Separating Familial Mixtures,
One Genotype at a Time

Northeastern Association of Forensic Scientists
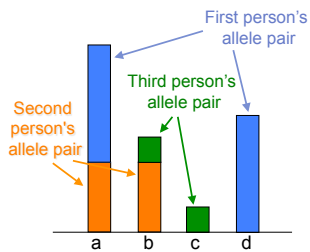November, 2014
Hershey, PA

Ria David, PhD, Martin Bowkley, MS,
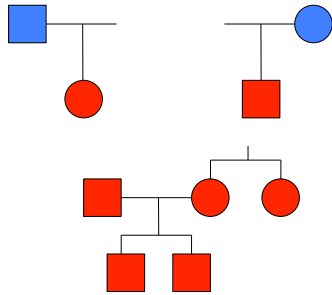and Mark W Perlin, PhD, MD, PhD
Cybergenetics, Pittsburgh, PA

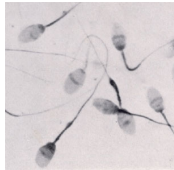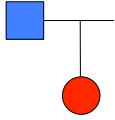Cybergenetics

Cybergenetics © 2003-2014

Relatives share alleles

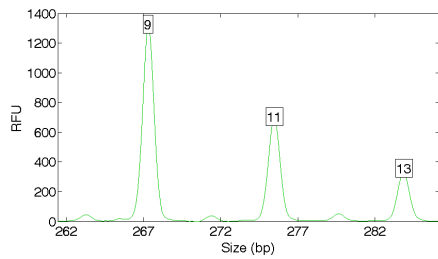---

Cybergenetics experience
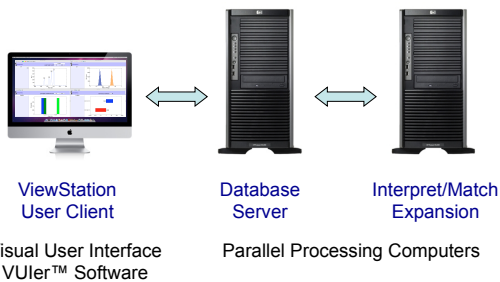
People of New York v Father of two daughters

Five semen stains on a comforter

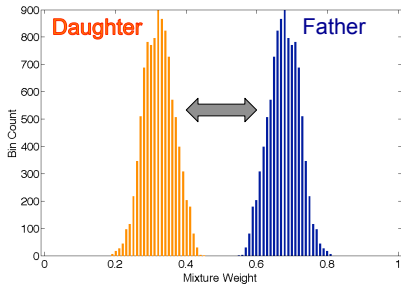## STR data

Quantitative peak heights at locus D16S539

TrueAllele® Casework

ViewStation
User Client

Database
Server

Interpret/Match
Expansion

Visual User Interface
VUIer™ Software

Parallel Processing Computers

Mixture separation

Daughter Father

Genotype separation

Consider every possible genotype solution

Explain the peak pattern

One person's allele pair

Another person's allele pair

Better explanation has a higher likelihood
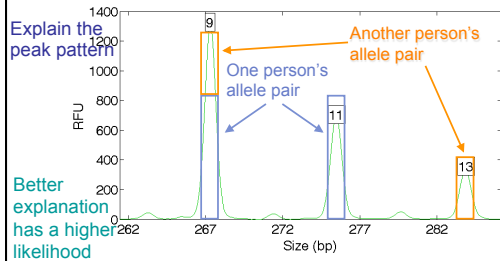
Minor genotype

Objective genotype determined solely from the DNA data.
Never sees a comparison reference.

99%

0.1%   0.1%

9,9   9,13   13,13
Allele Pair

Match statistic at locus

How much more does the victim match the evidence
than a random person?

15x

99%

Prob(evidence match)

Prob(coincidental match)
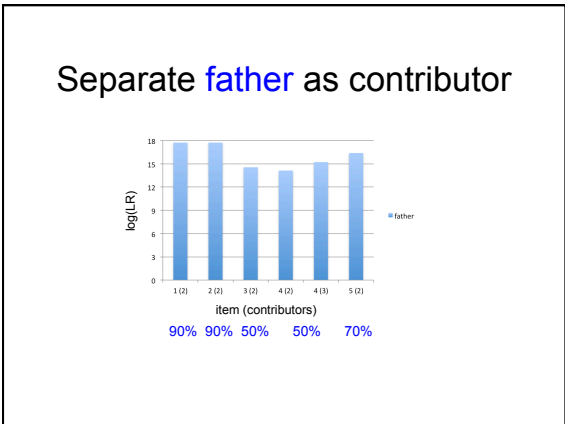
7%

Probability

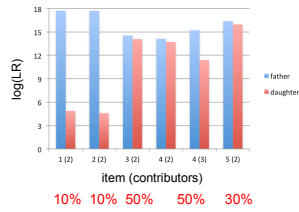9,9 9,13 13,13

Allele Pair
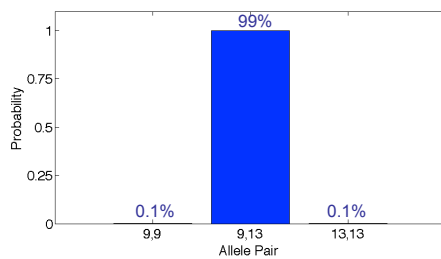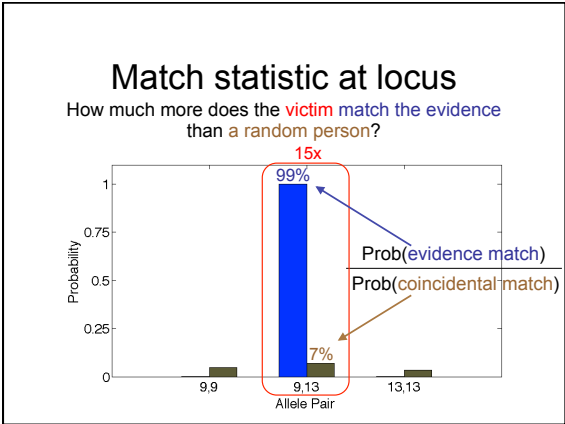
Reported match statistics

Multiply together the LR's at every locus

A match between the **comforter**
and **the young girl** is
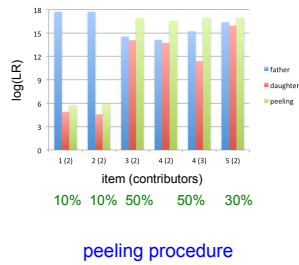186 quadrillion times more probable than coincidence

A match between the **comforter**
and **the girl's father** is
33 quadrillion times more probable than coincidence

Separate father as contributor

log(LR)

18
15
12
9
6
3
0

1 (2) 2 (2) 3 (2) 4 (2) 4 (3) 5 (2)

item (contributors)

90% 90% 50% 50% 70%

father

Separate daughter as contributor

Assume father, separate daughter

peeling procedure

Published 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):103-14.

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.

TrueAllele in the United States

Laboratory systems or case reports in 23 states
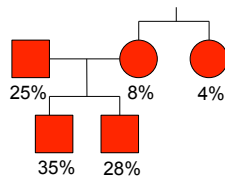

initial
final
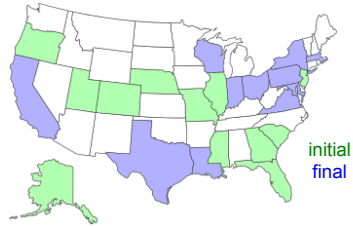
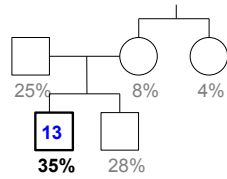## Court acceptance

- California
- Ohio
- Pennsylvania
- Virginia

- United Kingdom
- Australia

Appellate precedent in Pennsylvania

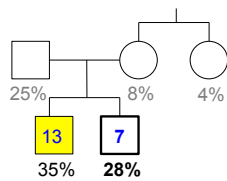## Separate out 5 relatives from a homicide mixture
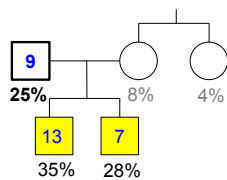

25%   8%   4%

35%   28%

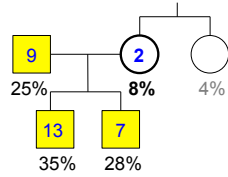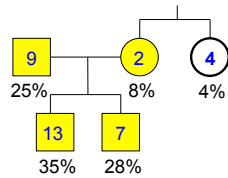Infer 5 unknown genotypes:
match 35% contributor

Assume 1 known, infer 4 unknown:
match 28% contributor

Assume 2 known, infer 3 unknown:
match 25% contributor

Assume 3 known, infer 2 unknown: match 8% contributor

9	**2**	○
25%	**8%**	4%
13	**7**	
35%	28%	

Assume 4 known, infer 1 unknown: match 4% contributor

9	**2**	**4**
25%	8%	4%
13	**7**	
35%	28%	

More information

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

Cybergenetics

You Tube

martin@cybgen.com