Aim of this research is to characterize genetic susceptibility to ulcerative colitis (UC), Crohn’s disease (CD) in Ukrainian cohort.

Materials and methods: 115 IBD patients (58.3% UC and 41.7% CD) and 22 healthy controls were recruited. Single-nucleotide polymorphism (SNP) of NOD2/CARD15 (3020insC, Gly908Arg), JAK2 (Val617Phe) were determined by polymerase chain reaction (PCR) with electrophoretic detection in 3% agarose gel.

Results: 72 (62.6%) IBD patients had NOD2/CARD15 single-nucleotide polymorphism. An association with CD for 3020insC was detected more often: 58.3% cases (p=0.001; OR =10.0; 95% CI=2.33-42.78), than with UC (37.3%, p=0.09). JAK2 mutations were revealed only in 11 (9.6%) patients with IBD and didn’t show any positive interaction with IBD. The interaction with SNPs and severity of IBD was revealed for CARD15 3020 insC.

Conclusions: Important role of genetic predisposition in pathogenesis of IBD was revealed. Polymorphism of NOD2/CARD15 correlated with IBD in Ukrainian cohort. More significant association with CD comparing to UC was determined for CARD15 (Gly908Arg). Patients with IBD had combined mutations of NOD2/CARD15, JAK2. Significant positive interaction between risk genes and the severity of CD and UC was determined.

**Keywords:** Inflammatory bowel diseases; NOD2/CARD15; JAK2

**Introduction**

The etiology of inflammatory bowel diseases (IBD) involves both genetic and environmental components. Genetic predisposition is the key factor for IBD [1-3]. At the same time, approximately in 50% of cases of disease incidence environmental triggers such as diet regimen and food quality, emotional stress, episodes of intestinal infections and industrial burdens are also important [2,4]. An influence of multiply combinations of environmental and non-environmental factors on health of population significantly increased in the last years. Changes of bacterial community are susceptible to different agents and can be caused by genetic and epigenetic factors. CARD15/NOD2 (nucleotide-binding oligomerization domain family, member 2) is responsible for recognizing of pathogen-associated molecular patterns play important role in innate immune host defense [2,3,5], Activation of Janus kinase-2 (JAK2) transcription pathway leads to expression of pro-inflammatory cytokines and induce immune response. JAK2 may be considered as a potential gene-candidate to IBD susceptibility. Genetic predisposition with polymorphism of NOD2/CARD15, JAK2 may have an influence on changes of gut microbiome, inducing of inflammatory process in the large and small intestine, and can play important role in pathogenesis of IBD.

**Aim**

Aim of this research is to characterize genetic susceptibility to ulcerative colitis (UC) and Crohn’s disease (CD) in Ukrainian cohort.