عنوان مقاله:

HBB FSC 36-37 (-T) Gene Mutation Detection in Carriers of Thalassemia Minor Using High Resolution Melting Analysis

محل انتشار:

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خلاصه مقاله:

Beta-thalassemia is one of the most common autosomal recessive disorders in the world population resulting from over 200 different mutations of HBB gene. Beta-thalassemias are caused by point mutations or, more rarely, deletions in the HBB gene leading to reduced (beta+) or absent (beta0) synthesis of the beta chains of hemoglobin (Hb). High-resolution melting of polymerase chain reaction (PCR) products can detect heterozygous and most homozygous mutations without electrophoretic or chromatographic separations. In the current study, blood samples collected from 20 individuals carrying minor thalassemia were genotyped using HRM technique. The genotype of each sample had been previously determined via the polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP)/ amplification-refractory mutation system (ARMS) or sequencing method. This study aimed to determine the specificity and sensitivity of HRM method in the diagnosis of carriers of FSC 36-37 (-T) mutation from carriers who do not have this mutation. DNA extraction from peripheral blood was performed and HRM method was used to genotype samples. The results were analyzed according to the normalized and difference plot. High-resolution melting analysis could correctly identify all carriers of FSC 36-37 (-T) from who did not have this mutation. In summary, HRM is a technique associated with high sensitivity and specificity. Therefore, HRM is an appealing technique for the identification of FSC 36-37 (-T) mutation.

کلمات کلیدی:

Beta thalassemia minor, Difference plot, Genotyping, Hemoglobin, HRM, Normalized plot