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SHORT COMMUNICATION

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Population study of eight novel Y-chromosome STRs (DYS460, DYS461, GATA-A10, GATA-C4, GATA-H4, DYS434, DYS437, DYS439) in a southeast Iberian population: looking for highly informative Y-chromosome haplotypes

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Abstract The present study analyses 8 recently described tetranucleotide microsatellites (DYS460, DYS461, GATA-A10, GATA-C4, GATA-H4, DYS434, DYS437, DYS439) in southeast Spain and out of a total of 76 individuals 67 showed different haplotypes. Out of the 67 different haplotypes, 63 were present once, 3 were found 2 times, and 1 was found 7 times (9.21%). By combining the allelic states of the present eight Y-chromosome STRs with those previously carried out on the same individuals, highly informative haplotypes could be obtained. The haplotype diversity using the basic set of Y-STRs (DYS19, DYS389-I, DYS389-II, DYS390, DYS391, DYS392, DYS393, DYS385 and DYX156Y) previously analyzed is 0.9844. For the same individuals, this value using the new set of Y-STRs is slightly lower (0.8949), while the haplotype diversity combining the two sets of primers significantly increase to 0.9868. The results obtained in the present work show the usefulness of these microsatellites for individual identification and paternity testing in forensic genetics.

Keywords Y-chromosome STRs · Y-haplotypes · Population study · Mediterranean

Introduction

During the last few years, the interest in Y chromosome polymorphisms has continuously increased, not only because of the application of Y chromosome variation in forensic or genealogical studies (Jobling et al. 1997; Roewer et al. 2000; Gill et al. 2001; Rolf et al. 2001), but also because of its use in other fields such as human evolutionary genetics (Jobling and Tyler-Smith 1995). Among the three main types of different polymorphisms describe to date on the Y chromosome (SNPs, STRs and minisatellites), Y-STRs are the most frequently used in the forensic field. Recently, White et al. (1999) and Ayub et al. (2000) described a set of Y-STRs which can be useful for forensic purposes (Zarrabeitia et al. 2002; Quintáns et al. 2002). The typing of these new Y-STRs as well as the estimation of the gene and haplotype frequencies in a population from southeast Spain, will allow us to determine the information provide by the combination of these new Y-STRs with the classical ones previously typed in the same individuals by Aler et al. (2001).

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Materials and methods

Blood samples from 76 autochthonous unrelated males from southeast Iberia previously used by Aler et al. (2001) were typed in the present work. Extraction procedure details were reported in Aler et al. (2001). The previously described loci GATA-A7.1 and GATA-A7.2 (White et al. 1999) are here referred to as DYS460 (GDB: 11498962), and DYS461 (GDB: 9996572) (see also Bosch et al. 2002), respectively, according to the recommendations of the ISFG (Gill et al. 2001). Two multiplexes were used to amplify the new Y-STRs. Multiplex I includes four systems: DYS460, GATA-A10, GATA-C4 and GATA-H4, multiplex II combines two STRs: DYS437 and DYS439 and DYS461 and DYS434 were amplified as singleplex reactions. The primers used were those described by Ayub et al. (2000) to amplify DYS434, DYS437, DYS439 and by White et al. (1999) to amplify GATA-A10, C4 and H4. For DYS460 and DYS461, primers were described in González-Neira et al. (2001). It was found that the primers described for GATA-A4 also amplify the same region as reported for DYS439 (González-Neira et al. 2001), therefore, only DYS439 was included in this study. Locus DYS461 was amplified following the conditions described by White et al. (1999). Details on multiplex and DYS434 loci amplification conditions can be provided by the authors on request. Amplification was carried out in a DNA thermocycler 2400 (PE Applied Biosystems, Foster City, CA). Typing of the amplified products was carried out using the Automatic Laser Fluorescent DNA sequencer (ALF-Express, APB). Fragment sizes were typed by comparison with sequenced allelic ladders. The nomenclature used in the present work is that proposed by Gusmão et al. (2002), which follows the recommendations of the DNA Commission of the International Society for Forensic Genetics (ISFG; Gill et al. 2001).

Gene frequency and haplotype diversity values were calculated as described by Nei (1987). Pairwise haplotype analysis was carried out using the Arlequin software (Schneider et al. 1997). The different combination of Y-STRs are referred to in the text as: SET A (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385 and DYXS156Y), SET B (DYS460, DYS461, GATA-A10, GATA-C4, GATA-H4, DYS434, DYS437, and DYS439), and SET C (SET A+SET B).

Results and discussion

The eight novel Y-chromosome STRs were investigated in a male population sample from a southeast Mediterranean region of Iberia. Allele frequencies and gene diversity computed in this population for the eight STRs are shown in Tables 1 and 2, respectively.

Table 1 Allele frequencies for 8 Y-STRs in 76 unrelated individuals from Southeast Iberia

Locus	Alleles	Frequency (sd)	Gene diversity
DYS460	9	0.079 (0.031)	0.6243
	10	0.355 (0.055)	
	11	0.487 (0.058)	
	12	0.079 (0.031)	
GATA-A10	14	0.263 (0.051)	0.5084
	15	0.645 (0.055)	
	16	0.079 (0.031)	
	17	0.013 (0.013)	
DYS461	12	0.013 (0.013)	0.6504
	13	0.066 (0.029)	
	14	0.184 (0.045)	
	15	0.526 (0.058)	
	16	0.184 (0.045)	
	17	0.026 (0.018)	
GATA-C4	21	0.013 (0.013)	0.6081
	22	0.053 (0.026)	
	23	0.145 (0.041)	
	24	0.105 (0.035)	
	25	0.592 (0.057)	
	26	0.079 (0.031)	
	28	0.013 (0.013)	
GATA-H4	25	0.013 (0.013)	0.4122
	26	0.013 (0.013)	
	27	0.039 (0.022)	
	28	0.342 (0.055)	
	29	0.539 (0.058)	
	30	0.053 (0.026)	
DYS434	10	0.053 (0.026)	0.1483
	11	0.921 (0.031)	
	12	0.026 (0.018)	
DYS437	14	0.408 (0.057)	0.5751
	15	0.500 (0.058)	
	16	0.092 (0.033)	
DYS439	10	0.026 (0.018)	0.6282
	11	0.276 (0.052)	
	12	0.526 (0.058)	
	13	0.132 (0.039)	
	14	0.026 (0.018)	
	15	0.013 (0.013)	

Table 2 Number of different haplotypes and haplotype diversity values for three sets of Y-STRs markers found in 76 unrelated individuals from southeast Iberia

	SET A	SET B	SET C
Number of different haplotypes	70	67	76
Frequency of the most common haplotype	0.039 (3/76)	0.092 (7/76)	0.013 (1/76)
No. of haplotypes shared	5	4	0
Haplotype diversity value	0.9844	0.8949	0.9868

SET A: DYS19, DYS389I, DYS389II, DSYS390, DYS391, DYS392, DYS393, DYS385, DYXS156Y
SET B: DYS460, DYS461, GATA-A10, GATA-C4, GATA-H4, DYS434, DYS437, DYS439
SET C: SET A+SET B

Gene diversity for each Y-STR analyzed is shown in Table 2. Almost all systems reached values of gene diversity (GD) higher than 0.5, except DYS434 (GD=0.1482; with only 3 alleles, the highest frequency of 0.921 for the allele with 11 repeats) and GATA-H4 (GD=0.4121). DYS461 showed the higher gene diversity (GD=0.6504). All the loci showed a unimodal allelic distribution except GATA-C4 (modal numbers 23 and 25).

Complete 8 Y-chromosomal STRs haplotypes were obtained in 76 individuals, among which 67 different haplotypes were observed. In Table 3 we show these haplotypes in combination with those previously described by Aler et al. (2001) for the same individuals. The most common haplotype was shared by approximately 9% (7 individuals) of the sample, while 63 haplotypes were unique. The most common haplotype 11–15–15–25–29–11–15–12 (DYS460, DYS461, GATA-A10, GATA-C4, GATA-H4, DYS434, DYS437, DYS439) had a frequency of 0.0921. The haplotype diversity was 0.89491.

The combination of the allelic states of the present eight Y-chromosome STRs with those previously carried out on the same individuals (see Aler et al. 2001), allowed us to construct highly informative haplotypes. The haplotype diversity using the basic SET A previously analyzed by Aler et al. (2001) was 0.9844 (discrimination of 92.1%, 70 out of 76). For the same individuals, this value using the new set of Y-STRs is slightly lower (0.8949; discrimination of 88.2%, 67 out of 76), while the haplotype diversity combining the two sets of primers significantly increases to 0.9868 (discrimination of 100%; all haplotypes were different). These results show the importance of implementing these Y-microsatellites for individual identification and paternity testing in forensic genetics in order to increase the discrimination power.

Due to the fact that population data on these new Y-STRs are scarce, and that there are no Iberian data available for these systems, no population comparison analysis can be performed.

Table 3 Y-STR haplotypes in 76 unrelated males from the southeast Iberia region. The code for each haplotype in the first column corresponds with the same used by Aler et al. (2001) for the same individuals. The systems typed in Aler et al. (2001) are also included in the present table

	DYS19	DYS389-I	DYS389-II	DYS390	DYS391	DYS392	DYS393	DYS385	DYXS156Y	DYS460	DYS461	GATA-A10	GATA-C4	GATA-H4	DYS434	DYS437	DYS439
Y-1	15	12	28	24	11	13	13	11/14	12	11	15	15	25	29	11	15	12
Y-2	14	13	29	25	10	13	13	11/14	12	10	16	15	26	28	11	15	13
Y-3	14	13	29	23	10	13	13	11/14	12	10	16	15	25	29	11	15	12
Y-4	14	14	30	24	11	13	13	12/14	12	10	16	15	25	28	11	14	11
Y-5	14	13	29	25	11	13	12	11/14	12	10	15	14	26	28	11	15	11
Y-6	14	13	28	23	11	13	13	11/14	12	11	16	15	23	29	11	15	12
Y-7	14	12	28	22	10	11	13	13/14	12	9	16	16	25	28	11	16	11
Y-8	15	13	29	24	10	13	13	11/14	12	11	16	15	25	30	11	14	12
Y-9	14	14	30	23	11	13	13	11/16	12	11	16	15	25	28	11	14	11
Y-11	14	14	30	23	11	13	13	11/13	12	10	17	15	25	29	11	15	12
Y-12	15	12	29	24	11	11	12	13/18	12	11	13	14	24	28	11	16	12
Y-13	16	12	28	24	10	11	12	13/16	12	11	13	14	23	28	11	16	12
Y-14	15	13	29	23	10	13	14	15/16	12	12	14	14	22	29	11	15	12
Y-15	13	13	30	25	10	11	13	15/18	12	9	15	16	24	28	11	14	13
Y-16	12	13	29	24	11	13	13	11/14	12	10	15	14	25	29	11	15	13
Y-17	15	13	29	23	10	11	12	10/17	12	10	15	17	25	28	11	15	11
Y-18	14	13	29	24	10	13	13	11/14	13	12	15	15	25	29	11	15	12
Y-19	14	11	27	24	11	13	13	11/14	12	12	15	15	25	28	11	15	11
Y-20	15	13	29	24	11	12	12	14/15	12	11	13	14	25	30	10	15	12
Y-21	14	13	29	23	11	13	13	11/15	12	11	15	15	25	28	11	14	13
Y-22	14	13	29	23	11	13	13	11/14	12	10	16	14	25	29	11	14	13
Y-23	16	12	29	22	10	11	14	15/15	12	10	14	14	22	29	11	16	12
Y-24	14	14	30	24	11	13	13	11/14	12	11	15	15	26	29	11	15	11
Y-25	15	13	29	23	10	13	14	12/14	12	10	14	15	25	29	11	15	12
Y-26	14	14	31	24	10	13	13	11/14	12	11	15	15	25	29	11	15	12
Y-27	14	13	28	23	11	15	12	11/16	12	10	15	14	25	28	11	14	12
Y-28	14	12	28	23	10	11	14	14/14	12	10	14	15	25	28	11	15	12
Y-29	14	13	29	24	10	13	13	11/15	12	10	15	15	24	28	11	15	11
Y-30	13	13	30	24	10	11	13	16/18	12	11	15	15	25	29	11	14	12
Y-31	15	12	28	24	10	12	12	12/17	12	9	15	15	25	29	11	14	12
Y-32	14	13	30	23	11	11	12	12/17	12	11	12	14	22	25	11	15	12
Y-33	14	14	30	24	11	13	13	10/15	12	11	14	15	23	28	11	14	11
Y-34	14	13	30	23	10	12	14	15/16	12	10	15	15	25	28	11	14	12
Y-35	14	13	29	24	10	13	13	11/16	12	11	15	14	24	28	11	14	11
Y-36	14	13	30	25	11	13	13	11/14	12	11	15	14	25	28	11	14	11
Y-37	14	13	29	24	10	13	13	11/11	12	11	15	15	25	29	11	15	11
Y-39	13	13	30	24	10	11	13	16/16	12	10	16	15	25	29	11	15	11
Y-40	15	14	30	23	11	11	13	11/14	12	12	15	15	25	29	11	15	11
Y-41	13	13	32	23	10	12	13	16/18	11	9	15	15	23	29	11	14	11
Y-42	15	12	28	24	10	11	12	16/18	12	10	16	14	23	28	11	15	12
Y-43	15	13	31	24	11	11	13	11/16	12	11	15	15	23	27	11	14	11

Table 3 (continued)

	DYS19	DYS389-I	DYS389-II	DYS390	DYS391	DYS392	DYS393	DYS385	DYXS156Y	DYS460	DYS461	GATA-A10	GATA-C4	GATA-H4	DYS434	DYS437	DYS439
Y-44	14	12	28	22	11	10	13	13/14	12	11	13	14	23	26	11	14	12
Y-45	15	13	29	25	9	14	13	11/14	12	11	14	15	25	30	11	14	10
Y-46	13	13	29	25	11	13	13	11/14	12	11	15	15	25	27	11	16	11
Y-47	16	13	29	23	9	11	12	13/16	12	11	15	15	26	29	11	15	13
Y-48	16	14	29	22	11	13	13	13/14	12	11	15	15	25	29	11	15	15
Y-49	14	13	29	24	11	13	13	11/14	12	10	16	15	24	28	11	14	11
Y-50	15	14	29	25	10	15	13	11/13	12	11	15	14	25	29	12	14	12
Y-51	16	13	31	22	10	11	12	14/15	12	12	14	16	24	28	11	15	11
Y-53	15	14	30	23	9	11	12	12/16	12	10	14	15	23	29	10	14	14
Y-54	14	13	29	25	11	13	13	11/14	12	10	17	15	21	30	10	14	12
Y-55	14	14	30	24	11	13	13	13/14	12	10	15	15	25	27	11	15	11
Y-56	14	14	30	24	10	13	13	11/14	12	11	16	15	25	28	11	14	13
Y-57	14	13	29	24	11	13	13	11/14	11	9	15	15	23	29	11	14	11
Y-58	17	13	31	25	11	11	13	13/14	12	11	15	15	23	29	11	14	12
Y-59	13	13	30	24	10	11	13	15/17	12	12	15	14	25	28	10	14	13
Y-60	15	12	29	22	10	11	13	14/14	12	11	15	14	25	28	11	14	12
Y-61	14	13	29	25	10	13	12	11/14	12	10	16	15	25	29	12	14	12
Y-62	14	13	29	24	11	13	13	11/15	12	11	15	15	25	29	11	15	12
Y-63	16	13	29	25	11	13	13	13/14	12	10	14	15	24	29	11	15	12
Y-64	14	14	31	25	11	13	13	11/14	12	10	16	15	25	29	11	14	12
Y-65	15	12	28	22	10	11	15	14/15	12	11	15	16	25	28	11	15	12
Y-67	14	13	29	23	12	13	13	11/15	12	10	14	14	23	29	11	16	13
Y-68	14	14	31	22	10	12	12	13/14	12	11	15	15	26	29	11	15	14
Y-69	15	13	29	23	11	12	14	15/15	12	10	14	16	25	29	11	14	10
Y-70	14	14	30	24	11	13	13	10/13	12	11	15	15	25	29	11	15	12
Y-71	14	13	28	23	10	13	13	13/16	12	9	13	16	24	29	11	14	12
Y-72	14	14	31	23	11	11	13	11/18	12	10	14	14	22	29	11	16	12
Y-73	14	14	30	24	9	11	13	13/14	11	11	15	15	25	28	11	15	12
Y-75	13	13	29	22	11	11	12	13/15	12	11	14	15	26	29	11	15	12
Y-76	14	13	29	23	10	13	13	12/14	12	10	14	14	25	29	11	15	13
Y-77	13	13	31	23	10	11	14	15/16	11	11	15	15	25	29	11	15	12
Y-78	14	13	29	23	12	13	13	11/14	12	11	15	15	25	29	11	15	12
Y-79	13	14	30	24	9	11	13	12/13	11	11	15	15	28	29	11	15	12
Y-80	14	13	29	24	11	11	12	13/17	12	11	15	15	25	29	11	15	12
Y-81	14	12	28	24	10	13	13	10/16	12	11	15	15	25	29	11	14	12

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