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SHORT COMMUNICATION

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Basque Country autochthonous population data on 7 short tandem repeat loci

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Abstract Blood samples from 202–208 unrelated Basque Country autochthonous individuals were amplified, typed and their allele frequencies were determined. Results demonstrate the assumption of independence within and between the loci analyzed. Therefore, a Basque population database can be used in identity testing to estimate the frequency of a multiple PCR-based locus DNA profile.

Key words Basque Country · Population database · PCR · Hardy-Weinberg equilibrium · Linkage equilibrium

Introduction

Before a new marker system can be introduced into forensic casework, a population database for the relevant population must be established for statistical evaluation of the evidence. Therefore, this report presents allele frequency data in a Basque Country autochthonous population sample (n = 202–208) for the seven STR loci HUMTH01 (11p15.5), HUMTPOX (2p13), HUMCSF1PO (5q33.5-q34), HUMVWA (12p12-pter), HUMFES/FPS (15q25-qter), HUMF13A01 (6p24-p25) and HUMF13B (1q31-q32.1).

Materials and methods

Whole blood was obtained from unrelated Basque autochthonous donors. Individuals were considered autochthonous if the eight surnames and birthplace of their grandparents were of Basque origin. Genomic DNA was extracted by the standard phenol/chloroform extraction procedure.

The HUMTH01, HUMTPOX, HUMCSF1PO and HUMVWA, HUMF13A01, HUMFES/FPS loci were amplified using two triplex systems. The locus HUMF13B was amplified individually. The reaction assay and the amplification conditions were performed using GenePrint STR Systems (Promega Corporation) according to the manufacturer's recommendations. The PCR products were typed as described previously [1].

Allele designations were made according to recommendations of the DNA Comission of the International Society for Forensic Haemogenetics [2].

Statistical evaluations were performed using a HWE-Analysis software package (C. Puers, Institute of Legal Medicine Münster). Analyses included the possible divergence from Hardy-Weinberg expectations and other parameters of forensic importance i.e. observed and expected heterozygosities [3], mean exclusion chance (MEC) [4], mean paternity exclusion probability (MEP) [5], polymorphic information content (PIC) [6] and discrimination power (DP) [8]. The possible associations between loci were tested using the computer program GDA (Genetic Data Analysis) (PO Lewis and D Zaykin). An R × C contingency table was used to test for homogeneity between various population samples.

Results and discussion

The observed allele frequencies for the seven STR loci in the Basque Country autochthonous population sample are shown in Table 1. The results of the different test procedures for testing the correspondence of the genotype frequencies with their HWE proportions are shown in Tables 2 and 3. The genotype frequency distributions for most of the loci show no deviations from HWE expectations based on the χ^2-test, the logarithmic likelihood ratio (G) test and the exact test [8] (in all cases, the data were shuffled 2000 times). HUMCSF1PO departs with the χ^2-test (marked with an asterisk) due to the presence of a 15,15 genotype in the sample, but this is not as meaningful a test for HWE as is the exact test. In addition, the test based on the number of distinct genotypes observed in the sample population shows that the observed numbers of distinct heterozygote and homozygote genotypes [9] are in accordance with their respective HWE predictions (Table 3).

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Table 1 Observed allele frequencies for STR loci

Allele	TH01 (n = 205)	TPOX (n = 206)	CSF1PO (n = 205)	VWA (n = 208)	FES/FPS (n = 208)	F13A01 (n = 208)	F13B (n = 202)
3.2						0.0313	
4						0.0409	
5						0.1755	
6	0.3049					0.3101	0.0718
7	0.0878	0.0024				0.3990	
8	0.1244	0.4927	0.0024			0.0024	0.2228
9	0.1976	0.0898	0.0122		0.0024		0.2401
9.3	0.2805						
10	0.0049	0.0874	0.3171		0.3486		0.4629
11		0.2743	0.2829		0.3029		0.0025
12		0.0534	0.3146		0.2620		
13			0.0585	0.0024	0.0745	0.0048	
14			0.0049	0.1154	0.0096	0.0120	
15			0.0073	0.1202		0.0048	
16				0.2404		0.0072	
17				0.3029		0.0072	
18				0.1587		0.0048	
19				0.0529			
20				0.0072			
Minimum frequency	0.0143	0.0132	0.0138	0.0141	0.0134	0.0133	0.0130

Table 2 HWE tests on the loci analyzed

	THO1	TPOX	CSF1PO	VWA	FES/FPS	F13A01	F13B
χ^2test	0.9520	0.8875	0.0070*	0.8575	0.5725	0.2220	0.2595
G test	0.9635	0.8215	0.1560	0.8265	0.4845	0.3360	0.2530
Exact test	0.9410	0.8220	0.1715	0.8825	0.6970	0.4320	0.3135

Number of random shuffles performed: 2 000

Table 3 Observed and expected numbers of heterozygous and homozygous classes for the PCR-based DNA loci

	Heterozygotes observed	Heterozygotes expected ± SE	Homozygotes observed	Homozygotes expected ± SE
TH01	12	11.58 ± 1.99	5	4.76 ± 0.90
TPOX	10	10.53 ± 1.87	5	4.05 ± 1.48
CSF1PO	13	12.99 ± 3.47	4	3.55 ± 1.07
VWA	18	17.95 ± 2.90	5	5.34 ± 1.19
FES/FPS	9	9.23 ± 0.95	3	3.71 ± 0.95
F13A01	22	21.13 ± 4.96	3	3.54 ± 1.28
F13B	7	6.85 ± 1.55	3	3.65 ± 0.94

Table 4 Statistical parameters of forensic importance

	H_{obs}	H_{exp}[a]	MEC	MEP	PIC	DP
TH01	0.8000	0.7680	0.5454	0.5411	0.7281	0.8999
TPOX	0.6893	0.6651	0.4230	0.3763	0.6149	0.8349
CSF1PO	0.7512	0.7185	0.4603	0.4575	0.6629	0.8579
VWA	0.7981	0.7966	0.5995	0.5928	0.7653	0.9283
FES/FPS	0.7260	0.7142	0.4535	0.4505	0.6577	0.8618
F13A01	0.7115	0.7126	0.4707	0.4479	0.6628	0.8706
F13B	0.6337	0.6750	0.4152	0.3906	0.6183	0.8433

[a] Expected heterozygosity is an unbiased estimate

Minimum allele frequencies for PCR-based loci, based on statistical and population genetics theory [10–12], were determined (Table 1). Thus, a greater confidence of the DNA profile frequency estimates can be attained with current database sizes.

Table 4 shows several statistical parameters of forensic importance, such as expected and observed heterozygosities, mean exclusion chance (MEC), mean paternity exclusion, polymorphic information content (PIC) and discrimination power (DP).

An interclass correlation test analysis demonstrated that there is no evidence for correlation between the alleles at any of the pairs of loci (Table 5). Moreover, correlations made with previous results obtained in the same sample for the HLA-DQA1 locus, the Polymarker set and the D1S80 locus [13, 14] demonstrated (data not shown) that there is little evidence for departures from independence for the sample population and support the view that the use of the product rule would provide a good approx-

Table 5 Two-loci interclass correlation test for the analyzed loci (*P* values)

TH01	–						
TPOX	0.9315	–					
CSF1PO	0.9780	0.9855	–				
VWA	0.5315	0.9930	0.8750	–			
FES/FPS	0.7600	0.8295	0.6335	0.7100	–		
F13A01	0.8910	0.5120	0.3945	0.9300	0.4015	–	
F13B	0.3865	0.5220	0.3070	0.8345	0.9490	0.1165	–
	TH01	TPOX	CSF1PO	VWA	FES/FPS	F13A01	F13B

Number of random shuffles performed: 2000

imation of the estimate of the rarity of a multiple locus profile.

A comparison of the allele frequencies in the population under study with those of previous studies in the Basque population revealed significant differences ($P < 0.001$) with the Basque resident data [15] but no significant differences ($P > 0.05$) with other Basque autochthonous data [16].

In conclusion, a Basque Country population database has been established for the STRs HUMTH01, HUMTPOX, HUMCSF1PO, HUMVWA, HUMFES/FPS, HUMF13A01 and HUMF13B. The combined power of exclusion was estimated as 99.041% and the combined power of discrimination as 99.999953%. These seven STR systems have been shown to be a useful tool for personal identification. The allele frequency data can be used for deriving estimates of multiple locus profile frequencies for identity testing purposes using the product rule.

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