CHAPTER 13: DNA TYPING NOW AND BEFORE

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EVIDENCE

Each of us is genetically unique, and there are many cases in which it is convenient to make use of our genetic individuality: for parentage analysis, identification of victims, and identification of criminals. DNA provides one of the most specific methods of "typing" a person, but many features of ideal data are being violated when evidence has been gathered for criminal prosecution.

SECTION 1

Motivation

Someone has committed a violent crime, and some blood was left at the crime scene. The blood type (presumed to be that of the assailant) is AB. The suspect also has blood type AB. Is this fact sufficient evidence of guilt? No, for several reasons. One reason is that AB is too common in the general population to warrant any conclusions as to the guilt of the suspect. If we have more information such as: the genotype of the blood at the scene is both AB and X1, and the suspect is both of these, it is now perhaps more likely that the suspect is the source of the crime-scene blood. However, it is necessary to know how common is the combination of AB and X1 in the population of possible assailants. If this combination is very common, then we still do not have much more information than we had with the AB blood type alone. We want enough information to know that the chance of finding a random person with that genotype is small. DNA gives us this specificity.

SECTION 2

What is DNA Typing?

DNA typing is a method in which our genetic material (DNA) is converted into a “barcode” that, ultimately distinguishes each of us from nearly everyone else on earth. DNA is easily recovered from many sources, so that criminals often unwittingly leave their DNA at crime scenes, and the DNA of victims is even sometimes carried away on the clothes of their assailants. By using DNA, we are thus often able to place individuals at crime scenes, and in the case of rape, are able to identify the man who "provided" the sperm.

Recent numbers. By 1990, DNA technology had been used in over 2000 court cases in the U.S., encompassing 49 states and Washington D.C. The October 12, 1991 Austin American Statesman reported that Williamson County's first use of DNA typing had just resulted in the conviction of a rape suspect, who was sentenced to 99 years in prison. Not all DNA typing has led to convictions, however, and the news nowadays more often reports the release of someone in prison (often having served more than 10 years) because DNA analysis of the old samples shows that he cannot have committed the crime. From any attempt to match a DNA fingerprint between suspect and forensic sample, three outcomes are possible. For the U.S. up to 1990, these outcomes (and their frequencies) were: (i) exclusion of the suspect (37%), (ii) inability to resolve the DNA fingerprint (20-25%), and declaration of a match (40%).

DNA typing is or can be used for many different crimes and circumstances: rape, assault and murder, body identification, and establishing parentage. It is also useful in conservation (establishing that meat came from an endangered species, for example, or that a set of antlers came from a deer poached on someone’s ranch). And a specific kind of DNA typing is used in molecular epidemiology, to identify the source of infectious agents.

SECTION 3

Sources of DNA

DNA occurs in all living cells of our bodies, with the exception of most red blood cells. However, because our blood also contains white blood cells, DNA can be obtained from blood samples, even tiny ones. DNA can also be obtained from saliva (saliva has cells from your mouth in it), so spitting on something or licking something allows your DNA to be typed. Hair has DNA. The root of the hair has cells adhering to it, which can be used to type a person, but the shaft of the hair has degenerate DNA which can be used for some DNA typing processes but not others. Skin has DNA, so touching an object can leave enough cells for DNA typing: someone who wielded a baseball bat in an assault was identified by the DNA left from holding the bat. Bone has DNA. Chances are that a criminal will not leave behind pieces of his bones, but bones are typically used to identify remains of bodies that may be even decades old. And last, even feces have your DNA – biologists often use "scat" samples to identify individuals (in studies of bears, for example).

DNA is extremely useful, but it does not last forever. Environmental samples can degrade, especially if wet. It is thus important for forensics labs to keep the samples frozen.

SECTION 4

The Typing Process

In the barely-20 years that DNA typing has existed, the technology of DNA typing has changed a great deal. Earlier versions of this chapter devoted many pages to the technology. We will abandon that emphasis on technology in this installment and restrict attention to the basics, as follows:

i. Obtain a tissue sample and extract the DNA

ii. "Xerox" the DNA with a technique known as PCR (polymerase chain reaction)

iii. Determine the DNA type with either of two methods

a. **STR (short tandem repeat):** This method generates the typical DNA bar code and is based on variable regions of DNA from your chromosomes. Chromosome regions have been found that are highly variable among us (even the two chromosome sets you have differ in these regions). Typing involves the assessment of about 5 of these regions. The method measures the length of the DNA on your chromosomes at those regions but does not determine the actual sequence.

b. **Mitochondrial Sequence:** Each of your cells (except red blood cells again) contains hundreds to thousands of organelles known as mitochondria. Mitochondria ultimately evolved from bacteria, and they have their own miniature chromosome. Unlike the case with your (nuclear) chromosomes, all of the mitochondria in your body are inherited from your mother – so you have a single type. The sequence of your mitochondrial DNA matches that of your mother's mitochondrial DNA and can be used as one form of a DNA type.

iv. Observe whether one sample has the same DNA type as the other sample (e.g., suspect). If so, calculate the odds of obtaining a match at random (as if the suspect had no association with the crime). The "random match probability (RMP) is typically less than 1 in a million for STR types, but it is much greater for mitochondrial DNA types (e.g., 1 in 100). For example, two full brothers (who aren't identical twins) will certainly have different STR types, but they will also certainly have the same mitochondrial DNA sequence as each other, as their mother, and her mother, and as any other siblings, as any siblings of their mother, etc. Even so, mitochondrial types are often useful, and they can be determined from samples whose DNA is too degraded for STR determination (because mitochondrial DNA is present in so many more copies than is nuclear DNA).

Errors

When DNA typing was a new technology, its introduction to the courts in the U.S. was hotly contested by some scientists. One objection was that the DNA typing process itself was not meeting ideal data criteria. Initially, there were NO rules for DNA labs, and there were no certification procedures. Databases for evaluating RMPs were inadequate. Many of the former problems have been resolved with database expansion and with technologies that removes the subjectivity in assigning DNA type to a sample, but problems still remain, at least for some labs. In summer of 2003, the Houston Crime Lab made the news by having such sloppy DNA procedures that even the local authorities recommended withdrawal of its certification. Dr. Larry Mueller's web page at U.C. Irvine (http://darwin.bio.uci.edu/~mueller/ go to "Forensic DNA Resources" at the bottom of the left menu) lists some of the lab errors that he has encountered in his experiences as an expert witness for the defense. Another, more recent and comprehensive site is http://www.scientific.org/. Since most or all of these errors favored the prosecutions' cases until they were discovered, there is no incentive for the government to maintain a public record of them.

The types of errors and problems most commonly encountered fall into a few types (sample mixups and bad data analyses are apparently the most prevalent):

Sample Mixup:

This is probably the most common source of false matches – the people in the lab mixed up the samples. Sample mixup is understandable simply because the technologies involve use of standardized tubes and other plasticware, and unless one is absolutely rigorous, it is very easy to accidentally grab the wrong tube, or load the wrong well with a sample. Ultimately, every sample is handled by a person before it gets processed, and this step of human handling is the vulnerable one.

Sample Contamination:

Some cases of sample contamination are similar to sample mixup. In other cases, sample contamination occurs because an officer touches the material with his/her hands, or the contamination may occur when the sample is deposited (e.g., if a blood stain gets bacteria in it).

DNA Degradation:

DNA degrades if it is not kept cold or dry. Thus, by the time the police arrive at a crime scene, the DNA in some of the samples may already be bad. Improper storage of samples also contributes to degradation. Degradation may lead to inaccurate DNA typing, though more so for the STR method than for the mitochondrial method.

Bad Data Analysis:

The calculation of RMP may be straightforward in many cases, and some software automatically calculates it for each STR. However, unusual cases require a deep understanding of probabilities (and statistics), which if often lacking.

SECTION 6

Ideal Data: What's Missing?

Lab error rates are typically regarded as being around 2%, although the labs do what they can to conceal errors (as well as avoid them). If the RMP is as low as 1 in a million, a lab error rate of 2% dominates the considerations of the significance of a match, so labs need to be striving for vastly lower error rates than they have had in the past. As outsiders, it is difficult to know what all the causes of these errors are, but we can get an idea from past exposures of these errors. A big unknown is the extent to which a lab actually follows its own protocols. The written protocol is only a model of what is done, and if the technicians deviate from the written protocol, it is difficult to uncover that after the fact.

1. Absence of external, blind proficiency tests (inadequate standards). The only way a lab can begin to correct its mistakes is to know how often and why they occur. Blind proficiency tests are the surest way to know the lab's error rate. Few labs submit to external, blind proficiency tests, though all labs now submit to some form of proficiency testing. (A blind test means that the lab does not realize they are being tested on the sample; a blind test is good because it means that the technicians are being no more careful in testing that sample than in testing any other sample.)

2. Sample identification is known when processing occurs (bad protocol: absence of blind). By knowing which samples belong to which people (or crimes), it is far easier to unintentionally produce a false match (perhaps by sample mixup or contamination).

3. Samples from the same crime are often processed together, in the same lab (bad protocol). This greatly increases the chance of sample mixup going undetected.

4. Inadequate replication (bad protocol). With the use of PCR, a single sample can be processed many times (which was not true of past methods). Ideally, samples should be split and sent to different labs for testing, which would greatly reduce sample mixups going undetected. Cost is probably the biggest impediment to this kind of replication.

5. Bad protocols for data analysis. People analyzing DNA data have not usually been trained adequately for assessing the true RMP. It is thus common for the RMP to be miscalculated (and the error may go in favor of or against the defendant).

External Links

DNA Fingerprinting:

Example 1

Example 2

Example 3