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Probabilistic genotyping of
evidentiary DNA typing results:
TrueAllele® admissibility

Federal Bureau of Investigation
& National Institute of Justice
June, 2019

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

Cybergenetics

Cybergenetics © 2003-2019

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Expert witness: Rule 702

a. Sufficient data

b. Reliable method

c. Reliably apply method to data

3

Reliability in science & law

1923. **Frye v. United States**
General acceptance

1993. **Daubert v. Merrell Dow**
Testable
Error rate
Peer review
General acceptance

Perlin MW. The Blairsville slaying and the dawn of DNA computing. In: Niapas A. ⁴
Death Needs Answers: The Cold-Blooded Murder of Dr. John Yelenic. Grelin; 2013.

2009. Pennsylvania v. Foley

World's first "probabilistic genotyping" Frye hearing



November 2007: Kevin Foley charged with murder

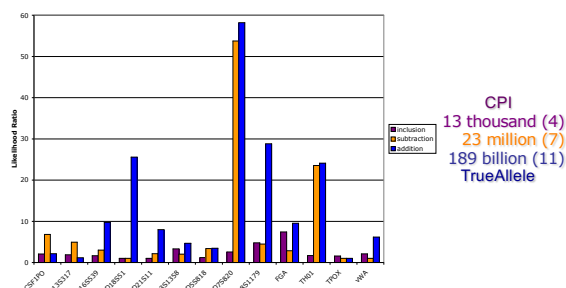
5

Different Statistical Methods

Data Used	<u>inclusion</u>	<u>subtraction</u>	<u>addition</u>
victim profile	NO	YES	YES
original data	NO	NO	YES

6

Different Match Statistics



Same Interpretation Principles

DNA data

A. Infer genotype

1. Data
2. Model
3. Compare
4. Probability

B. Match genotype

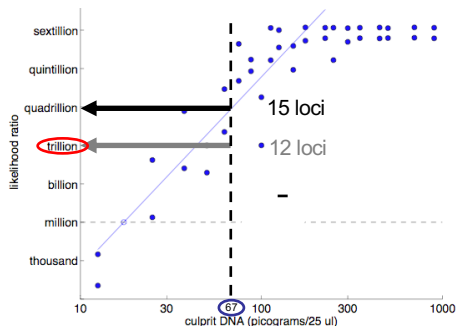
Likelihood ratio

Frye: General Acceptance in the Relevant Community

- Quantitative STR Peak Information
- Genotype Probability Distributions
- Computer Interpretation of STR Data
- Statistical Modeling and Computation
- Likelihood Ratio Literature
- Mixture Interpretation Admissibility
- Computer Systems for Quantitative DNA Mixture Deconvolution
- TrueAllele Casework Publications

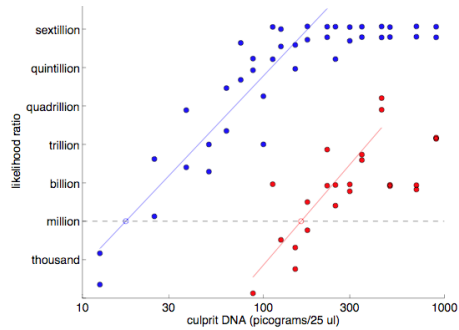
Perlin, M.W. and Sinelnikov, A. An information gap in DNA evidence interpretation. *PLoS ONE*, 4(12):e8327, 2009. 9

Expected TrueAllele Result



10

TrueAllele Addition vs. CPI Inclusion



11

Threshold: all or none



12

Quantitative: shades of gray



Cross Examination

- How can reliable DNA give different statistics?
- Why doesn't the computer use thresholds?
- Has this method ever been used before in court?

TrueAllele Admitted

COMMONWEALTH OF PENNSYLVANIA
 IN THE COURT OF COMMON PLEAS
 INDIANA COUNTY, PENNSYLVANIA
 NO. 1170 CRIM 2007
 KEVIN J. POLEY,
 Defendant.
 ORDER OF COURT
 MARTIN, P.J.
 AND NOW, this 2nd day of March 2009, this matter having come before the Court on the Defendant's Motion in Limine seeking to exclude the testimony of Dr. Robin Cotton and Dr. Mark Perlin and the Court having held a hearing thereon, it is hereby ORDERED and DIRECTED that the Motion in Limine is Denied.
 BY THE COURT,


Perlin MW, Galloway J. Computer DNA evidence interpretation in the Real IRA 15 Massereene terrorist attack. *Evidence Technology Magazine*. 2012;10(3):20-3.

2011. Massereene Barracks Attack

March 7, 2009 in Antrim, Northern Ireland



Homicide victims



Patrick Azimkar Mark Quinsey

Real Irish Republican Army claimed responsibility

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Abandoned Burned Car



17

DNA Evidence



passenger side
safety belt buckle



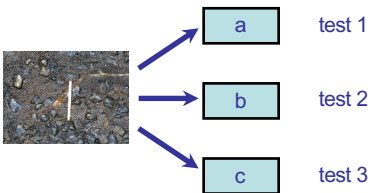
cell phone in
center console

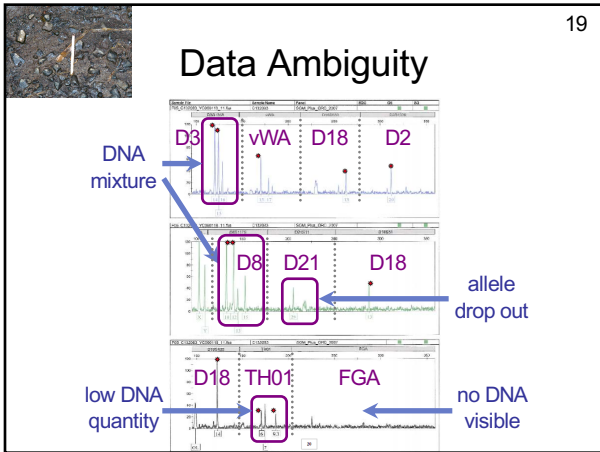
match stick
at side of road

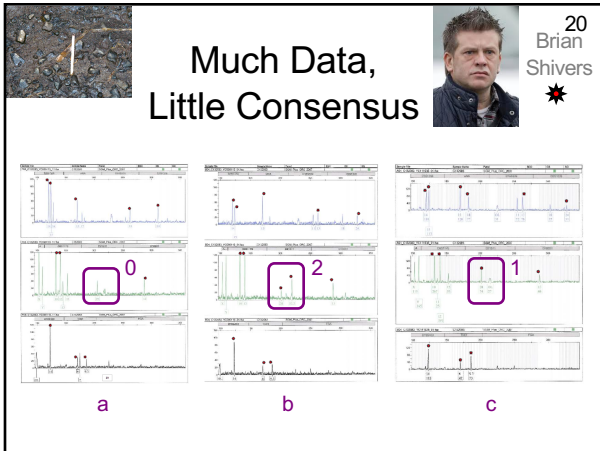


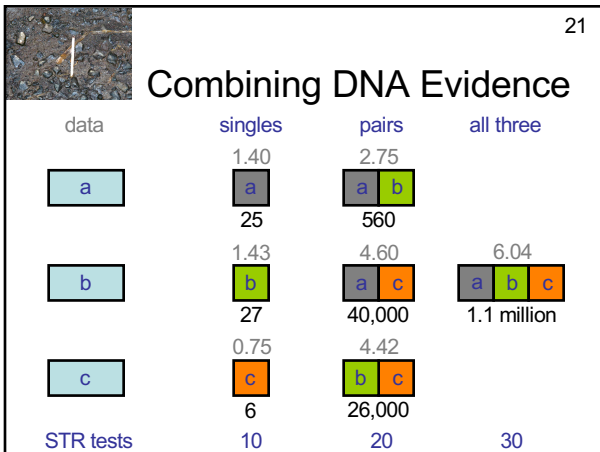
18

Match Stick



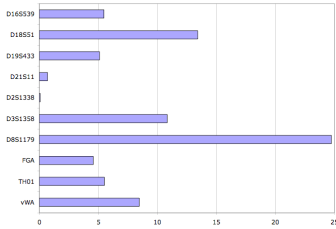









Statistic at 10 Loci



A match between the matchstick and Brian Shivers is 1.1 million times more probable than coincidence.



Direct Exam & Admissibility

- DNA Biology
- Mixture Evidence
- Human Review
- Probabilistic Genotypes
- TrueAllele Casework
- TrueAllele Validation
- Regulatory Approval
- Legal Precedent
- Published Method
- Readily Available
- Interpretation Admissibility
- Truth-Seeking Tool

Day 2: Cross Examination

DEFENSE: The People v Hector Espino, it was a DNA admissibility decision in which the judge said - this is not a legal point my Lord - he said: "We all understand the laws of physics apply from things, entire solar systems down to a grain of sand, but once you get to the atomic level all of a sudden all the rules change and the laws of physics don't apply and we have to start employing guesswork." Do you recognize the possibility that the theories that underpin your methods may not actually apply in the particular area that we are talking about?

WITNESS: No, I don't. Let me address your analogy. When you move from the world of Newton down to the world of Schroedinger at a sub-atomic level, you move from classical mechanics to quantum mechanics ... the laws of probability begin to apply to physics, and the answers to physics problems ... become probability distributions.

A 67 line answer, spanning 2 1/2 pages and taking several minutes.

Leaving out the historical context, and skipping ahead a page or so, the response concludes:

When you move from a classical model, whether it is in physics or in DNA mixtures, it is necessary, as we observed historically in physics and we see again happening in DNA, that in order to get reliable, accurate and reproducible results scientifically, whether in theory or in practice, you must move to a probability model. Your alternative is to simply discard all your data and not make any inference at all, however properly applied as confirmed by validation studies. You move from a deterministic classical world into a probability world that may be less comfortable initially to the practitioners, but is a better description of reality and makes more informative and accurate use of the data that is provided.

So I appreciate your analogy of moving from classical physics to quantum mechanics, because that's exactly what we see with DNA mixtures, as you move to uncertain data based on multiple individuals, as well as low template DNA.

Day 3: Cross Examination

WITNESS: That is correct.

DEFENSE: That's 33% isn't it? Isn't it?

WITNESS: I am looking at --

DEFENSE: Dr. Perlin, can you follow my questions?

PROSECUTION: Allow him to answer in his own way.

DEFENSE: You have reported on 33%.

JUDGE: Mr. O'Connor, please don't interrupt the witness.

DEFENSE: He had given an answer.

JUDGE: Please do not interrupt the witness.

DEFENSE: Do you want a calculator?

JUDGE: Don't ask any further questions until he has answered.

Motion to Exclude

27 page statement
99 paragraphs

Propositions.

1. P's assessments of LR's in this case are inadmissible for the following reasons:
 - a. no sufficiently reliable basis been provided to the Court for P's method of assessing LR's.
 - b. P has failed to fulfil his duty of candour as an expert witness.
 - c. P has at the very least negligently misled the Court upon important aspects of his evidence.
 - d. P lacks the necessary impartiality to provide admissible expert evidence.

Admissibility Ruling

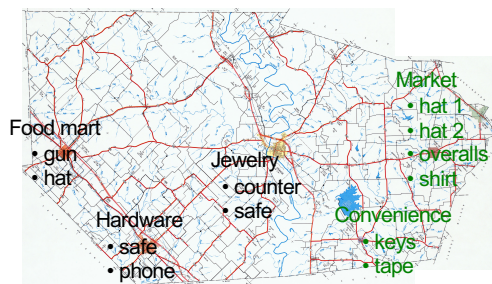
December 1, 2011

The Honorable Mr. Justice Hart

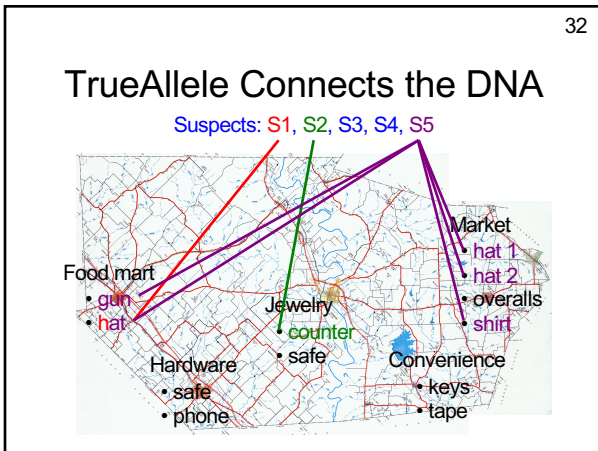
18 page ruling on TrueAllele, concluding:

I am satisfied that the stage has now been reached in the case of this system where it can be regarded as being reliable and accepted, and I am satisfied that Dr Perlin has given his evidence in a credible and reliable fashion. In the light of these conclusions I can see **no basis on which I could properly exercise my discretion ... to exclude this evidence**, and I therefore **admit it in evidence**.

2013. California v. Langston







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TrueAllele Validation Papers

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

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):in press.

Method Papers by Others

- Cowell RG, Lauritzen SL, Mortera J. *Identification and separation of DNA mixtures using peak area information*. Forensic Science International. 2007;166(1):28–34.
- Curran J. *A MCMC method for resolving two person mixtures*. Sci Justice. 2008;48(4):168–77.
- Tvedebrink T, Eriksen PS, Mogensen HS, Morling N. *Identifying contributors of DNA mixtures by means of quantitative information of STR typing*. J Comput Biol. 2011;18.

More TrueAllele Validation Studies

Allegheny County (PA): human vs. computer information
 Australia: extensive multi-axis validation
 Massachusetts State Police: internal validation
 National Institute of Standards and Technology (NIST)
 New York State Police Developmental Validation (2010)
 New York State Police Developmental Validation (2011)
 Suffolk County (NY): human vs. computer inconclusives
 UK Cellmark Forensics: volume crime (2,000 samples)
 Virginia Department of Forensic Science: casework study

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.

Kern County Validation Study

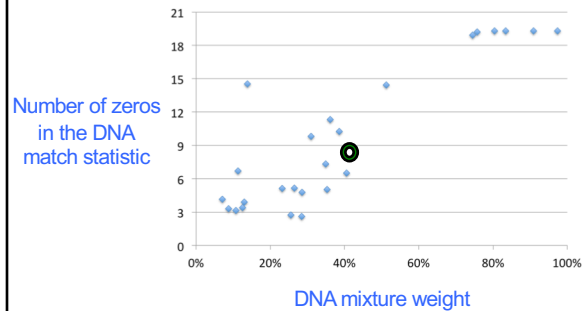
DNA mixtures: 2, 3, 4 & 5 contributors

- Casework
- Laboratory
 - 1 ng DNA amount
 - 200 pg low-template DNA amount
 - 10 second (longer) injection time

This case's DNA mixture:
 3 contributors in the mixture
 low-template DNA sample
 longer capillary injection

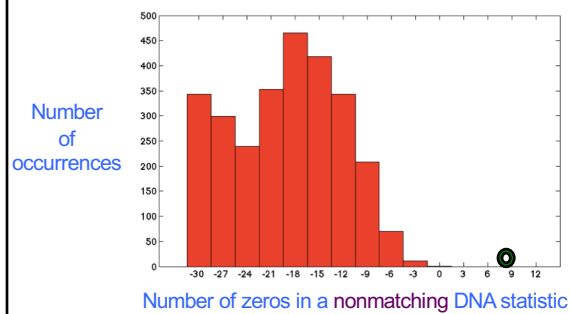
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Expected Match Statistic



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Specific Match Statistic



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TrueAllele in Criminal Trials

Over 100 case reports filed on DNA evidence

Court testimony:

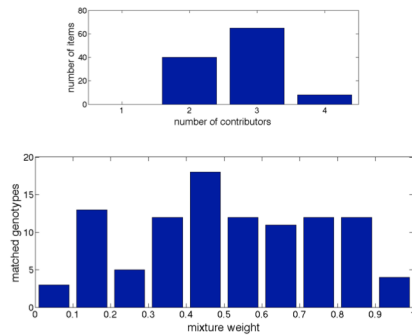
- state
- federal
- military
- foreign

Crimes:

- armed robbery
- child abduction
- child molestation
- murder
- rape
- terrorism
- weapons

43

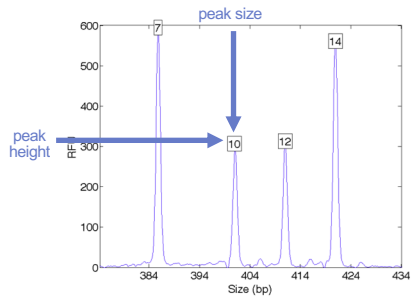
DNA mixture distribution



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DNA mixture data

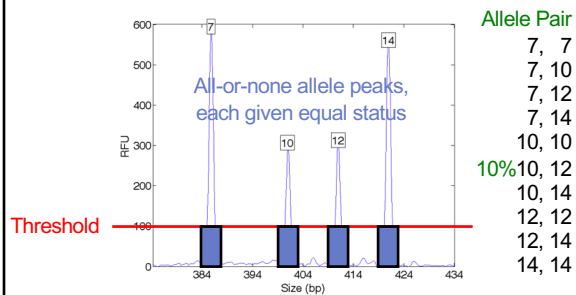
Quantitative peak heights at a locus



45

Data summary – “alleles”

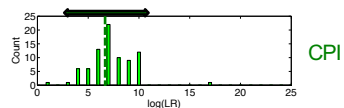
Over threshold, peaks are labeled as allele events



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CPI information

6.83 (2.22)
6.68 million

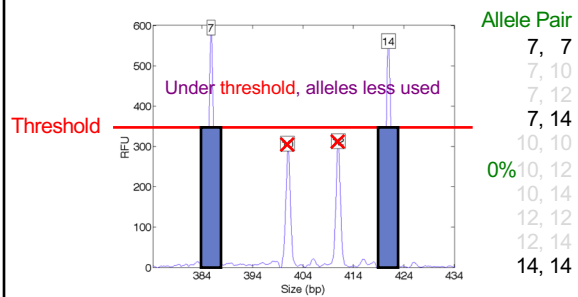


Combined probability of inclusion

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SWGIDAM 2010 guidelines

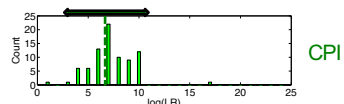
Higher threshold for human review



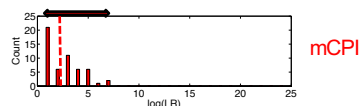
48

Modified CPI information

6.83 (2.22)
6.68 million



2.15 (1.68)
140

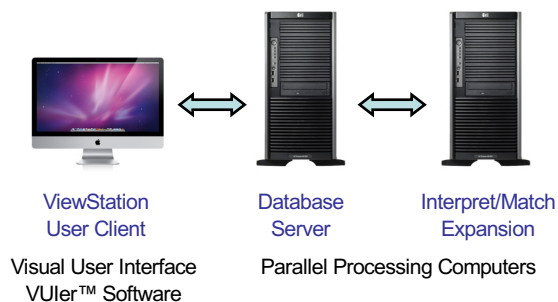


SWGDAM 2010 guidelines

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

TrueAllele Casework



Validated genotyping method

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

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):1430-47.

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):103-14.

Perlin MW, Belrose JL, Duceman BW. [New York State TrueAllele® Casework validation study](#). *Journal of Forensic Sciences*. 2013;58(6):in press.

52

Sensitivity

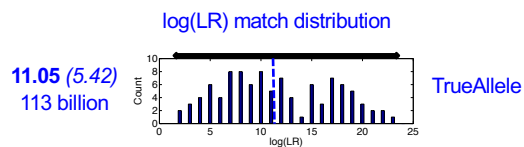
The extent to which interpretation identifies the correct person

True DNA mixture inclusions

101 reported genotype matches
82 with DNA statistic over a million

53

TrueAllele sensitivity



How do we know these are true matches?

54

Specificity

The extent to which interpretation does not misidentify the wrong person

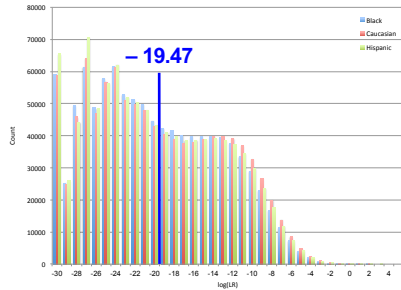
True exclusions, without false inclusions

101 matching genotypes x 10,000 random references
x 3 ethnic populations,
for over 1,000,000 nonmatching comparisons

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TrueAllele specificity

log(LR) nonmatch distribution



56

False positives

in over 1,000,000 comparisons per group

Tail distribution	Black	Caucasian	Hispanic
0	39	32	29
1	8	11	9
2	2	1	1
3	0	0	1
log(LR) > 0	49	44	40

false positive rate is under 1 in 20,000 (0.005%)
for LR > 100, error rate is 1 in 1,000,000 (0.0001)%

57

Reproducibility

The extent to which interpretation
gives
the same answer to the same question

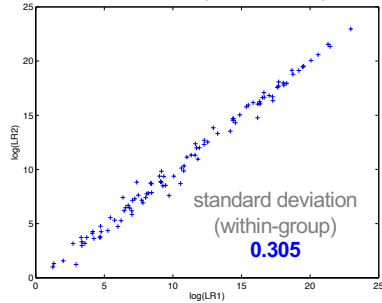
MCMC computing has sampling variation

duplicate computer runs
on 101 matching genotypes
measure log(LR) variation

58

TrueAllele reproducibility

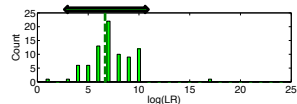
Concordance in two independent computer runs



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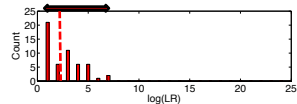
Comparison

6.83 (2.22)
6.68 million



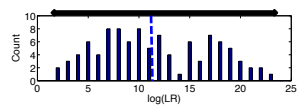
CPI

2.15 (1.68)
140



mCPI

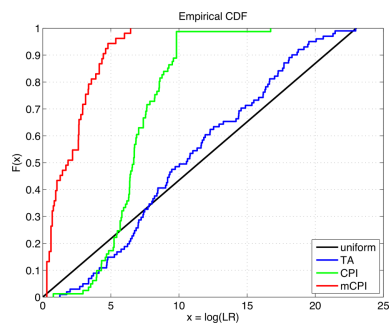
11.05 (5.42)
113 billion



TrueAllele

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Accuracy



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Conservative results

Five matches, TrueAllele less than CPI.
Ten comparisons, no statistical support:

Interpretation Method		Data Observations						
TrueAllele	CPI	mCPI	allele dropout	allele overlap	low peaks	peak imbalance	infeasible mixture	infeasible pattern
-10.64			3	4	1			1
-6.52			4	3	1			1
-5.05			4	3	1	1		1
-4.87				3		1	1	1
-4.86	3.48		4		1		1	
-3.22	6.04	6.34		2			1	1
-2.99	4.23		2		1		1	1
-2.18			2		1		1	
-1.41	4.08		1		1		1	
-0.67	2.95	0.60	1	2	1			

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Conservative results

Five matches, TrueAllele less than CPI.
Ten comparisons, no statistical support:

Interpretation Method		Data Observations						
TrueAllele	CPI	mCPI	allele dropout	allele overlap	low peaks	peak imbalance	infeasible mixture	infeasible pattern
-10.64			3	4	1			1
-6.52			4	3	1			1
-5.05			4	3	1	1		1
-4.87				3		1	1	1
-4.86	3.48		4		1		1	
-3.22	6.04	6.34		2			1	1
-2.99	4.23		2		1		1	1
-2.18			2		1		1	
-1.41	4.08		1		1		1	
-0.67	2.95	0.60	1	2	1			

CPI error rate 6% 2%

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More admissibility hearings

• **Australia** New South Wales Supreme Court admitted TrueAllele into evidence in Regina v Lian Bin 'Robert' Xie, case number 201101854, April 30, 2014. (Voir dire)

• **Ohio** trial court admitted TrueAllele into evidence in State v. Maurice Shaw, Cuyahoga County, case number CR-575691, October 10, 2014. (Daubert)

• **Louisiana** trial court admitted TrueAllele into evidence in State v. Chattley Chesterfield and Samuel Nicolas, East Baton Rouge Parish, case 01-13-0316 (II), November 6, 2014. (Daubert)

TrueAllele validation papers

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:e82837.

More admissibility rulings

• **New York** trial court admitted TrueAllele into evidence in *People v. John Wakefield*, Schenectady County, indictment number A-812-29, February 11, 2015. (Frye)

• **South Carolina** trial court admitted TrueAllele into evidence in *State v. Jaquard Aiken*, Beaufort County, case number 20121212-683, October 27, 2015. (Jones)

• **Massachusetts** trial court admitted TrueAllele into evidence in *Commonwealth v. Heidi Bartlett*, Plymouth County, case number PLCR2012-00157, May 25, 2016. (Daubert)

TrueAllele admissibility status

California (Kelly-Frye)
 Florida (Frye)
 Georgia (Harper-Daubert) [2]
 Indiana (Daubert) [4]
 Louisiana (Daubert) [3]
 Massachusetts (Daubert)
 Nebraska (Daubert)
 New York (Frye) [2]
 Ohio (Daubert) [2]
 Pennsylvania (Frye) appellate
 South Carolina (Jones)
 Tennessee (Daubert)
 Virginia (Spencer)
 Washington (Frye)
 United States (Daubert)
 Australia & Northern Ireland (Voir dire)

www.cybgen.com/information/admissibility/page.shtml
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TrueAllele® Admissibility

People of California v. Dupree Langston
 Kelly-Frye hearing
 Bakersfield, Kern County
 January 10, 2013
 Trial court admitted TrueAllele into evidence
 Case number BF139247B
[Download Ruling](#)

State of Florida v. Lajayvian Daniels
 Frye hearing
 Palm Beach County
 October 31, 2018
 Trial court admitted TrueAllele into evidence
 Case number 2015CF009320AMB
[Download Ruling](#)

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More TrueAllele information

<http://www.cybgen.com/information>



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<http://www.youtube.com/user/TrueAllele>
 TrueAllele YouTube channel


Cybergenetics

perlin@cybgen.com
