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Allelic variant frequency of the MALAT1 gene by rs619581 polymorphism in patients with transitional cell carcinoma of the bladder

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Objective. MALAT1 is a long non-coding RNA with over 3,500 identified polymorphisms, some of which impact the tumor metastasis and prognosis, but data on rs619581 variants in the bladder transitional cell carcinoma patients are lacking. Therefore, the aim of the study was to analyze the distribution of allele variants of the *MALAT1* gene by rs619581 polymorphism in the patients from Sumy region of Ukraine with transitional cell carcinoma of the bladder. **Methods.** Venous blood from 242 individuals was used for the study (142 patients with transitional cell carcinoma of the bladder and a control group of 100 individuals without this pathology). The studies followed the Helsinki Declaration's ethical principles, with patients providing written consent for molecular genetic research and data processing. Genotyping of patients for the rs619581 polymorphism of the *MALAT1* gene was conducted using Real-time PCR with the TaqMan assay C___1060479_10 (Catalog number: 4351379). The study results were analyzed using SPSS (version 25.0). The Pearson χ^2 test analyzed patient distribution differences, with $P \leq 0.05$ indicating significance. **Results.** The proportions of homozygotes for the major allele (A/A), heterozygotes (A/G), and homozygotes for the minor allele (G/G) in the group of patients with transitional cell carcinoma of the bladder were 130 (90.3%), 12 (8.3%), and 2 (1.4%) respectively. In the control group, the proportions were 95 (95%), 5 (5%), and 0% respectively. No significant difference in the distribution of genotypes was found between the comparison groups ($P = 0.29$; $\chi^2 = 2.473$). The distribution of alleles in bladder cancer patients: 272 (94.4%) A-alleles, 16 (5.6%) G-alleles; in controls: 195 (97.5%) A-alleles, 5 (2.5%) G-alleles. No significant differences ($P = 0.102$; $\chi^2 = 2.676$). Analysis of the genotype frequencies A/A and A/G+G/G among males demonstrated the following: in the patients with transitional cell carcinoma of the bladder, 106 (91.4%) had A/A and 10 (8.6%) had A/G+G/G; in the control group, the frequencies were 62 (93.9%) and 4 (6.1%) respectively. No significant differences were found between the comparison groups ($P = 0.55$; $\chi^2=1.212$). Genotype distribution among females: bladder cancer patients 24 (85.7%) and 4 (14.3%); healthy individuals 33 (97.1%) and 1 (2.9%). Differences were not statistically significant ($P = 0.103$; $\chi^2 = 2.665$). **Conclusions.** The distribution of genotypes and alleles by rs619581 polymorphism of the *MALAT1* gene does not significantly differ between the patients with transitional cell carcinoma of the bladder and practically healthy individuals living in the Sumy region of Ukraine. No sex-based difference in the polymorphism distribution was found. **Fundings.** SRW "Determination of the role of molecular genetic predictors in early diagnosis and prognosis of malignant tumor diseases of the urinary system", No.0123U101850.

Keywords: transitional cell carcinoma, bladder cancer, *MALAT1* gene, polymorphism.