

论著

我国汉族人群 $\alpha 1$ 基因数目可变串联重复序列分布特征的研究

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摘要 目的: 研究我国汉族人群免疫球蛋白 $\alpha 1$ 基因中3个数目可变串联重复序列(VNTR)多态性分布特征, 及其与已报道的高加索人群相比较的特点。方法: 从现存数据库中寻找 $\alpha 1$ 基因内的3个VNTR位点, 即 $\alpha 1$ 基因3'端的hs1,2增强子内的VNTR1、其上游6 Kb 的VNTR2和位于 $\alpha 1$ 基因第5外显子的E5VNTR。提取201例广东汉族人基因组DNA, PCR分别扩增含上述3个VNTR位点片段, 2%—3%凝胶电泳分带鉴定基因型, 并以测序证实。结果: 与高加索人群比较我国汉族人群 $\alpha 1$ 基因 VNTR1多态性分布特征表现为: C (3次重复) 等位基因频率明显升高 (10.0% vs 1.0%), A (1次重复) 等位基因频率偏低 (30.3% vs 36.1%-39.4%), 差异显著 ($\chi^2=72.85$, $P<0.01$)。基因型BB占37.8%, AB占32.3%, AA占12%, BC占11.4%, AC占4.5%, CC占2.0%, BC、AC基因型分布频率显著高于高加索人群, 而AB型分布频率显著低于高加索人群 ($\chi^2=73.77$, $P<0.01$)。另外两个数据库中报道的VNTR位点 (VNTR 2及E5VNTR) 在我们所测人群中呈均一分布, PCR产物长度分别为136 bp (VNTR 2) 和535 bp (E5VNTR)。结论: 我国汉族人群 $\alpha 1$ 基因 VNTR多态性分布与基因组数据库中基于高加索人群的资料不尽相同, 其中Ia1 hs1,2 VNTR1多态性不同于高加索人群, 突出表现为C等位基因频率及BC、AC基因型频率显著高于高加索人群。而VNTR2和E5VNTR在被检人群中未见多态性。本研究弥补了现存数据库中缺乏我国汉族人群相应数据的不足, 同时为以 $\alpha 1$ 基因为候选基因找寻疾病基因的研究提供了可靠的数据。

关键词 [免疫球蛋白A](#); [可变数量串联重复](#); [多态现象\(遗传学\)](#); [中国人](#)

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Distribution of $\alpha 1$ immunoglobulin gene VNTR polymorphisms in Chinese

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Abstract

AIM: To investigate the polymorphisms of the variable number tandem repeat (VNTR) in the $\alpha 1$ immunoglobulin gene in the Chinese population and to compare them with the Caucasians. METHODS: 3 VNTR loci (VNTR1: $\alpha 1$ gene hs1,2 enhancer VNTR; VNTR2: 6 kb forward (change to upstream or down stream) the VNTR1 and E5VNTR: located in the fifth exon) and their lengths in the $\alpha 1$ gene have been retrieved from the genetic databases and literature. Genomic DNA was extracted from 201 healthy Chinese Han subjects. The sizes of the 3 VNTRs were determined by polymerase chain reaction (PCR) and gel electrophoresis, and confirmed by sequencing randomly selected samples. RESULTS: The VNTR1 locus revealed three different lengthed alleles, designated as $\alpha 1A$, $\alpha 1B$ and $\alpha 1C$ for one, two and three repeat sequences, respectively, with the frequencies of 30.3%, 59.7% and 10.0%, respectively. 6 genotypes were formed from the alleles with the frequencies of 12% for the AA; 32.3% for the AB; 37.8% for the BB; 4.5% for the AC; 11.4% for the BC; and 2.0% for the CC, respectively. Compared with the reported Caucasian population the frequencies of the C allele and the BC、CC、AC genotypes were significantly higher ($P<0.01$), the A allele and the AB genotype frequencies were significantly lower ($P<0.01$). All the examined subjects showed the uniformed lengths of 136 bp for the VNTR2 and 535 bp for E5VNTR alleles. CONCLUSIONS: The repeat numbers of the VNTR1 of the $\alpha 1$ gene in the Chinese Han population are significantly different from the Caucasians with a higher C allele frequency and BC、AC genotypes, and lower A allele frequency and

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AB genotype. We could not find evidence of polymorphism in the VNTR2 and E5VNTR loci in the examined subjects. The results represent the first data from the Chinese population regarding the VNTR polymorphisms in the I alpha 1 gene, and provide a useful tool for the gene and the gene related studies.

Key words [Immunoglobulin A](#); [Variable numbers of tandem repeats](#); [Polymorphism \(genetics\)](#); [Chinese](#)

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