um mês de evolução marcado por plenitude pós-prandial, perda ponderal significativa (11 kg), além de parestesia de membros simétrica associada à instabilidade na marcha e bradipsiquismo. Ao exame físico observou-se também atrofia de papilas gustativas e a avaliação neurológica foi compatível com neuropatia periférica e redução da sensibilidade dolorosa e tátil do nível T8 até região suprapatelar bilateral. Laboratorialmente: anemia macrocítica (Hb 8,3 g/dL; Hto 24,7%; VCM 118 fL), índice de reticulócitos corrigido de 1,1, leucopenia com neutropenia (Leucócitos 2.240/mm^3 com 270 neutrófilos), trombocitopenia (28.000/mm^3), LDH muito elevada (4.597 UI/L) e bilirrubina indireta de 1,9 mg/dL. À hematoscopia: raros neutrófilos plurissegmentados, macrovalócitos e numerosos dacriócitos e esquizócitos. Embora a história clínica sugerisse o diagnóstico de deficiência de vitamina B12, os achados laboratoriais e da hematoscopia tornavam microangiopatia trombótica (MAT), especificamente púrpura trombocitopênica trombótica (PTTa), um preocupante diagnóstico alternativo (PLASMIC score: 6 pontos; French score: 2 pontos). Diante de um elevado VCM e da ausência de reticulocitose real, foi optado pelo início da reposição de vitamina B12 e pelo adiamento do início de plasmaférese. Após 48 horas da admissão, a dosagem de vitamina B12 foi liberada < 50 pg/mL. A paciente evoluiu com rápida melhora clínica e laboratorial. **Discussão:** A pseudo-MAT caracteriza-se por: anemia hemolítica, presença de esquizócitos e dacriócitos no sangue periférico, trombocitopenia grave, LDH muito elevada e haptoglobina consumida. Pacientes com deficiência grave de vitamina B12 podem apresentar um amplo espectro de manifestações hematológicas, dentre elas a pseudo-MAT é relatada em apenas 2,5% dos pacientes. Por ser achado incomum, de difícil reconhecimento e poder vir acompanhado de sintomas neurológicos, muitos pacientes recebem o diagnóstico de PTTa e são tratados desnecessariamente com plasmaférese. Diferentemente do que ocorre em outras formas de MATs, na pseudo-MAT o VCM costuma ser muito elevado, apesar da ausência de reticulocitose real, quando corrigimos pela anemia. O conhecimento dessa forma de apresentação e de suas peculiaridades pode evitar o diagnóstico incorreto com consequente instituição de tratamentos que agregam morbidade. O caso chama atenção para a necessidade de afastarmos deficiência de B12 quando diante de quadros clínicos semelhantes ao descrito.

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DIFFERENCES IN THE FREQUENCY OF POLYMORPHISMS IN THE HFE GENE (H63D AND C282Y) IN A POPULATION GROUP FROM NORTHWEST SÃO PAULO, BRAZIL

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Goals: Hereditary hemochromatosis (HH) is an autosomal recessive disease characterized by excess iron. People with polymorphisms in the *HFE* gene (*H63D* and *C282Y*) may develop inflammatory processes resulting from changes in iron metabolism. The ability of the HFE protein to make connections with transferrin receptors, located on cell membranes, allows it to help control the absorption of iron in the body. This work sought to investigate the frequency of *HFE* gene variants in a sample from the city of São José do Rio Preto, SP, Brazil. **Methodology:** DNA samples from 105 people were subjected to molecular analysis to confirm mutations in the *HFE* gene (*H63D* and *C282Y*), using PCR-RFLP. The group was separated by male and female gender, age group of 10 in 10 years, and ethnic groups. Statistical analyzes were performed using the IBM SPSS Statistics 20 software and data were presented as percentages, with a significance level of 0.05. The associations between the wild-type homozygote, heterozygote, and mutant homozygote genotypes for each mutation were tested by the Chi-Square test. Then, the allele frequencies in the genotypes were calculated. Fisher's exact test was performed to verify the association between gender, age group, and ethnicity with mutations in the *HFE* gene. **Results:** The sample included: 55 men (52.38%) and 50 women (47.62%), with ages ranging from 18 to 62 years, with an average of 30 years. A total of 210 alleles were evaluated and for the *H63D* mutation, 78 wild-type homozygotes, 26 heterozygotes, and 1 mutant homozygote were found. For the *C282Y* mutation, 99 wild-type homozygotes, 6 heterozygotes and no mutant homozygote were found. Allelic frequency analysis indicated the occurrence of significant differences between wild-type homozygous, heterozygous, and mutant homozygous genotypes for the mutations ($p < 0.05$). Fisher's exact test indicated no association between gender, age, and ethnicity for the *H63D* and *C282Y* polymorphisms ($p > 0.05$). The heterozygous genotype for *H63D* was more frequent in men (15-14%) than women 11 (10%). For *C282Y* equal frequencies (3-3%) were found for heterozygosity between men and women. *H63D* heterozygosity was more frequent in Caucasians (22-21%). In African-derived, only one individual was heterozygous for *H63D*. **Discussion:** The frequency of HH genotypes varies among different populations. The low incidence or absence of homozygous alleles is due to the different degrees of miscegenation in the formation of the Brazilian population. The *H63D* mutation is an important marker for the Brazilian population, being frequent in Africa, although it has low phenotypic expression. The *C282Y* is predominantly associated with homozygosity for clinical cases, however, it is rare in African populations and frequent in Caucasians. Our results corroborate the data in the literature, with a predominance of the *H63D* mutation in Caucasians, African-derived, and Afro-descendants, and *C282Y* in Caucasians. **Conclusion:** The frequency of *H63D* and *C282Y* polymorphisms in the *HFE* gene showed a higher occurrence of heterozygotes for *H63D* in this population group. Studies to characterize HH are needed to devise strategies for effective support to patients and, therefore, improve quality of life.

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ASSOCIAÇÃO ENTRE GRAUS DE OBESIDADE E DEFICIÊNCIA DE VITAMINA B12 E ÁCIDO FÓLICO: REVISÃO SISTEMÁTICA COM METANÁLISE

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