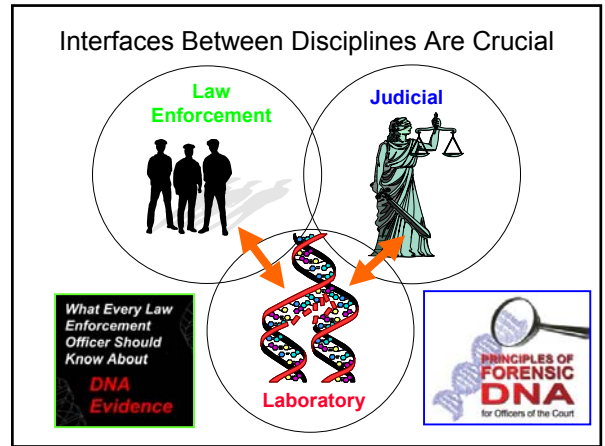


Human Identification Using DNA Analysis: Past, Present, and Future

John M. Butler

U.S. National Institute of Standards and Technology
International Symposium on the Forensic Sciences (ANZFSS 2006)
April 4, 2006 – Fremantle, Australia



Classroom to Courtroom

- We should encourage more scientist-lawyer interaction as both sides will benefit from understand the other perspective
- **DNA.gov website – Officers of the Court**
- Recent experience with the **American Prosecutors Research Institute (APRI)** (Jan 30 – Feb 1, 2006)

DNA Training for Officers of the Court

PRESIDENT'S **DNA** INITIATIVE *Advancing Justice Through DNA Technology*

- CD-ROM available from the U.S. National Institute of Justice (<http://www.ncjrs.gov>)
- On-line training available at <http://www.DNA.gov>

<http://www.dna.gov/training/otc/>

PRESIDENT'S **DNA** INITIATIVE

Principles of Forensic DNA for Officers of the Court

<ol style="list-style-type: none"> 1. Introduction 2. Biology of DNA 3. Practical Issues Specific to DNA Evidence 4. Forensic DNA Laboratory 5. Assuring Quality in DNA Testing 6. Understanding a Forensic DNA Lab Report 7. Statistics and Population Genetics 	<ol style="list-style-type: none"> 8. Mitochondrial DNA & Y-STR Analysis 9. Forensic DNA Databases 10. Collection of DNA Evidence 11. Pretrial DNA Evidence Issues 12. Victim Issues 13. Trial Presentation 14. Postconviction DNA Cases 15. Emerging Trends
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<http://www.dna.gov/training/otc/>

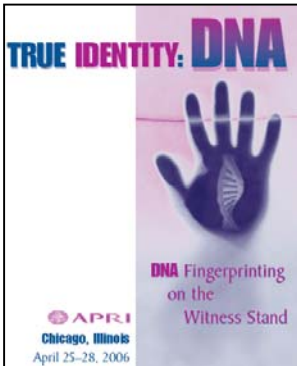
DNA Forensics Program

APRI Training Curriculum

http://www.ndaa-apri.org/apri/programs/dna/dna_home.html

- Case scenarios developed to train prosecutors in legal and scientific aspects of DNA testing
- **Example data and reports are being created**
- More scientist-lawyer interactions are needed
- **Training resources available on STRBase**

Upcoming Prosecutor Training Course



TRUE IDENTITY: DNA

DNA Fingerprinting on the Witness Stand

APRI
Chicago, Illinois
April 25-28, 2006

Topics to Include:

- Understanding Lab Reports
- Identification, Collection and Submission of DNA Evidence
- Nuclear DNA
- Mitochondrial DNA Analysis
- Y-STR Analysis
- Understanding Statistics
- Discovery
- Ethical Issues
- Defense Expert Witnesses
- Third Party Defense Motions
- Admissibility of DNA Evidence and Expert Scientific Testimony
- Preparation of the DNA Expert and Prosecutor for Trial

The National Advocacy Center

http://www.ndaa-apri.org/education/nac_index.html



<http://www.ndaa-apri.org/images/nac.jpg>



<http://www.sandlapper.org/advpic1.jpg>

The National Advocacy Center, situated on the University of South Carolina campus in Columbia, is a joint venture of the National District Attorneys Association (NDA) and the U.S. Department of Justice.



<http://www.sandlapper.org/advpic3.jpg>

Information Resources for Defense Attorneys

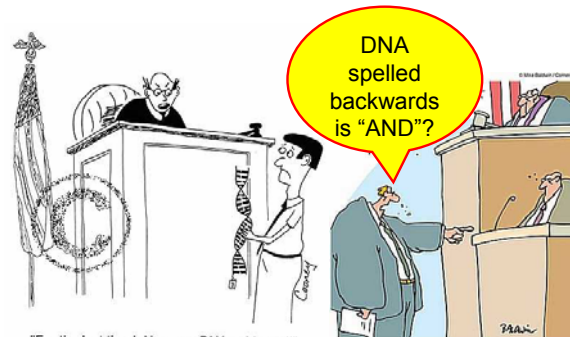
http://www.nlada.org/Defender/forensics/for_lib/Index/DNA/exhibits/index_html



Forensic Library

- DNA
- DNA Weblinks
- DNA Model Pleadings
- DNA Research (Scientific & Legal)
- DNA Government Expert Materials
- DNA Defense Expert Materials
- DNA Database Issues
- DNA Civil Rights Issues
- DNA Court Opinions
- DNA Training Materials
- DNA Misidentifications-Important Cases
- DNA Lab Procedures (QA, QC, SOPs, audits, etc.)
- DNA Lab Analysis (Fraud, Proficiency)
- DNA Lab Testing Kits and Software
- Y-STR Testing
- Mitochondrial DNA

What We Want to Avoid...




"For the last time! No more DNA evidence!"

He was good. Real good.


Impact of Forensic DNA Testing

Guilt




Colin Pitchfork


Innocence




Kirk Bloodworth



Josiah Sutton



Gary Coleman



The Innocence Project

<http://www.innocenceproject.org>



Calvin Willis
Year of Incident: 1981
Jurisdiction: Louisiana
Sentence: Life
Year of Exoneration: 2003
Sentence Served: 22 years

ROBERT CLARK
Exonerated in Georgia

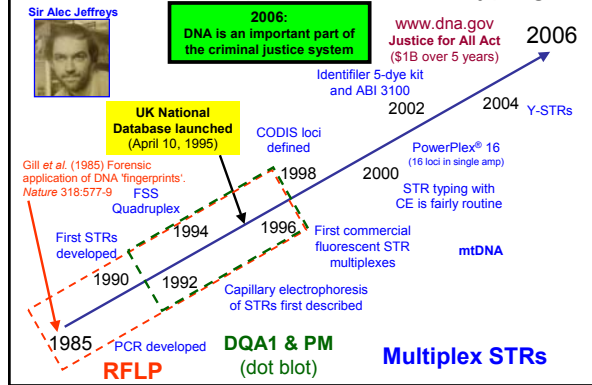
March 29, 2006
176 EXONERATED

Applications of Forensic DNA

- Forensic cases -- matching suspect with evidence
- Paternity testing -- identifying father
- Missing persons investigations
- Military DNA "dog tag"
- Convicted felon DNA databases
- Mass disasters -- putting pieces back together
- Historical investigations

Involves generation of DNA profiles usually with the same core STR (short tandem repeat) markers and then MATCHING TO REFERENCE SAMPLE

Historical Perspective on DNA Typing



Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition*, Box 1.3, p. 9

August 17, 1998
FBI Report on Analysis of Stain on Monica Lewinsky's Blue Dress



DNA History

Examiner Name: [REDACTED] Date: 08/17/98
 Unit: DNA Analysis 1 Phone No.: 202-324-4109
 File No.: 290-OIC-LR-35063 Lab No.: 980730902 & B0
 980603109 & B0

Results of Examination:
 Deoxyribonucleic acid (DNA) profiles for the genetic loci D2S44, D1S27, D1S1, D4S139, D10S24, D5S810 and D7S845 were developed from HaeIII digested high molecular weight DNA extracted from specimen K39 and Q3243-1 (a semen stain removed from specimen Q3243). Based on the results of these seven genetic loci, specimen K39 (CLINTON) is the source of the DNA obtained from specimen Q3243-1, to a reasonable degree of scientific certainty.

No DNA-RFLP examinations were conducted on specimen Q3243-2 (a semen stain removed from specimen Q3243).



RFLP - 1, 400, 000, 000, 000
 STR - 7, 8, 21, 400, 000, 000
 STR - 3, 140, 000, 000, 000
 STR - 973, 000, 000, 000

<http://www.law.umkc.edu/faculty/projects/frlabs/clinton/lewinskydress.html>



Tomb of the Unknown Soldier

DNA History

- Armed Forces DNA Identification Laboratory (AFDIL) (Rockville, MD)
- In June 1998 AFDIL identified Michael J. Blassie as the Vietnam Unknown in the Tomb of the Unknown Soldier (located in Arlington National Cemetery)
- There will be no more "unknown" soldiers.

Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition*, Box 10.1, pp. 250-251

Identifying Victims of Mass Disasters

Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition*, Chapter 24

POLICY FORUM

EPIDEMIOLOGY

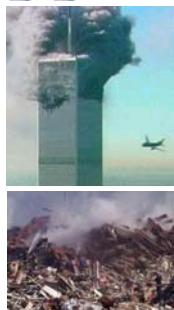
DNA Identifications After the 9/11 World Trade Center Attack

Leslie G. Breckenrider, Jean E. Bailey-Wilson, Jack Ballantyne, Howard Baum, Frederick K. Barber, Charles Brainerd, Bruce Budroff, John M. Butler, George Carroly, P. Michael Conneally, Barry Dorman, Arthur Eisenberg, Lisa Forman, Kenneth H. Kidd, Brent Leslie, Steven Mergende, Thomas J. Parsons, Elizabeth Pugh, Robert Shaler, Stephen T. Sherry, Amanda Sasser, Anna Walsh

Science (2005) 310: 1122-1123

Feb 23, 2005: Announcement that DNA identification efforts have been exhausted. Only 1,585 victims (58%) were identified from the 2,749 killed at the WTC site.

DNA History



The Present

Short Tandem Repeat (STR) Typing

National Commission on the Future of DNA Evidence

•Report published in Nov 2000

•Asked to estimate where DNA testing would be 2, 5, and 10 years into the future

Conclusions

STR typing is here to stay for a few years because of DNA databases that have grown to contain millions of profiles

<http://www.ojp.usdoj.gov/nij/pubs-sum/183697.htm>

- ### Advantages for STR Markers

 - Small product sizes are generally compatible with degraded DNA and PCR enables recovery of information from small amounts of material
 - Multiplex amplification with fluorescence detection enables high power of discrimination in a single test
 - Commercially available in an easy to use kit format
 - Uniform set of core STR loci provide capability for national and international sharing of criminal DNA profiles

National DNA Index System (NDIS)

FBI Laboratory

CODIS
COMBINED DNA INDEX SYSTEM

<http://www.fbi.gov/hq/lab/codis/index1.htm>

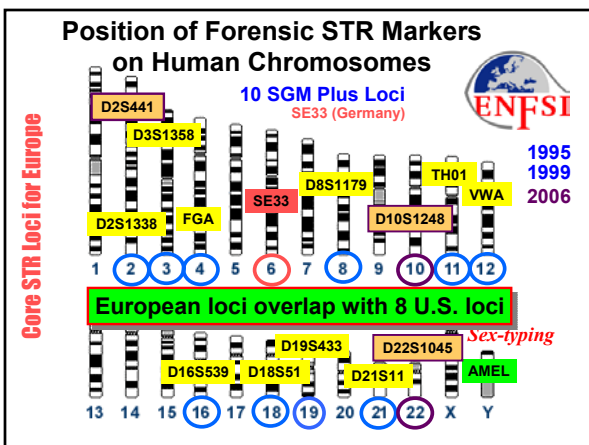
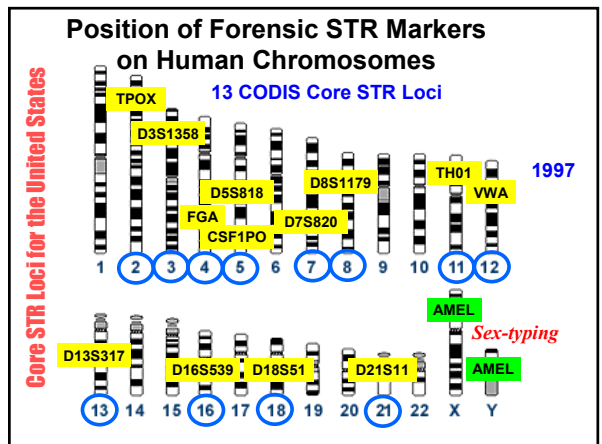
Combined DNA Index System (CODIS)

Launched in October 1998 and now links all 50 states
Used for linking serial crimes and unsolved cases with repeat offenders
Convicted offender and forensic case samples along with a missing persons index

Requires 13 core STR markers

>32,000 investigations aided nationwide as of Feb 2006

Contains more than 3.0 million DNA profiles



Review Article on Core STR Loci

J Forensic Sci, March 2006, Vol. 51, No. 2
 doi:10.1111/j.1556-4029.2006.00946.x
 Available online at: www.blackwell-synergy.com

John M. Butler,¹ Ph.D.

Genetics and Genomics of Core Short Tandem Repeat Loci Used in Human Identity Testing

Journal of Forensic Sciences 2006, 51(2): 253-265

- Reviews STR kits, genomic locations, mutation rates, potential genetic linkage, and known variant alleles for autosomal STR and Y-STR loci
- Covers characteristics of 18 autosomal loci (13 core CODIS loci, D2, D19, Penta D, Penta E, SE33) and 11 SWGDAM-recommended Y-STR loci

Locus Name	Chromosomal Location	Physical Position*
CSF1PO	5q33.1 cfms proto-oncogene, 6 th Intron	Chr 5 149.484 Mb
FGA	4q31.3 alpha fibrinogen, 3 rd Intron	Chr 4 156.086 Mb
TH01	11p15.5 tyrosine hydroxylase, 1 st Intron	Chr 11 2.156 Mb
TPOX	2p25.3 thyroid peroxidase, 10 th Intron	Chr 2 1.436 Mb
VWA	12p13.31 von Willebrand Factor, 40 th Intron	Chr 12 19.826 Mb
D3S1358	3p21.31	Chr 3 45.543 Mb
D5S818	5q23.2	Chr 5 123.187 Mb
D7S820	7q21.11	Chr 7 83.401 Mb
D8S1179	8q24.13	Chr 8 125.863 Mb
D13S317	13q31.1	Chr 13 80.52 Mb
D16S539	16q24.1	Chr 16 86.168 Mb
D18S51	18q21.33	Chr 18 59.098 Mb
D21S11	21q21.1	Chr 21 19.476 Mb

Position of Each CODIS STR Locus in Human Genome

Review article on core STR loci genetics and genomics published in March 2006

Butler, J.M. (2006) Genetics and genomics of core STR loci used in human identity testing. *J. Forensic Sci.* 51(2): 253-265

From Table 5.2, *Forensic DNA Typing*, 2nd Edition, p. 96 (J.M. Butler, 2005)

STRBase

Short Tandem Repeat DNA Internet Database

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

General Information

- Intro to STRs (downloadable PowerPoint)
- STR Fact Sheets
- Sequence Information
- Multiplex STR Kits
- Variant Allele Reports
- Training Slides

Forensic Interest Data

- FBI CODIS Core Loci
- DAB Standards
- NIST SRMs 2391
- Published PCR Primers
- Y-Chromosome STRs
- Population Data
- Validation Studies
- miniSTRs

Supplemental Info

Reference List >2500

- Technology Review
- Addresses for Scientists
- Links to Other Web Sites
- DNA Quantitation
- mtDNA
- New STRs
- Forensic SNPs

New information is added regularly...

Status of Genetic Marker Systems Used in Forensic DNA Testing

- **STRs** – widely used in national databases today
- **miniSTRs** – now in research; WTC use; kit under development (more detail in today's Biological Criminalistics section)
- **mtDNA** – used in specialty labs for highly degraded specimens
- **Y-STRs** – growing use due to kits now available
- **SNPs** – research; likely to be limited in use (more detail in today's Biological Criminalistics section)

The Future

- More Robotics**
- Expert Systems**
- Animal & Plant DNA**
- Physical Characteristics**
- Ethnicity Estimation**

A "Crystal Ball" to the Future

Improvements in Forensic DNA Analysis

- **Biology**
 - Improved DNA extraction with automation
 - New capabilities for recovery of information from degraded DNA samples (e.g., miniSTRs)
- **Technology**
 - Parallel processing of DNA with capillary arrays
 - Expert systems for automated data interpretation
- **Genetics**
 - Ethnicity estimations (with STRs and/or SNPs)
 - Larger Y-STR and mtDNA population databases

Effective Training is Needed in All Areas

Training Materials Available or Planned

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

- DNA Basics
- Validation
- **STR Analysis and Capillary Electrophoresis**
- Y-Chromosome Analysis
- Mitochondrial DNA Analysis
- Expert Systems
- Low-Copy Number (LCN) DNA Testing
- Statistics
- Mixture Interpretation

Training Materials and Review Articles

- Workshops on STRs and CE (ABI 310/3100)
 - Taught with Bruce McCord (Florida Int. Univ.)
 - NEAFS (Sept 29-30, 2004)
 - U. Albany DNA Academy (June 13-14, 2005)
 - **AAFS Feb 2006 Workshop #6 (February 20, 2006)**
- Other Workshops
 - **Validation** (August 24-26, 2005)
 - **mtDNA Analysis** (March 13-15, 2006)
 - **Expert Systems** (March 27, 2006)
- PowerPoint Slides from *Forensic DNA Typing*, 2nd Edition
 - >150 slides available now (~1,000 planned) for download
 - <http://www.cstl.nist.gov/biotech/strbase/FDT2e.htm>
- Review Articles
 - ABI 310 and 3100 chemistry – *Electrophoresis* 2004, 25, 1397-1412
 - **Core STR Loci – *J. Forensic Sci.* 2006, 51(2): 253-265**

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

AAFS Workshop #6 (Feb 2006, Seattle) Advanced Topics in STR DNA Analysis Instructors: John Butler and Bruce McCord

- **STR Biology, Markers, and Methods**
- **Capillary Electrophoresis Instrumentation: Theory and Application**
- **Validation Aspects to Consider in Bringing a New STR Kit “On-line”**
- **CE Troubleshooting**
- **STR Mixture Interpretation**
- **DNA Quantitation with Real-Time qPCR**
- **Low-copy Number Issues**
- **Y-STRs and mtDNA**

~500 PowerPoint Slides Available

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

NIST History and Mission

- National Institute of Standards and Technology (NIST) was created in 1901 as the National Bureau of Standards (NBS). The name was changed to NIST in 1988.
- NIST is **part of the U.S. Department of Commerce** with a mission to **develop and promote measurement, standards, and technology to enhance productivity, facilitate trade, and improve the quality of life.**
- NIST supplies over 1,300 Standard Reference Materials (SRMs) for industry, academia, and government use in **calibration of measurements.**



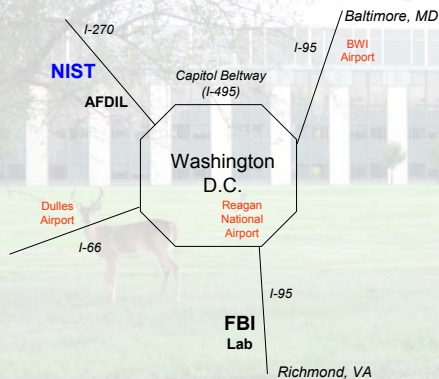
\$532 for 3 jars



DNA typing standard

- **NIST defines time for the U.S.**

Location of NIST (near Washington, DC)



NIST Gaithersburg Campus

Administration (Building 101)

Located in Gaithersburg, Maryland, on approximately 234 hectares (578 acres) just off Interstate 270 about **25 miles northwest of Washington, D.C.**

<http://www.nist.gov>

~2,500 staff

Advanced Chemical Sciences Laboratory (Building 227)

National Institute of Justice
The Research, Development, and Evaluation Agency of the U.S. Department of Justice

Current Areas of NIST Effort with Forensic DNA

- Standards**
 - Standard Reference Materials
 - Standard Information Resources (STRBase website)
 - Interlaboratory Studies
- Technology**
 - Research programs in SNPs, miniSTRs, Y-STRs, mtDNA, qPCR
 - Assay and software development
- Training Materials**
 - Review articles and workshops on STRs, CE, validation
 - PowerPoint and pdf files available for download

