ERAP1 and HLA-C interaction in inflammatory bowel disease in the Spanish population

Castro-Santos P.

Moro-García M.A.

Marcos-Fernández R.

Alonso-Arias R.

Díaz-Peña R.

Large genome-wide analysis studies (GWAS) and meta-analyses have dramatically increased our knowledge of the genetic risk factors of inflammatory bowel disease (IBD), identifying at least 163 loci. The endoplasmic reticulum aminopeptidase-2 (ERAP2) gene has been reported as a potential candidate gene for IBD. GWAS have also shown the potential associations between ERAP single nucleotide polymorphisms (SNP) loci and susceptibility to several autoimmune diseases, and ERAP1 and ERAP2 polymorphisms are related to HLA class I-associated diseases, including ankylosing spondylitis and Behçet's disease. Interestingly, these associations were confined to individuals carrying HLA class I-risk alleles. The aim of this study was to investigate the association of ERAP1 and ERAP2 SNPs with IBD in a Spanish population, analysing their possible interaction with specific HLA-C alleles to IBD susceptibility. A total of 367 individuals were divided into 216 IBD cases and 151 controls. SNP genotyping was performed using TaqMan® genotyping assays, whereas HLA-C typing was analysed by sequence-specific oligonucleotide probing. Herein, we report an association of the ERAP1 SNP rs30187 with the HLA-C?07 allele. The existence of shared inflammatory pathways in immunologically related diseases together with the understanding of ERAP1 function may offer clues to novel treatment strategies. © SAGE Publications.

Crohn's disease

ERAP1

HLA-C

inflammatory bowel disease

ulcerative colitis
aminopeptidase
ERAP1 protein, human
ERAP2 protein, human
HLA C antigen
minor histocompatibility antigen
antigen presentation
biological model
cytotoxicity
female
gene frequency
genetic association study
genetic predisposition
genetics
genotype
human
inflammatory bowel disease
male
molecular mimicry
risk
single nucleotide polymorphism
Spain
Aminopeptidases
Antigen Presentation
Cytotoxicity, Immunologic
Female

Gene Frequency
Genetic Association Studies
Genetic Predisposition to Disease
Genotype
HLA-C Antigens
Humans
Inflammatory Bowel Diseases
Male
Minor Histocompatibility Antigens
Models, Immunological
Molecular Mimicry
Polymorphism, Single Nucleotide
Risk
Spain