




## Forensic DNA Projects at NIST


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### NIST Human Identity Project Team


  
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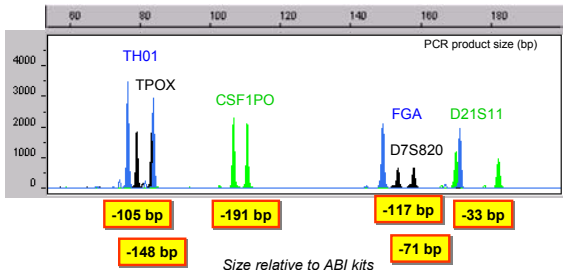
### Current Areas of NIST Research Effort

- Resources for "Challenging Samples"** (degraded DNA or mixtures)
  - miniSTRs (reduced size amplicons for CODIS loci) – *J. Forensic Sci.* 2003, 48, 1054-1064
  - New set of miniSTR loci under development
  - Autosomal SNP assays and U.S. population information – 70 Orchid SNPs
  - mtDNA coding SNP multiplex assays with AFDIL mtGenome sequencing
- Y-Chromosome Information, Assays, and Standards**
  - Y-STR 20plex, 11plex and U.S. population data including multi-copy DYS464
  - Y-SNP U.S. population data - *J. Forensic Sci.*, in press (July 2004)
  - Human Y-Chromosome Standard Reference Material (SRM 2395)
- DNA Quantitation**
  - Interlab study and standard reference material (SRM 2372)
  - Real-time PCR comparisons
- Resource Information to Aid Forensic Laboratories**
  - STRBase (STR fact sheets and variant allele lists)
  - Forensic SNP site
- Work with New Typing Technologies**
  - Microchip CE DNA separations

*J. Forensic Sci.* Sept 2003 issue  
John M. Butler, Ph.D.; Yin Shea, Ph.D.; and Bruce R. McCord Ph.D.<sup>2</sup>

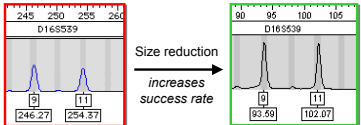
**The Development of Reduced Size STR Amplicons as Tools for Analysis of Degraded DNA\***

Describes new miniSTR primer sequences for all CODIS loci and initial assays developed



Size relative to ABI kits



### New DNA Tests with miniSTRs




Size reduction → increases success rate

Same result but with smaller region of DNA

New test developed to aid in identification of World Trade Center victims of 9/11/01 terrorist attacks

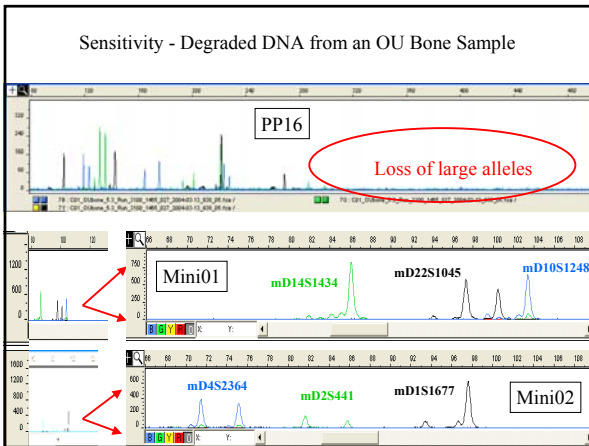
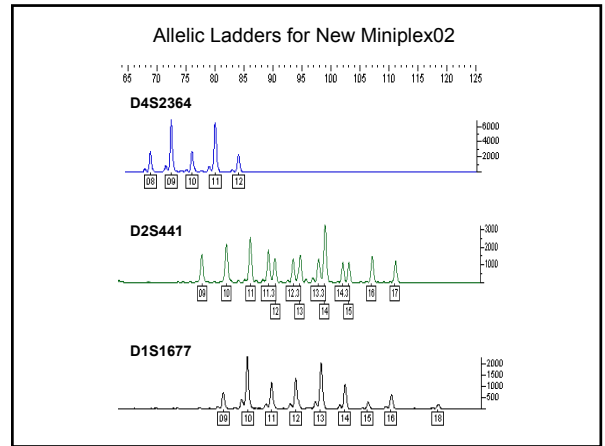
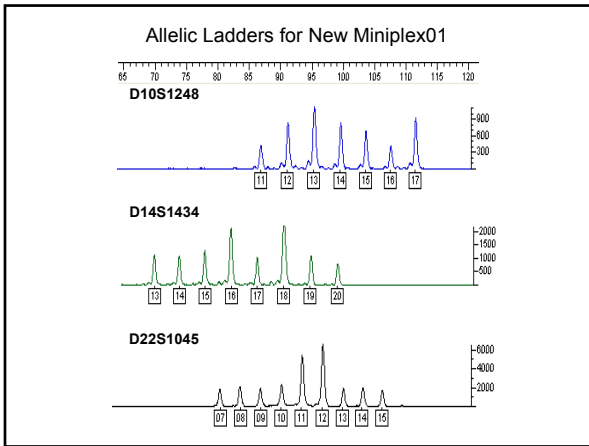


Has resulted in an increase in the number of WTC victims identified

### Initial Testing Results for New miniSTR Loci

```

>900 potential markers
  ↓
61 markers with "clean" flanking regions
  ↓
43 markers with amplicon size < 125bp
  ↓
18 markers for initial testing
  ↓
2 three loci miniplexes
    
```



**New Y-Chromosome NIST Standard Reference Material**

**Human Y-Chromosome DNA Profiling Standard**

- 5 male samples + 1 female sample (neg. control)
- 100 ng of each (50 µL at ~2 ng/µL) **\$245**
- 22 Y STR markers sequenced
- 9 additional Y STR markers typed
- 42 Y SNPs typed with Marigen kit

**Certified for all loci in commercial Y-STR kits:**

- Y-PLEX 6
- Y-PLEX 5
- Y-PLEX 12
- PowerPlex Y

**Minimal Haplotype loci and SWGDAM recommended loci:**  
 DYS19, DYS385 a/b, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS436, DYS439

**NIST STRBase**  
 National Institute of Standards and Technology  
 Short Tandem Repeat DNA Internet Database

... working with industry to develop and apply technology, measurements and standards.

**Recent Additions**

- Forensic SNP Information (will be official site for ISFG SNP information) .../SNP.htm
- NIST publications and presentations as pdf files .../NISTpub.htm

**We Regularly Update**

- Reference List
- Variant Alleles
- Addresses for Scientists
- Links to Other Web Sites
- Y-STR Information

We will continue to add downloadable PowerPoint files that can be used for training purposes

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

**ENFSI/ISFG SNP Site** (<http://www.cstl.nist.gov/biotech/strbase/SNP.htm>)

**Forensic SNP Information**

This site is intended to provide general information on single nucleotide polymorphisms (SNP) markers that may be of interest in human identification applications. Many of these markers come from The SNP Consortium (TSC) efforts or are already present in the NCEI SNP database. To submit a SNP marker for inclusion on this forensic SNP site, please provide the requested information on a standardized SNP fact sheet (click here to download) to John Butler via email: [john.butler@nist.gov](mailto:john.butler@nist.gov)

[Markers] [Assays] [SNP Typing Technologies]

Last Updated: 04/12/04

**Markers**

**Autosomal SNPs**

- TSC 0252540 (submitted by Peter Gill)
- TSC 1342445 (submitted by Peter Gill)
- TSC 0421768 (submitted by Peter Gill)
- TSC 0478751 (submitted by Peter Gill)
- TSC 0320706 (submitted by Peter Gill)
- TSC 0155410 (submitted by Peter Gill)
- TSC 0154197 (submitted by Peter Gill)

Marker Designation	Multiplex Panel	PCR Product Size (bp)	Chromosome Chr.	Position	Database Reference	SNP (db) ID	dbSNP ref
D1S1677	Y1a	100	1	111,848,000	Y1a	111848	111848
D2S441	Y1a	100	2	102,441,000	Y1a	102441	102441
D3S1358	Y1a	100	3	47,358,000	Y1a	47358	47358
D4S2364	Y1a	100	4	102,364,000	Y1a	102364	102364
D5S418	Y1a	100	5	171,418,000	Y1a	171418	171418
D6S1053	Y1a	100	6	15,105,000	Y1a	15105	15105
D7S820	Y1a	100	7	44,820,000	Y1a	44820	44820
D8S1179	Y1a	100	8	143,179,000	Y1a	143179	143179
D9S1125	Y1a	100	9	111,125,000	Y1a	111125	111125
D10S1248	Y1a	100	10	111,248,000	Y1a	111248	111248
D11S1871	Y1a	100	11	111,871,000	Y1a	111871	111871
D12S1739	Y1a	100	12	111,739,000	Y1a	111739	111739
D13S322	Y1a	100	13	111,322,000	Y1a	111322	111322
D14S1434	Y1a	100	14	111,434,000	Y1a	111434	111434
D15S658	Y1a	100	15	111,658,000	Y1a	111658	111658
D16S1961	Y1a	100	16	111,961,000	Y1a	111961	111961
D17S1196	Y1a	100	17	111,196,000	Y1a	111196	111196
D18S281	Y1a	100	18	111,281,000	Y1a	111281	111281
D19S253	Y1a	100	19	111,253,000	Y1a	111253	111253
D20S1117	Y1a	100	20	111,117,000	Y1a	111117	111117
D21S11	Y1a	100	21	111,11,000	Y1a	11111	11111
D22S1045	Y1a	100	22	111,1045,000	Y1a	1111045	1111045
D23S448	Y1a	100	23	111,448,000	Y1a	111448	111448
D24S241	Y1a	100	24	111,241,000	Y1a	111241	111241
D25S192	Y1a	100	25	111,192,000	Y1a	111192	111192
D26S195	Y1a	100	26	111,195,000	Y1a	111195	111195
D27S1329	Y1a	100	27	111,1329,000	Y1a	1111329	1111329
D28S1328	Y1a	100	28	111,1328,000	Y1a	1111328	1111328
D29S1327	Y1a	100	29	111,1327,000	Y1a	1111327	1111327
D30S1326	Y1a	100	30	111,1326,000	Y1a	1111326	1111326
D31S1325	Y1a	100	31	111,1325,000	Y1a	1111325	1111325
D32S1324	Y1a	100	32	111,1324,000	Y1a	1111324	1111324
D33S1323	Y1a	100	33	111,1323,000	Y1a	1111323	1111323
D34S1322	Y1a	100	34	111,1322,000	Y1a	1111322	1111322
D35S1321	Y1a	100	35	111,1321,000	Y1a	1111321	1111321
D36S1320	Y1a	100	36	111,1320,000	Y1a	1111320	1111320
D37S1319	Y1a	100	37	111,1319,000	Y1a	1111319	1111319
D38S1318	Y1a	100	38	111,1318,000	Y1a	1111318	1111318
D39S1317	Y1a	100	39	111,1317,000	Y1a	1111317	1111317
D40S1316	Y1a	100	40	111,1316,000	Y1a	1111316	1111316
D41S1315	Y1a	100	41	111,1315,000	Y1a	1111315	1111315
D42S1314	Y1a	100	42	111,1314,000	Y1a	1111314	1111314

### Variant Alleles Cataloged in STRBase

[http://www.cstl.nist.gov/biotech/strbase/var\\_tab.htm](http://www.cstl.nist.gov/biotech/strbase/var_tab.htm)

#### Off-Ladder Alleles

201 total variants reported as of 09/05/03

- CSF1PO (9)
- D2S1338 (2)
- D3S1338 (15)
- D5S818 (5)
- D7S820 (19)
- D8S1179 (4)
- D13S317 (6)
- D16S539 (10)
- D18S51 (27)
- D19S433 (4)
- D21S11 (19)
- FBSPPS (1)
- FGA (60)
- HUMTH01 (4)
- Penta E (2)
- TPOX (7)
- VWA (5)

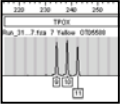
**Currently 201**  
at 13/13 CODIS loci

#### Tri-Allelic Patterns


49 total patterns reported as of 09/15/03

- CSF1PO (2)
- D3S1338 (4)
- D5S818 (1)
- D7S820 (0)
- D8S1179 (3)
- D13S317 (3)
- D16S539 (1)
- D18S51 (4)
- D21S11 (4)
- FGA (7)
- HUMTH01 (1)
- TPOX (12)
- VWA (7)

**Currently 49**  
at 12/13 CODIS loci



Total number of profiles: 1,692,004 (as of March 2004)




Convicted Offenders




Forensic Casework

Missing Persons

**FBI Laboratory**



**State and Local Forensic Laboratories**

	2,240	1,773	119
Investigations aided (thru March 2004)	2,240	1,773	119
Offender Profiles	174,392	181,769	16,924
Forensic Samples	5,790	3,060	1,007