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论文

轻度认知功能障碍与neprilysin基因单核苷酸多态性的相关性

徐明明，易咏红

广州医学院第二附属医院神经科学研究所， 广州 510260

摘要：

目的 分析轻度认知功能障碍（MCI）患者脑啡肽酶基因（NEP）rs3736187位点单核苷酸多态性，为MCI的防治提供理论依据。方法 参照美国精神病学会的精神障碍诊断和统计手册第4版（DSM-IV）的MCI诊断标准，应用聚合酶链反应-限制性片段长度多态性技术检测NEP基因多态性，采用病例-对照的关联分析方法，对NEP基因rs3736187位点进行基因型和等位基因频率分析。结果 NEP基因型频率和等位基因频率分布，MCI组与对照组间差异显著(P＜0.05)，T等位基因携带者出现MCI的危险性高于C等位基因携带者(OR=2.212, P＜0.05)，NEP基因基因型频率和等位基因频率分布，男性之间和女性之间差异不明显，女性MCI组与对照组间等位基因频率分布差异显著（P＜0.05）。结论 NEP基因的T等位基因可能是MCI的危险因素之一，在女性MCI的发病中可能起重要作用。

关键词： 认知障碍；多态性，单核苷酸；脑啡肽酶基因

Association between mild cognitive impairment and neprilysin gene rs3736187 polymorphism

XU Mingming， YI Yonghong

Department of Neurology， the Second Affiliated Hospital and Neuroscience Institute of Guangzhou Medical College， Guangzhou 510260， China

Abstract:

Objective To study the different distribution of the neprilysin (NEP) gene rs3736187 polymorphisms in patients with mild cognitive impairment (MCI) and normal healthy people. Methods 120 patients with MCI and 120 normal controls were examined with the neuropsychological test which included the mini-mental state examination (MMSE), activities of daily living scale (ADL), global deterioration scale(GDS) and Hachinski ischemic scale(HIS). NEP gene polymorphism was analyzed by PCR restriction fragment length polymorphism technique. Case-control analysis was adopted to analyze frequencies of the genotype and allele. Results (1) Distribution of genotypes and alleles of the NEP gene had significant differences between the MCI group and the control group. Frequency of the T allele in the MCI group was higher than that in the normal controls (P＜0.05). (2) People carrying the T allele had a higher incidence of MCI than people with the C allele (OR=2.212, P＜0.05). (3) The frequency of the T allele in female patients was higher than that in female controls (P＜0.05). Conclusion The T allele of NEP may be one of the risk factors of MCI, and may play a significant role in the incidence of MCI in females.

Keywords: Cognition disorders; Polymorphism， single nucleotide； Neprilysin gene

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通讯作者: 易咏红（1962- ），女，博士，教授，主要从事神经系统遗传性疾病及突触可塑性的研究。

作者简介: 徐明明（1981- ），男，博士研究生，主要从事脆性X综合征的研究。 Email：xmm7364@126.com

作者Email:

参考文献：

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