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## Genetic Association Study Between Vitamin D Receptor Gene and Temporomandibular disorders

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**Aim:** Temporomandibular joint internal derangement (TMJ-ID) is the imbalance of metabolic processes in the extracellular matrix (ECM) of the articular disc, which progressively degrades causing tissue breakdown. The Vitamin D receptor (VDR) gene polymorphisms have been investigated for their potential effects and functional significance on several pathological conditions particularly osteoarthritis (OA) and disc degeneration-linked pathologies. The aim of this study was to investigate the possible association of Fok1, Apa1 and Taq1 polymorphisms of VDR gene with TMJ-ID.

**Materials and methods:** The study included 49 unrelated TMJ-ID patients (31.7 ±7.9) and 70 healthy controls (28.22 ±5.9) without TMJ-ID. Additionally, TMJ-ID patients were evaluated as anterior disc displacement with reduction (ADDWR) (n=24) and anterior disc displacement without reduction (ADDWOR) (n=25). Blood samples were obtained and DNA was extracted by standard proteinase K/phenol-chloroform method. Fok1, Apa1 and Taq1 polymorphisms of VDR gene were investigated by a polymerase chain reaction (PCR) based restriction fragment length polymorphism (RFLP).

**Results:** The genotype and allele frequency distributions of Fok1/rs2228570 (C>T), Taq1/rs731236 (T>C) and Apa1/rs7975232 (A>C) did not show significant differences in TMJ-ID patients compared to the healthy group. In Fok1, carrying the TT genotype was almost 2 fold risk factor in TMJ-ID, ADDWR and ADDWOR patients compared to the healthy group (OR=1.72, OR=1.55, OR= 1.93 respectively) although not significant. In ADDWR, CT genotype was significantly different than CC genotype (OR=0.35, CI:0.12-1.02, p<0.05) as a protective factor. In Apa1, carrying the AC and CC genotype was almost 1.23-1.79 fold risk factor in TMJ-ID patients, in ADDWR and ADDWOR cases compared to the healthy group although not significant. There were no significant results in none of the groups in Taq1 polymorphism.

**Conclusion:** Our results suggest that Fok1 and Apa1 polymorphisms may be associated with TMJ-ID pathogenesis. Increasing the case and controls numbers is needed to further evaluate the genotype and allelic frequencies and risk factor ratios of VDR polymorphisms in TMJ-ID.

### Biography

Ayça Dilara YILMAZ has expertise in molecular biology and genetics. She has attended to different molecular biology departments working on hematology, pneumonia, asthma and epilepsy. She has done her master's degree in Hematodiagnosics department of Ankara University Faculty of Medicine. She has completed her Phd in Ankara University Biotechnology Institute Proteomics Department in 2012. She has been working as a researcher in the molecular biology laboratory of Dentistry Faculty since her graduate. She has been working with dentistry students and research fellows on many projects including rat models and zebra fish model. Research in periodontitis, maxillofacial genetics, PRP treatment for improving the health and wellbeing are her recent studies in the field.

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