

人类与医学遗传学

PON基因簇潜在功能多态位点与冠心病的关联研究

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摘要 在中国汉族人群PON基因簇序列筛查研究基础上, 系统探讨PON基因簇所有潜在功能多态位点与国人冠心病的关系, 以期明确PON基因簇序列变异是否国人冠心病的遗传危险因素。随机入选1997~1999年期间阜外心血管病医院病房收治的经冠状动脉造影确诊和/或有明确急性心肌梗塞病史男性冠心病患者474例及年龄(±2岁)匹配的男性健康对照475例。PCR产物直接测序法鉴定PON1基因-1076A/G、-908G/C、-831 G/A、-162G/A、-126 G/C和-107C/T多态基因型; 等位基因特异性扩增方法鉴定PON2基因的A148G和S311C多态; PCR-RFLP方法鉴定PON1基因R160G、Q192R和PON3基因-133C/A多态。单变量分析显示192Q, 160R, -162A和311C等位基因频率在病例组中显著高于对照组。以这四个多态性位点作为自变量的多元Logistic回归分析发现仅R160G和-162G/A多态仍然与冠心病显著关联(P值分别为0.0054和0.0002), 并独立于冠心病传统危险因素。不同多态组合的单体型分析进一步证实了单一SNP分析的结果, 只有包含160R或-162A等位基因的单体型在病例组中的频率显著高于对照组。中国北方汉族人群中, PON1基因-162G/A 和R160G多态与冠心病独立关联, 提示PON1基因可能是冠心病易感基因。

关键词 [冠心病; 遗传; 对硝苯磷酸酯酶; 连锁不平衡; 低密度脂蛋白氧化](#)

分类号

Association Analysis Between Polymorphisms of PON Gene Cluster with Coronary Heart Disease in Chinese

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Abstract

An extensive association analysis of PON gene cluster (PONs) with coronary heart disease (CHD) was performed in Chinese Han population. Eleven polymorphisms of PON1, PON2 and PON3 gene were investigated for association with CHD in 474 male patients and 475 controls. Univariate analyses showed the cases had significantly higher frequencies of PON1 192Q allele, 160R allele, -162A allele and PON2 311C allele than were seen in the controls. Logistic regression analyses revealed only the PON1 R160G and -162G/A polymorphisms remained significantly associated with CHD (P = 0.0054 and P = 0.0002). Haplotype analyses for various polymorphism combinations further confirmed the results of individual polymorphism analyses. Only the frequencies of haplotypes containing -162A allele were significantly higher, whereas only the frequencies of haplotypes containing 160G allele significantly lower in cases than those in controls in various polymorphism combinations. This extensive association study has identified the PON1 -162G/A and R160G polymorphisms to be independently associated with CHD in

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Chinese Han population, and warrants further study to elucidate the biological mechanism.

Key words [coronary heart disease](#) [genetics](#) [paraoxonase](#) [linkage disequilibrium](#) [LDL oxidation](#)

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