

Specific and straightforward molecular investigation of β -thalassemia mutations in the Malaysian Malays and Chinese using direct TaqMan genotyping assays

ABSTRACT

Beta-thalassemia is a life-threatening inherited blood disorder. Rapid characterization of β -globin gene mutations is necessary because of the high frequency of Malaysian β -thalassemia carriers. A combination real-time polymerase chain reaction genotyping assay using TaqMan probes was developed to confirm β -globin gene mutations. In this study, primers and probes were designed to specifically identify 8 common β -thalassemia mutations in the Malaysian Malay and Chinese ethnic groups using the Primer Express software. "Blind tests" using DNA samples from healthy individuals and β -thalassemia patients with different genotypes were performed to determine the specificity and sensitivity of this newly designed assay. Our results showed 100% sensitivity and specificity for this novel assay. In conclusion, the TaqMan genotyping assay is a straightforward assay that allows detection of β -globin gene mutations in less than 40 min. The simplicity and reproducibility of the TaqMan genotyping assay permit its use in laboratories as a rapid and cost-effective diagnostic tool for confirmation of common β -thalassemia mutations in Malaysia.

Keyword: β -thalassemia mutations; Malaysia; Malay; Chinese; TaqMan genotyping assay