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 β₀- and β⁺-thalassemia were found with a β₀-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.