talassemia é preciso melhorar a notificação do tratamento da doença pelas instituições de saúde.

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ASSOCIATION OF HUMAN NEUTROPHIL ANTIGENS WITH HAPLOTYPES IN SICKLE CELL ANEMIA

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Introduction: The SLC44A2 gene (rs2288094) encodes a choline-like membrane transporter expressed in various cells including blood cells. Genetic variants in this gene are associated with blood antigen polymorphisms, such as the human neutrophil antigens (HNA) HNA-3a/b and the erythroid antigen RIF. The HNA-3 polymorphism is associated with a single nucleotide polymorphism (SNP) at position 455G>A (rs2288904-G/A). Genome-wide association studies linked expression of HNA-3b (455A, Gln152) on the SLC44A2 protein with a decreased risk of venous thrombosis (VT). Notably, this antigen plays a critical role in the interaction of neutrophils with platelets and the von Willebrand factor. This is particularly relevant for patients with sickle cell anemia (SCA), whose condition can include different haplotypes. **Objective:** To investigate the association of HNA-3a and HNA-3b antigens with sickle cell anemia haplotypes. **Materials and methods:** This study included a Brazilian sample of 114 patients (44 men and 70 women) with a mean age of 22.5 years (15-42), all confirmed as HbSS by high-performance liquid chromatography and molecular analysis. DNA was extracted from leukocytes using the phenol/chloroform method and was used to confirm the genotype of Hb S and identify haplotypes by PCR/RFLP. The genotypes of the SLC44A2 gene were determined by the TaqMan assay. The association between variables was evaluated using the chi-square test. All statistical analyses were performed using RStudio software, with $p < 0.05$. **Results:** Genotyping of HNA antigens in HbSS patients revealed the presence of 81 individuals with HNA3a/a genotype, 5 with HNA3b/b genotype, and 28 with HNA3a/b genotype. Hardy-Weinberg equilibrium analysis demonstrated allele frequencies of 0.83 and 0.17, respectively (Chi2 = 1.52). In our study sample, six sickle cell anemia haplotypes were identified: 58 individuals with Bantu/Bantu haplotype, 8 with Benin/Benin, 7 with Bantu/Atypical, 2 with Benin/Atypical, 38 with Bantu/Benin, and 1 with Benin/Cameroon. The association of HNA genotypes with haplotypes showed the following distribution: 3 occurrences of AG Bantu/Atypical, 12 of AG Bantu/Bantu, 12 of AG Bantu/Benin, and 1 of AG Benin/Atypical. The GG genotype showed the following distribution: 44 individuals with GG Bantu/Bantu, 8 with GG Benin/Benin, 2 with GG Bantu/Atypical, 25 with GG Bantu/Benin, 1 with GG Benin/Cameroon, and 1 with GG Benin/Atypical. Finally, the AA genotype was observed in 2 individuals with AA Bantu/Bantu, 2 with AA Bantu/Atypical, and 1 with AA Bantu/Benin. Analyses revealed a significant association between genotypes and haplotypes ($p = 0.04$, $X2 = 18.30$, and $df = 10$). **Discussion:** It is known that the HNA3a antigen is associated with a significantly increased risk of thrombosis, while HNA3b may confer up to 30% protection against this risk. Additionally, SCA haplotypes are widely recognized for influencing disease severity and the frequency of associated complications. The absence of studies directly relating HNAs to disease haplotypes raises important questions about how the combination of these characteristics can enhance the impact on the progression and severity of sickle cell anemia. **Conclusion:** Identifying these associations is crucial to improving the understanding of the genetic bases of variations in blood alloimmunization phenotypes, contributing to more effective clinical and transfusion management strategies for patients with SCA.

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IMPACTO DA VARIANTE REGULADORA RS11865131 E CO-HERANÇA DE ALFA-TALASSEMIA NOS PARÂMETROS HEMATIMÉTRICOS E CRISES ÁLGICAS NA DOENÇA FALCIFORME

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Objetivos: Avaliar associação entre variante reguladora rs11865131 de genes HBA, co-herança de alfa-talassemia, parâmetros hematimétricos e crises álgicas na anemia falciforme (SS) e hemoglobinopatia SC. **Material e métodos:** Estudo observacional e transversal em indivíduos acompanhados no Hemocentro de Governador Valadares, MG. Extraíram-se dados demográficos, hematimétricos e clínicos de registros médicos do último ano anterior ao estudo. Genotipagens foram realizadas por qPCR com discriminação alélica por sondas TaqMan® de rs11865131 e Gap-PCR das mutações deleccionais de alfa-tal. **Resultados:** Avaliaram-se 58 indivíduos SC (média de idade 12,6 anos, mediana 12,8, 52% do sexo masculino, 5 em uso de hidroxiureia [HU]) e 110 SS (média de idade 12,2, mediana 12,4, 53,6% meninos, 55% em uso de HU). Média de Hb de SC 11,00 g/dL ± 0,769 significativamente maior que SS (8,28 g/dL ± 1,36). O uso de HU em SS aumentou significativamente Hb e HbFetal e reduziu leucócitos, neutrófilos e monócitos; em SC, reduziu leucócitos e neutrófilos. Co-herança de alfa-tal (α-3.7Kb) presente em 22,4% (SC) e em 19% (SS). Em ambos os grupos com co-herança de alfa-tal houve redução significativa nas médias de HCM e VCM ($p < 0,001$ - efeito de magnitude elevada > 0,80). Somente no grupo SC com co-herança de alfa-tal, houve redução significativa e efeito moderado a alto de leucócitos ($p = 0,017$; $€^2 = 0,0992$) e monócitos ($p = 0,003$; $€^2 = 0,1581$). Ao menos um episódio de crise álgica ocorreu em 57% (SC) comparado com 48,1% em SS. Não houve