

# Highly informative Y-chromosomal haplotypes of four Y-specific STR loci, DYS385, DYS446, DYS449 and DYS464

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## Introduction

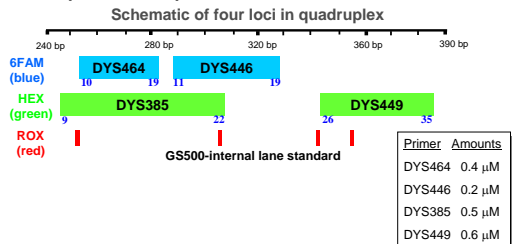
Typing of Y-specific STR has become very useful in population genetics and forensic casework, e.g. in deficiency paternity testing and in rape cases involving one or more semen donors. So far, numerous Y-STRs have been discovered and some have been used for forensic purposes and population studies to evaluate diversity of the haplotypes. More recently, new highly informative Y-STRs (DYS446, DYS449 and DYS464) have been identified and expected to be useful for the establishment of small but efficient subset of STRs. In this work, we examined the usefulness of DYS385, DYS446, DYS449 and DYS464 highly informative Y-STRs through determination of allele and haplotype diversity as well as through comparing that of minimal haplotype.

## Materials and Methods

### ◆ DNA samples

A sample of 301 male was obtained from unrelated Korean population. DNA was extracted from buccal swab samples using QIAamp DNA Mini Kit (Qiagen, Hilden, Germany).

### ◆ Multiplex PCR amplification conditions



PCR reaction: Total 10 μl reaction contained 1-2 ng of DNA, 1.4 μl of Gold STR buffer (Promega, Madison, WI), 2.0 U of AmpliTaq Gold polymerase (Applied Biosystems, Foster City, CA), and primers  
Cycling condition: 95°C 1'; 94°C 1'; 59°C 1'; 72°C 1' x 30; 60°C 45'

### ◆ Detection Systems

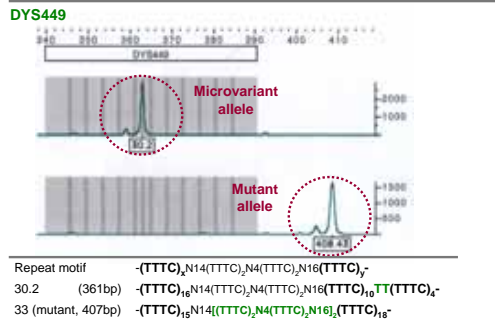
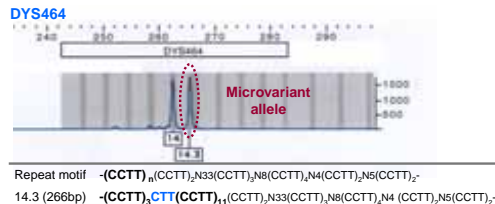
ABI prism 310 Genetic Analyzer and Gene Scan software 3.1 were used for sizing of each allele. The typing was performed using Genotyper 2.5 software (Applied Biosystems, Foster City, CA).

### ◆ Statistical analysis

Gene and haplotype diversity values were calculated using Arlequin version 2.0 software according to Nei. The discrimination capacity was determined by dividing the number of different haplotype by the total number of samples in a Korean population.

## Results

### ◆ Structure of variant alleles



### ◆ Single locus analysis

DYS446		DYS449	
Allele	Frequency	Allele	Frequency
11	0.0565	26	0.0033
12	0.1096	27	0.0299
13	0.3621	28	0.0532
14	0.2259	29	0.1130
15	0.1030	30	0.2525
16	0.0631	31	0.0199
17	0.0532	32	0.2027
18	0.0233	33	0.1561
19	0.0033	34	0.0963
<b>h<sup>a</sup></b>	<b>0.7873</b>	35	0.0631
		<b>h<sup>a</sup></b>	<b>0.8433</b>

<sup>a</sup> gene diversity

### Multi-copy Y STRs

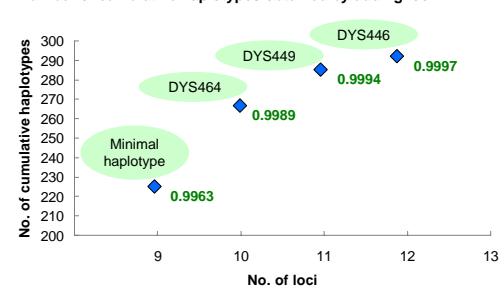
DYS385				DYS464			
Haplotype	Frequency	Haplotype	Frequency	Haplotype	Frequency	Haplotype	Frequency
9-18	0.0033	12-20	0.0100	12	0.0033	13-14-16	0.0698
9-19	0.0133	13-13	0.0233	13	0.0233	13-14-17	0.0166
9-20	0.0066	13-14	0.0033	16	0.0033	13-14-18	0.0033
10-16	0.0033	13-16	0.0100	17	0.0066	13-15-16	0.0365
10-17	0.0465	13-17	0.0133	12-13	0.0033	13-15-17	0.0133
10-18	0.1096	13-18	0.0664	12-14	0.0033	13-15-18	0.0199
10-19	0.0963	13-19	0.0266	12-16	0.0066	13-16-17	0.0332
10-20	0.0399	13-20	0.0332	13-14	0.0465	13-16-18	0.0033
10-21	0.0166	13-21	0.0066	13-15	0.0332	14-15-16	0.0066
11-11	0.0100	14-14	0.0033	13-16	0.0133	14-16-17	0.0133
11-12	0.0233	14-16	0.0033	13-17	0.0033	11-12-13-15	0.0033
11-13	0.0166	14-17	0.0100	14-14.3	0.0066	11-12-13-16	0.0033
11-14	0.0133	14-18	0.0133	14-15	0.0066	12-13-14-15	0.0432
11-15	0.0033	14-19	0.0033	14-16	0.0033	12-13-14-16	0.0299
11-16	0.0133	14-20	0.0166	15-16	0.0299	12-13-14-17	0.0133
11-17	0.0365	14-21	0.0100	15-17	0.0066	12-13-15-16	0.0100
11-18	0.0133	15-15	0.0033	16-17	0.0100	12-13-15-17	0.0066
11-19	0.0299	15-19	0.0100	16-18	0.0033	12-13-16-17	0.0033
11-20	0.0233	15-20	0.0100	17-18	0.0033	12-13-16-18	0.0100
12-12	0.0066	15-21	0.0166	10-14-16	0.0033	12-14-15-16	0.0166
12-13	0.0033	15-22	0.0066	11-12-17	0.0100	12-14-15-17	0.0066
12-14	0.0166	16-17	0.0033	11-13-14	0.0033	12-14-15-18	0.0033
12-16	0.0233	16-20	0.0033	11-13-15	0.0033	12-14-16-18	0.0033
12-17	0.0299	19-19	0.0033	12-13-14	0.0332	12-15-16-17	0.0133
12-18	0.0532	19-20	0.0033	12-13-15	0.0432	13-14-15-16	0.0133
12-19	0.0365			12-13-16	0.0033	13-14-15-17	0.0066
				12-13-17	0.0033	13-14-16-17	0.1063
				12-14-15	0.0233	13-14-16-18	0.0033
				12-14-16	0.0299	13-14-16-19	0.0033
				12-14-17	0.0033	13-14-17-18	0.0100
				12-15-16	0.0100	13-15-16-17	0.0199
				12-16-17	0.0033	13-15-16-18	0.0066
				12-16-18	0.0033	14-15-16-17	0.0100
				13-14-15	0.0465		
				<b>h<sup>a</sup></b>	<b>0.9668</b>		

<sup>a</sup> Haplotype diversity

### ◆ Haplotype analysis

	Minimal haplotype	New set of 4 loci	Combined set of 12 loci
Different haplotypes	230	269	292
Haplotype diversity	0.9963	0.9989	0.9997
Discrimination capacity	0.7641	0.8937	0.9701

### Number of cumulative haplotypes obtained by adding loci



## Conclusion

Analyzed allelic sequences indicated that DYS385 (GAAA), DYS449 (TTTC), DYS464 (CCTT) have tetramer repeat unit and that DYS464 (TCTCT) has pentamer unit. This study found intermediated alleles in both DYS464 (allele 14.3) and DYS449 (allele 30.2) and mutant allele in DYS449 (allele 33, but its 36bp flanking region is duplicated).

DYS464, DYS385, DYS449 and DYS446, in order of those, are highly diverse in a Korean population.

DYS464, DYS385, DYS449 and DYS446 are highly informative markers not only because the combination of these loci showed the higher capacity (0.9989) to distinguish paternal lineages than the minimal haplotype consisting of nine core Y-STRs loci (0.9963) but also because the addition of respective loci to minimal haplotype core set significantly increases the power of discrimination by increasing the number of different haplotypes in a Korean population.

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