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Incidence of fibroblast growth factor receptor 3 gene (*FGFR3*) A248C, S249C, G372C, and T375C mutations in bladder cancer

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ABSTRACT. Bladder cancer is the most frequent cancer of the urinary system. Fibroblast growth factor receptors (FGFR) belong to the tyrosine kinase family and have important roles in cell differentiation and proliferation and embryogenesis. *FGFR3* is located on chromosome 4p16.3, and missense mutations of *FGFR3* are associated with autosomal dominant human skeletal disorders and have some oncogenic effects. We examined the incidence of *FGFR3* thanatophoric dysplasia mutations located in exon 7, A248C and S249C, and in exon 10, G372C and T375C, and their correlation with clinical-pathological parameters in bladder carcinoma patients. Fifty-six paraffin-embedded specimens of transitional cell carcinoma of the urinary bladder were included in this study. Analysis of *FGFR3* thanatophoric dysplasia mutations located in exon 7, A248C and S249C, and in exon 10, G372C and T375C, was performed by PCR-

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restriction fragment length polymorphism (RFLP) analysis and DNA sequencing. *FGFR3* thanatophoric dysplasia mutations located in exon 7, A248C and S249C, and in exon 10, G372C and T375C, were detected in 33 of the 56 patients (heterozygous mutant). Among the 56 transitional cell carcinomas, missense point mutations were detected in seven of them at codon A248C, 28 of them at codon S249C, and three of them at codon T375C, similar to data from previous reports. When the results of the *FGFR3* thanatophoric dysplasia mutations located in exon 7, A248C and S249C and in exon 10, G372C and T375C, were analyzed one by one or as a group, despite the findings of previous research reports, our data suggest that these mutations are detected homogenously regardless of the tumor classification and tumor grade.

Key words: Bladder carcinoma; Fibroblast growth factor receptor 3; Mutation; PCR; DNA sequencing; Transitional cell carcinoma

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