CHAPTER V

CONCLUSION

The multiplex PCR was applied to detect the α-thalassemia-1 gene from 114 genomic DNA samples of patients with Hb H disease. Out of these 114 samples, 113 samples were heterozygous for $--^{SEA}$ deletion and only one was heterozygous for $--^{THAI}$ deletion. None of them has the $--^{FIL}$ deletion. Furthermore, we analyzed DNA of 33 Thai infants with the Hb Bart's hydrops fetalis (--/--) from Maharaj Nakorn Chiang Mai hospital. Only two out of 33 cases were of compound heterozygotes for the $--^{THAI}$ and $--^{SEA}$ mutants ($--^{THAI}/--^{SEA}$). The remaining 31 cases were homozygous for the common $--^{SEA}$ mutants ($--^{SEA}/--^{SEA}$). None of them has the compound heterozygotes for the $--^{THAI}$ or $--^{SEA}$ and $--^{FIL}$ mutants.

Although, only three α-thalassemia-1 deletions are included in the multiplex PCR assay at present, other α-thalassemia deletions, such as α-thalassemia-2: $-\alpha^{3.7}$ and $-\alpha^{4.2}$ deletions, should be easily incorporated into the assay if required. Moreover, the development of multiplex PCR protocols for detecting all of α-thalassemia-1 deletion types that reported in Thailand is of significant clinical importance for carrier screening and prenatal diagnosis of pregnancies at risk for Hb Bart's hydrops fetalis (86) at Maharaj Nakorn Chiang Mai hospital in the future.