

中国广东汉族人群核苷酸修复基因hMTH1 遗传多态性研究Genetic Polymorphisms of Nucleotide Repair Gene hMTH1 in Southern Chinese Han Population

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摘要

为研究中国南方汉族人群核苷酸修复基因hMTH1遗传多态性, 应用聚合酶链反应-单链构象多态性技术检测172名健康人外周血白细胞hMTH1基因启动子及全部5个外显子多态性, 并进行DNA测序。结果发现hMTH1基因启动子及外显子1序列保守, 未见突变; 外显子2第73位碱基存在T→C杂合型突变, 基因型TT和TC频率分别为93.02%、6.98%, 等位基因T和C频率分别为96.51%、3.49%; 外显子3第45位遗传密码存在T→C杂合型突变, 基因型TT和TC频率分别为95.35%、4.65%, 等位基因T和C频率分别为97.67%、2.33%, 该多态性为首次发现; 外显子4第83位遗传密码存在G→A杂合型突变, 基因型GG和GA频率分别为89.53%、10.47%, 等位基因G和A频率分别为94.77%、5.23%; 外显子5第119位氨基酸遗传密码存在C→T杂合型突变, 基因型CC和CT频率分别为95.93%、4.07%, 等位基因C和T频率分别为97.97%、2.03%。Abstract: In order to study the genetic polymorphisms of nucleotide repair gene hMTH1 in southern Chinese Han population, the polymorphisms of the gene's promoter and its five exons among peripheral blood lymphocytes of 172 Chinese Han people were analyzed with polymerase chain reaction-single strand conformation polymorphism (PCR-SSCP) and DNA sequencing. The sequences of the promoter and exon 1 of hMTH1 gene were conserved. A T to C polymorphism was detected at the 73th base in exon2. The genotype frequencies of TT and TC were 93.02% and 6.98%, respectively. The allelic frequencies of T and C were 96.51% and 3.49%, respectively. A T to C polymorphism was detected at codon 45 in exon3, which was first reported. The genotype frequencies of TT and TC were 95.35% and 4.65%, respectively. The allelic frequencies of T and C were 97.67% and 2.33%, respectively. A G to A polymorphism was detected at codon 83 in exon4. The genotype frequencies of GG and GA were 89.53% and 10.47%, respectively. The allelic frequencies of G and A were 94.77% and 5.23%, respectively. A C to T polymorphism was detected at codon 119 in exon5. The genotype frequencies of CC and CT were 95.93% and 4.07%, respectively. The allelic frequencies of C and T were 97.97% and 2.03%, respectively.

关键词 [hMTH1基因](#) [遗传多态性](#) [聚合酶链反应](#) [单链构象多态性](#) Key words [hMTH1 gene](#) [genetic polymorphism](#) [polymerase chain reaction](#) [single-strand conformation polymorphism](#)

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Abstract

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