MBL2 Polymorphisms and Celiac Disease

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Celiac Disease (CD) is a complex disorder, in which genetic and environmental factors play a fundamental role. An important genetic factor is Mannose Binding Lectin (MBL) that is involved in inflammatory processes and activates complement system. MBL also participates in the removal of apoptotic bodies of the organism, a possible mechanism which evokes CD pathogenesis. Our study evaluates the association between three SNPs (Single Nucleotide Polymorphisms) in *MBL2* exon 1 (codons 52, 54 and 57) and susceptibility to CD. These SNPs are related with low levels of MBL. SNPs were genotyped by real time PCR (melting temperature assay). Our study population consists of 54 CD patients from IMIP. The control group was formed by 57 close relatives of the CD patients. Another external control group comprehends of 165 healthy individuals. The three SNPs on *MBL2* exon 1 were grouped as "0" allele and the wild allele was denominated "A" allele. We observed a higher prevalence of 0 allele in CD patients (p=0.0417, OR=1.718) compared to control group. Any association between *MBL2* allele distribution of CD patients and their relatives were found (p=0.688). Despite genetic similarities, our study suggests that other features, possibly environmental factors, may influence disease insurgence on families of CD carriers. Moreover, low levels of MBL are involved with a defective removal of apoptotic bodies on intestinal villi, ratifying the role of *MBL2* SNPs. Thus, our study highlights the importance of *MBL2*, creating new perspectives on personalized treatments.

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