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SHORT COMMUNICATION

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Frequency data on the loci vWA, FES/FPS, F13A01, TH01, TPOX and CSF1P0 in a population from South Poland

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Abstract Allele and genotype frequencies for six short tandem repeat (STR) loci were determined in a sample of 124 inhabitants from South Poland with commercial PCR-based typing kits. No deviations from Hardy-Weinberg expectations were found. The combined power of discrimination for the six loci was 0.9999982. There was no genotypic disequilibrium between the loci except for vWA and F13A01. The set of PCR loci was validated as useful for paternity testing and individual identification in the Polish population.

Key words PCR · Short tandem repeats · Population genetics · Poland

Introduction

Short tandem repeat (STR) loci are polymorphic markers widely used for forensic identification and paternity testing. In recent years, allelic data for STR loci have been published from many different populations (Asmundo and Crino 1998; Crespillo et al. 1997; Furedi et al. 1996; Garofano et al. 1998; Halos et al. 1998; Hochmeister et al. 1995; Pinheiro et al. 1997). This paper presents allele frequencies for the STR loci vWA, FESFPS, F13A01, TH01, TPOX and CSF1P0.

Materials and methods

Blood samples were obtained from 124 unrelated individuals (62 males and 62 females), inhabitants of South Poland. DNA extraction was done by a standard salting out procedure (Miller et al. 1988). PCR amplification of the loci was done using two triplex reactions: vWA, FES/FPS, F13A01 and TH01, TPOX, CSF1P0. The conditions used were as recommended by the manufacturer (Promega, Madison, Wisc.) using denaturing PAGE on vertical gels (SA32, BRL, USA) and premixed 6% acrylamide/bisacrylamide with 6 M urea (GenePlus, Amresco, USA) in 1TBE buffer. Samples were denatured before electrophoresis in a formamide loading buffer included in the kits. The migration distance was 18 cm using constant power (35 W). Electrophoresis was stopped when the bromophenol blue dye front had reached the bottom of the gel plate. The amplified fragments were silver stained in the gel (Budowle et al. 1991). The results were scored in comparison with the allelic ladder supplied in the kits. Reproducible resolution was observed and scoring done by two independent observers revealed no ambiguous results.

The calculations of heterozygosity, Hardy-Weinberg equilibrium deviations and between loci linkage (Fisher's exact test) were performed using a computer program (Lewis and Zaykin 1997): Genetic Data Analysis computer program for the analysis of allelic data. Version 1.0. Free program distributed by the authors over internet from the GDA Home Page at http://chee.unm.edu/gda/).

Results and discussion

Allele frequencies for the six STR loci in the Polish population sample are shown in Table 1. The six systems showed heterozygosities ranging from 83.1% for vWA to 57.7% for TPOX. There were no significant deviations from the genetic equilibrium expectations in these systems (Table 2). The power of discrimination of the systems ranged from 0.812 for TPOX to 0.938 for vWA, while the polymorphism information content (PIC) was between 0.577 and 0.785 for the same loci. The probabilities predicting the usefulness of analysed systems in forensic studies are presented in Table 2. The combined power of discrimination for the system set was 0.9999982. Pairs of the loci were tested for genetic linkage (Table 3). When tested with Fisher's exact test, the linkage hypothesis could not be rejected for the pair vWA-F13A01 at $p = 0.026$. Expected heterozygosity of F13A01, calculated either as unbiased estimate or maximal likelihood estimate was significantly (over 3 standard deviations) less than observed. The distribution of alleles for F13A01, in which low frequency of larger alleles (14–16 repeats) composing

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Table 1 Allele frequencies for six STR loci in Poland

Allele	F13A01	vWA	TH01	FESFPS	TPOX	CSF1P0
3.2	0.069					
4	0.077					
5	0.169		0.008			
6	0.278		0.270			
7	0.371		0.105			
8			0.117	0.028	0.556	0.008
9			0.206	0.008	0.125	0.032
9.3			0.278			
10			0.016	0.325	0.081	0.355
11				0.411	0.214	0.266
12				0.183	0.024	0.262
13		0.004		0.045		0.060
14	0.012	0.101				0.004
15	0.020	0.145				0.012
16	0.004	0.153				
17		0.278				
18		0.222				
19		0.089				
20		0.008				

Table 2 Statistical parameters of forensic interest for six STR loci in Poland

Locus	Het.Obs.	Het. expected		St. error of het.	H-W eq.					
		Unbiased	MLE		Exact t.	PM	DP	PE	ME	PIC
vWA	0.831	0.814	0.811	0.009	0.084	0.354	0.938	0.662	0.750	0.785
FESFPS	0.732	0.692	0.689	0.015	0.967	0.333	0.848	0.488	0.402	0.634
F13A01	0.669	0.748	0.745	0.015	0.223	0.348	0.896	0.569	0.481	0.706
TH01	0.750	0.785	0.782	0.009	0.814	0.352	0.919	0.617	0.522	0.748
TPOX	0.577	0.624	0.622	0.026	0.063	0.327	0.812	0.428	0.368	0.577
CSF1P0	0.733	0.733	0.730	0.012	0.358	0.343	0.879	0.541	0.448	0.682
Comb. Loci						0.919506	0.9999982	0.9925	0.98706	

Expected heterozygosity was calculated as an unbiased estimate and as a maximum likelihood estimate. Standard error of heterozygosity estimates were similar for both methods.
Probability of genetic equilibrium for the locus following Hardy-Weinberg formula was calculated as a Fisher's exact test with 10 000 iterations.
PM, probability of random match; DP, discrimination power; PE, power of exclusion of paternity; ME, mean exclusion chance of paternity; PIC, polymorphism information content

Table 3 Linkage between the six loci

Locus	FESFPS	F13A01	TH01	TPOX	CSF1P0
vWA	0.661	0.026	0.228	0.254	0.063
FESFPS		0.617	0.836	0.454	0.905
F13A01			0.283	0.221	0.286
TH01				0.309	0.288
TP0X					0.621

a secondary peak of the bimodal distribution, suggest duplication or recombination as a possible source of the allelic variability. When analysed together with the system of the greatest polymorphism information content vWA, the F13A01 system showed disequlibrium, which can reflect a relatively recent origin of the large alleles. This hypothesis is further supported by less frequent heterozygosity in the F13A01 system. The results of disequilibrium tests for the other systems pairs were statistically insignificant. The set of single locus STR markers analysed proved to be very useful as a discrimination tool in forensic haemogenetics.

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