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9号染色体短臂上7个STR基因座在基因扫描中的信息表现

Information Behavior of 7 STR Loci on Chromosome 9p in Gene Scanning

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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 为了初步探讨7个位于染色体9p区域的短串联重复序列(short tandem repeat, STR)基因座: D9S288、D9S157、D9S1748、D9S171、D9S161、D9S1817和D9S1805在遗传学研究及法医学应用中的意义, 随机抽取225名湖南汉族无关个体, 复合PCR技术扩增上述基因座, ABI 377全自动测序仪进行基因分型, 共检出75种等位基因, 通过对基因型及等位片断频率分布的研究和数据统计分析, 7个基因座基因频率分布在0.002~0.800之间, 构成243种基因型。7个STR基因座基因型分布均符合Hardy-Weinberg平衡定律($P>0.05$), 杂合度(heterozygosity, H)介于0.347~0.844之间, 个体识别力(discernment power, DP)为0.346~0.841, 非父排除率(probabilities of paternity exclusion, PPE)为0.308~0.738, 多态信息含量(polymorphic information content, PIC)在0.328~0.822之间。种族比较结果显示, 湖南汉族与非洲黑人及欧洲白人在大多数基因座均存在显著差异($P<0.001$)。研究结果丰富了中华民族基因数据库, 在人类群体遗传学及法医学研究领域有重要应用价值。

Abstract: To get genotype and allele frequency distributions of seven short tandem repeat (STR) loci of chromosome 9p, D9S288, D9S157, D9S1748, D9S171, D9S161, D9S1817 and D9S1805 in Chinese Han population in Hunan area, blood samples were collected from the random Han individual in Hunan and the whole genomic DNA was extracted. STR loci were amplified by multiplex-PCR technique and genotyped by ABI 377 sequencer. Seventy-five alleles were detected, with frequencies ranging from 0.002 to 0.800, and constituted 243 genotypes. All the seven loci met Hardy-Weinberg equilibrium. The statistical analysis of seven STR loci showed H(heterozygosity) ranging from 0.347 to 0.844, DP (discernment power) ranging from 0.346 to 0.841, PPE(probabilities of paternity exclusion) ranging from 0.308 to 0.738 and PIC(polymorphic information content) ranging from 0.328 to 0.822. The result indicated that there was a significant difference between Han ethnic group and the white and the black.

关键词 [汉族](#) [STR基因座](#) [遗传多态性](#) [基因扫描](#) Key words [Han ethnic group](#) [STR](#) [genetic polymorphism](#) [genescan](#)

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