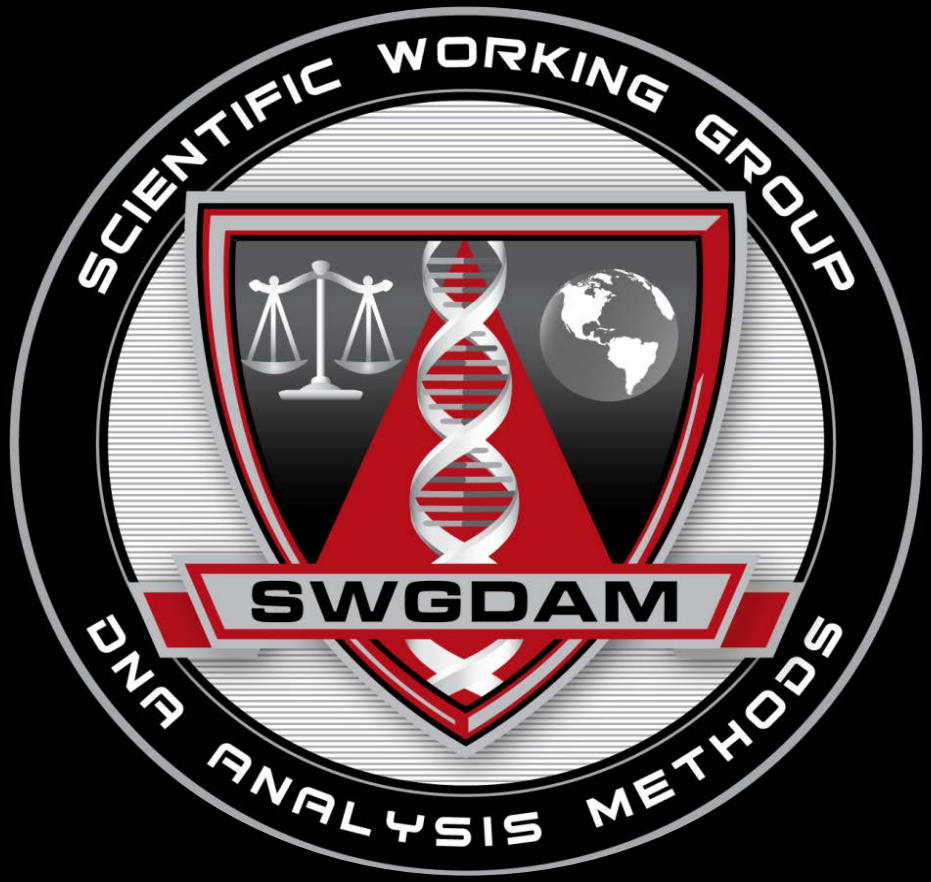
Revisions to the Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories



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Overview on Revisions

- Background
- Scope
- Highlights
- Timeline
- Future revision
- Acknowledgements

Scientific Working Group on DNA Analysis Methods Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories

Disclaimer – the revisions are still being finalized, so this presentation is simply to give an overview of what is coming

Background

- Soon after the 2010 Guidelines were approved and published, SWGDAM began working on revisions
- There have been a lot of events and issues raised since 2010
- These revisions are largely based on comments and questions sent to SWGDAM over the years from the community on these guidelines
  - What are guidelines and how should they be used?
  - Within many of the SWGDAM guidelines the statement is made that these guidelines are not intended to be used retroactively. What is the intent of this “retroactive” statement?
  - Are the 2010 SWGDAM Interpretation Guidelines applicable to all DNA mixtures?
  - When is it reasonable to assume on an item of evidence?
  - Should statistical calculations be conducted for every positive association?

Background

- There were also concerns that the 2010 guidelines were too vague
  - How does a laboratory go about validating these concepts?
  - More detail is needed with the guidelines in order to properly apply them in casework or incorporate them into an SOP

Background

- There is also the perpetual cycle we face in forensic DNA testing...
- …moving forward
  - Enhanced detection methods
  - Probabilistic genotyping
  - Rapid DNA technology
  - Next generation sequencing
- …while looking back
  - SOP changes to comply with new guidelines/revisions and having to address old or current cases
  - Example - CPI and the stochastic threshold

Scope

-excerpts from the new revisions

- These revisions specifically address the issues raised after the 2010 guidelines were published – they will supersede the 2010 Guidelines
- They like all other SWGDAM guideline documents are guidelines and not standards
- The FBI QAS and the QAS audit document have precedence over these guidelines
- These revisions are intended for using binary approaches to interpret electrophoresis-based data
  - RMP, CPI, and conventional LR
  - Interpreting this data using RFU for analytical and stochastic thresholds

Scope
-excerpts from the new revisions

- These new guidelines generally address the interpretation of single-source samples and mixtures of DNA from two people
- The basic concepts hold true as they relate to DNA mixtures of three or more contributors, those involving stochastic contributors, and with mixtures containing biologically related individuals
- However, there are nuances and limitations to binary interpretation of this more complex data which are addressed in these new revisions
  - Number of contributors
  - Additive effects of allele sharing
  - Stutter

Scope
-excerpts from the new revisions

- These revisions are intended to be applied prospectively and not retroactively
- With the underlying assumption that work (validation, training, analysis, interpretation) performed prior to the issuance of these revisions was appropriate and scientifically supportable, revision of the applicable guidelines is not intended to invalidate or call into question the previous work
- Laboratories are encouraged to review their standard operating procedures and validation data in light of these guidelines and to update their procedures as needed

Scope
-excerpts from the new revisions

- Only applicable to probabilistic genotyping, next generation sequencing, and/or rapid DNA technology in a limited capacity
- Why?
  - Most laboratories are still using binary interpretation in their casework
  - Much like Y-STRs, mtDNA, and enhanced detection methods, interpretation guidelines for next generation sequencing and rapid DNA will be addressed in other SWGDAM documents

Scope
-excerpts from the new revisions

- It is anticipated that these interpretation guidelines will evolve further with these new technologies and methodologies and be addressed in future revisions
- Keep in mind that many of the fundamental interpretation strategies for probabilistic genotyping are covered and still applicable with these current guidelines
  - Interpreting alleles versus artifacts
  - Stutter
  - Number of contributors
  - General considerations with making assumptions and formulating propositions with likelihood ratios

Highlights

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Sections are hyperlinked for quick path to desired topic of interest

Highlights

- What are the Core Elements?
  - Most notable guidelines from the document
  - What SWGDAM feels should be emphasized the most
  - These fundamental tenets are written as “shall” not “should”, since they are crucial for proper evaluation and interpretation of data to include determining conclusions and reporting results

Core Elements -examples

- “The primary goal of mixture interpretation shall be to determine the possible genotype combinations of the contributors.”
- “The interpretation of the evidentiary profile should determine the statistical approach used. It would be inappropriate to make inclusions or exclusions based on the statistical approach without first considering the interpretation of the profile.”
- Each core element is also hyperlinked to a particular section for further details.

Executive Summary

Core elements	Validation studies needed
Examine the profile and perform a quality assessment Core Elements I - VII (Section 1)	Analytical threshold Stochastic threshold
Categorize allele peaks Core Elements VIII - IX (Section 1)	Stutter thresholds Limit of linearity (off-scale data/pull-up)
Identify the minimum number of contributors to determine single source or mixture path Core Element X (Section 1)	Peak height ratio expectations
Mixture interpretation overview and strategies **The primary goal should be to determine all possible genotype combinations** Core Elements X1 - XIII (Section 2) • Assumptions • 2 person • >2 person • Major/minor • Known contributors • Potential stutter	Running applicable known mixtures (i.e., different contributor number, with different contributor proportions and template) to establish and assess protocols

Highlights

- It is important to note that the scope of this document is not intended to include validation, training, or reporting guidelines
- Validation strategies and examples are given in the document for illustrative purposes only, and are not meant to be all inclusive
- SWGDAM has other documents specific to these other areas for reference
- References and suggested readings are also listed at the end of this document which include additional resources for laboratories

Highlights

- Which binary statistical approach (RMP, conventional LR, or CPI) should my laboratory use?
- SWGDAM is not endorsing one model versus another in this document
- Instead, the intent is to illustrate the appropriate use and limitations for each model they currently use or intend to use

Timeline

- 2010-2015 – SWGDAM Autosomal STR Committee assigned to review and revise the document
  - In person meetings held twice/yr
  - Numerous conference calls and web-exes in between
  - Over a dozen SWGDAM members and invited guests involved from local, state, and federal labs to include input from ASCLD-LAB
  - Worked closely with OSAC liason to ensure document concordance
- Jan -Jul 2016 – reviewed and approved by the SWGDAM body to release for public comment
- Oct-Dec 2016 - the SWGDAM Autosomal STR Committee is currently reviewing the public comments and incorporating final changes
- Plan is to have a final version to SWGDAM at the January 2017 meeting for voting/approval

Future Revision

– to begin working on in 2017

- Guidance for interpreting data with Probablistic Genotyping software
- This will be an additional section added into the document

Acknowledgements

- Current Autosomal STR Committee
  - Joel Sutton (USACIL), Chair
  - Bruce Heidebrecht (MD), Co-chair
  - Jerrilyn Conway (FBI)
  - Sean Montpetit (San Diego PD)
  - Neil Fernando-Pulle (CFS-Toronto)
  - Russell Gettig (NYSP)
  - Tamyra Moretti (FBI)
  - Taylor Scott (FBI)

- Former members and invited guests (2011-2015)
  - John Butler (NIST) Former Chair
  - Gary Sims (Cal-DOJ), Former Co-chair
  - Tina Delgado (FBI)
  - Tim Zolandz (FBI)
  - Alan Giusti (FBI)
  - Cecilia Hageman (CFS)
  - Beth Ann Marne (PA)

Contact Information for SWGDAM

- Information and updates on any publications may be accessed on www.swgdam.org
- Comments and questions can also be uploaded to www.swgdam.org

Thank you for your time!

Questions?