

论著

CTLA-4启动子区多态性与江南地区汉族人群SLE临床表型的相关性研究

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摘要

目的 探讨细胞毒性T淋巴细胞相关抗原-4 (cytotoxic T lymphocyte-associated antigen 4, CTLA-4) 基因启动子区多态性与系统性红斑狼疮 (systemic lupus erythematosus, SLE) 表型间的相关性。方法 采用单纯病例研究, 按临床表型和基因型、单倍型对120例女性SLE病人分组, 分析基因与表型的相关性。结果 -1722T>C位点、-318C>T位点及单倍型与SLE各器官损害无关 (P>0.05)。在抗SSA抗体阳性患者中, -1722T>C位点TT基因型的频率升高 (P=0.020, OR=5.26, 95%CI: 1.30-21.32)。抗RNP抗体阳性患者中, -318C>T位点CT与TT合并基因型的频率降低 (P=0.016, OR=0.16, 95%CI: 0.04-0.71)。两位点等位基因间存在连锁不平衡 (P<0.05), 且在抗SSA抗体阳性患者中, T-C单倍型频率升高 (P=0.014, OR=1.90, 95%CI: 1.14-3.18), C-C单倍型频率降低 (P=0.028, OR=0.55, 95%CI: 0.32-0.94); 而在抗RNP抗体阳性患者中, T-T单倍型频率降低 (P=0.029, OR=0.20, 95%CI: 0.05-0.85)。结论 在江南地区汉族人群中, CTLA-4基因-1722T>C位点与抗SSA抗体的产生相关, -318C>T位点与抗RNP抗体的产生相关。

关键词 [系统性红斑狼疮 \(SLE\)](#); [细胞毒性T淋巴细胞相关抗原-4 \(CTLA-4\)](#); [单核苷酸多态性 \(SNP\)](#); [表型](#)

分类号

Analysis of association of cytotoxic T lymphocyte-associated antigen-4 gene promoter polymorphisms with the clinical feathers of systemic lupus erythematosus in Han nationality population in South of the Changjiang River

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Abstract

Objective To explore the association between polymorphisms within cytotoxic T lymphocyte-associated antigen-4 (CTLA-4) gene promoter and systemic lupus erythematosus (SLE) phenotypes in Han nationality population in South of the Changjiang River. Methods One hundred and twenty female cases were enrolled with the aim of case only design, and the genotype was determined by PCR-RFLP. Then, we calculated χ^2 and ORs for related study. Results Analysis of association between the 2 SNPs and organ damage indicated no significant relationship (P>0.05). The -1722T/T genotype frequency in patients with positive anti-SSA were higher than that of patients with negative anti-SSA (P=0.020, OR=5.26, 95%CI: 1.30-21.32). The -318C/T and T/T combined group in patients with positive anti-RNP antibody had lower frequency than that of patients with negative anti-RNP antibody (P=0.016, OR=0.16, 95%CI: 0.04-

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▶ 本文作者相关文章
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0.71). Meanwhile, there was a linkage disequilibrium between the -1722T>C and -318C>T, so we analyzed the association between haplotypes and clinical features. In patients with positive anti-SSA, there was a relative high proportion in T-C haplotype ($P=0.014$, $OR=1.90$, $95\%CI: 1.14-3.18$), and a less one in C-C haplotype ($P=0.028$, $OR=0.55$, $95\%CI: 0.32-0.94$). Patients with positive anti-RNP antibody was found to have a lower frequency of T-T haplotype compared to patients with negative anti-RNP antibody ($P=0.029$, $OR=0.20$, $95\%CI: 0.05-0.85$). Conclusions In Han nationality population in South of the Changjiang River, -1722T>C allele may play an important role in the production of anti-SSA, and -318C>T allele may play an important role in the production of anti-RNP antibody.

Key words [cytotoxic T lymphocyte-associated antigen-4 \(CTLA-4\)](#); [systemic lupus erythematosus\(SLE\)](#); [single nucleotide polymorphism \(SNP\)](#); [phenotypes](#)

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