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

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KEYWORDS Allele Specific PCR Assay. Beta-Globin Gene. β^+- Thalassemia. β^{-31A-G} Mutation. Genetic Origin. Haplotype Analysis

ABSTRACT Haplotype associated with the -31 (A-G) β^+ -thalassemia gene in seven Thai individuals were examined and compared with that described originally in Japanese. Seven polymorphic restriction sites within β-globin gene cluster were determined using allele specific polymerase chain reaction (ASPCR) methods newly developed for rapid β-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 β^+-thalassemia alleles were associated with the β-globin haplotype (+ - - - - - +), which is different from that described for a Japanese subject (- + + - + + -). This indicates two independent origins. As compared to the PCR-RFLP method, β-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 β-hemoglobinopathy.