DNA Mixture Interpretation Principles: Observations from a NIST Scientific Foundation Review AAFS 2019 Workshop #10 (February 18, 2019; Baltimore, MD)

What does a Forensic DNA practitioner need to know?

Robin Cotton

Biomedical Forensic Sciences
Boston University School of Medicine
rwcotton@bu.edu





We have educational requirements from the QAS-

- The required academic background for technical leaders and DNA analysts is described in the QAS.
- The topics fall into the areas of:
 - Molecular Biology
 - Biochemistry
 - Genetics
 - Statistics and/or Population Genetics
 - In DNA analysis we mainly use the principles of population genetics for evaluation and calculation of allele, genotype or haplotypes frequencies.
 - While both are important, there are many additional applications of statistics in the analysis of validation data with applications to casework methods.

Other Questions from the QAS-

- Do laboratory personnel have the education, training, and experience commensurate with the examination and testimony provided?
- Does the laboratory's training program teach and assess the technical skills and knowledge required to perform DNA analysis?
- Does the laboratory have a documented program to ensure that technical qualifications are maintained through continuing education?

But what constitutes the knowledge needed? To some degree this depend on what you do-

- What are you responsibilities in the laboratory?
 - Your position-
 - New analyst
 - Experienced analyst
 - Technical leader
 - QA Manager
 - Other responsibilities-
 - Technical review
 - Maintain instrumentation
 - Assist with validation
 - Provide court testimony

What information do you need from the laboratory?

Documents that are directly related to the processes and procedures.

Needed from the laboratory-

- Procedural manuals for kits, instruments, software:
- Laboratory validation data (not just the summary)-
 - What was done (i.e. what questions were addressed)?
 - What data analysis was conducted and what were the conclusions?
 - What do the data analysis results tell you about the method you are using?
- Therefore, what are the limitations and capabilities of a method?
 - Sensitivity and specificity
 - Comparison with previous method(s) & data in literature
- Used in development of Laboratory SOPs
- Used as basis of calculations used in testing and/or reporting.

What other information do you need to keep up to date in the field?

Information from the scientific literature as well as text book and journal resources that relate to laboratory procedures.

Found in the scientific literature-

- Foundational articles for DNA testing (DNA structure and chemistry, PCR, STRs, DNA sequencing, etc.)
- Method development articles-
 - Basis of methods, description of chemistry and detection of signal
- Validation articles-
 - For method, kits or instrumentation validation (from lab, industry or academic sources)
- Articles reporting on application to or performance in casework.
- New developments and procedures
 - Reports of new methods
 - Comparison with currently accepted methods

The scientific literature is growing....

- American Journal of Human Genetics
- Croatian Medical Journal
- Electrophoresis
- Frontiers in Genetics
- Investigative Genetics
- International Research Journal of Computer Science
- Nature
- Plos ONE

- Forensic Science Int.
- Forensic Science Int. Genetics
- International Journal of Legal Medicine
- Journal of Forensic Sciences
- Law, Probability & Risk
- Legal Medicine
- International Journal of Legal Medicine
- Science & Justice

Up-to-date knowledge and data analysis skills are critical-

- Scientific knowledge allows us to understand the molecular biology processes which are the basis for-
 - DNA extraction methods and method choice
 - DNA quantification using qPCR
 - DNA amplification for STR analysis or for DNA sequencing
 - Electrophoresis and CE instrument capabilities
- Data analysis skills allow us to understand methods for-
 - Understanding the extent of normal variation in each process step
 - Analysis of variant results from controls and samples
 - Make informed decisions for planning and use of validation data for analysis and reporting
 - Make informed decisions regarding the application of statistics to analysis methods for reporting results

How does this knowledge facilitate testimony?

- Since the prosecutor asks the witness to make DNA "simple"; why is in-depth background knowledge needed?
- It's difficult to correctly simplify information if your understanding is limited.
- For example-
- Confidence in court, in part, comes from knowledge of the scientific literature.
- Confidence does not come from knowing what the SOP or a colleague "says", because that's memorization.
- Confidence comes from understanding why the specifics of the SOP impact the success of sample processing.
 And that in-depth understanding comes from reading the literature.



Workshop participants

Contact information for RWC

rwcotton@bu.edu

617-358-1188

