

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