

22 Y - STR

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Genetic characteristics of 22 Y-STR loci in Koreans

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To construct a Korean Y-chromosomal STR database for 22 Y-STRs (DYS19, DYS385, DYS388, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS446, DYS447, DYS448, DYS449, DYS456, DYS458, DYS464, DYS635, and GATA H4.1), 708 DNA samples from unrelated Korean males were analyzed using three multiplex PCR systems. During analysis, thirty two microvariant alleles were observed at DYS449, DYS458, and DYS464, and duplicated alleles were occurred once each for DYS19, DYS390, and DYS447. In haplotype analysis for the 22 Y-STRs, a total of 693 different haplotypes were observed with overall haplotype diversity of 0.9999, and of these, 680 haplotypes were unique. By combining each marker for the extended SWGDAM haplotype, DYS458 was the most informative marker. In addition, the diversity of combined haplotypes of DYS447, DYS458, DYS635, GATA H4.1, and the SWGDAM Y-STR loci was comparable to haplotypes of 17 loci in the AmpFISTR Yfiler™ kit.

Key words : Y chromosome, STR, extended SWGDAM haplotype, AmpFISTR Yfiler™, microvariant allele, duplicated allele, Koreans

Y - STR

가

Y

(Short Tandem Repeat; STR)

DNA

가

on - line 가 Y - STR Haplotype
Reference Database (YHRD)⁶⁾ 464
9 Y - STR minimal
haplotype³⁾ 50,867 , 11 Y - STR extended
SWGDAM haplotype 23,981

가

4). Y

minimal haplotype

extended SWGDAM

5)

Y - STR

haplotype

7 - 14)

(haplotype)

Y - STR

(76.41% - 89.96%)

3.3%가 15 Y - STR

가

15)

Y - STR

Y - STR

discriminatory capacity 가 Y-STR , YHRD 22
 minimal haplotype diversity가 Y-STR 가 .
 Vienna, Alcamo, Ankara
 1.0000 Zriba 0.5699,
 Andon Poci 0.7076
 가 , minimal haplotype 1.
 extended SWGDAM haplotype Y-STR mutation ¹⁴⁾ 355
 discriminatory capacity 가 가 Y- 19 Y-STR ¹³⁾ 301
 STR 708 가
 , Y-STR autosomal STR 3 DNA 2-
 가 microvariant, duplication null - 20

2.
 가. DNA

kit AmpFISTR Yfiler™ QIAamp DNA Mini Kit (QIAGEN,
 17 Y-STR (DYS19, DYS385, Hilden, Germany) DNA
 DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DNA Picogreen dsDNA Quantitation Reagent (Molecular
 DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 Probes, Eugene, OR) TBS - 380 Mini -
 GATA H4.1) ^{13,14)} gene Fluorometer (Turner Biosystems, Sunnyvale, CA)
 diversity가 5 Y-STR (DYS388, DYS446, DNA
 DYS447, DYS449 DYS464) 22 Y- - 20
 STR 708 가

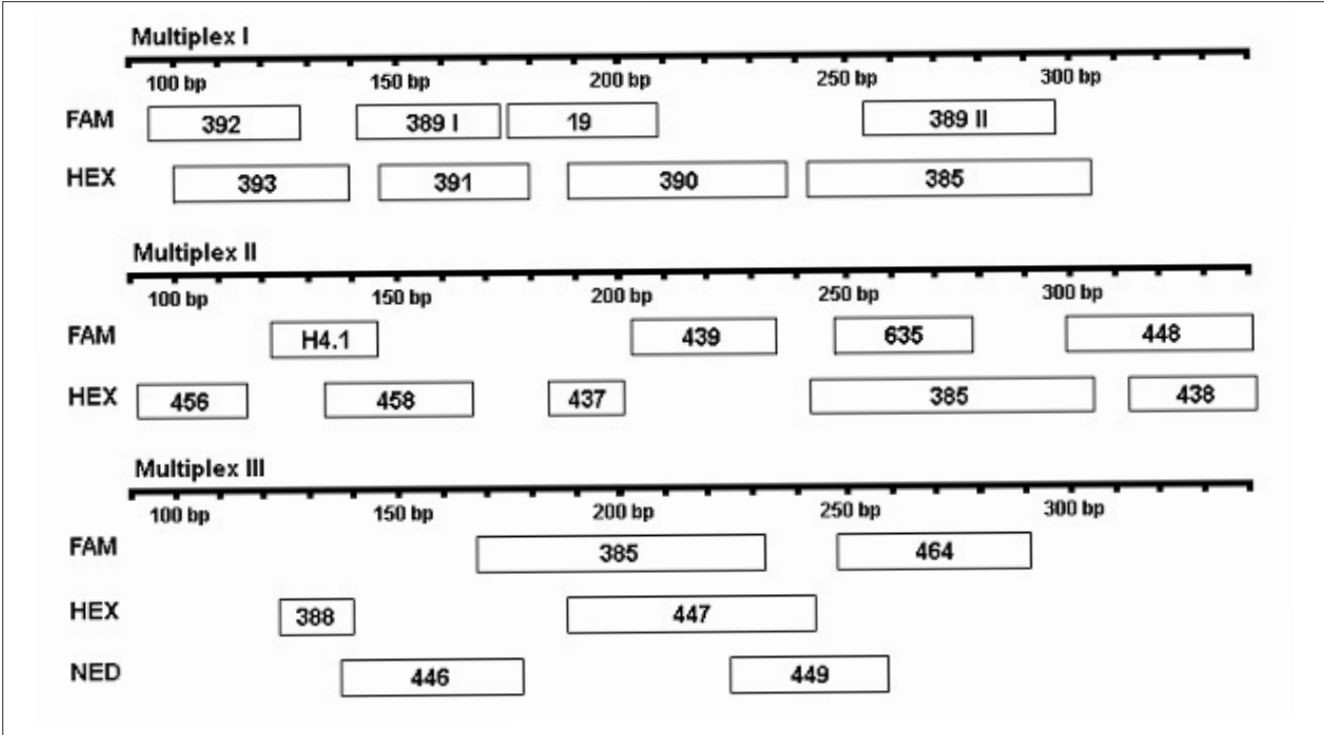


Fig. 1. Schematic of PCR product size for the loci in each multiplex. Marker names have been abbreviated (e.g. DYS392 is listed as 392)

22 Y-STR (multiplex PCR) primer (Fig. 1), PCR¹⁴⁾ kit (Applied Biosystems, Foster City, CA) ABI 310 PRISM Genetic Analyzer Duplicated PCR pGEM-T Easy Vector Systems I (Promega, Madison, WI)

PTC-200 DNA engine (MJ Research, Waltham, MA), PCR ABI PRISM 310 Genetic Analyzer (Applied Biosystems, Foster City, CA) capillary, GeneScan software 3.7 (Applied Biosystems, Foster City, CA) 가 Arlequin²⁰⁾, discriminatoty capacity

. *Allele ladder* PCR genotyping ladder minimal haplotype extended SWGDAM haplotype Terminator Cycle Sequencing kit (Applied Biosystems, Foster City, CA) ABI PRISM 310 Genetic Analyzer, International Society for Forensic Haemogenetics (ISFG)¹⁶⁾ PCR genotyping allele ladder BigDye extended SWGDAM discriminatory capacity GenoTyper Software 3.7 (Applied Biosystems, Foster City, CA) 가 haplotype Y-STR 가

GATA H4 GATA H4.1 Gusmão¹⁸⁾ AmpFISTR Yfiler™ kit¹⁷⁾ 1. microvariant 가 DYS447, DYS449, DYS458 DYS464 32 (Table 1). DYS447 (core repeat unit) TAAAA - (TAATA)_n (TAATA)_n - TAAAA가 가 6 가 microvariant DYS449 (TTTC) (27.2, 28.2, 29.2, 30.2) (25, 30.1, 42)

Table 1. Microvariant alleles observed in 708 Korean males

Locus	Allele ^a	Structure ^b	N
DYS447	18	(TAATA) ₇ TAAAA(TAATA) ₁₀ <u>TAAAA(TAATA)_n</u>	2
	19	(TAATA) ₇ TAAAA(TAATA) ₁₁ <u>TAAAA(TAATA)_n</u>	4
DYS449	25 (26)	(TTTC) ₁₂ -tctctctcctctc-(tttc) ₂ -N4-(tttc) ₂ -N12-cttc-(TTTC) ₁₄	1
	27.2	(TTTC) ₃ TC(TTTC) ₁₀ -tctctctcctctc-(tttc) ₂ -N4-(tttc) ₂ -N12-cttc-(TTTC) ₁₄	1
	28.2	(TTTC) ₃ TT(TTTC) ₁₁ -tctctctcctctc-(tttc) ₂ -N4-(tttc) ₂ -N12-cttc-(TTTC) ₁₄	1
	29.2	(TTTC) ₃ TC(TTTC) ₁₁ -tctctctcctctc-(tttc) ₂ -N4-(tttc) ₂ -N12-cttc-(TTTC) ₁₅	1
	30.1 (30)	(TTTC) ₁₆ -tctctctcctctc-(tttc) ₂ -N4-(tttc) ₂ -N12-cttc-(TTTC) ₁₄	1
	30.2	(TTTC) ₁₆ -tctctctcctctc-(tttc) ₂ -N4-(tttc) ₂ -N12-cttc-(TTTC) ₁₀ TT(TTTC) ₄	12
DYS458	42 (33)	(TTTC) ₁₅ -tctctctcctctc-[(tttc) ₂ -N4-(tttc) ₂ -N12-cttc] ₂ -(TTTC) ₁₈	1
	14.1	(GAAA) ₁₄ G	1
DYS464	17.2	(GAAA) ₁₅ AA(GAAA) ₂	1
	12.3	(CCTT) ₇ CCTT(CCTT) ₅	1
	14.3	(CCTT) ₃ CCTT(CCTT) ₁₁	5

^a The numbers in parentheses are genuine alleles identified by sequence analysis

^b Deletion and insertion are lined and underlined, respectively.

Table 2. Allele frequencies and gene diversities of single copy 19 Y-STR loci in a Korean population

Allele	DYS 19	DYS 388	DYS 389I	DYS 389II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 437	DYS 438	DYS 439	DYS 446	DYS 447	DYS 448	DYS 449	DYS 456	DYS 458	DYS 635	GATA H4.1
7						0.0014													
8						0.0268													
9						0.8475	0.0028	0.0014		0.0085	0.0028								
10		0.1271				0.1243	0.1554			0.5056	0.0438								
11		0.0028	0.0113				0.1017	0.3814		0.1525	0.2853	0.0763							
12		0.6850	0.3715				0.4901	0.4619	0.0014	0.0268	0.5141	0.1342				0.0169			
13	0.0240	0.1723	0.2359				0.2161	0.1073	0.6935	0.2980	0.1370	0.3573				0.0141	0.0085		
14	0.1540	0.0085	0.3729				0.0240	0.0424	0.2952	0.0085	0.0169	0.2175				0.1314	0.0085		
15	0.3799	0.0042	0.0071				0.0099	0.0056	0.0099	0.0791		0.0791				0.6766	0.0720		
16	0.3291									0.0678						0.1201	0.1511		
17	0.1102									0.0410				0.0339		0.0311	0.3107		
18	0.0014									0.0169			0.0028	0.3573		0.0099	0.2825		
19										0.0099			0.0056	0.2373		0.0099	0.1328	0.0523	0.0424
20					0.0014									0.2345			0.0254	0.2726	0.4266
21					0.0056									0.0960			0.0056	0.4661	0.4463
22					0.1229					0.0014				0.0297				0.1342	0.0819
23					0.4873					0.0071				0.1455				0.0466	0.0028
24					0.2444					0.2556				0.0028				0.0268	
25*					0.1243					0.3799				0.0028				0.0268	
26					0.0014	0.0113				0.1201				0.0042				0.0014	
27					0.0734	0.0014				0.0537				0.0325				0.0014	
28					0.2401					0.0212				0.0508				0.0014	
29					0.3983					0.0056				0.1144				0.0014	
30					0.2288					0.0212				0.2189				0.0014	
31					0.0523					0.0056				0.2105				0.0014	
32					0.0056					0.0028				0.1610				0.0014	
33										0.0960				0.0960				0.0014	
34										0.0664				0.0664				0.0014	
35										0.0155				0.0155				0.0014	
36										0.0028				0.0028				0.0014	
42*										0.0014				0.0014				0.0014	
Microvariants, duplicated and null alleles																			
14.1																			
17.2																			
27.2																			
28.2																			
29.2																			
30.1*																			
30.2																			
16-17	0.0014																		
22-23																			
23-24																			
Null																			
h	0.7119	0.4855	0.6681	0.7242	0.6731	0.2660	0.6789	0.6288	0.4324	0.6323	0.6342	0.7894	0.7523	0.7508	0.8521	0.5098	0.7783	0.6858	0.6112

* At DYS449, these alleles were determined by amplicon size (i.e. ostensible allele) and contain only one of ostensible allele 25.

. DYS458 DYS464
GAAA CCTT

Duplicated DYS19, DYS390 DYS447
(Fig. 2)

diversity 가 , DYS448
gene diversity 가
diversity 가 , extended
SWGDAM haplotype DYS458 가
DYS635 가 diversity
가 가 가 가 DYS447
GATA H4.1
diversity가 가 DYS447,
DYS458, DYS635 GATA H4.1 extended
SWGDAM haplotype 가 AmpFISTR
Yfiler™ haplotype diversity

2. Gene diversity Haplotype
single - copy Y - STR
gene diversity Table 2 multi - copy Y - STR
DYS385 DYS464 gene
diversity Table 3 . Single - copy
DYS449 가 가 gene diversity (0.8521)
, DYS446 (0.7894), DYS458 (0.7783)
가 DYS447 DYS448 가
gene diversity , DYS391 가 가
gene diversity (0.2660)
22 Y - STR
(http://forensic.yonsei.ac.kr) 가
708 693 가 . Y -
680 가 (98.12%) ISFG Y -
11 , 2 STR STR²¹⁾
. Minimal haplotype, extended , microvariant
SWGDAM haplotype AmpFISTR Yfiler™ haplotype 가
diversity 4
microvariant (Table 1), DYS449
27.2, 28.2, 29.2, 30.2 DYS458 14.1,
17.2 DYS464 12.3, 14.3
가 가 가
diversity 가 (Table 5). gene diversity microvariant GenoTyper

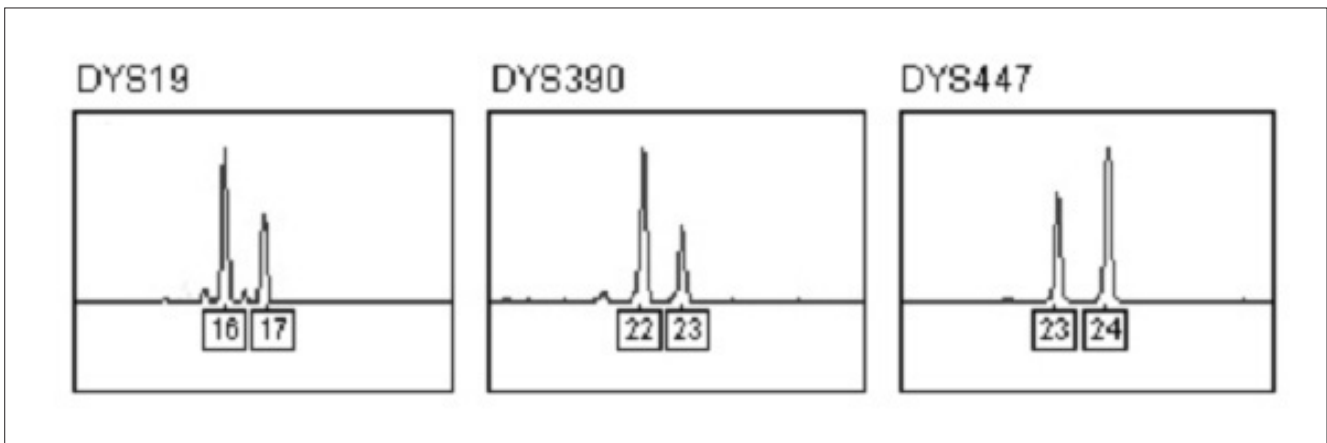


Fig. 2. Electropherogram for duplicated alleles at the single copy loci DYS19, DYS390 and DYS447

Table 3. Haplotype distribution and diversity of multi-copy Y-STRs in a Korean population

DYS385		DYS464					
Haplotype	Frequency	Haplotype	Frequency	Haplotype	Frequency	Haplotype	Frequency
8-20	0.0014	12-21	0.0014	12	0.0014	13-15-18	0.0127
9-16	0.0014	13-13	0.0226	13	0.0127	13-15-19	0.0014
9-18	0.0028	13-14	0.0071	14	0.0028	13-16-17	0.0282
9-19	0.0155	13-15	0.0028	15	0.0028	13-16-18	0.0113
9-20	0.0028	13-16	0.0169	16	0.0085	13-17-18	0.0028
10-10	0.0014	13-17	0.0212	17	0.0141	14-15-16	0.0042
10-16	0.0042	13-18	0.0537	11-17	0.0014	14-15-17	0.0028
10-17	0.0480	13-19	0.0381	12-13	0.0042	14-15-18	0.0014
10-18	0.1045	13-20	0.0254	12-14	0.0014	14-16-17	0.0071
10-19	0.0975	13-21	0.0028	12-16	0.0056	14-16-18	0.0028
10-20	0.0339	14-14	0.0028	13-14	0.0466	15-16-17	0.0014
10-21	0.0113	14-16	0.0028	13-15	0.0452	11-12-13-15	0.0014
11-11	0.0113	14-17	0.0141	13-16	0.0127	11-12-13-16	0.0014
11-12	0.0184	14-18	0.0212	13-17	0.0056	11-13-14-16	0.0014
11-13	0.0141	14-19	0.0113	13-18	0.0014	11-13-16-18	0.0014
11-14	0.0071	14-20	0.0155	14-15	0.0056	11-15-16-17	0.0014
11-15	0.0014	14-21	0.0085	14-16	0.0141	12-13-14-15	0.0353
11-16	0.0127	14-22	0.0042	14-17	0.0014	12-13-14-16	0.0311
11-17	0.0282	15-15	0.0028	15-16	0.0198	12-13-14-17	0.0113
11-18	0.0099	15-17	0.0014	15-17	0.0071	12-13-15-16	0.0113
11-19	0.0353	15-18	0.0014	16-17	0.0113	12-13-15-17	0.0056
11-20	0.0155	15-19	0.0071	16-18	0.0028	12-13-15-18	0.0014
11-21	0.0028	15-20	0.0184	17-18	0.0028	12-13-16-17	0.0028
12-12	0.0028	15-21	0.0127	14-14.3	0.0071	12-13-16-18	0.0042
12-13	0.0042	15-22	0.0085	10-13-15	0.0014	12-13-16-19	0.0014
12-14	0.0113	15-23	0.0014	10-14-16	0.0014	12-14-15-16	0.0311
12-15	0.0042	16-17	0.0014	11-12-17	0.0042	12-14-15-17	0.0056
12-16	0.0240	16-20	0.0028	11-13-14	0.0014	12-14-15-18	0.0014
12-17	0.0381	16-21	0.0028	11-13-15	0.0014	12-14-16-17	0.0028
12-18	0.0452	17-20	0.0028	11-14-16	0.0014	12-14-16-18	0.0014
12-19	0.0410	19-19	0.0014	12-13-14	0.0254	12-15-16-17	0.0071
12-20	0.0085	19-20	0.0014	12-13-15	0.0466	12-15-16-18	0.0014
				12-13-16	0.0042	12-15-17-18	0.0014
				12-13-17	0.0014	12.3-14-15-17	0.0017
				12-14-15	0.0254	13-14-15-16	0.0127
				12-14-16	0.0339	13-14-15-17	0.0099
				12-14-17	0.0056	13-14-15-18	0.0014
				12-15-16	0.0071	13-14-16-17	0.1045
				12-16-17	0.0028	13-14-16-18	0.0099
				12-16-18	0.0014	13-14-16-19	0.0014
				13-14-15	0.0494	13-14-17-18	0.0042
				13-14-16	0.0579	13-15-16-17	0.0282
				13-14-17	0.0198	13-15-16-18	0.0071
				13-14-18	0.0042	13-15-16-19	0.0014
				13-15-16	0.0254	14-15-16-17	0.0042
				13-15-17	0.0127		

h 0.9597 h 0.9669

가 TCTC가 T가 36 bp가
 가 , DYS449 duplication
 가 ISFG
 GenoTyper 25, 30.1 42 26, 30, 33
 , DYS449.1 DYS449.2

ISFG	30.1	haplotype	extended SWGDAM haplotype
DYS449.1*16 + DYS449.2*14 (U4Tins)		가	, extended
		SWGDAM haplotype	Y - STR 가
		discriminatory capacity	가 가
duplication triplication		DYS449	single - copy gene diversity가
22) DYS19, DYS385, DYS389I, DYS389II,		가	diversity 가 ,
DYS390, DYS391, DYS393, DYS437, DYS438, DYS439			germ line
DYS448	DYS19	¹⁴⁾	가
DYS385 duplication 가		extended SWGDAM haplotype	가
	DYS385	가	gene
duplication triplication		diversity	DYS446 diversity
DYS447 duplicated	23 - 24	-	가 homologous ^{14,23)}
germ line (23 - 24 24)가	¹⁴⁾		가 . DYS456
Y - STR		DYS446 가 X	homologous ²⁴⁾
가 duplication		, DYS388 DYS437	diversity
	duplication	가	
	peak가	discriminatory capacity 가	Y - STR
DNA profiles		. DYS448	AmpFISTR Yfiler™
, Y - STR	minimal	kit diversity	gene diversity 가

Table 4. Number of haplotypes and diversities for 22 Y-STRs in 708 Korean males

	Minimal 9 Y-STR haplotype	Extended SWGDAM 11 Y-STR haplotype	Y-filer™ 17 Y-STR haplotype	22 Y-STR haplotype
No. of haplotypes	485	558	657	693
No. of unique haplotypes	394	483	625	680
Discriminatory capacity (%)	68.5	78.8	92.8	97.9
Haplotype diversity (\pm SD)	0.9966 \pm 0.0005	0.9982 \pm 0.0004	0.9995 \pm 0.0002	0.9999 \pm 0.0001

Table 5. Numbers of haplotypes and haplotype diversities obtained by adding each marker to the extended SWGDAM haplotype

Haplotype	Gene diversity*	No. of haplotypes ^c	Discriminatory capacity (%)	Haplotype diversity
Extended SWGDAM haplotype		558	78.8	0.9982
Extended SWGDAM haplotype + DYS449	0.8521	612	86.4	0.9992
Extended SWGDAM haplotype + DYS446	0.7894	598	84.5	0.9989
Extended SWGDAM haplotype + DYS458	0.7783	616	87.0	0.9992
Extended SWGDAM haplotype + DYS447	0.7523	583	82.3	0.9986
Extended SWGDAM haplotype + DYS448	0.7508	565	79.8	0.9983
Extended SWGDAM haplotype + DYS635	0.6858	591	83.5	0.9988
Extended SWGDAM haplotype + GATA H4.1	0.6112	579	81.8	0.9985
Extended SWGDAM haplotype + DYS456	0.5098	574	81.1	0.9984
Extended SWGDAM haplotype + DYS388	0.4855	566	79.9	0.9983
Extended SWGDAM haplotype + DYS437	0.4324	568	80.2	0.9983
Extended SWGDAM haplotype + DYS458 + DYS635		639	90.3	0.9994
Extended SWGDAM haplotype + DYS458 + DYS447		630	89.0	0.9994
Extended SWGDAM haplotype + DYS458 + GATA H4.1		631	89.1	0.9993
Extended SWGDAM haplotype + DYS458 + DYS635 + DYS447		650	91.8	0.9996
Extended SWGDAM haplotype + DYS458 + DYS635 + GATA H4.1		649	91.5	0.9995
Extended SWGDAM haplotype + DYS458 + DYS635 + DYS447 + GATA H4.1		658	92.9	0.9996
Y-filer™ 17 Y-STR haplotype		657	92.8	0.9995

* Gene diversity of added marker to extended SWGDAM haplotype loci

(AZFc) , DYS448 가 azoospermia factor c microdeletion null
 가 Y 25-29)
 가
 , DYS464 multi-copy
 gene diversity 가
 DNA
 , DYS448 가 AZFc
 30,31) DYS448 null
 25) 가
 DYS458 single-copy 가 gene diversity 가 extended SWGDAM haplotype diversity
 가 가
 discriminatory capacity 가
 Y-STR , diversity 가
 가 DYS458, DYS635, DYS447, GATA H4.1
 4 extended SWGDAM haplotype
 가 15 17
 AmpFISTR Yfiler™ haplotype diversity 가 ,
 , 15 Y-STR haplotype
 AmpFISTR Yfiler™ haplotype discriminatory capacity 93% , Y-STR
 haplotype
 Y-STR 가
 708
 22 Y-STR
 ,
 . Microvariant 32 가
 DYS449 가 ,
 microvariant
 . DYS19, DYS390 DYS447
 duplicated 가 ,
 25) DYS385
 deletion AZFc
 DYS448 DYS464 deletion

가
 708 22 Y-STR 693
 680 가
 . Descimatory capacity 가
 extended SWGDAM haplotype
 DYS458 diversity가
 가 가 , DYS447, DYS458, DYS635,
 GATA H4.1 가 (15) 17
 AmpFISTR Yfiler™ haplotype
 discriminatory power 가 .

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