


Y-Chromosome and Mitochondrial DNA Analysis

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**The Human Y-Chromosome:  
Casework Examples  
and Resources**

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NEAFS 2006 Workshop  
Rye Brook, NY  
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Northeastern Association  
of  
Forensic Scientists

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Summary of 2006 CODIS Survey Questions  
Regarding Y-STRs **171 labs**

Questions #45a & #45b

- **Is your lab using or validating Y-STRs?**
  - **51 Yes** (30%)  
28 Yfiler, 15 PowerPlex Y, some both kits
  - 114 No
  - 6 no response

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Summary of 2006 CODIS Survey Questions  
Regarding Y-STRs **171 labs**

Question #50

- Y-STR data can be entered in CODIS similar to entering the current STR loci in CODIS. **Do you think CODIS should include Y-STR loci in Popstats calculations?**
  - Yes – 116 (68%)
  - No – 18
  - No response – 37

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# Casework with Y-STRs

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- ### Scenarios Where Y-STRs Can Aid Forensic Casework
- Sexual assaults by vasectomized or azoospermic males (no sperm left behind for differential extraction)
  - Extending length of time after assault for recovery of perpetrator's DNA profile (greater than 48 hours)
  - Fingernail scrapings from sexual assault victims
  - Male-male mixtures
  - Other bodily fluid mixtures (blood-blood, skin-saliva)
  - Gang rape situation to include or exclude potential contributors

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### Some Reported Casework Examples

*J.M. Butler (2005) Forensic DNA Typing, 2<sup>nd</sup> Edition; Table 9.7*

Kit/Loci Used	Reference	Comments
In-house assay with DYS19, DYS390, DYS389II	Prinz et al. (2001) <i>Forensic Sci. Int.</i> 120: 177-188	In one year at the New York City Office of the Chief Medical Examiner, Y-STR testing was performed in more than 500 cases with over 1000 evidence and reference samples examined. A full or partial profile was obtained on 81% of all tested evidence samples (740 worked/915 samples tested). Mixtures of at least two males were observed in 97 instances. In male-female mixtures of up to 1:4000, the male component could be clearly detected.
In-house assay with 9 Y-STR loci amplified in 3 PCR reactions	Dekairelle and Hoste (2001) <i>Forensic Sci. Int.</i> 118: 122-125	Y-STR typing was attempted on 166 semen traces from 89 cases that failed to yield a detectable male autosomal profile following differential extraction. About half of the cases had sufficient DNA to produce a Y-STR profile.
In-house assay with DYS393, DYS389II	Sibile et al. (2002) <i>Forensic Sci. Int.</i> 125: 212-216	Y-STR results could still be obtained more than 48 hours after the sexual assault in 30% of the cases examined. In 104 swabs collected with no evidence of sperm, Y-STRs could be detected in ~25% of the samples tested.
In-house assay with DYS19, DYS390, DYS389II	Prinz (2003) <i>Forensic Sci. Rev.</i> 15: 189-196	Six case studies are reviewed along with advantages and disadvantages of Y-STR testing in each case: (1) different semen donors on vaginal swab and underwear; (2) possible oligospermic perpetrator gave a nice Y-STR profile but failed to have a "male" fraction with differential extraction; (3) oral intercourse with no autosomal results—not possible to enrich male cell fraction with differential extraction in cases involving saliva; (4) presence of multiple semen donors created a complex autosomal mixture that could be sorted out with Y-STR results; (5) sperm cell fraction lacked amelogenin Y-specific peak due to known deletion—Y-STR results confirmed that the sperm cell fraction DNA was of male origin; and (6) Y-STR testing was used to rapidly screen 18 semen stains for comparison to 5 suspects and thus save the time of performing the differential extraction.
Y-PLEX 6 and Y-PLEX 5 kits	Sinha (2003) <i>Forensic Sci. Rev.</i> 15: 197-201	Five cases are reviewed: (1) criminal paternity case with a male fetus where the alleged father could not be excluded as the biological father; (2) autosomal STR test resulted in an uninterpretable mixture—suspect was excluded at 3 of the 7 Y-STR loci tested; (3) Y-PLEX 6 STR profile matched suspect with sweat stains on cloth found at crime scene; (4) fingernail cuttings from a victim matched a suspect at 11 Y-STR loci while another suspect was excluded at 2 loci; (5) semen positive stain with no sperm cells produced a Y-PLEX 6 profile consistent with the male suspect.
Y-PLEX 6 and Y-PLEX 5 kits	Sinha et al. (2004) <i>J. Forensic Sci.</i> 49: 691-700	Seven cases are reviewed (some are the same as Sinha 2003) and a list of cases where Y-STR results have been accepted in U.S. courts is provided.

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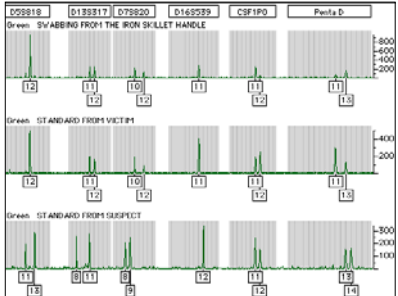
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Y-STR Analysis at The Missouri State Highway Patrol Crime Lab  
 CODIS Conference October 23, 2006  
 Brian K. Hoey

Our most interesting Y case to date




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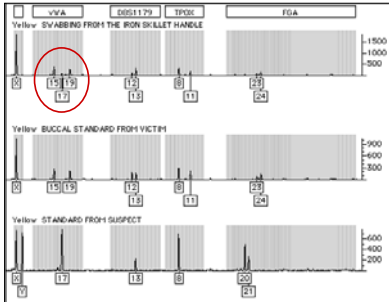
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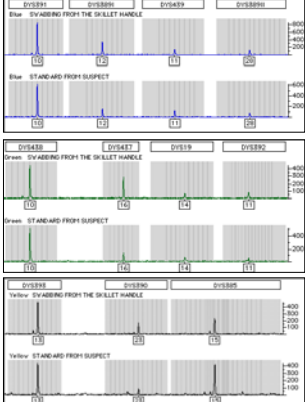
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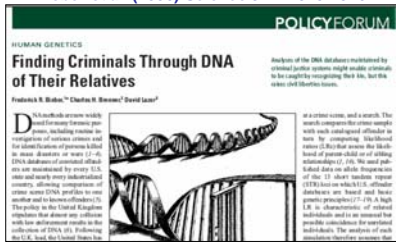
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### Use of Y-STRs to Aid Familial Searching

Bieber et al. (2006) *Science* 312: 1315-1316



- "...a seven-locus Y-STR haplotype analysis on the crime scene and the list of database leads would eliminate 99% of those not related by male lineage..."

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## Resources for Further Information

[http://www.cstl.nist.gov/biotech/strbase/y\\_strs.htm](http://www.cstl.nist.gov/biotech/strbase/y_strs.htm)

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### Y-Chromosome Publications from NIST (1)

pdf files available at <http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

- Butler, J.M., Schoske, R., Vallone, P.M., Kline, M.C., Redd, A.J., Hammer, M.F. (2002) A novel multiplex for simultaneous amplification of 20 Y chromosome STR markers. *Forensic Sci. Int.* 129: 10-24.
- Redd, A.J., Agellon, A.B., Kearney, V.A., Karalet, T., de Knijff, P., Park, H., Butler, J.M., Hammer, M.F. (2002) Forensic value of fourteen novel STRs on the human Y chromosome. *Forensic Sci. Int.* 130: 97-111.
- Butler, J.M. (2003) Recent developments in Y-short tandem repeat and Y-single nucleotide polymorphism analysis. *Forensic Sci. Rev.* 15:91-111.
- Schoske, R., Vallone, P.M., Ruitberg, C.M., Butler, J.M. (2003) Multiplex PCR design strategy used for the simultaneous amplification of 10 Y chromosome short tandem repeat (STR) loci. *Anal. Bioanal. Chem.*, 375: 333-343.
- Butler, J.M., Schoske, R., Vallone, P.M. Highly multiplexed assays for measuring polymorphisms on the Y-chromosome. (2003) *Progress in Forensic Genetics 9* (Brinkmann, B. and Carracedo, A., eds.), Elsevier Science: Amsterdam, The Netherlands, International Congress Series 1239, pp. 301-305.

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**Y-Chromosome Publications from NIST (2)**

pdf files available at <http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

- Schoske, R. (2003) The design, optimization and testing of Y chromosome short tandem repeat megaplexes. PhD dissertation, American University, 270 pp.
- Schoske, R., Vallone, P.M., Kline, M.C., Redman, J.W., Butler, J.M. (2004) High-throughput Y-STR typing of U.S. populations with 27 regions of the Y chromosome using two multiplex PCR assays, *Forensic Sci. Int.* 139: 107-121.
- Vallone, P.M. and Butler, J.M. (2004) Multiplexed assays for evaluation of Y-SNP markers in U.S. populations. *Progress in Forensic Genetics 10*, Elsevier Science: Amsterdam, The Netherlands, International Congress Series 1261, 85-87.
- Butler, J.M. and Schoske, R. (2004) Forensic value of the multi-copy Y-STR marker DYS464. *Progress in Forensic Genetics 10*, Elsevier Science: Amsterdam, The Netherlands, International Congress Series 1261, 278-280.
- Butler, J.M. and Schoske, R. (2004) Duplication of DYS19 flanking regions in other parts of the Y chromosome. *Int. J. Legal Med.*, 118: 178-183.

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**Y-Chromosome Publications from NIST (3)**

pdf files available at <http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

- Vallone, P.M. and Butler, J.M. (2004) Y-SNP typing of U.S. African American and Caucasian samples using allele-specific hybridization and primer extension. *J. Forensic Sci.* 49(4): 723-732.
- Butler, J.M. (2005) Constructing STR multiplex assays. *Methods in Molecular Biology: Forensic DNA Typing Protocols* (Carracedo, A., ed.), Humana Press: Totowa, New Jersey, 297: 53-66.
- Vallone, P.M., Fahr, K., Kostrzewa, M. (2005) Genotyping SNPs using a UV-photocleavable oligonucleotide in MALDI-TOF MS. *Methods in Molecular Biology: Forensic DNA Typing Protocols* (Carracedo, A., ed.), Humana Press: Totowa, New Jersey, 297: 169-178.
- Butler, J.M., Decker, A.E., Kline, M.C., Vallone, P.M. (2005) Chromosomal duplications along the Y-chromosome and their potential impact on Y-STR interpretation. *J. Forensic Sci.* 50(4): 853-859.

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**Y-Chromosome Publications from NIST (4)**

pdf files available at <http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

- Butler, J.M. and Schoske, R. (2005) U.S. population data for the multi-copy Y-STR locus DYS464. *J. Forensic Sci.* 50(4): 975-977.
- Butler, J.M., Appleby, J.E., Duewer, D.L. (2005) Locus-specific brackets for reliable typing of Y-chromosome short tandem repeat markers. *Electrophoresis* 26: 2583-2590.
- Butler, J.M., Decker, A.E., Vallone, P.M., Kline, M.C. (2006) Allele frequencies for 27 Y-STR Loci with U.S. Caucasian, African American, and Hispanic Samples. *Forensic Sci. Int.* 156: 250-260.
- Gusmão L, Butler JM, Carracedo A, Gill P, Kayser M, Mayr WR, Morling N, Prinz M, Roewer L, Schneider PM, Tyler-Smith C (2006) DNA Commission of the International Society of Forensic Genetics (ISFG): An update of the recommendations on the use of Y-STRs in forensic analysis. *Forensic Sci. Int.* 157: 187-197.

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### Y-Chromosome Publications from NIST (5)

pdf files available at <http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm>

- Gusmão, L., Butler, J.M., Carracedo, A., Gill, P., Kayser, M., Mayr, W.R., Morling, N., Prinz, M., Roewer, L., Tyler-Smith, C., Schneider, P.M. (2006) DNA Commission of the International Society of Forensic Genetics (ISFG): An update of the recommendations on the use of Y-STRs in forensic analysis. *Int. J. Legal Med.* 120: 191-200.
- Mulero, J.J., Budowle, B., Butler, J.M., Gusmão, L. (2006) Letter to the Editor--Nomenclature and allele repeat structure update for the Y-STR locus GATA H4. *J. Forensic Sci.* 51(3): 694.
- Decker, A.E., Vallone, P.M., Kline, M.C., Butler, J.M. (2006) Impact of additional Y-STR loci on resolving common haplotypes and closely related individuals. *FSI Genetics (in preparation)*.

**20 papers so far with 1 in preparation...**

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**Rich Schoske** (Air Force funded student working at NIST 2000-2003) – development of early Y-STR megaplexes

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