Gene analysis of Hemochromatosis

2002 Fiscal Year Final Research Report Summary

Gene analysis of Hemochromatosis

Research Project

Project/Area Number

13670501

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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2001 – 2002

Keywords

Hemochromatosis / HFE / gene analysis

Research Abstract

Background : Hereditary hemochromatosis (HH) is an autosomal recessive disorder caused by HFE mutations, mostly homozygosity for C282Y or compound heterozygosity for C282Y and H63D in Caucasian. In other populations, however, HH patients have no such mutations, and significance of HFE is unknown in these patients. Furthermore most of cases of hemochromatosis in Asian populations are sporadic ; no C282Y mutations, and only a few cases of H63D have been reported. However, there have been no studies analyzing the entire HFE coding regions to date ; the relationship between HFE and hemochromatosis in Asian populations remains unclear, Aim : To determine the significance of the HFE gene in hemochromatosis among Asian populations. Subjects & Methods : Unrelated nineteen Japanese patients with idiopathic hemochromatosis were tested. Genomic DNA was extracted from white blood cells, after performing PCR all coding regions were analyzed by direct sequencing. Results : No patients possessed C282Y mutations and only one patient was H63D heterozygous. Furthermore, no other casual mutations were identified in the HFE coding region. However, a high incidence of IVS2+4T/C mutation was noted. Twelve patients were C/C, five patients T/C, and none were T/T genotype. Discussion : This study suggests that HFE is not indicative for idiopathic hemochromatosis. However, a high prevalence of IVS2+4T/C mutation was found. Whether or not this mutation is contributes to hemochromatosis through abnormal splicing remains unclear. Further studies are necessary.

Research Products (4 results)

All Other

All Publications

[Publications] Yuhta Shiono, Hisao Hayashi, Toshihide Okada, et al.: "Iron Accumulation in the Liver of Male Patients With Wilson's Disease"The American Journal of Gastroenterology. 96・11. 3147-3151 (2001)

[Publications] Kaneko Y, Shiono Y, Suzuki A, Okada T, Mabuchi H.: "HFE Not Resposible for Idiopathic Hemochromatosis"Gastroenterology. 123(1)suppl. 61 (2002)

[Publications] Yuhta Shiono, Hrsao Hayashi Toshihide Okada, etal.: "Iron Accumulation in the Liver of Male Patients With Wilson's Disease."The American Journal of Gastroenterology. 96-11. 3147-3151 (2001)

[Publications] Kaneko Y, Shiono Y Suzuki A, Okada T, Mabuchi H.: "HFE Not Resposible for Idiopathic Hemochromatosis"Gastroenterology. 123(1) suppl. 61 (2002)

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