CHAPTER 5
CONCLUSION

Our study found no linkage disequilibrium between the fragile X mutation and all nearby polymorphic markers (microsatellites: DXS548 and FRAXAC1; SNPs: WEX5, ATL1, rs25731, IVS10, rs25702 and rs25723) among Thai FXS patients, suggesting no founder effect of the fragile X syndrome within the Thai population. We found, however, three strong associations among common CGG repeats, haplotype (FRAXAC1-WEX5-ATL1-rs25731-IVS10-rs25702-rs25723) and AGG interspersion patterns, the CGG-29 allele with AGG configuration of 9A9A9 was associated with haplotype 17-G-G-A-T-A-A (Hap A), the CGG-30 allele with AGG configuration of 10A9A9 was associated with haplotype 18-C-A-T-C-G-C (Hap B) and the CGG-36 allele with AGG configuration of 9A9A6A9 was associated with haplotype 17-C-G-T-T-A-A (Hap C). However, these common haplotypes were not statistically significant difference between normal and FXS groups. These findings indicate that the FXS mutation in Thais almost always arise from three common haplotype backgrounds. Finally, since Hap A and Hap C were evolutional derived with differently from Hap B, we hypothesize a model of repeat instability to provide a molecular explanation for multistep repeat expansions predisposing to the fragile X syndrome.