Scientific Working Group on DNA Analysis Methods

Public Meeting

November 2, 2009

Ted Staples

SWGDAM Chair

Outline

- Introduction
- Committee Updates
  - Quality Assurance
  - Missing Persons & Mass Disasters
  - CODIS
  - Mixture Interpretation
- Questions

Membership

- Group of forensic scientists (DNA Technical Leaders and CODIS Administrators) from international, federal, state and local laboratories
- Currently 23 Members and 30 Invited Guests from:
  - 3 International Forensic Laboratories (BKA, CFS Toronto, RCMP)
  - 5 Federal Laboratories/Agencies (ATF, AFDIL, Army Crime Lab, NIST and FBI Laboratory)
  - 23 State/Local Forensic Laboratories
  - 3 Academic Institutions

SWGDAM Executive Board

- SWGDAM Executive Board
  - **Ted Staples**, Chair, Georgia Bureau of Investigation
  - **Heather Seubert**, Vice-Chair, FBI Laboratory
  - **Angelo Della Manna**, AL Dept. of Forensic Sciences,
  - **Phil Kinsey**, MT Forensic Science Division
  - **Ken Konzak**, CA Dept. of Justice
  - **Peg Schwartz**, VT Forensic Laboratory
  - **Taylor Scott**, IL State Police

Tasks

- Recommend revisions, as necessary, to the Quality Assurance Standards
- Serve as a forum to discuss, share, and evaluate forensic biology services
- Recommend and conduct research to develop and/or validate forensic biology methods

Conduct of Business

- Semiannual meetings in January and July of each year
  - 3 days meeting with 1 day dedicated to Committees
  - Committees use conference calls, WebEx to work on projects during the year
- SWGDAM Bylaws also permit conference calls for SWGDAM Executive Board and e-mail voting
  - Used frequently over the past several years to complete revisions to the Quality Assurance Standards and Audit Documents

SWGDAM Bylaws

- Describe Mission, Membership, Executive Board, Committees, Meetings, Conduct of Business and Amendment of Bylaws

- Available at http://www.fbi.gov/hq/lab/fsc/backissu/april2003/swgdambylaws.htm.

SWGDAM Bylaws

- Members are appointed by the Chairman based on recommendation of the Membership Committee
  - Nominations from Members, Invited Guests and Laboratories
  - Representation from federal, state and local laboratories as well as geographic regions
  - FBI has 5 regular members for the 5 DNA Units/programs
- SWGDAM membership votes to select Executive Board members

Committees

- CODIS
- Mass Spectrometry
- Missing Persons and Mass Disasters
- Mitochondrial DNA
- Mixture Interpretation
- Quality Assurance (hiatus)
- Ad Hoc Group on Low Template DNA

Recent Work Products

- Revisions recommended to FBI Director for
  - *Quality Assurance Standards for Forensic DNA Testing Laboratories* and
  - *Quality Assurance Standards for DNA Databasing Laboratories*
- Recommended 2 Audit Documents:
  - FBI Quality Assurance Standards Audit for Forensic DNA Testing Laboratories
  - FBI Quality Assurance Standards Audit for DNA Databasing Laboratories
- Joint Policy Statement on Contamination with ENFSI (European Network of Forensic Science Institutes) (in press)

Updates

- New Committee Formed at July 2009 Meeting
- MtDNA committee members joined with Mass Spec members to assist them with first meeting

Goals of Mass Spec Committee

- Provide guidance to the forensic DNA community regarding:
  - The potential utility and value of mass spec technology for DNA analyses
  - Promotion of continued thorough and efficient audits
  - Validation and implementation of technology
  - Communication of concerns, desires, and requirements to technology provider

“Deliverables” for Mass Spec

- Reference list covering basic mass spec topics, terminology, & application biology and DNA
- Provide platform specific guidance related to July 2009 audit document
- Encourage expanded population studies
- Initiate communication with provider

Updates

Welcome input of DNA community on issues of interest and possible Committees

Quality Assurance Committee

Recognition of QA committee past

- Christine Tomsey
- Robyn Ragsdale
- Barbara Llewellyn
- Sindey Schueler
- David Freeman
- Eleni Levedakou
- Debbie Figarelli
- Kate Theisen
- Dorothy Catella
- David Coffman
- Melissa Smrz
- Jim Mudd
- Renee Romero

Recognition of QA committee present

- John Krebsbach
- Peg Schwartz
- Amy McGuckian
- Beth Ann Marne
- Jodi Dahl
- Eugene Lien
- Heather Seubert
- Richard Guerrieri

Accomplishments (since 2004)

- Revised Standards, Approved 7/2007
  - Public comments addressed
  - Public presentations made
- Audit Document, Approved 6/2009
  - Public comments addressed
  - Public presentations made

Missing Person / Mass Disaster Committee

MP/MD Committee

Old Business - Minifiler Validation

- Finalized Response for the request for more information from the NDIS Procedures Board.
- Provided CD of final documentation to SWGDAM Chair

Task Completed!

MP/MD Committee

Discussion - CODIS v.6.0

- Reviewed Disposition assignments for Missing Persons search returns for update in CODIS 6.0 SP1
- Discussed feasibility of “rare” mtDNA haplotype searching through use of single node pedigrees for limited family reference sample cases.

MP/MD Committee

Clarification of Specimen Category Definitions:

- A **Missing Person** sample is a sample whose origin is medically or legally documented as having come directly from the missing individual.
- A **Deduced Missing Person** sample is a sample that is presumed to have originated from the missing individual based on investigative information. It is recommended that these items are validated for use by kinship comparisons to familial references.

MP/MD Committee

- Committee Point of Contact: John Planz
  - 817-735-2397
  - Jplanz@hsc.unt.edu

CODIS Committee

CODIS Committee

Mission:

"To identify, evaluate and research issues relating to the use of CODIS in federal, state and local forensic laboratories."

CODIS Committee

Objectives:

- If in the course of the review of such issues, the CODIS committee determines that revisions to the FBI Director's Quality Assurance Standards for Forensic DNA Testing Laboratories and Convicted Offender DNA Databasing Laboratories are needed, the committee shall recommend such changes to the Chair of SWGDAM for consideration by SWGDAM.

- If the CODIS committee is presented with issues relating to the operation of CODIS, such as software functionality and performance, the committee shall forward such issues and their findings/recommendations through the SWGDAM chair to the Chief of the CODIS Unit.

- The CODIS committee will review issues requested by the NDIS Procedures Board through the SWGDAM chair and will provide their findings and recommendations through the SWGDAM chair to the Board.

CODIS Committee

Tasks/objectives accomplished:

(Since last meeting)

- Finalized Databasing QAS audit document after a score of WebEx meetings!

CODIS Committee

Tasks/objectives accomplished:

- Discussions of new ENFSI core loci
- Discussions of PopStats requirements
- 2009 CODIS Conference topics

CODIS Committee

Tasks/Objectives pending:

- PopStats discussions to continue
- Finalization of CODIS Conference Agenda

CODIS Committee

Committee Point of Contact:

Committee Chair:

Douglas Hares
703-632-8315
douglas.hares@ic.fbi.gov

Committee Co-Chair:

Elizabeth Johnson
404-469-7023
elizabeth.johnson4@us.army.mil

Mixture Interpretation Committee

Committee Member Backgrounds

- State Lab – CA (x2), OR, WA, MT, MN, CT, MA, MD
- State/Local Lab – CFS Toronto (early on PBSO)
- Canadian Lab – RCMP, CFS Toronto
- Federal Lab/Agency – FBI, NIST
- Academic – Jack Ballantyne, George Carmody

With 15 members, we represent almost one-third of SWGDAM

Mixture Committee Goals

- Conduct case summary analysis to see how many and what type of mixtures are being observed in crime labs
- **Complete interpretation guidelines** to aid mixture interpretation
- Prepare training materials

Gathered Case Summary Data

During 2007 and early 2008, **Ann Gross** (MN BCA) from the SWGDAM Mixture Interpretation Committee **coordinated the collection of case summary data** from **14 different forensic labs** who collectively reported on **4780 samples**.

A preliminary summary of this information is divided by crime classifications: sexual assault, major crime (homicide), and high volume (burglary). **Over half of the samples examined were single source and ~75% of all reported mixtures were 2-person.**

CFS Toronto Case Summary Data

Case type		# contributors				
N = 276		**1**	**2**	**3**	**4**	**>4**
Sexual Assault	N = 152	**42%**	52%	7%	1%	--
High Volume	N = 56	**69%**	16%	16%	--	--
Major Crime	N = 68	**59%**	34%	7%	--	--

Single source

Mixtures

Mixture Case Summaries

"Final" Data Set from 14 Different Labs

	minimum # of contributors					
Crime Class	**1**	**2**	**3**	**4**	**>4**	**N**
Sexual Assault	884	787	145	11	0	1827
Major Crime	1261	519	182	32	0	1994
High Volume	344	220	140	11	5	720
Total	2489	1526	467	54	5	4541
	54.8%	33.6%	10.3%	1.2%	0.1%	

Single source **mixtures**

Plan to conduct further data analysis and publish results

Mixture Interpretation Committee

Accomplished during and since July 2009 meeting:

- Worked on casework autosomal STR interpretation guidelines
  - Based on input from previous meeting efforts (July 2008 and Nov 2008) and FBI DNA Unit I
  - 2 full days + 3 WebEx meetings (4 hours each)
- **Initial draft guidelines were provided to SWGDAM body in July and comments were received back in August**
  - each comment has been addressed in subsequent WebEx meetings

Purpose and Scope of Document

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

This document provides guidelines for the interpretation of DNA typing results from short tandem repeats (STR) and **supersedes the Scientific Working Group on DNA Analysis Methods (SWGDAM) Short Tandem Repeat (STR) Interpretation Guidelines (2000).** Guidance is provided for **forensic casework** analyses on the identification and application of thresholds for allele detection and interpretation, and appropriate statistical approaches to the **interpretation of autosomal STRs with further guidance on mixture interpretation**. Laboratories are encouraged to review their standard operating procedures and validation data in light of these guidelines and to update their procedures as needed. It is anticipated that these guidelines will evolve further as future analytical technologies emerge.

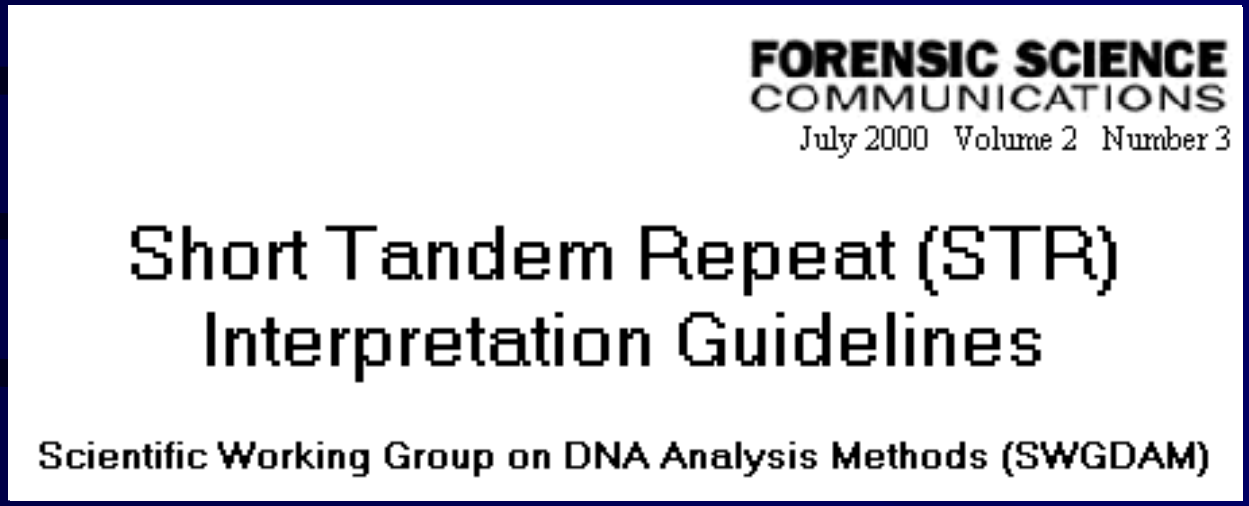
Preamble that Never Made It Out of Our "Constitutional" Committee…

Previously Proposed Preamble

We the Members of the Mixture Interpretation Committee of the Scientific Working Group on DNA Analysis Methods, in Order to form more perfect DNA interpretation guidelines for mixtures, establish Justice, insure laboratory and analyst consistency, provide for an appropriate prosecution of the guilty, prevent implementation of sequential unmasking, promote a better understanding of mixture interpretation, and secure the Blessings of Liberty to the innocent, do submit these Guidelines for use by the forensic DNA typing community.

Current SWGDAM (2000) STR Interpretation Guidelines

http://www.fbi.gov/hq/lab/fsc/backissu/july2000/strig.htm



1. **Preliminary Evaluation of Data**
2. **Designation**
3. **Interpretation of Results**
4. **Conclusions**
5. **Statistical Interpretation**
6. **References/Suggested Readings**

1. Preliminary Evaluation of Data

1.1. The laboratory should develop criteria to determine whether the results are of sufficient intensity/quality for interpretation purposes using methods appropriate for the detection platform. These criteria should be determined by evaluating data generated by the laboratory.

1.1.1. When quantitative results (e.g., peak amplitude) are used to evaluate STR profiles, the results should be examined to determine if they meet the laboratory's defined analytical and interpretational threshold(s).

1.1.1.1. The analytical threshold(s) is defined as the minimum and maximum intensity thresholds that are determined to assign alleles.

1.1.1.2. The interpretational threshold should be defined empirically.

1.1.2. When quantitative results are not used, the laboratory should establish criteria to interpret alleles based on visual inspection of gel images.

1.2. The laboratory should develop criteria to evaluate internal lane size standards and/or allelic ladders.

1.3. Controls are required to assess analytical procedures.

1.3.1. The laboratory should establish criteria for evaluation of the following controls, including but not limited to: reagent blank, amplification blank, and positive control.

1.3.2. The laboratory should develop criteria for the interpretation and documentation of results in the event that the controls do not perform as expected.

1.4. A laboratory using STR multiplexes that contain redundant loci should establish criteria regarding the concordance of such data.

http://www.fbi.gov/hq/lab/fsc/backissu/july2000/strig.htm

2. Designation

2.1. The laboratory should establish criteria to assign allele designations to appropriate peaks or bands.

2.1.1. *Locus Designation:* The laboratory should establish criteria to address locus assignment for alleles.

2.1.2. *Allele Designation:* The laboratory should designate alleles in accordance with Combined DNA Index System (CODIS) recommendations.

2.1.2.1. Whenever possible, allele designation should be based operationally on the number of repeat sequences contained within the allele and by comparison to an allelic ladder.

2.1.2.2. The designation of alleles containing an incomplete repeat motif (i.e., an off-ladder allele falling within the range spanned by the ladder alleles) should include the number of complete repeats and, separated by a decimal point, the number of base pairs in the incomplete repeat (e.g., FGA 18.2 allele).

2.1.2.3. If an allele falls above the largest or below the smallest allele of the allelic ladder, the allele should be designated as either greater than (>) or less than (<) the respective ladder allele, or when appropriate interpolation can be used.

2.2. Artifacts can occur and should be noted. These may include, but are not limited to, the following: pull-up, stutter, and nontemplate nucleotide addition. The laboratory should establish guidelines based on empirical data (obtained internally or externally) to address the interpretation of these and other artifacts.

http://www.fbi.gov/hq/lab/fsc/backissu/july2000/strig.htm

3. Interpretation of Results

3.1. The laboratory should define conditions in which the data would lead to the conclusion that the source of the DNA is either from a single person or more than one person. This may be accomplished by an examination of the number of alleles at each locus, peak height ratios, and/or band intensities.

3.1.1. *Single Contributor:* A sample may be considered to be from a single contributor when the observed number of alleles at each locus and the signal intensity ratios of alleles at a locus are consistent with a profile from a single contributor. All loci should be evaluated in making this determination.

Component Deconvolution

3.1.2. *Mixtures With Major/Minor Contributors:* A sample may be considered to consist of a mixture of major and minor contributors if there is a distinct contrast in signal intensities among the alleles. The difference is evaluated on a case-by-case context. All loci should be evaluated in making this determination.

Profile Subtraction

3.1.3. *Mixtures With a Known Contributor(s):* In some cases, when one of the contributors (e.g., the victim) is known, the genetic profile of the unknown contributor may be inferred. Depending on the profiles in the specific instance, this can be accomplished by subtracting the contribution of the known donor from the mixed profile.

CPE/CPI Approach

3.1.4. *Mixtures With Indistinguishable Contributors:* When major or minor contributors cannot be distinguished because of similarity in signal intensities or the presence of shared or masked alleles, individuals may still be included or excluded as possible contributors.

3.2. The laboratory should have guidelines for interpretation of partial profiles (i.e., profiles with fewer loci than tested) that may arise from degraded or limited quantity DNA or from the presence of polymerase chain reaction (PCR) inhibitors.

3.3. The laboratory should establish guidelines to interpret profiles that exhibit potential stochastic effects (e.g., allele dropout and/or substantial imbalance of alleles).

http://www.fbi.gov/hq/lab/fsc/backissu/july2000/strig.htm

4. Conclusions

4.1. The laboratory should prepare guidelines for formulating conclusions resulting from comparisons of single source samples and mixtures with known reference samples.

4.1.1. General categories of conclusions include, but are not limited to: inclusion or match, exclusion or nonmatch, inconclusive or uninterpretable, and no results.

5. Statistical Interpretation

5.1. The source of the population database(s) used should be documented. Relevant population(s) for which the frequency will be calculated should be identified.

5.2. The formulas used in calculating the frequency of a DNA profile should be defined for the following:

5.2.1. Heterozygote profiles

5.2.2. Homozygote profiles

5.2.3. Composite profiles (i.e., multiple locus profiles)

5.2.4. Minimum allele frequencies

5.2.5. Mixture calculations

5.2.6. Biological relationships, where appropriate

5.3. When used, criteria for the declaration of source attribution should be documented.

http://www.fbi.gov/hq/lab/fsc/backissu/july2000/strig.htm

6. References/Suggested Readings

Committee on DNA Forensic Science, National Research Council. *An Update: The Evaluation of Forensic DNA Evidence.* National Academy Press, Washington, DC, 1996.

DNA Advisory Board. Quality assurance standards for convicted offender DNA databasing laboratories (approved April 1999), *Forensic Science Communications* (July 2000) 2. Available at www.fbi.gov/programs/lab/fsc/backissu/july2000/codispre.htm

DNA Advisory Board. Quality assurance standards for forensic DNA testing laboratories (approved October 1998), *Forensic Science Communications* (July 2000) 2. Available at www.fbi.gov/programs/lab/fsc/backissu/july2000/codispre.htm

DNA Commission, ISFH. DNA recommendations: 1994 report concerning further recommendations regarding PCR-based polymorphisms in STR (short tandem repeat) systems, *Forensic Science International* (1994) 69:103–104.

Federal Bureau of Investigation. *National DNA Index System (NDIS) Procedures Manual.* U.S. Department of Justice, Washington, DC, February 1999 (revised).

Needed Revisions After a Decade…

Quality Assurance Standards (1998/1999) → **Quality Assurance Standards (2009)**

STR Interpretation Guidelines (2000) → **STR Interpretation Guidelines (2010)**

1066 words (4 pages)

9453 words (27 pages)

SWGDAM Interpretation Guidelines for Autosomal STR Typing

(including Mixture Interpretation)

Introduction/Background

1. Preliminary evaluation of data
2. Allele designation
3. Interpretation of DNA typing results
    1. Non-allelic peaks
    2. Application of peak height thresholds to allelic peaks
    3. Peak height ratio
    4. Number of contributors to a DNA profile
    5. Interpretation of DNA typing results for mixed samples
    6. Comparison of DNA typing results
4. Statistical analysis of DNA typing results
5. Statistical formulae
6. References and literature cited
7. Additional suggested readings

Glossary

Appendices (started but now removed): flowcharts & report wording examples

Points of Discussion for the Interpretation Guidelines

- **Scope**: autosomal STRs or just mixtures; both casework and databasing or just casework
- **Word use**: "must" versus "should" or "may" (how strong of a recommendation)
- **Detailed appendices**: full SWGDAM group recommended keeping the proposed Appendices: ~~Flowchart~~, Formulae, ~~Report Wording Examples for mixtures~~, Glossary

Review of Information contained in Section 3.2. Application of Peak Height Thresholds to Allelic Peaks

In Review:

- Introductory material (for educational purposes) addressing the need for a stochastic threshold
- *3.2.1.* How to establish a stochastic threshold
- *3.2.1.1.* When additional studies are needed
- *3.2.1.2.* Assumptions needed for use of data below stochastic threshold
- *3.2.2.* Must have supportive data and documentation for single threshold using alternative criteria to address potential stochastic effects

3.2. Application of Peak Height Thresholds to Allelic Peaks

Amplification of low-level DNA samples may be subject to stochastic effects, where two alleles at a heterozygous locus exhibit considerably different peak heights (i.e., peak height ratio generally <60%) or an allele fails to amplify to a detectable level (i.e., allelic dropout). Stochastic effects within an amplification may affect one or more loci irrespective of allele size. Such low-level samples exhibit peak heights within a given range which is dependent on quantitation system, amplification kit and detection instrumentation. A threshold value can be applied to alert the DNA analyst that all of the DNA typing information may not have been detected for a given sample. This threshold, referred to as a **stochastic threshold**, is defined as the value above which it is reasonable to assume that allelic dropout has not occurred within a single source sample. The application of a stochastic threshold to the interpretation of mixtures should take into account the additive effects of potential allele sharing.

Current wording (October 26, 2009 draft)

3.2. Application of Peak Height Thresholds to Allelic Peaks

- 3.2.1. The laboratory establishes a stochastic threshold based on empirical data derived within the laboratory and specific to the quantitation and amplification systems and the detection instrumentation used. It is noted that a stochastic threshold may be established by assessing peak height ratios across multiple loci in dilution series of DNA amplified in replicate; the RFU value at which substantial imbalance of alleles and/or allele dropout may tend to occur effectively constitutes the stochastic threshold.

Current wording (October 26, 2009 draft)

3.2. Application of Peak Height Thresholds to Allelic Peaks

- 3.2.1.1. The laboratory should perform additional studies to establish independent criteria for application of a separate stochastic threshold(s) if measures are used to enhance allelic height. Such measures may include increased amplification cycle number, increased injection time, and post-amplification purification/concentration of amplified products. The criteria should address the potential that stochastic effects may persist despite the enhancement measures.

Current wording (October 26, 2009 draft)

3.2. Application of Peak Height Thresholds to Allelic Peaks

- 3.2.1.2. For samples for which an assumption can be made as to the number of contributors, the laboratory should establish criteria for comparison of allelic peaks which fall below the stochastic threshold As an example, if a locus in an assumed single source sample exhibits two peaks, one or both of which are below the stochastic threshold, the laboratory may use that locus for matching purposes. Also, the presence of male DNA may be established based on a Y-allele at amelogenin that is below the stochastic threshold.

Current wording (October 26, 2009 draft)

3.2. Application of Peak Height Thresholds to Allelic Peaks

- 3.2.2. If a stochastic threshold based on peak height is not used in the evaluation of DNA typing results, the laboratory must establish alternative criteria (e.g., quantitation values) for addressing potential stochastic amplification. The criteria must be supported by empirical data and internal validation and must be documented in the standard operating procedures.

Some Other Example Statements

- 3.5.2. The laboratory should define and document what, if any, assumptions are used in a particular mixture deconvolution.

- 3.5.3. A laboratory may define other quantitative characteristics of mixtures (e.g., mixture ratios) to aid in further refining the contributors.

Current wording (October 26, 2009 draft)

3.5.8. Interpretation of Potential Stutter Peaks in a Mixed Sample

3.5.8.1. For mixtures in which minor contributors are determined to be present, a peak in stutter position (generally n-4) may be determined to be 1) a stutter peak, 2) an allelic peak, or 3) indistinguishable as being either an allelic or stutter peak. This determination is based principally on the height of the peak in the stutter position and its relationship to the stutter percentage expectations established by the laboratory.

3.5.8.2. When the height of a peak in the stutter position exceeds the stutter expectation for a given locus, that peak is consistent with being of allelic origin and should be designated as an allele.

3.5.8.3. If a peak is at or below this expectation, it is generally designated as a stutter peak. However, it should also be considered as a possible allelic peak, particularly if the peak height of the potential stutter peak(s) is consistent with (or greater than) the heights observed for any allelic peaks that are conclusively attributed (i.e., peaks in non-stutter positions) to the minor contributor(s).

Relates to ISFG (2006) Mixture Recommendation #6

ISFG (2006) Mixture Recommendation

- Recommendation 6: If the crime profile is a major/minor mixture, **where minor alleles are the same size (height or area) as stutters of major alleles, then stutters and minor alleles are indistinguishable…**

Gill *et al.* (2006) DNA Commission of the International Society of Forensic Genetics: Recommendations on the interpretation of mixtures. *Forensic Sci. Int.* 160: 90-101

Consideration of Peak in Stutter Position

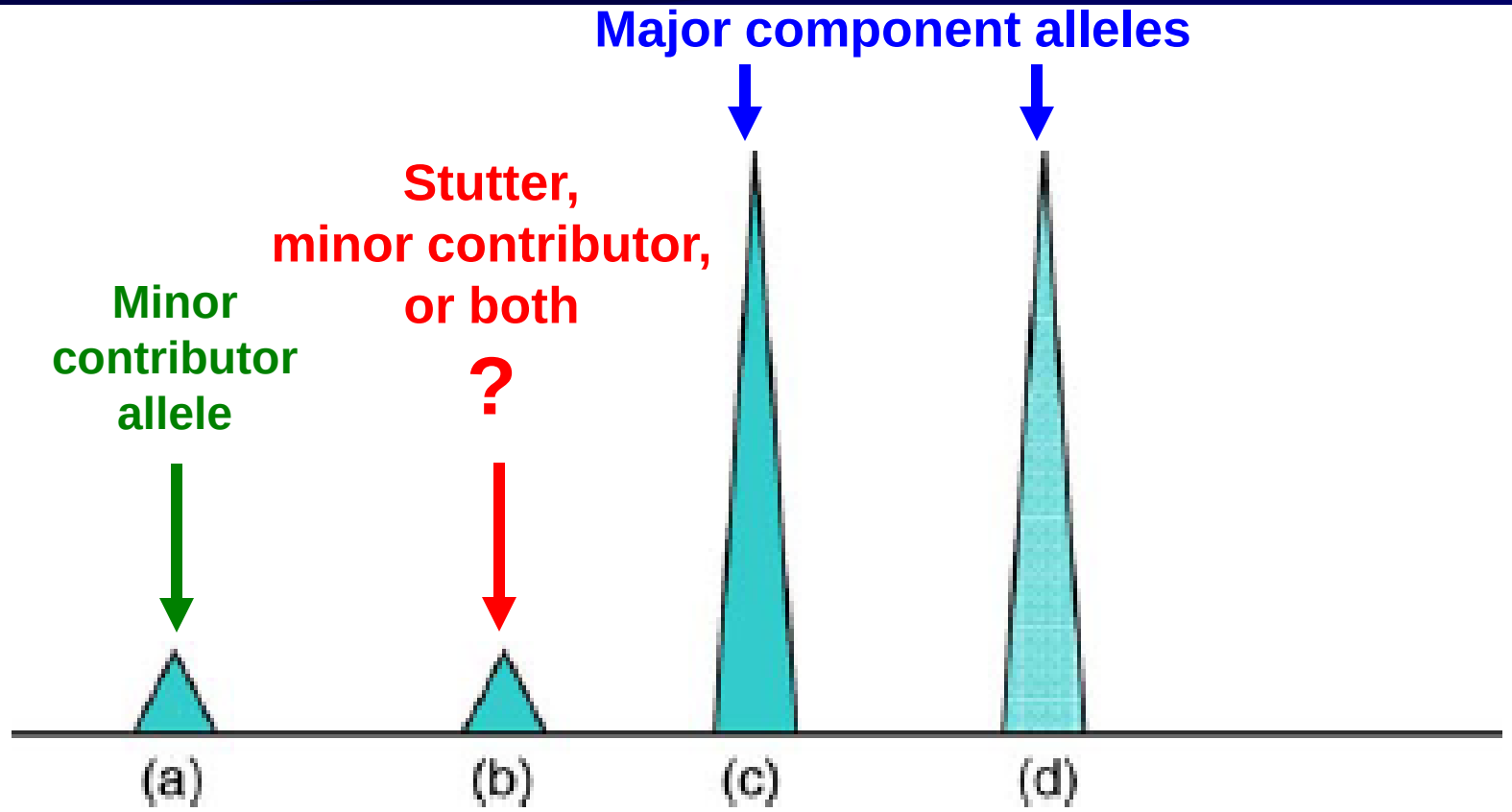


Fig. 4. *c* and *d* are unambiguous alleles, *b* is a minor allele in a stutter position and *a* is an unambiguous minor allele.

Gill *et al.* (2006) DNA Commission of the International Society of Forensic Genetics: Recommendations on the interpretation of mixtures. *Forensic Sci. Int.* 160: 90-101

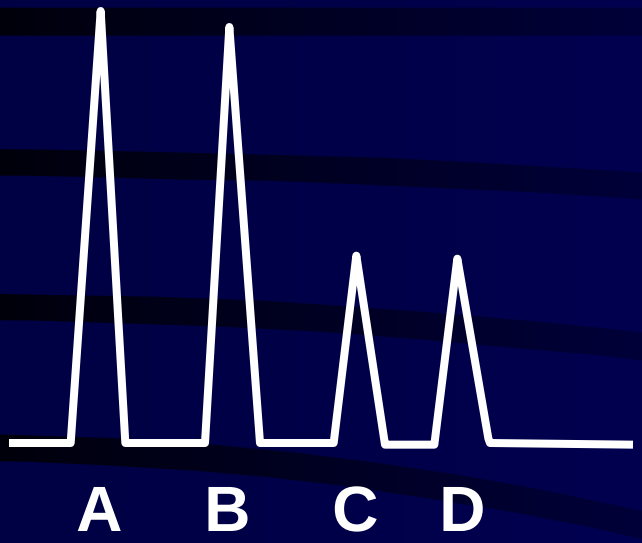
Current wording (October 26, 2009 draft)

Table 1 – Suitable Statistical Analyses for DNA Typing Results

Category of DNA Typing Result	RMP	CPE/CPI	LR (1)
Single Source	✔		✔
Single Major Contributor to a Mixture	✔		✔
Multiple Major Contributors to a Mixture	✔ (2)	✔ (2)	✔
Single Minor Contributor to a Mixture	✔	✔ (3)	✔
Multiple Minor Contributors to a Mixture	✔ (2)	✔ (3)	✔
Indistinguishable Mixture	✔ (1)	✔	✔

(1) Restricted or unrestricted
(2) Restricted
(3) All potential alleles identified during interpretation are included in the statistical calculation

Unrestricted vs. Restricted



Unrestricted

All combinations of alleles are deemed possible (relative peak height differences are not utilized)

AB + AC + AD + BC + BD + CD

Restricted

Based on relative peak heights, alleles are paired only where specific combinations of alleles are deemed possible

~~**AB + AC + AD + BC + BD + CD**~~

Articles Cited in the Guidelines

6. References and Literature Cited

Ayres, K.L. (2000) Relatedness testing in subdivided populations. *Forensic Sci. Int.* 114:107-115.

Bär, W., Brinkmann, B., Lincoln, P., Mayr, W. R., and Rossi, U. (1994) DNA recommendations – 1994 report concerning further recommendations of the DNA Commission of the ISFH regarding PCR-based polymorphisms in STR (short tandem repeat) systems. *Int. J. Legal Med.* 107: 159-160.

Bär, W., Brinkmann, B., Budowle, B., Carracedo, A., Gill, P., Lincoln, P., Mayr, W. R., and Olaisen, B. (1997) DNA recommendations – further report of the DNA Commission of the ISFH regarding the use of short tandem repeat systems. *Int. J. Legal Med.* 110: 175-176.

Committee on DNA Forensic Science, National Research Council. *An Update: The Evaluation of Forensic DNA Evidence*. National Academy Press, Washington, DC, 1996.

DNA Advisory Board. Quality Assurance Standards for Forensic DNA Typing Laboratories, *Forensic Sci. Comm.* 2 (3). See www.fbi.gov/programs/lab/fsc/backissu/july2000/codispre.htm

DNA Advisory Board (2000) Statistical and population genetic issues affecting the evaluation of the frequency of occurrence of DNA profiles calculated from pertinent population database(s). *Forensic Sci. Comm.* 2(3). See http://www.fbi.gov/programs/lab/fsc/backissu/july2000/dnastat.htm.

FBI Director (2009) Quality Assurance Standards for Forensic DNA Testing Laboratories. See http://www.fbi.gov/hq/lab/html/codis1.htm.

Fung, W.K. and Hu, Y.-Q. (2008) *Statistical DNA Forensics: Theory, Methods and Computation*. Wiley: Hoboken, NJ.

Scientific Working Group on DNA Analysis Methods (SWGDAM). Short Tandem Repeat (STR) Interpretation Guidelines, *Forensic Science Communications* 2 (July 2000). See http://www.fbi.gov/hq/lab/fsc/backissu/july2000/strig.htm

Useful articles for further information

7. Additional Suggested Readings

Bill, M., Gill, P., Curran, J., Clayton, T., Pinchin, R., Healy, M., and Buckleton, J. (2005) PENDULUM-a guideline-based approach to the interpretation of STR mixtures. *Forensic Sci. Int.* 148: 181-189.

Buckleton, J.S., Evett, I.W., Weir, B.S. (1998) Setting bounds for the likelihood ratio when multiple hypotheses are postulated. *Sci. Justice*. 38: 23-26.

Buckleton, J.S., Curran, J.M., Gill, P. (2007) Towards understanding the effect of uncertainty in the number of contributors to DNA stains. *Forensic Sci. Int. Genet.* 1:20-28.

Buckleton, J.S. and Curran, J.M. (2008) A discussion of the merits of random man not excluded and likelihood ratios. *Forensic Sci. Int. Genet.* 2: 343-348.

Budowle, B., Chakraborty, R., Carmody, G., Monson, K.L. (2000) Source attribution of a forensic DNA profile. *Forensic Sci. Commun.* 2(3). See http://www.fbi.gov/hq/lab/fsc/backissu/july2000/source.htm.

Budowle, B., Onorato, A.J., Callaghan, T.F., Della Manna, A., Gross, A.M., Guerrieri, R.A., Luttman, J.C., McClure, D.L. (2009) Mixture interpretation: defining the relevant features for guidelines for the assessment of mixed DNA profiles in forensic casework. *J. Forensic Sci.* 54: 810-821.

Clayton, T.M., Whitaker, J.P., Sparkes, R., Gill, P. (1998) Analysis and interpretation of mixed forensic stains using DNA STR profiling. *Forensic Sci. Int.* 91: 55-70.

Devlin, B. (1993) Forensic inference from genetic markers. *Stat. Methods Med. Res.* 2: 241-262.

Current wording (October 26, 2009 draft)

47 words defined

Glossary

Allelic dropout: failure to detect an allele within a sample or failure to amplify an allele during PCR

Analytical threshold: the minimum height requirement at and above which detected peaks can be reliably distinguished from background noise; peaks above this threshold are generally not considered noise and are either artifacts or true alleles.

Artifact: a non-allelic product of the amplification process (e.g., stutter, non-templated nucleotide addition, or other non-specific product), an anomaly of the detection process (e.g., pull-up or spike), or a by-product of primer synthesis (e.g., "dye blob")

Coincidental match: a match which occurs by chance.

Composite profile: a DNA profile generated by combining typing results from different loci obtained from multiple injections of the same amplified sample and/or multiple amplifications of the same DNA extract. When separate extracts from a given evidentiary item are combined prior to amplification, the resultant DNA profile is not considered a composite profile.

Conditional: as used here, an interpretation category that incorporates assumption(s) as to the number of contributors

CPE: combined probability of exclusion; produced by multiplying the probabilities of exclusion from each locus; 1-CPI.

CPI: combined probability of inclusion; produced by multiplying the probabilities of inclusion from each locus; 1-CPE.

Deconvolution: separation of contributors to a mixed DNA profile based on quantitative peak height information.

Mixture Interpretation Committee

Next Steps

- Discuss final committee document before the SWGDAM body at the January 2010 SWGDAM meeting and hopefully vote to accept

Mixture Interpretation Committee

Future Work

- **Publish guidelines (after SWGDAM discussion/vote)** in *Forensic Sci. Comm.*
- Data mine case summaries provided (and perhaps collect additional data) and submit results to *Forensic Sci. Comm*.
- Prepare training materials with examples?
  - Conduct training workshops
- Y-STR mixtures guidelines?

AAFS 2008 Mixture Workshop

- AAFS (February 19, 2008)
  - **DNA Mixture Interpretation:** Principles and Practice in Component Deconvolution and Statistical Analysis
  - **John Butler (NIST)**
  - **Ann Gross** (MN)
  - **George Carmody** (Carleton U.)
  - **Gary Shutler** (WA)
  - **Joanne Sgueglia** (MA)
  - Angela Dolph (Marshall U./NIST)
  - Tim Kalafut (USACIL)

196 page handout prepared

http://www.cstl.nist.gov/biotech/strbase/training/AAFS2008_MixtureWorkshop.htm

Laying Groundwork for Training Materials on Mixture Interpretation

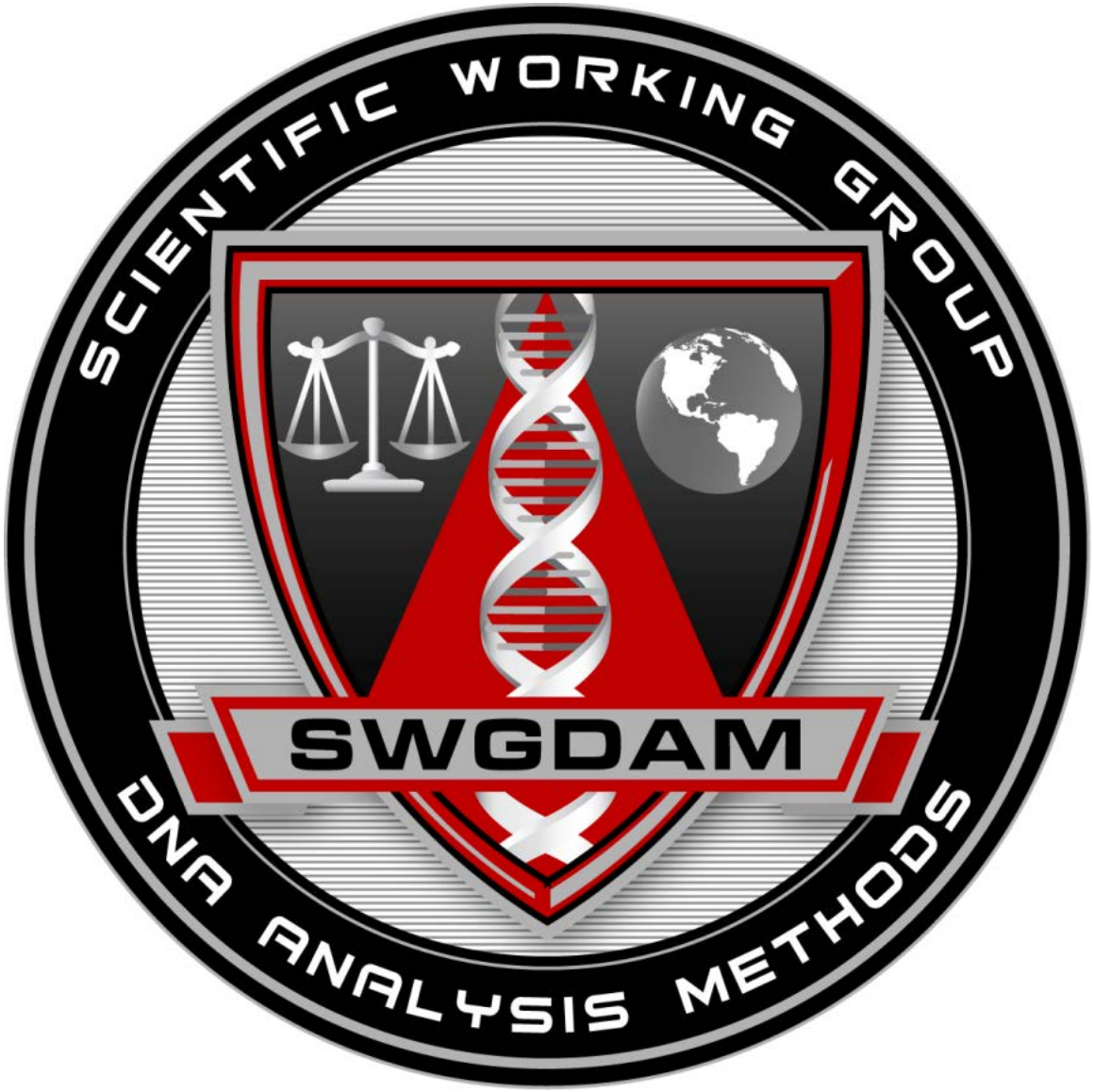
- CE User's Group Mixture Exercise (Dec 2008)
  - organized by Bruce Heidebrecht (MDSP) – involved ~50 analysts (16 labs)

- Training workshops/classes provided by John Butler (in the past year)
  - CODIS administrators closed session (Nov 2008)
  - AFDIL (Jan 2009)
  - Harris County, TX (Jan 2009)
  - NYC OCME (Mar 2009)
  - Wisconsin State Lab (May 2009)
  - Utah State Lab (May 2009)
  - Prosecutors at National Advocacy Center (May 2009)
  - New York BIO TWG (Jun 2009) – DNA tech leaders
  - Florida International University class (July 2009)
  - NWAFS (Sept 2009) – with Bruce Heidebrecht, Brian Burritt, Steven Myers
  - GWU graduate course (Oct 2009)
  - NERFI DNA Academy (Oct 2009)

Mixture Interpretation Committee

Committee Point of Contact:

- John Butler (chair) – john.butler@nist.gov
- Gary Sims (co-chair) – gary.sims@doj.ca.gov


SCIENTIFIC WORKING GROUP
SWGDAM
DNA ANALYSIS METHODS

Questions

Contact

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