

SHORT COMMUNICATION

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Spanish population data on the four STR loci D8S1179, D16S539, D18S51 and D21S11

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Abstract Population data were generated for four tetrameric short tandem repeat loci systems (D8S1179, D16S539, D18S51 and D21S11) for a Spanish Caucasian population sample ($n = 218$ – 219 individuals) using PCR. All loci were highly polymorphic, met Hardy-Weinberg expectations and the results demonstrated the assumption of independence of the loci analysed. The allele frequency data can be used in identity testing to estimate the frequency of a multiple PCR-based DNA profile in the Spanish population.

Key words PCR · STR · D8S1179 · D16S539 · D18S51 · D21S11 · Spain · Hardy-Weinberg equilibrium

Introduction

This study presents allele and genotype frequency data in a Spanish population sample for the four STR loci D8S1179 [1], D16S539 [GenBank G07925], D18S51 [2] and D21S11 [3, 4].

Materials and methods

DNA was extracted by the standard phenol/chloroform extraction procedure from 218–219 individuals residing predominantly in the following geographical areas of Spain: communities of Madrid, Castilla-La Mancha, and Castilla-León (Central Spain), Galicia and País Vasco (North Spain) and community of Valencia (Eastern Spain). PCR amplification was performed according to the manufacturer's recommendations using the Perkin Elmer AmpFISTR Profiler Plus Kit. Typing was performed on an ABI 377 sequencer according to the recommendations of the AmpFISTR Profiler Plus user's manual (Perkin Elmer), using the Genescan 2.1 analysis software and the Genotyper 2.0 analysis software. Allele designations were made according to recommendations of the DNA Commission of the International Society for Forensic Haemogenetics [5].

The statistical evaluations, including the possible divergence from Hardy-Weinberg expectations and other parameters of forensic importance, were performed using the HWE-Analysis, version 3.3. (Christoph Puers, Institute of Legal Medicine, University of Münster). The possible associations between loci were tested using the computer program GDA (Genetic Data Analysis: software for the analysis of discrete genetic data) distributed by the authors, Lewis and Zaykin (1996).

Results and discussion

The genotype frequency distributions showed no deviations from HWE expectations based on the χ^2 -test, the logarithmic likelihood ratio (G) test and the exact test [6] (Tables 1 and 2). The test based on the number of distinct genotypes observed in the sample population showed that the observed numbers of distinct heterozygote and homozygote genotypes [7] were in accordance with the respective HWE predictions (data not shown). Minimum allele frequencies for PCR-based loci, based on statistical and population genetics theory [8–10], were determined (Table 1). Thus, a greater confidence of DNA profile frequency estimates can be attained with current databases sizes. The heterozygosity rates, allele frequency distributions and other parameters of forensic importance (Table 3) were similar to those found in other Caucasians populations (data not shown) [11–14] [U.S. Caucasian databases (1997) Population Genetics. AmpFI STR Profiler Plus Amplification Kit. User's Manual. PE Applied Biosystems and U.S. Caucasian databases (1998) Population Genetics. AmpFI STR Cofiler Amplification Kit. User's Manual. PE Applied Biosystems]. An interclass correlation test analysis demonstrated that there was no evidence for correlation between the alleles at any of the pairs of loci (Table 4).

In conclusion, a Spanish population database has been established for the D8S1179, D16S539, D18S51 and D21S11 loci. The combined power of exclusion was estimated as 98.78% and the combined power of discrimination as 99.9993%. The allele frequency data can be used for deriving estimates of multiple locus profile frequencies for identity and paternity testing purposes using the product rule.

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Table 1 Observed allele frequencies for the STR loci

Allele	D8S1179 (n = 219)	D16S539 (n = 219)	D18S51 (n = 218)	D21S11 (n = 219)
8	0.0160	0.0251		
9	0.0068	0.1279	0.0023	
10	0.1050	0.0251	0.0138	
11	0.0822	0.2922	0.0115	
12	0.1416	0.3105	0.1514	
13	0.2717	0.1918	0.1009	
14	0.2260	0.0274	0.1628	
15	0.1164		0.1491	
16	0.0320		0.1628	
17	0.0023		0.1009	
18			0.0550	
19			0.0390	
20			0.0298	
21			0.0069	
22			0.0138	
24.2				0.0046
25.2				0.0023
27				0.0228
28				0.1187
29				0.2169
30				0.2489
30.2				0.0365
31				0.0776
31.2				0.1050
32				0.0046
32.2				0.0959
33.2				0.0594
34.2				0.0068
Minimum Frequency	0.0137	0.0129	0.0151	0.0137

n = sample size

Table 2 HWE tests on the STR loci

	D8S1179	D16S539	D18S51	D21S11
χ^2 test	0.2860	0.5700	0.4650	0.1645
G test	0.2835	0.7390	0.6410	0.1715
Exact test	0.3270	0.6940	0.6710	0.1875

Number of random shuffles performed: 2000

Table 3 Statistical parameters of forensic importance

	D8S1179	D16S539	D18S51	D21S11
H _{obs}	0.8219	0.7397	0.8853	0.8219
H _{exp}	0.8243	0.7648	0.8775	0.8471
MEC	0.6507	0.5439	0.7483	0.6951
PIC	0.7996	0.7251	0.8626	0.8277
DP	0.9454	0.9084	0.9683	0.9553

H_{obs} Observed heterozygosity [15]
 H_{exp} Expected heterozygosity [15]
 MEC Mean exclusion chance [16]
 PIC Polymorphism information content [17]
 DP Discrimination power [18]

Table 4 Two loci inter-class correlation test for STR loci in a Spanish population sample

	D8S1179	D16S539	D18S51	D21S11
D8S1179	–			
D16S539	0.5245	–		
D18S51	0.8895	0.5320	–	
D21S11	0.3445	0.1395	0.1775	–

References

1. Oldroyd NJ, Urquhart AJ, Kimpton CP, Millican ES, Watson SK, Downes T, Gill PD (1995) A highly discriminating octoplex short tandem repeat polymerase chain reaction system suitable for human individual identification. *Electrophoresis* 16: 334–337
2. Urquhart AJ, Oldroyd NJ, Kimpton CP, Gill P (1995) Highly discriminating heptaplex short tandem repeat PCR system for forensic identification. *Biotechniques* 18: 116–121
3. Sharma V, Litt M (1992) Tetranucleotide repeat polymorphism at D21S11 locus. *Hum Mol Genet* 1: 67
4. Möller A, Meyer E, Brinkmann B (1994) Different types of structural variation in microsatellites: HumFES/FPS, HumVWA and HUMD21S11. *Int J Legal Med* 106: 319–323
5. DNA recommendations (1994) Report concerning further recommendations of the DNA. Commission of the ISFH regarding PCR-based polymorphisms in STR (short tandem repeat) systems. *Int J Legal Med* 107: 159–160
6. Guo SW, Thompson EA (1992) Performing the exact test of Hardy-Weinberg proportion for multiple alleles. *Biometrics* 48: 361–372
7. Chakraborty R (1993) A class of population genetic questions formulated as the generalized occupancy problem. *Genetics* 134: 953–958
8. Budowle B, Monson KL, Chakraborty R (1996) Estimating minimum allele frequencies for DNA profile frequency estimates for PCR-based loci. *Int J Legal Med* 108: 173–176
9. Chakraborty R (1992) Sample size requirements for addressing the population genetic issues of forensic use of DNA typing. *Hum Biol* 64: 141–159
10. Weir BS (1992) Independence of VNTR alleles defined by fixed bins. *Genetics* 130: 873–887
11. Ribeiro T, Brito RM, Espinheira R, Geada H (1998) Duplex STR analysis of D19S253 and D18S51 in a Portuguese population. In: Olaisen B, Brinkmann B, Lincoln PJ (eds) *Progress in forensic genetics 7*. Elsevier, Amsterdam Lausanne New York, pp 285–287
12. Seidl C, Müller S, Kilp M, Seifried E (1998) Fluorescence based multiplex analysis of the STR polymorphism FIBRA (FGA), VWFA31 and D18S51 in German caucasoid individuals. In: Olaisen B, Brinkmann B, Lincoln PJ (eds) *Progress in forensic genetics 7*. Elsevier, Amsterdam Lausanne New York, pp 353–355
13. Mornhinweg E, Luckenbach C, Ritter H (1998) D3S1358 and D8S1179: analysis and allele frequencies in a south German population. In: Olaisen B, Brinkmann B, Lincoln PJ (eds) *Progress in forensic genetics 7*. Elsevier, Amsterdam Lausanne New York, pp 315–317
14. Phillips CP, Syndercombe Court D, Tamaki N (1998) UK caucasian databases for the short tandem repeat loci D21S11, HUMFIBRA (FGA) and D8S1179. In Olaisen B, Brinkmann B, Lincoln PJ (eds) *Progress in forensic genetics 7*. Elsevier, Amsterdam Lausanne New York, pp 398–400
15. Nei M (1978) Estimation of average heterozygosity and genetic distance from a small number of individuals. *Genetics* 89: 583–590
16. Wiegand P, Budowle B, Rand S, Brinkmann B (1993) Forensic validation of the STR systems SE33 and TC11. *Int J Legal Med* 105: 315–320
17. Botstein D, White RL, Skolnick M, Davis RW (1980) Construction of a genetic linkage map in man using restriction fragment length polymorphisms. *Am J Hum Genet* 32: 314–331
18. Jones DA (1972) Blood samples: probabilities of discriminations. *J Forensic Sci Soc* 12: 355–359