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SHORT COMMUNICATION

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Y-chromosomal STR haplotypes in a Romanian population sample

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Abstract A set of seven STR-loci mapping on the male-specific region of the human Y chromosome (DYS19, DYS385, DYS389 I/II, DYS390, DYS391, DYS392, DYS393) were typed by means of two multiplex PCR reactions and capillary electrophoresis in a Romanian population sample of 104 unrelated males. Among the 97 different haplotypes observed, 92 were unique, while 5 occurred more than once. The observed haplotype diversity was 0.989.

Keywords Y-chromosome · STRs · Y-haplotypes · Romanian population

Introduction

DNA typing for male-specific polymorphisms is a well established molecular analysis procedure in the forensic practice. The interest for the Y chromosome STR systems is continuously increasing [1, 2]. In 1997 an international multicenter study proposed a panel of 9 Y-STR loci (DYS19, DYS385(1+2), DYS389 I/II, DYS390, DYS391, DYS392, DYS393) for standard Y-haplotyping [3, 4, 5] and these loci define the minimal Y haplotype in the Y-STR reference database (YHRD, http://ystr.charite.de). Various European populations have already been investigated, while there is a lack of information regarding the Y-specific STR polymorphisms in the Romanian population. The Romanians represent a quite compact and large latin-speaking population, geographically located in the south-east of Europe which for centuries has had intense social and cultural interactions with various European populations living outside and inside the Romanian borders. Previous Y-chromosome studies, including Romanian male samples, were focused on binary marker polymorphisms and revealed, in terms of haplogroup frequencies, that the Carpathian region is a break point in the gene geography of eastern central Europe, acting as a genetic boundary [6]. The aim of this study was to establish a Y-haplotype database by means of a set of seven Y-STRs (minimal haplotype) in a representative Romanian population sample.

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Material and methods

DNA samples

Whole EDTA blood samples were collected from 104 unrelated Romanian males, randomly selected among the patients of a Bucharest hospital. All the males selected for the study had a birth place in Romania (different regions from all over the country) and Romanian surnames. Members of the Romanian minority populations living in Romania (Hungarians, Germans etc.) were excluded from the study.

STR typing

DNA was extracted from whole blood samples using a proteinase K-Chelex protocol [7]. Amplification of the Y-STR loci was carried out using two commercial kits, the *genRES* DYSplex-1 kit (including the DYS390, DYS391, DYS389 I/II, DYS385 and amelogenin loci) and the *genRES* DYSplex-2 kit (including the DYS392, DYS393, DYS389 I/II, DYS19 loci) according to the manufacturer's protocols (Serac, Bad Homburg, Germany). The PCR products were separated by capillary electrophoresis on an ABI Prism 310 Genetic Analyzer (Applied Biosystems) together with the allelic ladders provided with the kit [8].

Table 1 Y chromosome haplotypes observed in the Romanian population sample (n=104)

Haplotype	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS385I/II		n
1	13	12	29	24	10	11	13	16	19	1
2	13	13	30	24	10	11	13	15	17	1
3	13	13	30	24	10	11	13	16	18	1
4	13	13	30	24	10	11	13	16	19	1
5	13	13	30	24	10	11	13	16	20	1
6	13	13	30	24	10	11	13	17	18	1
7	13	13	30	24	10	11	14	17	19	1
8	13	13	30	25	9	11	14	17	17	1
9	13	13	30	25	10	11	13	16	16	1
10	13	13	30	25	10	11	13	16	17	1
11	13	13	30	25	10	11	13	16	18	2
12	13	13	31	24	10	11	13	16	17	2
13	13	13	32	25	11	11	13	16	18	1
14	13	14	30	22	10	13	14	13	16	1
15	13	14	31	24	10	11	13	15	18	1
16	13	14	31	24	10	11	13	17	19	1
17	13	14	32	23	10	15	13	15	17	1
18	14	12	28	22	10	11	13	14	14	2
19	14	12	28	22	11	12	13	14	14	1
20	14	12	28	23	10	11	13	13	14	1
21	14	12	29	22	10	11	12	12	15	1
22	14	12	29	22	10	11	13	13	14	1
23	14	13	28	24	11	13	12	12	15	1
24	14	13	29	22	11	11	12	15	18	1
25	14	13	29	23	10	11	12	14	17	1
26	14	13	29	23	10	11	14	13	17	1
27	14	13	29	23	10	14	13	12	13	1
28	14	13	29	24	10	14	13	12	12	1
29	14	13	29	24	11	13	13	11	14	1
30	14	13	29	24	11	15	13	11	14	1
31	14	13	29	24	12	11	12	11	11	1
32	14	13	29	26	10	13	13	11	14	1
33	14	13	30	24	10	11	13	15	18	1
34	14	13	30	24	10	13	12	11	15	1
35	14	13	30	24	11	13	12	11	14	1
36	14	13	30	25	10	13	12	11	14	1
37	14	13	31	23	10	11	12	14	18	1
38	14	13	31	23	10	11	13	13	17	1
39	14	14	30	22	9	11	12	13	18	1
40	14	14	30	23	10	11	12	14	17	1
41	14	14	30	24	11	12	13	11	14	1
42	14	14	30	25	11	13	12	10	15	1
43	14	14	31	23	10	11	12	10	16	1
44	14	14	31	23	10	11	12	13	16	1
45	14	14	31	24	10	11	13	17	17	1
46	14	14	31	24	10	13	12	11	14	1
47	14	14	32	23	10	11	12	13	16	1
48	15	12	28	24	10	11	12	14	17	1
49	15	12	28	25	10	11	12	10	18	1
50	15	12	28	25	10	11	12	14	14	1
51	15	12	29	22	10	11	12	13	14	1
52	15	12	29	22	10	11	13	12	14	1
53	15	13	29	22	10	12	12	13	14	1
54	15	13	29	23	9	11	12	13	16	1
55	15	13	29	24	10	11	12	13	18	1
56	15	13	29	24	11	13	14	11	14	1
57	15	13	29	25	10	11	13	12	14	1

Table 1 (continued)

Haplotype	DYS19	DYS389 I	DYS389 II	DYS390	DYS391	DYS392	DYS393	DYS385I/II		n
58	15	13	30	24	10	11	12	13	17	1
59	15	13	30	24	11	11	13	11	14	1
60	15	13	30	25	11	11	13	11	14	1
61	15	13	31	23	9	11	12	13	16	1
62	15	13	31	24	10	11	13	14	15	1
63	15	13	31	25	11	11	13	11	14	1
64	15	13	32	24	10	11	13	14	14	1
65	15	14	30	22	10	11	12	15	17	3
66	15	14	31	24	10	11	13	12	14	1
67	15	14	31	24	10	13	13	12	18	1
68	15	14	31	25	10	11	13	13	18	1
69	16	12	28	22	10	10	14	15	18	1
70	16	12	28	22	10	11	12	12	14	1
71	16	12	28	24	10	11	12	13	17	1
72	16	12	29	24	11	12	13	14	15	1
73	16	13	29	23	10	12	15	14	17	1
74	16	13	29	24	10	11	13	11	15	1
75	16	13	29	25	10	11	13	11	14	1
76	16	13	30	24	11	11	13	13	14	1
77	16	13	30	25	11	11	13	11	13	1
78	16	13	31	24	11	11	13	14	15	3
79	16	13	32	24	11	11	13	11	14	1
80	16	13	32	24	11	11	13	13	15	1
81	16	13	32	25	11	11	13	14	15	1
82	16	13	33	24	11	11	14	14	15	1
83	16	14	30	24	9	11	12	16	18	1
84	16	14	31	25	10	11	13	11	15	1
85	16	14	32	24	9	11	13	11	14	1
86	16	14	32	24	10	11	13	14	15	1
87	16	14	32	24	11	11	13	14	15	1
88	15,16	12	29	22	10	11	14	13	14	1
89	15,16	13	31	23	9	11	14	13	16	1
90	17	12	29	23	10	11	12	14	17	1
91	17	13	29	25	11	11	13	11	14	1
92	17	13	30	25	11	11	13	11	13	1
93	17	13	30	25	11	11	13	11	14	1
94	17	13	31	24	10	11	13	14	15	1
95	17	13	31	24	11	11	13	14	15	1
96	17	13	31	24	11	11	13	14	17	1
97	17	13	31	24	11	11	13	11	13	1

Statistical calculations

The allele and haplotype frequencies were determined according to Kayser et al. [3]. The haplotype diversity (HD) for all seven STR loci, corresponding to the power of discrimination or exclusion chance for unrelated males, was calculated with the equation as $HD=1-\Sigma q^2_i$, using haplotype frequencies q_i [9].

Results and discussion

The seven Y-STR loci investigated show allele frequency distributions very similar to those reported in other European populations [3, 4]. We found two duplications at the DSY19 locus, corresponding to two unrelated individuals. Table 1 summarizes the haplotypes observed in our random sample of unrelated Romanian males and the corresponding frequencies. From the total 104 chromosomes investigated, 97 revealed distinct haplotypes. A total of 92 haplotypes were unique (88.46%), while the others were shared by 2 or 3 individuals: 3 haplotypes were observed twice and another 2 occurred 3 times. The haplotype 14-13-29-24-11-13-13-11, 14 (see Table 1 for order of systems), which is the most frequent in the European database, was found once in our samples (haplotype no. 29 listed in Table 1). The overall discrimination capacity of the seven Y-STR loci haplotypes studied in this Romanian population was 93.26% (calculated as the percentage of different haplotypes) and the haplotype diversity was 98.87%.

Table 2 Comparison of the two Y-quadruplex systems

PCR multiplex	Number of haplotypes detected (n=104)	Haplotype diversity
DYSplex-1 (DYS390, DYS391, DYS389 I/II, DYS385)	86	0.9863
DYSplex-2 (DYS392, DYS393, DYS389 I/II, DYS19)	63	0.9791
7 STR loci haplotype	97	0.9887

The two commercial quadruplex PCR systems *genRES* DYSplex-1 and *genRES* DYSplex-2 (Serac, Bad Homburg, Germany) showed different discrimination capacities in our population sample (Table 2). DYSplex-1 alone detected 86 different haplotypes and revealed a high haplotype diversity of 98.63%, mainly because of the presence of the polymorphic locus DYS385. In comparison, the DYSplex-2 showed only 63 different haplotypes in the same samples and a haplotype diversity of 97.91%. The data presented in the paper will soon be available on-line in the Y-STR reference database (http://www.ystr.charite.de).

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