CHAPTER V

CONCLUSIONS

In this study, we investigated the TP63 mutations in all patients with non-syndromic hypodontia, non-syndromic orofacial clefts and syndromic hypodontia with/without orofacial clefts, by amplification of the DNA, and direct DNA sequencing. Of the 30 patients, the missense mutation at codon 227 within exon 6 of the TP63 gene (+/-c.R227P) was detected in a Thai family affected with EEC syndrome, a girl and her father. This mutation is novel. The interesting dental findings found in the affected father included congenital absence of the permanent mandibular canines, generalized microdontia, prominent marginal ridges of the permanent maxillary incisors, round-shaped permanent molars, barrel-shaped permanent maxillary central incisors, enamel hypoplasia of permanent mandibular first premolars, and extensive dental caries. The phenotype of the families with R227Q and the R227P mutations were compared. Micturition difficulties were common in those with R227Q. Both mutations appeared to cause high caries in the affected individuals. It is hoped that the results of this study will shed light on the understanding of the roles of p63 on the normal development of ectodermal derivatives and the effects of R227P mutation on tooth development.

However, TP63 mutation was not detected in the coding exons of patients with non-syndromic orofacial clefts, non-syndromic hypodontia, and other syndromic

hypodontia with/without orofacial clefts. All single nucleotide polymorphisms (SNPs) found in this study also have previously been reported.

The critique of this study was about the short working time period on the research project.