

Basque Country autochthonous population database on D2S1338, D19S433, Penta D, Penta E and SE33 loci

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Abstract. Blood samples from 204 unrelated Basque Country autochthonous individuals were 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. © 2005 Elsevier B.V. All rights reserved.

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1. 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 ($n=204$) for 5 STR loci. The loci are: D2S1338, D19S433, Penta D, Penta E and SE33. The combined power of exclusion is estimated as 99.76% and the combined power of discrimination is 99.999994%. These 5 STR systems have been shown to be useful tool for personal identification.

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Table 1
Observed allele frequencies for STR

Allele	D2S1338	D19S433	Penta D	Penta E	SE33
2.2			0.0025		
5				0.0417	
7				0.1961	
8			0.0049	0.0074	
9			0.1838	0.0074	
10			0.1348	0.1397	
11		0.0025	0.1838	0.1201	
12		0.1324	0.2230	0.2181	0.0049
13		0.1961	0.1838	0.0980	0.0123
13.2		0.0025			
14		0.3284	0.0735	0.0490	0.0613
14.2		0.0123			0.0025
15		0.1936	0.0074	0.0319	0.0588
15.2		0.0564			0.0049
16	0.0711	0.0466	0.0025	0.0270	0.0662
16.2		0.0098			0.0025
17	0.3211	0.0147		0.0270	0.1078
17.2		0.0025			0.0049
18	0.0539	0.0025		0.0221	0.0931
19	0.0907			0.0025	0.0515
19.2					0.0025
20	0.1471			0.0049	0.0294
20.2					0.0098
21	0.0319			0.0049	0.0270
21.2					0.0123
22	0.0098			0.0025	0.0123
22.2					0.0711
23	0.0956				0.0025
23.2					0.0221
24	0.0784				
24.2					0.0343
25	0.0931				
25.2					0.0196
26	0.0074				
26.2					0.0049
27.2					0.0392
28.2					0.0711
29.2					0.0711
30.2					0.0539
31.2					0.0319
32.2					0.0098
35					0.0025
36					0.0025
MAF	0.0155	0.0139	0.0149	0.0152	0.0157
χ^2 test	0.1100	0.3900	0.0895	0.2530	0.6210
H exp	0.8360	0.7948	0.8272	0.8653	0.9427
MEC	0.7496	0.5182	0.6814	0.7201	0.7794
DP	0.9509	0.9347	0.9392	0.9709	0.9896

2. Material and methods

Whole blood was obtained from unrelated Basque autochthonous donors. Individuals were considered autochthonous if the 8 surnames and birthplace of their grandparents were of Basque origin. Genomic DNA was extracted by the standard phenol/chloroform extraction procedure.

PCR amplification was performed according to the manufacturer's recommendations using the AmpFISTR Identifiler PCR Amplification kit (Applied Biosystems) and the PowerPlex 16/ES Monoplex Systems (Penta D, Penta E and SE33) (Promega Corporation). Samples were typed on an ABI310 sequencer.

Allele designations were made according to recommendations of the DNA Commission of the International Society for Forensic Genetics [1].

Statistical evaluations were performed using the computer program GDA (Genetic Data Analysis) and PowerStats. Analyses included the possible divergence from Hardy-Weinberg expectations and other parameters of forensic importance: expected heterozygosity, mean exclusion chance, polymorphic information content, discrimination power and the possible associations between loci.

3. Results and discussion

The observed allele frequencies and the result for the test procedure for testing the correspondence of the genotype frequencies with their HWE proportions for the 5 STR loci in the Basque Country autochthonous population sample are shown in Table 1. The genotype frequency distributions for the analyzed loci do not deviate from HWE expectations based on the χ^2 test (in all cases, the data were shuffled 2000 times). Minimum allele frequencies (MAF) for PCR-based loci, based on statistical and population genetics theory [2], were determined. Thus, a greater confidence of the DNA profile frequency estimates can be attained with current size databases. Table 1 shows several statistical parameters of forensic importance, such as expected heterozygosity, 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) and support the view that the use of the product rule would provide a good approximation of the estimate of the rarity of a multiple locus profile.

In conclusion, a Basque Country population database has been established for the D2S1338, D19S433, Penta D, Penta E and SE33 loci. The combined power of exclusion is estimated as 99.76% and the combined power of discrimination is 99.999994%. These 5 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.

References

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