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Serum ferritin and transferrin saturation levels in β^0 and β^+ thalassemia patients

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ABSTRACT. There have been few studies on the mutations that cause heterozygous beta-thalassemia and how they affect the iron profile. One hundred and thirty-eight individuals were analyzed, 90 thalasemic β^0 and 48 thalasemic β^+, identified by classical and molecular methods. Mutations in the hemochromatosis (*HFE*) gene, detected using PCR-RFLP, were found in 30.4% of these beta-thalassemic patients; heterozygosity for H63D (20.3%) was the most frequent. Ferritin levels and transferrin saturation were similar in beta-thalassemics with and without mutations in the *HFE* gene. Ferritin concentrations were significantly higher in men and in individuals over 40 years of age. Transferrin saturation also was significantly higher in men, but only in those without *HFE* gene mutations. There was no significant difference in the iron profile among the β^0 and β^+ thalassemics, with and without *HFE* gene mutations. The frequency of ferritin values above 200 ng/mL in women and 300 ng/mL in men was also similar in β^0 and β^+ thalassemics ($P > 0.72$). Our conclusion is that ferritin levels are variable in the beta-thalassemia, trait regardless of the type of beta-globin mutation. Furthermore, *HFE* gene polymorphisms do not change the iron profile in these individuals.

Key words: Ferritin; Beta-thalassemia; Hyperferritinemia; Transferrin saturation

