

INVESTIGATION OF METHYLATION AND EXPRESSION STATUS OF PAX1 GENE VIA EPIGENOMIC PROFILING IN ORAL MALIGNANT LESIONS

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ABSTRACT

Introduction: Our previously study (TUBITAK-SBAG-114S497) showed hypermethylation of the *PAX1* gene in Turkish oral squamous cell carcinoma (OSCC) patients via methylation-array results. In this study, we aimed to validate its potential as an epigenetic biomarker for OSCC by analyzing the methylation status and the changes in methylation-related gene expression of the *PAX1* gene in Turkish OSCC patient and healthy groups.

Material and Methods: Expression and methylation levels of *PAX1* gene were analyzed by Quantitative Real Time-PCR and Quantitative Methylation Specific PCR methods respectively, in tissues and body-fluid samples of 30 OSCC patients and 30 healthy individuals.

Results: The methylation rates of the *PAX1* gene in matched-normal tissue, tumors, serum and saliva samples of OSCC patients were 50%, 73%, 40%, and 66%, respectively. The methylation rates of the *PAX1* gene in normal mucosa, serum and saliva samples of healthy individuals were found to be 6%, 0 and 10%, respectively. In the malignant group, 59% patients in whom methylation was observed in the promoter region of the *PAX1* gene in the tumor tissue was found to have decreased expression levels in the tumor tissues compared to the matched-normal tissues.

Conclusion: DNA hypermethylation of *PAX1* gene may play a role in oral carcinogenesis and may be used as an epigenetic biomarker.

Keywords: OSCC, Hypermethylation, Epigenetic Biomarker, *PAX1*