

# Forensic evaluation and haplotypes of 19 Y-chromosomal STR loci in Koreans

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## Y-chromosomal STRs



### STR Markers

DYS19  
DYS389I/II  
DYS390  
DYS391  
DYS392  
DYS393  
DYS385  
DYS388  
DYS434  
DYS435  
DYS436  
DYS437  
DYS438  
DYS439  
⋮

### Applications

- forensic investigations
- genealogical purposes
- evolutionary studies

### Advantages to Human Identity Testing

- male component isolated without differential extraction
- paternal lineages

### Needs

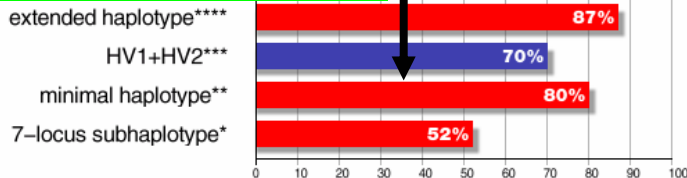
- population studies to evaluate diversity of haplotypes
- robust assay for accurate characterization of Y markers

# Minimal haplotype loci

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**Y STR**  
**DATABASE**
**Y-STR Haplotype Reference Database**
<http://www.ystr.org/asia>
**9 best studied Y STRs**

DYS19 DYS389I DYS389II DYS390 DYS391 DYS392 DYS393 DYS385

**Percentage of unique haplotypes**


\* n = 2576 7-locus Asian haplotypes, DYS385 excluded, logged in the database

\*\* n = 2576 minimal Asian haplotypes logged in the database

\*\*\* n = 850 European mt-DNA D-Loop sequences (from Legal Medicine Magdeburg, Germany)

\*\*\*\* n = 551 extended Asian haplotypes logged in the database

# Previously study of Y-STRs

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International Journal of Legal Medicine 115 (2001) 109-117

**Y-chromosome multiplexes and their potential for the DNA profiling of Koreans.**

D. J. Shin, H. J. Jin, K. D. Kwak, J. W. Choi, M. S. Han, P. W. Kang, S. K. Cho, W. Kim

Kor. J. Legal Med 27 (2003) 62-72

**Polymorphism and Haplotypes of 10 Y-STR Loci in Koreans**

Zhe-Jia Zheng, Yong-Ji Zhang, Jin-Cheol Park, Hai-Yu Lin, S—Young Kim, Young-Tae Choi, Jung Bin Lee

Forensic Science International 136 (2003) 89-91

**Allele frequencies and haplotypes of six new Y-specific STR loci in Koreans**

Hwan Young Lee, Ji-Eun Oh, Gil-Ro Han, Kyoung-Jin Shin

**DYS434, DYS435, DYS436**  
**DYS437, DYS438, DYS438**

# Selection of new Y-STRs

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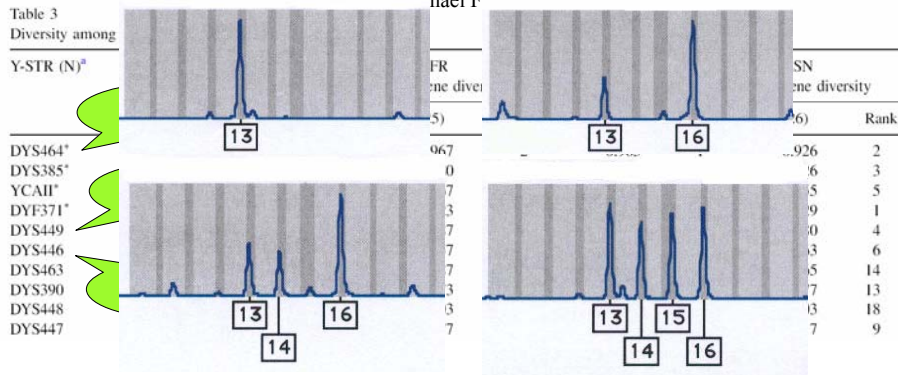
Forensic Science International 126 (2002) 97-111

## DYS464 : four-copy locus

## the human Y chromosome

Learney, Veronica A. Contreras,  
de Knijff, John M. Butler,

### DYS464 a/b/c/d



# Object

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- Usefulness of **DYS464, DYS449, DYS446**
- **Gene/haplotype diversity and discriminatory power** of 19 STR loci containing **minimal haplotype loci, DYS388, DYS434, DYS435, DYS436, DYS437, DYS438 and DYS439**
- Assess of contribution of respective Y-STRs to the **increase of haplotype diversity**

# Materials and Methods

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- DNA samples
  - 301 buccal swab samples from unrelated Korean population
  - DNA was extracted using QIAamp DNA Mini Kit
- Multiplex PCR
  - Total 10  $\mu$ l PCR reaction: 1-2 ng of DNA, 1.4 -1.6  $\mu$ l of Gold STR buffer, 2.0 U of AmpliTaq Gold polymerase and primers
  - Cycling condition: 95°C 11'; 94°C 1' 55-59°C 1' 72°C 1' x 30; 60°C 45'
- Detection Systems
  - ABI prism 310 genetic analyzer, Gene Scan software 3.1, and Genotyper 2.5 software (PE Applied Biosystems, CA, USA)



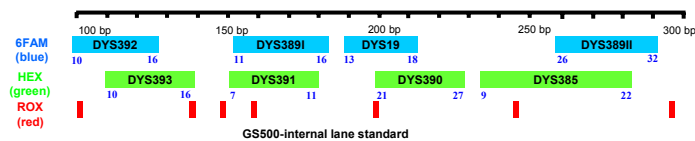
## Statistical analysis

- Gene, haplotype diversity and linkage disequilibrium values were calculated using Arlequin version 2.0 software.

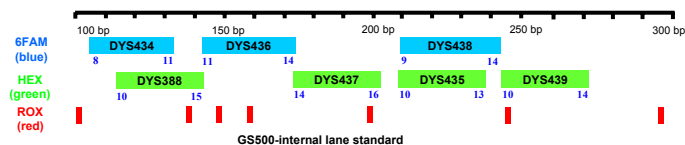
# Schematic of multiplex PCR

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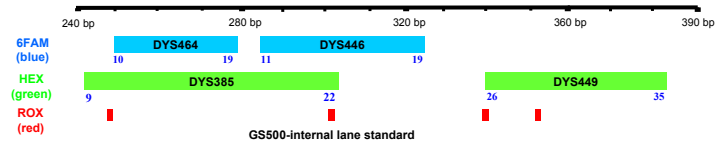
## Multiplex I (minimal haplotype)



## Multiplex II (DYS388+previously studied 6 Y-STRs)

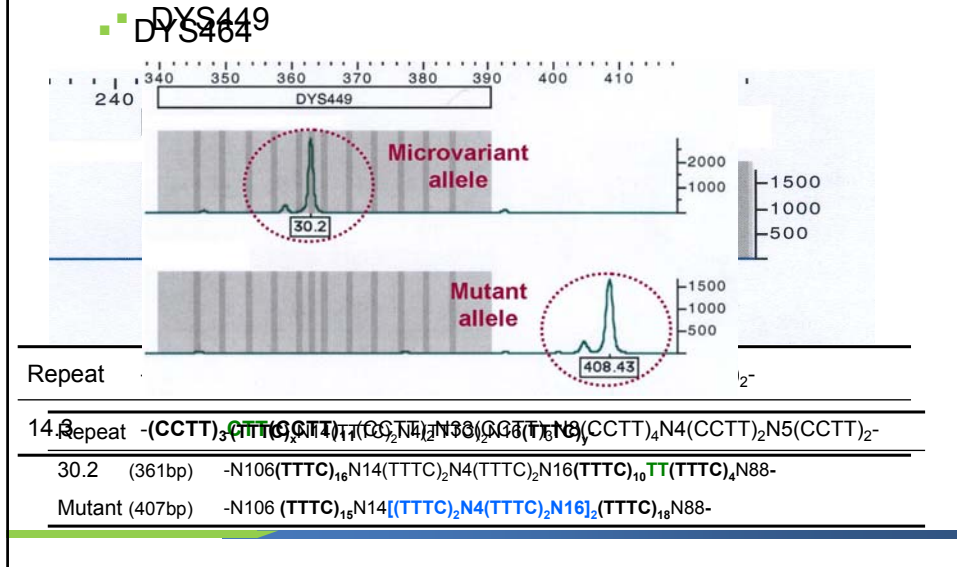


## Multiplex III (DYS464, DYS446 and DYS385)



## Allelic structure

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## Single locus analysis

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Marker	Motif	#Allele seen	# Repeat unit	Gene diversity
DYS19	TAGA	6	13 – 18	<b>0.7167</b>
DYS389I	(TCTG)(TCTA)	6	11 – 16	<b>0.6659</b>
DYS389II	(TCTG)(TCTA)	7	26 – 32	<b>0.7257</b>
DYS390	(TCTA)(TCTG)	7	21 – 27	<b>0.6689</b>
DYS391	TCTA	4	7, 9 – 11	<b>0.2924</b>
DYS392	TAT	7	10 – 16	<b>0.6931</b>
DYS393	AGAT	6	10, 12 – 16	<b>0.6338</b>
<b>DYS385</b>	GAAA	51	9-18 – 19-20	<b>0.9590</b>
DYS388	ATT	5	10, 12 – 15	<b>0.5083</b>
<b>DYS446</b>	TCTCT	9	11 – 19	<b>0.7873</b>
<b>DYS449</b>	TTTC	11	26-30, 30.2, 31 – 35	<b>0.8433</b>
<b>DYS464</b>	CCTT	67	12 – 14-15-16-17	<b>0.9668</b>

## Haplotype analysis

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Haploes	H12	H254	1	16	14	29	25	10	11	15	10,21	12	11	11	12	14	10	13	17	32	12,14,16			
H11	H33	H1	Haplotypes	N	DYS-19	DYS-389I	DYS-389II	DYS-390	DYS-391	DYS-392	DYS-393	DYS-385	DYS-388	DYS-434	DYS-435	DYS-436	DYS-437	DYS-438	DYS-439	DYS-446	DYS-449	DYS-464		
H12	H34	H2	Y-STR marker																				13,14,17	
H13	H35																							13,14,17
H14	H36																							12,15,16,17
H15	H37																							
H16	H38																							
H17	H39		Multiplex I (minimal haplotype)																					17
H18	H40																							
H19	H41																							13,14
H20	H42																							
H21	H43		Multiplex II																					15,17
H22	H44																							
H23	H45																							12,15,16
H24	H46																							
H25	H47		Multiplex III																					13,15,18
H26	H48																							13,15,18
H27	H49																							13,15,18
H28	H50		DYS385+DYS464																					13,15,18
H29	H51																							12,14,15,16
H30	H52																							13,15,16
H31	H53		19 Y-STRs haplotype																					12,13,14,16
H32	H54																							12,14,17
H33	H55																							12,14,16
H34	H56	H1278		1	17	12	29	25	11	13	12	13,21	12	9	12	12	14	10	11	12	31			13,15,17
H35	H57	H1279		1	17	12	30	24	10	13	12	15,21	13	9	12	12	14	10	11	11	30			12,14,15,16
H36	H58	H1280		1	17	12	30	24	11	13	12	14,20	13	9	12	12	14	10	11	11	31			13,14,15,16
H37	H59	H1281		1	17	13	28	23	10	13	13	10,18	12	9	11	12	14	13	12	14	33			13,15,16,17
H38	H60	H1282		1	17	13	28	23	10	14	13	10,18	12	9	11	12	14	13	12	13	31			13,14,15,17
H39	H61	H1283		1	17	13	29	24	10	13	13	14,20	13	9	12	12	15	10	13	11	32			12,14,16

## Contribution of Y-STR loci to haplotype diversity

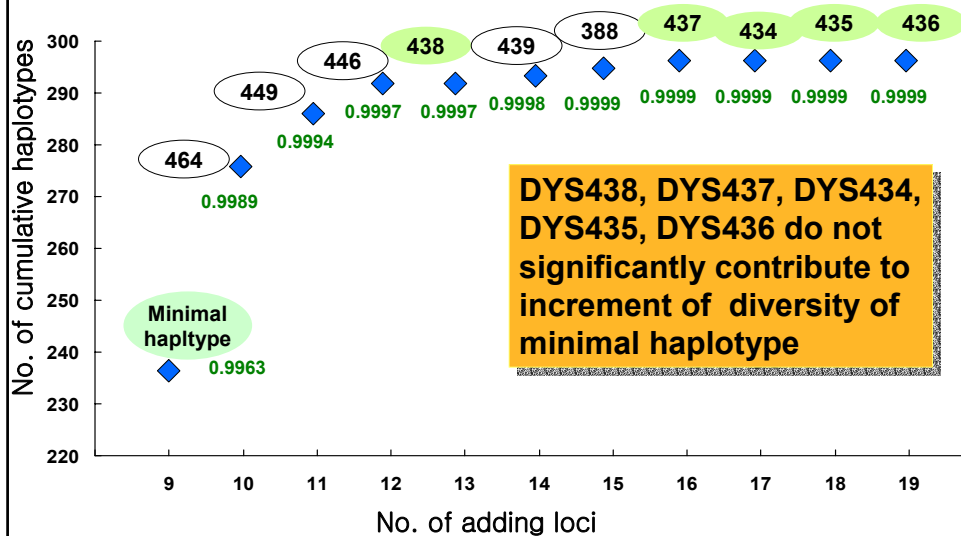
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Y-STR marker	# of LD loci <sup>a</sup>	Gene diversity	# of haplotype	Haplotype diversity
Minimal haplotype	—		<b>230</b>	<b>0.9963</b>
Minimal haplotype + <b>DYS388</b>	7	0.508	239	0.9968
Minimal haplotype + <b>DYS434</b>	7	<b>0.361</b>	<b>233</b>	<b>0.9967</b>
Minimal haplotype + <b>DYS435</b>	6	<b>0.259</b>	<b>234</b>	<b>0.9966</b>
Minimal haplotype + <b>DYS436</b>	5	<b>0.102</b>	<b>233</b>	<b>0.9967</b>
Minimal haplotype + <b>DYS437</b>	7	<b>0.413</b>	<b>236</b>	<b>0.9968</b>
Minimal haplotype + <b>DYS438</b>	8	<b>0.672</b>	<b>232</b>	<b>0.9964</b>
Minimal haplotype + <b>DYS439</b>	6	<b>0.610</b>	<b>255</b>	<b>0.9980</b>
Minimal haplotype + <b>DYS446</b>	7	<b>0.787</b>	<b>250</b>	<b>0.9980</b>
Minimal haplotype + <b>DYS449</b>	8	<b>0.843</b>	<b>258</b>	<b>0.9985</b>
Minimal haplotype + <b>DYS464</b>	8	<b>0.967</b>	<b>276</b>	<b>0.9989</b>

<sup>a</sup> Number of minimal haplotype core loci in significant linkage disequilibrium with each added Y-STR locus.

## Contribution of Y-STR loci to haplotype diversity

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## Conclusions

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- Haplotype diversity of 19 Y-STR loci was **0.9999** and **297** different haplotypes was identified.
- **DYS464 (0.9668)**, **DYS385 (0.9590)**, **DYS449 (0.8433)** and **DYS446 (0.7873)**, in order of those, are highly diverse in a Korean population
- DYS464, DYS449 and DYS446 are significantly increase the haplotype diversity of minimal haplotype.
- **DYS434, DYS435, DYS436, DYS437, DYS438** did not significantly increase the number of different haplotypes and the haplotype diversity.