

CHAPTER V

CONCLUSION

1. The molecular bases of β -thalassemia intermedia and β -thalassemia major in Maharaj Nakorn Chiang Mai Hospital were characterized and three genetic modifying factors, including types of the β -thalassemia mutations, the α -thalassemia 1 (SEA type) and the $XmnI$ - γ polymorphism, were considered.
2. Eight β -thalassemia mutations producing both β^0 - and β^+ -thalassemia were found with a β^0 -thalassemia producing mutation, the 4-bp (-TTCT) deletion, predominated followed by the HbE gene. The presence of $XmnI$ - γ polymorphism was less frequent than the absent one. The α -thalassemia (SEA type) was the least frequent genetic modulating factor observed.
3. No consistent relationships between the three analysed genetic factors and the clinical severity in the studied β -thalassemic patients was demonstrated.
4. Other genetic and non-genetic modifying factors particularly those associated with increased HbF and F cell production are needed to be further elucidated.