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Allele frequencies of the Profiler Plus™ STR loci in Canary Islands (Spain)

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Abstract

Polymorphic short tandem repeat (STR) loci are the most informative PCR-based genetic markers for attempting to individualize biological material. For this purpose, it is necessary to have a database representative of the reference population. The Canarian archipelago is a crossroad for populations from Europe, Africa and South America. This makes it a particularly busy zone for criminal activities and, therefore, for laboratory investigations. In this work, the allele distribution data for the nine Profiler Plus™ tetrameric STR loci (D3S1358, vWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317 and D7S820) were determined from a survey of 240 individuals from the Canary Islands population.

Keywords: Allele frequencies; Population study; Short Tandem Repeat; Canary Islands

1. Samples

Blood stain samples were randomly taken from routine forensic investigations or paternity casework, and saliva samples were collected from volunteers. DNA was isolated from all samples using a phenol–chloroform method, followed by concentration with Microcon-100® devices (Millipore). The amplification was performed by the AmplFlSTR® Profiler Plus™ kit (Applied Biosystems), following the manufacturer's recommendations. The amplified products were analyzed by capillary electrophoresis using an ABI Prism™ 310 Genetic Analyzer.

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Allele designations were determined by comparison of the sample fragments with those of the allelic ladders. The frequency of each allele for each locus was calculated from the number of each genotype in the sample set (i.e., the gene count method). The statistical analysis was aided by two software packages [1,2]. Unbiased estimates of expected heterozygosity were computed as described by Nei [3]. Possible divergence

Table 1
Observed allele frequencies for the STR loci in Canary Islands population (Min. freq., minimum frequencies)

Allele	D3S1358 ($N=239$)	VWA ($N=240$)	FGA ($N=238$)	D8S1179 ($N=240$)	D21S11 ($N=240$)	D18S51 ($N=236$)	D5S818 ($N=240$)	D13S317 ($N=240$)	D7S820 ($N=238$)
7									0.0063
8				0.0208			0.0104	0.1354	0.1554
9				0.0104		0.0021	0.0479	0.0667	0.1218
10				0.0979		0.0085	0.0458	0.0646	0.3151
11				0.0812		0.0085	0.3521	0.2917	0.2101
12	0.0021			0.1208		0.1102	0.3437	0.2667	0.1702
13	0.0063	0.0083		0.2562		0.1038	0.1917	0.1167	0.0189
13.2						0.0021			
14	0.0795	0.0979		0.2416		0.1610	0.0062	0.0583	0.0021
15	0.2636	0.1521		0.1312		0.1716	0.0021		
16	0.2510	0.2562		0.0312		0.1589			
17	0.2008	0.2542		0.0062		0.1208			
18	0.1904	0.1521	0.0168	0.0021		0.0572			
19	0.0063	0.0604	0.0735			0.0445			
20		0.0167	0.1197			0.0318			
21		0.0021	0.1702			0.0084			
21.2			0.0021						
22			0.1996			0.0084			
22.2			0.0042						
23			0.1471		0.0020	0.0021			
23.2			0.0063						
24			0.1534						
25			0.0735						
26			0.0294						
27			0.0021		0.0271				
28			0.0021		0.1479				
29					0.2312				
29.2					0.0042				
30					0.2458				
30.2					0.0396				
31					0.0750				
31.2					0.0812				
32					0.0104				
32.2					0.0812				
33					0.0021				
33.2					0.0396				
34.2					0.0104				
35					0.0021				
Min. freq.	0.0124	0.0123	0.0125	0.0127	0.0120	0.0129	0.0117	0.0124	0.0118

Table 2
Statistical parameters of forensic interest

	D3S1358	VWA	FGA	D8S1179	D21S11	D18S51	D5S818	D13S317	D7S820
H_{obs}	0.8033	0.7958	0.8067	0.8375	0.7708	0.8340	0.7333	0.8083	0.7353
H_{exp}[a]	0.7862	0.8116	0.8615	0.8281	0.8430	0.8772	0.7181	0.8015	0.7898
χ^2 test	0.2985	0.1190	0.4480	0.5725	0.8015	0.1065	0.9230	0.8965	0.1965
Exact test	0.1610	0.1460	0.0535	0.7870	0.3640	0.0505	0.9230	0.9015	0.0740
MEC	0.6053	0.5913	0.6116	0.6704	0.5461	0.6637	0.4817	0.6146	0.4849
PIC	0.7499	0.7834	0.8437	0.8046	0.8232	0.8613	0.6670	0.7727	0.7568
DP	0.9120	0.9351	0.9613	0.9465	0.9556	0.9694	0.8678	0.9314	0.9230

H_{obs}, observed heterozygosity; H_{exp}, expected heterozigosity; MEC, mean exclusion chance; PIC, polymorphic information content; DP, discrimination power. Number of random shuffles performed: 2000.

[a] Expected heterozygosity is an unbiased estimate.

from Hardy–Weinberg expectations was tested by calculating the unbiased estimate of the expected heterozygote frequencies and the exact test [4], based on 2000 shuffling experiments.

2. Results and discussion

The distributions of observed allele frequencies for the STR loci are shown in Table 1. Likewise, some statistical parameters of forensic interest are shown in Table 2, indicating no deviation from the HWE ($p>0.05$). Independence between loci was also confirmed (not shown). The accumulated discrimination power for all loci was 99.9999999989%, and the combined chance of exclusion was 99.9684%.

Some of the samples had been previously analysed by silver-stained electrophoresis gels using Triplex GenePrint™ Systems (Promega), and the concordance in some loci between both data pools was checked as a validation study. No discrepancies were found, except for the locus D7S820 that was wrongly assigned in silver-stained gels in 1 out of 56 cases, mainly due to the band compression in the longer markers. This confirms the superiority of capillary electrophoresis both in automation and precision.

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