Online journal ISSN - 1676-5680
GMR Genetics and Molecular Research
EVOLUTION AND TECHNOLOGY

Frequency of the S65C mutation in the hemochromatosis gene in Brazil

V.C. Oliveira[1,2], F.A. Caxito[1], K.B. Gomes[3], A.M. Castro[1], V.C. Pardini[1] and A.C.S. Ferreira[1,2]

[1]Departamento de Genética Humana,
Instituto Hermes Pardini, Belo Horizonte, MG, Brasil
[2]Laboratório de Genética Animal e Humana,
Instituto de Ciências Biológicas,
Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[3]Faculdade de Farmácia,
Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil

Corresponding author: V.C. Oliveira
E-mail: vanessa.oliveira@labhpardini.com.br

Genet. Mol. Res. 8 (3): 794-798 (2009)
Received December 3, 2008
Accepted May 13, 2009
Published July 14, 2009

ABSTRACT. Development of hereditary hemochromatosis is associated with the C282Y, H63D or S65C mutations in the hemochromatosis gene. Though there is extensive knowledge about the former two, there is little information on the mechanism of action and the allelic frequency of the S65C mutation. We examined the prevalence of the S65C mutation of the hemochromatosis gene in Brazilians with clinical suspicion of hereditary hemochromatosis. Genotyping for this mutation was carried out in 633 individuals with clinical suspicion of hereditary hemochromatosis, using the polymerase chain reaction, followed by enzymatic digestion. The sample comprised 77.1% men and 22.9% women, giving a ratio of approximately 3:1; the mean age was 48.8 $\pm$ 13.8 years. More than half (57.3%) of the individuals in the sample were 41 to 60 years old. The frequency of heterozygotes for this mutation was 0.016; no homozygous mutant patients were found.

This is the first analysis of the S65C mutation in individuals suspected of having hereditary hemochromatosis in Brazil.

Key words: Hereditary hemochromatosis; HFE gene; S65C mutation; Brazil

