Forensic DNA Projects at NIST

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Cyprus
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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
  - 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

New DNA Tests with miniSTRs

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

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

ENFSI Meeting (Nicosia, Cyprus)
Allelic Ladders for New Miniplex01

<table>
<thead>
<tr>
<th>Allele</th>
<th>Size (bp)</th>
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<tbody>
<tr>
<td>D10S1248</td>
<td>80</td>
</tr>
<tr>
<td>D14S1434</td>
<td>100</td>
</tr>
<tr>
<td>D22S1045</td>
<td>120</td>
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Allelic Ladders for New Miniplex02

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<th>Allele</th>
<th>Size (bp)</th>
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<tbody>
<tr>
<td>D4S2364</td>
<td>80</td>
</tr>
<tr>
<td>D2S441</td>
<td>100</td>
</tr>
<tr>
<td>D1S1677</td>
<td>120</td>
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</tbody>
</table>

Sensitivity - Degraded DNA from an OU Bone Sample

PP16

Loss of large alleles

Mini01

Mini02

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)
- 22 Y STR markers sequenced
- 9 additional Y STR markers typed
- 42 Y SNPs typed with Marligen 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, DYS438, DYS439

STRBase

Short Tandem Repeat DNA Internet Database

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

STRBase (http://www.cstl.nist.gov/biotech/strbase)

ENFSI Meeting (Nicosia, Cyprus)
Variant Alleles Cataloged in STRBase

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

<table>
<thead>
<tr>
<th>Off-Ladder Alleles</th>
<th>Tri-Allelic Patterns</th>
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</thead>
<tbody>
<tr>
<td>201 total patterns reported as of 09/2003</td>
<td>49 total patterns reported as of 09/2003</td>
</tr>
<tr>
<td>Currently 201 at 12/13 CODIS loci</td>
<td>Currently 49 at 12/13 CODIS loci</td>
</tr>
</tbody>
</table>

- CSF1PO (9)
- D8S1179 (13)
- D3S1358 (13)
- D16S539 (12)
- D5S818 (12)
- D7S820 (12)
- D3S1358 (12)
- D21S11 (12)
- vWA (12)

- CSF1PO (9)
- D8S1179 (9)
- D3S1358 (5)
- D16S539 (5)
- D5S818 (5)
- D7S820 (5)
- D3S1358 (5)
- D21S11 (5)
- vWA (5)

**FBI Laboratory**

Investigations aided (thru March 2004):
- Convicted Offenders: 2,240
- Forensic Casework: 1,773
- Missing Persons: 119

**State and Local Forensic Laboratories**

- Convicted Offenders: 174,392
- Forensic Samples: 3,060
- Missing Persons: 1,007
- Total number of profiles: 1,692,004 (as of March 2004)