
**Abstract**

BACKGROUND: Heterozygotes for the C282Y mutation of the HFE gene may have altered hematology indices and higher iron stores than wild-type subjects.

METHODS: We performed a cross-sectional analysis of 1488 females and 1522 males 20-79 years of age drawn from the Busselton (Australia) population study to assess the effects of HFE genotype, age, gender, and lifestyle on serum iron and hematology indices.

RESULTS: Male C282Y heterozygotes had increased transferrin saturation compared with the wild-type genotype. Neither male nor female heterozygotes had significantly increased ferritin values compared with the wild-type genotype. Younger (20-29 years) wild-type males, but not heterozygous males, had significantly lower ferritin values than wild-type males in the older age groups. Compound heterozygous subjects had increased means for serum iron, transferrin saturation, corpuscular volume, and corpuscular hemoglobin compared with the wild-type genotype, and the males also had increased ferritin values (medians 323 vs 177 microg/L; P = 0.003). In both male and female wild-type subjects, an increased body mass index was associated with decreased serum iron and transferrin saturation and increased ferritin values. There was a significant increase in ferritin concentrations in both genders with increasing frequency of red meat consumption above a baseline of 1-2 times per week and alcohol intakes >10 g/day.

CONCLUSIONS: Male C282Y heterozygotes had significantly increased transferrin saturation values. Compound heterozygous (C282Y/H63D) subjects formed a separate category of C282Y heterozygotes in whom both iron and red cell indices were significantly increased compared with the wild-type genotype.