ONLINE ISSN : 1880-1404
PRINT ISSN : 0916-717X

Biomedical Research on Trace Elements
Vol. 17 (2006) , No. 4 406-411

[PDF (756K)] [References]

Familial Genetic Analysis of Copper Transporting P-type ATPase (ATP7B)

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(Received: July 19, 2006)
(Accepted: October 18, 2006)

Abstract:
We have analyzed the copper-transporting P-type ATPase (ATP7B) gene responsible for Wilson's disease to provide an explanation for the early onset of acute hepatitis. The ATP7B coding sequence, including the intron-exon boundaries, has been screened for mutations by direct sequence analysis. The genetic data in this study indicate that the patient has been proven to carry both R778L and 2871del.C, each as one of the known disease-causing mutations. The R778L and 2871del.C were inherited from the father and the mother, respectively. Therefore, the patient was confirmed as a compound heterozygote for these mutations. This compound heterozygous mutation resulted in severe disruption of the ATP7B function. In sibs, however, the suspicion of Wilson's disease could be rejected because the compound heterozygous mutation was not found in them. The evidence suggests that the familial genetic analysis provides integral information to the genetic counseling for the disease in families with these patients.

Key words: Wilson's disease, copper-transporting P-type ATPase (ATP7B), genetic analysis, molecular diagnosis, familial screening

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