Analysis of α1 and α2 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 -α3.7/αCd59α mutation, while the -α3.7/αCd59α mutation accounted for 2 samples (5.6%) and the remaining 10 (27.8%) samples were α/αCd59α. All 36 samples were found to have the Hb Adana mutation on the a2-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 a2-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