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Alpha-Thalassemia patterns in Saudi population: a single-center study from Jeddah city, Saudi Arabia

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Abstract

Alpha thalassemia is a genetic disorder characterized by a decreased synthesis of alpha-globin chains. It is most often caused by a deletion mutation in one or more alpha-globin chains. To date, no comprehensive studies have been conducted on α- thalassemia in Jeddah. Therefore, the purpose of this research is to identify the spectrum of genetic mutations responsible for α-thalassemia in our region. All individuals with microcytic, hypochromic RBCs, and normal hemoglobin A2 were included. IDA was excluded. A total of 20 samples for individuals suspected of alpha-thalassemia were selected for further genetic analysis. Multiplex ligation-dependent probe amplification (MLPA) assay was used to detect deletion mutations in alpha genes. Among all samples tested, the -α3.7deletion mutation was detected in 19 (95%) of cases, and no deletion mutation was detected in 1(5%). In addition, 3 (15 %) individuals were heterozygous for -α3.7, while -α3.7 homozygosity was found in 80 % of the analyzed cases. Furthermore, no significant variants related to anemia or α-thalassemia were detected in the sample that showed no deletion mutations. On the other hand, the hematological characteristics of α3.7 subjects were significantly lower compared to the control group in the mean of Hb, Hct, MCV, MCH, and MCHC ($P< 0.001$). These data support the importance of including alpha thalassemia screening in the premarital screening program in Saudi Arabia. The results are consistent with previous studies that indicated that α3.7 deletion is most common among Saudis. Routine screening for this mutation will improve the premarital screening program in Saudi Arabia. Thus, it contributes to reducing the spread of genetic diseases

Biography

Sarah completed her bachelor's degree in the College of Applied Medical Sciences at the age of 21 from Umm Al-Qura University and postmaster study in Hematology and Blood Transfusion from the College of Applied Medical Sciences at King Abdulaziz University at the age of 31. She is a supervisor of transfusion service in East Jeddah Hospital for 7 years.

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