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

Biomedical Research on Trace Elements
Vol. 15 (2004) , No. 1 88-90

[Image PDF (203K)] [References]

A young child case of Wilson disease with normoceruloplasminemia

R Adachi[1)], K Nihei[1)], Y Takeshita[1)], Y Yamaguchi[1)], N Shimizu[1)], N Kato[2)] and T Aoki[1)]

1) Second Department of Pediatrics, Toho University School of Medicine
2) Toho University School of Medicine

(Accepted: February 16, 2004)

Abstract:
A 5-year-old girl presented with chronic liver dysfunction. Her serum ceruloplasmin level was within normal range. Urinary copper excretion was slightly elevated. We performed ATP7B gene analysis. And mutations were found intron 4 and exon 18. One splicing donor site mutation, 1708-5 t to g, and missense mutation, N1270S, were detected. We thus diagnosed her to have Wilson disease with normo-ceruloplasminine-
mia. And we confirmed her diagnosis by measurement of hepatic copper content. Then, familial analysis was performed by gene analysis. Three carriers (parents and older brother) were detected. We conclude that molecular diagnosis is very useful for atypical cases of Wilson disease.

Key words: [in Japanese]

[Image PDF (203K)] [References]

Download Meta of Article[Help]
RIS
BibTeX

To cite this article:

R Adachi, K Nihei, Y Takeshita, Y Yamaguchi, N Shimizu, N Kato and T Aoki, “A young child case of Wilson disease with normoceruloplasminemia”, Biomedical Research on Trace Elements, Vol. **15**, pp.88-90 (2004) .