Mining the Mixture:
A DNA Analyst Explains

New York State
Judicial Summer Seminars
The New York State Judicial Institute
June, 2019
Rye Brook, NY

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Pittsburgh, PA

Cybergenetics

JUSTICE
THROUGH
SCIENCE

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Dr. Mark W. Perlin

Curriculum Vitae

Mark W. Perlin, PhD, MD, PhD
DNA evidence interpretation and the likelihood ratio

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Positions Held

Cybergenetics, Corp.	chief scientist & executive	1996-present	Comput. Bioscience
Carnegie Mellon University	senior research scientist	1995-1996	Computer Science
Carnegie Mellon University	research computer scientist	1992-1995	Computer Science
Carnegie Mellon University	research associate	1988-1992	Computer Science
Carnegie Mellon University	visiting researcher	1986-1988	Computer Science
Pittsburgh NMR Institute	research scientist	1985-1986	Comput. Radiology
Mercy Hospital, Pittsburgh, PA	transitional resident	1984-1985	Medicine/Radiology
IBM/Watson Research Yorktown, NY	post-doctoral fellow	1984-1984	Mathematics

Education and Training

Carnegie Mellon University, Pittsburgh, PA	Ph.D.	1991	Computer Science
The University of Chicago Pritzker School of Medicine	M.D.	1984	Medicine
City University of New York Graduate School	Ph.D.	1982	Mathematics
Harpur College/SUNY, Binghamton, NY	B.A.	1977	Chemistry

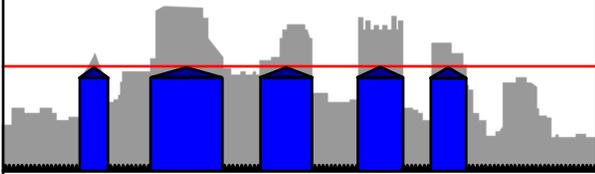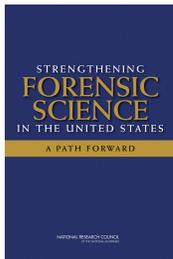
DNA mixture

eye of newt   toe of frog

Double, double toil and trouble

Crime lab analysts
simplify data to interpret mixtures

Applying a threshold unreliably gives
the same heights & vacant lots

National Academy of Sciences

STRENGTHENING
FORENSIC
SCIENCE
IN THE UNITED STATES
A PATH FORWARD

NATIONAL RESEARCH COUNCIL

Trouble in River
City
2009

Where's the
"science" in
Forensic Science?

When DNA Is Not a Gold Standard: Failing to Interpret Mixture Evidence

F orensic science connects evidence through shared characteristics. Markings on a bullet can appear to match grooves in the barrel of a gun. Latent fingerprints left at a crime scene may be similar to ridge patterns on a suspect's hand. Tracks in the mud may mirror the treads of a shoe or tire. Police gather forensic evidence to help build a case, and police dramas on television convey the myth of forensic infallibility through the "CSI" effect.[1]

In 2009, the National Academy of Sciences (NAS) published its seminal report titled *Strengthening Forensic Science in the United States*.[2] The NAS report reviewed many forensic modalities and questioned their scientific validity. The interpretation of forensic data is often unreliable. Match statistics are needed to gauge the strength of match between items, relative to coincidence. But forensic statistics are typically absent or incorrect. Human bias can skew answers by unconsciously selecting favorable data, using knowledge about defendant characteristics, or by trying to please stakeholders who have a desired criminal justice outcome.

Deoxyribonucleic acid (DNA) evidence seems immune to such criticism, long serving as a gold standard for other forensic disciplines. Abundant DNA from one person produces pristine data signals. Interpreting these clear signals yields an unambiguous genetic type ("genotype"). Comparing definite genotypes, relative to a random person, yields a reliable match statistic that numerically conveys the probative force of DNA evidence. But most crime scene DNA is now a mixture of two or more people, with good data but less certain interpretation. As the NAS report noted, there may be problems with how the DNA was interpreted, such as when there are mixed samples.

Simplistic interpretation of DNA mixture data often fails to produce an accurate match statistic or give any answer at all. While the limitations and liabilities of unscientific DNA mixture interpretation were recognized early on,[3] only recently has this profound forensic failure come to the fore. Crime laboratories in Austin, Texas, and Washington, D.C., have been shuttered in large part because of failed DNA mixture interpretation.[4] Virginia re-evaluated DNA match statistics for mixture evidence in hundreds of cases.[5] Texas is reviewing 24,000 criminal cases for flawed interpretation of DNA mixture evidence.[6] The New York State Police (NYSP) has suppressed reliable DNA mixture interpretation methods that could expose its crime laboratory's mistakes in thousands of cases.[7] These numbers extrapolate to hundreds of thousands of mixture items throughout the United States, and the national press has taken notice.[8]

This failure of forensic DNA interpretation is of broad concern. Pervasive errors in DNA match statistics undermine public trust in science and erode confidence in government agencies that misuse science to obtain convictions. A failed DNA gold standard portends little hope for fledgling forensic fields. Perhaps the greatest loss is true justice in a free society. Misinterpreting DNA evidence causes injustice for defendants denied potentially exculpa-

BY MARK W. PERLIN, PH.D., M.D., PH.D.

Unreliable DNA mixture statistics

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

Some Differences in Reporting Statistics

LabID	Kits Used	Caucasians	Case1 African Americans	Hispanics
90	ProPlus/Cofiler	1.18E+15	2.13E+14	3.09E+15
34	ProPlus/Cofiler	2.40E+11		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		1,350,000
16	ProPlus/Cofiler	434,600	31,710	399,100

Remember that these labs are interpreting the same MIX05 electropherograms

When not "inconclusive":

213 trillion (14)

31 thousand (4)

Forensic DNA labs put on notice 15 years ago

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Falsely identify innocent people

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**

Biased DNA workflow

(1)
Choose data

(3)
Person decides

IN
OUT

(2)
Calculate statistic
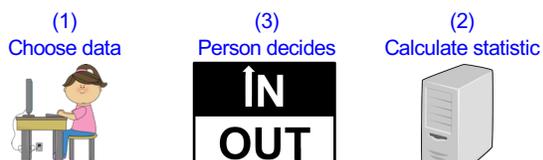
• Put people in the process
• To overcome software limits
• And introduce human bias

Good evidence, wrong answers

Misled courts for 15 years on most DNA mixtures

Human mixture interpretation

- Inaccurate. Disagrees with true information

- Subjective. Workflow introduces human bias

- Widespread. Hundreds of thousands of cases

- Opaque. Choices use only some of the data

- Biased. Can only include – or give no answer

Inconclusive

MathWorks Newsletters: Technical Article. 2013:1-4.

Cybergenetics TrueAllele Technology Enables Objective Analysis of Previously Unusable DNA Evidence

By Dr. Mark W. Perlin, Cybergenetics

Dr. John Yelenic was found murdered in his Blairsville, Pennsylvania home in 2006. His fingernails contained largely his own DNA, but also a small amount of DNA from someone else—possibly deposited when he scratched his assailant in self-defense. Indeed, this minor component of the DNA mixture tied suspect Kevin Foley to the crime, with a match statistic a forensic expert said was 13,000.

DNA mixture data can be hard for human experts to interpret. Their laboratory protocols simplify such data and typically understate the match number. Foley's defense attorney said that the fingernail evidence did not rule out other suspects, since there was a one in 13,000 chance that the DNA came from someone other than his client.

Human expert evaluation of DNA evidence can be challenging, even on simpler samples. The analyst performing the examination requires significant training, and the review process is slow and tedious. Human interpretation methods may not eliminate natural examination bias. Heuristic approaches that truncate data can rob the evidence of much probative value.

Today, most DNA samples are not simple. They can contain little DNA, exhibit degradation, or mix together the DNA of several people. These factors compound the data analysis difficulties. Sometimes expert analysts are unable to draw a conclusion, despite expending considerable effort. As a result, valuable evidence to convict the guilty or exonerate the innocent becomes unusable in court.

TrueAllele® technology

VUIer™ Software

Parallel Processing
& Database Servers

Fully automated
Uses all the data
No thresholds or choices
No calibration – case data

No human information from mixture

Crime laboratory DNA report
Crime lab user fee: $5,000

Conclusions:

Item 1 – Swab of textured areas from a handgun

The data indicates that DNA from four (4) or more contributors was obtained from the swab of the handgun. Due to the complexity of the data, no conclusions can be made regarding persons A and B as possible contributors to this mixture.

Probabilistic genotyping

Cybergenetics TrueAllele report
Match statistics provide information

Unmix the mixture

Contributor
1
2
400,000
3
4
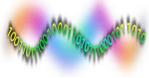
Person A excluded

Person B included

TrueAllele® computer technology

- Accurate. 35+ validation studies, 7 published

- Objective. Workflow removes human bias

- Accepted. Reported in 43 states, used by labs

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

- Neutral. Can statistically include or exclude

Informative

Objective workflow

(1)
Enter all data

(2)
Calculate statistic

(3)
Math decides

IN
OUT

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

How is TrueAllele used?

- Prosecution
- Defense
- Investigation
- Post-conviction
- Mass disaster
- Touch DNA
- Complex mixtures
- Kinship, paternity
- DNA database
- Preventing crime

World Trade Center

18,000
victim remains

2,700
missing people

match

September 11, 2001 – New York City

Pennsylvania v. Kevin Foley

Apr 2006: Blairsville Dentist John Yelenic murdered

Nov 2007: Trooper Kevin Foley charged with crime

February 2008: Defense questions 13,000 DNA match score

March 2009: Jury hears 189,000,000,000 TrueAllele statistic

Pennsylvania v. Joshua 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

Virginia v. David 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

Maryland v. Nelson Clifford

11% contributor 1	82% contributor 2	7% contributor 3
Victim	Elimination	Nelson Clifford
23.1 thousand	32 trillion	182 thousand

match statistic
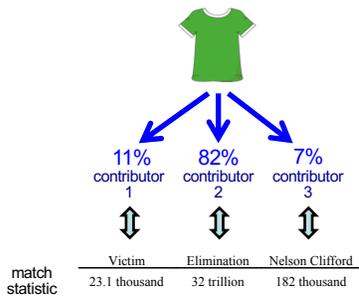
California v. Billy Ray Johnson

8 mixture items vs. 5 victims + 1 suspect

Description	Item	C0428X	C0459X	C0460X	C0475X	C0820X	C1147X
Purse strap	C0431X	18.73	5.91				1.63
Phone cord	C0432X						4.53
Phone	C0984X	18.74	2.06				4.62
Stain on pants	C0801S				16.39		6.24
Stain from pants	C0802S				17.42		6.73
Back of shirt	C0806S			6.69	17.43		8.86
Bathtub handle	C0929X					5.80	2.74
Zip tie	C0937X					4.55	20.32

BRJ

DNA match statistics
corroborate victim statements

Wolfe sisters homicide

On February 6, 2014, Susan Wolfe (44)
and her younger sister Sarah (38, left)
were killed in their East Liberty home in Pittsburgh.

Pennsylvania v. Allen Wade

Thresholds failed to interpret most DNA mixtures

Hat	No conclusions
Cup	Insufficient data
Fingernails	Contamination, insufficient data
Gear shift	Insufficient data
Seat lever	Cannot be excluded
Knit hat	Insufficient data
Sock	Too complex, no conclusions

Pennsylvania v. Allen Wade

The crime lab reported 5 DNA mixture matches
TrueAllele found 17 matches on the same data

Hat	65.3 thousand	Allen Wade
Cup	20.5 thousand	Susan Wolfe
Fingernails	6.06 trillion	Allen Wade
Gear shift	9.37 million	Sarah Wolfe
Seat lever	385 billion	Sarah Wolfe
Knit hat	25.7 thousand	Allen Wade
Sock	300	Sarah Wolfe

Reported DNA match statistics

A match between the right fingernails
and Allen Wade is:

6.06 trillion times more probable than
a coincidental match to an unrelated Black person

32.5 trillion times more probable than
a coincidental match to an unrelated Caucasian person

8 trillion times more probable than
a coincidental match to an unrelated Hispanic person

Allen Wade Found Guilty On All Counts In East Liberty Sisters' Slaying

CBS News, May 23, 2016

PITTSBURGH (KDKA/AP)
- A man accused of killing two sisters who lived next door to him in East Liberty has been found guilty on all counts
- Allen Wade was accused of shooting Sarah and Susan Wolfe after they returned from work, apparently to steal bank card
- On Monday morning, a jury found Wade guilty of first-degree murder, robbery, burglary and theft by unlawful taking

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Pennsylvania v. Allen Wade

Thresholds failed to interpret DNA mixture
TrueAllele succeeded on the same data

A hat left from a burglary of the Wolfe sister's home
six weeks before the murder matched
Allen Wade with a 65.3 thousand statistic

Preventable Crime

feature

Hidden DNA Evidence: Exonerating the Innocent

Mark W. Perlin, Ph.D., M.D.

In 1989, northwest Indiana was plagued by bump-and-rob road crimes of escalating violence. In the darkness of a cold December night, a woman's car was rear ended on highway I-65. Upon exiting her car, she was dragged into another vehicle, then stripped and raped by five strangers. The men left her in her car, draped by green coveralls. The same night, coworkers Darryl Pinkins, Roosevelt Glenn and William Durden had engine trouble along that highway. They parked their car on the roadside, and went to get help and motor oil. On their return, they found shattered side windows and their work coveralls gone.

Traced to their employer by the crime scene coveralls, Pinkins, Glenn and Durden, along with two other coworkers, were arrested for the I-65 bump-and-rape. RFLP testing of semen DNA left on the victim's jacket and sweater excluded the defendants. But nonspecific serology testing, along with faulty hair evidence and tainted eye witness identification, led to Pinkins' and Glenn's wrongful convictions. Pinkins was found guilty of all charges in May 1991, and sentenced to 65 years in prison. Glenn's 1992 jury deadlocked, but on retrial he was convicted of rape in 1993, and sentenced to 36 years. Despite their incarceration, the bump-and-rob and rape crimes continued unabated. The men's exoneration by science would not happen soon.

Darryl Pinkins imprisoned

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 released

April 25, 2016

CBS News *48 Hours*
"Guilty Until Proven Innocent"

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feature

Suspect-centric Bias in DNA Mixture Interpretation

Mark W. Perlin, Ph.D., M.D.

Bias abounds in criminal justice. Predictive policing can bake bias into software, reflecting and reinforcing prior beliefs. Bail-risk computer programs may entrench pre-trial detention disparity. Human judgment pervades the process. Prosecutor and defender alike passionately argue their client's case, drawing opposite conclusions from identical facts.

Science is above the fray. Objective data suggest forensic match between crime scene and suspect. Statistical data analysis yields incontrovertible numbers for the strength of match. Cold DNA facts are presented as confirmed theories in court.

But what if DNA analysts could pick and choose their data? Or adjust software parameters to suit their theories? Changing data and parameters will alter forensic match results. Quantitatively, subjective manipulation can artificially inflate match strength. Qualitatively, some DNA evidence that excludes a suspect may be statistically twisted to include him.

Suspect-centric bias has long plagued forensic science. The mythic infallibility of fingerprint analysis was shattered when the FBI misidentified Brandon Mayfield in the Madrid bombing case. Confirmation bias just puts a number to a foregone match conclusion. Suspect-centric thought twists forensic facts to suit prosecution theories.

Subjectivity and bias in forensic DNA mixture interpretation

Itiel E. Dror [a,b,*], Greg Hampikian [c]

[a] Institute of Cognitive Neuroscience, University College London (UCL), London, UK
[b] Cognitive Consultants International (CCI), London, UK
[c] Departments of Biology and Criminal Justice, Boise State University, USA

ABSTRACT

The objectivity of forensic science decision making has received increased attention and scrutiny. However, there are only a few published studies experimentally addressing the potential for contextual bias. Because of the esteem of DNA evidence, it is important to study and assess the impact of subjectivity and bias on DNA mixture interpretation. The study reported here presents empirical data suggesting that DNA mixture interpretation is subjective. When 17 North American expert DNA examiners were asked for their interpretation of data from an adjudicated criminal case in that jurisdiction, they produced inconsistent interpretations. Furthermore, the majority of 'context free' experts disagreed with the laboratory's pre-trial conclusions, suggesting that the extraneous context of the criminal case may have influenced the interpretation of the DNA evidence, thereby showing a biasing effect of contextual information in DNA mixture interpretation.

New York v. Nick Hillary

Garrett Phillips (12)
Died from strangulation
October 24, 2011

Oral "Nick" Hillary
Arrested for murder
May 15, 2013

DNA evidence and results

150 biological **evidence** items,
with focus on DNA under victim's fingernails

Other software finds match statistic of ten million,
connecting victim's fingernails to defendant's DNA

Unknown **minor** contributor is 0.4% or 1:250,
a very, very small amount of DNA

F.R.E. Rule 702

a. Sufficient data

b. Reliable method

c. Reliably apply method to data

Sufficient data

Mixture
• ratio is 1:250
• less than 1 cell

Fingernail data show
low mixture amount
& low peak heights
for minor contributor

Peak height
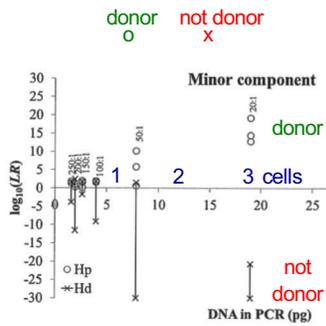• 30 to 70 rfu

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Reliable method

Mixture (validation)
• ratio of 1:25
• many cells

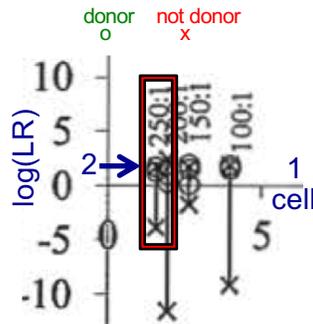**Peak threshold**
• 30 rfu in study

Reliably apply method to data

Mixture (case)
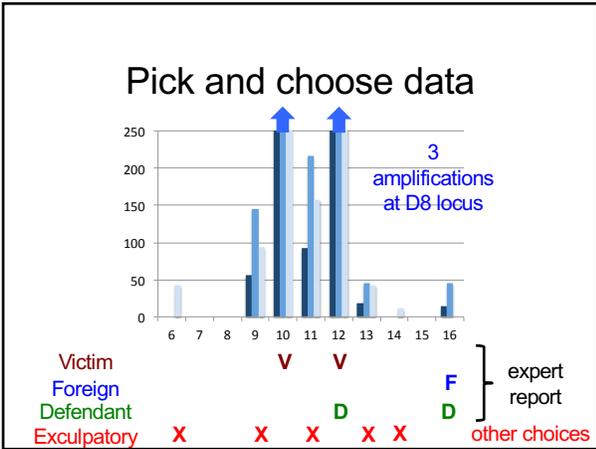• ratio is 1:250
• less than 1 cell

Peak threshold
• 30 rfu in study
• 50 rfu in case

Choose threshold level

		LR	
RFU	**Data choices**	**All stutters**	
80	0	9	
70	30	51	
60	250	1,660	
50	15,500,000	69,200	include
40	0	0	exclude
30	0	0	

Different choices, different answers
Software does not agree with itself

Pick and choose data

3 amplifications at D8 locus

		6	7	8	9	10	11	12	13	14	15	16
Victim						V		V				
Foreign												F
Defendant							D				D
Exculpatory	X			X		X		X X		

expert report

other choices

STRmix™ results precluded

Dr. Buckleton conceded at the hearing that no internal validation studies were performed by the New York State Police crime lab for the use of STRmix on casework samples developed at the lab.

As a result Dr. Buckleton was forced to **pick and choose** data from different "reliable sources" and input parameters into the program in such a way that he believed the system would tolerate.

ORDERED that the defendant's motion to preclude the prosecution from calling an expert witness to testify on their direct case regarding any conclusion reached by the use of STRmix is granted as the prosecution cannot lay a foundation for the introduction of evidence that had not been internally validated.

Hillary acquitted

The New York Times

Oral Nicholas Hillary Acquitted in Potsdam Boy's Killing

September 28, 2016

"Who Killed Garrett Phillips?"
HBO on July 9, 2019