
**Abstract**

BACKGROUND & AIMS: Two major mutations are defined within the hemochromatosis gene, HFE. Although the effects of the C282Y mutation have been well characterized, the effects of the H63D mutation remain unclear. We accessed a well-defined population in Busselton, Australia, and determined the frequency of the H63D mutation and its influence on total body iron stores.

METHODS: Serum transferrin saturation and ferritin levels were correlated with the H63D mutation in 2531 unrelated white subjects who did not possess the C282Y mutation.

RESULTS: Sixty-two subjects (2.1%) were homozygous for the H63D mutation, 711 (23.6%) were heterozygous, and 1758 (58.4%) were wild-type for the H63D mutation. Serum transferrin saturation was significantly increased in male and female H63D homozygotes and heterozygotes compared with wild-types. Serum ferritin levels within each gender were not influenced by H63D genotypes. Elevated transferrin saturation > or = 45% was observed in a greater proportion of male H63D carriers than male wild-types. Male H63D homozygotes (9%) and heterozygotes (3%) were more likely to have both elevated transferrin saturation and elevated ferritin > or = 300 ng/mL than male wild-types (0.7%). Homozygosity for H63D was not associated with the development of clinically significant iron overload.

CONCLUSIONS: Presence of the H63D mutation results in a significant increase in serum transferrin saturation but does not result in significant iron overload. In the absence of the C282Y mutation, the H63D mutation is not clinically significant.

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