

Abstract

Alpha thalassemia is the most common genetic abnormalities in hemoglobin synthesis which is characterized by hypochromic microcytic anemia, and presents the variable clinical phenotype from asymptomatic to lethal hemolytic anemia. Alpha thalassemia is autosomal recessive disorder and according to more than 70% of marriages in Sistan and Balouchistan are consanguineous marriages, furthermore endemic malaria region which leads to natural selection of thalassemia gene, it seems essential to research on alpha thalassemia prevalence and common mutations in this province which can provide a valuable foundation for rapid carrier thalassemia screening, genetic counseling and prenatal diagnosis. This study conducted on 3254 couples in Zahedan and Zabol were referred to the central laboratories. At first people were screened based on hematology indices (MCV<83, MCH<29 and normal HbA2), and then the common deletion mutations (single and double gene deletion $-\alpha^{3.7}$, $-\alpha^{4.2}$, $-\alpha^{20.5}$ and MED) with Gap-Multiplex-PCR method and non-deletion mutation (α^{Cd19} , IVS-I α^{-5nt}) have been determined with Amplification Mutation Refractory Systems Polymerase Chain Reaction (AMRS-PCR). Data have been analyzed by T-Test, Chi-square and ANOVA tests. The results have been shown that prevalence of alpha-thalassemia in Zabol and Zahedan cities were 14.6%. The $-\alpha^{3.7}$ mutation with 48.7% was the most mutation in this region and followed by α^{-5nt} (7.9 %), α^{Cd19} (1.9%) and $-\alpha^{4.2}$ (1.6%) . Based on hematological indices we found that, 36% of subjects have MCV less than 83 and 74.8% of people MCH less than 29 and 34.3 subjects have MCV<93 & MCH<29. So keeping these two indices together with HbA2 can be more useful in thalassemia screening.

Key Words: Alpha thalassemia, mutation, Sistan and Balouchistan province, Prenatal Diagnosis