



# 신인학술 수상자 논문

## Polymorphism analysis of OFC1 gene in Korean nonsyndromic cleft lip and palate individuals

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This study was performed to identify the characteristics of the the OFC1 gene (locus: chromosome 6p24.3) in Korean that is assumed for the major gene on nonsyndromic cleft lip and palate. The samples were consisted of 80 subjects: 40 nonsyndromic cleft lip and palate patients (proband, 20 males and females, mean age 14.2 years); 40 normal adults (20 males and 20 females, mean age 25.6 years). Using PCR-based assay, the OFC1 gene was amplified, sequenced, and then searched for similar protein structures. Results were as follows;

1. The OFC1 gene contains the microsatellite marker 'CA' repeats. The number of the reference's 'CA' repeats was 21 times, and formed as TA(CA)11TA(CA)10. But, in Korean, the number of tandem 'CA' repeats were varied from 17 to 30, and 'CA' repeats consisted of TA(CA)n.
2. Fourteen allelic variants were found. Distribution of the OFC1 allele was similar between patient and control group.
3. There was a replacement of the base 'T' to 'C' after 11 tandem 'CA' repeats in Korean, comparing with 'ABI linkage map 2'. Methionine amino acid of reference was changed into valine in Korean.
4. Although allelic variants were varied, however, predicted amino acid alignments only three types with a ruled repeat((CA)(14+3n), (CA)(15+3n), and (CA)(16+3n),). Of those with cleft lip and palate, 75%(30/40) were heterozygotic amino acid alignment, compared with 66%(27/40) of control group in this study.
5. In the Pedant-Pro database analysis, predictable protein structure of the OFC1 gene had at least one transmembrane region, and one non-globular region.