Selection of treatment in Wilson's disease based on gene analysis

2004 Fiscal Year Final Research Report Summary

Selection of treatment in Wilson's disease based on gene analysis

Research Project

Project/Area Number

15590632

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)

2003 – 2004

Keywords

Wilson's disease / genotype-phenotype correlation / hepatic failure

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 failure among the patients with WD. Subjects : DNA was isolated from peripheral blood collected from 45 unrelated Japanese families including 55 patients with WD. Methods : 1)Each exon of ATP7B was amplified by PCR, and the products were screened by SSCP. When abnormal bands were detected by SSCP, patient DNA was directly sequenced to iden ⋯▾ More

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