

FY 2012 Scientific Working Group Semi-Annual Report

SWG Name: Scientific Working Group on DNA Analysis Methods (SWGDM)

Meeting Date: January 17-19, 2012

Meeting Location: Marriott Residence Inn and Conference Center, Fredericksburg, Virginia

I. Agenda:

Please see Attachment A.

II. Meeting Synopsis:

Tuesday, January 17, 2012

SWGDM Chairman, Anthony Onorato, welcomed the attendees and announced that the next meeting would be held the week of July 9th, 2012. The Chair reviewed the tasks completed at the last meeting, including approval of amendments to the SWGDAM Bylaws and the Conflict of Interest Statement. The Chair requested that all SWGDAM Members and Invited Guests participating in SWGDAM Committees review and sign the Conflict of Interest Statements provided in their folders to the Executive Secretary.

The Chair also updated the attendees on the proceedings of the Scientific Working Groups' Chair meeting held in December, 2011: development of standardized bylaws for use by all Scientific Working Groups; establishment of a process for the review of all standards documents by standard setting organizations; and requirement for approval by the funding agency (National Institute of Standards and Technology) for any changes to the standardized bylaws.

The Chair also noted that much work had been done on the creation of a SWGDAM web site and invited the attendees to review the site at www.SWGDAM.org over the next few days and provide the Chair with comments or feedback on Thursday (January 19th). The Chair noted that additional sections were planned for information on previous meetings (meetings archives) and a public comment section that could be used to obtain input from the DNA community on SWGDAM draft documents.

The Chair introduced Dawn Herkenham for an update on the request for guidance for the manufacturers of consumables in participating in employee quality control DNA databases for contamination detection purposes. While the Genetic Information

Nondiscrimination Act of 2008 (GINA) provides an exception that permits employers that engage in DNA testing for law enforcement purposes as a forensic laboratory or for purposes of human remains identification to request or require genetic information from their employees to analyze DNA identification markers for quality control to detect sample contamination, this exception does not appear to include the manufacturers of consumables used by the forensic and missing person laboratories. Before proceeding to pursue the establishment of employee DNA databases for such purposes, SWGDAM voted at the July 2011 meeting to request a non-binding informal advisory opinion from the Equal Employment Opportunity Commission (EEOC) to determine if it is legally permissible for the manufacturing companies to obtain DNA samples from their employees for this purpose. A draft request was distributed to the Members and Invited Guests for review prior to the January meeting and this draft, with a minor revision, was unanimously approved by the membership to send to EEOC.

Following the review of the EEOC request letter, there was considerable discussion on the use of employee DNA samples for validation, population studies, and research purposes and whether such use was permissible under GINA. Ms. Herkenham noted that the forensic and missing person laboratory exception has been narrowly construed for quality control purposes to detect sample contamination and thus, it did not appear that these other purposes would be recognized as falling within the quality control purpose exception under GINA. Ms. Herkenham recommended that agencies make their legal representatives aware of this issue.

Following a theme initiated several meetings earlier at which there were presentations on the mixture tools that are currently available and being used by forensic laboratories, the Chair introduced Steven Myers of the California Department of Justice to describe his mixture deconvolution tool. Mr. Myers noted that his tool, "Mix Master," was designed to perform quantitative interpretation calculations for two person mixtures. He explained that his tool was intended to standardize approaches for addressing mixtures and facilitate the efficient preparation of documentation. Mr. Myers noted that "Mix Master" is currently not available outside of CA DOJ and any requests for the software program should be addressed to Gary Sims.

The Chair introduced the main topic for discussion relating to SWGDAM's examination of the most appropriate confidence interval to be used with lineage markers. The Chair noted that there will be presentations on the mtDNA and YSTR perspective as well as a presentation from Dr. Bruce Weir followed by questions and discussion.

Ms. Susan Cropp of the FBI Laboratory's MtDNA Unit described the calculation of confidence intervals for mtDNA haplotypes noting that the goal is to provide to the court an unbiased estimate of the true population frequency of a particular mitochondrial DNA haplotype. Ms. Cropp explained the calculations for each method (normal approximation and exact confidence interval) as well as how the two methods differ. Ms. Cropp also described the option of using a 1-tailed test or 2-tailed test. Ms. Cropp proposed that for lineage markers, that a 2-tailed exact confidence limit (Clopper & Pearson) (for population group sizes ≥ 100) be calculated, regardless of the number of observations in the database. Ms. Cropp also recommended there be renewed efforts to increase database size, including sequence range as well as the number of samples.

Dr. John Butler of the National Institute of Standards and Technology and Dr. Jack Ballantyne of the University of Central Florida presented issues on Y-STR profile frequency estimation. In response to an inquiry by Dr. Butler on the number of laboratories currently performing Y-STR testing, approximately one-quarter of the attendees indicated that their laboratories were conducting Y-STR testing. Dr. Butler described the work of the previous SWGDAM Y-STR Committee, including selection of Y markers and publication of Y-STR interpretation guidelines. Dr. Butler explained the differences between Y-STRs and mtDNA; the Y-STR loci and kits available; the two Y-STR databases: YHRD and USYSTR; and the approaches to profile frequency estimation. Dr. Jack Ballantyne provided detailed information on the USYSTR database, including contributors to the database and advantages/disadvantages of the two Y-STR databases. Dr. Butler concluded with predictions for the future of Y-STR analysis, including an example of "being able to put a lineage name to an unknown Y-STR profiles using on-line genetic genealogy information."

Dr. Bruce Weir of the University of Washington presented on haplotype frequency estimation. Dr. Weir distinguished the match probability (from the profile probability) as the probability that a randomly chosen person would have the profile given that it has been seen in the evidence. Dr. Weir suggested the use of "an exact upper confidence limit when n individuals have x copies of A between them." Dr. Weir explained calculations for Y-STR profiles, including the value of θ and theories advanced by , Warren Ewens and Charles Brenner.

There was additional discussion following Dr. Weir's presentation of the various formulae. Additionally, there was discussion of the Y-STR databases and in response to an inquiry, those attendees performing Y-STR testing indicated that the overwhelming majority of laboratories (16) use the USYSTR database with a few laboratories using the YHRD (2 laboratories) and Yfiler (1 laboratory) databases. Discussion continued regarding the use of the USYSTR database and how to communicate support for this database. Representatives of the National Institute of Justice (NIJ) attending the meeting on Tuesday noted that a letter in support of the database would be helpful. After the discussion, it was the general sense of SWGDAM to support the use of the USYSTR database and its continued growth and communicate this to NIJ. Preparation of a letter was referred to the Committees meeting jointly to discuss the lineage markers.

Dr. Walther Parson of the Institute of Legal Medicine presented an update on the European Mitochondrial DNA Population Database (EMPOP), a database of mtDNA control region haplotypes collected from around the world designed to provide population data on random samples to the forensic community. The EMPop database currently contains over 16,000 haplotypes. The haplotype submissions are rechecked before inclusion in the database and the data is also reviewed with each new release. Dr. Parson emphasized that regional sampling is important for the database.

The Chair noted that it would not be necessary for the Committees to provide an update on what they will be working on during this meeting as that would be covered in their presentations on Thursday. The SWGDAM Executive Board met on Tuesday evening to discuss pending requests for information and the need for additional Committees.

Wednesday, January 18, 2012

The following Committee meetings were held during the day:

CODIS Committee

Enhanced Detection Methods and Interpretation Committee

Mass Spectrometry/MtDNA Committee

Missing Person and Mass Disaster Committee

Mixture Committee

Quality Assurance Committee

Rapid DNA Committee

For a portion of Wednesday, the Mass Spectrometry/MtDNA and Missing Person and Mass Disaster Committees met jointly (along with most of the presenters from Tuesday) to discuss appropriate calculations for lineage markers.

A round table discussion was held in the evening and included a discussion of proficiency test issues. Members of the Proficiency Review Committee (Ann Marie Gross and Phil Kinsey) and the Chair of the NDIS Audit Review Panel (Jodi Dahl) were present to respond to questions from the attendees. Proficiency test issues were discussed relating to the interpretation of Standard 13.1.1 on manual and automated methodologies. The understanding of the group was that methods may be performed on individual samples and does not need to be performed for the entire proficiency test set.

Thursday, January 19, 2012

Committee meetings continued on Thursday morning and the general session resumed at noon.

The Chair introduced the Committee updates and each Committee provided an update of its progress toward the tasks and objectives.

Following the Committee updates, the Chair introduced Dr. John Butler for an update on the National Institute on Standards and Technology (NIST). Dr. Butler updated attendees on the following NIST projects: NIST SRM 2391c; Insertion/Deletion (InDel) Markers; Kit Concordance Comparisons; TrueAllele Mixture Software Evaluation; Rapid PCR and Rapid DNA Testing; ABI 3500 Validation Studies; Performance Assessment of PlexID; and Characterizing New STR Loci. Dr., Butler also described public criticism directed at the expanded CODIS core loci efforts and shared data relating to success rates of loci for profiles stored in Forensic Index of CODIS based on PCR product size. Additionally, Dr. Butler emphasized the importance of an expanded set of core loci to avoid adventitious hits on large and growing DNA databases; the limited “electrophoretic real-estate” in constructing STR multiplex assays for 5-dye instruments; the number of females in DNA database and missing persons cases for which Y-STRs will provide no information.

Dr. Douglas Hares, NDIS Custodian with the FBI Laboratory's CODIS Unit provided an update on the progress of the CODIS Core Loci Working Group. Dr. Hares described the creation of the CODIS Core Loci Working Group, the reasons necessitating expansion of the CODIS core loci and the criteria identified for evaluation of any proposed loci. Dr. Hares reviewed the proposed CODIS core loci and noted the announcement was published in the January issue of FSI: Genetics. Additionally, Dr. Hares stated that an addendum to the proposed core has been accepted for publication in FSI:Genetics and that it removes Penta D and Penta E from the listing of proposed CODIS core loci. Dr. Hares explained that the Working Group has been working on a timeline and implementation plan that includes the following phases: selection of laboratories to participate in validation studies; validation of proposed additional CODIS core loci; selection of CODIS core loci; and implementation of new CODIS core loci into NDIS operations. Dr. Hares noted that this plan will be published on the FBI's Internet site under CODIS in the next few weeks and invited attendees to provide him with any feedback or comments on the proposed plan and timeline.

Ms. Dawn Herkenham of SAIC provided an update on legislative and litigation. Ms. Herkenham updated attendees on the following: two states passed arrestee legislation in 2011 – Illinois and New Jersey – bringing the total to 27 States, the Federal government and Department of Defense as authorized to collect DNA samples from certain categories of arrestees; all 50 states are now authorized to collect DNA samples from all felony offenders with the passage of all felon legislation in Idaho; challenges to arrestee sample collection laws continue (*Shavlovsky*, *Mario O.*, *Thomas* and *Mitchell* decisions); a recap of familial searching legislation and one-house approval of a Pennsylvania bill to authorize modified DNA searches (SB 775); and an update on the Supreme Court's review of confrontation clause cases (*Williams v. Illinois*) as they relate to forensic reports and supporting testimony.

In closing, the Chair reminded attendees that the July meeting would be a full three day meeting as it appeared that a number of Committees were finalizing documents for consideration by the membership. The Chair also requested input on the SWGDAM web site and thanked everyone for all their work. The meeting was adjourned at approximately 2:20 p.m.

III. Attendees:

Please see Attachment B.

IV. Status Update on Progress toward Goals and Objectives:

The Committees continue work on their tasks for the next meeting in July, 2012 with some Committees planning to hold teleconferences in the intervening months.

V. Current and Planned Standards/Best Practices/Guidelines Development Activities:

The CODIS Committee continues to work on their tasks that do not, at this time, include standards or guidelines.

The Enhanced Detection Methods Committee has completed its work revising the SWGDAM Validation guidelines document and it be forwarded to the Chair for consideration before submission to the membership for review and comment.

The Mass Spectrometry/MtDNA Committee and Missing Person and Mass Disaster Committee completed an addendum to the Y-STR and MtDNA guidelines on calculation of a confidence interval to more appropriately reflect the degree of uncertainty surrounding the statistics and that addendum will be provided to the Chair for consideration before submission to the membership for review and comment. The Mass Spectrometry/MtDNA Committee is also working on the revision of mtDNA interpretation guidelines.

The Missing Persons and Mass Disaster and Mixture Committees continue to work on training documents for the DNA community.

The QA Committee has completed its substantive revisions to a 2001 SWGDAM Training guidelines document and will be finalizing formatting issues over the next few months.

The Rapid DNA Committee is working on validation and pilot test parameters for the R-DNA prototypes.

VI. Next Meeting:

The week of July 9th, 2012 has been confirmed for the next SWGDAM meeting.

VII. Additional Information:

SWGDM Semi-Annual Report

NIJ Interagency Agreement 2006-DN-R-094

None at this time.





SWGDM REGULAR MEETING

January 17 - 19, 2012

AGENDA

Tuesday, January 17, 2012

7:00 AM Chair and Vice Chair Meeting

8:00 AM Committee Chair Meeting

OPENING BUSINESS SESSION

9:00 AM Review of GINA Advisory Opinion Request Letter – Dawn Herkenham

9:30 AM Mixture Deconvolution Tool: MixMaster – Steven Myers

10:15 AM BREAK

TECHNICAL SESSION: FORENSIC HAPLOTYE TESTING

10:45 AM Current Methods for the Calculation of mtDNA Profile Frequency Estimates – Les McCurdy

11:30 AM Current Methods for the Calculation of Y-STR Profile Frequency Estimates – John Butler/Jack Ballantyne

12:15 PM Q & A Session – Les McCurdy, John Butler, and Jack Ballantyne

12:30 PM LUNCH

1:30 PM Haplotype Frequency Estimation

Dr. Bruce Weir
Chair and Professor of Biostatistics
Adjunct Professor of Genome Sciences
University of Washington

3:00 PM BREAK

3:30 PM Haplotype Frequency Calculation Panel Discussion – Les McCurdy, John Butler, Jack Ballantyne, and Bruce Weir



SCIENTIFIC WORKING GROUP

DNA ANALYSIS METHODS

Tuesday, January 17, 2012 (con't)

- 4:30 PM European Mitochondrial DNA Population Database (EMPOP) Update
Dr. Walther Parson
Professor
Institute of Legal Medicine
Innsbruck Medical University
- 5:15 PM Committee Kick-off
- 6:30 PM Executive Board Meeting

Wednesday, January 18, 2012

- 9:00 AM Committee Break-Out Sessions
- 12:00 PM LUNCH
- 1:30 PM Committee Break-Out Sessions
- 6:00 PM Round Table Session
- Informal Open Proficiency Testing Discussion: What's Actually Expected and Required by the QAS & PRC vs What's Perceived as Being Expected and Required by the QAS & PRC
- Facilitators: Phil Kinsey, Ann Gross, Jodi Dahl, and Cathy McCord

Thursday, January 19, 2012

- 8:30 AM Committee Breakout Sessions (con't)
- 11:00 AM LUNCH
- 12:00 PM Committee Session Updates
- CODIS – Doug Hares
- Enhanced Detection Methods – Eugene Lien
- Mass Spectrometry/mtDNAU – Les McCurdy
- Missing Person/Mass Disaster – John Tonkyn



SCIENTIFIC WORKING GROUP

DNA ANALYSIS METHODS

Mixture – John Butler

Quality Assurance – Jodi Dahl

Rapid DNA – Tom Callaghan

UPDATE SESSION

1:00 PM Legislative Update – Dawn Herkenham

NIST Update – John Butler

CODIS Core Loci Expansion Update – Doug Hares

CLOSING BUSINESS SESSION

2:00 PM Meeting Close-out

2:30 PM ADJOURN

**SWGDM January 2012 Meeting
Attendees**

SWGDM Chair

Anthony Onorato
FBI Laboratory

Todd Bille (IG)
Bureau of Alcohol, Tobacco and Firearms

SWGDM Vice-Chair

Philip Kinsey
Montana Forensic Science Division

John Butler (IG)
National Institute of Standards and
Technology

SWGDM Executive Board

Angelo Della Manna
Alabama Department of Forensic Sciences

Thomas Callaghan (IG)
FBI Laboratory

Eric Pokorak
FBI Laboratory

Theresa Caragine (IG)
Office of the Chief Medical Examiner of the
City of New York

Margaret (Peg) Schwartz
Vermont Forensic Laboratory

Chris Carney (IG)
FDLE – DNA Database

Taylor Scott
Illinois State Police

Amber Carr (IG)
FBI Laboratory

Gary Sims
California Department of Justice

Michael Coble (IG)
NIST

SWGDM Executive Secretary

Dawn Herkenham (IG)
SAIC

Jerrilyn Conway (IG)
FBI-NMRCFL

SWGDM Members and Invited Guests

Jack Ballantyne (IG)
University of Central Florida

Susan Cropp (IG)
FBI Laborator

Jodi Dahl (IG)
FBI Laboratory

Brian Hoey
Missouri State Highway Patrol
Crime Laboratory

Tina Delgado (IG)
FBI Laboratory

Deedra Highes (IG)
Mississippi Crime Lab

Neil Fernandopulle (IG)
Centre of Forensic Sciences

Clark Jaw (IG)
FBI Laboratory

Connie Fisher (IG)
FBI Laboratory

Elizabeth Johnson (IG)
USA CIL

Julie French (IG)
Michigan State Police

Ken Konzak
California Department of Justice

Russell Gettig (IG)
NY State Police Forensic Investigation
Center

Catherine Knutson (IG)
Minnesota Bureau of Criminal
Apprehension

Ann Marie Gross
Minnesota Bureau of Criminal
Apprehension

Sylvain Lalonde (IG)
Royal Canadian Mounted Police

Richard Guerrieri
FBI Laboratory

George Li
Virginia Department of Forensic Science

Douglas Hares (IG)
FBI Laboratory

Eugene Lien
NYC Office of the Chief Medical Examiner

Bruce Heidebrecht
Maryland State Police

Beth Ann Marne
Pennsylvania State Police

Cathy McCord
Texas Department of Public Safety

Gina Sola (IG)
FBI Laboratory

Amy McGuckian
Palm Beach Sheriff's Office

Joel Sutton (IG)
USA CIL

Jeff Modler (IG)
RCMP Forensic Laboratory

John Tonkyn
California Department of Justice

Shawn Montpetit (IG)
San Diego Police Department

Peter Vallone (IG)
National Institute of Standards and
Technology

Lilly Moreno (IG)
FBI Laboratory

Russell Vossbrink
Arizona Department of Public Safety

Tamyra Moretti (IG)
FBI Laboratory

Jennifer Wendel
FBI Laboratory

Steven Myers
California Department of Justice

Tim Zolandz (IG)
FBI Laboratory

Nicole Nicklow (IG)
FBI Laboratory

Jeffrey Nye (IG)
Michigan State Police – FSD

Dixie Peters (IG)
University of North Texas Health Science
Center

Juliet Rolando (IG)
FBI Laboratory