EVALUATION OF THE ALLELE FREQUENCY OF THE HFE GENE POLYMORPHISMS IN PATIENTS WITH HIPERFERRITINEMIA

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Introduction: The Hereditary Hemochromatosis (HH) is a genetic disorder characterized by the progressive deposition of iron and injury in multiple organs because of a mutation in the gene encoding a regulatory protein (HFE), resulting in excess of this ion in the organism[1]. The C282Y mutation, together with the H63D and S65C mutation, match the most recurrent mutations in HH. **Objectives:** Investigation of mutations C282Y, H63D and S65C in HFE gene and allele frequency of these genes in patients with persistent hiperferritinemia met in the center of Oncology and Hematology of Mossoró-RN and Blood Center. **Material and Methods:** Were analyzed 120 patients (66 men and 54 women) with persistent hyperferritinemia, the PCR-RFLP was performed on extracted DNA to detect polymorphisms. **Results and Discussion:** We analyzed 120 cases and 120 controls for data comparison, both groups were represented by 66 men and 54 women. The allele frequency obtained in patients with hyperferritinemia was n=15 (6.25%) n=44 (18.3%) and n =7 (2.91%) respectively for the C282Y, H63D and S65C. The controls, was found the allele frequency of n =7 (2.91%), n =32 (13.33%) and n =2 (0.83%) respectively the mutations in the same order. And the frequency found in this casuistry most consistent data published in Brazil and abroad. Among all surveyed individuals, iron overload was observed only in those who had the HH clinically manifest and the following alleles: 7 C282Y; 4 H63D and 0 S65C. **Conclusions:** Showed greater the C282Y allele penetrance in relation to other on HH. The H63D mutation was the second most frequent when associated with disease Due to the high prevalence of HH, its simple, effective and inexpensive treatment, the molecular diagnosis of HH becomes an important aid in the control of complications from the disease, especially in risk groups.

KEYWORDS: Hereditary Hemochromatosis; HFE; hyperferritinemia; Iron; Molecular diagnosis.

1. Leão, G.D; Freire, J.L; Fernandes, A.L.A.C, et al. Analysis of HFE Genes C282Y, H63D, and S65D in Patients With Hyperferritinemia From Northeastern Brazil. Journal of Clinical Laboratory Analysis. Natal – RN. v.01, p.1–8. 2014.