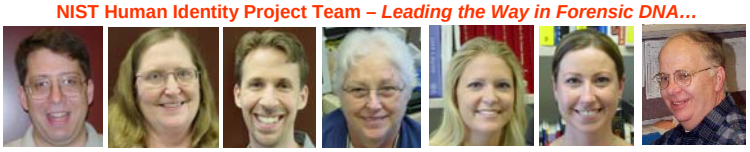
NIST Human Identity Project Team – *Leading the Way in Forensic DNA…*



John Butler | Margaret Kline | Pete Vallone | Jan Redman | Amy Decker | Becky Hill | Dave Duewer

NIST Human Identity Team Projects

John M. Butler, Ph.D.
National Institute of Standards and Technology

EDNAP and 26th ENFSI DNA Working Group Meeting

April 17-20, 2007
Krakow, Poland

National Institute of Justice
The Research, Development, and Evaluation Agency of the U.S. Department of Justice

Current Areas of NIST Effort with Forensic DNA

- **Standards** **http://www.cstl.nist.gov/biotech/strbase**
  - Standard Reference Materials
  - Standard Information Resources (STRBase website)
  - Interlaboratory Studies
- **Technology**
  - Research programs in SNPs, miniSTRs, Y-STRs, mtDNA, qPCR
  - Assay and software development, expert system review
- **Training Materials**
  - Review articles and workshops on STRs, CE, validation
  - PowerPoint and pdf files available for download

Team Impact

- **26 publications** from Jan-Dec 2006
- **45 presentations** and **10 workshops** to the community from Jan-Dec 2006

All NIST publications and presentations available on STRBase: http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm

Some Workshops Conducted This Past Year

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

- **qPCR workshop** **by Vallone and Orrego** (July 2006)
  slides available on STRBase
  - **http://www.cstl.nist.gov/biotech/strbase/qPCRworkshop.htm**
- **LCN workshop** **by Butler, Caragine, and Gill** (May 2006)
  Butler slides available on STRBase
  - **http://www.cstl.nist.gov/biotech/strbase/training.htm**

  Peter Gill's talk covered LoComatioN software (see *Forensic Sci. Int*. 2007, 116: 128-138)
- **Y-STR and mtDNA workshop** **by Butler and Coble** (Nov 2006)
  >600 slides available on STRBase
  - **http://www.cstl.nist.gov/biotech/strbase/YmtDNAworkshop.htm**

Training Workshops Planned

- ISFG Meeting (August 2007, Copenhagen, Denmark)
  - **CE Fundaments and Troubleshooting**
  - **Validation**

Technology: NIST Research Programs

- miniSTRs
- Y-chromosome STRs
- mtDNA
- SNPs
- qPCR for DNA quantitation
- DNA stability studies
- Variant allele characterization and sequencing
- Software tools
- Expert System review
- Assay development with collaborators

Apparent Null Alleles Observed During Concordance Studies

New Section of STRBase (launched to track MiniFiler discordance and allele dropout frequency):
http://www.cstl.nist.gov/biotech/strbase/NullAlleles.htm

Locus	STR Kits/Assays Compared	Results	Frequency of Primer Binding Site Mutation	Source
CSF1PO	MiniFiler vs ID vs PP16	MF: 11,11 and ID: 11,11.1 One base insertion in Identifiler amplicon outside of MiniFiler and PP16 primers	1/1308	Hill *et al.* (2007)
CSF1PO	PP16 vs COfiler	Loss of allele 14 with COfiler; fine with PP16	2/1537	Budowle *et al.* (2001)
FGA	SGM vs SGM Plus	Loss of allele 26 with SGM Plus; weak amp of same allele with SGM		Cotton *et al.* (2000)
FGA	PP16 vs ProPlus	Loss of allele 22 with ProPlus; fine with PP16		Budowle and Sprecher (2001)
TH01	PP16 vs COfiler	Loss of allele 9 with COfiler; fine with PP16	1/1537	Budowle *et al.* (2001)
TH01	SGM vs SGM Plus	Loss of allele 6 with SGM Plus; fine with SGM	1/4245	Clayton *et al.* (2004)
VWA	PP1.1 vs ProPlus	Loss of allele 19 with ProPlus; fine with PP1.1	2/1483	Kline *et al.* (1998) and Walsh (1998)
VWA	PP16 vs ProPlus	Loss of alleles 15 and 17 with ProPlus; fine with PP16	2/1537	Budowle *et al.* (2001)
VWA	ID vs miniplexes	Loss of alleles 12, 13, and 14 with miniplex assay; fine with ID	9/532	Drabek *et al.* (2004)

Variant Allele Sequencing Service (Free)

Send us any unusual variant or null alleles and we will sequence them…

Address http://www.cstl.nist.gov/biotech/strbase/STRseq.htm

Variant allele characterization

Send 10-20 ng of DNA (or 2-3 FTA bloodstain punches)
Contact margaret.kline@nist.gov or john.butler@nist.gov
Information will be posted on **STRBase …/STRseq.htm**
Sequence details provided back to sender

Locus	Variant Allele	Sample Source	Comments
			Deletion of a "G" that is 157 bp from the repeat region under
D18S51	40	Nebraska State Crime Lab	DNA sequence analysis showed 40 GAAA repeats
D18S51	"5.3"	DNA Solutions	DNA sequence analysis revealed a 9 bp deletion beyond the end of the 8th repeat unit to produce a "5.3" allele
DYS392	"10.2"	AFDIL	DNA sequence analysis revealed a C-to-G transversion 180 bp upstream of the STR repeat region; the mutation causes an apparent mobility shift of approximately 0.75 bp such that the allele falls outside of the +/-0.5 bp genotyping bin
DYS635	21.3	NIST U.S. population samples	DNA sequence analysis revealed a deletion of a "T" in the repeat region; full repeat was $[TCTA]_4(TGTA)_2[TCTA]_2(TGTA)_2[TCTA]_2(TGTA)_2[TCTA]_3$ TC-A $[TCTA]_2$
Penta D	18	DNA Solutions	DNA sequence analysis confirmed 18 repeats
Penta D	"8.2"	Peter de Knijff's lab at Leiden University	DNA sequence analysis revealed a 13 bp deletion prior to a $[AAAGA]_{11}$ repeat
Penta D	6	Peter de Knijff's lab at Leiden University	DNA sequence analysis confirmed 6 repeats


Pete Vallone


Dave Duewer

Software Tools from NIST

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

- AutoDimer – multiplex PCR primer screening tool

http://www.cstl.nist.gov/biotech/strbase/AutoDimerHomepage/AutoDimerProgramHomepage.htm

- mixSTR – mixture component resolution tool
- **Multiplex_QA** – quality assessment tool for monitoring instrument performance over time
- Tools to aid Expert System data review
  - **STR_ConvertFormats.xls** (converts data format)
  - **STR_MatchSamples.xls** (compares samples)

Mixture Interpretation Efforts

- NIST-led MIX05 interlaboratory study revealed that variation exists currently in the community in abilities and approaches for mixture interpretation
- SWGDAM started a mixture interpretation subcommittee in January 2007 with John Butler-NIST (chair) and Gary Sims-CA DOJ (co-chair)
- NIST entered into collaboration with US Army Crime Lab to evaluate and help make more user-friendly an in-house Excel-based program developed by Tom Overson
- Training workshops on mixture interpretation are planned…

Acknowledgments

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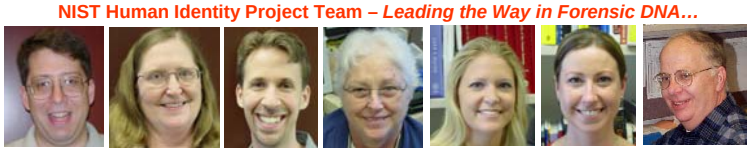

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Past and Present Collaborators (also funded by NIJ):

Mike Hammer and Alan Redd (U. AZ) for Y-chromosome studies
Tom Parsons, Rebecca Just, Jodi Irwin (AFDIL) for mtDNA coding SNP work
Sandy Calloway (Roche) for mtDNA LINEAR ARRAYs
Bruce McCord and students (FL Int. U.) for miniSTR work
Marilyn Raymond and Victor David (NCI-Frederick) for cat STR work
Artie Eisenberg and John Planz (U. North Texas) for miniSTR testing on bones
Murray Brilliant (U. AZ) for phenotype markers
Ken Kidd (Yale U.) for SNP typing population samples
Sree Kanthaswamy (UC Davis) for dog STR multiplex assay
Tom Reid (DNA Diagnostics Center) for father-son samples

Disclaimer: Points of view are those of the authors and do not necessarily represent the official position or policies of the US Department of Justice. 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.

Scientific Working Group on DNA Analysis Methods

Committee Updates

January 2007 SWGDAM Meeting

W. David Coffman, Chair

Presented to EDNAP and ENFSI DNA Working Group
by John M. Butler (NIST)
April 2007

Committees

CODIS
Expert Systems
Missing Person and Mass Disaster
Mitochondrial DNA
Mixture Interpretation
Quality Assurance
Serology

SWGDAM Contact

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