

中国汉族群体vWF基因40内含子VNTR

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收稿日期 修回日期 网络版发布日期 接受日期

摘要 本文将Amp-FLP与RFLP结合起来, 检测vWF基因40内含子VNTR(variable number of tandem repeats)。调查长沙地区156名无亲缘关系汉族人, 发现v WF基因40内含子VNTR基因136个。基因型153个, 全部为杂合子, 分布符合Hardy-Weinberg平衡定律。此VNTR的杂合率为0.9884, PIC为0.9883, 是现已检出Amp-FLP中多态性最高的遗传标记。家系分析显示VNTR无遗传重组, 依照常染色体共显性遗传。用高分辨聚丙烯酰胺凝胶电解质梯度电泳, 分离扩增的杂合子个体vWF基因40内含子VNTR两条DNA, 回收后再次扩增, AluI消化, 电泳直接检测了VNTR基因型, 解决了Amp-FLP-RFLP分析中无法判定基因的难题。首次将SSCP应用于微卫星DNA亚型的检测, 检出一个VNTR亚型。

关键词 [vWF基因](#) [可变数量串联重复](#) [单链构型多态性](#)

分类号

Investigation on VNTR in Intron 40 of vWF Gene in Chinese Han Population

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Abstract

The VNTR in the intron 40 of vWF gene was investigated with a method of combination of Amp-FLP and RFLP. Two amplified fragments from a heterozygous person were separated by a non-hydratable polyacrylamide electrolyte gradient electrophoresis. After elution from gel, the preliminary products were amplified again, the amplified fragments were digested with AluI, then the genotype could be directly determined through electrophoresis. For the first time SSCP was used to detect the subtypes of microsatellite DNA. 136 alleles were found from 156 unrelated Hans in Changsha area. There were 153 genotypes in this sample and all of them were heterozygous. The calculated heterozygosity was 0.9884. The distribution of genotypes was in accordance with the Hardy-Weinberg equilibrium. The polymorphic information content (PIC) was 0.9883. The polymorphism of this VNTR was the highest one in the Amp-FLPs that had been found before. Our data did not show any recombination event in 2 generations of 66 families and 3 generations of 5 families. Aco-dominant segregation was demonstrated.

Key words [vWF gene VNTR \(variable number of tandem repeats\)](#) [SSCP \(single-strand conformation polymorphism\)](#)

DOI:

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