

## CHAPTER V

### CONCLUSION

1. Optimization of single ARMS-PCR for detection of  $\beta$ -thalassemia mutations including codon 17 (A-T), codons 41/42 (-TTCT), codons 71/72 (+A), IVS I-nt 1 (G-T) and Hb E was successfully performed.
2. Multiplex ARMS-PCR of four combinations of ARMS-PCR including codon 17 (A-T) + codons 41/42 (-TTCT), codon 17 (A-T) + Hb E, codons 41/42 (-TTCT) + Hb E and codons 41/42 (-TTCT) + codons 71/72 (+A) + codon 17 (A-T) were set up to detect  $\beta$ -thalassemia mutations.
3. The lowest levels of WBC numbers that can yield amplified products enough to visualize by naked eyes after agarose gel electrophoresis was 21,000 to 33,000 cells for the single ARMS-PCR and 10,500 to 16,500 cells for multiplex ARMS-PCR.
4. The set-up ARMS-PCR is highly applicable in the  $\beta$ -thalassemia heterozygote screening in the northern Thailand.
5. The potential application of ARMS-PCR in Hb E screening was perfect.
6. The ARMS-PCR technique were successfully applicable in prenatal diagnosis of  $\beta$ -thalassemia and  $\beta$ -hemoglobinopathies.
7. The combination of ARMS-PCR technique and nucleotide sequencing in prenatal diagnosis are alternative choice for diagnosis  $\beta$ -thalassemia major and  $\beta$ -thalassemia / Hb E disease.