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THE ADMISSIBILITY OF DNA TESTING*

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In contrast to the widespread acceptance of red blood cell grouping, blood serum protein and enzyme analysis, and HLA typing,¹ the evidentiary status of forensic applications of recombinant-DNA technology is in flux. A proper evidentiary analysis must attend to the fact that there is no single method of DNA typing. As with the more established genetic tests, the probative value of the laboratory findings depends both on the procedure employed and the genetic characteristics that are discerned. This paper describes some of these procedures and the theory that lies behind them,² and then considers the developing case law.³

I. THEORY AND METHODS OF DNA TESTING

DNA is a long molecule with two strands that spiral around one another, forming a double helix.⁴ Within the double helix are molecules, called nucleotide bases, that link one strand to the other, like the steps of a spiral staircase. There are four of these bases, which can be referred to by their initials, A, T, G, and C. The A on one strand pairs with T on the other, and the G bonds to C. The lengthy sequence of AT and GC “stairs” within the DNA contained in human

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¹ See, e.g., MCCORMICK ON EVIDENCE § 205 (E. Cleary 3d ed. 1984).
² See generally FORENSIC DNA TECHNOLOGY (M. Farley & J. Harrington eds. 1991); DNA FINGERPRINTING: AN INTRODUCTION (Kirby ed. 1990); OFFICE OF TECHNOLOGY ASSESSMENT, GENETIC WITNESS: FORENSIC USES OF DNA TESTS (1990) [hereinafter GENETIC WITNESS].
cells includes all the genes and control sequences (for turning certain genes on and off). The genes are stretches of base pairs whose order determines the composition of proteins and related products synthesized by various cells. Oddly enough, however, much of the DNA has no known function.\(^5\)

Examining cell surface antigens (such as the ABO and HLA systems) or blood serum enzymes or proteins gives some information about the DNA sequences that code for these particular substances; if the markers differ, then the underlying DNA must differ. In contrast, DNA analysis is not limited to identifying variations in these coding sequences. With appropriate "DNA probes," one can detect differences in the base pair sequences anywhere in the DNA. A probe is a short piece of a single strand of DNA with a radioactive or other readily identifiable component attached, like a sticker or tag on a suitcase. If the bases in the target DNA are in an order matching those in the probe, the probe will bind to the target DNA.\(^6\)

Because 99.9% of the DNA sequence in any two people is identical, the technical challenge is to detect the relatively rare stretches of DNA, sometimes called alleles, that vary among individuals. Two detection procedures are currently in use. In one, the DNA is "amplified" by heating and cooling it with an enzyme called DNA polymerase.\(^7\) Even if the sample contains only one or two copies of the allele, the polymerase induces a chain reaction that increases the number to about 10 million. The amplified DNA is "spotted" onto a membrane, and a probe is added. If the sequence complementary to the probe is there, it will be tagged. If a radioactive element is used for the tag, for example, the spot will become radioactive and a dark dot will appear when the membrane is placed on X-ray film. The analyst simply looks to see whether the dot, and hence the allele, is present. This test resembles serologic tests in giving a categorical answer: either the allele is present or it is not.

The great advantage of the polymerase chain reaction over conventional immunogenetic and other DNA typing techniques is that it

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6 The probe binds to a strand of DNA whose base pairs are complementary to its own. For example, a probe that includes the sequence ATGCAG will hybridize to a target strand that includes the complementary sequence TACGTC.

requires very little biological material. As with serologic tests, however, a single allele may be common in the population, and hence not especially revealing. Of course, a series of probes may narrow the percentage of the population that could have been the source of the sample, but the procedure cannot identify any one individual as the only possible source.

The more frequently used procedure for identifying DNA variations ("polymorphisms") involves "digesting" DNA into fragments with enzymes ("restriction enzymes") from bacteria, separating the restriction fragments according to length by gel electrophoresis, blotting the array of fragments onto a nylon membrane, tagging the fragments with a probe, then placing X-ray film to the membrane to give an image with dark bands at the locations of the tagged fragments. The pattern of bands is the DNA "print" or "profile."

How many people have a given DNA profile (and hence, how

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9 A restriction enzyme binds to DNA when it encounters a certain short sequence (usually four to eight base pairs) and cleaves the DNA at a specific site within that sequence. Digesting a sample of DNA with such an enzyme usually gives rise to fragments ranging from several hundred to several thousand base pairs in length.

10 The broken pieces of DNA are loaded into small holes cut into one end of a slab of gel. Because DNA fragments have a negative charge, applying an electric field to the gel pulls all the fragments toward the positive pole. Larger fragments have more difficulty moving through the gel, so after a while, the smaller fragments migrate farther. When the electric current is turned off, equally long pieces of DNA will lie in a band near their starting point, and equally short pieces will be in a band toward the other end of the gel. The length of any particular fragment can be measured by comparing the distance it has travelled to the distances that standard fragments of known size placed in a parallel slot in the gel have migrated.

11 Here, however, the purpose of the probe is not to test for the presence of a given sequence, but merely to mark a fragment so that its length can be measured. Although the underlying polymorphism consists of differences in the sequence of nucleotide bases, only the length of the fragments is ascertained by using a particular restriction enzyme and probe. In this sense, the measured "alleles" are just the different lengths of the restriction fragments. Baird, Balazs, Giusti, Miyazaki, Nicholas, Wexler, Kanter, Glassberg, Allen, Rubinstein & Sussman, *Allele Frequency Distribution of Two Highly Polymorphic DNA Sequences in Three Ethnic Groups and Its Application to the Determination of Paternity*, 39 Am. J. Hum. Genetics 489 (1986). This usage has been criticized, see Kobinsky & Levine, *Recent Application of DNA Analysis to Issues of Paternity*, 33 J. Forensic Sci. 1107, 1108 (1988) (letter), and other researchers use the term "allele" to designate variations in base pair sequence.

valuable that profile is for identification) depends on where the restriction enzyme cuts the DNA (the restriction sites) and on the probe that picks out some of the resulting fragments. Suppose that some people have two restriction sites 32,000 bases apart, while others have an extra site located 12,000 bases inside this 32,000 base region. If the probe recognizes a sequence that occurs only on the shorter side of the extra restriction site, people with the extra site will have a profile consisting of one band for the 12,000 base fragment. People without the extra site also will have one band, but it will correspond to a fragment 32,000 bases long. Because the shorter 12,000 base band will migrate farther down the gel during electrophoresis, by placing DNA from two samples in parallel lanes on the same gel, an observer can tell whether one sample produces the smaller fragment while the other does not. The extra site thus gives rise to a restriction fragment length polymorphism ("RFLP") detectable with a particular enzyme-probe combination. However, there may be many people with each of the two possible bands, and this one simple site RFLP may not be very revealing.

Other enzyme-probe combinations generate many more possible length measurements within a population. Suppose that the 32,000 base pair sequence differs among individuals, not by a change in a single base pair at a given locus, creating or deleting an interior restriction site, but instead by the insertion of a short sequence starting at this locus and repeating itself many times. The more tandem repeats there are inside the restriction fragment, the longer it will be. A probe that detects the core repetitive sequence starting at this single site will detect these variable length fragments. A person with a single copy of the core sequence will have a band at the 32,000 point, someone with a hundred repeats of a core sequence ten base pairs long will have a band at the 33,000 point, and so on. Because the number of repeating units at a "variable number tandem repeat locus" ("VNTR locus") can vary greatly within a population, the probes that detect this type of repetitive DNA are generally much more informative than probes for simple site polymorphisms.

In addition to single-locus probes for VNTR loci, multiple-locus probes for VNTR loci have been developed and employed for forensic purposes. Some core sequences have tandem repeats not just at one locus, but in many places. Under proper conditions, a probe based on these core sequences detects length polymorphisms from all these loci scattered amidst many fragments, all at once. The developers of such probes call the set of 10 to 20 bands obtained with them "finger-
II. ADMISSIBILITY OF DNA TESTING

Initial journalistic\textsuperscript{15} and judicial praise for applications of RFLPs in homicide, rape, paternity, and other cases has been effusive. Indeed, one judge proclaimed “DNA fingerprinting” to be “the single greatest advance in the ‘search for truth’ . . . since the advent of cross-examination.”\textsuperscript{16} In this first wave of cases, expert testimony for the prosecution rarely was countered, and courts readily admitted RFLP findings.\textsuperscript{17}

Yet, the early enthusiasm for these techniques has led to second thoughts.\textsuperscript{18} The problems arise at two levels: controlling the experi-


\textsuperscript{14} However, with multilocus probes, it is difficult or impossible to tell which fragment comes from which locus. Some workers contend that the complexity of the patterns makes the procedure more subject to variation under experimental conditions, and the population genetics required for the statistical analysis of the results may be more controversial. \textit{Genetic Witness}, supra note 2, at 69.


\textsuperscript{17} Andrews v. State, 533 So. 2d 841 (Fla. Dist. Ct. App.), aff'd, 533 So. 2d 851 (1988); Wesley, 140 Misc. 2d 306, 333 N.Y.S.2d 643; \textit{In re “Baby Girl S,”} 140 Misc. 2d 299, 532 N.Y.S.2d 634 (County Sur. Ct. 1988) (admissible as “blood test” pursuant to statute governing paternity suit); Spencer v. Commonwealth, 238 Va. 295, 314, 384 S.E.2d 785, 797 (1989), cert. denied, 110 S. Ct. 1171 (1990) (DNA evidence properly admitted where “[i]t is not the record is replete with uncontradicted expert testimony that no ‘dissent whatsoever [exists in the scientific community]”’); State v. Woodall, 385 S.E.2d 253 (W. Va. 1989) (taking judicial notice of general scientific acceptance where there was no expert testimony, but holding that inconclusive results were properly excluded as irrelevant); \textit{Genetic Witness}, supra note 2, at 158-72 (collecting unreported as well as reported cases); cf. King v. Tanner, 142 Misc. 2d 1004, 539 N.Y.S.2d 617 (Sup. Ct. 1989) (slender action over an attribution of paternity); Alexander v. Alexander, 42 Ohio Misc. 2d 30, 537 N.E.2d 1310 (County Probate Ct. 1988) (permitting disinterment for “a DNA test” by an illegitimate child claiming an inheritance).

mental conditions of the analysis, and interpreting the results. Declaring matches or non-matches among the RFLPs due to VNTR loci in two samples is not always trivial. Furthermore, many forensic calculations of the probability of a coincidentally matching pattern have been oversimplified.

Despite these concerns, most recent cases continue to find forensic RFLP analyses to be generally accepted, and a number of states have provided for admissibility of DNA tests by legislation. Concerted attacks by defense experts of impeccable credentials, however, have produced a few cases rejecting specific proffers on the ground that the testing was not sufficiently rigorous.

In evaluating the general acceptance (as well as the validity and reliability of any DNA analysis), one must recognize that most of the probes used in criminal and paternity cases have no other medical or scientific application.

For a comprehensive survey of possible sources of error and ambiguity, see Thompson & Ford, The Meaning of a Match: Sources of Ambiguity in the Interpretation of DNA Prints, in FORENSIC DNA TECHNOLOGY 93 (Farley & Harrington eds. 1991).


Melson, Legal and Ethical Considerations, in DNA FINGERPRINTING: AN INTRODUCTION at 189, 199-200 (L. Kirby ed. 1990).

Caldwell v. State, 260 Ga. 278, 289-90, 393 S.E.2d 436, 443-44 (1990) (because laboratory’s calculation that frequency of profile in population was 1/24,000,000 rested on assumption inconsistent with its data base, the more conservative figure of 1/250,000 derived from that data base would have to be used); State v. Schwartz, 447 N.W.2d 422, 428 (Minn. 1989) (“DNA typing has gained general acceptance in the scientific community,” but “the laboratory in this case did not comport” with appropriate standards); People v. Castro, 144 Misc. 2d 956, 974, 545 N.Y.S.2d 985, 996 (Sup. Ct. 1989) (principles of DNA testing generally accepted, but “[i]n a piercing attack upon each molecule of evidence presented, the defense was successful in demonstrating to this court that the testing laboratory failed in its responsibility to perform the accepted scientific techniques and experiments”); cf. Commonwealth v. Cumin, 409 Mass. 218, 226, 565 N.E.2d 440, 444-45 (1991) (error to allow testimony that profile frequency was 1/59,000,000 in light of criticisms of the derivation of this figure).

But see Jeffrey, Wilson, Thein, Weatherall & Ponder, DNA “Fingerprints” and Segreg-
zymes followed by separation by gel electrophoresis and radioactive tagging of the fragments is a well established and fruitful research and diagnostic tool in medical genetics. 25 In most such applications, however, there is no need to measure precisely the position of bands or to estimate the frequency of these bands in the population; moreover, the information available in diagnostic work makes spurious or missing bands a much less serious problem. 26 Consequently, the outcome of an inquiry into general acceptance depends largely on the generality with which the question is posed. If all that need be accepted is the theoretical basis for DNA identification, there is no doubt that the technique is potentially admissible. 27 If proof that molecular biologists and geneticists believe that DNA analysis of possibly contaminated samples with probes of largely forensic interest are as infallible as some forensic analysts have maintained, then general acceptance is far more doubtful. 28 Given the ongoing debate over the standards and controls that should be used, perhaps the findings of laboratories that have yet to establish a track record on independently administered, blind proficiency tests should be inadmissible, 29 while proof that a lab-


26 GENETIC WITNESS, supra note 2, at 61-62; Lander, supra note 18, at 501; Thompson & Ford, supra note 3.

27 See, e.g., Commonwealth v. Curnin, 409 Mass. at 219, 565 N.E.2d at 441; GENETIC WITNESS, supra note 2, at 59 ("molecular and genetic principles underlying DNA techniques are solid and can be successfully applied to forensic casework"); id. at 66 ("That basic scientific principles of population genetics can be applied to forensic DNA analysis is not in question, but how best to apply which principles to single-locus RFLP analysis is under debate.").

28 GENETIC WITNESS, supra note 2, at 63 ("At present, scientists agree on the necessity for some controls, but not others . . . . Determining the type of controls necessary to ensure confidence in the results of any single DNA typing of a forensic specimen is of the highest priority"); Thompson & Ford, supra note 19 (adequate standards, controls and validation research for forensic DNA testing are not yet in place). But see GENETIC WITNESS, supra note 2, at 59 ("Forensic uses of DNA tests are valid"); id. at 60 ("properly performed, DNA technologies per se are reliable"); Budowle, Baechtel & Adams, Validation With Regard to Environmental Insults of the RFLP Procedure for Forensic Purposes, in FORENSIC DNA TECHNOLOGY 83 (M. Farley & J. Harrington eds. 1991).

29 At least one trial court, in an unreported criminal case in Arizona, held test results inadmissible when the FBI refused to disclose records of its internal proficiency testing. Thompson & Ford, supra note 19, at 145 n.124. It has been observed that only one meaningful
oratory has participated successfully in blind proficiency tests and has applied a similar or more rigorous protocol to the samples at bar should satisfy the threshold test for admissibility. In addition, the performance of the laboratory on the proficiency tests should accompany the results provided to the judge or jury. In this way, the trier of fact will be better positioned to assess the ability of the laboratory, using whatever type of DNA analysis and quality controls it has adopted, to obtain and interpret DNA profiles correctly.\textsuperscript{30}

\textsuperscript{30} For a description of currently available proficiency tests, see \textit{id.} at 142-45 (identifying "serious concerns"). Figures on the risk of a false positive are especially important to a fair evaluation of the weight of a match when estimates of the phenomenally small frequencies of the matching profile in the general population are introduced. \textit{See, e.g.,} Martinez v. State, 549 So.2d 694, 695 (Fla. App. 1989) ("one individual in 234 billion . . . would have the same banding pattern").

Professor Richard Lempert, in his oral remarks at this conference on Decision and Inference in Litigation, suggested that under certain conditions, evidence of proficiency testing could reinforce the tendency to misconstrue the probability of a coincidentally matching profile as a complete expression of the probative force of the DNA findings. He pointed out that given the relatively small numbers of samples in a proficiency test, many laboratories can be expected to achieve perfect scores even though the true risk of a false positive may be many orders of magnitude greater than the relative frequency of a matching profile. Rather than stating or implying that the probability of a false positive is zero in these circumstances, the proficiency data should be presented in a way that makes the effect of the limited sample size clear. That is, some procedure should be employed to estimate the upper bound on the error rate. An upper bound could be set, for example, by considering what the proportion of false positives would have been reported had the laboratory tested one more sample and falsely reported a match. No doubt, more suitable and sophisticated procedures are available. If these are pursued, the basic point—that even a player who bats a thousand in the first game has a non-zero chance of striking out later in the season—should be comprehensible to a jury.