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未定

β-纤维蛋白原基因启动子区单体型与缺血性脑卒中的关联分析

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摘 要 目的 研究b β-纤维蛋白原（FgB）基因启动子区单体型与缺血性脑卒中的关系。方法 采用聚合酶链反应-限制性片段长度多态性法（PCR-RFLP）、等位基因特异聚合酶链反应及核苷酸序列测定法分析160例缺血性脑卒中患者和130例健康对照个体的FgB基因启动子区的－1420G/A、-993C/T、-854G/A、－455G/A、-249C/T、-148C/T单核苷酸多态性(SNPs)和基因型，用EH+程序分析核苷酸多态性的连锁不平衡关系及单体型，用卡方检验分析病例组和对照组之间的基因型频率、等位基因频率及单体型频率的差异，用MatInspector 程序预测序列中的顺式作用元件。结果 脑卒中组与对照组之间的-993C/T、－455G/A、-148C/T 三个位点的基因型频率和等位基因频率存在显著性差异（p< 0.01），由－1420G、-993C、-854G、－455G、-249C、-148C构成的单体型H1在对照组中的频率高与病例组（P<0.05），由－1420A、-993T、-854G、－455A、-249T、-148T构成的单体型H14在病例组中的频率高于对照组（P<0.01）。野生型序列存在6个不同于突变型的顺式作用元件，突变型序列出现9个不同于野生型的顺式作用元件。结论 海南汉族人群中单体型H14可能是与缺血性脑卒中相关的危险因素，单体型H1可能是降低缺血性脑卒中发生风险的保护性因素，单体型与缺血性脑卒中的相关性可能是由于顺式作用元件改变所致。

关键词

分类号

Haplotypes in the promoter region of b β-fibrinogen gene and their relationship to ischemic stroke in Hainan Han population

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Abstract Objective To analyze the haplotypes in promoter region of beta-fibrinogen gene and their relationship to with ischemic stroke(IS). Method Genotypes were determined by PCR-RFLP, allelic specific-PCR and sequencing at polymorphisms -1420G/A、-993C/T、-854G/A、-455G/A、-249C/T、-148C/T in the promoter region of beta-fibrinogen gene in 160 cases with ischemic stroke and 130 healthy individuals from Hainan Han population. Pairwise linkage disequilibrium was calculated and haplotypes were estimated by the EH+ program. Statistical differences of allele, genotype and haplotype frequencies were obtained by Chi square test. Cis-elements were predicted by MatInspector program. Results There were highly significant differences in genotype frequencies and allelic frequencies at polymorphisms -993C/T、-455G/A、-148C/T between the IS group and control(P<0.01). The frequency of haplotype H1 with -1420G、-993C、-854G、-455G、-249C and -148C were higher in the control than in the IS group (P<0.05), whereas haplotype H14 with 1420A、-993T、-854G、-455A、-249T and -148T were higher in the IS group than in the control(P<0.01). Six cis-elements absent in the mutant sequences were found in the wild type sequence and nine cis-elements absent in the wild type sequences were found in the mutant sequence. Conclusion The results indicated that haplotype H14 may be a risk factor associated with IS and haplotype H1 a protective factor in reducing the risk of ischemic stroke in Hainan Han population. The association of haplotype with IS may be due to the change of cis-elements in the promoter region.

Key words

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