Evaluation of alpha globin gene mutations among different ethnic groups in Khuzestan Province (#13)

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Introduction: Alpha thalassemia is considered as one of the most common inherited hemoglobin disorders in the world. In addition to large deletions, over 50 different point mutations have been detected in alpha thalassemia around the world, for which patients show different phenotypes with regard to genotype. The aim of this study was to evaluate the genetic frequency of alpha thalassemia in Khuzestan Province to help implement premarital and prenatal screening programs.

Materials and Methods: In this descriptive study, data from 114 people referred to genetic laboratory of Shafa Hospital in Ahvaz for prenatal diagnosis (PND) or pre-marital screening in 2012 were studied. Genomic DNA was purified by salting out and tested using ARMS PCR and Multiplex Gap.

Results: In general, 11 mutations were found in alpha gene. According to gene frequency, the most common mutant allele was α3.7 (71. 3%) followed by two-gene deletion of MED (9.7%). Other common mutations included Cd19, PA-1 and -5nt with 8.4%, 2.8% and 2% frequency, respectively. In addition, a case of extremely rare mutation in Cd21 was found. Assessment of different ethnic groups showed similar results, except for Cd19, which was the second most common mutation in Lur ethnic group.

Conclusion: The results of this study are critical for correct diagnosis of alpha thalassemia carriers, pre-marriage counseling and prenatal diagnosis. The findings of this study suggest that the distribution of mutations in alpha globin gene is rather different with other provinces as well as among the ethnic groups in this province.

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