

强直性脊柱炎的新易感基因识别研究 Recognition and study of susceptible gene to ankylosing spondylitis

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摘要

为了研究中国人群中TNF α 基因与强直性脊柱炎(ankylosing spondylitis, AS)病理发生的潜在关系,我们通过对中国南方75名AS患者的TNF α 基因启动子进行扫描分析,发现-850处突变型T等位基因出现频率较高(39.3%)。经Case-Control研究发现TT突变基因型在AS组中的分布显著高于对照组(10.7% vs 2.1%, P=0.003);突变型T等位基因携带者在AS组与对照组间分布差异极其显著(68.0% vs 21.4%, P=7.928 \times 10⁻¹³)。按性别分组后,发现TX基因型和非TX基因型在AS组和对照组之间的分布差异同样具有统计学意义(男性:P=1.029 \times 10⁻¹⁰;女性:P=0.001),此多态位点在男性和女性中都存在显著性关联。经文献查新未见本突变位点在国内外有与AS存在相关的报道。本研究证实了我们的研究假设,TNF α 基因启动子-850C \rightarrow T的突变可能是AS发生的新易感基因。

Abstract: To study the potential correlations between variances of TNF α gene and onset of ankylosing spondylitis in Chinese population, We scanned and analyzed the promoters of TNF α genes in 75 AS patients from south of China and found -850 T mutation allele frequency rather high (39.3%). By case-control study, the distribution of TT genotype is significantly higher in AS patients than that in normal subjects (10.7% VS 2.1%, P=0.003); Mutation T allele has a remarkable difference between AS group and normal control (68.0% vs 21.4%, P=7.928 \times 10⁻¹³). The difference in distribution of TX genotype and non -TX genotype is also significant statistically between different genders (male: P=1.029 \times 10⁻¹⁰; female: P=0.001). The result suggests that this variation has a strong association with AS in males and females. No similar reports about the association between AS and the T mutation allele have been acquired. Therefore, our hypothesis can be supported by our results on the whole and the -850C \rightarrow T mutation allele in the region on promoter of TNF α gene is likely one of susceptible genes to AS.

关键词 [易感基因](#) [强直性脊柱炎](#) [TNF \$\alpha\$ 启动子](#) Key words [susceptible genes](#) [ankylosing spondylitis](#) [promoter of TNF \$\alpha\$](#)

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