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 hemoglobinopathies worldwide. Alpha thalassemia patients may represent a 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 the north of Iran.

Methods: In this descriptive study, 412 cord blood samples of neonates from Amir Mazandari hospital 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, ααanti3.7 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 HbH disorder in the region.

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

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