

Analysis of $\alpha 1$ and $\alpha 2$ globin genes among patients with hemoglobin Adana in Malaysia

Abstract

Hemoglobin (Hb) Adana [HBA2: c179G>A (or HBA1); p.Gly60Asp] is a non-deletional α -thalassemia variant found in Malaysia. An improvement in the molecular techniques in recent years has made identification of Hb Adana much easier. For this study, a total of 26 Hb Adana α -thalassemia intermedia and 10 Hb Adana trait blood samples were collected from patients. Common deletional and non-deletional α -thalassemia genotypes were determined using multiplex gap polymerase chain reaction (PCR) and multiplex ARMS PCR techniques. Identification of the Hb Adana location on the α -globin gene was carried out using genomic sequencing and the location of the mutation was confirmed via restriction fragment length polymorphism-PCR. Among the 36 samples, 24 (66.7%) had the $-\alpha 3.7/\alpha Cd59\alpha$ mutation, while the $-\alpha 3.7/\alpha Cd59\alpha$ mutation accounted for 2 samples (5.6%) and the remaining 10 (27.8%) samples were $\alpha/\alpha Cd59\alpha$. All 36 samples were found to have the Hb Adana mutation on the $\alpha 2$ -globin gene. The position of the α -globin gene mutation found in our cases was similar to that reported in Indonesia (16%) but not to that in Turkey (0.6%). Our results showed that the Hb Adana mutation was preferentially present in the $\alpha 2$ -globin genes in Malays compared to the other ethnicities in Malaysia. Thus, the Malays might have similar ancestry based on the similarities in the Hb Adana position.

Keyword: Hb Adana; α -thalassemia; α -globin genes; RFLP-PCR; Genotyping