Le laboratoire d'hématologie médico-légale
14 October 2016
Bordeaux, FRANCE

Risks and Errors in DNA Identification

John M. Butler, Ph.D.

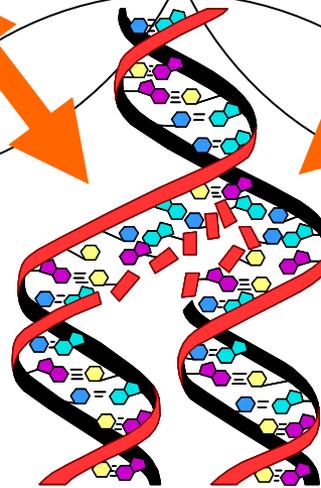*NIST Fellow & Special Assistant to the Director for Forensic Science*

U.S. National Institute of Standards and Technology

Communication Across the Criminal Justice System is Important

Law Enforcement

Judicial

What Every Law Enforcement Officer Should Know About **DNA Evidence**

Laboratory

PRINCIPLES OF FORENSIC DNA for Officers of the Court

https://forensic.training.nij.gov/

TRAINING : *Forensic DNA for Officers of the Court*

Home | Glossary | Resources

https://forensic.training.nij.gov/

- + **01 Introduction**
- + **02 Biology of DNA**
- + **03 Practical Issues Specific to DNA Evidence**
- + **04 Introduction to the Forensic DNA Laboratory**
- + **05 Assuring Quality in DNA Testing**
- + **06 Understanding a Forensic DNA Lab Report**
- + **07 Statistics and Population Genetics**
- + **08 Mitochondrial DNA & Y-STR Analysis**
- + **09 Forensic DNA Databases**
- + **10 Collection of DNA Evidence from Suspects and Arrestees**
- + **11 Pretrial DNA Evidence Issues**
- + **12 Victim Issues**
- + **13 Trial Presentation**
- + **14 Postconviction DNA Cases**
- + **15 Emerging Trends**

Login

Security Upgrades

You should already have noticed and agreed to a new disclaim
made to make our registration process and courses more secu

- Your password must now be at least **8** characters and inc
  letters, numbers, and special characters (e.g., ~!@&#$%^
- You will be asked to reset your password every **90** days.
- You may not reuse any of your past **6** passwords.

Please Login

Username [   ]

Password [   ]

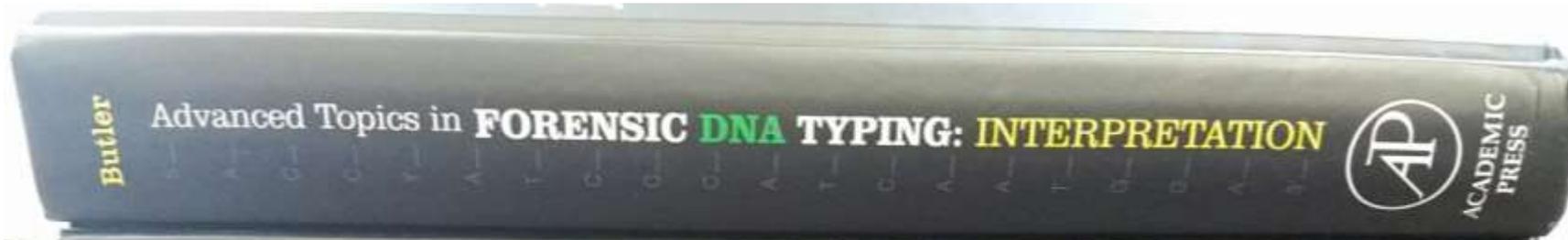[Login]

Don't have an account? Register Now

Forgot your username? Retrieve your username now.

Butler Books on Forensic DNA Typing

2015 — Advanced Topics in FORENSIC DNA TYPING: INTERPRETATION — ACADEMIC PRESS

2012 — Advanced Topics in FORENSIC DNA TYPING: METHODOLOGY — ACADEMIC PRESS

2010 — Fundamentals of Forensic DNA Typing — ACADEMIC PRESS

2005 — BUTLER — FORENSIC DNA TYPING — SECOND EDITION — ELSEVIER ACADEMIC PRESS
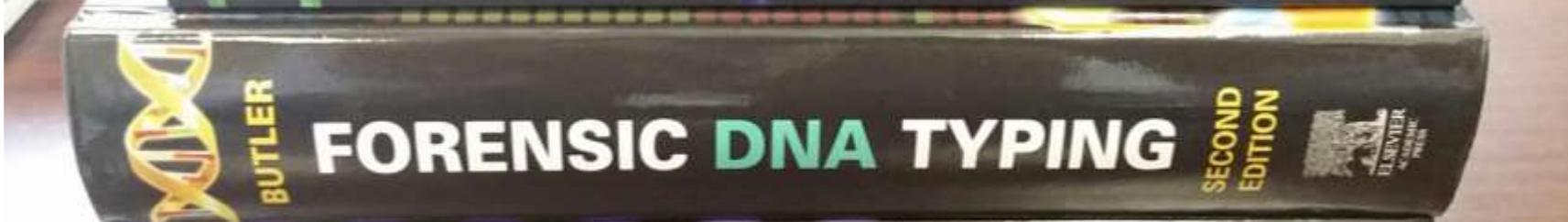
2001 — FORENSIC DNA TYPING — BUTLER — ACADEMIC PRESS

DNA Capabilities
to Aid Forensic Investigations

1. The **ability to identify the perpetrator**

2. Weight-of-evidence based on established genetic principles and statistics (Hardy-Weinberg 1908)

3. Established characteristics of genetic inheritance enables close **biological relatives** to be used for reference points using kinship associations

4. Superb **sensitivity** with PCR amplification (opens the possibility for contamination)

5. Well-established **quality assurance measures**

6. New **technology development** aided by genomics

Successful interpretation of DNA (Q-to-K comparison) depends on quality of the crime scene evidence (Q) and availability of suitable reference samples (K)

Concerns have been Raised over Potential for DNA Contamination

Professor Peter Gill

Misleading DNA Evidence
Reasons for Miscarriages of Justice

Peter Gill

Previous articles by Peter Gill on this topic:

- Gill, P. (1997). The utility of 'substrate controls' in relation to 'contamination'. *Forensic Science International,* 85(2):105-111.

- Gill, P., & Kirkham, A. (2004). Development of a simulation model to assess the impact of contamination in casework using STRs. *Journal of Forensic Sciences,* 49(3): 485-491.

- Gill, P., et al. (2010). Manufacturer contamination of disposable plastic-ware and other reagents—an agreed position statement by ENFSI, SWGDAM and BSAG. *Forensic Science International: Genetics,* 4(4): 269-270.
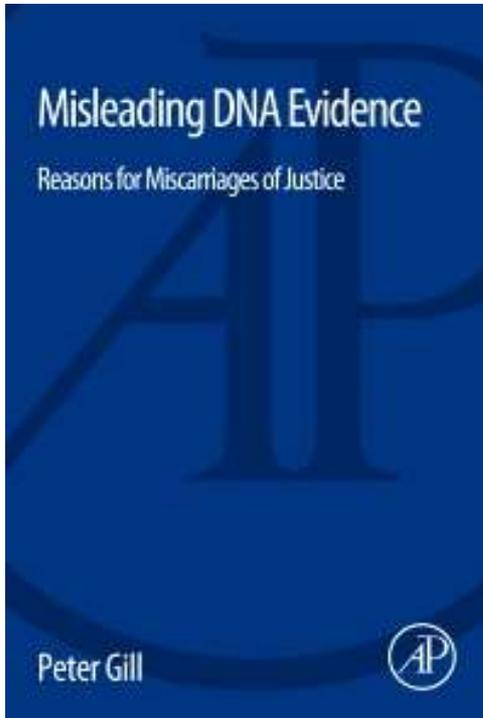
Discusses the Amanda Knox case DNA results

Forensic DNA Testing in the United States

- We have **~200 public (state and local government) laboratories** performing forensic DNA analysis
  - Two large private companies (Bode Cellmark and Sorenson Forensics) and a few smaller ones perform forensic DNA analysis
- Almost 15 million DNA profiles in the national DNA database (NDIS: National DNA Index System) run by the FBI Laboratory
  - Since 1998, the U.S. has included 13 core STR (short tandem repeat) markers; starting in 2017, this number will increase to 20 required STR loci
- Laboratories have many different protocols and in some cases, submitting the same sample to two different laboratories could result in two different results
  - Efforts are underway to improve standardization in the field

Critical Challenges Faced Today

- **Success of DNA testing** → significant growth in sample submissions → sample backlogs
  - Laboratory automation and expert system data review
  - Restrictive case acceptance policies to avoid law enforcement investigator 'swab-athons' at crime scenes

- **Greater detection sensitivity** → more complex DNA mixtures and low-template DNA with 'touch' evidence
  - Probabilistic genotyping to cope with increase in data interpretation uncertainty
 - Use of a complexity threshold to avoid "skating on thin ice"

Landmark Report Gives DNA Testing a Pass

The U.S. National Research Council of the National Academies issued a major report on forensic science in Feb. 2009.

*"**With the exception of nuclear DNA analysis**, no forensic method has been rigorously shown to have the capacity to consistently, and with a high degree of certainty, demonstrate a connection between evidence and a specific individual or source." (p. 41)*

STRENGTHENING

FORENSIC SCIENCE

IN THE UNITED STATES

A PATH FORWARD

NATIONAL RESEARCH COUNCIL
OF THE NATIONAL ACADEMIES

p. 100 mentions limitations with DNA mixtures
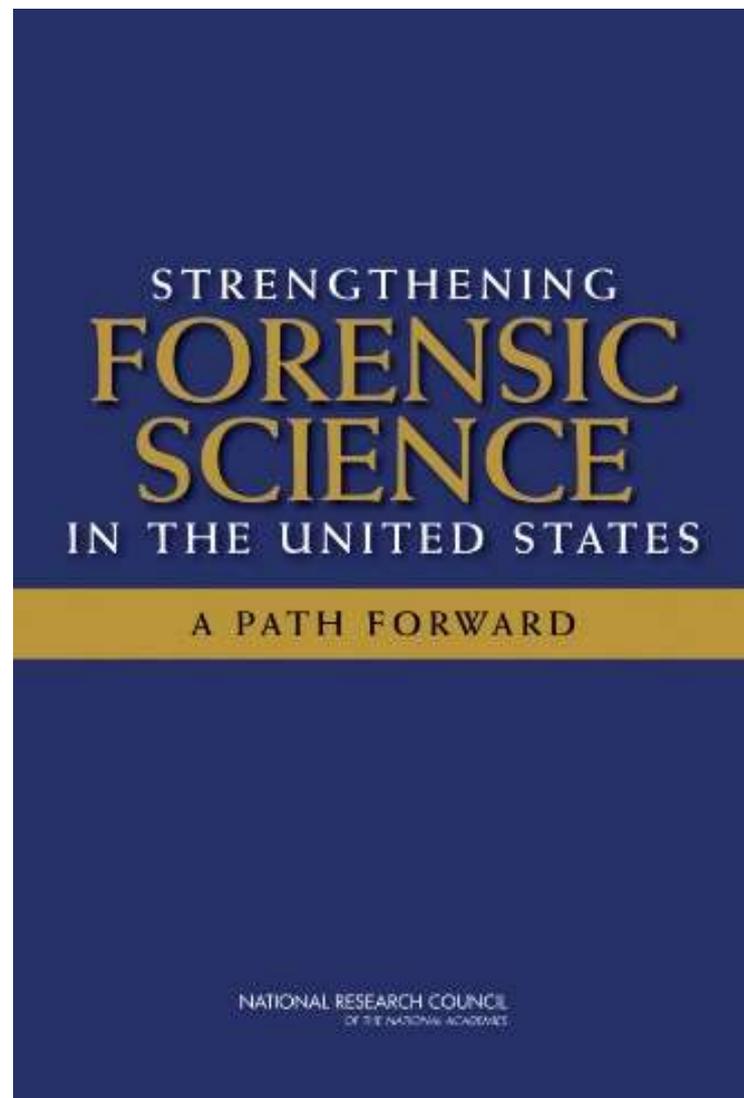
PCAST Report Comments on Forensic DNA

Released September 20, 2016

- Supports appropriate use of single-source and simple mixture DNA analysis

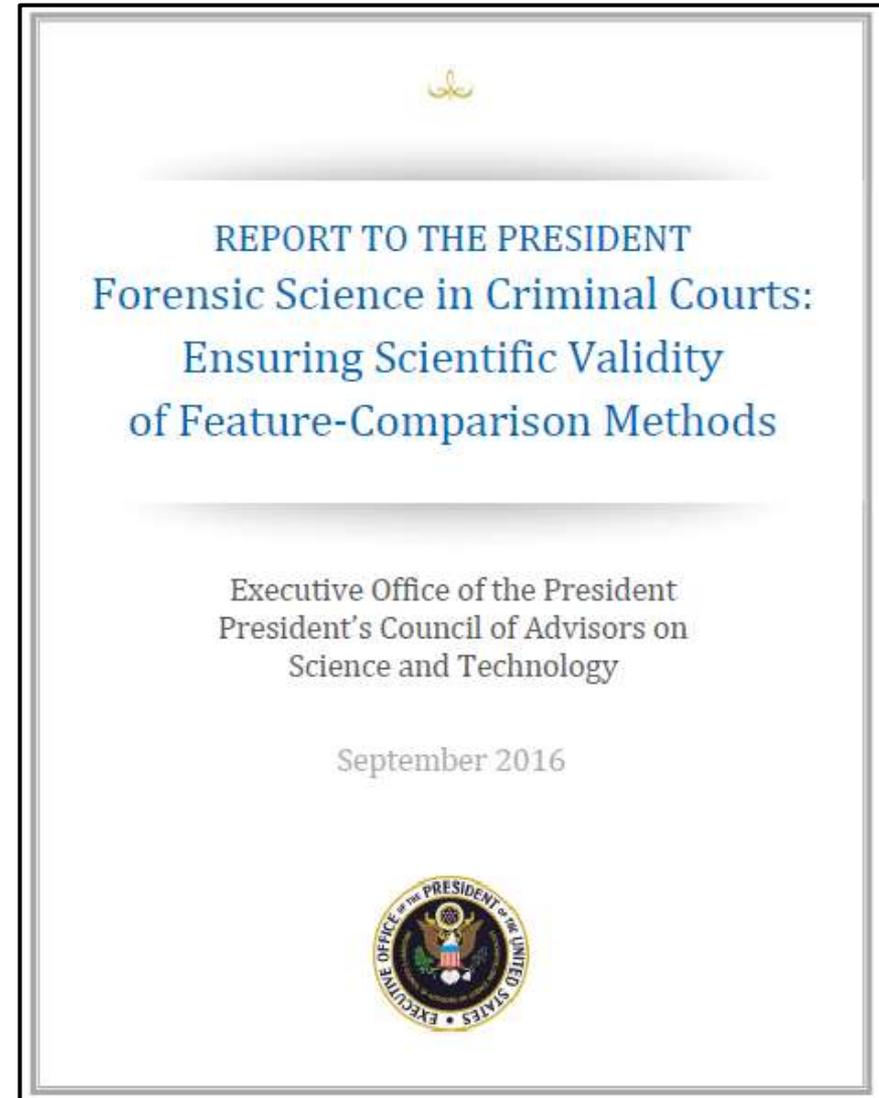- **Expresses reservations with complex DNA mixtures** (≥3 contributors)

PCAST Co-Chairs

Eric Lander   **John Holdren**

REPORT TO THE PRESIDENT

Forensic Science in Criminal Courts:
Ensuring Scientific Validity
of Feature-Comparison Methods

Executive Office of the President
President's Council of Advisors on
Science and Technology

September 2016

Recent Forensic Problems in the News

Director of D.C.'s embattled DNA lab
resigns after suspension of testing

Max M. Houck had been the director since the lab opened in
2012. Auditors found major problems there.

Keith L. Alexander and Julie Zauzmer | Local | Apr 30, 2015

April 2015

District could spend nearly $1 million for outside lab to test
DNA evidence

The District is scrambling to find an alternative after the D.C. lab was ordered to cease
DNA testing.

Keith L. Alexander | Crime | Apr 29, 2015

National accreditation board suspends all
DNA testing at D.C. crime lab

Two audits of the District lab found inadequate procedures for
DNA testing and poorly trained employees.

Keith L. Alexander | Crime | Apr 28, 2015

The Washington Post

Texas DNA Mixture Case Review

August 2015

http://www.fsc.texas.gov/texas-dna-mixture-interpretation-case-review

http://www.tdcaa.com/journal/changing-state-dna-analysis

Broward County Florida DNA Lab

July 2016

http://www.browardpalmbeach.com/news/bso-crime-lab-could-be-mishandling-crucial-dna-evidence-whistleblower-says-7881208

International conference
The hidden side of DNA profiles.
Artifacts, errors
and uncertain evidence
Auditorium, Università Cattolica del Sacro Cuore
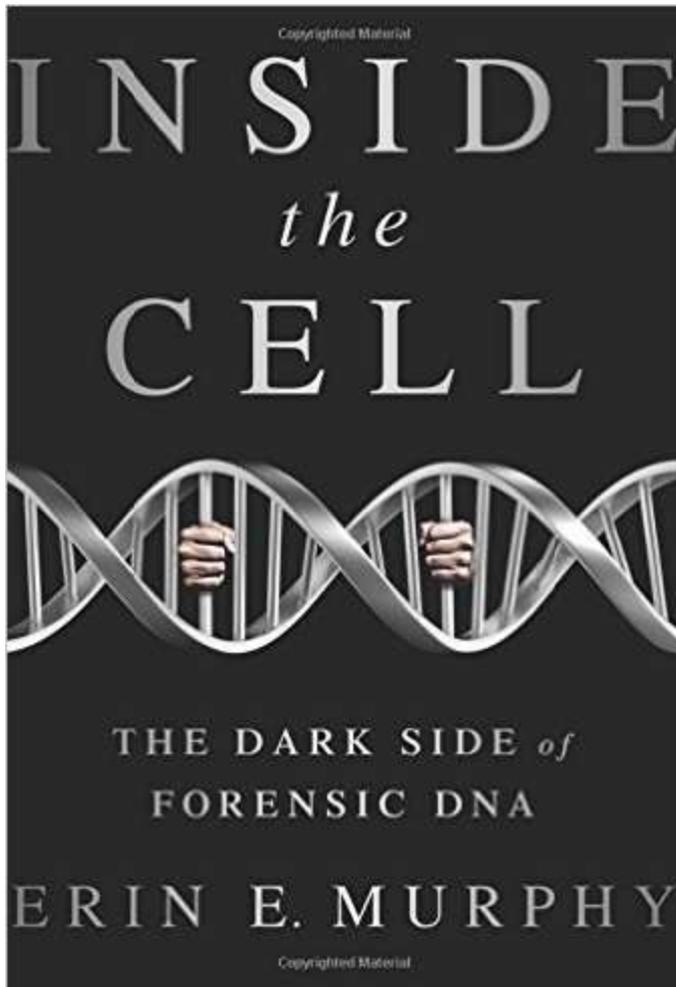**Rome, 27-28 April, 2012**

David Balding: "Low-template DNA cases are coming to court with limited abilities for <u>sound</u> interpretation. ... There are dangers with LTDNA but we know how to handle and manage them. Unfortunately, proper management is not a universal practice."

Peter Schneider: "If you cannot explain your evidence to someone that is not from the field (like a judge) – and you need a lot of technical excuses to report something – then the result is not good. You should leave it on your desk and not take it to court. This is a very common sense approach to this problem."

New Book by Law Professor Erin Murphy

400 pages

INSIDE *the* CELL

THE DARK SIDE *of* FORENSIC DNA

ERIN E. MURPHY

(Nation Books, Oct 2015)

Information from Chapter 7 of my New Book
Advanced Topics in Forensic DNA Typing: Interpretation

CHAPTER

7

Low-Level DNA and Complex Mixtures

"The limits of each DNA typing procedure should be understood, especially when the DNA sample is small, is a mixture of DNA from multiple sources, or is contaminated with interfering substances."
NRC I, 1992, p. 8

"For the complex DNA profile, there is no predominant or overarching standard interpretation method."
Peter Gill (Gill et al. 2012, report to the UK Forensic Science Regulator, p. 18)

"The limits of each DNA typing procedure should be understood, especially when the DNA sample is small, is a mixture of DNA from multiple sources…" **(NRC I, 1992, p. 8)**

Updated Guidelines to Help with DNA Mixture Interpretation

Scientific Working Group on
DNA Analysis Methods
Interpretation Guidelines for
Autosomal STR Typing
by Forensic DNA Testing
Laboratories
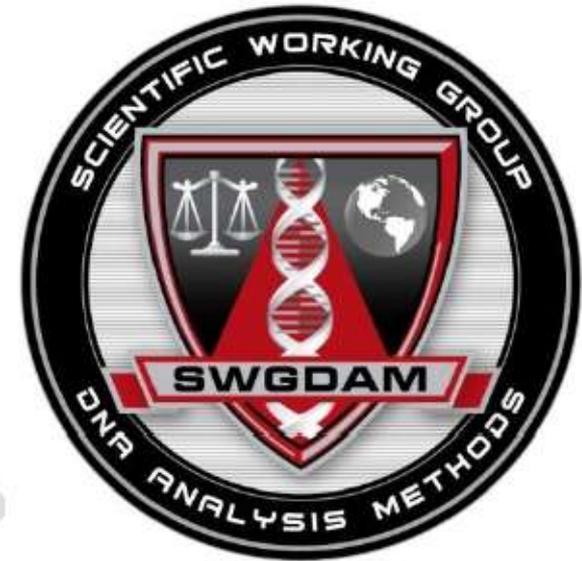
Current draft available for review is 90 pages long

http://www.swgdam.org/

5 Reasons that DNA Results Are Becoming More Challenging to Interpret

1. **More sensitive DNA test results**

2. **More touch evidence samples** that are poor-quality, low-template, complex mixtures

3. **More options exist** for statistical approaches involving probabilistic genotyping software

4. **Many laboratories are not prepared** to cope with complex mixtures

5. **More loci being added** because of the large number of samples in DNA databases

Improved Sensitivity is a Two-Edged Sword

"As sensitivity of DNA typing improves, laboratories' abilities to examine smaller samples increases. This improved sensitivity is a two-edged sword. **With greater capabilities comes greater responsibilities to report meaningful results.** Given the possibility of DNA contamination and secondary or even tertiary transfer in some instances, **does the presence of a single cell (or even a few cells) in an evidentiary sample truly have meaning?...**"

Butler, J.M. (2015) *Advanced Topics in Forensic DNA Typing: Interpretation* (Elsevier Academic Press: San Diego), p. 458

More Touch Evidence Samples

The DNA Field Experiment:
Cost-Effectiveness Analysis of the Use of DNA in the Investigation of High-Volume Crimes

John K. Roman
Shannon Reid
Jay Reid
Aaron Chalfin
William Adams
Carly Knight

Expanded DNA testing for burglary cases

NIJ April 2008 Research Report

DNA Solves Property Crimes (But Are We Ready for That?)
by Nancy Ritter

NIJ Journal October 2008 (vol. 261, pp. 2-12)

- **More poor-quality samples are being submitted**
  - Samples with <100 pg of DNA submitted in Belgium:
    19% (2004) → 45% (2008)
    (Michel 2009 FSIGSS 2:542-543)

- AAFS 2014 presentations showed poor success rates
  - NYC (**A110**): **only 10% of >9,500 touch evidence swabs from 2007 to 2011 produced usable DNA results**
  - Allegheny County (**A114**): examined touch DNA items processed from 2008 to 2013 across different evidence types (e.g., 6 of 56 car door handles yielded "resolvable profiles")
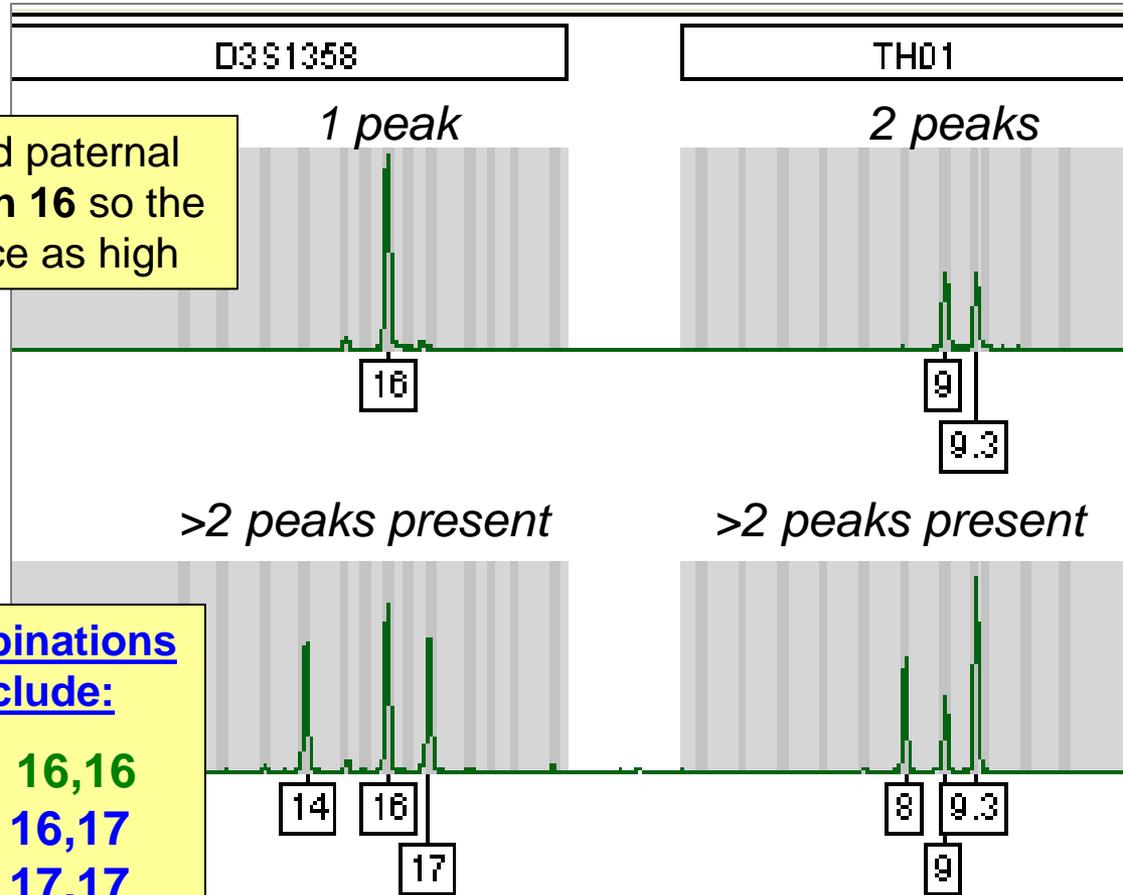
New Options Exist for Statistical Analysis

- Increase in approaches to try and cope with potential allele dropout → number of **probabilistic genotyping** methods have grown since Balding & Buckleton 2009 article

- Many possible choices for **probabilistic genotyping software** with commercial interests at stake

Balding, D.J. & Buckleton, J. (2009) Interpreting low template DNA profiles. *Forensic Sci. Int. Genet.* 4(1):1-10.

Gill P, Whitaker J, Flaxman C, Brown N, Buckleton J. (2000) An investigation of the rigor of interpretation rules for STRs derived from less than 100 pg of DNA. *Forensic Sci. Int.* 112(1):17-40.

Single-Source Sample vs Mixture Results

Maternal and paternal allele are **both 16** so the signal is twice as high

Possible combinations at D3S1358 include:

14, 17 with 16,16
14,14 with 16,17
14,16 with 17,17

Multiple possible combinations could have given rise to the mixture observed here
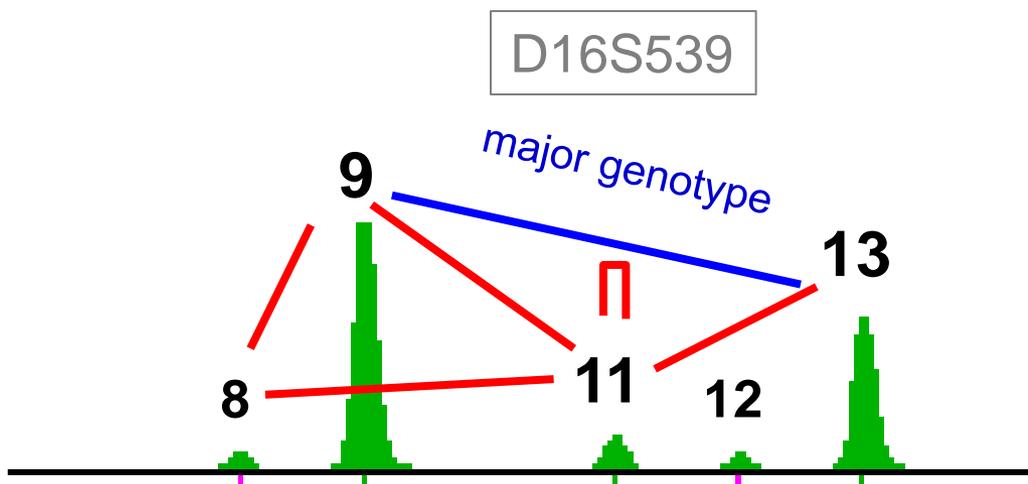
Probabilistic Genotyping
via Modeling Simulations

Mathematical Modeling of the Data

Typically thousands of simulations are performed

(MCMC)

Probable *Genotypes* to explain the mixture

PHR, mix ratio, stutter, etc…

D16S539

major genotype

9

13

∏

11

8

12

Minor Contributor Possible Genotypes   **Probability**

Minor Contributor Possible Genotypes	Probability
9,11	76%
11,11	15%
11,13	2%
8,11	2%
8,9	<1%
…	<1%

- Quantitative computer interpretation using numerous Markov Chain Monte Carlo (MCMC) simulations
- Models peak uncertainty and infers possible genotypes
- Results are presented as the Combined LR
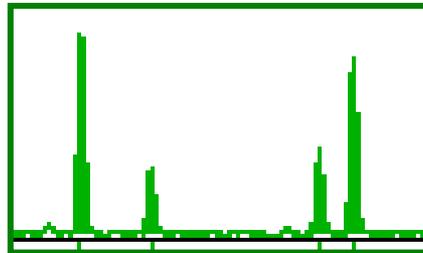
Math Analogy to DNA Evidence

$$2 + 2 = 4$$

$$2x^2 + x = 10$$

$$\int_{x=0}^{\infty} f(x)\,dx$$

Basic Arithmetic   **Algebra**   **Calculus**
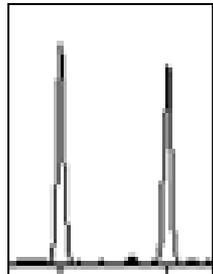
**Single-Source
DNA Profile**

(DNA databasing)

Sexual Assault Evidence
(2-person mixture with
high-levels of DNA)
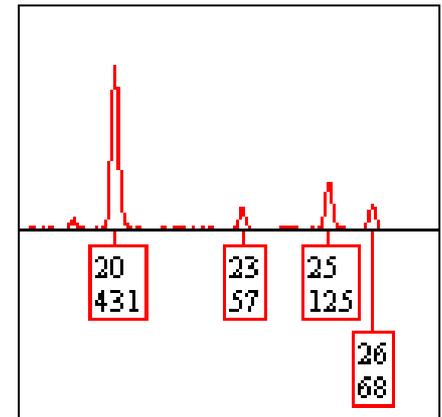
Touch Evidence
(>2-person, low-level,
complex mixtures
perhaps involving
relatives)

Many laboratories are not prepared
to cope with complex mixtures

- Have **appropriate validation studies** been performed to inform proper interpretation protocols? (curriculum & classroom instruction)

- Are **appropriately challenging proficiency tests** being given? (graded homework assignments)

- **Would we want to go into a calculus exam only having studied algebra and having completed homework assignments involving basic arithmetic?**

Netherlands Forensic Institute (NFI) Article on Forensic DNA Error Rates
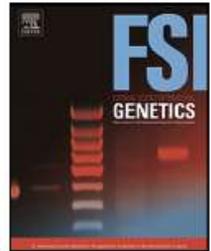
Error rates in forensic DNA analysis: Definition, numbers, impact and communication

Ate Kloosterman [a,b,c,*], Marjan Sjerps [b,d], Astrid Quak [a]

[a] Department of Human Biological Traces (HBS), Netherlands Forensic Institute, P.O. Box 24044, 2490 AA The Hague, The Netherlands
[b] Department of Science, Interdisciplinary Research, Statistics and Knowledge Management (WISK), Netherlands Forensic Institute, P.O. Box 24044, 2490 AA The Hague, The Netherlands
[c] Institute for Biodiversity and Ecosystem Dynamics, University of Amsterdam, Science Park 904, 1098 XH Amsterdam, The Netherlands
[d] Korteweg-de Vries Institute for Mathematics, University of Amsterdam, Science Park 904, 1098 XH Amsterdam, The Netherlands

Reported DNA Error Rates

	year	# tests	# errors	1 in	%
Plebani & Carraro [33]	1997 (3 mo.)	40,490	189	**214**	0.47%
Carraro & Plebani [36]	2007 (3 mo.)	51,746	160	**323**	0.31%
Stahl et al. [34]	1998 (3 yr.)	676,564	4,135	**164**	0.61%
Hofgärtner & Tait [35]	1999 (1 yr.)	88,394	293	**302**	0.33%

Medical Laboratory Literature Data (label for rows above)

notifications

	year	# tests	# notifications	1 in	%
NFI DNA casework	2008	66,391	328	**202**	0.49%
NFI DNA casework	2009	82,896	329	**252**	0.40%
NFI DNA casework	2010	89,977	435	**207**	0.48%
NFI DNA casework	2011	100,407	526	**191**	0.52%
NFI DNA casework	2012	132,456	572	**232**	0.43%

Kloosterman et al. 2014 *FSI Genetics* (label for rows above)

Kloosterman et al. (2014) Error rates in forensic DNA analysis: definition, numbers, impact and communication. *FSI Genetics* 12: 77-85

	year	# tests	# errors	1 in	%
NIST Identifiler JFS 2003 population data	2003	11,200 genotypes	7	**1600**	0.06%
FBI errata JFS 2015 population data	1999	30,550 alleles	51	**599**	0.17%

Population Data (label for rows above)

Even with single-source, pristine samples, the error-rate is not zero!

Not all quality issue notifications (aka "errors") are equal

Table 3

Types of quality issue notifications (QINs) at the NFI in the years 2008–2012. In 2011 it was decided to no longer incorporate the type c QIN: opportunities for improvement ($n = 2$ in 2011 and $n = 10$ in 2012) in the yearly totals of this overview.

	2008	2009	2010	2011	2012
a. External origin	23	10	23	54	100
b. External contamination	3	0	5	24	22
c. Room for improvement	11	6	3	(2)	(10)
d. Positive response	19	9	11	6	17
e. Clerical (no adverse outcome)	29	25	92	77	82
f. Not related to case work	13	9	20	10	5
g. Other (NFI related)	230	270	281	355	346
Total	328	329	435	526	572

Kloosterman et al. (2014) Error rates in forensic DNA analysis: definition, numbers, impact and communication. *FSI Genetics* 12: 77-85

Checks and Controls on Forensic DNA Results

Community	FBI DNA Advisory Board's Quality Assurance Standards *(also interlaboratory studies)*
Laboratory	ASCLD/LAB, ANAB, A2LA Audits and Accreditation
Analyst	Proficiency Tests & Continuing Education
Method/Instrument	**Validation of Analytical Performance** *(with aid of **traceable reference materials**)*
Protocol	Standard Operating Procedure is followed
Data Sets	Allelic ladders, positive and negative amplification controls, and reagent blanks are used
Individual Sample	Internal size standard present in every sample
Interpretation of Result	**Second review by qualified analyst/supervisor**
Court Presentation of Evidence	Defense attorneys and experts with power of discovery requests

Forensic Conference Organized by NIST

FORENSIC SCIENCE ERROR MANAGEMENT
INTERNATIONAL FORENSICS SYMPOSIUM
JULY 20-24, 2015 · WASHINGTON, DC

Planning has started for a second Symposium
Date: July 24-28, 2017
Location: Gaithersburg, MD
Sponsors that have been approached
DoD, FBI, NIST

http://www.nist.gov/director/international_forensics_home.cfm

National Institute of Standards and Technology

- Science agency **part of the U.S. Department of Commerce**
- Started in 1901 as the **National Bureau of Standards**
- Name changed in 1988 to the **National Institute of Standards and Technology (NIST)**
- Forensic science research activities dating back to 1920s
- Partnership since 2013 with U.S. Department of Justice to create the National Commission on Forensic Science (NCFS) and the Organization of Scientific Area Committees (OSAC)
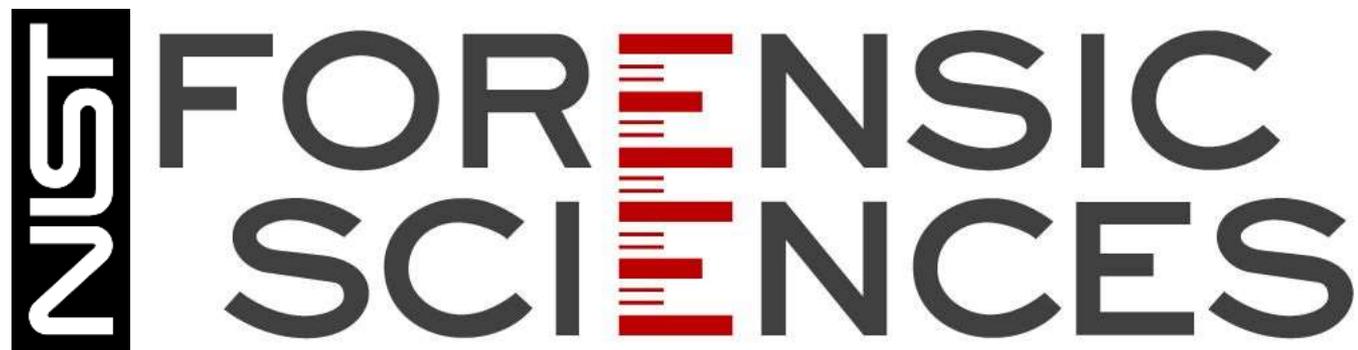
- Primary campus in Gaithersburg, Maryland (near Washington, D.C.)
- >3,400 employees and >3,700 associates
- Supplies >1300 reference materials
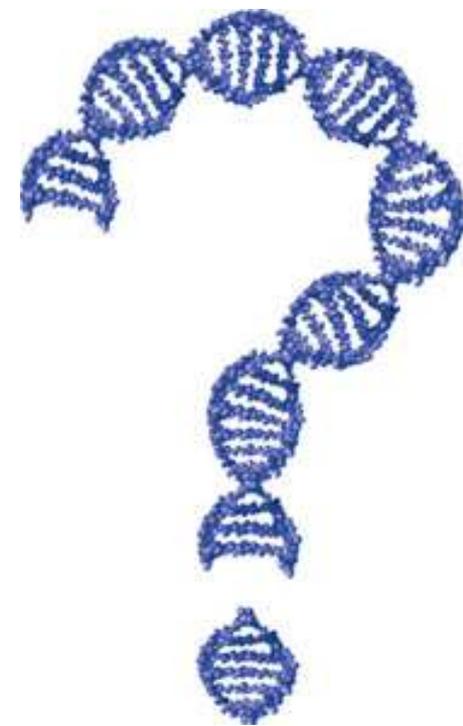- Defines official time for the U.S.

DNA reference material

NIST FORENSIC SCIENCES

www.nist.gov/forensics

+1-301-975-4049   **john.butler@nist.gov**