ISSN 0972-3757

International Journal of HUMAN GENETICS

© Kamla-Raj 2014 PRINT: ISSN 0972-3757 ONLINE: 2456-6360 Int J Hum Genet, 14(1): 9-15 (2014) DOI: 10.31901/24566330.2014/14.01.02

Haplotype Analysis of *TNFA* Gene in Peptic Ulcer Patients

Aleksandra Salagacka, Marta Zebrowska, Agnieszka Jelen, Marek Mirowski and Ewa Balcerczak

Laboratory of Molecular Diagnosis and Pharmacogenomics, Department of Pharmaceutical Biochemistry, Medical University of Lodz, Muszynskiego 1, 91-151 Lodz, Poland

KEYWORDS TNFA. Polymorphism. Haplotype. Helicobacter pylori. Peptic Ulcer

ABSTRACT The *TNFA* gene product TNF- α is a cytokine promoting an immune response after bacterial infections. An excessive production of the cytokine is thought to be connected with *Helicobacter pylori*-induced disorders like gastritis, peptic ulcer disease (PUD), intestinal metaplasia, or even gastric cancer. The synthesis of TNF-á is controlled at transcriptional level and dependent on single nucleotide polymorphisms (SNPs) of *TNFA* promoter region. SNPs assembled in haplotype have been implicated as potential risk factors for various autoimmune and infectious diseases. The aim of this study was to determine the frequencies of *TNFA* haplotypes composed of the four common single-nucleotide polymorphisms of the gene (-1031C>T: rs1799964, -863C>A: rs1800630, -857C>T: rs1799724, -308G>A: rs1800629) in peptic ulcer patients and to assess the significance of the haplotypes as the risk factors of *H. pylori* infection development. 203 peptic ulcer patients were genotyped using polymerase chain reaction-restriction fragment length polymorphism method. Haplotypes and degree of linkage disequilibrium (LD) were inferred with PHASE 2.1 and EMLD software. There was no statistically significant difference in haplotype frequencies between the *H. pylori*-infected and -uninfected peptic ulcer cases (p=0.62). Analogous association was also absent in the subgroups: peptic ulcer woman (p=0.69), peptic ulcer men (p=0.17). The locus pairs -308_-857, -863_-1031, and -857_-1031 was found to be in very strong LD, -308_-1031 and -857 and -863 in strong LD, and -308_-863 - in modest LD. *TNFA* haplotype structure is not connected with individual differences in succeptibility to development of *H. pylori* infection in peptic ulcer patients.