THE ART IN THE SCIENCE OF DNA: A LAYPERSON’S GUIDE TO THE SUBJECTIVITY INHERENT IN FORENSIC DNA TYPING

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INTRODUCTION

Serious concerns pervade the use of forensic evidence in criminal cases. Despite efforts by a dedicated coterie of scholars who have long endeavored to expose fraudulent forensic methods, it perhaps took the DNA-exoneration cases to finally bring the breadth and depth of the problems to light. Much of the critical attention has centered upon what I have elsewhere termed “first-generation forensic techniques”—methods such as ballistics, handwriting analysis, and tool or bite mark analyses.

Although the judicial and executive branches have been slow to respond, some incremental changes have finally started to occur. Scrutiny has revealed that the methodological foundations that purport to legitimate many...
longstanding methods either do not exist or are woefully inadequate.\(^5\) While renewed efforts to scientifically validate some techniques—such as fingerprint typing\(^6\)—may prove effective, other methods have already fallen into disrepute. For instance, after a national expert panel discredited the forensic technique of bullet lead analysis, the Federal Bureau of Investigation (FBI) issued a statement that it would no longer conduct such testing.\(^7\) Similarly, courts have also recently called into question other long-accepted forensic techniques.\(^8\)

Often omitted from critiques of forensic methods, however, is a robust discussion of nuclear DNA typing. If anything, DNA typing is typically held out as the pinnacle of “good” forensic evidence, in that it exemplifies the kind of scientific rigor that first-generation techniques lack.\(^9\) After all, DNA analysis emerged from scientific processes, and it is a testable, reproducible, and falsifiable technique.\(^10\) DNA analysts even testify to their findings with expressions of statistical probabilities rather than arbitrary and unsupportable statements of certain identity.\(^11\)

Without question, this praise is well-deserved. DNA typing represents a marked advance beyond the shamanistic “sciences” of the first generation. Yet the seeming corollary—that DNA typing is therefore an exercise in purely objective, indisputable science—does not hold true. This is not to suggest that DNA has no basis in objective science, or even that it is as subjective as other forensic techniques; comparing most first-generation methods to DNA typing is like comparing astrology to neuroscience. Nevertheless, not unlike neuroscience, the fact that DNA typing is scientifically grounded does not mean that there are not plenty of things that we still do not understand about it,
and plenty of instances in which the best conclusions we can draw are nonetheless tentative ones.

Many people know this, but a surprising number of laypersons, and even lawyers, do not.\textsuperscript{12} To be clear, I am not saying that DNA typing done \textit{poorly} entails an exercise of subjective judgment. Rather, DNA typing—done perfectly and precisely according to protocol—still often entails making discretionary calls and choices. But just because DNA typing is not wholly objective does not mean that it is wholly indeterminate—it simply means that it may be more like meteorology than mathematics.

Why is it important to understand this discretionary aspect of DNA typing? Because the perception that DNA typing is an exercise in purely objective testing and observation obscures many of the difficult issues raised by this forensic method. If DNA typing is viewed as a practice that is as objective as algebra, then the only concerns it presents are those of human error. While human error is a serious problem—ranging from innocent laboratory mistakes to more pernicious or structural deficiencies\textsuperscript{13}—eliminating human error (even assuming that were possible) would not alter the fundamental fact that most forensic cases will involve some exercise of discretion. And acknowledging this fact is a critical step in any appraisal of the proper scope and application of forensic genetic testing.

The purpose of this Essay is neither to provide a comprehensive explanation of the mechanics of forensic DNA typing nor to offer a checklist for attorneys or judges seeking to test the reliability of such evidence. Other

\textsuperscript{12} William C. Thompson, \textit{Tarnish on the 'Gold Standard': Understanding Recent Problems in Forensic DNA Testing}, CHAMPION, Jan.–Feb. 2006, at 10, 15 ("DNA evidence is difficult to challenge in the courtroom because most people think it is virtually infallible. It is not just jurors, fed on a media diet of \textit{CSI}-style fantasies, who think so. Most members of the academic and legal community believe it as well. Even scholars who are critical of other areas of forensic identification science have argued that DNA is an exception—calling DNA testing 'a model for scientifically sound identification science.'").

\textsuperscript{13} See, e.g., William C. Thompson et al., \textit{Part 1: Evaluating Forensic DNA Evidence: Essential Elements of a Competent Defense Review}, CHAMPION, Apr. 2003, at 16, 18 [hereinafter Evaluating Forensic DNA Evidence (Part One)] ("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."); William C. Thompson et al., \textit{Part 2: Evaluating Forensic DNA Evidence: Essential Elements of a Competent Defense Review}, CHAMPION, May 2003, at 24, 25 [hereinafter Evaluating Forensic DNA Evidence (Part Two)] (detailing types of inadvertent errors).
sources have already achieved both of these objectives quite successfully. Indeed, this Essay will not consider the many ways in which DNA typing can go wrong—such as contamination of samples, intentionally malfeasant analysts, or statistical misrepresentations. Nor is it an essay that deals with cutting-edge issues in forensic typing, such as new methods for obtaining results from incredibly small amounts of DNA, or continuing debates about the optimal way of calculating probabilities in certain cases.

Rather, this Essay is about DNA typing done absolutely correctly. It is intended for the forensic science outsider—the person perhaps casually acquainted with forensic techniques—who may even suspect that bite, tool, or handwriting analyses are faulty, but assumes that DNA typing is science in its purest, most objective form. It provides an answer to questions like: why do DNA analysts often demand a suspect’s sample before reaching definitive conclusions about what profiles are contained in the crime scene sample, or why should genetic attribution statements that may be made with a high degree of confidence nevertheless not be confused with those made with absolute certainty?

This Essay proceeds as follows: Part I provides a superficial overview of nuclear DNA typing as the basis for the remainder of the discussion. Part II presents, in as clear and simple terms as possible, a sampling of the kinds of discretionary decisions that analysts often confront when interpreting crime scene samples. Part III wraps up with remarks about current disputes in forensic DNA typing, and how recognition of its inherent subjectivity might inform and illuminate these debates.

I. FUNDAMENTALS OF DNA TYPING

To understand the nature of the subjective decision making required by forensic DNA typing, it is first necessary to have some general notion of how it works. At the outset, it is worth spending a moment to resolve what, for many readers, may be a nagging preliminary contradiction. That is, some may wonder why criticisms of forensic DNA typing used to inculpate suspects should not likewise call into question the legitimacy of DNA typing used to

14 See Evaluating Forensic DNA Evidence (Part One), supra note 13; Evaluating Forensic DNA Evidence (Part Two), supra note 13; William A. Tobin & William C. Thompson, Evaluating and Challenging Forensic Identification Evidence, CHAMPION, July 2006, at 12 (presenting a general framework for evaluating the claims of forensic experts).
exculpate people. After all, DNA typing has grabbed headlines for its use to exonerate wrongfully convicted persons, and thus the suggestion that it might have shortcomings understandably causes alarm. However, the use of DNA typing to inculpate a person—by which I mean to say that a suspect is the likely source of a sample—fundamentally differs from its use to exculpate. The simplest analogy is to blood typing. Imagine a murder scene at which police find a blood sample certain to belong to the killer. Crime scene technicians test the blood sample and show that it is type O+. Later, the police find and draw blood from two suspects. One suspect is type AB; the other is type O-. We can, with unreserved confidence, say that the first suspect is not the killer, but regarding the second suspect, we can only say that she is included within the class of people that includes the killer. The probability that she is the actual killer turns on how many other people have that blood type, along with any other evidence that we might be able to adduce.

DNA typing works in a similar fashion. When a genetic profile is generated, it is far easier to determine with confidence those individuals from whom the sample could not have come than to identify with certainty the individual to whom the sample absolutely belongs. The difficulty is in determining the parameters of inclusion—how to define the characteristics and size of the class of persons to whom it may belong. To provide another analogy, one can imagine that a witness glimpses a six-digit license plate and detects some symbols, but not others, which can be written as follows: “??2 3?6” or “??2 3?8.” That makes a suspect out of all those with plates of “XX2 3X6” and “XX2 3X8” (using “X” to represent the variable), but it does not call into question the certainty with which we can exclude all those with plates of “XX8 4X9.”

Next, it is important to note that the general labels “DNA typing” or “forensic DNA” comprise a variety of techniques and methodologies. Most generally, forensic DNA typing currently consists largely of two different forms—nuclear DNA and mitochondrial DNA (mtDNA). As the name suggests, nuclear DNA typing looks at the strands of DNA found in the nucleus of cells—specifically those pieces of DNA found on a person’s chromosomes. However, mtDNA typing examines DNA found in the

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15 See Murphy, supra note 2, at 731 n.33 (recognizing that there are differences between exculpatory and inculpatory DNA typing).
16 JOHN M. BUTLER, FORENSIC DNA TYPING: BIOLOGY, TECHNOLOGY, AND GENETICS OF STR MARKERS 20 (2d ed. 2005). Professor Butler’s book is the canonical text on forensic DNA typing and is an excellent general reference.
mitochondria—organelles in the cell outside of the nucleus.\textsuperscript{17} The greatest advantage of mtDNA is that it is present in copious quantities and, therefore, may be more resilient to cell degradation.\textsuperscript{18} However, because mtDNA typing is both more time-intensive and produces less specific results (in terms of individuating persons), it is typically used only when nuclear DNA typing is not possible.\textsuperscript{19} For this reason, and for reasons of clarity, this Essay focuses only on nuclear DNA typing; however, it is worth noting that mtDNA also entails a large number of subjective determinations.\textsuperscript{20}

Lastly, even within the practice of nuclear DNA typing, many different individuation techniques have emerged. When DNA typing first appeared in 1986, a method known as restriction fragment length polymorphism (RFLP) was used to measure the length of certain parts of the DNA strand known as variable number tandem repeats (VNTRs).\textsuperscript{21} Ultimately, the technique that has prevailed and reflects contemporary practice is known as short tandem repeat multiplexing (STR), and thus it is that technique which this Essay discusses.\textsuperscript{22} The attractiveness of this particular method stems largely from its efficacy; it offers a high degree of discriminatory power at a rapid speed and relatively low cost.\textsuperscript{23}

A. Basics of STR Typing

How does STR typing work? I have explained that nuclear DNA typing looks at pieces of the genome found on chromosomes within the nucleus of the cell. The whole human genome is incredibly long—if unraveled it would stretch to more than six feet.\textsuperscript{24} Although examining the entire genome would be an excellent means of individuating persons, it would take a very long time and expend a great deal of resources. It would also be largely unnecessary:

\bibitem{EMORY}{Id. at 241-42.}
\bibitem{EMORY}{Id. at 242.}
\bibitem{EMORY}{See id. at 4–5 (pointing out that mtDNA has "the lowest power of discrimination and longest sample processing time").}
\bibitem{EMORY}{See, e.g., Frederika A. Kaestle et al., Database Limitations on the Evidentiary Value of Forensic Mitochondrial DNA Evidence, 43 AM. CRIM. L. REV. 53, 55 (2006).}
\bibitem{EMORY}{\textsc{Butler}, supra note 16, at 3.}
\bibitem{EMORY}{Id. at 5.}
\bibitem{EMORY}{Id. at 5, 30.}
\bibitem{EMORY}{Wellcome Trust Sanger Institute, Human Genome Project, 20 Facts About the Human Genome (Feb. 20, 2008), http://www.sanger.ac.uk/HGP/draft2000/facts.shtml.}
over 99% of the genomes of two human beings are identical.\textsuperscript{25} In fact, human DNA is roughly 98% identical to that of a chimpanzee.\textsuperscript{26}

It is on some of these regions of difference among people, then, that DNA typing focuses. Specifically, STR typing looks to particular regions of the genome where certain known sequences of the four DNA base pairs (GATC) repeat themselves, and then measures how many times those repeats occur.\textsuperscript{27} These repetitive sections are particularly useful because, presently, they have no known function. That is why they are sometimes called “junk” DNA—because unlike sections (called genes) of the DNA strand that “code” for something and have a purpose (such as determining hair color), these genes are “non-coding.”\textsuperscript{28}

In light of the standard set by the FBI when it put together the national database of genetic profiles, known as CODIS (Combined DNA Index System),\textsuperscript{29} most crime laboratories look at thirteen different places (or “loci,” the plural of “locus”) where these repeats occur.\textsuperscript{30} To make matters somewhat more complicated, at each place there are two possibilities for the number of repeats. The reason for this is that individuals have two sets of chromosomes—one from their father and one from their mother—and the number of repeats they have in a particular section might be different on each chromosome.\textsuperscript{31} In the end, a genetic profile is expressed as a list of twenty-six numbers—two numbers for each of the thirteen places.\textsuperscript{32} Those numbers represent the repeats present at the thirteen specific sites on both chromosomes.\textsuperscript{33}

For example, looking at a particular locus on the gene strand, an analyst might observe six repeats on one chromosome, and fifteen on another. The

\textsuperscript{25} Id. (noting that only 0.2% of DNA differs among humans).
\textsuperscript{26} Id.
\textsuperscript{27} BUTLER, supra note 16, at 85–88.
\textsuperscript{28} Id. at 22. But see Simon A. Cole, Is the “Junk” DNA Designation Bunk?, 102 NW. U. L. REV. COLLOQUIY 54, 56–60 (2007) (reviewing debate over use of the term “junk” to refer to noncoding DNA, and citing reports suggesting that it may turn out that the CODIS STRs have utility).
\textsuperscript{29} BUTLER, supra note 16, at 94.
\textsuperscript{30} Id. at 441.
\textsuperscript{31} See id. at 23 (“A DNA profile is the combination of genotypes obtained for multiple loci.”). If the number is the same, the person is said to be homozygotic at that locus; if the number is different, the person is heterozygotic. Id.
\textsuperscript{32} Id.
\textsuperscript{33} Id. at 26.
person would therefore be a 6, 15 at that locus. The repeats at a particular locus are called the alleles. Thus, in the example, the person would have a 6 allele and a 15 allele. All twenty-six alleles together—the two alleles from each of the thirteen loci—constitute the forensic DNA profile.

By crude analogy, you can imagine a DNA profile as a composition of the size measurements of a person. If we measure precisely, we might find a person—let’s call her Jean—has a 34-inch bust, a 28-inch waist, 38-inch hips, and a 34-inch inseam. We could then think of Jean’s profile as 34x28x38x34. By comparison, Jean’s friend Dana might have a profile of 28x28x32x26. If we had a big pile of clothing, and we knew both Jean’s and Dana’s profiles, then we could probably separate the clothes accordingly.

Similarly, DNA typing allows us to take the measurements of the individual in two respects at thirteen different places and then compare them to the measurements of another individual. Like the example given above, the range of “sizes” at each spot is not limitless—there is a finite degree of possibilities. Of course, the variation in body sizes is both less diverse and less precise than the variation in genetic material, but the general idea is the same.

B. Crime Scene Sample Processing

Where, then, does the subjectivity come in? Shouldn’t a person either possess or not possess a particular allele? Why might such information be the subject of dispute? The simplest answer would be that all of these assumptions are true: testing conducted under idyllic conditions often eliminates a significant degree of the necessary interpretive discretion. But drawing blood

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34 See id. ("A sample containing two alleles, one with 13 and the other with 18 repeat units, would be said to have a genotype of '13,18'.").
35 Id. at 23.
36 See id.
37 The most popular DNA typing kits use markers that have between six and twenty-eight allele possibilities, and seven of the thirteen loci have ten or fewer variations. Id. at 103, tbl.5.4 (listing number of observed alleles for different kits made by two major commercial manufacturers, and showing ten or fewer alleles at loci TH01, TPOX, D3, D5, D7, D13, and D16).
38 Indeed, this analogy helps to illuminate one of the longstanding debates in DNA typing—the question of linkage. Following the example above, the size of a person’s inseam may correlate to his or her shoe size. It would be unusual, although not unheard of, to have a tall inseam and tiny feet. The question of whether there is a similar kind of association or link between alleles across loci is one that has been debated in forensic DNA testing. See, e.g., Bruce S. Weir, The Rarity of DNA Profiles, 1 ANNALS APPLIED STAT. 358, 369 (2007) ("DNA profiles are genetic entities with evolutionary histories that impose dependencies among profiles.").
for the purpose of DNA testing allows an analyst to exercise a great deal of control that is lacking in the typical forensic context. In a clinical setting, the sample derives from a single source, and it can be quantified and pristinely preserved to optimize the results. And, if anything should go wrong, another sample can easily be taken and tested again.

But forensic DNA testing rarely occurs in such idyllic conditions. Crime scene DNA samples do not come from a single source obtained in immaculate conditions; they are messy assortments of multiple unknown persons, often collected in the most difficult conditions. The samples can be of poor quality due to exposure to heat, light, moisture, or other degrading elements. They can be of minimal or insufficient quantity, especially as investigators push DNA testing to its limits and seek profiles from a few cells retrieved from cigarette butts, envelopes, or soda cans. And most importantly, forensic samples often constitute a mixture of multiple persons, such that it is not clear whose profile is whose, or even how many profiles are in the sample at all. All of these factors make DNA testing in the forensic context far more subjective than simply reporting test results, as the following section elaborates more carefully.

When a crime scene sample arrives at a laboratory, an analyst must check to see if the item contains any usable biological material—most typically blood, semen, or saliva. Once detected, the DNA must be "extracted" from the item, a delicate process that creates a high risk of sample contamination or further degradation. The analyst then measures the amount of DNA present to select an optimal quantity for testing, and to ensure that the sample is human DNA.

Once the sample is ready, the first step is to amplify the genetic material so that testing it is easier. Again, to use a crude illustration, imagine you saw something that looked like a termite in your house. You want to know if it was a termite or just an ant, whether there are more termites, and if so, where they are. But it is very hard to figure that out from observing just one termite. If you saw one hundred termites, then you could not only more readily conclude that, in fact, you have a termite problem, but it would also be easier to

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40 See Evaluating Forensic DNA Evidence (Part Two), supra note 13, at 25.
41 Id. at 39–41; see also FBI LAB., SHORT TANDEM REPEAT ANALYSIS PROTOCOL §§ 4.1-4.3 (2002) [hereinafter FBI PROTOCOL] (detailing extraction procedures for blood, semen, and saliva).
43 See id. at 50 (explaining how the analyst determines whether the DNA is human).
determine the specific type of termite and where they have nested. DNA amplification, through a process known as polymerase chain reaction (PCR), takes the DNA strand, cuts it, and then creates identical copies of only the part of the DNA strand important for testing. Thus, it not only eliminates the irrelevant parts of the genetic strand, but it also amplifies, through replication, the relevant bit so it is easier to measure its characteristics.

The actual testing and measuring of the DNA sample is conducted using kits made by commercial companies and a process called electrophoresis. Detailed explanation of this process is not necessary for the purposes of this Essay, and even a brief description can be surprisingly complex. What matters for our purposes is that the results of this process are interpreted by computer software that separates out the relevant pieces of information. In this phase, DNA testing appears to the analyst not as a list of numbers, but rather as a kind of graph with peaks and valleys. Different computer software then assigns values to those peaks and valleys, based on a template of information. For a single-source sample—that is, a sample known to contain the DNA of only one person like that collected in clinical conditions from prisoners or parolees—the graphical representation is typically a fairly clean and easily interpreted chart, such as Figure 1.

44 Id. at 7, 63–79. Multiplexing PCR essentially executes this process for multiple parts of the strand at the same time. Id. at 73.
45 Id. at 63–79. Notably, the ability to amplify parts of the DNA strand particularly helps forensic DNA testing, because crime scene samples frequently are poor in both quality and quantity. Id. at 63–65. But the sensitivity of the amplification process can also raise problems, both in terms of risk of contamination (not discussed in this Essay), id. at 79, and in terms of later ambiguity in interpretive results. See id. at 68 (discussing stochastic effects).
46 There are two major types of electrophoresis: slab-gel and capillary. Id. at 321.
47 See generally id. at 313–23 (providing background on the electrophoresis process).
48 Id. at 373–74.
49 Id. at 376.
When the DNA of more than one person is involved, this entire process can become a bit more complicated. To return to the clothing analogy above, imagine that both Jean and Dana take their clothes to the same dry cleaner. Unfortunately, the cleaner is having a bad day and instead of putting the clothes in the “to clean” bag, he accidentally throws them into the garbage. Fortunately, he realizes his mistake in time and grabs the bag before it leaves the shop. Also fortunately, the cleaner knows the relative sizes of Dana and Jean, and that both of them brought in five different items: a skirt, a blouse, slacks, a jacket, and a sweater.

In trying to straighten things out, the cleaner might first sort through the pile—separating out the trash from the clothes, and then separating out skirts from pants and so on. The cleaner hopes, at this point, to have one of each—and to only have to figure out which belongs to Dana and which to Jean. Assume that the cleaner finishes separating and identifying the relevant clothes, but the cleaner still does not know to whom each article of clothing belongs. The next step he might undertake is to assess the relative sizes of each of the items in each category—distinguishing the larger from the smaller skirt, blouse, slacks, a jacket, and a sweater.

But our cleaner still is not finished. He has not yet reached his goal of ultimate attribution. He has separated the clothes from the garbage, and even
the types of clothes from one another. He has figured out which is the larger and which is the smaller of each kind of item. But recall that he knows both Jean and Dana’s relative sizes. Now he must take what he knows and make inferences about to whom each piece of clothing belongs. For instance, he should be able to surmise that the longer pants belong to Jean and the shorter ones to Dana. Or the tighter fitting blouse is likely to fit only Dana, and thus the looser one is probably Jean’s. And in most cases, he will be right.

This somewhat silly example of the bumbling cleaner mirrors the process of DNA typing. In the first stage, a machine and computer software work together to amplify and then observe the relevant characteristics of the pieces of genetic material passing through the machine. This is akin to the cleaner looking into the trash bag and distinguishing between both clothes and trash, and between types of clothes, like skirts and pants. In the second stage, the software assigns values to those characteristics based on an internal sizing standard that tells it how to quantify what it detected. This is akin to the cleaner looking at the clothing and determining that the inseam on one pair of pants is about 28” and on another it is about 34”.

The last part—where the cleaner determines that the shorter pants go with Dana and the longer pants with Jean—is done not by a machine, but by the forensic analyst. And although as a general rule such determinations can be made with confidence, they are nonetheless fraught with the potential for erroneous inferences. The cleaner, for instance, might be wrong in assuming that just because Jean is taller she prefers longer pants. Perhaps the shorter pants belong to Jean, and Dana, in turn, buys her pants long so that she can roll them up.

Still more complicated, imagine that the cleaner accidentally overlooked a third pair of pants that were in the trash bag. Suppose further that this overlooked pair in fact belonged to Jean, and so not only was the cleaner wrong in assuming that the longer pants were Jean’s, but also that there were only two people’s clothing mixed up in the pile. Or maybe he looked carefully, but somehow only one pair of pants was found in the bag—he now knows that he has lost one set of pants, but he is not sure whose they were or how they got lost.

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50 Id. at 373–77.
51 Id. at 376.
52 See id. at 378–82 (listing factors affecting genotype results).
These examples of the mixed-up cleaner help to illustrate how this act of sorting and labeling may in fact be more difficult than it seems at first blush. In much the same way, forensic DNA typing churns out an unsorted mass of information, which must be deciphered by an analyst, and which is not always amenable to only one interpretation. This leads us to the necessary subjectivity of forensic DNA typing.

II. SUBJECTIVE DETERMINATIONS IN DNA TYPING

The job of the DNA analyst, like the mixed-up cleaner, often involves taking the information processed by the computer and attributing it meaning. As in the case of the cleaner, this process relies largely on reasoning abilities, processes of elimination, subjective judgment calls, and inferences; it is not a mathematically certain, objective enterprise. If it was, we would not need DNA analysts at all because there would be no need for interpretation of DNA results. This is not to say that interpretation involves unbounded discretion—a DNA analyst works within a range of assumptions and knowledge that forms the basis of the inferences and conclusions drawn. But DNA interpretation is a discretionary act—more like stepping outside and predicting the afternoon weather than like reciting multiplication tables.

The remainder of this Part gives brief overviews of the various kinds of subjective determinations that an analyst may make in interpreting DNA typing results. In providing the examples below, I want to make clear that I have focused on ordinary, run-of-the-mill issues that arise in forensic DNA typing. I have specifically avoided discussion, for instance, of contamination—either at the crime scene or in the laboratory—even though it constitutes a significant problem.53 I have also avoided discussion of transfer—the phenomenon by which DNA may appear to be present in a place where it was not directly deposited.54 For example, my DNA may show up on a towel simply because I shook hands with a person who later grabbed that towel himself.55 And lastly, I am not discussing any ongoing methodological

53 See, e.g., Murphy, supra note 2, at 767–74 (providing examples of both questionable methodological assertions and erroneous technical applications); DNA Testing Mistakes at the State Patrol Crime Labs, SEATTLE POST-INTELLIGENCER, July 22, 2004, http://seattlepi.nwsource.com/local/183018_crimalabboxesweb22.html (cataloging errors at state DNA labs).
54 See Evaluating Forensic DNA Evidence (Part Two), supra note 13, at 25–26 (discussing innocent DNA transfer).
55 See id.
disputes or vanguard technologies, such as "low copy number" (LCN) DNA typing.56

Rather, my focus in this Part is on forensic samples collected in typical crime scene conditions, and the subjective determinations that often go into interpreting them. Figure 1 illustrated the typical clarity of a single-source forensic DNA result. But for a forensic sample collected from more chaotic conditions and with an unknown number of contributors, the results can be a bit more difficult to interpret. Consider, for instance, the image reproduced below as Figure 2.

Figure 2

![Image of a DNA profile diagram](image)

At each locus—identified by the boxed-off information at the top of different sections and typically abbreviated to the first few letters or numbers (for instance, locus "vWA" on the top row)—there are multiple identifiable peaks representing the alleles. For instance, locus vWA shows at least four identifiable peaks with clearly observable alleles—16, 17, 18, and 19. The amount of the genetic material (the quantity, not the length) is reflected by the numbers off to the right. At locus vWA, there is a large amount of material for the 16, 18, and 19 alleles, and much less for the 17 allele. The differences in

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quantity can be significant for typing purposes; a large amount of material may reflect that one person has two of the same allele at that locus or that multiple persons share the same allele. On the contrary, very little material may indicate any number of things—for instance, it could mean nothing; it could mean that the peak is “spurious” (not a real indication of genetic material); or it could mean simply that fewer persons in the mixture possess that allele than others. That is why, for instance, there is an unlabeled peak to the left of the 16 allele.

This is the essential difficulty in interpreting forensic DNA samples—determining what to count and what not to count, while also recognizing that the logical inference is not necessarily the only or correct inference. When analysts are given the known suspect’s profile—as opposed to being asked what profiles are possible, given the results they have generated—the risk of erroneous attribution becomes heightened. An analyst may unwittingly fall prey to confirmation bias—seeing in the results what she expects to see, rather than what may or may not be there.\footnote{See, e.g., Evaluating Forensic DNA Evidence (Part One), supra note 13, at 18 ("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.").} The paragraphs below give additional examples of the ways in which even the most conscientious forensic analyst may make the kind of subjective calls that risk an erroneous interpretation of DNA test results.

A. Deciding Which Peaks Should Count: Peak-Height Thresholds

As explained in the previous section, the first thing an analyst must do is determine what information matters. The peaks and valleys of a forensic DNA sample are not always indisputably clear, but instead require interpretation that labels some as legitimate and others as spurious. The height of the peak (literally, how tall it looks) reflects the amount of genetic material measured at that allele. The amount of measured material can be a function of many different things, some of which are covered in the following sections. Regardless, analysts typically aim for a peak height within an optimal range to ensure that what is observed is genuine genetic material. For instance, the FBI Protocol allows only peaks over 200 relative fluorescence units (RFU) to “be considered conclusive for match purposes,” though it recommends interpretation of any peak over 50 RFU, including for exculpatory purposes.\footnote{FBI PROTOCOL, supra note 41, §§ 10.1, 10.2.3.}
The makers of commercial equipment for DNA testing recommend a minimum of 150 RFU.\textsuperscript{59} Other laboratories allow thresholds as low as 50 RFU for inculpation.\textsuperscript{60}

The importance of peak heights in interpreting DNA results is several-fold. If the threshold is set too low, then many small peaks that do not represent true alleles, but appear for other reasons,\textsuperscript{61} will be treated as representing genetic material. Without minimum thresholds, an analyst might think that a peak belongs to the suspect, when in fact it is not from the suspect. Conversely, a very large quantity of genetic material raises risks of a spillover effect, wherein additional peaks that are relics of the excess of material, rather than a reflection of a true allele, are created.\textsuperscript{62} A very large peak might also (correctly or erroneously) suggest that one person has two copies of the same allele at that locus (recall that each locus has genetic material from the father and mother), or that multiple persons share that allele.\textsuperscript{63} Almost always, a forensic profile does not contain only perfectly formed peaks that reflect true genetic alleles; rather, before making any conclusions, an analyst must first determine which peaks should factor into consideration at all.

**B. Choosing How to Interpret Peaks of Different Sizes: Peak-Height Imbalance**

When interpreting which peaks are significant, the analyst is typically guided by some rules of thumb. For instance, a genetic profile should have relatively balanced material across the loci. A pristine, single-source sample collected in clinical conditions will often reflect relatively consistent peak heights, with the primary exception being loci where the individual has two copies of the same allele at that locus (known as homozygosity).\textsuperscript{64} In those places, the peak height should be roughly twice the size of the other peaks because twice as much genetic material was observed for that allele.\textsuperscript{65}

\textsuperscript{59} Evaluating Forensic DNA Evidence (Part One), supra note 13, at 24–25.
\textsuperscript{60} See, e.g., Jason R. Gilder et al., Run-Specific Limits of Detection and Quantitation for STR-Based DNA Testing, 52 J. FORENSIC SCI. 97, 97 (2007).
\textsuperscript{61} See infra Part III.C.
\textsuperscript{62} See infra Part III.C.
\textsuperscript{63} BUTLER, supra note 16, at 155–57.
\textsuperscript{64} Id. at 23.
\textsuperscript{65} Typically, in a single-source sample only two alleles should appear per locus, but reproducible tri-allelic patterns have been observed. Id. at 132, 383.
But perfect balance can be difficult to achieve with crime scene samples. Such samples, as previously noted, often arrive at the crime laboratory in imperfect condition, both in terms of quantity and quality. For a variety of reasons, some of which are explored below, peak heights can vary dramatically. As a result, an analyst often must interpret that variation and decide what, if any, significance it carries.

For instance, suppose the analyst knows that the defendant is homozygotic at a locus. The analyst might then interpret a very large peak as evidence of that homozygosity, and dismiss any additional peaks at that locus as spurious. Even if the peak is not extremely high, the analyst might still claim that it represents a “true” allele that matches the defendant, while at the same time dismissing another peak for various reasons. Such determinations would not necessarily be wrong, but they also would not necessarily be right. The key is that these decisions involve the exercise of (hopefully reasoned) discretion, and no analyst can state without reservation which peaks will always, and which will never, count as “true.”

C. Ignoring Some Peaks Altogether: Artifacts and Stochastic Effects

What are some of the reasons, then, that an analyst might ignore or dismiss a peak—even a peak that is taller (and thus suggests a significant quantity of material) than other peaks in the same profile? There are numerous responses, but I will only discuss a few. Notably, at times an analyst may not have any explanation for the presence of a peak, but often the presence of a particular peak may be explained by one of several recognizable phenomena. These phenomena are typically lumped under the label “artifacts”—because they create spurious peaks rather than true alleles, or “stochastic effects”—meaning that they occur as a result of a random variable. Such effects have very expressive names like stutter, pull-up or bleed through, spikes, and blobs.

\[66\] Id. at 145–46 (noting that crime scene samples are often degraded or limited in quantity, although commenting that PCR can generate typeable results from less than one nanogram of material); see also Jeanette M. Wallin et al., TWGDAM Validation of the AmpFISTR Blue PCR Amplification Kit for Forensic Casework Analysis, 43 J. FORENSIC Sci. 854, 863–64 (1998) (reporting typeable results at or above 0.25 nanograms).

\[67\] See infra Part III.C.

\[68\] See, e.g., BUTLER, supra note 16, at 68.

\[69\] “Stutter” refers to a phenomenon well-described by its label. Frequently, a peak will appear in a position just short or just long of the true peak’s position, due to difficulties in the amplification process. Id. at 123–24, 382.
Some artifacts are "reproducible," meaning that repeat testing should generate identical artifacts, while others are not. The occurrence of artifacts, however, requires human interpretation of DNA testing results in order to separate true peaks from spurious peaks. Again, to be clear, a good laboratory will have standards and protocols for undertaking this effort, but even such standards are apt to be mere guidelines—not precise, unalterable rules. Ultimately, the decision to dismiss a peak as an artifact, or label it a true allele, is a discretionary decision left to the analyst.

D. Assigning Meaning to Missing Peaks: Allelic Dropout

At times, the difficulty in interpreting DNA evidence is not the presence of extraneous peaks, but the absence of an expected peak or allele. One way to interpret such an absence would be to identify it as an exclusion; that is, if the suspect has a particular profile that matches the crime scene sample in all but one place, it might simply be assumed that the suspect is then excluded as the source. Commonly, however, analysts will explain the absence of an anticipated or expected allele as a function of "allelic dropout." Dropout often occurs when the DNA is not properly amplified so that it can be detected. This commonly results from degraded or low quantity DNA samples, where some material fails to amplify. A similar result can also occur as a result of genetic mutations that cause an allele not to amplify properly, creating what is commonly referred to as a null allele.

The fact that an allele may simply fail to appear—for any number of reasons—obviously causes problems in interpreting DNA typing results. A

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70 Pull-up occurs as a result of problems with the instruments’ reading of the dye colors that distinguish various bits of DNA. As Butler explains, "If the matrix color deconvolution does not work properly than the baseline can be uneven or a phenomenon known as 'pull-up' can occur. Pull-up is the result of a color bleeding from one spectral channel into another, usually because of off-scale peaks." Id. at 336–37.

71 Spikes (or voltage spikes) are typically steep, tall peaks (as the name suggests) that are not reproducible and are a product of physical conditions in the instrument, such as air bubbles. Id. at 383.

72 As Butler explains, "Dye blobs . . . occur when fluorescent dyes come off of their respective primers and migrate independently through the capillary." Id.

73 Id. at 382.

74 Id. (“A laboratory needs to establish criteria to identify a true allele because a DNA typing analyst must decide which peaks contribute to a donor(s) profile(s) and which are due to an artifact.”).

75 Id. at 133.

76 Id. at 133–34.

77 Id. at 145–46.

78 Id. at 134.

79 Id.
missing allele could mean any number of things; for example, it could represent grounds for excluding a suspect as a possible source because the suspect, unlike the sample, has a typeable allele at that locus. Or it could simply generate ambiguity in the possible interpretations of the results because it is not known with certainty why the allele did not appear. What matters is that the analyst reading the crime scene profile has the opportunity to ascribe significance to the missing material. It is like evidence that the perpetrator lives at 55 Jones Street; if a suspect in fact lives at 55 Jones Court, but matches the evidence in every other way, we might not be too worried. But if that address is the only evidence, we might not feel comfortable concluding it was just a trivial mistake.

E. Labeling Peaks as Belonging to Particular Profiles: Mixture Deconvolution

In the area of mixture deconvolution, forensic DNA analysts have another major opportunity for interpretive discretion. This is simply a fancy way of saying that crime scene samples often contain the DNA of multiple persons, and analysts frequently attempt to reach conclusions about the number and characteristics of the various contributors. Recall Figure 2, which contained the DNA typing results of a fictive crime scene stain. The interpreted data does not arrive labeled with the number of contributors or their particular profiles; rather, the technology simply analyzes all the DNA present and identifies the results indiscriminately. It is up to the analyst to try to separate out and ascribe relevant profiles, much like our bumbling cleaner had to do with the mixed up clothing.

Because deciphering mixtures constitutes such an important part of forensic DNA typing, many efforts are underway to improve the available technology. One recent, significant advance is the capacity to separate male DNA from female DNA, and then focus on amplifying only the male fragment. This technique, known as Y-STR typing, is particularly helpful in rape cases, in which samples are commonly a mixture of DNA (frequently, a male

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80 See Murphy, supra note 2, at 753–54 & n.148 (discussing the difficulty in handling mixtures and how such issues lead to interpretation by the analyst).
82 For instance, the National Institute of Justice recently requested proposals for projects concerning mixture deconvolution, see U.S. DEP'T OF JUSTICE, SOLICITATION: FORENSIC DNA RESEARCH AND DEVELOPMENT (2006), available at http://www.ncjrs.gov/pdffiles1/nij/s000803.pdf, and efforts are already underway to develop software and algorithms that might automatically decipher mixtures according to their component ratios. BUTLER, supra note 16, at 164.
perpetrator and a female victim). However, because this form of typing only examines half of the quantity of material as a full DNA profile (i.e., just the male fraction), it is less discriminating and therefore less determinate than a full profile.

As with most aspects of DNA interpretation, the discretion exercised by analysts is not limitless. There are principles and guidelines that instruct analysts on how to separate out a mixture, assign the number of contributors, and perhaps even tentatively identify major or minor contributor profiles. The FBI Protocol, for instance, permits an analyst to label a major and minor contributor in a mixed specimen sample "if there is a distinct contrast in peak intensities between the alleles, and the alleles contributing the largest peak height values satisfy the conditions of a single source specimen."

The problem with mixtures is that they exacerbate all of the above issues regarding the interpretation of stutter, pull-ups, spikes, blobs, or dropout. Not only may it be more difficult to distinguish true from false peaks, but even in the cleanest of profiles, contributors can readily mask one another's contributions. Studies have shown that roughly 3% of three-person mixtures are easily misidentified as two-person mixtures, and that over 70% of four-person mixtures would be wrongly labeled two- or three-person mixtures. And even where the number of contributors is known, there may be an extraordinarily large number of ways of interpreting the evident genetic material to create possible individual profiles.

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83 BUTLER, supra note 16, at 201-04.
84 Id. at 213-14.
85 Id. at 158-60.
86 FBI PROTOCOL, supra note 41, § 10.4.2.1.
87 BUTLER, supra note 16, at 157. It becomes similarly difficult to separate out contributor profiles where the quantity of material from one source compared to the quantity of material from a second source appears in a sharply skewed ratio. Id. at 155 (noting that the "minor component of a mixture is usually not detectable for mixture ratios below the 5% level or 1:20").
88 David R. Paoletti et al., Empirical Analysis of the STR Profiles Resulting from Conceptual Mixtures, 50 J. FORENSIC SCI. 1361, 1361 (2005) ("The relatively small number of alleles detectable at most CODIS loci and the fact that some alleles are likely to be shared between individuals within a population can make the maximum number of different alleles observed at any tested loci an unreliable indicator of the maximum number of contributors to a mixed DNA sample.").
89 For example, using the genotypes of 959 individuals, it is possible to construct roughly 146,536,159 possible different three-person mixtures. Id. at 1363.
III. POLICY IMPLICATIONS

Although the primary purpose of this Essay is to highlight the subjectivity inherent in even the most favorable conditions of forensic DNA typing, it seems only fitting to conclude with some brief meditations on how acknowledging the existence of this discretion might affect policymaking in the area. Forensic DNA testing has developed rapidly and expansively in the past twenty years, and all evidence suggests that it will continue its rapid growth. This is a good thing: DNA typing offers a powerful means of identifying suspects and bolstering the evidence in a large number of otherwise difficult cases. At the same time, difficult questions about the proper use of forensic DNA presently confront our society. Legal and ethical controversies swarm around the proper scope for collecting DNA samples, searching DNA databases, and investigating and prosecuting cases based on the results of DNA typing.

With regard to the collection of DNA samples, legislatures and courts continue to confront concerns about the limits on collecting DNA from various classes of persons, ranging from convicted felons and misdemeanants to simple arrestees. A recent bill signed into law by President Bush, authorizing a committee to set guidelines regarding the preservation of blood samples taken from newborns, has spurred concerns about the quiet creation of a national DNA database. Complaints have also arisen with regard to law enforcement’s use of informal means of collecting DNA—whether through DNA dragnets in which large swaths of a community are asked to volunteer a sample, surreptitious collection by police of discarded or abandoned items from a suspect, or even outright trickery.

With regard to searches of DNA databases, there are two primary debates about the permissible breadth of such searches. The first concerns what are commonly called cold hits—these arise when a database is trawled for a match to a crime scene sample. Cold hits increasingly generate convictions in cases that are many years old and in which little or no additional evidence is adduced. Moreover, there is not yet a consensus as to how to calculate match

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91 See Murphy, supra note 2, at 736 n.63 (citing examples).
probabilities in such cases. Nevertheless, no court has ruled that a conviction cannot stand on a cold hit alone.

Secondly, and more recently, localities have pushed to use “low stringency” or “familial” search techniques in DNA databases. Such queries seek not the exact profile of a suspect, but rather a similar profile that is likely to correspond to a relative. Such familial searches purport to help narrow the class of suspects in cases in which the actual perpetrator is not in the database, but relatives or family members are. Apart from the scientific legitimacy of this approach, the legal repercussions of such searches have yet to be fully explored, including whether a familial match should suffice for probable cause, or even reasonable suspicion, to engage in additional intrusions against either the family member or the suspect himself or herself.

It is beyond the scope of this Essay to outline all the arguments in favor of or against certain approaches to DNA collection, databasing, and investigation, much less to offer any specific policy prescriptions. Rather, the modest point made here is that a clear-eyed understanding of the inherent subjectivity and discretion involved in forensic DNA typing is essential to any debate. All too often, the conversation about DNA methods seems to presume an almost mathematical rigor, as though a crime scene DNA test result was as indisputable as the product of two times four.

In contrast, an appreciation of the subjectivity required in forensic typing might enhance the quality of deliberation on all of these topics. It might also enable us to see greater parallels between scientific evidence and other, more traditional forms of evidence, like information from eyewitnesses or informants. For example, we have an intuition in the criminal justice system about the quanta of evidence required before the police may undertake certain actions; searching a home typically requires a warrant and probable cause, and investigatory detentions typically demand at least reasonable, articulable suspicion. In giving content to those terms, we in turn have a notion that

93 See Murphy, supra note 2, at 741 n.87 (noting, however, an unreported case from the United Kingdom in which the court reversed the conviction on the basis of an insufficiently strong probability calculation).
certain types of information are more reliable than others—for instance, a known informant’s report of criminal activity is more reliable than that of an anonymous tipster.95

The same kinds of questions arise in the context of genetic typing, but in a far less familiar guise. How many loci are necessary to constitute enough probative evidence to justify a search warrant, an arrest, or a conviction? What confidence level must we have in the accuracy of the claimed genetic profile? When are the profiles extracted from a mixture or found in a familial search ones in which we can rely on with reasonable confidence, versus conclusions that give us no more than a hunch or a line along which to pursue further investigation? All too often, those discussing legal regulations simply assume that the results of DNA testing will be indisputably certain and complete, and overlook the possibility that there may be shades of gray or areas of dispute. In short, DNA typing is not an exercise in pure, objective observation. Some DNA typing results are more probative than others.

Similarly, the specter of erroneous attribution, for reasons beyond issues of human error, should loom largely in the minds of policymakers as they approve expansive forms of DNA databasing and searching. In allowing investigations to proceed on the basis of partial matches—such as trawls and familial searches in DNA databases—we must acknowledge the very real concern that we expose certain individuals to the risk of arrest or intrusive investigation on the basis of coincidental, and only coincidental, matches. This is particularly true given that many efforts to expand collection and investigation policies uniquely affect politically disadvantaged racial, ethnic, or socioeconomic groups. In a criminal justice system indisputably fractured upon such demographic lines, the determination to permit familial searches, for instance, disproportionately exposes particular segments of society to intrusion or harassment. When predominantly white, affluent legislators cite scientific certainty as the basis for promoting and adopting intrusive policies shouldered predominantly by minority, low-income individuals, there is cause for reflection and concern. Either our confidence in such forensic techniques should be so great that we should not bristle at universal application, or we should recognize that the fallibility and subjectivity of such methods spark doubts that should dampen the fever for limitless expansion.

95 See Florida v. J.L., 529 U.S. 266 (2000) (holding that an anonymous tip was insufficient grounds for frisk).
CONCLUSION

DNA typing is a powerful, useful, and revolutionary forensic technique. Many of the benefits of its development have already been realized in the closing of otherwise impossible-to-solve cases and in the exoneration of wrongly convicted individuals. Yet despite its formidable power, it is not a perfect and purely objective science. Good inculpatory DNA methods nonetheless entail significant exercises of discretion on the part of forensic analysts, and whenever there is discretion there is always the heightened risk of error or mistake. As we enter this new era of scientific proof, it behooves us not only to remember that we must be vigilant in revisiting our settled expectations of forensic proof so as to ensure their continued viability, but also that we must display the proper degree of humility and restraint in expressing our confidence in these powerful, but still evolving, methods.