DNA Mixture Interpretation Principles: Observations from a NIST Scientific Foundation Review

Chair: **John M. Butler** (NIST)
Co-Chair: **Sheila Willis** (NIST guest researcher)

DNA Mixture Interpretation Principles: Observations from a NIST Scientific Foundation Review

AAFS 2019 Workshop #10 (February 18, 2019; Baltimore, MD)

Introduction

Melissa K. Taylor

National Institute of Standards and Technology

UPDATED SLIDE

Workshop Disclaimer

Points of view in this workshop are those of the presenters and do not necessarily represent the official position or policies of the National Institute of Standards and Technology.

Certain commercial equipment, instruments, and materials are identified in order to specify experimental procedures as completely as possible. In no case does such identification imply a recommendation or endorsement by NIST, nor does it imply that any of the materials, instruments, or equipment identified are necessarily the best available for the purpose.

NEW SLIDE

Shutdown Contingency Plans

- Plan A = The best case plan.
- Plan B = Backup plan.
- Plan C = Contingency plan. The backup to the backup plan.

  Plan D = Danger plan. At this point, you've had three plans fall through.

  Plan E = Emergency plan.

  Plan F = Plan to fail.

  Plan G = Gangsta plan.

  Plan H = Plan from Hell or Heaven. Purgatory plan.

  Plan I = iPlan.

  Plan J = Jump plan.

  Plan KIT = Any plan to keep in touch.
- LOL Plan = A laughable plan.
- Plan M = Master plan.
- Plan N = "Plannin'".
- Plan O.G. = Original Gangsta plan.
- Plan P = Power plan.
- Plan Q = Quick plan.
- Plan R = Raw plan.
- Plan S = Summer plans.
- Plan TTYL = A plan to catch up some other time.
- Plan U = Under plan. A way to be flexible with your planning (and/or to be unprepared).
- Plan V = Vulture plan. A plan left for dead.
- Plan W = A questionable plan.
- Plan X = Explicit plan. Usually a plan revealing too much information.
- Y Plan? = When plans are unnecessary.
- ZzZzz Plan = … plan.

NEW SLIDE

Four Potential Options for Our AAFS Workshop

1. **The government does not shut down and we have a normal workshop on February 18.** There are currently 103 people registered for the workshop in addition to the 18 presenters. (the preferred option)

2. If a government shutdown occurs on February 15, then three options are possible:
   a. **Shutdown exemption is granted**, **and the workshop can continue as planned**. We are in the process of seeking approval for an exemption to host the workshop regardless of the shutdown. We may not know if this approval has been granted until February 15.
   b. **An alternative version of the workshop occurs** with non-federal employees using slides already prepared by the non-federal participants. If this happens, then the focus of the workshop would need to be changed and the NIST study would not be discussed. We need to discuss how time slots could be expanded in the event of this option.
   c. **The workshop is canceled** because it is focused on a federal government activity and without the NIST participants it should not be held. This decision would be made by the co-chairs in consultation with NIST management.

Note: If a federal government shutdown occurs and persists through the morning of February 18 and we are unable to obtain approval for the exemption, then invitational travel authorizations will be canceled, and reimbursements will not be provided.

UPDATED SLIDE

Purpose of This Workshop

- Review scientific principles and approaches used in DNA mixture interpretation including those involving probabilistic genotyping software
- Explore some of the foundational literature supporting these principles
- Report on the current NIST scientific foundation review underway with DNA mixture interpretation

We had originally planned on having a draft of our report completed before this workshop instead we are going to share information we have learned during this study *(in progress)*

NOTE: the U.S. government shutdown in Dec 2018 and Jan 2019 has impacted final preparation for this workshop

UPDATED SLIDE

Review of Workshop Agenda

Time	Topic	Speaker(s)
8:30 – 9:00	Introduction, Background, Historical Overview	Sheila, Melissa, John
9:00 – 9:45	Establishing SOPs in Your Laboratory	Bruce, Jen, Eugene
9:45 – 10:30	Performance-Based Validation Data	Hari
10:30 – 10:45	**BREAK**	
10:45 – 11:30	Exploring Capabilities and Limitations	Keith, Lisa
11:30 – 12:00	Potential of New Technologies	Pete

LUNCH 12:00 – 1:00 pm

Time	Topic	Speaker(s)
1:00 – 1:30	DNA Transfer Studies	Sheila
1:30 – 2:00	Case Examples	Roger, Charlotte
2:00 – 2:30	Exploring Capabilities and Limitations	Eugene, Joel
2:30 – 3:00	Core Literature & Principles	John
3:00 – 3:15	**BREAK**	
3:15 – 3:45	Training and Establishing Expertise	Robin, Jack, Ray
3:45 – 4:00	NIST Study & Report	John, Rich
4:00 – 4:45	Panel Discussion	Sheila and others
4:45 – 5:00	Next Steps, Q&A, Summary	John, Melissa

UPDATED SLIDE

Additional Slides in Final Presentations

- After the workshop, final versions of the workshop slides will be posted at **http://strbase.nist.gov**

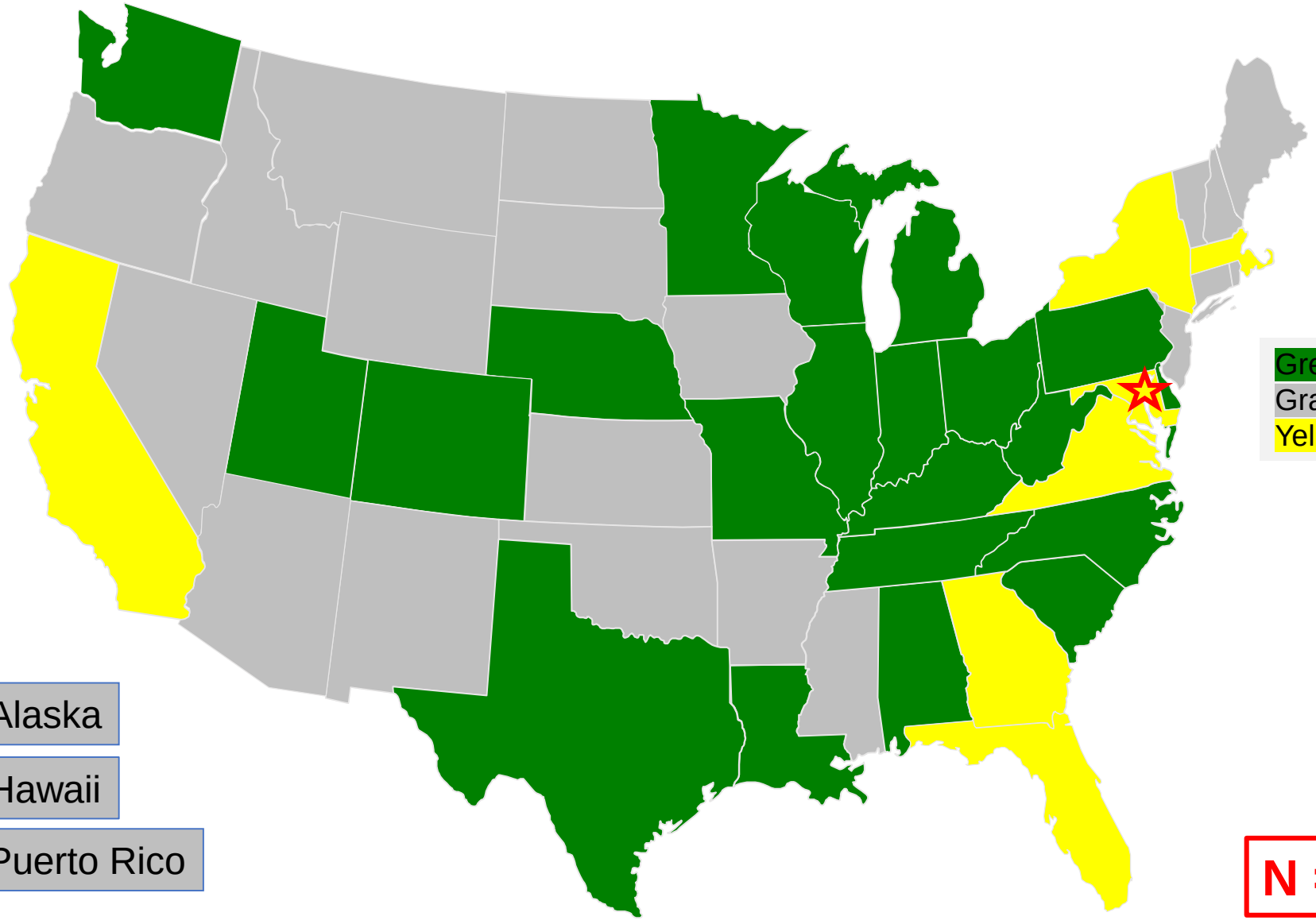
- Where to sign up for NIST information:
  - **https://www.nist.gov/topics/forensic-science**

NOTE: the U.S. government shutdown in Dec 2018 and Jan 2019 has impacted final preparation for this workshop

NEW SLIDE

AAFS 2019 Mixture Workshop Registrants

27 states and 6 other countries



Canada
Italy
Japan
Korea
Nepal
New Zealand

Green = participants
Gray = no attendees
Yellow = presenters

Alaska

Hawaii

Puerto Rico

<u>Those registered include</u>:
DNA analysts
DNA technical leaders
Academic researchers
Prosecutors
Defense attorneys
Commercial suppliers

N = 103 + 17 presenters

NEW SLIDE

Handling Questions

- Write down your questions
  - Bring them up to the front at a break or at lunch
- Can be answered during the panel discussion at the end or at the end of a group of talks if there is time

DNA Mixture Interpretation Principles: Observations from a NIST Scientific Foundation Review

AAFS 2019 Workshop #10 (February 18, 2019; Baltimore, MD)

Historical Overview and Study Background

John M. Butler

National Institute of Standards and Technology

What Has Changed Since the 2008 and 2011 AAFS Workshops Were Conducted?

Washington, DC (**2008**)

DNA Mixture Interpretation:
Principles and Practice in Component Deconvolution and Statistical Analysis

Background and Introductory Information

AAFS 2008 Workshop #16
Washington, DC
February 19, 2008

John M. Butler
Ann Marie Gross
Gary G. Shutler

Chicago, IL (**2011**)

DNA Mixture Analysis:
Principles and Practice of Mixture Interpretation and Statistical Analysis Using the SWGDAM STR Interpretation Guidelines

Background and Introductory Information

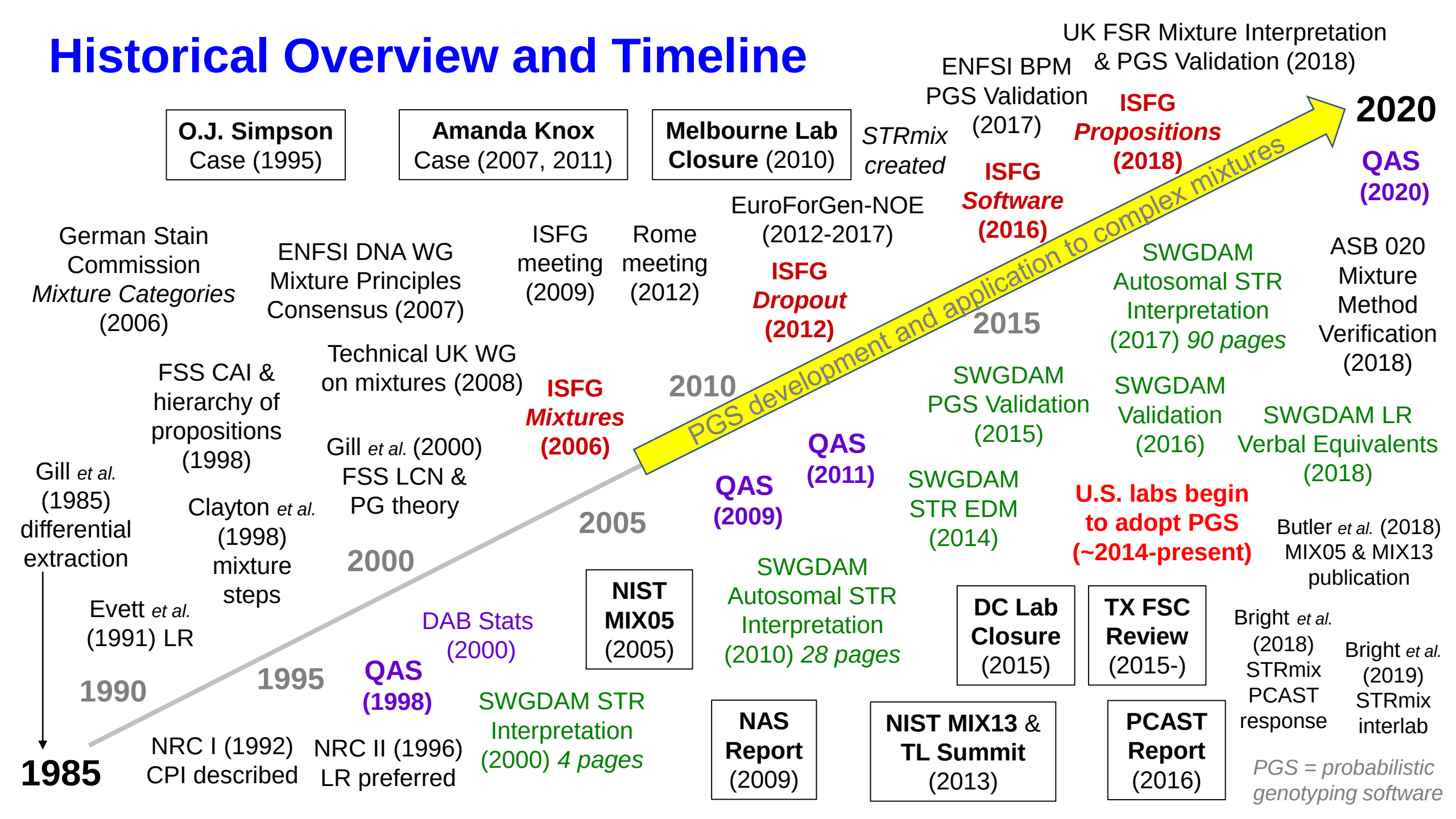
Chair: John M. Butler
Co-Chairs:
Michael D. Coble & Todd W. Bille

AAFS 2011 Workshop #17
Chicago, IL
February 22, 2011

Changing Landscape, Guidelines, and Approaches for DNA Mixture Interpretation Over the Years

	2008	2011	2019
Types of mixtures seen in laboratories	Mostly 2-person (most sexual assault)	Increasingly >2-person (more burglary)	Predominantly >2-person ("touch DNA" swabs)
SWGDAM interpretation guidelines in place	2000 (4-pages)	2010 (28-pages)	2017 (90-pages)
Statistical approach used in U.S. labs	RMP (when mixture was deconvoluted) or CPI (often with no stochastic threshold)	Mostly CPI (increasing use of stochastic thresholds)	Still have CPI but a growing use of likelihood ratios (LRs) and probabilistic genotyping software (PGS)

Historical Overview and Timeline



What are the biggest obstacles you face in your lab in terms of mixture interpretation?

(responses from a **November 2007** mixture workshop at NEAFS)

- Trying to be consistent in my interpretation and with coworkers
- **Consistency between analysts**
- No consistency – based on analysts discretion/experience; due to lack of consistent training
- Vague SOP leading to inconsistency between analysts due to differences in how “conservative” each analyst is
- There is a lot of “individual interpretation” in our lab
- Varying opinions between interpreting analysts due to lack of uniform guidelines
- Resistance to change from other analysts/supervisors
- Getting management to commit to guidelines that will be followed by everyone
- Where to draw the line without throwing away valuable data
- Partial minor contributors
- Stochastic effects in minor components
- STATS and presenting them in court so that the jury will understand them
- When to do stats and what stats to do in different cases
- Lack of concrete/uniform guidelines from statisticians

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Steps in DNA Analysis and Interpretation

swab

Gathering the Data

Understanding Results Obtained

Collection/Storage/ Characterization → Extraction/ Quantitation → Amplification/ Marker Sets → Separation/ Detection → Data → Stats → Report → result

Interpretation

Probabilistic Genotyping Software (PGS)

Inter-laboratory and Intra-laboratory Studies

Started with stains	NIST Mixed Stain Study #1 (1997) and #2 (1999)	1p, 2p, 3p mixtures
Started with DNA extracts	NIST Mixed Stain Study #3 (2001)	1p, 2p, 3p mixtures
Started with data files, analyst inputs, approaches varied	NIST MIX05 (2005)	2p mixtures
Started with data files, analyst inputs, approaches varied	NIST MIX13 (2013)	2p, 3p, 4p mixtures
Started with common PGS, fixed inputs (same AT)	STRmix (2019)	3p, 4p mixtures

Ideally, **we would like to characterize uncertainty for the entire system** (swab-to-result), but most interlaboratory studies only provide a partial picture of the variability because of the difficulty of generating and providing consistent mixture samples

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DNA Mixture Interpretation Approaches

- **Binary methods with simple** (mostly 2-person) **mixtures**
  - Statistical approaches: **LR** (Evett 1991, NRC 1996), **CPI** (NRC 1992, Budowle 2009, Bieber 2016)
  - Deconvolution/interpretation: (Clayton 1998, Evett 1998, Bill 2005)

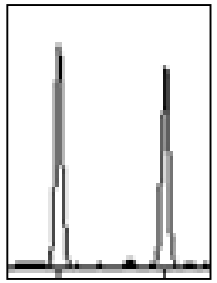
- **Probabilistic genotyping**
  - Theory: probability of drop-out and drop-in (Gill 2000, Balding 2009)
  - Early implementation: LoComationN (Gill 2007), gamma model (Cowell 2007)
  - Current PG software: **TrueAllele** (Perlin 2011), **FST** (Mitchell 2012), **LRmix** (Gill 2013), **STRmix** (Taylor 2013), **likeLTD** (Balding 2013), **LiRa** (Puch-Solis 2014), **Lab Retriever** (Inman 2015), **DNAmixtures** (Cowell 2015), **EuroForMix** (Bleka 2016), **CEESIt** (Swaminathan 2016), ***Kongoh*** (Manabe 2017), **GenoProof Mixture 3** (Götz 2017), **DNA Mixture Solution**, **LRmix studio**, **MaSTR**

NEW SLIDE

Math Analogy to DNA Evidence

$2 + 2 = 4$

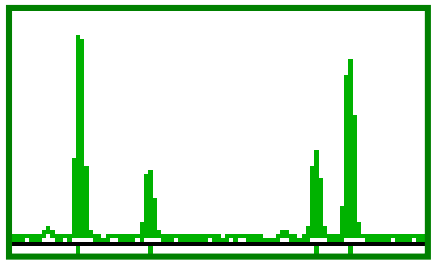
Basic Arithmetic



Single-Source DNA Profile
(DNA databasing)

$2\,x^2 + x = 10$

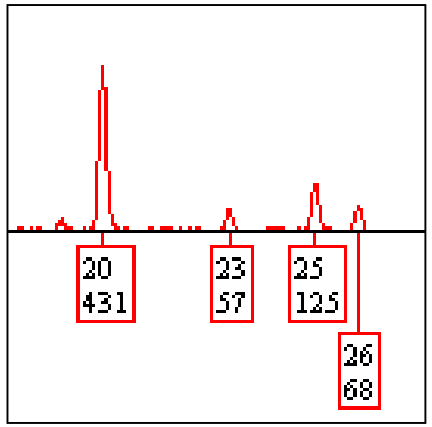
Algebra



Sexual Assault Evidence
(2-person mixture with high-levels of DNA)

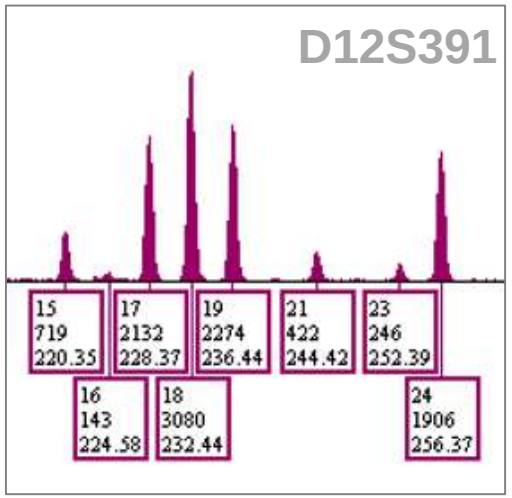
$$\int_{x=0}^{\infty} f(x)\,dx$$

Calculus



Touch Evidence
(>2-person, low-level, complex mixtures perhaps involving relatives)

[15,21] [18,19] [17,24]



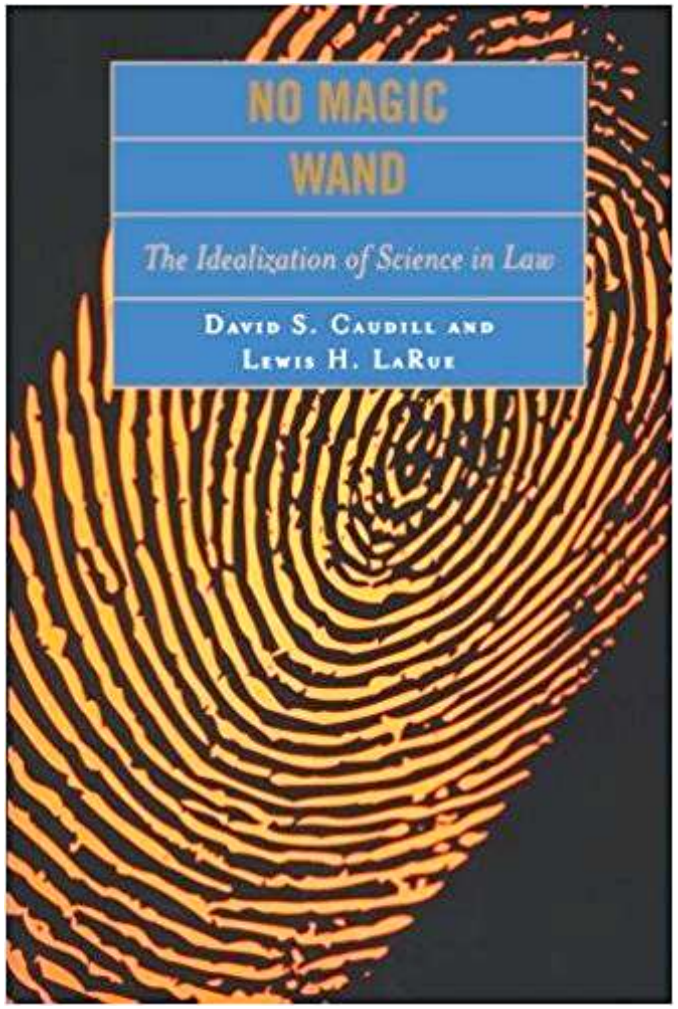
Complex Mixture

PROVED-It GlobalFiler data
3-person 1:4:4 (750pg)
Used in *FSIG* (2019) 40:1-8 interlab

NIST Scientific Foundation Reviews

Seeds for this activity were planted with two NCFS documents regarding “Technical Merit Evaluation of Forensic Science Methods and Practices” and Congressional funding to conduct these studies comes from money previously spent on the NCFS

Court Admissibility ≠ "Scientific Validity"



Rowman & Littlefield Publishers (2006)

170 pages

- This book discusses the *Daubert* trilogy "criteria" and issues that can arise because **science is often idealized by the law**

- Likewise, the law can sometimes be idealized by forensic scientists
  - **no one is justified in stating that a method being used is "scientifically valid" *because* it was accepted in court**…

Federal Rules of Evidence (FRE) 702 discussions are ongoing in an attempt to provide more clarity on this issue

Purpose of NIST Scientific Foundation Review

- Views of the National Commission on Forensic Science on Technical Merit Evaluation of Forensic Science Methods and Practices (June 2016): “It is the view of the NCFS that an institutional entity assigned a permanent independent scientific evaluation function would **facilitate the gathering of scientific research**, knowledge and expertise over time, **creating a service resource for forensic science**, technology research, and user communities. Development of a trusted and impartial process of evaluating technical merit of forensic practices and the presentation of data will ensure that all decisions rendered by the justice system are based on sound and current science.”

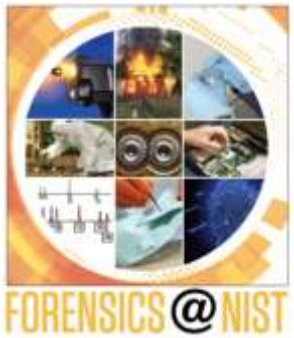
https://www.justice.gov/archives/ncfs/file/881796/download

NIST Forensic Science Activities

Conduct Research and Collaborate

Intramural Research

DNA
Digital
Fingerprints
Firearms
Footmarks
Statistics
Toxins
Trace



Extramural Research
funding a NIST Center of Excellence in Forensic Science (CSAFE: since 2014)

1920s - present

Partner with Community to Strengthen Policies and Practices

National Commission on Forensic Science (NCFS) with DOJ

2013 - 2017

Organization of OSAC Scientific Area Committees for FORENSIC SCIENCE

2013 - present

Convene Meetings to Examine Issues

Human Factors Working Groups (with NIJ)

2009 - present

Explore Scientific Foundations

Initial efforts with DNA mixture interpretation and bitemark analysis


NISTIR 8225 DRAFT

NIST Scientific Foundation Reviews

2017 - present

NIST FORENSIC SCIENCES

NIST National Institute of Standards and Technology U.S. Department of Commerce

NIST Scientific Foundation Review

- Requested and funded by Congress to examine forensic disciplines

- **Initial pilot study on DNA mixture interpretation**
  - Project begun in September 2017

- **6 NIST team members meet weekly with regular input from 13 forensic practitioners/researchers** (our "DNA Mixture Resource Group")

- **Examining the literature and studying issues…**
  - **>500 articles collected on DNA mixture interpretation**
  - Seeking to compile underlying principles and assess claims

- Report has taken longer to write than initially expected
- Government shutdown (Dec 2018 to Jan 2019)

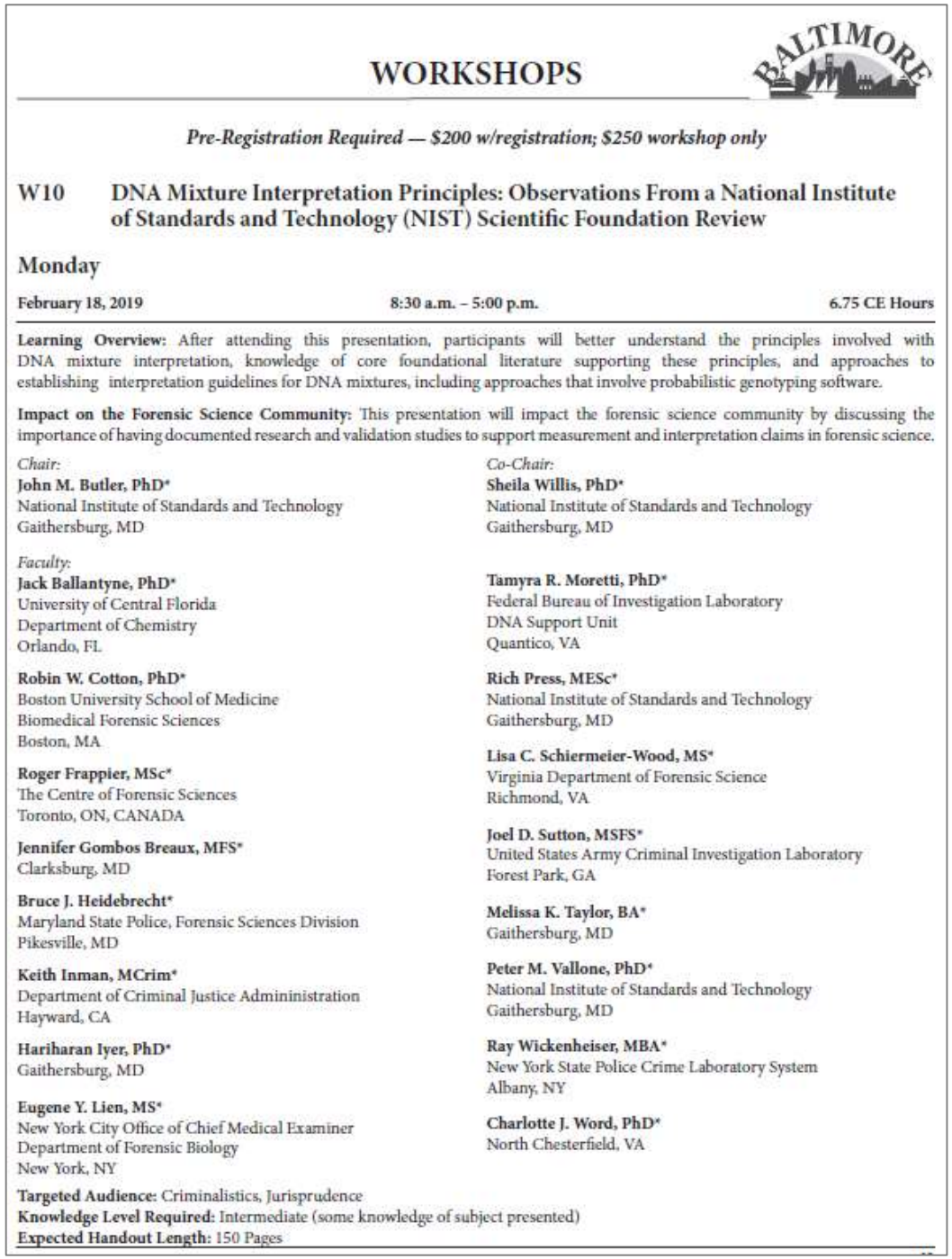
- **Report is being written for release** (as a draft) **later this year**
  - Plan to collect public comment on the report and reactions to its findings
  - AAFS 2019 workshop to discuss the topic and report contents

NEW SLIDE

Input Provided by a DNA Mixture Resource Group

Name	Affiliation
Jack Ballantyne	University of Central Florida
Todd Bille	ATFE Laboratory, DNA Technical Leader
Jennifer Breaux	Montgomery County Police Crime Lab
Robin Cotton	Boston University School of Medicine
Roger Frappier	Centre of Forensic Sciences - Toronto
Bruce Heidebrecht	Maryland State Police, DNA Technical Leader
Keith Inman	Cal State East Bay & forensic DNA consultant
Eugene Lien	NYC OCME, DNA Technical Leader
Tamyra Moretti	FBI Laboratory, DNA Support Unit
Lisa Schiermeier-Wood	Virginia Department of Forensic Sciences
Joel Sutton	Defense Forensic Science Center, USACIL
Ray Wickenheiser	NYSP Laboratory Director (former ASCLD President)
Charlotte Word	forensic DNA consultant

9 practitioners (3 Federal, 3 state, 2 local, 1 Canadian), **4 academics/consultants**



WORKSHOPS

Pre-Registration Required — $200 w/registration; $250 workshop only

W10 **DNA Mixture Interpretation Principles: Observations From a National Institute of Standards and Technology (NIST) Scientific Foundation Review**

Monday

February 18, 2019 **8:30 a.m. – 5:00 p.m.** **6.75 CE Hours**

Learning Overview: After attending this presentation, participants will better understand the principles involved with DNA mixture interpretation, knowledge of core foundational literature supporting these principles, and approaches to establishing interpretation guidelines for DNA mixtures, including approaches that involve probabilistic genotyping software.

Impact on the Forensic Science Community: This presentation will impact the forensic science community by discussing the importance of having documented research and validation studies to support measurement and interpretation claims in forensic science.

Chair:
John M. Butler, PhD*
National Institute of Standards and Technology
Gaithersburg, MD

Co-Chair:
Sheila Willis, PhD*
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Gaithersburg, MD

Faculty:
Jack Ballantyne, PhD*
University of Central Florida
Department of Chemistry
Orlando, FL

Robin W. Cotton, PhD*
Boston University School of Medicine
Biomedical Forensic Sciences
Boston, MA

Roger Frappier, MSc*
The Centre of Forensic Sciences
Toronto, ON, CANADA

Jennifer Gombos Breaux, MFS*
Clarksburg, MD

Bruce J. Heidebrecht*
Maryland State Police, Forensic Sciences Division
Pikesville, MD

Keith Inman, MCrim*
Department of Criminal Justice Admininistration
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Hariharan Iyer, PhD*
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Virginia Department of Forensic Science
Richmond, VA

Joel D. Sutton, MSFS*
United States Army Criminal Investigation Laboratory
Forest Park, GA

Melissa K. Taylor, BA*
Gaithersburg, MD

Peter M. Vallone, PhD*
National Institute of Standards and Technology
Gaithersburg, MD

Ray Wickenheiser, MBA*
New York State Police Crime Laboratory System
Albany, NY

Charlotte J. Word, PhD*
North Chesterfield, VA

Targeted Audience: Criminalistics, Jurisprudence
Knowledge Level Required: Intermediate (some knowledge of subject presented)
Expected Handout Length: 150 Pages

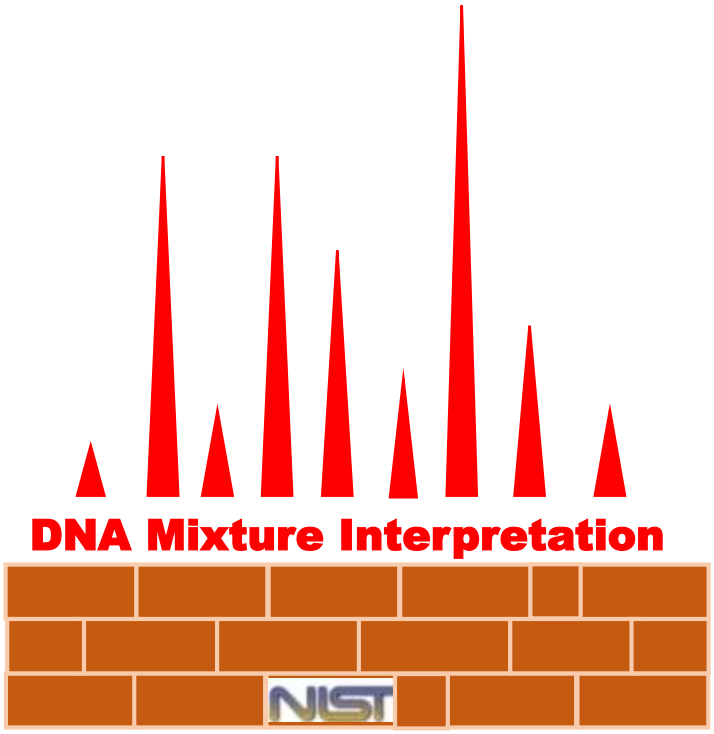
AAFS 2019 Workshop (W10) to Discuss Study Findings and Process Used

"DNA Mixture Interpretation Principles: Observations from a NIST Scientific Foundation Review"

- **Monday, February 18, 2019, Baltimore Convention Center**

- **Program Description**: Presenters will share observations from a scientific foundation review conducted this past year by a NIST review team with input from a resource group of practitioners and technical leaders.

Thank you for your attention!



NIST FORENSIC SCIENCES

www.nist.gov/forensics

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