Classroom to Courtroom

- We should encourage more scientist-lawyer interaction as both sides will benefit from understanding 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

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

Principles of Forensic DNA for Officers of the Court

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
8. Mitochondrial DNA & Y-STR Analysis
9. Forensic DNA Databases
10. Collection of DNA Evidence Issues
11. Pretrial DNA Evidence Issues
12. Victim Issues
13. Trial Presentation
14. Postconviction DNA Cases
15. Emerging Trends

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

APRI Training Curriculum

- 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

http://www.ndaa-apri.org/apri/programs/dna/dna_home.html
Upcoming Prosecutor Training Course

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

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

Information Resources for Defense Attorneys
http://www.nlada.org/Defender/forensics/for_lib/index/DNA/exhibits/index_html

Impact of Forensic DNA Testing

Guilt
Colin Pitchfork
Kirk Bloodsworth
Josiah Sutton
Gary Coleman

Innocence

What We Want to Avoid…
DNA spelled backwards is "AND"?

"For the last time! No more DNA evidence!"
He was good. Really good.

The Innocence Project
http://www.innocenceproject.org

The Innocence Project of the Benes & Selden School of Law & National Liberty Foundation helps convicts win exonerations. Since 1992, the organization has helped free more than 200 people, including Colin Pitchfork, who had been convicted of rape and murder. The Innocence Project also works with law enforcement agencies to prevent wrongful convictions.
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

- 1985: PCR developed
- 1990: RFLP
- 1992: First STRs developed
- 1994: First commercial fluorescent STR multiplexes
- 1996: CODIS loci defined
- 1998: STR typing with CE is fairly routine
- 2000: PowerPlex® 16 (10 loci in single assay)
- 2002: Identifier 5-plex kit and ABI 3100
- 2006: DNA is an important part of the criminal justice system

Identifying Victims of Mass Disasters


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.
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

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

Position of Forensic STR Markers on Human Chromosomes

Review Article on Core STR Loci

- 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

http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm
Position of Each CODIS STR Locus in Human Genome

<table>
<thead>
<tr>
<th>Locus Name</th>
<th>Chromosomal Location</th>
<th>Physical Position</th>
</tr>
</thead>
<tbody>
<tr>
<td>CSFPO</td>
<td>19p13.1</td>
<td>Chr 1: 145,654,454</td>
</tr>
<tr>
<td>HLA</td>
<td>6p21.1</td>
<td>Chr 6: 16,265,064</td>
</tr>
<tr>
<td>VWA</td>
<td>19q13.13</td>
<td>Chr 19: 11,226,273</td>
</tr>
<tr>
<td>IPG</td>
<td>1q21.3</td>
<td>Chr 1: 11,963,826</td>
</tr>
<tr>
<td>STRS</td>
<td>19p13.1</td>
<td>Chr 19: 11,494,044</td>
</tr>
<tr>
<td>STRS2</td>
<td>1q21.3</td>
<td>Chr 1: 12,234,617</td>
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<tr>
<td>STRS3</td>
<td>1q21.3</td>
<td>Chr 1: 12,23,617</td>
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<tr>
<td>STRS4</td>
<td>1q21.3</td>
<td>Chr 1: 12,65,623</td>
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<tr>
<td>STRS5</td>
<td>1q21.3</td>
<td>Chr 1: 12,90,526</td>
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<td>STRS9</td>
<td>1q21.3</td>
<td>Chr 1: 19,45,042</td>
</tr>
</tbody>
</table>

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


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

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 28-30, 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

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.

• NIST defines time for the U.S.

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

Location of NIST (near Washington, DC)

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

NIST Gaithersburg Campus

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

Genetic Markers Under Examination

New miniSTRs

CORE STR Loci

Variant allele sequencing

Y-chromosome STRs

Genetic Markers Under Examination

New miniSTRs

CORE STR Loci

Variant allele sequencing

Y-chromosome STRs

A miniSTR is a reduced size STR amplicon that enables higher recovery of information from degraded DNA samples

D18S51 Null Allele from Kuwait Samples with ABI Primers

PowerPlex 16

Identifier

mutation

Allele 18 drops out

Clayton et al. (2004) Primer binding site mutations affecting the typing of STR loci contained within the AMPFISTR SGM Plus kit, Forensic Sci Int. 139(2-3): 255-259

Initial Testing Results with Potential miniSTR Loci


We have characterized 27 new miniSTR loci

http://www.cstl.nist.gov/biotech/strbase/newSTRs.htm

http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm
The DNA Field Moves Forward…

The Past

RFLP

500 – 25,000 bp

The Present

STRs

100 - 500 bp

The Future

miniSTRs

50 - 150 bp

Acknowledgments

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- Mike Hammer and Alan Redd (U. AZ) for Y-chromosome studies
- Tom Parsons, Rebecca Just, Jodi Irwin (AFDIL) for mtDNA coding SNP work
- Sandy Calloway (Roche) for mtDNA LINEAR ARRAYS
- Bruce McColl and students (FL Int. U.) for miniSTR work
- Marilyn Raymond and Victor David (NCI-Frederick) for cat STR work
- Artie Eisenberg and John Planz (U. North Texas) for miniSTR testing on bones
- Murray Brilliant (U. AZ) for phenotype markers
- Ken Kidd (Yale U.) for SNP typing population samples

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