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

Biomedical Research on Trace Elements
Vol. 16 (2005) , No. 4 315-317

[PDF (239K)] [References]

Molecular diagnosis for presymptomatic patients with Wilson disease

Norikazu Shimizu[1)], Yukiko Takeshita[1)2)], Atsuko Watanabe[1)], Hiromichi Hemmi[3)], Hiroyuki Shimatake[3)] and Tsugutoshi Aoki[1)]

1) Second Department of Pediatrics, Toho University School of Medicine
2) Sanno Hospital
3) Laboratory of Molecular Biology, Toho University School of Medicine

(Accepted: October 20, 2005)

Abstract:
Wilson disease is a genetic disorder of copper metabolism characterized by hepatic and/or neurological manifestations. This disease is caused by mutations in the gene of copper transporting ATPase (ATP7B). Early diagnosis is very important to improve the prognosis of this disease. However, biochemical studies are not sufficiently effective for the definitive diagnosis of young patients. This study presents the molecular diagnosis of presymptomatic patients with Wilson disease. Three patients, two infants and one young child without any symptoms, and one carrier, were diagnosed by ATP7B gene analysis. We conclude that the molecular diagnosis of Wilson disease is very useful for the identification of young patients and familial analysis.

Key words: Wilson disease, ATP7B, gene analysis, presymptomatic diagnosis, familial analysis

[PDF (239K)] [References]

Download Meta of Article[Help]

To cite this article:

Norikazu Shimizu, Yukiko Takeshita, Atsuko Watanabe, Hiromichi Hemmi, Hiroyuki Shimatake and Tsugutoshi Aoki, “Molecular diagnosis for presymptomatic patients with Wilson disease”, Biomedical Research on Trace Elements, Vol. **16**, pp.315-317 (2005) .

JOI JST.JSTAGE/brte/16.315