

Haplotype analysis of β -Thalassaemia major and carriers with Filipino β^0 -Deletion in Sabah, Malaysia

ABSTRACT

Objective: The Filipino β^0 -deletion has been reported as a unique mutation in East Malaysia with a severe phenotype due to the complete absence of β -globin chain synthesis. In this study, the haplotype patterns of the β -globin gene cluster were used to relate the human genetic variation to this specific β -thalassaemia mutation. **Methods:** The 376 study subjects included 219 β -thalassaemia major (β -TM) patients with homozygous Filipino β^0 -deletion and 157 carriers with heterozygous Filipino β^0 -deletion from 10 government hospitals in different regions of Sabah. Genomic DNA was isolated from whole blood using silica membrane based DNA purification protocol. Polymerase chain reaction restriction fragment length polymorphism analysis (PCR-RFLP) was conducted on five markers within the β -globin gene cluster to construct the haplotype patterns. **Results:** Four haplotypes (Haplotype I–IV) were identified with Haplotype I as the predominant haplotype with the highest frequency of 0.98, followed by Haplotype II, III and Haplotype IV with 0.02. Haplotype I was strongly linked with the Filipino β^0 -deletion among the indigenous population. **Conclusion:** Haplotype I as the predominant haplotype suggests the patients with the Filipino β^0 -deletion in Sabah have a similar origin.

Keyword : Haplotypes; β -thalassaemia major; β -thalassaemia carrier; Filipino β^0 -deletion; Sabah, Malaysia