DIABETOLOGY & METABOLIC SYNDROME

MEETING ABSTRACT Open Access

Haplotype analysis of DQ2.5 and DQ8 by simple nucleotide polymorphism technique (TAG-SNP) in type 1 diabetes and/or celiac disease patients

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From 20th Brazilian Diabetes Society Congress
Porto Alegre, Brazil. 11-18 November 2015

Background

Celiac disease (CD) is a chronic and permanent enteropathy triggered by ingestion of gluten proteins, present in wheat, rye and barley, in genetically predisposed individuals, as type 1 diabetes mellitus (T1D) patients, who have a higher prevalence of the disease compared to the general population. Both diseases have similar autoimmune origin, being associated to the histocompatibility complex class II antigen (HLA), mainly DQ2.5 and/or DQ8 haplotypes, allowing estimating negative predictive value accurately. Nevertheless, these genetic tests are expensive, making its implementation a challenge in routine clinical practice.

Objective

The aim of the study was the evaluation of the frequency of DQ2.5 and DQ8 haplotypes in T1D and CD patients, using a simple nucleotide polymorphism technique (Tag-SNP).

Materials and methods

The study enrolled 365 individuals, being 296 with T1D (without CD=265 and with CD=31) and 69 with only CD. The HLA-DQA1* 0501 and DQB1* 0201alleles of DQ2.5 and HLA-DQB1*0302 allele of DQ8 were analyzed by HLA Tag SNP and its frequency compared between T1D and CD.

Results

The presence of DQ2.5 alleles was found in 57.1% (169/296) among T1D, being significantly more frequent among T1D individuals with CD (without CD 54.3% and with CD 80.6%, p=0.006). There was no significant difference in the comparison among T1D with CD and CD individuals without diabetes (80.6% and 62.3%, p=0.054). The presence of the DQ8 allele was found 54.2% (160/295) among all T1D (without CD 51.6% and with CD 48.4%,p=0.569).The DQ8 allele was significantly higher among T1D with CD when compared to CD individuals without diabetes (72.6% and 29.5%, p<0.001).

Conclusion

Our data evidenced a higher frequency of HLA-DQB1*0302 allele of DQ8 in T1D than CD individuals without diabetes, which did not exclude CD in this group of patients. However, the analysis of HLA-DQA1* 0501 and DQB1* 0201 alleles of DQ2.5 is useful in the evaluation of the risk of CD in predisposed individuals as T1D patients.

Published: 11 November 2015

doi:10.1186/1758-5996-7-S1-A222
Cite this article as: Bastos *et al.*: Haplotype analysis of DQ2.5 and DQ8 by simple nucleotide polymorphism technique (TAG-SNP) in type 1 diabetes and/or celiac disease patients. *Diabetology & Metabolic Syndrome* 2015 **7**(Suppl 1):A222.

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