DNA

Ed Ungvarsky, Special Counsel to the Director,
Public Defender Service for the District of Columbia,
Washington, D.C.

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DUE DILIGENCE IN A DNA CASE
Edward J. Ungvarsky, July 3, 2006

1. Read *Champion* articles. Learn as much as you can in advance (esp. basic vocabulary).

2. Get **all** the files.
   a. Determine what kind of DNA testing was conducted (mitochondrial or nuclear).
   b. Send DNA discovery letter.

3. Review **all** the files.
   a. Keep a set of all pages in file in order it was given (bate-stamp).
   b. Consult with more DNA-experienced attorney to identify files and to identify what might be missing.

4. Get the raw data (information contained on CD-ROM)
   a. Make a copy of cd-rom; never work on original.

5. Send copy of CD-ROM and paperwork to expert for independent consultation.

6. Review **all** the files again and **view** all the evidence.
   a. Look for inconsistencies, typos, and indications of third parties in the findings.
   b. Read the analyst's notes – all the notes.

7. Compare what the lab did with their standard operating procedures and what has been scientifically validated.

8. Consult with more DNA-experienced attorney regarding what the lab has done in other cases (consistent or inconsistent) and what do other labs do (check the Government experts CV for "puffery" and lies).

9. **Interview the analyst.**

10. After completing steps 1-9, assess significance of DNA evidence.
    a. Motion to Exclude?
    b. Independent DNA testing (do it at all? of what? which lab?)?
    c. Trial strategy (theory; cross; defense expert)?

IDENTIFYING, LOCATING AND PREPARING EXPERTS

The following is an attempt to summarize suggestions about how to best locate and make effective use of experts. Much of this information was taken from an outline that Julia Leighton (Public Defender Service in Washington D.C.) and I prepared for a clinic in which we prepared experts to testify and attempted to educate experts about how to effectively work with criminal defense lawyers. This summary is not comprehensive and may not apply to all situations. It is intended to be used as a guide primarily for cases in which the expert will be the focus of the defense, but the suggestions are useful in any case in which you will call an expert to testify. Discovery-related issues and concerns regarding what to give your expert or what not to give your expert are beyond the scope of this article. This guide may also help you prepare to cross-examine a prosecution expert. Much of the content of this article comes from lessons learned through mistakes I have made.

The goal is to present effective expert testimony: testimony from an unbiased expert with specific expertise in the relevant subject. Expertise is demonstrated, in part, by an ability to talk knowledgeably about the research and the literature. Such an expert has not changed positions on the basis of the questions asked or the case, but only upon new research or further developments in the field.

I. Locating the Appropriate Expert

We have many resources available to us in this office. I maintain a list of experts that is always expanding and being updated. I also try to keep relevant information about experts that has been provided to me by lawyers in the office. There are also many other experienced, knowledgeable lawyers who have worked with numerous experts during their careers in the office. The PDNET, is a good place to go to seek information from the lawyers in our office. The internet is another good place to locate experts. However, keep in mind that the internet is a starting place. If you identify an expert from a search on the net, you should fully research the expert you locate. This includes "googling" the expert, running his or her name through LEXIS or WESTLAW to see if the expert is mentioned in any published decisions, or perhaps checking "Verdicts and Settlements" on Lexis to get names of lawyers who have used the expert. Finally, we are extremely lucky to live in Los Angeles, a city with a large number of universities, which are wonderful sources for experts.

II. Where to Begin Once You Have Located an Expert

I suggest that the first thing you do is obtain a CV (curriculum vitae) from the expert. Then you will need to vet the CV. What do I mean? Check out the expert's web page to see if it has any inappropriate material on it. Attempt to determine whether the expert has any past criminal convictions. Assess the expert's receipt of funding from the defense versus prosecution, business interests, academic appointments and grant sources. Assess the expert's objectivity as demonstrated by attendance at defense versus prosecution conferences, and assess the expert's organizational, academic and personal affiliations - defense versus prosecution. Finally, be prepared to discuss any problem areas with the expert when you meet

him/her.

Why take the above steps? In a courtroom, the relevant issue for a judge and for jurors is as much about bias as it is about science, and as much about appearance as it is about science. In a courtroom, it is all about credibility - which side is deemed more credible, which side is to be believed, which expert's testimony should be credited. Jurors and judges may not understand the science, but in a courtroom, they are the sole determiners of who to believe and who to credit. Any fact that tends to show that an expert might have a bias in favor of one view or one side is admissible evidence. In other words, the expert's past fee arrangements, business interests and organizational affiliations are potentially fair game for cross-examination. Expert witnesses are often viewed with skepticism by judges and juries. The perception that an expert witnesses' testimony may be purchased is a prevalent attitude among lay jurors. Keep in mind that an expert can be discredited during his or her testimony if opposing counsel (1) can expose an apparent bias by the expert for one side, (2) can expose a lack of expertise in a specific area, and/or (3) can expose unexplained or unexplainable changes in the expert's opinion over time.

III. Meet With the Expert

Before even considering having an expert appointed, meet with him/her in person. It is important to meet with the expert to be able to see what the expert looks like and how the expert relates to you. Most experts will be happy to give you 30-45 minutes of their time. This will give you an opportunity to see the expert and also to get an idea about what the expert thinks about your issue. Finally, you will be able to ask the expert if he/she has any "baggage" about which you should be aware. This should be done in every case and as delicately as possible.

IV. Preparing an Expert to Testify

Before calling an expert to testify, it is important to collect and review all relevant articles, book chapters, letters, abstracts and publicly-presented PowerPoints written by the expert, and to categorize them by field. Assess the expert's education, categorize it by field and ask the expert how many times he/she has been qualified in the area of expertise relevant to your case. Review the expert's CV for areas of expertise and gaps in expertise. Talk with the expert about the areas in which you want to qualify him/her as an expert, and ask him/her the types of questions that will be asked on voir dire by the opposing lawyer, if permitted.

You should review all the expert's articles related to the opinion he/she will render. Review prior transcripts related to the opinion to be rendered. Review all prior affidavits related to the opinion. Talk with the expert about the basis of his/her opinion. What data, scientific articles and/or scientific research supports the expert's view? Talk with the expert about which articles and what research contradicts his/her opinion. Why don't those articles or research affect his/her opinion?

It is important to only attempt to qualify the expert in areas in which he/she is "expert." A psychologist, John Briere, suggests: "Remember the acronym HELP which stands for honesty, evenhandedness, **limit expertise** and preparation." In order to effectively prepare

your expert to testify, you will need to know who will testify for the prosecution and who else you will call as an expert. Make sure to inform the expert of the areas that you will ask of the other experts.

There are a number of tasks the expert must do before he/she testifies. The expert should develop an opinion based on science - i.e. on research in the field of study and the views of credible peer-reviewed work by (and conversations with) others. The expert must know the literature and know what he/she has said in the past (the exact wording) regarding the relevant subject matter. If the expert's views have changed, the expert should tell you this and be prepared to explain the basis for the change in his/her opinion.

Any opinion should be grounded in good science. Good science should be published in the peer-reviewed literature and should be supported by other scientists. Very few experts are of sufficient reputation to be an "expert of one." Experts must know the literature and the research and be able to back up their opinions with those of other respected scientists, particularly those who all scientists would agree are world-renowned in the topic at issue.

Lawyers should explain and discuss the following with all experts regardless of their experience in testifying: The expert must know whether his/her involvement in the case is confidential, with whom the expert may discuss involvement in the case, issues relating to the case and what the expert's response should be if he/she receives a communication from anyone about the case. An expert may not present information pertaining to a particular case, even if the case is not specifically identified, at a meeting, in publications, to the media or on email listserves without explicit permission from the attorney. The lawyer should be prepared to answer the following questions: Can the expert communicate with the lawyer via email or in writing while working on this case? Should the expert prepare any reports, make notations? What materials can the expert rely on while testifying (notes, reports, etc.)? What materials should the expert bring to court? The expert should be informed that information should never be disclosed by him/her to the other side. The exchange of information should always be done by the lawyers. The expert should also be advised that before creating any documents, the expert should confer with the lawyer to ensure that such documents accurately and completely reflect the expert's opinion. (See "Nuts and Bolts" by Terri Towery in the August issue of the PDQ for a more detailed discussion of issues regarding "draft reports".)

You should explain and discuss with the expert the background, experience and practice of the particular judge hearing the case, the background, experience and practice of the prosecutor, and the make-up and sensibilities of the jurors.

You should explain leading versus direct questions and how to respond when objections are raised. You should also discuss how to respond if the questioner cuts the expert's answer off, and how to respond if the questioner does not permit the expert to explain an answer. Finally, you should have a discussion with your expert regarding attire, demeanor and the use of humor in the courtroom. While there are some occasions where humor may be appropriate, the expert must recognize that, in general, the court is not a place for humor, informal language or off-handed remarks.

Effective preparation takes time. Both the lawyer and the expert must plan accordingly. Assess how much time it will take the expert to complete his/her review of the information. Add several hours that will allow you to discuss the issues in the case with the expert. Add several more hours to go over the anticipated direct examination and to discuss likely lines of cross-examination. Be sure that the initial court order for appointment realistically anticipates this time. Obtain supplemental funds as needed and make sure that your experts does not exceed the funds provided by the orders. Be sure to assist your expert with billing and payment through PACE management. This will help assure that the expert will be willing to help you or another member of the office on a future case.

Reasonable Doubt in the Age of CSI: Experts and Science in the Courtroom

Edward J. Ungvarsky[1]

The nature of criminal defense practice is changing. Prosecutors are presenting ever-increasing amounts and types of scientific evidence in the courtroom. A reluctance to deal with such evidence is understandable. Few defense lawyers went to law school to be scientists. Most prefer to practice that which is comfortable, such as more traditional challenges to the perceptions and biases of civilian and police officer witnesses.

Nonetheless, scientific evidence exists, and, as a committed defense attorney, you must be prepared to confront it. When you hear the prosecutor say that the crime laboratory identified your client's DNA on the scene, you may recoil at having to deal with the science, with the word "plea" flashing through your mind. Yet, because the last thing anyone wants is to become a "plea lawyer," it is time to regroup. It is time to evaluate the scientific evidence with the same inquisitive and skeptical nature applied to all other evidence.

This article is intended to provide assistance in how to evaluate and challenge such evidence. The article discusses how to approach (A) the case where the prosecution fails to present scientific evidence and (B) the case where the prosecution presents scientific evidence that must be explained before the jury.

A. Reasonable Doubt Includes the Absence of Scientific Evidence

To start, it is important to observe that there are allies in the evaluation of scientific evidence: those twelve ordinary citizens sitting in the jury box who, like you, are questioning the prosecution's evidence, and, as is key here, the lack of evidence, thus holding the prosecution to its constitutional burden.

The fact that jurors now expect scientific evidence in criminal cases provides the defense a powerful tool.[2] When prosecutors fail to bring forward such evidence, you can argue that the failure to obtain and present such evidence undermines the prosecution's proof and provides a reason to doubt. Reminding the jury that it is the prosecution's heavy burden to prove its case beyond reasonable doubt, and that the absence of evidence indicates that it has not met its burden can be a powerful argument in a case based on circumstantial or testimonial evidence. In fact, these simple but compelling arguments are succeeding in courtrooms across the country.[3]

Not surprisingly, prosecutors are grumbling that they are not getting convictions where, in *their* opinions, the defendants are clearly guilty. They complain to the press that they should not be expected to bring forward all possible evidence, just the evidence they believe necessary to prove their case. They complain about the "CSI Effect," saying that jurors are holding them to a standard of science fiction, not demonstrable scientific facts.

Do not buy into this claim. Nobody is arguing science fiction: In weapons cases, fingerprints and DNA *can* be obtained from the handle of a gun. In vehicular homicide cases, the speed of the automobile *can* be estimated by skid and other tire markings. In drug cases, the precise chemical concentration of the residue on the crack pipe found in the client's jacket *can* be calculated and contrasted with the concentration of the rocks of cocaine found ten feet away from the client who is a user not a dealer.

Prosecutors cannot be allowed to turn the legitimate "CSI Effect" – jurors' proper awareness that law enforcement has scientific tools at its disposal and has elected not to present such evidence to the jury – into a prosecutorial argument that low expectations should be enough to meet constitutional requirements.

In making this claim, you need to be wary of an inclination in which judges side with the prosecution and erroneously limit questioning and summations. Judges sometimes incorrectly view such absence-of-evidence arguments as premised in supposition or as improper "missing evidence" arguments. You should correct such misunderstandings by explaining that you are not asking the jurors to draw an adverse inference from the absence of evidence equally available to both sides but rather are asking the jurors to hold the prosecution to its burden to prove its case beyond reasonable doubt, including providing physical and scientific evidence that corroborates the testimony of its witnesses.[4]

To make this claim effectively, you also must know the available forensic science. For example, if your client is charged with a shooting and is arrested shortly after the event without possession of a handgun, you need to know about the availability and effectiveness of gunpowder residue testing. With this knowledge, you can cross-examine the he arresting officer on the facts that such testing is available and was not used, and then argue that the absence of that evidence weakens the prosecution's case during summation. Here, the laboratory's own manuals, outlining their own best practices, can be powerful tools, with which you highlight to the jury that the prosecution's failure to comply with its own policies significantly weakens its case. Arguments about the absence of scientific evidence require as much preparation as arguments about the meaning of the results of scientific testing that was done. Therefore, from first assignment, think about the types of forensic evidence that might apply in your case and prepare to confront such evidence or argue its absence.

B. **Challenges to Expert Witnesses at Trial**

Prosecutors' more genuine response to the "CSI Effect" is reflected by their efforts to obtain scientific and forensic evidence with far greater frequency than even five years ago. This is where the instinct to flee science runs up against the passion for trial. The challenge becomes how we prepare yourself for a trial where scientific evidence is central to the case.

1. The Standard of Admissibility

The first step is to know the law of admissibility of scientific evidence. In interpreting Texas Rule of Criminal Procedure 702, Texas essentially has adopted the *Daubert*[5] test for determining the admissibility of scientific and other expert testimony.[6] The trial court acts as the gatekeeper to ensure that the proposed evidence is reliable and that the proposed expert is qualified to render an opinion. In making its reliability determination, the court considers a wide variety of factors.[7]

2. *Discovery*

The second step is to obtain full and complete discovery. Typically, the prosecutor only discloses a one or two page report, providing the expert's ultimate opinion and conclusion. This report is not full disclosure. Just as cross-examination of an eyewitness is ineffective without access to a witness' prior statements to the police and the grand jury, so too a competent cross-examination of an expert witness requires access to all his or her notes and reports. You must obtain every single note, recording, photograph, and computer file related to the expert's work on the case. This includes but is not limited to all correspondence between the expert and the prosecutor or the lead detective. If it is a DNA case, you also need the electronic data from the running of the software, typically handed over in CD-Rom form. You would not cross-examine about an out-of-court photographic identification procedure without copies of the photographs and some understanding of how the procedure was conducted. Similarly, in a case involving DNA or other forensic evidence, you need to know exactly how the expert conducted any forensic analysis in your case.

It is not easy to obtain full discovery. Some prosecutors and experts are unaware of their obligations and must be educated; others are simply unconcerned. Few, if any, have as complete an understanding of criminal discovery as the Constitution and rules demand. Do not be discouraged by their misunderstanding. Send carefully written, comprehensive discovery letters, serve detailed subpoenas, and submit probing Freedom of Information Act requests. Where compliance is lacking or limited, you should file timely motions to compel discovery and to enforce the subpoenas, grounded in the law that provides your right to the material. If your jurisdiction's courthouse practice affords minimal discovery, take advantage of the Houston Crime Lab scandal and other similar scandals across the country[8] and litigate for broader, fairer discovery.

3. *Laboratory Manuals, Protocols, and Quality Control/Assurance Procedures*

Discovery is not limited to materials connected directly with the analysis of the evidence. Whenever the prosecution presents evidence analyzed by a crime laboratory, obtain the laboratory protocols governing the type of testing done. Immediately request this information through discovery and by subpoena.

There are a number of benefits to this information. For one, use the manual to see if the prosecution's analyst followed the protocols, and, if not, cross-examine about the excuse for the deviation. Pointing out that an expert failed to follow protocols in his or her own

manual is particularly effective. For example, in the first DNA case that I tried, we established that the laboratory had ignored its protocols, with the result that the laboratory inappropriately neglected to include the presence of a third-party perpetrator's DNA on the scene.

Additionally, not all laboratory procedures are comprehensive or even adequate. If you compare your testing laboratory's procedures to those at other laboratories, you can demonstrate that the expert's laboratory has low expectations and minimal guidelines, which are hardly objective and scientific. Finally, because many experts have not read beyond their own guidelines, reading the manuals teaches you the extent of the expert's knowledge, which you can contrast with what you learn from other, more comprehensive sources, as discussed below.

4. Investigate the Expert

You must investigate the expert witness no less critically than you investigate a cooperating informant. Credibility is as much the linchpin of an expert's testimony as it is of cooperators or jailhouse informants. If you undermine credibility, you have demolished the opinion.

The first step in undermining the credibility of the expert is a careful reading and investigation of the expert's curriculum vitae, obtained as part of discovery. It is no secret that experts exaggerate and outright lie about their experiences and qualifications. Investigate every single line of the resume. Do not assume the truth of the representations. Did the pathologist really receive that medical degree? What were the expert's grades? Did the expert really take that continuing education class and how rigorous was it? Does the resume reflect the taking and passing of any proficiency tests? Are there standards to join the expert's impressive-sounding professional organizations or do annual dues payment suffice? I recently watched a trial where the defense lawyer had two different sets of resumes for the same expert. The resumes were written several years apart, and, as it turns out, the more recent one expanded a number of educational claims reported in the earlier resume. When all was said and done, the expert admitted that he had puffed, and his credibility was lost.

The more you know about an expert, the more you know how to approach him or her on cross-examination. The public record is full of information about experts. Get everything you can. Run a criminal records check. Run searches through Westlaw, Lexis, and other databases. "Google" the expert. When has the expert testified before, and what did he or she have to say? Perhaps the defense lawyers in such previous cases can provide some intelligence. Ask around about the expert to anyone you think might have useful information.

Experts are professional witnesses. You should seek to obtain transcripts of previous testimony. Transcripts are the best kind of expert investigation because they provide a preview of the expert's opinion and alert you to previously undisclosed surprises. When you obtain transcripts, you learn that the expert's opinion may have changed from case to

case. Impeaching a witness about an expert's change in an opinion goes a long way in undercutting the credibility of the expert's opinion. To a jury, science should not change, and such impeachments indicate that the expert's opinion is no more than a subjective interpretation guided by his or her allegiance to the prosecutor and to the prosecutor's view of the facts. Think about laying the transcripts out on your table before starting cross-examination for the expert to see. The mere threat of impeachment often leads to a far more compliant witness, willing to provide that desired string of affirmative answers in response to your questions.

5. *Interview the Expert*

In every single case you absolutely must interview the prosecution's expert face-to-face. You learn a wealth of information about the expert, the expert's opinion, and the bases of the opinion during that interview.

Do not worry about giving a preview of your case to the expert. You control the interview, and you control what information about yourself and your case that you will reveal to the expert. Further, do not worry if the expert refuses to meet with you. Scientists are supposed to be objective and even-handed, and an unwillingness to explain to you the analysis in advance of trial is fodder for effective bias cross-examination.

Treat the interview as a deposition, not as a preliminary cross-examination. The whole point is to gather information about the expert – how he or she will come across before your jury – and about the science. The interview is not a chance for you to show off how smart you are. The interview should be done in a non-confrontational manner. Get the expert to talk and keep him or her talking. The more the expert talks, the more you learn. The interview should feel like a classroom in which the expert is the teacher and you are the student. You get to show off what you have learned during cross-examination before the jury, when it matters.

6. *Investigate the Scientific Evidence*

It is critical that you get out of your office and investigate the forensic evidence. Go to the crime scene and view the physical evidence at the police property room. If the case involves an autopsy and time of death is an issue, take a trip out to the medical examiner's office to see how and where the bodies are stored when brought into the morgue. If the case involves the interview of a child complainant in a sex case, visit the hospital or advocacy center where the interview took place. Observe the physical environment and learn where the interviewer was sitting in relation to the complainant – perhaps the physical circumstances affected that child during the interview.

You also must read all reports, statements, and expert's notes with great care and attention. Look for inconsistencies. Look for ways in which the expert's opinion differs over time and from other evidence. For example, blood pattern experts may opine about the physical evidence at the crime scene in a way wildly inconsistent with how eyewitnesses claim an assault occurred.

7. *Investigate the Science*

You need to know the science as well as, if not better than, the prosecution's expert. You must control the witness, and to do so, you must be prepared to exercise control. By meeting with the expert, you will have learned everything that the expert knows about the scientific evidence. But you must know more. The internet is a terrific resource to learn about the different scientific disciplines. Use it as your first resource, and then follow up by reading relevant articles and treatises.

Obtain copies of the leading treatises in the discipline. It is not hard to get an expert to acknowledge on the witness stand a particular learned treatise as an authority. Upon doing so, go the next step and show how the prosecution's expert opinion conflicts with that of the treatise. During deliberations, the jury will likely listen to you and follow the admitted authority and reject the discredited testimony.

Besides reading as much as you can, talk to other lawyers familiar with the science. Attend forensic conferences[9] and participate in forensic evidence listservs.[10] Reach out and identify other defense lawyers who have dealt with the subject. For instance, the Forensic Evidence Committee of the National Association of Criminal Defense Lawyers (NACDL) has experienced attorneys a telephone call away ready to help you. Local attorneys in San Antonio and elsewhere in Texas serve the same role.

In response to the growing use of scientific evidence, NACDL and the National Legal Aid and Defender Association (NLADA) have created a partnership to sponsor an online Forensics Library -- an ongoing, comprehensive, national repository of defense-oriented forensic science information. The NLADA-NACDL Forensics Library, at www.nlada.org/Defender/forensics/, provides a forum for defense attorneys to share many kinds of materials. Each area contains subfolders with weblinks, model pleadings, research bibliographies and articles, expert transcripts and affidavits from defense and prosecution witnesses, relevant court opinions, scientific standards or best practices, and much more. It is a great place for preliminary research.[11]

8. *Retain a Defense Expert*

As a general rule of thumb, if the prosecution has an expert working on an aspect of the case, you should have your own independent expert as a consultant and, potentially, as a testifying witness. Think creatively and consult with experts whenever you identify an area of expertise that could inform the defense theory. Independent experts are invaluable because they educate you about relevant scientific concepts, assist in discovery requests, help you figure out what you need to find out from the prosecutor's expert pre-trial, assist in preparing your cross-examination of the prosecution's expert, present affidavits or testimony in admissibility challenges, and testify in the defense case.

The effective use of a defense expert merits an article of its own, but several points can be made in this context. First, identify an expert by referrals from colleagues and your

investigation of professional associations, universities, web searches, and review of the scientific literature. Second, be aware that your communications with your expert and your expert's notes are protected by attorney-client work product privilege only until you decide that you will call the expert at trial, at which time all those notes are fair game for disclosure. The best practice is to have separate experts for consulting and testifying purposes. If you are going to use the same person in both roles, explain each role and how you want the expert to memorialize his or her work. Finally, you should provide the expert as much information as necessary to form an opinion and remember that you, not the expert, are ultimately responsible for case strategy. Do not defer to your expert.

Conclusion

The "CSI Effect" creates both challenges and opportunities for defense lawyers. As science increasingly enters the courtroom, we must become increasingly comfortable with the science. Scientific evidence is no different than any other kind of evidence. Investigation and preparation remain the key ingredients to effective and successful representation of criminal defendants in cases involving such evidence.

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[1]Edward J. Ungvarsky received his B.A. from Wesleyan University and his J.D. from Yale Law School. After law school, Mr. Ungvarsky clerked for the Honorable Frank M. Johnson, Jr., on the U.S. Court of Appeals for the Eleventh Circuit. Mr. Ungvarsky then practiced criminal appellate and capital post-conviction law in Arizona and New York before joining the Trial Division of the Public Defender Service for the District of Columbia (PDSDC). Mr. Ungvarsky has since tried all levels of criminal cases. He is currently Special Counsel to the Director at PDSDC, where he directs PDSDC's Forensic Practice Group and has developed an expertise in the litigation of DNA, eyewitness identification, false confession, and other forensic evidence. He is a Vice-Chair of the National Association of Criminal Defense Lawyers' Forensic Evidence Committee and is an administrator of the NLADA/NACDL Forensics Library. Mr. Ungvarsky can be reached at eungvarsky@pdsdc.org or (202) 824-2301. © Edward J. Ungvarsky

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[2] Jamie Stockwell, *Defense, Prosecution Play to New "CSI" Savvy*, WASH. POST (May 22, 2005); Kit R. Roane & Dan Morrison, *The CSI Effect*, U.S. NEWS & WORLD REPORT (Apr. 25, 2005); Richard Willing, *"CSI Effect" Has Juries Wanting More Evidence*, USA TODAY (Aug. 8, 2004).

[3] *See* Andrew Blankstein & Jean Guccione, *The Blake Verdict and the "CSI Effect*," CHI. TRIB. (Mar. 22, 2005) (reporting observations that jury acquitted because prosecution failed to use forensic evidence to resolve reasonable doubts). *Contrast* Simon Cole & Rachel Dioso, *Do TV Shows Really Affect How Juries Vote? Let's Look at the Evidence*, WALL. ST. J. (May 13, 2005) (observing that television shows may make juries more inclined to convict because they falsely portray forensic evidence as unambiguous and certain).

[4] *See, e.g.*, *Bishop v. United States*, 107 F.2d 297, 303 (D.C. Cir. 1939) ("Reasonable doubt is doubt arising from the evidence or lack of evidence in the case."); *United States v. Iredia*, 866 F.2d 114, 117 (5th Cir. 1989) (error for prosecution to respond to defense's challenge to government's failure to produce handwriting expert by noting that "[the defense has] the opportunity to present evidence if they wish"). *See also Allen v. State*, 693 S.W.2d 380, 383-84 (Tex. Crim. App. 1984) (recognizing as proper defense argument that absence of fingerprinting can be reason to doubt); *Greer v. United States*, 697 A.2d 1207, 1210 (D.C. 1997) ("[I]in assessing whether the government has met its burden of proving guilt beyond a reasonable doubt, the jury may properly consider not only the evidence presented but also the lack of any evidence that the government, in the particular circumstances of the case, might reasonably be expected to present. For this reason, defense counsel may appropriately comment in closing argument on the failure of the government to present corroborative physical evidence.").

[5] *Daubert v. Merrill Dow Pharm., Inc.*, 509 U.S. 579 (1993). *See* F.R.E. 702 & 703.
[6] *E.I. du Pont de Nemours & Co., Inc. v. Robinson*, 923 S.W.2d 549 (Tex. 1995). *See also Kelly v. State*, 824 S.W.2d 568 (Tex. Crim. App. 1992) (pre-*Daubert* decision whose reasoning was largely adopted in *Robinson*).
[7] As set forth in *Robinson*:

> There are many factors that a trial court may consider in making the threshold determination of admissibility under Rule 702. These factors include, but are not limited to:
> (1) the extent to which the theory has been or can be tested;
> (2) the extent to which the technique relies upon the subjective interpretation of the expert;
> (3) whether the theory has been subjected to peer review and/or publication;
> (4) the technique's potential rate of error;
> (5) whether the underlying theory or technique has been generally accepted as valid by the relevant scientific community; and
> (6) the non-judicial uses which have been made of the theory or technique.

923 S.W.2d at 557 (citation and footnote omitted).
[8] *See, e.g.*, *Commission Created to Look Into Crime Labs*, HOUSTON CHRON. (June 20, 2005); Roma Khanna & Steve McVicker, *HPD Crime Lab Officials Resign to Avoid Firing*, HOUSTON CHRON. (June 12, 2003). *See also* Maurice Possley, Steve Mills & Flynn McRoberts, *Scandal Touches Even Elite Labs*, CHICAGO TRIB. (Oct. 21, 2004) (recounting problems with Virginia crime lab).
[9] For example, Forensic Bioinformatics, a private DNA consulting, is hosting its Fourth Annual Conference, The Science of DNA Profiling: An Expert Forum, on August 12-14, 2005. *See* http://www.bioforensics.com/conference05/index.html. PDSDC is sponsoring its Third Annual Forensic Science Conference, a DNA Cross-Examination College, on September 17, 2005. *See* http://www.pdsdc.org/Calendar/TrainingDetails.asp?id=19.
[10] NACDL and NLADA members should join the NACDL forensics listserv and eyewitness listserv. *See* http://www.nacdl.org/listserv. Numerous local listservs also exist.
[11] Other useful websites include http://www.texasdefender.org/expert%20witness%20directory.htm, http://www.corpus-delicti.com/forensic_fraud.html, http://www.bioforensics.com/kruglaw/, http://www.scientific.org/, and http://www.ncstl.org.

DATABASE LIMITATIONS ON THE EVIDENTIARY VALUE OF FORENSIC MITOCHONDRIAL DNA EVIDENCE

Frederika A. Kaestle, Ricky A. Kittles, Andrea L. Roth, & Edward J. Ungvarsky*

ABSTRACT: Mitochondrial DNA (mtDNA) typing is increasingly being offered in criminal jury trials as proof that the defendant is a possible contributor of DNA found at a crime scene. As a prerequisite to introducing such evidence, the prosecution typically must estimate the frequency in the general population of the mtDNA sequence found in the defendant and the crime scene so that jurors can evaluate the probative value of the defendant's inclusion as a potential contributor. The government estimates sequence frequencies by comparing the observed sequence to sequences listed in a racially categorized mtDNA database developed and maintained by the Federal Bureau of Investigation and the Scientific Working Group on DNA Evidence. While mtDNA evidence has significant potential as a law enforcement tool, the SWGDAM database is currently too small and insufficiently representative to provide meaningful estimates of sequence frequencies. Most importantly, the database fails to account for historic and recent human migration patterns that, because mtDNA is maternally inherited and not recombinant, have resulted in significant regional differences in sequence frequencies. With further sampling and study, large regional databases may prove to be an effective and feasible improvement upon the current forensic database for the calculation of meaningful frequency estimates. However, until such databases and meaningful frequency estimates exist, mtDNA evidence is not yet ready for admission in criminal cases to permit inferences that suspects left mtDNA at crime scenes.

INTRODUCTION

Although the public and the legal community are now familiar with nuclear

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* Dr. Frederika A. Kaestle is Assistant Professor of Anthropology at Indiana University in Bloomington, Indiana, and a Fellow of the Indiana University Institute of Molecular and Cellular Biology. Dr. Ricky A. Kittles is Associate Professor of Molecular Virology, Immunology and Medical Genetics, and of Anthropology at Ohio State University in Columbus, Ohio. Andrea L. Roth and Edward J. Ungvarsky are attorneys who specialize in forensic DNA evidence with the Public Defender Service for the District of Columbia in Washington, D.C. The views in this Article are those of the authors alone and do not represent those of the affiliate organizations.

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DNA, first admitted as evidence in United States courts almost twenty years ago,[1] nuclear DNA has a less famous counterpart found in the mitochondria of human cells, known as mitochondrial DNA ("mtDNA"). In recent years, law enforcement has increasingly used mtDNA evidence as a tool of both exclusion and inclusion of individuals as suspects in criminal cases. Specifically, because mtDNA exists in greater copy numbers per cell than nuclear DNA, mtDNA is becoming the primary type of forensic evidence extracted and reported from hair shaft samples and degraded DNA. A person may be excluded as a suspect if his mtDNA "profile," i.e., his mtDNA sequence in particular regions of his full mtDNA strand or "genome," differs from the profile of the crime scene sample. Such exclusion evidence may be relevant both at the pretrial investigation stage – to prevent wrongful accusation and conviction of innocent persons – and in post-conviction proceedings, such as Innocence Project DNA exonerations, to correct wrongful convictions. Use of mtDNA typing for exclusion purposes has been widely embraced in the scientific and legal communities because it preserves law enforcement resources by removing red herrings from investigations and focusing attention on the true perpetrator(s).[2]

As a tool of inclusion, however, mtDNA typing is more controversial. In criminal cases involving mtDNA, the prosecution typically reports that a defendant is included as a suspect if his mtDNA profile is consistent with, or "matches,"[3] the profile in a crime scene sample.[4] Most courts also require the prosecution to present an estimate of this shared mtDNA profile's frequency in the relevant population,[5] on grounds that, without such an estimate, jurors cannot meaningfully assess the probative value, if any, of the defendant's inclusion as a potential

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1. The first use of nuclear DNA in a criminal trial was in a Florida sexual assault trial in 1987. *See* NORAH RUDIN & KEITH INMAN, AN INTRODUCTION TO FORENSIC DNA EVIDENCE 186, 187 (2d ed. 2001) (discussing *Florida v. Andrews*).

2. *See, e.g.*, Max M. Houck & Bruce Budowle, *Correlation of Microscopic and Mitochondrial DNA Hair Comparisons*, 47 J. FORENSIC SCI. 1, 4 (2002) (noting that mtDNA has been used to exclude suspects who were originally included falsely based on microscopic hair analysis).

3. The term "match" in the context of forensic mtDNA typing is misleading because mtDNA is maternally inherited and nonrecombinant and therefore is not a unique identifier. *See* discussion *infra* at 58-99.

4. *See, e.g.*, DAVID L. FAIGMAN ET AL., 3 MODERN SCIENTIFIC EVIDENCE § 25-1.2.1 (2d ed. 2002) (listing criminal cases admitting mtDNA evidence against defendant). The federal government is in the process of greatly expanding its use of mtDNA typing in criminal cases. *See The FBI Selects 4 Regional MtDNA Laboratories*, 6 FORENSIC SCI. COMM. (Jan. 2004), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/jan2004/shortcomm/2004_01_short02.htm ("As laboratories become operational during the next two years, the effect will be to double the FBI's capacity to deliver no-cost mtDNA analysis to the criminal justice system.").

5. *See, e.g.*, United States v. Porter, 618 A.2d 629, 640 (D.C. 1992) ("[I]t is the probability feature which is at the very core of the DNA evidence."); United States v. Cuff, 37 F. Supp. 2d 279, 282 (S.D.N.Y. 1999) (same). *See generally* David H. Kaye & George F. Sensabaugh Jr., *Reference Guide on DNA Evidence*, *in* REFERENCE MANUAL ON SCIENTIFIC EVIDENCE 545 (2d ed. 2000) (citing cases for proposition that "many courts have held that a DNA match is inadmissible unless the expert attaches a scientifically valid number to the figure"); NATIONAL RESEARCH COUNCIL, THE EVALUATION OF FORENSIC DNA EVIDENCE 192 (1996) (discussing the statistical basis for interpretation) [hereinafter NRC II (1996)]; NATIONAL RESEARCH COUNCIL, DNA TECHNOLOGY IN FORENSIC SCIENCE 74-75 (1992) (discussing the meaning of "match") [hereinafter NRC I (1992)].

contributor. To estimate such frequencies, an analyst typically compares the suspect's mtDNA profile to a forensic reference database compiled and maintained by the Scientific Working Group on DNA Evidence ("SWGDAM"), a group sponsored by the Federal Bureau of Investigation ("FBI").[6] In comparing a suspect's sequence to the SWGDAM database, the analyst counts the number of times the sequence appears in various sub-databases organized by the self-reported "race"[7] of the sample contributors, then uses this number to estimate the true frequency of the sequence in each race-based population.[8] Because of the small size of these databases and the diversity of mtDNA profiles,[9] this approach usually yields zero "hits" in the database. Thus, the estimated frequency of the sequence reported out is often less than 0.1%.[10] Such a low estimate is potent evidence in a criminal jury trial because it suggests that a randomly selected individual has only a 1 in 1000 chance of sharing the profile observed in the crime scene sample.

Of course, a frequency estimate is only as good as the statistical method used to calculate it. If the method is invalid, such as if the database is not representative of the relevant population, the resulting frequency estimates may be inaccurate and, if so, will not give the jury a meaningful way to evaluate the probative value of the reported inclusion. In turn, if the probative value is unknown or inaccurately reported, most courts would rule that the evidence of inclusion is inadmissible based on rules of relevance and novel scientific evidence. Thus, whether the SWGDAM database is a valid tool to estimate mtDNA sequence frequencies is a critical question courts should ask in determining whether to admit evidence of mtDNA inclusions. Neither forensic scientists nor attorneys frequently present these questions to courts in a considered way. This Article is an attempt to

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6. *See Bylaws of the Scientific Working Group on DNA Analysis Methods*, 5 FORENSIC SCI. COMM. (Apr. 2003), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/april2003/swgdambylaws.htm (noting that FBI Director charged SWGDAM with reviewing DNA laboratory protocols, sets terms of SWGDAM members, receives SWGDAM recommendations, and provides SWGDAM with resources, including staff, travel and lodging budget). The version of the SWGDAM database currently used in forensic testing was made public in 1999. *See* Mark R. Wilson et al., *Further Discussion of the Consistent Treatment of Length Variants in the Human Mitochondrial DNA Control Region*, 4 FORENSIC SCI. COMM. (Oct. 2002), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/oct2002/wilson.htm.

7. While the SWGDAM database classifies individuals by "race," we believe that the more accurate classification for mtDNA profiles is by ancestry and use that terminology in discussing classifications in genetic lineages.

8. *See* Keith L. Monson et al., *The MtDNA Population Database: An Integrated Software and Database Resource for Forensic Comparison*, 4 FORENSIC SCI. COMM. (Apr. 2002), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/april2002/miller1.htm (explaining database comparison method); FBI Laboratory DNA Unit II, Mitochondrial DNA Sequencing Protocol § 11.4.1 (2004) [hereinafter FBI MtDNA Protocols (2004)] (explaining counting method used by FBI when comparing profile to database).

9. *See* discussion *infra* at 76-78.

10. Because the prosecution bases its frequency estimate solely on the number of "hits" in the database, the number reported to the jury in *any* case involving a mtDNA sequence not observed in the SWGDAM database, regardless of the geographical origin, ancestry, or other characteristic of the suspect, will be identical. *See* FBI MtDNA Protocols, *supra* note 8, at § 11.4.4 (stating FBI reports 95% confidence interval around number of "hits" in every case, without consideration of other variables, such as ancestry or geographical origin).

encourage the legal and scientific communities to view these questions with a more critical eye.

Part I of this Article briefly discusses the fundamentals of mtDNA biology and forensic typing methods and how they differ significantly from nuclear DNA biology and typing methods. Part II explains mtDNA's forensic applications and the particular methods used by the FBI to type mtDNA sequences and estimate sequence frequencies using population databases.

Part III contends that reported mtDNA frequency estimates are currently misleading because the SWGDAM database from which the estimates are calculated is neither representative of the general population nor of the various sub-populations it professes to characterize. First, the SWGDAM database is an incomplete, non-random, non-representative collection of mtDNA profiles compiled without regard to geographic patterns of genetic clustering that have resulted from cultural, political, historical, and economic forces. Second, current reliance in criminal cases on estimates derived from comparisons to the SWGDAM database is misplaced given the poor quality control measures of the database and the manner in which the assessment method is skewed toward reporting an inclusion.

Part IV examines both the general principles governing the admissibility of scientific evidence in most jurisdictions and courts' treatment of mtDNA inclusion evidence. Part IV argues that, because the estimation of profile frequencies using the SWGDAM database is currently controversial and of questionable validity, evidence of mtDNA inclusions does not yet meet most jurisdictions' legal standards for admissibility.

The Article concludes by prescribing various measures to improve the quality and integrity of forensic mtDNA typing. It is the expectation of the authors that, once mtDNA evidence is properly understood in its full scientific context, and once statistically valid databases can place a true probative value on mtDNA evidence, it will be reliable, highly relevant, and properly used in criminal investigations and prosecutions. Today, however, the state of mtDNA evidence presents an unacceptable risk of accusing or convicting the innocent based on inaccurate and misleading scientific evidence.

I. THE BASICS OF MTDNA TYPING AND HOW IT DIFFERS FROM NUCLEAR DNA TYPING

A. Differences in the Biology of MtDNA and Nuclear DNA

Deoxyribonucleic acid ("DNA") exists in every human cell and contains genetic codes inherited from previous generations. Humans have two types of DNA: nuclear DNA ("nDNA") and mtDNA. The mtDNA genome is distinct from the nDNA genome, and the two types of DNA differ in terms of their location within the body, genome size, and genetic makeup. While nDNA is bundled within chromosomes in the nucleus of most human cells, mtDNA exists outside the

nucleus in energy-producing organelles called mitochondria. The mtDNA genome is also much smaller than that of nuclear DNA; while the nuclear genome consists of approximately three billion base pairs,[11] the mtDNA genome contains approximately 16,569 base pairs.[12] The mtDNA genome consists of two primary regions: a *coding region*, which regulates the production of various biological molecules, and a *control region*, which regulates replication of the mtDNA molecule itself.[13] The control region, approximately 1125 base pairs long,[14] is the only significant portion of the mtDNA strand that does not code for genes.[15] In contrast, the nDNA genome contains coding regions spread throughout the twenty-three chromosomes that are known to have a genetic purpose, surrounded by regions of so-called "junk" nDNA, for which scientists have yet to find a genetic purpose.

B. Differences in Forensic Typing of MtDNA and Nuclear DNA

To distinguish one individual's DNA from that of another, forensic scientists look to particular locations within non-coding regions of the nuclear and mtDNA genomes that are highly variable among humans and therefore have discriminating power. In nuclear DNA typing, scientists typically look to thirteen locations along an individual's nDNA strand identified by the FBI as particularly suitable for forensic testing and used by the FBI to generate the profiles contained in its Combined DNA Index System (CODIS). A person's forensic nDNA "profile" consists of the twenty-six alleles he exhibits at these thirteen "CODIS loci."[16]

To compare individuals' mtDNA strands, most forensic scientists focus on two regions within the mtDNA control region – "Hypervariable Region I" ("HVI") and "Hypervariable Region II" ("HVII") – that together encompass approximately 610 base pairs and that exhibit high mutation rates and high amounts of variation from person to person.[17] A person's mtDNA "profile" consists of a list of the differences in HVI and HVII between that person's sequence and a reference sequence called the Cambridge Reference Sequence ("CRS") or "Anderson sequence," so named

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11. Base pairs consist of pairs of nucleotides that are bound to each other across the double helix of DNA (Adenine pairing with Thymine, Guanine pairing with Cytosine). The order of the base pairs encodes the genetic instructions.

12. *See* JOHN M. BUTLER, FORENSIC DNA TYPING, 242 (2d ed. 2005).

13. *Human Mitochondrial DNA – Amplification and Sequencing Standard Reference Materials* 1-2, NAT'L INST. OF STANDARDS AND TECH SPEC. PUB. NO. 260-155 (Sept. 2003).

14. *See* Stephen Anderson et al., *Sequence and Organization of the Human Mitochondrial Genome*, 290 NATURE 457, 457-65 (1981); Mitchell M. Holland & Thomas J. Parsons, *Mitochondrial DNA Sequence Analysis: Validation and Use for Forensic Casework*, 11 FORENSIC SCI. REV. 21, 24 (Feb. 1999).

15. Anderson et al., *supra* note 14, at 457; Thomas Parsons & Michael Coble, *Increasing the Forensic Discrimination of Mitochondrial DNA Testing Through Analysis of the Entire Mitochondrial DNA Genome*, 42 CROATIAN MED. J. 304, 304 (2001).

16. *See, e.g.*, Bruce Budowle et al., *Genotype Profiles for Six Population Groups at the 13 CODIS Short Tandem Repeat Core Loci and Other PCR-Based Loci*, 1 FORENSIC SCI. COMM. (July 1999), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/july1999/budowle.htm.

17. Holland & Parsons, *supra* note 14, at 24.

because the mtDNA genome of a particular individual, Stephen Anderson, was completely sequenced in 1981 by biochemist Fredrick Sanger in Cambridge, England.[18]

Scientists have begun to sequence nucleotides other than those in HVI and HVII in an effort to provide additional means of distinguishing different individuals' mtDNA.[19] They have focused, for instance, on a region referred to as "Hypervariable Region III" ("HVIII"),[20] also in the control region. In addition, even greater discrimination may be possible by typing certain nucleotides in the coding region called Single Nucleotide Polymorphisms ("SNPs"), single locations along the mtDNA genome exhibiting hypervariability.[21] Most forensic analysts do not currently type these additional locations along the mtDNA genome both because they know little about the variation outside HVI and HVII and because of convenience and cost concerns.[22] Exceptions exist, however; the Armed Forces DNA Identification Laboratory ("AFDIL") is currently attempting to detect levels of variation throughout the entire mtDNA genome for forensic purposes.[23]

MtDNA typing also differs significantly from nDNA typing because of the manner in which mtDNA is inherited. A child inherits twenty-three chromosomes from both of his parents; each set of chromosomes contains a full complement of nDNA strands with 3.2 billion base pairs. Thus, at each location along the nDNA

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18. Anderson et al., *supra* note 14, at 457; Richard M. Andrews et al., *Reanalysis and Revision of the Cambridge Reference Sequence for Human Mitochondrial DNA*, 23 NATURE GENETICS 147 (Oct. 1999).

19. *See, e.g.*, Carla Bini & Stefania Ceccardi et al., *Different Informativeness of Three Hypervariable Mitochondrial DNA Regions in the Population of Bologna (Italy)*, 135 FORENSIC SCI. INT'L 48 (2003); Sabine Lutz & Holger Wittig et al., *Is It Possible to Differentiate MtDNA By Means of HVIII In Samples That Cannot Be Distinguished By Sequencing the HVI and HVII Regions?*, 113 FORENSIC SCI. INT'L 97 (2000).

20. Lutz & Wittig, *supra* note 19, at 97.

21. *See* Michael D. Coble et al., *Single Nucleotide Polymorphisms Over the Entire MtDNA Genome that Increase the Power of Forensic Testing in Caucasians*, 118 INT'L J. LEGAL MED. 137, 143-44 (2004) (discussing typing of SNPs, including position 16519, which is outside of HVI and HVII and has greatest variability in entire mtDNA genome); Luisa Pereira et al., *Evaluating the Forensic Informativeness of MtDNA Haplogroup H Sub-Typing on a Eurasian Scale*, FORENSIC SCI. INT'L, *available at* http://www.sciencedirect.com (2005) (discriminating otherwise identical haplotypes by sequencing eight coding region SNPs); Yong-Gang Yao et al., *Phylogeographic Differentiation of Mitochondrial DNA in Han Chinese*, 70 AM. J. HUM. GENETICS 635, 648 (2002) (noting that "[c]oding region information is indispensable for phylogenetic analysis of mtDNA").

22. *See* Coble et al., *supra* note 21, at 143-44 (discussing how further typing of regions outside HVI and HVII would require development of new databases and other costly and laborious efforts).

23. Parsons & Coble, *supra* note 15, at 305. *See also* Coble et al., *supra* note 21, at 137 (discussing the study about sequencing entire mtDNA genome to increase forensic discrimination); Rebecca S. Just et al., *Toward Increased Utility of MtDNA in Forensic Identifications*, 146S FORENSIC SCI. INT'L S147 (2004) (discussing preliminary results of large-scale databasing project targeting populations underrepresented in current forensic mtDNA databases); Thomas J. Parsons et al., Report for U.S. Dep't of Justice, Office of Justice Programs, Progress of Project 2000-IJ-CX-K010, Homogeneous Fluorescent PCR Assays over the MtDNA Genome, up to June 30, 2005, provided in response to OJP FOIA No. 05-00258 (on file with authors) (explaining that, from 2000 to 2005, AFDIL has sequenced hundreds of full mtDNA genomes in the African-American, Hispanic, and Central Asian populations in an effort to better account for documented regional variation in mtDNA sequence frequencies).

strand, each individual has two genetic forms or "alleles" – one from each parent.[24] Of course, the parents each have two alleles at each location as well; which one of these two alleles each parent passes on is random. At each of the thirteen locations along the human nDNA genome used in forensic testing, only a limited number of possible alleles have been observed. As recombination occurs with each successive generation, different combinations of alleles are created over time. In contrast, scientists generally believe that the human mtDNA genome is only passed on from mother to child.[25] Consequently, all biological children of one woman will, absent mutations, have identical mtDNA profiles, and, going back generations, *all* relatives within the maternal lineage, absent mutations, will share the same mtDNA sequence.

While mtDNA's lack of recombination makes mtDNA sequences relatively static compared to nDNA, mtDNA exhibits a high rate of mutation between generations. Some regions of mtDNA evolve at rates five to ten times faster than single-copy nuclear genes.[26] Consequently, while each member of a maternal line should theoretically exhibit identical mtDNA profiles, the high mutation rate of mtDNA means that the profiles of members of the same maternal line, particularly over generations, may be slightly different.

Individuals are "homoplasmic" with respect to their nuclear DNA profile, meaning that all nuclear DNA strands found in a person's body contain the identical genetic material and do not differ from cell to cell. As recently as ten years ago, most scientists considered the vast majority of individuals to be "homoplasmic" with respect to mtDNA as well.[27] Scientists now understand, however, that most, if not all, individuals are actually "heteroplasmic" with respect to mtDNA, meaning that an individual's mtDNA sequence can differ among

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24. BUTLER, *supra* note 12, at 20.

25. *See* D. Andrew Merriweather & Frederika A. Kaestle, *Mitochondrial Recombination? (Continued)*, 285 SCI. 835 (1999) (concluding mtDNA is maternally inherited and finding no evidence for mtDNA recombination in humans). Other scientists have reported observations of "recombination" – mixing between maternal and paternal mtDNA in offspring. *See* Adam Eyre-Walker, Noel Smith & John Maynard Smith, *How Clonal Are Human Mitochondria?*, 266 PROC.: BIOLOGICAL SCI. 477 (1999). These observations are disputed. *See, e.g.*, Joanna L. Elson et al., *Analysis of European MtDNAs for Recombination*, 68 AM. J. HUM. GENETICS 145, 145 (2001) (disagreeing with Eyre-Walker results and concluding "that there is no compelling reason to overturn the standard paradigm of clonal mtDNA transmission in humans"); Peter Forster, *To Err Is Human,* 67 ANNALS OF HUM. GENETICS 2 (2003) (same). There have been some observations of paternal inheritance, *see* Marianne Schwartz & John Vissing, *Paternal Inheritance of Mitochondrial DNA*, 347 NEW ENGLAND J. OF MED. 576, 579 (Aug. 22, 2002) (observing paternal inheritance of pathogenic mtDNA), but the phenomenon of paternal inheritance is an exception, if not a well understood one, to the general rule of maternal inheritance.

26. Bruce Budowle et al., *Forensics and Mitochondrial DNA: Applications, Debates, and Foundations*, 4 ANN. REV. GENOMICS & HUM. GENETICS 119, 121 (2003).

27. *See id.* at 128 ("A decade ago, most individuals were thought to be homoplasmic."); Terry Melton, *Mitochondrial DNA Heteroplasmy*, 16 FORENSIC SCI. REV. 1, 3 (Jan. 2004).

locations in the body, or even within the same cell.[28] Although heteroplasmy is routinely observed, its causes are not fully known.[29] The chance of detecting heteroplasmy depends on the sequencing chemistry and techniques used.[30] Some body tissues, such as hairs, tend to show more variability in mtDNA sequence than others.[31]

Heteroplasmy and high mutation rates complicate forensic mtDNA analysis in two respects. On the one hand, samples from a suspect and a crime scene may, because of heteroplasmy, exhibit mtDNA sequence differences even when the two are, in fact, from the same individual or lineage, thus leading to potentially false exclusions. On the other hand, the suspect and crime scene samples may exhibit sequence commonalities even when the two are, in fact, from different individuals.

One other important difference in the state of current nDNA and mtDNA typing is the existence of population databases from which accurate frequency estimates can be generated. Scientists have conducted many population studies to generate population frequency estimates for nDNA and have reached some agreement that, using modifications to account for population inter-relatedness, reliable frequency estimates are possible.[32] In stark contrast, the frequency and distribution of mtDNA sequences in the population are not yet known. These population substructure issues with respect to mtDNA are the focus of nascent, but already quite active, scholarship among genetic anthropologists and forensic scientists.[33]

What *is* known about the frequency of mtDNA profiles in the population suggests that, unlike nuclear DNA, mtDNA profiles are far from randomly distributed. Groups of people with similar mtDNA within a circumscribed range of variation are called "haplogroups"; variation within a haplogroup divides people into "haplotypes," or particular mtDNA sequences. Researchers have named haplogroups observed in certain populations, based on the presence of certain

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28. Walter Bär et al., *Guidelines for Mitochondrial DNA Typing*, 79 VOX SANGUINIS 121, 122 (2000) ("[I]t is now thought that all individuals are heteroplasmic at some level."); Melton, *supra* note 27, at 2 ("[I]t is also certain that some degree of heteroplasmy exists in all individuals.").

29. *See* John Buckleton, Simon Walsh, & Sallyann Harbison, *Nonautosomal Forensic Markers*, *in* JOHN BUCKLETON, CHRISTOPHER M. TRIGGS & SIMON J. WALSH, EDS., FORENSIC DNA EVIDENCE INTERPRETATION 303 (2005) (discussing various theories); Peter D'Eustachio, *High Levels of Mitochondrial DNA Heteroplasmy in Human Hairs by Budowle et al.*, 130 FORENSIC SCI. INT'L 63, 63 (2002) ("Major unresolved issues include the molecular mechanisms responsible for the occurrence of heteroplasmy to different extents in different tissues, and the possibility that heteroplasmy levels in an individual might vary with age.").

30. Buckleton et al., *supra* note 29, at 304 (stating routine-sequencing methods cannot detect heteroplasmy above sequencing background noise unless it approaches 20%).

31. Mark R. Wilson et al., *A Family Exhibiting Heteroplasmy in the Human Mitochondrial DNA Control Region Reveals Both Somatic Mosaicism and Pronounced Segregation of Mitotypes*, 100 HUM. GENETICS 167, 167 (1997).

32. The NRC's 1992 and 1996 reports on forensic DNA typing both contain lengthy discussions of population substructure in the database used by the FBI to generate nDNA frequency tables and, using the product rule, a "random match probability" over the thirteen locations used in forensic nDNA testing. NRC II (1996), *supra* note 5, at 122-23; NRC I (1992), *supra* note 5, at 74-77.

33. *See* discussion *infra* at III.

combinations of variations.[34] For example, ten mtDNA haplogroups have been identified at significant frequencies in the European and U.S. European-American sub-populations.[35]

II. THE USE OF FORENSIC MTDNA TESTING AS A TOOL OF INCLUSION IN CRIMINAL TRIALS

To be clear, mtDNA typing is used as a tool of identification in many fields unrelated to inculpation of suspects in criminal trials. Because mtDNA is maternally inherited and found in higher copy number than nDNA, mtDNA analysis is particularly helpful in conducting population studies for medical,[36] genealogical,[37] and anthropological purposes,[38] and has been used by the military to identify casualties of war and terrorism.[39] For example, through mtDNA analysis, scientists have been able to identify victim remains from the World Trade Center tragedy, the Oklahoma City bombing, the Bosnian War, natural disasters, and plane crashes.[40] The demonstrated utility of mtDNA testing in these contexts stems

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34. Marc W. Allard et al., *Characterization of the Caucasian Haplogroups Present in the SWGDAM Forensic MtDNA Dataset for 1771 Human Control Region Sequence*, 47 J. FORENSIC SCI. 1215, 1219 (2002).

35. *Id. See also* Sarah A. Tishkoff et al., *Genetic Analysis of African Populations: Human Evolution and Complex Disease*, 3 NATURE REVIEWS - GENETICS 611 (2002).

36. *See, e.g.*, Douglas C. Wallace, *Mitochondrial Disease in Man and Mouse*, 283 SCI. 1482, 1482 (1999).

37. *See, e.g.*, Mark Shriver & Rick Kittles, *Genetic Ancestry and the Search for Personalized Genetic Histories*, 5 NATURE REVIEWS - GENETICS 611, 611 (2004); Anne C. Stone, James E. Starrs, & Mark Stoneking, *Mitochondrial DNA Analysis of the Presumptive Remains of Jesse James*, 46 J. FORENSIC SCI. 173, 173 (2001) (using mtDNA to determine if particular remains could be those of Jesse James); Lev A. Zhivotovsky, *Recognition of the Remains of Tsar Nicolas II and His Family: A Case of Premature Identification?*, 26 ANNALS HUM. BIOLOGY 569, 569 (1999).

38. *See, e.g.*, Rebecca L. Cann et al., *Mitochondrial DNA and Human Evolution*, 325 NATURE 31, 31 (1987); Frederika A. Kaestle & K. Ann Horsburgh, *Ancient DNA in Anthropology: Methods, Applications, and Ethics*, 45 YEARBOOK OF PHYS. ANTHROP. 92 (2002); Frederika A. Kaestle & David Glen Smith, *Ancient Mitochondrial DNA Evidence for Prehistoric Population Movement: The Numic Expansion*, 115 AM. J. PHYS. ANTHROP. 1-12 (2001) (using ancient and modern mtDNA to test hypothesis that modern Native American inhabitants of Nevada are recent arrivals who replaced previous inhabitants); Ripan S. Malhi et al., *Patterns of MtDNA Diversity in Northwestern North America*, 76 HUM. BIOLOGY 33, 33-34 (2004) (using ancient and modern mtDNA to show significant migration from sub arctic and Pacific coast into Columbian Plateau region); Stéphanie Plaza et al., *Insights into the Western Bantu Dispersal: MtDNA Lineage Analysis in Angola*, 115 HUM. GENETICS 439 (2004) (using mtDNA to clarify spread of Bantu populations throughout Africa and to trace movement of slaves into Brazil from Angola).

39. The Armed Forces DNA Identification Laboratory engages in such identification procedures using mtDNA testing for casualties from wartime and disasters. *See AFDIL Mitochondrial DNA (MtDNA) Section*, http://www.afip.org/Departments/oafme/dna/afdil/mito.html (last visited Jan. 20, 2006).

40. *See, e.g.*, Sarah Koenig, *DNA Identification Is a Daunting Task* (Sept. 20, 2001), http://www.baltimoresun.com/news/custom/attack/bal-te.dna20sep20,1,242800.story?coll=bal-attack-utility (discussing use of mtDNA typing to identify remains from September 11 bombings, the Oklahoma City bombings, and plane crashes); Yasser Daoudi et al., *Identification of Missing Individuals from Bosnia and Herzegovina Using Mitochondrial DNA Analysis*, presented at 11th International Symposium on Human Identification (2000), http://www.promega.com/geneticidproc/ussymp11proc/abstracts/daoudi.pdf. *See generally* http://www.dna.gov/uses/m_person (discussing the President's DNA Initiative, including use of mtDNA in identifying missing persons from natural disasters and crimes).

largely from its ability to identify maternal lines and to exclude individuals whose profiles differ from questioned samples. For similar reasons, mtDNA is appropriately used in criminal cases to exonerate persons whose mtDNA profiles are revealed inconsistent with crime scene samples.[41]

The FBI has led the field in the use of mtDNA evidence to *inculpate* criminal suspects. The FBI began studying mtDNA technology in 1992 and conducting mtDNA casework in 1996,[42] and is now one of a handful of public and private laboratories in the United States that conduct forensic mtDNA testing.[43] Because the FBI is the federal government's forensic laboratory, and because SWGDAM, under the auspices of the FBI, maintains the sole database used in the United States in forensic mtDNA analysis, this Article discusses its procedures as illustrative of all forensic mtDNA laboratories. What follows is a description of the FBI's methods for developing a suspect's mtDNA profile, determining whether the suspect should be "included" as a potential contributor because his profile is consistent with the evidence sample profile, and calculating the statistical significance of an inclusion through estimation of sequence frequencies in the population using the SWGDAM database.[44]

An analyst first sequences the HVI and HVII regions of a sample found at the crime scene. Next, assuming a suspect has been identified and has submitted a DNA sample, the analyst sequences the HVI and HVII regions of the suspect's sample and compares the two profiles against each other. The FBI, according to its protocols, does not automatically exclude a suspect if his profile differs from that of the evidence sample. Indeed, the FBI will only definitively exclude a suspect if there are two or more base pair differences between the samples with no evidence of heteroplasmy, on the theory that one difference may be the result of heteroplasmy.[45]

While the FBI declares an automatic exclusion only in cases involving two or more differences, the FBI will declare an *inclusion* (called a "failure to exclude")

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41. *See* Peter Neufeld, *Preventing the Execution of the Innocent: Testimony Before the House Judiciary Committee*, 29 HOFSTRA L. REV. 1155, 1161-62 (2001).

42. Alice A. Isenberg, *Forensic Mitochondrial DNA Analysis*, FBI LAW ENFORCEMENT BULLETIN 16 (August 2002), *available at* http://www.fbi.gov/publications/leb/2002/august02leb.pdf.

43. Forensic mtDNA testing is far more specialized, expensive, and time-consuming than nDNA testing. Over one hundred laboratories in the United States are authorized to conduct forensic nuclear DNA analysis. Government forensic mtDNA laboratories include the FBI and AFDIL. Commercial forensic mtDNA laboratories include Bode Technology Group, Inc., in Springfield, Virginia; Laboratory Corporation of America, in Research Triangle Park, North Carolina; Mitotyping Technologies, LLC, in State College, Pennsylvania; Orchid Cellmark Dallas, in Dallas, Texas; Reliagene Technologies, Inc., in New Orleans, Louisiana; and Serological Research Institute, in Richmond, California.

44. Drs. Kaestle and Kittles have become familiar with forensic laboratories' procedures for analyzing and typing mtDNA sequences through their anthropological and genetic research involving mtDNA sequencing. Ms. Roth and Mr. Ungvarsky have become familiar with such procedures through trial and appellate litigation involving the United States' use of mtDNA typing conducted by the FBI and other laboratories as evidence in criminal trials.

45. FBI MtDNA Protocols (2004), *supra* note 8, at § 11.3.3.

under several different scenarios.[46] Thus, if the examiner determines that the profiles of the suspect and evidentiary samples are identical at each of the bases in HVI and HVII, the suspect is included as a possible contributor.[47] The examiner also will not exclude the suspect if the profiles have a one base-pair difference and either sample displays heteroplasmy.[48] If the profiles have a one-base pair difference in HVI and HVII and no evidence exists that the suspect or evidence sample is heteroplasmic, the result is "inconclusive," and the suspect will again not be excluded.[49] As explained in Part I, the FBI does not sequence outside HVI and HVII to determine whether other differences exist besides those already observed that could exclude the suspect as a potential contributor to the evidence sample.

If the suspect's profile is consistent with the evidence profile, the analyst then compares this shared profile to the SWGDAM database.[50] The entire database contains 5071 profiles.[51] The database is subdivided into fourteen so-called "racial" sub-populations.[52] The database profiles come from samples collected by

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46. While this Article focuses on the sequence comparison protocols followed by the FBI, it is worth noting that other mtDNA typing laboratories differ in their treatment of heteroplasmy when comparing the suspect's profile to the evidence sample profile. *See* Statement of Dr. M. Thomas P. Gilbert, submitted in *United States v. Chase*, D.C. Super. Ct. Crim. No. F-7330-99 (July 9, 2004) (on file with authors) (reviewing protocols for all major mtDNA testing laboratories and observing that "forensic laboratories come to no consensus as to how to interpret heteroplasmic sequences. . . . [T]he interpretation guidelines vary when determining what would be labeled as 'inconclusive' and what would be labeled as an 'exclusion.'").

47. FBI MtDNA Protocols (2004), *supra* note 8, at § 11.3.3.

48. *Id.*

49. *Id.*

50. For convenience purposes, forensic laboratories do not search all 610 bases of the HVI and HVII regions. Rather, the sample is first compared to the revised CRS. Differences between the two are then searched against the profiles in the SWGDAM database. Alice R. Isenberg & Jodi M. Moore, *Mitochondrial DNA Analysis at the FBI Laboratory,* 1 FORENSIC SCI. COMM. 1 (1999), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/july1999/dnalist.htm.

51. *See* FBI MtDNA Protocols (2004), *supra* note 8, at § 12.1. The SWGDAM database has grown from 1393 sequences in 1998 to its current size of 5071. *See* Bruce Budowle et al., *Mitochondrial DNA Regions HVI and HVII Population Data*, 103 FORENSIC SCI. INT'L 23, 25 (1999) [hereinafter Budowle et al. (1999)] (1393 sequences in 1998); Isenberg & Moore, *supra* note 50, at 1 (2426 sequences in 1999); Constance Fisher & Bruce Budowle, Presentation, *Mitochondrial DNA: Today & Tomorrow*, 11th Annual Int'l Symposium on Human Identification (2000), *available at* http://www.promega.com/geneticidproc/ussymp11proc/content/fisher.pdf (4142 sequences in 2000). The database has not grown at all since at least April 2003. *See* FBI MtDNA Protocols (2004), *supra* note 8, at § 12.1 (stating that the database as of April 14, 2003, "contain[s] 5071 individuals").

52. None of the sub-databases has more than 1814 profiles; ten have fewer than two hundred profiles; and five have fewer than one hundred profiles. The populations and the number of profiles within each are as follows:

Race	Number of Profiles
African-Americans	1148
Apaches	180
Caucasians	1814
Chinese/Taiwanese	356
Egyptians	48

paternity testing laboratories, blood banks, FBI agents, and scientific research groups.[53] The classifications of the sequences are based on the self-reporting of the individuals who agreed to give the samples. The FBI does not claim that these samples are geographically diverse, randomly selected, or representative. Rather, the samples were obtained in an *ad hoc*, non-random manner from a few locations.[54]

To generate frequency estimates from the SWGDAM database, forensic scientists count the number of times that the shared profile "matches" a profile in each of the sub-population databases (the "counting method").[55] The analysts count only the number of appearances of the profile in the database and not the appearances of the profile in the suspect and in the evidence sample. Because the SWGDAM database omits the vast majority of mtDNA profiles and because more than 50% of the profiles in the SWGDAM database appear only once in the database,[56] this approach most often results in a count of zero observations or "hits."

The analyst next estimates the rarity of the profile in various "racial" populations based on the number of observations in each of several sub-databases categorized by self-reported ancestry. If the analyst sees at least one observation,

Race	Number of Profiles
Guam	87
Hispanics	759
India	19
Japanese	163
Koreans	182
Navajos	146
Pakistan	8
Sierra Leone	109
Thai	52
TOTAL	5071

Monson et al., *supra* note 8, at "Release Notes," *available at* http://www.fbi.gov/hq/lab/fsc/backissu/april2002/mtDNA_popDB1.2ReleaseNotes.pdf.

53. *See* Budowle et al. (1999), *supra* note 51, at 25 (explaining origins of SWGDAM database profiles).

54. *See id.* (listing geographical origins of SWGDAM profiles).

55. FBI MtDNA Protocols (2004), *supra* note 8, at § 11.1 (stating the FBI reports the number of "hits" in each racial database regardless of the suspect's putative race).

56. *See* Allard et al., *supra* note 34, at 2 (stating 72% of profiles in SWGDAM Caucasian database as of 2000 appear only once); Parsons & Coble, *supra* note 15, at 305:

> [C]omparing the 2000 database with the 1998 database shows that the number of sequences occurring once is decreasing, going from 63% in 1998 to 54% in 2000. The percentage of single occurrences will continue to decrease as more mtDNA samples are typed. However, the number of individuals who must be sequenced to reach the limit of mtDNA diversity is unknown [as] the overall distribution of mtDNA types is highly skewed toward rare types.

this process involves dividing the number of observations by the size of the database.[57] For example, if the profile were observed once in the African-American database (n = 1148), the frequency would be 1/1148 or 0.0008711. The analyst would then place a 95% confidence interval around that number as a margin of error in estimating the frequency in the larger population,[58] and the laboratory would report the upper-bound frequency. For an observed frequency of 0.0008711, the upper confidence limit is 0.002577, or 0.2577%, and the laboratory would report that about 99.74% of African Americans are excluded as potential contributors of the sample.

Because all people sharing a common maternal lineage are expected to have the same mtDNA sequence (excluding considerations of intergenerational mutation), the FBI acknowledges that examiners cannot declare identity based on mtDNA analysis alone. Yet such small reported probabilities of inclusion calculated from the SWGDAM database can suggest to juries that the consistency between the mtDNA profiles is a "match" amounting to a statement of identity.[59] Based on such small frequency estimates, mtDNA evidence has thus become a powerful tool of prosecution. A closer examination suggests, however, that such frequency estimates are based on faulty scientific assumptions that do not meet prevailing legal standards for admissibility of scientific evidence, and should not, in their current state of development, be admitted against criminal defendants at trial.

III. THE DUBIOUS RELIABILITY OF FREQUENCY ESTIMATES ASSOCIATED WITH FORENSIC MTDNA

The current SWGDAM database suffers from several structural problems that make it incapable of producing reliable estimates of mtDNA profile frequencies in particular geographical and ancestral populations. *First*, the database is a statistically unsound sample set from which to estimate mtDNA sequence frequencies

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57. Laboratories use a slightly different statistical calculation when the sequence is not observed in the database. *See* Holland & Parsons, *supra* note 14, at 31-32.

58. A 95% confidence interval means that, if a series of such margins of error were constructed in estimating the frequency of the sequence in the population, approximately 95% of them should include the true frequency of the sequence in the population. Alternatively stated, there is approximately a 5% chance that the margin of error does not contain the true frequency of the sequence in the population. *See* ROBERT S. WITTE, STATISTICS 215 (2d ed. 1985). As the sample size grows, the confidence interval will become narrower, indicating 95% confidence in a smaller range of possible values for the frequency. *Id.* at 216. Ninety-nine percent confidence intervals are also "prevalent" in statistical calculations. *Id.* at 221. None of the forensic literature or forensic laboratory protocols reviewed by the authors discusses why a 95% confidence interval, as opposed to a more conservative interval like 99%, is used in forensic casework. Indeed, the FBI uses a 99% confidence interval when determining whether to label a nuclear DNA profile as "unique" in the population. *See* FBI Laboratory Unit I, Short Tandem Repeat Analysis Protocols § 10.6 at 10-10, 10-11 (Apr. 1, 2002).

59. *See, e.g.*, United States v. Coleman, 202 F. Supp. 2d 962, 964, 967 (E.D. Mo. 2002) (stating 99.93% of all persons excluded from contributing mtDNA sample); Lewis v. State, 889 So. 2d 623, 673 (Ala. Crim. App. 2003) (stating 99.8% of Caucasians excluded); State v. Pappas, 776 A.2d 1091, 1104 (Conn. 2001) (stating 99.75% of Caucasians excluded); Magaletti v. State, 847 So.2d 523, 587 (Fla. Dist. Ct. App. 2003) (stating 99.93% of all persons excluded).

because it does not account for geographic and ancestral clustering of identical or related mtDNA profiles. Specifically, the manner in which the samples are collected – samples taken from a handful of arbitrarily selected regions of the United States – assumes, incorrectly, that mtDNA profiles are randomly distributed in the population. But unlike nuclear DNA, which always reflects inheritance of certain of the mother's and father's unique influences and thus varies even between siblings (except identical twins), mtDNA is maternally inherited, does not recombine, and is far from randomly distributed in the population. Moreover, the "racial" categories in the databases, do not sufficiently take into account the intra- and inter-ethnic diversity resulting from well-documented ancestral migration patterns and clustering of profiles into identifiable haplogroups. *Second*, the SWGDAM database is too small in relation to the general populations it purports to represent to estimate such frequencies adequately. *Third*, even if the database were representative and large enough, significant previously unaddressed quality control problems undermine its reliability. *Fourth*, as applied, the "counting method" potentially understates frequency estimates systematically through assumptions biased against suspects. While each of these problems can be remedied, their existence suggests that reliance upon the current SWGDAM database is currently unfounded and that such issues should, in any event, be addressed by attorneys and the courts.

A. MtDNA Is Not Randomly Distributed in the Population

1. The Statistical Validity of the SWGDAM Database Is Premised on False Assumptions about the Distribution of MtDNA Profiles in the Population

Many of SWGDAM's sequences came from the same samples that existed in the FBI's STR databases used to generate match statistics in forensic nDNA typing.[60] In nuclear DNA testing, forensic scientists typically obtain miniscule random match probability statistics generated by comparison of alleles at each of the thirteen standard STR locations.[61] The nDNA STR databases, and the validation studies thereof, were based on the premise that, particularly given the miniscule associated statistics, only statistically insignificant genetic linkage exists within the populations sampled. Accordingly, for nDNA, it was concluded that sampling from a small number of locations was acceptable.[62]

This assumption of randomness is not valid with respect to mtDNA sequences. As explained below, because mtDNA is maternally inherited and not recombinant,

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60. *See* Budowle et al. (1999), *supra* note 51, at 25.

61. *See* BUTLER, *supra* note 12, at 502 ("Often the rarity of a calculated [nuclear] DNA profile goes beyond one in billions (10^9) or trillions (10^{12}) to numbers that are not frequently used because they are so large."); *id.* at 504, tbl.21.3 (listing values such as quadrillion (10^{15}), quintillion (10^{16}), and google (10^{100})).

62. *See* NRC II (1996), *supra* note 5, at 30 (stating STR database consists of convenience samples from "blood banks, paternity-testing laboratories, laboratory personnel, clients in genetic-counseling centers, law-enforcement officer, and people charged with crimes"); United States v. Bridgett, 120 Daily Wash. L. Rptr. 1697, 1700 n.12 (D.C. Super. Ct Aug. 11, 1992) (same).

mtDNA profiles are not randomly distributed. The distribution of a particular mtDNA sequence is primarily a function of the migration of women. A child and his maternal great-great-great-great-grandmother, or a child and all of his mother's sisters' children, are expected, absent mutations, to have identical mtDNA profiles. Over generations, profiles stay intact or mutate to a very similar sequence. In addition, the high mutation rates characteristic of the HVI and HVII regions create unique variants, including more recently created ones that have not had time to spread from their location of origin. This creates geographical areas where certain haplogroups or haplotypes are prevalent, and other areas where those same haplogroups and constituent haplotypes are wholly or largely nonexistent.

2. *Phylogeographic Studies Confirm That MtDNA Haplogroups Exist and Are Geographically Stratified*

Dozens of phylogeographic[63] studies have been performed to identify the geographic distribution of mtDNA haplotypes in countries all over the world, although such studies are extremely limited in the United States. These studies demonstrate that mtDNA is not randomly distributed and that different haplogroups and haplotypes are concentrated within certain populations that vary geographically.[64] Scientists rarely come across new nDNA gene types when studying new population subgroups; however, the same is not true for mtDNA sequences. While certain haplogroups of mtDNA sequences are widely distributed throughout the population,[65] many exist only within certain geographic clusters.[66]

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63. Phylogeography "is a field of study concerned with the principles and processes governing the geographic distributions of genealogical lineages, especially those within and among closely related species [and] deals with historical, phylogenetic components of the spatial distributions of gene lineages. In other words, time and space are the jointly considered axes of phylogeography onto which (ideally) are mapped particular gene geneaologies of interest." JOHN C. AVISE, PHYLOGEOGRAPHY: THE HISTORY AND FORMATION OF SPECIES 3 (2000).

64. *See, e.g.*, Ripan S. Malhi et al., *The Structure of Diversity Within New World Mitochondrial DNA Haplogroups: Implications for the Prehistory of North America*, 70 AM. J. HUM. GENET. 905, 906 (2002) [hereinafter Malhi et al. (2002)] ("significant geographic variation in frequency distributions across North America" existed for nearly five hundred Native American haplotypes and "haplogroup frequency distribution was correlated with geography"); Dan Mishmar et al., *Natural Selection Shaped Regional MtDNA Variation in Humans*, 100 PROC. NATL. ACAD. SCI. 171 (Jan. 7, 2003) ("extensive global population studies have shown that there are striking differences in the nature of the mtDNAs found in different geographic regions").

65. In North America, the C and D haplogroups, present in Native American populations, are widely distributed across the continent. *See* Malhi et al. (2002), *supra* note 64, at 909-11 & figs. 2, 3, 4. The H haplogroup occurs in 20% to 25% of the population in the Near East, 50% in Europe, and nearly 60% in the Basque country of Spain. *See* Martin Richards et al., *In Search of Geographical Patterns in European Mitochondrial DNA*, 71 AM. J. HUM. GENETICS 1168, 1170 (2002).

66. While *phylogenetic* analysis – reconstructing genetic relationships within a population – has been conducted on many of the SWGDAM racial sub-databases, such studies only show, at most, that a particular database accurately reflects most of the haplogroups that exist in the relevant population, e.g., that the Caucasian database contains all major haplogroups in the Caucasian population. *See* Allard et al., *supra* note 34, at 8. Such studies do not, however, take into account the *geographical distribution* of the sequences within the population, and thus cannot be cited as evidence that a database accurately reflects the frequency of a profile in a particular geographic area. Only *phylogeographic* studies – those that focus on the spectrum and area-specificity of major

Non-random mtDNA haplotype distributions also exists *within* geographic locations, because of often subtle linguistic, religious, or economic/caste distinctions.[67]

Distinctive mtDNA haplotype distributions are not limited to rare or ancient populations; today, different geographic regions demonstrate strikingly different mtDNA patterns.[68] For example, a particular cluster of mtDNA sequences called haplogroup J is widely distributed in western and central Europe, but is rare in the Iberian Peninsula.[69] A sub-haplogroup of that cluster has been observed primarily in Britain, with one other occurrence from an ancestor in Italy.[70] A mutation that has an 8% frequency within the Canary Islands has never been found outside the Islands.[71] One study related to the natives of Mozambique as compared to those in the Americas identified a considerable number of matches between Mozambique and American sequences from African haplogroups, including some sequences

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haplogroups and the haplotypes within them – can accurately determine true frequencies. *See* Juan C. Rando et al., *Phylogeographic Patterns of MtDNA Reflecting the Colonization of the Canary Islands*, 63 ANNALS HUM. GENETICS 413, 424 (1999).

67. *See, e.g.*, Michael Bamshad et al., *Genetic Evidence on the Origins of Indian Caste Populations*, 11 GENOME RESEARCH 994 (2001) (discussing economic and caste distinction); Ranjan Dutta et al., *Patterns of Genetic Diversity at the Nine Forensically Approved STR Loci in the Indian Populations*, 74 HUM. BIOL. 33 (2002) (same); D. Andrew Merriwether et al., *Mitochondrial DNA Is an Indicator of Austronesian Influence in Island Melanesia*, 110 AM. J. PHYS. ANTHROPOL. 243 (1999) (linguistic distinctions); Pavao Rudan et al., *Anthropological Research of Hvar Islanders, Croatia – From Parish Registries to DNA Studies in 33 Years*, 28 COLLEGIUM ANTHROPOLOGICUM 321 (2004) (religious); Lev A. Zhivotvsky et al., *The Forensic DNA Implications of Genetic Differentiation Between Endogamous Communities*, 119 FORENSIC SCI. INT'L 269 (2001) (no obvious subdivision).

68. *See, e.g.*, DAVID BALDING, WEIGHT-OF-EVIDENCE FOR FORENSIC DNA PROFILES 105-06 (2005):

> [M]aternally-related individuals might be expected to be tightly clustered, possibly on a fine geographical scale. Reports of F_{ST} estimates for mtDNA drawn from cosmopolitan European populations typically cite low values, reflecting the fact that this population is reasonably well-mixed, as well as the effects of high mtDNA mutation rates. However, researchers rarely are able to focus on the fine geographic scale that may be relevant in forensic work, and there are some large F_{ST} estimates at this scale.

see also Anita Brandstätter et al., *Mitochondrial DNA Control Region Sequences from Nairobi (Kenya): Inferring Phylogenetic Parameters for the Establishment of a Forensic Database*, 118 INT'L J. LEGAL MED. 294 (2004) (describing new forensic database containing sequences from Nairobi and finding that there were significant differences in mtDNA compositions of this new database and the African-American SWGDAM database, as well as of published sequences from Sierra Leone, Mozambique, and United States); Peter Forster et al., *Continental and Subcontinental Distributions of MtDNA Control Region Types*, 116 INT'L J. LEGAL MED. 99, 99 (2002); Kaestle & Horsburgh, *supra* note 38, at 95 ("[M]itochondrial markers are also often geographically specific, and in some cases are limited in distribution to a single tribe (private polymorphisms)."); Rick A. Kittles & Shomarka O. Keita, *Interpreting African Genetic Diversity*, 16 AFRICAN ARCHEOL. REV. 87, 87 (1999); Luisa Pereira et al., *Prehistoric and Historic Traces in the MtDNA of Mozambique: Insights into the Bantu Expansions and the Slave* Trade, 65 AM. J. HUM. GENETICS 439 (2001); Rando et al., *supra* note 66, at 424; Antonio Salas et al., *The African Diaspora: Mitochondrial DNA and the Atlantic Slave Trade,* 74 AM. J. HUM. GENETICS 454 (2004); Yao et al., *supra* note 21, at 649.

69. Richards et al., *supra* note 65, at 255 (discussing J Haplogroup).

70. *Id.* at 254 (discussing J1b1 Haplogroup).

71. Rando, *supra* note 66, at 420, 424.

that had never been observed outside Mozambique, as well as others observed only in the American populations.[72] From 2000 to the present, the AFDIL has been documenting such regional differences through a DOJ-funded effort to create databases of mtDNA control region sequences for African-origin, Hispanic, and Central Asian individuals. As of July 2005, AFDIL had databased "249 African-American, 646 U.S. Hispanic, and nearly 2500 Central Asia samples."[73] The five-year project "is intended to . . . investigate the potential for forensically significant regional variation within U.S. racial/ethnic groups" and has successfully unearthed such significant results, particularly with respect to U.S. Hispanics.[74] Based on these observed differences, AFDIL reported to the National Institute of Justice that its "work establishes that there is highly significant geographic variation of mtDNA types among individuals classified as 'Hispanics' in the United States" and that "[t]his has serious implications for the appropriate structuring of forensic mtDNA population databases."[75] To the AFDIL researchers, "[i]t seems very unlikely that reference to a single Hispanic database can be justified in evaluating the significance of mtDNA matching in the Hispanic population."[76]

Similarly, a study of the Han Chinese revealed dramatic regional differences in haplogroup frequencies among a population that constitutes 93% of the Chinese population and nearly 20% of the world's population.[77] Researchers examined 263 unrelated Han Chinese samples taken from six different provinces. They observed that, while certain haplogroups made up almost 20% of the population in a certain province, the haplogroup was nonexistent in a different province. Ultimately, the clustering in particular provinces was so pronounced that the authors concluded that an East Asian database, or even "Northern Han" and "Southern Han" databases, would grossly underestimate the frequency of certain groups of se-

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72. Pereira et al., *supra* note 68, at 452:

> There remain a large number of sequences from African haplogroups sampled in the Americas and Europe for which no match can be found in the current African database. This may be due in part to the fact that the main regions from where slaves were taken, such as Angola and the Slave Coast remain uncharacterized.

See also Joseph Lorenz et al., *African-American Lineage Markers: Determining the Geographic Source of MtDNA and Y Chromosomes* (Apr. 15, 2004), http://www.physanth.org/annmeet/aapa2004/ajpa2004.pdf (discussing study suggesting that there is large proportion of unexamined, undocumented mtDNA variability among individuals indigenous to sub-Saharan Africa).

73. Parsons, *supra* note 23, at 1.

74. Parsons et al., *supra* note 23, at 4-5 ("U.S. Hispanics are a complex admixture of Native American, European, and African lineages, making the regional variation of U.S. Hispanics important in a forensic context."); *id.* (listing dramatic differences in frequency of various haplotypes among Hispanics in different regions of the United States).

75. *Id.* at 5.

76. *Id.*

77. Yao et al., *supra* note 21, at 635.

quences that themselves are highly common in surrounding regions.[78]

MtDNA population genetic linkage in North America – discussed in detail in the next two sections – is also well documented in scientific research.[79] Whether the heterogeneous geographic distribution of mtDNA lineages reflects genetic clustering, inadequate sampling, or some combination of the two, it appears clear that the sampling of mtDNA profiles must take into account geographic heterogeneity and stratification in order to create representative databases for use in forensic typing.

3. Significant Ancestry-Related Population Substructure Exists in the Distribution of MtDNA Sequences in the African-American Population

The SWGDAM database also fails to account for ancestry-related clustering of haplogroups in the United States, particularly with respect to the collective experience of African Americans, whose post-slavery era migration patterns are well documented.[80] The oldest mtDNA profiles stem from Africa, whose popula-

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78. *See id.* at 649:

> The comparison of the regional Han mtDNA samples revealed an obvious geographic differentiation in the Han Chinese, as shown by the haplogroups-frequency profiles. . . . Hence, the grouping of different Han populations into just "Southern Han" and "Northern Han" or the use of one or two Han regional populations to stand for all Han Chinese . . . does not appropriately reflect the genetic structure of the Han.

79. *See, e.g.,* David Biello, *Skulls Suggest Differing Stocks for First Americans* (Dec. 13, 2005), http://www.sciam.com/print_version.cfm?articleID=000E8538-F33D-139D-B33D83414B7F0000 ("Today, no South American native group presents the X [mitochondrial DNA] lineage, which is universal among North American native groups.") (alteration in original); Jason Eshleman et. al., *Mitochondrial DNA Studies of Native Americans: Conceptions and Misconceptions of the Population Prehistoric of the Americas*, 12 EVOL. ANTHROPOL. 7-18 (2003) (noting that while Haplogroup X is found in low frequency in Europe and Western Asia, Native American variant is significantly different, possessing mutation that distinguishes it from Old World versions); Lynn B. Jorde & Stephen P. Wooding, *Genetic Variation, Classification, and "Race,"* 36 NATURE GENETICS S28, S29 (Nov. 2004) ("[I]ndividuals tend to cluster according to their ancestry or geographic origin."); Malhi et al. (2002), *supra* note 64, at 3-5 (stating native Americans have haplogroups whose frequencies varies greatly among Canada, United States, and Mexico); Esteban J. Parra, Rick A. Kittles et al., *Ancestral Proportions and Admixture Dynamics in Geographically Defined African Americans Living in South Carolina*, 114 AM. J. PHYS. ANTHROPOL. 118 (2001) [hereinafter Parra & Kittles (2001)]; Estaban J. Parra, Amy Marcini et al., *Estimating African-American Admixture Proportions by Use of Populations-Specific Alleles*, 63 AM. J. HUM. GENETICS 1839 (1998); Sarah A. Tishkoff & Kenneth K. Kidd, *Implications of Biogeography of Human Populations for "Race" and "Medicine,"* 36 NATURE GENETICS S21, S26 (Nov. 2004) (stating that frequency of mtDNA haplogroups are unevenly distributed within and among geographic regions and "knowledge of ethnicity (not just broad geographic ancestry) and statistical tests of substructure are important proper design of case control association studies"). *Cf.* Terry Melton et al., *Diversity and Heterogeneity in Mitochondrial DNA of North American Populations*, 46 J. FORENSIC SCI. 46 (2001) (arguing that North American population is homogeneous, and identifying, without exploring, population of Hispanics in Pennsylvania who differed significantly from any other population in study).

80. The authors' focus on the migration patterns of African Americans should not be taken as a statement that ancestry-related substructure in mtDNA profile distribution does not exist with respect to other groups in the United States. Rather, the focus reflects the fact that the African-American population exhibits high genetic heterogeneity, and the most infrequently occurring haplotypes, compared to other ethnic groups in the United States. As descendants of enslaved Africans, African Americans possess a diverse gene pool that is mainly of west and central African origin but also of substantial European and Native American admixture. As a result, it is of

tion displays great regional diversity and heterogeneity in mtDNA profiles.[81] In some regions, specific mtDNA profiles are common; in others, the same mtDNA profiles are rare or nonexistent.[82] Scientific studies in Africa repeatedly uncover more unknown and previously unexamined mtDNA sequences, and far more is left to learn about regional differences that exist both now and hundreds of years ago.

During the period of slavery in the United States, the forced migration of Africans to the New World brought these regional differences to the United States and led to significant regional differences in the ethnic and geographic ancestry of African Americans.[83] Various political, economic, and cultural factors associated with the implementation of slavery contributed to these regional differences. For instance, during the period of slavery in the South, plantation owners in South Carolina primarily grew rice. These owners sought West Africans who already knew how to grow rice and therefore imported enslaved Africans from the "Grain Coast" of Africa.[84] In contrast, in Virginia, plantation owners primarily sought to grow tobacco.[85] The area surrounding the tobacco farms was swampy, and with the swamps, mosquitoes and malaria were common.[86] Neither Native American nor European American workers had genetic resistance to malaria and were dying in large numbers. Plantation owners sought enslaved Africans resistant to malaria and turned to the "Gold Coast" – modern day Ghana and Benin.[87] Similarly, because

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significant scientific importance, both in forensic science and biomedicine, to understand the genetic consequences of this unique population history. The authors' emphasis also reflects the significant number of studies on African-American migration in particular and the relative familiarity of the lay public with the historical post-slavery era migration of African Americans.

81. Rebecca L. Cann, Mark Stoneking, & Allan C. Wilson, *Mitochondrial DNA and Human Evolution*, 325 NATURE 31 (1987). *See also* Philip D. Curtin, *From Guesses to Calculations, in* THE ATLANTIC SLAVE TRADE: A CENSUS (David Northrup ed., 1994). Curtin's calculations were later refined by David Northrup. Paul E. Lovejoy, *Curtin's Calculations Refined but Not Refuted, in* THE ATLANTIC SLAVE TRADE 50-59 (David Northrup ed., 1994). *See also* Elizabeth E. Watson et al., *MtDNA Sequence Diversity in Africa*, 59 AM. J. HUM. GENETICS 437 (1996).

82. *See, e.g.*, Terry Melton et al., *Extent of Heterogeneity in Mitochondrial DNA of sub-Saharan African Populations*, 42 JOURNAL OF FORENSIC SCI 582, 588-89 (1997) (finding numerous haplotypes with occurrences of sequence-specific oligonucleotides (SSO) – particular base pair variations in certain parts of the mtDNA control region – of more than 10% in a particular African population and "substantial subpopulation heterogeneity" in "continental African populations"). The authors conclude that "control region sequencing would be a good alternative for forensic identifications in African or African-derived populations where there is uncertainty about whether subpopulations are present, at least until further populations are studied." *Id.* at 589.

83. *See generally* Salas et al., *supra* note 68, at 455-56.

84. Parra & Kittles (2001), *supra* note 79, at 19.

85. PHILLIP D. MORGAN, SLAVE COUNTERPOINT: BLACK CULTURE IN THE EIGHTEENTH CENTURY CHESAPEAKE AND LOWCOUNTRY 33-44 (1998).

86. *Id.* at 34-36.

87. Fatimah Jackson, *Concerns and Priorities in Genetic Studies: Insights from Recent African-American Biohistory*, 27 SETON HALL L. REV. 951, 961-62 (1997); Parra, Marcini et al., *supra* note 79, at 1839 (listing countries of Africa by economic region). This very same resistance makes African Americans whose ancestors come from the Gold Coast more likely to carry the sickle cell trait and sickle cell disease. A. Muniz et al., *Sickle-Cell-Anemia and Beta-Gene Cluster Haplotypes in Cuba*, 49 AM. J. OF HEMATOLOGY 163 (1995); Gabriella Pante-De Sousa et al., *Betaglobin Hapolytpes Analysis in Afro-Brazilians from the Amazon Region: Evidence for a Significant Gene Flow from Atlantic West Africa*, 26 ANNALS OF HUM. BIO. 365 (1999).

Portuguese and French slave traders were the primary slave traffickers in New Orleans, many of the enslaved Africans brought to Louisiana were from Angola.[88] Thus, the forced migration of enslaved Africans to the United States led to geographic variation in this country similar to that of regional African variation.

Once in the United States, the clusters of African Americans either remained in their geographical origins or migrated in distinct groups, as family members joined family members, friends followed friends, and neighbors encouraged neighbors to emigrate.[89] This patterned migration resulted in further geographic variation throughout the United States. For the most part, this took place during the "Great Migration" – roughly 1910 to 1930 – when African Americans in the rural south traveled north for better jobs in light of World War I and a boll weevil crop infestation in the South.[90] These migrations took predictable routes: African Americans from Mississippi, Alabama, and Louisiana largely followed the Mississippi River and migrated to the great cities of the Midwest, such as Detroit, Chicago, Cleveland, and Kansas City; and African Americans from the Carolinas and Virginia tended to travel up the coastline to Washington, D.C., Philadelphia, and New York.[91] Notwithstanding the effects of this large-scale migration, most African Americans have remained in the southern part of the United States, in the crescent-shaped region ranging from Washington, D.C. to Louisiana.[92] Today, scientists observe genetic variation among African Americans in different regions of the country based upon the routes of those African Americans who migrated there and based on the variable levels of mixing with European Americans in different parts of the United States.

Heterogeneity also exists in African-American mtDNA profiles as one moves westward across the country. African Americans living in the western United States tend to exhibit larger percentages of European and Native American ancestry than those living in the South, Mid-Atlantic, and Midwest.[93] These phenomena may be the result of history, in that western territories and states had less restrictive social mores with respect to interracial relationships at the time of greatest migration. Additionally, the number of Native Americans surviving European settlement living in western states was significantly higher than in the

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88. Curtin, *supra* note 81, at 83.

89. *See generally* JAMES R. GROSSMAN, LAND OF HOPE: CHICAGO, BLACK SOUTHERNERS, AND THE GREAT MIGRATION (1991).

90. *Id.* at 28-30.

91. *Id.* at 112-13 (describing migration from Mississippi delta to Chicago); NICHOLAS LEMANN, THE PROMISED LAND: THE GREAT BLACK MIGRATION AND HOW IT CHANGED AMERICA 119-20 (1991) (alluding to migration from Carolinas and Virginia up East Coast).

92. *See* http://www.censusgov/prod/cen2000/dp1/2khus.pdf (displaying a pictorial depiction of geographical distribution of African Americans in United States).

93. *See* Parra, Mancini et al., *supra* note 79, at 1845-47; Ranajit Chakraborty, *Gene Admixture in Human Populations: Models and Predictions*, 29 YEARBOOK OF PHYS. ANTHROP. 1-43 (1986); David C. McLean, Jr. et al., *Three Novel MtDNA Restriction Site Polymorphisms Allow Exploration of Population Affinities of African Americans*, 75 HUM. BIOLOGY 147-61 (2003).

east, which helps to explain the Native-American "admixture" in African-American mtDNA profiles.[94]

A further critical dimension of regional variation in mtDNA profiles is a result of the variation in the level of "admixture" between African Americans and other groups around the country. For example, while African Americans living in Charleston, South Carolina, possess about 6.5% of European maternal ancestry, this figure is much higher in Baltimore (14.94%), New York (9.11%), and Pittsburgh (9.9%).[95] To determine the frequency of an mtDNA sequence at a Charleston crime scene, for example, a forensic scientist should use a database that takes into account the types of mtDNA profiles that exist in Charleston. As further illustration, Jamaican Americans, whose mtDNA is on average 12.93% derived from European ancestry, have quite different mtDNA profiles from African Americans in most American cities – information that should be known to the forensic scientist in electing to which mtDNA database to compare the questioned profile.[96] The SWGDAM database does not account for, or reflect, these regional differences.

4. Population Substructure with Respect to MtDNA Sequences in the United States and Other, Non-African-American Ancestral Populations

The SWGDAM database also fails to account for regional variation in other U.S. ethnic groups. For example, while the database has a Hispanic category, most geneticists agree that the term "Hispanic" is primarily a language-based categorization, not a genetic one.[97] Not surprisingly, then, individuals in the linguistic category "Hispanic" display tremendous amounts of genetic variation.[98] One cannot reasonably claim, for example, that Hispanics living in South Florida (largely of Cuban and Puerto Rican ancestry) are genetically representative of Hispanics living in California (largely Mexican in ancestry). Yet the design of the SWGDAM mtDNA database assumes that mtDNA profiles of Hispanic-

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94. *See American, Indian, Eskimo, and Aleut Persons* (last visited Feb. 17, 2006) http://www.census.gov/geo/www/mapGallery/images/americanindian.jpg (displaying visual depiction of heavy Native-American clustering in western part of United States); STELLA U. OGUNWOLE, THE AMERICAN INDIAN AND ALASKA NATIVE POPULATION: 2000 4-6 (U.S. Census Bureau Feb. 2002) (noting that 43% of American Indians lived in West, 31% lived in South, 17% lived in Midwest, and 9% lived in Northeast United States).

95. Parra, Marcini et al., *supra* note 79, at 1845. The admixture study reports *two* results from Philadelphia, based on two independent sample sets taken from patients in two separate hypertension studies. These sample sets exhibited significant differences in their percentage of admixture. *Id.* Thus, even within a single city, different groups of African Americans display significantly different mtDNA profiles.

96. *Id.* at 1845-47.

97. *See* Carolina Bonilla et al., *Admixture in the Hispanics of the San Luis Valley, Colorado and Its Implications for Complex Trait Gene Mapping*, 68 ANNALS HUM. GENETICS 139, 140 (2004) (stating that the term "Hispanic" applies to individuals from several continents with "diverse cultural features and genetic backgrounds").

98. *See id.* (reporting differences in admixture among Puerto Rican, Cuban, and Mexican groups, as well as within smaller region of San Luis Valley).

Americans are randomly distributed. This failure to account for genetic diversity is particularly troubling given that the FBI *does* distinguish between Southeast and Southwest Hispanics in its *nuclear* DNA database, presumably to account for population substructure within the "Hispanic" population.[99] The FBI's attempt to subcategorize its (more recombinant) nDNA database to account for substructure is laudable, but the lack of recombination in mtDNA inheritance makes geographic clustering all the more critical in designing a representative mtDNA population database.

A simple contrast between the SWGDAM database and various compilations of mtDNA sequences observed in the published literature highlights the database's lack of geographic representation. For example, the SWGDAM database contains only two categories of Native-American mtDNA profiles, Apache and Navajo, which contain 180 and 146 mtDNA sequences, respectively. These SWGDAM sub-databases are incomplete and unrepresentative. Haplogroup D exists in Apache anthropological databases but is completely missing from the SWGDAM Apache database.[100]

The frequency of Haplogroup X in studies in the academic literature is four-to-five times greater than in the corresponding SWGDAM database.[101] More generally, the collections of mtDNA sequences in the research literature – which are approximately one-third the size of the SWGDAM database – report entire haplogroups not present in the SWGDAM database and significantly different percentages of the kinds of haplotypes represented in the SWGDAM database.[102]

The same issue of disproportionate representation of sub-populations is also reflected in SWGDAM's East Asian databases. The database fails to account for the proportional ancestry of East Asian Americans. The 753 individuals in the SWGDAM East Asian database are from China, Korea, Japan and Thailand, with almost half from China.[103] Significant disconnect exists between the SWGDAM

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99. *See* Bruce Budowle et al., *Population Data on the STR Loci D2S1338 and D19S433*, FORENSIC SCI. COMM. (July 2001), *available at* http://www.fbi.gov/hq/lab/fsc/backissu/july2001/budowle2.htm.

100. Ripan S. Malhi et al., *Native American MtDNA Prehistory in the American Southwest*, 120 AM. J. PHYS. ANTHROP. 108, 113 (2003).

101. *Id.* Additionally, the Navajo and Apache are not representative of the variation present in haplotypes/ haplogroups among all North American Native Americans. Tribal groups in the United States share few haplotypes. *See* Malhi et al. (2002), *supra* note 64, at 914, tbl.2 (estimating sharing at about 29%).

102. Malhi et al., *supra* note 100, at 121-22.

103. The primary published analysis of this database concerns only the Chinese samples and while the analysis suggests that the frequencies of the haplogroups in the dataset are similar to those in another Han Chinese dataset of 263 individuals, the authors' data reveal significant differences in almost all cases. Marc W. Allard, Mark R.Wilson et al., *Control Region Sequences for East Asian Individuals in the Scientific Working Groups on DNA Analysis Methods Forensic MtDNA Data Set*, 6 LEGAL MED. 11, 18, fig.2 (2004). Other studies also show significant genetic variation among and within Asian populations. *See, e.g.*, Toomas Kivisild et al., *The Emerging Limbs and Twigs of the East Asian MtDNA Tree*, 19 MOL. BIOL. EVOL. 1737 (2002) (noting other Asian populations not represented in the SWGDAM East Asian database have significantly different frequencies of mtDNA haplogroups than those in the database); Terry Melton & Mark Stoneking, *Extent of Heterogeneity in*

database and the 2000 U.S. Census figures.[104] For example, based on the 2000 Census, 18.3% of the U.S. Asian population is Filipino, while there are no Filipinos in the SWGDAM database.[105] Similarly, the percentage of Chinese and Korean individuals in the SWGDAM database is double that reflected in the Census, while the database's percentage of Asian-Indians is one-seventh of their true percentage of the population in 2000. Simply put, the SWGDAM database is not representative of the distribution of Asian-American source populations.[106]

The Caucasian database is also problematic in its apparent failure to account for non-random distribution of ancestral haplogroups. Those who argue that SWGDAM's Caucasian database is representative point to the fact that its samples include 44.2% of the H Haplogroup, which appears in approximately the same proportion in certain Western European countries.[107] But the percentage of "H" varies widely outside a handful of countries in Western Europe, such as countries in Scandinavia, Eastern Europe, and parts of West Europe such as France, Northern Germany, and Scotland – areas where, of course, many American

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Mitochondrial DNA of Ethnic Asian Populations, 41 J. FORENSIC SCI. 591-602 (1996) (same); Yao et al., *supra* note 21, at 636 (combining all Han Chinese would be inappropriate).

104. *See* Terrance J. Reeves & Claudette E. Bennett, WE THE PEOPLE: ASIANS IN THE UNITED STATES, Pub. No. CENSR-17, U.S. Census Bureau, Dep't of Commerce 1, tbl.1 (2004) (listing major Asian groups in U.S., many of which are not included in SWGDAM Asian databases), *available at* http://www.census.gov/prod/2004pubs/censr-17.pdf.

105. *Id.* at 4, fig.1.

106. A chi-square analysis conducted by the authors comparing the SWGDAM and 2000 Census frequencies of Asian subpopulations (converted to sample sizes in both cases) rejects the hypothesis that the SWGDAM database is a random sample of the Census Asian Populations, with an extremely significant p value of less than 10^{-20}:

Asian Population	2000 Census	SWGDAM Database
Asian Indian	16.1%	2.4%
Cambodian	1.7%	0
Chinese	23.8%	45.7%
Filipino	18.3%	0
Hmong	1.7%	0
Japanese	7.8%	20.9%
Korean	10.6%	23.3%
Laotian	1.7%	0
Pakistani	1.6%	1%
Thai	1.1%	6.7%
Vietnamese	10.9%	0
Other Asian	4.7%	0

107. *See* Allard et al., *supra* note 34, at 1219-20.

families originated.[108]

In sum, the SWGDAM database appears to misrepresent the regional genetic diversity of mtDNA profiles to ignore studies showing tremendous intra-group diversity in the various macro-ethnic categories represented in the database. The database combines internally heterogeneous groups under broad rubrics without a demonstration that the deviation from homogeneity would have negligible consequences in reporting mtDNA match significance. If the database is not representative, it does not serve its intended purpose of providing a random selection of the relevant population from which reliable sequence frequency estimates may be calculated. In turn, if the database does not produce reliable frequency estimates, its admissibility under the prevailing rules for admission of scientific evidence would, as discussed below in Part IV, seem to be questionable at best.[109]

B. The Size of the SWGDAM Database

Given the degree of known mtDNA genetic variation, the SWGDAM database, whether the surveyed category contains 8 profiles (the Pakistani category of the database), 1148 profiles (the African-American category of the database), or 5071 profiles (the total number of profiles in the database), is too small to provide meaningful estimates of sequence frequencies. Extrapolation from small sample sizes inhibits the ability to make meaningful mtDNA profile frequency estimates, and statistical claims about frequency estimates based on databases smaller than one hundred profiles are particularly questionable.[110] While the larger categories such as the Caucasian, African-American, and Hispanic databases may appear to contain a considerable number of profiles, such appearances are misleading. Even the relatively larger databases are insufficient, both because the validation work

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108. *See* Wojciech Branicki, Ksenia Kalista et al., *Distribution of MtDNA Haplogroups in a Population Sample from Poland*, 50 J. FORENSIC SCI. 732, 733 (2005) (noting H Haplogroup was observed in 37.8% of samples in population from Southern Poland); Vincent Dubut et al., *MtDNA Polymorphisms in Five French Groups: Importance of Regional Sampling*, 12 EUR. J. HUM. GENETICS 293, 296 (2004) (showing that within France alone, frequency of H varies between 35% and 50% in two separate communities in Brittany); Ana M. Gonzalez et al., *Mitochondrial DNA Affinities at the Atlantic Fringe of Europe*, 120 AM. J. PHYS. ANTHROPOL. 391, 394 (recording 26.3% in Norway, 34% in England, 36.4% in Northern Germany, 38.5% in France and 42.2% in Galicia); Boris A. Malyarchuk et al., *Mitochondrial DNA Variability in Bosnians and Slovenians*, 67 ANNALS HUM. GENETICS 412-25 (2003) (illustrating frequency of H haplogroup is 24% in Finland, 26.8% in Scotland, and 45% in Poland). *See also* Pereira et al., *supra* note 21, at 7 (noting the use of SNPs to more closely examine haplogroups demonstrates significant inter-relatedness below the haplogroup level and suggests that "phylogenetic dissection of mtDNA haplogroups is revealing gradients previously hidden on the Eurasian scale").

109. That the SWGDAM database is not representative also arguably invalidates the FBI's use of a confidence interval to extrapolate from the database to a sub-population. Use of such margins of error presupposes random distribution in the population. WITTE, *supra* note 58, at 214.

110. *See* NRC II (1996), *supra* note 5, at 34 (noting that database of "a few hundred persons" is necessary even to have "some statistical accuracy" in estimating nDNA frequencies); RUDIN & INMAN, *supra* note 1, at 147 ("[T]he mtDNA databases are not yet large enough to be confident that an occurrence of a particular type divided by the number of people in the database gives a reasonable estimate of the frequency.").

underlying those databases is incomplete[111] and because individuals in those categories come from a much wider geographic range than the smaller, more narrowly targeted databases.

Take the African-American database, containing 1148 profiles, as an example. Assume, for the sake of illustration, that the database contains 1200 profiles and is representative of the relevant population. Given that many mtDNA profiles occur with a frequency of at least 1 per 1000, or 0.1%, one can state that there is a 99.9% chance that any particular person in the relevant population will *not* exhibit this profile. In a 1200-sequence database presumed to be representative, the probability of completely missing a profile that is as common as 1 in 1000 is 30.1%.[112] In an *unrepresentative* database, where the distribution of sequence frequencies is unknown, such a probability cannot even be calculated. To predict the frequency of a given mtDNA frequency, the database needs to be both representative *and* sufficiently large.

Any determination of how large the database must be to provide accurate frequency estimates must take into account what scientists presently know about mtDNA haplotype frequencies. For example, over 50% of known mtDNA sequences have been observed just one time.[113] Therefore, in a database of one thousand samples, one would expect that five hundred of the samples occur only once in the database. If an additional 1000 individuals were sampled, the likelihood that those same five hundred samples would each be observed only once decreases significantly. In addition, given that mtDNA sequence frequencies in the population are yet unknown, ancestral categories not yet accounted for in the SWGDAM database may have significantly higher or lower frequencies of unique sequences. So as not to underestimate the frequency of a suspect's profile in determining the probative value of a "match," databases should be constructed to

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111. Melton, *supra* note 79, at 46 and Budowle et al., *supra* note 51, at 31-32, argue that the SWGDAM database is valid based on the "lack" of significant population structure in the sequence-specific oligonucleotide ("SSO") types and the similarity of the haplogroup frequencies in the database to various non-U.S. populations. This argument has two fundamental flaws. First, these studies look only to haplo*group* or SSO diversity, not to haplo*type* diversity, even though it is at the haplo*type* level that many population differences are detected and that the FBI generates its inclusion statistics. *See, e.g.*, Malhi et al. (2002), *supra* note 64, at 906. Second, their use of the F_{ST} – an ostensible measure of genetic population differentiation – is controversial. Melton (2001) relies on the F_{ST} to tout the homogeneity of North American populations. But the assumptions of F_{ST} make the statistic inherently biased against the detection of diversity among populations. Jeffrey C. Long & Rick A. Kittles, *Human Genetic Diversity and the Nonexistence of Biological Races*, 75 HUM. BIOLOGY 449, 450 (2003). Even if the use of F_{ST} indicates no significant subdivision in a haplogroup or SSO, such evidence is irrelevant to whether significant differences exist in frequencies of haplo*types* in various populations.

112. To calculate the cumulative probability that a database containing 1200 samples should miss a particular such profile, one must use the product rule to determine the probability that an event with probability .999 will not occur in 1200 trials. As it turns out, $(0.999)^{1200} = 0.301$.

113. *See* Tishkoff & Kidd, *supra* note 79, at S25. Of the 1771 individuals in the SWGDAM Caucasian database as of 2000, 72% of the profiles appeared only once in the database. Allard et al., *supra* note 34, at 1216; Parsons & Coble, *supra* note 15, at 305 (noting rare profiles exceed 50% of observed sequences in SWGDAM database). *See generally* Kittles & Keita, *supra* note 68, at 88-89.

minimize the probability of missing a rare profile and to maximize the probability that the database represents all possible profiles in the relevant population.

To the authors' knowledge, no published article on mtDNA typing has suggested how large a database would have to be to generate accurate mtDNA sequence frequency statistics for forensic use, and the authors do not speculate in this article as to how large such a forensic database would have to be. Scientists are still in the process of collecting mtDNA samples and determining the extent of geographic and ancestry-related substructure in the distribution of mtDNA profiles. The scientific community must arrive at an accurate estimate of the level of mtDNA variation, and the distribution of profiles, in the population before determining how many, and what type of, samples would be necessary to create a statistically sound forensic mtDNA database.

C. Quality Control Issues

There is reason to believe that the SWGDAM database contains errors that potentially affect the quality of the data and therefore potentially undercut the ability of the database to generate accurate statistical estimates. Not surprisingly, mtDNA sequencing – which involves typing the 600-base HVI/II region and hand-transcribing the positions at which the sequence differs from the CRS – results in many more transcription errors than does nDNA testing.[114] Human errors also stem from incorrect recording or exchanging of laboratory samples or misreading of machine outputs.[115] Outside scientists have found several transcription errors in the SWGDAM database.[116] Upon the publishing of such errors, the FBI has conducted its own studies and found even more errors.[117]

The lack of quality control has subjected the SWGDAM database to interna-

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114. *See, e.g.*, Carina Dennis, *Error Reports Threaten To Unravel Databases of Mitochondrial DNA*, 421 NATURE 773, 773-74 (2003) (reporting observation by Dr. Neil Howell that Dr. Forster's error-detection method may underestimate number of errors in databases); Forster, *supra* note 25, at 2:

> In forensics, accurate comparative mtDNA database are needed to assess the probability that an mtDNA profile from a crime stain is likely to derive from a suspect rather than from any other member of the population, so the number of errors in forensic journals listed in Table 1 does not engender confidence.

Corinna Herrnstadt et al., *Errors, Phantom and Otherwise, in Human MtDNA Sequences*, 72 AM. J. HUM. GENETICS 1585, 1585 (2003).

115. Hans-Jurgen Bandelt et al., *Detecting Errors in MtDNA Data by Phylogenetic Analysis*, 115 INT'L J. LEGAL MED. 64, 64 (2001).

116. *See, e.g.*, Hans-Jurgen Bandelt, Antonio Salas, & Lutz-Bonengal, *Artificial Recombination in Forensic MtDNA Population Database*, 118 INT'L J. LEGAL MED. 267 (July 2004).

117. Kevin Miller & Bruce Budowle, *A Compendium of Human Mitochondrial DNA Control Region: Development of an International Standard Forensic Database*, 42 CROATIAN MED. J. 315, 316 (2002). Miller and Budowle found that "a few substitutions in some published and SWGDAM sequences were clearly reviewed to be anomalous." *Id.* Sources for confusion in the data, according to the authors, include failure to conform to a standardized numbering system, non-recordation of insertions and deletions in the polycystine stretch of HVI, and differences between the number of sequences in the literature and the number of sequences in GenBank/EMBL. *Id.* While the authors subsequently recommended that the public data containing the errors be used for

tional criticism. For example, several European scholars, after inspecting the database for errors, have written about its "poor quality," saying that reliance on its utility "inhibit[s] the generation of a new reliable mtDNA database in the United States."[118]

For the African-American database, the researchers conducted a phylogenetic analysis – the use of computer algorithms to detect inconsistencies in the evolutionary position of individual sequences resulting from miscoding or recombining of computer records during data entry – and observed "a number of major deficiencies," suggesting a confusion of specimens in the laboratory or faulty data entry.[119] Because the FBI's raw data is not publicly available for reexamination and correction by independent scientists, the European scientists were unable to manually look for errors; instead, the FBI was notified of their findings with the hope that the FBI would conduct a comprehensive investigation into the accuracy of the database profiles.[120]

Instead, the FBI's response to these reports of errors was to conduct only a partial inquiry for error correction.[121] Specifically, when notified of the findings of error by the European scholars, the FBI conducted a phylogenetic analysis and partial manual verification of its database, uncovering additional errors. Despite finding a number of errors in the manual verification process, the FBI only checked a small percentage of the SWGDAM profiles manually. For example, only 196 of the 1148 sequences in the African-American database were so checked,[122] leaving unknown how many other errors exist in the African-American database or in the other subpopulation databases. Given that only the FBI has the ability to conduct a complete inquiry into errors in its database, and that the FBI currently refuses to grant non-employees uncontrolled access to its raw data,[123] no apparent means exist for independent reviewers to ensure that the SWGDAM database is now free of such errors.[124]

"investigative or research purposes only and not for the assignment of weight regarding forensic matches," the authors did not address how, if at all, the FBI addressed the SWGDAM sequencing errors. *Id.*

118. Hans-Jurgen Bandelt, Antonio Salas, & Antonio Bravi, *Problems in FBI MtDNA Database*, 305 SCIENCE 1402, 1403-04 (Sept. 2004).

119. *Id.*

120. *Id.*

121. *Id.*; Bruce Budowle et al., *Addressing the Use of Phylogenetics for Identification of Sequences in Error at the SWGDAM Mitochondrial DNA Database*, 49 J. FORENSIC SCI. 1, 1256 (Nov. 2004).

122. Budowle et al., *supra* note 121, at 1259 tbl.2, 1260 tbl.3.

123. *See* D. Michael Risinger & Michael J. Saks, *Rationality, Research and Leviathan: Law Enforcement-Sponsored Research and the Criminal Process*, 2003 MICH. ST. L. REV. 1023, 1047 & n.107 (2003) (noting that current FBI rules require an FBI employee to be a co-author on any article as a condition for gaining access to agency data).

124. AFDIL researchers have already suggested possible improvements to quality control in mtDNA testing. *See* Coble et al., *supra* note 21, at 139 Table (minimizing human intervention in process and reviewing all results by two individuals who conducted independent evaluations); Just, et al., *supra* note 23, at S148-49 (noting that implementation of a high-throughput robotic system for population databasing backed up by multiple scientists

D. Problems with the Counting Method

The present method of estimating the frequency of a sequence in the population – counting the number of hits in the SWGDAM forensic database and calculating an upper-bound confidence interval around that number – does not take into account the observations of the profile in the suspect sample and in the evidence sample. In cases in which the suspect sample and the evidence sample are consistent for reasons other than the suspect being the source, this approach systematically underestimates the frequency of questioned mtDNA profile in the relevant population. This issue should be addressed and resolved by attorneys and courts, given the effect such underestimations can have on the presumption of innocence.

Again, consider the hypothetical in which a suspect's mtDNA sequence is consistent with that found at the crime scene, and the shared profile is compared to the SWGDAM African-American database, where the number of profiles is 1148 ($n = 1148$) and where the database is assumed to be representative and random. If an examiner searches the database and does not find the shared profile, she reports the number of hits as zero, reporting that the suspect's profile has never been seen before in the database. But the suspect's profile has arguably been seen twice: once in the suspect himself and once in the evidence sample. Failing to count the observation of the profile in the suspect as a hit in the database seems to ignore useful and relevant information. The suspect is, after all, part of the relevant population, and there appears to be no reason to consider him outside the scope of the database of profiles. Thus, rather than conduct a frequency estimate based upon zero hits in a database of 1148 samples, the examiner should, at the very least, conduct a frequency estimate based upon one hit in a database of 1149 ($n+1$) samples.

Moreover, the election to disregard the observation of the profile in the suspect's sample and in the evidence sample may implicate the presumption of innocence. Most jurisdictions have a jury instruction providing that, unless and until the suspect is proven guilty by the prosecution and the jury renders a verdict of guilty, the suspect in a criminal trial is presumed innocent in the eyes of the jury.[125] If that

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checking key laboratory steps increases the size and range of current mtDNA databases and decreases potential sources of error in creating databases).

125. *See, e.g.*, Pattern Jury Instr. (criminal cases) First Circuit § 3.02 (1998) ("It is a cardinal principle of our system of justice that every person accused of a crime is presumed to be innocent unless and until his/her guilt is established beyond a reasonable doubt. The presumption is not a mere formality. It is a matter of the most important substance."); Manual of Model Crim. Jury Instr. Eighth Circuit § 3.05 (2005) ("The presumption of innocence alone is sufficient to find the defendant not guilty and can be overcome only if the Government proves, beyond a reasonable doubt, each essential element of the crime charged."); Conn. Prac., Crim. Jury Instr. § 2.8 (3d ed. 2005) ("[T]he accused is presumed to be innocent until he is proved guilty. That means that at the moment when he was presented before you for trial, he stood before you free of any bias, prejudice or burden arising from his position as the accused."); Criminal Jury Instr. for the District of Columbia § 2.08 (2004) ("This presumption of innocence remains with the defendant throughout the trial unless and until s/he is proven guilty beyond a

presumption is taken seriously, the evidence sample and the suspect's sample should not be presumed to confirm the same person. After all, the question of whether or not the suspect actually contributed the sample is the very question to which the mtDNA frequency estimate is relevant. Thus, the most appropriate assumption is that the suspect's profile has been seen twice: once in the evidence sample and once in the suspect's sample. Therefore, in the hypothetical, the FBI should conduct a frequency estimate based upon two hits in a sample of 1150 (n+2) sequences.[126] Such modifications to the "counting method" affect – in some cases, dramatically so[127] – the reported frequency estimates and therefore should be provided to the jury.

IV. COMMUNICATION OF SWGDAM DATABASE ISSUES TO COURTS AND JURIES THROUGH ADMISSIBILITY CHALLENGES

Notwithstanding these inherent problems with using the SWGDAM database to calculate the statistical significance of a mtDNA "inclusion," the burgeoning use of mtDNA evidence in criminal trials throughout the country has continued unabated. The reason, as discussed below, is not that large numbers of courts have rejected challenges based on these problems; rather, courts and defense counsel are generally not *aware* of the existence or scope of the scientific dispute over the reliability of the FBI's methods. It is critical that courts be apprised of these issues through admissibility challenges so that they may have the opportunity to examine the issues and ensure that only reliable scientific evidence is used against criminal

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reasonable doubt."); Haw. Crim. Jury Instr. § 3.02 (2005) ("You must presume the defendant is innocent of the charge against him/her. This presumption remains with the defendant throughout the trial of the case, unless and until the prosecution proves the defendant guilty beyond a reasonable doubt."); Tenn. Pattern Jury Instr. Criminal § 2.01 (2005) ("This presumption [of innocence] remains with the defendant throughout every stage of the trial, and it is not overcome unless from all the evidence in the case you are convinced beyond a reasonable doubt that the defendant is guilty."); Va. Model Jury Instr. Crim. § 2.100 (2005) ("This presumption of innocence remains with the defendant throughout the trial and is enough to require you to find the defendant not guilty unless and until the Commonwealth proves each and every element of the offense beyond a reasonable doubt.").

126. Other commentators have advocated such modification to the counting method. *See* BALDING, *supra* note 68, at 99 ("to make some allowance for sampling variability, it is advantageous to include both the crime scene and the defendant profiles with those of the population database."); Marlan D. Walker, Note, *Mitochondrial DNA Evidence in* State v. Pappas, 43 JURIMETRICS J. 427, 437 (2003) ("[T]he fact that T has been observed once in a sample of 1220 individuals (the 1219 in the database plus the defendant) suggests that the sample frequency of 0/1219 used to form the confidence interval may be understated.").

127. Using the FBI's formula for calculating confidence intervals, *see* FBI MtDNA Protocols (2004), *supra* note 8, at § 11 at 10, the upper bound 95% confidence limit for 0/1148 is 0.26%. The upper bound 95% confidence limit for 2/1150 is still small, but almost twice as great, 0.42%. Using a more conservative 99% confidence interval increases the frequency estimates a bit more. The upper bound 99% confidence limit for 0/1148 is 0.4%. The upper bound 99% confidence limit for 2/1150 is 0.49%. The smaller the database, the more pronounced the effects of such modifications. The SWGDAM Chinese/Taiwanese population database has 356 profiles. The upper bound 95% confidence limit for 0/356 is 0.84%. The upper bound 95% confidence limit for 2/358 is 1.33%. Changing the confidence limits to 99% causes the upper bound confidence limits increase to 1.3% and 1.6%, respectively. While the practical effects of these modifications may be marginal in most cases, the difference may be material in a particular case, and accuracy is a worthy goal in itself.

defendants.

A. Legal Standards Governing Admission of Scientific Evidence

Most U.S. courts employ one of two tests to determine admissibility of novel scientific evidence. A number of jurisdictions follow *Frye v. United States,*[128] admitting novel scientific evidence only when the methodology used is "generally accepted" in the relevant scientific community.[129] Still others, including the federal courts, follow the test set forth by the Supreme Court in *Daubert v. Merrell Dow Pharmaceuticals, Inc.*,[130] in which the Court construed the Federal Rule of Evidence governing admission of expert testimony to require that trial judges make an independent judicial determination whether the scientific evidence is sufficiently reliable.[131] Still other courts follow some combination of the two dominant approaches or another, more idiosyncratic approach.[132] Because the vast majority of jurisdictions subscribe to either *Frye*, *Daubert*, or a hybrid standard, this section focuses on the two dominant standards.

The *Frye* standard requires that the scientific community approve of a technique before it may be used in courts. If scientists, significant either in number or expertise, publicly oppose a new technique as unreliable, the trial judge must exclude the evidence.[133] The *Frye* standard is inherently conservative in that it requires those experts who are in a position best to understand and review a procedure to pass on its reliability. While the waiting period that scientific evidence and techniques must endure before gaining legal acceptance under *Frye*

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128. Frye v. United States, 293 F. 1013 (D.C. Cir. 1923).

129. *Id.* at 1014:

> Just when a scientific principle or discovery crosses the line between the experimental and demonstrable stages is difficult to define. Somewhere in this twilight zone the evidential force of the principle must be recognized, and while courts will go a long way in admitting expert testimony deduced from a well-recognized scientific principle or discovery, the thing from which the deduction is made must be sufficiently established to have gained general acceptance in the particular field in which it belongs.

130. Daubert v. Merrell Dow Pharmaceuticals, Inc., 509 U.S. 579 (1993).

131. *Id.* at 589. The *Daubert* test was subsequently refined by the Court and incorporated into the relevant Federal Rule of Evidence. *See* Kumho Tire v. Carmichael, 526 U.S. 137, 138 (1999) (extending *Daubert* more generally to non-scientific expert testimony); General Electric Co. v. Joiner, 522 U.S. 136, 136-37 (1997) (applying abuse-of-discretion standard of review to trial court's determination of admissibility under *Daubert*); Comment to Fed. R. Evid. 702 (discussing 2000 amendments to Federal Rules of Evidence in response to *Daubert* that codify the requirement that "the testimony is the product of reliable principles and methods" and that "the witness has applied the principles and methods reliably to the facts of the case").

132. *See generally* Andrew R. Stolfi, Note, *Why Illinois Should Abandon* Frye*'s General Acceptance Standard for the Admission of Novel Scientific Evidence*, 78 CHI.-KENT L. REV. 861 (2003) (discussing standards for admission of scientific evidence in all U.S. jurisdictions).

133. People v. Pizarro, 3 Cal. Rptr. 3d 21, 44 (Cal. Ct. App. 2003); United States v. Porter, 618 A.2d 629, 633-34 (D.C. 1992).

has generated criticism,[134] the *Frye* standard remains the standard for admission of scientific evidence in a number of jurisdictions.

In *Daubert*, the Supreme Court rejected the *Frye* standard for use in federal court and concluded that that the Federal Rules of Evidence require judges to act as gatekeepers, making a "preliminary assessment of whether the reasoning or methodology underlying the testimony is scientifically valid and of whether that reasoning or methodology properly can be applied to the facts at issue."[135] The Supreme Court set out a flexible and nonexclusive list of factors for courts to consider, including (1) whether the science underlying the questioned evidence can be, and has been, empirically tested; (2) whether the science has been subjected to peer review and publication; (3) whether there is a known or potential rate of error; (4) whether standards exist and are maintained to control the technique's operation; and (5) whether the scientific proposition at issue has been generally accepted.[136] If, after considering these and any other relevant factors, the trial judge determines that the scientific methodology is both "reliable" and "fit[s]" the circumstances of the case,[137] the evidence is admissible.[138] While *Daubert* involved interpretation of a Federal Rule, many states have adopted the *Daubert* standard.[139]

As numerous commentators have observed and courts have held, evidence of a DNA inclusion or match has little meaning without a sense of the frequency of the profile in the population.[140] If the frequency of a profile from evidence at the crime scene were found in 99% of the general population, for example, the fact that the defendant is a potential contributor would have only nominal probative value. And while expert testimony as to frequency statistics may not be necessary where the issue is the frequency of people with blond hair or other information easily accessible through common human experience, most jurors have little if any

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134. *See* MICHAEL J. SAKS ET AL., ANNOTATED REFERENCE MANUAL ON SCIENTIFIC EVIDENCE, SECOND 73 (2004) (stating that "*Frye* is often criticized as overly conservative, for it imposes a protracted waiting period that valid scientific evidence and techniques must endure before gaining legal acceptance.").

135. *Daubert*, 509 U.S. at 592-93.

136. *Id.* at 593-94.

137. *Id.* at 597-98.

138. *See id.* (stating that "[p]ertinent evidence based on scientifically valid principles" satisfies the Federal Rules of Evidence).

139. *See* SAKS ET AL., *supra* note 134, at 78 & n.9 (listing jurisdictions that have adopted or declined to reject *Daubert*).

140. *See* BUTLER, *supra* note 12, at 270 ("When 'failure to exclude' is the interpretation for reference and evidence samples, then a statistical estimate of the significance of a [mtDNA] match is needed."); *Porter*, 618 A.2d at 640 (indicating that a statistical assessment of significance of DNA is at the core of its admission and "underlying method of arriving at that calculation must pass muster under [*Frye*]"). *See generally* Kaye & Sensabaugh, *supra* note 5, at 545 & n.269 (noting a number of courts that have concluded that reliable statistical methodology must accompany science to be admissible). *But see id.* at 546 & n.275 (advocating that jurors need not be presented with a particular number if the profile is shown to be exceedingly rare by admissible scientific methodology).

intuition as to the relative frequencies of mtDNA sequences in the population.[141] Thus, as a legal matter, the statistical methodology used to estimate the significance of an inclusion or match must independently pass muster under either the *Frye* or *Daubert* standard of admissibility for scientific evidence.

Neither *Daubert* nor *Frye* sets a more "lenient" threshold for admissibility than the other; both require a pretrial showing of scientific validity. The principal distinction between the two lies in the question, "who decides the issue of scientific validity?" Under *Frye*, the trial court defers to the opinions of scientists as to whether a particular scientific advancement is valid. Under *Daubert*, the court considers the views of scientists but, ultimately, the court itself determines the scientific validity of the evidence. Under either standard, evidence with a strong scientific foundation will likely be admitted; evidence lacking such a foundation will likely be excluded.[142] For the reasons given in Part III, this Article submits that forensic mtDNA does not yet have a strong enough scientific underpinning to satisfy either the *Frye* or *Daubert* standards for admissibility, and courts should take great care prior to admitting such evidence – whether by the prosecution seeking to inculpate a defendant or by the defense seeking to convince the jury that an uncharged third party is the true perpetrator.

B. The Importance of Well-Litigated Admissibility Challenges to the Reliability of MtDNA Evidence Used Against Criminal Defendants

The stakes in admitting mtDNA evidence without a proper scientific foundation are high; because reliance on the SWGDAM database potentially underestimates the frequency statistic, jurors typically hear that the defendant belongs to the less than 1% of the population who could have contributed the mtDNA, and, therefore, are given the sense that the defendant likely committed the offense.[143] An additional concern is that jurors will confuse mtDNA with nDNA, which has an

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141. This point should not obscure forensic mtDNA testing's effectiveness in the exoneration of suspects. The issue when *excluding* an individual is simply whether the sequencing was done properly at each stage. But when mtDNA typing is used to *include* an individual as a suspect, the information has no meaning without determining the statistical significance of the inclusion.

142. *See* SAKS ET AL., *supra* note 134, at 96-97 (comparing *Daubert* and *Frye* tests).

143. One dilemma facing courts is whether to consider the defendant's apparent ancestry in determining which database to use to determine a frequency estimate. At least one court has suggested that the presumption of innocence requires that no assumptions be made as to the perpetrator's ancestry and, thus, the statistics reported to the jury should not assume that the perpetrator is of a particular ancestry. *See, e.g.*, People v. Prince, 36 Cal. Rptr. 3d 300, 305-07 (Cal. App. 5th Dist. 2005) (reaffirming and following *Pizarro*'s disapproval of how the prosecution, when it presented the Hispanic profile frequency, impermissibly assumed the *perpetrator*, like the defendant, was Hispanic). On the other hand, where a perpetrator is likely from a particular ethnic community, it may be to the defendant's advantage to consider the perpetrator's ancestry, because the observed haplotype may be quite common in a particular ancestral sub-population, even though very uncommon in the general U.S. population. *See* Julian Adams, *Nuclear and Mitochondrial DNA in the Courtroom*, 13 J. L. & POL'Y 69, 88 (2005) (giving example of *Passino* case in which perpetrator of crime in remote trailer camp was likely of Abnaki ancestry).

aura of infallibility from its coverage in the popular press and is viewed as having an almost mysterious quality as a tool of identification. In fact, recent jury studies suggest that jurors may confuse different types of DNA, and may see all DNA evidence as infallible.[144]

If litigants do not timely apprise trial judges of these scientific issues, courts will not have the opportunity to determine whether, in light of such issues, the proffered mtDNA inclusion statistics that will be reported to the jury are scientifically unsound or are not generally accepted in the scientific community. Due in large part to the fact that mtDNA evidence is often admitted against a defendant without any challenge to its admissibility, most courts are simply unaware of the unique aspects of mtDNA typing as compared to nDNA typing, and have not been exposed to the scientific literature discussing the problems with the SWGDAM database. While the majority of the few courts that *have* ruled on the admissibility of mtDNA evidence have approved of its use against a defendant, judicial consideration of this form of evidence is in its infancy. To date, only two published opinions from federal courts address the admissibility of mtDNA evidence, along with a handful of published state trial and appellate decisions. Significantly, no state court of last resort has yet ruled mtDNA evidence admissible under a *Frye*-type standard.[145] Only one federal appellate court and two state supreme

144. In December 2003, the Public Defender Service for the District of Columbia commissioned a jury poll through Lake Snell Perry and Associates, an independent polling firm, of 1000 potential D.C. jurors. *See* http://www.pdsdc.org/SpecialLitigation/SLDSystemResources/Brady%20Poll%20Results,%20December%202003.pdf. The poll showed that, on a scale of 0 to 10, 10 being most persuasive, the mean response for DNA evidence was 9, as compared to 6.6 for the testimony of the accuser. *Id.* at 2. Thirty-one percent of those polled stated that DNA evidence can never "be wrong." *Id.* at 4. Thirty percent stated that mistakes are made "almost never" with respect to DNA evidence. *Id.* When asked which was more reliable, nuclear or mtDNA, 32% answered that the two forms of DNA are "equally" reliable, while 11% answered that mtDNA was *more* reliable. *Id.* A study commissioned by the National Institute of Justice showed that, after sitting through a mock trial involving mtDNA, a large percentage of jurors fell victim to fallacies and misperceptions about DNA in general, and mtDNA in particular. *See generally* B. MICHAEL DANN ET AL., TESTING THE EFFECTS OF SELECTED JURY TRIAL INNOVATIONS ON JUROR COMPREHENSION OF CONTESTED MTDNA EVIDENCE, FINAL TECHNICAL REPORT 41-54 (Dec. 30, 2004), *available at* http://www.ncjrs.gov/pdffiles1/nij/grants/211000.pdf. *Cf.* Michael J. Saks & Jonathan J. Koehler, *The Coming Paradigm Shift in Forensic Identification Science*, 309 SCIENCE 892, 893 (2005) ("[E]rroneous forensic science expert testimony is the second most common contributing factor to wrongful convictions, found in 63% of those cases.").

145. For state courts that have admitted mtDNA evidence under *Frye*, see Magaletti v. State, 847 So. 2d 523, 526-29 (Fla. Dist. Ct. App. 2003); Wagner v. Maryland, 864 A.2d 1037, 1044-50 (Md. Ct. Spec. App. 2005); People v. Holtzer, 660 N.W.2d 405, 409-11 (Mich. Ct. App. 2003); People v. Ko, 757 N.Y.S.2d 561, 563 (N.Y. App. Div. 2003); People v. Klinger, 713 N.Y.S.2d 823, 831 (N.Y. Sup. Ct. 2000). *See also* Adams v. State, 794 So. 2d 1049, 1057 (Miss. Ct. App. 2001) (stating in one sentence that mtDNA expert was qualified and that, because expert claimed evidence was "generally accepted," defense challenge to testimony was denied). *Contrast* RUDIN & INMAN, *supra* note 1, at 195 (reporting on *State v. Crow*, No. 96-1156 (Fla. 18th Cir. Ct. May 14, 1998), in which the trial judge "ruled that the results of an mtDNA test did not meet the *Frye* standard and were inadmissible as evidence. He based his opinion on his understanding that the FBI database was too small and was insufficient to provide reliable statistical conclusions. [The judge] further found that the 'counting method' failed to provide meaningful comparison that would assist, rather than confuse, the jury.")

courts have ruled mtDNA admissible under a *Daubert*-like standard.[146] In many of these cases, the evidence was admitted without the trial court ever hearing a witness contrary to the government's forensic scientist vouching for admission of the evidence.[147] None of the published decisions discusses in a meaningful way problems with the content of the databases used to generate frequency statistics.[148] In addition, scholars in legal academia, while recognizing general issues with respect to admission of mtDNA evidence in criminal trials, have not yet begun to discuss the database problems at length.[149]

The lack of awareness on the part of courts and other participants in the legal system with respect to the problems with the SWGDAM database and other important scientific issues is compounded by the fact that most practicing forensic scientists are not part of, and perhaps not even aware of, the conversation taking place among medical geneticists, evolutionary biologists, and molecular anthropologists concerning the extent of genetic individuality and diversity of mtDNA.[150] Additionally, because most criminal defendants do not have the resources to mount complicated, expensive admissibility challenges to forensic mtDNA evidence, the

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146. For federal courts that have admitted mtDNA under a *Daubert* standard, see United States v. Beverly, 369 F.3d 516, 527-31 (6th Cir. 2004); United States v. Coleman, 202 F. Supp.2d 962 (E.D. Mo. 2002). For state courts that have admitted mtDNA under a *Daubert* standard, see Lewis v. State, 889 So. 2d 623, 668-74 (Ala. Crim. App. 2003); State v. Pappas, 776 A.2d 1091, 1100-13 (Conn. 2001); State v. Underwood, 518 S.E.2d 231 (N.C. App. 1999); State v. Council, 515 S.E.2d 508 (S.C. 1999). *Cf.* State v. Scott, 33 S.W.3d 746, 759-60 (Tenn. 2000) (admitting mtDNA evidence by statute that looks at reliability of proposed evidence).

147. *See Beverly*, 369 F.3d at 530-31 (stating court heard only from prosecution's expert); *Lewis*, 889 So. 2d at 673-74 (same); *Magaletti*, 847 So. 2d at 526-27 (same); *Klinger*, 713 N.Y.S.2d at 824 (same); *Scott*, 33 S.W.3d at 752 (upholding denial of hearing as not abuse of discretion and deeming harmless trial court's failure to give defendant funds to hire DNA expert).

148. The Public Defender Service for the District of Columbia raised many of the issues discussed in this Article through testimony, affidavits, and articles from statisticians, genetic anthropologists, molecular biologists, and population geneticists at a *Frye* hearing in D.C. Superior Court in July 2004. While the trial judge admitted the mtDNA evidence, the jury rendered a verdict of not guilty on all counts – perhaps reflecting the exculpatory nature of some of the mtDNA evidence and/or the challenges to the prosecution's frequency statistics brought out at trial. *Compare* Memorandum & Order, United States v. Chase, No. F-7730-99, 2005 WL 757259 (D.C. Super. Jan. 10, 2005) (denying defendant's motion to exclude mitochondrial DNA test results in the Ida Chase case), *with* Henri E. Cauvin, *Woman Acquitted in 1996 Slaying of Md. Salesman*, WASH. POST, Feb. 10, 2005, at B01 (reporting Mrs. Chase's acquittal).

149. *See, e.g.*, Adams, *supra* note 143, at 87-89 (focusing on heteroplasmy issues; touching upon potential relevance of perpetrator's ancestry in determining mtDNA frequency estimate); Kiran Bisla, *It All Came Down to a Single Hair: The Probability of Exclusion vs. the Probability of Guilt Through the Use of Mitochondrial DNA Evidence in* State v. Pappas, 26 WHITTIER L. REV. 263, 296-98 (2004) (analyzing *Pappas* decision and briefly noting that court dismissed arguments relating to convenience samples and fact that "FBI was in complete control of the contents of the database"); Edward K. Cheng, *Mitochondrial DNA: Emerging Legal Issues*, 13 J. L. & POL'Y 99, 107-18 (2005) (focusing on advantages of mtDNA testing over microscopic hair analysis and on privacy issues in mtDNA databanks); Paul C. Giannelli, *Mitochondrial DNA*, 19 CRIM. JUST. 54, 54-56 (Winter 2005) (primarily discussing problems with contamination and chain of custody); FAIGMAN ET AL., *supra* note 4, at § 25 (discussing legal and scientific issues with DNA typing).

150. *See, e.g.*, Pereira et al., *supra* note 21, at *2 (study is "an attempt to extend the use of phylogeographic approaches to mtDNA forensics"); RUDIN & INMAN, *supra* note 1, at 196 ("Forensic scientists tend to be insular and self-reliant to a fault.").

emerging and accumulated knowledge of scientists who regularly work outside the legal system appears not to be making its way into courtrooms before judges and juries. Professors Mildred Cho and Pamela Sankler express the need to bridge the gap between science inside, and outside, the courtroom:

> [The] series of arguments and counterarguments about the association between 'race' and patterns of DNA markers [that] has been unfolding in the medical genetics literature over the last four years . . . are relevant to, and should include, forensic geneticists. . . . These conversations are directly relevant to the forensics genetics community[, but they] have not been widely extended into this group. There is an urgent need to expand this debate into the field of forensics.[151]

While hiring experts to testify at admissibility hearings may be beyond a particular defendant's means, individual attorneys and defender institutions should at the very least bring to courts' attention the scientific studies discussed in articles such as this one, so that courts will begin to take notice of evidence of both a lack of general acceptance in the scientific community of using the SWGDAM database to generate mtDNA frequency estimates, which may render the mtDNA evidence inadmissible under *Frye*, and a lack of scientific validity in using the database, evidence that, if accepted, would potentially render the mtDNA evidence inadmissible under a *Daubert*-like standard.

CONCLUSION

What can be done to ensure that mtDNA evidence is used fairly and effectively as "inclusion" evidence? In the first instance, both forensic scientists and the broader scientific community should work to improve the science of mtDNA frequency statistics to provide a sufficient scientific underpinning for mtDNA to support its admission in criminal cases. Scientists should also follow the lead of laboratories such as AFDIL by increasing the discriminatory power of mtDNA typing and decrease the chance of a false inclusion, by typing additional locations in both the control region and coding region of the mtDNA genome, and by taking advantage of SNP technology.

Additionally, the SWGDAM forensic mtDNA database should be corrected and expanded. Specifically, before mtDNA evidence is used as inclusion evidence in the courtroom, the scientific community should collect more data on human migration patterns, including migration patterns within the United States, to identify possible locations of mtDNA clusters based on historical developments. Upon generating that data, the scientists should conduct phylogeographic as well as phylogenetic studies to determine not only the full diversity of mtDNA

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151. Mildred K. Cho & Pamela Sankar, *Forensics Genetics and Ethical, Legal, and Social Implications Beyond the Clinic*, Vol. 36 No. 11 NATURE GENETICS SUPP. S-8, S-9 (Nov. 2004).

sequences in the population, but the geographic distribution of such sequences as well. Upon understanding the geographic distribution of such sequences, the criminal justice community should develop *regional* databases that reflect uneven geographic distribution of mtDNA sequences. In doing so, efforts should be made to vastly increase the number of sequences in those databases over the current size, by additional sampling and by accessing the raw data from the published academic studies and adding that data to the regional forensic databases.

Furthermore, because databases are only useful if their data are accurate, forensic scientists should develop and implement an additional quality assurance procedure to create a systematic means of identifying, minimizing, and correcting database errors, including the implementation of quality control measures urged by AFDIL and other reputable laboratories. Forensic scientists should also open up the raw data of the sequences in the SWGDAM database to independent researchers to permit outside review of the database, and immediately implement a complete manual verification of every sequence in the database, double-checked visually and by hand, base-pair by base-pair. Errors should not be accepted.

Moreover, we suggest modification in the counting method, by accounting for the observation of the profile in the suspect and the evidence sample when determining the number of "hits" in the database, and using a 99% confidence interval to reflect the grave need to avoid error in criminal trials. Such a process accords more with a constitutional presumption of innocence.

Finally, prosecutors, defense attorneys, and judges must be vigilant in ensuring the reliability of mtDNA evidence admitted in criminal trials. As Professor Michael Saks notes, the true "'revolution' of *Daubert* lies" in the fact that "[j]udges and lawyers, long insulated from the scientific revolution, are now obligated to become familiar with the methods and culture of science."[152] As scientists and legal professionals work to foster an understanding of the "methods and culture of science" in the courtroom, genuinely reliable evidence will appear before juries, promoting greater confidence in the outcome of criminal trials.

152. SAKS ET AL., *supra* note 134, at 78.

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Forensics
By Richard S. Schmechel

Defending With (And Against) Forensic Evidence:A Call To Share Resources

As the number of cases with forensic evidence expands markedly and as jurors become increasingly aware of, and fascinated by, forensic evidence, defense attorneys nationwide are rising to the challenge of educating themselves about how to use science in the courtroom.1

There are many difficulties inherent in learning to use forensic evidence. The quantity of information is vast, the literature is full of complicated scientific jargon, and the available research studies testing this evidence are generally uncritical. The sheer speed with which new technologies are adopted outpaces serious inquiries into their reliability. The goal of presenting accurate, meaningful, and reliable information to jurors suffers – and innocent persons are convicted.

Prosecutors circumvent many of these difficulties by accessing government forensic crime laboratory scientists, who are, not surprisingly, the strongest proponents for admission of forensic evidence. Prosecutors further rely upon the technical support, training conferences, and resource libraries offered by various national law enforcement organizations. How can defense attorneys, carrying burdensome caseloads and having only a fraction of the resources, hope to even the playing field to ensure only reliable scientific evidence appears before jurors?

The Answer

The defense bar responds best when all members work together and pool the dispersed experiences of institutional and individual defenders: collaboration.

Apart from face-to-face interaction at professional conferences2 and participation in forensic evidence listservs,3 the most fundamental need has been for an ongoing, comprehensive, national repository of defense-oriented forensic science information. Perceiving this need, the National Association of Criminal Defense Lawyers (NACDL) and the National Legal Aid and Defender Association (NLADA) created a partnership to sponsor an online Forensics Library.4

The NLADA-NACDL Forensics Library at www.nlada.org/Defender/forensics/ provides a forum for defense attorneys to share many kinds of materials. The site's nearly twenty topics include DNA evidence, allegations of child sexual abuse, and arson. Each area contains subfolders with weblinks, model pleadings, research bibliographies and articles, expert transcripts and affidavits from defense and prosecution witnesses, relevant court opinions, scientific standards or best practices, and much more.

How To Use The NLADA- NACDL Forensics Library

The Forensics Library is very user-friendly. To use it, go to the Web site and either type a keyword into the "Search" box appearing on each screen in the library, or browse through the topical folders and sub-folders on the site. Descriptions of Adobe and Word documents will appear on your screen that are subject to three levels of control.

No password is required. Many items are available to the general public. Others, of greater sensitivity, are available only to those who click on the item and certify they are practicing defense attorneys. A few of the most sensitive documents (e.g., transcripts of defense experts) are available only to those who click on the item and complete a short, individualized, online request form for the document. This three-tiered system maximizes accessibility to all users while limiting more sensitive information to defense attorneys. NACDL and NLADA members receive individually requested documents on an expedited basis.

The Forensics Library is already the first place anyone should look if they have forensic evidence in a case. Yet, although the library already has hundreds of documents, to maximize the impact of the library, help is needed to add more materials.

Submitting a brief, transcript, or any non-copyrighted material to the library is easy. Just click the "Submit a Document" button at the bottom of any Forensics Library page. In thirty seconds, you can give the document a title, designate the level of access you want it to have, browse your computer's hard drive for the document, and completely upload the document. Alternatively, documents may be faxed to a number listed on the "Submit a Document" screen.5 It may take a few days for library administrators to review submissions, but very soon your document will be available to help other attorneys in your jurisdiction and nationally who face similar issues.

Conclusion

Many other steps should be and are being taken by the defense bar to improve its use of forensic science, not the least of which is the regular attention to these matters in The Champion. The NLADA-NACDL Forensics Library, however, is an important tool for defenders seeking relevant, accessible legal and scientific information on

forensic issues for their cases. Through collaborations such as this, the defense bar will bring to light bad forensic practices and return some fairness into criminal courtrooms.

Notes

1. Jamie Stockwell, Defense, Prosecution Play to New "CSI" Savvy, Wash. Post (May 22, 2005); Kit R. Roane & Dan Morrison, The CSI Effect, US News & World Report (Apr. 25, 2005); Richard Willing, "CSI Effect" Has Juries Wanting More Evidence, USA Today (Aug. 8, 2004).

2. Many readers likely benefited from NACDL's Mid-Winter meeting on "CSI-Style Defenses" this past February. Forensic Bioinformatics, a private DNA consulting firm in which Dr. Thompson is a principal, is hosting its Fourth Annual Conference, the Science of DNA Profiling: An Expert Forum, on August 12-14, 2005. See http://www.bioforensics.com/conference05/index.html. PDSDC is sponsoring its Third Annual Forensic Science Conference, a DNA Cross-Examination College, on September 17th, 2005.

3. For example, we encourage NACDL and NLADA members to join the NACDL forensics listserv and eyewitness listserv. See http://www.nacdl.org/listserv.

4. The initial development of the Forensics Library was supported by a grant from the Gideon Project of the Open Society Institute to William C. Thompson.

5. You may fax documents to a dedicated line at 202-824-2929. While the library submission system is as technologically simple and obvious as possible, submitters may also mail hard copies of materials to the authors of this article for inclusion in the library. n

National Association of Criminal Defense Lawyers (NACDL)
1150 18th St., NW, Suite 950, Washington, DC 20036
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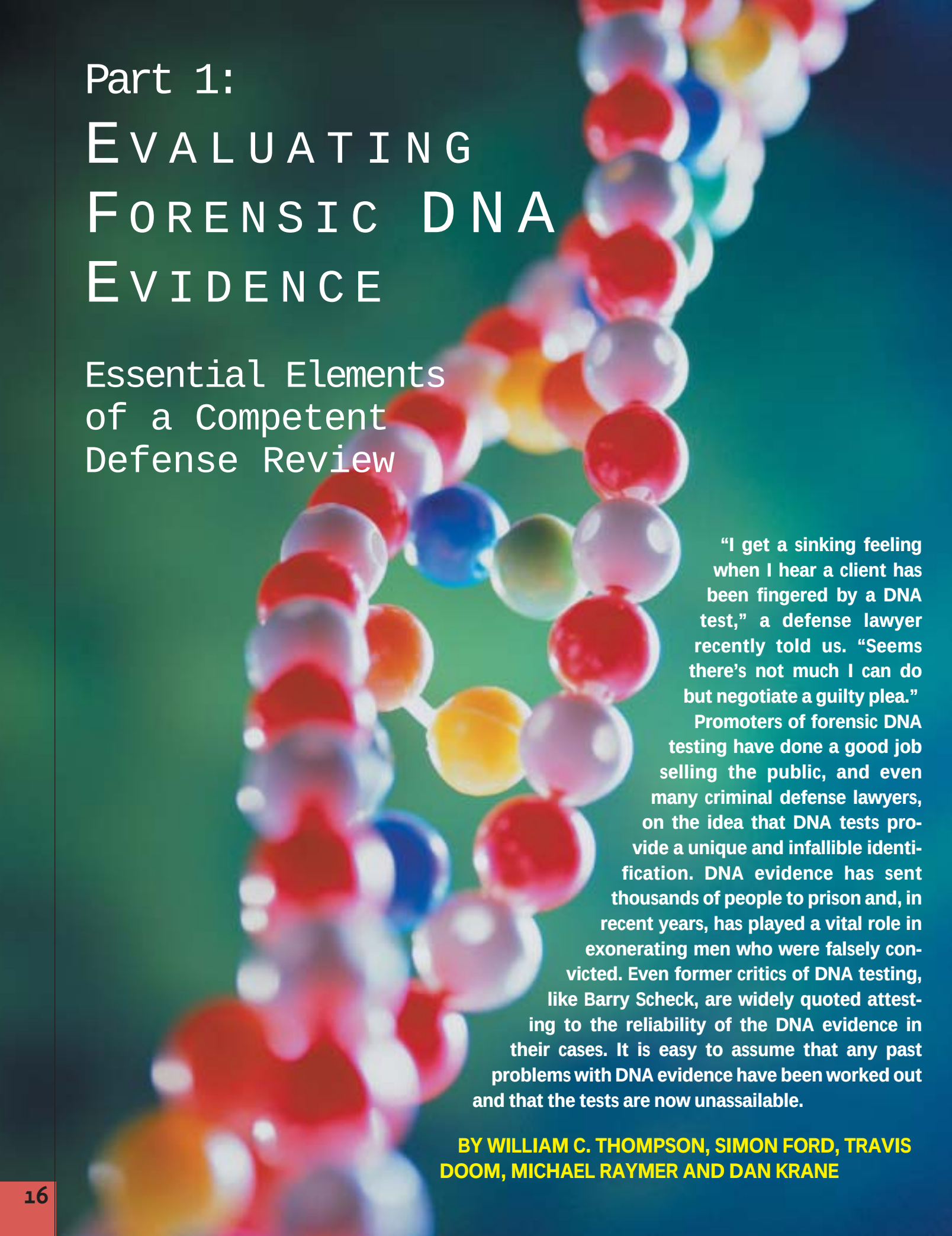


Part 1: Evaluating Forensic DNA Evidence

Essential Elements of a Competent Defense Review

"I get a sinking feeling when I hear a client has been fingered by a DNA test," a defense lawyer recently told us. "Seems there's not much I can do but negotiate a guilty plea."

Promoters of forensic DNA testing have done a good job selling the public, and even many criminal defense lawyers, on the idea that DNA tests provide a unique and infallible identification. DNA evidence has sent thousands of people to prison and, in recent years, has played a vital role in exonerating men who were falsely convicted. Even former critics of DNA testing, like Barry Scheck, are widely quoted attesting to the reliability of the DNA evidence in their cases. It is easy to assume that any past problems with DNA evidence have been worked out and that the tests are now unassailable.

BY WILLIAM C. THOMPSON, SIMON FORD, TRAVIS DOOM, MICHAEL RAYMER AND DAN KRANE

The problem with this assumption is that it ignores case-to-case variations in the nature and quality of DNA evidence. Although DNA technology has indeed improved since it was first used just 15 years ago, and the tests have the potential to produce powerful and convincing results, that potential is not realized in every case. Even when the reliability and admissibility of the underlying test is well established, there is no guarantee that a test will produce reliable results every time it is used. In our experience there often are case-specific issues and problems that greatly affect the quality and relevance of DNA test results. In those situations, DNA evidence is far less probative than it might initially appear.

The criminal justice system presently does a poor job of distinguishing unassailably powerful DNA evidence from weak, misleading DNA evidence. The fault for that serious lapse lies partly with those defense lawyers who fail to evaluate the DNA evidence adequately in their cases. This article describes the steps that a defense lawyer should take in cases that turn on DNA evidence in order to ascertain whether and how this evidence should be challenged.

Our focus here is on the most widely used form of DNA testing, which examines genetic variants called short tandem repeats, or STR's. Our goal is to explain what you need to know, why you need to know it, and how you get the materials and help you need. We leave for a future article discussion of another less common and even more problematic form of DNA testing, which examines mitochondrial DNA (mtDNA).

Understanding the lab report

The first item you need in a DNA case is the lab report. The report should state what samples were tested, what type of DNA test was performed, and which samples could (and could not) have a common source. Reports generally also provide a "table of alleles" showing the *DNA profile* of each sample. The *DNA profile* is a list of the *alleles* (genetic markers) found at a number of *loci* (plural for "locus," a position) within the human genome. To understand DNA evidence, you must first understand the table of alleles.

Figure 1 shows a table of alleles, as represented in a typical lab report. This table shows the DNA profiles of five samples — blood from a crime scene and reference samples from four suspects. These samples were tested with an automated instrument called the ABI Prism 310 Genetic Analyzer™ using a set of genetic probes called ProfilerPlus™. A company called Applied Biosystems, Inc. (ABI) developed this system for typing DNA. It is currently the most widely used method for forensic DNA typing in the United States, used by about 85 percent of laboratories that do forensic DNA testing.[1]

Across the top of the table are the names of the various loci examined by the test. The ProfilerPlus™ system examines ten loci. (Labs sometimes also run another set of genetic probes, called Cofiler™, which includes four additional loci). The alleles that the test detected at each locus are identified numbers. Thus, at locus D3S1358, the test detected alleles 15 and 16. At each locus, a person has two alleles, one inherited from each parent. In some cases, only one allele is detected, which is interpreted as meaning that by chance the person inherited the same allele from each parent. (See in Figure 1, e.g., Suspect 1's profile at locus D3S1358 and Suspect 4's profile at locus D8S1179). However, most samples will have two different alleles at each locus, as seen in Figure 1.

Each allele is a short fragment of DNA from a specific location on the human genome known as an STR (short tandem repeat). STRs are places in human DNA where a short section of the genetic code repeats itself. Everyone has these repeating segments, but the number of repetitions (and hence the length of these segments) varies among individuals. The numbers assigned to the alleles indicate the number of repetitions of the core sequence of genetic code. ProfilerPlus™ identifies and labels fragments of DNA that contain STRs. The Genetic Analyzer then measures their length and thereby determines which alleles are present.

By examining the DNA profiles, one can tell whether each suspect could or could not have been the source of the blood. Suspects 1, 2 and 4 are ruled out as possible sources because they have different alleles than the blood at one or more loci. However, Suspect 3 has exactly the same alleles at every locus, which indicates he could have been the source of the blood. In a case like this, the lab report will typically say that Suspects 1, 2 and 4 are "excluded" as possible sources of the blood, and that Suspect 3 "matches" or is "included" as a possible donor.

One of the loci analyzed is called amelogenin (Amel) and is used for typing the sex of a contributor to a sample. Males have X and Y versions of the alleles at that locus; females have only the X because they inherit two copies of the X chromosome. All of the profiles shown in Figure 1 appear to be of males.

Lab reports generally also contain estimates of the statistical frequency of the matching profiles in various reference populations (which are intended

FIGURE 1: TABLE OF ALLELES

Which suspect is a possible source of the blood? Only one of the four suspects has a DNA profile that matches the DNA profile observed in the blood sample

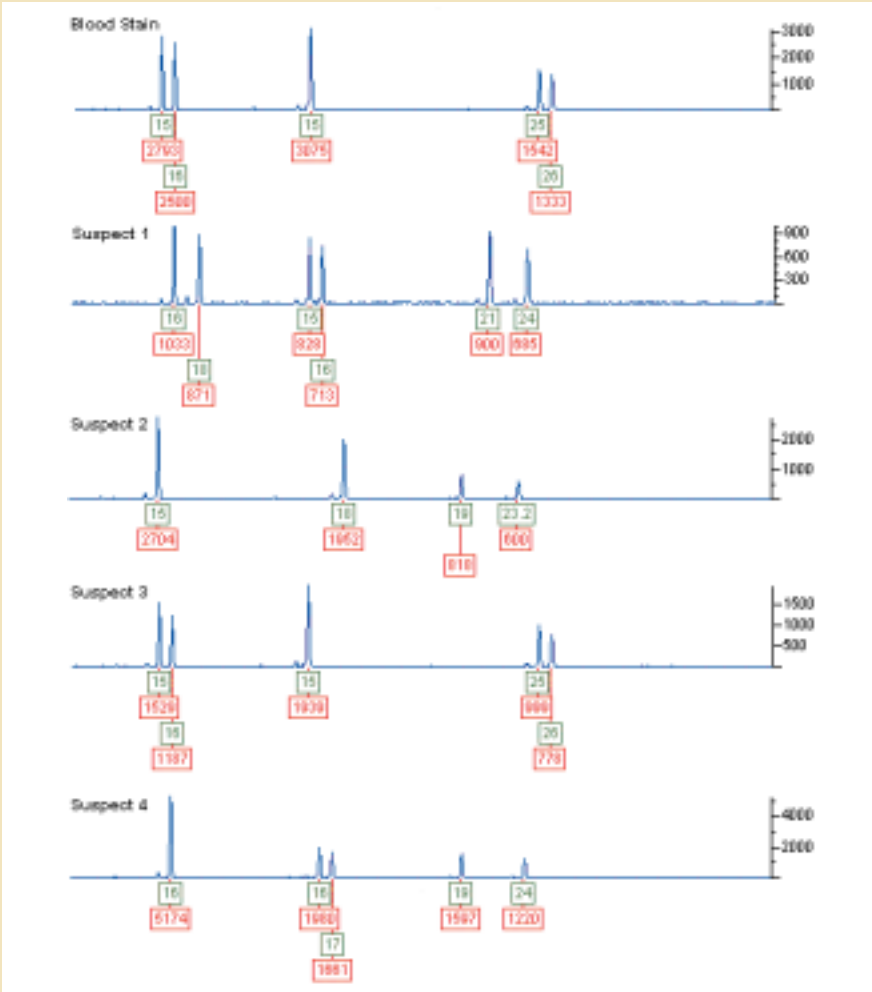
	D3S1358	VWA	FGA	Amel	D8S1179	D21S11	D18S51	D5S818	D13S317	D7S820
BloodStain	15, 16	15, 15	25, 26	XY	12,13	27,30	13,14	10,11	9,12	10,12
Suspect 1	16, 18	15, 16	21, 24	XY	12,14	27,28	13,17	11,12	8,11	8,12
Suspect 2	15, 15	18, 18	19, 23.2	XY	13,15	29,30	17	11	8,9	9,10
Suspect 3	15, 16	15, 15	25, 26	XY	12,13	27,30	13,14	10,11	9,12	10,12
Suspect 4	16, 16	16, 17	19, 24	XY	14	30,30	13,16	9,11	10,11	9,10

to represent major racial and ethnic groups). Crime labs compute these estimates by determining the frequency of each allele in a sample population, and then compounding the individual frequencies by multiplying them together. If 10% (1 in 10) of Caucasian Americans are known to exhibit the 14 allele at the first locus (D3S1358) and 20% (1 in 5) are known to have the 15 allele, then the frequency of the pair of alleles would be estimated as 2 x 0.10 x 0.20 = 0.04, or 4% among Caucasian Americans. The frequencies at each locus are simply multiplied together (sometimes with a minor modification meant to take into account the possibility of under-represented ethnic groups), producing frequency estimates for the overall profile that can be staggeringly small: often on the order of 1 in a billion to 1 in a quintillion, or even less. Needless to say, such evidence can be very impressive.

When the estimated frequency of the shared profile is very low, some labs will simply state "to a scientific certainty" that the samples sharing that profile *are* from the same person. For example, the FBI laboratory will claim two samples *are* from the same person if the estimated frequency of the shared profile among unrelated individuals is below one in 260 billion. Other labs use different cut off values for making identity claims. All of the cut-off values are arbitrary: there is no scientific reason for setting the cut off at any particular level just as there is no formally recognized way of being "scientifically certain" about anything. Moreover, these identity claims can be misleading because they imply that there could be no alternative explanation for the "match," such as laboratory error, and they ignore the fact that close relatives are far more likely to have matching profiles than unrelated individuals. They can also be misleading in that the DNA tests themselves are powerless to provide any insight into the circumstances under which the sample was deposited and are generally unable to determine the type of tissue that was involved.

FIGURE 2: ELECTROPHEROGRAMS

Showing the Results of ProfilerPlus™ Analysis of Five Samples at Three Loci (D3S1358, vWa and FGA). Which suspect is a possible source of the blood? Boxes immediately below the peaks label



Looking behind the lab report: Are the laboratory's conclusions fully supported by the test results?

Many defense lawyers simply accept lab reports at face value without looking behind them to see whether the actual test results fully support the laboratory's conclusions. This can be a serious mistake.

In our experience, examination of the underlying laboratory data frequently reveals limitations or problems that would not be apparent from the laboratory report, such as inconsistencies between purportedly "matching" profiles, evidence of additional unreported contributors to evidentiary samples, errors in statistical computations and unreported problems with experimental controls that raise doubts about the validity of the results. Yet forensic DNA analysts tell us that they receive discovery requests from defense lawyers in only 10-15% of cases in which their tests incriminate a suspect.

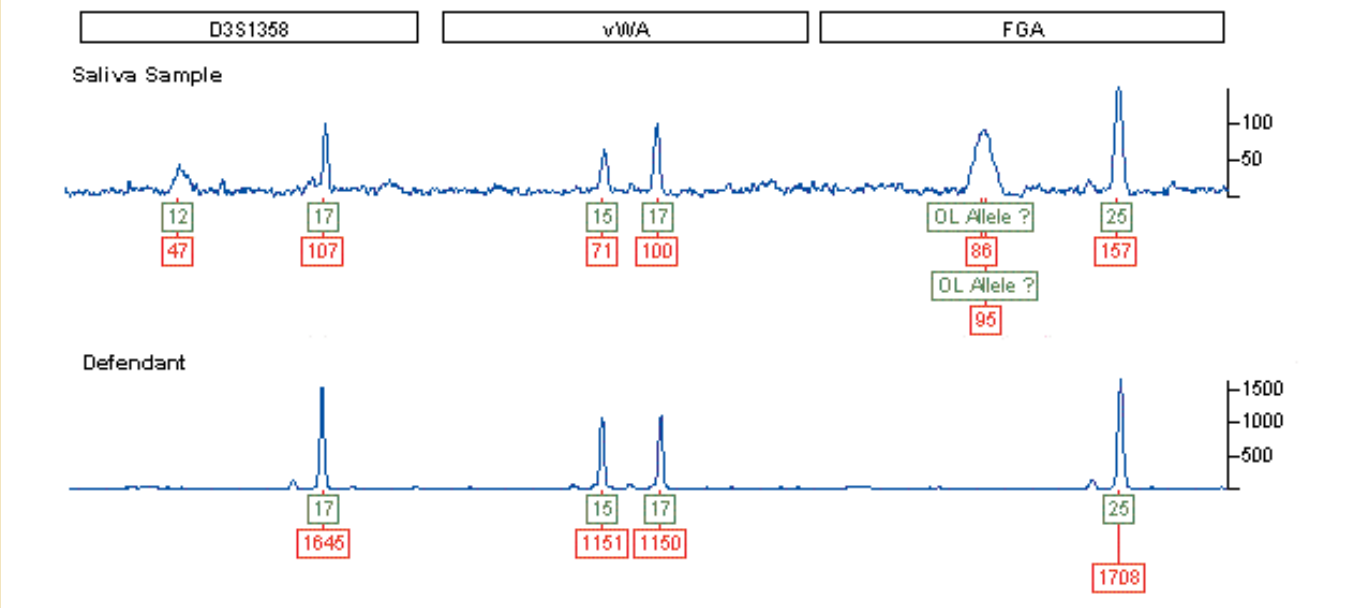
Although current DNA tests rely heavily on computer-automated equipment, the interpretation of the results often requires subjective judgment. When faced with an ambiguous situation, where the call could go either way, crime lab analysts frequently slant their interpretations in ways that support prosecution theories.[2]

Part of the problem is that forensic scientists refuse to take appropriate steps to "blind" themselves to the government's expected (or desired) outcome when interpreting test results. We often see indications, in the laboratory notes themselves, that the analysts are familiar with facts of their cases, including information that has nothing to do with genetic testing, and that they are acutely aware of which results will help or hurt the prosecution team. A DNA analyst in one case wrote:

Suspect-known crip gang member — keeps 'skating' on charges-never serves time. This robbery he gets hit in head with bar stool — left blood trail. [Detective] Miller wants to connect this

FIGURE 3: ELECTROPHEROGRAMS OF DEFENDANT AND A "SALIVA SAMPLE" FROM AN EVIDENCE SWAB

Electropherograms showing a DNA profile for the D3, vWA and FGA loci for two samples. Top sample is



guy to scene w/DNA …

In another case, where the defense lawyer had suggested that another individual besides the defendant had been involved in the crime, and might have left DNA, the DNA laboratory notes include the notation: "Death penalty case. Need to eliminate [other individual] as a possible suspect."

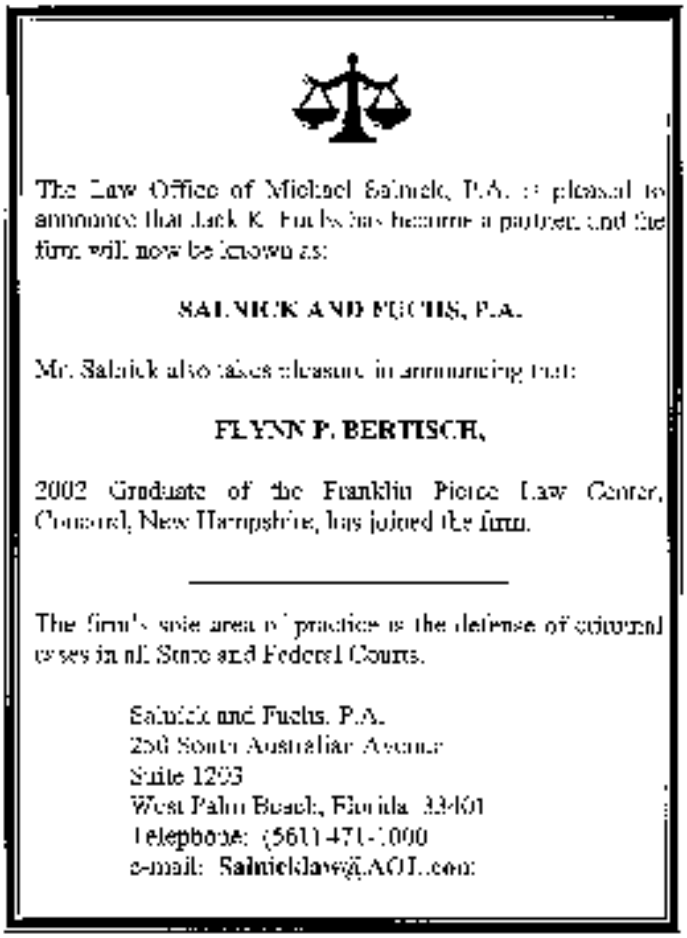
It is well known that people tend to see what they expect (and desire) to see when they evaluate ambiguous data.[3] This tendency can cause analysts to unintentionally slant their interpretations in a manner consistent with prosecution theories of the case. Furthermore, some analysts appear to rely on non-genetic evidence to help them interpret DNA test results. When one of us questioned an analyst's interpretation of a problematic case, the analyst defended her position by saying: "I know I am right — they found the victim's purse in [the defendant's] apartment." Backwards reasoning of this type (*i.e.*, "we know the defendant is guilty, so the DNA evidence must be incriminating") is another factor that can cause analysts to slant their reports in a manner that supports police theories of the case. Hence, it is vital that defense counsel look behind the laboratory report to determine whether the

HOW ELECTROPHEROGRAMS ARE PRODUCED

ProfilerPlus™ uses "primers" to identify the relevant STR-DNA segments and then "amplifies" (replicates) these segments using a process called polymerase chain reaction (PCR). Each locus is "labeled" with a colored dye (either blue, yellow or green). The Genetic Analyzer measures the length of the DNA segments by using an electrical current to impel them through a narrow capillary tube, wherein the shorter fragments move more quickly than the longer fragments. Under laser light, the colored dyes produce florescent light, signaling the presence of DNA. A computer-operated camera detects the light as the fragments reach the end of the capillary. The "peaks" on the electropherogram record these flashes of light. Based on the color of the light, and the time it took the DNA to pass through the capillary, a series of computer programs determines which alleles are present at each locus.

Figure 2 show the results for three loci that were labeled with blue dye. The position of the peaks on the graph (how far left or right) indicates how long it took the allele to pass through the capillary, which indicates the length of the underlying DNA fragment. From this, the computer program infers which allele is represented and generates the appropriate label.

The height of the peaks corresponds to the quantity of DNA present. The unit of measurement for peak heights is the RFU, or "relative fluorescent unit," which reflects the intensity of the fluorescent light detected by the computer-operated camera. Peaks representing alleles from the same person are expected to have roughly the same heights measured in RFUs throughout a given sample, although *peak height imbalances* occasionally occur.



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lab's conclusions are well supported, and whether there is more to the story than the report tells.

Behind the Table of Alleles Detected (Figure 1) is a set of computer-generated graphs called *electropherograms* that display the test results. When evaluating STR evidence, a defense lawyer should always examine the electropherograms because they sometimes reveal unreported ambiguities and, fairly frequently, evidence of additional, unknown contributors. The electropherograms shown in Figure 2 display the results for the crime scene blood and four suspects discussed above at three of the ten loci summarized in Figure 1.

The "peaks" in the electropherograms indicate the presence of human DNA. The peaks on the left side of the graphs represent alleles at locus D3S1358; those in the center represent alleles at locus vWA; and those on the right represent alleles at locus FGA. The numbers under each peak are computer-generated labels that indicate which allele each peak represents and how high the peak is relative to the baseline.

By examining the electropherograms in Figure 2, one can readily see that the computerized system detected two alleles in the blood from the crime scene at locus D3S1358. These are alleles 14 and 15, which are reported in the Table of Alleles (Figure 1). The other alleles reported in the allele chart (Figure 1) can also be seen. Our initial examination of these electropherograms reveals no obvious problems of interpretation in this case.

However, other cases are not so clearcut. Consider the electropherogram in Figure 3, which shows the DNA test results that purportedly "matched" a defendant to a saliva

FIGURE 4: PRESENCE OF MORE THAN TWO ALLELES AT A LOCUS INDICATES A MIXTURE.

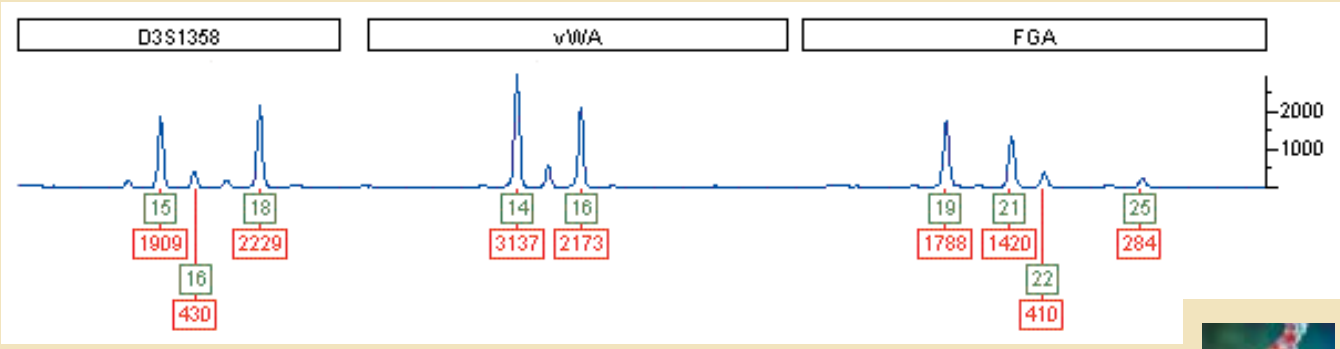
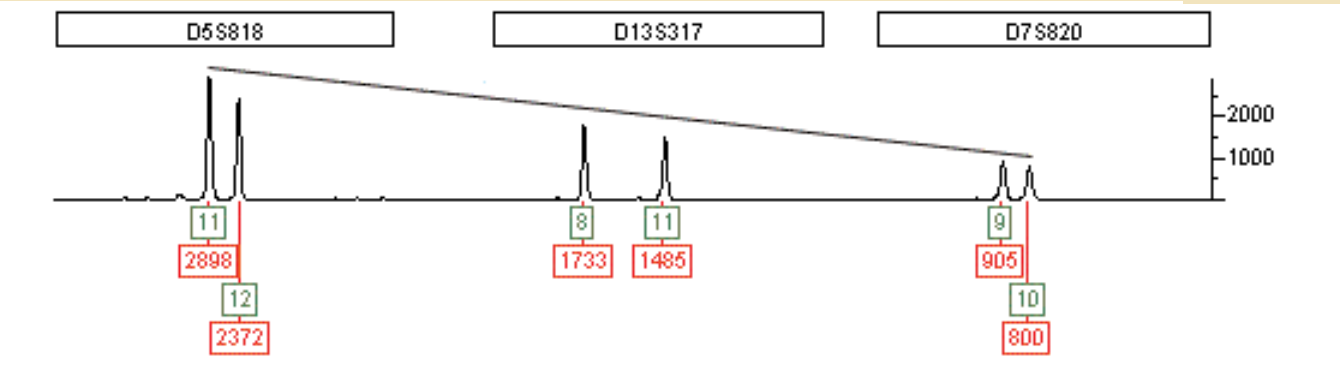


FIGURE 5: THE PROGRESSIVELY SMALLER PEAK HEIGHTS



sample taken from the breast of an alleges sexual assault victim. Although the laboratory report stated that the same alleles were found in both samples at these three loci, close examination of the electropherograms supports a significantly different conclusion. There are two additional "peaks" in the saliva sample that the laboratory failed to report — a peak labeled "12" (indicating allele 12) at locus D3S1358, and a peak labeled "OL Allele" (indicating a possible "off-ladder," or unclassified, allele) at locus FGA. The laboratory decided to ignore these two peaks and never mentioned them in its report. A defense lawyer who failed to examine the underlying test results would never have known about them. However, they clearly complicate the interpretation of the evidence — raising the possibility, for example, that the DNA on the breast swab is from a person with alleles 12 and 17 at locus D3S1358, rather than just allele 17, which would exclude the defendant as a possible contributor.

Sources of ambiguity in STR interpretation

A number of factors can introduce ambiguity into STR evidence, leaving the results open to alternative interpretations. To competently represent an individual incriminated by DNA evidence, defense counsel must uncover these ambiguities, when they exist, understand their implications, and explain them to the trier-of-fact.

Mixtures. One of the most common complications in the analysis of DNA evidence is the presence of DNA from multiple sources. A sample that contains DNA from two or more individuals is referred to as a *mixture*. A single person is expected to contribute at most two alleles for each locus. If more than two peaks are visible at any locus, there is strong reason to believe that the sample is a mixture.

By their very nature mixtures are difficult to interpret. The number of contributors is often unclear. Although the presence of three or more alleles at any locus signals the presence of more than one contributor, it often is difficult to tell whether the sample originated from two, three, or even more individuals because the various contributors may share many alleles. If alleles 14, 15 and 18 are observed at a locus, they could be from two individuals, A and B, where A contributed 15 and B contributed 14, 18. Alternatively, A could have contributed 14, 15 while B contributed 15, 18, and so on. There might also be three contributors. For example A could have contributed 14, 15, while B contributed 15, 18 and C contributed 15. Many other combinations are also consistent with the findings. A study of one database of 649 individuals found over 5 million three-way combinations of individuals that would have shown four or fewer alleles across all 12 commonly tested STR loci.[5]

Some laboratories try to determine which alleles go with which contributor based on peak heights. They assume that the taller peaks (which generally indicate larger quantities of DNA at the start of the analysis) are associated with a "primary" contributor and the shorter peaks with a "secondary" contributor. In Figure 4, for example, a laboratory analyst might conclude that allele 15 in the left locus, and alleles 10 and 13 in the right locus are associated with a primary contributor while alleles 14 and 18 in the left locus, and allele 12 in the right locus are associated with a secondary contributor. But these inferences are often problematic because a variety of factors, other than the quantity of DNA present, can affect peak height. Moreover, labs are often inconsistent in the way they make such inferences, treating peak heights as a reliable indicator of DNA quantity when doing so supports the government's case, and treating them as unreliable when it does not.

These interpretive ambiguities make it difficult, and sometimes impossible, to estimate the statistical likelihood that a randomly chosen individual will be "included" (or, could not be "excluded") as a possible contributor to a mixed sample. Defense lawyers should look carefully at the way in which laboratories compute statistical estimates in mixture cases because these estimates often are based on debatable assumptions that are unfavorable to the defendant.

Degradation. As samples age, DNA like any chemical begins to break down (or *degrade*). This process occurs slowly if the samples are carefully preserved but can occur rapidly when the samples are exposed for even a short time to unfavorable conditions, such as warmth, moisture or sunlight.

Degradation skews the relationship between peak heights and the quantity of DNA present. Generally, degradation produces a downward slope across the electropherograms in the height of peaks because degradation is more likely to interfere with the detection of longer sequences of repeated DNA (the alleles on the right side of the electro-

pherogram) than shorter sequences (alleles on the left side).

Degraded samples can be difficult to type. The process of degradation can reduce the height of some peaks, making them too low to be distinguished reliably from background "noise" in the data, or making them disappear entirely, while other peaks from the same sample can still be scored. In mixed samples, it may be impossible to determine whether the alleles of one or more contributors have become undetectable at some loci. Often analysts simply guess whether all alleles have been detected or not, which renders their conclusions speculative and leaves the results are open to a variety of alternative interpretations. Further, the two or more biological samples that make up a mixture may show different levels of degradation, perhaps due to their having been deposited at different times or due to differences in the protection offered by different cell types. Such possibilities make the interpretation of degraded mixed sample particularly prone to subjective (unscientific) interpretation.

Allelic Dropout. In some instances, an STR test will detect only one of the two alleles from a particular contributor at a particular locus. Generally this occurs when the quantity of DNA is relatively low, either because the sample is limited or because the DNA it contains is degraded, and hence the test is near its threshold of sensitivity. The potential for allelic dropout complicates the process of interpretation because analysts must decide whether a mismatch between two profiles reflects a true genetic difference or simply the failure of the test to detect all of the alleles in one of the samples.

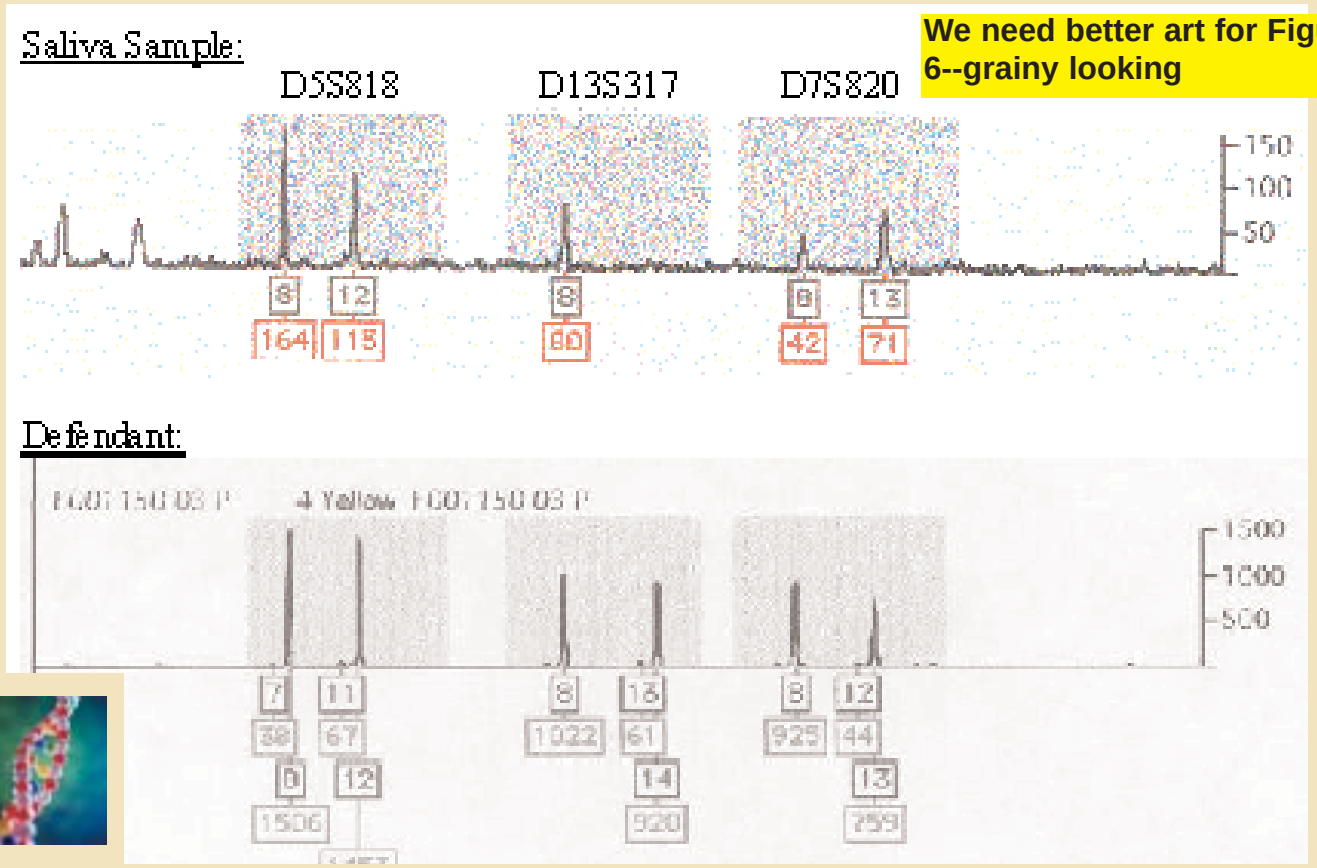
Figure 6 shows three additional loci from the case shown in Figure 3, in which a defendant's profile was "matched" to the profile of a saliva sample from a woman's breast. The laboratory reported that the DNA profile of the saliva sample shown in Figure 6 was consistent with the defendant's profile, despite the absence of the defendant's 10 allele at locus D13S317 because it assumed that the 10 allele had "dropped out." However, the occurrence of "allelic dropout" is cannot be independently verified — the only evidence that this phenomenon occurred is the "inconsistency" that it purports to explain. Obviously, there is another possible interpretation that is more favorable for this defendant — i.e., that police arrested the wrong man.

Spurious Peaks. An additional complication in STR interpretation is that electropherograms often exhibit spurious peaks that do not indicate the presence of DNA. These extra peaks are referred to as "technical artifacts" and are produced by unavoidable imperfections of the DNA analysis process. The most common artifacts are *stutter*, *noise* and *pull-up*.

Stutter peaks are small peaks that occur immediately before (and, less frequently, after) a real peak. Stutter occurs as a by-product of the process used to amplify DNA from evidence samples. In samples known to be from a single source, stutter is identifiable by its size and position. However, it is sometimes difficult to distinguish stutter bands from a secondary contributor in samples that contain (or might contain) DNA from more than one person.

"Noise" is the term used to describe small background peaks that occur along the baseline in all samples. A wide variety of factors (including air bubbles,

FIGURE 6: ALLELIC DROPOUT OF THE WRONG MAN?



We need better art for Figure 6--grainy looking

urea crystals, and sample contamination) can create small random flashes that occasionally may be large enough to be confused with an actual peak or to mask actual peaks.

Pull-up (sometimes referred to as bleed-through) represents a failure of the analysis software to discriminate between the different dye colors used during the generation of the test results. A signal from a locus labeled with blue dye, for example, might mistakenly be interpreted as a yellow or green signal, thereby creating false peaks at the yellow or green loci. Pull-up can usually be identified through careful analysis of the position of peaks across the color spectrum, but there is a danger that pull-up will go unrecognized, particularly when the result it produces is consistent with what the analyst expected or wanted to find.

Although many technical artifacts are clearly identifiable, standards for determining whether a peak is a true peak or a technical artifact are often rather subjective, leaving room for disagreement among experts. Furthermore, analysts often appear inconsistent across cases in how they apply interpretive

Not all sentences end with a period.

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FIGURE 7: ELECTROPHEROGRAM

Contains technical artifacts called stutter that may mask the presence of true alleles present in an evidence sample

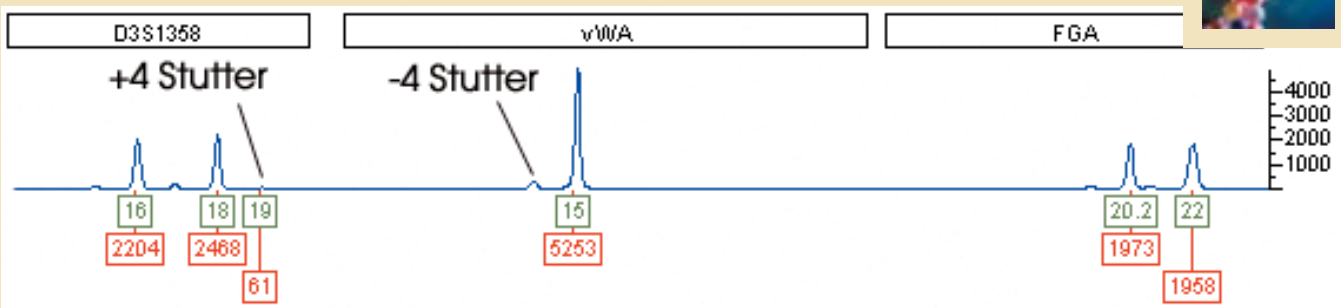
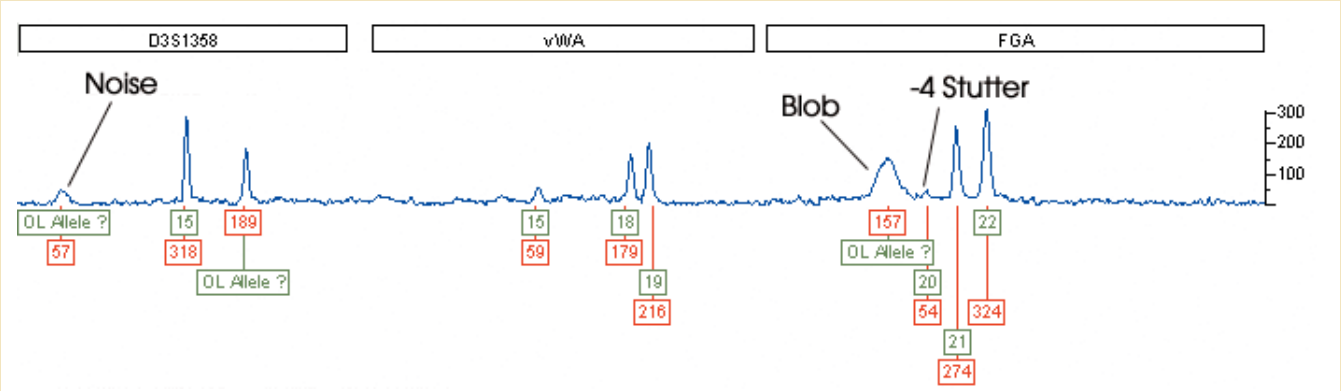


FIGURE 8: BLOBS AND OTHER FALSE PEAKS

May hide the presence of true alleles



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standards — accepting that a signal is a "true peak" more readily when it is consistent with the expected result than when it is not. Hence, these interpretations need to be examined carefully.

Spikes, blobs and other false peaks. A number of different technical phenomena can affect genetic analyzers, causing spurious results called "artifacts" to appear in the electropherograms. *Spikes* are narrow peaks usually attributed to fluctuation in voltage or the presence of minute air bubbles in the capillary. Spikes are usually seen in the same position in all four colors. *Blobs* are false peaks thought to arise when some colored dye becomes detached from the DNA and gets picked up by the detector. Blobs are usually wider than real peaks and are typically only seen in one color. The "OL Allele" shown in Figure 8 below may be a blob.

Spikes and blobs are not reproducible, which means that if the sample is run through the genetic analyzer again these artifacts should not re-appear in the same place. Hence, the correct way to confirm that a questionable peak is an artifact is to rerun the sample. However analysts, to save time, often simply rely on their "professional experience" to decide which results are spurious and which are real. This practice can be problematic because no generally accepted objective criteria have yet been established to discriminate between artifacts and real peaks (other than retesting).

Threshold Issues: Short Peaks, "Weak" Alleles. When the quantity of DNA being analyzed is very low (as indicated by low peak-heights) the genetic analyzer may fail to detect the entire profile of a contributor. Furthermore, it may be difficult to distinguish true low-level peaks from technical artifacts. Consequently, most forensic laboratories have established peak-height thresholds for "scoring" alleles. Only if the peak-height (expressed in RFU) exceeds a standard value will it be counted.

There are no generally accepted thresholds for how high peaks must be to qualify as a "true allele." Applied Biosystems, Inc., which sells the most widely used system for STR typing (the ABI Prism 310 Genetic Analyzer™ with the ProfilerPlus™ system) recommends a peak-height threshold of 150 RFU, saying that peaks below this level must be

FIGURE 9: DEFENSE EXAMINATION OF ELECTRONIC DATA

interpreted with caution. However, many crime laboratories that use the ABI system have set lower thresholds (down to 40 RFU in some instances). And crime laboratories sometimes apply their standards in an inconsistent manner from case to case or even within a single case. Hence, a defendant may be convicted in one case based on "peaks" that would not be counted in another case, or by another lab. And in some cases there may be unreported peaks, just below the threshold, that would change the interpretation of the case if considered.

Finding and evaluating low-level peaks can be difficult because labs can set their analytic software to ignore peaks below a specified level and can print out electropherograms in a manner that fails to identify low-level alleles. The best way to assess low-level alleles is to obtain copies of the electronic data files produced by the genetic analyzer and have them re-analyzed by an expert who has access to the analytic software.

Figure 9 shows electropherograms from a rape/homicide case. The defendant admitted having intercourse with the victim, but contended another man had subsequently raped and killed her. The crime lab reported finding only the defendant's profile in vaginal samples from the victim; the lab report stated that the second man was "excluded" as a possible source of the semen collected from the victim's body. However, a review of the electronic data by a defense expert revealed low-level alleles (peaks) consistent with those of the second man, which significantly helped the defense case. Notice how these low-level alleles are obscured in the upper electropherogram (which the lab initially provided in response to a discovery request) due to the use of a large scale (0-2000 RFU) on the Y-axis. These low peaks are revealed in the lower electropherogram, where the defense expert set the software with a lower threshold of detection and produced an electropherogram with a lower scale (0-150 RFU).

Notes

1. Bureau of Justice Statistics, *Survey of DNA Crime Laboratories, 2001*. National Institute of Justice, NCJ 191191, January 2002.<http://www.ojp.usdoj.gov/bjs/pub/pdf/sdnacl01.pdf>

2. *See*, William C. Thompson, *Subjective interpretation, laboratory error and the value of DNA evidence: Three case studies*, 96 GENETICA 153 (1995); William C. Thompson, Accepting Lower Standards: *The National Research Council's Second Report on Forensic DNA Evidence.* 37 JURIMETRICS 405 (1997); William C. Thompson, *Examiner Bias in Forensic RFLP Analysis,* SCIENTIFIC TESTIMONY: AN ONLINE JOURNAL, *www.scientific.org*.

3. *See* D. Michael Risinger, Michael J. Saks, William C. Thompson, & Robert Rosenthal, *The Daubert/Kumho Implications of Observer Effects in Forensic Science: Hidden Problems of Expectation and Suggestion*. 90 CAL.L.REV. 1 (2002).

4. For more background information on STR testing, *see* John M. Butler, FORENSIC DNA TYPING: BIOLOGY AND TECHNOLOGY BEHIND STR MARKERS (2001).

5. For more information about this study, contact Dan Krane.

About the Authors

William C. Thompson, J.D., Ph.D., is a professor in the Department of Criminology, Law & Society at the University of California, Irvine and a member of the California Bar. He has been a member of the NACDL DNA Strikeforce and has represented a number of defendants in cases that turned on DNA evidence.

William C. Thompson
Department of Criminology,
Law & Society
University of California
Irvine, Calif. 92697
949-824-6156
Fax: 561-679-0890
E-MAIL wcthomps@uci.edu

Simon Ford, Ph.D. was trained in molecular biology and biochemistry. He is President of Lexigen Science and Law Consultants, a firm that specializes in providing advice to lawyers about genetic evidence. He frequently consults on DNA cases and has conducted workshops for a number of agencies on computer analysis of STR test results.

Need photo of Simon

Simon Ford
Lexigen Science & Law Consultants, Inc.
2261 Market St, Box 302
San Francisco, CA 94114
(415) 865-0600
E-MAIL sford@lexigen.com

Travis Doom, Ph.D., is an assistant professor in the Department of Computer Science and Engineering at Wright State University in Dayton, Ohio. He played a key role in development of Genophiler™, the automated system for re-analysis of STR data discussed in this article.

Travis Doom
Department of Computer
Science and Engineering
Wright State University
Dayton, Ohio 45431-0001
(937) 775-5105
E-MAIL travis.doom@wright.edu

Michael Raymer, Ph.D., is an assistant professor in the Department of Computer Science and Engineering and the Program in Bioinformatics at Wright State University in Dayton, Ohio. He also played a key role in development of Genophiler™.

Michael Raymer
Department of Computer
Science and Engineering
Wright State University
Dayton, Ohio 45431-0001
(937) 775-5110
E-MAIL mraymer@cs.wright.edu

Dan Krane, Ph.D., is a professor in the Department of Biological Sciences at Wright State University. He helped develop Genophiler™ and is founder and president of Forensic Bioinformatic Services, Inc. A leading authority on forensic DNA evidence, he has testified as an expert witness in over 50 cases.

Dan Krane
Department of Biological Sciences
Wright State University
Dayton, Ohio 45435-0001
(937) 426-9270
Fax: 937-426-9271
E-MAIL Dan.Krane@wright.edu

Part 2: Evaluating Forensic DNA Evidence

Essential Elements of a Competent Defense Review

Breaking open the black box: How to review the electronic data

Reviewing the electronic files produced by the ABI Prism 310 Genetic Analyzer™ (or similar equipment) has a number of additional benefits beyond revealing unreported low-level peaks. The software that controls these devices creates a complete record of all operations the device performs while typing samples in a particular case and records the results for each sample. These records can reveal a variety of problems in testing that a forensic laboratory may fail to notice or choose not to report, such as failure of experimental controls, multiple testing of samples with inconsistent results, re-labeling of samples (which can flag potential sample mix-ups or uncertainty about which sample is which), and failure to follow proper procedures. We know of several cases in which review of electronic data has revealed that the laboratory failed to run all of the necessary control samples needed to verify the reliability of the test results, or that the laboratory ran the control samples under different conditions than the analytical samples (a major breach of good scientific practice).

BY WILLIAM C. THOMPSON, SIMON FORD, TRAVIS DOOM, MICHAEL RAYMER AND DAN E. KRANE

The electronic files are also useful for producing trial exhibits. An expert with the right software can convert the files from their proprietary format into Adobe Acrobat™ files containing images that can easily be inserted into Powerpoint™ and Microsoft Word™ documents.

It is easy for crime laboratories to produce the electronic data that underlie their conclusions. All that is necessary is to copy the files produced in the case onto a CD-ROM or other storage medium. CD-ROMs are generally preferred because they create an unalterable record of the data produced by the laboratory. Copying files to a CD-ROM is a simple point and click operation that can be accomplished in fifteen minutes or less in most cases. CD-ROM burners compatible with any laboratory computer are available commercially for under $200. There is no legitimate excuse for refusing to turn over electronic data for defense review. In a few instances laboratories have resisted producing electronic files, or have even destroyed the files, but the great majority of trial courts will not tolerate such obstructive behavior.

The electronic data produced by the ABI 310 Genetic Analyzer™ is in a proprietary format that can only be read and interpreted by ABI's Genescan™ and Genotyper™ software. Defense lawyers seeking a review of electronic data must find an expert who has access to this software. The review process typically takes a minimum of 3-4 hours, and may take much longer in an even moderately complicated case. The recent development of "expert system" software for analyzing Genescan™ and Genotyper™ data (see Sidebar) provides another option for analysis of electronic data.

Are there innocent explanations for the lab's findings?

In many cases, careful review of the underlying laboratory notes, electropherograms and electronic data will reveal no significant problems. Defense lawyers should never forget, however, that even clear-cut DNA test results may have innocent explanations.

Sample handling errors. Accidental mix-up or mislabeling of samples is a possibility that always must be considered. We have encountered a number such errors while reviewing case work.[1] In most instances the mix-ups readily come to light (and are caught by the lab) because they produce unexpected results: Samples that are supposed to be from a man show a female DNA profile, two samples known to be from the same person show different DNA profiles, and so on. The real danger arises when sample mix-ups produce plausible results. In these instances, forensic analysts may overlook subtle clues that something is amiss because they expected to find the very result produced by their error.

For example, after reviewing the laboratory notes in a Philadelphia rape case, one of the authors noticed some clues (later confirmed by additional testing) that the Philadelphia Police Crime Laboratory had mixed up the reference samples of the defendant and the rape victim. This mix up had falsely incriminated the defendant because the lab found what it thought was the defendant's DNA profile in a vaginal swab from the victim. In fact, it was the victim's own profile, and was mistakenly matched to the defendant due to the mix up.[2] Similar errors have come to light in other cases. Cellmark Diagnostics mistakenly mixed up the victim and defendant in a San Diego rape case, thereby mistakenly incriminating the defendant.[3]

The Las Vegas Crime Laboratory made the same error in a recent Las Vegas rape case.[4] This error, which came to light in April 2002, sent the wrong man to jail for over a year. In both cases the error came to light only after a defense expert noticed inconsistencies in the laboratory records.

It is not always possible to tell from the laboratory records whether samples *actually* were mixed up or cross-contaminated. However, careful review of the laboratory records will usually provide important information about whether such errors *could have happened*. For example, evidence that a reference sample from the defendant was handled or processed in close proximity to samples from the crime scene can support the theory that a sample handling error explains incriminating results. In one case, review of a criminalist's notes showed that the defendant's trousers, collected at his home, were transported to the laboratory in the same box that contained a number of items from the crime scene that were saturated with the victim's blood. This fact cast important new light on a seemingly incriminating result: blood from victim was detected on the defendant's trousers.

We suggest that defense lawyers obtain and review complete copies of all records related to evidentiary samples collected in the case (see Appendix for a model discovery request). It should be possible to document the complete history of every sample from the time it was initially collected through its ultimate disposition.

Inadvertent transfer of DNA

One of the most striking developments in forensic DNA testing in recent years is the testing of ever smaller biological samples. Whereas the original DNA tests required a fairly large amount (*i.e.* a blood stain the size of a dime) of biological material to get a result, current DNA tests are so sensitive that they can type the DNA found in samples containing only a few cells. There is likely to be enough of your DNA on the magazine you are reading right now for your DNA profile to be determined by a crime lab.

The increasing sensitivity of DNA tests has affected the nature of criminal investigations and has created a new class of DNA evidence. Analysts talk of detecting "trace DNA," such as the minute quantities of DNA transferred through skin contact. DNA typing is currently being applied, with varying degrees of success, to samples such as doorbells pressed in home invasion cases, eyeglasses found at a crime scene, handles of knives and other weapons, soda straws, and even single fingerprints.

These developments will bring more DNA evidence to court in a wider variety of cases and may well open new lines of defense. A key issue will be the potential for inadvertent transfer of small amounts of DNA from one item to another, a process that could easily incriminate an innocent person. Studies have documented the presence of typeable quantities of human DNA on doorknobs, coffee cups and other common items.[5]

Studies have also documented the inadvertent transfer of human DNA from one item to another.[6] *Primary transfer* occurs when DNA transferred from a person to an item. *Secondary transfer* is when the DNA deposited on one item is transferred to a second item. *Tertiary transfer* is when the DNA on the second item is, in turn, transferred to a third. There are published studies that document secondary transfer of DNA (in quantities that can be detected by STR tests) from items that people simply touched to other items.

A recent study commissioned by a wealthy defendant was used to show that tertiary transfer of DNA could have occurred in a manner that falsely incriminated the defendant. Dr. Dirk Greineder, a prominent physician and adjunct Harvard professor, was accused

Federal Circuit Oral Argument Panel

NACDL members with an oral argument scheduled in a federal court of appeal can hone their skills before a panel of mock judges consisting of attorneys, professionals, and law students. At least six weeks prior to oral argument, members wishing to use this service should submit a one-page "bench memo" to the coordinator(s) listed below in the circuit where the case will be heard. Coordinators will screen cases and arrange for a panel of mock judges. The panel will attempt to accommodate every request, but given the volume of federal appeals, the coordinators will have the discretion to prioritize cases, favoring those that raise significant appellate issues.

CIRCUIT COORDINATORS

Please contact the coordinator in the circuit where the case will be heard.

PANEL CO-CHAIRS

Howard M. Srebnick
Black, Srebnick & Kornspan, PA
201 S. Biscayne Blvd, Ste. 1300
Miami, FL 33131
(305) 371-6421
Fax (305) 358-2006
hsrebnick@royblack.com

Scott A. Srebnick
2400 S. Dixie Hwy.
Ste. 200
Miami, FL 33133
(305) 285-9019
Fax (305) 854-8782
srebnick@aol.com

1st Circuit
Kim Homan
Boston, MA
(617) 227-8616
homanlaw@aol.com

Rachel Brill
Hato Rey, PR
(787) 753-6131
rabrill@attglobal.net

2nd Circuit
Susan Necheles
New York, NY
(212) 997-7595
srn@hafetzlaw.com

3rd Circuit
Ted Simon
Philadelphia, PA
(215) 563-5550
tsimonesq1@aol.com

4th Circuit
Terry Reed
Alexandria, VA
(703) 299-8734

5th Circuit
Jancy Hoeffel
New Orleans, LA
(504) 865-5895
jhoffel@law.tulane.edu

William Kim Wade
Dallas, TX
(972) 233-1100
kwade@wadelaw.com

6th Circuit
John Feldmeier
Cincinnati, OH
(513) 721-4876

7th Circuit
Allan Ackerman
Chicago, IL
(312) 332-2891
doctrips@aol.com

8th Circuit
Grant Shostak
St. Louis, MO
(314) 725-3200
bshostak@aol.com

9th Circuit
Shereen Charlick
San Diego, CA
(619) 234-8469
shereen_charlick@fd.org

Martin Sabelli
San Francisco, CA
(415) 621-2400
piropo@msn.com

10th Circuit
Norman Mueller
Denver, CO
(303) 831-7364
nrmueller@hmflaw.com

11th Circuit
Ellen Podgor
Atlanta, GA
(404) 651-2087
epodgor@gsu.edu

Scott A. Srebnick
Miami, FL
(305) 285-9019
srebnick@aol.com

DC Circuit
Neil Jaffee
Washington, DC
(202) 208-7500

of killing his wife. A DNA profile similar to Greineder's was found, mixed with his wife's profile, on gloves and a knife found near the crime scene. Greineder denied touching these items, which appeared to have been used by the killer. But how did his DNA get on them?

Greineder offered a two-pronged defense.

First, he challenged the conclusion that his DNA matched that on the gloves, noting inconsistencies between his profile and the profile on the gloves. The crime laboratory had shifted its threshold for scoring alleles in a manner that allowed it to count alleles that matched with Greineder, while ignoring some that did not. And the lab had to evoke the theory of "allelic drop out" to explain why some of Greineder's alleles were not found.

Greineder's second line of defense is our focus here. He argued that his DNA could have gotten onto the glove through tertiary transfer. He and his wife had shared a towel the morning of the murder — perhaps his DNA was transferred from his face to the towel, and from the towel to his wife's face. His wife was later attacked by a glove-wearing stranger who struck her on the face, strangled her, and stabbed her, in the process transferring Greineder's DNA from his wife's face to the gloves and the knife. According to this theory, the telltale extra alleles on the gloves and knife that matched neither Greineder nor his wife were those of the killer.

To support the theory that his DNA could have been transferred innocently to the instruments of murder, Greineder commissioned a study. Forensic scientists Marc Taylor and Elizabeth Johnson, of Technical Associates (an independent laboratory in Ventura, California) simulated the sequence of events posited by the defense theory: A man wiped his face with a towel, then a woman wiped her face with the towel, then gloves and a knife like those used in the murder were rubbed against the woman's face. DNA tests on the gloves and knife revealed a mixture of DNA from the man and woman — exactly what was found in the Greineder case.[7] Taylor was allowed to present his findings to the jury. Although the jury ultimately convicted Greineder (there was other incriminating evidence besides the DNA) the case is a good example of how the amazing sensitivity of contemporary DNA profiling methods facilitate a plausible explanation for what might at first seem to be a damning DNA test result.

Ethics Hotline

Need advice on ethical questions arising in your practice?

Contact the NACDL Ethics Advisory Committee. All requests are handled in confidence, covered within the attorney-client privilege. Ethics Hotline available for emergency situations. Formal written opinions available upon request.

John Wesley Hall
Chair, Ethics Advisory Committee
Phone (501) 371-9131
Fax (501) 378-0888
forhall@aol.com

Whenever possible, requests for assistance should be in writing, setting forth all relevant facts and circumstances. In urgent situations, oral requests will be accepted.

Finding experts

The complexity of short tandem repeat (STR) testing makes it difficult if not impossible for a lawyer to evaluate the evidence without expert assistance. Defense lawyers generally need expert assistance to look behind the laboratory report and evaluate whether its conclusions are fully supported by the underlying data. Defense lawyers may also need expert assistance to develop and assess alternative theories of the evidence. Experts can also be helpful, and often are necessary, to assess whether laboratory error or inadvertent transfer of DNA might plausibly account for the incriminating results.

In our experience, the best experts for evaluating whether the lab's finding are supported by the underlying data are academic scientists in the fields of molecular biology, biochemistry, bioinformatics, molecular evolution, genetics (particularly human and population genetics), and related fields. It is not essential that the expert have had experience analyzing forensic samples. In fact, we find that forensic scientists often (but not always) make poor defense experts because they tend to accept too readily the goal-directed subjective judgments and circular reasoning of their crime lab

colleagues.

Academic scientists generally have much stronger training in scientific methods and, as a result, demand that test results be interpreted in a scientifically rigorous and unbiased manner. They often are appalled at the willingness of some forensic scientists to rely on subjective judgment and guesswork to resolve ambiguities in scientific data and their unwillingness to utilize blind procedures when making such judgments.

Having the electronic data analyzed by a company like Forensic Bioinformatics Services can make it easier to work with an expert. The FBS analysis eliminates the need for the expert to do several hours of tedious work that requires specialized software, making it possible for the expert to get to the heart of the matter more quickly. The FBS reports also highlight potential issues and problems that the attorney can use to get the interest of an expert.

Conclusions

Careful review of DNA evidence can reveal a variety of potential weaknesses, making it possible in some cases to challenge the government's conclusions and offer alternative interpretations. In order to provide effective representation to a client incriminated by DNA evidence, the defense attorney must do more than simply read the laboratory's conclusions. It is important to obtain and review the underlying scientific records, including electronic data, in order to determine whether the laboratory's conclusions are fully supported by the test results. It is also important to evaluate alternative explanations for the test results, to determine whether there are plausible innocent explanations. Promoters of DNA testing have effectively used the media to convince most people, including potential jurors, that the tests are virtually infallible. As DNA testing becomes more common in the justice system, it is vital that defense lawyers give it careful scrutiny in order to detect and expose those cases where genetic evidence deserves less weight than it is otherwise likely to receive.

Notes

1. *See*, William C. Thompson, Franco Taroni, and Colin G. Aitken, *How the probability of a false positive affects the value of DNA evidence,* J. Forensic Sci. (January 2003, in press).

2. *See Id.* for further discussion of this case. Copies of the laboratory reports may be obtained from William C. Thompson.

3. *Id.*

4. Glen Puit, *DNA Evidence: Officials admit error, dismiss case. LV lab put wrong name on sample,* Las Vegas Review-Journal, April 18, 2002.

5. *See*, van Oorschot *DNA fingerprints from fingerprints*, Nature, June 19,1997, 767; Findlay, *et al.*, *DNA fingerprinting from single cells,* Nature, October 9, 1997, 555-556; Ladd, *et al.*, *A systematic analysis of secondary DNA transfer*, 1999 J. Forensic Sci. 44(6): 1270.

6. Ladd, *et al.*, *supra* note 5.

7. An unpublished report on this study may be obtained from William C. Thompson.

About the Authors

William C. Thompson, J.D., Ph.D., is a professor in the Department of Criminology, Law & Society at the University of California, Irvine and a member of the California Bar. He has been a member of the NACDL DNA Strikeforce and has represented a number of defendants in cases that turned on DNA evidence.

William C. Thompson

Department of Criminology,
Law & Society
University of California
Irvine, Calif. 92697
949-824-6156
Fax: 561-679-0890
E-MAIL wcthomps@uci.edu

Simon Ford, Ph.D. was trained in molecular biology and biochemistry. He is President of Lexigen Science and Law Consultants, a firm that specializes in providing advice to lawyers about genetic evidence. He frequently consults on DNA cases and has conducted workshops for a number of agencies on computer analysis of STR test results.

Simon Ford

Lexigen Science & Law Consultants, Inc.
2261 Market St, Box 302
San Francisco, CA 94114
(415) 865-0600
E-MAIL sford@lexigen.com

Travis Doom, Ph.D., is an assistant professor in the Department of Computer Science and Engineering at Wright State University in Dayton, Ohio. He played a key role in development of Genophiler™, the automated system for re-analysis of STR data discussed in this article.

Travis Doom

Department of Computer
Science and Engineering
Wright State University
Dayton, Ohio 45431-0001
(937) 775-5105
E-MAIL travis.doom@wright.edu

Michael Raymer, Ph.D., is an assistant professor in the Department of Computer Science and Engineering and the Program in Bioinformatics at Wright State University in Dayton, Ohio. He also played a key role in development of Genophiler™.

Michael Raymer

Department of Computer
Science and Engineering
Wright State University
Dayton, Ohio 45431-0001
(937) 775-5110
E-MAIL mraymer@cs.wright.edu

Dan E. Krane, Ph.D., is an associate professor in the Department of Biological Sciences at Wright State University. He helped develop Genophiler™ and is founder and president of Forensic Bioinformatic Services, Inc. A leading authority on forensic DNA evidence, he has testified as an expert witness in over 50 cases.

Dan E. Krane

Department of Biological Sciences
Wright State University
Dayton, Ohio 45435-0001
(937) 426-9270
Fax: 937-426-9271
E-MAIL Dan.Krane@wright.edu

APPENDIX: MODEL DISCOVERY REQUEST FOR STR TEST RESULTS

DISCOVERY REQUEST

This is a request for disclosure of scientific materials pertaining to DNA testing performed in the case of [case name] ([County, Case Number]). This request applies to all DNA testing that has been, is currently being, or will be performed in the instant case. The request is ongoing. In the event that new materials responsive to this request are produced, discovered, or otherwise come into the possession of the prosecution or its agents, said materials should be provided to the defendant without delay.

In the event that there is a charge for reproducing any of these materials please include an itemized list indicating the number of items (for example number of pages of documents, number of photographs, X-ray films, number of CD-ROMs, etc.) and the cost of copying per item.

1. Case file: Please provide a complete copy of the case file including all records made by the laboratory in connection with this case. If the file includes photographs, please include photographic quality copies.

2. Laboratory Protocols: Please provide a copy of all standard operating protocols (SOPs) used in connection with the testing in this case. To minimize any burden of duplicating these items, we invite you to provide them in electronic form.

3. Chain of custody and current disposition of evidence: Please provide copies of all records that document the treatment and handling of biological evidence in this case, from the initial point of collection up to the current disposition. This information should include documentation which indicates where and how the materials were stored (temperature and type of container), the amount of evidence material which was consumed in testing, the amount of material which remains, and where and how the remaining evidence is stored (temperature and type of container).

4. Software: Please provide a list of all commercial software programs used in the DNA testing in this case, including name of software program, manufacturer and version used in this case.

5. Macros: If the results produced by the software are dependent on the instructions contained in macros, please provide copies of any macros used. (For analyses performed with GeneScan and Genotyper, these macros are contained in Genotyper output files in order to allow analysts to interpret the results. Simply providing a copy of the Genotyper output files in response to request 6 will satisfy this request as well).

6. Data files: Please provide copies of all data files used and created in the course of performing the testing and analyzing the data in this case. These files should include all data necessary to, (i) independently reanalyze the raw data and (ii) reconstruct the analysis performed in this case. For analyses performed with GeneScan and Genotyper, these materials should include

   (6.1) All collection files (such as injection lists and log files for an ABI 310 analysis).

   (6.2) All Genescan files, including sample files and project files.

   All Genotyper files, including templates/macros (see Request 5).

7. STR frequency tables: Please provide copies of any allelic frequency tables relied upon in making statistical estimates in this case. If the laboratory relied upon published or publicly available data, this request can be satisfied by providing a specific reference to the source.

8. Instances of Unintended DNA Transfer or Sample Contamination: Please provide copies of all records maintained by the laboratory that document instances of unintended transfer of DNA or sample contamination, such as any instances of negative controls that demonstrated the presence of DNA or the detection of unexpected extra alleles in control or reference samples, and any corrective measures taken.

9. Accreditation: Please provide copies of all licenses or other certificates of accreditation held by the DNA testing laboratory.

10. Laboratory personnel: Please provide background information about each person involved in conducting or reviewing the DNA testing performed in this case, including:

    Current resume
    Job description
    A summary of proficiency test results

Investigating and Litigating DNA Claims

Complex Litigation Seminar
(Administrative Office of the U.S. Courts, Office of Defender Services Training Branch)

Atlanta, GA September 8, 2006

Edward J. Ungvarsky, Special Counsel
(202) 824-2301, eungvarsky@pdsdc.org

Public Defender Service for the District of Columbia

Introduction/Goals

- First Reaction: What to do when you hear there is DNA evidence in your case
- Pretrial motions practice to limit admission of DNA evidence
- Pretrial preparation
- Trial: Presenting DNA evidence with jurors in mind

What to do when you hear there is DNA evidence in your case

Flight or Fight Response?

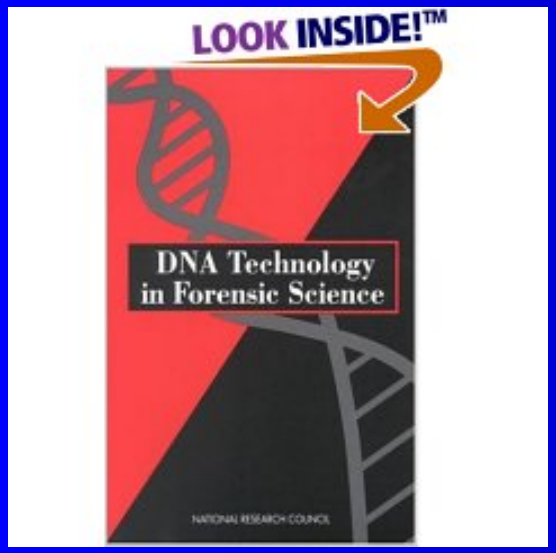
Talk About Issues With Experts and Experienced Attorneys

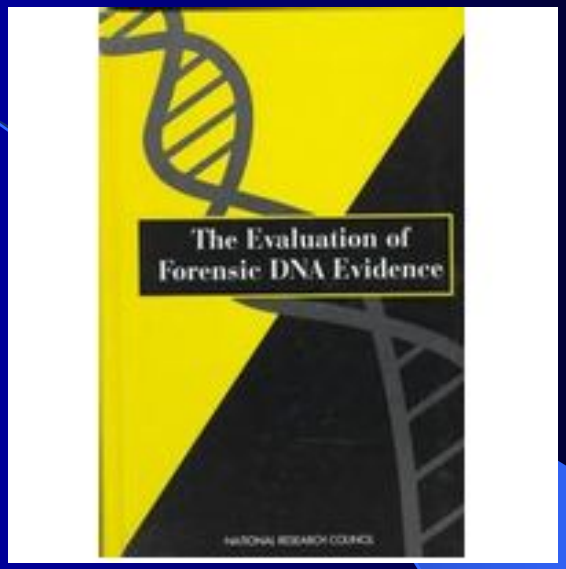
- Meetings
  - National meetings
    - Bioinformatics, Chicago PD, Local trainings
- Experts
  - Referrals
  - Develop own
- Email

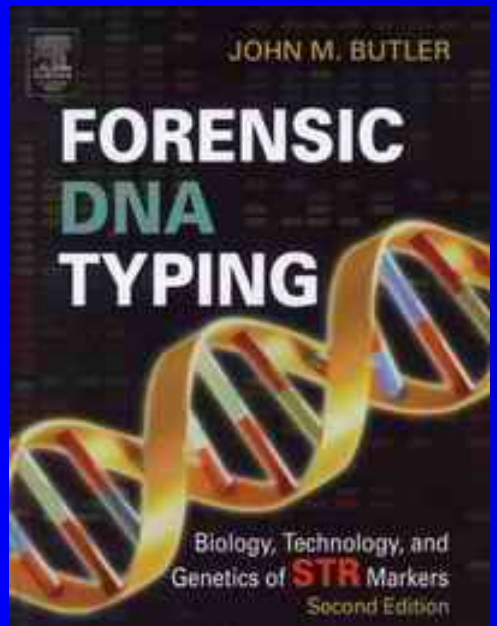
How to Find Experts

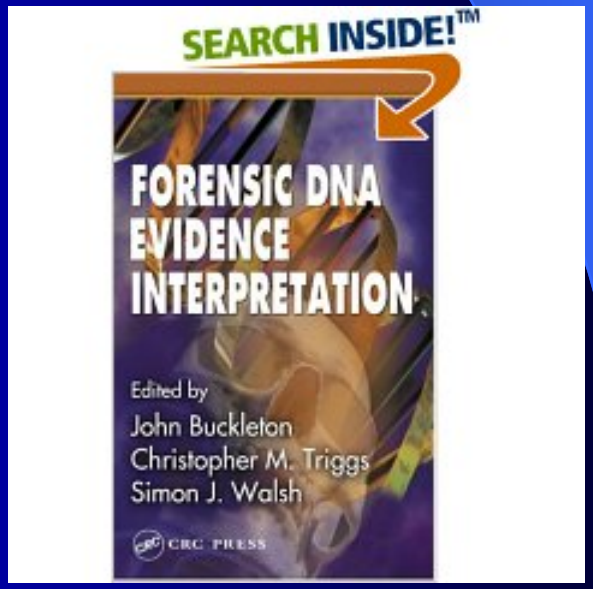
- Colleagues
- Websites
- Professional Associations
- Universities
- Expert Referrals
- Authors of Books and Articles

Own the Literature









Motions Practice: Keeping the evidence out

Areas Ripe for Admissibility Challenges and Motions in Limine

- Statistics
- MtDNA
  - *Database Limitations on the Evidentiary Value of Forensic Mitochondrial DNA Evidence*, 43 Am. Crim. L. Rev. 53 (2006)
- Y-STR
- Low Copy Number
- Cold Hit DNA Evidence
- Mixture DNA

Do You Want a Hearing?

- Make the prosecution carry its burden
- Learn about the evidence against the accused, under oath
- Exclude prejudicial evidence
- Always cite 5th, 6th, 8th, and 14th Amends.

- Hearing versus trial?
- How extensive a hearing?
- How does DNA evidence affect trial evidence?

Preparing to Attack and Present DNA Evidence Before the Jury

Jurors

- Jurors don’t understand forensic DNA profiling
- Jurors see DNA as most reliable forensic science
- Jurors CAN be persuaded that DNA is not infallible (n.b.: necessity for double negative)
- Different statistical approaches have demonstrably different effects

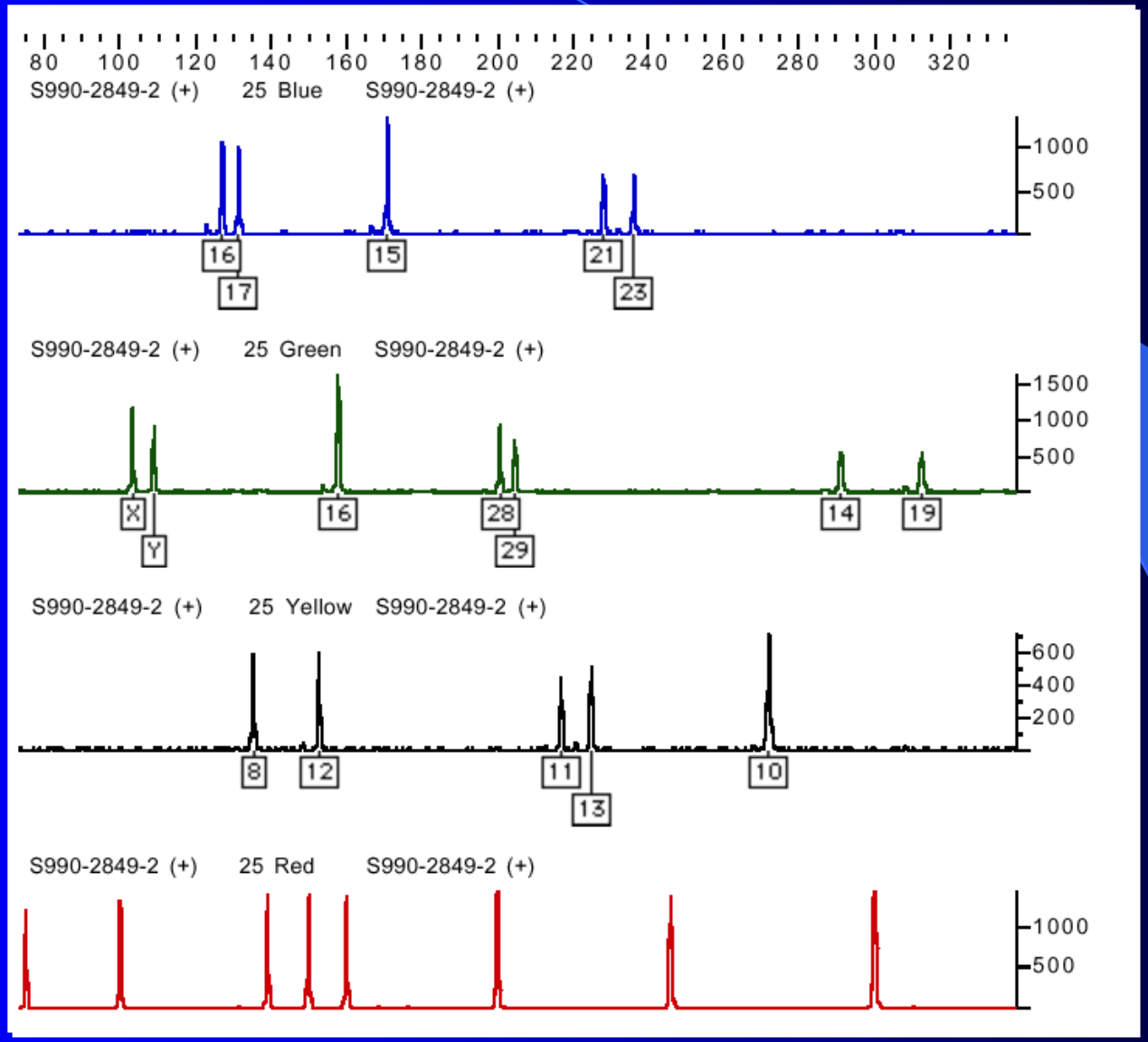
General Myths That Need Debunking

DNA is unique	Difference between uniqueness of genome and 13-location forensic profile
Gene type-calling is an objective science	Gene type-calling requires human interpretation
Scientists are neutral and laboratories are well-run	Houston, VA, FBI, Cellmark, …
Prosecution is just presenting DNA as it sees it	DNA is no different than any other type of evidence

Obtain and Review **ALL** Discovery

- Final Report
- Property/Evidence Reports
- Chain of Custody Reports
- Bench Notes
- Communications
- Printouts
- **Raw Electronic Data!**
- Protocols
- QA/QC Manuals
- Proficiency Test Results
- Error Logs
- Audit Reports
- Validation Studies
- Database Information
- Serology Report

What DNA Results Look Like



Consult with Independent Expert

- Another set of eyes
- Expertise and experience
- Not replacement for own assessment of DNA evidence

Investigate the Expert

- Curriculum vitae – Puffing/Outright lies
- http://www.corpus-delicti.com/forensic_fraud.html
- NACDL-NLADA Forensics Library
- Google
- Criminal records check
- Colleagues
- Transcripts
- Meet with the expert!!!!!!

Put Yourself in Jurors' Shoes

- How is the DNA evidence significant to the charges?
- How does it relate to the remainder of the evidence?
- What is an innocent explanation for the DNA evidence?
  - MUST have jurors think about DNA from the perspective of the defense

Evaluating DNA Evidence at Trial From Defense Perspective

- **Your client *is* the source of the DNA**
  - Was involved in the offense but was legally justified: consent, self-defense, innocent presence
  - Was not involved in the offense: DNA transfer, prior contact, malfeasance, contamination
- **Your client is *not* the source of the DNA**
  - Coincidental “match”
  - False reported “match” or inclusion (failure to properly call gene types, etc.)
  - Inflation of statistical significance of “match”

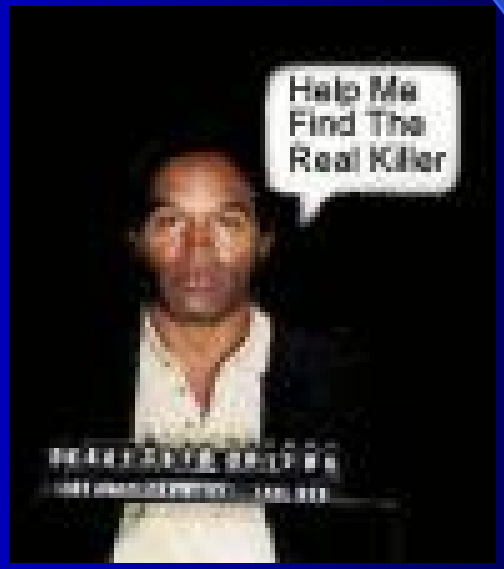
Transfer

- Transfer: through towels, laundry basket, brushing against someone on the subway
- Other reasons for client's saliva, skin cells, blood, semen, hair to be at scene (e.g., frequent visitor at decedent's home)

DNA really can fly!

Malfeasance

Deliberate contamination with client’s profile in testing process

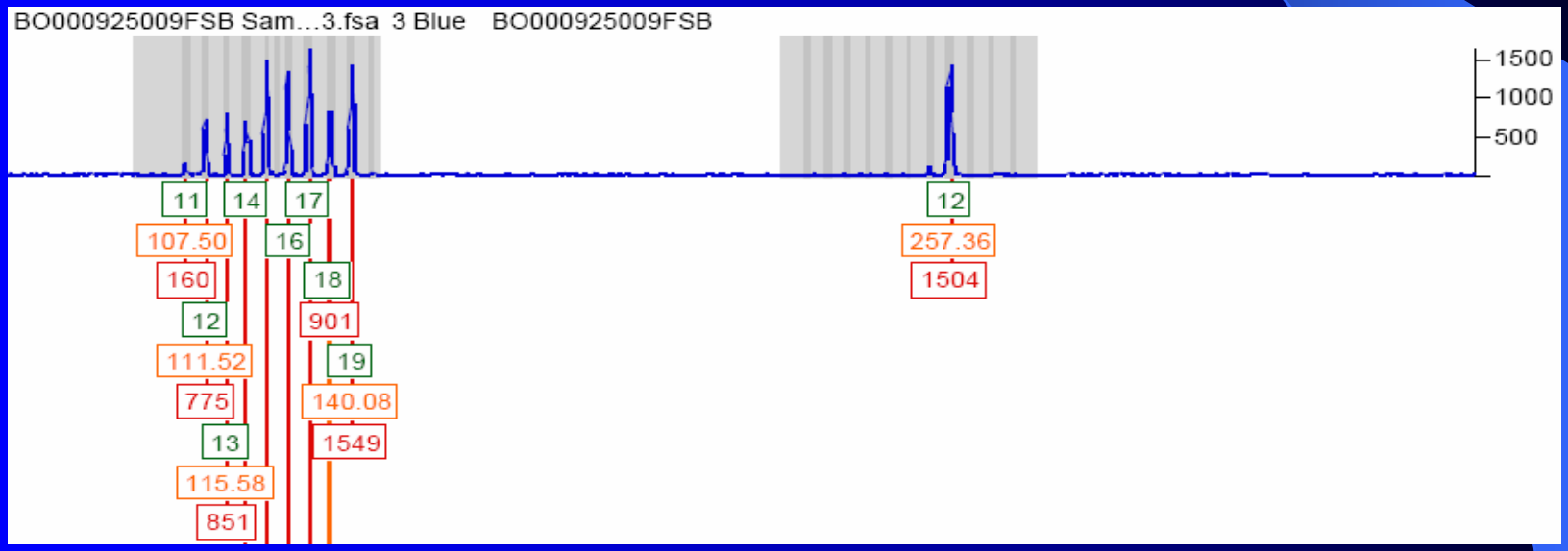


Contamination

- Can occur at collection, extraction, amplification, injection (earlier in the process, the more of a problem)
- Degraded or low copy # DNA increases risk
- Look to see if laboratory's protocols were followed
- Look at sufficiency of protocols, proficiency tests, and reviewing process
- Have a theory for why client's DNA would be near sample during collection or testing

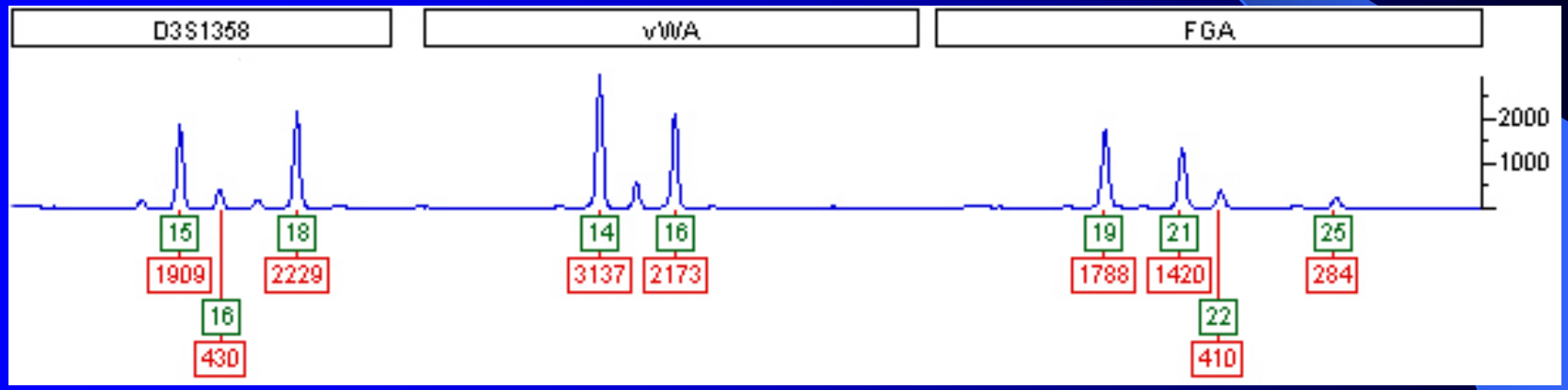
Contamination

Contaminated Positive Control



Inaccurate Calling of Data

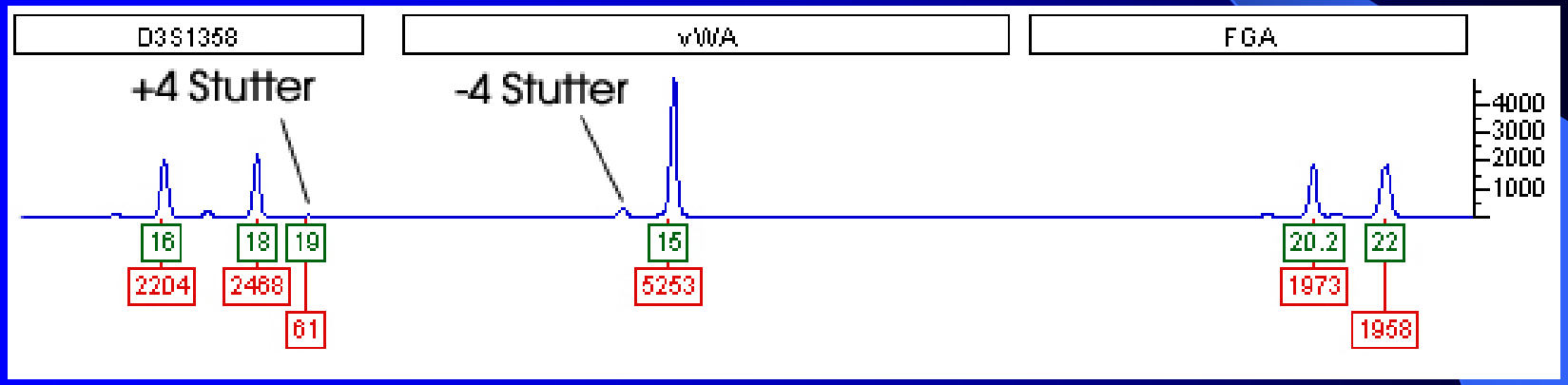
Masked contributors (misinterpretation of mixtures)



Inaccurate Calling of Data

Miscalled gene types (interpretation, not science):

stutter



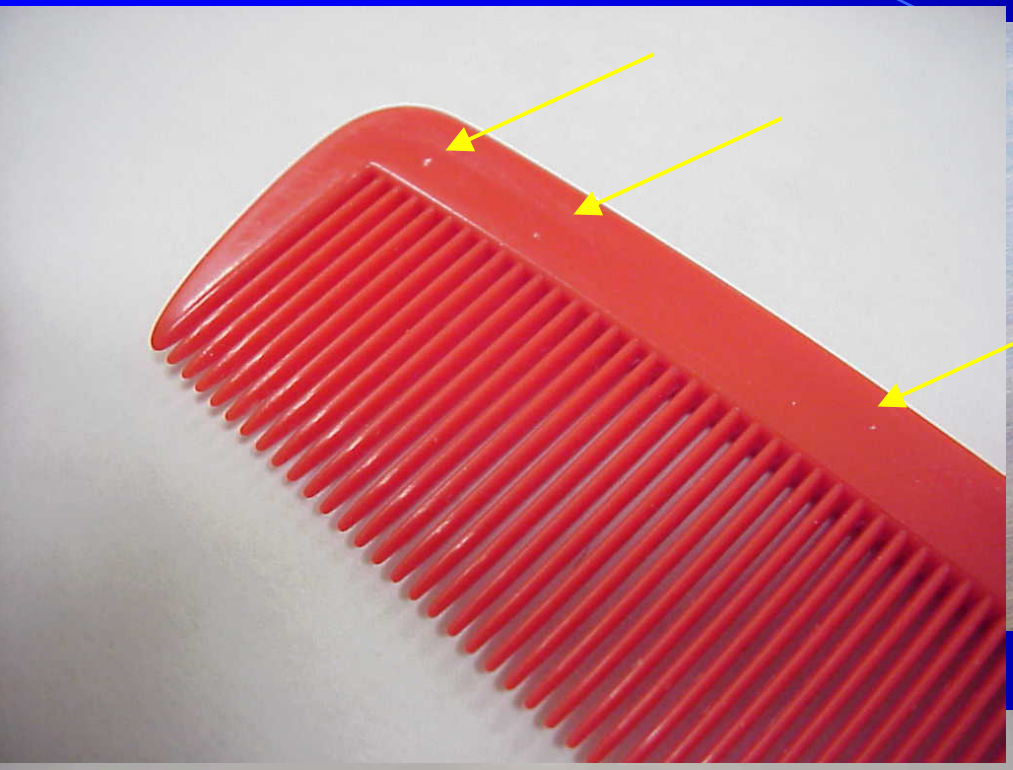
False Positives: Josiah Sutton

- Complainant ID'd Sutton as rapist; Sutton convicted and sentenced to 25 years
- Rain from holes in roof of lab may have caused contamination
- Recent retesting exonerated Sutton

Coincidental Matches/ Exaggerated Statistics

- There is a match, but it is coincidental
- Perpetrator could be a relative (still difficult)
- Perpetrator could be a non-relative (concentrate on lowering the RMP through statistical arguments)
- Statistics are far less compelling than prosecution expert suggests
- Different statistical methodologies to evidence

Possible DNA Sources



Independent Testing

- Different expert than consulting expert who reviews prosecution's work
- Results of independent testing
  - Whether to conduct independent testing is complicated judgment call
  - Face possibility that exclusion may not occur
  - Assume jury will find out you tested
    - Lab discloses
    - Prosecution witness or attorney blurts out

Presentation of the DNA Evidence at Trial with Jurors' Perspectives in Mind

Trial Tactics

- Fit expert cross-examination into theory of innocence
- Jury voir dire
- Opening statement
- Cross-examination
- Defense case
- Jury instructions
- Closing argument

Presentation of DNA: Voir Dire

- Jury Selection
  - What do jurors think about DNA evidence
  - What do jurors think about OJ Simpson case
  - Learn about your jurors
  - Educate your jurors
  - Open-ended questions
  - Questionnaires
  - Get commitment from jurors that they will not convict unless they UNDERSTAND the DNA evidence

Presentation of DNA: Opening Statement

- Put forward your understanding of the DNA
- Incorporate DNA evidence with rest of anticipated evidence
- Don’t give away secrets
- Don’t talk about what you don’t understand
- Don’t bore the jury

Cross-Examination

- "Bias is Always Relevant"
  - Don't put witness on pedestal, treat like any other witness
  - "Who is your client?"
- Determine what the LIMITED scope of C-X should be based on your defense theory and what you are planning to question
  - i.e., don't question credentials of prosecution witness who will admit to DNA transfer
- Question the SCOPE of expert's expertise
  - Forensic scientist v. statistician

Cross-Examination Practice Pointers

- Short, targeted cross-examination
  - One fact per question, building upon each other, lead
  - Use the jargon correctly – Don't Use Jargon
  - Keep it to tight (2 days = 2 hour jury deliberation, conviction)
- Turn prosecution expert into your expert
- What was not collected/tested that could/should have been collected/tested?
- Preempt fallacies
  - "Now doctor, saying that RMP is 1 in 100 does NOT mean that there is 1 in 100 chance that the suspect is not the source of the DNA, right?
  - "And that false assumption is referred to in the literature as the prosecutor's fallacy, right

Cross-Examination of Prosecution DNA Expert

Do not spar!

Don’t try to out-expert the expert; you’re an intelligent, well-informed layperson, not scientist. You will lose.

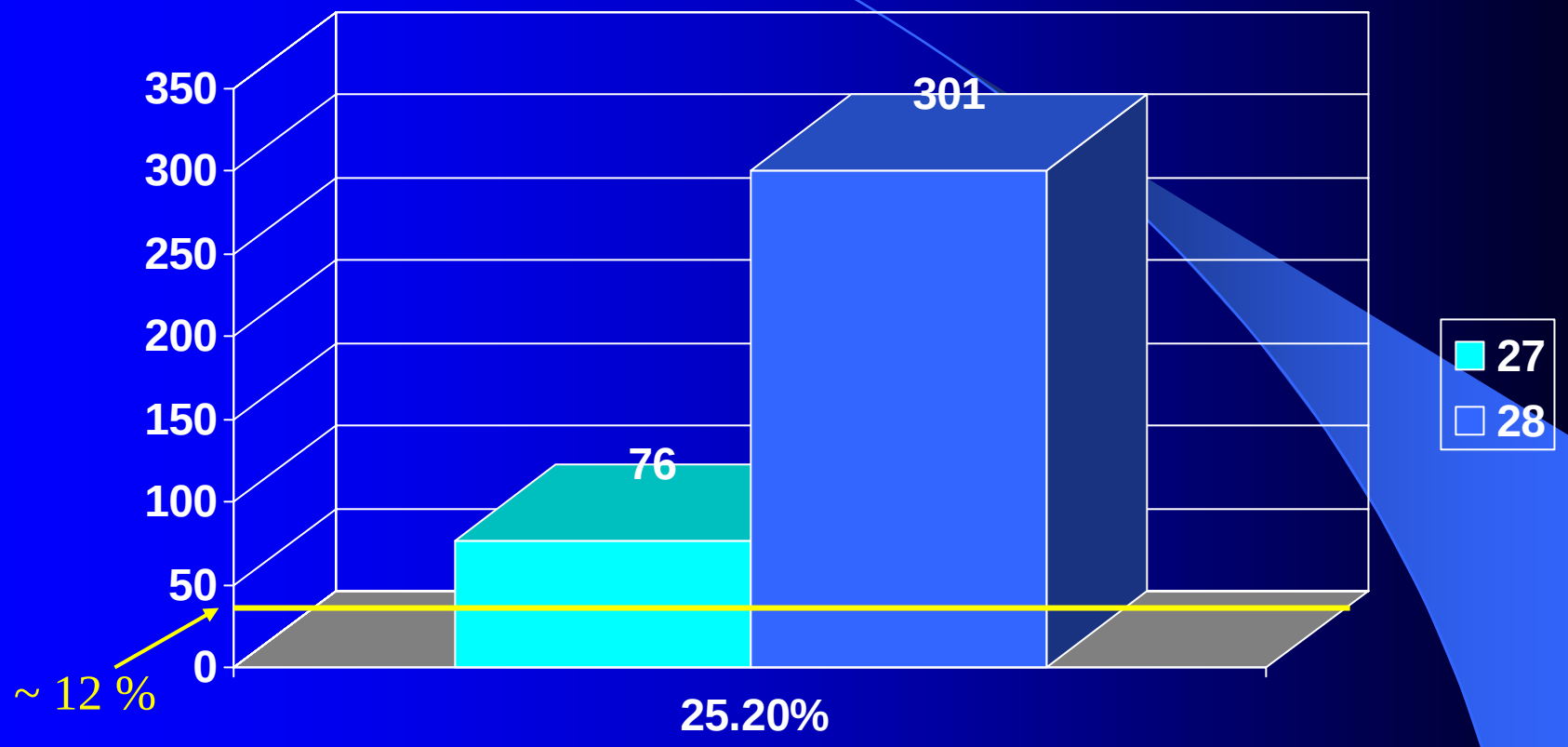
Do You Call Your Own Expert?

- Different witness than your consult
- Counter opinion/interpretation of DNA
- Prosecution bears burden of proof
- Risk – what can prosecution get from your expert that reinforces prosecution's case?
- Set Up battle of the experts
  - What will your expert say?
  - You must understand DNA and talk about testimony with expert before expert testifies

Direct Examination

- Direct Examination should be tight
- Use analogies and visuals
  - TV, not radio, generation
- Explain through questioning difference between your expert's area of expertise and prosecution's, and why that matters
- Prepare expert for cross-examination
- Prepare re-direct examination – LAST WORD!

Evidence of 3rd Party

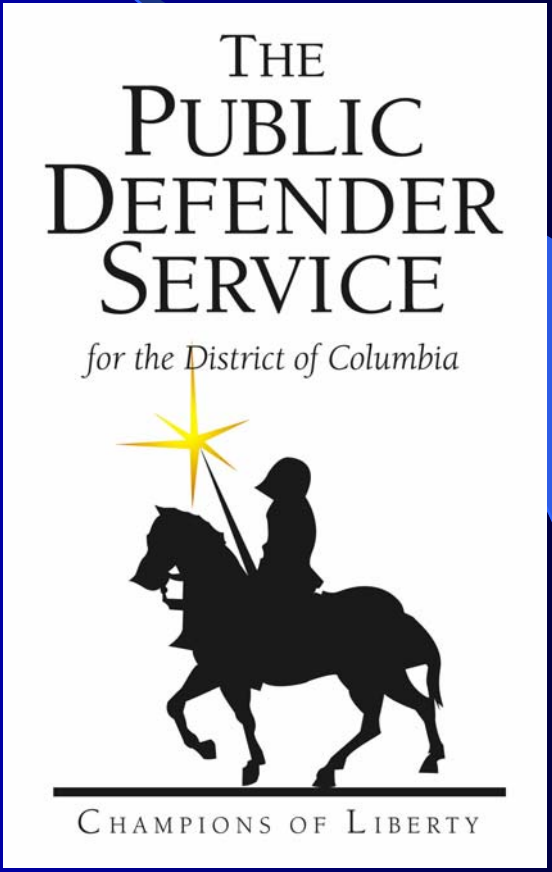


- FBI protocols: % > 12 = genetic profile
- FBI analyst: “technical artifact”

Closing Argument

- Visuals? Yes, yes, yes.
- PowerPoint? No, no, no.
  - Breaks down
  - Inflexible
  - Focus should be on the lawyer and the connection you have made with jury over course of case
- Simple – Do not get overly technical
- Demystify; bottom line, science v. art
- Empower jurors

Questions?

- **Edward J. Ungvarsky**
  (202) 824-2301
  eungvarsky@pdsdc.org

- **PowerPoint and Case materials**
  http://www.nlada.org/Defender/forensics/