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# Allelic frequencies of 13 STR loci in autochthonous Basques from the province of Vizcaya (Spain)

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## Abstract

Allelic frequencies of 13 STR loci (D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D16S539, TH01, TPOX, CSF1PO, and D7S820) were estimated from a sample of 73 unrelated healthy donors natives of the Spanish Basque province of Vizcaya. These STR loci constitute the core of polymerase chain reaction (PCR)-based DNA genetic markers in the US Combined DNA Index System (CODIS). All STR loci analysed met Hardy–Weinberg expectations. Based upon the allelic frequencies, forensically important parameters including gene diversity (GD), polymorphism information content (PIC) and power of discrimination (PD) were calculated.

*Keywords:* Short tandem repeats; Allelic frequencies; AmpF*l*STR Profiler Plus; AmpF*l*STR Cofiler; CODIS; Basque Country

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**Population**: Whole blood samples were collected from 73 unrelated healthy Basque autochthonous donors natives of the Spanish Basque province of Vizcaya. Basque surnames, and birthplaces of individuals and ancestors (recorded back to the third generation) were the criteria employed to define local autochthony. Some previous population genetic studies have shown that genetic diversity in Basques is spatially structured [1–5]. This suggests the need to consider the different Basque "historical territories" (Provinces) as independent populations. If subpopulation structure is reflected in the STR loci sanctioned by CODIS, databases from different Basque regions may be justified.

**DNA extraction**: Blood was collected in EDTA vacuum tubes. Adherence to ethical guidelines was followed as stipulated by each of the institutions involved. DNA was extracted by standard phenol:chloroform extraction procedure and ethanol precipitation.

**PCR**: PCR amplifications of the 13 CODIS core STR loci (D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D16S539, TH01, TPOX, CSF1PO, and D7S820) were performed using AmpF*l*STR Profiler Plus and AmpF*l*STR Cofiler amplification kits (PE Applied Biosystems, Foster City, CA, USA). This process was performed using 1.0–2.5 ng of genomic DNA in a final PCR volume of 12 μl.

**Typing**: DNA fragment analysis was performed in an ABI Prism 377 (PE Applied Biosystems). ABI GeneScan 500 Rox was used as internal lane standard. Genotyping was

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made with the software Genotyper® 3.7 NT by comparison with reference sequenced ladders following the guidelines of the DNA Commission of the International Society for Forensic Haemogenetics [6].

**Results**: For results see Tables 1 and 2 as well as Fig. 1.

**Analysis of data**: HWE using the exact-*p* method [7], polymorphism information content (PIC) [8], and power of discrimination (PD) [9] analyses were performed.

**Remarks**: No significant departure from Hardy–Weinberg Equilibrium (HWE) expectations was found in any of the STR loci analysed. The combined value of the power of discrimination (PD) estimated for the Spanish Basque province of Vizcaya was robust (0.999999999999995) which strongly supports the use of these 13 STR loci for personal identity testing. As expected, the most polymorphic loci were also the most discriminating loci: FGA (PD: 96.19%), D21S11 (PD: 94.65%) and D18S51 (PD: 95.59%).

In addition, the following points should be underscored: (i) alleles 13 and 20 of the VWA locus, 32 of D21S11, 28 of D18S51, and 14 of CSF1PO were not detected in the Basque population from Vizcaya. Several previous studies involving Basques groups have reported similar findings [10–12]. Interestingly, these alleles have been identified in many different populations from the Iberian Peninsula (i.e., Spanish and Portuguese populations); (ii) the Basque sample under study showed the lowest frequency for allele 28 of the D21S11 locus within the context of the Iberian Peninsula. This is in agreement with previous results [12]. Yet, both of these results must be considered with caution bearing in mind the small sample size. Based on $F_{ST}$ values, the autochthonous Basque population from Vizcaya examined in the present study is not significantly different from other Spanish populations (Resident Basques [13], Northeast of Spain [14], Andalusia [15], Extremadura [16] and Canary Islands [17]). Yet, significant differences were observed between Vizcaya and Portugal [18] ($F_{ST} = 0.00109$; $P < 0.00001$). Also, a comparative analysis using the Neighbor Joining (NJ) method including bootstrap values was

Table 1
Allelic frequencies of the 13 STR CODIS core loci in autochthonous Basques from the province of Vizcaya (Spain)

| *N* | D3S1358 (*n* = 73) | VWA (*n* = 72) | FGA (*n* = 70) | D8S1179 (*n* = 69) | D21S11 (*n* = 69) | D18S51 (*n* = 69) | D5S818 (*n* = 69) | D13S317 (*n* = 71) | D7S820 (*n* = 73) | D16S539 (*n* = 73) | TH01 (*n* = 73) | TPOX (*n* = 73) | CSF1PO (*n* = 73) |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 5 | | | | | | | | | | | 0.0206 | | |
| 6 | | | | | | | | | | | 0.2534 | 0.0069 | |
| 7 | | | | | | | | | 0.0206 | | 0.0890 | 0.0206 | |
| 8 | | | | 0.0362 | | | | 0.2324 | 0.0616 | 0.0069 | 0.1781 | 0.5069 | |
| 9 | | | | 0.0290 | | | 0.0290 | 0.0423 | 0.1027 | 0.1164 | 0.2055 | 0.1507 | 0.0069 |
| 9.3 | | | | | | | | | | | 0.2534 | | |
| 10 | | | | 0.0870 | | | 0.1304 | 0.0775 | 0.3767 | 0.0137 | | 0.0616 | 0.2534 |
| 11 | | | | 0.0217 | | | 0.2899 | 0.2676 | 0.2329 | 0.3151 | | 0.1986 | 0.3151 |
| 12 | | | | 0.0870 | | 0.1812 | 0.3623 | 0.2958 | 0.1233 | 0.3082 | | 0.0548 | 0.3219 |
| 13 | 0.0069 | | | 0.3188 | | 0.0870 | 0.1739 | 0.0775 | 0.0616 | 0.1849 | | | 0.0959 |
| 14 | 0.0616 | 0.1181 | | 0.2391 | | 0.1449 | 0.0145 | 0.0070 | 0.0206 | 0.0548 | | | |
| 15 | 0.3288 | 0.2014 | | 0.1594 | | 0.2536 | | | | | | | 0.0069 |
| 16 | 0.1918 | 0.1944 | | 0.0145 | | 0.1449 | | | | | | | |
| 17 | 0.1986 | 0.2639 | | 0.0073 | | 0.0942 | | | | | | | |
| 18 | 0.1849 | 0.2153 | 0.0214 | | | 0.0290 | | | | | | | |
| 19 | 0.0274 | 0.0069 | 0.0929 | | | 0.0435 | | | | | | | |
| 20 | | | 0.2357 | | | 0.0145 | | | | | | | |
| 21 | | | 0.1214 | | | 0.0073 | | | | | | | |
| 22 | | | 0.1000 | | | | | | | | | | |
| 23 | | | 0.1643 | | | | | | | | | | |
| 24 | | | 0.0929 | | | | | | | | | | |
| 25 | | | 0.1429 | | | | | | | | | | |
| 26 | | | 0.0286 | | | | | | | | | | |
| 27 | | | | | 0.0217 | | | | | | | | |
| 28 | | | | | 0.0870 | | | | | | | | |
| 29 | | | | | 0.2174 | | | | | | | | |
| 30 | | | | | 0.3044 | | | | | | | | |
| 30.2 | | | | | 0.0145 | | | | | | | | |
| 31 | | | | | 0.0652 | | | | | | | | |
| 31.2 | | | | | 0.0797 | | | | | | | | |
| 32.2 | | | | | 0.1159 | | | | | | | | |
| 33.2 | | | | | 0.0870 | | | | | | | | |
| 34.2 | | | | | 0.0073 | | | | | | | | |