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Genetic association study between ESR1 and temporomandibular joint internal derangement

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Temporomandibular joint internal derangement (TMJ-ID) is the imbalance of metabolic processes in the extracellular matrix (ECM) of the articular disc that progressively degrades causing tissue breakdown. Estrogen receptor alpha (ESR1) is found in the intra-articular cartilage and ESR1 polymorphisms are candidates for association with the disorder. The aim of this study was to investigate the association of XbaI and PvuII polymorphisms with TMJ-ID disorder among 48 unrelated TMJ-ID patients (31.7 ±7.9) (38 female, 10 male) and 70 healthy controls (28.22 ±5.9) (33 female, 37 male) without TMJ-ID. Also, TMJ-ID patients were grouped as anterior disc displacement with reduction (ADDWR) (n=23) and anterior disc displacement without reduction ADDWOR (n=25). Blood samples were obtained and DNA was extracted by standard proteinase K/phenol-chloroform method. PvuII and XbaI polymorphisms of ESR1 gene were investigated by a polymerase chain reaction (PCR) based restriction fragment length polymorphism (RFLP). In PvuII polymorphism, TMJ-ID patients, ADDWR and ADDWOR cases carrying the Pp/pp genotype had 1.28/1.27, 1.43/1.90 and 1.18/0.81 fold risk for developing the disorder although not significant. P allele was found to be 1.33 fold risk factor in ADDWR cases (CI: 0.68-2.60, p=0.4) compared to the healthy group. In XbaI polymorphism, ADDWR cases carrying the Xx and xx genotype had 1.50 and 1.85 fold risk for developing the disorder although not significant. Carrying the x allele in ADDWR cases had a 1.33 fold risk compared to the control group (CI: 0.68-2.62, p=0.4). Female TMJ-ID patients were compared to healthy female; the difference in genotype/allelic distributions and the odds ratios were not significant for PvuII and XbaI polymorphisms. The finding that PvuII and XbaI polymorphisms has high risk for developing TMJ-ID disorder needs to be further evaluated by increasing the case and controls numbers. A polymorphism in the ESR1 gene may be associated to TMJ-ID.

Biography

Ayça Dilara Yilmaz has completed her PhD from Ankara University, Biotechnology department and Post-doctoral studies from Ankara University, Faculty of Dentistry. She is a Post-doctoral Researcher in the Molecular Biology Laboratory of the faculty.

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