2.2 Executive Summary

Grant Number: RMU4880009

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Title of the Project: Molecular basis of phenotypic variability in paediatric patients with Haemoglobin E / β-thalassaemia

Progress Report Period : Final report

Summary :

During the period of this study, our research team have established a set of extensive molecular analyses to identify several common α and β globin gene mutations in Thai population using a combination of molecular techniques including reverse dot blot hybridization, multiplex-GAP polymerase chain reaction (PCR) analyses, multiplex amplification refractory mutation system (ARMS-PCR), in order to identify more than 30 different α and β mutations in studied samples. All the setting up and test validation have been a great success and all techniques are now fully established. In addition, the PCR-restriction fragment length polymorphism (RFLP) test for analysing the important *cis*-regulatory single nucleotide polymorphism (SNP); *Xmn* I site of the $^{G}\gamma$ globin promoter was established and working well in the laboratory . We have successfully analysed 256 cases of Hb E/ β thalassaemia. The data on setting up the experiments, genotype data, clinical phenotype and association analysis are presented.