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Evaluation of seven autosomal STR loci in a German population

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Abstract. The autosomal tetranucleotide short tandem repeat loci D4S2366, D6S474, D14S608, D19S246, D20S480, D21S226, and D22S689 were evaluated with regard to their use for forensic applications. Allele frequencies for these loci were investigated in a sample of 189 unrelated German individuals. The loci showed no significant deviations from Hardy-Weinberg equilibrium except for D14S608. All genotyped alleles were cloned, sequenced and an allelic nomenclature consistent with the ISFG recommendations was defined. © 2005 Elsevier B.V. All rights reserved.

Keywords: DNA typing; Short tandem repeats; Population study

1. Introduction

Polymorphic short tandem repeats (STR) are powerful genetic markers to distinguish individuals and still preferred for forensic applications and paternity testing. Evaluations of new STR markers or primer redesign for conventional STR systems are useful tools to obtain additional information and to complement conventional STR analysis [1]. There is a growing interest in polymorphic markers unlinked to the current CODIS loci [1]. Multiplex PCR based on tetranucleotide STRs allows fast and reliable genotyping. Here we report biostatistical data and the variable repeat sequence motifs of seven autosomal loci, which were analyzed in a German population using two multiplex PCR systems.

2. Materials and methods

Blood samples and buccal swab samples from 189 healthy unrelated German individuals from the area around were collected for DNA extraction with written informed

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Table 1
Chromosomal location and accession number for all STR loci from this study

Locus	Chromosomal location	Accession number
D4S2366	4p16-15.2	G08339
D6S474	6q21-22	G08540
D14S608	14q11.1-11.2	G09052
D19S246	19q13.3-13.4	L13118
D20S480	20q	G08049
D21S226	21q22.1	M93147
D22S689	22q11.2-12.1	G08087

consent. Two screening multiplexes with fluorescently labeled primers were developed and used for population study.

The PCR products were analyzed by capillary electrophoresis using an ABI PRISM 310 Genetic Analyzer. The STR genotypes of the population were analyzed by calculation the observed heterozygosity (HET), the polymorphic information content (PIC), the power of discrimination (PD), the mean exclusion chance (MEC) and the deviation from the Hardy-Weinberg equilibrium (HWE) based on χ^2-test.

Furthermore, all different alleles for the loci were cloned into a plasmid vector. Sequencing of both strands of the DNA inserts was performed using a commercial kit. An allelic nomenclature consistent with the ISFG recommendations was defined based on the variable repeat motifs [2].

3. Results and discussion

Chromosomal location and accession number for all loci are given in Table 1. Allele frequencies of unrelated individuals from the area around Dresden were investigated in two screening

Table 2
Repeat sequences of different alleles for all loci

Locus	Alleles found	Repeat structure
Simple repeats		
D14S608	4–15	$(TCTA)_{4–15}$
Simple repeats with sequence variants		
D21S226	7–11	$(TAAA)_{7,8,11}$
		$(TAAA)_8(TAGA)/(TAAA)_9CAAA$
Compound repeat sequences		
D4S2366	9–15	$(ATAG)_{5–11}(ATTG)_{2–3}(ATAG)_{1–2}$
D6S474	13–18	$(TAGA)_5TGA(TAGA)_{8–13}$
D20S480	12–19	$(TATC)_{10–17}ATC(TATC)_2$
D22S689	12–20	$(TAGA)_{8–16}(CAGA)_3(TAGA)_{0–1}$
Complex repeat sequences		
D19S246	27–39	
	39	$(ATAG)_4ATAC(ATAG)_{13}(ATAC)_{10}ACAGATAC$
		$(ATAG)_3ACAG(ATAG)_2AT(TAGA)_3$

Table 3
Biostatistical parameters for the seven STR loci

Parameters	D4S2366	D6S474	D14S608	D19S246	D20S480	D21S226	D22S689
PIC	0.760	0.740	0.800	0.820	0.740	0.530	0.680
HET	0.795	0.737	0.820	0.857	0.815	0.492	0.735
PD	0.919	0.918	0.938	0.951	0.901	0.780	0.877
MEC	0.758	0.745	0.758	0.825	0.739	0.535	0.678
HWE (p-value)	0.867	0.997	0.000	0.997	0.986	0.229	0.979

multiplexes. A triplex assay for D4S2366, D6S474 and D14S608 was developed whereas alleles for D19S246, D20S480, D21S226 and D22S689 were amplified in a quadruplex assay. The repeat motifs of the seven new STRs could be divided into simple, compound and complex repeat sequences (Table 2). There was no evidence for incomplete repeat motifs. D19S246 showed a complex repeat sequence with variable and non-variable tetranucleotide repeats as well as a conserved AT motif as indicated for allele 39 (Table 2).

The allele frequencies for all loci were determined and the population data were statistically analyzed. The PD values of the STR loci ranged from 0.78 for D21S226 to 0.951 for D19S246 (Table 3).

We found significant deviation from HWE for D14S608. Deviations from HWE can point either to mistyping genotypes, sampling bias or natural selection. The primer pair, which was used in PCR for D14S608, gave amplicons shorter than 230 bp and all peaks could be clearly assigned in capillary electrophoresis. So far, we can exclude mistyping errors.

Due to high degrees of heterozygosity (Table 3) and high discrimination power most of the 7 STR markers except for D21S226 can be efficiently used for kinship analyzes or forensic purposes in German populations. All markers except for D21S226 and D19S246 are located on different chromosomes than or separated by at least 50 cM from STR loci, which are currently used in STR databases. Consequently, these markers are probably unlinked to common STRs and useful to obtain additional information in STR analysis for forensic casework and paternity testing.

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