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Abstract

Crohn‘s disease (CD) is ranked among inflammatory bowel diseases. The etiology of the disease is not completely understood yet. However, it is assumed that genetic predispositions may play important role in the etiology of CD. This work is a part of a project studying causality of single nucleotide polymorhisms within candidate genes for CD in the Czech population. We tested in total 333 patients with CD and 499 healthy subjects for SNPs: c.3020insC, p.Gly908Arg, p.Arg702Trp within the gene *CARD15*, g.-308G>A within the gene *TNFA*, p.Arg381Gln within the gene *IL23R* a p.Ala197Thr within the gene *ATG16L1*. For determination of the genotypes we used alellic discrimination in TaqMan sonds format. Variants c.3020insC, p.908Arg within the gene *CARD15* were significantly associated with CD (OR = 4,4; 95%CI 3,0 – 6,4, OR = 2,7; 95%CI 1,4 – 5,0, respectively). SNP p.702Trp was associated with CD after adjustation for other two polymorphisms within the gene *CARD15* (OR = 1,7; 95%CI 1,0 – 2,7). We found the protective effect of the p.381Gln in the gene *IL23R* (OR = 0,6; 95%CI 0,3 – 1,0). Variant p.197Ala within the gene *ATG16L1* increased the risk of CD (OR = 1,3; 95%CI 1,0 – 1,9). We did not detect association between g.-308A in *TNFA* gene and CD. The genotype-phenotype analysis carried only on the patients detected association of c.3020insC with early onset disease ($p = 0,001$), with ileal involvement ($p < 0,001$) and its protective effect on the colon involvement ($p < 0,001$). Variant p.381Gln has protective effect on the upper gastrointestinal involvement ($p = 0,032$). This work contributed to the knowledge of the genetic background of CD among Czech patients.