High throughput molecular confirmation of β-Thalassemia mutations using novel Taqman probes

ABSTRACT

β-Thalassemia is a public health problem where 4.5% of Malaysians are β-thalassemia carriers. The genetic disorder is caused by defects in the β-globin gene complex which lead to reduced or complete absence of β-globin chain synthesis. Five TaqMan genotyping assays were designed and developed to detect the common β-thalassemia mutations in Malaysian Malays. The assays were evaluated with 219 “blinded” DNA samples and the results showed 100% sensitivity and specificity. The in-house designed TaqMan genotyping assays were found to be cost- and time-effective for characterization of β-thalassemia mutations in the Malaysian population.

Keyword: β-thalassemia; Malaysia; Malay; Quantitative real-time PCR; TaqMan