



NGT National Institute of Standards and Technology • U.S. Department of Commerce

(a)



STRBase and Information **Resources on Forensic DNA**

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NIST Fellow & Applied Genetics Group Leader

Forensics@NIST 2012 Meeting

Gaithersburg, MD November 28, 2012



Information Gathering and Sharing

- We live in the information age and need to share what we learn as scientists with others
- Sharing information impacts validation of techniques, which impact court use of the technique
- DNA is often referred to as the "gold standard" in forensic science because of the scientific studies performed and information sharing that has occurred
- You need a good library (information collection) to be successful in developing any scientific discipline
- Knowing the literature provides a solid foundation for research and future work





Presentation Outline

Information Input

- Information gathering efforts within our group
- Forensic DNA literature
- Meetings where we learn from others (& present)

Information Output

- STRBase website its origin and content
- Training workshops
- Forensic DNA Typing textbooks

Committee contributions





Forensic DNA Library Books Located in 227/B250 and B224

Have purchased >300 books on topics related to forensic DNA analysis as of November 2012



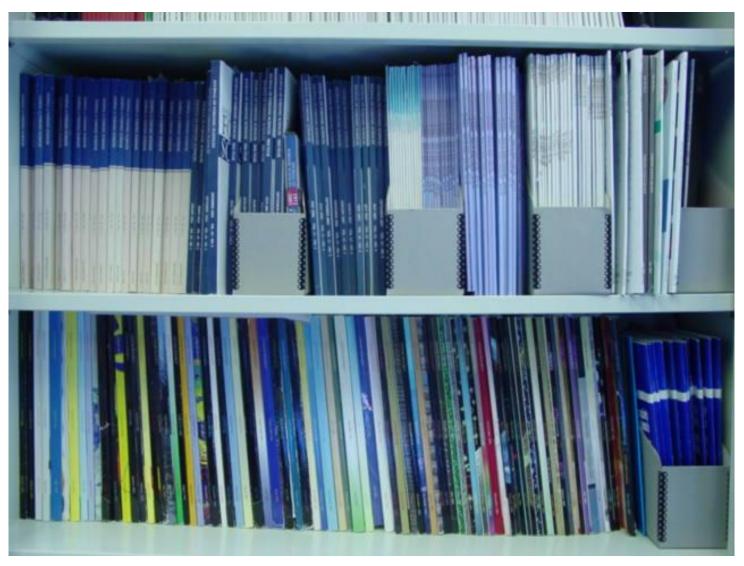


Initially funded from 2002-2007 by PECASE award money



Collection of Journals in Our Group Library

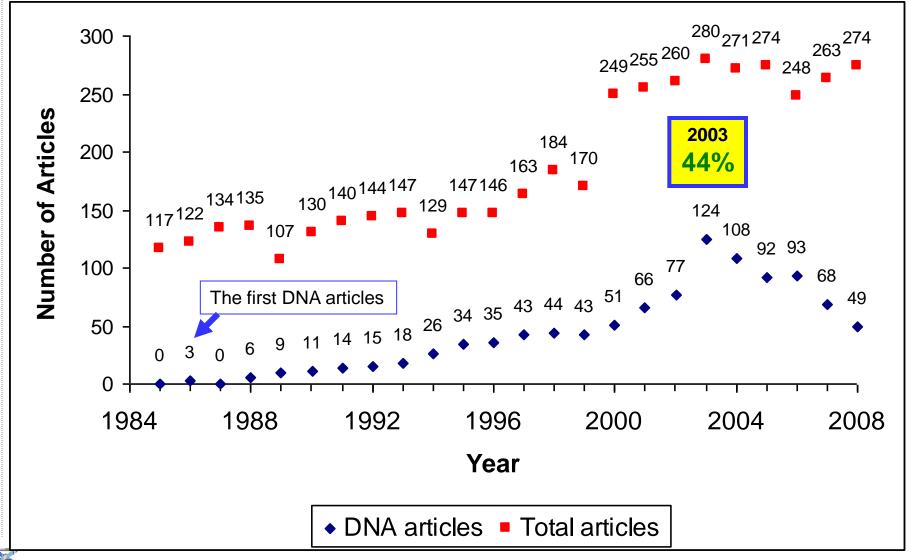
We now have on-line access to all forensic DNA journals







Journal of Forensic Sciences DNA publications vs total articles



From PubMed searches performed Feb 2009 using http://www.ncbi.nlm.nih.gov/sites/entrez



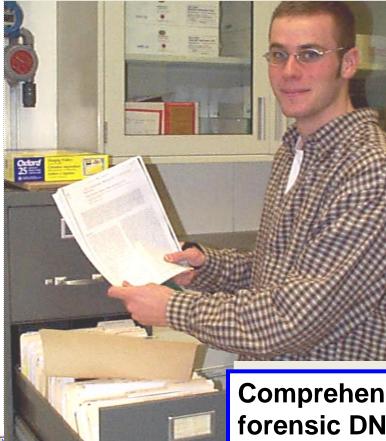
Forensic Science Publications





Our Project Team Library

>9,000 references gathered and cataloged in Reference Manager



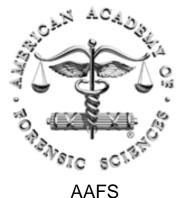
 Started by Christian Ruitberg •Maintained now by John Butler •Updated regularly and provided to **NIST Human Identity Project Team**

💱 Reference Manager - Reference List - STR_REF Database: Generic Reference ID 364									
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Title 1	Gene Print ^{TM STR Systems Technical Manual}								
Authors 1									
Pub Date 1* 🛛 🛅	/ /1995 Other								
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Agrawal,									

Comprehensive set of forensic DNA articles



We Attend & Contribute Each Year to Numerous Scientific Meetings



ESFG

International Society of Forensic Genetics



SWGDAM



National CODIS Conference



Advanced DNA Technical Workshop









Arlington, Virginia





AAFS 2009 Topics Regarding Forensic DNA

From abstracts of presentations at AAFS meeting in Denver, CO (Feb 2009)

- Improved DNA extraction
- Predicting hair color and ancestry with SNPs
- X-chromosome STRs
- Familial searching
- Y-STRs and mixtures
- Low level DNA samples
- miniSTRs
- DNA screening assays
- Optimizing database labs
- Microfluidic biochip systems

- Use with property crimes
- Recovery from handguns
- DNA from IEDs
- Expert systems
- Automation with robotics
- DNA quantitation qPCR
- PCR directly from blood
- mtDNA
- RNA
- Non-human DNA (dogs & cows)
- Mixture interpretation



Collection of Notes from Meetings Attended

Detailed notes are taken at every meeting we attend and shared with the group when we return to NIST (presentations are made to our group at the next group meeting)







Good information input improves output... Some Fruits of a Good Literature Collection

Review Articles

J Forensic Sci, March 2006, Vol. 51, No. 2 doi:10.1111/j.1556-4029.2006.00046.x Available online at: www.blackwell-synergy.com

John M. Butler,¹ Ph.D.

Genetics and Genomics of Core Short Tandem Repeat Loci Used in Human Identity Testing

Anal. Chem. 2007, 79, 4385-4384

Analytical Chemistry (June 15, 2007 issue)

Forensic Science

T. A. Brettell*

Department of Chemical and Physical Sciences, Cedar Crest College, 100 College Drive, Allentown, Pennsylvania 18104-6196

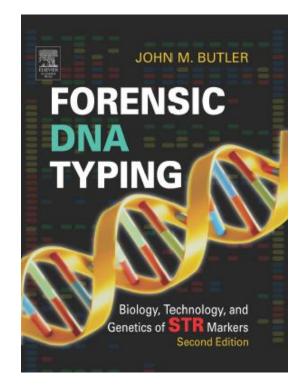
J. M. Butler

Biochemical Science Division, National Institute of Standards and Technology, Gaithersburg, Maryland 20899-8311

J. R. Almirall

Department of Chemistry and Biochemistry and International Forensic Research Institute, Florida International University, University Park, Miami, Florida 33199

<u>Textbooks</u>

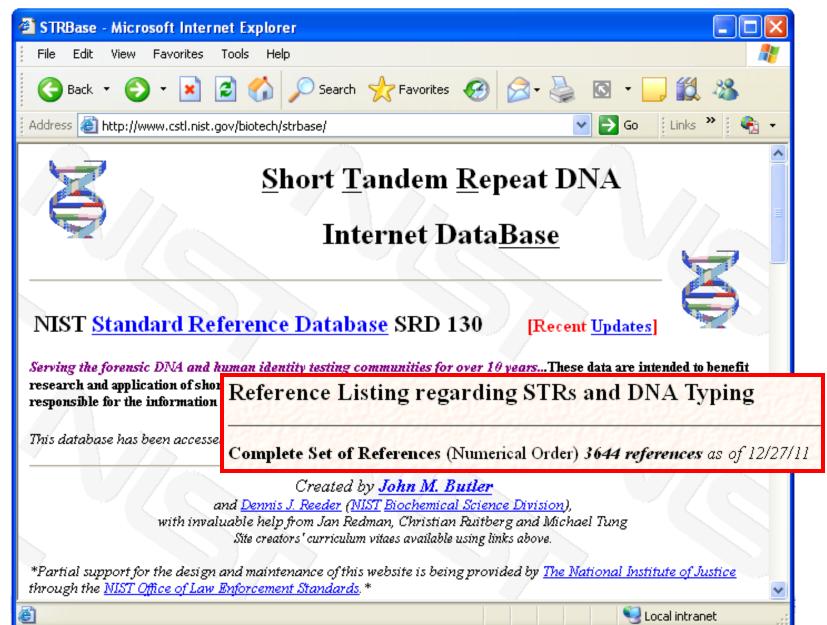


2nd Edition 688 pp. Feb 2005



And a Useful Reference Website...

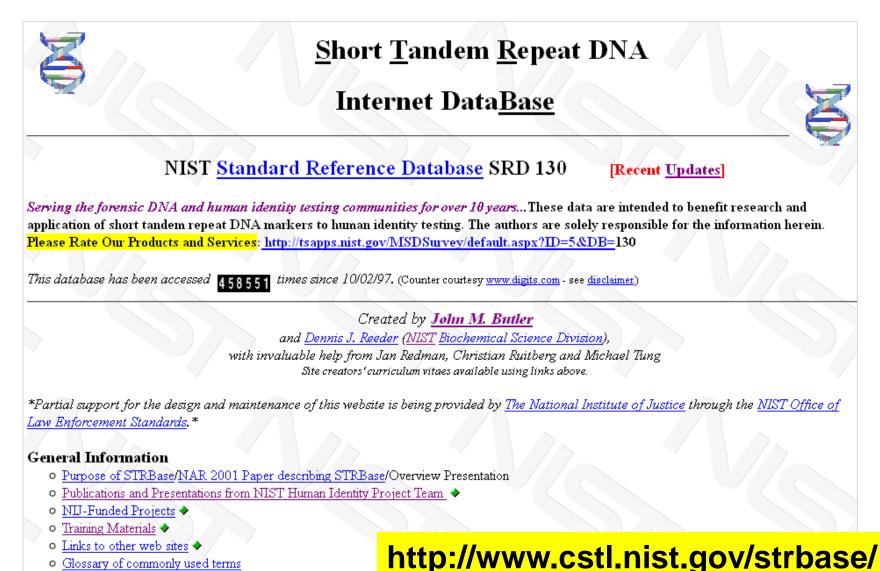
Applied Genetics





NIST STRBase Website

Serving the Forensic DNA Community for >15 Years





A Brief History of the STRBase Website

- Initial information was collected on STR markers while working on my PhD dissertation in 1993-1995
- Started a review article in 1996 while a NIST postdoc but wanted to create a dynamic rather than an out-of-date resource
- Created hundreds of individual web pages that were hyperlinked together
- Website launched in July 1997
- Became a NIST Standard Reference Database (SRD 130) because of its high visibility
- I continue to update the website (via an HTML editor)...





Benefits of Website like STRBase http://www.cstl.nist.gov/strbase

- Develops expertise when collecting information
- Requires NIST to stay up-to-date with field
- Provides transparency to our team's work
- Training tool and resource for the world
- Respected resource for >15 years
- ~10,000 pages of information available now
- >450,000 hits cumulative
- Method for sharing information (PowerPoint files, population data, etc.)

STRBase be a model for other forensic disciplines in sharing information with the forensic science community





Review of Some Recent STRBase Additions

Forensic STR Information

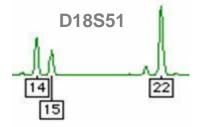
- o STRs101: Brief Introduction to STRs
- <u>Core Loci</u>: FBI CODIS Core STR Loci and European Core Loci
- STR Fact Sheets (observed alleles and PCR product sizes)
- <u>Multiplex STR kits</u>
- <u>Sequence Information (annotated)</u>
- o <u>Variant Allele Reports</u> 🔶
- o <u>Tri-Allelic Patterns</u> 🔶
- Mutation Rates for Common Loci
- <u>Published PCR primers</u>
- o <u>Y-chromosome STRs</u> ◆
- Low-template DNA Information
- Mixture Interpretation
- <u>Kinship Analysis</u>
- o <u>miniSTRs (short amplicons)</u> 🔶
- Null Alleles discordance observed between STR kits
 - STR Reference List now 3687 references





Information on Variant Alleles http://www.cstl.nist.gov/strbase/var_tab.htm

- We collect contributions from all over the world where unusual results have been observed with STR data
- Enables laboratories to check if others have seen a specific variant allele or tri-allelic pattern
- Currently (as of Oct 31, 2012 update)
 657 variants at 41 loci
 326 tri-allelic patterns at 33 loci



Type 1 tri-allelic pattern

From D2S1338 Variants Table (http://www.cstl.nist.gov/strbase/var_D2S1338.htm)

Allele Designation	Allele Size	Instrument	Amp Kit*	Contributor	Verification/Conformation Method(s)	Notes	Frequency
11	290.61	ABI 310	ID	Cintia alves, IPATIMUP, Porto, Portugal	Re-extracted and re-amplified	Portuguese Caucasian sample	1 in 780



Mixture Section of STRBase

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

- **Training workshop slides** (thousands of slides of training materials available from 7 workshops)
- SWGDAM Mixture
 Committee resource page
 (contains worked mixture examples
 by Bruce Heidebrecht, Maryland State
 Police DNA Technical Leader)
- Links to mixture
 interpretation software
 (currently 12 links)
- Literature references (currently 144 articles)

Literature listing by topic for 144 articles

Topic category	# References
Mixture Principles & Recommendations	13
Setting Thresholds	11
Stutter Products & Peak Height Ratios	19
Stochastic Effects & Allele Dropout	18
Estimating the Number of Contributors	15
Mixture Ratios	9
Statistical Approaches	23
Low Template DNA Mixtures	8
Separating Cells to Avoid Mixtures	3
Software (plus 12 websites)	7
Probabilistic Genotyping Approach	11
General Information on Mixtures	7





Outreach via Presentations

from the NIST Applied Genetics Group

- Present our research at scientific conferences
- Conduct training workshops at forensic labs
- Visit universities with forensic science programs
- Teach via webinars on specific topics
 - NIST will host a mixture interpretation webinar on April 12, 2013 (goal is to reach DNA analysts world-wide)

In the past 2 years (Oct 2010-Nov 2012): 138 talks & posters 20 training workshops

Slide handouts and posters available at http://www.cstl.nist.gov/strbase/NISTpub.htm





Scientific Conferences (Past 2 Years)





Crime Labs Visited (Past 2 Years)

Presentations were given in each lab reviewing NIST DNA research and providing requested training on forensic DNA topics



<u>ř</u>

April 2012



June 2012

Sept 2012



Universities Visited (Past 2 Years)



ТНЕ	GEORGE
WASI	HINGTON
UNI	VERSITY
W A S H	INGTON DC





April 2011 Sept 2011 March 2012

May 2012* *commencement address



April 2011 May 2012

April 2012



April 2011



April 2011 Nov 2011





March 2011



Nov 2011





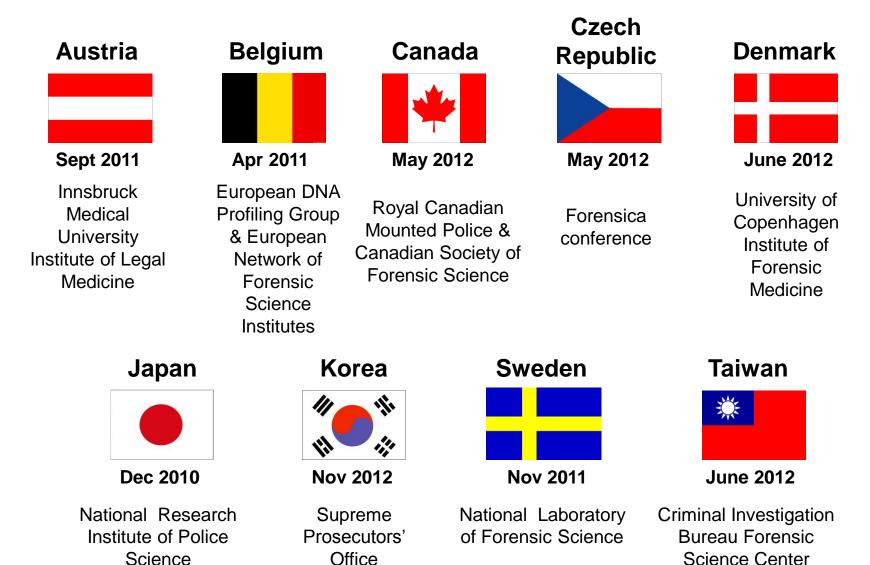
ENCE CENTER

Nov 2011

Nov 2011



Other Countries Visited (Past 2 Years)



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NIST Applied Genetics Group Hosted a **Spanish Guest Researcher** (Jan 2011 – July 2012)





Dr. Manuel Fondevila



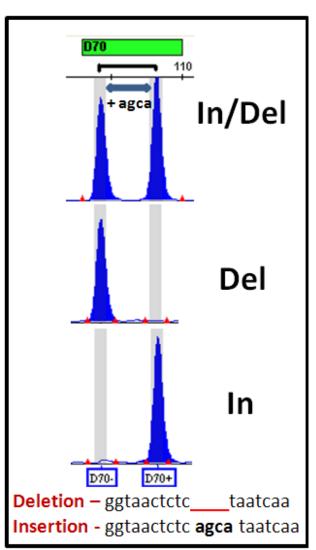
Studied **commercial 30plex** (Qiagen DIPlex) and a **home-brew 38plex** in **U.S. population samples**

Int J Legal Med (2012) 126:725-737 DOI 10.1007/s00414-012-0721-7 Int. J. Legal Med. (2012) 126: 725-737

ORIGINAL ARTICLE

Forensic performance of two insertion-deletion marker assays

M. Fondevila • C. Phillips • C. Santos • R. Pereira • L. Gusmão • A. Carracedo • J. M. Butler • M. V. Lareu • P. M. Vallone





Publications on Forensic DNA

from the NIST Applied Genetics Group

144 publications since 2002

- 40 in the past 2 years
- Includes journal articles, book chapters, and textbooks
- References are all listed on STRBase
 - http://www.cstl.nist.gov/strbase/NISTpub.htm
 - Many are available directly from STRBase



Most of our articles are published in *Forensic Sci. Int. Genetics* – currently the highest impact journal in the field

136 page report written by Kevin Kiesler

NIST Report to the FBI: Plex-ID Electrospray Time-of-Flight Mass Spectrometer for Mitochondrial DNA Base Composition Profiling

Experiments performed and report written by: Kevin Kiesler, M.S. (NIST)

Under the direction of: Dr. Peter Vallone (NIST)



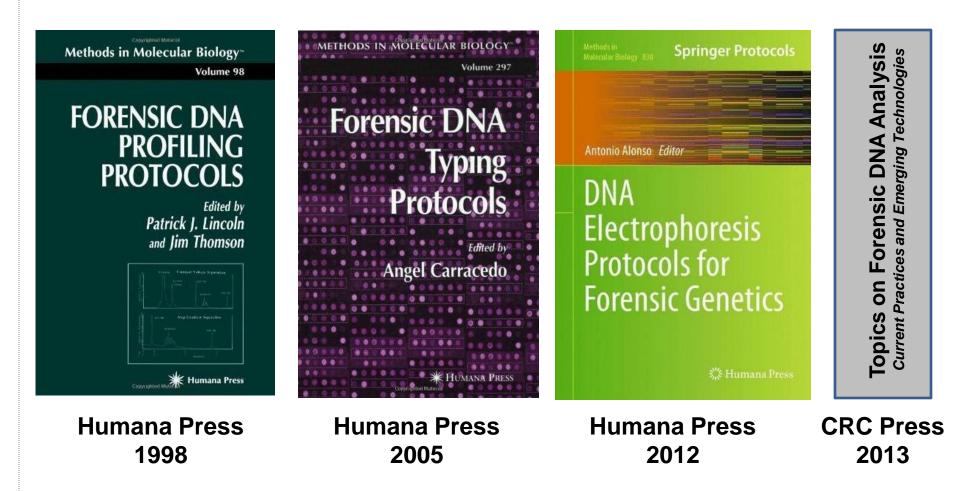
Application Review on Forensic Science

appears every other year in June 15 issue of Analytical Chemistry

Anal. Chem. 2005, 77, 3839–3860		Year Published	Years Covered	# Articles Reviewed	# DNA Articles Reviewed			
Forensic Science		2005	2003 & 2004	789	250			
T. A. Brettell* Anal. Chem. 2007, 79, 4365–4384			2007	2005 & 2006	560	181		
Office of Forensic 1200 Negron Roa	Forensic Science		е	2009	2007 & 2008	552	163	
J. M. Butler	T. A. Brettell*	Anal. Chem. 2009, 8		2011	2009 & 2010	575	122	
National Institute (R. Saferstein	Department of Cha Allentown, Pennsy J. M. Butler		c Scie	nce				
Box 1334, Mount	Biochemical Scien J. R. Almirall Department of Che University Park, M			chemist		2011 , <i>83</i> , 4539	-4556 REVIEW pubs.acs.org/ac	
Oniversity Fails, in J. M. Butler Forensic Science Biochemical Scien T. A. Brettell J. R. Almirall Department of Chemical and Physical Sciences, Cedar Crest College, 100 College Drive, Allentown, Pennsylvania 18104						wn, Pennsylvania 18104-6196,		
Department of Cr University, Univer J. M. Butler				r Science Division, National Institute of Standards and Technology, Gaithersburg, Maryland 20899-8312, United States				
Department of Chemistry and Biochemistry and International Forensic Research Institute, Florida International University, University Park, Miami, Florida 33199, United States					a International University,			



Contributing to Book Compilations

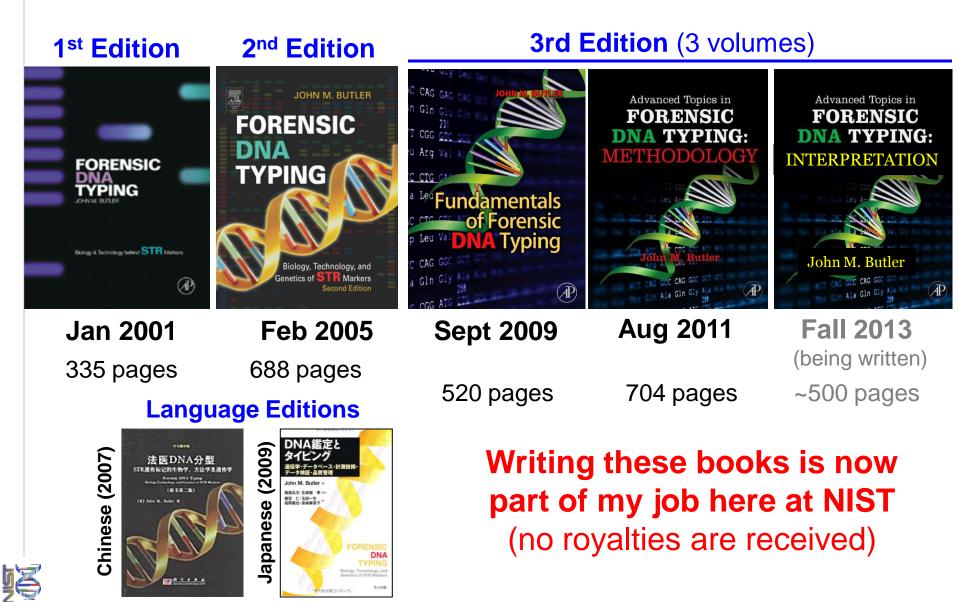


The NIST Applied Genetics Group has contributed to every major book compilation regarding forensic DNA over the past 15 years





Forensic DNA Typing Textbooks Have Set the Standard for the Field





Current Committee Contributions



SWGDAM (Scientific Working Group on DNA Analysis Methods)

http://www.swgdam.org

John Butler – Mixture Committee (chair) Pete Vallone – Rapid DNA Committee (chair) Mike Coble – Enhanced Detection Methods & Interpretation Committee

Held each Jan & July (Fredericksburg, VA)

NIST/NIJ Technical Working Group on Biological Evidence Preservation

http://www.nist.gov/oles/forensics/crime_scene.cfm

Margaret Kline serves as a member

Virginia Department of Forensic Science – Scientific Advisory Committee

http://www.dfs.virginia.gov/about/saCommittee.cfm

John Butler serves as a member of the biology sub-committee

North Carolina State Bureau of Investigation Forensic Science Advisory Board http://www.ncdoj.gov/About-DOJ/State-Bureau-of-Investigation/Crime-Lab/Forensic-Advisory-Board.aspx

Mike Coble serves as vice-chairman and member





Summary and Lessons Learned

- Need a good team to become the experts in the field – having the right people is crucial
- Comprehensively gather information, create standardized information formats that are useful, share what is learned through multiple avenues
- Be plugged into the community and willingly help meet their needs (visits to labs to conduct training workshops benefits labs and our understanding of their needs and challenges)





Acknowledgments

Community feedback and contributors to the NIST STRBase website

Forensic DNA Team



Butler



Coble



Margaret

Kline



Dave Duewer DNA Biometrics Team



Pete Vallone



Kevin Kiesler

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

Hill

Funding from the **FBI S&T Branch** through NIST Information Access Division

Butts



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Points of view are those of the presenters and do not necessarily represent the official position of the National Institute of Standards and Technology or the U.S. Department of Justice.



Thank you for your attention!

