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Population genetics of Y chromosomal STR haplotypes in south Spain (Andalusia)

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Abstract

We present a study of 109 unrelated males from an Andalusian population (southern Spain) for minimal haplotype using in parallel the Y-Plex™ 6 STR kit (Reliagene Technologies) and amplification and typing protocols according to Elmoznino and Prinz (http://ystr.charite.de). Different parameters of forensic interest have been calculated.

Keywords: Y-STRs; Minimal haplotype; Population studies

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1. Materials and methods

1.1. Population

Blood samples were selected from 109 unrelated individuals from casework, paternity cases and staff, living in Andalusia, southern Spain. All the samples have been typed previously for the whole set of CODIS STRs.

1.2. Extraction

Either with phenol–chloroform–isoamylalcohol followed by purification–concentration with Microcon 100 (Millipore, Bedford, MA, USA) or with GFX genomic purification columns (Amersham-Pharmacia-Biotech, Piscataway, NJ, USA).

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Table 1
YSTR haplotypes in 109 males from Andalusia

	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS385	*n*
1	13	10	26	22	10	15	13	14–16	1
2	13	10	26	23	11	14	12	11–15	1
3	13	10	27	23	10	11	13	17–18	1
4	13	10	27	24	10	11	13	16–18	1
5	13	10	27	25	10	11	13	16–19	1
6	13	11	27	24	9	11	13	13–13	2
7	13	11	27	24	9	11	13	13–14	3
8	13	11	27	25	10	13	13	12–14	1
9	13	11	28	24	10	11	12	16–18	1
10	13	11	28	24	9	11	13	13–15	1
11	13	9	25	24	9	12	13	13–14	1
12	14	10	25	24	10	13	13	11–14	1
13	14	10	26	22	11	13	12	11–14	1
14	14	10	26	23	10	11	12	13–18	1
15	14	10	26	23	10	13	13	11–14	1
16	14	10	26	23	10	9	12	13–17	1
17	14	10	26	24	10	13	13	11–11	1
18	14	10	26	24	10	13	13	11–14	5
19	14	10	26	24	10	13	13	12–14	2
20	14	10	26	24	11	13	13	11–13	1
21	14	10	26	24	11	13	13	11–14	5
22	14	10	26	24	11	13	13	11–15	2
23	14	10	26	24	11	13	13	12–14	1
24	14	10	26	24	11	13	13	16–16	1
25	14	10	26	24	11	7	13	11–14	1
26	14	10	26	25	10	13	13	11–14	1
27	14	10	26	25	10	13	13	11–15	1
28	14	10	26	25	10	14	13	11–14	1
29	14	10	26	25	11	11	12	14–17	1
30	14	10	26	25	11	13	12	10–15	1
31	14	10	26	25	11	13	13	12–14	1
32	14	10	27	24	10	14	13	12–14	1
33	14	10	27	24	11	11	13	11–14	1
34	14	10	27	24	11	13	13	11–13	1
35	14	10	27	24	11	13	13	11–14	2
36	14	10	27	24	11	13	13	11–16	1
37	14	10	27	24	11	13	13	12–14	1
38	14	10	28	23	10	11	12	13–17	1
39	14	10	28	23	10	12	14	14–14	1
40	14	10	28	25	11	13	13	11–14	1
41	14	10	31	24	10	13	13	11–14	1
42	14	11	26	23	11	13	13	11–14	1
43	14	11	27	23	10	11	12	13–19	1
44	14	11	27	23	11	11	12	15–17	1
45	14	11	27	23	11	11	13	12–12	1

Table 1 (*continued*)

	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS385	*n*
46	14	11	27	24	10	13	12	11–13	1
47	14	11	27	24	10	13	13	11–14	1
48	14	11	27	24	10	13	13	12–14	1
49	14	11	27	24	11	13	13	11–13	1
50	14	11	27	24	11	13	13	11–14	4
51	14	11	27	24	11	13	13	12–14	1
52	14	11	27	24	12	13	13	11–17	1
53	14	11	27	25	11	13	13	11–14	1
54	14	11	27	25	11	13	13	11–14	1
55	14	11	28	23	10	11	12	13–16	3
56	14	11	28	23	10	11	13	19–21	1
57	14	11	28	23	11	10	12	11–18	1
58	14	11	28	24	11	13	13	11–14	1
59	14	12	29	23	11	11	12	13–16	1
60	14	9	25	23	10	11	13	13–15	1
61	14	9	25	24	10	13	14	11–14	1
62	14	9	25	24	11	13	13	11–14	1
63	14	9	25	24	11	13	13	10–14	1
64	14	9	26	22	9	11	14	14–15	1
65	14,15	11	30	22	10	11	14	13–13	1
66	15	10	25	24	11	13	13	11–14	1
67	15	10	26	24	11	13	13	11–14	1
68	15	10	27	22	10	11	14	12–14	1
69	15	10	27	23	10	11	12	12–18	1
70	15	10	27	23	10	13	13	11–15	1
71	15	10	27	23	11	15	13	16–17	1
72	15	10	27	23	9	11	12	14–16	1
73	15	10	28	21	10	13	14	13–16	1
74	15	10	28	22	10	11	13	13–15	1
75	15	11	26	24	11	14	13	11–14	1
76	15	11	27	22	10	11	12	15–17	1
77	15	11	27	22	10	11	12	16–17	1
78	15	11	29	23	10	12	14	14–16	1
79	15	9	27	23	10	12	14	14–14	1
80	15,16	9	25	22	10	12	12	14–14	1
81	16	10	25	23	10	11	13	12–12	2
82	16	10	26	23	9	11	12	13–16	1
83	16	10	26	24	11	13	13	11–14	1
84	16	10	27	25	11	11	13	11–14	1
85	16	11	26	22	10	11	13	11–12	1
86	16	9	26	22	10	11	14	13–14	1
87	16	9	27	21	10	11	14	14–15	1
88	17	10	25	23	10	11	13	12–13	1
89	17	10	26	23	10	11	13	12–13	1

1.3. Quantification

Quantiblot, Human DNA Quantitation kit (PE Applied BioSystems, Foster City, CA, USA).

1.4. PCR

According to manufacturers' protocols of the commercially available Y-Plex™ 6 STR kit (Reliagene Technologies, New Orleans, LA, USA) (DYS19, DYS389II, DYS390,

Table 2
Allele frequencies and gene diversity values of the systems

Locus	Allele	Frequency	Diversity value
DYS19	13	0.1261	0.5588
	14	0.6306	
	15	0.1441	
	16	0.0811	
	17	0.0180	
DYS389 I	9	0.0917	0.5589
	10	0.5688	
	11	0.3303	
	12	0.0092	
DYS389 II	25 (28)	0.1009	0.6968
	26 (29)	0.3670	
	27 (30)	0.3761	
	28 (31)	0.1284	
	29 (32)	0.0183	
	30 (33)	0.0092	
DYS390	21	0.0183	0.6519
	22	0.1009	
	23	0.2661	
	24	0.5046	
	25	0.1101	
DYS391	9	0.0917	0.5847
	10	0.4862	
	11	0.4158	
	12	0.0092	
DYS392	7	0.0092	0.6126
	9	0.0092	
	10	0.0092	
	11	0.3578	
	12	0.0459	
	13	0.5046	
	14	0.0459	
	15	0.0183	
DYS393	12	0.2018	0.4519
	13	0.7064	
	14	0.0917	

DYS391, DYS393, DYS385) and amplification and typing protocols according to Elmoznino and Prinz (http://ystr.charite.de): Triplex (DYS19, DYS389 and DYS390) and Pentaplex (DYS19, DYS391, DYS392, DYS393, DYS385).

1.5. Typing

ABI PRISM™ 310; sequenced ladders from Reliagene, sequenced ladders for DYS19, DYS390, DYS391 and DYS393, kindly supplied by Dr. Carracedo, and a set of reference consensus samples previously typed in interlaboratory exercises. Genescan®2.1 and Genotyper®2.0 softwares were used for the analysis of typing results.

Table 3
DYS385 phenotype frequencies (haplotype diversity value: 0.8607)

Phenotype	Frequency
10–14	0.0092
10–15	0.0092
11–11	0.0092
11–12	0.0092
11–13	0.0367
11–14	0.3394
11–15	0.0459
11–16	0.0092
11–17	0.0092
11–18	0.0092
12–12	0.0275
12–13	0.0183
12–14	0.0917
12–18	0.0092
13–13	0.0275
13–14	0.0459
13–15	0.0275
13–16	0.0550
13–17	0.0183
13–18	0.0092
13–19	0.0092
14–14	0.0275
14–15	0.0183
14–16	0.0275
14–17	0.0092
15–17	0.0183
16–16	0.0092
16–17	0.0183
16–18	0.0183
16–19	0.0092
17–18	0.0092
19–21	0.0092

1.6. Nomenclature

According to Kayser et al. [1], for DYS389II, we have included in brackets the nomenclature used by Reliagene (according to Roewer), which considers the additional stretch $[TCTG]_3$ common to all the alleles.

1.7. Quality control

Proficiency testing of the GEP-ISFG working group.

1.8. Calculations

The gene and haplotype diversity values have been calculated as $1 - \Sigma pi^2$.

2. Results and discussion

Haplotypes are listed in Table 1. Allele frequencies and gene diversity are shown in Tables 2 and 3.

No discrepancies have been observed between the results obtained with the three typing systems (Y-Plex, Triplex and Pentaplex) for the shared loci. Double peaks (n and $n+a$) have been observed in DYS389II ladder from Reliagene. Better amplification results were obtained for Pentaplex when both the amounts of DNA template and BSA were increased over the recommended values. DYS390 yielded lower efficiency than the other loci in Pentaplex.

Two individuals with a duplication in DYS19 (frequency 0.018) and two rare alleles in DYS392 (alleles 7 and 9, already described in Y-STR database) have been observed. No other cases of segmental duplication or null alleles have been observed.

Out of 109 individuals 89 different haplotypes have been found (diversity value: 0.9846).

Reference

[1] M. Kayser, et al., Evaluation of Y-chromosomal STRs: a multicenter study, Int. J. Legal Med. 110 (1997) 125–133, Appendix 141–149.