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DNA Law

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> Hogan's r-right whin he says: 'Justice is blind.' Blind she is, an' deef an' dumb an' has a wooden leg. (Finley Peter Dunne, *Mr. Dooley's Opinions,* 1900)

> Facts are stubborn things; and whatever may be our wishes, our inclinations, or the dictates of our passions, they cannot alter the state of facts and evidence. (John Adams, December 1770)

In September 1943, Joan Berry gave birth to a baby daughter, Carol Ann. The father, she would claim in court, was Charlie Chaplin, something ardently denied by the great movie star. The court decided against Chaplin, who was ordered to pay for Carol Ann's support. Charlie Chaplin's blood group was O, Joan Berry's was A. Chaplin could not have been the biological father of Carol Ann, who had blood group B. The verdict was nevertheless affirmed on appeal, although with the vigorous dissent of Justice McComb, who argued that

> modern science [has] brought new aids . . . [that] have revised the judicial guessing game of the past into an institution approaching accuracy in portraying the truth as to the actual fact. . . . If the courts do not utilize these unimpeachable methods for acquiring accurate knowledge of pertinent facts they will neglect the employment of available, potent agencies which serve to avoid miscarriages of justice. (*Berry v. Chaplin*)

There are 3×10^9 nucleotide base pairs (bp) in the DNA of a human gamete. The two gametes from which a person develops differ from each other at $3–10 \times 10^6$ bp. It follows that the probability that a person will by chance produce two identical gametes is much smaller than 10^{-100}. Thus no two humans can ever be genetically identical (monozygotic twins excepted). The DNA is a fail-safe ID that uniquely identifies each person in the world, as law-enforcement agencies in the United States and other countries have been quick to notice.

It has been known since 1892 that the set of ten fingerprints is unique to each person. Dermatoglyphics has more extensive forensic applications than DNA typing simply because fingerprints are more likely than body fluids to be left at the scene of crimes such as burglaries and homicides. But in rapes and contact crimes, the perpetrator's blood, semen, saliva, or hair is often present on the victim or at the scene of the crime; moreover, blood stains or hairs from a victim may show in the criminal's clothing. DNA typing is particularly definitive when it *excludes* a suspect, something that cannot be accomplished with fingerprints. An additional difference between DNA typing and fingerprinting is that the only information that latent fingerprints can provide about a person is the person's identity, whereas DNA typing conveys medical and other information.

Molecular biology has provided access to DNA sequences. Obtaining the complete DNA sequence of an individual is beyond the possibilities of current technology, but DNA polymorphisms are so extensive that a few DNA fragments may yield distinctive individual patterns. A.J. Jeffreys and col-

leagues developed in 1985 a DNA "fingerprinting" method readily applicable to forensics (Jeffreys et al. 1985; Gill et al. 1985). There are regions in human DNA that consist of variable numbers of tandem repeats (VNTRs) of units that are each a few nucleotides long. The number of repeats identifies alleles at a given locus or DNA region. The alleles are manifested as bands by Southern probing of gels with restricted DNA. As many as 100 alleles may occur at a VNTR locus, so the population frequency of any given genotype is low. A few very polymorphic loci may then be sufficient to fingerprint or "type" an individual. The efficiency of the method can be increased by using Southern probes that hybridize to several VNTR loci in a single gel.

The technique saw its first forensic application in 1985 in the United Kingdom and in 1986 in the United States. Data were obtained at first in commercial laboratories, but the Federal Bureau of Investigation (FBI) set up its own DNA forensic laboratories in 1988.

The VNTR method is plagued by two kinds of problems. There is an issue of quality control: the reliability of the technical methods and interpretation of the band patterns. There are also difficulties in estimating the probability with which a particular DNA fingerprint can be unambiguously attributed to a person.

These problems were apparent to a number of observers from the start. Members of the Board on Biology of the National Research Council (NRC, an operating arm of the National Academy of Sciences and the National Academy of Engineering) alerted some federal agencies and proposed to study the issues and provide a report that might guide forensic practice. In the late 1980s, however, DNA typing was leading to a number of criminal convictions. Law-enforcement agencies remained blind to the potential problems and apparently perceived that the academy's study would just delay, and perhaps complicate, the forensic uses of DNA typing. The situation, however, would soon change, as defense lawyers obtained expert testimony that challenged conclusions reached by law-enforcing agencies. The technical challenges made criminal convictions increasingly difficult and some earlier convictions were overturned by the courts on appeal. By late 1989, the FBI and other federal agencies saw the need for an authoritative document from the National Academy of Sciences and offered the necessary financial support. Under the sponsorship of the Board on Biology, the work was started in January 1990 by a distinguished committee chaired by geneticist Victor McKusick of Johns Hopkins University. The committee's report was made public in the spring of 1992.

A litany of technical hurdles handicaps forensic DNA typing. The tissue samples available are small and may be degraded or commingled. Even under the best of circumstances, separation, probing, and matching of digested DNA fragments are demanding techniques that may yield confusing and conflicting results. The repetitions by which experimental scientists resolve ambiguities and repair failures are not options available to forensic scientists. DNA amplification by the polymerase chain reaction (PCR) has provided access to tissue samples that are too small for direct analysis, but PCR is a technique notoriously sensitive to experimental conditions and to contamination. The two most prominent commercial enterprises engaged in DNA typing (Lifecodes Corporation of Valhalla, New York, and Cellmark Diagnostics of Germantown, Maryland) have a deplorable record of mangled data, mixed samples, and disingenuous testimony. *DNA Technology in Forensic Science* recommends that a "National Committee on Forensic DNA Typing" be created to oversee technical correctness.

(The report's recommendation is that the committee on forensic DNA typing "provide expert advice . . . on scientific and technical issues," but we are not told who would be the recipients of the advice: forensic scientists and laboratories? regulatory agencies that will set standards? the courts? defense lawyers and prosecutors? law-enforcement agencies? *DNA Technology* is artlessly written, but the chapter on "Technical Considerations" is particularly egregious—exuberant with platitudinous statements, emphatic qualifiers, ambiguous antecedents, repetitions, and other insults to the language and to the reader's intelligence. I wonder why the NRC would not see that expertise in the expression and communication of ideas should be represented in the membership or supporting staff of its committees, and that draft reports should be held to language standards.)

A vexing problem in DNA forensics is how to determine that a DNA sample comes from a certain individual and nobody else. When identical DNA configurations are observed in a forensic sample and in a suspect, how do we ascertain that the sample came from the suspect rather than from some other individual with the same DNA pattern? (This question does not arise for exclusions: an individual can be definitely eliminated as the source of a sample if the DNAs are different.) I have said above that no two persons have identical DNA makeup, but only a small fraction of the DNA is examined in forensics.

In forensic practice, DNA identity between sample and suspect is not established with absolute certainty, although it may be "proved" with a proba-

bility high enough to meet the "beyond reasonable doubt" standard required in the courts of law. The issue is how the probability of identity is calculated. If the population frequency of a certain VNTR allele is 0.10, the probability that an individual will be homozygous for the allele is 0.01. DNA forensic laboratories typically examine four or five VNTR loci. Assume that at three additional loci the probabilities of the particular genotypes found in a sample are 0.02, 0.02, and 0.01. The expected frequency of the four-locus genotype is calculated as the product of the four frequencies, 4×10^{-8}, or one in 25 million. The odds against a suspect that has that particular genetic configuration would seem sufficiently high to support a conviction.

The calculus I have just illustrated makes, however, assumptions that may be erroneous; namely, that the genotype frequencies at the four loci are independent and that we know the relevant population frequencies. The NRC committee invested much effort and no little acrimony grappling with these assumptions.

How the issue of independence may affect the calculus of probabilities can be illustrated with a simple example. Assume that 10% of adult males in New York are blond and 20% have blue eyes. If the two characters are independent, the expected frequency of a blond blue-eyed man would be 2%; in fact, it is likely to be very nearly 10% since most blond males also are blue-eyed. Even when the loci are on different chromosomes, the genotypic frequencies may be correlated as a consequence of population subdivision.

Genotypic frequencies are disparate in different subpopulations: the frequency of blonds in New York is not the same among Caucasians, Blacks, and Hispanics. The expected probabilities obtained by multiplying the frequencies observed for the different loci may therefore be grossly off if the frequencies come from a subpopulation other than the suspect's.

Human populations differ in the frequency of blood groups, antigens, and other extensively sampled genetic markers. There are no theoretical grounds to anticipate that population variation is less with respect to VNTRs than it is for those better-known systems. VNTRs exhibit higher mutation rates and are subject to lower selective pressures than protein-encoding loci, which increases the chances that different human subpopulations might have radically different VNTR allelic frequencies.

One way out of this uncertainty would seem to be to measure allelic frequencies separately in different ethnic groups, such as Caucasians, Native Americans, Blacks, and Hispanics (Chakraborty and Kidd 1991; Weir 1992) But this approach can hardly suffice, since each of these ethnic groups is in turn very heterogeneous (Lewontin and Hartl 1991). I was born in Spain and thus am ethnically Hispanic, but I have not shared a common ancestor for at least 16,000 years with one of my Mexican students, who is pure Mayan.

The problem of ascertaining genetic frequencies in the relevant subpopulations would seem all but hopeless, since it is not even clear how to define the boundaries of the subpopulations relevant to particular cases, let alone how to obtain accurate estimates of their genetic frequencies. *DNA Technology* proposes an ingenious solution to this quandary, which it calls the "ceiling principle." The preliminary step is to determine the allelic frequencies at marker loci in 15–20 populations that are each fairly homogeneous and collectively representative of the various ethnic groups. A random sample of 100 persons from each population would suffice for the purpose. For each allele, select its highest frequency in any population or 0.05, whichever is higher. Use these allele "frequencies" for estimating the expected population frequency of the genotypes in the sample. This method overestimates the expected frequency of the genotypes and thus the probability of the composite genotype in a random sample from any population. It is a very conservative method, but any excesses are in the direction of decreasing the chances that the wrong suspect might be convicted. If the odds need to be increased in a particular situation, this can be accomplished by examining more loci. Until such a time as data for the recommended 15–20 populations become available, the NAS committee urges that the allelic frequency used in the calculations be the highest one so far observed in any population, but never less than 0.10 for any allele.

Expert witnesses in common law were first used in England in the 14th century as court-appointed witnesses. In America, by the 19th century experts were usually hired by the disputing parties. However, the courts reined the experts, limiting their role to conveying the consensual view of their profession. This practice became sanctioned in *Frye v. United States* by a federal appellate court that ruled in 1923 that only testimony founded on theories, methods, and procedures generally accepted by other experts in the same field could be presented in court. This "general acceptance" rule, known as the *Frye* test, is one of the two principles followed by the U.S. courts in deciding the admissibility of expert testimony as evidence. The other principle, known as the "helpfulness standard," comes from the Federal Rules of Evidence, first codified in 1975. Rule 702 states that expert testimony is allowed when "scientific, technical, or other special-

ized knowledge will assist the trier of fact to understand the evidence or to determine a fact."

The abuses perpetrated by scientific "experts" who present unsound testimony as trial evidence, and the undue damage they cause to innocent individuals, public health, the economy, and the legal process, have been recently described with authority and eloquence in a book by Peter W. Huber (1991) appropriately subtitled *Junk Science in the Courtroom*. Legal scholars are currently engaged in a debate concerning adherence to the *Frye* rule, which some see as the cure to the junk-science malaise, but which is ignored by many courts. There is little evidence that adherence to the *Frye* test is decisive. Courts that have rejected *Frye* often have been quite effective in controlling scientific testimony, whereas courts applying *Frye* have at times accepted questionable evidence (Ayala and Black in press).

The NAS committee points out that the *Frye* test sometimes prevents valid scientific evidence from being presented to a jury because it has not achieved a sufficient history in some discipline. The committee favors the helpfulness standard because it provides the court with more flexibility. (The committee's preference for Rule 702 over *Frye* would not surprise those familiar with the writings and judicial practice of the distinguished legal scholar Judge Jack B. Weinstein, who was a member of the committee.) In any case, the committee asserts that the theoretical basis of DNA typing is sound and recommends that the courts judicially notice this appropriateness by reference to *DNA Technology in Forensic Science*. But before permitting the introduction of DNA evidence, "the judge should determine . . . that appropriate standards have been followed, that tests were adequately performed by a reliable laboratory, and that the appropriate protocols . . . were fully complied with." The committee notices that a number of states have already enacted statutes that affirm the admissibility of DNA evidence; other states have implicitly acknowledged the same by statutorily creating DNA banks of convicted felons.

The creation of databanks, as the committee notes, raises constitutional issues of considerable legal import that merit sustained scrutiny. The potential uses and abuses of DNA typing extend well beyond the judicial system: identification of missing persons, detection of genetic ailments, invasion of privacy, unreasonable search or seizure, and many, many more. The wealth of legal, moral, and social issues that is at stake can provide enough subject matter to sustain the scholarly efforts of several academic departments for many years. For now, however, judicial applications will cast a large shadow. DNA typing will help convict many guilty parties and acquit still more. On May 3, 1992, the *New York Times* (p. 15, national edition) carried the story of Glen Dale Woodall, who after 4 years in prison was cleared by DNA tests. Mr. Woodall had been sentenced to two life prison terms without parole, plus 335 years, after being convicted in 1987 of kidnapping and raping two women. Both victims had identified Woodall as their assailant, but semen found on the women, tested after years of legal struggle, showed that Mr. Woodall could not have committed the crime.

There remain legislators and government officials who challenge the appropriateness of using the public purse for supporting basic research that lacks defined social objectives at the time when the research is proposed.

References

Ayala FJ, Black B (in press) The nature of science: a primer for the legal consumer of scientific information. Paper presented at a conference on "Science and the Law," Washington, DC, November 1–2, 1992

Berry v. Chaplin 169 P 2d, 453 (1946) Cited by Peter W Huber, Galileo's revenge: junk science in the courtroom. Basic Books, New York (1991), p 168

Chakraborty R, Kidd KK (1991) The utility of DNA typing in forensic work. Science 254:1735–1739

Gill P, Jeffreys AJ, Werrett DJ (1985) Forensic application of DNA "fingerprints." Nature 318:577–579

Huber PW (1991) Galileo's revenge: junk science in the courtroom. Basic Books, New York

Jeffreys AJ, Wilson V, Thein SL (1985) Individual-specific "fingerprints" of human DNA. Nature 316:76–79

Lewontin, RC, Hartl DL (1991) Population genetics in forensic DNA typing. Science 254:1745–1750

Weir BS (1992) Independence of VNTR alleles defined as fixed bins. Genetics 130:873–887