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MEETING ABSTRACT Open Access

Polymorphisms and genetic susceptibility of type 1 diabetes mellitus and celiac disease

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Background

Type 1 diabetes mellitus (T1D) is a chronic autoimmune disease characterized by pancreatic beta-cell destruction, hyperglycemia and progressive insulin deficiency, affecting mainly children and adolescents genetically predisposed. About 10% (2.4-16.4%) of T1D individuals develop celiac disease (CD), an immune-mediated enteropathy triggered by gluten exposure. Both diseases have a common autoimmune origin and share a similar genetic Background, the major histocompatibility complex class II antigen (HLA-DQ). Some genetic association studies have also identified susceptibility polymorphisms non-HLA associated to both diseases, located in different genes: RGS1 (rs2816316), IL2-IL21 (rs6822844), BACH2 (rs11755527) and IL18RAP (rs917997).

Objective

The aim of the present study was to determine the allelic and genotypic frequencies of polymorphisms in RGS1, IL2-IL21, BACH2 and IL18RAP genes in a sample of 317 T1D individuals and to compare the frequency of the risk alleles in patients with negative (N=264) or positive (N=53) serology for CD.

Materials and methods

Saliva or blood sample was collected and DNA extraction and genotyping performed by PCR Real-Time. All polymorphisms were in Hardy-Weinberg equilibrium. The comparison between the allele and genotype frequencies was calculate by Chi-square and Fisher's exact test.

Results

The frequency of the allele risk for the genes RGS1 (allele A); IL2-IL21 (allele C); BACH2 (allele C) e IL18RAP (allele T) in T1D individuals and negative serology for CD was respectively: 95.5%, 97.4%, 79.8% and 46.6%. In T1D with seropositive, the frequency of the same alleles were: 98.1%(p=0.703), 100.0% (p=0.604), 86.0% (p=0.433) and 46.2%(p=1.000). Both genotype and allele frequencies were not significantly between T1D with negative or positive serology for CD.

Conclusion

Our data did not evidence differences between the polymorphisms non-HLA analyzed in T1D with or without seropositive for CD, being not possible to assure that the presence of these polymorphism increase or decrease the predisposition to celiac disease.

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