Understanding Forensic DNA Testing for the Juvenile Law Community

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What is DNA?

- DNA is **deoxyribonucleic acid**
- Genetic blueprint for living organisms
- DNA encodes instructions needed for growth, survival, and reproduction
- DNA is found in nearly all cells of the body
How is DNA Transferred?

**Biological Materials**

- Sweat
- Blood
- Saliva
- Semen
- Vaginal secretions
- Hair
- Bone and teeth
- Bodily tissues
- Skin cells
Why is DNA important in criminal investigations?

- **Characteristics of DNA**
  - Unique
  - Transferrable
  - Persistent

- **DNA establishes connections**
  - Suspect to crime scene
  - Suspect to victim
  - Suspect to an object from the crime scene
  - Victim to the suspect or suspect’s possessions
  - Multiple scenes to one other
  - Human remains to family references
Why is DNA important in criminal investigations?

Criminal Activity → Evidence Collection → DNA Testing

- No known suspect
- Upload DNA Profile to Database
- Compare DNA Evidence to Suspect DNA Profile
- Suspect identified

CODIS (Combined DNA Index System)
HCIFS Forensic Genetics Laboratory (FGL)

Conducts DNA testing primarily for:

- Law enforcement agencies
- HCIFS medical examiners and forensic anthropologists to assist in positively identifying decedents

Cases submitted for DNA testing include:
- Crimes against person
  - Sexual assaults
  - Homicides
  - Robbery
- Crimes against property
  - Burglaries
  - Theft
Case Submissions by Offense Type (2018)

- Property: 467 (18%)
- Sexual Assault: 943 (37%)
- Assault/Robbery: 707 (27%)
- Homicide/Death Investigation: 316 (12%)
- Other: 151 (6%)

N=2,584
Cases Likely to Involve Juveniles

- **Sexual Assaults**
  - Juvenile complainants and/or juvenile suspects
  - Allegations of fondling or groping of small children
  - Sex trafficking and prostitution of teenage minors

- **Robberies and Assaults**
  - Often involve juvenile suspects
  - Car jackings
  - Robberies of businesses
    - Gas stations, cell phone storefronts, fast food restaurants

- **Homicides and Death Investigations**
  - May involve juvenile decedents and/or juvenile suspects
Common Types of Evidence

- Sexual assault evidence collection kit
- Fingerprints
- Underwear
- Finger nails
Common Types of Evidence
Common Types of Evidence
Common Types of Evidence
Serology and DNA Testing Workflow

Serology

DNA Operations

Interpretation
What Testing is Performed by the FGL?

- **Serology**
  - Examine physical evidence for presence of biological stains
  - Identify biological materials, including **blood and semen**
  - Select samples for DNA testing

- **DNA Operations**
  - Extract and purify nuclear DNA from evidence and reference samples
  - Generate and detect DNA profiles for further interpretation

- **Interpretation**
  - Evaluate DNA profiles for interpretation
  - Compare reference samples to evidence DNA profiles when possible
  - Select DNA profiles for entry into local and national databases
Serology Laboratory
Serology Laboratory
Serology Laboratory

- **Blood Testing**
  - Presumptive color test indicates presence of blood
  - Confirmed by immunological test
Serology Laboratory

- **Semen Testing**
  - Presumptive color test
  - Presumptive immunological test
  - Confirmed by microscopic identification of sperm cells
DNA Operations Laboratory
### DNA Testing Process

- **Extraction**
  - Enzymes and heat release DNA from cells

- **Purification**
  - Remains of the cell and substrate are removed from the sample extract, leaving purified DNA

- **Quantification**
  - Amount of DNA in the sample extract is determined
DNA Testing Process

- **Amplification**
  - Polymerase chain reaction (PCR)
  - Specific fragments of DNA are replicated and labeled with a fluorescent marker
  - 500 pg or less is needed to generate a DNA profile

- **Detection**
  - Replicated portions of DNA are separated by size and detected using an instrument that measures the amount of fluorescent light emitted by each DNA fragment
  - Light signals are translated into a graph display by the instrument
How many individuals may have contributed to the DNA profile?

Can the person of interest be a possible contributor?

What is the weight of the DNA evidence?
Evidence DNA Profile
Single Source

Can the suspect be a contributor to the evidence DNA profile?
Interpretation of DNA Evidence

**Complexity of DNA Evidence**

- **DNA from a single individual** is discrete, simple to interpret, and highly discriminating.

- **Mixtures of DNA are more complex, more difficult to interpret, and often less discriminating**
  - More complex due to shared or overlapping DNA characteristics
  - Difficult or impossible to unambiguously resolve each separate DNA profile
  - Assumed contributor in a mixture can assist in resolution of a foreign DNA profile
  - FGL typically does not interpret mixtures of more than three individuals
Interpretation of DNA Evidence

**Complexity of DNA Evidence**

- **Contributors to a mixture may be present at similar or very different levels**
  - “Major” or “predominant” contributors are present at high levels
  - “Minor” or “trace” contributors are present at low levels

- **Predominant contributors in a mixture are easier to interpret**
  - High levels of DNA mean less ambiguity in the DNA profile
  - Highly discriminating results still possible

- **Trace contributors can be very difficult to interpret conclusively**
  - Low levels of DNA cause “stochastic effects”
  - Partial or missing genetic information
  - DNA profile of trace contributors can be masked by predominant contributors
Can the suspect be a contributor to the evidence DNA profile?
Evidence DNA Profile

Three-Person Mixture

Suspect Reference

Can the suspect be a contributor to the evidence DNA profile?
Interpretation of DNA Evidence

Weight of Evidence

- Statistical weight of evidence conveys the strength of the association between evidence DNA profile and POI DNA profile

- Previous methods of calculating weight of evidence
  - Random match probability (RMP)
    - How often is the DNA profile expected to occur if we sampled at random from the population?
  - Combined probability of inclusion (CPI)
    - How often would we expect to find an individual who could be a contributor to the DNA mixture if we sampled at random from the population?

- Neither RMP nor CPI consider possibility of “stochastic effects”
  - More appropriate for high level DNA profiles and predominant contributors
  - Not well suited for low level DNA or very complex mixtures
  - Much information was lost due to inadequacies of RMP and CPI
Interpretation of DNA Evidence

**STRmix Software**

- Interpretation of data involving stochastic effects requires a method to systematically estimate probabilities of complex events

- STRmix software adopted by FGL in 2018
  - Developed by New Zealand and Australian forensic laboratories ESR and FSSA
  - First introduced in 2012
  - Currently in use by ~50 forensic laboratories in the US
Interpretation of DNA Evidence

**STRmix Software**

- How STRmix assists in DNA interpretation
  - STRmix estimates probable combinations of DNA profiles that would result in the evidence DNA profile
  - Numerous conceptual DNA profiles are built by the software and compared to the evidence DNA profile
  - Combines biological knowledge of DNA behavior and an iterative mathematical algorithm to model the evidence DNA profile
  - Weight of evidence in STRmix is expressed as a likelihood ratio
Interpretation of DNA Evidence

**Likelihood Ratios (LRs)**

- Comparison of probability of DNA evidence under two competing propositions
  - $H_p$ or $H_1$: What is the likelihood of observing DNA evidence if POI is a true contributor?
  - $H_d$ or $H_2$: What is the likelihood of observing DNA evidence if POI is not a true contributor?

- If POI DNA profile and evidence DNA profile have a strong fit, LR value will support $H_1$

- If POI DNA profile and evidence DNA profile do not have a strong fit, LR value will support $H_2$

- Equivalent support for $H_1$ and $H_2$ results in $LR = 1$
**Likelihood Ratios (LRs)**

\[
\frac{\Pr(H_p \mid E)}{\Pr(H_d \mid E)} = \frac{\Pr(E \mid H_p)}{\Pr(E \mid H_d)} \times \frac{\Pr(H_p)}{\Pr(H_d)}
\]

Posterior odds  Likelihood Prior odds ratio
Interpretation of DNA Evidence

**LR Support Occurs on a Spectrum**

- Magnitude of LR reflects relative degree of support
  - Driven by interpretability, quality, and complexity of DNA evidence

- LR values stratified into categories of verbal support
  - Limited support = 2-99
  - Moderate support = 100-9,999
  - Strong support = 10,000-999,999
  - Very strong support = >1,000,000

- Each category is based on frequency of adventitious support
  - Limited support ➔ higher occurrence of false support
  - Very strong support ➔ very low occurrence of false support
Reporting of a LR in Support of $H_1$

- “The DNA mixture is approximately 10 trillion times more likely to have originated from the Complainant and another individual than to have originated from two unknown individuals. This analysis provides very strong support for the proposition that the Complainant is a contributor to this DNA mixture.”

- “The DNA mixture is approximately 10 times more likely to have originated from the Suspect and another individual than to have originated from two unknown individuals. This analysis provides limited support for the proposition that the Suspect is a contributor to this DNA mixture.”
Reporting of a LR in Support of $H_2$

- “The DNA mixture is approximately $1,000$ times more likely to have originated from three unknown individuals than to have originated from the Suspect and two other individuals. This analysis provides moderate support for the proposition that the Suspect is excluded as a contributor to this DNA mixture.”

- “The DNA mixture is approximately $15,000$ times more likely to have originated from three unknown individuals than to have originated from the Complainant and two other individuals. This analysis provides strong support for the proposition that the Complainant is excluded as a contributor to this DNA mixture.”
Interpretation of DNA Evidence

**Limits of DNA Evidence**

- **Transfer**
  - DNA testing does not prove mode of transfer
    - Touch DNA or bodily fluid transfer?
    - Direct contact or secondary contact?

- **Time**
  - DNA testing does not prove when DNA was deposited on an item

- **Activity**
  - DNA testing does not prove the nature of activity that resulted in DNA deposition
    - Is DNA present on item due to criminal activity or regular use?

- **Order of Deposition**
  - DNA testing does not prove order that individuals deposited DNA
Thank you!

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