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Molecular analysis of polimorphisms in *HLA-CLASS II DRB1** and *IL7Rα* possibly associated with multiple sclerosis susceptibility in a population sample of Rio de Janeiro

Análise molecular de polimorfismos nos genes HLA-ClASSE II DRB1* e IL7Rα possivelmente associados com a susceptibilidade à esclerose múltipla em uma amostra da população do Rio de Janeiro

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ABSTRACT

Multiple sclerosis (MS) is an inflammatory and degenerative disease of the central nervous system (CNS) that affects mainly young adults. MS seems to be a polygenic and multifactorial disease, and genetic susceptibility has been associated mainly with the major histocompatibility complex (MHC), which in humans is the human leukocyte antigen (HLA). Among non-HLA genes is the alpha chain of interleukin 7 receptor gene (*IL7R**α***) at the 5p12-14 locus, also known as CD127. The aim of this study was to evaluate the correlations between polymorphism in the *IL7R**α*** (*rs6897932C*) gene, *HLA-class II DRB1** haplotypes and susceptibility to multiple sclerosis in patients with Recurrent Remitting form (RRMS). **METHOD:** In this study, peripheral blood samples were taken from 50 patients diagnosed using the diagnostic criteria for MS according to Polman (MacDonald) et al (2011). The patients were monitored at the Clinic of Neurology, Hospital Universitário Clementino Fraga Filho, along with 100 healthy control subjects matched for ancestry, sex and age. After DNA extraction by organic method, polymorphism *+244 *C* (*rs6897932*) was assessed by PCR followed by capillary electrophoresis on the ABI PRISM® 3500 Genetic Analyzer (Applied Biosystems, USA) platform. **RESULTS:** The results indicated a significant association between the CC haplotype and RRMS (p=0.02 , OR=2.14), as well as an association between the **C* allele (*CC* and *CT*) and RRMS (p=0.042, OR=2.15). The same C allele was more frequent in the sample, both in patients (0.82), and in the control group (0.71). The sample, control group and patients included, was in Hardy- Weinberg equilibrium. The correlation between the presence of the *CC* genotype and *HLA-DRB1* 15:01* was significant (OR=3.6, p=0.034). **CONCLUSION:** These results reinforce the polygenic/multifactorial characteristic or genetic heterogeneity of MS, indicating a relationship between putative polymorphism *+244*C* (*CC* genotype) in the *IL7R**α*** gene and susceptibility to MS in the sample.

Keywords: multiple sclerosis, genetic, IL7RA, HLA-DRB1* 15:01, EMRR.