Everything a Trial Judge Needs to Know about DNA (in a nutshell)

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What Topics Would You Like to Explore?

From 6/21/13 audience

• Key concepts
• How to know if experts are qualified
• Rapid DNA capabilities
• New information in “junk DNA” regions
• How to make DNA understandable to juries
• Should data be accepted without stochastic threshold
• Why racial categories when reporting profile statistics
• Review of DNA basics
Interfaces Between Disciplines Are Crucial

Law Enforcement

Judicial

Laboratory

What Every Law Enforcement Officer Should Know About DNA Evidence

http://www.dna.gov/
Progress Since 1995…

Almost 8 weeks needed to get results

O.J. Simpson DNA testing was performed with RFLP

Now <8 hours to get results
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.
Standard Reference Materials

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

Traceable standards to ensure accurate measurements in our nation’s crime laboratories

SRM 2391b – CODIS STRs
SRM 2392-I – mtDNA
SRM 2395 – Y-STRs
SRM 2372 – DNA quantitation

Calibration with SRMs enables confidence in comparisons of results between laboratories

Helps meet DAB Std. 9.5 and ISO 17025
Information Resources

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

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

- 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
Lessons from the First Case Involving DNA Testing

Describes the first use of DNA (in 1986) to solve a double rape-homicide case in England; about 5,000 men asked to give blood or saliva to compare to crime stains

- Connection of two crimes (1983 and 1986)
- Use of DNA database to screen for perpetrator (DNA only done on 10% with same blood type as perpetrator)
- Exoneration of an innocent suspect
- DNA was an investigative tool – did not solve the case by itself (confession of accomplice)

A local baker, Colin Pitchfork, was arrested and his DNA profile matched with the semen from both murders. In 1988 he was sentenced to life for the two murders.
308 exonerated as of June 19, 2013

4 in Maryland
New Handbook on Biological Evidence Preservation


73 page handbook that makes recommendations for evidence retention, safe handling, packaging and storage, chain-of-custody and tracking, and appropriate disposal once evidence retention is no longer required by law.

Table III-2: Long-Term Storage Conditions Matrix

<table>
<thead>
<tr>
<th>Type of Evidence</th>
<th>Frozen</th>
<th>Refrigerated</th>
<th>Temperature Controlled</th>
<th>Room Temperature</th>
</tr>
</thead>
<tbody>
<tr>
<td>Liquid Blood</td>
<td>Never</td>
<td>Best</td>
<td>Best</td>
<td>Best</td>
</tr>
<tr>
<td>Urine</td>
<td></td>
<td>Best</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Dry Biological Stained Items</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Bones</td>
<td></td>
<td>Best</td>
<td>Best (dried)</td>
<td>Acceptable</td>
</tr>
<tr>
<td>Hair</td>
<td></td>
<td>Best</td>
<td>Best</td>
<td></td>
</tr>
<tr>
<td>Swabs with Biological Material</td>
<td></td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Vaginal Smears</td>
<td></td>
<td>Best</td>
<td>Best</td>
<td></td>
</tr>
<tr>
<td>Feces</td>
<td></td>
<td>Best</td>
<td>Best</td>
<td></td>
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<tr>
<td>Buccal Swabs</td>
<td></td>
<td>Best</td>
<td>Acceptable (dried)</td>
<td></td>
</tr>
<tr>
<td>DNA Extracts</td>
<td>Best (liquid)</td>
<td>Acceptable (liquid)</td>
<td>Acceptable (dried)</td>
<td></td>
</tr>
</tbody>
</table>

Released April 2013
National Academies Report on Forensic Science

• Released February 18, 2009
• Entitled “Strengthening Forensic Science in the United States: A Path Forward”
• 13 recommendations provided to Congress
• Recommends establishing a National Institute of Forensic Science (NIFS)
• NIST and the U.S. Department of Justice announced plans on February 15, 2013 to establish a National Commission on Forensic Science
“[DNA analysis] has set the bar higher for other forensic science methodologies, because it has provided a tool with a higher degree of reliability and relevance than any other forensic technique.”

p.41
Methods for Human Identification

Fingerprints have been used since 1901

DNA since 1986
Historical Perspective on DNA Typing

2013: DNA is an important part of the criminal justice system

- 1985: PCR developed
- 1990: First STRs developed
- 1992: Capillary electrophoresis of STRs first described
- 1994: First STRs developed
- 1996: CODIS loci defined
- 1998: First commercial fluorescent STR multiplexes
- 2000: STR typing with CE is fairly routine
- 2002: Identifiler 5-dye kit and ABI 3100
- 2004: PowerPlex 16 (16 loci in single amp)
- 2006: NDIS launched (October 13, 1998)
- 2013: UK National Database launched (April 10, 1995)
- 2013: www.dna.gov
- 2013: President’s DNA Initiative
- 2013: Debbie Smith Act Backlog Reduction (> $1B from 2004-2010)

- 1990: RFLP
- 1992: DQA1 & PM (dot blot)
- 1994: FSS
- 1996: Quadruplex
- 2000: miniSTRs
- 2004: Y-STRs
- 2006: mtDNA
- 2013: Multiplex STRs

- 2013: DNA is an important part of the criminal justice system
<table>
<thead>
<tr>
<th>Stages</th>
<th>Time Frame</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Exploration</td>
<td>1985-1995</td>
<td>Beginnings, different methods tried (RFLP and early PCR)</td>
</tr>
<tr>
<td>Stabilization</td>
<td>1995-2005</td>
<td>Standardization to STRs, selection of core loci, implementation of Quality Assurance Standards</td>
</tr>
<tr>
<td>Growth</td>
<td>2005-2013</td>
<td>Rapid growth of DNA databases, extended applications pursued</td>
</tr>
<tr>
<td>Sophistication</td>
<td>The Future</td>
<td>Expanding tools available, confronting privacy concerns</td>
</tr>
</tbody>
</table>

**Stages of Forensic DNA Progression**
We are finding new ways to use DNA...

We’re taking back your first place ribbon. —We found traces of your parents’ DNA all over your science fair project.
**Paternity Testing**

Father's Profile? 11,14

Alleged Father(s) is asked to donate DNA sample

STR Alleles from D13S317
Lab Procedures
Steps in DNA Analysis

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

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

**Genetics**
- Blood Stain
- Buccal swab
- Sample Collection & Storage
- 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

**Technology**
- DNA Extraction
- DNA Quantitation
- Multiplex PCR Amplification
- DNA separation and sizing

**Biology**
- STR Typing

**Database**
- Search

**Notes**
- Male: 13,14-15,16-12,13-10,13-15,16
- 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

**Samples**
- Blood Stain
- Buccal swab

**Quantitation**
- Slot Blot
  - 1 ng
  - 0.3 ng
  - No DNA
  - 0.5 ng
  - 0.7 ng
  - 1 ng
Crime Scene Collection of Evidence

• Police officers and crime scene investigators respond to the scene of a crime to collect biological evidence to be used in forensic DNA testing

• Investigators must be careful not to contaminate the evidence with their own DNA

http://projects.nfstc.org/gallery/main.php?g2_itemId=626
DNA Evidence Received in the Lab

- Evidentiary samples (commonly in the form of cotton swabs) are brought or shipped to the DNA laboratory after collection from the crime scene or victim.

- Sexual assault evidence collection kits provide swabs and bags for clothing collections from the victim.
DNA Collection

- Cotton swabs are commonly used to collect biological material from bloodstains or semen from sexual assault victims.

- The amount of DNA needed has decreased dramatically in the past decade due to the sensitivity of the PCR process (which makes millions of copies of targeted regions).
Sources of Biological Evidence

- Blood
- Semen
- Saliva
- Urine
- Hair
- Teeth
- Bone
- Tissue

Blood Sample

Only a very small amount of blood is needed to obtain a DNA profile.

best results with >100 cells, but DNA profiles can be recovered from as little as a single cell.
DNA Reference Sample from Suspect

- Blood samples may be collected but require a phlebotomist to draw blood

- Easier to collect a buccal swab from the inside of an individual’s mouth, which scrapes off some cheek cells
Buccal Swab DNA Collection

- The inside of the check is scrubbed to collect cells
- Less invasive than drawing blood
- Swab must be dried before storing and shipping to lab to avoid mold and bacterial growth
DNA Extraction

- DNA is extracted from proteins that protect it in the nucleus of a cell

- Chemicals are added to digest the protecting proteins and produce “naked” DNA molecules

- The final solution looks like a tube of water
DNA Quantitation

- DNA quantitation is important to determine how much human DNA (as opposed to bacterial DNA) is present in a sample.

- A commonly used DNA quantitation kit is called Quantifiler (sold by Applied Biosystems).

ABI 7500: an instrument used to perform “real-time quantitative PCR”
Impact of DNA Amount into PCR

Reason that DNA Quantitation is Important Prior to Multiplex Amplification

- Too much DNA
  - Off-scale peaks
  - Split peaks (+/- A)
  - Locus-to-locus imbalance

- Too little DNA
  - Heterozygote peak imbalance
  - Allele drop-out
  - Locus-to-locus imbalance

Stochastic effect when amplifying low levels of DNA produces allele dropout
PCR Amplification

• PCR = polymerase chain reaction

• Process that copies a particular region of DNA using two “primers” (short pieces of DNA)

• Each strand of DNA is used as a template to create a replicate that permits a doubling of the number of target molecules with each cycle of heating and cooling
PCR Process

Separate strands (denature)

Starting DNA Template 80-500 bases

Add primers (anneal)

Forward Primer

Reverse Primer

Make copies (extend primers)

Repeat Cycle, Copying DNA Exponentially
PCR Amplification (Thermal Cycling)

- The polymerase chain reaction (PCR) copies sections of DNA through heating and cooling the sample.
  - Each DNA strand is copied with each temperature cycle.
  - A thermal cycler heats and cools DNA samples (usually 28 cycles).
DNA in the Cell

The vast majority of DNA is the same from person to person

chromosome 22 pairs + XX or XY

cell nucleus

Double stranded DNA molecule

~3 billion total base pairs

Only a Small Varying Region is Targeted and Probed for Each DNA Marker Examined
<table>
<thead>
<tr>
<th>Printed Information</th>
<th>Genetic Information</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Library</strong></td>
<td><strong>Body</strong></td>
</tr>
<tr>
<td><strong>Book</strong></td>
<td><strong>Cell</strong></td>
</tr>
<tr>
<td><strong>Chapter</strong></td>
<td><strong>Nucleus</strong></td>
</tr>
<tr>
<td><strong>Page Number</strong></td>
<td><strong>Chromosome</strong></td>
</tr>
<tr>
<td><strong>Line on Page</strong></td>
<td><strong>Locus</strong> (part of chromosome)</td>
</tr>
<tr>
<td><strong>Word</strong></td>
<td><strong>Short DNA sequence</strong></td>
</tr>
<tr>
<td><strong>Letter</strong></td>
<td><strong>DNA nucleotides</strong></td>
</tr>
</tbody>
</table>
Characteristics of DNA

- Each person has a unique DNA profile (except identical twins).
- Each person's DNA is the same in every cell.
- An individual’s DNA profile remains the same throughout life.
- Half of your DNA comes from your mother and half from your father.
Our DNA Comes from our Parents

Father’s Sperm

Child’s Cell

Mother’s Egg
Inheritance Pattern of DNA Profiles
Basis of DNA Profiling

The genome of each individual is unique (with the exception of identical twins) and is inherited from parents.

Probe subsets of genetic variation in order to differentiate between individuals (statistical probabilities of a random match are used).

DNA typing must be performed efficiently and reproducibly (information must hold up in court).

Current standard DNA tests DO NOT look at genes – little/no information about race, predisposition to disease, or phenotypical information (eye color, height, hair color) is obtained.
What is a DNA Profile?

Human Genome
23 Pairs of Chromosomes

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

The copied fragments now contain fluorescent dyes for detection


Nuclear DNA
3.2 billion bp
Short Tandem Repeat (STR) Markers

An accordion-like DNA sequence that occurs between genes

TCCCAAGCTCTTCCCTCTTCCCTAGATCAATACAGACAGAAGACA
GGTG\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}\textbf{GATA}

= 11 GATA repeats ("11" is all that is reported)

\begin{itemize}
  \item 7 repeats
  \item 8 repeats
  \item 9 repeats
  \item 10 repeats
  \item 11 repeats
  \item 12 repeats
  \item 13 repeats
\end{itemize}

The number of consecutive repeat units can vary between people

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

Target region
(short tandem repeat)
Position of Forensic STR Markers on Human Chromosomes

13 CODIS Core STR Loci

- TPOX
- D3S1358
- D5S818
- FGA
- CSF1PO
- D8S1179
- D7S820
- TH01
- VWA
- D13S317
- D16S539
- D18S51
- D21S11
- AMEL
- AMEL

Sex-typing

Core STR Loci for the United States 1997
Short Tandem Repeat (STR) Typing

Fluorescent dye-labeled primer

STR Repeat Region

forward primer hybridization region

reverse primer hybridization region

(size in bp)

75…80…100…120…140…160…180…200…220…240…260…..

RFUs

1000

500

DNA Separation and Detection

139bp

147bp

6

8
DNA Reaction Setup

• DNA sample is added (about 1 ng based on DNA quantitation performed) – 10 µL

• PCR primers and other reaction chemicals from an STR typing kit are added – 15 µL

Strip of 8 tubes containing ~25 µL of solution
STR Typing Kit

• Kit Components:
  – Primer mix
  – PCR Reaction Buffer and Building Blocks
  – DNA Polymerase (Taq Gold)

• Most expensive reagent

• Common kits used:
  – Identifiler (Applied Biosystems)
  – Profiler Plus/COFiler (Applied Biosystems)
  – PowerPlex 16 (Promega)
What is in an STR Typing Kit?

• Primer mix
  – containing fluorescently labeled oligonucleotides used to target specific regions of the human genome
  – Applied Biosystems has not published their primer sequences
  – PowerPlex 16, which amplifies 16 genomic sites, contains 32 PCR primers
## PCR Primers in an STR Kit

### Locus Dye Promega PP16 Primer Sequences

<table>
<thead>
<tr>
<th>Locus</th>
<th>Dye</th>
<th>Promega PP16 Primer Sequences</th>
</tr>
</thead>
<tbody>
<tr>
<td>D3S1358-F</td>
<td>FL</td>
<td>ACTGCAGTCCAATCTGGGT</td>
</tr>
<tr>
<td>D3S1358-R</td>
<td>FL</td>
<td>ATGAAATCAACAGAGGCTTGC</td>
</tr>
<tr>
<td>TH01-F</td>
<td>FL</td>
<td>GTGATTCCCATTGGCCTGTTC</td>
</tr>
<tr>
<td>TH01-R</td>
<td>FL</td>
<td>ATTCCTGTGGGTGAAGCCTCT</td>
</tr>
<tr>
<td>D21S11-F</td>
<td>FL</td>
<td>ATATGTGAGTCAATCCCAG</td>
</tr>
<tr>
<td>D21S11-R</td>
<td>FL</td>
<td>TGTATTAGTCAATGTGTTCTCAGAGAC</td>
</tr>
<tr>
<td>D18S51-F</td>
<td>FL</td>
<td>TTCTTGAGCCCAGAGGTTA</td>
</tr>
<tr>
<td>D18S51-R</td>
<td>FL</td>
<td>ATTCCTACAGCAACACACAAAAATAAAC</td>
</tr>
<tr>
<td>PentaE-F</td>
<td>FL</td>
<td>ATTACCAACATGAAAGGTACACATAA</td>
</tr>
<tr>
<td>PentaE-R</td>
<td>FL</td>
<td>TGGGTTATATTGGAGAAACTCCTCATAAATTT</td>
</tr>
<tr>
<td>D5S818-F</td>
<td>JOE</td>
<td>GGTGATTTTCCTTTTGTTATCC</td>
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<tr>
<td>D5S818-R</td>
<td>JOE</td>
<td>AGCCACAGTTTACAACATTGGTAC</td>
</tr>
<tr>
<td>D13S317-F</td>
<td>JOE</td>
<td>ATTACAGAAGTCTGGATGGAGGAG</td>
</tr>
<tr>
<td>D13S317-R</td>
<td>JOE</td>
<td>GCCAGCCCCAAAAGACAGA</td>
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<tr>
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<td>JOE</td>
<td>ATGTGGTGCAGGCTGACTATG</td>
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<tr>
<td>D7S820-R</td>
<td>JOE</td>
<td>ATTCCACATTTTACCTCATGAC</td>
</tr>
<tr>
<td>D16S539-F</td>
<td>JOE</td>
<td>GGGGGTCTAAGAAGCTTGTAAGAAG</td>
</tr>
<tr>
<td>D16S539-R</td>
<td>JOE</td>
<td>GCTTGCTGTGTGCATCTGTAAAGCATGTAC</td>
</tr>
<tr>
<td>CSF1PO-F</td>
<td>JOE</td>
<td>CCAGGAGGTAAGGTTCTTTAAGT</td>
</tr>
<tr>
<td>CSF1PO-R</td>
<td>JOE</td>
<td>ATTTCTGTCAGACCCCTTTT</td>
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<td>PentaD-F</td>
<td>JOE</td>
<td>GAAGGTCGAAAGCTGAAGTG</td>
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<tr>
<td>AMEL-F</td>
<td>TMR</td>
<td>CCCTGGGCTCGTAAAGA</td>
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<td>AMEL-R</td>
<td>TMR</td>
<td>ATCACAGCTTAAACTGGGAAGCTG</td>
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<td>vWA-F</td>
<td>TMR</td>
<td>GCCCTAGTGGATGATAAAGAATACATCAGATGTG</td>
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<td>vWA-R</td>
<td>TMR</td>
<td>GGGAGCAGATGATAAATACATGAGGATGGATGG</td>
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<tr>
<td>D8S1179-F</td>
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<td>ATGCAACCTATATGTATTTTGTATTTTATTTTCATG</td>
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<tr>
<td>D8S1179-R</td>
<td>TMR</td>
<td>ACCAAATTGTGTCTGAGTATATGTTTTTC</td>
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<tr>
<td>TPOX-F</td>
<td>TMR</td>
<td>GCACAGAAAGCAGCATTAGG</td>
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<td>FGA-F</td>
<td>TMR</td>
<td>GGCTGCAGGGCATAACATTA</td>
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<tr>
<td>FGA-R</td>
<td>TMR</td>
<td>ATTCTATGACCTTGCGCTCAGGA</td>
</tr>
</tbody>
</table>
The polymerase chain reaction (PCR) is used to amplify STR regions and label the amplicons with fluorescent dyes using locus-specific primers.
Transfer of DNA Samples

- Following PCR, a small portion of the sample is transferred for analysis.

- This aliquot of the sample is mixed with a molecular size marker (termed an internal size standard) that permits calibration of sizing measurements.
Sample Plates Spun Down via a Centrifuge

- Sample plates are spun to remove bubbles that would interfere with the injection (loading) process onto the capillary electrophoresis instrument.
ABI 3130xl DNA Analysis Instrument

- Import sample names
- Determine run conditions (voltages and times to be used based on laboratory protocols)
Data Collection on ABI 3130xl Instrument

• Data analysis is performed on an Applied Biosystems (ABI) 3130xl capillary electrophoresis instrument

DNA Profile
Capillary Electrophoresis Instrumentation

ABI 310
single capillary

ABI 3100
16-capillary array
Steps in STR Typing with ABI 310/3100

A DNA Profile is Produced by Separating DNA Molecules by Size and Dye Color

The labeled fragments are separated (based on size) and detected on a gel or capillary electrophoresis instrument ~2 hours or less.

Peaks represent labeled DNA fragments separated by electrophoresis. This ‘profile of peaks’ is unique for an individual – a DNA type.
### DNA Profile Frequency with all 13 CODIS STR loci

<table>
<thead>
<tr>
<th>Locus</th>
<th>allele</th>
<th>value</th>
<th>Locus</th>
<th>allele</th>
<th>value</th>
<th>1 in</th>
<th>Combined</th>
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<tbody>
<tr>
<td>D3S1358</td>
<td>16</td>
<td><strong>0.2533</strong></td>
<td>17</td>
<td>17</td>
<td><strong>0.2152</strong></td>
<td>9.17</td>
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<td>VWA</td>
<td>17</td>
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<td>18</td>
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<tr>
<td>FGA</td>
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<td>D8S1179</td>
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<td>0.1854</td>
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<td>13</td>
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<td>9.25</td>
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<td>D13S317</td>
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<td>14</td>
<td>0.0480</td>
<td>30.69</td>
<td>1.38 x 10^9</td>
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<tr>
<td>D7S820</td>
<td>9</td>
<td>0.1772</td>
<td></td>
<td></td>
<td></td>
<td>31.85</td>
<td>4.38 x 10^{10}</td>
</tr>
<tr>
<td>D16S539</td>
<td>9</td>
<td>0.1126</td>
<td>11</td>
<td>11</td>
<td>0.3212</td>
<td>13.8</td>
<td>6.05 x 10^{11}</td>
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<tr>
<td>THO1</td>
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<td>0.2318</td>
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<td>18.62</td>
<td>1.13 x 10^{13}</td>
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<tr>
<td>TPOX</td>
<td>8</td>
<td>0.5348</td>
<td></td>
<td></td>
<td></td>
<td>3.50</td>
<td>3.94 x 10^{13}</td>
</tr>
<tr>
<td>CSF1PO</td>
<td>10</td>
<td>0.2169</td>
<td></td>
<td></td>
<td></td>
<td>21.28</td>
<td>8.37 x 10^{14}</td>
</tr>
</tbody>
</table>

The Random Match Probability for this profile in the U.S. Caucasian population is 1 in 837 trillion (10^{12}).

What would be entered into a DNA database for searching:
- 16,17-
- 17,18-
- 21,22-
- 12,14-
- 28,30-
- 14,16-
- 12,13-
- 11,14-
- 9,9-
- 9,11-
- 6,6-
- 8,8-
- 10,10
The Same 13 Locus STR Profile in Different Populations

1 in 837 trillion

1 in 0.84 quadrillion \((10^{15})\) in U.S. Caucasian population (NIST)
1 in 2.46 quadrillion \((10^{15})\) in U.S. Caucasian population (FBI)*
1 in 1.86 quadrillion \((10^{15})\) in Canadian Caucasian population*

1 in 16.6 quadrillion \((10^{15})\) in African American population (NIST)
1 in 17.6 quadrillion \((10^{15})\) in African American population (FBI)*

1 in 18.0 quadrillion \((10^{15})\) in U.S. Hispanic population (NIST)

These values are for unrelated individuals assuming no population substructure (using only \(p^2\) and \(2pq\))


*http://www.csfs.ca/pplus/profiler.htm
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)
Differential extraction used to separate sperm (male fraction) from vaginal epithelial cells (female fraction).

Four samples are typically associated with a sexual assault forensic DNA case...
Three Possible Outcomes of Evidence Examination

- **Exclusion** (no match)

- **Inclusion** (match)

- **Inconclusive result** (or a complex mixture)

\[ \text{Unable to make } Q \rightarrow K \text{ comparison} \]
Rapid DNA Efforts

Accelerated Nuclear DNA Equipment (ANDE) developed by NetBio

- Evaluating ANDE (NetBio) and IntegenX rapid DNA instruments
  - both instruments are capable of swab in → STR profile out in less than 90 minutes without user intervention

- Exploring rapid DNA techniques including direct PCR and rapid PCR
  - STR profiles generated in <2 hours with standard lab equipment and rapid protocols
  - See ISHI 2012 poster available on STRBase “Rapid DNA Testing Approaches for Reference Samples”

Fastest results swab-to-profile (Identifiler): 57 minutes
Time Line Showing the Potential for DNA Deposition/Transfer

Opportunity for DNA Transfer from Perpetrator

Opportunity for Adventitious Transfer

Crime Event

Potential to “Contaminate”

Discovery

Laboratory analysis

Analysis completed

Investigators arrive, detect, and recover evidentiary material

Higher sensitivity techniques are most likely to pick up previously deposited (background) DNA

Adapted from Gill, P. (2002) *BioTechniques* 32(2): 366-385, Figure 5
DNA Mixture Interpretation
April 12, 2013 Webcast


• 8-hours of DNA mixture interpretation training

• 11 presentations from five different presenters
  – John Butler, Mike Coble, Robin Cotton, Bruce Heidebrecht, Charlotte Word

• 20 poll questions asked via SurveyMonkey (>600 participated)
  – Addressed additional questions sent via email or Twitter

• >1000 participants (almost entire U.S. represented and >10 countries)

• Available for viewing or download for at least six months (storage costs may limit longer-term storage)

Left to right:
Gladys Arrisueno (NIST, Twitter feed monitor & poll questions)
John Paul Jones (NIST, webcast organizer)
Mike Coble (NIST, presenter)
John Butler (NIST, presenter & organizer)
Charlotte Word (Consultant, presenter)
Robin Cotton (Boston University, presenter)
Bruce Heidebrecht (Maryland State Police Lab, presenter)
Forensic DNA Typing Textbooks Have Set the Standard for the Field

1st Edition
Jan 2001
335 pages
Language Editions
Chinese (2007)

2nd Edition
Feb 2005
688 pages

3rd Edition (3 volumes)
Sept 2009
520 pages
Aug 2011
704 pages
Fall 2014
(being written)
~500 pages

Advanced Topics in Forensic DNA Typing:
Methodology
Advanced Topics in Forensic DNA Typing:
Interpretation
NIST STRBase Website
Serving the Forensic DNA Community for >15 Years

Short Tandem Repeat DNA
Internet Database

NIST Standard Reference Database SRD 130

Serving the forensic DNA and human identity testing communities for over 10 years... These data are intended to benefit research and application of short tandem repeat DNA markers to human identity testing. The authors are solely responsible for the information herein.

Please Rate Our Products and Services: http://tsapps.nist.gov/MSDSurvey/default.aspx?ID=5&DB=130

This database has been accessed 458551 times since 10/02/97. (Courtesy courtesy www.digits.com - see disclaimer)

Created by John M. Butler
and Dennis J. Reeder (NIST Biochemical Science Division),
with invaluable help from Jan Redman, Christian Raubes and Michael Tsung
Site creators' curriculum vitae available using links above.

*Partial support for the design and maintenance of this website is being provided by The National Institute of Justice through the NIST Office of Law Enforcement Standards.*

General Information

- Purpose of STRBase/NAR 2001 Paper describing STRBase/Overview Presentation
- Publications and Presentations from NIST Human Identity Project Team
- NIJ-Funded Projects
- Training Materials
- Links to other web sites
- Glossary of commonly used terms

http://www.cstl.nist.gov/strbase/
Additional Resources

Beginning with the development or/and revision of its next draft guidance document(s), SWGDAM will make a “Draft for Comment” or other work product available for the purpose of receiving comments from the general public. This “Draft for Comment” solicitation will be open for a minimum of 60 days, usually through SWGDAM.org. SWGDAM will make all reasonable efforts to advise the forensic DNA community of the open comment period for a proposed guidance document or standard, guideline, best practice, study, or other recommendation and/or finding via as many avenues as possible to include posting notices through discipline-specific and related professional organizations. SWGDAM strongly encourages all interested parties to regularly monitor SWGDAM.org for the posting of such draft documents as well. All public comments received by SWGDAM will forwarded to the appropriate SWGDAM Committee for review and consideration as a part of its formal business practice for the development of the guidance documents or other work product.

The following information resources have been produced and reviewed by members of the Mixture Committee of SWGDAM and are available at

http://www.cstl.nist.gov/biotech/strbase/mixture/SWGDAM-mixture-info.htm
Thank you for your attention

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

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