CHAPTER VI CONCLUSIONS

This study has identified the novel mutation (c.646-663Del18) and previously reported missense mutations (p.Arg156Cys and p.Ala349Thr) in Thai patients affected with XLHED. Moreover, the p.Glu164Ala mutation, which has previously been reported to contribute to XLHED, can also contribute to non-syndromic hypodontia in this study. This study found the p.Arg334His variant in three unrelated healthy female controls with the same ethnic background. Therefore, the p.Arg334His variant could be either pathogenic mutation or non-pathogenic, requiring other methods to classify. Regarding the hypothesis of this study, the author concluded that *EDA* mutations were detected in patients affected with XLHED and in patients affected with non-syndromic hypodontia in the Thai population. Furthermore, some *EDA* mutations have differential effects on the pathogenesis of XLHED and non-syndromic hypodontia.

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