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**The clinical importance of genetic diversity of the  $\beta$ 2-adrenergic receptor gene assessed by mapping of the encoding region and by single-based polymorphisms present in a Brazilian population**

Luiz Werber-Bandeira and Aldaberto Santos

General Hospital-Santa Casa da Misericórdia, Brazil

**Background:** The gene encoding the adrenergic  $\beta$ 2 receptor (ADR $\beta$ 2) located on chromosome 5q31-32 presents a high allelic diversity characterized by the presence of several single nucleotide polymorphisms (SNPs) associated with different receptor activities at the cellular levels. 45 SNPs have been identified along this gene, some with significant effect on clinical response to  $\beta$ 2-agonists. The allelic distribution varies with ethnicity and due to alterations in sensitization and down regulation of these receptors. The existing studies refer to ethnically homogeneous populations, with no data on the distribution of these SNPs in ethnically mixed populations, although certain haplotypes appear to directly affect receptor function.

**Objective:** The objective of this study was to determine the prevalence of previously described SNPs in the coding region of the ADR $\beta$ 2 gene, as well as to identify new SNPs in a group of individuals living in Rio de Janeiro through sequencing technique.

**Patients & Methods:** DNA samples from 162 individuals were subjected to PCR amplification and genotyping by sequencing.

**Results:** Analysis of the sequences generated identified the presence of nine different SNPs 46 (G>A), 66 (C>T), 79 (C>G), 252 (G>A), 455 (T>G), 470 (T>G), 491 C>T, 523 (C>A) e 579 (C>T) within the sequenced region (899 bp), from which, three: 455 (T>G), 470 (T>G), and 579 (C>T) were not yet described, and the characterization of 14 different haplotypes. The SNPs in codons 27 and 164, two of the most studied, showed different frequencies from those observed in other populations. The SNP in codon 27 was: 8.1% versus 24% in Caucasian-Americans and versus 18.7% in African-Americans. The SNP in codon 164 was: 4.6% versus 1% in Caucasian-Americans and versus 2% African-Americans. The SNP at codon 16, Arg16Gly, showed similar frequency to the one found in the literature in Caucasians and Chinese ethnicity: 54.9% vs. 39%, and 42% and 51% respectively.

**Conclusion:** The results of this study, although represent a partial mapping of the coding region of the ADR $\beta$ 2 gene, demonstrated the diversity of this gene, of great clinical importance. The results represent the first data on the partial mapping of the gene ADR $\beta$ 2 in an ethnically mixed population.

**Biography**

Luiz Werber-Bandeira is the Head of Clinical and Experimental Immunology Unit - Santa Casa de Misericórdia do Rio de Janeiro, Brazil. He has a degree in Medicine; completed his Post-doctorate in Immuno-Genetics and; PhD in Medicine-Immunology-Dermatology at Federal University of Rio de Janeiro. He is also specialized in Clinical Immunology-Allergy at Federal University of Rio de Janeiro. He is Reorganizer of the Clinical and Experimental Immunology Unit - Santa Casa da Misericórdia, Rio de Janeiro.

werberbandeira@globo.com

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