TECHNICAL NOTE

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Allelic Distribution of Four Tetranucleotide Repeat Loci (D3S1358, D18S51, D19S253, and FGA) in a Population from Porto (North Portugal)

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ABSTRACT: Allele frequencies for four short tandem repeat loci were determined in a population sample from Porto (North Portugal), using the polymerase chain reaction (PCR), in order to investigate possible genetic differences between populations from the center and north of Portugal. After denaturing PAGE electrophoresis, nine alleles were identified for D3S1358 ($n = 256$), 13 alleles for D18S51 ($n = 235$), 10 alleles for D19S253 ($n = 238$), and 15 alleles for FGA ($n = 181$). No deviations from Hardy-Weinberg equilibrium were found. The allele frequencies observed are similar to those of the Portuguese population compared except for the D3S1358 system.

KEYWORDS: forensic science, DNA typing, population genetics, allele frequencies, Portugal, D3S1358, D18S51, D19S253, FGA

Tetranucleotide short tandem repeats (STRs) are useful markers for genetic characterization of individuals (e.g., for forensic purposes such as criminal investigations or paternity testing) and human populations. In order to obtain population data from North Portugal, four STR systems (D3S1358, D18S51, D19S253, and FGA.) have been analyzed in a sample from Porto. The results were compared with those obtained from other Portuguese populations.

Materials and Methods

DNA was extracted from bloodstain samples using the phenol-chloroform-isoamyl alcohol method (1). PCR of D3S1358, D18S51, and D19S253 was achieved using primers described by Hua et al. (2), Straub et al. (3), and Weber et al. (4), respectively. Triplex PCR amplifications were performed according to the method described by Gené et al. (5). PCR amplification and primer sequences for the FGA STR locus were used according to Barber et al. (6). Separation and genotyping of all PCR products were carried out on polyacrylamide denaturing high-performance DNA sequencing gels in the automated laser fluorescent (ALF) DNA sequencer.

Possible divergence from Hardy-Weinberg equilibrium (HWE) was determined by calculating the exact test proposed by Guo and Thompson (7). Statistical parameters of medicolegal interest for the STR systems such as the power of discrimination (PD) (8), the heterozygosity value (h) (9), and the "a priori" chance exclusion value (CE) (10) were also calculated. The Porto data were compared with other Portuguese populations using an R×C contingency table χ^2 test for homogeneity.

Results and Discussion

Allele frequencies for the four STR systems obtained in the Portuguese sample are shown in Table 1. For D3S1358 a total of 26 different genotypes and nine alleles were observed in 256 individuals. A total of 53 genotypes from 13 alleles were observed for D18S51 locus in 235 individuals. At the locus D19S253, 31 genotypes from ten alleles were observed in 238 individuals, and 43 genotypes and 15 alleles were observed in 181 individuals typed at the FGA locus. The distribution of the genotypes for all loci were in Hardy-Weinberg equilibrium. In order to know if there was genetic heterogeneity between the north and the center of Portugal, the allele frequencies from Porto were compared with those of the other Portuguese population. For the D3S1358 and FGA systems our data were compared with those observed by Santos et al. (11), and for D18S51 and D19S253, Porto was compared with data analyzed by Ribeiro et al. (12). Our results showed no significant differences for the systems D18S51 ($p = 0.5823$), D19S253 ($p = 0.9963$), and FGA ($p = 0.4217$). Nevertheless, significant differences were observed for the D3S1358 system ($p = 0.0350$) between Porto and the Portuguese population (11).

Theoretical values of medicolegal interest, such as the heterozygosity index, power of discrimination, and chance exclusion value were calculated from the gene frequencies obtained in our study (Table 1). The four systems demonstrated a highly polymorphic allele distribution, leading to a high forensic efficiency, and with a combined chance exclusion of 0.988.

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TABLE 1—*Allele frequency distribution for D3S1358, D18S51, D19S253, and FGA in a Porto population and statistical parameters for the four STR systems.*

Allele	D3S1358 $n = 256$	D18S51 $n = 235$	D19S253 $n = 238$	FGA $n = 181$
5	…	…	0.252	…
7	…	…	0.036	…
8	…	…	…	…
9	…	…	0.010	…
10	…	0.006	…	…
11	…	0.013	0.025	…
12	0.006	0.132	…	…
13	0.004	0.140	0.105	…
14	0.078	0.142	…	…
15	0.232	0.136	0.323	…
16	0.260	0.155	…	…
17	0.213	0.102	0.181	0.003
18	0.184	0.087	…	0.022
19	0.017	0.049	0.052	0.061
20	0.006	0.028	…	0.135
21	…	0.006	0.013	0.177
22	…	…	…	0.188
23	…	…	0.002	0.149
24	…	…	…	0.130
25	…	…	…	0.099
26	…	…	…	0.019
27	…	…	…	0.003
28	…	…	…	0.003
33	…	0.002	…	…
41	…	…	…	0.003
42	…	…	…	0.005
45	…	…	…	0.003
HWE = Exa Exact Test	$p = 0.472$	$p = 0.478$	$p = 0.088$	$p = 0.743$

System	h	PD	CE
D3S1358	0.744	0.925	0.589
D18S51	0.881	0.973	0.753
D19S253	0.701	0.922	0.585
FGA0.881	0.965	0.720	

h = heterozygosity value.
PD = Power of discrimination.
CE = Chance of exclusion.

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