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ORIGINAL ARTICLE

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Y-chromosomal STR haplotypes in a population sample from southwest Germany (Freiburg area)

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Abstract In the present study Y-chromosomal short tandem repeat (Y-STR) haplotypes of 433 unrelated male individuals from southwest Germany (Freiburg area) were analyzed. Population data for the loci DYS19, 389I, 389II, 390, 391, 392, 393, 385 and YCA II are given. Analysis of these loci provides the data that is defined as the "extended haplotype" and is available in the on-line Y-STR haplotpye reference database (YHRD) at http://ystr.org. Special emphasis is placed on the sequence and nomenclature of YCA II.

Keywords Y-STR · YCA II · Haplotype · Nomenclature

Introduction

In recent years Y-chromosomal short tandem repeats (Y-STRs) [1, 2, 3] have received increasing interest in forensic medicine. The specifically male patterns facilitate the assessment of mixed stains, e.g., in sexual assault cases [4, 5, 6, 7]. As an analogue to mitochondrial DNA, Y-chromosomal polymorphisms are also suitable for evaluating paternal lineages in kinship testing, especially in deficiency cases and for the development of models and theories in evolutionary genetics [8, 9, 10]. The number of Y-STRs that are suitable for forensic purposes is rising and their inclusion in forensic casework is a matter of discussion [11, 12], but the loci which form the so-called minimal and extended haplotypes can be referred to as the basis for forensic Y-STR haplotyping [13]. In this study we present population data for southwest Germany (Freiburg area) that were collected from 433 unrelated male individuals.

Within the context of forensic applications, the collection and databasing of as many Y-chromosomal haplotypes as possible is essential for reliable frequency estimates, especially for rare haplotypes. Another aim is the coverage of a maximum of different populations [13]. For appropriate databasing the application of standardized nomenclatures for the loci that are routinely used is crucial and should follow approved guidelines [14]. The nomenclature of the highly informative diallelic locus YCA II has not yet been standardized [2, 3, 15, 16, 17]. This is partly due to the different sequences that have been published for this locus [15, 16, 18], and partly due to considerations of whether or not to assign fragments to certain loci (YCAII a/b) as previously suggested [2, 10, 17].

Materials and methods

Whole blood samples or buccal swabs were collected from 433 unrelated male individuals. As a prerequisite, the individuals tested as well as their fathers had to have German surnames, and their place of birth had to be in the Freiburg area in southwest Germany. Genomic DNA was prepared according to standard procedures [19, 20]. For each sample a quadruplex (DYS19, 389I, 389II, 390) and a triplex (DYS391, 392, 393) reaction were performed on a thermocycler (PTC-200, MJ Research Inc.) as decribed elsewhere [3]. For DYS385 and YCA II single reactions were performed as previously described [16, 21]. PCR fragments were separated and detected on an ABI 373A DNA sequencer (Applied Biosystems, Weiterstadt, Germany).

Allele and haplotype frequencies for each allele/haplotype were obtained by simple counting and division methods. Estimates of gene diversity and haplotype diversity values were calculated according to Nei [22].

Results and discussion

Among the 433 unrelated male individuals, 340 different extended Y-chromosomal haplotypes were found (Table 1) and Table 2 shows the 15 most common Y-STR haplotypes found in the study sample. The majority of haplotypes (86.47%) were found only once and haplotype frequencies ranged between 0.23% and 3.93%. By analyzing

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Table 1 Extended Y chromosomal STR haplotypes found among unrelated male individuals from south west Germany (*N*=433). Allelic information is given in the format DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385, YCAII (*HT* haplotype number, *n* number of individuals observed for each given haplotype)

HT	Allelic information	n
1	13, 12, 28, 22, 10, 16, 11, 12–17, 18–22	1
2	13, 12, 28, 23, 11, 13, 13, 11–14, 19–23	1
3	13, 12, 29, 22, 11, 11, 12, 13–16, 19–22	1
4	13, 12, 29, 25, 9, 11, 12, 12–18, 19–22	1
5	13, 13, 28, 22, 10, 15, 13, 15–16, 19–19	1
6	13, 13, 28, 25, 10, 13, 14, 11–14, 19–23	1
7	13, 13, 29, 24, 10, 13, 13, 11–14, 19–23	1
8	13, 13, 29, 24, 11, 13, 13, 11–11, 19–23	1
9	13, 13, 29, 24, 11, 13, 13, 11–14, 19–23	1
10	13, 13, 30, 22, 10, 11, 13, 17–18, 19–21	1
11	13, 13, 30, 23, 10, 11, 12, 17–17, 19–21	1
12	13, 13, 30, 24, 10, 11, 13, 14–18, 19–21	1
13	13, 13, 30, 24, 10, 11, 13, 16–17, 19–21	1
14	13, 13, 30, 24, 10, 11, 13, 16–18, 19–21	2
15	13, 13, 30, 24, 10, 11, 13, 16–19, 19–21	2
16	13, 13, 30, 24, 10, 11, 13, 17–18, 19–21	1
17	13, 13, 30, 24, 10, 13, 13, 11–14, 19–22	1
18	13, 13, 30, 24, 11, 13, 13, 11–11, 19–23	1
19	13, 13, 30, 25, 10, 11, 14, 16–18, 19–21	1
20	13, 13, 30, 25, 12, 11, 14, 16–17, 19–21	1
21	13, 13, 31, 22, 10, 11, 12, 12–16, 19–21	1
22	13, 13, 31, 24, 10, 11, 12, 17–18, 19–21	1
23	13, 13, 31, 24, 10, 11, 13, 16–20, 19–21	1
24	13, 13, 31, 24, 10, 13, 13, 11–14, 19–23	1
25	13, 13, 31, 24, 11, 11, 13, 16–19, 21–21	1
26	13, 13, 31, 24, 9, 11, 13, 13–14, 19–22	1
27	13, 14, 30, 24, 10, 11, 13, 14–14, 19–22	1
28	13, 14, 30, 24, 9, 11, 13, 13–14, 19–22	1
29	13, 14, 31, 24, 10, 10, 13, 16–18, 19–21	1
30	13, 14, 31, 24, 10, 11, 13, 16–18, 19–21	2
31	13, 14, 32, 25, 12, 11, 13, 16–17, 19–21	1
32	14, 12, 27, 22, 10, 11, 13, 13–14, 19–21	1
33	14, 12, 27, 24, 11, 13, 13, 11–14, 19–23	2
34	14, 12, 27, 24, 11, 13, 14, 11–14, 19–23	1
35	14, 12, 27, 24, 11, 13, 14, 12–14, 19–23	1
36	14, 12, 28, 22, 10, 11, 13, 12–13, 19–21	1
37	14, 12, 28, 22, 10, 11, 13, 12–15, 19–21	1
38	14, 12, 28, 22, 10, 11, 13, 13–13, 19–21	2
39	14, 12, 28, 22, 10, 11, 13, 13–14, 19–21	3
40	14, 12, 28, 22, 10, 11, 13, 13–15, 19–21	1
41	14, 12, 28, 22, 10, 11, 13, 13–17, 18–21	1
42	14, 12, 28, 22, 10, 11, 13, 14–14, 19–21	1
43	14, 12, 28, 22, 10, 11, 13, 14–15, 19–21	1
44	14, 12, 28, 22, 10, 11, 14, 13–14, 19–21	3
45	14, 12, 28, 22, 10, 12, 13, 12–13, 19–21	1
46	14, 12, 28, 22, 11, 10, 13, 12–15, 19–21	1
47	14, 12, 28, 22, 11, 11, 12, 13–14, 19–21	1
48	14, 12, 28, 22, 11, 11, 13, 13–14, 19–21	1
49	14, 12, 28, 22, 11, 11, 13, 13–15, 19–21	1
50	14, 12, 28, 22, 11, 11, 13, 14–14, 19–21	1
51	14, 12, 28, 22, 13, 11, 13, 13–14, 19–21	1
52	14, 12, 28, 23, 10, 11, 13, 13–13, 19–21	1
53	14, 12, 28, 23, 10, 11, 13, 13–14, 18–21	1
54	14, 12, 28, 23, 10, 11, 13, 13–14, 19–21	4
55	14, 12, 28, 23, 10, 11, 13, 14–14, 19–21	1
56	14, 12, 28, 23, 10, 11, 13, 14–15, 19–21	1
57	14, 12, 28, 23, 10, 11, 15, 13–14, 19–21	1
58	14, 12, 28, 23, 10, 13, 12, 11–15, 19–23	1
59	14, 12, 28, 24, 10, 13, 13, 11–15, 19–20	1
60	14, 12, 28, 24, 11, 13, 13, 11–11, 18–23	1
61	14, 12, 28, 24, 11, 13, 13, 11–14, 19–23	1
62	14, 12, 28, 24, 11, 13, 13, 11–15, 19–23	1
63	14, 12, 28, 24, 11, 13, 13, 12–14, 19–23	1
64	14, 12, 29, 22, 10, 11, 13, 13–14, 19–21	2
65	14, 12, 29, 22, 11, 12, 13, 13–13, 19–21	1
66	14, 12, 29, 23, 11, 12, 13, 12–14, 19–23	1
67	14, 12, 29, 25, 11, 13, 13, 11–15, 19–23	1
68	14, 12, 30, 22, 10, 11, 14, 14–16, 18–20	1
69	14, 12, 30, 23, 10, 11, 13, 14–14, 19–21	1
70	14, 13, 28, 22, 11, 13, 13, 11–14, 19–23	2
71	14, 13, 28, 23, 10, 13, 11, 11–15, 19–23	1
72	14, 13, 28, 23, 10, 13, 13, 11–15, 19–23	1
73	14, 13, 28, 23, 11, 13, 14, 11–15, 19–23	1
74	14, 13, 28, 24, 10, 13, 13, 11–14, 19–23	1
75	14, 13, 28, 24, 10, 13, 13, 12–13, 19–23	1
76	14, 13, 28, 24, 11, 13, 12, 11–14, 19–23	1
77	14, 13, 28, 24, 11, 13, 13, 11–14, 19–23	1
78	14, 13, 28, 24, 11, 13, 13, 12–13, 19–23	1
79	14, 13, 28, 24, 11, 13, 15, 11–14, 19–23	1
80	14, 13, 28, 25, 11, 12, 13, 11–14, 19–19	1
81	14, 13, 29, 22, 10, 11, 13, 13–14, 19–21	1
82	14, 13, 29, 22, 10, 11, 13, 13–15, 19–21	1
83	14, 13, 29, 22, 10, 11, 13, 14–15, 19–21	1
84	14, 13, 29, 22, 10, 11, 15, 12–13, 17–17	1
85	14, 13, 29, 23, 10, 11, 12, 13–16, 19–22	2
86	14, 13, 29, 23, 10, 11, 12, 13–16, 21–22	1
87	14, 13, 29, 23, 10, 11, 12, 14–15, 19–23	1
88	14, 13, 29, 23, 10, 11, 13, 13–14, 19–21	1
89	14, 13, 29, 23, 10, 13, 12, 11–14, 19–23	1
90	14, 13, 29, 23, 10, 13, 13, 11–14, 19–22	1
91	14, 13, 29, 23, 10, 13, 13, 11–14, 19–23	4
92	14, 13, 29, 23, 10, 13, 13, 11–16, 19–23	1
93	14, 13, 29, 23, 10, 13, 13, 12–14, 19–23	1
94	14, 13, 29, 23, 10, 14, 13, 11–14, 19–19	1
95	14, 13, 29, 23, 10, 14, 13, 11–14, 19–23	2
96	14, 13, 29, 23, 11, 13, 12, 11–13, 19–22	1
97	14, 13, 29, 23, 11, 13, 13, 10–14, 19–19	1
98	14, 13, 29, 23, 11, 13, 13, 11–11, 19–22	1
99	14, 13, 29, 23, 11, 13, 13, 11–12, 19–23	2
100	14, 13, 29, 23, 11, 13, 13, 11–13, 19–23	3
101	14, 13, 29, 23, 11, 13, 13, 11–14, 19–21	1
102	14, 13, 29, 23, 11, 13, 13, 11–14, 19–22	1
103	14, 13, 29, 23, 11, 13, 13, 11–14, 19–23	10
104	14, 13, 29, 23, 11, 13, 13, 11–15, 19–23	2
105	14, 13, 29, 23, 11, 13, 13, 14–15, 23–23	1
106	14, 13, 29, 23, 11, 13, 14, 11–13, 18–23	1

Table 1 (continued)

HT	Allelic information	n
107	14, 13, 29, 23, 11, 13, 14, 11–14, 19–23	1
108	14, 13, 29, 23, 11, 13, 14, 11–15, 19–23	1
109	14, 13, 29, 23, 11, 14, 13, 11–14, 19–23	1
110	14, 13, 29, 23, 12, 13, 13, 11–14, 19–21	1
111	14, 13, 29, 24, 10, 13, 12, 11–14, 19–23	1
112	14, 13, 29, 24, 10, 13, 13, 11–14, 19–23	9
113	14, 13, 29, 24, 10, 13, 13, 11–15, 19–23	2
114	14, 13, 29, 24, 10, 13, 13, 11–16, 19–22	1
115	14, 13, 29, 24, 10, 13, 13, 12–14, 19–23	1
116	14, 13, 29, 24, 10, 13, 13, 12–15, 19–23	1
117	14, 13, 29, 24, 10, 13, 14, 11–14, 19–23	1
118	14, 13, 29, 24, 10, 13, 14, 11–15, 19–23	1
119	14, 13, 29, 24, 10, 14, 13, 10–15, 19–23	1
120	14, 13, 29, 24, 10, 14, 13, 11–14, 19–24	1
121	14, 13, 29, 24, 10, 14, 13, 11–15, 19–23	1
122	14, 13, 29, 24, 11, 12, 14, 11–16, 19–22	2
123	14, 13, 29, 24, 11, 13, 12, 11–12, 19–23	2
124	14, 13, 29, 24, 11, 13, 12, 11–14, 19–23	2
125	14, 13, 29, 24, 11, 13, 13, 11–11, 19–23	2
126	14, 13, 29, 24, 11, 13, 13, 11–14, 18–23	1
127	14, 13, 29, 24, 11, 13, 13, 11–14, 19–22	1
128	14, 13, 29, 24, 11, 13, 13, 11–14, 19–23	17
129	14, 13, 29, 24, 11, 13, 13, 11–14, 19–24	1
130	14, 13, 29, 24, 11, 13, 13, 11–15, 19–23	6
131	14, 13, 29, 24, 11, 13, 13, 11–16, 19–23	3
132	14, 13, 29, 24, 11, 13, 13, 12–15, 19–23	1
133	14, 13, 29, 24, 11, 13, 13, 14–14, 19–23	1
134	14, 13, 29, 24, 11, 13, 14, 11–14, 19–22	1
135	14, 13, 29, 24, 11, 13, 14, 11–14, 19–23	1
136	14, 13, 29, 24, 11, 15, 13, 12–15, 19–23	1
137	14, 13, 29, 24, 12, 13, 13, 11–14, 19–23	1
138	14, 13, 29, 24, 12, 13, 13, 11–16, 19–23	1
139	14, 13, 29, 24, 12, 13, 13, 13–14, 19–23	1
140	14, 13, 29, 24, 9, 11, 12, 12–16, 19–23	1
141	14, 13, 29, 24, 9, 13, 13, 11–15, 19–23	1
142	14, 13, 29, 25, 10, 11, 12, 12–12, 22–23	1
143	14, 13, 29, 25, 10, 13, 13, 11–13, 19–23	1
144	14, 13, 29, 25, 10, 14, 13, 11–14, 19–23	1
145	14, 13, 29, 25, 11, 13, 13, 11–14, 19–23	3
146	14, 13, 29, 25, 11, 13, 13, 11–15, 19–24	1
147	14, 13, 29, 25, 12, 13, 13, 11–15, 19–23	1
148	14, 13, 29, 27, 11, 14, 13, 11–14, 19–23	1
149	14, 13, 30, 22, 10, 11, 14, 11–13, 17–17	1
150	14, 13, 30, 22, 11, 13, 13, 11–13, 19–23	1
151	14, 13, 30, 23, 10, 11, 12, 11–16, 22–22	1
152	14, 13, 30, 23, 10, 11, 12, 13–18, 21–22	1
153	14, 13, 30, 23, 10, 11, 12, 13–18, 22–22	1
154	14, 13, 30, 23, 11, 11, 12, 13–15, 22–22	1
155	14, 13, 30, 23, 11, 13, 13, 11–14, 19–21	2
156	14, 13, 30, 23, 11, 13, 13, 11–14, 19–23	3
157	14, 13, 30, 24, 11, 11, 13, 11–14, 19–23	1
158	14, 13, 30, 24, 11, 13, 13, 11–14, 19–23	2
159	14, 13, 30, 24, 11, 13, 14, 11–13, 19–23	1
160	14, 13, 30, 25, 10, 13, 13, 11–11, 19–22	1
161	14, 13, 30, 25, 11, 13, 11, 10–14, 18–23	1

Table 1 (continued)

HT	Allelic information	n
162	14, 13, 30, 25, 12, 13, 13, 11–14, 19–23	1
163	14, 13, 31, 23, 10, 11, 14, 17–17, 19–22	1
164	14, 13, 31, 24, 11, 11, 13, 16–18, 19–21	1
165	14, 13, 31, 24, 11, 13, 14, 12–14, 19–23	1
166	14, 14, 29, 23, 12, 13, 13, 11–14, 19–23	1
167	14, 14, 30, 22, 10, 11, 13, 13–14, 17–21	1
168	14, 14, 30, 23, 10, 11, 12, 12–18, 18–22	1
169	14, 14, 30, 23, 10, 12, 13, 11–15, 19–21	1
170	14, 14, 30, 23, 10, 12, 13, 12–13, 19–21	1
171	14, 14, 30, 23, 11, 13, 13, 11–14, 19–22	1
172	14, 14, 30, 23, 11, 13, 13, 11–14, 19–23	3
173	14, 14, 30, 23, 11, 14, 13, 11–14, 19–23	1
174	14, 14, 30, 23, 11, 14, 14, 11–13, 18–20	1
175	14, 14, 30, 23, 12, 13, 13, 11–13, 21–23	1
176	14, 14, 30, 24, 10, 11, 12, 13–15, 19–22	1
177	14, 14, 30, 24, 10, 13, 13, 11–14, 19–22	1
178	14, 14, 30, 24, 10, 13, 13, 11–14, 19–24	1
179	14, 14, 30, 24, 11, 13, 13, 11–11, 19–23	1
180	14, 14, 30, 24, 11, 13, 13, 11–14, 19–23	1
181	14, 14, 30, 24, 11, 13, 13, 12–14, 19–23	2
182	14, 14, 30, 24, 11, 13, 14, 10–14, 19–23	1
183	14, 14, 30, 24, 11, 13, 14, 11–14, 19–23	1
184	14, 14, 30, 24, 11, 14, 13, 11–14, 19–23	1
185	14, 14, 30, 24, 12, 13, 13, 11–14, 18–23	1
186	14, 14, 31, 23, 10, 11, 12, 13–17, 19–25	1
187	14, 14, 31, 23, 10, 13, 13, 11–13, 19–23	1
188	14, 14, 31, 23, 11, 13, 13, 12–14, 19–23	1
189	14, 14, 31, 24, 11, 13, 13, 11–14, 19–23	1
190	14, 14, 31, 24, 11, 13, 13, 12–14, 19–23	1
191	14, 14, 31, 25, 10, 11, 12, 13–15, 19–22	1
192	14, 14, 32, 23, 10, 12, 14, 15–16, 19–21	1
193	14, 14, 32, 23, 11, 11, 12, 13–19, 19–22	1
194	14, 15, 31, 24, 11, 13, 14, 10–14, 19–23	1
195	(14, 15), 13, 29, 24, 11, 13, 13, 11–14, 19–23	1
196	15, 12, 27, 24, 10, 11, 13, 14–17, 20–20	1
197	15, 12, 28, 22, 10, 11, 13, 12–14, 19–20	1
198	15, 12, 28, 22, 10, 11, 13, 13–14, 19–21	2
199	15, 12, 28, 22, 10, 11, 13, 14–14, 19–21	2
200	15, 12, 28, 22, 10, 11, 13, 14–14, 20–20	1
201	15, 12, 28, 22, 9, 11, 14, 14–14, 20–20	1
202	15, 12, 28, 23, 10, 11, 13, 13–14, 19–21	1
203	15, 12, 28, 23, 10, 11, 13, 13–15, 19–21	1
204	15, 12, 28, 23, 10, 11, 13, 14–14, 19–23	1
205	15, 12, 28, 23, 10, 11, 14, 14–14, 19–21	1
206	15, 12, 28, 23, 10, 14, 13, 13–14, 19–23	1
207	15, 12, 28, 24, 10, 11, 12, 13–14, 19–20	1
208	15, 12, 28, 24, 10, 11, 12, 13–17, 19–20	1
209	15, 12, 28, 24, 10, 11, 12, 13–17, 20–20	2
210	15, 12, 28, 24, 10, 11, 12, 14–17, 19–20	1
211	15, 12, 28, 24, 10, 11, 12, 15–17, 19–20	1
212	15, 12, 28, 24, 11, 13, 13, 11–13, 19–23	1
213	15, 12, 28, 24, 11, 13, 13, 11–14, 19–23	3
214	15, 12, 28, 24, 11, 13, 13, 11–15, 20–23	1
215	15, 12, 28, 25, 10, 11, 12, 14–18, 19–20	1
216	15, 12, 28, 25, 11, 11, 13, 11–14, 19–19	1

Table 1 (continued)

HT	Allelic information	n
217	15, 12, 29, 22, 10, 11, 13, 12–14, 19–20	1
218	15, 12, 29, 22, 10, 11, 13, 13–14, 19–21	1
219	15, 12, 29, 22, 10, 11, 13, 14–14, 20–20	1
220	15, 12, 29, 22, 10, 11, 14, 12–14, 19–20	1
221	15, 12, 29, 22, 10, 11, 14, 13–14, 20–20	1
222	15, 12, 29, 22, 10, 11, 14, 14–14, 19–19	1
223	15, 12, 29, 22, 10, 11, 14, 14–14, 20–20	2
224	15, 12, 29, 22, 10, 11, 14, 14–15, 20–20	1
225	15, 12, 29, 22, 10, 11, 15, 13–14, 20–20	1
226	15, 12, 29, 23, 10, 7, 13, 13–15, 19–21	1
227	15, 12, 29, 24, 10, 11, 12, 10–17, 19–20	1
228	15, 12, 29, 24, 10, 11, 12, 15–18, 19–20	1
229	15, 12, 29, 24, 11, 13, 13, 11–13, 19–23	1
230	15, 12, 29, 25, 10, 11, 13, 11–14, 19–23	1
231	15, 12, 30, 21, 11, 11, 14, 12–14, 19–20	1
232	15, 12, 30, 22, 10, 11, 13, 12–13, 19–20	1
233	15, 12, 30, 22, 10, 11, 14, 14–14, 20–20	1
234	15, 12, 30, 24, 10, 11, 13, 19–19, 19–22	1
235	15, 12, 30, 25, 11, 11, 13, 11–14, 19–23	1
236	15, 13, 28, 21, 11, 13, 13, 11–14, 19–23	1
237	15, 13, 28, 23, 11, 13, 13, 11–14, 21–23	1
238	15, 13, 28, 24, 11, 13, 13, 11–11, 19–23	1
239	15, 13, 28, 24, 11, 14, 13, 11–14, 19–23	1
240	15, 13, 28, 25, 10, 11, 14, 11–14, 19–23	1
241	15, 13, 29, 22, 10, 11, 13, 14–14, 19–21	1
242	15, 13, 29, 22, 10, 11, 14, 14–15, 20–20	1
243	15, 13, 29, 22, 10, 13, 14, 16–17, 19–21	1
244	15, 13, 29, 23, 10, 11, 12, 13–18, 19–22	1
245	15, 13, 29, 23, 10, 11, 13, 14–14, 19–21	1
246	15, 13, 29, 23, 10, 12, 15, 16–16, 19–21	1
247	15, 13, 29, 23, 10, 13, 13, 11–15, 19–23	1
248	15, 13, 29, 23, 11, 12, 14, 15–16, 19–19	1
249	15, 13, 29, 23, 11, 13, 13, 11–14, 19–23	2
250	15, 13, 29, 23, 11, 13, 13, 11–15, 19–23	1
251	15, 13, 29, 23, 11, 13, 14, 11–14, 19–23	2
252	15, 13, 29, 23, 9, 11, 12, 13–16, 20–22	1
253	15, 13, 29, 24, 10, 12, 15, 16–16, 19–19	1
254	15, 13, 29, 24, 10, 13, 13, 11–15, 19–23	1
255	15, 13, 29, 24, 11, 13, 13, 10–14, 19–24	1
256	15, 13, 29, 24, 11, 13, 13, 11–14, 19–23	3
257	15, 13, 29, 24, 9, 11, 12, 13–16, 19–22	1
258	15, 13, 29, 24, 9, 11, 12, 13–17, 19–22	1
259	15, 13, 29, 26, 10, 11, 14, 11–14, 19–23	1
260	15, 13, 30, 22, 10, 11, 13, 14–14, 19–21	1
261	15, 13, 30, 22, 10, 11, 14, 13–14, 19–20	1
262	15, 13, 30, 23, 10, 11, 12, 12–16, 19–22	1
263	15, 13, 30, 23, 11, 13, 13, 11–15, 19–23	1
264	15, 13, 30, 23, 11, 14, 14, 11–13, 18–20	1
265	15, 13, 30, 25, 10, 11, 13, 11–14, 19–23	2
266	15, 13, 30, 25, 10, 11, 13, 15–15, 19–23	1
267	15, 13, 30, 25, 11, 11, 13, 11–14, 19–21	1
268	15, 13, 31, 23, 10, 12, 14, 15–15, 19–21	2
269	15, 13, 31, 26, 10, 11, 13, 11–14, 19–23	1
270	15, 13, 32, 25, 11, 11, 13, 11–15, 19–23	1
271	15, 14, 29, 24, 10, 14, 13, 11–14, 19–23	1
272	15, 14, 30, 22, 10, 11, 12, 15–16, 19–21	1
273	15, 14, 30, 22, 10, 11, 12, 15–17, 19–21	1
274	15, 14, 30, 22, 10, 11, 12, 15–18, 19–21	1
275	15, 14, 30, 23, 10, 12, 15, 15–15, 19–21	1
276	15, 14, 30, 23, 11, 13, 13, 15–16, 23–23	1
277	15, 14, 30, 24, 10, 13, 13, 11–14, 19–23	1
278	15, 14, 30, 24, 11, 14, 14, 11–13, 18–20	1
279	15, 14, 30, 24, 9, 11, 13, 12–12, 11–21	1
280	15, 14, 30, 24, 9, 11, 13, 13–14, 19–21	1
281	15, 14, 31, 23, 10, 12, 17, 11–14, 19–21	1
282	15, 14, 31, 23, 10, 13, 13, 14–16, 23–24	1
283	15, 14, 31, 23, 11, 13, 13, 11–14, 19–23	1
284	15, 14, 31, 25, 11, 11, 13, 11–15, 23–23	1
285	15, 14, 31, 25, 12, 11, 13, 11–13, 23–23	1
286	15, 14, 32, 22, 10, 12, 14, 14–15, 19–21	1
287	15, 14, 32, 23, 10, 12, 14, 15–15, 19–21	1
288	15, 14, 33, 23, 10, 12, 15, 15–15, 19–21	1
289	16, 12, 28, 23, 10, 11, 13, 14–14, 19–21	1
290	16, 12, 28, 24, 10, 11, 12, 14–16, 19–20	1
291	16, 12, 28, 24, 10, 11, 13, 13–16, 19–19	1
292	16, 12, 28, 25, 11, 12, 13, 13–18, 19–19	1
293	16, 12, 29, 21, 11, 11, 15, 13–16, 19–20	1
294	16, 12, 29, 22, 10, 11, 14, 12–14, 20–20	1
295	16, 12, 29, 24, 10, 11, 12, 14–17, 19–20	1
296	16, 12, 29, 24, 10, 11, 13, 13–17, 19–19	1
297	16, 12, 30, 26, 10, 13, 13, 11–14, 19–23	1
298	16, 13, 28, 22, 11, 11, 13, 12–12, 11–21	1
299	16, 13, 28, 23, 10, 11, 13, 12–12, 11–21	1
300	16, 13, 28, 23, 10, 11, 13, 12–13, 11–21	1
301	16, 13, 29, 25, 10, 11, 13, 11–14, 19–23	1
302	16, 13, 29, 26, 10, 11, 13, 11–14, 19–23	1
303	16, 13, 30, 24, 10, 11, 13, 10–14, 19–23	1
304	16, 13, 30, 24, 11, 11, 13, 11–14, 19–22	1
305	16, 13, 30, 24, 11, 11, 13, 11–14, 19–23	2
306	16, 13, 30, 25, 10, 11, 13, 10–13, 19–23	1
307	16, 13, 30, 25, 10, 11, 13, 11–14, 19–23	1
308	16, 13, 30, 25, 10, 11, 14, 11–14, 19–23	1
309	16, 13, 30, 25, 11, 11, 13, 11–13, 19–23	1
310	16, 13, 30, 25, 11, 11, 13, 11–14, 19–23	2
311	16, 13, 30, 25, 11, 11, 13, 11–15, 19–23	1
312	16, 13, 31, 24, 10, 11, 13, 14–15, 21–21	1
313	16, 13, 31, 24, 11, 11, 13, 14–15, 21–21	2
314	16, 13, 31, 24, 11, 11, 13, 15–15, 21–21	1
315	16, 13, 31, 25, 10, 11, 13, 10–14, 19–23	1
316	16, 13, 31, 25, 10, 11, 13, 11–14, 19–23	1
317	16, 13, 31, 25, 11, 11, 13, 10–16, 19–23	1
318	16, 14, 30, 25, 10, 11, 13, 11–15, 19–23	1
319	16, 14, 31, 25, 10, 11, 13, 11–14, 19–23	1
320	16, 14, 33, 24, 10, 11, 13, 15–15, 21–21	1
321	16, 15, 32, 24, 10, 11, 13, 11–14, 19–23	1
322	17, 12, 28, 22, 11, 11, 14, 14–15, 20–20	1
323	17, 12, 28, 25, 10, 11, 12, 13–17, 19–20	1
324	17, 12, 28, 25, 11, 11, 14, 13–17, 19–19	1
325	17, 12, 28, 25, 11, 12, 13, 13–17, 19–19	1
326	17, 12, 29, 25, 10, 11, 13, 14–16, 19–19	1

Table 1 (continued)

HT	Allelic information	n
327	17, 13, 28, 23, 10, 11, 13, 11–12, 11–21	1
328	17, 13, 29, 25, 10, 11, 13, 10–14, 19–23	1
329	17, 13, 29, 25, 10, 11, 13, 11–11, 19–21	1
330	17, 13, 29, 25, 10, 11, 13, 11–14, 19–23	1
331	17, 13, 30, 21, 10, 11, 14, 18–19, 19–21	1
332	17, 13, 30, 25, 10, 11, 12, 14–14, 19–23	1
333	17, 13, 30, 25, 10, 11, 13, 10–14, 19–23	2
334	17, 13, 30, 25, 11, 11, 13, 11–14, 19–23	1
335	17, 13, 30, 26, 11, 11, 13, 11–14, 19–23	1
336	17, 13, 31, 25, 10, 11, 13, 10–14, 19–23	1
337	17, 14, 29, 25, 10, 11, 13, 10–14, 19–23	1
338	17, 14, 31, 24, 9, 13, 13, 13–14, 18–23	1
339	17, 14, 32, 23, 10, 12, 15, 15–16, 17–21	1
340	17, 14, 32, 24, 10, 11, 13, 14–15, 21–21	1

Table 2 The 15 most common Y chromosomal STR haplotypes found among unrelated male individuals from southwest Germany (*N*=433). Allelic information is given in the format DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385, YCAII (*n* number of individuals observed for each given haplotype, *HT* haplotype number)

Nr.	Allelic information	HT	n
1	14, 13, 29, 24, 11, 13, 13, 11–14, 19–23	128	17
2	14, 13, 29, 23, 11, 13, 13, 11–14, 19–23	103	10
3	14, 13, 29, 24, 10, 13, 13, 11–14, 19–23	112	9
4	14, 13, 29, 24, 11, 13, 13, 11–15, 19–23	130	6
5	14, 12, 28, 23, 10, 11, 13, 13–14, 19–21	54	4
6	14, 13, 29, 23, 10, 13, 13, 11–14, 19–23	91	4
7	14, 12, 28, 22, 10, 11, 13, 13–14, 19–21	39	3
8	14, 12, 28, 22, 10, 11, 14, 13–14, 19–21	44	3
9	14, 13, 29, 23, 11, 13, 13, 11–13, 19–23	100	3
10	14, 13, 29, 24, 11, 13, 13, 11–16, 19–23	131	3
11	14, 13, 29, 25, 11, 13, 13, 11–14, 19–23	145	3
12	14, 13, 30, 23, 11, 13, 13, 11–14, 19–23	156	3
13	14, 14, 30, 23, 11, 13, 13, 11–14, 19–23	172	3
14	15, 12, 28, 24, 11, 13, 13, 11–14, 19–23	213	3
15	15, 13, 29, 24, 11, 13, 13, 11–14, 19–23	256	3

the extended haplotype including YCA II, 294 out of 433 Y-chromosomes (67.9%) could be distinguished compared to 274 out of 433 Y-chromosomes (63.3 %) by minimal haplotyping (9 loci without YCAII). Accordingly, typing of YCAII led to the sub-division of 14 minimal haplotypes into 31 different extended haplotypes. The estimate for haplotype diversity was 0.9956 for the extended haplotypes versus 0.9947 for the minimal haplotypes. Allele ranges and gene diversity values for the examined loci are given in Table 3. For DYS385 and YCAII the number of observed allelic classes is given. One duplication (14, 15) was found in DYS19.

Part of the data presented in this study has already been submitted to the YHRD (Y-STR haplotype reference database) and the data were analyzed in comparison to other, mostly European, populations. The different frequencies of the haplotypes given in Table 2 within the population examined in this study and the European YHRD (by 12.2002) are illustrated in Fig. 1. However, statistical analysis failed to prove any significant differences. Due to a very homogeneous European Y-STR pool, interpopulation variability is very low [13] and so far, significant differences have only been shown for Polish paternal lineages [23].

Table 3 Observed allele ranges for the analyzed loci (*N*=433). For the diallelic loci DYS385 and YCAII the number of allelic classes is provided. Gene diversity was estimated according to Nei [22]

Y-STR	Allele range (number of allelic classes)	Gene diversity [22]
DYS19	13–17	0.626
DYS389I	12–15	0.549
DYS389II	27–33	0.710
DYS390	21–27	0.719
DYS391	9–13	0.553
DYS392	7–16	0.596
DYS393	11–17	0.454
DYS385	(43)	0.861
YCAII	(24)	0.705

For the diallelic locus YCAII several sequences have been published [15, 16] or submitted to the GenBank (AY059068 [16], AF140640 [18]). A numeric nomenclature with allele 1 as $(CA)_{19}$ was given by Quintana-Murci et al. [17]. In this nomenclature as in other studies [2, 10] the larger fragment of the electrophoretic pattern is assigned to locus YCAIIa, the smaller fragment to locus YCAIIb. However, the position and structure of the two YCAII loci have not yet been sufficiently clarified.

The numeric nomenclature for YCA II established by Kayser et al. [3] utilized the shortest allele observed in their study as the point of reference, a $(CA)_{17}$ allele with 146 bp (sequenced). However, several research groups have identified shorter alleles [16, 17], which can clearly not be included in this nomenclature in a meaningful manner. This emphasized the need for standardized repeat-based nomenclatures for STR loci used in the forensic context [14].

The repeat-based nomenclature used in this study is based on the revised sequence of YCAII as previously reported [16]. The differences between the published sequences are shown in Fig. 2: the sequence published by Matsumoto et al. [15] lacks a thymine at position 2 (first base of priming site according to Mathias et al. [2]). A second base is lacking within the adenosine-stretch at position 21–26 which might be due to inadequate base calling by the sequencing software. Bergen et al. [18] submitted the sequence of a YCA II clone to the GenBank. There is an additional cytidine at the end of the submitted sequence (G at position 1 of the complementary strand shown in Fig. 2), which is not included in the cited primer or primer-binding site [2]. This surplus base might be part of the vector that was used for cloning.

Based on the revised sequence of YCAII a repeat-based nomenclature for this locus could be proposed and has recently been adopted by the YHRD. In this way, another

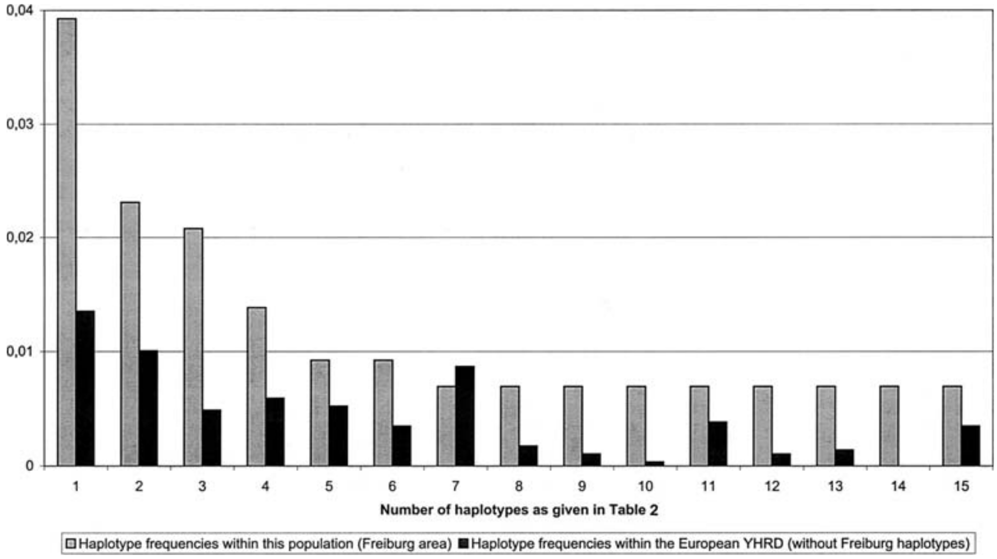

Fig. 1 Haplotype frequencies in the examined population (Freiburg area) as compared to the European YHRD

Fig. 2 Sequences of YCAII as previously published, citations are mentioned in square brackets. If available, GenBank accession numbers are given. Differences between the sequences are indicated by dahses (-). The revised sequence as published by the authors [16] is 150 bp long (allele 19, previously 3)

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                          *         20         *         40         *
[15]           --ATCGATGTAATGTTATATTAAAA-ATGCACACACACACACACACACACA  :  48
AY059068[16]:  -TATCGATGTAATGTTATATTAAAAAATGCACACACACACACACACACACA  :  50
AF140640[18]:  GTATCGATGTAATGTTATATTAAAAAATGCACACACACACACACACACACA  :  51

                        60         *         80         *        100
[15]           CACACACACACACACACAGCTAACCTTATGGTGAAAGACTGACAGATTTTACT  :  99
AY059068[16]:  CACACACACACACACACAGCTAACCTTATGGTGAAAGACTGACAGATTTTACT  : 101
AF140640[18]:  CACACACACACACACACAGCTAACCTTATGGTGAAAGACTGACAGATTTTACT  : 102

                        *        120         *        140         *
[15]           CTAAGAGCAAAAACAAGGATGCCTGTTTTCACTACTTCTATTTAATATA    : 148
AY059068[16]:  CTAAGAGCAAAAACAAGGATGCCTGTTTTCACTACTTCTATTTAATATA    : 150
AF140640[18]:  CTAAGAGCAAAAACAAGGATGCCTGTTTTCACTACTTCTATTTAATATA    : 151
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step towards a standardized and extensive databasing of Y-STR haplotypes for forensic, genealogical, and evolutionary purposes has been accomplished.

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