
**Abstract**

BACKGROUND: Women who inherit heterozygosity for the C282Y mutation of the HFE gene may have increased serum iron indices and hemoglobin and are less likely to develop iron deficiency compared with women with the wild-type genotype.

METHODS: We performed a cross-sectional analysis of 497 women 20-44 years of age and 830 women >51 years of age drawn from the Busselton (Australia) population study to assess the effects of the HFE genotype on serum iron and hematology indices.

RESULTS: Heterozygosity for the C282Y mutation occurred in 13.8% of the study population, comprising 11.8% C282Y wild-type heterozygotes and 2.0% C282Y/H63D compound heterozygotes. In the younger age group, C282Y wild-type women did not have significantly increased serum iron, transferrin saturation, or hemoglobin values, and were not protected from developing iron deficiency, compared with women of the same age with the wild-type genotype. Young compound heterozygous women had higher means for serum iron (25.0 vs 16.9 micromol/L; P <0.001), transferrin saturation (42.0% vs 25.6%; P <0.05), hemoglobin (139.4 vs 132.3 g/L; P <0.05), and corpuscular volume (91.1 vs 87.7 fL; P <0.05), and a higher median ferritin (53 vs 44 microg/L; P <0.05) compared with the wild-type genotype. Similar results were observed for compound heterozygotes in the >51 years age group.

CONCLUSIONS: Women with the compound heterozygous HFE genotype C282Y/H63D, but not the C282Y wild-type genotype, had increased values for serum iron and transferrin saturation, and the younger age group also had increased hemoglobin values. We conclude that the compound heterozygous genotype may have a beneficial effect in protecting women from iron deficiency.