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# AGT基因单倍型与原发性高血压

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摘要 选取血管紧张素原（angiotensinogen, AGT）基因启动子区 -217, -152, -20, -6, 内含子1 的+31, 第二外显子T174M（3889）和 T235M（4072）共7个位点，对497例的样本（高血压患者298例，血压正常对照199例）用PCR-RFLP、和最大期望值（expectation maximization，EM）算法为基础的最大似然法(maximum likelihood estimate，MLE)检测和估算，本群体AGT基因A-6G，C+31T，T235M三位点两两存在完全连锁不平衡(D′=1)；G-217A和G-152A位点，G-152A和3889T位点平衡传递。存在7种单倍型，单倍型H2(-217: A, -152: G, -20: A, -6: G, +31: T, 174: T, 235: M) 在正常血压个体中的频率高于高血压组。研究结果提示AGT基因中H2单倍型可能与控制血压的保护性因素连锁不平衡。此外，本研究结果支持基因剂量效应可能存在于单倍型中，而不与单个位点直接关联。

关键词 血管紧张素原 连锁不平衡 单倍型 限制性片段长度多态性

分类号



## Haplotypes Extending Across AGT are Associated Essential Hypertension

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**Abstract**

Present studies examined the DNA polymorphisms in the AGT genes in a Chinese population in Henan province of central China. By using PCR-RFLP and maximum likelihood estimation (MLE), we estimated the pattern of intragenic linkage disequilibrium and the haplotype structure and explored the possible association between the polymorphisms of AGT gene and essential hypertension in a case-control study. Seven polymorphic sites (SNPs) and seven major haplotypes of AGT gene were analyzed. Among the individual SNP pairs examined, the A-6G, C+31T and M235T are nearly completely disequilibrium. All those single polymorphism loci were individually not associated with hypertension. But we found the frequency of haplotype H2 (-217: G, -152: G, -20: A, -6: G, +31: T, 174: T, 235: M) was significantly higher in controls than patients (P=0.010). Our study suggested that few haplotypes derived seven polymorphism loci could account for the most of the variation in AGT gene in Chinese Hans. The haplotype H2 of AGT gene might represent or be in disequilibrium with a genetic protective factor against EH. <br>

**Key words** Rennin angiotensinogen system linkage disequilibrium haplotype restriction fragment length polymorphism

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