

Overview of the Applied Genetics Group

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FORENSICS @ NIST

#NISTForensics



Applied Genetics Group – Forensic Genetics



Group Leader
Peter
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Michael
Coble



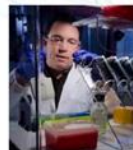
Becky
Steffen



Erica
Romsos



Katherine
Gettings



Kevin
Kiesler



Margaret
Kline



Lisa
Borsuk



Sarah
Riman



David
Duewer

Advancing technology and traceability through quality genetic measurements to aid work in Forensic Genetics

A core competency of our group is the application of *nucleic acid-based methods*
PCR – Genotyping – Sequencing – Real-time PCR – Digital PCR - DNA based SRMs

Past and Present Funding



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

Past and Present Collaborations





APPLIED GENETICS GROUP

Standards for the Forensics Human Identification Community

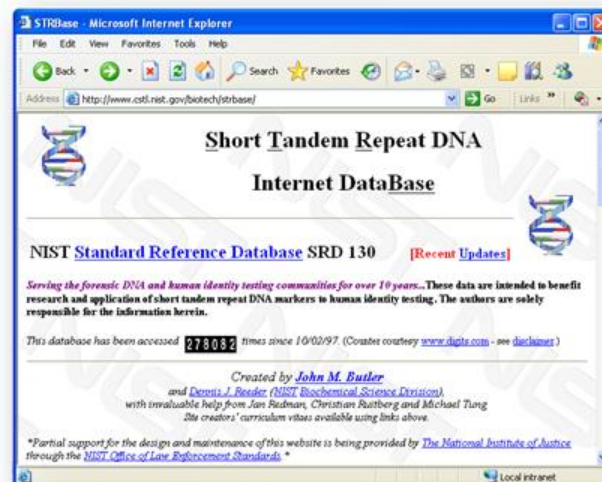
SRM 2391c: enables forensic laboratories to meet the FBI DNA Quality Assurance Standard. **All U.S. labs uploading and searching DNA profiles to the National DNA Database (NDIS) are required to run the NIST SRM annually and when validating new typing methods**



Photos Matt Delorme

SRM 2372: for the quantification of human DNA. Used to calibrate DNA quantitation kits (an essential step in the DNA typing workflow)

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



SRMs enable US manufacturers to provide accurate DNA typing kits





New Core short tandem repeat (STR) Loci in the US database

The U.S. national DNA database currently has 12 million profiles and continues to grow quickly each year

13 to 20 STR loci in the National DNA Database

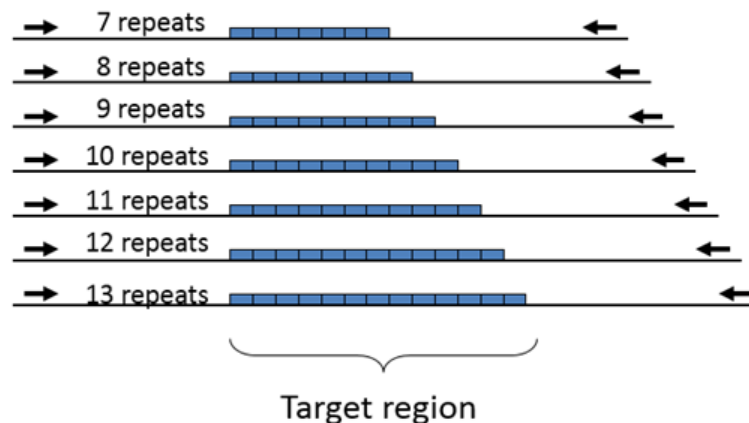
Three of the seven new STR loci were discovered and characterized by NIST

Forensic labs in the U.S. will need to adopt these new core STR loci by 2017

New markers = new commercial STR kits

We have facilitated in the beta testing of over 20 new commercial STR kits with industry partners (Promega, Qiagen)

Short tandem repeat (STR)
a repeat sequence region in the human genome



We have provided the allele frequencies of these 20 STR markers that allow for the calculation of matching statistics

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Letter to the Editor

U.S. population data for 29 autosomal STR loci

Dear Editor,

run and population statistics were confirmed using the PowerMarker v3.25 statistics program [10]. There were 14 instances where statistically significant deviations from Hardy–Weinberg expectations based on the exact test



Rapid DNA Profiling

Current methods for forensic DNA typing of short tandem repeat markers (STRs) require on the order of **6-8 hours** with trained laboratory analysts

The potential to generate a DNA profile **'hands off'** in the less than 2 h will open up new applications of forensic DNA typing (non-lab environments such as booking stations and borders)

- Supporting US commercial R-DNA developers
- Funding from FBI and DHS stakeholders for the implementation and assessment of R-DNA

The FBI goal is to have one of these instruments in every booking station for enrollment of arrestees into the national DNA database

Silver Medal Award: for the development of rapid forensic DNA typing techniques that enable state-of-the-art human identity testing and DNA biometrics



NetBio | Rapid DNA Analysis™ Solutions



First-generation R-DNA prototypes



Next Generation Sequencing (NGS) for Human Identity Testing

The emerging technology of next generation sequencing allows for the sequencing forensically relevant genetic markers

- The core US STRs (short tandem repeats)
- Genetic markers for biogeographical ancestry and eye/hair color estimation
- Currently sequencing approximately 1000 well-characterized samples to provide **allele frequencies** for the downstream application of the new technology
- Existing Standard Reference Materials (SRMs) are being updated for the expanded markers in support of the calibration and adoption of NGS



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 [AGAT]13 [AGAC]7 AGAT
 [AGAT]13 [AGAC]8
 [AGAT]14 [AGAC]6 AGAT
 [AGAT]14 [AGAC]7

STR sequences



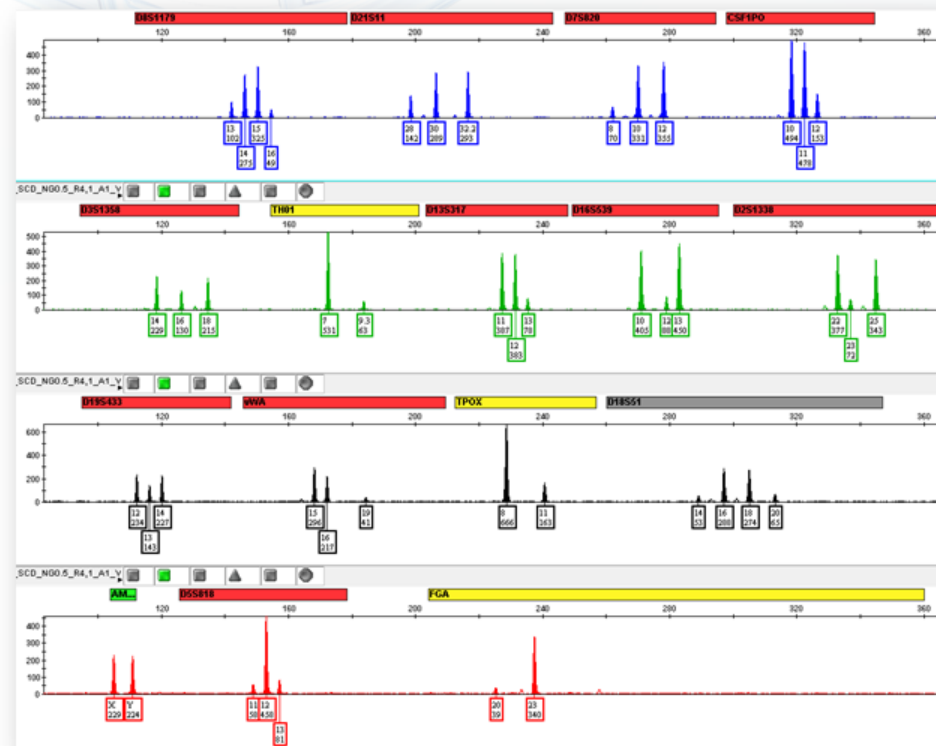


Interpretation of Complex DNA Mixtures

DNA mixtures of two or more individuals are among the most challenging cases to interpret for the DNA laboratory. Software tools to help the analyst to interpret challenging DNA mixtures are now available.

- The Applied Genetics group at NIST is currently exploring and testing both commercially available and open source software tool for complex DNA mixture interpretation.
- Education and training via publications and webcasts have been developed and presented to the forensic DNA community,
- Future inter-laboratory exercises to help laboratories to identify areas of improvement are planned.

Mix 05 and Mix 13 interlaboratory studies



Thank you!

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