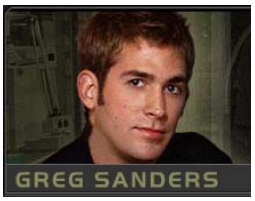
Forensic DNA:

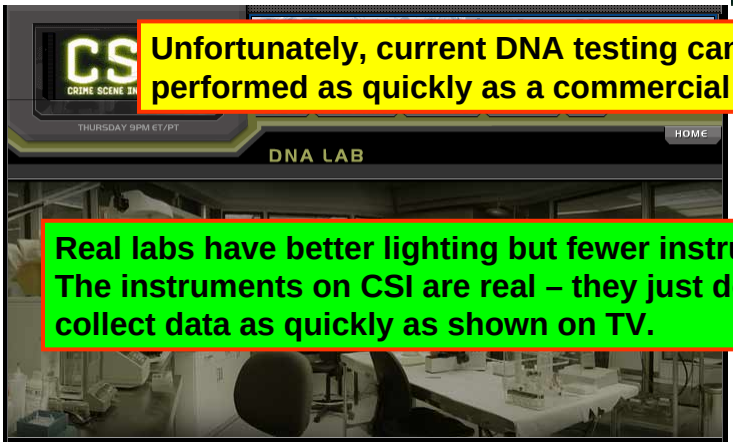
Where Have We Come From and Where Are We Going?

John M. Butler, Ph.D.
National Institute of Standards and Technology

Jersey City, New Jersey
September 27, 2008

County Prosecutors Association
of New Jersey





Unfortunately, current DNA testing cannot be performed as quickly as a commercial break…

Real labs have better lighting but fewer instruments. The instruments on CSI are real – they just do not collect data as quickly as shown on TV.

New Jersey State Police
Office of Forensic Sciences
DNA Laboratory

http://www.state.nj.us/njsp/divorg/invest/dna-lab.html

Growth and Impact of DNA Databases

http://www.fbi.gov/hq/lab/codis/stats.htm#NewJersey

New Jersey Statistics (Sept 2008)	**Total**
Offender Profiles	168,646
Forensic Samples	5,785
Investigations Aided	2,094

National DNA Index System:
>6.3 million samples
~75,000 investigations aided

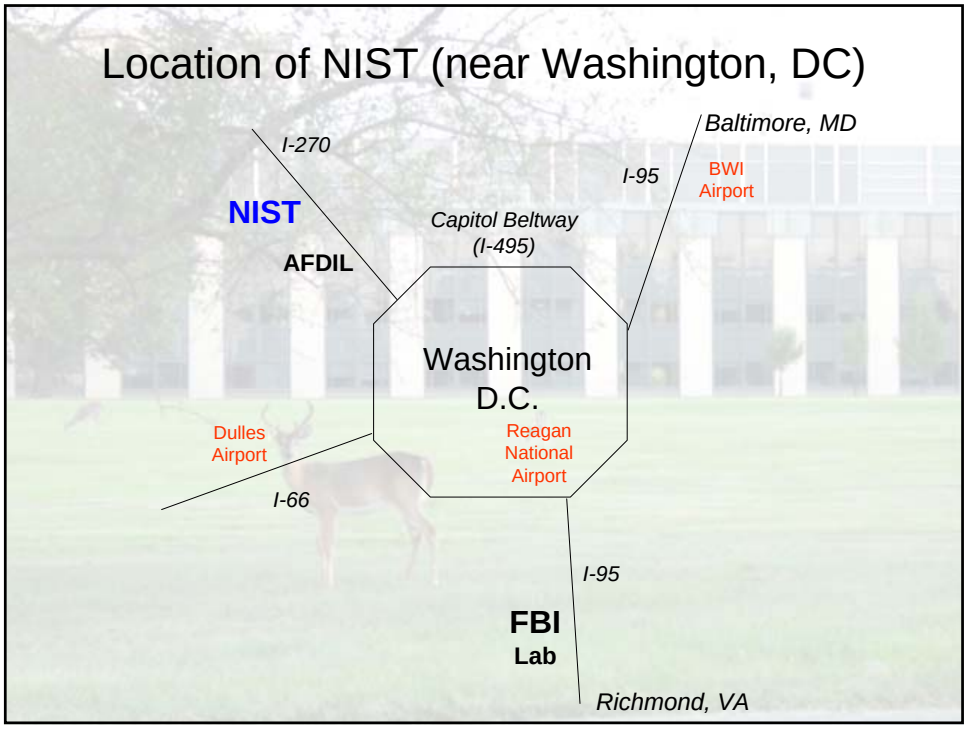
NIST Background

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.**

$573 for 3 jars

DNA typing standard


Location of NIST (near Washington, DC)
I-270
Baltimore, MD
I-95
BWI Airport
NIST
AFDIL
Capitol Beltway (I-495)
Washington D.C.
Dulles Airport
Reagan National Airport
I-66
I-95
FBI Lab
Richmond, VA


http://www.eeel.nist.gov/oles/
Office of Law Enforcement Standards
Public Safety and Security Technologies
Weapons and Protective Systems
Detection, Inspection, and Enforcement Technologies
Helping law enforcement, corrections, criminal justice, and public safety agencies ensure that the equipment they purchase and the technologies they use are safe, dependable, and effective.
Public Safety Communications Systems
Forensic Sciences
Critical Incident Technologies

Forensic Science

Computer Forensics

Computer Forensics:

Computer Forensic Tool Testing (CFFT)

- **Write Block Devices**: **20 reports**
- **Write Block Tools**: **9 reports**
- **Disk Imaging Tools**: **9 reports**
- **Deleted File Recovery**: **1 report**

www.cftt.nist.gov

Forensics Research at NIST

- Computer (digital evidence) forensics
- Ballistics
- Fingerprints
- Arson investigation
- **DNA**

SRM 2461
Standard Casing

For more information, contact:

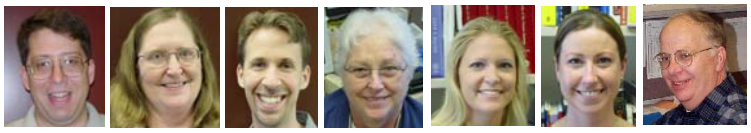
Susan Ballou
Program Manager for Forensic Sciences
susan.ballou@nist.gov
301-975-8750

Evaluating Error within Forensic Disciplines

Some NIST Goals:

- To convene a panel of experts and administer research projects to investigate sources of error in forensic pattern recognition analysis,
- To identify methods to eliminate or mitigate sources of error, and
- To evaluate various approaches to numerically quantifying error within forensic pattern recognition disciplines

NIST Human Identity Project Team



John Butler | Margaret Kline | Pete Vallone | Jan Redman | Amy Decker | Becky Hill | Dave Duewer

Publications and presentations available on STRBase:
http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm

FY 2007 Achievements:
14 publications
44 presentations
9 workshops

Since 2000:
100 publications
258 presentations
30 workshops

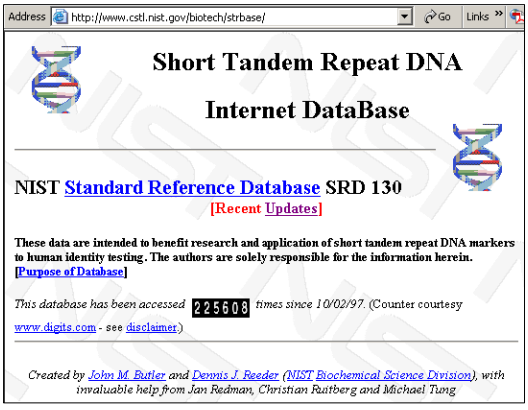
National Institute of Justice
The Research, Development, and Evaluation Agency of the U.S. Department of Justice

Current Areas of NIST Effort with Forensic DNA

- **Standards** http://www.cstl.nist.gov/biotech/strbase/
  - 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

Information Resources

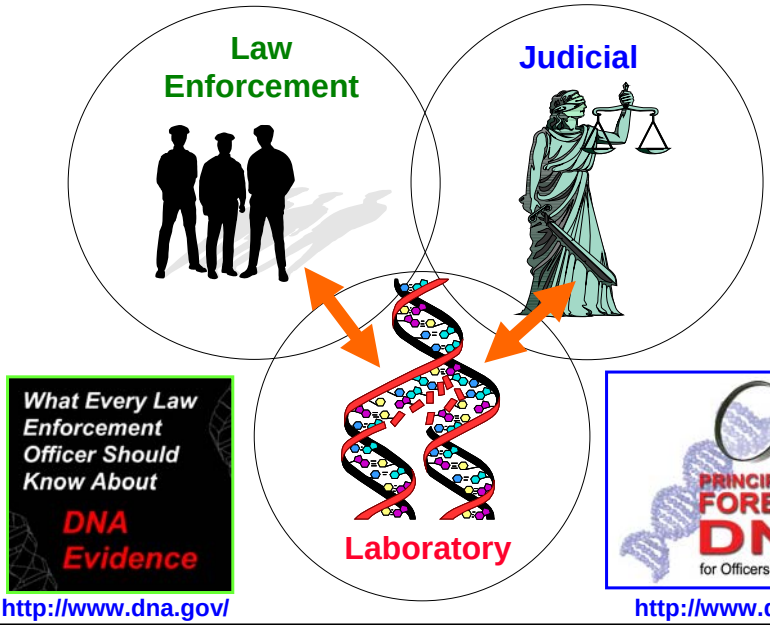
http://www.cstl.nist.gov/biotech/strbase



Includes information on:
- Core STR loci
- Validation
- STR reference list
- NIST publications
- miniSTRs
- Forensic SNPs
- Variant STR alleles
- Population data resources
- Addresses of scientists

Provides up-to-date information and has been used in court cases to support application of DNA technology

Interfaces Between Disciplines Are Crucial

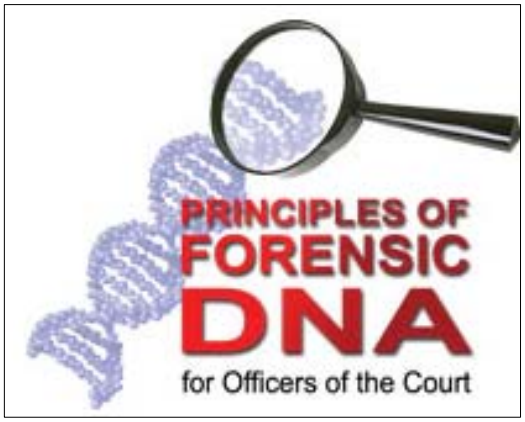


http://www.dna.gov/

http://www.dna.gov/

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/

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
11. Pretrial DNA Evidence Issues
12. Victim Issues
13. Trial Presentation
14. Postconviction DNA Cases
15. Emerging Trends

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

Information Resources for Defense Attorneys

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

Defense Lawyers and Experts are becoming more united and informed

Common Defense Attacks

Compiled from Forensic Bioinformatics website

http://www.bioforensics.com

- Contamination
- Statistical Weight of a Match
- Degradation/PCR Inhibition of "True" Perp
- Artifacts and "Complications" to DNA Profiles
- **Thresholds Set Too High (missing peaks)**
- Examiner Bias
- **Improper Mixture Interpretation**
- Meaning of a Database Hit
- Protocol Violations

Forensic Bioinformatics
7th Annual Conference
The Science of DNA Profiling: A National Expert Forum
August 15 - 17, 2008
Dayton, OH

See http://www.bioforensics.com/conference08/index.html

How Are DNA Results Obtained?

Steps Involved

- Collection
- Specimen Storage
- Extraction
- Quantitation
- Multiplex PCR
- STR Typing
- Interpretation of Results
- Database
  Storage & Searching
- Calculation of Match Probability

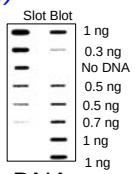
Steps in DNA Analysis

Usually 1-2 day process (a minimum of ~5 hours)

Blood Stain Buccal swab
Sample Collection & Storage

Biology

DNA Extraction



DNA Quantitation

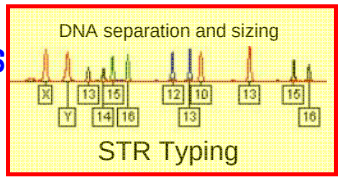
Multiplex PCR Amplification

Genetics

If a match occurs, comparison of DNA profile to population allele frequencies to generate a case report with probability of a random match to an unrelated individual

DNA Database Search

Technology



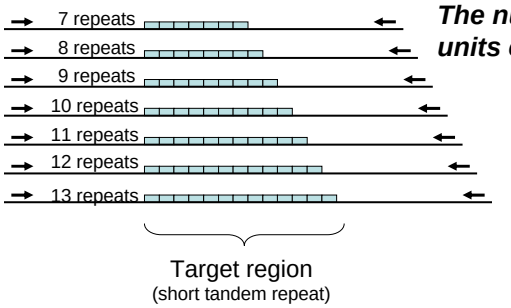
Male: 13,14-15,16-12,13-10,13-15,16
Interpretation of Results

Short Tandem Repeat (STR) Markers

An accordion-like DNA sequence that occurs between genes

TCCCAAGCTCTTCCTCTTCCCTAGATCAATACAGACAGAAGACA
GGTG**GATAGATAGATAGATAGATAGATAGATAGATAGATAGA**
TAGATAGATATCATTGAAAGACAAAACAGAGATGGATGATAGAT
ACATGCTTACAGATGCACAC

= 12 GATA repeats ("12" is all that is reported)

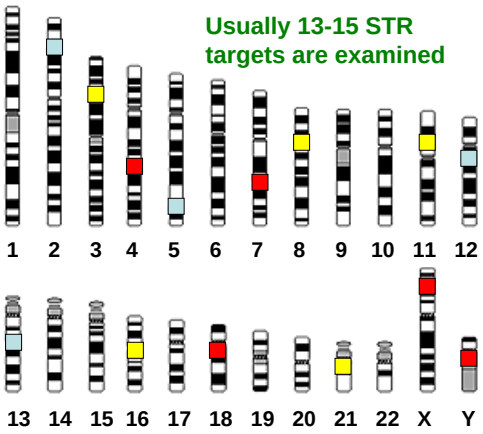


The number of consecutive repeat units can vary between people

The FBI has selected **13 core STR loci** that must be run in all DNA tests in order to provide a common currency with DNA profiles

What is a DNA Profile?

Human Genome
23 Pairs of Chromosomes (~3 billion bp)

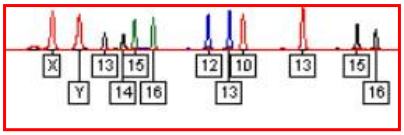


Unique regions of the human genome are targeted

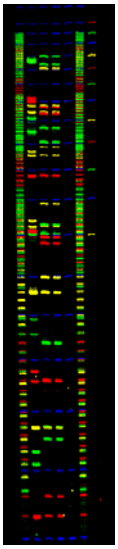
These regions consist of a few hundred base pairs

The regions are copied by the **polymerase chain reaction (PCR)** – billions of exact copies are made

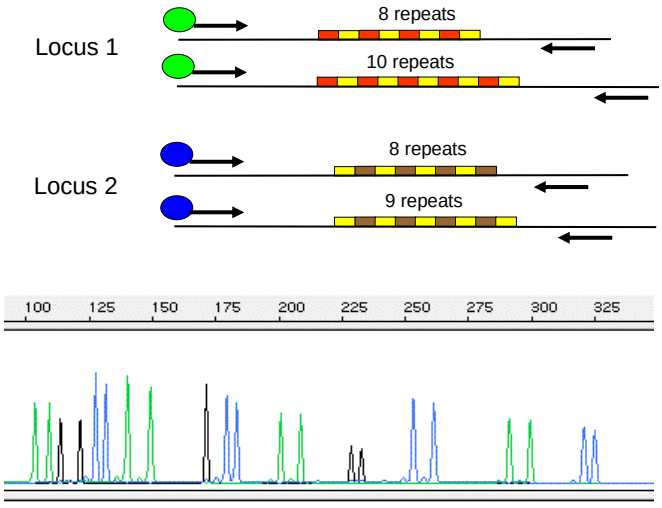
The copied fragments are labeled with fluorescent dyes for detection purposes



STR Allele Separation Can Be Performed by Gel or Capillary Electrophoresis with Detection of Fluorescent Dyes Labeling Each PCR Product



Scanned **Gel Image**



Capillary Electropherogram

STR Data is Tabulated by Genotype Calls for Each Locus

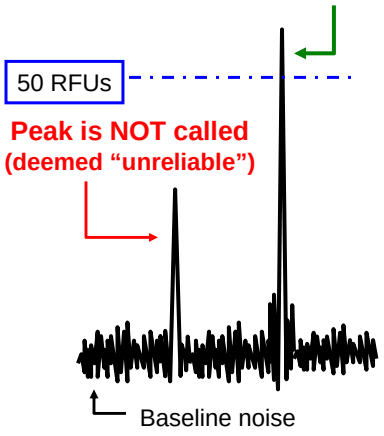
	AMEL	CSF1PO	FGA	TH01	TPOX	VWA	D3S1358	D5S818
Ind(1)	X,Y	11,12	19,21	6,7	8,8	15,18	14,18	10,13

The number of repeats observed for each locus is tabulated

This data format is stored in databases and used for comparisons/matches

Finally a case report is written based on tabulated STR genotype calls

Thresholds for Measuring DNA Data

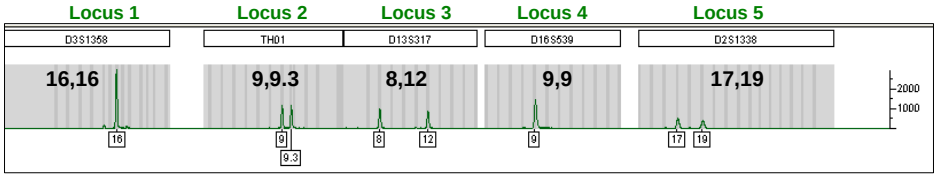


These thresholds for reliable data are determined through validation studies

- **Detection (analytical) threshold**
  - Dependent on instrument sensitivity ~50 RFU (relative fluorescence units)
  - Impacted by instrument baseline noise
- **Dropout (stochastic) threshold**
  - Dependent on biological sensitivity ~150-200 RFU
  - **Important in mixture interpretation**

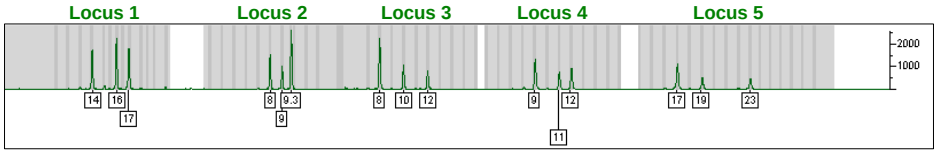
Single Source vs. Mixture Samples

Single Source Sample



One or two peaks observed at each locus (tested DNA region)

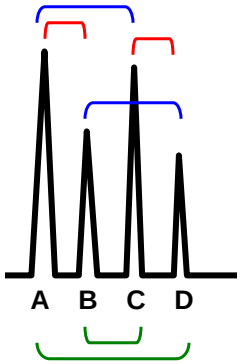
Mixture Sample



More than two peaks observed at more than two loci (tested DNA regions)

Different possible combinations could have given rise to the particular mixture observed

With Some Mixtures, Multiple Genotype Combinations Are Possible

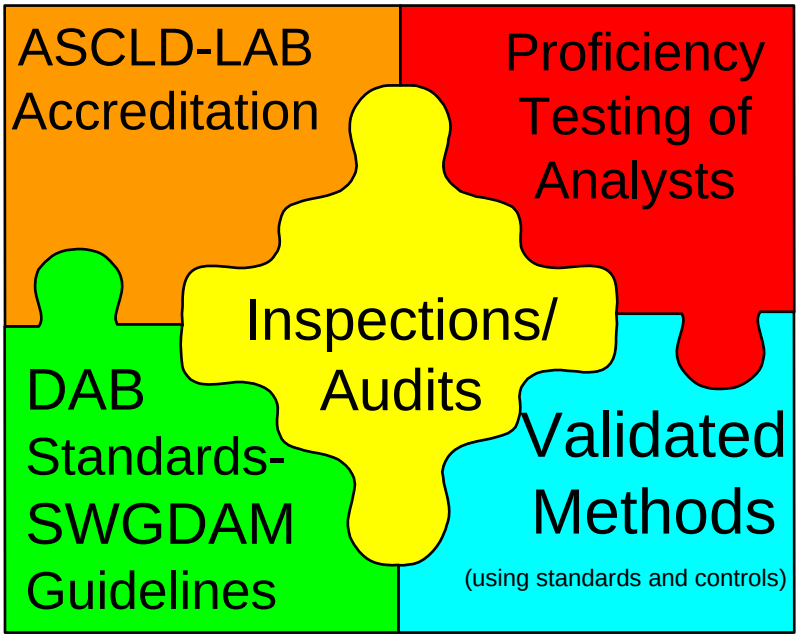


AC
BD
AB
CD
BC
AD

Depends on PHR and proportion of mixture components from the various contributors

Peak Height Ratios (PHR)
Minimum Peak Height (mPH)
Proportion (p) or mixture proportion (M_x)

Ensuring Accurate Forensic DNA Results



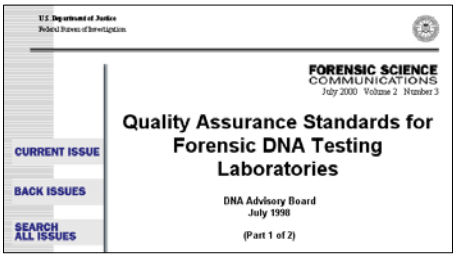
Scientific Working Group on DNA Analysis Methods (SWGDAM)

- Organized originally by FBI Laboratory as Technical Working Group on DNA Analysis Methods (TWGDAM) in 1988
- Meets semiannually – each January and July
- **Organized into eight subcommittees**:
  - Quality Assurance, CODIS, mtDNA, Mass Disasters/Missing Persons, Expert Systems, Serology, Y-STRs, and Mixture Interpretation
- Membership (usually ~50 attend) from public forensic DNA laboratories around the U.S.

DAB Standards issued in 1998-1999

Quality Assurance Standards (QAS)

1. SCOPE
2. DEFINITIONS
3. QUALITY ASSURANCE PROGRAM
4. ORGANIZATION AND MANAGEMENT
5. PERSONNEL
6. FACILITIES
7. EVIDENCE *(SAMPLE)* CONTROL
8. VALIDATION
9. ANALYTICAL PROCEDURES
10. EQUIPMENT CALIBRATION AND MAINTENANCE
11. REPORTS
12. REVIEW
13. PROFICIENCY TESTING
14. CORRECTIVE ACTION
15. AUDITS
16. SAFETY
17. SUBCONTRACTOR OF ANALYTICAL TESTING FOR WHICH VALIDATED PROCEDURES EXIST



Revised Quality Assurance Standards will go into effect July 1, 2009

http://www.fbi.gov/hq/lab/fsc/backissu/july2000/codis2a.htm
http://www.fbi.gov/hq/lab/fsc/backissu/july2000/codis1a.htm

Checks and Controls on DNA Results

Community	FBI DNA Advisory Board's Quality Assurance Standards *(also interlaboratory studies)*
Laboratory	ASCLD/LAB Accreditation and Audits
Analyst	Proficiency Tests & Continuing Education
Method/Instrument	**Validation of Performance** ***(along with traceable standard sample)***
Protocol	Standard Operating Procedure is followed
Data Sets	Allelic ladders, positive and negative amplification controls, and reagent blanks are used
Individual Sample	Internal size standard present in every sample
Interpretation of Result	Second review by qualified analyst/supervisor
Court Presentation of Evidence	Defense attorneys and experts with power of discovery requests

DNA Testing Requires a Reference Sample

A DNA profile by itself is fairly useless because it has no context…

DNA analysis for identity only works by comparison – **you need a reference sample**

Crime Scene Evidence compared to **Suspect(s)** (Forensic Case)
Child compared to **Alleged Father** (Paternity Case)
Victim's Remains compared to **Biological Relative** (Mass Disaster ID)
Soldier's Remains compared to **Direct Reference Sample** (Armed Forces ID)

The Three Possible Outcomes of Evidence Examination

	"Suspect" Known (K) Sample	"Evidence" Question (Q) Sample
• **Exclusion** (no match)	11 12	13
• **Non-exclusion** – "Match" or "inclusion"	11 12	11 12
• **Inconclusive result**	11 12	No result (or a complex mixture)

Emerging Trends/Issues

Things that are coming…

(or in development as possibilities)

Forensic Science Journals

Analytical Chemistry Application Review

Forensic Science

June 15, 2005 issue of *Analytical Chemistry*

T. A. Brettell*

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R. Saferstein

Box 1334, Mount Laurel, New Jersey 08054

Review Contents

250 articles referenced covering forensic DNA analysis during 2003-2004

2007 Article describes 181 forensic DNA articles published in 2005 and 2006 (**560 references** covering DNA, trace evidence, drugs and poisons)

PRESIDENT'S DNA INITIATIVE

Principles of Forensic DNA for Officers of the Court

1. **Introduction**
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12. **Victim Issues**
13. **Trial Presentation**
14. **Postconviction DNA Cases**
15. **Emerging Trends**

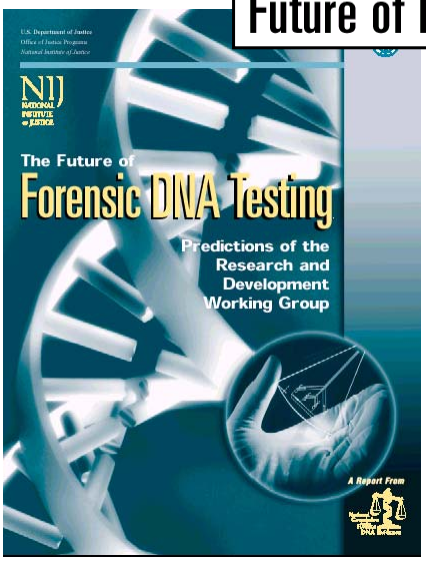
http://www.dna.gov/training/otc/

Content of Section 15 "Emerging Trends" from *Officers of the Court*

- Topic 1 :: **Single Nucleotide Polymorphisms (SNPs)**
- Topic 2 :: Automation
  - Microarrays (Chip Technology)
  - **Portable DNA Typing Laboratory**
  - Low Copy Number DNA Analysis
- Topic 3 :: Microbial Forensics and DNA Testing
- Topic 4 :: Other **Non-human Forensic DNA Analysis**
- Topic 5 :: **DNA Typing and Physical Appearance**
  - Biogeographical Ancestry
  - Approximate Age Determination

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

National Commission on the Future of DNA Evidence



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

- 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

Status of Genetic Marker Systems Used in Forensic DNA Testing

- **STRs** – widely used in casework and national databases world-wide
- **miniSTRs** – smaller versions of STR loci that can work well on degraded DNA
- **Y-STRs** – permits examination of male-only DNA
- **mtDNA** – used in specialty labs for highly degraded specimens or hair that contains limited amounts of DNA
- **SNPs** – potential for identifying ethnicity of evidence sample; still in research and likely to be limited in use

Portable DNA Testing Devices

NYC Forensic DNA "X-Prize"

January 17, 2008 Press Release
From Mayor Bloomberg's **STATE OF THE CITY ADDRESS**

"The City will establish **a six-figure prize** for anyone who can invent **a device** tailored to the NYPD **which analyzes the DNA of potential suspects right at the crime scene** - so that officers can release innocent suspects before they are arrested, and track down promising leads more quickly"

http://home2.nyc.gov/html/om/html/2008a/pr017-08.html

Efforts towards Portable/Mobile DNA Devices

- NEC (Japan)
  - Poster at Promega meeting in Hollywood, CA (Oct 1-4, 2007)
  - Press release on October 15, 2007 (http://www.nec.co.jp/press/en/0710/1501.html)
- Network Biosystems (based on Dan Ehrlich's work at Whitehead)
  - http://www.netbio.com
- Mathies group at UC-Berkeley and Microchip Biotech
  - Publications… in *Analytical Chemistry*, *FSI Genetics*, etc.
  - http://www.microchipbiotech.com
- Landers group at UVA and MicroLab Diagnostics
  - Publications… *Proc Natl Acad Sci USA* 2006; 103:19272-19277
  - http://www.microlabdiagnostics.com

Available online at www.sciencedirect.com
ScienceDirect
Forensic Science International: Genetics xxx (2008) xxx–xxx
ELSEVIER
FSI GENETICS
www.elsevier.com/locate/fsig

Real-time forensic DNA analysis at a crime scene using a portable microchip analyzer

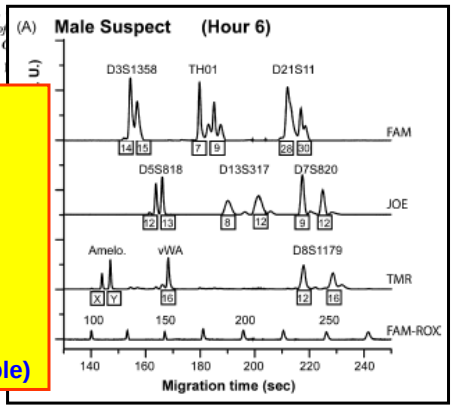
Peng Liu [a], Stephanie H.I. Yeung [a], Karin A. Crenshaw [c], Cecelia A. Crouse [c], James R. Scherer [b], Richard A. Mathies [a,b,*]

[a] UCSF/UC Berkeley Joint Graduate Group in Bioengineering,
[b] Department of Chemistry, MS 1460, University of
[c] Palm Beach County Sheriff's Office Crime Laboratory, 3228

Received 6 February 2008; received in revised form

Field Trial Results
7:00 a.m. Arrived and set up mock crime scene
7:10-7:30 a.m. Samples collected by CSI
7:30-9:30 a.m. DNA extraction
9:30-10:00 a.m. PCR set-up
10 a.m. – 12 p.m. PCR performed
12 – 12:30 p.m. DNA separation
12:30-12:50 p.m. CODIS search of local database

6 hours from sample collection to the generation of the CODIS hit (for one sample)



DNA Degradation

Intact sample

Target region for PCR

300 base pair PCR product can be produced

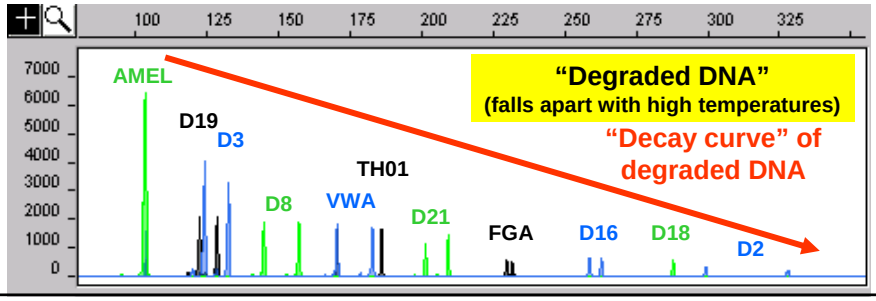
Degraded sample

Target region for PCR is fragmented

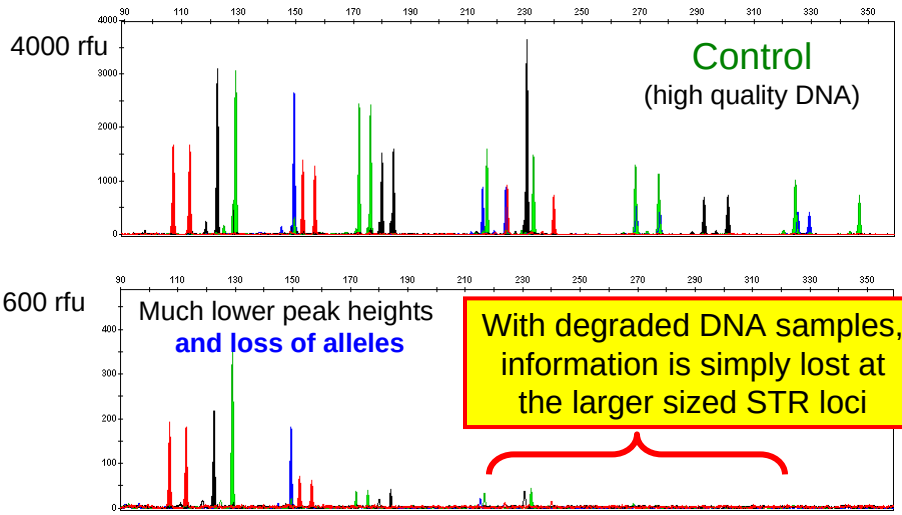
300 base pair PCR product can not be produced or only in limited quantities

Degraded DNA

Larger segments of DNA cannot be recovered when DNA molecules have fragmented into small pieces (caused by heat, water, or bacteria)



DNA Degradation Means **Less Loci Work**



Impact of Degraded DNA Samples

- Comparison to a phone number (string of 13 numbers)

001-301-975-4049

- If you only had "4049"…this information would be of limited value since it is not as specific (and could match other phone numbers from different area codes)
- DNA profiles are essentially a string of numbers – **if the DNA is damaged, then the string of numbers is shorter and less informative…**

----------4049 or ----301-9-------

The Statistic (Determining the Weight of the Evidence) Should Be **Calculated from the Evidence**

Evidence (partial profile):

	Type	Statistic
Locus 1	16,17	1 in 9
Locus 2	17,18	1 in 9
Locus 3	21,22	1 in 12
Locus 4	12,14	1 in 16
Locus 5	28,30	1 in 11

	Product =	1 in 171,000

Match Observed at All Loci that May Be Compared

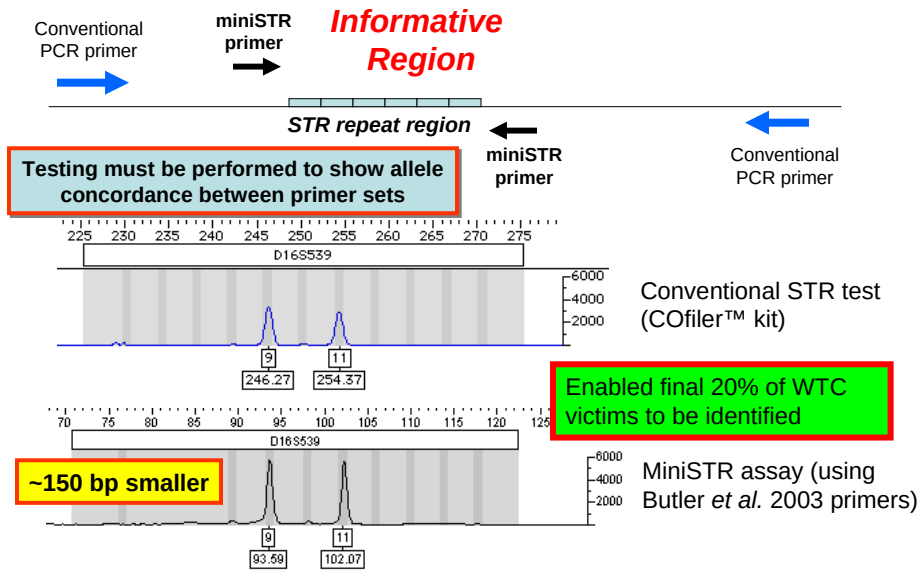
Reference (full profile):

	Type	Statistic
Locus 1	16,17	1 in 9
Locus 2	17,18	1 in 9
Locus 3	21,22	1 in 12
Locus 4	12,14	1 in 16
Locus 5	28,30	1 in 11
Locus 6	14,16	1 in 26
Locus 7	12,13	1 in 9
Locus 8	11,14	1 in 31
Locus 9	9,9	1 in 32
Locus 10	9,11	1 in 14
Locus 11	6,6	1 in 19
Locus 12	8,8	1 in 3
Locus 13	10,10	1 in 21

	Product =	1 in 665 trillion

The reference sample is still a "match" – just not as much information is available from the evidence for comparison

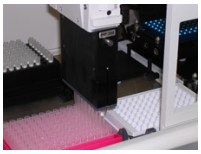
A miniSTR is a reduced size STR amplicon that enables higher recovery of information from degraded DNA samples



Butler, J.M. (2005) *Forensic DNA Typing, 2nd Edition,* Figure 7.2, ©Elsevier Science/Academic Press

The Future

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



http://www.manastungare.com/publications/genetic/dna.gif

Non-Human Forensic DNA Analysis

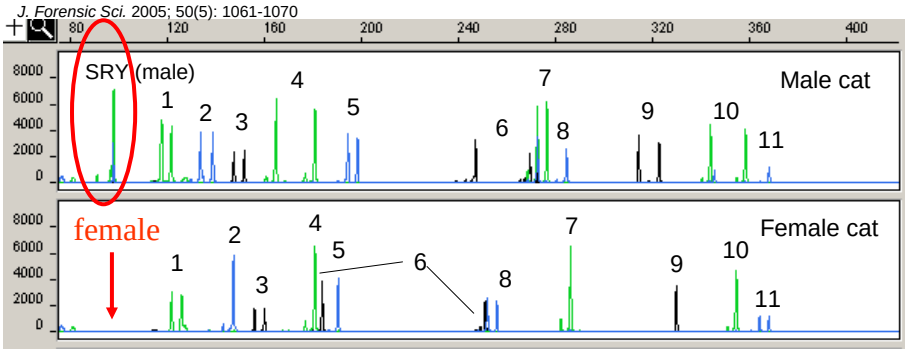
Non-Human DNA Testing

- Cat DNA
- Dog DNA

Animal as
1) Victim (abuse case)
2) Perpetrator (dog bite)
3) Silent witness (crime scene linkage)

- Other uses of non-human DNA
  - Plant DNA – for possible linkage to crime location
  - Marijuana DNA – tracking drug sources

New DNA Test for Cats Developed at NIST



Animal DNA Testing

- Most non-human DNA tests are specialty tests that will be rarely used by public labs and thus typically will be performed through outsourcing to a contract lab

- QuestGen Forensics
  - http://www.questgen.biz/
- UC Davis Vet Gen Lab
  - http://www.vgl.ucdavis.edu/

Challenges with Presenting Non-Human DNA in Court (or other novel DNA methods)

Sensabaugh and Kaye (1998) *Jurimetrics* 38: 1-16

- Novelty of the application
- Validity of the underlying scientific theory
- Validity of any statistical interpretations
- Relevant scientific community to consult in assessing the application may be limited

- **New methods may not have undergone the scientific scrutiny of regular forensic human DNA testing techniques**

What It Means to "Validate" An Analysis Process

- How **reproducible** is an analysis?
  - If the same evidence is examined multiple times, would the same conclusion be reached each time?
- How **robust** is an analysis?
  - Are results obtained every time (or a high percentage of the time) evidence is tested?
- How **reliable** is an analysis?
  - If known samples are examined, are the expected results obtained?
  - Are situations with insufficient evidence reaching conclusions of "no results"?

DNA Typing and Physical Appearance

Biogeographical Ancestry
Approximate Age Determination

Biogeographical Ancestry

- **Shriver, M.D. *et al.* (2003) Skin pigmentation, biogeographical ancestry and admixture mapping. *Hum. Genet.* 112(4):387-99**
- **From abstract:** Ancestry informative markers (AIMs) are genetic loci showing alleles with large frequency differences between populations. AIMs can be used to estimate biogeographical ancestry at the level of the population, subgroup (e.g. cases and controls) and individual…. **This work indicates that it is possible to estimate the individual ancestry of a person based on DNA analysis with a reasonable number of well-defined genetic markers.**

Biogeographical Ancestry (2)

- Mark Shriver's work on ancestry informative markers has been commercialized through the company **DNAPrint Genomics**
- http://www.dnaprint.com
- http://www.ancestrybydna.com
- Used in Derrick Todd Lee (Louisiana serial killer) case to overcome faulty eyewitness testimony of a Caucasian perpetrator…

Pigmentation (Skin Color, etc.) Prediction

- **Lamason, R.L. *et al.* (2005) SLC24A5, a putative cation exchanger, affects pigmentation in zebrafish and humans. *Science* 310:1782-1786**
- **From abstract:** Lighter variations of pigmentation in humans are associated with diminished number, size, and density of melanosomes, the pigmented organelles of melanocytes. **The variant allele is nearly fixed in European populations**, is associated with a substantial reduction in regional heterozygosity, **and correlates with lighter skin pigmentation** in admixed populations, suggesting a key role for the SLC24A5 gene in human pigmentation.

Approximate Age Determination

- **Alvarez, M. and Ballantyne, J. (2006) The identification of newborns using messenger RNA profiling analysis. *Anal. Biochem.* 357(1):21-34.**
- **From abstract:** In theory, it may be possible to determine patterns of gene expression that are age specific, thereby permitting the distinction among tissue samples originating from individuals of different ages (e.g., newborn, adolescent, middle-age, elderly). **We have discovered two novel isoforms of gamma hemoglobin messenger RNA, designated HBG1n and HBG2n, which exhibit an extremely restricted pattern of gene expression, being confined to newborn individuals.** Multiplex quantitative reverse transcription PCR (qRT-PCR) assays incorporating these novel mRNAs have been designed, tested, and evaluated for their potential forensic use. The results indicate that the assays provide the ability to determine whether a bloodstain originated from a newborn.

Age of Bloodstain Deposition

- **Anderson, S., Howard, B., Hobbs, G.R., Bishop, C.P. (2005) A method for determining the age of a bloodstain. *Forensic Sci. Int.* 148(1):37-45**
- **From abstract:** If there were independent evidence that the biological sample was deposited at the time of the crime, then its age would reveal when the crime occurred. If the time of the crime were known through another means, then the age of the biological sample could potentially exclude the human source as a suspect. **We have used real-time reverse transcriptase PCR to show that the ratio between different types of RNA (mRNA versus rRNA) changes over time** in a linear fashion when dried human blood from eight individuals was examined over the course of 150 days.

Determination of Body Fluid Type

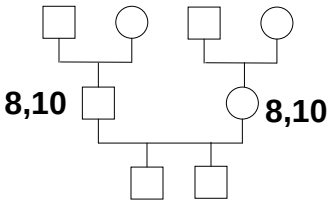
- **Juusola, J. and Ballantyne, J. (2005) Multiplex mRNA profiling for the identification of body fluids. *Forensic Sci. Int.* 152(1):1-12**
- **From abstract:** We report the development of a multiplex reverse transcription-polymerase chain reaction (RT-PCR) method for the **definitive identification of the body fluids that are commonly encountered in forensic casework analysis, namely blood, saliva, semen, and vaginal secretions**. Using selected genes that we have identified as being expressed in a tissue-specific manner we have developed a multiplex RT-PCR assay which is composed of eight body fluid-specific genes and that is optimized for the detection of blood, saliva, semen, and vaginal secretions as single or mixed stains. The genes include beta-spectrin (SPTB) and porphobilinogen deaminase (PBGD) for blood, statherin (STATH) and histatin 3 (HTN3) for saliva, protamine 1 (PRM1) and protamine 2 (PRM2) for semen, and human beta-defensin 1 (HBD-1) and mucin 4 (MUC4) for vaginal secretions.

Partial Matching/Familial Searching

- Current searching software not designed for partial matches
- Need Y-STRs along with autosomal STR information to help sort through false positive matches obtained with single allele sharing hits
- See Bieber *et al.* (2006) Finding criminals through DNA of their relatives. *Science* 312:1315-1316

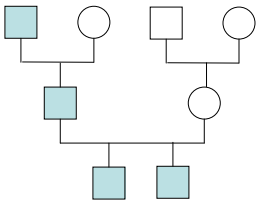
Why Y-STRs Are Needed for Familial Searching

Autosomal STRs



8,8 10,10

Y-Chromosome STRs



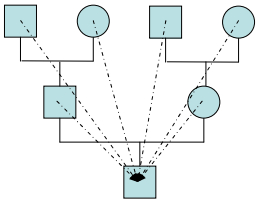
Y-STRs match

For brothers, autosomal STRs may not match at a locus (or even share a single allele)

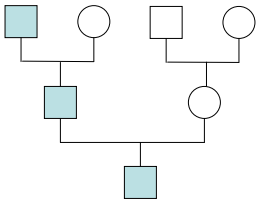
Different Inheritance Patterns

Lineage Markers

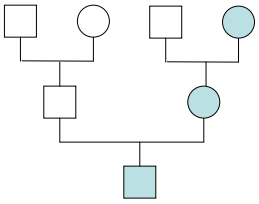
CODIS STR Loci



Autosomal
(**passed on in part**, from all ancestors)



Y-Chromosome
(**passed on complete**, but only by sons)



Mitochondrial
(**passed on complete**, but only by daughters)

Butler, J.M. (2005) ***Forensic DNA Typing, 2nd Edition,*** Figure 9.1, ©Elsevier Science/Academic Press

Some Final Thoughts

- "DNA" + "Match" ➔ "Guilty" in the minds of many jurors
- Be careful to state assumptions going into the weight of the evidence particularly for mixtures
- General population (i.e., jury pool) is becoming more informed regarding DNA testing thanks to genetic genealogy and TV shows like CSI
- Low-level DNA recovered from a crime scene may not be relevant to the committed crime

Conclusions

- This is an exciting time to be involved in forensic DNA testing
- However, it is a little scary because technology is advancing so rapidly on some fronts
- Thus, training for both the scientific and legal communities is vital to make the most effective use of the wonderful power of DNA technology

Thank you for your attention…

Our team publications and presentations are available at:
http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm

Questions?

See also http://www.dna.gov/research/nist

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

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