Two Independent Genetic Origins of $\beta^+-$Thalassemia Due to -31 A to G Mutation in Thai and Japanese Populations

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ABSTRACT Haplotype associated with the -31 (A-G) $\beta^+-$thalassemia gene in seven Thai individuals were examined and compared with that described originally in Japanese. Seven polymorphic restriction sites within $\beta$-globin gene cluster were determined using allele specific polymerase chain reaction (ASPCR) methods newly developed for rapid $\beta$-globin haplotyping. A concordant result of DNA polymorphisms examined using ASPCR and conventional PCR-restriction fragment length polymorphism (PCR-RFLP) method was observed. It was found that all these seven Thai $\beta^+-$thalassemia alleles were associated with the $\beta$-globin haplotype (+ - - - - - +), which is different from that described for a Japanese subject (- + + - + + -). This indicates two independent origins. As compared to the PCR-RFLP method, $\beta$-globin haplotyping using ASPCR developed is easier, rapid, less time-consuming and requires no restriction digestion. The methods should also prove useful in population genetic study and linkage analysis of $\beta$-hemoglobinopathy.