19th International Forensic Science Managers Symposium
INTERPOL Headquarters, Lyon, France
10 October 2019

Recent Advances in Forensic Biology and Forensic DNA Typing 2016-2019

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Review was written with Sheila Willis, Ph.D.
(former director of Forensic Science Ireland and a NIST guest researcher from 2017-2019)

NIST

Acknowledgments and Disclaimer

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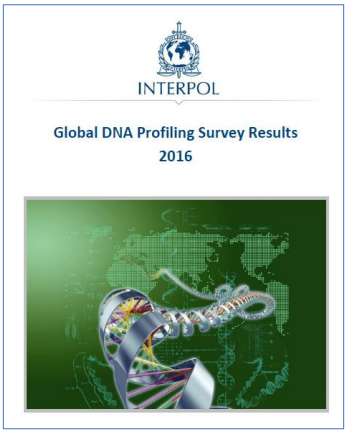
Feedback on the written review from Tom Callaghan (FBI Laboratory) and Erica Romsos (NIST Applied Genetics Group)

Points of view are the presenter and do not necessarily represent the official position or policies of the National Institute of Standards and Technology.

Certain commercial equipment, instruments and materials are identified in order to specify experimental procedures as completely as possible. In no case does such identification imply a recommendation or endorsement by the National Institute of Standards and Technology nor does it imply that any of the materials, instruments or equipment identified are necessarily the best available for the purpose.

Status of DNA Use in INTERPOL Member Countries Through 2016 (95 of 190 member countries responded)



https://www.interpol.int/content/download/4875/file/GlobalDNASurvey.pdf

Estimated global total DNA profiles was **35.4 million** (at end of 2016)

84 countries use DNA in criminal investigations

Region	Member Country
AFRICA	Algeria
	Botswana
	Egypt
	Ghana
	Seychelles
	South Africa
	Sudan
	Swaziland
	Tunisia
TOTAL	9
AMERICAS	Brazil
	Canada
	Chile
	Colombia
	Costa Rica
	Ecuador
	El Salvador
	Honduras
	Nicaragua
	Panama
	St. Lucia
	United States of America
	Uruguay
TOTAL	13
ASIA & SOUTH PACIFIC	Australia
	Bahrain
	Brunei
	China
	India
	Iraq
	Japan
	Kuwait
	Lebanon
	Malaysia
	Nepal
	New Zealand
	Philippines
	Saudi Arabia
	Singapore
	Syria
	Turkmenistan
	Uzbekistan
	Vietnam
TOTAL	19

Region	Member Country
EUROPE	Albania
	Andorra
	Armenia
	Austria
	Azerbaijan
	Belarus
	Belgium
	Bosnia & Herzegovina
	Bulgaria
	Cyprus
	Czech Republic
	Denmark
	Estonia
	Finland
	France
	FYR Macedonia
	Georgia
	Germany
	Greece
	Hungary
	Iceland
	Ireland
	Israel
	Latvia
	Liechtenstein
	Lithuania
	Luxembourg
	Malta
	Monaco
	Montenegro
	Netherlands
	Norway
	Portugal
	Romania
	Russia
	Serbia
	Slovakia
	Slovenia
	Spain
	Sweden
	Switzerland
	Ukraine
	United Kingdom
TOTAL	43

69 countries possess a national DNA database

Region	Member country
AFRICA	Botswana
	Egypt
	South Africa
	Sudan
	Tunisia
TOTAL	5
AMERICAS	Brazil
	Canada
	Chile
	Colombia
	Costa Rica
	Ecuador
	El Salvador
	Honduras
	Panama
	St Lucia
	United States of America
	Uruguay
TOTAL	12
ASIA & SOUTH PACIFIC	Australia
	Bahrain
	China
	Iraq
	Japan
	Kuwait
	Lebanon
	Malaysia
	New Zealand
	Philippines
	Saudi Arabia
	Singapore
	Syria
	Vietnam
TOTAL	14

Region	Member country
EUROPE	Albania
	Austria
	Azerbaijan
	Belarus
	Belgium
	Bulgaria
	Cyprus
	Czech Republic
	Denmark
	Estonia
	Finland
	France
	FYR Macedonia
	Georgia
	Germany
	Greece
	Hungary
	Iceland
	Ireland
	Israel
	Latvia
	Liechtenstein
	Lithuania
	Luxembourg
	Malta
	Montenegro
	Netherlands
	Norway
	Portugal
	Romania
	Russia
	Slovakia
	Slovenia
	Spain
	Sweden
	Switzerland
	Ukraine
	United Kingdom
TOTAL	38

73 countries use Y-STRs; **31 countries** use mtDNA

Overview of Papers Reviewed

19th International Forensic Science Managers Symposium

Years Examined	2016, 2017, 2018, 2019 (through July 2019)
Number of Papers	**235**
Number of Journals	35
Topics Covered	12 *(see next slide)*

17th INTERPOL Review on DNA (2010-2013) examined **114 articles** (Jolicoeur 2013)
18th INTERPOL Review on DNA (2013-2016) examined **75 articles** (Laurent & Pene 2016)

Topics Covered: Forensic Biology and DNA Typing

1. Core Loci Expansion
2. Rapid Analysis of STR Markers
3. Investigative Genetic Genealogy
4. Next-Generation Sequencing
5. DNA Mixture Interpretation and Probabilistic Genotyping Software
6. DNA Transfer and Activity Level Evaluations
7. Forensic Biology and Body Fluid Identification
8. DNA Phenotyping
9. Privacy and Ethical Issues
10. Guidance Documents (SWGDAM, OSAC, ASB, ENFSI, UK FS Regulator)
11. Contamination Avoidance and DNA Success Rates
12. Recent Special Issues and Review Articles of Note

Journals (Alphabetical Listing)

Traditional forensic science journals are in **bold font**

1. Analytical Chemistry
2. Applied Spectroscopy
3. BMC Bioinformatics
4. BMC Genetics
5. Current Issues in Criminal Justice
6. Developing World Bioethics
7. Electrophoresis **40 citations**
8. **Forensic Science International** **12 citations**
9. **Forensic Science International: Genetics** **102 citations**
10. **Forensic Science International: Genetics Supplement Series**
11. **Forensic Science Review** **1 citation** **5 citations**
12. Frontiers in Genetics
13. Genes (Basel) **11 citations**
14. Genetics in Medicine
15. Genome Biology
16. Gerontology
17. Human Genetics
18. Human Genomics
19. **International Journal of Legal Medicine** **16 citations**
20. **Investigative Genetics**
21. Journal of Clinical Microbiology
22. **Journal of Forensic Sciences** **8 citations**
23. Journal of Investigative Dermatology
24. MicroRNA
25. Molecular and Cellular Probes
26. New Genetics and Society
27. PLoS Biology
28. PLoS ONE **4 citations**
29. **Proceedings of INTERPOL Forensic Science Managers Symposium**
30. **Proceedings of International Symposium on Human Identification**
31. Proceedings of the National Academy of Sciences U.S.A.
32. Science
33. **Science & Justice** **6 citations**
34. Sensors (Basel)
35. Talanta

Most journals only have a single citation in this review

Relevant (Physical and Virtual) **Special Issues**

ELECTROPHORESIS

October 2016

Volume 37, Issue 21
Special Issue: Forensic Analysis

20 articles

November 2018

Volume 39, Issue 21
Special Issue: Novel Applications of Massively Parallel Sequencing (MPS) in Forensic Analysis

20 articles

November 2017 to December 2018

GCAT TACG GCAT genes

Virtual Issue on ***Forensic Genomics***

11 articles

September 2018 to January 2019

Virtual Issue on ***Trends and Perspectives in Forensic Genetics 2018***

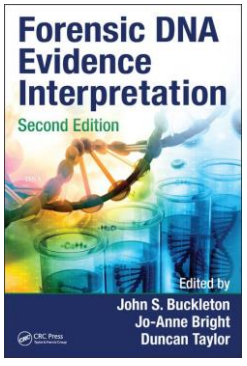
11 articles

May 2019 to August 2019

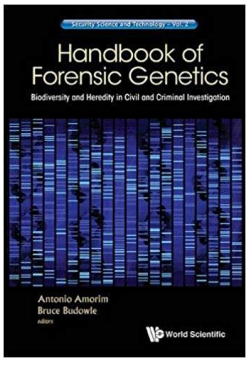
Virtual Issue on ***Cold Cases***

3 articles
on DNA topics

Recent Books and Major Conferences on Forensic DNA (2016-2019)



CRC Press, 2016



World Scientific, 2016

International Symposium on Human Identification

2016

2017

2018

2019

International Society for Forensic Genetics

Seoul, Korea
Sept **2017**

237 extended abstracts published

Prague, Czech Republic
Sept **2019**

>350 extended abstracts published

ISFG

American Academy of Forensic Sciences

ISFG Proceedings published in *Forensic Science International: Genetics Supplement Series*

Topics Covered: Forensic Biology and DNA Typing

1. **Core Loci Expansion**
2. Rapid Analysis of STR Markers
3. Investigative Genetic Genealogy
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Core Loci Expansion

- In January 2017, the FBI required expansion of the U.S. core loci from 13 short tandem repeat (STR) loci **to 20 STR loci**
- Commercial kits that amplify more than 20 STR loci have now been adopted in many countries worldwide
- These kits enable more international sharing of DNA data with increased compatibility between STR data going into national DNA databases

(original 13 CODIS core STR loci, established in November 1997):
CSF1PO FGA TH01 TPOX vWA D3S1358 D5S818 D7S820 D8S1179 D13S317 D16S539 D18S51 D21S11

(effective January 1, 2017): **D1S1656 D2S441 D2S1338 D10S1248 D12S391 D19S433 D22S1045**

See https://www.fbi.gov/services/laboratory/biometric-analysis/codis

Topics Covered: Forensic Biology and DNA Typing

1. Core Loci Expansion
2. **Rapid Analysis of STR Markers (Rapid DNA)**
3. Investigative Genetic Genealogy
4. Next-Generation Sequencing
5. DNA Mixture Interpretation and Probabilistic Genotyping Software
6. DNA Transfer and Activity Level Evaluations
7. Forensic Biology and Body Fluid Identification
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Rapid DNA

ANDE 6C
(5 samples per run)

RapidHIT ID
(1 sample per run)

- Integrated instruments (sample-to-result) can produce reliable DNA results for single-source samples in <90 minutes
- Size-based analysis of 15 to 23 autosomal STR markers
- Success rates are typically >80%
  https://www.nist.gov/sites/default/files/documents/2018/11/14/3_romsos.pdf
  https://promega.media/-/media/files/products-and-services/genetic-identity/ishi-29-oral-abstracts/romsos.pdf
- Reagent costs are approximately 10 times conventional testing (≈$300 per sample)
- Rapid DNA Act of 2017 signed into U.S. law on August 18, 2017
- 13 published evaluation or validation studies

Rapid DNA Instrument Validation and Evaluation Studies (1)

Publication	Instrument	STR Primer Set	Tests Performed and Success Rates Reported
Turingan et al. 2016	DNAscan/ANDE 4C	PowerPlex 16	Evaluation of swabs from drinking containers, blood and buccal cells on FTA paper, blood and buccal cells on untreated paper, dried blood on ceramic tile, dried blood and dried semen on clothing, chewing gum, cigarette butt, cellphones, and bones to assess reproducibility, accuracy/ concordance, sensitivity, precision, resolution, and peak height ratios
Della Manna et al. 2016	DNAscan/ANDE 4C	PowerPlex 16	SWGDAM developmental validation (across 8 laboratories, >2300 swabs): species specificity, sensitivity, stability, inhibitors, reproducibility, mixtures, precision, accuracy, and concordance; success rate (1362 samples with 13 CODIS core loci) = 84% (auto) → 91% (manual)
Date-Chong et al. 2016	RapidHIT 200	GlobalFiler Express	Evaluation of 34 known buccal samples and 23 negative controls; success rate = 50% (auto)
Moreno et al. 2017	DNAscan/ANDE 4C	PowerPlex 16	SWGDAM internal validation: contamination assessment, consistency and reliability, sizing precision, peak height ratio determination, noise and average peak height assessment, stutter percent calculation, sensitivity and interpretation threshold calculations, and stability studies; success rate (193 samples) = 75% (auto) for 13 CODIS core loci with no incorrect calls
Wiley et al. 2017	RapidHIT ID	GlobalFiler Express	SWGDAM internal validation: contamination assessment, reliability, swab re-analysis, sensitivity, inhibitor, mixture, swab stability, precision, concordance and reliability, swab substrate, standards/controls, and bridge studies; success rate (50 samples) = 72% (auto) → 90% (manual)
Salceda et al. 2017	RapidHIT ID	GlobalFiler Express	SWGDAM developmental validation: thermal cycling parameters, mock inhibition, species specificity, sensitivity, concordance and carryover, swab retrieval and re-extraction, repeatability and reproducibility, electrophoresis sizing accuracy, stutter calculations and precision studies

Rapid DNA Instrument Validation and Evaluation Studies (2)

Publication	Instrument	STR Primer Set	Tests Performed and Success Rates Reported
Boiso et al. 2017a, 2017b	RapidHIT 200	NGM SElect Express	28 runs performed in total (with 7 samples each); problems encountered with hardware, software, and consumables; found the system was not suitable for crime scene samples in its current design; success rate (155 samples) = 77% gave complete DNA profiles with samples involving (1, 2, or 5) µL blood spotted on swabs
Buscaino et al. 2018	RapidHIT ID	GlobalFiler Express and NGM SElect Express	Evaluation of thermal cycling parameters, sensitivity, carryover contamination risks, repeatability and reproducibility, mixtures, and mock crime scene samples
Amick & Swiger 2019	RapidHIT ID	GlobalFiler Express	SWGDAM internal validation: known and database-type samples, reproducibility, precision, sensitivity, stochastic effects, mixtures, contamination assessment, and concordance studies
Carney et al. 2019	ANDE 6C	FlexPlex (6-dye, 27plex STR assay)	SWGDAM developmental validation (across 6 labs, 2045 swabs, 13 instruments): species specificity, limit of detection, stability, inhibitors, reproducibility, reference material, mixtures, precision, concordance, signal strength, peak height ratio, stutter, non-template addition, resolution, and contamination assessment; first-pass success rate (1338 samples with 20 CODIS core loci) = 92%; successfully interpreted >2000 samples with over 99.99% concordant alleles; data package led to receiving NDIS approval in June 2018
Shackleton et al. 2019a	RapidHIT 200	NGM SElect Express	Development studies that included process optimization, sensitivity, repeatability, contamination checks, inhibition, swab age, concordance, and overall performance; success rate (124 samples) = 84.5% gave a full profile
Shackleton et al. 2019b	RapidHIT 200	NGM SElect Express	Protocol adjustments that extended the overall run times were made to enhance slightly sensitivity with mock crime scene samples (dilutions of blood and cell line DNA)

Rapid DNA in Police Stations?

https://www.nytimes.com/2019/01/21/science/dna-crime-gene-technology.html

The New York Times

January 21, 2019

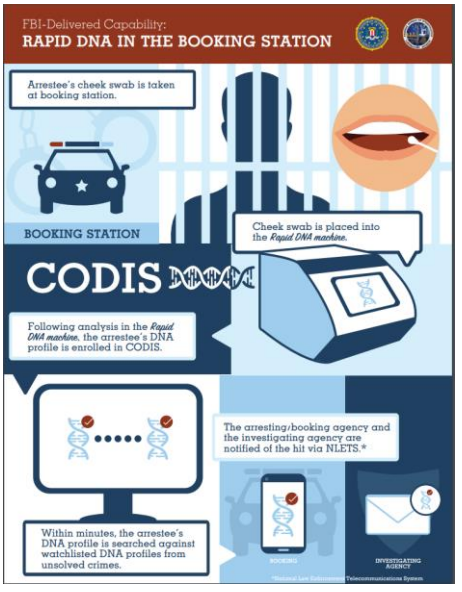
Coming Soon to a Police Station Near You: The DNA 'Magic Box'

With Rapid DNA machines, genetic fingerprinting could become as routine as the old-fashioned kind. But forensic experts see a potential for misuse.

A Rapid DNA machine can reveal whether an individual's DNA matches genetic evidence collected from a local crime. Mark Makela for The New York Times

By Heather Murphy

Jan. 21, 2019



https://www.fbi.gov/services/laboratory/biometric-analysis/codis/rapid-dna

The capability exists but how much rapid DNA will be used remains to be seen…

Non-CODIS Rapid DNA Considerations and Best Practices for Law Enforcement Use

https://www.fbi.gov/file-repository/non-codis-rapid-dna-best-practices-092419.pdf/view

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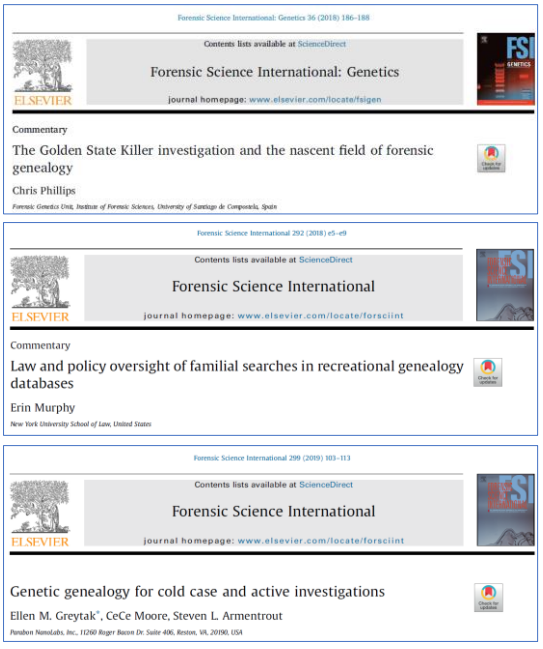

Forensic Science International: Genetics 36 (2018) 186–188
Contents lists available at ScienceDirect
Forensic Science International: Genetics
journal homepage: www.elsevier.com/locate/fsigen
Commentary
The Golden State Killer investigation and the nascent field of forensic genealogy
Chris Phillips
Forensic Genetics Unit, Institute of Forensic Sciences, University of Santiago de Compostela, Spain

Forensic Science International 292 (2018) e5–e9
Contents lists available at ScienceDirect
Forensic Science International
journal homepage: www.elsevier.com/locate/forsciint
Commentary
Law and policy oversight of familial searches in recreational genealogy databases
Erin Murphy
New York University School of Law, United States

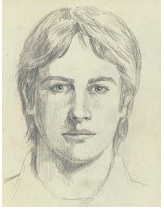
Forensic Science International 299 (2019) 103–113
Contents lists available at ScienceDirect
Forensic Science International
journal homepage: www.elsevier.com/locate/forsciint
Genetic genealogy for cold case and active investigations
Ellen M. Greytak*, CeCe Moore, Steven L. Armentrout
Parabon NanoLabs, Inc., 11260 Roger Bacon Dr. Suite 406, Reston, VA, 20190, USA

Investigative Genetic Genealogy

- Arrest of the alleged Golden State Killer Joseph DeAngelo in April 2018 opened the doors to solving cold cases using genetic genealogy (non-law-enforcement) DNA databases
- Has raised genetic privacy concerns
- Subject of active discussions at recent forensic DNA and genetic genealogy conferences

https://time.com/collection/100-most-influential-people-2019/

See also J.M. Butler (2019) https://www.nist.gov/blogs/taking-measure/national-dna-day-and-birth-investigative-genetic-genealogy



Sketch of the Original Night Stalker (also known as Visalia Ransacker and East Area Rapist who is thought to have operated from 1974 to 1986)

Joseph James DeAngelo (April 24, 2018 mugshot)

Golden State Killer Case

- **Crimes: 13+ murdered, 50+ raped, 120+ burgled** (many linked by DNA evidence over the years)
- DNA from a **crime scene sample** (sexual assault kit) was examined with **genetic genealogy DNA markers**
- Data obtained was uploaded to a **genetic genealogy database** (GEDmatch) to locate potential distant relatives of the perpetrator
- **Family trees were created** from possible links and **detective work** reduced number of possible suspects
- **Confirmation DNA sample** was collected from a door handle and later a tissue in the garbage to verify that the **suspect's DNA profile matched the crime scene DNA profile**
- DeAngelo (age 72) was arrested at his home outside of Sacramento, CA on April 24, 2018 and is awaiting trial

Sources: numerous news articles and Wikipedia

>60 Cold Cases in the United States Have Been Solved with Genetic Genealogy since April 2018

POLICY FORUM *Science* (2018) 360:1078-1079

GENETICS AND PRIVACY

Genealogy databases and the future of criminal investigation

The police can access your online family-tree research—and use it to investigate your relatives

By Natalie Ram,[1] Christi J. Guerrini,[2] Amy L. McGuire[2]

The 24 April 2018 arrest of Joseph James DeAngelo as the alleged Golden State

tigators used genealogical DNA matches to develop and pursue a suspect in the Golden State Killer case itself. A year before investigators zeroed in on DeAngelo, they subpoenaed another genetic testing company for

PLOS BIOLOGY (2018) 16(10): e2006906

Of 1587 members of the general public questioned in May 2018, **91% said yes**

PERSPECTIVE

Should police have access to genetic genealogy databases? Capturing the Golden State Killer and other criminals using a controversial new forensic technique

Christi J. Guerrini, Jill O. Robinson, Devan Petersen, Amy L. McGuire

Center for Medical Ethics and Health Policy, Baylor College of Medicine, Houston, Texas, United States of America

Forensic Science International 301 (2019) 107–117

Contents lists available at ScienceDirect

Forensic Science International

journal homepage: www.elsevier.com/locate/forsciint

ELSEVIER

Using genetic genealogy databases in missing persons cases and to develop suspect leads in violent crimes

Debbie Kennett

Research Department of Genetics, Evolution and Environment, University College London, Gower Street, London WC1E 6BT, United Kingdom

SCIENCE The Atlantic

The Messy Consequences of the Golden State Killer Case

Tools meant to reunite families are now being "used essentially to get families to put their members in jail."

SARAH ZHANG OCT 1, 2019

https://www.theatlantic.com/science/archive/2019/10/genetic-genealogy-dna-database-criminal-investigations/599005/

DOJ Policy on Investigative Genetic Genealogy

Released September 24, 2019 *(goes into effect November 1, 2019)*

THE UNITED STATES DEPARTMENT of JUSTICE

UNITED STATES DEPARTMENT OF JUSTICE
INTERIM POLICY
FORENSIC GENETIC GENEALOGICAL DNA ANALYSIS AND SEARCHING

https://www.justice.gov/olp/page/1204386/download

This interim policy applies to:

1) all criminal investigations run by the Department of Justice (DOJ)
2) any criminal investigation in which DOJ provides funding
3) any criminal investigation in which DOJ employees or contractors conduct genealogical research
4) any federal agency or any unit of state, local, or tribal government that receives DOJ grant award funding

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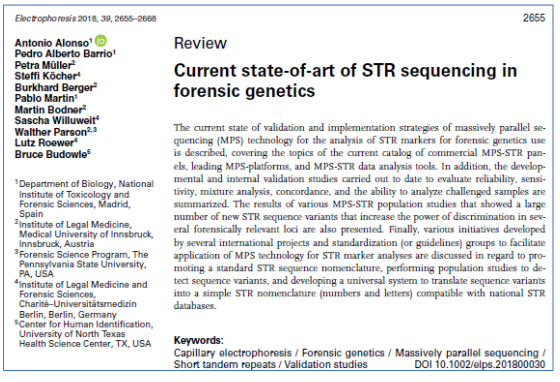
Next-Generation DNA Sequencing



Electrophoresis 2018, *39*, 2655–2668 2655

Review

Current state-of-art of STR sequencing in forensic genetics

Antonio Alonso[1], Pedro Alberto Barrio[1], Petra Müller[2], Steffi Köcher[4], Burkhard Berger[2], Pablo Martin[1], Martin Bodner[2], Sascha Willuweit[4], Walther Parson[2,3], Lutz Roewer[4], Bruce Budowle[5]

[1] Department of Biology, National Institute of Toxicology and Forensic Sciences, Madrid, Spain
[2] Institute of Legal Medicine, Medical University of Innsbruck, Innsbruck, Austria
[3] Forensic Science Program, The Pennsylvania State University, PA, USA
[4] Institute of Legal Medicine and Forensic Sciences, Charité–Universitätsmedizin Berlin, Berlin, Germany
[5] Center for Human Identification, University of North Texas Health Science Center, TX, USA

The current state of validation and implementation strategies of massively parallel sequencing (MPS) technology for the analysis of STR markers for forensic genetics use is described, covering the topics of the current catalog of commercial MPS-STR panels, leading MPS-platforms, and MPS-STR data analysis tools. In addition, the developmental and internal validation studies carried out to date to evaluate reliability, sensitivity, mixture analysis, concordance, and the ability to analyze challenged samples are summarized. The results of various MPS-STR population studies that showed a large number of new STR sequence variants that increase the power of discrimination in several forensically relevant loci are also presented. Finally, various initiatives developed by several international projects and standardization (or guidelines) groups to facilitate application of MPS technology for STR marker analyses are discussed in regard to promoting a standard STR sequence nomenclature, performing population studies to detect sequence variants, and developing a universal system to translate sequence variants into a simple STR nomenclature (numbers and letters) compatible with national STR databases.

Keywords:
Capillary electrophoresis / Forensic genetics / Massively parallel sequencing / Short tandem repeats / Validation studies DOI 10.1002/elps.201800030

Forensic Science International: Genetics (May 2016) 22:54-63

Massively parallel sequencing of forensic STRs: Considerations of the DNA commission of the International Society for Forensic Genetics (ISFG) on minimal nomenclature requirements

Walther Parson[a,b,*], David Ballard[c], Bruce Budowle[d,e], John M. Butler[f], Katherine B. Gettings[f], Peter Gill[g,h], Leonor Gusmão[i,j,k], Douglas R. Hares[l], Jodi A. Irwin[l], Jonathan L. King[d], Peter de Knijff[m], Niels Morling[n], Mechthild Prinz[o], Peter M. Schneider[p], Christophe Van Neste[q], Sascha Willuweit[r], Christopher Phillips[s]

[a] Institute of Legal Medicine, Innsbruck Medical University, Innsbruck, Austria
[b] Forensic Science Program, The Pennsylvania State University, University Park, PA, USA
[c] Faculty of Life Sciences, King's College, London, UK
[d] Institute of Applied Genetics, Department of Molecular and Medical Genetics, University of North Texas Health Science Center, Fort Worth, TX, USA
[e] Center of Excellence in Genomic Medicine Research (CEGMR), King Abdulaziz University, Jeddah, Saudi Arabia
[f] National Institute of Standards and Technology, Gaithersburg, MD, USA

- **Additional markers** can be run simultaneously (≈10 times as many as current CE systems) with higher information content
  - May enable additional capabilities (e.g., phenotyping)
  - Privacy concerns with additional genomic information
- Involves **more sample preparation** steps and requires **more extensive data analysis**
  - Expensive per run although cost per marker is lower
  - STR allele nomenclature challenges to keep backwards compatibility
  - Data handling and storage issues
- Agreeing upon and implementing a standard STR allele nomenclature is critical for future progress

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DNA Mixture Interpretation

- Deciphering the various components present in a mixture and assigning an appropriate weight to the evidence can be challenging
- Improper use of DNA mixture interpretation approaches led to closure of several U.S. forensic DNA laboratories in 2015 and 2016
- The past few years have seen an increase in the use of probabilistic genotyping software (PGS) to assist DNA mixture interpretation
  - PGS systems use either (1) **"discrete"** (sometimes called "semi-continuous") models that use the presence or absence of peaks along with probabilities of allele drop-out or drop-in or (2) **"continuous"** (sometimes called "fully-continuous") models that take peak heights into account as well as the presence or absence of peaks along with probabilities of allele drop-out or drop-in

Probabilistic Genotyping Software (PGS) – as of July 2019

	Program Name	Type (Model)	Creator(s)	Availability	References
1	CEESIt	Continuous	Catherine Grgicak	Open-source software: https://lftdi.camden.rutgers.edu/	Swaminathan et al. 2016
2	DNAmixtures	Continuous	Therese Graversen	Open-source software: http://dnamixtures.r-forge.r-project.org/	Cowell et al. 2015
3	DNA Mixture Solution	Continuous	Charles Brenner	Commercial product: http://dna-view.com/dnaview.htm	Brenner 2015
4	eDNA	Discrete & Continuous	Available through subscription service: Bullet (Semi-continuous; uses LRmix math) and BulletProof (Fully-continuous; uses EuroForMix math) http://ednalims.com/probabilistic-genotyping/		
5	EuroForMix	Continuous	Øyvind Bleka, Peter Gill	Open-source software: http://www.euroformix.com/	Bleka et al. 2016
6	**FST**	Discrete	NYC OCME	Proprietary to NYC OCME Department of Forensic Biology	Mitchell et al. 2012
7	GenoProof Mixture 3	Continuous	Frank Götz	Commercial product: https://www.qualitype.de	Götz et al. 2017
8	Kongoh	Continuous	Sho Manabe	Open-source software: https://github.com/manabe0322/Kongoh/releases	Manabe et al. 2017
9	**Lab Retriever**	Discrete	David Balding; maintained by Norah Rudin and colleagues	Open-source software: https://scieg.org/lab-retriever/	Inman et al. 2015
10	likeLTD	Discrete & Continuous	David Balding	Open-source software: https://sites.google.com/site/baldingstatisticalgenetics/software/likeltd-r-forensic-dna-r-code	Balding 2013
11	LiRa/ LiRa-HT	Discrete/ Continuous	Roberto Puch-Solis	Proprietary to LGC (now Eurofins)	Puch-Solis & Clayton 2014
12	LRmix; LRmix studio	Discrete/ Discrete	Hinda Haned, Peter Gill; Jeroen de Jong	Open-source software: https://sites.google.com/site/forensicdnastatistics/PCR-simulation/lrmix; http://lrmixstudio.org/	Gill & Haned 2013
13	MaSTR	Continuous	Teresa Snyder-Leiby	Commercial product: https://softgenetics.com/MaSTR.php	Adamowicz et al. 2018
14	**STRmix**	Continuous	Duncan Taylor, Jo-Anne Bright, John Buckleton	Commercial product: https://strmix.esr.cri.nz/	Taylor et al. 2013
15	**TrueAllele**	Continuous	Mark Perlin	Commercial product: http://www.cybgen.com	Perlin et al. 2011

Topics Covered: Forensic Biology and DNA Typing

1. Core Loci Expansion
2. Rapid Analysis of STR Markers
3. Investigative Genetic Genealogy
4. Next-Generation Sequencing
5. DNA Mixture Interpretation and Probabilistic Genotyping Software
6. **DNA Transfer and Activity Level Evaluations**
7. Forensic Biology and Body Fluid Identification
8. DNA Phenotyping
9. Privacy and Ethical Issues
10. Guidance Documents (SWGDAM, OSAC, ASB, ISO, ENFSI, UK FS Regulator)
11. Contamination Avoidance and DNA Success Rates
12. Recent Special Issues and Review Articles of Note

DNA Transfer and Persistence and Activity Level Propositions

The importance of DNA transfer and activity propositions has increased in recent years with use of highly sensitive DNA testing methods. There is a growing body of literature on this topic in the past three years due to the desire to address not only the source of the DNA but how the DNA got there. Several reviews highlight ***how little we know with certainty about how DNA transfers*** from the donor, the range of variables that affect transfer and persistence, the value of activity propositions, and a proposal for a more systematic approach to data collection.

- **Promotion of activity propositions** (2018) Taylor, D., Kokshoorn, B. and Biedermann, A. Evaluation of forensic genetics findings given activity level propositions: A review. *Forensic Sci. Int. Genet.* 36: 34-49.
- **Mechanisms on how DNA is transferred from donor** (2019) Burrill, J., Daniel, B., and Frascione, N. A review of trace "touch DNA" deposits: variability factors and an exploration of cellular composition. *Forensic Sci. Int. Genet.* 39: 8-18.
- **Comprehensive review of all aspects of transfer** (2019) van Oorschot, R.A.H., Szkuta, B., Meakin, G.E., Kokshoorn, B. and Goray, M. DNA transfer in forensic science: a review. *Forensic Sci. Int. Genet.* 38: 140-166.
- **Scheme for more systematic approach to data collection** (2019) Gosch, A. and Courts, C. On DNA transfer: the lack and difficulty of systematic research and how to do it better. *Forensic Sci. Int. Genet.* 40: 24-36.

The overall takeaway from this literature is that sub-source LR results in isolation cannot automatically be assumed to be connected with the crime

Topics Covered: Forensic Biology and DNA Typing

1. Core Loci Expansion
2. Rapid Analysis of STR Markers
3. Investigative Genetic Genealogy
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12. Recent Special Issues and Review Articles of Note

Organizations Providing Guidance Documents on Forensic DNA

35 issued in the past three years

SWGDAM — Scientific Working Group DNA Analysis Methods

OSAC — Organization of Scientific Area Committees for Forensic Science

ASB — AAFS Standards Board

The United States Department of Justice

ISO

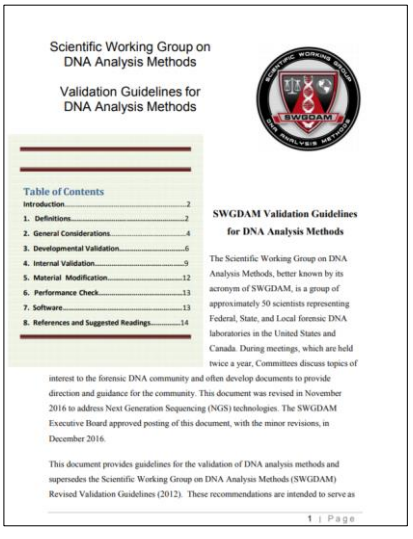
ENFSI — European Network of Forensic Science Institutes

ISFG — International Society for Forensic Genetics

Forensic Science Regulator — Overseeing Quality

SWGDAM Documents

Validation Guidelines



Scientific Working Group on DNA Analysis Methods

Validation Guidelines for DNA Analysis Methods

Table of Contents

Introduction....2
1. Definitions....2
2. General Considerations....4
3. Developmental Validation....6
4. Internal Validation....9
5. Material Modification....12
6. Performance Check....13
7. Software....13
8. References and Suggested Readings....14

SWGDAM Validation Guidelines for DNA Analysis Methods

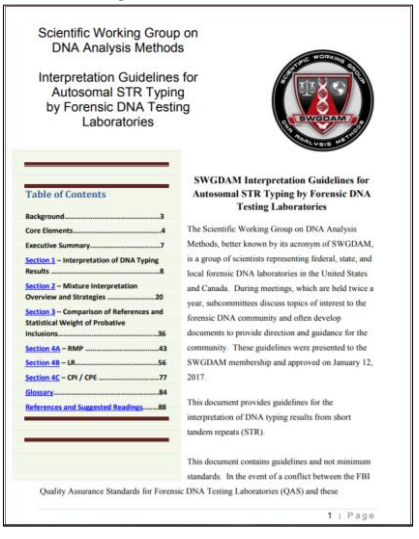
The Scientific Working Group on DNA Analysis Methods, better known by its acronym of SWGDAM, is a group of approximately 50 scientists representing Federal, State, and Local forensic DNA laboratories in the United States and Canada. During meetings, which are held twice a year, Committees discuss topics of interest to the forensic DNA community and often develop documents to provide direction and guidance for the community. This document was revised in November 2016 to address Next Generation Sequencing (NGS) technologies. The SWGDAM Executive Board approved posting of this document, with the minor revisions, in December 2016.

This document provides guidelines for the validation of DNA analysis methods and supersedes the Scientific Working Group on DNA Analysis Methods (SWGDAM) Revised Validation Guidelines (2012). These recommendations are intended to serve as

1 | Page

December 2016

Interpretation Guidelines



Scientific Working Group on DNA Analysis Methods

Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories

Table of Contents

Background....3
Core Elements....4
Executive Summary....7
Section 1 – Interpretation of DNA Typing Results....8
Section 2 – Mixture Interpretation Overview and Strategies....20
Section 3 – Comparison of References and Statistical Weight of Probative Inclusions....36
Section 4A – RMP....43
Section 4B – LR....56
Section 4C – CPI / CPE....77
Glossary....84
References and Suggested Readings....88

SWGDAM Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories

The Scientific Working Group on DNA Analysis Methods, better known by its acronym of SWGDAM, is a group of scientists representing federal, state, and local forensic DNA laboratories in the United States and Canada. During meetings, which are held twice a year, subcommittees discuss topics of interest to the forensic DNA community and often develop documents to provide direction and guidance for the community. These guidelines were presented to the SWGDAM membership and approved on January 12, 2017.

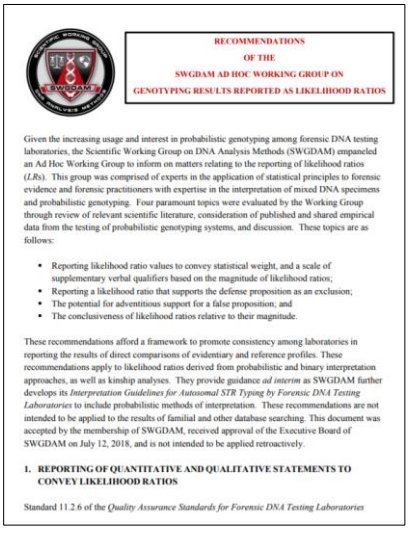
This document provides guidelines for the interpretation of DNA typing results from short tandem repeats (STR).

This document contains guidelines and not minimum standards. In the event of a conflict between the FBI Quality Assurance Standards for Forensic DNA Testing Laboratories (QAS) and these

1 | Page

January 2017

LR Verbal Equivalents



RECOMMENDATIONS OF THE SWGDAM AD HOC WORKING GROUP ON GENOTYPING RESULTS REPORTED AS LIKELIHOOD RATIOS

Given the increasing usage and interest in probabilistic genotyping among forensic DNA testing laboratories, the Scientific Working Group on DNA Analysis Methods (SWGDAM) empaneled an Ad Hoc Working Group to inform on matters relating to the reporting of likelihood ratios (*LRs*). This group was comprised of experts in the application of statistical principles to forensic evidence and forensic practitioners with expertise in the interpretation of mixed DNA specimens and probabilistic genotyping. Four paramount topics were evaluated by the Working Group through review of relevant scientific literature, consideration of published and shared empirical data from the testing of probabilistic genotyping systems, and discussion. These topics are as follows:

- Reporting likelihood ratio values to convey statistical weight, and a scale of supplementary verbal qualifiers based on the magnitude of likelihood ratios;
- Reporting a likelihood ratio that supports the defense proposition as an exclusion;
- The potential for adventitious support for a false proposition; and
- The conclusiveness of likelihood ratios relative to their magnitude.

These recommendations afford a framework to promote consistency among laboratories in reporting the results of direct comparisons of evidentiary and reference profiles. These recommendations apply to likelihood ratios derived from probabilistic and binary interpretation approaches, as well as kinship analyses. They provide guidance *ad interim* as SWGDAM further develops its *Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories* to include probabilistic methods of interpretation. These recommendations are not intended to be applied to the results of familial and other database searching. This document was accepted by the membership of SWGDAM, received approval of the Executive Board of SWGDAM on July 12, 2018, and is not intended to be applied retroactively.

1. REPORTING OF QUANTITATIVE AND QUALITATIVE STATEMENTS TO CONVEY LIKELIHOOD RATIOS

Standard 11.2.6 of the *Quality Assurance Standards for Forensic DNA Testing Laboratories*

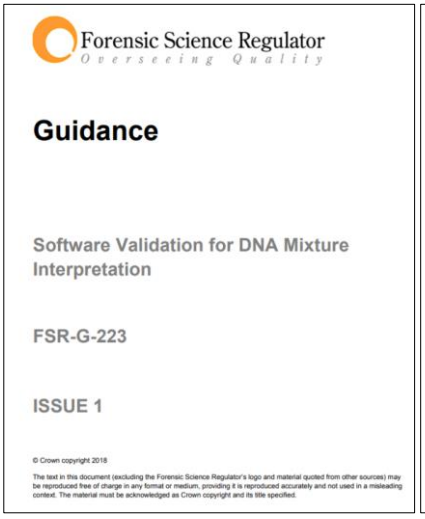
July 2018

UK Forensic Science Regulator

Codes of Practice and Conduct

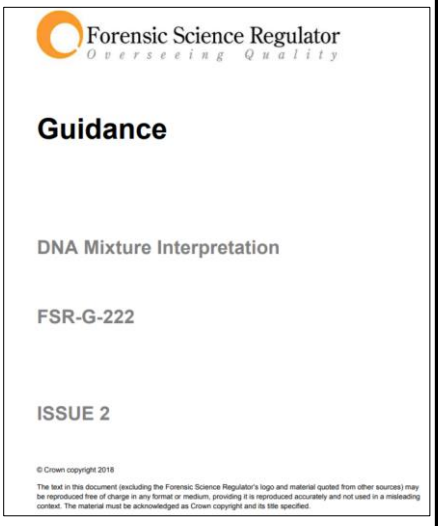
October 2017

PGS Software Validation



September 2018

DNA Mixture Interpretation



October 2018

Guidance Documents Related to Forensic DNA (1)

Organization	Publication Date	Title
SWGDAM	December 2016	Recommendations for the Efficient DNA Processing of Sexual Assault Evidence Kits https://docs.wixstatic.com/ugd/4344b0_4daf2bb5512b4e2582f895c4a133a0ed.pdf
SWGDAM	December 2016	Validation Guidelines for DNA Analysis Methods https://docs.wixstatic.com/ugd/4344b0_813b241e8944497e99b9c45b163b76bd.pdf
SWGDAM	January 2017	Contamination Prevention and Detection Guidelines for Forensic DNA Laboratories https://docs.wixstatic.com/ugd/4344b0_c4d4dbba84f1400a98eaa2e48f2bf291.pdf
SWGDAM	January 2017	Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories https://docs.wixstatic.com/ugd/4344b0_50e2749756a242528e6285a5bb478f4c.pdf
SWGDAM	July 2018	Recommendations of the SWGDAM Ad Hoc Working Group on Genotyping Results Reported as Likelihood Ratios https://docs.wixstatic.com/ugd/4344b0_dd5221694d1448588dcd0937738c9e46.pdf
SWGDAM	April 2019	Addendum to "SWGDAM Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories" to Address Next Generation Sequencing https://docs.wixstatic.com/ugd/4344b0_91f2b89538844575a9f51867def7be85.pdf
SWGDAM	April 2019	Interpretation Guidelines for Mitochondrial DNA Analysis by Forensic DNA Testing Laboratories https://docs.wixstatic.com/ugd/4344b0_f61de6abf3b94c52b28139bff600ae98.pdf
SWGDAM	January 2018	Quality Assurance Standards for Forensic DNA Testing Laboratories (draft) https://docs.wixstatic.com/ugd/4344b0_d4c50d6204b240d3ab23e388b5f6591a.pdf
SWGDAM	February 2019	FBI Quality Assurance Standards Audit for Forensic DNA Testing Laboratories (draft) https://docs.wixstatic.com/ugd/4344b0_7b03780db7244a5b9a93b3bdd59345b5.pdf
SWGDAM	February 2019	Quality Assurance Standards for DNA Databasing Laboratories (draft) https://docs.wixstatic.com/ugd/4344b0_bf68274461f3425888adce9399115099.pdf
SWGDAM	February 2019	FBI Quality Assurance Standards Audit for DNA Databasing Laboratories (draft) https://docs.wixstatic.com/ugd/4344b0_990aee2783af4a82b4d21358e0bd1c53.pdf

Guidance Documents Related to Forensic DNA (2)

Organization	Publication Date	Title
US DOJ	September 2018	Department of Justice Uniform Language for Testimony and Reports for Forensic Autosomal DNA Examinations Using Probabilistic Genotyping Systems https://www.justice.gov/olp/page/file/1095961/download
US DOJ	September 2018	Department of Justice Uniform Language for Testimony and Reports for Forensic Mitochondrial DNA Examinations https://www.justice.gov/olp/page/file/1095966/download
US DOJ	September 2018	Department of Justice Uniform Language for Testimony and Reports for Forensic Y-STR Data Examinations https://www.justice.gov/olp/page/file/1095976/download
US DOJ	September 2018	Department of Justice Uniform Language for Testimony and Reports for Forensic Serological Examinations https://www.justice.gov/olp/page/file/1095971/download

Released September 24, 2019 ***(goes into effect November 1, 2019)***

THE UNITED STATES DEPARTMENT of JUSTICE

UNITED STATES DEPARTMENT OF JUSTICE
INTERIM POLICY
FORENSIC GENETIC GENEALOGICAL DNA ANALYSIS AND SEARCHING

https://www.justice.gov/olp/page/file/1204386/download

Guidance Documents Related to Forensic DNA (3)

Organization	Publication Date	Title
ASB	September 2018	Standard for Validation Studies of DNA Mixtures, and Development and Verification of a Laboratory's Mixture Interpretation Protocol https://asb.aafs.org/wp-content/uploads/2018/09/020_Std_e1.pdf
ISO/TC 272	February 2016	ISO 18385:2016 Minimizing the Risk of Human Contamination in Products Used to Collect, Store and Analyze Biological Material for Forensic Purposes – Requirements https://www.iso.org/standard/62341.html?browse=tc
ISO/TC 272	August 2018	ISO 21043-1:2018 Forensic Sciences – Part 1: Terms and Definitions https://www.iso.org/standard/69732.html?browse=tc
ISO/TC 272	August 2018	ISO 21043-2:2018 Forensic Sciences – Part 2: Recognition, Recording, Collecting, Transport and Storage of Items https://www.iso.org/standard/72041.html?browse=tc
ISO/CASCO	November 2017	ISO/IEC 17025:2017 General Requirements for the Competence of Testing and Calibration Laboratories https://www.iso.org/standard/66912.html

Guidance Documents Related to Forensic DNA (4)

Organization	Publication Date	Title
ISFG DNA Commission	January 2016	Massively parallel sequencing of forensic STRs: Considerations…on minimal nomenclature requirements (Parson et al. 2016) https://www.isfg.org/files/d5ccd549ee232596c75ad8a0b435190e7dba3035.parson2016_str.recommendations.pdf
ISFG DNA Commission	June 2016	Recommendations…on quality control of autosomal short tandem repeat allele frequency databasing (STRidER) (Bodner et al. 2016) https://www.isfg.org/files/db9864824b44997f1014a62a0321f0d25ef6cf98.bodner2016_strider.pdf
ISFG DNA Commission	September 2016	Recommendations on the validation of software programs performing biostatistical calculations for forensic genetic applications (Coble et al. 2016) https://www.isfg.org/files/225be64835df624d1ddac70b95a2e7354f916fbb.coble_software_validation_fsigen2016.pdf
ISFG DNA Commission	May 2017	Guidelines on the use of X-STRs in kinship analysis (Tillmar et al. 2017) https://www.isfg.org/files/eea3394d1595b83aeb59e093725518fb94691e78.tillmar2017_x.str.recommendations.pdf
ISFG DNA Commission	July 2018	Assessing the value of forensic biological evidence – guidelines highlighting the importance of propositions. Part I: evaluation of DNA profiling comparisons given (sub-) source propositions (Gill et al. 2018)

Guidance Documents Related to Forensic DNA (5)

Organization	Publication Date	Title
UKFSR	October 2017	Codes of Practice and Conduct for Forensic Science Providers and Practitioners in the Criminal Justice System (Issue 4) https://www.gov.uk/government/publications/forensic-science-providers-codes-of-practice-and-conduct-2017
UKFSR	March 2016	Validation: Use of Casework Material (FSR-P-300) https://www.gov.uk/government/publications/protocol-using-casework-material-for-validation-purposes
UKFSR	July 2016	Sexual Assault Referral Centres and Custodial Facilities: DNA Anti-Contamination https://www.gov.uk/government/publications/sexual-assault-referral-centres-and-custodial-facilities-dna-anti-contamination
UKFSR	July 2016	Crime Scene DNA: Anti-Contamination Guidance https://www.gov.uk/government/publications/crime-scene-dna-anti-contamination-guidance
UKFSR	September 2018	Software Validation for DNA Mixture Interpretation (FSR-G-223) https://www.gov.uk/government/publications/software-validation-for-dna-mixture-interpretation-fsr-g-223
UKFSR	October 2018	DNA Mixture Interpretation (FSR-G-222) https://www.gov.uk/government/publications/dna-mixture-interpretation-fsr-g-222

Guidance Documents Related to Forensic DNA (6)

Organization	Publication Date	Title
ENFSI	May 2017	Best Practice Manual for the Internal Validation of Probabilistic Software to Undertake DNA Mixture Interpretation http://enfsi.eu/wp-content/uploads/2017/09/Best-Practice-Manual-for-the-internal-validation-of-probabilistic-software-to-undertake-DNA-mixture-interpretation-v1.docx.pdf
ENFSI DNA WG	April 2017	DNA Contamination Prevention Guidelines http://enfsi.eu/wp-content/uploads/2017/09/DNA-contamination-prevention-guidelines-v2.pdf
ENFSI DNA WG	April 2017	DNA Database Management Review and Recommendations http://enfsi.eu/wp-content/uploads/2017/09/DNA-databasemanagement-review-and-recommendations-april-2017.pdf

OSAC Organization of Scientific Area Committees for Forensic Science

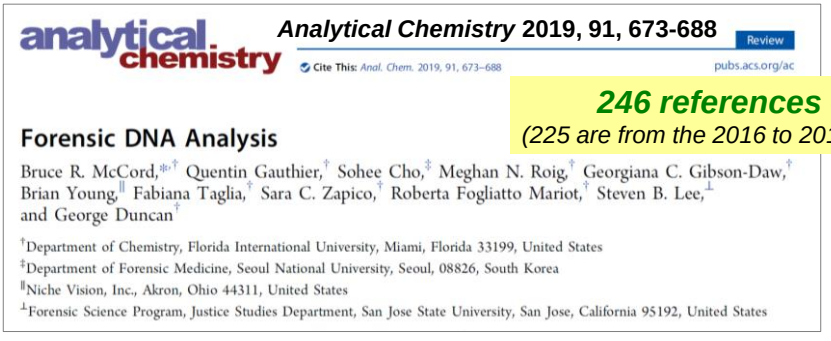
Biological Methods Subcommittee
13 work products to SDO
14 additional documents under development

Wildlife Forensics Subcommittee
8 work products to SDO
9 additional documents under development

Biological Data Interpretation and Reporting Subcommittee
5 work products to SDO
14 additional documents under development

https://www.nist.gov/topics/organization-scientific-area-committees-forensic-science

Another Recent Review Article on Forensic DNA



246 references cited
(225 are from the 2016 to 2018 time frame)

Topics covered:

- Forensic serology
- Chemical & spectroscopic methods
- Body fluid identification via RNA typing
- Proteomic body fluid identification
- Epigenetics
- DNA extraction & sample recovery
- Genotyping methods using STRs
- Mixtures and probabilistic genotyping
- Estimating the number of contributors
- Y-STRs and X-STRs; Y-SNPs and X-SNPs
- SNPs; Insertion/Deletions; Mitochondrial DNA
- Ancient DNA, bones, and teeth
- Improving DNA extractions from teeth and bone
- Nonhuman DNA; Wildlife forensics
- Drug sourcing
- Massively parallel sequencing
- The microbiome as a source of DNA
- Postmortem interval

Thank you for your attention!

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http://strbase.nist.gov/NISTpub.htm

Any questions?