# Forensic DNA Mixture Interpretation

**MAFS Workshop** 

Milwaukee, WI **September 25, 2012** 



Dr. John M. Butler & Dr. Michael D. Coble
National Institute of Standards and Technology

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# Forensic DNA Mixture Interpretation

# Introductions

### **MAFS Workshop**

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# Outline for Today's Workshop

- Introductions (John, Mike & each of you!)
- Mixture Fundamentals & Literature Review (John)

### **BREAK**

Statistical Approaches for Mixtures (Mike)

### LUNCH BREAK

Thresholds & Low Level Mixtures (John)

### BREAK

Probabilistic Genotyping & Software (Mike)



# Dr. John M. Butler

http://www.cstl.nist.gov/biotech/strbase/butler.htm

### **Experience**

- University of Virginia/FBI Laboratory (1992-1995)
  - Work performed in Bruce McCord's lab
- NIST NRC Postdoc (1995-1997)
- GeneTrace Systems Inc (1997-1999)
- NIST Human Identity Project Leader (1999-present)
- Ph.D. dissertation (Aug 1995): "Sizing and quantitation of polymerase chain reaction products by capillary electrophoresis for use in DNA typing"
- Forensic DNA Typing textbook (now in its 2<sup>nd</sup> Edition)
- STRBase website: http://www.cstl.nist.gov/biotech/strbase/
- Family: wife Terilynne and 6 children
- Hobbies: reading, writing, and making PowerPoint slides

Contact Information john.butler@nist.gov 301-975-4049







# Dr. Michael D. Coble

http://www.nist.gov/mml/biochemical/genetics/michael\_d\_coble.cfm

### **Experience**

- George Washington University/AFDIL (1996-2004)
  - Work performed with Tom Parsons
- NIST NRC Postdoc (2004-2006)
- AFDIL Research Section Chief (2006-2010)
- NIST Applied Genetics Group (2010-present)
- Ph.D. dissertation (Jan 2004): "The Identification of Single Nucleotide Polymorphisms in the Entire Mitochondrial Genome to Increase the Forensic Discrimination of Common HV1/HV2 Types in the Caucasian Population"
- Family: wife Karen and 3 children
- Hobbies: Fantasy baseball and football team owner

Contact Information
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# **NIST History and Mission**

- National Institute of Standards and Technology (NIST) was created in 1901 as the National Bureau of Standards (NBS). The name was changed to NIST in 1988.
- NIST is a non-regulatory agency within the U.S. Department of Commerce with a mission to develop and promote measurement, standards, and technology to enhance productivity, facilitate trade, and improve the quality of life.
- NIST supplies over 1,300 Standard Reference Materials (SRMs) for industry, academia, and government use in calibration of measurements.
- NIST defines time for the U.S.

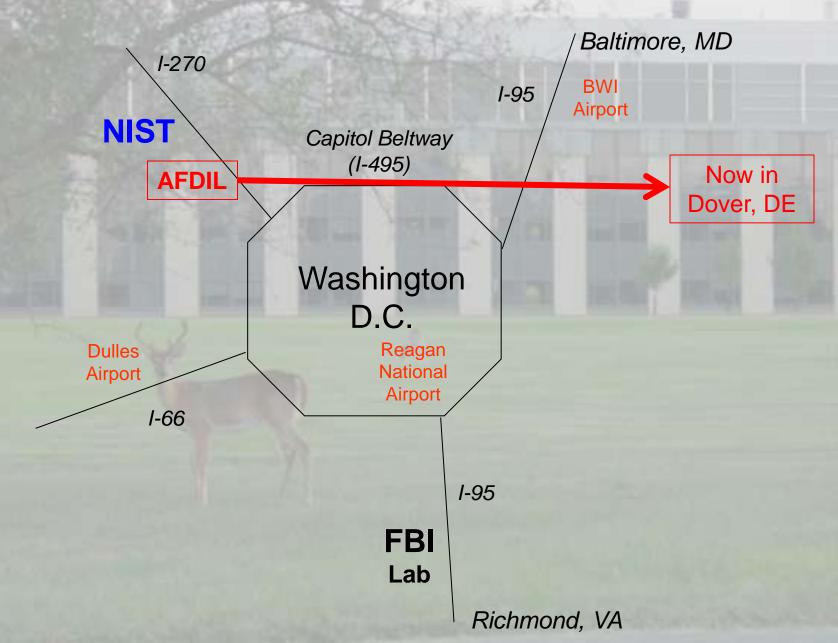


\$686 for 3 jars

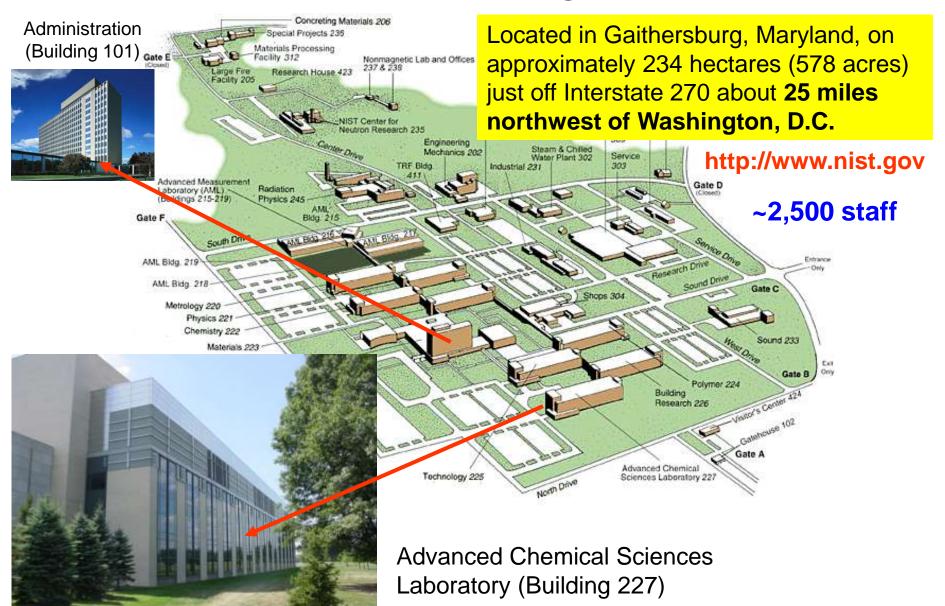


**DNA typing standard** 

# Location of NIST



# NIST Gaithersburg Campus





### Group Leader

# NIST Applied Genetics Group



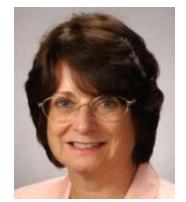




Mike Coble



Margaret Kline



Marcia Holden



Pete Vallone



Patti Rohmiller Office Manager



Becky Hill



Ross Haynes



Erica Butts



Kevin Kiesler



Bringing calibration to clinical DNA diagnostics, speed to DNA testing, and technology to the scales of justice



# APPLIED GENETICS Group

# Major Programs Currently Underway

### Forensic DNA

- STRBase website
- New loci and assays (26plex)
- STR kit concordance & new autosomal STR loci
- Ancestry SNP assays
- Low-template DNA studies
- Mixture interpretation research and training
- Variant allele cataloging and sequencing
- ABI 3500 validation
- Training workshops to forensic DNA laboratories
- Y-STR markers
- Validation experiments, information and software tools
- Textbooks 3<sup>rd</sup> ed. (3 volumes)

### Clinical Genetics

- Huntington's Disease SRM
- CMV SRM
- Exploring future needs

### Ag Biotech

 "universal" GMO detection/ quantitation (35S promoter)

### DNA Biometrics

- Rapid PCR methods
- Efforts to standardize testing of future portable DNA systems
- Kinship analysis

### Cell Line Authentication

ATCC documentary standard



# NIST Human Identity Project Teams within the Applied Genetics Group

### Forensic DNA Team

**Guest Researcher** 

### **DNA Biometrics Team**

Funding from the FBI S&T Branch

through NIST Information Access Division

Funding from the **National Institute of Justice (NIJ)** through NIST Office of Law Enforcement Standards



John Butler



Mike Coble



Becky Hill



Margaret Kline

Returned to Spain in July 2012

Manuel **Fonde**vila Alvarez

Data

Analysis

Support



Pete Vallone



Erica Butts



Kevin Kiesler

STRBase, Workshops & Textbooks

Mixtures, mtDNA & Y

Concordance & LT-DNA

SRM work, variant alleles & Cell Line ID



Dave Duewer

Rapid PCR, ABI 3500
Direct PCR & DNA
& Biometrics Extraction

PLEX-ID & NGS Exploration





Office Manager Patti Rohmiller



http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm

# Forensic DNA Typing Textbook

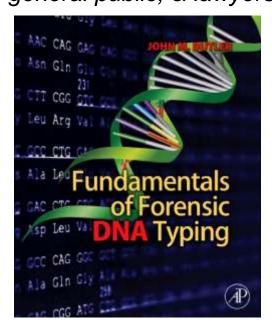
# 3<sup>rd</sup> Edition is Three Volumes

Now part of my job at NIST (no royalties are received)



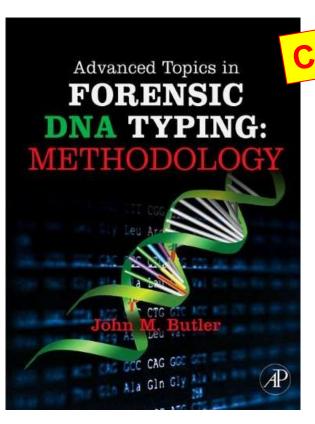
John Butler

For beginning students, general public, & lawyers



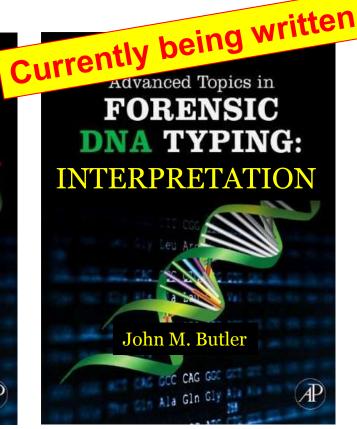
**Sept 2009** 

~500 pages



August 2011

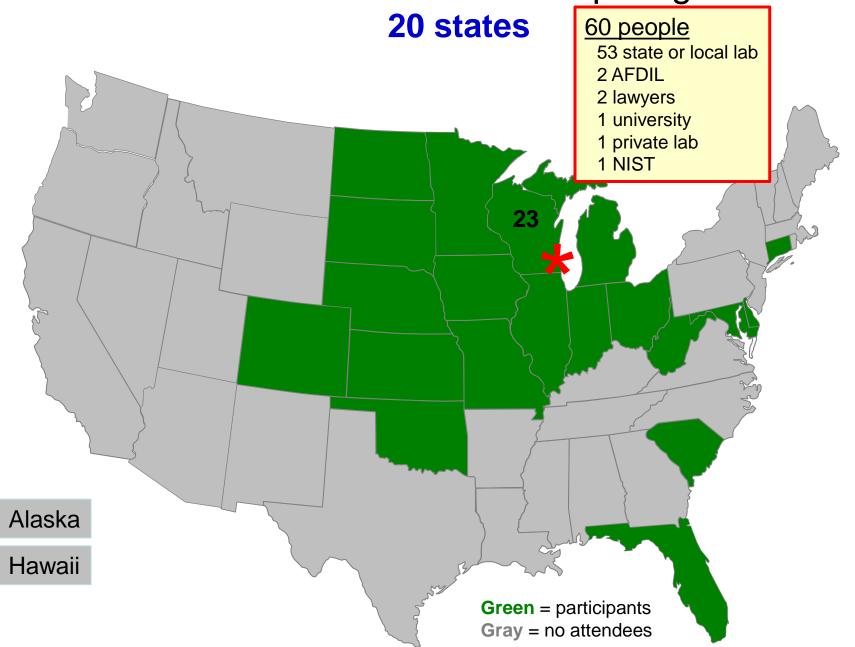
~700 pages



Fall 2013

~500 pages

# MAFS 2012 Mixture Workshop Registrants



# Understanding the Audience Here

## Name, Laboratory, what you hope to learn in workshop

- Where is everyone from?
  - State lab?
  - Local lab?
  - Private lab?
- Experience level?
  - Less than 1 year?
  - 1-3 years?
  - >3 years?

### **Benefits of this exercise:**

- 1) We can tailor remarks to your interests and needs
- 2) You feel more comfortable asking questions and being involved when you know others

- STR kits in use?
  - Profiler Plus/COfiler
  - Identifiler (Direct, Plus)
  - PowerPlex 16 (HS)
  - Y-STRs?
- Instrumentation is use?
  - ABI 310
  - ABI 3100/3130/3130xl
  - ABI 3500/3500xl
- Software in use?
  - GeneMapperID
  - GeneMapperID-X
  - Other?

# Contributors to These Workshop Slides













Becky Hill

Erica Butts

Bruce McCord

Robin Cotton

Catherine Grgicak

Charlotte Word

**NIST** 

**NIST** 

Florida International University Boston University Boston University

Consultant

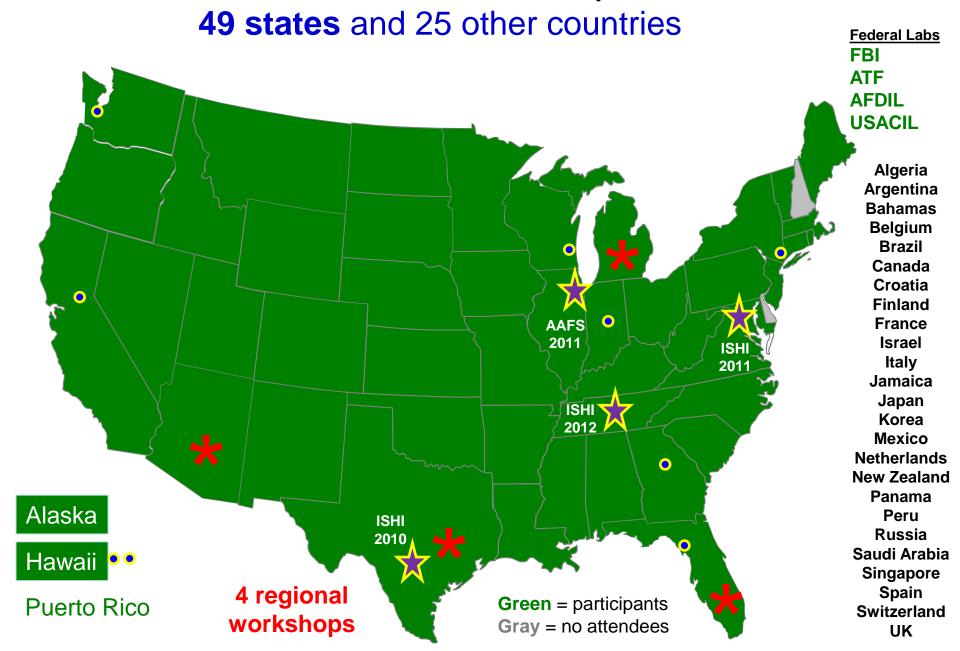
**STRs** 

**ABI 3500** 

CE

**Mixture Interpretation** 

# 2010-2012 Mixture Workshop Attendees



# STRBase Mixture Section

http://www.cstl.nist.gov/strbase/mixture.htm

- Slides from various mixture workshops
- SWGDAM Mixture Committee Resource Page
  - http://www.swgdam.org/resources.html
- Information and links to mixture software
- Literature listing by topic

# NIST and NIJ Disclaimer

<u>Funding</u>: Interagency Agreement 2010-DN-R-7121 between the National Institute of Justice and NIST Office of Law Enforcement Standards

Points of view are the presenters and do not necessarily represent the official position or policies of the US Department of Justice or 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 the National Institute of Standards and Technology nor does it imply that any of the materials, instruments or equipment identified are necessarily the best available for the purpose.

### SWGDAM and FBI CODIS Loci WG Disclaimer...

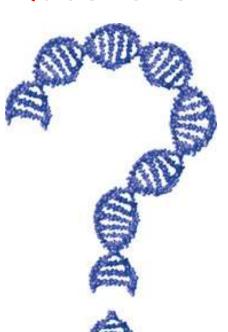
# Now on with the workshop...

Our team publications and presentations are available at: http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm

Funding from the National Institute of Justice (NIJ) through NIST Office of Law Enforcement Standards



**Questions?** 



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