September 5, 2011 - Innsbruck, Austria

STRBase, Textbooks, and NIST Research Efforts:

Developing Tools to Aid the Forensic DNA Community

John M. Butler

NIST Human Identity Project Team

National Institute of Standards and Technology

Gaithersburg, Maryland USA

Presentation Topics

- Introduction to NIST and to Our Group
- STRBase website
- Textbooks
  - *Advanced Topics in Forensic DNA Typing* (3rd edition)
- Group Research Overview
  - Standard Reference Materials (SRMs)
  - STR loci characterization
  - STR kit concordance studies - BECKY
  - ABI 3500 validation work - ERICA
  - TrueAllele software examination – MIKE
  - Rapid PCR amplification - PETE
- ABI 3500 open letter status update

NIST Human Identity Project Teams
within the Applied Genetics Group

Forensic DNA Team

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

John Butler

Workshops & Textbooks

Mike Coble

Mixtures, mtDNA & Y

Becky Hill

Concordance & LT-DNA

Margaret Kline

SRM work, variant alleles & Cell Line ID

Guest Researcher

Manuel **Fonde**vila Alvarez

Data Analysis Support

Dave Duewer

DNA Biometrics Team

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

Pete Vallone

Rapid PCR, Direct PCR & Biometrics

Erica Butts

ABI 3500 & DNA Extraction

Kristen Lewis O'Connor

D12/vWA & Kinship Analysis

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

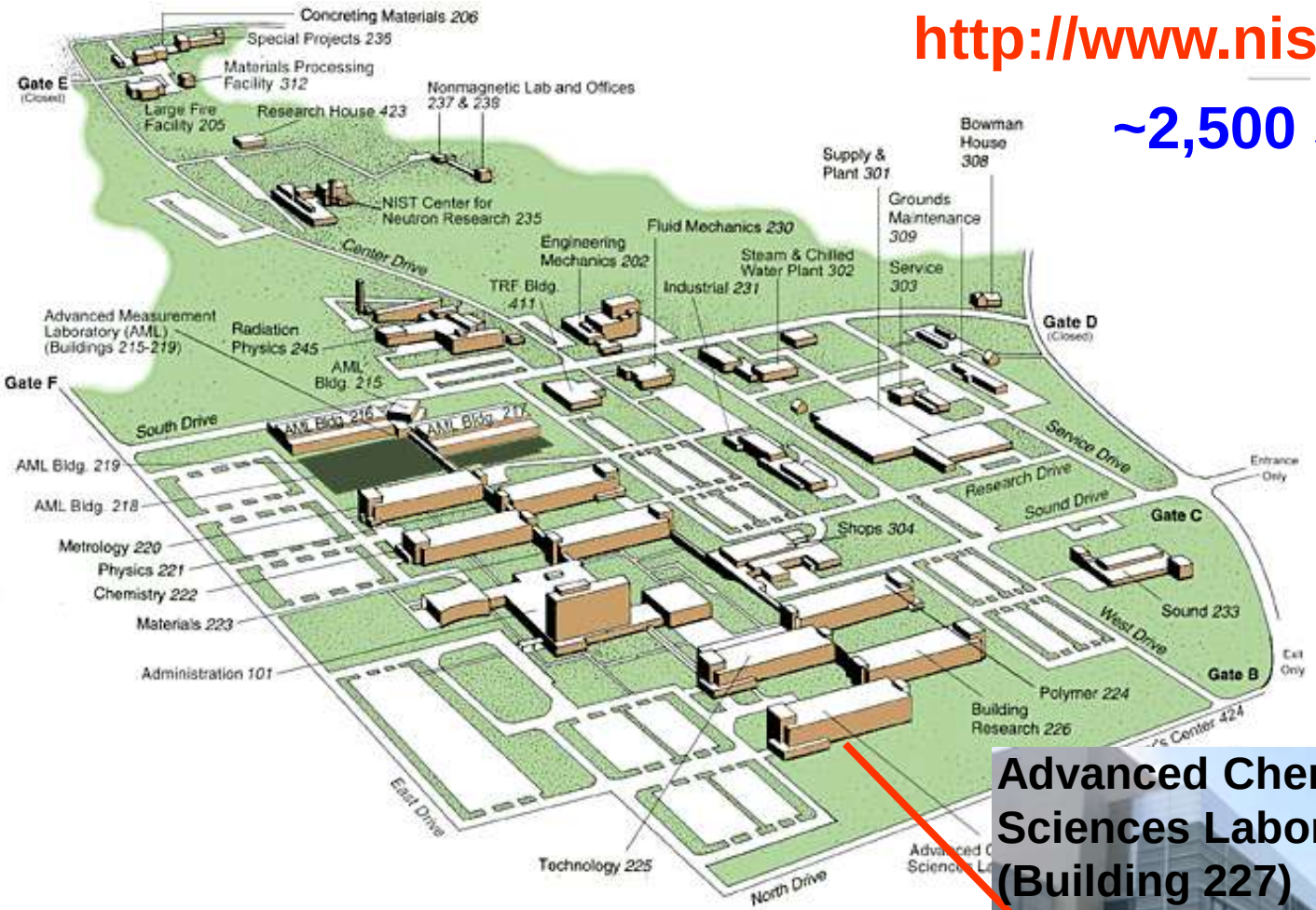
National Institute of Standards & Technology (NIST)

- **Non-regulatory agency** established in 1901 in the US Department of Commerce.
- Mission to promote US innovation and industrial competitiveness by advancing measurement science, standards & technology.
- NIST develops a wide variety of physical standards, test methods, and standard reference data.

NIST Gaithersburg Campus

http://www.nist.gov

~2,500 staff

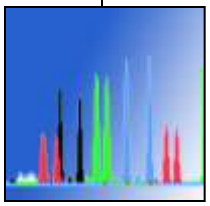


Located about 40 km Northwest of Washington, DC (8 km from AFDIL)

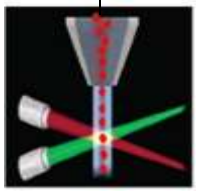
NIST Biochemical Science Division

Laurie Locascio
Division Chief

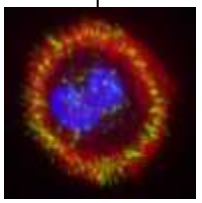
Process Sensing
Dean Ripple
Group Leader



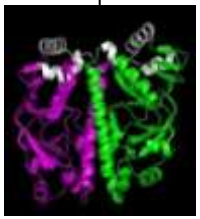
Applied Genetics
John Butler
Group Leader



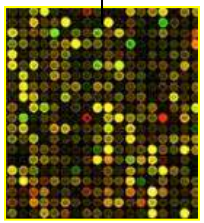
Bioassay Methods
Steve Choquette
Group Leader



Cell Systems Science
Anne Plant
Group Leader



Macromolecular Structure & Function
John Marino
Group Leader



Multiplexed Biomolecular Science
Marc Salit
Group Leader

Doing some Next Generation Sequencing using ABI SOLID

NIST Applied Genetics Group

Group Leader

John Butler

Marcia Holden

Margaret Kline

Pete Vallone

Mike Coble

Ross Haynes

Becky Hill

Erica Butts

Kristen O'Connor

Starts Sept 12

Kevin Kiesler

NIST
Applied Genetics

APPLIED GENETICS Group

Major Programs Currently Underway

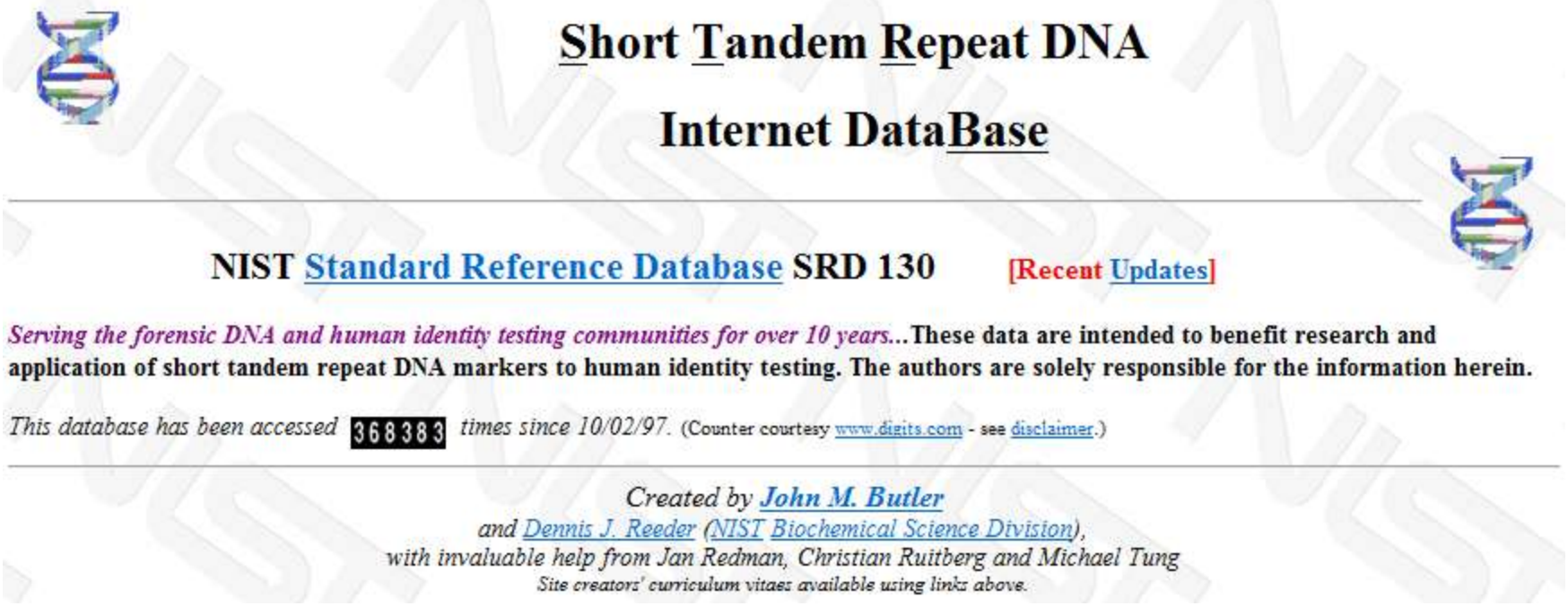
- **Forensic DNA**
  - **STRBase website**
  - New loci and assays (26plex)
  - **STR kit concordance**
  - Ancestry SNP assays
  - Low-template DNA studies
  - **Mixture interpretation research** and training
  - STR nomenclature
  - Variant allele cataloging and sequencing
  - Expert systems review
  - Training workshops to forensic DNA laboratories
  - **Validation experiments**, information and software tools
  - **Textbooks** – 3rd 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**

STRBase

Not str-base! ☺

NIST STRBase Website:
Forensic DNA Information Resources

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



I invite your lab to supply information on variant and tri-alleles observed

History of STRBase Website

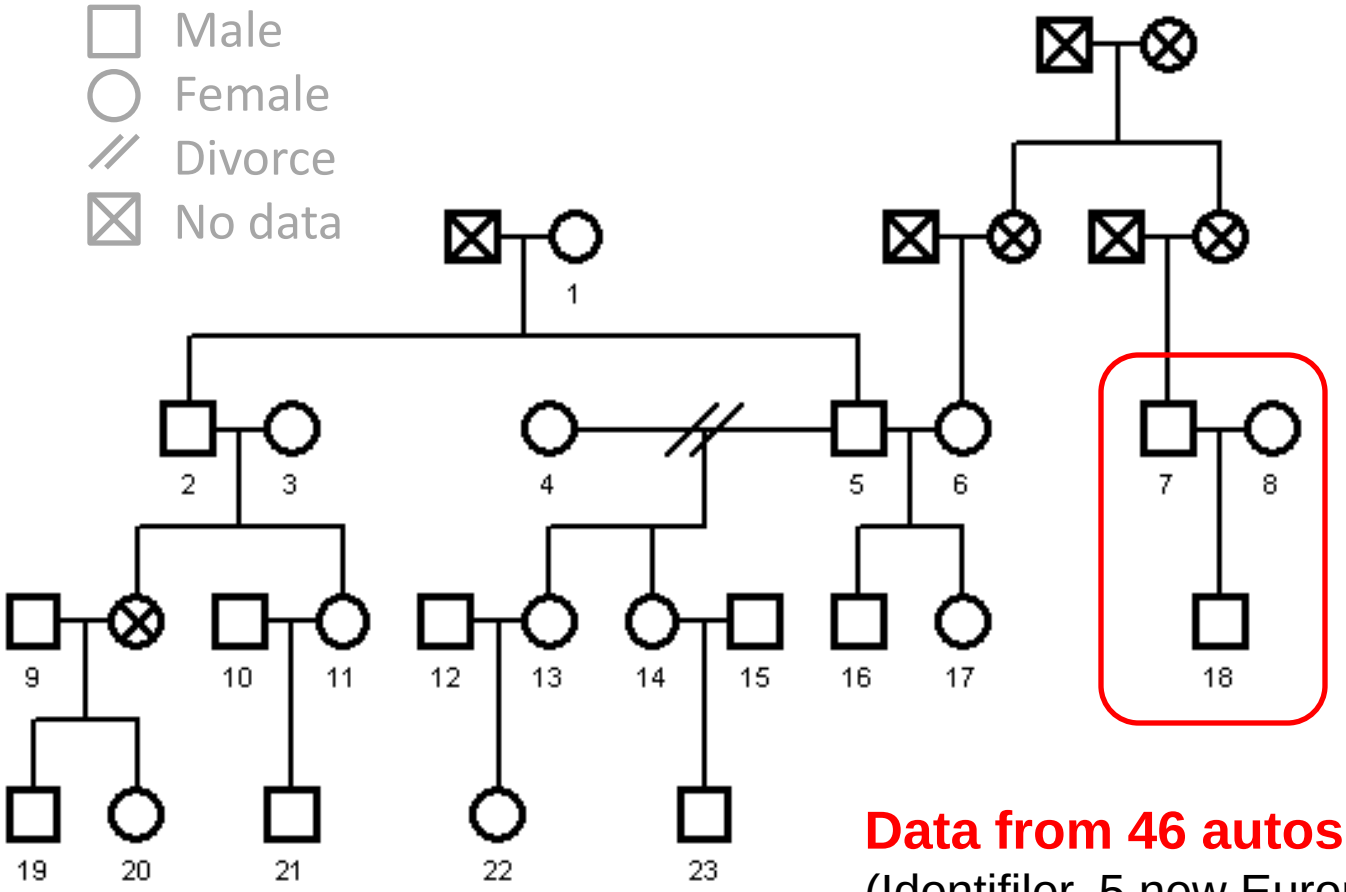
- Information was collected on STR markers while working on my PhD dissertation in 1993-1995
- Started a review article in 1996 while a NIST postdoc but wanted to create a dynamic rather than an out-of-date resource
- Created hundreds of individual web pages that were hyperlinked together
- Website launched in July 1997
- **I continue to update the website (via an HTML editor)…**

New STRBase Sections

Forensic STR Information

- STRs101: Brief Introduction to STRs
- ***Core Loci***: FBI CODIS Core STR Loci and European Core Loci
- STR Fact Sheets (observed alleles and PCR product sizes)
- **Multiplex STR kits**
- Sequence Information (annotated)
- Variant Allele Reports ◆
- Tri-Allelic Patterns ◆
- Mutation Rates for Common Loci
- Published PCR primers
- Y-chromosome STRs ◆
- Low-template DNA Information *Updated*
- Mixture Interpretation *NEW*
- Kinship Analysis *NEW*
- miniSTRs (short amplicons) ◆
- Null Alleles - discordance observed between STR kits ◆
- STR Reference List - *now 3400 references* ◆

NIST Standard Reference Family Pedigree



Paternity Trio Individuals 7,8,18	
Locus	**PI Formula (AABB Appendix 8)**
D8S1179	1
D21S11	9
D7S820	8
CSF1PO	13
D3S1358	14
TH01	8
D13S317	2
D16S539	3
vWA	7
TPOX	15
D18S51	4
D5S818	4
FGA	mutation

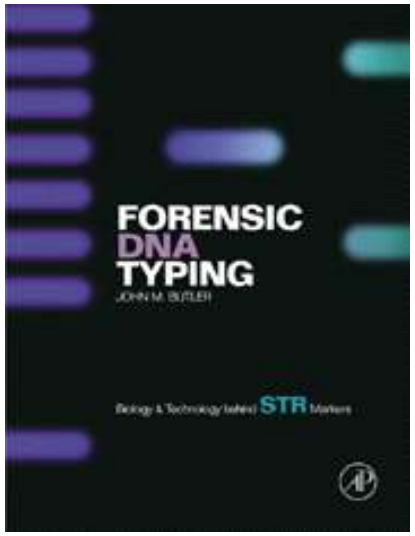
Data from 46 autosomal STRs
(Identifiler, 5 new European loci, SE33, NIST 26plex) **and 17 Y-STRs** (Yfiler)

Data available for testing software programs:
http://www.cstl.nist.gov/biotech/strbase/kinship.htm

Textbooks

Forensic DNA Typing Textbook

1st Edition



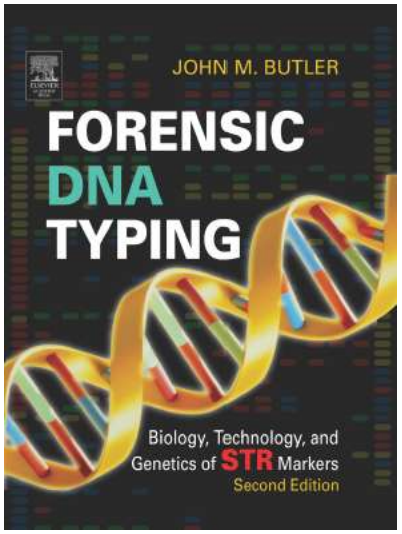
Jan 2001

335 pp.

17 chapters

With Y. Fukuma
(Japanese translator)

2nd Edition



Feb 2005

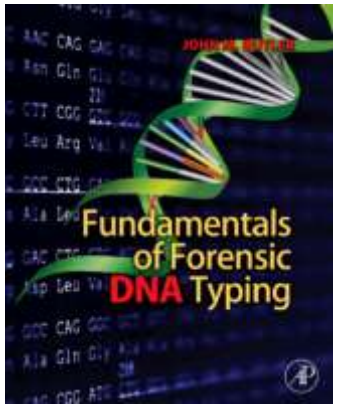
688 pp.

24 chapters

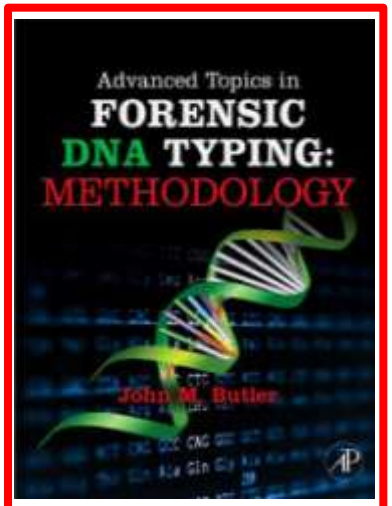
Now available in **Chinese**
(Yiping Hou)

Japanese in preparation
(Yoshiya Fukuma)

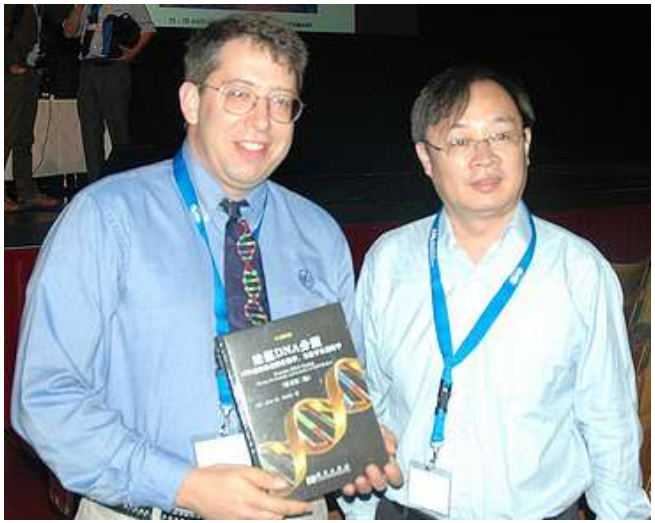
3rd Edition



Sept 2009



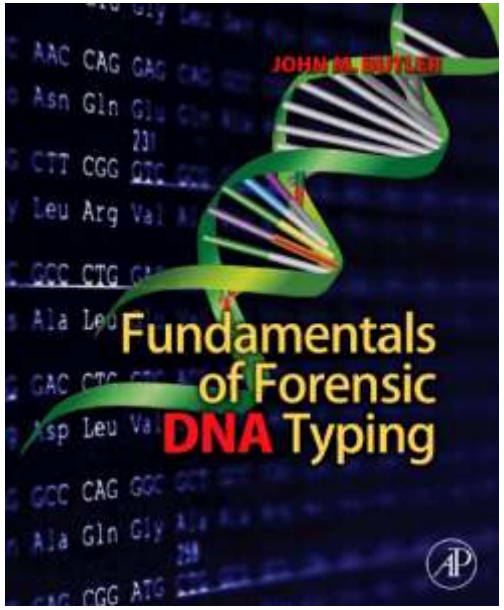
Sept 2011



With Y. Hou (Chinese translator)

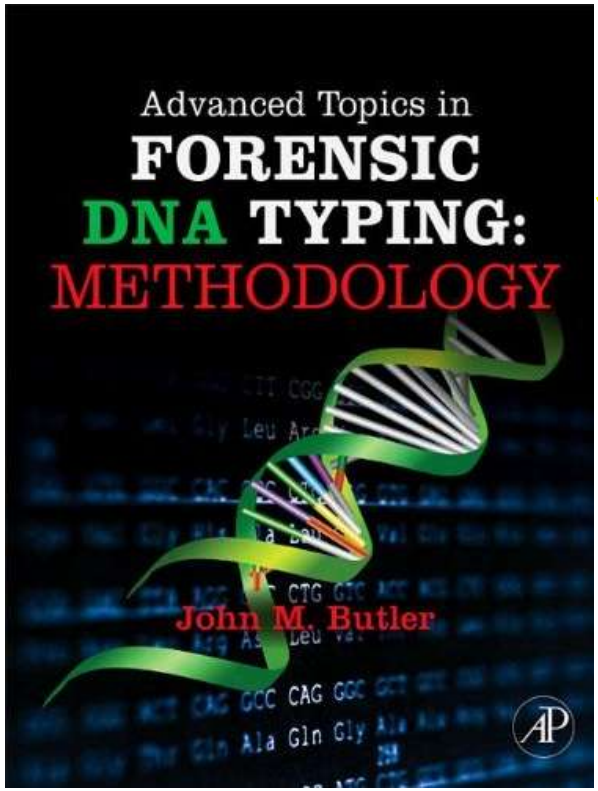
Forensic DNA Typing Textbook

3rd Edition is Three Volumes



Sept 2009

~500 pages



Sept 2011

~700 pages

Currently being written

Advanced Topics in
Forensic
DNA Typing:
INTERPRETATION

Fall 2012

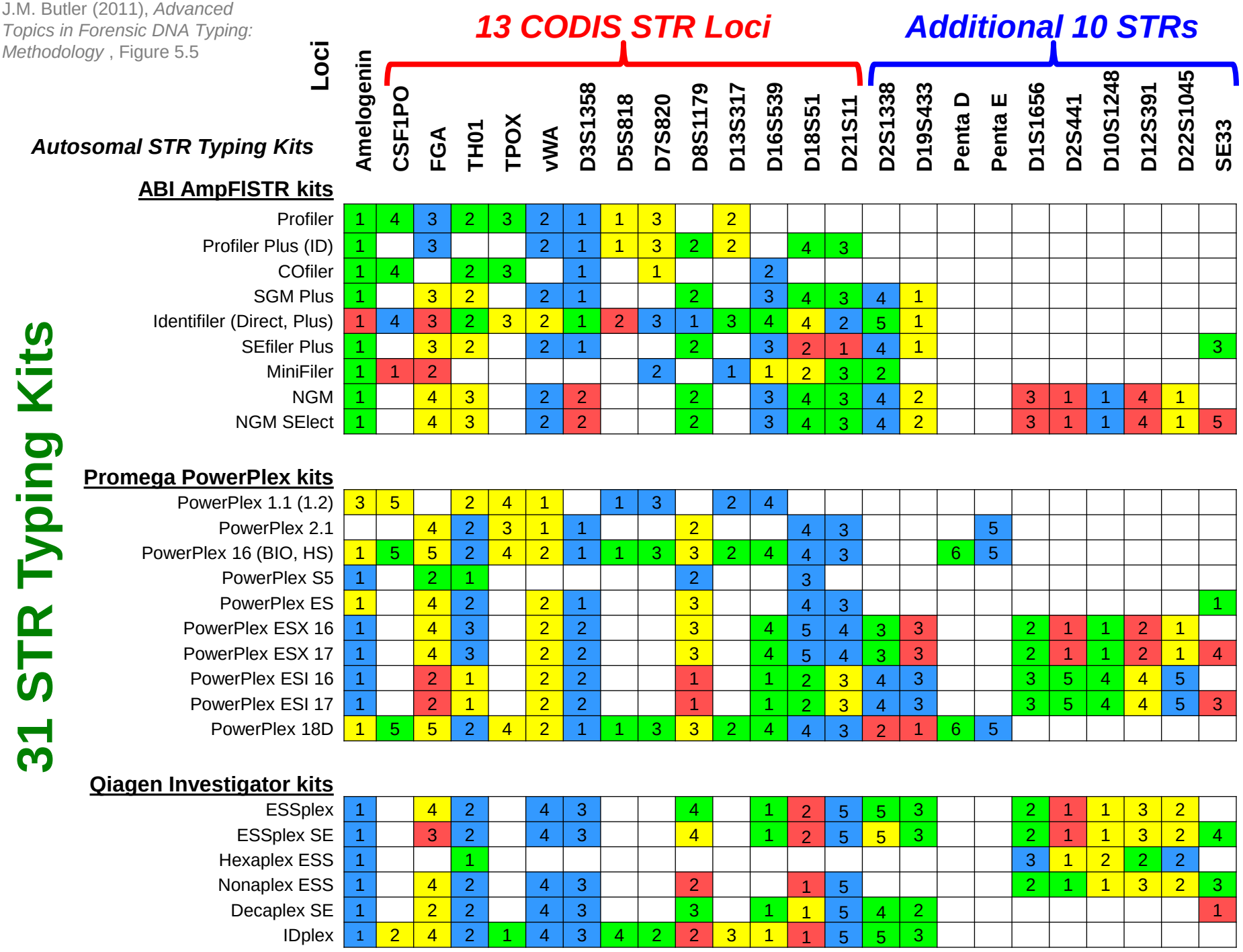
~500 pages

New Materials in *Advanced Topics* book

Released August 2011

- Cites >1500 new references (>2800 ref. total)
- New chapter on legal aspects
  - expert witness prep, perspectives from lawyers
- New chapter on X-chromosome markers
- Extensive updates on LCN, Y-STRs, miniSTRs, mtDNA, SNPs, non-human DNA, and database issues
- Coverage of all the new STR kits
- Listing of all known STR alleles for all 23 kit loci

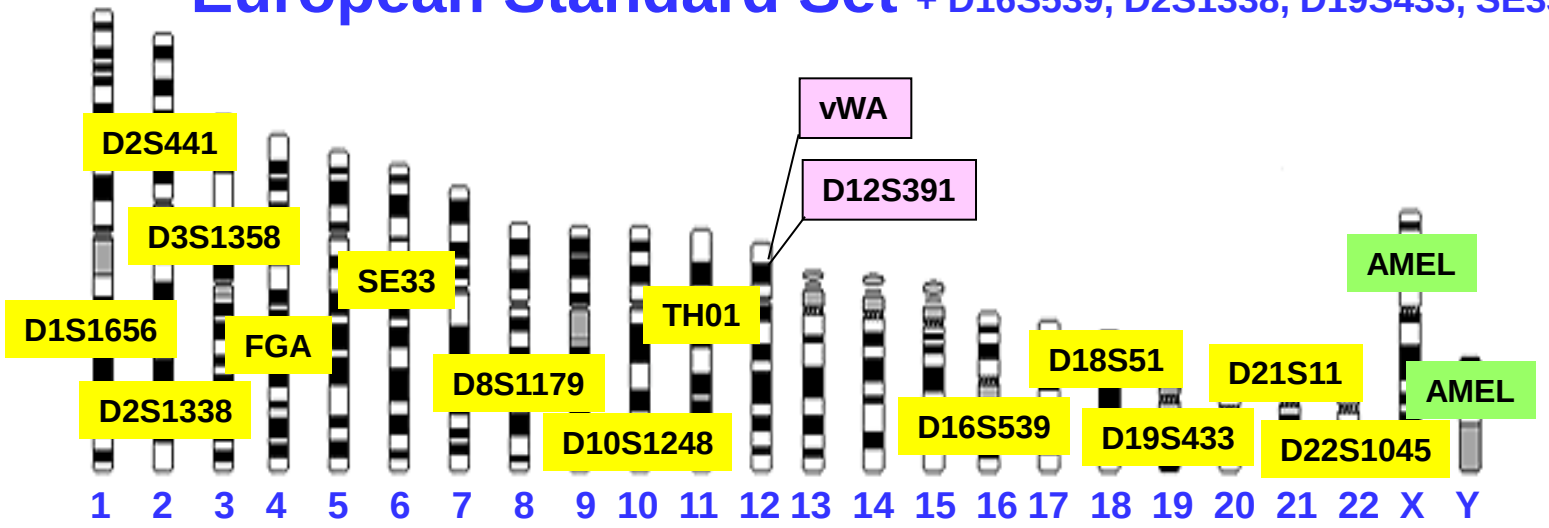
J.M. Butler (2011), *Advanced Topics in Forensic DNA Typing: Methodology* , Figure 5.5



Common Forensic STR Loci

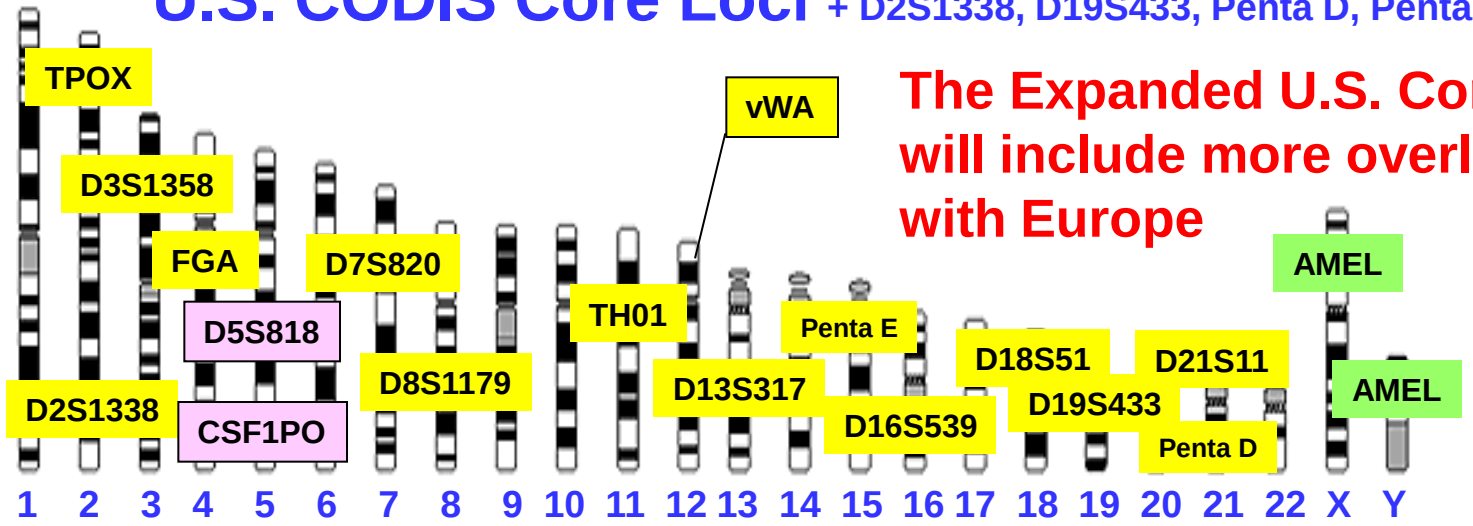
Europe

European Standard Set + D16S539, D2S1338, D19S433, SE33



United States

U.S. CODIS Core Loci + D2S1338, D19S433, Penta D, Penta E



The Expanded U.S. Core Set will include more overlap with Europe

The 10 STR Loci Beyond the CODIS 13

STR Locus	Location	Repeat Motif	Allele Range*	# Alleles*
D2S1338	2q35	TGCC/TTCC	10 to 31	40
D19S433	19q12	AAGG/TAGG	5.2 to 20	36
Penta D	21q22.3	AAAGA	1.1 to 19	50
Penta E	15q26.2	AAAGA	5 to 32	53
D1S1656	1q42	TAGA	8 to 20.3	25
D12S391	12p13.2	AGAT/AGAC	13 to 27.2	52
D2S441	2p14	TCTA/TCAA	8 to 17	22
D10S1248	10q26.3	GGAA	7 to 19	13
D22S1045	22q12.3	ATT	7 to 20	14
SE33	6q14	AAAG‡	3 to 49	**178**

5 new European loci

**Allele range and number of observed alleles from Appendix 1, J.M. Butler (2011) Advanced Topics in Forensic DNA Typing: Methodology; ‡SE33 alleles have complex repeat structure*

SE33 (58 alleles observed)

343 genotypes observed
Heterozygosity = 0.9377

Allele	Total #	Total %	Populations, % Af Am	Asian	Cauc	Hisp
6.3						
7						
8						
10.2						
11	1	0.0			0.1	
11.2	2	0.1	0.2			
12	11	0.4	0.3		0.5	0.4
12.2	4	0.1	0.2			0.3
13	31	1.1	1.1		1.5	1.0
13.2	9	0.3	1.0			
14	85	2.9	5.1	0.2	2.5	2.4
14.2	10	0.3	0.4		0.4	0.3
15	102	3.5	3.9	1.2	3.9	3.9
15.2	8	0.3	0.3			0.7
16	144	5.0	4.8	4.7	4.0	6.7
16.2	5	0.2	0.3		0.1	0.1
16.3	2	0.1				0.3
17	205	7.1	9.3	4.0	6.2	7.3
17.2	1	0.0	0.1			
17.3	5	0.2	0.1		0.2	0.3
18	268	9.3	12.1	5.0	7.2	**11.0**
18.3	1	0.0			0.1	
19	250	8.7	**12.4**	6.2	6.6	8.0
19.2	8	0.3		0.2	0.4	0.4
20	216	7.5	10.9	9.2	5.4	4.8
20.2	20	0.7	0.3	1.2	1.1	0.3
21	108	3.7	4.6	6.7	2.4	2.7
21.2	48	1.7	1.1	1.7	2.4	1.3
22	42	1.5	1.3	1.7	1.5	1.3
22.2	65	2.3	0.4	3.2	3.8	1.9
23	12	0.4	0.6	1.0	0.2	0.1
23.2	91	3.2	2.2	4.2	4.3	2.1
24	1	0.0			0.1	
24.2	74	2.6	1.3	6.2	2.2	2.5
25.2	109	3.8	2.6	6.9	4.0	3.1
26	1	0.0	0.1			
26.2	163	5.6	6.1	5.2	4.3	7.1
27	1	0.0				0.1
27.2	225	7.8	4.3	**10.4**	**9.5**	8.6
27.3	2	0.1				0.3
28	2	0.1	0.1	0.2		
28.2	180	6.2	4.4	7.9	7.4	6.1
28.3	2	0.1	0.1		0.1	
29	1	0.0		0.2		
29.2	147	5.1	2.7	5.7	6.3	6.3
29.3	1	0.0		0.2		
30	1	0.0				0.1
30.2	111	3.8	1.6	3.2	5.8	4.6
31	3	0.1	0.1		0.2	
31.2	52	1.8	1.5	2.5	2.2	1.3
32	1	0.0			0.1	
32.2	25	0.9	0.4	0.7	1.3	0.9
33	2	0.1			0.1	0.1
33.2	11	0.4	0.3		0.5	0.4
34	9	0.3	0.3		0.7	
34.2	1	0.0			0.1	
35	1	0.0	0.1			
36	2	0.1	0.2			

Loci sorted on Probability of Identity (P_I) values

STR Locus	Alleles Observed	Genotypes Observed	Het. (obs)	P_I value N = 938
SE33	53	292	0.9360	0.0069
Penta E*	20	114	0.8799	0.0177
D2S1338	13	68	0.8785	0.0219
D1S1656	15	92	0.8934	0.0220
D18S51	21	91	0.8689	0.0256
D12S391	23	110	0.8795	0.0257
FGA	26	93	0.8742	0.0299
Penta D*	16	71	0.8754	0.0356
D21S11	25	81	0.8358	0.0410
D19S433	16	76	0.8124	0.0561
D8S1179	11	45	0.7878	0.0582
vWA	11	38	0.8060	0.0622
D7S820	11	32	0.8070	0.0734
TH01	8	24	0.7580	0.0784
D16S539	9	28	0.7825	0.0784
D13S317	8	29	0.7655	0.0812
D10S1248	12	39	0.7825	0.0837
D2S441	14	41	0.7772	0.0855
D3S1358	11	30	0.7569	0.0873
D22S1045	11	42	0.7697	0.0933
CSF1PO	9	30	0.7537	0.1071
D5S818	9	34	0.7164	0.1192
TPOX	9	28	0.6983	0.1283

23 STR Loci present in STR kits

rank ordered by their variability

Better for mixtures (more alleles seen)

There are several loci more polymorphic than the **current CODIS 13 STRs**

Better for kinship (low mutation rate)

NIST Projects

A Short Overview…

Congress Passed **the DNA Identification Act of 1994** (Public Law 103 322)

Formalized the FBI's authority to establish a national DNA index for law enforcement purposes.

FBI's DNA Advisory Board

Quality Assurance Standards for Forensic DNA Testing Laboratories

(October 1, 1998)

STANDARD 9.5

The laboratory shall check its DNA procedures annually or whenever substantial changes are made to the protocol(s) against an appropriate and available NIST standard reference material or standard traceable to a NIST standard.

NIST DNA Reference Materials

Date of release or certificate revision (r)

Forensic Applications

- **STR PCR DNA Profiling** (**SRM 2391c**) – 1995, r2008, **2011**
- Mitochondrial DNA Sequencing (SRM 2392-I, 2392) – 1999, 2003, r2009
- **Human Y-Chromosome DNA Profiling** (SRM 2395) – 2003, r2008
- RFLP DNA Profiling (SRM 2390) – 1992, r2001, *now obsolete*

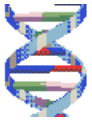
Clinical Applications

- Fragile X Human DNA Triplet Repeat (SRM 2399) – 2004, r 2007
- Huntington's Disease CAG Repeats (**SRM 2393**) – **2011**
- Cytomegalovirus (**SRM 2366**) – **2011**

Platform Testing

- **Human DNA Quantitation** (SRM 2372) - 2007
- Heteroplasmic mtDNA Mutation Detection (SRM 2394) - 2004
- DNA Sequence Library for External RNA Controls (SRM 2374) – in process

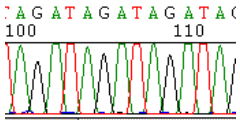
A few others are in early stages of development

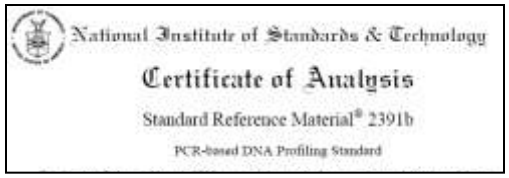


Steps Involved in SRM Production

Attend conferences, read the literature, talk to potential customers

Sequence & Copy Number





Receive input on priorities for projects and potential SRMs

Research potential properties and samples to be characterized and measurement method to be used

Obtain candidate components/make measurements

Decide on number of SRM units to produce (impacts price/unit), sample packaging, concentration, etc.

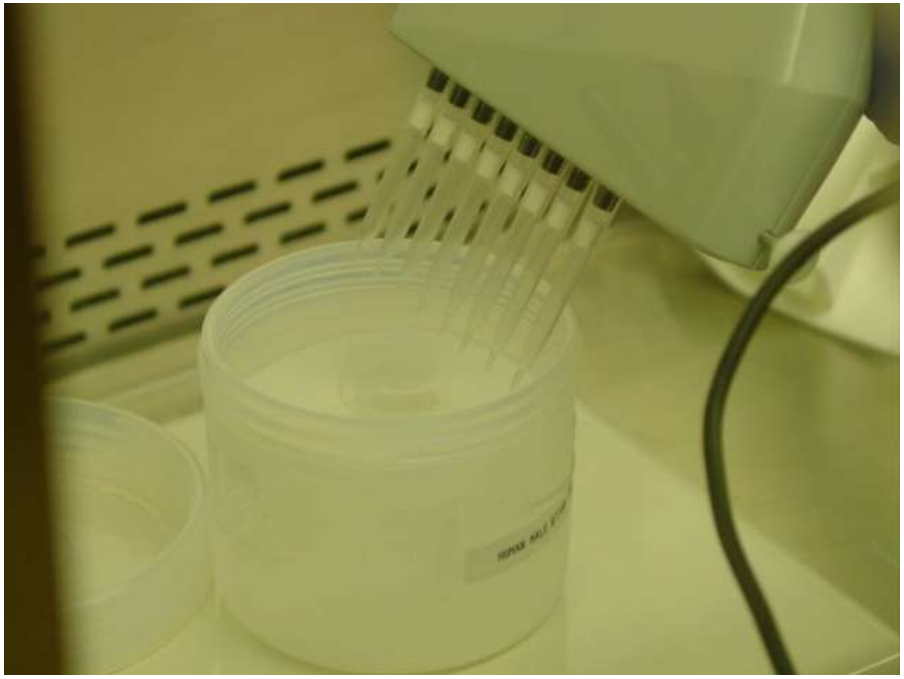
Bottle components and conduct homogeneity and stability studies; finalize uncertainty

Write Report of Analysis and Certificate of Analysis

Certificate Reviewed and Approved by NIST Measurement Services Division

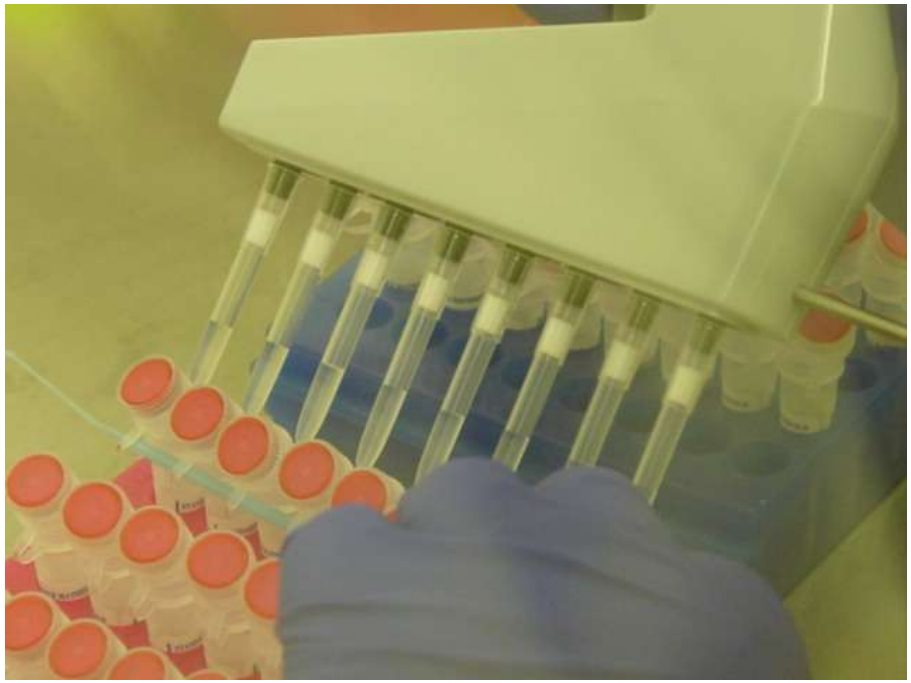
SRM Made Available for Purchase http://www.nist.gov/srm

Bottling SRM 2372 Materials



Teflon container holding ≈ 250 mL of ***Candidate*** SRM 2372.

It's not an SRM until it passes all testing.



With a multi-channel pipettor 8 tubes can be filled at a time. That's ≈ 214 reps to fill 1700 tubes per component.

The assembly line closing the recently filled tubes

Protecting the SRM Product from the Staff : Lab Coats, Masks and Hair nets or full face shields **P**ersonal **P**rotective **E**quipment (PPE) or **P**roduct **P**rotective **E**quipment.

Safety Considerations:
The Blister Brigade

Closing the 1,700 component A tubes (SRM 2372) caused some blisters even while wearing gloves.

Safety resolution: Band-aids applied prior to closing SRM component tubes the next session helped reduce the number of blisters formed!

NIST Standard Reference Material (SRM) for Forensic DNA Testing

SRM 2391b (2003-2011)

- **48 autosomal STR loci** with certified values
- **10 liquid genomic DNA** components + **2 punches** (cells on 903 paper)
- All single source samples
- 4 males + 6 females
- 9947A & 9948 included

SRM 2391c (2011-future)

- **23 autosomal STR loci and 17 Y-STRs** certified
- **4 liquid genomic DNA** components + **2 punches** (cells on **FTA** & 903 paper)
- 5 single source + 1 mixture
- 3 males + 2 females (unique)
- All new samples
  - no 9947A or 9948

SRM 2391c to replace SRM 2391b and SRM 2395 (for Y-STRs)

Commercially Available STR Kits

Applied Biosystems (17)

- ~~AmpFlSTR Blue (1996)~~
- ~~AmpFlSTR Green I (1997)~~
- Profiler (1997)
- Profiler Plus (1997)
- COfiler (1998)
- SGM Plus (1999)
- **Identifiler** (2001)
- Profiler Plus ID (2001)
- ~~SEfiler (2002)~~
- Yfiler (2004)
- MiniFiler (2007)
- SEfiler Plus (2007)
- Sinofiler (2008) – China only
- **Identifiler Direct** (2009)
- NGM (2009)
- **Identifiler Plus** (2010)
- NGM SElect (2010)

Promega Corporation (13)

- PowerPlex 1.1 (1997)
- PowerPlex 1.2 (1998)
- PowerPlex 2.1 (1999)
- **PowerPlex 16** (2000)
- PowerPlex ES (2002)
- PowerPlex Y (2003)
- PowerPlex S5 (2007)
- **PowerPlex 16 HS** (2009)
- PowerPlex ESX 16 (2009)
- PowerPlex ESX 17 (2009)
- PowerPlex ESI 16 (2009)
- PowerPlex ESI 17 (2009)
- PowerPlex 18D (2011)

Qiagen (2010)

Primarily selling kits in Europe

Due to patent restrictions cannot sell in U.S.

- ESSplex
- ESSplex SE
- Decaplex SE
- IDplex
- Nonaplex ESS
- Hexaplex ESS
- HD (Chimera)
- Argus X-12
- Argus Y-12
- DIPlex (30 indels)

~1/3 of all STR kits were released in the last two years

STR Kits Tested with SRM 2391c

Applied Biosystems (12)
Identifiler
Identifiler Plus
NGM
NGM SElect
COfiler
Profiler
Profiler Plus
Profiler Plus ID
SGM Plus
SEfiler Plus
MiniFiler
Yfiler

Promega (9)
PowerPlex 16
PowerPlex 16 HS
PowerPlex Y
PowerPlex ESX 17
PowerPlex ESI 17
PowerPlex ES
PowerPlex S5
PowerPlex Y
FFFL

Qiagen (2)
ESSplex
IDplex

NIST assays
26plex
miniSTRs

Alleles sequenced:
SE33
D12S391
D1S1656
Penta D
Penta E
D8S1115

23 commercial STR kits examined

NIST developed 26plex and miniplexes

No discordant results observed on SRM 2391c samples

Kit Concordance Comparisons

Kits compared	Samples	Loci compared	Comparisons	# Differences	Concordance (%)
SGM-ID	1436	11	15,796	1	99.994
ID-ProPlus	1427	10	14,270	1	99.993
ID-IDplex	669	16	10,704	19	99.822
ID-PP16	662	14	9,268	4	99.957
ID-MiniFiler	1308	9	11,772	27	99.771
SGM-NGM	1436	11	15,796	4	99.975
ID-NGM	1449	11	15,939	3	99.981
ProPlus-NGM	1427	10	14,270	4	99.972
SGM-ESI	1436				99.968
ProPlus-ESX	1427				99.970
ESI-ESX	1455				99.939
ESI-ESSplex	1445				99.875
ESX-ESSplex	1445				99.870
ESI-NGMSElect	715				99.860
ESX-NGMSElect	715				99.942
ESS-NGMSElect	663	17	11,271	17	99.849
		TOTAL	240,156	186	99.923

> 1 million comparisons

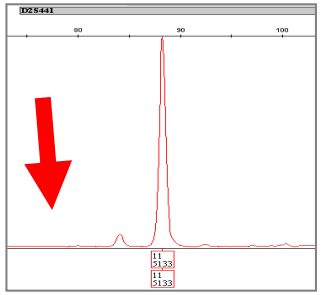
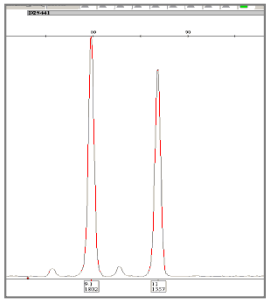
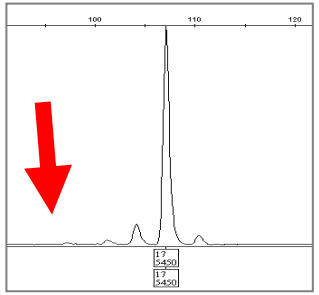
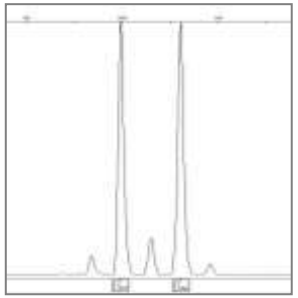
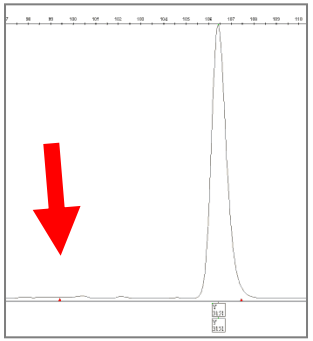
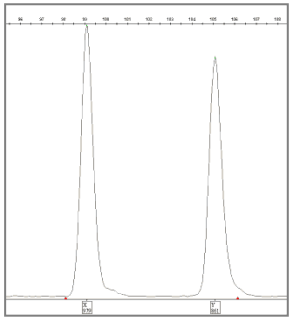
>1100 differences observed

~99.9% concordance

(many corrected now)

*Kits (except Identifiler) were kindly provided by **Applied Biosystems, Promega, and Qiagen** for concordance testing performed at NIST*

Extra (Degenerate) Primers Added with NGM SElect

	NGM (original)		NGM SElect and NGM’	
D2S441 9.1 allele missing in 7 Asians		**11,11**		**9.1,11**
D22S1045 15 allele missing in 4 samples		**17,17**		**15,17**
Amelogenin X allele missing in 3 samples		**Y,Y**		**X,Y**

NIST Standard Sample Sets

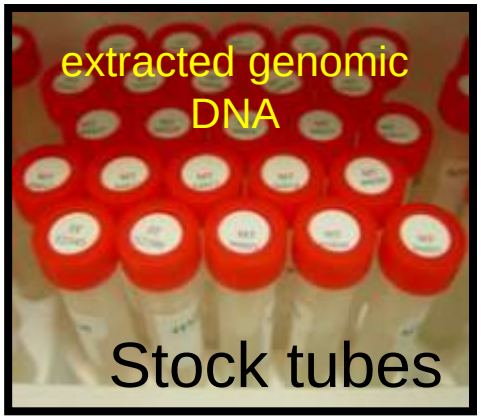
- **U.S. Population Samples** (**663 samples**)
  - Previously studied with Identifiler, MiniFiler, Yfiler, PP16, PP ESX/ESI 17, NGM, miniSTRs, and 23plex *(>200,000 allele calls)*
  - 260 African Americans, 260 Caucasians, 140 Hispanics, and 3 Asians

- **U.S. Father/Son pairs** (**800 samples**)
  - Previously studied with Identifiler, MiniFiler, Yfiler, PP ESX/ESI 17, NGM, 23plex
  - ~**100 fathers/100 sons for each group**: African Americans, Caucasians, Hispanics, and Asians

- **NIST SRM 2391b** PCR DNA Profiling Standard (**12 samples**)
  - Components 1-10 (includes 9947A and 9948): *well characterized*
  - ABI 007 and K562

>1450 total samples

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



D8S1179

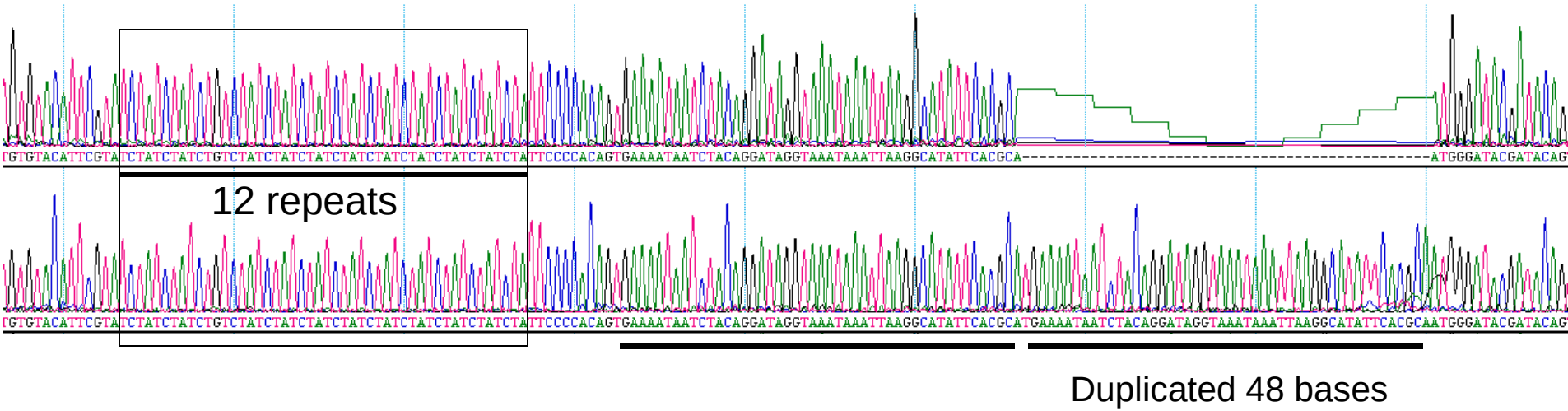
All Previously Known Alleles

Many alleles sequences are not known

We just set the new world record for the largest D8 allele ("24")

Allele (Repeat #)	Promega PowerPlex 16	ABI Identifiler	Repeat Structure $[TCTR]_n$	Reference
6	199 bp	119 bp	Not published	STRBase
7	203 bp	123 bp	$[TCTA]_7$	Griffiths *et al.* (1998)
8	207 bp	127 bp	$[TCTA]_8$	Barber and Parkin (1996)
9	211 bp	131 bp	$[TCTA]_9$	Barber and Parkin (1996)
10	215 bp	135 bp	$[TCTA]_{10}$	Barber and Parkin (1996)
10.1	216 bp	136 bp	Not published	STRBase
10.2	217 bp	137 bp	Not published	STRBase
11	219 bp	139 bp	$[TCTA]_{11}$	Barber and Parkin (1996)
12	223 bp	143 bp	$[TCTA]_{12}$	Barber and Parkin (1996)
12.1	224 bp	144 bp	Not published	STRBase
12.2	225 bp	145 bp	Not published	STRBase
12.3	226 bp	146 bp	Not published	STRBase
13 (a)	227 bp	147 bp	$[TCTA]_1[TCTG]_1[TCTA]_{11}$	Barber and Parkin (1996)
13 (b)	227 bp	147 bp	$[TCTA]_2[TCTG]_1[TCTA]_{10}$	**Kline *et al.* (2010)**
13 (c)	227 bp	147 bp	$[TCTA]_1[TCTG]_1TGTA[TCTA]_{10}$	**Kline *et al.* (2010)**
13 (d)	227 bp	147 bp	$[TCTA]_{13}$	**Kline *et al.* (2010)**
13.1	228 bp	148 bp	Not published	STRBase
13.2	229 bp	149 bp	Not published	STRBase
13.3	230 bp	150 bp	Not published	STRBase
14	231 bp	151 bp	$[TCTA]_2[TCTG]_1[TCTA]_{11}$	Barber and Parkin (1996)
14.1	232 bp	152 bp	Not published	STRBase
14.2	233 bp	153 bp	Not published	STRBase
15	235 bp	155 bp	$[TCTA]_2[TCTG]_1[TCTA]_{12}$	Barber and Parkin (1996)
15.1	236 bp	156 bp	Not published	STRBase
15.2	237 bp	157 bp	Not published	STRBase
15.3	238 bp	158 bp	Not published	STRBase
16	239 bp	159 bp	$[TCTA]_2[TCTG]_1[TCTA]_{13}$	Barber and Parkin (1996)
16.1	240 bp	160 bp	Not published	STRBase
17	243 bp	163 bp	$[TCTA]_2[TCTG]_2[TCTA]_{13}$	Barber and Parkin (1996)
17.1	244 bp	164 bp	Not published	STRBase
17.2	245 bp	165 bp	Not published	STRBase
18	247 bp	167 bp	$[TCTA]_2[TCTG]_1[TCTA]_{15}$	Barber and Parkin (1996)
19	251 bp	171 bp	$[TCTA]_2[TCTG]_2[TCTA]_{15}$	Griffiths *et al.* (1998)
20	255 bp	175 bp	Not published	**STRBase**

D8S1179 12,“24”



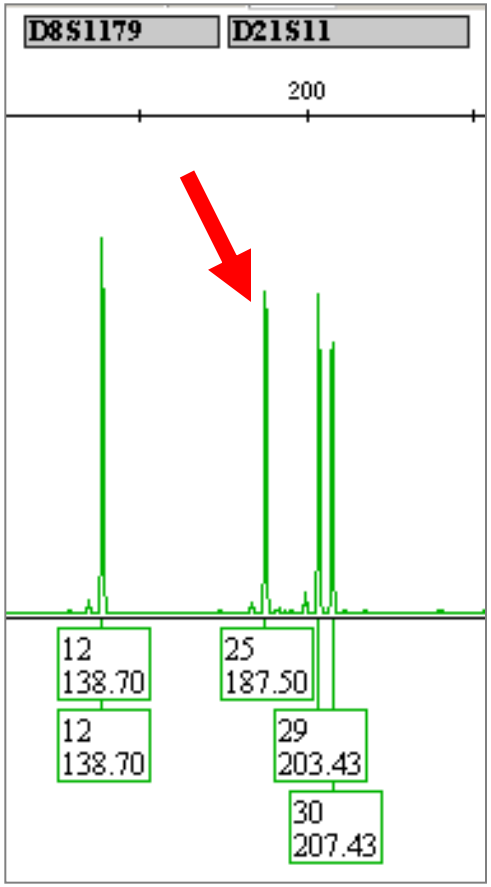
Allele 12 : $[TCTA]_2$ TCTG $[TCTA]_9$

Allele “24” : $[TCTA]_2$ TCTG $[TCTA]_9$ **duplication of the 48 bases 10 bases downstream of the repeat**

Result with This Large D8S1179 Allele

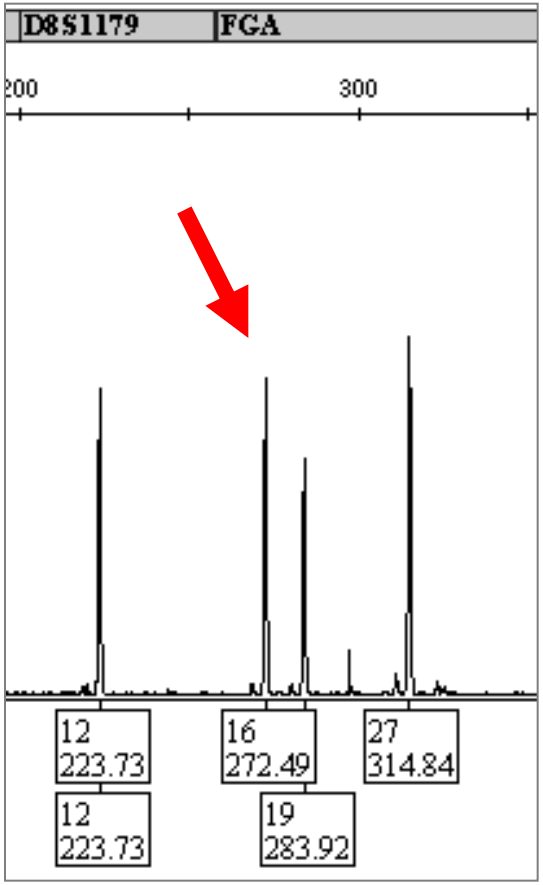
Using European STR Kits

NGM SElect



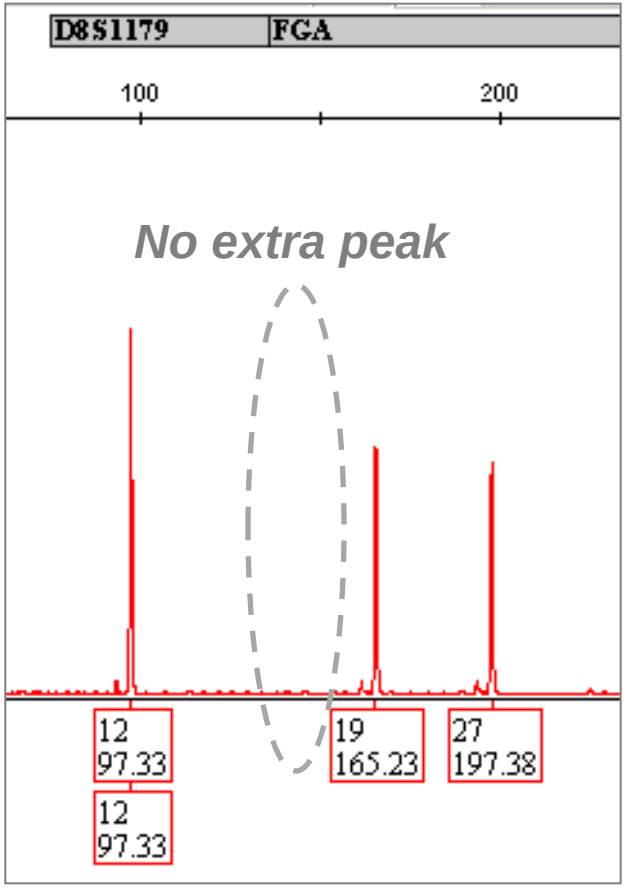
False D21S11 tri-allele

PP ESX 17



False FGA tri-allele

PP ESI 17



Reverse primer internal to duplicated flanking region

ABI 3500 Genetic Analyzer

Status Update on Open Letter to Applied Biosystems

Open Letter to Applied Biosystems on Concerns with ABI 3500

- **3/14/11 - emailed ~900 forensic DNA scientists** (SWGDAM, forens-dna, ENFSI, EDNAP) inviting them to sign onto a letter that will be sent to Applied Biosystems expressing concern with ABI 3500
- **Very positive response with 101 who agreed to sign the letter**
- Letter was sent March 31 to the president of ABI and scientists involved with the ABI 3500
- **Community will be notified of ABI's response**

A Sampling of Feedback I Received…

- **People did not just sign the letter but many have an opinion about the issues or concern about ABI customer support (I have received >100 emails – often with some very strong thoughts)**

- "I think that the AB3500 related issues most likely represent the beginning of a sea of problems, against which every independent lab must take arms. **It is not up to the manufacturer of a machine to decide the basic procedures of a lab - it is up to the lab**" (4/29/11)

- "I greatly appreciate your advocacy on behalf of our community. **Hopefully we will be heard**." (4/1/11)

Concerns Expressed in Open Letter

- RFID tags
- New .hid file structure requires new software
- Short shelf life of reagents – would like to see data for expiration times

Hopefully a change will result…

A desire for greater communication with the community – the 3500 FAQ sheet is a good start but does not directly address all of the concerns raised

What was learned from ABI visit to NIST on May 11, 2011

- RFID over-ride is possible (their R&D lab has instrument that can use “expired” reagents)
- New software is required for 3500 .hid or .fsa files due to new file structure
- They do not have ANY data to support short shelf life of 3500 reagents
  - hard stops keep labs from having failures that lead to ABI having to replace arrays
- ABI 31xx instruments have a 4X signal reduction

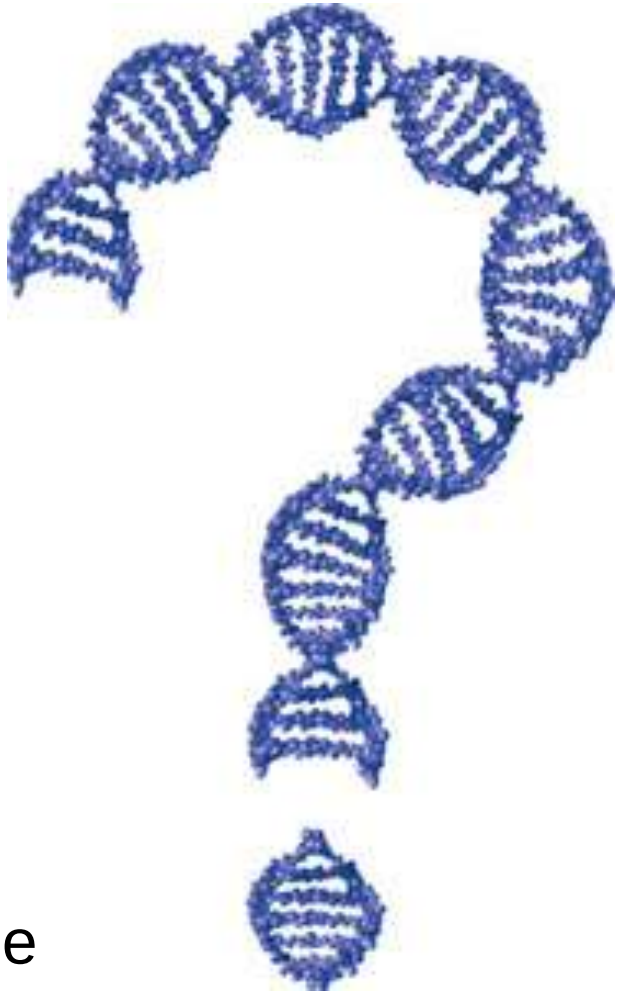
Support to the Community

…Bringing traceability and technology to the scales of justice…

- Conduct interlaboratory studies
- Perform beta-testing of new human identity testing products
- **We collaborate with other NIJ grantees**
- We provide input to (or have aided):
  - Scientific Working Group on DNA Analysis Methods (**SWGDAM**)
  - Department of Defense Quality Assurance Oversight Committee for DNA Analysis
  - Virginia DFS Science Advisory Committee
  - American Prosecutor's Research Institute (**APRI**) DNA Forensics Program "Course-in-a-Box" for training lawyers
  - WTC Kinship and Data Analysis Panel (**KADAP**) and Hurricane Katrina efforts
  - NIJ Expert System Testbed (**NEST**) Project

Thank you for your attention

Acknowledgments: Applied Biosystems, Promega, and Qiagen for STR kits used in concordance studies



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

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