



FORENSICS @ NIST

#NISTForensics

# Forensic Genetics

Peter M. Vallone, PhD

Leader, Applied Genetics Group



# Applied Genetics Group – Forensic & Clinical Genetics



Becky  
Steffen



Erica  
Romsos



Katherine  
Gettings



Kevin  
Kiesler



Margaret  
Kline



Lisa  
Borsuk



Sarah  
Riman



David  
Duewer



Megan  
Cleveland

One aspect of the Applied Genetics group is focused on forensic genetics. Using DNA–based technologies, the AG group ***develops standards and assesses emerging forensic methods in support of the human identity community.***

These activities provide a foundation to ensure accurate measurements and validations performed by the forensic DNA typing community.



FORENSICS @ NIST

#NISTForensics

# Applied Genetics Group – Forensic Genetics

## Standard Reference Materials

SRM 2372a: Human DNA Quantitation Standard (March 2018) →

***Under production*** SRM 2391d PCR-based DNA Standard



Five components

**A-C three single-source components**

**D one mixture; approximately 3:1 (F:M)**

**E one component: cells spotted on FTA paper (from cell lines)**

- Components A-D are DNA extracted from blood (not cell lines)
- Certified allele calls for U.S. core STR loci
- Characterized by CE- and **NGS-based methods (SNPs, mitochondrial genome)**
- Supports the FBI Quality Assurance Standards



**FORENSICS @ NIST**

**#NISTForensics**

# Applied Genetics Group – Forensic Genetics

## Next Generation Sequencing

ELSEVIER  
OPEN ACCESS

Forensic Science International: Genetics 37 (2018) 106–115

Contents lists available at ScienceDirect

Forensic Science International: Genetics

journal homepage: [www.elsevier.com/locate/fsigen](http://www.elsevier.com/locate/fsigen)

Research paper

Sequence-based U.S. population data for 27 autosomal STR loci

Katherine Butler Gettings\*, Lisa A. Borsuk, Carolyn R. Steffen, Kevin M. Kiesler, Peter M. Vallone

*U.S. National Institute of Standards and Technology, Biomolecular Measurement Division, 100 Bureau Drive, Gaithersburg, MD 20899, USA*



Allele frequencies from NIST population samples enable the use of sequence-based methods for typing STR markers

## ELECTROPHORESIS

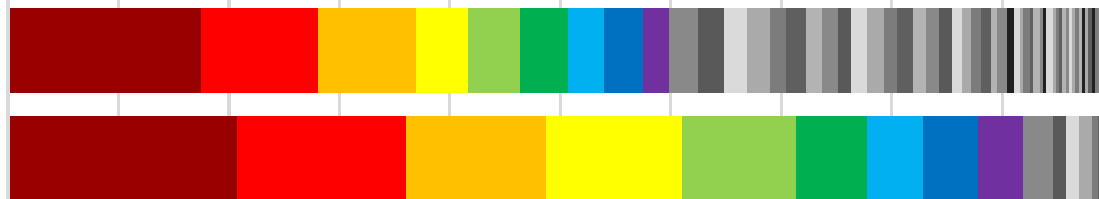
Research Article | [Full Access](#)

Sequence-based U.S. population data for the SE33 locus

Lisa A. Borsuk✉, Katherine B. Gettings, Carolyn R. Steffen, Kevin M. Kiesler, Peter M. Vallone

First published: 07 May 2018 | <https://doi.org/10.1002/elps.201800091>

Sequence  
D12S391  
Length



89 alleles  
24 alleles



# FORENSICS @ NIST

## #NISTForensics

# Applied Genetics Group – Forensic Genetics

## Next Generation Sequencing

Forensic Science International: Genetics 31 (2017) 111–117

Contents lists available at ScienceDirect



ELSEVIER

Forensic Science International: Genetics

journal homepage: [www.elsevier.com/locate/fsigen](http://www.elsevier.com/locate/fsigen)

STRSeq – cataloging STR alleles at NCBI

<https://www.ncbi.nlm.nih.gov/bioproject/380127>

Research paper

STRSeq: A catalog of sequence diversity at human identification Short Tandem Repeat loci



Katherine Butler Gettings<sup>a,\*</sup>, Lisa A. Borsuk<sup>a</sup>, David Ballard<sup>b</sup>, Martin Bodner<sup>c</sup>, Bruce Budowle<sup>d,e</sup>, Laurence Devesse<sup>b</sup>, Jonathan King<sup>d</sup>, Walther Parson<sup>c,f</sup>, Christopher Phillips<sup>g</sup>, Peter M. Vallone<sup>a</sup>

Forensic Science International: Genetics 34 (2018) 162–169

Contents lists available at ScienceDirect



Forensic Science International: Genetics

journal homepage: [www.elsevier.com/locate/fsigen](http://www.elsevier.com/locate/fsigen)



Providing STR nomenclature support

“The devil’s in the detail”: Release of an expanded, enhanced and dynamically revised forensic STR Sequence Guide



C. Phillips<sup>a,\*</sup>, K. Butler Gettings<sup>b</sup>, J.L. King<sup>c</sup>, D. Ballard<sup>d</sup>, M. Bodner<sup>e</sup>, L. Borsuk<sup>b</sup>, W. Parson<sup>e,f</sup>



FORENSICS @ NIST

#NISTForensics

# Applied Genetics Group – Forensic Genetics

## STRBase 2.0

<https://strbase-b.nist.gov/>

Under construction

Enhanced searching, sorting, downloading

The screenshot shows the STRBase 2.0 website. At the top left is the NIST logo (National Institute of Standards and Technology, U.S. Department of Commerce). The main header is "STRBase 2.0 (Short Tandem Repeat DataBase)". Below the header is a navigation menu with "Forensic Markers", "NIST Resources", "Community Resources", and "About". A search bar is on the right. On the left side, there are four buttons: "Commonly Used Auto STRs", "Other Auto STRs", "X-Chromosome STRs", and "Y-Chromosome STRs". The main content area has an "Introduction" section. The introduction text states: "STRBase is a resource for Short Tandem Repeat and other human identification markers. Within this site, users can navigate, search, and download locus information such as reported variant alleles, tri-allele, and general information including genomic coordinates, allele size ranges, sequence motifs. Information is also available by kit or core set. Registered users can upload newly observed length-based variant alleles and receive alerts of new information on pages of interest." Below this, it says: "Additionally, STRBase hosts content produced by NIST Applied Genetics: publications, presentations, population data, sample data sets, and information regarding Standard Reference Materials of interest to the Forensic DNA community." Further down, it mentions: "STR data produced via next generation sequencing is cataloged separately in the STRSeq BioProject at NCBI, with sequence-specific tools and resources forthcoming at [strseq.nist.gov](http://strseq.nist.gov)". At the bottom of the introduction section is a "Learn More" button. Below the introduction are two buttons: "Acknowledgments" and "Citation Guide". On the right side of the page, there is a photograph of a "2391c PCR-Based DNA Profiling Standard" kit and several small vials. Below the photo is a "News:" section with a date "15-Oct-18" and the text "STRBase 2.0 Launches beta test site!".

**NIST** National Institute of Standards and Technology  
U.S. Department of Commerce

## STRBase 2.0 (Short Tandem Repeat DataBase)

Search

Forensic Markers ▾ NIST Resources ▾ Community Resources ▾ About ▾

[Commonly Used Auto STRs](#)

[Other Auto STRs](#)

[X-Chromosome STRs](#)

[Y-Chromosome STRs](#)

### Introduction

STRBase is a resource for Short Tandem Repeat and other human identification markers. Within this site, users can navigate, search, and download locus information such as reported variant alleles, tri-allele, and general information including genomic coordinates, allele size ranges, sequence motifs. Information is also available by kit or core set. Registered users can upload newly observed length-based variant alleles and receive alerts of new information on pages of interest.

Additionally, STRBase hosts content produced by NIST Applied Genetics: publications, presentations, population data, sample data sets, and information regarding Standard Reference Materials of interest to the Forensic DNA community.

STR data produced via next generation sequencing is cataloged separately in the STRSeq BioProject at NCBI, with sequence-specific tools and resources forthcoming at [strseq.nist.gov](http://strseq.nist.gov).

[Learn More →](#)

[Acknowledgments](#) [Citation Guide](#)

News:

15-Oct-18 - **STRBase 2.0** Launches beta test site!

2391c  
PCR-Based DNA Profiling Standard  
NIST National Institute of Standards and Technology  
U.S. Department of Commerce

Photo: Matt DeLorme

# Applied Genetics Group – Forensic Genetics

## Other projects/activities

- Assessing DNA extraction efficiency
- Digital PCR for quantifying DNA
  - Understanding bias in qPCR measurements
- Use of probabilistic genotyping software for mixture analysis
- The use of SNP markers for ancestry and eye/hair color prediction
- IARPA – **Proteos** project - QC of DNA materials and eval. of extraction procedures
  - The use of proteins for human identification
- Various group member participating in forensic working groups
  - FBI-SWGDAM, OSAC, Forensic Laboratory Needs-TWG, NIJ-TWG, FBI RDNA task force



FORENSICS @ NIST

#NISTForensics

# Applied Genetics Group – Forensic Genetics

Today

Characterization of noise in targeted sequencing of STR markers: Sarah Riman

Results from the 2018 Rapid DNA Maturity Assessment: Erica Romsos

Sequencing and standards for characterization of the mitochondrial genome: Kevin Kiesler

## Funding

NIST Special Programs Office: *Forensic DNA*

FBI Biometrics Center of Excellence: *Forensic DNA Typing as a Biometric tool.*

NIJ: *STRSeq and Nomenclature*

*Contact - Peter.Vallone@nist.gov*



FORENSICS @ NIST

