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Short communication

The impact of additional Y-STR loci on resolving common haplotypes and closely related individuals☆

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Abstract

Commercial Y-STR kits have permitted laboratories to go beyond the original nine minimal haplotype loci (MHL) and to discover the advantage of additional Y-STR loci in resolving common haplotypes. In an effort to examine the impact of Y-STR markers beyond the 17 loci now available in commercial kit form, new Y-STR loci are being investigated on a common set of samples representative of the major U.S. population groups. Additional Y-STRs can also increase the power of discrimination between closely related male individuals, which is important not only in forensics but also in the paternity and genetic genealogy communities.

Keywords: Short tandem repeat; STR; Y-chromosome; Y-STR; DYS449; DYS570; DYS576; DYS534; NIST

1. Introduction

Y-chromosome STR analysis is a valuable tool to add to the arsenal of techniques available for human identification. In forensics, Y-STRs prove useful in sexual assault situations where there is little male DNA in the presence of high amounts of female DNA. Paternity and genetic genealogy laboratories are discovering the value of adding new Y-STR loci beyond what is commercially available in an attempt to resolve closely related individuals such as fathers and sons and for tracing paternal lineages. Therefore it is necessary to evaluate these loci to determine their value and possible future adoption by commercial kit providers and the genetic genealogy community.

Two commercial Y-chromosome STR kits are widely available for amplifying 12 [1] or 17 [2] Y-STR loci. Using these commercial kits as a starting point, additional loci were screened to determine their ability to resolve U.S. population samples. A primary evaluation of 27 Y-STR loci (including DYS635 in the Yfiler kit) and their allele frequencies was reported previously [3]. After testing, 20 of these loci were evaluated in conjunction with the Yfiler loci on their ability to resolve the most common haplotype observed in 656 U.S. population samples. With the addition of these loci, all samples with the most common type were able to be completely resolved.

2. Materials and methods

A total of 656 male U.S. population samples were typed with the 37 loci listed in Table 1. The Yfiler kit contains the loci in bold. The GenBank accession for each locus is listed along with the PCR product size and the location of the locus on the Y-chromosome.

The loci evaluated were either from the Yfiler kit [2] or were selected based on best candidates from previous studies [4,5]. Using the available Genome Database (GDB) primers (http://www.gdb.org), a BLAST search was conducted to locate the sequence region containing the loci. The Y-chromosome locations were determined using BLAT (http://genome.ucsc.edu/cgi-bin/hgBlat) and the updated March 2006 human

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Table 1
Characteristics of 37 Y-STR loci examined in this study using GDB primers

Locus	GenBank accession	Reference allele	Amplicon size (bp)	PCR product size range (bp)	Allele range	Chromosomal position (MB)
DYS19	AC017019	$[TAGA]_3$tagg$[TAGA]_{12}$ = 15	252	232–268	10–19	10.132
DYS385a/b	AC022486	$(GAAA)_{11}$	369	353–425	7–25	19.261
DYS389I/II	AC004617	$[TCTG]_5[TCTA]_{12}$ . . . $[TCTG]_3[TCTA]_9$	247, 367	239–259, 347–387	10–15, 24–34	13.122
DYS390	AC011289	$[TCTG]_8[TCTA]_{11}[TCTG]_1$ $[TCTA]_4$ = 24	215	191–227	18–27	15.784
DYS391	AC011302	$(TCTA)_{11}$	287	271–295	7–13	12.613
DYS392	AC011745	$(TAT)_{13}$	254	236–269	7–18	21.043
DYS393	AC006152	$(AGAT)_{12}$	119	103–135	8–16	3.191
DYS437	AC002992	$(TCTA)_{10}[TCTG]_2(TCTA)_4$ = 16	192	180–196	13–17	12.977
DYS438	AC002531	$(TTTTC)_{10}$	221	211–236	8–13	13.376
DYS439	AC002992	$(GATA)_{13}$	252	232–260	10–15	13.025
DYS444[a]	AC007043	$(ATAG)_{14}$	308	292–316	10–16	17.736
DYS446	AC006152	$(TCTCT)_{14}$	308	278–343	8–21	3.192
DYS448	AC025227	$[AGAGAT]_{11}$ . . . $[AGAGAT]_8$ = 19	294	282–324	17–24	22.775
DYS449	AC051663	$(TTTC)_{15}$ . . . $(TTTC)_{14}$ = 29	355	335–387	24–37	8.278
DYS456	AC010106	$(AGAT)_{15}$	149	141–161	13–18	4.331
DYS458	AC010902	$(GAAA)_{16}$	123	115–139	14–20	7.928
DYS463	AC007275	$(AAAGG)_7(AAGGG)_{15}(AAGGA)_2$ = 24	254	219–269	17–28	7.704
DYS485	AC009233	$(TTA)_{16}$	278	263–284	10–18	20.559
DYS495	AC004474	$(AAT)_{15}$	212	203–221	12–18	13.521
DYS505	AC012078	$(TCCT)_{12}$	174	162–186	9–15	3.701
DYS508	AC006462	$(TATC)_{11}$	177	165–193	8–15	16.303
DYS520	AC007275	$(ATAG)_{10}(ATAC)_{10}$ = 20	179	171–203	18–26	7.790
DYS522	AC007247	$(GATA)_{10}$	352	344–372	8–17	7.476
DYS532	AC016991	$(CTTT)_{14}$	479	459–491	9–17	8.439
DYS533	AC053516	$(ATCT)_{12}$	214	202–222	9–14	16.903
DYS534	AC053516	$(CTTT)_{15}$	208	188–228	10–20	16.903
DYS540	AC010135	$(TTAT)_{12}$	266	258–274	10–14	17.075
DYS556	AC011745	$(AATA)_{11}$	211	203–219	9–13	21.011
DYS557	AC007876	$(TTTC)_{16}$	196	176–220	11–22	21.644
DYS570	AC012068	$(TTTC)_{17}$	256	236–280	12–23	6.921
DYS576	AC010104	$(AAAG)_{17}$	191	175–207	13–21	7.113
DYS594[a]	AC010137	$(AAATA)_{10}$	264	259–284	9–14	20.116
DYS635	AC004772	$[TCTA]_4(TGTA)_2[TCTA]_2(TGTA)_2[TCTA]_2$ $(TGTA)_2[TCTA]_9$ = 23	176	152–192	17–27	12.890
DYS643	AC007007	$(CTTTT)_{11}$	145	125–165	7–15	15.936
Y-GATA-H4	AC011751	$(TAGA)_{12}$	368	352–372	8–13	17.253

Commercial kit loci in bold.
[a] Updated nomenclature following International Society of Forensic Genetics (ISFG) guidelines [7].

genome reference sequence. The loci were then divided into small multiplexes of four or five loci based on size and tested with population samples to determine allele ranges and gene diversity values. Conditions for PCR amplification, electrophoresis, typing and sequencing were as previously described [3].

3. Summary

We have typed over 656 U.S. population samples with 37 Y-STR loci, 17 of which are commercially available in the Yfiler kit along with 20 additional Y-STRs. Typing of these samples revealed a common haplotype that was observed 26 times with only the minimal haplotype loci [6]. Therefore about 4% of the samples were unresolved using these nine loci. Adding DYS438 and DYS439 loci [6] breaks these samples into three groups. The additional locus DYS437 in the Powerplex Y kit further resolves the samples into seven groups. The Yfiler kit adds five more loci, separating all but three samples with the most common type. Adding either DYS522 or DYS576 to the Yfiler loci resolves all 26 samples with the most common type.

In this population set, 7 of the 20 additional Y-STRs have the same ability to resolve sample haplotypes as all 20 loci and therefore demonstrate promise for future multiplexing. These loci include DYS449, DYS505, DYS522, DYS532, DYS534, DYS570, and DYS576. Future studies will evaluate other Y-STRs beyond these 37 loci that may also be useful to Y-STR testing communities.

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