

论文

ERAP1基因与山东汉族人群强直性脊柱炎的相关性

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摘要:

目的 利用基于群体的病例对照关联分析方法, 探讨ERAP1基因是否为北方汉族人群的易感基因。方法 从山东省立医院收集了311例临床确诊的强直性脊柱炎病例及320例正常对照资料, 利用Taqman探针法对ERAP1基因内的两个单核苷酸多态性位点rs7711564和rs27434分型, 利用PLINK软件行Hardy-Weinberg遗传平衡检验、等位基因及基因型频率分布分析。样本统计学效能采用CaTS软件计算。结果 两个位点在病例组及对照组中均达到遗传平衡, 其中rs7711564位点罕见等位基因频率在病例组及对照组分别为0.4309和0.4891(OR=1.26, P=0.04), rs27434位点罕见等位基因频率在病例组和对照组分别为0.4839与0.4219(OR=0.78, P=0.03)。结论 ERAP1基因内的遗传变异与北方汉族人群强直性脊柱炎相关, 进一步证实了该基因是强直性脊柱炎的易感基因。

关键词: 强直性脊柱炎; ERAP1基因; 关联分析; 汉族人群

Association of polymorphisms in the ERAP1 gene with ankylosing spondylitis in the Han Chinese population

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Abstract:

Objective To validate the association of ERAP1 gene polymorphisms with ankylosing spondylitis (AS) in the northern Han Chinese population. Methods 311 AS patients and 320 healthy controls from Shandong Provincial hospital were enrolled in this case-control association study. The Taqman SNP genotyping method was used to genotype 2 SNPs: rs7711564 and rs27434 in the ERAP1 gene. The differences of allele and genotype frequencies were analyzed using PLINK 1.07. Results The distributions of the 2 SNPs were in Hardy Weinberg equilibrium (P>0.05) for both patients and controls. The allelic and genotypic distributions of the 2 SNPs were significantly different in the case and control groups. The risk of AS was increased with the C allele of rs7711564 (OR=1.26, P=0.04) and the A allele of rs27434 (OR=0.78, P=0.03). Conclusions The genetic variation of the ERAP1 gene may be associated with ankylosing spondylitis in the northern Han Chinese population, and the results confirmed ERAP1 is a susceptible gene for ankylosing spondylitis.

Keywords: Ankylosing spondylitis; ERAP1 gene; Association study; Chinese Han

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