

The diagram shows a horizontal chromosome segment with two orange semi-circular ends. The left end is labeled 'p' and the right end is labeled 'q'. A green cross-hatched rectangular area is located between the 'p' and 'q' arms, with the word 'heterochromatin' written below it.

# Y-STR Data Interpretation

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Science Advisory Committee

Richmond, VA  
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## NIST Activities with Y-STRs

- SRM 2395 (Human Y Chromosome Standard)
  - <http://www.cstl.nist.gov/biotech/strbase/SRM2395.htm>
- Characterized duplications and deletions
  - Butler et al. (2005) *J. Forensic Sci.* 50(4): 853-859
- Sequenced variant alleles
  - <http://www.cstl.nist.gov/biotech/strbase/STRseq.htm>
- Supplied ~20% of Yfiler 3561 database
  - <http://www.cstl.nist.gov/biotech/strbase/NISTpop.htm>
- Measured mutation rates with Yfiler loci
  - Decker et al. (2008) *FSI Genetics* 2(3): e31-e35

**26 publications since 2001 on NIST Y-chromosome work**

<http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

## Value of Y-STRs to Forensic Casework

**Y-STRs can extend range of potential solvable forensic cases**

- **Enabling detection of male DNA when mixed with excess female DNA**
  - Sexual assaults by vasectomized or azoospermic males (no sperm left behind to enable differential extraction)
  - Fingernail scrapings from sexual assault victims
  - Other bodily fluid mixtures (blood-blood, skin-saliva)
  - Extending length of time after assault for recovery of perpetrator's DNA profile (greater than 48 hours)
- Dealing with multiple male contributors
  - Gang rape situation to include or exclude potential contributors
- Gender clarification (with amelogenin Y null alleles)
- Extension of power of discrimination (with partial profiles)

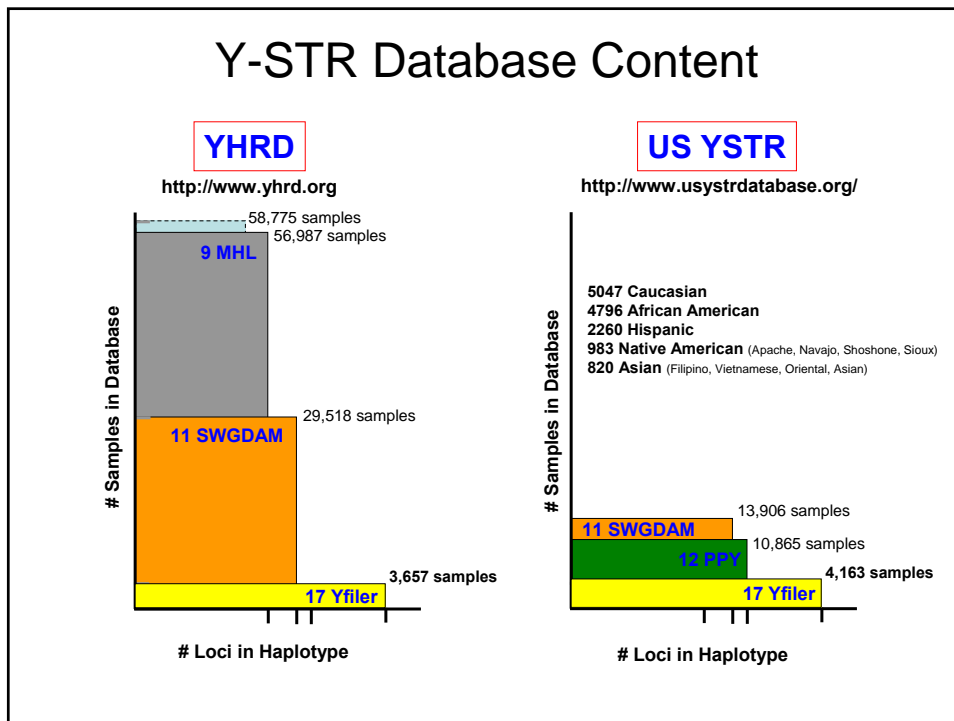
## Forensic Advantages of Y-STRs

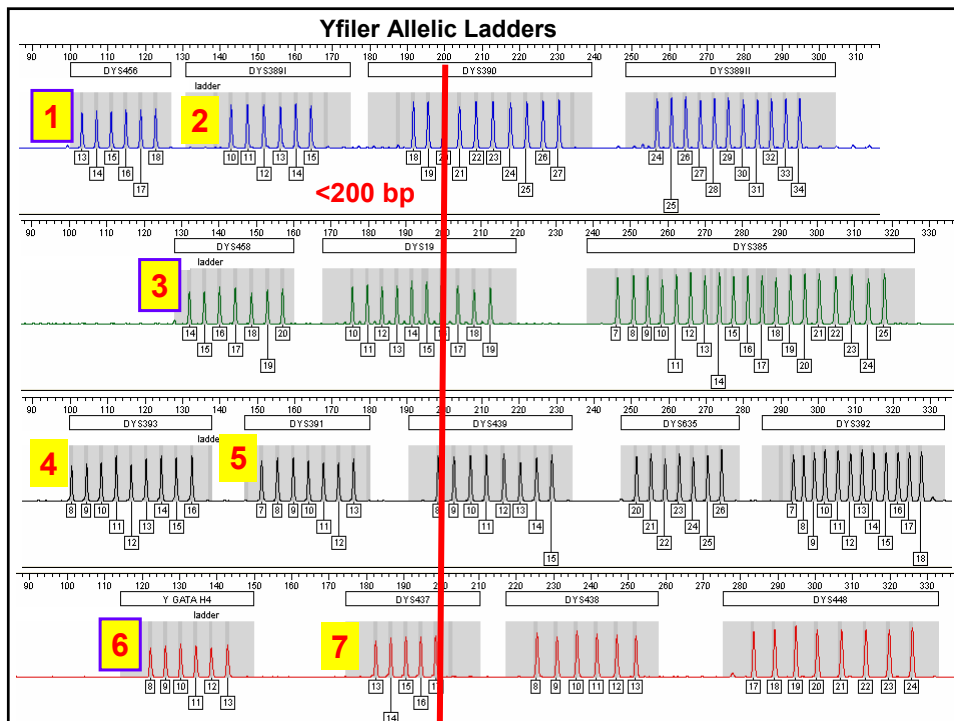
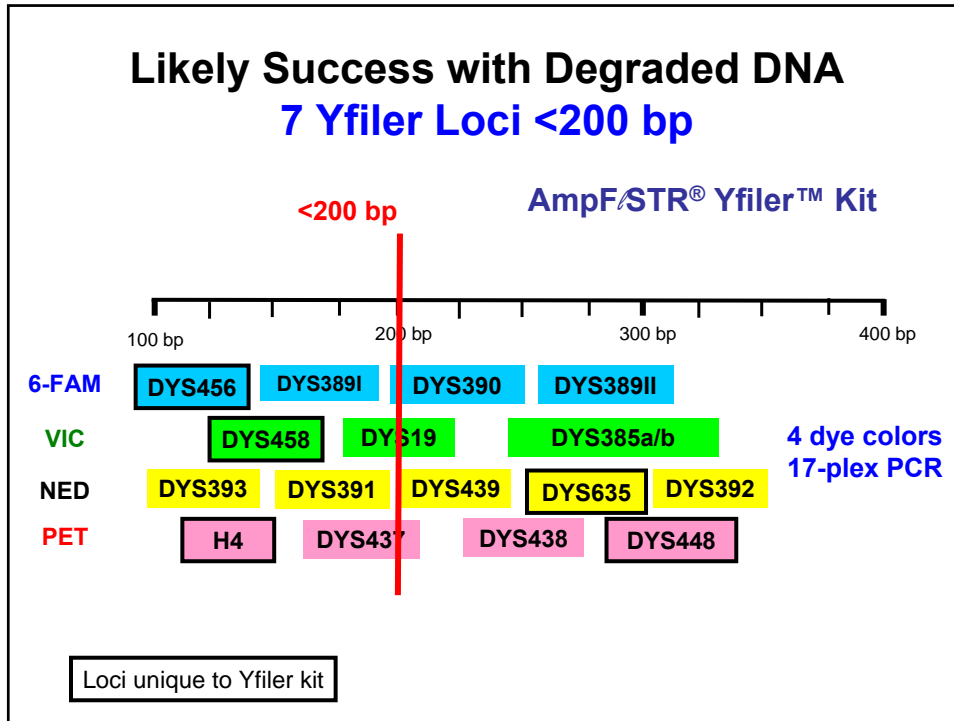
- **Male-specific amplification** extends range of cases accessible to obtaining probative DNA results (e.g., fingernail scrapings, sexual assault without sperm)
- **Technical simplicity due to single allele profile**; can potentially recover results with lower levels of male perpetrator DNA because there is not a concern about heterozygote allele loss via stochastic PCR amplification; number of male contributors can be determined
- **Courts have already widely accepted STR typing**, instrumentation, and software for analysis (Y-STR markers just have different PCR primers)
- **Acceptance of statistical reports using the counting method** due to previous experience with mtDNA

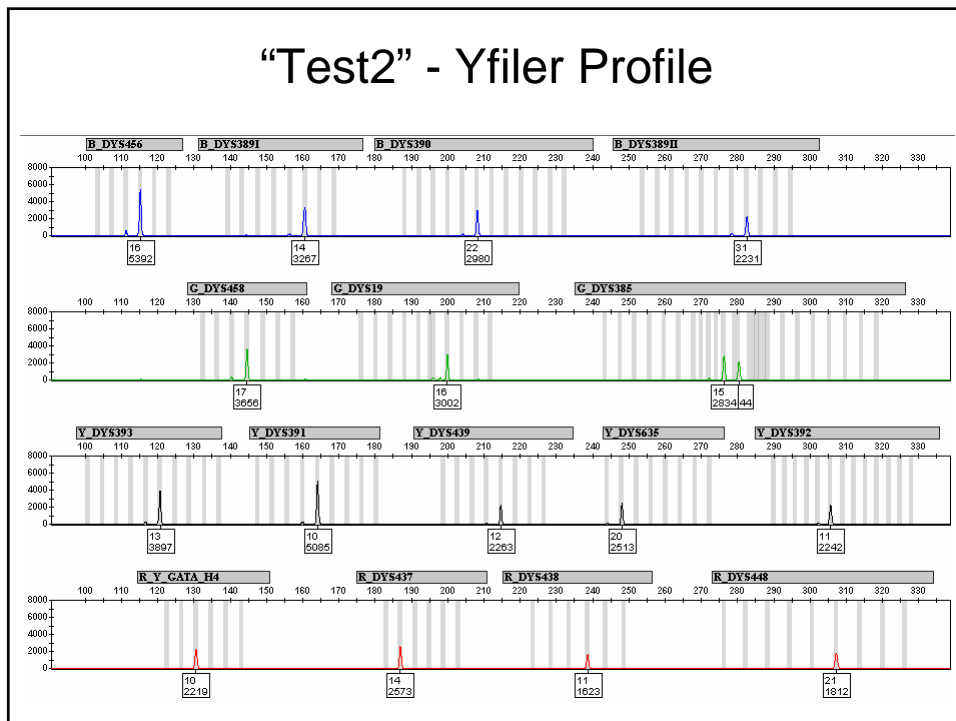
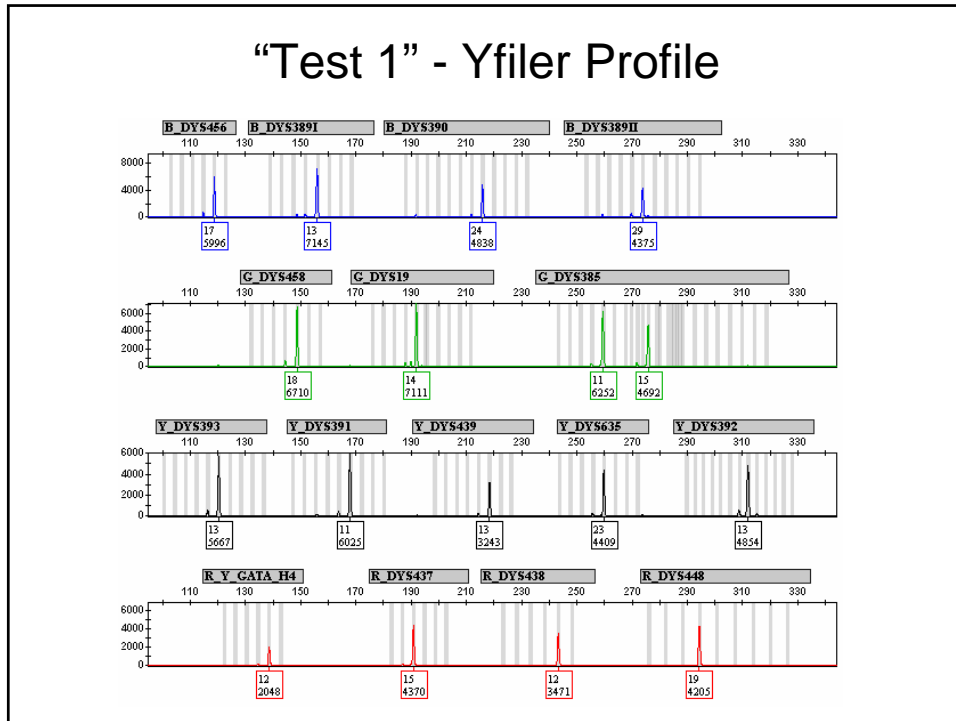
### Available Y-STR Loci, Kits and Databases

<u>Loci</u>	<u>Grouping (# Loci)</u>	<u>Available Data</u>
DYS19 DYS389I DYS389II DYS390 DYS391 DYS392 DYS393 DYS385 a/b	Minimal Haplotype (9)	<a href="http://www.YHRD.org">http://www.YHRD.org</a> <b>58,775 haplotypes</b> (499 populations from around the world) NIJ-funded <b>US Database at UCF:</b> <b>13,906 haplotypes</b> <a href="http://www.usystrdatabase.org/">http://www.usystrdatabase.org/</a>
DYS438 DYS439	SWGDM Core (11)	<a href="http://www.YHRD.org">http://www.YHRD.org</a> 29,518 haplotypes
DYS437	PowerPlex Y (12)	Promega website: 4004 haplotypes
DYS448 DYS456 DYS458 DYS635 GATA-H4	Yfiler (17)	Applied Biosystems website: <b>3561 haplotypes</b>

**~400 additional Y-STRs currently known**  
 Hanson & Ballantyne, *Legal Med* 2006;8(2):110-20







## Search Results with a Single Y-STR Locus

### Search Result with 1 locus (DYS456 allele 17)

Ancestry	# of Haplotypes	Number of Haplotypes (with Selected Alleles)	Frequency	Frequency Upper Bound (95%)
African American	2138	162	0.075772	0.086989
Asian	596	48	0.080537	0.102384
Caucasian	2472	331	0.133900	0.147324
Hispanic	1106	119	0.107595	0.125857
Native American	444	46	0.103604	0.131950
Total	6756	706	0.104499	0.111794

~1 in 9

### Search Result with 1 locus (DYS456 allele 16)

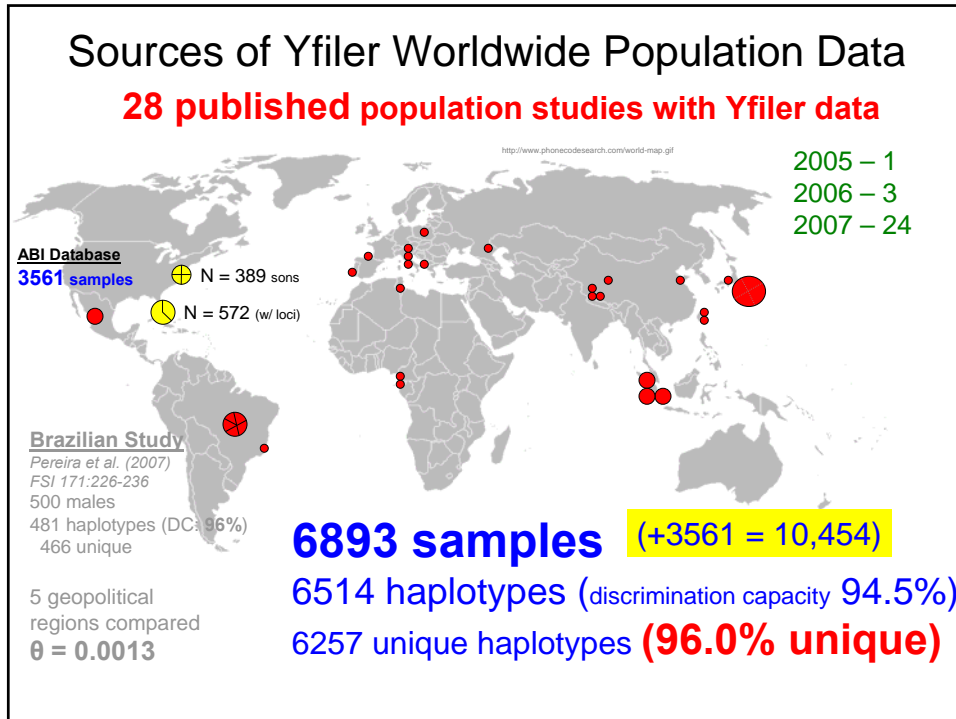
Ancestry	# of Haplotypes	Number of Haplotypes (with Selected Alleles)	Frequency	Frequency Upper Bound (95%)
African American	2138	553	0.258653	0.277214
Asian	596	106	0.177852	0.208552
Caucasian	2472	787	0.318366	0.336729
Hispanic	1106	323	0.292043	0.318841
Native American	444	97	0.218468	0.256903
Total	6756	1866	0.276198	0.286860

~1 in 3

## Y-STR Haplotype Search Results

	YHRD Search				US YSTR Search	
	17 Yfiler	11 SWGDAM	9 MHL	7 Yfiler <200bp	17 Yfiler	7 Yfiler <200bp
test1	0 of 3657	17 of 29,518	295 of 56,987	5 of 3657	0 of 4163	11 of 6601
test2	0 of 3657	2 of 29,518	3 of 56,987	1 of 3657	0 of 4163	6 of 6601
MCT	4 of 3657	243 of 29,518	1114 of 56,987	36 of 3657	9 of 4163	93 of 6601

~95% of worldwide Yfiler profiles (N=10,454) are unique  
 More loci are helpful in reducing "matches" (MHL – SWGDAM – Yfiler)



### Various Theta Values with a Partial Profile

$$f = \theta + (1 - \theta)p = p + \theta(1 - p)$$

$$p = \frac{x}{n}$$

**For  $p < \theta$ ,  $\theta$  bounds the equation**

Counting Method  
 where p = the frequency of a specific haplotype based on count (x) of haplotype in a database of size n

<u>n</u>	<u>x</u>	<u>p</u>	<u>+95%CI</u>	<u>theta calc</u>	<u>theta</u>
6601	11	0.001666	<b>0.00265</b>	<b>0.101500</b>	0.1
6601	11	0.001666	<b>0.00265</b>	<b>0.011650</b>	0.01
6601	11	0.001666	<b>0.00265</b>	<b>0.002964</b>	0.0013
6601	11	0.001666	<b>0.00265</b>	<b>0.001766</b>	0.0001

## Why a Y-STR theta correction is not needed (nor should be implemented)

- Full 17-locus profiles are unique ~95% of the time; partial profiles will have a higher degree of matches making a small theta irrelevant in many cases
- **No one else is doing it** (and no consensus in the community that it is required or necessary)
  - counting method is sufficiently conservative
- Would be too complicated to accurately employ on a routine basis – how would the “appropriate” population of interest be determined?

## Y-STR Expert Panels on Which I Have Served

- **ISFG DNA Commission**
  - Met in Nov 2004 in Berlin at Forensic Y Users Group
  - Drafted document in 2005 via email (nomenclature focus)
  - Published recommendations in 2006 in FSI & IJLM
- **SWGAM Y-STR Subcommittee**
  - Initiated in July 2002; recommended loci in Jan 2003
  - Completed interpretation guidelines in Jan 2008
  - Guidelines to be published in FSC in Jan 2009



## NIST Y-Chromosome Publications and Other Useful References

Handout for VA DFS SAB Meeting (August 5, 2008)

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## ISFG Recommendations

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## Databases

- STRBase Listing of Y-STR Databases: [http://www.cstl.nist.gov/biotech/strbase/y\\_strs.htm](http://www.cstl.nist.gov/biotech/strbase/y_strs.htm)
- Y-Chromosome Haplotype Reference Database (YHRD): <http://www.yhrd.org>
- U.S. Y-STR Database: <http://www.usystrdatabase.org/>
- Genealogy Y-STR Database: <http://www.ysearch.org/>
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## Population Variation and Data Interpretation

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