Medical or Research Professionals/Clinicians

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Topic: 1. Genomics, genetic basis of disease and antigen presentation

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HLA CLASS II IN PARAGUAYAN IMMUNE-MEDIATED INFLAMMATORY PATIENTS

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Background: Immunemediated inflammatory disease (IMID) is a concept used to describe a group of conditions that share common inflammatory pathways leading to systemic inflammation. The best-known genetic factor for IMID susceptibility is the human leukocyte antigen (HLA) haplotypes. Nowadays, there is a lack of information about HLA profile in Paraguayan patients with IMIDs.

Objectives: To identify HLA alleles associated with susceptibility to develop an IMID in Paraguayan patients controlled in a reference center

Methods: Paraguayan IMID patients were recruited from the Rheumatology Department of Hospital de Clínicas, Paraguay. IMID *HLA II* frequencies where compared with a control group of 50 unrelated individuals without disease and from the same geographic origin. Genotyping for HLA was performed using Luminex PCR technology. The association analysis with the IMIDs risk was performed using the chi-square allelic test

Results: 249 IMID patients (95 lupus,104 rheumatoid arthritis and 50 systemic sclerosis) where included. Of these 84,4% were women with an average age of 43,4 (±14). Comparing the haplotypes profiles for the 5 HLA class II genes between the patients and the healthy controls, in the risk association analysis, the association of the known risk allele was corroborated HLADRB1*03:01 (p =2e-06, OR:14,97). A significant association was identified between the allele HLADRB1*08:02 (p= 0.0271, OR:0,13) and HLADRB1*08:07 (p= 0.0133, OR: 0.08). In the gene *HLADQA1*, 1 allele associated with the IMIDs were found, the HLADQA1*04:01 (p=1.4e-05, OR: 0.06). In the *HLADPB1* gene 3 alleles associated with the IMIDs were identified: HLADPB1*02:01 (p= 4.2e-05, OR: 82.91), HLADPB1*03:01 (p=2e-06, OR: 14.97), HLADPB1*04:01 (p= 1.5e-05 OR: 34.55). Different associations between IMIDs and alleles was identify (Table 1)

Table 2. List of associated alleles stratified by disease

ALLELE (Systemic Lupus Erythematosus Cohort)	P-valor	OR
HLADQA1*02:01	0.0253	14
ALLELES (Rheumatoid Arthritis Cohort)	P-valor	OR
HLADRB1*08:02	0.0367	0.11
HLADPB1*02:01	0.0003	54.62
HLADPB1*03:01	0.0010	7.92
HLADPB1*04:01	0.0001	29.69
HLADQA1*02:01	0.0467	0.24
HLADQA1*04:01	0.002	0.07
HLADPA1*02:01	0.0022	0.029
ALLELES (Scleroderma Cohort)	P-valor	OR
HLADPB1*02:01	0.0003	82.33
HLADPB1*03:01	0.0094	8.44
HLADPB1*04:01	0.0001	47.50
HLADQA1*04:01	0.0013	0.06

Conclusions: In the genetic association analysis, already known associations have been replicated and new ones previously unpublished have been identified in Paraguayan IMIDs patients. This is the first genetic association study in IMID patients Paraguayan origin.

Disclosure of Interest: None declared



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CERTIFICADO

Otorgado a

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BIOMARCADORES GENÉTICOS DE SUSCEPTIBILIDAD DE PADECER LUPUS ERITEMATOSO SISTÉMICO EN PACIENTES DE ORIGEN PARAGUAYO.

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