STRBase

Short Tandem Repeat DNA Internet Database

Recent Additions
- Forensic SNP information (will be official site for ISFG SNP Information)
- NIST publications and presentations as pdf files

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

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

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

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

Outline for Presentation
- STRBase Updates
- Variant Allele Cataloging and Characterization
- SRM 2395 Y-Chromosome Standard
- mtDNA Typing Work
- miniSTR Work
- Future Work
- Another NIST Interlaboratory Study

Variant Alleles Cataloged in STRBase

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

Off-Ladder Alleles

Currently 201 at 13/13 CODIS loci

Tri-Allelic Patterns

Currently 49 at 12/13 CODIS loci

Large D18 Allele Characterized at NIST

Sample from Christine Moraczewski, NE State Patrol Crime Lab

PowerPlex 16

D18

Penta E

FGA

CSF

D18S51 monoplex

Size difference = 102 bp

Shoulder on Penta E allele

Allele 14

Allele 40

Allele 12

Typical D18S51 allele range (8-27)

Size difference = 105 bp

ABI D18S51 monoplex gave same result

Analysis of Common STR Variant Alleles

- We have monoplex primers for all common STR loci and kits
- We have sequencing primers that bind outside of STR kit primer sequence positions to enable view of polymorphic nucleotides that cause primer binding site mutations
- NIJ has funded us to characterize STR variants for the forensic DNA community

Margaret Kline
New Y-Chromosome NIST SRM

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 S
- Y-PLEX 12
- PowerPlex Y

$245

Come see Poster #32 on NIST SRM 2395 and Other Y-Chromosome Work

SRM 2395

NIST Y-Chromosome DNA Profiling Standard

Helps establish allele nomenclature and confirm/calibrate typing data

- 22 Y STRs sequenced
- 42 Y SNPs typed

Come see Poster #27 on NIST SRM 2395 and Other Y-Chromosome Work

Warning with SRM 2395 Component C and DYS385 Locus Using Original Y-PLEX 6 Macro

DYS385 allele 20 is called correctly with new Y-PLEX 12 kit

DYS385 allele 20 is called correctly with new Y-PLEX 12 kit

NIST mtDNA Work

Roche Linear Arrays
(probes for HV1/HVII)

Data from Population Study

Coding Region mtSNP 11plex
(minisequencing assay)

Developed with AFDIL to resolve mtDNA most common types

Int. J. Legal Med., submitted

Semi-Automation of mtDNA LINEAR ARRAYS

Agilent Bioanalyzer 2100
(quantifies PCR products)

Tecan Problot – processes sample through wash steps

Analysis of probe results is still manual!

Come see Poster #30 on Semi-Automation of mtDNA Arrays

Some Results with Roche mtDNA LINEAR ARRAYS

Typing frequencies for 666 NIST population samples

<table>
<thead>
<tr>
<th>Sample</th>
<th>% Types</th>
<th>% People</th>
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>82 different types
>185 were unique (occurred only once)
>61 samples had "Most Common Type"

Accurate Detection of Heteroplasmy at 16093

“Most Common Type” evaluated further with mtDNA coding region SNP assay

Margaret Kline
mtDNA Coding Region 11plex SNAPSHOT Assay

Result from 1 pg (genomic DNA)

11plex PCR and 11plex SNP detection
Sites are polymorphic in Caucasians (H1) and useful in resolving most common HV1/HV2 types
Multiplex PCR used to co-amplify all regions of interest at once
PCR product sizes kept under 200 bp to enable success with degraded DNA samples

"Most Common mtDNA Type"

Examined with mtDNA Coding SNP 11plex

51 samples were identical by Roche mtDNA Linear Arrays

111111AT

12 haplogroups were observed
4 haplogroups were unique

miniSTR Work

- miniSTRs (a.k.a. BodePlexes) are being used successfully in WTC effort
- Collaboration with Bruce McCord (NIJ-funded) to further develop reduced size STR amplicons
- Starting a new project this fall with Mike Coble (NRC postdoc) to develop new miniSTR loci

Future Work

- Quality assurance testing software
  - Dave Duewer in collaboration with NCBI
- Quantitation standard SRM 2372
- Evaluation of real-time PCR methodologies for DNA quantitation
- More miniSTR work for degraded DNA
- Locus-specific brackets for alternative Y-STR typing
- Comparison of new SNP markers to STRs in NIST U.S. population sample set

NIST U.S. Population Samples

As of 06/2003: 666 males (anonymous; self-identified ethnicities)

- 260 Caucasians
- 260 African Americans
- 143 Hispanics
- 3 Asians

Whole blood received from Interstate Blood Bank (Memphis, TN)

Working tubes/plates: 1 ng/uL

To date: (~50,000 allele calls)

- Identifier (15 autosomal markers + Amelogenin) (10,608)
- Roche Linear Arrays (HV1/HV2 10 regions) (6,630)
- Y STRs 22 loci—27 amplicons (17,388)
- Y SNPs 50 markers on sub-set of samples (11,498)

Another NIST Interlaboratory Test

- Solicitation for 2004 NIST Quantitation Study
  - Plan to ship in Nov 2003
  - Data due by March 1, 2004
- Purpose to examine accuracy of current DNA quantitation methods and measure stability of prototype NIST quantitation SRM
- Open to all human identity testing labs
- Will involve only quantifying 6-8 samples
- Handouts available
Acknowledgments

**Interagency Agreement between National Institute of Justice and NIST Office of Law Enforcement Standards**

**NIST Project Team:**
- John Butler (project leader)
- Pete Vallone
- Jan Redman
- Rich Schoske (American U)
- Dave Dueser

**Collaborators:**
- Mike Hammer and Alan Redd (U. AZ) for Y STR sequences
- Tom Parsons and Mike Cable (AFDIL) for mtDNA coding SNP work
- Sandy Calvesso (Roche) for mtDNA linear arrays
- Bruce McGill and students (Ohio U) for miniSTR work

**Funding:**
- Interagency Agreement between National Institute of Justice and NIST Office of Law Enforcement Standards

Available as pdf files from [http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm](http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm)

**Publications from Our Group this Past Year**


This presentation available as pdf file from [http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm](http://www.cstl.nist.gov/biotech/strbase/NISTpub.htm)