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MVPs of Forensic DNA: Examining Some of the Most Valuable Publications in the Field

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Experts Need Up-to-Date Knowledge in Their Field

Dr. Gillian Tully, the UK Forensic Science Regulator at the time, stated in her 2017 annual report:

“It is a clear expectation of the courts that expert evidence is presented by people who are indeed experts in their field. This necessitates **an up-to-date knowledge of developments in the relevant field**, which in turn necessitates access to scientific literature and sufficient time **to ensure that each expert has the current relevant knowledge that they need.**”

<https://www.gov.uk/government/publications/forensic-science-regulator-annual-report-2017>

(published January 19, 2018, quote from page 10)

Development of Expert Knowledge

DNA analysts benefit from at least three different levels of expert knowledge:

- 1. Education in basic science** covering biochemistry, biology, chemistry, genetics, molecular biology, population genetics, and statistics
- 2. Training in forensic science** and specific methods and protocols used in their laboratory to develop competency needed to perform casework
- 3. Continued education and professional development** to keep up-to-date as the field evolves and new methods become available

#3 involves knowing the ever-growing scientific literature

A Constantly Growing DNA Literature...



INTERPOL Review 2016-2019

Discussed 235 references
from 35 journals
across 12 categories

Review of forensic biology and DNA publications from 2016 to 2019 (12 categories):

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 (SWGDM, OSAC, ASB, ENFSI, UK Regulator)
11. Contamination Avoidance and DNA Success Rates
12. Recent Special Issues and Review Articles of Note

INTERPOL Review 2019-2022 (in progress) = **1884+ articles** across 32 categories

- Scopus and Web of Science searches with “forensic DNA” and then removing duplicates and non-English articles
- Manual searches of non-indexed journals such as FSI Reports, WIREs Forensic Science

Some Improvements That Could Be Beneficial to the Forensic DNA Community

An AAFS 2021 workshop was intended as a start

From deliberations and discussions of NIST team members and Resource Group in connection with the Scientific Foundation Review on DNA Mixture Interpretation (see Appendix 2 in NISTIR 8351-DRAFT published in June 2021)

1. **An agreed upon, defined body of knowledge for DNA analysis and interpretation** and a means to update and remove outdated information as methods evolve
2. **Access to appropriate relevant literature** for technical leaders and analysts
3. **Dedicated time in the workday to read the literature** so that technical leaders and analysts can keep up-to-date with developments
4. **Uniformly documented knowledge assessment**
5. **A method to acknowledge competence in a specific area** to allow true expertise in testimony (e.g., DNA transfer and activity assessments, see van Oorschot et al. 2019)
6. **Additional training for technical leaders in experimental design and data analysis** to assist with validation studies and protocol development

Creating a Most Valuable Publications (MVP) List

- Experience with an extensive examination of the literature for the NIST Scientific Foundation Review on DNA Mixture Interpretation (2018-2021) – *collecting and studying >1,000 articles*
- Consideration of the new SWGDAM Training Guidelines (published in July 2020)
 - **129 references** in five categories + 6 websites
- Developed initial MVP list in September 2020
 - My assistance was requested from an OSAC task group (I expanded on their initial efforts)
- **Created 26 category groups** (labeled A-to-Z) for various forensic DNA topics
 - Subjective selection of a **#1 article** followed by reference citations defined by date of publication
 - Selected a **total of 480 articles plus 17 books = 497 MVPs**
- **Conducted a virtual workshop in February 2021 with three co-presenters**
 - Input on articles from experienced practitioners and researchers including Robin Cotton, Mecki Prinz, Charlotte Word, Amy Brodeur, Teresa Cheromcha, and Phil Danielson
- **Reduced MVP list from 480 articles to 85 articles** for AAFS 2022 workshop

SWGDAM (2020): “This list is not meant to be all inclusive. The laboratory should develop a list tailored to its specific needs.”

(MVP 2021 List) Category A: Plain Language Guides to Forensic DNA Analysis

(MVP 2022 List)

A1. Sense about Science (2017) Making Sense of Forensic Genetics.

A 40-page plain language guide available at <https://senseaboutscience.org/activities/making-sense-of-forensic-genetics/>.

A2. Jobling, M.A. and Gill, P. (2004) Encoded evidence: DNA in forensic analysis. *Nature Reviews: Genetics* 5(10): 739-751.

A3. The Royal Society (2017) *Forensic DNA Analysis: A Primer for Courts*.

A 60-page plain language guide available at <https://royalsociety.org/-/media/about-us/programmes/science-and-law/royal-society-forensic-dna-analysis-primer-for-courts.pdf>.

A4. Press, R. (2019) DNA Mixtures: A Forensic Science Explainer.

Available at <https://www.nist.gov/featuredstories/dna-mixtures-forensic-science-explainer>. (see also *Forensic Science Review* 31: 87-91 available at [http://forensicsciencereview.com/Abstract/31\(2\)-\(R&C\)%20Full%20text.pdf](http://forensicsciencereview.com/Abstract/31(2)-(R&C)%20Full%20text.pdf))

A1.



A3.



Informative Forensic DNA Reviews and Research Studies (A-to-Z)

Articles

Category Group	Topic(s) Covered	480 (2021)	85 (2022)
A	Plain Language Guides to Forensic DNA Analysis	4	2
B	Serology and Body Fluid Identification	24	3
C	Collection and Storage of Biological Material	25	2
D	DNA Extraction/Purification, Differential Extraction	18	2
E	DNA Quantitation, Degraded DNA	10	2
F	PCR Amplification, Inhibition, and Artifacts	13	3
G	Capillary Electrophoresis Separation and Detection	12	2
H	Assessing Sample Suitability & Complexity, Low-Template	7	2
I	Estimating the Number of Contributors	12	4
J	Data Interpretation, Mixture Deconvolution, Interlab Studies	12	4
K	Interpretation: Binary Approaches (CPI, RMP, LR)	11	5
L	Interpretation: Probabilistic Genotyping Software	44	4
M	Report Writing and Technical Review	8	4

Informative Forensic DNA Reviews and Research Studies (A-to-Z)

Articles

Category Group	Topic(s) Covered	480 (2021)	85 (2022)
N	Court Testimony, Communication, Juror Comprehension	22	5
O	Autosomal STR Markers and Kits	29	2
P	Mitochondrial DNA Testing	11	3
Q	Y-Chromosome and X-Chromosome Testing	17	4
R	DNA Databases and Investigative Genetic Genealogy	14	3
S	Statistical Analysis	11	2
T	Population Genetics	11	2
U	DNA Phenotyping (Ancestry, Appearance, Age)	24	2
V	New Technologies (Rapid DNA, Massively Parallel Sequencing)	35	5
W	DNA Transfer and Activity Level Reporting	57	8
X	Non-Human DNA Testing	15	2
Y	Method Validation, Quality Control, and Human Factors	23	5
Z	General Forensic Science Topics	11	3

(MVP 2022 List) Category W:

DNA Transfer and Activity Level Reporting

1. van Oorschot, R.A.H., Szkuta, B., Meakin, G.E., Kookshoorn, B., Goray, M. (2019) DNA transfer in forensic science: a review. *Forensic Science International: Genetics* 38: 140-166.
2. Taylor, D., Abarno, D., Rowe, E., Rask-Nielsen, L. (2016) Observations of DNA transfer within an operational Forensic Biology Laboratory. *Forensic Science International: Genetics* 23: 33-49.
3. Kokshoorn, B., Blankers, B.J., de Zoete, J., Berger, C.E.H. (2017) Activity level DNA evidence evaluation: On propositions addressing the actor or the activity. *Forensic Science International* 278: 115-124.
4. Taylor, D., Kokshoorn, B. and Biedermann, A. (2018) Evaluation of forensic genetics findings given activity level propositions: A review. *Forensic Science International: Genetics* 36: 34-49.
5. Burrill, J., Daniel, B., Frascione, N. (2019) A review of trace “touch DNA” deposits: Variability factors and an exploration of cellular composition. *Forensic Science International: Genetics* 39:8-18.
6. Gosch, A. and Courts, C. (2019) On DNA transfer: the lack and difficulty of systematic research and how to do it better. *Forensic Science International: Genetics* 40: 24-36.
7. Gosch, A., Euteneuer, J., Preuss-Wossner, J., Courts, C. (2020) DNA transfer to firearms in alternative realistic handling scenarios. *Forensic Science International: Genetics* 48: 102355.
8. van Oorschot, R.A.H., Meakin, G.E., Kookshoorn, B., Goray, M., Szkuta, B. (2021) DNA transfer in forensic science: recent progress towards meeting challenges. *Genes* 12: 1766. Available [open access] at <https://www.mdpi.com/2073-4425/12/11/1766>.



How We Examined MVPs in the 2021 Workshop

1. Discuss important principles involved with the category topic (e.g., DNA extraction or PCR amplification)
2. In each examined category, briefly review the number and types of articles in our reference list and number of times cited in Google Scholar (as of January 2021)
3. Focus on one or a few specific articles and the findings reported
4. Summarize and review key takeaways

116-page handout
240 slides and a 33-page reference list

https://strbase.nist.gov/pub_pres/AAFS2021-W19-Handouts.pdf

#1 MVP(s) on PGS

L1. Coble, M.D. and Bright, J.-A. (2019) Probabilistic genotyping software: An overview. *Forensic Science International: Genetics* 38: 219-224.

The screenshot shows the article page for "Probabilistic genotyping software: An overview" by Michael D. Coble and Jo-Anne Bright. The page includes the Elsevier logo, the journal title "Forensic Science International: Genetics", and the article title. It also features a "Check for updates" button and the authors' affiliations.

Forensic Science International: Genetics 38 (2019) 219–224

Contents lists available at ScienceDirect

Forensic Science International: Genetics

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

Probabilistic genotyping software: An overview

Michael D. Coble^{a,*}, Jo-Anne Bright^b

^a Center for Human Identification, Department of Microbiology, Immunology, and Genetics, University of North Texas Health Science Center, 3500 Camp Bowie Blvd., Fort Worth, TX 76107, USA

^b Institute of Environmental Science and Research Limited, Private Bag 92021, Auckland, 1142 New Zealand

Google Scholar
Cited 40 times
(8 Jan 2021)

Google Scholar
Cited 80 times
(10 Jan 2022)

- **Why is this article valuable?**

- Provides a historical perspective and overview on the movement from binary methods of interpretation to probabilistic methods of interpretation

Interpretation: Probabilistic Genotyping Software - PGS (Discrete, Continuous) (Category L – 44 articles)

• Reviews:



- DNA Commission on allele drop-out/in (L11)
- PGS overview and history (L1)
- Comparison of statistical models (L17)
- Historical: 20 years of R&D (L26)
- Paradigm shift (L34)
- Statistical evaluation of forensic evidence (L24)

• Validation:

- Definitions and recommended tests (L25, L32)
- EuroForMix studies (L29, L39)
- TrueAllele studies (L5, L8, L27, L28, L43)
- STRmix theory and studies (L14, L18, L19, L20, L21, L30, L33, L36, L42, L44)
- STRmix collaborative exercise (L40)
- Comparing model performance (L22, L29, L38, L41)

• Continuous Models:

- Early work (L3, L4, L6, L7, L9, L10)
- Modeling stutter (L13, L15)
- Low template profiles (L23, L31)
- Information gain from peak heights (L37)

• Likelihood Ratios:

- Framework for addressing questions (L2)
- Exploring nondonor distributions (L12, L44)
- Calibration and method validation (L16, L35)

2022 MVP List reduced to 4 articles
(2021 L1, L26, L32) + new 2021 article

ISFG DNA Commission Articles

Several of the #1 MVPs are ISFG DNA Commission articles:

DNA mixture interpretation

- **K1.** Gill, P., Brenner, C.H., Buckleton, J.S., Carracedo, A., Krawczak, M., Mayr, W.R., Morling, N., Prinz, M., Schneider, P.M. and Weir, B.S. (2006) DNA Commission of the International Society of Forensic Genetics: Recommendations on the interpretation of mixtures. *Forensic Science International* 160: 90-101.

mtDNA

- **P1.** Parson, W., Gusmão, L., Hares, D.R., Irwin, J.A., Mayr, W.R., Morling, N., Pokorak, E., Prinz, M., Salas, A., Schneider, P.M., Parsons, T.J. (2014) DNA Commission of the International Society for Forensic Genetics: revised and extended guidelines for mitochondrial DNA typing. *Forensic Science International: Genetics* 13: 134-142.

Y-STRs

- **Q1.** Roewer, L., Andersen, M.M., Ballantyne, J., Butler, J.M., Caliebe, A., Corach, D., D'Amato, M.E., Gusmão, L., Hou, Y., de Knijff, P., Parson, W., Prinz, M., Schneider, P.M., Taylor, D., Vennemann, M., Willuweit, S. (2020) DNA commission of the International Society of Forensic Genetics (ISFG): Recommendations on the interpretation of Y-STR results in forensic analysis. *Forensic Science International: Genetics* 48: 102308.

Non-human DNA testing

- **X1.** Linacre, A., Gusmão, L., Hecht, W., Hellmann, A.P., Mayr, W.R., Parson, W., Prinz, M., Schneider, P.M., Morling, N. (2011) ISFG: recommendations regarding the use of non-human (animal) DNA in forensic genetic investigations. *Forensic Science International: Genetics* 5(5): 501-505.

- **These are freely available on the ISFG website:**

- <https://www.isfg.org/Publications/DNA+Commission>



genes

an Open Access Journal by MDPI

Special Issue (Niels Morling, editor): **Advances in Forensic Genetics**

https://www.mdpi.com/journal/genes/special_issues/Advances_Forensic_Genetics

24 articles published (2021-2022), topics covered include:



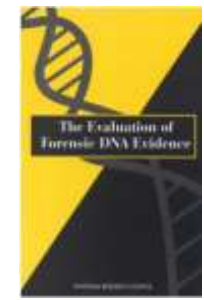
PGS Review: EuroForMix, DNASstatistX, STRmix

- OpenArray for forensic phenotyping
- Skin pigmentation and genetic ancestry
- Eye color prediction
- Ancestry informative markers (VISAGE)
- Single cell analysis for forensic phenotyping
- Animal forensic genetics
- Predicting visible traits in dogs (CaDNAP)
- Single cell analysis for mixture interpretation
- New STR panel for cross-species bird DNA
- Ancient DNA methods improve Korean/WW2 IDs



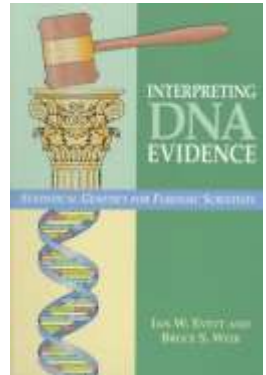
DNA transfer review and recent progress

- Bayesian Networks for DNA transfer questions
- SNP markers for investigative genetic genealogy (FORCE panel)
- **DNA sampling in burglary investigations**
- Body fluid ID and tissues
- Microbiome analysis
- **Software options for forensic sequencing**
- ChrY and mtDNA statistics/assessment
- Ethical decision-making as lived practice
- DNA from aged rootless hair shafts in Romanov relics

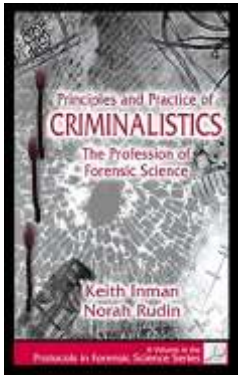


NRC II
1996

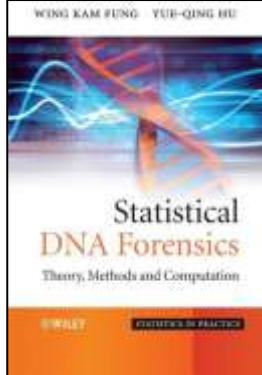
(17) Informative Textbooks on Forensic DNA



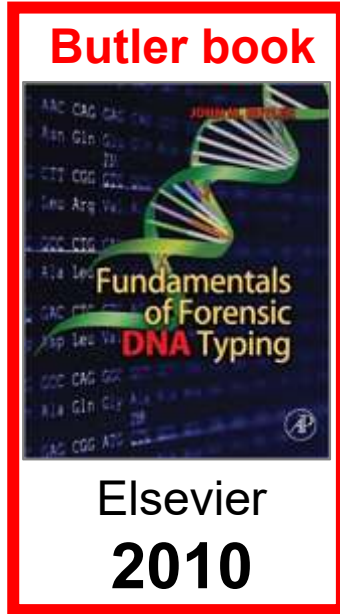
Sinauer
1998



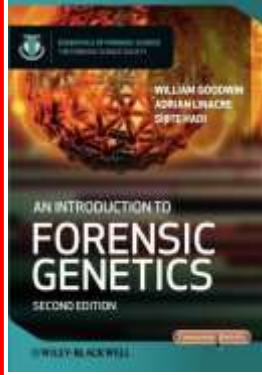
CRC Press
2001



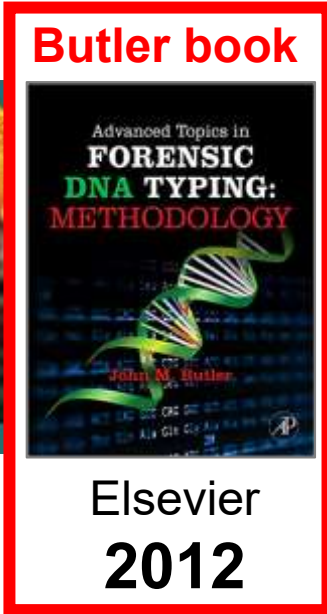
Wiley
2008



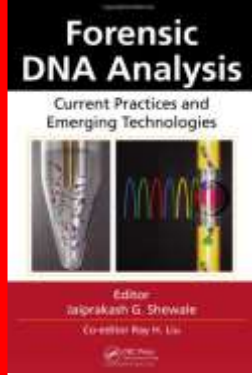
Elsevier
2010



Wiley
2011



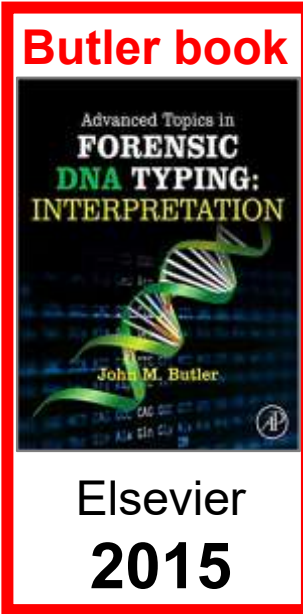
Elsevier
2012



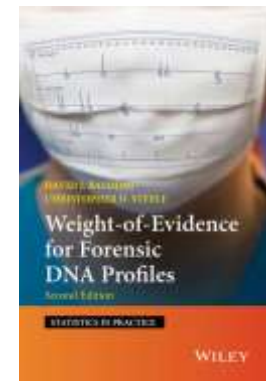
CRC Press
2013



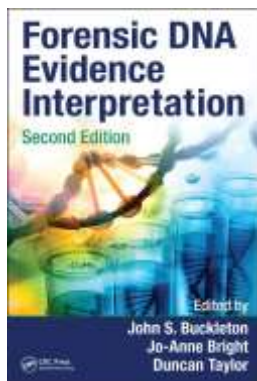
Elsevier
2014



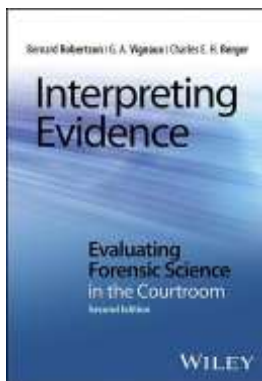
Elsevier
2015



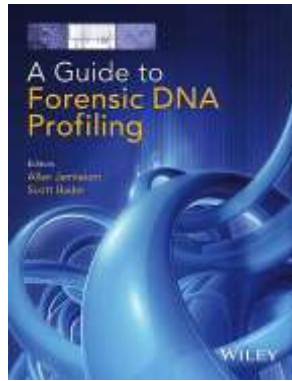
Wiley
2015



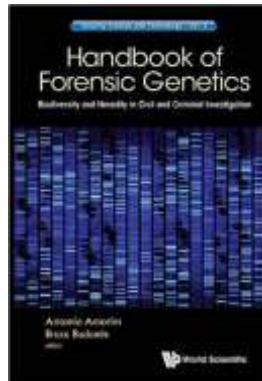
CRC Press
2016



Wiley
2016



Wiley
2016



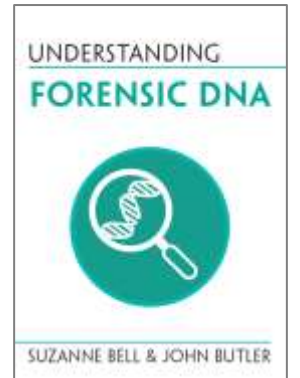
World Scientific
2016



CRC Press
2020



Elsevier
2020



Cambridge
University Press
2022

Testing the Current 26 MVP “A-to-Z” Categories with the 2019-2022 INTERPOL Review

**Starting with
4,087 articles**

Scopus & Web of Science “forensic DNA” searches
January 2019 to March 2022 (with some additions)

Removed duplicates
and **sorted**
into 26 categories
MVP “A to Z”
+ 6 additional ones

**1,884
articles**

version
May 16, 2022

MVP	# articles	MVP	# articles
A	9	N	18
B	56	O	49
C	116	P	95
D	100	Q	117+25
E	27	R	77
F	38	S	54
G	5	T	147
H	3	U	172
I	10	V	105+32
J	20	W	57
K	6	X	126
L	63	Y	22
M	2	Z	18

Additional Categories

Human Remains ID (DVI)

92

Microbial & Viral DNA

59

Microhaplotypes/InDels

53

Proteomics

15

Sexual Assault Policy

33

Other Applications

48

(+12 unsorted)

Some Final Thoughts

1. No selection criteria or reference list will be perfect or complete
 - continuing research and future review articles add knowledge to our field
 - some references could be removed to focus content in various categories
2. We would love to hear your ideas on how to best maintain an updated list to benefit the community
 - Are there other category groups that should be included in MVP lists?
3. How could a national/international MVP list benefit future training?
 - Would it be worth conducting an AAFS or EAFS survey on this topic?
 - It would be nice to have all of the articles available as PDF files that could be freely shared
 - If we understand the need, then we can lay the groundwork for future possibilities in funding

Thank you for your attention!

MVPs of DNA

2021 (480): https://strbase.nist.gov/pub_pres/AAFS2021-W19-Handouts.pdf (pp. 3-35)

2022 (85): https://strbase.nist.gov/pub_pres/AAFS2022-W2-NIST-Forensic-DNA-Activities-FINAL.pdf (pp. 77-84)

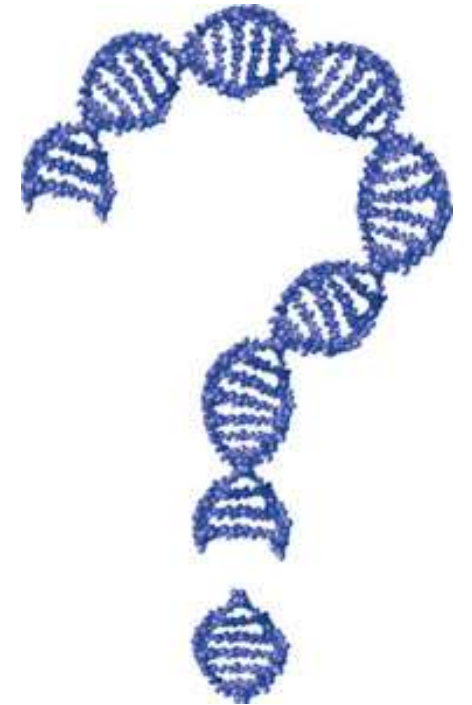
John Butler

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<https://www.nist.gov/topics/forensic-science>



RESEARCH. STANDARDS. FOUNDATIONS.



Questions?