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Alpha-Thalassemia patterns in Saudi population: a single-center study

Sarah S Alswat[2,3], Raed Alserihi[1,2]

1 Center of Innovation in Personalized Medicine (CIPM), King Abdulaziz University, Jeddah, Saudi Arabia
2 Department of Medical Laboratory Technology, Faculty of Applied Medical Sciences, King Abdulaziz University
3 Department of Laboratory, East Jeddah Hospital (EJH), Ministry of Health (MOH)

Introduction

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. Although alpha-thalassemia in Saudi Arabia has among the highest rates of public health disorders, there is little outdated knowledge about the molecular spectrum of alpha-thalassemia among Saudis. Therefore, the purpose of this research is to determine the spectrum of alpha thalassemia genotypes in individuals undergoing premarital screening. Thus, the premarital screening program can be improved to reduce the prevalence of genetic disorders.

Methodology

Multiplex ligation-dependent probe amplification (MLPA) assay was used to detect mutations in alpha genes in 20 blood samples control/ 20 blood samples suspected of having alpha thalassemia based on hematological features. In addition, next-generation sequencing was used to detect a non-deleted mutation in the alpha genes.

Results

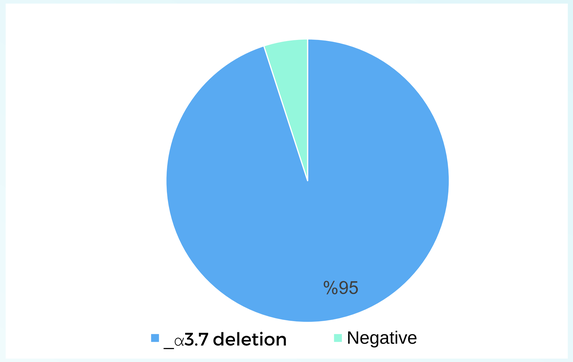

Fig. 1: *HBA* gene deletion screening results

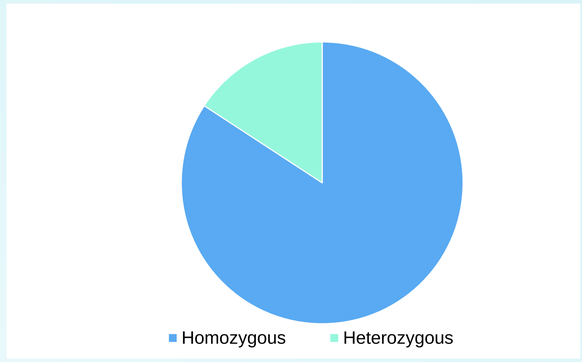

Fig. 2: Type of *HBA* gene deletion genotype

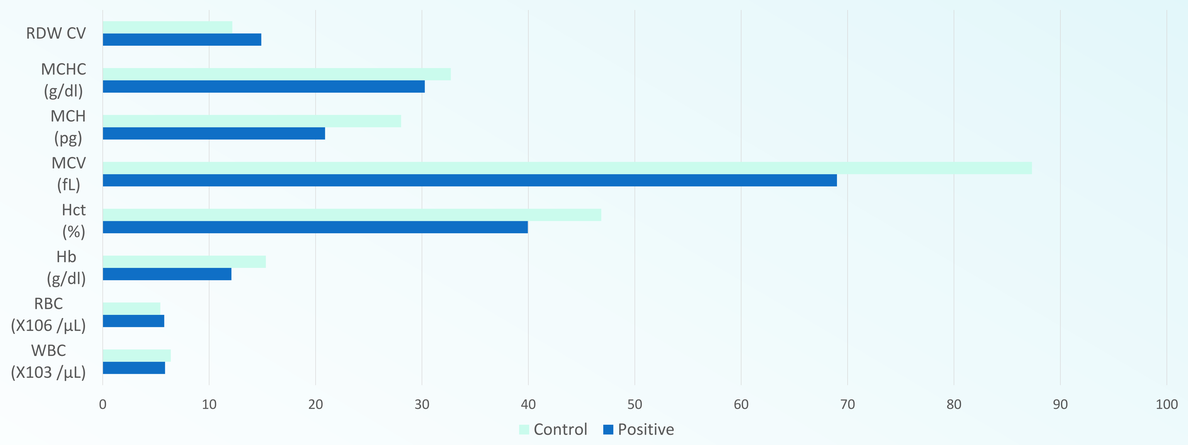

Fig.3 Comparison of haematological parameters between control group and alpha thalassemia subjects.

Results

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 (15.7%) individuals were heterozygous for -α3.7, while -α3.7 homozygosity was found in 84.3% of the positive cases. Furthermore, for the deletion-negative sample, no significant variants related to anemia or alpha thalassemia were detected by NGS. On the other hand, the hematological characteristics of α3.7 subjects were significantly lower compared to the control group in mean Hb, Hct, MCV, MCH, and MCHC ($P < 0.001$).

Conclusion

These data support the importance of including alpha thalassemia screening in the premarital screening program in Saudi Arabia. Morever, there is a need to determine the spectrum of α-thalassemia mutations that are common in the Saudi population. This will be useful to identify at-risk couples and to prevent severe forms of α-thalassemia such as HbH and Hb Barts.

References

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Contact email: saralswat1410@gmail.com