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Announcement of population data

Y chromosome STRs in Croatians

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Abstract

Eight Y chromosome short tandem repeat (STR) polymorphisms (DYS19, DYS388, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393) were analyzed in the sample of 457 unrelated Croatian men. A general STR allelic frequency pattern in Croatians corresponds to other European populations with the exception of the loci DYS19 and DYS389II. The most frequent DYS19 allele was 16, while at the DYS389II the most frequent were alleles 30 and 31. The most frequent Y chromosome haplotype (16-13-13-31-24-11-11-13) was found in 33 individuals (7.22%). One hundred and seventy-four haplotypes (38.07%) were observed in single copies.

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Keywords: Y chromosome; Short tandem repeats (STR); Haplotypes; Croatia

Population: The studied sample included 457 unrelated healthy Croatian men (109 were from mainland Croatia and 348 from four Adriatic islands: Krk, 74; Brač, 49; Hvar, 91 and Korčula, 134).

Extraction: The whole blood was obtained by venipuncture and collected into EDTA tubes, followed by DNA extraction using the macromethod of Poncz et al. [1].

PCR: Two trinucleotide (DYS388 and DYS392) and six tetranucleotide loci (DYS19, DYS389I, DYS389II, DYS390, DYS391 and DYS393) were typed. PCR reactions were carried out in the thermocycler “Biometra UNO II” and performed in 20 µl reaction volume containing 100 ng genomic DNA, 2.5 mM MgCl₂, 75 mM Tris–HCl (pH 9.0), 100 µM each dNTP, 0.1 µM of each primer, and 2 U Taq DNA polymerase. The PCR cycling conditions used were those described by de Knijff et al. [2].

Typing: PCR products were resolved and detected on the 6% denaturing ReproGelTM High Resolution gels (Amersham Pharmacia Biotech) by using the Automatic

Laser Fluorescence ExpressTM DNA sequencer (Pharmacia Biotech AB). The Allele Links Version 1.00 software (Pharmacia Biotech) was used to analyze fragment sizes. The alleles were named according to the number of repeat units which was based on the sequenced allele leaders as suggested by de Knijff et al. [2].

Results: The results are shown in Tables 1 and 2.

Data analysis: Allelic frequencies were estimated by gene counting method. Haplotype diversity within population was calculated using Arlequin 2.0 Package [3], according to the formula:

$$H = \frac{n}{n-1} \left(1 - \sum_{i=1}^k p_i^2 \right),$$

where n is the number of gene copies, k the number of haplotypes, and p_i is the sample frequency of the i th haplotype.

Locus diversity was estimated according to the same equation using allele frequencies instead of haplotype frequencies.

Other remarks: Majority of the eight studied loci showed the allelic frequency distribution (Table 1) similar to those previously reported in other European populations [4,5]. The exceptions were found at the loci DYS19 and DYS389II. At the DYS19 locus the allele 16 had the highest

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Table 1
Allele frequencies at eight Y chromosome STRs in Croatians

Locus	DYS19	Frequency	DYS388	Frequency	DYS389I	Frequency	DYS389II	Frequency	DYS390	Frequency	DYS391	Frequency	DYS392	Frequency	DYS393	Frequency
Alleles	13	0.109	11	0.046	12	0.096	28	0.044	19	0.002	9	0.013	10	0.004	11	0.011
	14	0.116	12	0.427	13	0.786	29	0.214	21	0.004	10	0.468	11	0.821	12	0.096
	15	0.184	13	0.483	14	0.116	30	0.311	22	0.103	11	0.490	12	0.042	13	0.823
	16	0.501	14	0.022	15	0.002	31	0.311	23	0.105	12	0.028	13	0.081	14	0.070
	17	0.090	15	0.015			32	0.105	24	0.521			14	0.007		
			16	0.007			33	0.015	25	0.243			15	0.044		
									26	0.022			16	0.002		

Table 2
Y STR haplotypes in Croatian population

Haplotype	<i>n</i>	<i>F</i>	DYS19	DYS388	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
1	1	0.0022	13	11	13	29	22	9	15	13
2	7	0.0153	13	11	13	29	22	10	15	13
3	4	0.0088	13	11	13	30	22	10	15	13
4	1	0.0022	13	11	13	30	22	11	15	13
5	1	0.0022	13	11	13	30	24	10	15	13
6	1	0.0022	13	11	13	30	25	10	15	13
7	1	0.0022	13	12	12	28	24	10	11	12
8	1	0.0022	13	12	12	29	24	11	11	13
9	1	0.0022	13	12	12	30	24	10	11	13
10	2	0.0044	13	12	13	29	22	10	15	13
11	1	0.0022	13	12	13	29	24	10	11	13
12	1	0.0022	13	12	13	29	24	11	13	12
13	1	0.0022	13	12	13	30	22	10	15	13
14	2	0.0044	13	12	13	30	22	10	15	14
15	1	0.0022	13	12	13	30	23	10	11	13
16	2	0.0044	13	12	13	30	23	11	11	13
17	1	0.0022	13	12	13	30	23	11	13	13
18	1	0.0022	13	12	13	30	24	10	11	12
19	3	0.0066	13	12	13	30	24	10	11	13
20	3	0.0066	13	12	13	30	24	11	11	13
21	1	0.0022	13	12	13	30	24	12	13	13
22	4	0.0088	13	12	13	30	25	10	11	13
23	1	0.0022	13	12	13	31	24	10	11	13
24	1	0.0022	13	12	13	31	24	11	11	14
25	1	0.0022	13	12	13	32	25	11	11	13
26	1	0.0022	13	12	14	30	22	11	13	13
27	1	0.0022	13	13	13	31	24	10	11	13
28	1	0.0022	13	13	13	31	25	11	11	13
29	1	0.0022	13	14	12	29	22	10	11	13
30	1	0.0022	13	15	14	31	23	10	11	11
31	1	0.0022	13	15	14	31	25	10	11	12
32	1	0.0022	14	12	12	28	23	10	11	12
33	1	0.0022	14	12	12	28	23	10	11	14
34	1	0.0022	14	12	12	28	25	10	13	13
35	1	0.0022	14	12	12	29	24	10	12	13
36	1	0.0022	14	12	13	28	24	11	13	13
37	2	0.0044	14	12	13	29	24	11	13	13
38	1	0.0022	14	12	13	29	24	11	13	14
39	1	0.0022	14	12	13	29	24	11	14	14
40	1	0.0022	14	12	13	29	24	11	16	13
41	1	0.0022	14	12	13	30	23	11	13	13
42	1	0.0022	14	12	13	30	24	10	11	13
43	1	0.0022	14	12	13	30	24	10	13	13
44	1	0.0022	14	12	13	30	24	11	11	12

Table 2 (Continued)

Haplotype	<i>n</i>	<i>F</i>	DYS19	DYS388	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
45	2	0.0044	14	12	13	30	24	11	13	12
46	3	0.0066	14	12	13	30	25	10	13	12
47	1	0.0022	14	12	13	31	22	9	11	13
48	1	0.0022	14	12	14	30	23	11	13	13
49	2	0.0044	14	12	14	30	24	10	13	14
50	1	0.0022	14	12	14	31	23	11	11	14
51	1	0.0022	14	12	14	31	24	11	13	13
52	1	0.0022	14	12	15	31	24	10	13	14
53	1	0.0022	14	13	12	28	22	10	11	13
54	1	0.0022	14	13	12	30	22	10	11	13
55	1	0.0022	14	13	13	30	24	11	11	13
56	1	0.0022	14	13	13	31	24	10	11	13
57	3	0.0066	14	13	13	31	24	11	11	13
58	1	0.0022	14	13	13	32	24	11	11	12
59	1	0.0022	14	13	13	32	24	11	11	13
60	1	0.0022	14	13	14	30	22	10	11	13
61	1	0.0022	14	13	14	30	22	11	11	11
62	1	0.0022	14	13	14	30	22	11	11	13
63	1	0.0022	14	13	14	30	23	10	11	13
64	2	0.0044	14	13	14	30	23	11	11	13
65	1	0.0022	14	13	14	30	24	10	13	13
66	1	0.0022	14	13	14	32	24	11	11	13
67	2	0.0044	14	14	12	28	23	10	11	13
68	2	0.0044	14	14	12	28	24	10	11	13
69	1	0.0022	14	14	12	29	23	10	11	13
70	1	0.0022	14	14	13	30	23	11	13	14
71	1	0.0022	14	15	14	30	24	10	14	12
72	1	0.0022	14	16	13	29	23	9	11	12
73	2	0.0044	14	16	13	29	24	10	11	11
74	1	0.0022	15	11	12	29	21	11	11	13
75	1	0.0022	15	12	12	30	21	11	11	13
76	1	0.0022	15	12	13	28	24	11	13	13
77	3	0.0066	15	12	13	29	24	10	13	13
78	4	0.0088	15	12	13	29	25	10	11	13
79	1	0.0022	15	12	13	29	25	10	13	13
80	6	0.0131	15	12	13	29	25	11	11	13
81	1	0.0022	15	12	13	29	26	10	11	13
82	1	0.0022	15	12	13	30	22	11	11	14
83	1	0.0022	15	12	13	30	23	10	11	13
84	1	0.0022	15	12	13	30	23	11	11	13
85	1	0.0022	15	12	13	30	24	10	11	14
86	1	0.0022	15	12	13	30	24	12	11	13
87	3	0.0066	15	12	13	30	25	10	11	13
88	1	0.0022	15	12	13	30	25	11	11	13
89	1	0.0022	15	12	13	31	24	10	11	13
90	1	0.0022	15	12	13	31	24	11	11	13
91	1	0.0022	15	12	13	31	25	11	12	13
92	1	0.0022	15	12	13	32	24	11	11	13
93	1	0.0022	15	12	13	32	24	11	12	13
94	1	0.0022	15	12	14	31	25	11	11	13
95	1	0.0022	15	12	14	32	25	11	11	13
96	2	0.0044	15	13	12	28	22	10	11	13
97	1	0.0022	15	13	12	28	22	10	12	13
98	7	0.0153	15	13	12	29	22	10	11	13
99	1	0.0022	15	13	12	29	22	10	11	14
100	1	0.0022	15	13	12	29	22	10	12	13
101	1	0.0022	15	13	12	29	22	11	11	13

Table 2 (Continued)

Haplotype	<i>n</i>	<i>F</i>	DYS19	DYS388	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
102	1	0.0022	15	13	12	30	22	10	11	13
103	3	0.0066	15	13	13	29	23	10	12	13
104	1	0.0022	15	13	13	29	23	11	11	13
105	3	0.0066	15	13	13	30	24	10	11	13
106	1	0.0022	15	13	13	30	24	10	11	14
107	2	0.0044	15	13	13	30	24	11	11	13
108	1	0.0022	15	13	13	30	26	11	13	12
109	1	0.0022	15	13	13	31	24	10	11	12
110	2	0.0044	15	13	13	31	24	10	11	13
111	6	0.0131	15	13	13	31	24	11	11	12
112	9	0.0197	15	13	13	31	24	11	11	13
113	1	0.0022	15	13	13	31	24	11	13	12
114	2	0.0044	15	13	13	32	24	11	11	13
115	1	0.0022	15	13	14	30	24	11	11	13
116	1	0.0022	15	14	12	29	23	10	11	13
117	1	0.0022	15	15	12	28	23	10	12	12
118	1	0.0022	15	15	12	28	24	10	11	12
119	2	0.0044	16	11	13	31	24	11	11	13
120	1	0.0022	16	11	13	32	24	11	11	13
121	1	0.0022	16	11	14	31	24	11	11	13
122	1	0.0022	16	12	12	29	22	10	11	13
123	1	0.0022	16	12	12	29	23	11	11	13
124	1	0.0022	16	12	12	29	23	11	14	12
125	1	0.0022	16	12	12	29	24	11	11	13
126	1	0.0022	16	12	13	29	23	10	12	14
127	1	0.0022	16	12	13	29	24	10	11	13
128	2	0.0044	16	12	13	29	24	10	13	13
129	9	0.0197	16	12	13	29	25	10	11	13
130	1	0.0022	16	12	13	29	25	10	12	13
131	1	0.0022	16	12	13	29	25	10	13	12
132	1	0.0022	16	12	13	29	25	10	13	13
133	5	0.0109	16	12	13	29	25	11	11	13
134	1	0.0022	16	12	13	29	25	11	11	14
135	2	0.0044	16	12	13	30	23	10	11	14
136	1	0.0022	16	12	13	30	23	11	11	12
137	1	0.0022	16	12	13	30	24	10	10	13
138	3	0.0066	16	12	13	30	24	10	11	13
139	1	0.0022	16	12	13	30	24	10	13	12
140	1	0.0022	16	12	13	30	24	11	11	13
141	9	0.0197	16	12	13	30	25	10	11	13
142	2	0.0044	16	12	13	30	25	10	11	14
143	7	0.0153	16	12	13	30	25	11	11	13
144	1	0.0022	16	12	13	30	25	12	11	13
145	1	0.0022	16	12	13	30	26	10	11	14
146	1	0.0022	16	12	13	30	26	11	10	13
147	1	0.0022	16	12	13	30	26	11	11	13
148	1	0.0022	16	12	13	31	23	12	11	13
149	1	0.0022	16	12	13	31	24	10	11	12
150	3	0.0066	16	12	13	31	24	10	11	13
151	1	0.0022	16	12	14	30	24	11	13	13
152	1	0.0022	16	12	14	31	23	11	11	13
153	1	0.0022	16	12	14	31	23	12	11	13
154	1	0.0022	16	12	14	31	24	10	11	13
155	1	0.0022	16	12	14	31	24	12	11	13
156	8	0.0175	16	12	14	31	25	10	11	13
157	1	0.0022	16	12	14	31	25	10	11	14
158	2	0.0044	16	12	14	31	25	11	11	13
159	1	0.0022	16	12	14	32	23	10	11	13

Table 2 (Continued)

Haplotype	<i>n</i>	<i>F</i>	DYS19	DYS388	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
160	2	0.0044	16	12	14	32	24	10	11	13
161	1	0.0022	16	12	14	32	24	10	12	13
162	1	0.0022	16	12	14	32	24	11	11	13
163	2	0.0044	16	12	14	32	25	10	11	13
164	2	0.0044	16	12	14	32	25	11	11	13
165	1	0.0022	16	13	12	28	23	11	11	13
166	1	0.0022	16	13	12	30	22	10	11	13
167	1	0.0022	16	13	12	30	24	11	11	13
168	1	0.0022	16	13	13	28	19	11	11	13
169	1	0.0022	16	13	13	29	23	10	11	14
170	1	0.0022	16	13	13	29	24	10	11	13
171	6	0.0131	16	13	13	29	25	10	11	13
172	1	0.0022	16	13	13	30	23	10	11	13
173	7	0.0153	16	13	13	30	24	10	11	13
174	1	0.0022	16	13	13	30	24	11	11	12
175	12	0.0263	16	13	13	30	24	11	11	13
176	1	0.0022	16	13	13	30	24	11	12	13
177	1	0.0022	16	13	13	30	25	11	11	13
178	1	0.0022	16	13	13	31	22	10	11	13
179	1	0.0022	16	13	13	31	22	11	11	13
180	2	0.0044	16	13	13	31	24	9	11	12
181	1	0.0022	16	13	13	31	24	9	11	13
182	11	0.0241	16	13	13	31	24	10	11	13
183	3	0.0066	16	13	13	31	24	11	11	12
184	33	0.0722	16	13	13	31	24	11	11	13
185	2	0.0044	16	13	13	31	24	11	11	14
186	2	0.0044	16	13	13	31	24	11	12	13
187	2	0.0044	16	13	13	31	24	12	11	12
188	2	0.0044	16	13	13	31	24	12	11	13
189	2	0.0044	16	13	13	31	25	10	11	13
190	2	0.0044	16	13	13	31	25	11	11	12
191	2	0.0044	16	13	13	31	25	11	11	13
192	1	0.0022	16	13	13	32	22	11	11	12
193	1	0.0022	16	13	13	32	23	10	11	13
194	2	0.0044	16	13	13	32	23	11	11	12
195	1	0.0022	16	13	13	32	23	12	11	13
196	5	0.0109	16	13	13	32	24	10	11	13
197	10	0.0219	16	13	13	32	24	11	11	13
198	1	0.0022	16	13	13	32	24	11	12	13
199	1	0.0022	16	13	13	32	25	10	11	13
200	1	0.0022	16	13	13	32	25	11	11	13
201	4	0.0088	16	13	13	33	24	11	11	13
202	1	0.0022	16	13	14	31	24	10	11	13
203	1	0.0022	16	13	14	31	24	11	11	13
204	1	0.0022	16	13	14	31	25	10	11	13
205	1	0.0022	16	13	14	31	25	11	11	13
206	1	0.0022	16	13	14	32	24	11	11	12
207	1	0.0022	16	13	14	32	24	11	11	13
208	1	0.0022	16	14	13	31	24	11	11	13
209	1	0.0022	16	14	13	33	24	11	11	13
210	1	0.0022	16	15	12	28	23	10	12	11
211	1	0.0022	16	15	13	30	24	11	11	13
212	1	0.0022	17	11	13	29	25	11	11	13
213	1	0.0022	17	12	13	29	24	11	11	13
214	1	0.0022	17	12	13	29	25	10	11	13
215	1	0.0022	17	12	13	29	25	11	11	13
216	1	0.0022	17	12	13	30	24	11	11	13
217	2	0.0044	17	12	13	30	25	10	11	13

Table 2 (Continued)

Haplotype	<i>n</i>	<i>F</i>	DYS19	DYS388	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393
218	1	0.0022	17	12	13	30	25	10	11	14
219	1	0.0022	17	12	13	30	25	11	11	13
220	5	0.0109	17	12	13	30	26	10	11	13
221	1	0.0022	17	12	13	31	24	10	11	13
222	1	0.0022	17	12	13	31	24	11	11	13
223	1	0.0022	17	12	13	31	25	10	11	13
224	1	0.0022	17	12	13	32	23	11	11	13
225	1	0.0022	17	12	13	33	22	10	12	14
226	1	0.0022	17	12	14	31	25	10	11	13
227	1	0.0022	17	13	12	28	24	11	11	14
228	1	0.0022	17	13	12	31	24	11	11	14
229	1	0.0022	17	13	13	29	23	11	12	13
230	2	0.0044	17	13	13	29	24	11	11	13
231	1	0.0022	17	13	13	29	25	10	11	14
232	1	0.0022	17	13	13	29	25	11	11	14
233	1	0.0022	17	13	13	30	23	11	13	13
234	3	0.0066	17	13	13	30	24	11	11	13
235	1	0.0022	17	13	13	31	23	12	11	13
236	1	0.0022	17	13	13	31	24	10	11	13
237	3	0.0066	17	13	13	31	24	11	11	13
238	1	0.0022	17	13	13	31	25	11	11	13
239	2	0.0044	17	13	13	32	24	11	11	13
240	1	0.0022	17	13	13	32	24	12	11	13
241	1	0.0022	17	13	13	33	24	11	11	13
LDV			0.683	0.582	0.361	0.749	0.649	0.540	0.317	0.310

n: number of individuals; *F*: frequency of each haplotype; LD: locus diversity value.

frequency in Croatians (0.501) as opposed to the reported highest frequency of allele 14 in Europe [5]. At the locus DYS389II the most frequent allele previously reported in Europe was 29 [4,5], but we noticed 30 and 31 as the most common in Croatian sample.

Haplotypes and locus diversity for the total investigated sample is presented in Table 2. The highest diversity that corresponds to the highest discrimination values were found for the loci DYS389II (0.749), followed by DYS19 (0.683) and DYS390 (0.649). If compared with values observed for some European populations, e.g. Spain, Italy and Germany, diversity values found for loci DYS19 and DYS389II are higher, whereas values for DYS392 and DYS393 are lower [6–8]. The rest of the diversity values are in concordance with the previously reported results [6–8].

Out of the total sample 241 different Y chromosome haplotypes were identified, 172 (37.64%) were observed in single copies. The most frequent one 16-13-13-31-24-11-11-13 (DYS19–DYS388–DYS389I–DYS389II–DYS390–DYS391–DYS392–DYS393) was found in 33 copies (7.22%), followed by haplotypes 16-13-13-30-24-11-11-13 and 16-13-13-31-24-10-11-13 in 12 and 11 copies, i.e. 2.6 and 2.4%, respectively. Haplotype diversity using all loci for the total sample was estimated at 0.9896 ± 0.0017 . In searching short tandem repeat (STR) database <http://www.ystr.org> [9] the most frequent haplotype

(16-13-13-31-24-11-11-13) is the most frequent in a sample of 150 individuals from Croatia and it is also present in central and eastern Europe populations.

This study is reported by following the guidelines for publication of population data requested by the journal [10].

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