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The codis system in the Basque Country resident population studied with multiplex systems

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Abstract

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 resident population sample ($n > 200$) for 13 STR loci. The loci are: TH01, TPOX, CSF1PO, D3S1358, FGA, VWA, D5S818, D13S317, D7S820, D8S1179, D21S11, D18S51 and D16S539. The combined power of exclusion is estimated as 99.9998% and the combined power of discrimination is >99.99999%. These 13 STR systems have been shown to be 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.

Keywords: Forensic science; DNA typing; Short tandem repeat; Population genetics; Basque Country

1. Materials and methods

Genomic DNA was extracted by standard phenol-chloroform extraction procedures. PCR amplification was performed according to the manufacturer's recommendations using the AmpFlSTR Profiler Plus and Cofiler amplification kit. Samples were denatured for 3 min at 95 °C and loaded onto 5% denaturing Long Ranger gels. The gels were run for 3 h

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Table 1
Observed allele frequency for STR loci

	TH01	TPOX	CSF1PO	D3S1358	VWA	FGA	D5S818	D13S317	D7S820	D8S1179	D21S11	D18S51	D16S539
	N=243	*N*=243	*N*=241	*N*=248	*N*=248	*N*=248	*N*=248	*N*=248	*N*=248	*N*=206	*N*=206	*N*=206	*N*=206
6	0.2140		0.0021										
7	0.1296	0.0020	0.0021						0.0161	0.0024			
8	0.1564	0.4918	0.0041				0.0141	0.1593	0.1573	0.0133			0.0266
9	0.1975	0.1296	0.0124				0.0383	0.0504	0.1250	0.0076		0.0024	0.1358
9.3	0.2963												
10	0.0062	0.0679	0.2759				0.0766	0.0504	0.2742	0.1234		0.0122	0.0266
11		0.2860	0.3091				0.3347	0.3065	0.2379	0.0530		0.0170	0.3033
12		0.0226	0.3195	0.0020			0.3347	0.2581	0.1512	0.1320		0.1624	0.3215
13			0.0622		0.0020		0.1935	0.1190	0.0363	0.3035		0.1087	0.1596
14			0.0062	0.1028	0.1290		0.0081	0.0504	0.0020	0.2144		0.1544	0.0266
15			0.0062	0.2923	0.1371			0.0020		0.1214		0.1325	
16				0.2440	0.2056					0.0266		0.1655	
17				0.1431	0.2681					0.0024		0.1098	
18				0.1956	0.1714	0.0222						0.0532	
18.2						0.0020							
19				0.0202	0.0726	0.0706						0.0532	
20					0.0141	0.1512						0.0266	
21						0.1512						0.0021	
21.2						0.0020							
22						0.1653							

22.2						0.0081							
23						0.1593							
23.2						0.0060							
24						0.1452							
25						0.0827							
26						0.0202							
27						0.0101					0.0266		
28						0.0040					0.1174		
29											0.2024		
30											0.2518		
30.2											0.0532		
31											0.0719		
31.2											0.1065		
32											0.0042		
32.2											0.1065		
33.2											0.0532		
34.2											0.0063		
Min. freq.	0.0120	0.0108	0.0116	0.0118	0.0119	0.0123	0.0111	0.0121	0.0116	0.0132	0.0135	0.0133	0.0138
χ^2 test	0.4895	0.0515	0.2380	0.8715	0.3725	0.1755	0.4285	0.3345	0.3310	0.2355	0.1745	0.4680	0.7865
Exact test	0.1725	0.0540	0.5755	0.8400	0.3145	0.2660	0.2455	0.2120	0.1495	0.2730	0.2355	0.8820	0.1635
H obs.	0.7778	0.6296	0.7178	0.7863	0.7944	0.8306	0.7016	0.8145	0.7621	0.7241	0.8517	0.8966	0.8421
H exp.	0.7877	0.6558	0.7236	0.7869	0.8172	0.8693	0.7324	0.7935	0.8050	0.7987	0.8422	0.8822	0.8478
MEC	0.5585	0.3280	0.4564	0.5738	0.5886	0.6571	0.4308	0.6263	0.5307	0.4666	0.6812	0.7884	0.6794
DP	0.9169	0.8229	0.8712	0.9208	0.9394	0.9660	0.6850	0.9172	0.9318	0.9191	0.9478	0.9620	0.9030

at constant voltage (3000 V) on an ABI 377 sequencer. Allele designations were made according to recommendations of the DNA Commission of the International Society for Forensic Haemogenetics [1].

Statistical evaluations were performed using the computer program Genetic Data Analysis (GDA) as previously described [2]. Analyses included the possible divergence from Hardy–Weinberg expectations and other parameters of forensic importance: observed and expected heterozygosities, mean exclusion chance (MEC) and discrimination power (DP).

2. Results and discussion

The observed allele frequencies and the results of the different test procedures for testing the correspondence of the genotype frequencies with their HWE proportions for the 13 STR loci are shown in Table 1. The genotype frequency distributions for most of the loci do not deviate from HWE expectations based on the χ^2 test and the exact test (in all cases, the data were shuffled 2000 times). Minimum allele frequencies for PCR-based loci, based on statistical and population genetics theory [3], were determined. Table 1 shows several statistical parameters of forensic importance, such as expected and observed heterozygosities, mean exclusion chance (MEC), 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 (data not shown).

In conclusion, a Basque Country resident population database has been established for the analysed systems. The combined power of exclusion is estimated as 99.9998% and the combined power of discrimination is >99.99999% The allele frequency data can be used for deriving estimates of multiple locus profile frequencies for identity testing purposes using the product rule.

References

[1] DNA recommendations, Report concerning further recommendations of the DNA Commission of the ISFH regarding PCR-based polymorphisms in STR (short tandem repeat) systems, Int. J. Leg. Med. 107 (1994) 159–160.

[2] O. García, P. Martín, B. Budowle, C. Albarrán, A. Alonso, Basque Country autochthonous population data on 7 short tandem repeat loci, Int. J. Leg. Med. 111 (1998) 162–164.

[3] B. Budowle, K.L. Monson, R. Chakraborty, Estimating minimum allele frequencies for DNA profile frequency estimates for PCR-based loci, Int. J. Leg. Med. 108 (1996) 173–176.