

# ACE基因I/D多态与冠心病的遗传流行病学研究——表型不一致同胞对分析和传递不平衡检验 The Insertion/Deletion Polymorphism of Angiotensin I Converting Enzyme Gene and Coronary Heart Disease——Discordant Sib Pair Analysis and Transmission/Disequilibrium Test

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**摘要** 用表型不一致同胞对分析(DSP)和传递不平衡检验(TDT), 在冠心病家系中探讨血管紧张素转换酶(ACE)基因内含子16中的插入/缺失(I/D)多态是否为冠心病的遗传易患因素。方法: 1998年10月~1999年2月期间收集先证者一级亲属中至少有1例冠心病患者的家系45个, 其中完整核心家系、父母一方、双方基因型缺失家系分别为21、2与22个, 调查对象212人。PCR-RFLP方法鉴定ACE基因I/D多态性基因座基因型。条件Logistic回归进行DSP分析, TDT-STDT 1.1程序进行TDT、STDT检验。结果表明, 45个冠心病家系共组成106对DSP, 单变量条件Logistic回归及调整传统危险因素后的多变量条件logistic回归均未发现II、ID和DD基因型在表型不一致同胞对中的分布存在差别。对13个满足要求的核心家系进行TDT检验, 杂合子父母传递给患病子代的D等位基因频率未显著偏离50% (P>0.05); 24个满足要求的同胞组进行STDT检验亦未发现受累子代与非受累子代D等位基因分布有显著差异 (P>0.05)。结论: 在冠心病家系中未发现ACE基因I/D多态与冠心病存在关联或与疾病基因座存在连锁, 说明该基因座可能不是国人冠心病的遗传易患基因。

**Abstract:** To investigate whether the insertion/deletion polymorphism of the human angiotensin I converting enzyme gene increased the risk of coronary heart disease (CHD) in CHD pedigrees, discordant sib pair analysis (DSP) and transmission/disequilibrium test (TDT) were used. Forty-five CHD pedigrees with at least one CHD patient in the first degree relatives of probands were recruited during Oct. 1998 to Feb. 1999, of which parental genotype known, one or both parental genotype missing was 21, 2 and 22 respectively. ACE genotype was measured by PCR technique. Conditional Logistic regression was used to analyze the DSP, and TDT-STDT program 1.1 was used for TDT and STDT. Univariable conditional Logistic regression did not find significant difference of the distribution of three different ACE genotypes in the 106 discordant sib pairs obtained from the 45 pedigrees. After adjusting effects of traditional risk factors of CHD, no significant difference of the distribution was found by multiple Logistic regression model. Neither the TDT for 13 nuclear families or STDT (sib transmission/disequilibrium test) for 24 sibships showed significant difference between the transmitted and untransmitted ACE gene D allele distributions. Our results show that the insertion/deletion polymorphism of ACE gene is not associated or linked with CHD in Chinese population.

**关键词** [冠心病](#) [遗传易感性](#) [血管紧张素转换酶基因](#) **Key words** [coronary heart disease](#) [genetic susceptibility](#) [ACE gene](#)

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## Abstract

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