Analysis of Alu-mediated genomic deletion in a case with hemeooxygenase-1 deficiency

2002 Fiscal Year Final Research Report Summary

Analysis of Alu-mediated genomic deletion in a case with hemeooxygenase-1 deficiency

Research Project

Project/Area Number

13670788

Research Category

Grant-in-Aid for Scientific Research (C)

Allocation Type

Single-year Grants

Section

一般

Research Field

Pediatrics

Research Institution

Kanazawa university

Principal Investigator

SAIKAWA Yutaka Kanazawa university hospital, Pediatrics, Assistant Professor, 医学部附属病院, 講師 (60283107)

Project Period (FY)

2001 – 2002

Keywords

Human hemeoxygenase-1 / hereditary disease / gene correction / Alu repeat / homologous recombination / site-specific recombinase

Research Abstract

To investigate the pathomechanisms of Alu-mediated genomic deletion observed in a case with hemeoxygenase-1 (HO-1) deficiency, following analyses have been performed ;
1. Functional analyses of topoisomerase II-binding sites (TBSs) located in the introns and Alu repeats of the human HO-1 gene.
The role of Alu-mediated homologous recombination has been established as a pathomechanism in some hereditary diseases and cancers. Since chromosomal double-strand breaks (DSBs) play a crucial role in the homologous recombination processes, potential TBSs found in the introns and Alu repeats, surrounding HO-1 exon 2, could be the target sites of homologous recombination. To test this hypothesis, determination of functional TBSs in the

HO-1 gene was performed. HO-1^<+/+> LCLs established from the normal controls were treated with the inhibitors (VP-16 and doxorubicin) of topoisomearase II. The DNA fragments produced by the inhibition of endogenous topoisomerase II resulting in the creation of DSBs w ···▾ More

Research Products (11 results)

All | Other

All | Publications (11 results)

[Publications] Tomoko Toma: "HO-1 production by monocytes an a stress regulator and its clinical relevance"International Journal of Hematology. 73,suppl.. 64 (2001)

[Publications] Yoshinori Goto: "A novel single-nucleotide polymorphism in the 3'-untranslated region of the human dihydrofolate reductase gene with enhanced expression"Clinical Cancer Research. Vol.7. 1952-1956 (2001)

[Publications] Lijie Yue: "A functional single-nucleotide polymorphism in the human cytidine deaminase gene contributing to ara-C sensitivity"Pharmacogenetics. Vol.13. 29-38 (2003)

[Publications] Yutaka Saikawa: "Emergent properties of feedback regulation and stem cell behavior in a granulopoiesis model as a complex system"Complex Systems. Vol.14. 45-61 (2003)

[Publications] Rui Wang, Ed.: "Carbon Monoxide and Cardiovascular Functions(CRC Press, Boca Raton, Florida)"Human HO-1 Deficiency and Cardiovascular Dysfunction. Section IV. Molecular Pathology,. 320 (2002)

[Publications] Tomoko Toma: "HO-1 production by monocytes as a stress regulator and its clinical relevance"International Journal of Hematology. 73, suppl.. 64 (2001)

[Publications] Yoshinori Goto: "A novel single-nucleotide polymorphism in the 3'-untranslated region of the human dihydrofolate reductase gene with enhanced expression"Clinical Cancer Research. 7. 1952-1956 (2001)

[Publications] Lijie Yue: "A functional single-nucleotide polymorphism in the human cytidine deaminase gene contributing to ara-C sensitivity"Pharmacogenetics. 13. 29-38 (2003)

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[Publications] Rui Wand, Ed.: "Human HO-1 deficiency and cardiovascular dysfunction. Section IV. Molecular pathology"Carbon Monoxide and Cardiovascular Functions CRC Press, Boca Raton, Florida. 320 (2002)

[Publications] Nader G. Abraham: "Human heme oxygenase (HO-1) deficiency and die oxidative injury of vascular endothelial cells"Heme Oxygenase in Biology and Medicine Kluwer Academic/Plenum Publishers. 515 (2002)

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