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Allele Frequencies of 15 STR loci (Identifiler™ kit) in Basque-Americans

Abstract: Individuals with Basque ancestry form a historically and culturally important minority of the population of the western United States. Allele frequencies for the 15 autosomal STRs in the AmpFI STR® Identifiler® PCR Amplification Kit (Applied Biosystems) from 156 unrelated self-identified Basque individuals born in the United States are presented. Allele frequencies were used to calculate parameters commonly used in genetics and forensics including power of discrimination (PD), power of exclusion (PE), polymorphic information content (PIC), and expected heterozygosity (He). The sample population was also compared with the European Basque population and the major American ethnicities.

Keywords:

Short Tandem Repeats (STR)

Allele Frequency

American Basques

Forensic DNA

Population:

The Basque are a linguistic isolate and ethnic group, inhabiting the Basque Country (Euskal Herria) of northern Spain and southwestern France; they have been the subjects of numerous genetic studies [1-8]. Once considered genetic isolates, they are now known to be more closely related to neighboring European populations than other world populations [1], although they can be differentiated from other Europeans with high-density SNP genotyping [2]. Basque migrants

settled in the region of the Intermountain West in the late 1800's, especially the regions of Southwestern Idaho, Southeastern Oregon, and Northern Nevada. The 2000 census registered a total of 57,793 people of Basque ethnicity residing in the USA [9].

Buccal swab samples (Whatman® Omni Swabs) were collected at the Jaialdi and San Inazio Basque Festivals, in Boise, ID, USA. Samples were taken from 156 unrelated individuals, who identify themselves as Basque and were born in the United States. Informed consent was obtained from each individual as well as information on their ethnic background.

In previous studies it has been shown that genetic diversity in European Basque populations is spatially structured [3-5], and it has been suggested that it is necessary to consider different Basque populations as independent populations [6].

Extraction:

DNA was extracted from buccal swabs using the QIAamp® DNA Mini Kit (QIAGEN) following manufactures protocol for DNA purification from buccal swabs.

PCR:

PCR was performed using AmpFISTR® Identifiler® PCR Amplification Kit (Applied Biosystems, Thermofisher Scientific) following the manufacturer's protocol.

Typing:

Multiplex PCR samples from each individual and controls were analyzed via capillary gel electrophoresis using the ABI 3130 Genetic Analyzer (Applied Biosystems). The internal lane

standard used was GeneScan™ 500 LIZ™ Size Standard (Applied Biosystems). Genotyping was carried out using GeneMapper ID-X v1.0 software (Applied Biosystems).

Quality control:

The positive control DNA provided in the AmpFISTR® Identifiler® PCR Amplification Kit, and negative extraction controls were run along with all experimental samples. Individuals analyzing the samples on the ABI 3130 received advanced GeneMapper ID-X training from Applied Biosystems “HID University 300”. In a previous study [7], our laboratory underwent a proficiency testing quality control check, in conjunction with submission to the Y-HRD database [10, 11].

Analysis of data:

Arlequin v3.5 software [12] was used to determine observed heterozygosity (H_o), expected heterozygosity (H_e) [13], p-values (p) for Hardy-Weinberg Equilibrium exact test [14] using a Markov chain, and pair-wise population comparisons of genetic distances (F_{st}) [15] between the sample population and the European Basque population [16], as well as with the main ethnicities of the United States of America [17] (10,000 permutations; significance level 0.05). Powerstats v1.2 software [18] was used to calculate match probability (MP), power of discrimination (PD) [19], power of exclusion (PE), polymorphic information content (PIC) [20] and Typical Paternity Index (TPI) for each locus.

Results:

Statistical parameters and allele frequencies for the 15 STR loci of self-identified Basque individuals born in the United States (N=156) are reported in Table 1. F_{st} values for the pair-wise comparison with other populations of interest are reported in Table 2. Average F_{st} values have been calculated for each comparison [15, 21].

Access of data:

Available upon request to greghampikian@boisestate.edu.

Other remarks:

The results indicate that all loci are in Hardy-Weinberg equilibrium, considering Bonferroni correction ($0.05/15 = 0.003$) [22]. The most variable loci are D18S51 (PD: 0.963), D21S11 (PD: 0.943), FGA (PD: 0.963) and D2S1338 (PD: 0.962). The average level of expected heterozygosity (H_e) for the overall population is 0.7815. The combined match probability (MP) and power of exclusion (PE) for all 15 STR loci of American Basques is 3.300×10^{-17} and 0.999995 respectively.

The average F_{st} index between the sampled American Basques and the European Basques indicated very little difference between these two populations. A close affinity between the American Basque population and the Hispanic and Caucasian ethnicities was noted. On the other hand, the average F_{st} values indicated a significant genetic difference between American Basques and African Americans, and between American Basques and American Asians (see Table 2 for F_{st} values).

Data obtained from this study display that most of the loci have a high power of discrimination and exclusion, confirming their use for individual identification.

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Table 1: Allelic frequencies for American Basques population. (N=156)

| Allele | D8S1179 | D21S11 | D7S820 | CSF1PO | D3S1358 | TH01 | D13S317 | D16S539 | D2S1338 | D19S433 | vWA | TPOX | D18S51 | D5S818 | FGA |
|------------|---------|--------|--------|--------|---------|-------|---------|---------|---------|---------|-------|-------|--------|--------|-------|
| 6 | - | - | - | - | - | 0.234 | - | - | - | - | - | - | - | - | - |
| 7 | - | - | 0.026 | - | - | 0.151 | - | - | - | - | - | - | - | - | - |
| 8 | 0.022 | - | 0.125 | 0.006 | - | 0.096 | 0.170 | 0.022 | - | - | - | 0.519 | - | - | - |
| 9 | 0.010 | - | 0.090 | 0.022 | - | 0.218 | 0.038 | 0.071 | - | - | - | 0.119 | - | 0.029 | - |
| 9.3 | - | - | - | - | - | 0.288 | - | - | - | - | - | - | - | - | - |
| 10 | 0.071 | - | 0.337 | 0.237 | - | 0.013 | 0.048 | 0.029 | - | - | - | 0.058 | 0.010 | 0.093 | - |
| 11 | 0.042 | - | 0.260 | 0.343 | - | - | 0.353 | 0.256 | - | - | - | 0.256 | 0.013 | 0.365 | - |
| 12 | 0.080 | - | 0.131 | 0.343 | - | - | 0.199 | 0.365 | - | 0.119 | - | 0.048 | 0.212 | 0.324 | - |
| 13 | 0.343 | - | 0.022 | 0.045 | 0.010 | - | 0.099 | 0.247 | - | 0.218 | 0.003 | - | 0.106 | 0.183 | - |
| 13.2 | - | - | - | - | - | - | - | - | - | 0.006 | - | - | - | - | - |
| 14 | 0.269 | - | 0.010 | 0.003 | 0.099 | - | 0.093 | 0.010 | - | 0.359 | 0.103 | - | 0.112 | 0.006 | - |
| 14.2 | - | - | - | - | - | - | - | - | - | 0.019 | - | - | - | - | - |
| 15 | 0.151 | - | - | - | 0.308 | - | - | - | - | 0.160 | 0.080 | - | 0.183 | - | - |
| 15.2 | - | - | - | - | - | - | - | - | - | 0.054 | - | - | - | - | - |
| 16 | 0.010 | - | - | - | 0.250 | - | - | - | 0.080 | 0.032 | 0.247 | - | 0.122 | - | - |
| 16.2 | - | - | - | - | - | - | - | - | - | 0.013 | - | - | - | - | - |
| 17 | 0.003 | - | - | - | 0.147 | - | - | - | 0.237 | 0.019 | 0.269 | - | 0.138 | - | - |
| 18 | - | - | - | - | 0.167 | - | - | - | 0.051 | - | 0.231 | - | 0.064 | - | 0.022 |
| 19 | - | - | - | - | 0.019 | - | - | - | 0.125 | - | 0.067 | - | 0.019 | - | 0.122 |
| 20 | - | - | - | - | - | - | - | - | 0.147 | - | - | - | 0.003 | - | 0.099 |
| 21 | - | - | - | - | - | - | - | - | 0.029 | - | - | - | 0.010 | - | 0.192 |
| 22 | - | - | - | - | - | - | - | - | 0.019 | - | - | - | 0.010 | - | 0.115 |
| 22.2 | - | - | - | - | - | - | - | - | - | - | - | - | - | - | 0.006 |
| 23 | - | - | - | - | - | - | - | - | 0.096 | - | - | - | - | - | 0.135 |
| 24 | - | - | - | - | - | - | - | - | 0.096 | - | - | - | - | - | 0.106 |
| 24.2 | - | 0.003 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 25 | - | - | - | - | - | - | - | - | 0.109 | - | - | - | - | - | 0.128 |
| 25.2 | - | - | - | - | - | - | - | - | - | - | - | - | - | - | 0.003 |
| 26 | - | 0.003 | - | - | - | - | - | - | 0.010 | - | - | - | - | - | 0.051 |
| 27 | - | 0.022 | - | - | - | - | - | - | - | - | - | - | - | - | 0.019 |
| 28 | - | 0.093 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 29 | - | 0.234 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 29.2 | - | 0.003 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 30 | - | 0.314 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 30.2 | - | 0.035 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 31 | - | 0.048 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 31.2 | - | 0.054 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 32 | - | 0.010 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 32.2 | - | 0.138 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 33.2 | - | 0.032 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| 34.2 | - | 0.010 | - | - | - | - | - | - | - | - | - | - | - | - | - |
| Ho | 0.763 | 0.750 | 0.782 | 0.699 | 0.814 | 0.795 | 0.769 | 0.699 | 0.872 | 0.808 | 0.750 | 0.673 | 0.833 | 0.660 | 0.846 |
| He | 0.776 | 0.813 | 0.780 | 0.708 | 0.785 | 0.785 | 0.788 | 0.736 | 0.868 | 0.781 | 0.794 | 0.647 | 0.862 | 0.721 | 0.879 |
| p | 0.658 | 0.164 | 0.012 | 0.119 | 0.548 | 0.177 | 0.549 | 0.246 | 0.237 | 0.841 | 0.077 | 0.877 | 0.860 | 0.032 | 0.005 |
| PD | 0.918 | 0.943 | 0.913 | 0.852 | 0.911 | 0.911 | 0.925 | 0.883 | 0.962 | 0.922 | 0.920 | 0.820 | 0.963 | 0.865 | 0.963 |
| PE | 0.532 | 0.510 | 0.566 | 0.426 | 0.625 | 0.590 | 0.543 | 0.426 | 0.738 | 0.613 | 0.510 | 0.388 | 0.662 | 0.369 | 0.687 |
| PIC | 0.740 | 0.790 | 0.750 | 0.650 | 0.750 | 0.750 | 0.760 | 0.690 | 0.850 | 0.750 | 0.760 | 0.600 | 0.840 | 0.670 | 0.860 |
| MP | 0.082 | 0.057 | 0.087 | 0.148 | 0.089 | 0.089 | 0.075 | 0.117 | 0.038 | 0.078 | 0.080 | 0.180 | 0.037 | 0.135 | 0.037 |
| TPI | 2.110 | 2.000 | 2.290 | 1.660 | 2.690 | 2.440 | 2.170 | 1.660 | 3.900 | 2.600 | 2.000 | 1.530 | 3.000 | 1.470 | 3.250 |

Ho: Observed Heterozygosity – He: Expected Heterozygosity – p: Exact Test of Hardy-Weinberg – PD: Power of Discrimination – PE: Power of Exclusion– PIC: Polymorphic Information Content – MP: Match Probability – TPI: Typical Paternity Index.

Table 2: F_{st} values calculated for each locus, comparing the differentiation between the sampled America Basque (AB) population and other populations of interest: European Basques (EB), American (all ethnicities) (US), African American (AfA), Caucasian Americans (CA), Hispanic Americans (HA), Asian Americans (AsA).

| <i>STR locus</i> | <i>AB vs EB</i> | <i>AB vs US</i> | <i>AB vs AfA</i> | <i>AB vs CA</i> | <i>AB vs HA</i> | <i>AB vs AsA</i> |
|------------------------------------|------------------------|------------------------|-------------------------|------------------------|------------------------|-------------------------|
| D8S1179 | 0.00066 ^{ns} | 0.00620 | 0.01313 | 0.01226 | 0.00295 ^{ns} | 0.02173 |
| D21S11 | 0.00516 | 0.00690 | 0.02993 | 0.00455 | 0.00078 ^{ns} | 0.00417 ^{ns} |
| D7S820 | 0.00616 | 0.00141 ^{ns} | 0.00837 | 0.00807 | -0.00122 ^{ns} | 0.00922 |
| CSF1PO | -0.00195 ^{ns} | 0.00227 ^{ns} | 0.00892 | -0.00005 ^{ns} | 0.00112 ^{ns} | 0.01197 |
| D3S1358 | 0.00599 | 0.00355 | 0.01153 | 0.00132 ^{ns} | 0.00060 ^{ns} | 0.01418 |
| TH01 | 0.00022 ^{ns} | 0.01839 | 0.07540 | 0.00728 | 0.01717 | 0.07621 |
| D13S317 | 0.00573 | 0.01442 | 0.04665 | 0.00556 | 0.02298 | 0.01727 |
| D16S539 | -0.00071 ^{ns} | 0.02310 | 0.03811 | 0.00779 | 0.02290 | 0.09458 |
| D2S1338 | -0.00179 ^{ns} | 0.00804 | 0.02417 | 0.00129 ^{ns} | 0.00744 | 0.02822 |
| D19S433 | -0.00103 ^{ns} | 0.00430 | 0.02192 | 0.00084 ^{ns} | 0.00131 ^{ns} | 0.01956 |
| vWA | 0.00233 ^{ns} | 0.00197 ^{ns} | 0.01058 | 0.00098 ^{ns} | 0.00327 ^{ns} | 0.01353 |
| TPOX | -0.00113 ^{ns} | 0.00132 ^{ns} | 0.02473 | -0.00211 ^{ns} | 0.00117 ^{ns} | 0.00025 ^{ns} |
| D18S51 | 0.00106 ^{ns} | 0.00859 | 0.02047 | 0.00458 | 0.00559 | 0.03338 |
| D5S818 | -0.00334 ^{ns} | 0.00145 ^{ns} | 0.01353 | 0.00295 ^{ns} | 0.00393 ^{ns} | 0.02614 |
| FGA | 0.00596 | 0.00689 | 0.01029 | 0.00830 | 0.00214 ^{ns} | 0.01819 |
| Average F_{st} | 0.00580 | 0.01004 | 0.02385 | 0.00730 | 0.01522 | 0.02955 |

ns: not significant.

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