Gene analysis of Wilson's disease

2000 Fiscal Year Final Research Report Summary

Gene analysis of Wilson's disease

Research Project

Project/Area Number

11670487

Research Category

Grant-in-Aid for Scientific Research (C)

Allocation Type

Single-year Grants

Section

一般

Research Field

Gastroenterology

Research Institution

Kanazawa University

Principal Investigator

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Project Period (FY)

1999 – 2000

Keywords

Wilson's disease / gene analysis / genotype phenotype correlation / Fulminant hepatic failure / ATP7B / mutation

Research Abstract

Background : Fulminant hepatic failure is rare but fatal manifestation of Wilson's disease (WD). Although many mutations of the gene WD (ATP7B) have been reported, genotype-phenotype correlation in WD was not completely investigated and specific mutations related to fulminant hepatic failure have not been found. Aims : In this study, we, detected mutations of ATP7B among Japanese patients with WD including patients with fulminant hepatic failure and sought the correlation between mutations and phenotypes. We also sought to determine if genotypic assignment according to the types of protein-product could be related to the prevalence of fulminant hepatic failureamong the patients with WD.Subjects : NDA was isolated from peripheral blood collected from 45 unrelated Japanese families including 51 patients with WD.Methods : 1) Each exon of ATP7B was amplified by PCR, and the products were screened by SSCP.When abnormal bans were detected by SSCP, patient DNA was directly sequenced to identi ⋯▾ More

Research Products (7 results)

All Other

All Publications

[Publications] Toshihide Okada: "High prevalence of fulminant hepatic failure among the patients with mutant alleles for trancation of ATP7B in Wilson's disease"Hepatology . 32(4). 412A (2000)

[Publications] Toshihide Okada: "Mutation analysis of ATP7B and genotype-phenotype correlation in Japanese with Wilson's disease"Human Mutation. 15・5. 454-462 (2000)

[Publications] Toshihide Okada: "A new variant deletion of a copper-transporting P-type ATPase gene found in patients with Wilson's disease presenting with hepatic failure"Journal of Gastroenterology. 35・4. 278-283 (2000)

[Publications] 岡田俊英: "日本におけるWilson病-遺伝子解析に基づいて-"総合臨床. 49・4. 651-656 (2000)

[Publications] 岡田俊英: "Wilson病と遺伝子異常"日本臨床分子医学会雑誌. 37・63. 63 (2000)

[Publications] Toshihide Okada, et al.: "A new variant deletion of a copper-transporting P-type ATPase gene found in patients with Wilson's disease presenting with fulminant hepatic failure."J Gastroenterol. 35(4). 278-283 (2000)

[Publications] Toshihide Okada, et al.: "Mutational analysis of ATP7B and genotype-phenotype correlation in Japanese with Wilson's disease."Hum Mutat. 14(5). 454-462 (2000)

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