

研究报告

吉林人群强直性脊柱炎6号染色体短臂上的HLA区域遗传易感基因定位研究

王雅文^{1, 2}, 朱小泉¹, 宋玉国², 孙亮³, 杨泽¹

1. 北京医院, 北京卫生部老年医学研究所, 北京100730;
2. 北华大学附属医院临床免疫研究室, 吉林 132001;
3. 中国科学院遗传与发育生物学研究所, 北京 100101

收稿日期 2006-10-30 修回日期 2007-5-21 网络版发布日期 2007-7-11 接受日期

摘要

为了寻找中国人群中与强直性脊柱炎相关的新的易感基因及其所在位置, 在与强直性脊柱炎强连锁的6号染色体短臂上的HLA基因区域内选取11个SNPs多态位点, 通过对中国吉林地区79名AS患者和132名正常对照者进行case-control分析, 发现TNF-a -850处TT突变基因型在AS组中的分布高于正常对照组 (P=0.027), 突变型T等位基因在AS组和正常对照组中的分布差异更为显著 (P=0.002)。通过多位点之间的连锁不平衡分析发现, LTA基因、TNF-a基因、LST1基因和NCR3基因中的5个SNPs多态位点之间存在连锁不平衡, 范围是15 kb, 在这5个SNPs多态位点组成的单体型中, TCTTC单体型在AS组和正常对照组中的分布有显著差异 (c2=7.406, P=0.0065), 并且该单体型中含有具有统计学意义的TNF-a -850的突变型等位基因T。提示在LTA、TNF-a、NCR3和LST1这4个基因构成的15 kb范围内可能存在增加AS患病易感性的位点, 可能是TNF-a -850 C→T突变, 也可能是在TNF-a -850附近的其他位点。

关键词 [强直性脊柱炎](#) [易感基因](#) [基因型频率](#) [连锁不平衡](#)

分类号

Mapping study of ankylosing spondylitis on HLA region in Jilin population of China

WANG Ya-Wen¹, ZHU Xiao-Quan¹, SONG Yu-Guo², SUN Liang³, YANG Ze¹

1. Institute of Geriatrics, Ministry of Health, Beijing Hospital, Beijing 100730, China;
2. Department of Clinic Immune, the Affiliated Hospital of Beihua University, Jilin 132001, China;
3. Institute of Genetics and Developmental Biology, Chinese Academy of Sciences, Beijing 100101, China

Abstract

<P>To find the susceptible genes of ankylosing spondylitis in Chinese population, we select 11 SNPs on gene HLA gene family that has strong linkage of ankylosing spondylitis in 6p21.3. By case-control study in 79 AS patients and 132 healthy subjects, the distribution of TNF-a -850 genotype TT is higher in AS group than that in normal control group (P=0.027); Mutational allele T has a significant statistically difference between AS group and normal control group (P=0.002). By linkage disequilibrium study, there are 5 SNPs present the linkage disequilibrium and the region is 15 kb, including gene LTA, TNF-a, LST1 and NCR3; In the haplotypes of the 5 SNPs, the distribution of haplotype TCTTC has statistical difference between AS group and normal control group (c2=7.406, P=0.0065), the haplotype contains mutational allele T of TNF-a -850. The result hints that there may be susceptible sites of AS in this 15 kb region, which may be TNF-a -850 C→T mutation or other sites that around the TNF-a -850.</P>

Key words [ankylosing spondylitis](#) [susceptible gene](#) [the frequency of genotype](#) [linkage disequilibrium](#)

扩展功能	
本文信息	
▶ Supporting info	
▶ PDF(0KB)	
▶ [HTML全文](0KB)	
▶ 参考文献	
服务与反馈	
▶ 把本文推荐给朋友	
▶ 加入我的书架	
▶ 加入引用管理器	
▶ 复制索引	
▶ Email Alert	
▶ 文章反馈	
▶ 浏览反馈信息	
相关信息	
▶ 本刊中 包含“强直性脊柱炎”的 相关文章	
▶ 本文作者相关文章	
· 王雅文	
· 朱小泉	
· 宋玉国	
· 孙亮	
· 杨泽	

通讯作者 王雅文 yawenjin@sohu.com