Evaluating Forensic DNA Evidence: Essential Elements of a Competent Defense Review

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Introduction

"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.

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% 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.

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
Blood Stain	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

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 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.

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 guy to scene w/DNA …

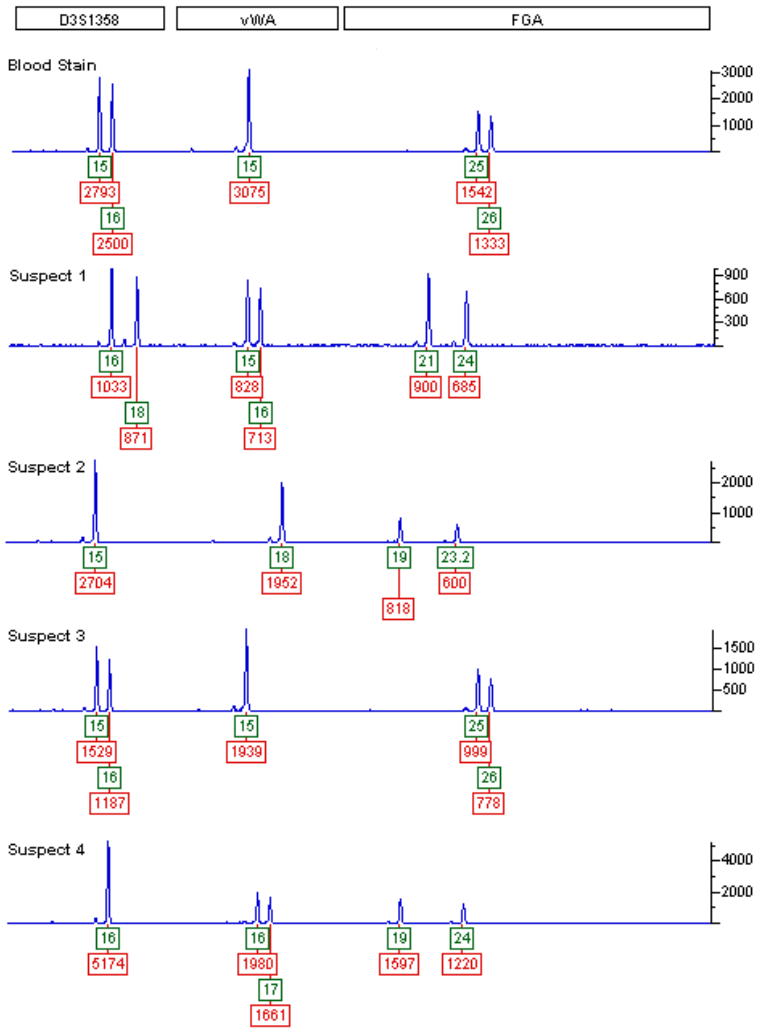
In another case, where the defense lawyer had suggested that another individual besides the defendant had been involved in the crime, and might have left DNA, the DNA laboratory notes include the notation: "Death penalty case. Need to eliminate [other individual] as a possible suspect."

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 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.

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 the name of the alleles seen while boxes below indicate their heights in RFUs



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.

[Note: Sidebar #1 should appear about here. Sidebar #1 includes more details about how STR testing is done, but is separated from the main text of the article to avoid bogging down readers who aren't interested in so much detail. The sidebar is in bold type]

Sidebar #1: How Electropherograms Are Produced[4]

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.

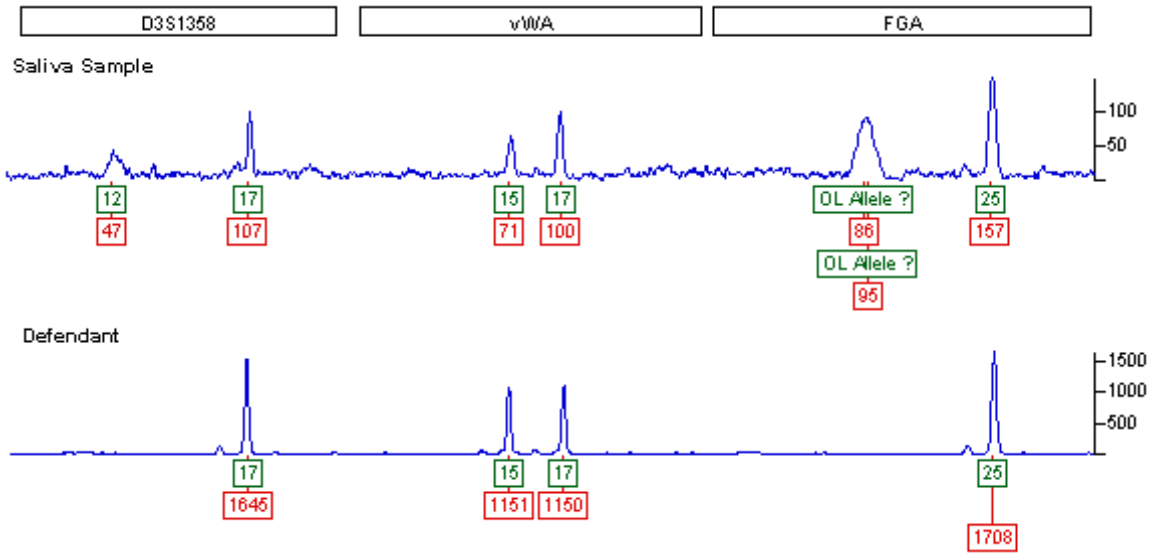
Figure 2 show the results for three loci that were labeled with blue dye. The position of the peaks on the graph (how far left or right) indicates how long it took the allele to pass through the capillary, which indicates the length of the underlying DNA fragment. From this, the computer program infers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

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 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.

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 from a swab of a woman's breast that the defendant is said to have licked. Bottom sample is the defendant's profile. Boxes below the peaks label the name of the alleles seen while boxes below indicate their heights in RFUs.



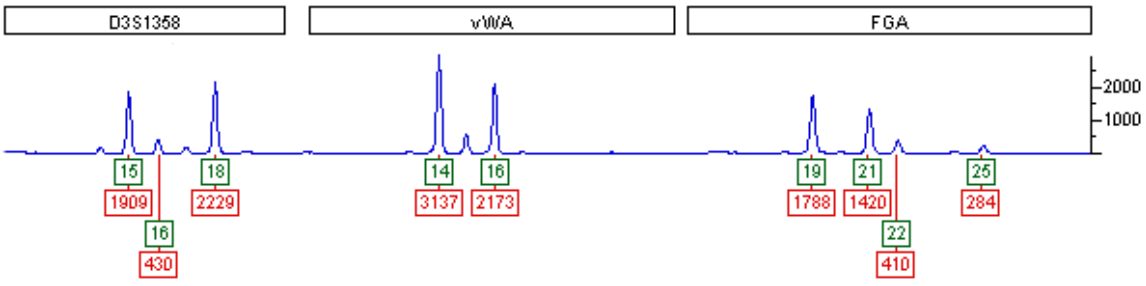
Sources of Ambiguity in STR Interpretation

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]

Figure 4: Presence of more than two alleles at a locus indicates a mixture.



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

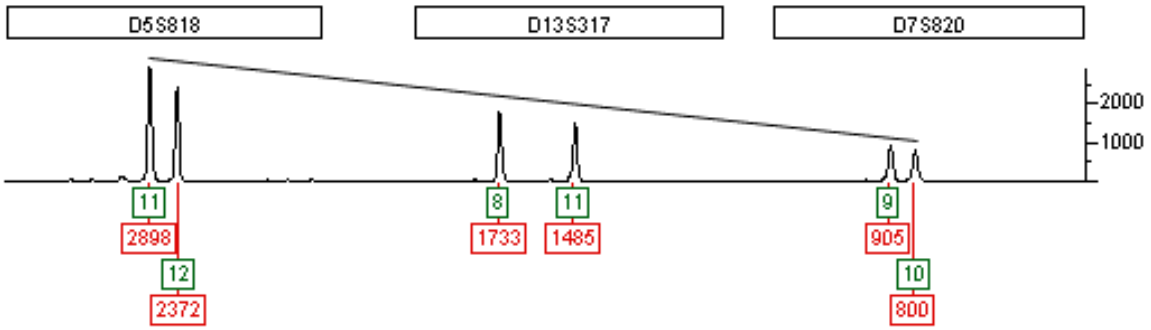
make such inferences, treating peak heights as a reliable indicator of DNA quantity when doing so supports the government's case, and treating them as unreliable when it does not.

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

Figure 5: The progressively smaller peak heights in this sample from left to right are indicative of degradation.



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 possi-

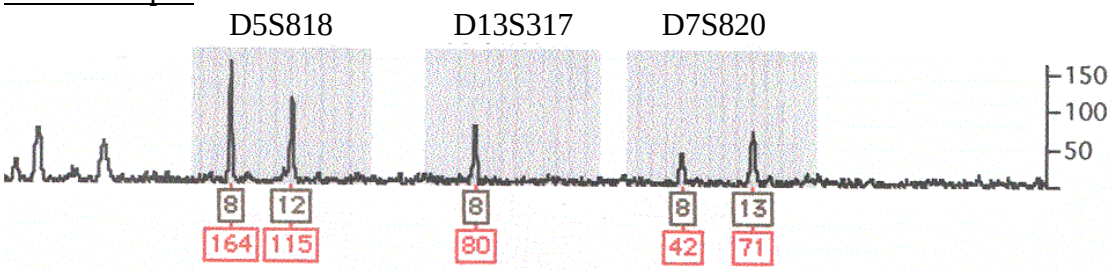
bilities make the interpretation of degraded mixed sample particularly prone to subjective (unscientific) interpretation.

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.

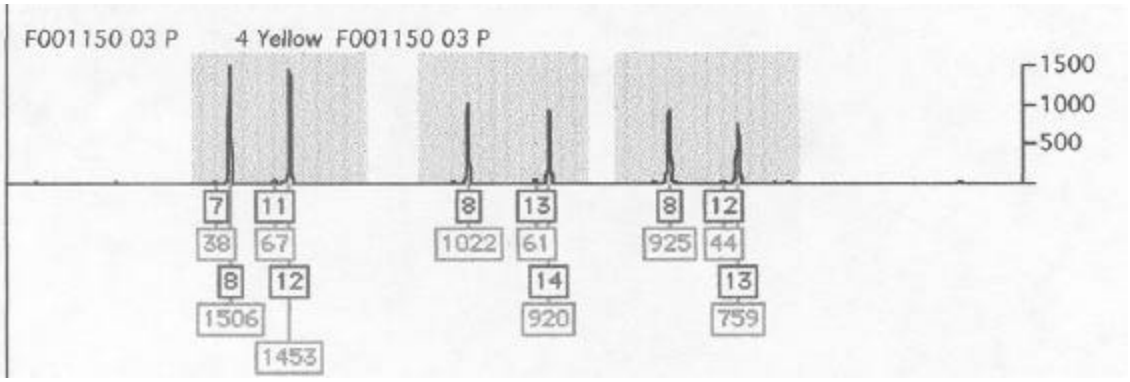
Figure 6 shows three additional loci from the case shown in Figure 3, in which a defendant's profile was "matched" to the profile of a saliva sample from a woman's breast. The laboratory reported that the DNA profile of the saliva sample shown in Figure 6 was consistent with the defendant's profile, 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.

Figure 6: Allelic Dropout or the Wrong Man?

Saliva Sample:

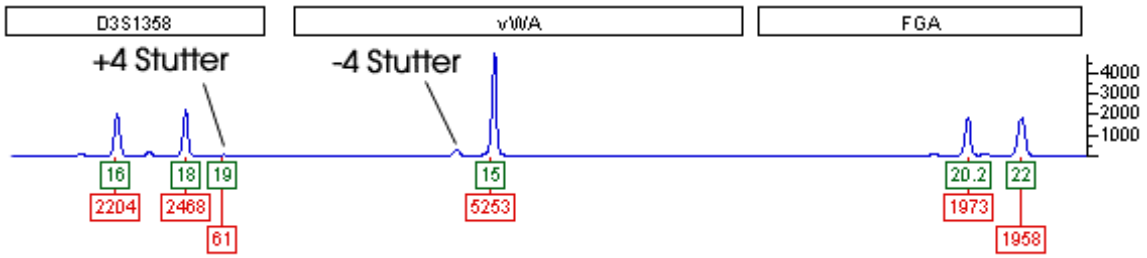


Defendant:



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*.

Figure 7: This electropherogram contains technical artifacts called stutter that may mask the presence of true alleles present in an evidence sample.



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, 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.

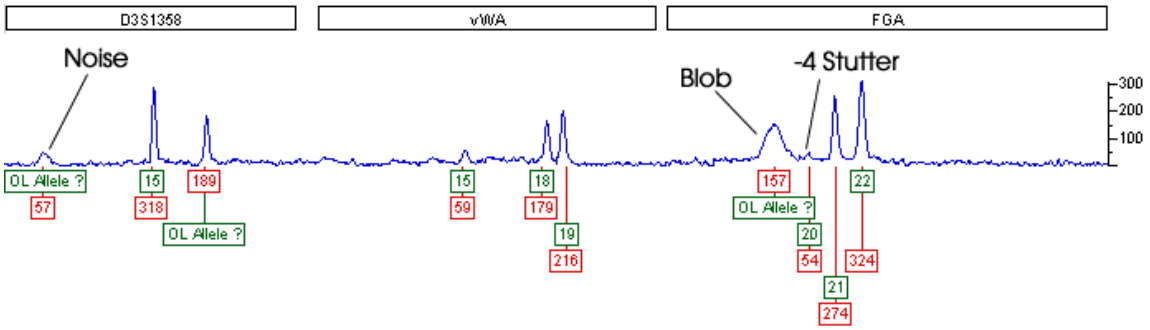
Pull-up (sometimes referred to as bleed-through) represents a failure of the analysis software to discriminate between the different dye colors used during the generation of the test results. A signal from a locus labeled with blue dye, for example, might mistakenly be interpreted as a yellow or green 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 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).

Figure 8: Blobs and other false peaks may hide the presence of true alleles.



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 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.

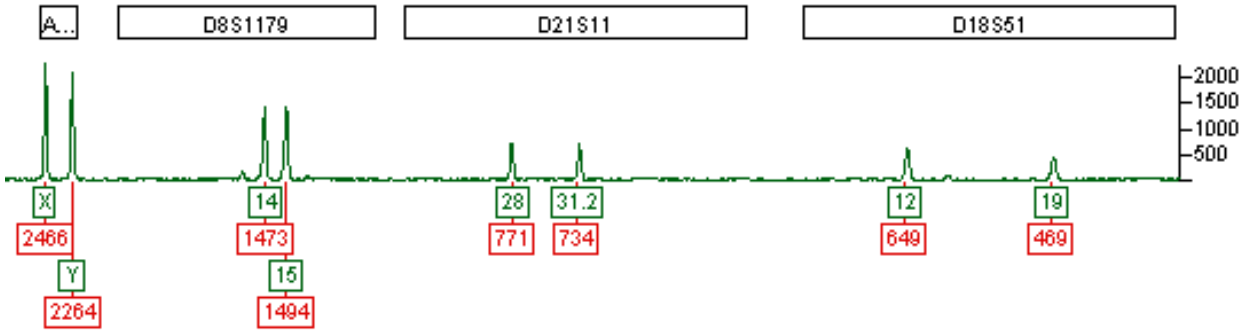
Finding and evaluating low-level peaks can be difficult because labs can set their analytic software to ignore peaks below a specified level and can print out electropherograms in a manner that fails to identify low-level alleles. The best way to assess low-level alleles is to obtain copies of the electronic 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).

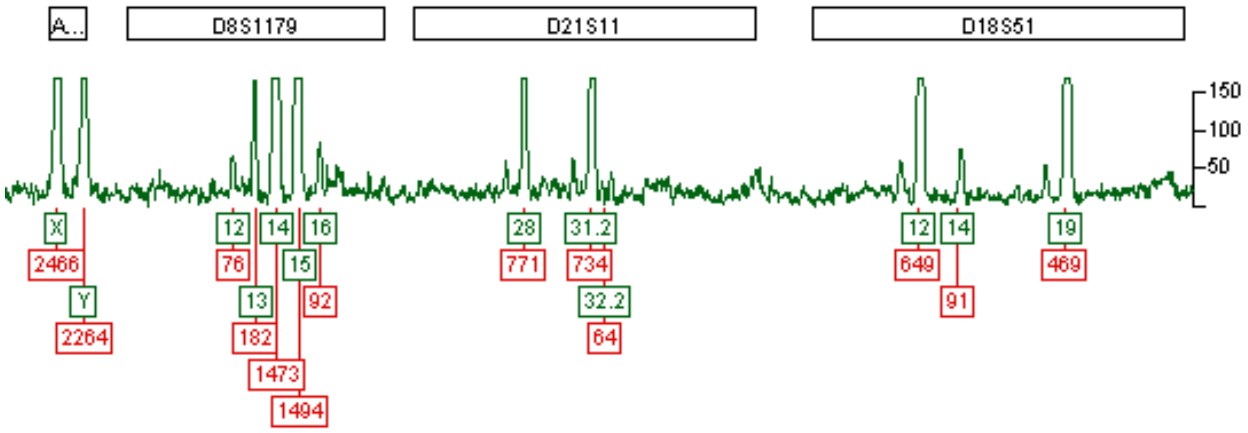
Figure 9: Defense Examination of Electronic Data Reveals Second Contributor to Vaginal Sample (After Crime Lab Reported the Second Man Had Been "Excluded")

	D8S1179	D21S11	D18S15
Defendant	**14,15**	**28, 31.2**	**12,19**
Second Man	**13,16**	**28, 32.2**	**14,14**

Vaginal Swab Profile (Showing Alleles Consistent with Defendant, but None Consistent with Second Man)



Vaginal Swab Profile After Defense Reanalysis of Electronic Data (Showing Additional Low-Level Alleles Consistent with "Excluded" Man)



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).

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 #2) provides another option for analysis of electronic data.

[Sidebar #2 Should Appear About Here]

Sidebar #2: Using Genophiler™ to Examine Electronic STR Data

Another option for review of electronic data is a service provided by Forensic Bioinformatics Services (FBS), a company established and operated by the authors of this article. FBS uses Genescan™ and Genotyper™ to analyze electronic data according to a systematic protocol that was designed to detect ambiguities, problems, and evidence supporting alternative interpretations. FBS is able to do the work at relatively low cost by using an automated "expert system" called Genophiler™. Genophiler™ is a computer program that operates Genescan and Genotyper the way a highly sophisticated human operator would--but faster and more systematically. Genophiler™ extracts all necessary information, analyzes it, and produces various reports of its results.

Defense lawyers can use these reports to rapidly determine whether there are any significant issues or problems in a case. Defense experts can use these reports as a basis for their own analysis and assessment of the case. All of the electropherograms and other critical data are automatically converted to Adobe Acrobat format, so that the defense expert need not have access to Genescan™ and Genotyper™ software to review and evaluate the electronic files. An example of Genophiler's™ outputs and reports can be found at the FBS web site at www.bioforensics.com.

Genophiler™ was developed by faculty in the Bioinformatics Program at Wright State University, with guidance and input from Simon Ford and William Thompson.

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.[6] 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.[7] 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.[8] The Las Vegas Crime Laboratory made the same error in a recent Las Vegas rape case.[9] This error, which came to light in April, 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 I 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.[10] Studies have also documented the inadvertent transfer of human DNA from one item to another.[11] *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 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 tell-tale 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.[12] 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.

Finding Experts

The complexity of 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, bio-informatics, 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 (see Sidebar) 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.

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Appendix: Model Discovery Request for STR Test Results

DISCOVERY REQUEST

INTRODUCTION**:**

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.
(6.3) 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:

(10.1) Current resume
(10.2) Job description
(10.3) A summary of proficiency test results

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.

[6] See, William C. Thompson, Franco Taroni, F. and Colin G. Aitken, How the probability of a false positive affects the value of DNA evidence. Journal of Forensic Sciences (January 2003, in press).

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

[8] Id.

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

[10] See, van Oorschot "DNA fingerprints from fingerprints," Nature, June 19,1997 at 767; Findlay, et al, "DNA fingerprinting from single cells" Nature, Volume 389, October 9, 1997 (555-556); Ladd, et al, "A Systematic Analysis of Secondary DNA Transfer," 1999 Journal of Forensic Science 44(6): 1270-12272;

[11] Ladd, et al., supra note 9.

[12] An unpublished report on this study may be obtained from William C. Thompson.