MISMATCH REPAIR GENE POLYMORPHISMS IN ASSOCIATION WITH LUNG CANCER RISK IN SLOVAK POPULATION

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Objective: *hMLH1* and *hMSH2* are two of the main members of the mismatch repair genes family. Some polymorphisms of mismatch repair genes are associated with the risk of developing sporadic and hereditary tumours. In the present case-control study, we investigated promoter polymorphisms of selected mismatch repair genes: *hMLH1* (rs1800734), *hMSH2* (rs2303425) and the risk they present with regards to the lung cancer development in Slovak population.

Material and methods: The study includes 422 lung cancer cases, 511 controls for *hMLH1* gene and 486 controls for *hMSH2* gene. Polymorphisms were investigated by PCR-RFLP method. The development risk was evaluated in additive as well as in dominant and recessive genetic model. Statistical analysis was carried out using SNP & Variation Suite v7.6.11 software.

Results: The evaluation of the association between polymorphisms rs1800734 and lung cancer risk in dominant model study, showed a statistically significant increase of risk of dominant genotype among lung cancer cases (OR=1,4; 95% CI=1,08-1,82; P=0,012). These findings were further highlighted in the group of women (OR=1,99; 95% CI=1, 23-3,25; P=0,006). Our results for polymorphism rs2303425 study revealed also significant correlation of variant genotype CC (OR=2,27; 95%CI=1, 12-4, 63; P=0,024) in recessive model.

Conclusions: We conclude that the genotype of the mismatch repair genes underscore the risk of lung cancer development in Slovak population.

Key words: single nucleotide polymorphism, promoter, human mutL homolog 1, human mutS homolog 2, lung cancer risk