CHAPTER V

CONCLUSION

The prevalence of alpha-thalassemia was determined and the molecular types of alpha-thalassemia were characterized using DNA hybridization techniques in 112 subjects from rural areas of northern Thailand. The alpha-thal-2 haplotype was found in 33 samples (0.183); 29 of these were of the rightward type (0.165) and the remaining four were of the leftward type (0.018). All the rightward alpha-thal-2 mutations belonged to the subtype I. Alpha-thal-1 was found on 13 of 224 chromosomes, and all of these were of the common Southeast Asian type (0.058). Twelve cases were subtypeI and the remaining two were subtypeII. Deletions of the entire zeta alpha globin gene cluster were not detected in the present study. The Rsa I polymorphic site was present on 54 of 170 normal chromosomes and on 3 of 37 alpha-thalassemic chromosomes.The common nondeletional alpha-thal-2, Hb Constant Spring was found in 4 samples (gene frequency 0.018).

From these distribution data, the following expected frequencies of alpha-thalassemia syndromes in the rural areas of northern Thailand are calculated: Hb Bart's hydrops fetalis 0.0034 or approximately 1:290, Hb H disease 0.021 or approximately 1:48 and Hb CS-Hb H disease 0.002 or approximately 1:500. These figures indicate that the clinically severe alpha-thalassemia syndromes are of considerable public health importance in northern Thailand. That the genetic counselling by the prenatal diagnosis at DNA level is needed.