

The Evaluation of Forensic DNA Evidence*

The following is an excerpt from the *Executive Summary* of the National Research Council Report.

INTRODUCTION

Nearly a decade has passed since DNA typing methods were first used in criminal investigations and trials. Law enforcement agencies have committed substantial resources to the technology; prosecutors, defense counsel, and judges have struggled with the terminology and ideas of molecular biology, genetics, and statistics. In 1992, a broad-ranging report released by the National Research Council attempted to explain the basics of the relevant science and technology, to offer suggestions for improving forensic DNA testing and its use in law enforcement, and to quiet the controversy that had followed the introduction of DNA profiling in court. Yet, the report did not eliminate all controversy. Indeed, in propounding what the committee regarded as a moderate position—the *ceiling principle* and the *interim ceiling principle*—the report itself became the target of criticism from scientists and lawyers on both sides of the debate on DNA evidence in the courts. Moreover, some of the statements in the 1992 report have been misinterpreted or misapplied in the courts.

This committee was formed to update and clarify discussion of the principles of *population genetics* and *statistics* as they apply to DNA evidence. Thus, this second report is much narrower than the 1992 report. Issues such as confidentiality and security, storage of samples for future use, the desirability and legality of data banks on convicted felons, and the international exchange of information are not in our charge. Rather, this report deals mainly with the computation of probabilities used to evaluate the implications of DNA test results that incriminate suspects. It focuses on situations where the *DNA profile* of a suspect (or sometimes a victim) apparently matches that of the *evidence DNA*. (We use the phrase “evidence DNA” to refer to the sample of biological material, such as blood or semen, usually taken from the crime scene or from the victim). The central question that the report addresses is this: What information can a forensic scientist, population geneticist, or statistician provide to assist a judge or jury in drawing inferences from the finding of a match?

To answer this question, the committee reviewed the scientific literature and the legal cases and commentary on DNA profiling, and it investigated the various criticisms that have been voiced about population data, statistics, and laboratory error. Much has been learned since the last report. The technology for DNA profiling and the methods for estimating frequencies and related statistics have progressed to the point where the reliability and validity of properly collected and analyzed DNA data should not be in doubt. The new recommendations presented here should pave the way to more effective use of DNA evidence.

This report describes both the science behind DNA profiling and the data on the frequency of profiles in the human populations, and it recommends procedures for providing various statistics that may be useful in the courtroom. The procedures are based on population genetics and statistics, and they render the ceiling principle and the interim ceiling principle unnecessary.

This *Executive Summary* outlines the structure and contents of the full report, and it gives the recommendations. This summary does not constitute a complete exposition, and it is no substitute for a careful reading of the chapters that follow. As the report will reveal, the committee agrees with many recommendations of the 1992 report but disagrees with others. Since the committee has not attempted to review all the statements and recommendations in the 1992 report, the lack of discussion of any statement should not be interpreted as either endorsing or rejecting that statement.

CONTENTS OF THE REPORT

Overview. The report begins with an extended summary of the chapters that make up the full report. This overview describes the essentials of the subject with the minimum of jargon, statistics, and technical details, and it includes a numerical example that illustrates how the procedures that are discussed and recommended would apply in a typical case. The main report offers fuller explanations, details, and justifications.

Chapter 1. The first chapter describes the 1992 report, the changes since that report, the uses and validity of DNA typing, differences between DNA typing in criminal cases and in civil paternity litigation, reasons for seemingly contradictory probability estimates that different experts sometimes present in court, and the committee’s approach to the issue of “population structure.”

Chapter 2. The second chapter describes the genetic and molecular basis of DNA typing. It introduces the fundamental concepts of genetics, and it surveys the genetic systems and the technologies used in DNA profiling.

Chapter 3. The third chapter concerns laboratory performance. Although our focus is on the statistics that can be used to characterize the significance or implications of a match between two DNA samples, these statistics do not float in a vacuum. They relate to specific claims or hypotheses about the origin of the DNA samples. If DNA from an evidence sample and DNA from a suspect share a profile that has a low frequency in the population, this suggests that the samples came from the same person; the lower the frequency, the stronger the evidence. But the possibility remains that the match is only apparent—that an error has occurred and the profiles differ from what the laboratory has reported. This chapter describes ways that errors can arise and how their occurrence might be minimized. It contains recommendations regarding quality control and assurance, laboratory accreditation, proficiency tests, and confirmatory testing.

Chapter 4. Much of the controversy about the forensic use of DNA has involved population genetics. The fourth chapter explains the generally applicable principles, then considers the implications of the fact that the population of the United States includes different groups and subgroups with different mixes of genes. The chapter develops and illustrates procedures for taking this fact into account in computing *random-match probabilities* for an incriminating DNA profile in a population or a subgroup of a population.

Chapter 5. The fifth chapter considers how the estimated frequency of an incriminating DNA profile relates to conclu-

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Abbreviations and technical terms are defined in the glossary. The underlying concepts are explained in the overview and in appropriate chapters in the body of the report.

sions about the source of the DNA in the evidence sample. It discusses how the frequencies are interpreted as probabilities and related quantities, the degree of uncertainty in such estimates, and the type of calculations that might indicate that a profile is unique. It concludes that the abundance of data in different ethnic groups within the major races and the methods outlined in *Chapters 4 and 5* imply that the 1992 report's suggested ceiling principle and interim ceiling principle are unnecessary. In addition, it makes recommendations to help assure the accuracy of estimates for what are known as *VNTR profiles* and to handle the special situation in which the suspect was identified as a result of a search through a database of DNA profiles of known offenders.

Chapter 6. The sixth and final chapter discusses the legal implications of the conclusions and recommendations. It describes the most important legal rules that affect the use of DNA evidence, identifies the questions of scientific fact that have been disputed in court, reviews case law on the admissibility of DNA evidence, and explains how the conclusions and recommendations might be used in applying and developing the law. The report makes no recommendations on matters of legal policy, but it does suggest that the formulation of such policy might be assisted by behavioral research into the various ways that DNA test results can be presented in the courtroom.

Appendices. A glossary of scientific terms is provided at the end of this report.

RECOMMENDATIONS

Major conclusions and recommendations are given at the end of the chapter in which the subject is discussed. For convenience, the report also lists them as a group at the end of the overview. This *Executive Summary* lists the recommendations only.

Recommendations to Improve Laboratory Performance

Recommendation 3.1. Laboratories should adhere to high quality standards (such as those defined by the Technical Working Group on DNA Analysis Methods and the DNA Advisory Board) and make every effort to be accredited for DNA work (by such organizations as the American Society of Crime Laboratory Directors—Laboratory Accreditation Board).

Recommendation 3.2. Laboratories should participate regularly in proficiency tests, and the results should be available for court proceedings.

Recommendation 3.3. Whenever feasible, forensic samples should be divided into two or more parts at the earliest practicable stage and the unused parts retained to permit additional tests. The used and saved portions should be stored and handled separately. Any additional tests should be performed independently of the first by personnel not involved in the first test and preferably in a different laboratory.

Recommendations for Estimating Random-Match Probabilities

Recommendation 4.1. In general, the calculation of a profile frequency should be made with the product rule. If the race of the person who left the evidence-sample DNA is known, the database for the person's race should be used; if the race is not known, calculations for all the racial groups to which possible suspects belong should be made. For systems such as VNTRs, in which a heterozygous locus can be mistaken for a homozygous one, if an upper bound on the frequency of the genotype at an apparently homozygous locus (single band) is desired, then twice the allele (bin) frequency, $2p$, should be used instead of p^2 . For systems in which exact genotypes can be determined, $p^2 + p(1 - p)\theta$ should be used for the frequency at such a locus instead of p^2 . A conservative value of θ for the

U.S. population is 0.01; for some small, isolated populations, a value of 0.03 may be more appropriate. For both systems, $2p_i p_j$ should be used for heterozygotes.[†]

Recommendation 4.2. If the particular subpopulation from which the evidence sample came is known, the allele frequencies for the specific subgroup should be used as described in *Recommendation 4.1*. If allele frequencies for the subgroup are not available, although data for the full population are, then the calculations should use the population-structure equations 4.10 for each locus, and the resulting values should then be multiplied.

Recommendation 4.3. If the person who contributed the evidence sample is from a group or tribe for which no adequate database exists, data from several other groups or tribes thought to be closely related to it should be used. The profile frequency should be calculated as described in *Recommendation 4.1* for each group or tribe.

Recommendation 4.4. If the possible contributors of the evidence sample include relatives of the suspect, DNA profiles of those relatives should be obtained. If these profiles cannot be obtained, the probability of finding the evidence profile in those relatives should be calculated with formula 4.8 or 4.9.

Recommendations on Interpreting the Results of Database Searches, on Binning, and on Establishing the Uniqueness of Profiles

Recommendation 5.1. When the suspect is found by a search of DNA databases, the random-match probability should be multiplied by n , the number of persons in the database.

Recommendation 5.2. If floating bins are used to calculate the random-match probabilities, each bin should coincide with the corresponding match window. If fixed bins are employed, then the fixed bin that has the largest frequency among those overlapped by the match window should be used.

Recommendation 5.3. Research into the identification and validation of more and better marker systems for forensic analysis should continue with a view to making each profile unique.

Recommendation on Research on Juror Comprehension

Recommendation 6.1. Behavioral research should be carried out to identify any conditions that might cause a trier of fact to misinterpret evidence on DNA profiling and to assess how well various ways of presenting expert testimony on DNA can reduce any such misunderstandings.

GLOSSARY

Allele one of two or more alternative forms of a gene

Chromosome the structure by which hereditary information is physically transmitted from one generation to the next; the organelle that carries the genes

Deoxyribonucleic acid (DNA) the genetic material of organisms, usually double-stranded—composed of two complementary chains of nucleotides in the form of a double helix; a class of nucleic acids characterized by the presence of the sugar deoxyribose and the four bases adenine, cytosine, guanine, and thymine

Diploid having two sets of chromosomes, in pairs

[†]The $2p$ rule involves replacing the quantity p^2 for a *single-banded VNTR locus* with the much larger quantity $2p$ in the product rule. This substitution accounts for cases in which one VNTR band from a heterozygote is not detected, and the person is mistakenly classified as a homozygote. The substitution also ensures that the estimate of the profile frequency will be larger than an estimate from a more precise formula that accounts for the population structure explicitly. The technology for *PCR-based systems* however, does not have these problems, and the $2p$ rule is inappropriate for these systems. Therefore, *Recommendation 4.1* calls for using $p^2 + p(1 - p)\theta$ (rather than $2p$) in place of p^2 for such systems.

DNA deoxyribonucleic acid

DNA databank (database) a collection of DNA typing profiles of selected or randomly chosen individuals

DNA probe a short segment of single-stranded DNA labeled with a radioactive or chemical tag that is used to detect the presence of a particular DNA sequence through hybridization to its complementary sequence

Gene the basic unit of heredity; a sequence of DNA nucleotides on a chromosome

Genotype the genetic makeup of an organism, as distinguished from its physical appearance or phenotype

Heterozygote a diploid organism that carries different alleles at one or more genetic loci on its homologous chromosomes

Heterozygous having different alleles at a particular locus; for most forensic DNA probes, the autoradiogram displays two bands if the person is heterozygous at the locus

Homozygote a diploid organism that carries identical alleles at one or more genetic loci on its homologous chromosomes

Homozygous having the same allele at a particular locus; for most forensic DNA probes, the autoradiogram displays a single band if the person is homozygous at the locus

Locus (plural loci) the specific physical location of a gene on a chromosome

Marker a gene with a known location on a chromosome and a clear-cut phenotype that is used as a point of reference in the mapping of other loci

PCR polymerase chain reaction

Polymerase chain reaction (PCR) an *in vitro* process that yields millions of copies of desired DNA through repeated cycling of a reaction that involves the enzyme DNA polymerase

Population a group of individuals occupying a given area at a given time

Proficiency tests tests to evaluate the competence of technicians and the quality performance of a laboratory; in open tests, the technicians are aware that they are being tested, but in blind tests, they are not aware; internal proficiency tests are conducted by the laboratory itself, and external tests are conducted by an agency independent of the laboratory being tested

Variable number of tandem repeats (VNTR) repeating units of a DNA sequence for which the number varies between individuals

VNTR variable number of tandem repeats

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Commission on DNA Forensic Science: An Update

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PETER J. BICKEL, University of California, Berkeley

MICHAEL T. CLEGG, University of California, Riverside

BURTON SINGER, Princeton University, Princeton, New Jersey

NRC Staff

PAUL GILMAN, Executive Director

Committee on DNA Forensic Science: An Update

JAMES F. CROW, PH.D.‡§ (Chair)

Professor Emeritus of Genetics

University of Wisconsin

Madison, WI

MARGARET A. BERGER, J.D.

Professor of Law

Brooklyn Law School

Brooklyn, NY

SHARI S. DIAMOND, J.D., PH.D.

Professor of Psychology and Criminal Justice

University of Illinois

Chicago, IL

DAVID H. KAYE, J.D.

Regents' Professor of Law

College of Law

Arizona State University

Tempe, AZ

HAIG H. KAZAZIAN, JR., M.D.§

Chairman of the Department of Genetics and

Seymour Gray Professor of Molecular Medicine in Genetics

University of Pennsylvania School of Medicine

Philadelphia, PA

ARNO G. MOTULSKY, M.D.‡§

Professor of Medicine and Genetics

University of Washington

Seattle, WA

THOMAS A. NAGYLAKI, PH.D.

Professor of Ecology and Evolution and of Genetics

University of Chicago

Chicago, IL

MASATOSHI NEI, PH.D.

Evan Pugh Professor of Biology and

Director Institute of Molecular Evolutionary Genetics

Pennsylvania State University

University Park, PA

GEORGE F. SENSABAUGH, JR., D.CRIM.

Professor of Forensic Science and Biomedical Sciences

School of Public Health

University of California

Berkeley, CA

DAVID O. SIEGMUND, PH.D.

Professor of Statistics and

Associate Dean for the Natural Sciences

Stanford University

Stanford, CA

STEPHEN M. STIGLER, PH.D.

Ernest DeWitt Burton Distinguished Service Professor of Statistics

University of Chicago

Chicago, IL

National Research Council Staff

Advisor

VICTOR A. MCKUSICK, The Johns Hopkins Hospital, Baltimore, Maryland

NRC Staff

ERIC A. FISCHER, Study Director

LEE R. PAULSON, Senior Staff Officer

MIRON L. STRAF, Senior Staff Officer

JOHN R. TUCKER, Senior Staff Officer

PAULETTE A. ADAMS, Senior Project Assistant

NORMAN GROSSBLATT, Editor

‡Member, National Academy of Sciences.

§Member, Institute of Medicine.