

# DNA Identification Science: The Search for Truth

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

Summer Research Symposium  
Duquesne University  
July, 2010



Cybergenetics © 2003-2010

---

---

---

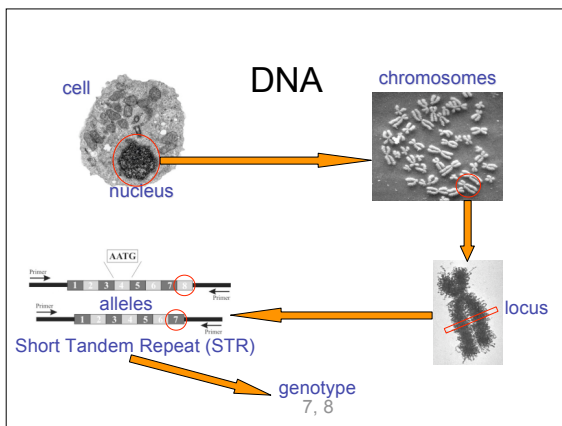
---

---

---

---

---




---

---

---

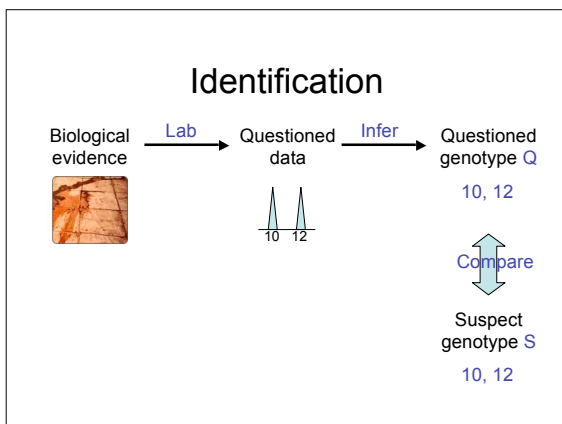
---

---

---

---

---




---

---

---

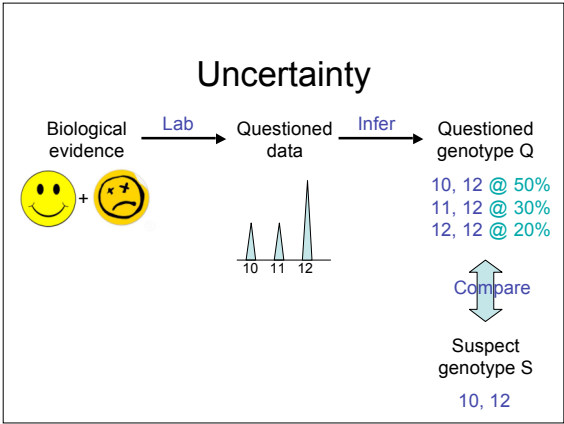
---

---

---

---

---




---

---

---

---


---

---

---

---


### Science



Prof Jevons, 1874

Scientific method

1. Propose hypothesis
2. Examine consequences
3. Compare with data



Prof Feynman, 1965

Agree: hypothesis likely  
Sort of agree: sort of likely  
Disagree: unlikely

---

---

---

---


---

---

---

---

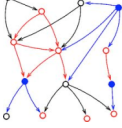
### Search for Truth



Rev Bayes, 1765

Bayes Theorem

Our belief in a hypothesis after we have seen data is proportional to how well that hypothesis explains the data times our initial belief.



Computers, 1985

All hypotheses must be considered.  
Need computers to do this properly.

Find the probability of causes by examining effects.

---

---

---

---

---

---

---

---

## Information Gain (LR)

identification hypothesis:  
the suspect contributed to the evidence

$$\text{information gain (likelihood ratio)} = \frac{\text{Odds(hypothesis | data)}}{\text{Odds(hypothesis)}}$$

↑ data  
after  
before

Additive information units: log(LR)  
Order of magnitude, powers of ten

---

---

---

---

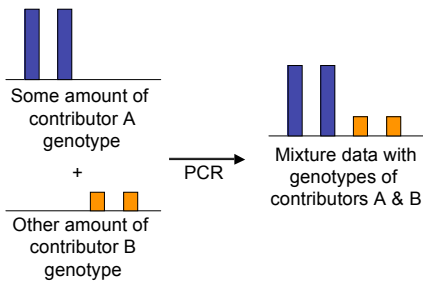
---

---

---

---

## DNA Mixture Data




---

---

---

---

---

---

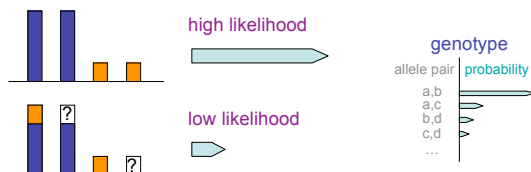
---

---

## Quantitative Mixture Interpretation

Step 1: infer genotype

- consider every possible allele pair
- compare pattern with DNA data
- Rule: *better fit's more likely it*




---

---

---

---

---

---

---

---

## Information Gain (LR)

Step 2: match genotypes

At the suspect's genotype allele pair,  
what is the locus *information gain*?

$$\text{information gain (likelihood ratio)} = \frac{\text{Prob(allele pair | data)}}{\text{Prob(allele pair)}}$$

after  
 ↑ data  
 before  
 (population)

Computer objectivity:  
 (Step 1) infer evidence genotype from data  
 (Step 2) compare genotype with suspect

---

---

---

---

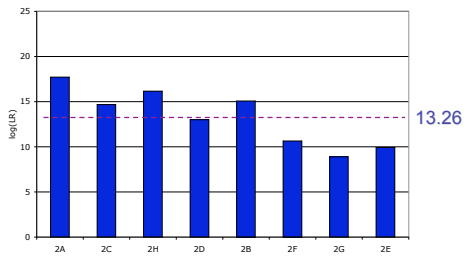
---

---

---

---

## Efficacy (2 unknown)




---

---

---

---

---

---

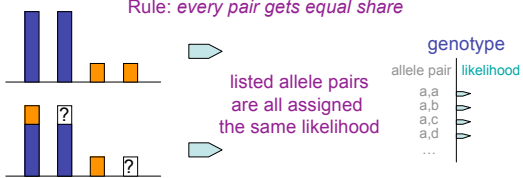
---

---

## Qualitative Manual Review

Step 1: infer genotype

Rule: every pair gets equal share



Step 2: match genotype

lower probability means lower information gain (LR)

---

---

---

---

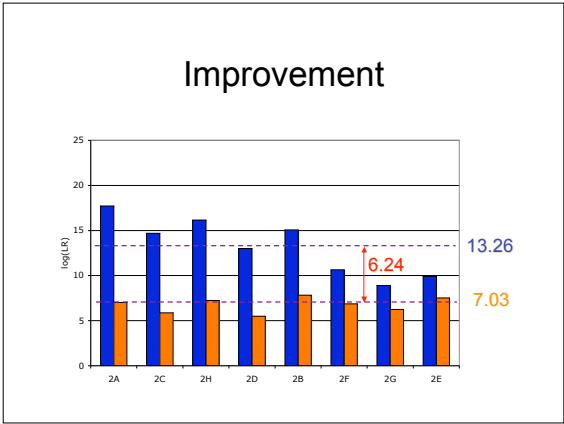
---

---

---

---

### Improvement




---

---

---

---

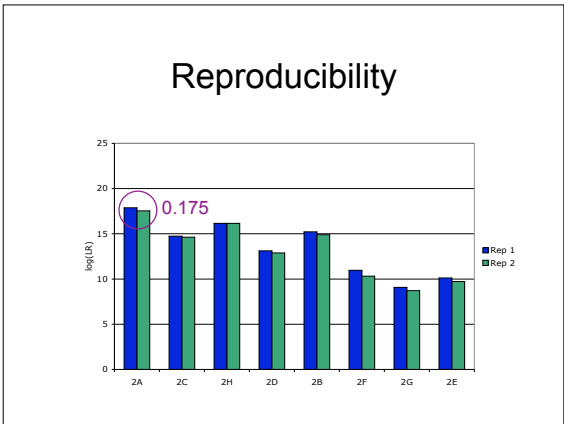
---

---

---

---

### Reproducibility




---

---

---

---

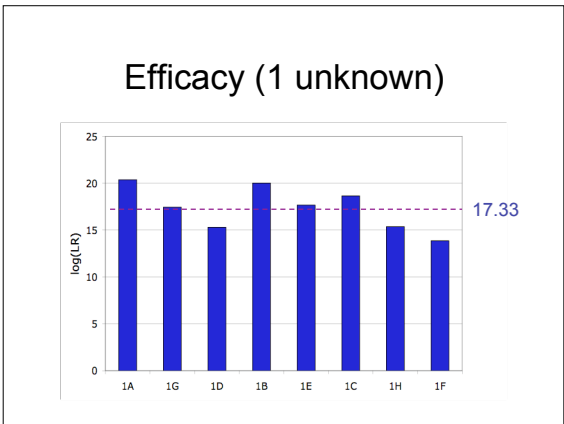
---

---

---

---

### Efficacy (1 unknown)




---

---

---

---

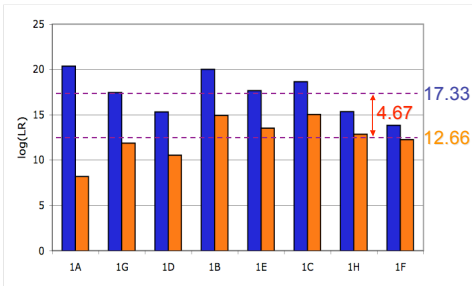
---

---

---

---

## Improvement




---

---

---

---

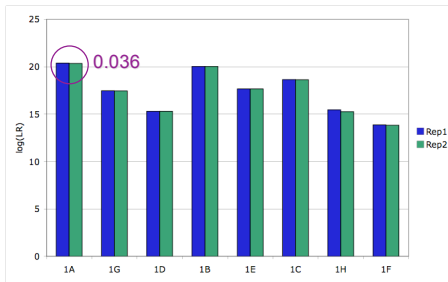
---

---

---

---

## Reproducibility




---

---

---

---

---

---

---

---

## Validation Summary

interpretation method	two unknown (without victim)	one unknown (with victim)
quantitative computer	13.26 (0.175) (ten trillion)	17.33 (0.036) (hundred quadrillion)
qualitative human	7.03 (ten million)	12.66 (five trillion)
improvement	6.24 (one million)	4.67 (fifty thousand)

---

---

---

---

---

---

---

---

## Commonwealth vs. Foley

Apr 2006: Blairsville Dentist John Yelenic murdered

Nov 2007: Trooper Kevin Foley charged with crime



Feb 2008: Defense questions 13,000 DNA match score

---

---

---

---

---

---

---

---

## DNA Evidence

- DNA from under victim's fingernails (Q83)
- two contributors to DNA mixture
- 93.3% victim & 6.7% unknown
- 1,000 pg DNA in 25 ul
- STR analysis with ProfilerPlus®, Cofiler®
- know victim contributor genotype (K53)
- TrueAllele® computer interpretation (using genotype addition method)  
infer unknown contributor genotype
- only after having inferred unknown, compare with suspect genotype (K2)

---

---

---

---

---

---

---

---

## Three DNA Match Statistics

<u>Score</u>	<u>Method</u>
13 thousand	inclusion
23 million	subtraction
189 billion	addition

- Why are there different match results?
- How do mixture interpretation methods differ?
- What should we present in court?

---

---

---

---

---

---

---

---

## Different Interpretation Methods

Data Used	inclusion	subtraction	addition
victim profile	NO	YES	YES
quantitative data	NO	NO	YES

---

---

---

---

---

---

---

---

## 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

---

---

---

---

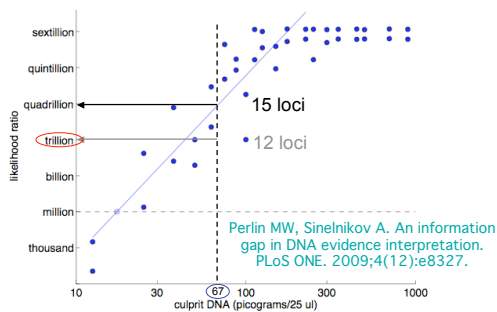
---

---

---

---

## Expected Result




---

---

---

---

---

---

---

---



## Expert Testimony

Dr. Perlin explained to the jury why these apparently different results were expected by DNA science. "The less informative methods ignored some of the data," said Dr. Perlin, "while the TrueAllele computation considered all of the available DNA data."

"A scientist may look at the same slide using the naked eye, a magnifying glass, or a microscope," analogized Dr. Perlin. "A computer that considers all the data is a more powerful DNA microscope."

---

---

---

---

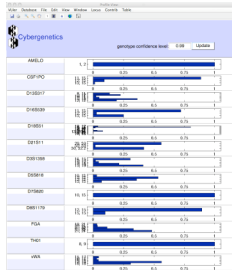
---

---

---

---

## Inferred Genotype



---

---

---

---

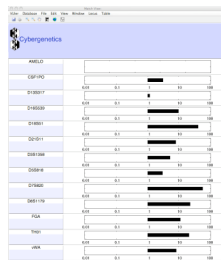
---

---

---

---

## log(LR) Match Information



---

---

---

---

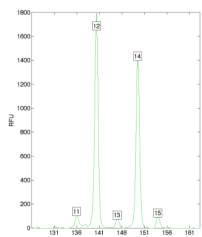
---

---

---

---

### Locus D8S1179 Data



---

---

---

---

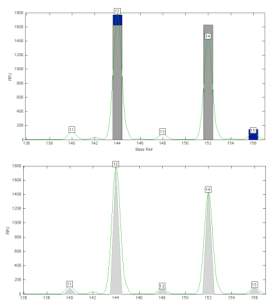
---

---

---

---

### Explain D8S1179 Genotype



---

---

---

---

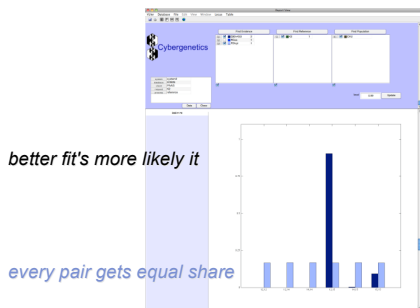
---

---

---

---

### Likelihood Comparison



---

---

---

---

---

---

---

---

## Generate Report

locus	allele pair	Q	R	S	LR	log(LR)
CSF1PO	12, 13	0.091	0.8518	1	1.755	0.244
D13S317	8, 11	0.136	0.8683	1	1.900	0.299
D16S539	11, 13	0.722	0.8928	1	7.775	0.891
D18S51	12, 13	0.803	0.8354	1	22.683	1.356
D21S11	29, 30	0.561	0.8277	1	6.388	0.885
D2S1328	15, 18	0.213	0.8839	1	2.538	0.485
D5S818	12, 13	0.358	0.1077	1	3.324	0.522
D7S820	10, 13	1	0.8226	1	44.188	1.645
D8S1179	12, 15	0.895	0.8365	1	24.525	1.390
FGA	21, 24	0.483	0.8514	1	9.388	0.973
TH01	8, 9	1	0.8450	1	22.201	1.346
VWA	17, 18	0.562	0.1199	1	4.689	0.671

Locus information gain is genotype probability ratio:  
LR = after/before

Joint information is the sum of the locus information

---

---

---

---

---

---

---

---

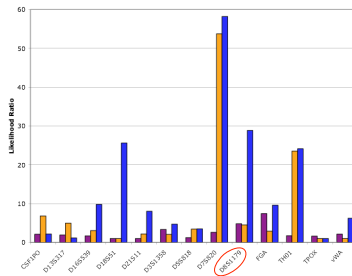
---

---

---

---

## More Data In, More Information Out



13 thousand (4)  
23 million (7)  
189 billion (11)

---

---

---

---

---

---

---

---

---

---

---

---

## Case Observations

- objective review never saw suspect
- easy to testify about in court
- understandable to judge and jury
- have precedent: admitted, testified
- preserve match information in data
- rapid response to attorney
- multiple match scores presented  
all information to the triers of fact –  
nothing was withheld from the jury  
this should be standard practice

---

---

---

---

---

---

---

---

---

---

---

---

Indiana Gazette.com 08 17 09 41  
 IN PRINT DAILY. ONLINE ALWAYS.

HOME SUBSCRIBERS MARKETPLACE NEWS OBITUARIES SPORTS BUSINESS MULTIMEDIA  
 FYI JOURNALS

ARCHIVES > NEWS One Verdict

Print | E-mail | Comment (No comments posted.) | RSS-Feeds | Text Size

### Jury convicts trooper of dentist slaying

Published: Thursday, March 19, 2009 12:46 AM EDT

An Indiana County Court jury this evening convicted state trooper Kevin Foley of first degree murder in the April 13, 2006, slashing death of Blairsville dentist John Yelenic.

"John Yelenic provided the most eloquent and poignant evidence in this case," said the prosecutor, senior deputy attorney general Anthony Krastek. "He managed to reach out and scratch his assailant," capturing the murderer's DNA under his fingernails.

The DNA Investigator Newsletter. Same Data, More Information - Murder, Match and DNA, Cybergenetics, 2009.  
[www.cybgen.com/information/newsletters/CybgenNews1.pdf](http://www.cybgen.com/information/newsletters/CybgenNews1.pdf)

---

---

---

---

---

---

---

---

---

---

---

---

## Public Safety

- DNA databases of criminal offenders
- police investigation: DNA database hits
- prevent crime by catching criminals
- could prevent 100,000 stranger rapes
- ensure conviction of the guilty
- avoid implicating the innocent

DNA public policy assumes that crime labs preserve DNA identification information

---

---

---

---

---

---

---

---

---

---

---

---

## Information Loss

Discarding DNA identification information instead of preserving it with science

The left chart is a bar graph with 'Information' on the y-axis (0 to 25) and categories 2A through 2E on the x-axis. Each category has two bars: a blue one and a red one. The blue bars are consistently higher than the red bars, with values ranging from approximately 10 to 18. The right chart is a scatter plot with 'culprit DNA (pgrams/25 ul)' on the x-axis (logarithmic scale from 10 to 1000) and 'information' on the y-axis (logarithmic scale from thousand to trillion). Blue dots represent higher information levels (trillion to quadrillion) and red dots represent lower information levels (million to billion). Two regression lines are shown: a blue one for the higher information points and a red one for the lower information points.

---

---

---

---

---

---

---

---

---

---

---

---

## No Information Incentive

### Crime lab evaluation

- NIJ funding based on other metrics
- ASCLD/ISO accreditation standards
- FBI/SWGDAM method guidelines

### Actual metrics and incentives

- eliminating backlogs
- passing audits
- testifying comfort

DNA identification information is not assessed

---

---

---

---

---

---

---

---

## No Information Consistency

National Institute of Standards and Technology  
Two Contributor Mixture Data, Known Victim

Some Differences in Reporting Statistics

LabID	Kits Used	Cases			
		Caucasians	Afr.	Hispanics	Hispanics
30	ProPlus/Collier	1.18E+15	2.13E+14	3.09E+15	
34	ProPlus/Collier	2.40E+11	7.66E+09	3.93E+10	
33	ProPlus/Collier	2.94E+08	1.12E+08	1.74E+09	
6	ProPlus/Collier	40,000,000	3,500,000	260,000,000	
9	ProPlus/Collier	1.14E+07	1.97E+07	1.54E+08	
79	ProPlus/Collier	930,000	47,000	1,350,000	
16	ProPlus/Collier	434,000	31,710	999,100	

213 trillion (14)

31 thousand (4)

Remember that these labs are interpreting the same MIX05 electropherograms

---

---

---

---

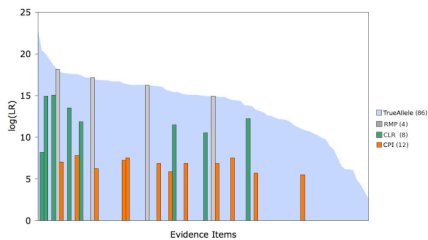
---

---

---

---

## Failure to Identify




---

---

---

---

---

---

---

---

## Scientist Task

- practice [identification science](#), not forensic art
- teach [public about scientific methods](#)
- learn more [mathematics \(probability theory\)](#)
- research [accurate and objective methodology](#)

---

---

---

---

---

---

---

## Acknowledgements

[Northeast Regional  
Forensics Institute](#)  
Jamie Belrose

[Cybergenetics](#)  
William Allan  
Meredith Clarke  
Matthew Legler  
Jessica Smith  
Cara Spencer



[Cybergenetics](#)

[New York State Police](#)  
Barry Duceman  
Melissa Lee  
Shannon Morris  
Elizabeth Staude

[Boston University](#)  
Robin Cotton

[Carnegie Mellon](#)  
Jay Kadane

---

---

---

---

---

---

---