Original Paper

Carrier frequency of alpha thalassemia mutations among newborns in northern Iran

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Abstract

Background and Objective: Alpha Thalassemia is one of the most prevalent hemaglobinophaties worldwide. Alpha thalasseima patients may represent wide spectrum of symptoms ranging from asymptomatic to severe life threatening anemia. This study was done to assess the carrier frequency of alpha globin gene mutations among newborns in north of Iran.

Methods: In this descriptive study, 412 cord blood samples of neonate from Amir Mazandari hospitali were randomly selected during 2012. Genomic DNA was extracted using phenol-chloroform method. Multiplex Gap- PCR and PCR-RFLP methods were applied in order to detect three common gene deletions, one triplication and one point mutation.

Results: Total allelic frequency of investigated mutations was 0.0825. The -α3.7 deletion with allelic frequency of 0.0485 was the most prevalent mutation among 412 neonates. Allelic frequencies of -α4.2, αααanti3.7 triplication and α-5nt mutations were 0.0206, 0.0109 and 0.0024; respectively and -Med double gene deletion was not detected.

Conclusion: Most mutated cases had single gene deletion that is asymptomatic while -Med double gene deletion was not detected among the neonates. Therefore, there is low probability of a child birth with Hb H disorder in the region.

Keywords: Alpha Thalassemia, Alpha globin, Gene Mutation, Newborn, Iran

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