

Original Paper

Prevalence of common point mutations of alpha globin gene in Babol, Iran (2005-09)

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Abstract

Background and Objective: Alpha thalassemia is one of the most common hemoglobin disorders. Some combination of alpha globin gene mutations may cause HbH disease with severe anemia or intermediate thalassemia. genotype common deletions are routinely tested for suspicious alpha thalassemia couples but because of lack of information about the nature and frequency of point mutations and higher expenosor of sequencing, less attention was paid to them. This study was done to determine the prevalence of common point mutations of alpha globin gene in Babol, Iran.

Materials and Methods: This descriptive study was carried out on DNA of 153 adult suspected to α -thalasemia with deleted α - golobolin gene referred to genetic laboratory in Babol, Iran during 2005-09. $\alpha 1$ and $\alpha 2$ genes were amplified by using specific biotinilated primers by PCR method. PCR products were assayed using 11 specific probs corresponding to common point mutations in alpha gene (C19, IVSI (-5nt), C59, Hb constant spring, Hb Icaria, Hb seal Rock, IVSI (148), C14, poly A (-2bp), poly A2, Poly A1) and fixed on byodine C membrabe. Hybridization between the probes and PCR products was visualized after a colorimetric reaction using of conjugated streptavidin peroxidase and TMB (tetra methyle Benzidine) and H^2O^2 .

Results: The prevalence of point mutations in poly A2, 5nt, Hb constant spring and poly A1 were 28.75%, 14.38%, 7.84% and 2.61%, respectively.

Conclusion: Point mutation in alpha globin genes was detected in %53.60 out of 153 adults suspected with alpha thalassemia without common deletion mutations.

Keywords: Alpha Thalassemia, Point mutation, Reverse Dot Blot

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