DNA: TrueAllele®
Statistical Analysis,
Probabilistic Genotyping

Indiana Prosecuting Attorneys Council
IPAC Winter Conference
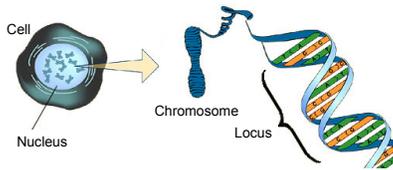**December, 2016**
Indianapolis, IN

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Cybergenetics

Cybergenetics © 2003-2016

DNA biology

Cell

Chromosome

Nucleus

Locus

Short tandem repeat
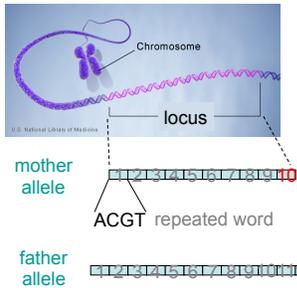
DNA locus paragraph

Take me out to the ball game
take me out with the crowd
buy me some peanuts and Cracker Jack
I don't care if I never get back
let me
root root root root root root root root root root
for the home team,
if they don't win, it's a shame
for it's one, two, three strikes, you're out
at the old ball game

23 volumes in cell's
DNA encyclopedia

"root" repeated 10 times, so
allele length is 10 repeats

DNA genotype

Chromosome

locus

U.S. National Library of Medicine

mother allele

1 2 3 4 5 6 7 8 9 10

ACGT repeated word

father allele

1 2 3 4 5 6 7 8 9 10 11 12
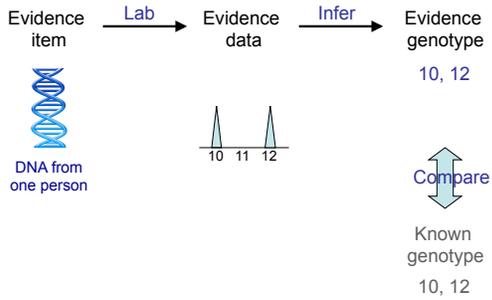
A genetic locus has two DNA sentences, one from each parent.

An allele is the number of repeated words.

A genotype at a locus is a pair of alleles.

10, 12

Many alleles allow for many many allele pairs. A person's genotype is relatively unique.

DNA evidence interpretation

Evidence item → Lab → Evidence data → Infer → Evidence genotype

10, 12

DNA from one person

10 11 12

Compare

Known genotype

10, 12

National Academy of Sciences

STRENGTHENING FORENSIC SCIENCE IN THE UNITED STATES
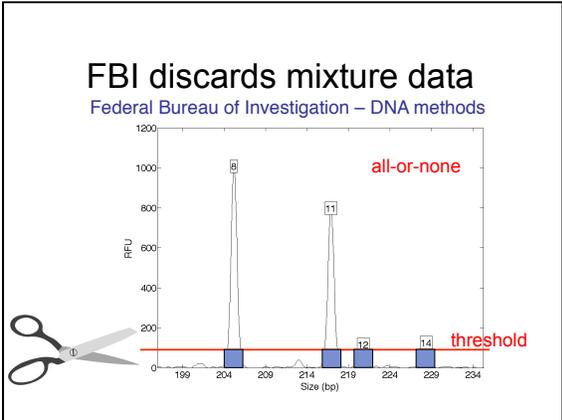A PATH FORWARD

"Strengthening Forensic Science: A Path Forward" (2009)

• Human examination bias
• Statistics & reporting
• Underlying scientific basis

Among existing forensic methods, only nuclear DNA …

However, ... there may be problems ... with how the DNA was ... interpreted, such as when there are **mixed samples**

FBI discards mixture data
Federal Bureau of Investigation – DNA methods

all-or-none

threshold

Unreliable DNA mixture statistics
National Institute of Standards and Technology (NIST)
2005 Commerce Department study
Two contributor mixture data, known victim

Some Differences in Reporting Statistics

		Case1		
LabID	Kits Used	Caucasians	African Americans	Hispanics
90	ProPlus/Cofiler	1.18E+15	2.13E+14	3.09E+15
34	ProPlus/Cofiler	2.40E+11	7.00E+08	9.80E+10
33	ProPlus/Cofiler	2.94E+08	1.12E+08	1.74E+09
6	ProPlus/Cofiler	40,000,000	3,500,000	280,000,000
9	ProPlus/Cofiler	1.14E+07	1.97E+07	1.54E+08
79	ProPlus/Cofiler	930,000	17,888	1,350,000
16	ProPlus/Cofiler	434,600	31,710	399,100

Remember that these labs are interpreting
the same MIX05 electropherograms

Most "inconclusive."
Some gave statistics:

213 trillion (14)

31 thousand (4)

Forensic DNA labs put on notice ten years ago

SWGDAM 2010 guidelines
Scientific Working Group on DNA Analysis Methods

FBI & NIST make crime labs
discard even more DNA data

stochastic threshold
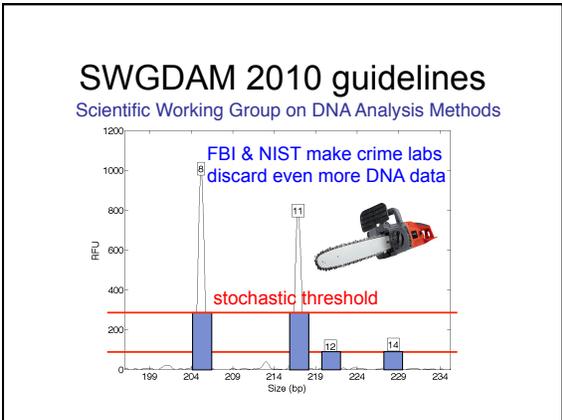
Falsely identify innocent people

MIX13 Case 5 Outcomes with Suspect C
(whose genotypes were <u>not present in the mixture</u>)

# Labs	Report Conclusions	Reasons given
6	Exclude Suspect C	detailed genotype checks (ID+); TrueAllele negative LR (ID+); assumed major/minor and suspects did not fit (ID+); 3 labs noted Penta E missing allele 15 (PP16HS)
3	Inconclusive with C only (A & B included)	All these labs used PP16HS
21	Inconclusive for A, B, and C	
70	<u>Include</u> **& provide CPI statistics**	*All over the road…*

<u>Range of CPI stats for Caucasian population</u>:
FBI allele frequencies: **1 in 9** to **1 in 344,000**

Mixture statistics shut down labs

"National accreditation board suspends all
DNA testing at D.C. crime lab"
𝕿𝖍𝖊 𝖂𝖆𝖘𝖍𝖎𝖓𝖌𝖙𝖔𝖓 𝕻𝖔𝖘𝖙 *April 27, 2015*
Did not comply with FBI standards

"New protocol leads to reviews of
'mixed DNA' evidence"
𝕿𝖍𝖊 𝕿𝖊𝖝𝖆𝖘 𝕿𝖗𝖎𝖇𝖚𝖓𝖊 *September 12, 2015*
24,468 lab tests affected

Mixture statistics block CODIS

CODIS handles only simple DNA
Most DNA evidence is mixtures
Less than 10% uploaded to CODIS

Investigative DNA failure
Can't solve crimes
Prosecutor, defense, police, innocence

Statistics lack scientific basis

Inherently invalid, no correct way to use method

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Biased DNA workflow

(1)
Choose data

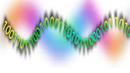**(3)**
Person decides

(2)
Calculate statistic

- Put people in the process
- To overcome software limits
- And introduce human bias

Unbiased DNA workflow

(1)
Enter all data

(2)
Calculate statistic

(3)
Math decides

- Keep people out of the process
- Because software is robust
- And eliminate human bias

TrueAllele computer technology

- Accurate. 34 validation studies, 7 published

- Objective. Workflow removes human bias

- Accepted. 10 Daubert/Frye challenges

- Transparent. Give math, software (4GB DVD)

- Neutral. Can statistically include or exclude

- Database. Upload all evidence genotypes

SWGDAM 2010 guidelines

3.2.2. If a stochastic threshold based on peak height is not used in the evaluation of DNA typing results, the laboratory must establish alternative criteria (e.g., quantitation values or use of a probabilistic genotype approach) for addressing potential stochastic amplification. The criteria must be supported by empirical data and internal validation and must be documented in the standard operating procedures.

Use TrueAllele® Casework for DNA mixture statistics

Validated genotyping method

Perlin MW, Sinelnikov A. An information gap in DNA evidence interpretation. *PLoS ONE*. 2009;4(12):e8327.

Ballantyne J, Hanson EK, Perlin MW. DNA mixture genotyping by probabilistic computer interpretation of binomially-sampled laser captured cell populations: Combining quantitative data for greater identification information. *Science & Justice*. 2013;53(2):103-114.

Perlin MW, Hornyak J, Sugimoto G, Miller K. TrueAllele® genotype identification on DNA mixtures containing up to five unknown contributors. *Journal of Forensic Sciences*. 2015;60(4):857-868.

Greenspoon SA, Schiermeier-Wood L, Jenkins BC. Establishing the limits of TrueAllele® Casework: a validation study. *Journal of Forensic Sciences*. 2015;60(5):1263-1276.

Perlin MW, Legler MM, Spencer CE, Smith JL, Allan WP, Belrose JL, Duceman BW. Validating TrueAllele® DNA mixture interpretation. *Journal of Forensic Sciences*. 2011;56(6):1430-1447.

Perlin MW, Belrose JL, Duceman BW. New York State TrueAllele® Casework validation study. *Journal of Forensic Sciences*. 2013;58(6):1458-1466.

Perlin MW, Dormer K, Hornyak J, Schiermeier-Wood L, Greenspoon S. TrueAllele® Casework on Virginia DNA mixture evidence: computer and manual interpretation in 72 reported criminal cases. *PLOS ONE*. 2014;(9)3:e92837.