OP10 EVALUATION OF MSX1 GENE AS THE COMMON CANDIDATE GENE OF NON-SYNDROMIC CONGENITAL HYPODONTIA AND CLEFT LIP AND PALATE

Ayşe Tuba Altuğ[1], Aslı Şenol[1], Özlem Özkepir[2], Serdar Ceylaner[3], Erhan Özdiler[1], [1]Department of Orthodontics, University of Ankara, Faculty of Dentistry , [2]Department of Orthodontics, Gölbaşı Oral and Dental Health Center, Ankara and [3]Intergen Genetic Diagnosis and Research Center, Ankara, Turkey

AIMS: Non-syndromic cleft lip/palate (CLP) is a congenital anomaly and all types of this anomaly are always observed on the path between the maxillary canine and lateral incisor. The maxillary lateral incisors are the most common congenitally missing teeth. Absence of those teeth is also observed unilaterally or bilaterally similar to CLP and they also are located on the same suture with oral clefting. Additionally, both anomalies are observed more frequently on the left side. Therefore; the aim of this study was to investigate if there is a common genetic pattern between the occurrence of cleft lip/palate and congenitally missing maxillary lateral incisors (CMML).

MATERIALS AND METHOD: The common candidate gene of hypodontia and cleft lip/palate MSX1, encodes a homeobox protein and is involved in multiple epithelial-mesenchymal interactions during tooth development, therefore this gene has an important role in craniofacial development. In this study, the role of MSX1 as a candidate gene for CLP and CMML was evaluated. The CLP and CMML groups were consisted of 51 and 48 subjects, respectively. Three cubic centilitre blood samples with ethylenediaminetetraacetic acid were collected and genomic DNAs were isolated. In order to screen for putative mutations, two exons of MSX1 gene as well as their exon–intron boundaries, were amplified by the PCR and analyzed with the Sanger sequencing method.

RESULTS: In both groups, the same SNP (c.*6C>T, rs 8670) which is localized in 3'untranslated region of MSX1 gene was detected. Minor allele frequency, heterozygosity, $\chi(2)$ test for Hardy-Weinberg equilibrium at c.*6C>T variation were computed. The expected wild type, heterozygous and homozygous allele frequencies of c.*6C>T variation were 65.61, 30.78 and 3.61 per cent, respectively. However, the frequencies were 47.9, 45.8 and 6.3 per cent in the CMML group and 80.4, 11.8 and 7.8 per cent in the CLP group. These frequencies were diverted from normal for both groups and the differences between the groups were statistically significant $P < 000.1$ (Chi-square test).

CONCLUSION: The existence of common polymorphisms and diversions from the normal population in the 3'untranslated region of the MSX1 gene supports the hypothesis of a possible relationship between a CLP and CMML.