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Forensic
Science
International

Announcement of Population Data

A Basque Country autochthonous population study of 11 Y-chromosome STR loci

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Abstract

Haplotype, allele frequencies and population data of 11 Y-chromosome STR loci DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS438 and DYS439 were determined from a sample of 168 unrelated autochthonous male individuals from the Basque Country. The eight surnames and birth places of the grandparents of all analyzed individuals were of Basque origin. A total of 89 haplotypes were identified by the 11 Y-STR loci. The haplotype diversity (97.49%) and discrimination capacity (52.98%) were calculated. Comparisons were made with previously published haplotype data on other Iberian population samples and significant differences were found.
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Keywords: Y-chromosome; STR; Basque Country; Population data

Population: Blood samples were obtained from unrelated healthy autochthonous Basque donors.

N: 168.

Extraction: DNA was extracted from blood by a standard phenol/chloroform extraction procedure and quantified by slot-blot hybridization using the Quantiblot® Human DNA Quantitation kit (Applied-Biosystems, Foster City, CA).

PCR: PCR amplification was performed using at least 1 ng target DNA in a 25 μl final reaction volume. The loci DYS19, DYS385, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS438 and DYS439 were analyzed by using the Y-Plex 6™ and Y-Plex 5™ kits (Reliagene, New Orleans, LA) following the amplification conditions recommended by the manufacturer.

Detection system: The ABI310 and ABI377 sequencers (Applied-Biosystems, Foster City, CA) were used for genetic typing.

Quality control: The proficiency was successfully achieved by the proficiency testing of the GEP-ISFG working group [1] and the Y-STR haplotyping quality assurance exercise 2003 (http://www.ystr.org) for the markers included in this study.

Access to the data: gobies01@sarenet.es.

Analysis of data: Haplotype and allele frequencies were estimated by gene counting. Gene and haplotype

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Table 1
Allele frequencies and gene diversities at 11 Y-STRs in a Basque Country autochthonous population sample (168 individuals)

Allele	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS438	DYS439	Allelic class	DYS385
9					0.024			0.012		10, 11	0.006
10					0.286	0.006		0.107		11, 11	0.042
11					0.679	0.101			0.310	11, 12	0.012
12	0.006	0.167			0.012	0.024	0.036	0.869	0.536	11, 13	0.048
13	0.060	0.494				0.851	0.869	0.012	0.149	11, 14	0.607
14	0.827	0.333				0.018	0.071		0.006	11, 15	0.042
15	0.065						0.024			11, 16	0.012
16	0.018	0.006								12, 12	0.018
17	0.024									12, 13	0.006
18										12, 14	0.077
19										12, 15	0.024
20										12, 17	0.006
21										13, 14	0.018
22				0.018						13, 15	0.028
23				0.012						13, 16	0.006
24				0.155						13, 17	0.006
25				0.738						13, 19	0.006
26				0.077						14, 14	0.012
27			0.006							14, 15	0.006
28			0.143							15, 17	0.006
29			0.435							15, 18	0.006
30			0.333							16, 17	0.006
31			0.077							16, 19	0.006
32			0.006							17, 18	0.006
NA	6	4	6	5	4	5	4	4	4		24
GD (%)	30, 67	61, 70	67, 36	42, 48	45, 72	26, 43	23, 78	23, 30	59, 50		61, 73

NA: Number of alleles/allele classes; GD: Gene Diversity, estimated according to Nei [2].

diversities were calculated according to Nei [2] using the Arlequin software, version 2.000 [3] and the discrimination capacity was the percentage proportion of the different haplotypes.

Analysis of molecular variance (AMOVA) results were summarized in the form of R_{st} values and assessed for statistical significance using a Monte-Carlo test as implemented in the Arlequin software [3]. In population comparisons, DYS385 was not considered and the number of repeats in DYS389I was subtracted from DYS389II.

Results: Results are shown in Tables 1 and 2. Haplotype results were obtained for the 11 loci selected by The Scientific Working Group on DNA Analysis Methods (SWGDAM) for forensic DNA analysis in the US, the same group of markers included in Reliagene kits Y-PLEX[TM] 6 and Y-PLEX[TM] 5, as well as in Y-PLEX[TM] 12. Using this set of Y-STR markers, a total of 89 different haplotypes were identified, 62 of them being singletons. The observed haplotype diversity value (0.9749) can be considered low when compared with the ones obtained for two other Iberian samples from Spain (0.9972) [4] and northern Portugal (0.9975) [5], for the same set of markers.

The most frequent haplotype (H34) was found in 21 individuals. Considering just the 'minimal haplotype' included in the YHRD (Y-STR haplotype reference database; http://www.ystr.org), this haplotype matches the most fre-

quent haplotype in Europe (2.83%) and has a 5.18% frequency in Iberia (varying from 2.68% in southern Portugal to 8.33% in Zaragoza). In the present sample from the Basque Country it reaches 19.05%, the highest frequency ever found. This haplotype also matches the group 1 (G1) haplotypes (that haplotype together with its one step neighbours) and described by Gusmão et al. [6] which presents an increasing gradient from east towards the west European continent, with the highest frequency found in Ireland (37.36%). Once more, G1 haplotypes present the highest frequency (56.55%) in Basques. On the contrary, the second most frequent haplotype group in Iberia (G2, described by Gusmão et al. [6] as an indicator of north African male contribution) appeared just once in our sample (matching H6).

Other remarks: The present haplotype data were compared with the previously published data available for the same set of Y-STR loci in samples from Spain [4] and northern Portugal [5]. In contrast to what was previously found by the comparison of the Iberian minimal haplotype data available at the the YHRD, the Basque Country population presented significant R_{st} values ($P < 0.0001$) in both pairwise comparisons (with northern Portugal, pairwise $R_{st} = 0.07115$; with Spain, pairwise $R_{st} = 0.04894$).

Pairwise comparison between this and another Basque Country sample, previously described for haplotypes including DYS19, DYS389 I and II, DYS390, DYS391, DYS392

Table 2
List of 89 Y-chromosome STR haplotypes detected in 168 unrelated autochthonous males from the Basque Country

Haplotypes	DYS19	DYS385	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS438	DYS439	N
H1	12	11, 15	13	30	24	10	13	13	12	13	1
H2	13	11, 14	12	29	24	10	13	15	12	13	1
H3	13	11, 14	14	30	25	11	12	13	12	12	1
H4	13	11, 14	14	31	24	10	13	13	12	11	1
H5	13	11, 14	16	32	23	11	13	13	12	11	1
H6	13	13, 14	14	30	24	9	11	13	10	11	1
H7	13	15, 17	13	30	23	10	11	13	10	12	1
H8	13	15, 18	13	31	24	11	11	13	10	13	1
H9	13	16, 17	13	31	24	11	11	13	10	12	1
H10	13	16, 19	13	30	25	10	11	13	10	12	1
H11	13	17, 18	12	29	25	10	11	12	10	11	1
H12	14	10, 11	13	29	23	11	13	13	12	13	1
H13	14	11, 11	13	29	25	9	13	14	12	12	1
H14	14	11, 11	14	30	23	10	13	13	12	13	3
H15	14	11, 11	14	30	23	11	13	13	12	12	1
H16	14	11, 11	14	30	23	11	13	13	12	13	1
H17	14	11, 11	14	30	24	10	13	13	12	12	1
H18	14	11, 12	13	29	25	11	13	13	12	11	1
H19	14	11, 13	12	28	24	11	13	13	12	12	2
H20	14	11, 13	13	29	24	10	13	12	12	13	1
H21	14	11, 13	13	29	25	10	13	12	12	13	1
H22	14	11, 13	14	30	24	10	13	13	12	12	1
H23	14	11, 13	14	30	24	11	13	13	12	11	1
H24	14	11, 14	12	28	24	11	13	13	12	11	3
H25	14	11, 14	12	28	24	11	13	13	12	12	7
H26	14	11, 14	12	28	24	11	13	13	13	12	1
H27	14	11, 14	12	28	24	11	14	13	12	12	2
H28	14	11, 14	12	28	24	12	13	13	12	12	1
H29	14	11, 14	13	29	23	10	13	13	12	11	1
H30	14	11, 14	13	29	23	10	13	13	12	12	2
H31	14	11, 14	13	29	23	11	13	13	12	12	1
H32	14	11, 14	13	29	24	10	13	13	12	12	5
H33	14	11, 14	13	29	24	11	13	13	12	11	8
H34	14	11, 14	13	29	24	11	13	13	12	12	21
H35	14	11, 14	13	29	24	11	13	13	12	13	2
H36	14	11, 14	13	29	24	11	13	13	12	14	1
H37	14	11, 14	13	29	24	11	13	14	12	12	3
H38	14	11, 14	13	29	24	11	13	15	12	11	1
H39	14	11, 14	13	29	25	10	13	13	12	12	2
H40	14	11, 14	13	29	25	11	13	13	12	12	1
H41	14	11, 14	13	30	24	11	13	13	12	11	2
H42	14	11, 14	13	30	24	11	13	13	12	12	5
H43	14	11, 14	13	30	24	11	13	13	13	12	1
H44	14	11, 14	14	30	23	11	13	13	12	13	1
H45	14	11, 14	14	30	23	11	13	14	12	13	2
H46	14	11, 14	14	30	24	10	13	13	12	11	1
H47	14	11, 14	14	30	24	10	13	13	12	12	2
H48	14	11, 14	14	30	24	10	14	13	12	12	1
H49	14	11, 14	14	30	24	10	13	13	12	12	1
H50	14	11, 14	14	30	24	11	13	13	12	11	8
H51	14	11, 14	14	30	24	11	13	13	12	12	1
H52	14	11, 14	14	30	25	11	13	13	12	12	1
H53	14	11, 14	14	31	24	10	13	13	12	11	2
H54	14	11, 14	14	31	24	10	13	13	12	12	3
H55	14	11, 14	14	31	24	11	13	13	12	12	2
H56	14	11, 15	12	28	23	11	13	14	12	13	1
H57	14	11, 15	13	30	24	12	13	13	12	12	1

Table 2 (*Continued*)

Haplotypes	DYS19	DYS385	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS438	DYS439	N
H58	14	11, 15	14	30	24	10	13	13	12	11	1
H59	14	11, 15	14	30	24	11	13	13	12	11	3
H60	14	11, 16	13	29	25	11	13	13	12	12	1
H61	14	12, 13	14	30	24	11	13	13	12	13	1
H62	14	12, 14	13	29	24	11	13	13	12	11	5
H63	14	12, 14	13	29	24	11	13	13	12	12	3
H64	14	12, 14	14	30	24	11	13	13	12	12	2
H65	14	12, 14	14	30	25	10	13	13	12	12	1
H66	14	12, 14	14	31	25	10	13	13	12	12	1
H67	14	12, 15	12	28	23	11	13	14	12	13	1
H68	14	12, 15	14	30	24	11	13	13	12	11	3
H69	14	12, 17	14	30	22	9	11	13	10	12	1
H70	14	13, 14	13	29	24	11	13	13	12	11	1
H71	14	13, 15	14	30	24	11	13	13	12	11	1
H72	14	13, 19	14	31	23	11	11	12	10	11	1
H73	14	14, 14	14	30	24	11	13	13	12	11	1
H74	15	11, 13	12	18	21	10	11	14	10	11	1
H75	15	11, 13	13	28	24	11	13	13	12	13	1
H76	15	11, 14	12	28	24	10	13	13	12	13	1
H77	15	11, 14	13	29	23	11	13	13	12	13	2
H78	15	11, 16	14	30	24	11	12	13	12	13	1
H79	15	12, 14	13	28	24	11	13	13	12	13	1
H80	15	13, 14	12	30	22	10	10	14	10	11	1
H81	15	13, 15	12	29	21	10	11	14	10	13	1
H82	15	13, 15	12	29	21	10	11	15	10	12	1
H83	15	13, 17	12	28	23	10	11	12	9	12	1
H84	16	12, 12	12	27	23	10	11	13	10	12	1
H85	16	13, 16	13	29	24	9	11	12	9	12	1
H86	16	14, 14	12	29	23	10	12	15	10	12	1
H87	17	11, 12	13	28	23	10	11	13	10	11	1
H88	17	12, 12	14	29	23	10	11	13	10	12	2
H89	17	14, 15	14	31	24	10	12	14	10	11	1

and DYS393 [7], resulted in a low non-significant R_{st} value ($R_{st} = 0.00402$; $P = 0.35135 \pm 0.0642$).

AMOVA results (grouping Spain and northern Portugal in one group and Basques in another) revealed a high percentage of variation among groups (5.61%; $P < 0.0001$).

In conclusion, a Basque Country Y-STR haplotype database should be used in the forensics field rather than a general Spanish population one, in clear contrast to what was until now observed for other Iberian samples (either from Portugal or Spain).

This paper follows the guidelines for publication of population data requested by the journal [8].

Acknowledgements

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