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HLA alleles and cutaneous hypersensitivity to carbamazepine

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The Human Leucocyte Antigen (HLA) genes are involved in susceptibility to inflammatory, infectious and autoimmune diseases. HLA genes are relevant in pharmacogenetics, as several of their alleles are associated with hypersensitivity to specific drugs, such as carbamazepine (CBZ) in patients of Asian ancestry. Despite the importance of identifying and relating HLA alleles to clinical conditions, there are very few databases dedicated to characterizing HLA alleles in different populations. This is partly due to the high costs of determining HLA genotypes. In this context, we aim to determine the distribution of HLA subtypes in the Brazilian population and in patients with cutaneous hypersensitivity to CBZ. The HLA genotyping was performed using the Trusight HLA v2 Sequencing Panel (Illumina), which provides an assay to obtain ultrahigh resolution sequencing of 11 HLA Loci. DNA libraries are loaded onto a MiSeq Sequencer (Illumina) and data is analyzed with the TruSight HLA Assign 2.0 software. Our preliminary results of 26 control subjects shows some frequent alleles: A*02:01:01 (37%), B*51:01:01 (25%), C*04:01:01 (24%), DPA1*01:03:01 (74%), DQA1*01:02:01 (20%), DQB1*02:01:01 (19%), DRB1*03:01:01 (23%), DRB3*02:02:01 (50%) and DRB4*01:03:01 (81%). Currently, there is great interest in the study of cutaneous drug reaction to CBZ, since it is a widely used medication for the treatment of epilepsy and other neurological conditions. Furthermore, our data will be compared to that generated by high-density SNP-panels in the same sample and other population, helping to establish a high-resolution map of the HLA region in the Brazilian population, which will be publicly available at www.bipmed.org. Supported by: FAPESP.

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