) in a Costa Rican population, as well as to determine whether there are differences in the CYP2D6-predicted phenotype frequencies among three Costa Rican groups with different ethnic backgrounds. Additionally, these frequencies of PMs and UMs obtained were compared with Ibero-American populations published data. Finally, we also aimed to describe allele frequencies among different Costa Rican ethnic groups. This research has been undertaken within the framework of the RIBEF CEIBA Consortium studies on Latin American populations. A total of 385 individuals were included in the study: 139 mestizos, 197 Amerindians, and 49 Afro-Caribbeans. CYP2D6 genotypes were determined by XL-PCR and Real-Time PCR. The CYP2D6 variant alleles *2, *3, *4, *5, *6, *10, *17, *29, *35, and *41 were also determined. For the entire Costa Rican population, the frequency of PMs and UMs was 6% and 6.5%, respectively. The percentage of UMs in the mestizo population was higher than in the Amerindian population. CYP2D6 UMs vary from 3.6% to 10.1% and PMs from 1.4% to 10.2% among three Costa Rican groups. The highest frequencies of UMs (10.1%) and PMs (10.2%) were found in the mestizo and Amerindian populations, respectively. In conclusion, the frequencies of UMs and PMs for CYP2D6 varied widely across the mestizo, Amerindian and Afro-Caribbean Costa Rican populations. Future research in this population should be oriented to identify new CYP2D6 variants through sequencing methods, as well as to determine CYP2D6 phenotype, in order to establish the phenotype-genotype relation. Finally, further studies involving genetic markers of ancestry are needed in the Costa Rican population. Rev. Biol. Trop. 62 (4): 1659-1671. Epub 2014 December 01.

Keywords

CYP2D6, Costa Rica, Amerindian, Afro-Caribbean, mestizo, populations, Poor Metabolizers, Ultra-rapid Metabolizers.