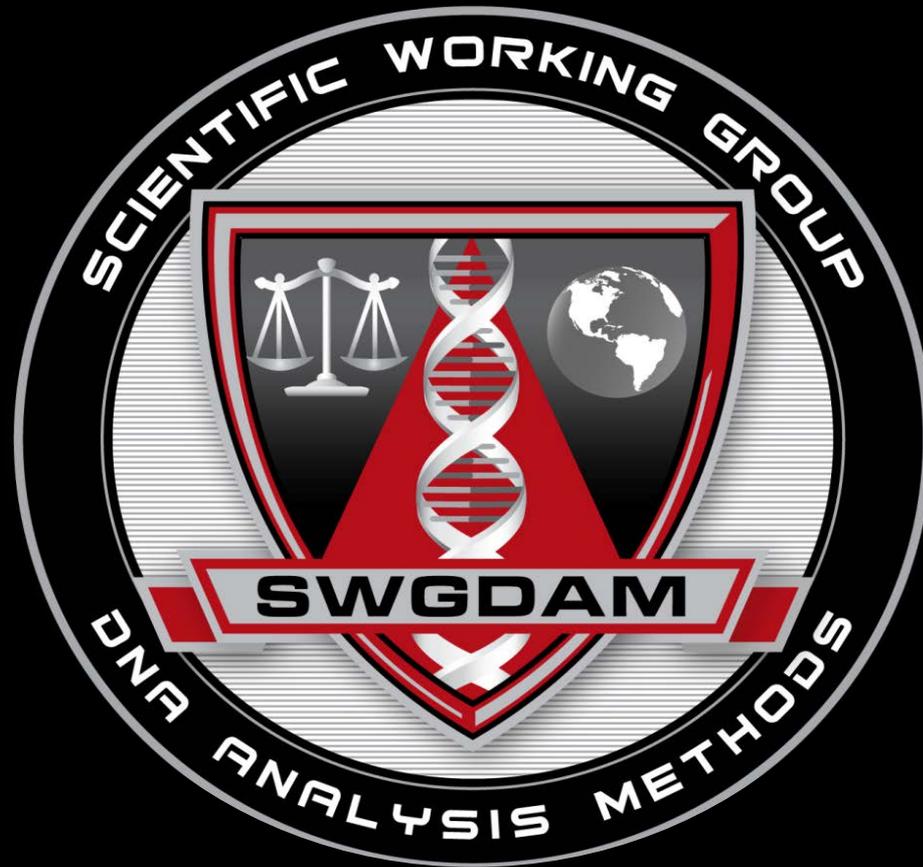


# SWGDM Update for ENFSI



**April 2011**



# SWGAM Update

Scientific Working Group on DNA Analysis Methods

## Mission:

- (1) to recommend revisions, as necessary, to the *Quality Assurance Standards for Forensic DNA Testing Laboratories* and the *Quality Assurance Standards for DNA Databasing Laboratories*;
- (2) to serve as a forum to discuss, share, and evaluate forensic biology methods, protocols, training, and research to enhance forensic biology services; and
- (3) to recommend and conduct research to develop and/or validate forensic biology methods.



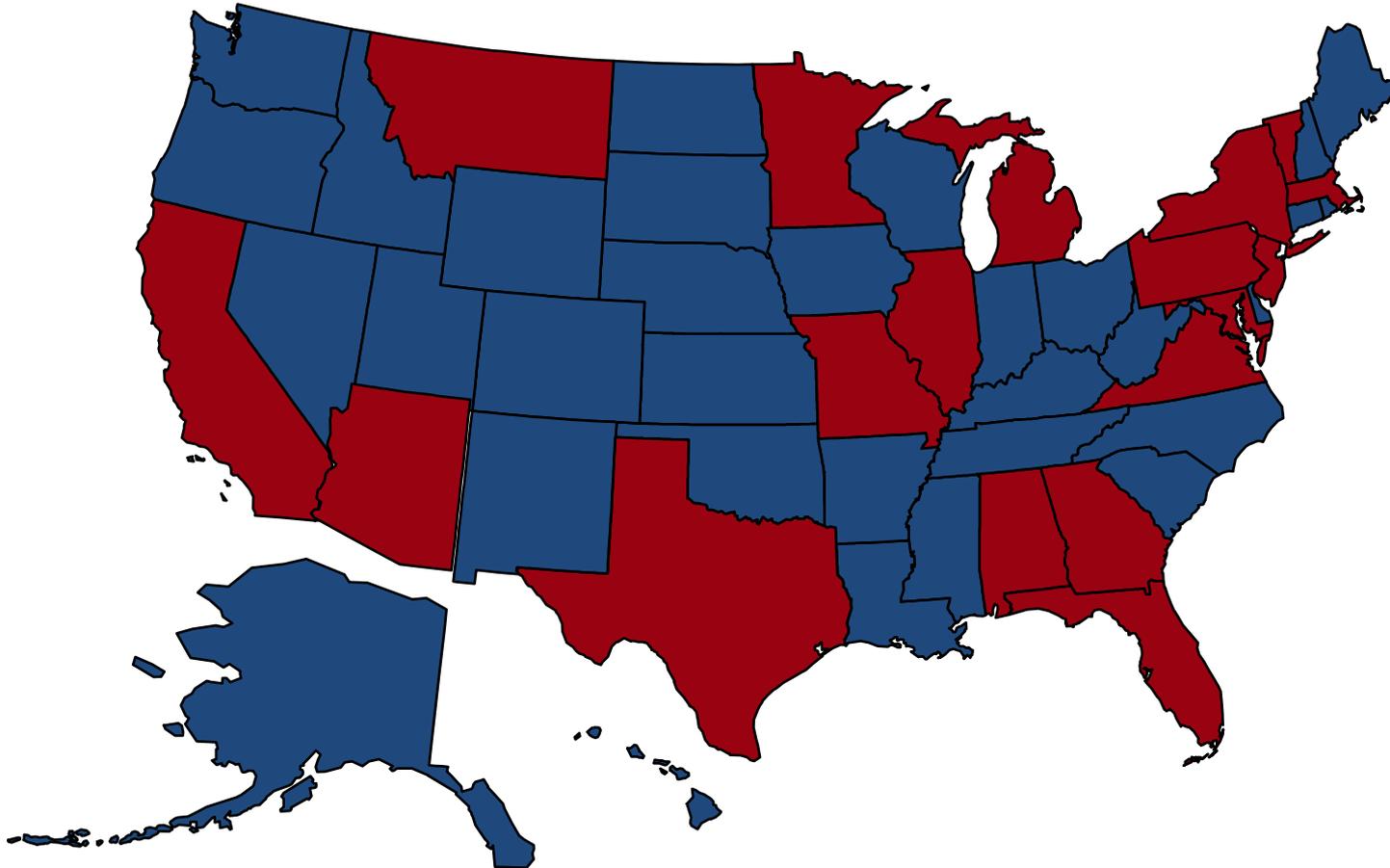
# SWGAM Update

## Membership

- Members are technical leaders and/or CODIS Administrators from local, state and federal crime laboratories
  - Membership is for 3 year term
  - Appointed by Chair
  - 19 Regular Members
  - 21 Invited Guests (Including ENFSI representative)
- Chair: Anthony Onorato (*appointed by FBI Director*)
- Vice Chair: Phil Kinsey (*appointed by Chair*)
- Executive Board: Angelo Della Manna, Eric Pokorak, Margaret Schwartz, Taylor Scott, Gary Sims



# Current SWGDAM Representation

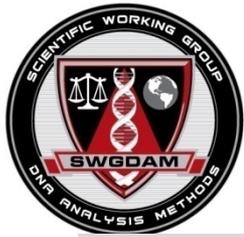




# SWGAM Update

## Committees

- CODIS: *Douglas Hares*
- EDM (*new name*): *Arthur Eisenberg and Eugene Lien*
- Mass Spectrometry/Mito: *Les McCurdy*
- Missing Persons & Mass Disaster: *John Planz*
- Mixture Interpretation: *John Butler*
- Rapid DNA: *Thomas Callaghan (acting)*
- Ad Hoc Working Group on Communication



# QAS Update

- FBI posted SWGDAM recommended revisions for the *Quality Assurance Standards on Forensic DNA Testing Laboratories* for public comment until March 1, 2011 at [www.fbi.gov/about-us/lab/codis](http://www.fbi.gov/about-us/lab/codis)
- Current QAS available at FBI web site at CODIS <http://www.fbi.gov/about-us/lab/codis>
- Current QAS also published in October 2008 issue of *Forensic Science Communications* at <http://www.fbi.gov/about-us/lab/forensic-science-communications>



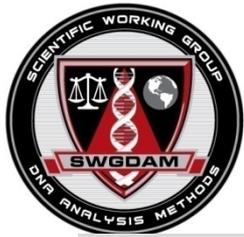
# Contamination Paper Published

- Published in July 2010 issue of FSI Genetics
- Gill, P., Rowland, D., Tully, G., Bastisch, Staples, T., Scott, P. (2010) **Manufacturer contamination of disposable plastic-ware and other reagents—An agreed position statement by ENFSI, SWGDAM and BSAG.** Forensic Science International: Genetics 4: 269–270



# Interpretation Guidelines

- **SWGDAM Interpretation Guidelines for Autosomal STR Typing by Forensic DNA Testing Laboratories**
- Available at FBI web site at <http://www.fbi.gov/about-us/lab/codis> (NEW)



# CODIS Bulletin on YSTR Issue

## **ATTENTION Y-STR Laboratories this is a bulletin from SWGDAM:**

For those laboratories using the database at [www.usystrdatabase.org](http://www.usystrdatabase.org) for calculating statistics for YSTR DNA analyses, please note that the method used for calculating the statistics has been changed.

Effective March 26, 2010, the database now calculates exact confidence intervals according to C.J. Clopper and E.S. Pearson, "The use of confidence or fiducial intervals illustrated in the case of the binominal" at *Biometrika* 26; p. 64-69 (1934). Additional information, including the exact formula used, can be found at the US Y-STR database site under "User Directions".

As a result of this change in the way the confidence interval is calculated, the results no longer reflect the interval calculation described in the published SWGDAM Y-STR Interpretation Guidelines, although the results are still consistent with the intent to provide an appropriate confidence interval.

SWGDM is in the process of reviewing this issue.



# SWGDM

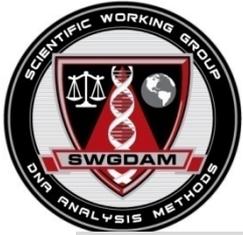
## Committees and Ad Hoc Working Group Updates



# CODIS Committee

## Mission:

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



# CODIS Committee

Tasks/objectives accomplished:

- Worked on CODIS match and rank disposition definitions for CODIS 7.0



# CODIS Committee

## Dispositions

**Arrestee Hit**

**Benchmark Match**

**Candidate Match**

**Conviction Match**

**Detainee Hit**

**Duplicate**

**Duplicate Match**

**Forensic Hit**

**ID Confirmed**

**ID Pending**

**Insufficient Data**

**Investigative Information**

**Legal Index Hit**

**Maternal Relatives**

**No Match**

**No Profile**

**Offender Duplicate**

**Offender Hit**

**Pending**

**Pending Local Disposition**

**Paternal Relatives**

**Requesting More References**

**Siblings**

**Twins**

**User Defined #1**

**User Defined #2**

**User Defined #3**

**Waiting for More Data**



# EDM Committee

- For the guidelines:
  - Include definitions such as “single source.”
  - Include general statements for extraction procedures and quantitation procedures. Start setting guidelines with amplification.
  - Include sample retention statement
  - Recommend using a sensitivity test as a “litmus test” to see whether or not a protocol should be considered an EDM.



# EDM Committee

- For the guidelines (continued):
  - Provide guidance on what must be addressed when validating an EDM. (Assessing stutter percentage, stochastic threshold, peak height imbalance, locus-to-locus imbalance, etc.)
  - QC Measures will be covered in document (gowning, cleanliness, etc)
  - Make a note that following manufacturer recommendations can still be EDM or LC/LT DNA Testing.



# EDM Committee

- For the guidelines (continued):
  - Conversely, NOT following manufacturer recommendations may NOT be EDM or LC/LT DNA Testing.
  - Include specific examples of EDMs



# EDM Committee

- **Enhanced Detection Methods** are those employed during or subsequent to the STR amplification step that increase the sensitivity of analysis and are typically employed with low quantity and/or low quality samples. These methods include, but are not limited to, increased cycle number, increased injection time, and post-amplification desalting. When using enhanced detection methods, the potential for stochastic effects (i.e., higher stutter, allele drop-out, and intra-locus peak imbalance) may increase. Stochastic effects can be addressed through appropriate interpretation guidelines and relevant thresholds (e.g., an increased injection time may require the adjustment of the stochastic threshold). Therefore, prior to any enhanced detection protocol being employed, which may include one or more enhanced detection methods, appropriate validations must be performed to address the potential increase in stochastic effects. Appropriate validations shall include assessments of stutter percentages, peak-height ratios, stochastic thresholds, locus-to-locus balances, and non-reproducible alleles.



# EDM Committee

- **Low Template or Low Copy DNA Analyses** are types of Enhanced Detection Methods that, in addition to the increased potential for stochastic effects, have an increased potential for non-reproducible alleles. Non-reproducible alleles shall only be identified through replicate analysis.



# Mass Spec/Mito Committee

- Mass spec
- Bridge gaps with technology provider...
  - Technical differences between T-5000 and Plex-ID
    - Increased observation of pp2923 dropout with Plex-ID
  - Ability to export data for sharing, comparison, discovery purposes
  - Security of data
  - Limited shelf-life of reagents



# Mass Spec/Mito Committee

- mtDNA
- 3500 concerns related to run modules and capillary lengths
- Clopper/Pearson:
  - More detailed discussions specific for mtDNA
  - Initiate comparison between Clopper/Pearson & current stats practices based on normal distribution assumption
    - Available macros?



# Missing Persons - Mass Disaster Committee

- Committee members developed and delivered an introductory training block on Missing Persons processes at the Fall 2010 National CODIS Conference in Salt Lake City UT
- Feedback indicated the presentations were very well received and it is expected that future, more advanced presentations will be requested supporting the deployment of CODIS 7.0



# Missing Persons - Mass Disaster Committee

- Suggestions for preparing a “Guidelines” style document to aid laboratories in establishing and interacting in missing persons activities within the CODIS umbrella were discussed at the conference
- Committee discussed the structure and content of such a work product



# Missing Persons - Mass Disaster Committee

- Committee outlined a three (3) part guidelines document plan to progressively cover the relevant issues laboratories need to consider in missing persons casework in conjunction with the new functionality provided with CODIS 7.0



# Missing Persons - Mass Disaster Committee

**Document 1:** Sample procedures  
(Reference sample vs Remains),  
Metadata, Client/Lab communications  
and resources

**Document 2:** Missing persons searches,  
Search statistics, association  
management and dispositions

**Document 3:** Missing persons reporting,  
Pedigree statistics, Information release



# Mixture Committee

- Help the community to better understand and implement the SWGDAM Autosomal STR Interpretation Guidelines
  - Request for training at Public SWGDAM (Nov 2010)
- Y-STR Guideline revisions incorporating Clopper-Pearson (section 5.3.1.2)
- Casework expert systems discussed



# Mixture Committee

- **Reviewed recent training materials & workshops on mixtures**
  - **Bruce Heidebrecht (voice-over slides)**, George Carmody, Steven Myers
  - Promega 2010 mixture workshop, AAFS 2011 workshop
- **Revision to Y-STR interpretation guidelines**
  - (5.3.1.2) Clopper-Pearson equation – **will impact mtDNA as well**
- **Discussed requests for advise on guidelines**
  - Email to Ted from San Diego Police Department on interpretation issues
  - Email to John from Oklahoma State Lab on setting thresholds
- **Discussed potential *Brady* issues from protocol changes**
  - Orange County, CA prosecutors concern over “retroactive” terminology



# Mixture Committee

<http://www.cstl.nist.gov/biotech/strbase/mixture.htm>

## Information on DNA Mixture Interpretation

### Workshops, Presentations, and Training Information

**Promega 2010 workshop  
(handouts ~500 slides)**

- [Mixture Interpretation: Principles, Protocols, and Practice](#) (Workshop: J.M. Butler, M.D. Coble, R.W. Cotton, C.M. Grgicak, C.J. Word, ISHI 2010)
- [SWGAM Autosomal STR Interpretation Guidelines](#) (Talk: John Butler, ISHI 2010) - [\[link to guidelines\]](#)
- [DNA Mixture Interpretation: Principles and Practice in Component Deconvolution and Statistical Analysis](#) (Workshop, AAFS 2008)

### Links to software programs or information

*TO BE DEVELOPED...*

**Plan to add information about  
GeneMapperID-X, FSS, USACIL,  
and TrueAllele software programs**

### Literature References on Elements of Mixture Interpretation

**>100 papers cited currently**

### Mixture Principles & Recommendations

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



# Mixture Committee

5.3.1.2. The haplotype has been observed in the database:

A conservative formula for calculating the upper 95 percent confidence limit in this case would be

$$\sum_{k=0}^x \binom{n}{k} p_0^k (1 - p_0)^{n-k} = 0.05$$

where  $n$  = database size,  $x$  = the number of observations of the haplotype in the database,  $k = 0, 1, 2, 3 \dots x$  observations, and  $p$  = the haplotype frequency at which  $x$  or fewer observations are expected to occur 5% of the time.

This cumulative binomial distribution formula is solved for  $p$  through serial iterations and therefore requires the use of a computer program.

[Add Clopper-Pearson \(1934\) to reference list](#)

Clopper CJ, Pearson ES. The use of confidence or fiducial limits illustrated in the case of the binomial. *Biometrika* 1934; 26:404–413.

**No other changes at this time – a future version 2.0 to update Y-STR mixtures info**

5.3.1.1. The haplotype has not been previously observed in the database:

The formula for calculating the upper 95 percent confidence limit in this case would be

$$1 - (0.05)^{1/n}$$

where  $n$  is the size of the database.

5.3.1.2. The haplotype has been observed in the database:

The formula for calculating the upper 95 percent confidence limit in this case would be

$$p + 1.96 \sqrt{\frac{(p)(1-p)}{n}}$$

where  $p$  is  $x/n$ ,  $n$  = database size, and  $x$  = the number of observations of the haplotype in the database.

5.4. For Y-STR mixtures that cannot be deconvoluted, calculations may be performed for the probability of exclusion and likelihood ratios.

5.5. If both autosomal and Y-STR data are collected on a sample, the product rule may be used to combine the autosomal STR genotype match probability and Y-STR haplotype frequency information.

5.6. It is recognized that population substructure exists for Y-STR haplotypes. Studies with current population databases have shown that the  $F_{ST}$  values are very small for most populations. Thus the use of the counting method that incorporates the upper-bound estimate of the count proportion offers an appropriate and conservative statistical approach to evaluating the probative value of a match.

## Current Y-STR Interpretation Guidelines

## Proposed replacement

These sections need work (a future v2.0) as well as further data and expert input



# Mixture Committee

- Continue to produce training materials
  - Training slides from committee members (Bruce Heidebrecht)
  - AAFS 2011 mixture workshop
- Validation assistance
  - Experimental design and thought behind setting thresholds
- Y-STR Interpretation Guidelines v2.0
  - Will require more data and input from additional experts
  - Provide further guidance on Y-STR mixtures
- Casework expert systems



# Rapid DNA Ad Hoc

**Sept 22 Biometric Consortium Conference, Tampa**

**Valone Briefing on 4 Channel Machine Evaluation (>60%)**

**Development status discussion of each initiative**

**QAS discussion**

**NCC Mtg**

**Nov 16 CODIS Conference**

**Recommendation for full SWGDAM Committee status**

**Failure to identify issues that would prevent QAS validation  
for NDIS Laboratory use**

**State Administrator Meeting Presentation**

**Vision Statement discussion**

**Jan '11 SWGDAM**

**Possible Committee membership**

**Discussion on establishing validation requirements**

**Committee Report Presentation**



# Rapid DNA Ad Hoc

<b>Taylor Scott</b>	<b>Illinois State Police</b>
<b>Chris Carney</b>	<b>FDLE</b>
<b>Peter Valone</b>	<b>NIST</b>
<b>Tom Callaghan</b>	<b>FBI</b>
<b>Valerie Evanoff</b>	<b>FBI</b>

**SDIS & LDIS Labs use R-DNA first, following demonstrated performance**  
**Single Source Buccal Swabs**  
**Mature System in NDIS Labs prior to Booking Suites (Phase Integration)**  
**Pilots:**

- 2 swab collection**
- Maximize data from each run (time and expense)**
- Phases for Pilots (5% QA, Confirm Hits, Drive Swabs, EX'r in BS)**
- TL coordination L/SDIS (Casework QAS)**

**Performance & Validation**

- ANDE Coordination (NIST Set of Samples)**
- Record of every sample run (Perm Machine Log)**
- Independent evaluation of ExpSys with CE data**
- New Artifacts**
- #of Unique samples determined after initial performance**  
**(1000 / 200 / 5 art)**
- Expert System Spike issue**



# Communication Ad Hoc

“Provide communication from SWGDAM based upon requests for information from the Interagency Working Groups, in addition to fostering improved communication and outreach to the DNA community.”



# Communication Ad Hoc

- Respond to Interagency Working Group on Education, Ethics, and Terminology
- Review existing SWGDAM publications
  - Training Guidelines
  - Validation Guidelines
  - Y-STR Guidelines

Make recommendations to Chair, if major changes are required.



# Contact Info

- Anthony Onorato
- SWGDAM Chair
- [Anthony.Onorato@ic.fbi.gov](mailto:Anthony.Onorato@ic.fbi.gov)