Int J Legal Med (2005) 119: 241–246
DOI 10.1007/s00414-005-0546-8

SHORT COMMUNICATION

Burkhard Berger · Alexandra Lindinger · Harald Niederstätter · Petra Grubwieser · Walther Parson

Y-STR typing of an Austrian population sample using a 17-loci multiplex PCR assay

Received: 23 December 2004 / Accepted: 23 March 2005 / Published online: 21 April 2005

Abstract Y-chromosomal STR haplotypes were determined from a sample of 135 unrelated men and 70 sons from Tirol (Austria) using the AmpFlSTR Yfiler PCR amplification kit (Applied Biosystems) that coamplifies 17 Y-STRs. The panel of markers includes the 9-loci European minimal haplotype (minHt) and, in addition, the markers DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 (Y GATA C4) and Y GATA H4. A total of 130 different haplotypes (125 were unique) were identified by the 17 Y-STR markers, an increase of 19 compared with the minHt. The gene diversity of DYS635, DYS456 and DYS458 exceeded 0.75 and only that of the duplicated marker DYS385 (0.86) was higher. Consistently high haplotype diversity values were found in all tested Y-SNP haplogroups. Because the simultaneous analysis of 17 Y-STR systems offers a high power of discrimination at minimum sample consumption, the Yfiler kit is a promising tool for forensic applications.

Keywords Y chromosome · Short tandem repeats · Y-haplotypes · Y-SNP · Haplogroups · Population data · Austria

Introduction

The analysis of Y-chromosomal short tandem repeats (Y-STRs) has become a widely accepted tool for human identification, often complementing autosomal STR results. Due to the absence of inter-chromosomal meiotic recombination between Y-chromosomal markers located in the male-specific region of the Y-chromosome [1], the discrimination capacity of Y-STRs is limited. A practical approach is to combine a number of polymorphic markers to a highly discriminating set, which is usually analyzed by a (single) PCR multiplex assay. The most common set of Y-STRs is the European minimal haplotype (minHt) consisting of the loci DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385 [2, 3]. Until 2004 the Y Chromosome Haplotype Reference Database [4] contained more than 26,000 minHt entries on a worldwide basis (YHRD release 14; http://www.yhrd.org). Despite the high utility of the minHt, additional Y-STR loci are required in order to improve the potential to distinguish between different paternal lineages. In 2003, the Scientific Working Group on DNA Analysis Methods (SWGDAM) recommended the minHt plus the loci DYS438 and DYS439 [5]. A number of new Y-specific STRs have been characterized and studied in different populations during the last years [6–14], meaning that today more than 100 Y-STRs are available for forensic investigations [15]. Recently, Y-STR multiplex kits have become commercially available and have been validated for forensic casework [5, 16]. Empirically, the Y-STR markers of commercial multiplex assays have become widely used by the forensic community, and therefore population data of these loci are of high interest for forensic purposes. Here we present a population study that was carried out on west Eurasian individuals using a test version of the recently commercially released AmpFlSTR Yfiler PCR amplification kit (Applied Biosystems) containing 17 Y-STR loci. We determined allele frequencies and haplotype diversity data. In addition, the STR data were interpreted in the context of the Y-SNP haplogroup structure.

Materials and methods

DNA samples of 135 west Eurasian men from Tirol (Austria) and 70 sons (confirmed by autosomal STR analysis; paternity index >1,000) were the same as used in a previous study [17].

Amplification of 3 ng template DNA was carried out with the AmpFlSTR Yfiler PCR amplification kit (Applied

B. Berger · A. Lindinger · H. Niederstätter · P. Grubwieser · W. Parson (✉)
Institute of Legal Medicine, Innsbruck Medical University, Müllerstraße 44,
6020 Innsbruck, Austria
e-mail: walther.parson@uibk.ac.at
Tel.: +43-512-5073303
Fax: +43-512-5072764

Table 1 The 17-loci Y-STR haplotypes in the Austrian population sample (n=135)

Sample	Haplogroup	Minimal haplotype	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y GATA H4
24/25	F*(xK)	14 12 28 22 10 11 14 13–14	16	10	11	20	14	15	22	11
39/41	P*(xR1ab)	14 13 29 23 11 13 10 11–14	15	12	13	19	15	15	24	12
77/78	F*(xK)	15 12 28 22 10 11 14 15–16	16	10	11	21	15	17	21	12
113/114	P*(xR1ab)	15 14 30 24 11 13 13 12–15	15	12	13	19	15	19	23	12
125/127	R1a*	16 13 30 25 10 11 13 11–14	14	11	10	20	17	15	23	12
1	P*(xR1ab)	12 12 28 25 11 13 12 11–14	15	12	13	19	16	18	23	12
2	F*(xK)	13/15 13 29 22 10 11 14 13–13	16	10	13	21	17	18	22	12
3	E3b	13 12 29 23 10 11 13 16–17	14	10	12	20	16	17	21	12
4	F*(xK)	13 13 30 22 10 11 12 13–19	15	9	12	21	16	15	21	13
5	E3b	13 13 30 23 11 11 13 16–17	14	10	12	20	17	14	21	12
6	E3b	13 13 30 24 10 11 13 15–18	14	10	13	19	17	15	22	12
7	E3b	13 13 30 24 10 11 13 16–17	14	10	12	19	17	15	23	11
8	E*(xE3b)	13 13 30 24 10 11 13 16–18	14	10	12	20	17	15	23	11
9	E*(xE3b)	13 13 30 246 10 11 13 16–18	14	10	12	20	15	16	22	11
10	E*(xE3b)	13 13 30 24 10 11 13 16–18	14	10	12	20	16	15	23	11
11	E*(xE3b)	13 13 30 24 10 11 13 16–18	14	10	12	20	16	15	21	13
12	E*(xE3b)	13 13 30 24 10 11 13 16–18	14	10	12	20	16	15	21	11
13	E3b	13 13 30 24 10 11 13 17–17	14	10	12	20	17	16	22	11
14	E3b	13 13 30 24 11 11 13 17–19	14	10	13	20	18	15	22	12
15	E3b	13 13 30 25 10 11 13 15–17	14	10	11	20	17	15	22	12
16	E3b	13 13 31 23 10 11 12 14–19	14	10	12	20	16	16	23	11
17	E3b	13 13 31 24 10 11 13 16–18	14	10	12	20	16	15	21	12
18	F*(xK)	13 13 31 24 10 11 13 17–19	14	10	11	20	17	16	24	11
19	E3b	13 13 31 24 11 11 13 15–16	14	10	12	19	15	18	20	13
20	F*(xK)	14 11 27 23 10 11 13 13–15	16	10	11	20	14	17	22	11
21	F*(xK)	14 12 28 22 10 11 12 12–19	14	10	12	21	15	20.2	22	11
22	F*(xK)	14 12 28 22 10 11 13 13–15	16	10	11	20	15	15	22	11
23	F*(xK)	14 12 28 22 10 11 13 13–15	16	10	12	20	14	16	22	11
26	F*(xK)	14 12 28 23 10 11 13 13–13	16	10	10	20	14	15	23	10
27	F*(xK)	14 12 28 23 10 11 13 13–14	16	10	11	21	14	15	21	11
28	P*(xR1ab)	14 12 28 23 11 13 13 11–14	15	12	12	19	16	18	23	12
29	F*(xK)	14 12 28 23 9 11 12 13–14	16	10	11	21	14	15	21	11
30	P*(xR1ab)	14 12 28 24 10 13 13 11–14	15	12	12	20	16	17	23	12
31	F*(xK)	14 12 29 22 10 11 13 13–14	16	10	11	20	14	16	21	11
32	F*(xK)	14 12 30 22 10 11 13 13–14	16	10	11	20	14	16	21	11
33	K*(xN3.P)	14 12 30 26 11 13 13 11–14	15	12	13	19	15	18	23	14
34	P*(xR1ab)	14 13 28 23 11 11 13 11–13	15	12	11	19	16	17	23	11
35	P*(xR1ab)	14 13 28 24 11 13 13 12–14	15	12	12	20	15	17	23	11
36	F*(xK)	14.1 13 29 22 10 11 14 14–15	16	10	11	21	14	16	19	13
37	F*(xK)	14 13 29 23 10 11 12 13–17	15	9	11	19	16	15	24	11
38	P*(xR1ab)	14 13 29 23 11 13 10 11–14	15	12	12	19	15	15	24	13
40	P*(xR1ab)	14 13 29 23 11 13 10 11–14	15	12	13	19	16	15	24	12
42	P*(xR1ab)	14 13 29 23 11 13 13 11–13	15	12	11	19	16	17	23	12
43	P*(xR1ab)	14 13 29 23 11 13 13 11–13	15	12	12	19	18	19	23	11
44	P*(xR1ab)	14 13 29 23 11 13 13 11–14	15	12	12	19	16	16	23	12
45	P*(xR1ab)	14 13 29 23 11 13 13 11–14	15	12	12	19	16	17	23	12
46	P*(xR1ab)	14 13 29 23 11 13 13 11–14	15	12	11	19	15	18	23	12
47	P*(xR1ab)	14 13 29 23 11 13 13 11–14	15	12	12	19	17	16	23	12
48	P*(xR1ab)	14 13 29 24 10 13 13 11–14	15	12	12	19	16	16	23	12
49	P*(xR1ab)	14 13 29 24 10 14 13 11–14	15	12	13	19	16	17	23	12
50	R1b	14 13 29 24 11 13 12 11–14	15	12	12	19	15	17	23	12
51	P*(xR1ab)	14 13 29 24 11 13 13 11–14	15	12	12	19	16	20	23	11
52	P*(xR1ab)	14 13 29 24 11 13 13 11–14	15	12	12	19	13	18	23	12
53	P*(xR1ab)	14 13 29 24 11 13 13 11–14	15	13	12	19	16	17	23	11

Table 1 (continued)

Sample	Haplogroup	Minimal haplotype	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y GATA H4
54	P*(xR1ab)	14 13 29 24 11 13 13 11–14	15	13	12	19	15	18	23	12
55	P*(xR1ab)	14 13 29 24 11 13 13 11–15	15	12	11	19	16	18	23	12
56	P*(xR1ab)	14 13 29 24 11 13 14 11–13	15	12	12	19	16	15	25	12
57	P*(xR1ab)	14 13 29 24 11 14 13 11–13	15	12	11	19	17	18	23	12
58	P*(xR1ab)	14 13 29 25 10 13 13 11–14	15	12	11	19	17	18	23	12
59	P*(xR1ab)	14 13 29 25 11 13 13 11–14	15	12	11	19	15	17	23	10
60	P*(xR1ab)	14 13 30 22 11 13 13 11–13	15	12	12	19	19	19	23	12
61	F*(xK)	14 13 30 23 11 11 12 17–23	14	10	12	21	15	19.2	20	12
62	E3b	14 13 30 24 10 10 13 17–19	14	10	12	20	14	17	20	12
63	E3b	14 13 30 24 10 11 13 16–18	14	10	12	20	16	16	22	11
64	P*(xR1ab)	14 13 30 24 10 13 13 11–14	14	12	11	19	15	16	25	12
65	P*(xR1ab)	14 13 30 24 10 13 13 11–14	15	12	12	19	15	18	23	12
66	P*(xR1ab)	14 13 30 25 10 14 13 11–14	15	12	12	19	16	18	23	11
67	F*(xK)	14 13 31 23 10 11 12 13–16	15	9	12	20	17	14	21	10
68	E3b	14 13 31 24 11 11 13 15–15	14	10	13	20	16	17	21	12
69	P*(xR1ab)	14 13 31 24 11 13 13 11–14	14	12	11	19	16	18	23	12
70	P*(xR1ab)	14 14 30 23 11 13 13 11–13	15	12	11	20	17	17	22	12
71	P*(xR1ab)	14 14 30 23 12 13 13 11–14	16	12	13	19	17	17	23	12
72	P*(xR1ab)	14 14 30 25 11 13 14 11–14	15	12	12	19	14	18	23	13
73	P*(xR1ab)	14 14 31 23 11 13 13 11–14	15	12	12	19	16	17	25	12
74	F*(xK)	15 12 28 21 10 11 13 14–14	16	10	11	21	16	15.2	20	12
75	F*(xK)	15 12 28 22 10 11 14 12–16	16	10	12	19	15	18	21	12
76	F*(xK)	15 12 28 22 10 11 14 13–14	14	10	11	21	15	15	20	12
79	F*(xK)	15 12 28 24 10 11 12 14–17	16	9	12	19	14	16	22	11
80	F*(xK)	15 12 28 24 11 11 12 13–17	16	9	12	19	13	16	22	11
81	F*(xK)	15 12 28 24 11 11 12 14–17	16	9	12	19	14	16	21	11
82	P*(xR1ab)	15 12 28 24 11 13 13 12–14	15	12	11	18	16	18	23	12
83	F*(xK)	15 12 28 25 10 11 12 13–17	16	9	11	19	13	16	21	11
84	F*(xK)	15 12 29 22 10 11 13 14–14	16	10	11	20	15	16	20	11
85	F*(xK)	15 12 29 22 10 11 13 14–14	16	10	11	21	16	18	20	12
86	F*(xK)	15 12 29 22 10 11 13 14–14	16	10	11	21	15	16	21	12
87	F*(xK)	15 12 29 22 10 11 14 13–14	16	10	11	21	15	16	20	12
88	F*(xK)	15 12 29 23 10 11 13 14–14	16	10	11	20	14	18	21	11
89	F*(xK)	15 12 29 23 10 11 13 15–15	16	10	11	19	14	17	21	11
90	F*(xK)	15 12 29 23 10 11 14 14–14	16	10	11	21	15	16	22	13
91	F*(xK)	15 12 29 23 10 12 15 14–15	16	10	12	21	15	17	20	12
92	F*(xK)	15 12 29 24 10 11 14 14–14	16	10	11	21	15	16	23	13
93	F*(xK)	15 12 30 21 10 11 14 14–15	16	11	12	21	16	16	20	13
94	F*(xK)	15 12 30 22 10 11 14 14–14	16	10	12	21	15	16	20	11
95	R1a1	15 13 27 25 10 11 13 11–14	14	11	11	20	16	16	25	13
96	F*(xK)	15 13 29 22 10 11 12 13–15	15	9	12	21	14	17	21	11
97	F*(xK)	15 13 29 22 10 11 13 13–14	16	10	11	20	15	16	22	11
98	F*(xK)	15 13 29 23 11 11 12 13–16	14	8	12	20	16	17	22	12
99	P*(xR1ab)	15 13 29 23 11 13 13 11–14	15	12	13	18	18	17	23	12
100	N3	15 13 29 23 11 13 14 11–14	14	10	10	19	14	17	22	11
101	P*(xR1ab)	15 13 29 24 11 13 12 11–14	15	12	12	19	15	18	25	12
102	P*(xR1ab)	15 13 29 24 11 13 14 11–14	15	12	12	17	16	17	24	11
103	R1a1	15 13 29 25 10 11 13 11–15	14	11	12	20	17	15	23	12
104	R1a1	15 13 30 24 10 11 13 12–14	14	11	10	20	17	15	23	11
105	R1a1	15 13 30 26 10 10 13 11–14	14	11	11	20	17	15	23	12
106	R1a1	15 13 30 26 11 11 13 11–14	14	11	10	20	16	15	23	12
107	F*(xK)	15 13 31 23 10 11 12 13–15.2	15	9	11	20	15	16	21	11
108	F*(xK)	15 13 31 23 10 12 14 15–15	14	10	13	20	15	15	21	11
109	P*(xR1ab)	15 13 31 24 10 13 12 12–14	15	12	14	19	15	16	23	12

Table 1 (continued)

Sample	Haplogroup	Minimal haplotype	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y GATA H4
110	F*(xK)	15 13 31 24 11 11 13 15–15	15	10	12	20	15	17	22	11
111	R1a1	15 13 31 25 10 11 13 11–14	14	11	10	20	15	15	24	12
112	F*(xK)	15 14 30 24 10 12 15 15–16	14	10	11	19	14	14	21	12
115	F*(xK)	15 14 32 23 10 11 13 13–16	16	10	11	21	17	19	24	12
116	F*(xK)	16 12 28 24 10 11 12 13–17	16	9	12	19	13	16	22	11
117	F*(xK)	16 12 28 24 11 11 12 14–17	16	9	12	19	13	18	23	11
118	F*(xK)	16 12 29 22 10 10 14 15–15	16	10	13	21	16	16	24	12
119	F*(xK)	16 12 29 23 10 12 14 13–14	15	10	11	21	15	15	21	13
120	R1a1	16 13 29 25 10 11 13 11–14	14	11	11	20	16	16	23	12
121	R1a*	16 13 30 24 10 11 13 11–14	14	11	11	20	17	17	23	12
122	R1a1	16 13 30 24 11 11 13 10–14	14	11	11	19	15	16	24	12
123	R1a1	16 13 30 24 11 11 13 11–15	14	11	10	20	15	15	23	11
124	F*(xK)	16 13 30 24 11 11 13 14–15	15	10	12	20	15	17	22	11
126	R1a*	16 13 30 25 10 11 13 11–14	14	11	13	20	17	15	23	12
128	R1a*	16 13 30 25 10 11 13 11–14	14	11	10	20	18	15	23	12
129	R1a1	16 13 32 25 11 11 13 11–14	14	11	10	20	17	15	23	12
130	R1a1	16 14 30 24 11 11 13 11–14	14	12	10	20	16	16	23	13
131	R1a1	16 14 30 25 11 11 13 11–14	14	11	10	20	16	15	24	12
132	R1a1	16 14 30 26 11 11 13 10–14	14	11	10	20	16	15	23	12
133	R1a*	16 14 31 25 11 11 13 11–14	14	11	10	20	17	15	23	12
134	F*(xK)	17 12 29 24 10 11 14 12–14	15	10	13	20	17	18	24	11
135	F*(xK)	17 13 31 24 11 11 13 13–15	15	10	13	20	15	17	20	11

The loci of the minHt are (from left to right) DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393 and DYS385

Biosystems) in a TETRAD2 thermal cycler (MJ Research) using the thermal cycling conditions recommended by the manufacturer. The amplification products were analyzed on an ABI PRISM 3100 Genetic analyzer using 36-cm capillary arrays, POP-6 and the dye set G5vb module. Analysis of the data was performed using GeneScan software v. 3.7 and Genotyper v. 3.7NT. Fragment sizing was performed by means of an internal size standard (GeneScan-500 LIZ size standard) and the amplified samples were compared with the provided allelic ladder (AmpFlSTR Yfiler allelic ladder) to designate the specific alleles present at each locus.

Gene and haplotype diversity (*H*) values and estimates of the sampling variance were computed with the Arlequin software [18].

Y-SNP data were taken from [19]. The nomenclature of the Y-chromosomal haplogroups found followed the YCC 2003 Tree [20, 21]. Due to limited space, the Y-SNP haplogroup structure of the Austrian population sample is presented on a reduced level of resolution in this article (detailed haplogroup-related information is available from the authors upon request).

Results and discussion

Y-STR haplotypes were determined from a sample of 205 men (135 unrelated men and 70 sons) from Tirol (Austria) using the AmpFlSTR Yfiler PCR amplification kit (Applied Biosystems) that includes the 9-loci European minHt, the 11-loci SWGDAM core set (minHt plus DYS438 and DYS439) and, in addition, the markers DYS437, DYS448, DYS456, DYS458, DYS635 (Y GATA C4) and Y GATA H4. All minHts determined with the Yfiler kit were concordant with previously published results [17, 22].

We analyzed the 17 markers of the Yfiler kit in 70 meioses from confirmed father–son pairs. Only a single mutation was found for the marker DYS390 (allele 24 to allele 25). No mutation was detected in the eight Y-STR loci that are not included in the minHt.

The 17-loci Y-STR haplotypes of the 135 samples are shown in Table 1 and the relative frequencies of the STR loci are listed in Table 2. A total of 130 different haplotypes (125 of them were unique) were identified, corresponding to a haplotype diversity (*H*) of 0.9994 (see Table 3). This means an increase of 19 Y lineages compared with the 111 different minHts (*H*=0.9952). By adding the data for the highly polymorphic locus DYS464 [17], it was only possible to differentiate the lineages of samples 24 and 25, but none of the other men showed identical Y-STR haplotypes with the AmpFlSTR Yfiler kit (see Table 1). The haplotype diversity of the loci ranged from 0.506 (DYS391) to 0.866 (DYS385) for the entire sample set. Apart from the multicopy STR DYS385 the loci DYS458 (*H*=0.788), DYS456 (*H*=0.769) and DYS635 (*H*=0.752) showed the highest *H* values of the Yfiler marker set (Table 3). Some of the Y-STR loci showed variable *H* values in different Y-SNP haplogroups, e.g. DYS437, DYS438, DYS448, whereas other markers revealed a pattern with minor differences, e.g. DYS456 and DYS458. A Y-SNP-haplogroup dependency of *H* values can also be seen for the minHt and the

Table 2 Relative allele frequencies of the Yfiler markers (except for DYS385) in the population sample (n=135) as determined by simple counting

Allele	DYS19	DYS389I	DYS389II	DYS390	DYS391	DYS392	DYS393	DYS437	DYS438	DYS439	DYS448	DYS456	DYS458	DYS635	Y GATA H4
8									0.0074						
9					0.0074				0.0815						
10					0.5704	0.0222	0.0296		0.4370	0.1037					0.0222
11		0.0074			0.4148	0.6222			0.1407	0.3407					0.3556
12	0.0074	0.3037			0.0074	0.0296	0.1481		0.3185	0.4074					0.5259
13	0.1259	0.6000				0.3037	0.6519		0.0148	0.1407		0.0370			0.0889
14	0.3926	0.0889				0.0222	0.1556	0.3481		0.0074		0.1407	0.0222		0.0074
14.1	0.0074														
15	0.3111						0.0148	0.3778				0.3037	0.2889		
15.2													0.0074		
16	0.1333							0.2741				0.2963	0.2444		
17	0.0148										0.0074	0.1852	0.2148		
18											0.0148	0.0296	0.1630		
19											0.3926	0.0074	0.0370	0.0074	
19.2													0.0074		
20											0.4148		0.0074	0.0889	
20.2													0.0074		
21				0.0148							0.1704			0.1852	
22				0.1704										0.1704	
23				0.2667										0.4148	
24				0.3852										0.0963	
25				0.1333										0.0370	
26				0.0296											
27			0.0148												
28			0.1852												
29			0.3556												
30			0.3185												
31			0.1111												
32			0.0148												
13/15	0.0074														

For DYS385 data, see [19]

Table 3 Estimated haplotype/gene diversities (H), sampling variances (SV) and numbers of different lineages of the Yfiler set, the SWGDAM core set, the minHt and of the individual markers in the entire population sample and in four haplogroups representing the population structure at a low level of resolution

Marker	All samples, n=135			E, n=18			F*(xK), n=53			K*(xR1a), n=45			R1a, n=19		
	H	SV	Lineages	H	SV	Lineages	H	SV	Lineages	H	SV	Lineages	H	SV	Lineages
Yfiler	0.9994	±0.0010	130	1.0000	±0.0185	18	0.9985	±0.0040	51	0.9980	±0.0052	43	0.9942	±0.0193	18
SWGDAM	0.9970	±0.0016	118	0.9346	±0.0524	14	0.9964	±0.0048	49	0.9909	±0.0072	38	0.9825	±0.0259	17
minHt	0.9952	±0.0019	111	0.9346	±0.0524	14	0.9956	±0.0049	48	0.9788	±0.0106	33	0.9649	±0.0358	16
DYS385	0.8656	±0.0251	28	0.8366	±0.0751	9	0.9194	±0.0177	17	0.4737	±0.0826	5	0.4561	±0.1317	4
DYS458	0.7878	±0.0134	10	0.7059	±0.0877	5	0.7779	±0.0351	9	0.7616	±0.0356	6	0.4327	±0.1167	3
DYS456	0.7693	±0.0154	7	0.7124	±0.0743	5	0.7337	±0.0352	5	0.7091	±0.0440	7	0.6842	±0.0702	4
DYS635	0.7516	±0.0246	7	0.7582	±0.0504	4	0.7576	±0.0304	6	0.4162	±0.0857	4	0.3684	±0.1254	3
DYS390	0.7381	±0.0193	6	0.3856	±0.1280	3	0.6967	±0.0306	5	0.6404	±0.0416	5	0.6023	±0.0881	3
DYS389II	0.7305	±0.0173	6	0.4510	±0.1174	3	0.7257	±0.0333	6	0.6081	±0.0625	4	0.5322	±0.1296	5
DYS19	0.7204	±0.0207	8	0.2941	±0.1193	2	0.6509	±0.0534	7	0.3354	±0.0771	3	0.4561	±0.0852	2
DYS439	0.6924	±0.0204	5	0.3856	±0.1280	3	0.5871	±0.0443	4	0.6657	±0.0449	5	0.5556	±0.1030	4
DYS438	0.6859	±0.0230	6	0.0000	±0.0000	1	0.3940	±0.0693	4	0.1293	±0.0667	3	0.1053	±0.0920	2
DYS437	0.6659	±0.0087	3	0.0000	±0.0000	1	0.4949	±0.0668	3	0.1687	±0.0727	3	0.0000	±0.0000	1
DYS448	0.6493	±0.0182	5	0.2941	±0.1193	2	0.6459	±0.0276	3	0.2475	±0.0828	4	0.1053	±0.0920	2
Y GATA H4	0.5929	±0.0259	5	0.6601	±0.0593	3	0.5965	±0.0516	4	0.4374	±0.0809	5	0.3743	±0.1296	3
DYS389I	0.5438	±0.0316	4	0.1111	±0.0964	2	0.4913	±0.0576	4	0.4081	±0.0816	3	0.3509	±0.1112	2
DYS393	0.5318	±0.0427	5	0.1111	±0.0964	2	0.7003	±0.0210	4	0.4485	±0.0855	4	0.0000	±0.0000	1
DYS392	0.5226	±0.0336	5	0.1111	±0.0964	2	0.1771	±0.0677	3	0.1687	±0.0727	3	0.1053	±0.0920	2
DYS391	0.5062	±0.0175	4	0.3660	±0.1124	2	0.2932	±0.0721	3	0.3354	±0.0771	3	0.5146	±0.0517	2

SWGDAM core set with lower values for haplogroup E (see Table 3). With the 17-loci Yfiler set these small differences are compensated.

The simultaneous analysis of 17 Y-STR systems offers a high power of discrimination for minimum sample consumption. Therefore, the Yfiler kit is a promising tool for forensic applications. Evaluation work describing forensically critical parameters such as sensitivity, robustness and reproducibility and mixture analysis as well as investigation of different types of forensic casework samples and degraded DNA is ongoing.

References

1. Skaletsky H, Kuroda-Kawaguchi T, Minx PJ et al (2003) The male-specific region of the human Y chromosome is a mosaic of discrete sequence classes. Nature 423:825–837
2. Kayser M, Caglia A, Corach D et al (1997) Evaluation of Y-chromosomal STRs: a multicenter study. Int J Leg Med 110:125–129
3. Pascali VL, Dobosz M, Brinkmann B (1999) Coordinating Y-chromosomal STR research for the courts. Int J Leg Med 112:1
4. Roewer L, Krawczak M, Willuweit S et al (2001) Online reference database of European Y-chromosomal short tandem repeat (STR) haplotypes. Forensic Sci Int 118:106–113
5. Krenke BE, Viculis L, Richard ML et al (2005) Validation of a male-specific, 12-locus fluorescent short tandem repeat (STR) multiplex. Forensic Sci Int 148:1–14
6. Beleza S, Alves C, Gonzalez-Neira A, Lareu M, Amorim A, Carracedo A, Gusmao L (2003) Extending STR markers in Y chromosome haplotypes. Int J Leg Med 117:27–33
7. Bosch E, Lee AC, Calafell F, Arroyo E, Henneman P, de Knijff P, Jobling MA (2002) High resolution Y chromosome typing: 19 STRs amplified in three multiplex reactions. Forensic Sci Int 125:42–51
8. Butler JM, Schoske R, Vallone PM, Kline MC, Redd AJ, Hammer MF (2002) A novel multiplex for simultaneous amplification of 20 Y chromosome STR markers. Forensic Sci Int 129:10–24
9. Gusmao L, Alves C, Beleza S, Amorim A (2002) Forensic evaluation and population data on the new Y-STRs DYS434, DYS437, DYS438, DYS439 and GATA A10. Int J Leg Med 116:139–147
10. Quintans B, Beleza S, Brion M, Sanchez-Diz P, Lareu M, Carracedo A (2003) Population data of Galicia (NW Spain) on the new Y-STRs DYS437, DYS438, DYS439, GATA A10, GATA A7.1, GATA A7.2, GATA C4 and GATA H4. Forensic Sci Int 131:220–224
11. Redd AJ, Agellon AB, Kearney VA et al (2002) Forensic value of 14 novel STRs on the human Y chromosome. Forensic Sci Int 130:97–111
12. Schoske R, Vallone PM, Kline MC, Redman JW, Butler JM (2004) High-throughput Y-STR typing of U.S. populations with 27 regions of the Y chromosome using two multiplex PCR assays. Forensic Sci Int 139:107–121
13. Uchihi R, Yamamoto T, Usuda K et al (2003) Haplotype analysis with 14 Y-STR loci using 2 multiplex amplification and typing systems in 2 regional populations in Japan. Int J Leg Med 117:34–38
14. Zarrabeitia MT, Riancho JA, Gusmao L, Lareu MV, Sanudo C, Amorim A, Carracedo A (2003) Spanish population data and forensic usefulness of a novel Y-STR set (DYS437, DYS438, DYS439, DYS460, DYS461, GATA A10, GATA C4, GATA H4). Int J Leg Med 117:306–311
15. Kayser M, Kittler R, Erler A et al (2004) A comprehensive survey of human Y-chromosomal microsatellites. Am J Hum Genet 74:1183–1197
16. Sinha SK, Budowle B, Arcot SS et al (2003) Development and validation of a multiplexed Y-chromosome STR genotyping system, Y-PLEX 6, for forensic casework. J Forensic Sci 48:93–103
17. Berger B, Niederstätter H, Brandstätter A, Parson W (2003) Molecular characterization and Austrian Caucasian population data of the multi-copy Y-chromosomal STR DYS464. Forensic Sci Int 137:221–230
18. Schneider S, Roessli D, Excoffier L (2000) Arlequin ver.2.000: a software for population genetics data analysis. Genetics and Biometry Laboratory, University of Geneva, Switzerland
19. Niederstätter H, Berger B, Oberacher H, Brandstätter A, Huber CG, Parson W (2004) Separate analysis of DYS385a and b versus conventional DYS385 typing: is there forensic relevance? Int J Leg Med 119:1–9
20. Jobling MA, Tyler-Smith C (2003) The human Y chromosome: an evolutionary marker comes of age. Nat Rev Genet 4:598–612
21. Y Chromosome Consortium (2002) A nomenclature system for the tree of human Y-chromosomal binary haplogroups. Genome Res 12:339–348
22. Parson W, Niederstätter H, Köchl S, Steinlechner M, Berger B (2001) When autosomal short tandem repeats fail: optimized primer and reaction design for Y-chromosome short tandem repeat analysis in forensic casework. Croat Med J 42:285–287