Beta-Defensin 1 Genetic Polymorphisms and Atopic Dermatitis

Addobbati, C.J.C.[1], Brandão, L.A.C[1], Guimarães, R.L.[1], Rocha, C.R.C[1], Arraes, L.C.[1], Crovella, S.[1], Lima-Filho J.L[1].

[1]Laboratório de Imunopatologia Keizo Asami, UFPE

Atopic Dermatitis (AD) is a chronic inflammatory disease of skin related to environmental and immunogenetic factors. Unfortunately, AD patients are more susceptible to infections, principally due to the continuously scratching of skin allowing the entrance of allergens and pathogens which aggravate the disease. The skin produces antimicrobial peptides, such as the beta-defensin 1 (hbd1), capable to destroy pathogens by disrupting their membrane. DEFB1 gene, which encodes hbd1, can present functional single nucleotide polymorphisms (SNPs) at 5'UTR previously described to be associated with AD in others population. In order to check the role of a SNP at 5'UTR (-44 position; C→G) of DEFB1 gene in pathogenesis of AD and their future complications this work was performed. We enrolled 93 Brazilians children with AD from IMIP and 115 healthy subjects as control group. Genotyping was performed using Real Time PCR trough allele specific primers targeting -44 (C→G) alleles at 5'UTR of *DEFB1.* Statistical analyses were performed using Fisher exact Test. The G allele was more frequent among AD carriers (28% VS 14%; *P-value*=0.001, OR=2.25; 95% CI = 1.34 a 3.81). G/G genotype was predominant among the patients (10% VS 2%; *p-value*=0.0043). However, no association was found between severity of the disease and the polymorphism on *DEFB1.* Thus, our findings also ratify the role of DEFB1 polymorphisms on AD pathogenesis. The investigation of genetic factors could help the comprehension of physiopathology of AD, making possible the development of more accurate diagnostic tests and new treatment alternatives.

Key words: *Atopic Dermatitis, Innate Immunity, DEFB1, SNP, Real Time PCR.*

Supported by: FACEPE, UFPE-LIKA, CNPq, IMIP.