

TrueAllele® Casework Validation on PowerPlex® 21 Mixture Data

Australian and New Zealand
Forensic Science Society
September, 2014
Adelaide, South Australia

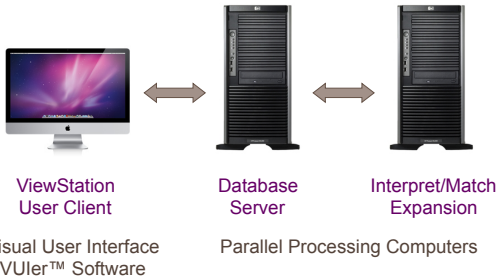
Jennifer Hornyak, MS, William P. Allan, MS,
and Mark W. Perlin, PhD, MD, PhD
Cybergenetics, Pittsburgh, PA, USA



Cybergenetics

Cybergenetics © 2003-2014

TrueAllele® Casework



Peer-reviewed validation studies

Perlin MW, Sinehnikov 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):103-14.

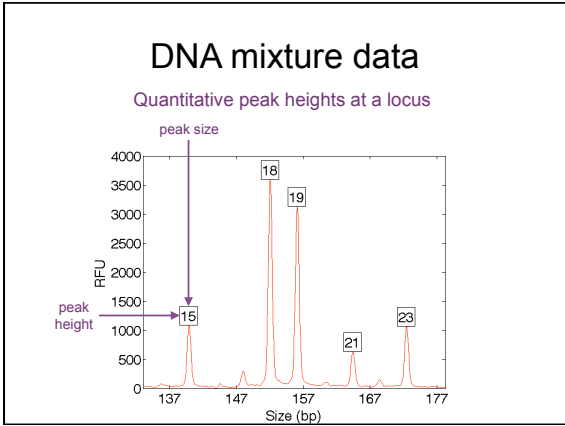
Perlin MW, Hornyak J, Sugimoto G, Miller K. TrueAllele® genotype identification on DNA mixtures containing up to five unknown contributors. *Journal of Forensic Sciences*. 2015;in press.

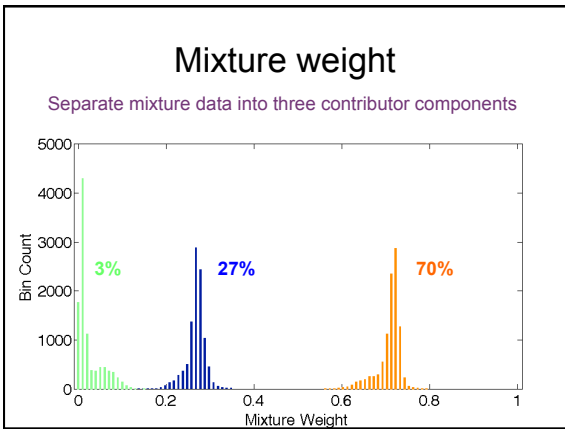
Greenspoon SA, Schiermeier-Wood L, Jenkins BC. Establishing the limits of TrueAllele® Casework: a validation study. *Journal of Forensic Sciences*. 2015;in press.

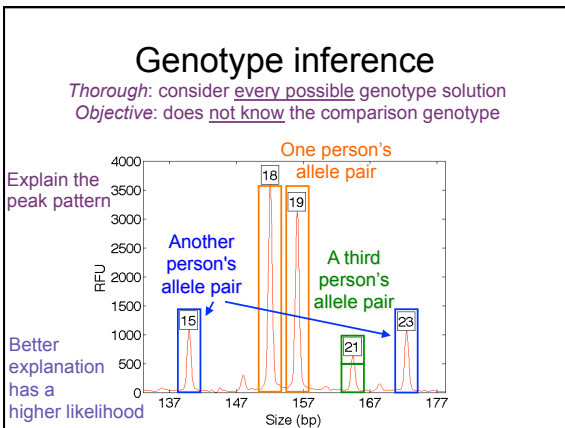
Perlin MW, Legler MM, Spencer CE, Smith JL, Allan WP, Belrose JL, Duceman BW. Validating TrueAllele® DNA mixture interpretation. *Journal of Forensic Sciences*. 2011;56(6):1430-47.

Perlin MW, Belrose JL, Duceman BW. New York State TrueAllele® Casework validation study. *Journal of Forensic Sciences*. 2013;58(6):1458-66.

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.







Study design

ncon	Item total	Genotype total
2	28	56
3	24	72
4	10	40
Overall	62	168

Measure in log(LR)

- sensitivity
- specificity
- reproducibility

log(LR) = 6 → 1 followed by 6 zeros → 1 million

STR data

PowerPlex® 21 kit

- 20 STR loci + sex-typing marker
- more discriminating power than 16 locus kits

Mixtures of Known Composition

- 2, 3, and 4 contributors in known ratios
- developed by NSW Forensic and Analytical Science Service
- ABI 3500xl genetic analyzer

TrueAllele operator

STR evidence data

.hid genetic analyzer files

- 2, 3, or 4 unknown mixture contributors
- 100K/100K MCMC sampling
- Replicate computer runs for each item

Evidence genotypes

probability distributions

Match statistic

information measured as log(LR)

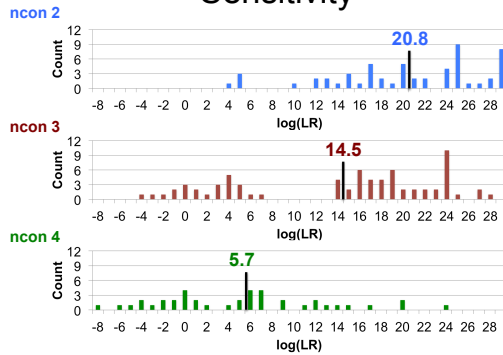
Sensitivity

The extent to which an interpretation method correctly includes a true contributor

168 genotype match statistics from 62 mixture items

Plotting the frequencies produces a log(LR) match distribution...

Sensitivity



False exclusions

False negative tail distribution

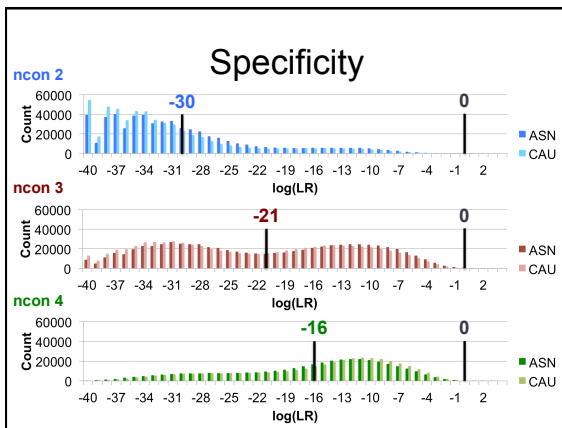
log(LR)	ncon		
	2	3	4
-1	0	2	2
-2	0	1	2
-3	0	1	1
-4	0	1	2
-5	0	0	1
-6	0	0	1
-7	0	0	0
-8	0	0	1
Total	0	5	10
N =	56	72	40

Specificity

The extent to which an interpretation method correctly excludes a non-contributor

168 genotypes x 10,000 random references x 2 ethnic populations

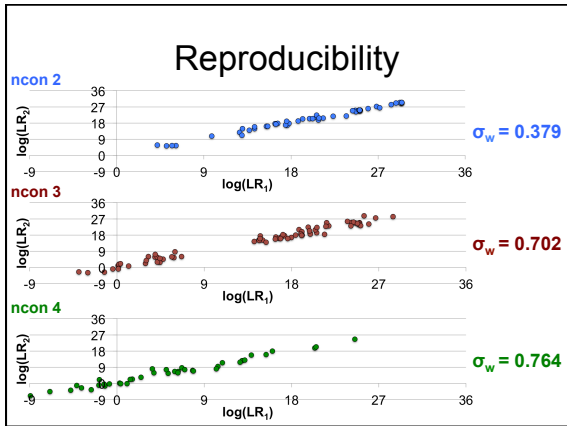
Plotting the frequencies produces a log(LR) mismatch distribution...



False inclusions

False positive tail distribution

log(LR)	ncon		
	2	3	4
0	31	824	655
1	10	273	212
2	1	82	59
3	1	26	13
4	0	2	0
Total	43	1,207	939
N=	1,120,000	1,440,000	800,000



Study conclusions

TrueAllele Casework computer DNA mixture interpretation on PowerPlex® 21 mixture data with 2, 3, and 4 contributors is:

- objective
- sensitive
- specific
- reproducible

Criminal Cases

Mixtures of family members

- child rape
- homicide
- 3 person mixture
- 5 person mixture

Over 200 case reports

- Many states & countries

On-line in crime labs

- California
- Virginia
- Middle East

TrueAllele today

Invented math & algorithms	20 years
Developed computer systems	15 years
Support users and workflow	10 laboratories
Used routinely in casework	3 labs
Validate system reliability	20 studies
Educate the community	50 talks
Train & certify analysts	200 students
Go to court for admissibility	5 hearings
Testify about LR results	20 trials
Educate lawyers and laymen	1,000 people
Make the ideas understandable	200 reports

Learn More About TrueAllele

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



- Courses
- Newsletters
- Newsroom
- Presentations
- Publications

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



Cybergenetics



jennifer@cybgen.com
