

Estimation of Allele Frequencies and Population Incidence of Wilson Disease in Brazil

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Abstract

The present paper deals with the estimation of the overall frequency of *ATP7B* alleles determining Wilson disease (WD) and the population frequency of the condition in Brazil. Genealogical, demographic, and molecular data from 83 WD probands, studied at three distinct WD referral centers, were used for obtaining a population estimate for the overall frequency of alleles that in homozygous or compound heterozygous state determine the frequency of the condition in Brazil; the method we used exploits the relatively high proportion of consanguinity among parents of affected individuals compared with the proportion in the general population. The value we obtained for the overall allele frequency was $q = 0.006$, with a 95% bootstrap confidence interval of 0.004 to 0.012. The corresponding value for the disease incidence or frequency was $P(\text{WD}) = 0.000041$