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Genetic association study between vitamin d receptor gene and temporomandibular disorders

Ayça Dilara Yilmaz
Ankara University Faculty of Dentistry, Turkey

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 Apa1polymorphisms 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.

ayca_kasapoglu@hotmail.com