CHAPTER 6

CONCLUSIONS

The mutation analysis of *BRCA1* gene obtained from 31 Thai breast cancer patients residing in the sounthern part of Thailand and one healthy volunteer was performed by PCR-SSCP analysis and direct sequencing. The result showed a relative low number of *BRCA1* mutation in Thai breast cancer patients. One mutation IVS+34_47delTTCTTTTCTTTTTT was identified from the patient ID 17 who has been diagnosed with breast cancer at age 56. In addition to heterozygous, including small novel deletions (IVS7+50_63delTTCTTTTTTTTTTT on the sense strand and IVS7+34_47delAAGAAAAGAAAAAA on the antisense strand) and point substitution (IVS7+38T > C) were identified in the 25 years old patient ID17's healthy daughter. However, the number of probands was small and the family selection criteria were not stringent. Hence, a much stronger family history would be necessarily for selection criteria (at least two breast cancer cases diagnosed at any age among first degree relatives).

It is essential to estimate the contribution of *BRCA1* mutations in hereditary breast cancer through the screening of a large series in Thai patients. Large population-based screening studies are needed to establish the frequency, importance and penetrance of the broad spectrum of variations in the sequence of *BRCA1* observed in both affected and unaffected women in the Thai population. Further insight into the structure and function of the *BRCA1* gene and its protein product will facilitate the development of more comprehensive mutation-detection strategies and improve the interpretation of alterations in the sequence of the gene. These studies will contribute to the assessment of the prevention program for mutation carrier including post-test counseling and developing an individualized risk management plan by physicians. This prevention program can be useful to decrease the incidence of familial breast cancer in our country.