

desnutrição grave, 25,7% e 21% respectivamente, os casos de anemia não foram correlacionados, sendo mais predominante no Sudeste com 45,8% dos casos.

<https://doi.org/10.1016/j.htct.2024.09.006>

HYPERFERRITINEMIA ASSOCIATED WITH HFE GENE MUTATIONS (H63D, C282Y, AND S65C) IN BRAZIL

TF Ribeiro^a, ACP Flausino^a, OR Júnior^b,
A Lorenzetti^b, CR Bonini-Domingos^a

^a Universidade Estadual Paulista Júlio de Mesquita Filho (UNESP), São José do Rio Preto, SP, Brazil

^b Hemocentro de São José do Rio Preto, São José do Rio Preto, SP, Brazil

Objectives: Intracellular iron overload, resulting from excessive iron accumulation in ferritin, is a primary cause of hyperferritinemia, often linked to genetic diseases such as hereditary hemochromatosis (HH). Most HH cases are associated with the HFE gene, specifically H63D, S65C, and C282Y polymorphisms. Hyperferritinemia related to hemochromatosis can exacerbate symptoms due to excessive iron. This study aimed to evaluate changes in serum iron, ferritin, and transferrin saturation (TS) in patients with HFE gene mutations (H63D, C282Y, and S65C). **Methodology:** Blood samples from 20 adults with hyperferritinemia were collected to investigate the inheritance of HFE gene polymorphisms (H63D, C282Y, and S65C); the group were categorized based on serum iron, ferritin, and TS levels into low, normal, and high. DNA samples underwent molecular analysis using PCR-RFLP to confirm mutations. Statistical analyses were performed using IBM SPSS Statistics 20, with $p = 0.05$. Associations between wild-type homozygous, heterozygous, and mutant homozygous genotypes for each polymorphism were tested using the Chi-Square test. Associations between sex and age with alterations in serum iron, ferritin, and TS levels were tested using Fisher's exact test. ANOVA was performed to assess the association between serum iron, ferritin, and TS with HFE gene mutations. **Results:** The group consisted of 8 women (40%) and 12 men (60%), aged 19 to 87 years, with a mean age of 58.5 years. Of the 80 alleles evaluated, 14 heterozygous and 3 homozygous mutants were identified for the H63D mutation. For C282Y, 2 heterozygous and 3 homozygous mutants were found. No genotypes were detected for S65C. Men showed a higher prevalence of ferrocytosis (55%) compared to women (20%). Normal iron levels were more common in women (20%) than in men (5%). High TS levels predominated in men (60%), with only one woman showing low TS (TS of 5%). No significant differences were found when comparing sex and age with iron profile ($p > 0.05$). Men exhibited higher mean levels of iron, ferritin, and TS, with 204.7 $\mu\text{g/dL}$, 2593.6 ng/mL , and 77.3%, respectively. Women had mean values of 181.2 $\mu\text{g/dL}$, 1931.9 ng/mL , and 70.1%. Homozygous genotypes for H63D and C282Y were found only in men. Individuals with heterozygous and homozygous H63D had higher mean iron and ferritin levels (197.8 $\mu\text{g/dL}$ and 2251.9 $\mu\text{g/dL}$ for heterozygotes, 216.7 $\mu\text{g/dL}$ and 2633.3 $\mu\text{g/dL}$ for homozygotes) compared to

C282Y. The TS was highest in C282Y heterozygous (TS of 83%). No significant differences were observed in the mean iron profile concerning HFE gene mutations ($p > 0.05$). **Discussion:** Higher mean iron levels in men suggest a greater predisposition to iron overload in this population group. The H63D mutation was more frequent in this group, indicating a possible risk factor for iron overload. Homozygosity C282Y, known for causing severe overload, is predominantly found in Caucasians. The low incidence of homozygous alleles in the Brazilian population may be attributed to genetic admixture. Lower TS values in C282Y heterozygotes can be attributed to low values in women. Iron loss during pregnancy and menstruation may influence disease manifestation in women. **Conclusion:** A high frequency of the H63D genotype, associated with elevated iron profile levels, particularly in men, who exhibited a predominance of mutations. Elevated ferritin levels in H63D suggest a specific marker for increased iron absorption.

<https://doi.org/10.1016/j.htct.2024.09.007>

PREVALÊNCIA DE ANEMIA EM CRIANÇAS MENORES DE 5 ANOS QUE FIZERAM ACOMPANHAMENTO NAS UNIDADES BÁSICAS DE SAÚDE DO MUNICÍPIO DE ITAPETININGA EM 2023

VM Irineu, WMS Junior, RAMN Godoy, NM Vital

Universidade Municipal de São Caetano do Sul
(USCS), São Caetano do Sul, SP, Brasil

A anemia em crianças menores cinco anos de idade é frequente no Brasil, com uma prevalência estimada em 25 a 68%. Considerada um problema de saúde pública, a negligência dos cuidados da anemia na infância poderá comprometer a saúde física e mental do indivíduo durante toda sua vida. As deficiências nutricionais são a principal causa de anemia nessa faixa etária e medidas simples e orientações nutricionais podem ajudar a diminuir o problema. Neste sentido, a investigação quanto à prevalência de anemia em crianças menores de 5 anos na cidade de Itapetininga, se faz importante para avaliar o diagnóstico situacional local e traçar o perfil epidemiológico da doença possibilitando estruturar medidas de prevenção e promoção de saúde nessa faixa etária. **Objetivo:** Os objetivos do estudo foram identificar o perfil epidemiológico da anemia entre crianças menores de 5 anos atendidas nas Unidades Básicas de Saúde (UBS) do Município de Itapetininga no ano de 2023 e entender os fatores de risco associados à presença de anemia nesta população. **Material e método:** Realizado estudo transversal por meio da coleta de dados dos prontuários das UBS de referência de menores de cinco anos no período de janeiro a dezembro de 2023. Foram coletados dados referentes a identificação (sexo, idade), anamnese (informações sobre aleitamento materno e suplementação de ferro dos seis aos vinte e quatro meses de vida) e exame físico (dados antropométricos). Na consulta de retorno para avaliação dos exames foram coletados dados do hemograma e, exames correlatos quando disponíveis (ferritina, eletroforese de hemoglobina). Foram excluídos do estudo crianças fora da faixa etária de interesse