

1

Opening the DNA Past with TrueAllele® Automation

Virginia Association of Commonwealth's Attorneys
Justice & Professionalism Meeting

Mark W Perlin, PhD, MD, PhD
Cybergenetics, Pittsburgh, PA



Cybergenetics

Cybergenetics © 2003-2021



2

Failed DNA data interpretation

The Washington Post

Virginia reevaluates DNA evidence in 375 cases

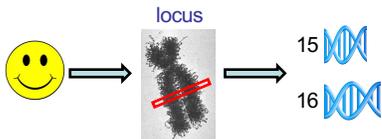
July 16, 2011

"Mixture cases are their own little nightmare," says William Vosburgh, director of the D.C. police's crime lab. "It gets really tricky in a hurry."

"If you show 10 colleagues a mixture, you will probably end up with 10 different answers"
Dr. Peter Gill, Human Identification E-Symposium, 2005

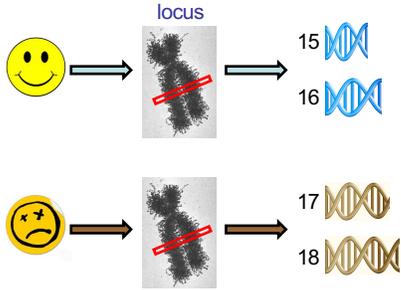
3

One person, one genotype



4

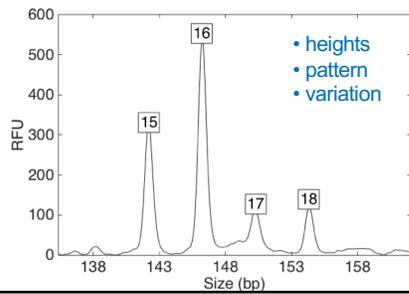
Two people, two genotypes



5

DNA mixture data

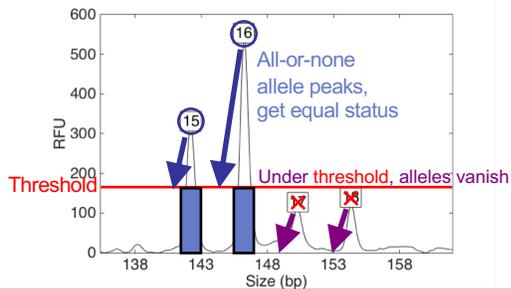
At a genetic locus, peak heights show allele amounts



6

Human review simplifies data

Arbitrary thresholds lose identification information



Unreliable DNA mixture statistics

NIST (US Commerce Department) study in 2005
Two contributor mixture data, known victim

Some Differences in Reporting Statistics

LabID	Kits Used	Caucasians	African-Americans	Hispanics
80	ProPlus/Cofiler	1.19E+15	2.13E+14	3.09E+15
34	ProPlus/Cofiler	2.40E+11	2.00E+10	9.80E+10
33	ProPlus/Cofiler	2.94E+08	1.12E+08	1.74E+09
6	ProPlus/Cofiler	40,000,000	3,600,000	280,000,000
9	ProPlus/Cofiler	1.14E+07	1.97E+07	1.54E+08
79	ProPlus/Cofiler	930,000	47,896	1,360,000
16	ProPlus/Cofiler	438,600	51,710	999,100

Remember that these labs are interpreting the same MIX05 electropherograms

When not "inconclusive":
213 trillion (14)
31 thousand (4)

Forensic science put on notice 15 years ago

Biased DNA workflow

- (1) Choose data
- (2) Person decides
- (3) Calculate statistic

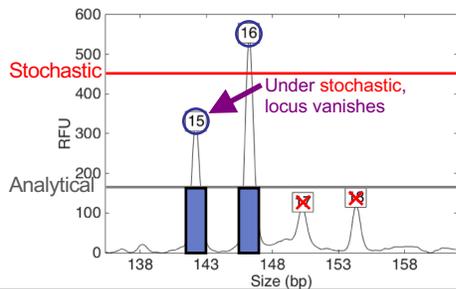


- Put people in the process
- To overcome software limits
- And introduce human bias



NIST: Stochastic threshold

Under threshold, discard the locus DNA test entirely



NIST: Thresholds misidentify

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

# 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	Include & provide CPI statistics	All over the road...

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

Statistics lack scientific basis

CPI has an analyst count up "included" loci

J Pathol Inform

Research Article
Inclusion probability for DNA mixtures is a subjective one-sided match statistic unrelated to identification information
Mark William Perlin¹

¹Cybergenetics, Pittsburgh, USA
E-mail: *Dr. Mark William Perlin - perlin@cygn.com
*Corresponding author

Received 14 July 2015 Accepted 21 September 2015 Published 28 October 2015

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

Human mixture interpretation

- Incomplete. Discard data, apply thresholds
- Inaccurate. Disagrees with true information
- Subjective. Workflow introduces human bias
- Inoperative. Hundreds of thousands of cases
- Opaque. Choices use only some of the data
- Biased. Can only include – or give no answer

Inconclusive

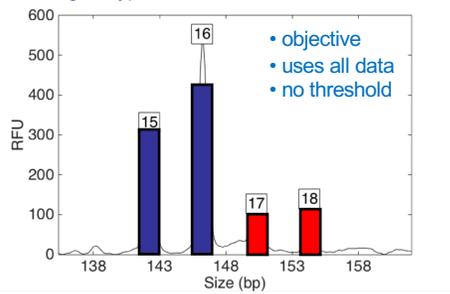
TrueAllele® computer solution

- Complete. Use all data, no thresholds
- Accurate. 42 validation studies, 8 published
- Objective. Workflow removes human bias
- Accepted. Reported in 45 states, WTC, labs
- Transparent. Give math, software (4GB DVD)
- Neutral. Can statistically include or exclude

Informative

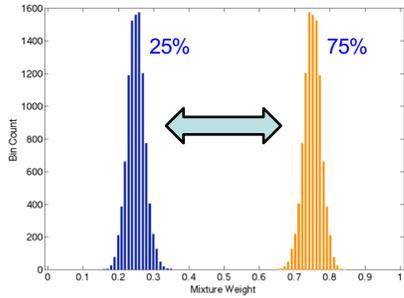
TrueAllele explains the data

Considers all genotype combinations, doesn't see the "suspect"



TrueAllele unmixes mixtures

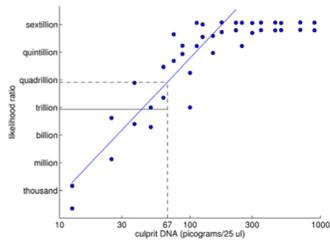
Separates the mixture data into contributor components



TrueAllele predictability

An Information Gap in DNA Evidence Interpretation

Mark W. Perlin^{1*}, Alexander Sinenikov² 2009



Daubert reliability prongs

1. Is testing possible? Has it been tested?
2. Error rates & standards
3. Subjected to peer review and publication
4. General acceptance in relevant scientific community

19

TrueAllele testing

TrueAllele Casework on Virginia DNA Mixture Evidence: Computer and Manual Interpretation in 72 Reported Criminal Cases

Mark W. Perlin^{1*}, Kiersten Dormer¹, Jennifer Hornyak¹, Lisa Schiermeier-Wood², Susan Greenspoon²

1 Cybergenetics, Pittsburgh, Pennsylvania, United States of America, 2 Department of Forensic Science, Richmond, Virginia, United States of America

 2014

Validation axes

- sensitive
- specific
- reproducible

20

TrueAllele (& human) error rates

TrueAllele specificity (million samples)
From noncontributor distribution, for LR > 100:

Error rate = 1 in 1,000,000 (0.0001)%

CPI – analytical threshold
5 false positives in 81 comparisons
Error rate = 5 in 81 (6%)

mCPI – stochastic threshold
17 inconclusive results
1 false positive in 53 comparisons
Error rate = 1 in 53 (2%)

21

TrueAllele peer review

Perlin MW, Sineelnikov 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.

Bauer DW, Butt N, Hornyak JM, Perlin MW. Validating TrueAllele® interpretation of DNA mixtures containing up to ten unknown contributors. *Journal of Forensic Sciences*. 2020; 65(2):380-398.

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.

22

JOURNAL OF FORENSIC SCIENCES

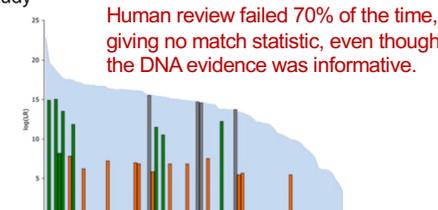
J. Forensic Sci., November 2013, Vol. 58, No. 6
doi: 10.1111/1556-4029.12223
Available online at: onlinelibrary.wiley.com

PAPER

CRIMINALISTICS

Mark W. Perlin,¹ M.D., Ph.D.; Jamie L. Belrose,² M.S.; and Barry W. Duceman,³ Ph.D.

New York State TrueAllele® Casework Validation Study*



Human review failed 70% of the time, giving no match statistic, even though the DNA evidence was informative.

23

TrueAllele general acceptance

Invented math & algorithms	25 years
Developed computer systems	20 years
Support users and workflow	10 laboratories
Routinely used in casework	10 crime labs
Validate system reliability	42 studies
Educate the community	100 talks
Train or certify analysts	400 students
Admissibility challenges	29 rulings, 14 states
Testify about LR results	102 trials
Educate lawyers and public	1,000 people
Make the ideas understandable	1,000 cases, 45 states

24

29 US admissibility rulings

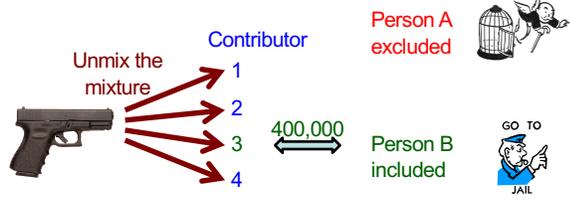
Commonwealth of Pennsylvania v Kevin Foley (admitted, 2009; appellate precedent, 2012)
 People of California v Dupree Langston (admitted, 2013)
 Commonwealth of Virginia v Matthew Brady (admitted, 2013)
 State of Ohio v Maurice Shaw (admitted, 2014)
 State of Louisiana v Chattley Chesterfield & Samuel Nicolas (admitted, 2014)
 People of New York v John Wakefield (admitted, 2015; appellate precedent, 2019)
 State of South Carolina v Jaquard Aiken (admitted, 2015)
 Commonwealth of Massachusetts v Heidi Bartlett (admitted, 2016)
 State of Indiana v Dugniqio Forest (admitted, 2016)
 State of Indiana v Malcolm Wade (admitted, 2016)
 State of Washington v Emanuel Fair (admitted, 2017)
 State of Louisiana v Harold Houston (admitted, 2017)
 State of Indiana v Randal Coalter (admitted, 2017)
 State of Nebraska v Charles Simmer (admitted, 2018; appellate precedent, 2019)
 State of Indiana v Vaylen Glazebrook (admitted, 2018)
 State of Ohio v David Mathis (admitted, 2018)
 State of Florida v Lajavian Daniels (admitted, 2018)
 State of Tennessee v Demontez Watkins (admitted, 2018)
 State of Georgia v Thaddus Nundra (admitted, 2019)
 State of Georgia v Monte Baugh & Thaddeus Howell (admitted, 2019)
 State of Louisiana v Kyle Russ (admitted, 2019)
 People of New York v Casey Wilson (admitted, 2019)
 State of Georgia v Alexander Battle (admitted, 2019)
 United States v Lenard Gibbs (admitted, 2019)
 State of Georgia v Guy Sewell (admitted, 2019)
 State of Georgia v Adedojah Bah (admitted, 2019)
 State of Georgia v Nathaniel Day (admitted, 2019)
 State of Tennessee v Abdullah Powell (admitted, 2021)
 United States v Curtis Johnson (admitted, 2021)

Virginia v. Black

Item	Description	David Black	Bonnie Black	BettyAnn Armstrong	Craig Black	Eleanora Black
08	Baseball hat velcro strap	32.5 quintillion	16.1 billion		1/1.83 thousand	1/62.6
94	Master bedroom light switch	364 million	8.14 million			
95	Master bathroom light switch	1/19.5	554 million			3.63 million

Pennsylvania v. McBride

Cybergenetics TrueAllele reanalysis
Match statistics provide information



Indiana v. Pinkins & Glenn

1989 – 5 men raped an Indiana woman
Darryl Pinkins and 2 others misidentified
1991 – wrongfully convicted, 65 year sentence

2001 – DNA mixture evidence
2 contributors found, not the accused
but 5 were needed, post-conviction relief denied

TrueAllele Pinkins findings

- 1. compared *evidence with evidence*
- 2. calculated *exclusionary match statistics*
- 3. revealed 5% *minor mixture contributor*
- 4. *jointly analyzed DNA mixture data*
- 5. showed three perpetrators were *brothers*

found **5 unidentified genotypes**,
defendants not linked to the crime

Pinkins exonerated



Georgia v. Gates

1977, Johnnie Lee Gates admits to murder,
is convicted and sentenced to death

- Mental deficiency
- Brought to crime scene for confession, touched items
- Prosecutor struck all black jurors in several capital cases
- Two newly discovered ligatures – four-contributor mixtures
- Lab's human review finds DNA mixtures "inconclusive"

TrueAllele match statistics

Item	Description	Johnny Lee Gates
76C2573-032	robe belt side 1 swab	one in 1.5 million
76C2573-033	robe belt side 2 swab	one in 134 thousand
76C2573-034	front of black tie swab	one in 4.33 million
76C2573-035	back of black tie swab	one in 963 million
76C2573-042	robe belt M-vac filter	one in 902 trillion
76C2573-044	black tie M-vac filter	one in 825 billion

DNA doesn't "fall off"

New trial, released from prison

Georgia Innocence Project
May 9 at 1:23pm · 🌐

Like Page

For an update on Johnny Lee Gates' Extraordinary Motion For New Trial, check out the Columbus Ledger Enquirer with video at the top and photos at the bottom.



He's been a convicted killer for 40 years. Columbus court will decide if racism put him in prison

Johnny Lee Gates was convicted in the Nov. 30, 1976, rape and murder of Katharina Wright, 19, found bound and shot in the head in the Broadway...

Texas v. Grant

2011, Houston lab fails to interpret DNA mixture from fingernails of 2010 murder victim; crime lab testifies DNA is inconclusive

Lydell Grant convicted of murder and receives a life sentence

2019, Innocence Project of Texas sends DNA data to Cybergeneics

Match statistics

Item	Description	12.1.1	13.1
		Aaron Scheerhoorn	Lydell Grant
12.2.1.1	right hand fingernails	18.61	-12.93

Non-matching evidence genotype

TrueAllele also inferred a **non-matching evidence genotype** from the right-hand fingernails. The probabilistic genotype of this unknown contributor has an expected LR match statistic of 18.2 trillion.

A **CODIS-searchable allele list** was derived from the probabilistic genotype at a 90% credible level.

Should additional reference genotypes become available, **Cybergenetics can compare** them with the probabilistic genotype to calculate DNA match statistics.

Unprecedented CODIS search

- In 2019, TrueAllele crime lab searches CODIS
- Search finds the unknown fingernail person
- Confronted in Georgia, killer confesses to crime

Government was not pleased by our use of better science to reveal the truth in this case

Grant released, exonerated

- Grant eventually released from prison
- In 2021, Lydell Grant finally exonerated



New York v. Robinson

Crime laboratory reported “inconclusive” results

Prosecutor: from “inconclusive” to “guilty”

TrueAllele	Victim	Defendant
fingernail	57.2 septillion	one in 1.18 trillion

Crime laboratory later reported a weak exclusion

Pennsylvania v. Huber

Item	Description	Melissa Zuk	Derek Schindler	Joshua Huber
13E	Living room wall bloodstain	1 in 160 million	1 in 36 thousand	11.6 quintillion
29A	Schindler's right hand fingernails		1.37 quintillion	53.8 thousand
1603461-13A	Left hand fingernails of Melissa Zuk	17.4 billion		3.35 thousand

California v. Lopez

Man accused of rape and murder of girlfriend's toddler son

- Facing the death penalty, or life in prison.
- The child was 2 years and 10 months old.
- There were bruises to his face, genitals, and rectum.
- An autopsy showed brain swelling, skull fracture, cheek bruises, and asphyxia.
- A rectal swab from the boy showed semen.
- The swab matched the defendant's DNA.

County Crime Lab (1 of 3)

Item	Sister	Brother	Mother	Defendant	Brother	Victim	Person in	Person in	Female in	Person in	Person in	Male in
	2 REF	3 REF	23	24	25	35	21A-EC	21D-SP	45D-EC	7B-4-SP	8C-SP	7-S1
5A												
7-S1												
7-S2												
7A-2-EC												
7B-1-SP												
7B-2-SP												
7B-3-EC												
7B-4-EC												
7B-4-SP												
7B-5-EC												
8A												
8B-EC												
8B-SP												
8C-EC												
8C-SP												
8D-EC												
8D-SP												
8E-EC												
8E-SP												
8F-EC												
8G-EC												
8G-SP												
8H-EC												
8H-SP												
8I-EC												

Cybergenetics TrueAllele (1 of 3)⁴²

Item	Sister	Brother	Mother	Defendant	Brother	Victim	Person in	Person in	Female in	Person in	Person in	Male in
	2 REF	3 REF	23	24	25	35	21A-EC	21D-SP	45D-EC	7B-4-SP	8C-SP	7-S1
5A												
7-S1												
7-S2												
7A-2-EC												
7B-1-SP												
7B-2-SP												
7B-3-EC												
7B-4-EC												
7B-4-SP												
7B-5-EC												
8A												
8B-EC												
8B-SP												
8C-EC												
8C-SP												
8D-EC												
8D-SP												
8E-EC												
8E-SP												
8F-EC												
8G-EC												
8G-SP												
8H-EC												
8H-SP												
8I-EC												

Two strange puzzles

- **Where's Mom's DNA?**

Lots of different people left lots of DNA, but the primary caretaker left none.

- **Rectal DNA conflict**

Why was the defendant's DNA found in the **initial** hospital rectal swabs (Item 16), but **not later** at autopsy (Item 39)?

Final verdict

The prosecution was target-driven.
The defense was nontarget-driven.
Cybergenetics experts educated the jury.

The nontargeted scenario better explained the evidence.
The jury acquitted the defendant of all charges.

The county no longer seeks the death penalty.

Human review has failed

- Inaccurate, unvalidated, biased
- Inconclusive, uninformative
- Unreliable, irrelevant, fails prongs

Inclusion probability for DNA mixtures is a subjective one-sided match statistic unrelated to identification information (JPI)

Hundreds of thousands of past cases have **informative DNA evidence**, but:

- meaningless DNA statistics
- **no DNA statistics**
- no exclusionary statistics

TrueAllele automation succeeds

- Accurate, validated, unbiased
- Gives stats & error rates, informative
- Reliable, relevant, passes prongs

TrueAllele mixture interpretation
is a reliable method (PLoS):
objective, sensitive, specific, reproducible, accurate

The **automated** TrueAllele computer process
can be run on **large batches** of DNA cases
with minimal human effort

How to open the past

Commonwealth's Attorneys can:

1. Request electronic DNA data from state lab
2. Send the data to Cybergeneitics for processing
3. Get back accurate identification information

TrueAllele can report all DNA match results
(without cutoffs or limits, for or against)
because it gives exact *error rates* on every match statistic

Better science leads to better justice
