

中国南方汉族人群6个Y-STR基因座遗传多态性及法医学应用 Genetic Polymorphisms of 6 Y-Chromosome Specific STR Loci in the southern Chinese Han Population and Its Application in Forensic Science

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摘要

为研究中国南方汉族人群DYS393等6个Y-STR基因座的遗传多态性并用于法医学鉴定, 通过采用PCR复合扩增和基因测序仪荧光检测方法, 检查204个无关男性个体, 调查南方汉族的6个Y-STR基因座的单倍型频率, 并对93对真父子和38对非父子的亲子鉴定样本进行检测。结果DYS393基因座检出5个等位基因, DYS19基因座检出6个等位基因, DYS389 II 基因座检出8个等位基因, DYS390基因座检出6个等位基因, DYS391基因座检出4个等位基因, DYS385 基因座检出44个等位基因, 共检出176种单倍型。93对真父子中, 观察到2例分别有1个基因座突变。检测38对非父子, 有1个或2个Y-STR基因座排除的案例各有1例 (2.6%); 有3个和3个以上的Y-STR基因座可以排除父子关系的案例为35例 (92.1%); 6个Y-STR基因座不能排除父子关系的为1例。结果表明6个Y-STR基因座具有丰富的遗传多态性, 可用于法医学个体识别和亲子鉴定。Abstract: To study the genetic polymorphisms of six Y-chromosome specific STR loci in the southern Chinese Han population and apply it in forensic science, six Y-STR loci were amplified by multiple PCR and the PCR products were detected by using ABI Prism™ 377 Sequencer. The haplotype frequencies at 6 Y-STR loci were determined in a total of 204 unrelated males from southern Han population of China. Ninety-three father/son pairs with demonstrated paternity and thirty-eight non-paternity father/son pairs were detected by using our Y-STR system. As a result, the number of alleles for DYS393, DYS19, DYS389 II, DYS390, DYS391 and DYS385 were 5, 6, 8, 6, 4 and 44, respectively. A total of 176 haplotypes at 6 Y-STR loci were found. Two father/son pairs with single Y-STR mutation were observed in the 93 father/son pairs with demonstrated paternity. Among the 38 non-paternity father/son pairs, one case with one Y-STR exclusion of paternity, one case with two Y-STR exclusions and 35 cases with 3 or more Y-STR exclusions were observed. Non-exclusion of paternity at 6 Y-STR loci was found only in one case. This result indicated that the six Y-STR loci were highly polymorphic and are suitable for personal identification and paternity testing.

关键词 [Y染色体](#) [短串联重复序列 \(STR\)](#) [单倍型](#) [遗传多态性](#) Key words [Y-chromosome](#) [short tandem repeat \(STR\)](#) [haplotype](#) [genetic polymorphism](#)

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