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Haplotype Analysis of *ABCB1* in Patients with Peptic Ulcer–Predisposition to Diseases Development

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ABSTRACT The aim of this study was to determine the significance of *ABCB1* haplotypes composed of the three most common single-nucleotide polymorphisms (c.1236C>T: rs1128503, c.2677G>T/A: rs2032582, c.3435C>T: rs1045642) in the context of peptic ulcer risk and *Helicobacter pylori* infection development in this condition. The *ABCB1* gene product P-glycoprotein is a membrane protein that functions as an ATP-dependent exporter of xenobiotics from cells. The function of the transporter is determined by the assembles of *ABCB1* polymorphism called haplotypes. Particular haplotypes have been reported as determining individual susceptibility to conditions such as inflammatory bowel diseases, colorectal cancer.202 peptic ulcer patients and 96 healthy subjects were genotyped. Genotyping was performed for c.1236C>T and c.2677G>T/A by automated sequencing and for c.3435C>T by polymerase chain reaction-restriction fragment length polymorphism method. Haplotypes and degree of linkage disequilibrium (LD) were inferred using PHASE 2.1 and EMLD software. The groups of peptic ulcer and healthy subjects differed significantly in haplotype frequencies (p=0.04). Moreover, there was a statistically significant difference in haplotype frequencies between the *H. pylori*-infected and -uninfected peptic ulcer cases (p=0.01), and between *H. pylori*-infected and -uninfected peptic ulcer men (p=0.03). In the peptic ulcer group 1236-2677 locus pair was observed to be in modest LD (D'=0.187), 1236-3435 and 2677-3435 pairs were almost in linkage equilibrium (D'=0.036 and 0.051, respectively). Haplotype structure could be responsible for individual differences in peptic ulcer predisposition and in development of *H. pylori* infection in peptic ulcer patients.