



Forensic and Legal
Applications



FOREWORD BY

NOBEL LAUREATE

DR. JAMES WATSON AND

DR. JAN WITKOWSKI

Lawrence Kobilinsky Thomas F. Liotti Jamel Oeser-Sweat

# DNA: Forensic and Legal Applications

## Lawrence Kobilinsky

John Jay College of Criminal Justice The City University of New York

Thomas F. Liotti

Law Offices of Thomas F. Liotti

Jamel Oeser-Sweat

Law Offices of Jamel Oeser-Sweat



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# DNA: Forensic and Legal Applications

## Foreword

The Banbury Center of Cold Spring Harbor Laboratory was the place to be in November 1988, if you were interested in forensic applications of DNA fingerprinting. Molecular geneticists, population geneticists, forensic scientists, lawyers, and bioethicists came to Banbury for what turned out to be an historic meeting. It was only three years earlier that Alec Jeffreys had published an esoteric study describing a curious DNA sequence that he had found in the myoglobin gene. It ceased to be an arcane topic when Jeffreys and his colleagues showed how this DNA sequence could be used for individual identification and demonstrated its use in an immigration case. And when, in 1987, Jeffreys was called in to assist in a notorious murder case, DNA fingerprinting attracted worldwide attention.

In the two years prior to the Banbury Center meeting, DNA fingerprinting was taken up eagerly by the forensic community, eager to have an identification method that was of far greater applicability than fingerprinting (traces of DNA are left everywhere); appeared to be of far greater precision (identifications could be made with figures like one in ten billion); and it came with the cachet of molecular genetics. But by 1988 controversies had arisen and DNA fingerprinting was under attack; the numerical estimates of identity were challenged; the competence of forensic laboratories doing DNA testing was called into question; and concerns were raised about the collection, retention, and use of DNA samples.

Alec Jeffreys was one of the participants who came to Banbury that November, together with others who were to become well known in the coming years for their involvement with DNA profiling: Bruce Budowle, Tom Caskey, Rockne Harmon, John Hicks, Eric Lander, Henry Lee, Peter Neufeld, and Barry Scheck, to name a few. The meeting proved historic because it was the first to subject the forensic practice of DNA fingerprinting to critical scrutiny. It was controversial, and discussions

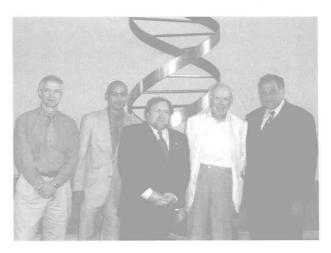
were frequently heated, but it initiated a comprehensive review of DNA fingerprinting including studies by independent bodies such as the National Academy of Sciences. These in turn led to improvements in technique; refinements in statistical analysis; the establishment of quality controls; and a change in way DNA evidence is regarded in the courtroom. Indeed, it may yet be that the rigorous scrutiny of the theory and practice of DNA fingerprinting will come to be applied to other forms of forensic evidence.

Scientific findings move slowly from the laboratory bench to application in the wider world, so the rapidity with which DNA fingerprinting moved from Jeffreys' laboratory to become the gold standard in evidence is unprecedented. And, just as the DNA double helix became emblematic of scientific research, so, through exposure on television programs such as *CSI*, DNA fingerprinting has become an icon for forensic science.

Last year was the 50th anniversary of the DNA double helix, and there was a grand, black tie gala at the Waldorf. One speaker was Marvin Anderson, who had been sentenced to 210 years, and had served 15 years, for a crime he did not commit. DNA evidence had exonerated him, and now he was addressing a gathering of Nobel laureates, eminent scientists, and celebrities. He came down from the stage to the table where Alec Jeffreys was sitting and the two embraced. A fitting and moving testimony to the power of DNA fingerprinting.

DNA: Forensic and Legal Applications is a comprehensive and invaluable guide to the field, covering topics ranging from collecting samples in the field to presenting the complex results to a jury. We are sure that it will play its part in promoting this most powerful tool in the forensic scientist's armamentarium.

JAMES D. WATSON JAN A. WITKOWSKI Cold Spring Harbor Laboratory



Left to Right: Dr. Jan Witkowski, Jamel Oeser-Sweat, Dr. Lawrence Kobilinsky, Dr. James Watson, Thomas F. Liotti

# Preface

The recently developed techniques that permit human identification by analysis of specific regions of DNA within the human genome have emerged as powerful evidentiary tools for the criminal justice system. The realization that a person can be "individualized" by analyzing his or her DNA has been heralded as one of the greatest revelations of the twentieth century. The number of clinical, scientific, and forensic uses for DNA grows with each passing day. As scientists strive to elucidate the many mysteries locked in the code that comprises DNA, we begin to understand why nature has made it the medium for storage of its blueprint for life, the genetic code. In DNA lies not merely the story of our evolution but also who we are, what lies in store for us in the future, and perhaps even the reason for our very existence.

Forensic DNA analysis has had a major impact on our criminal justice system and on the law during the last decade of the twentieth century. It has been employed in criminal law to help prove guilt or innocence, in family law to prove paternity, and in immigration law to prove blood relationships to establish citizenship. Its usefulness as a human identification tool is clear. Accordingly, in recent years, our legal system has given forensic DNA analysis the credibility that nature has given it as the blueprint of life. However, the examination of DNA can become compromised when environmental factors intervene, leading to deterioration, destruction, or contamination of the evidence, or when human error results in incorrect conclusions. These factors are crucial in determining what weight to give DNA evidence. Determining whether these factors exist and, if so, the extent to which they have corrupted the evidence or compromised the analysis are important tasks for a lawyer. A lawyer must not only understand what is advantageous about the science of DNA but what can go wrong and how to detect and prevent procedural errors. Attorneys facing trials in which DNA evidence will be offered must understand the underlying

science and technology on which DNA testing is based. This book guides attorneys and judges through the complexities of the biochemical sciences to help them understand the methodology of DNA analysis. It will provide them with this knowledge so that, at trial, they can ask appropriate questions and understand the responses that are given. This book has been written for students of science and law, for criminal justice practitioners, and for those forensic scientists who do not currently work in the field of DNA identification but who seek to learn more about the scientific and legal procedures involved. It is assumed that the reader knows very little about DNA and written in a style that is easy to read and comprehend.

The first chapter will provide the reader with the background necessary to understand the science underlying the common tests employing DNA. While much can be written regarding the chemistry, uses, and functions of DNA, this discussion is limited to an overview of its chemistry, structure, and its ability to replicate, which provides the information necessary to explore more advanced topics including the molecular biology and forensic applications of DNA in later portions of the book.

The second chapter provides the reader with information on the techniques employed by criminalists on the path from crime scene to final result; evidence is recognized at a crime scene and samples are collected, documented, packaged and brought to the laboratory, and analysis begins. It explores several issues that are relevant to each of the above procedures, as well as the impact of environmental factors, contamination, aging, and so forth on DNA evidence and test results. It also reviews methods of chemical and/or physical identification of common items of biological evidence.

The third chapter familiarizes the reader with procedures used to analyze biological evidence to determine its origin and if an association can be made between a suspect, victim, and/or crime scene. The different kinds of human DNA identification tests are reviewed, beginning with DNA fingerprinting, which was developed in the mid-1980s and used effectively into the mid-1990s. The chapter continues with a discussion of tests based on reverse dot blot technology, amplified fragment-length polymorphism (AmpFLP) analysis, and finally, the two current state-of-the-art techniques, known as PCR-STR analysis and mitochondrial DNA sequencing. Each of these tests differs markedly from the other, and each is a product of scientific and technological advancements. The advantages, drawbacks, and significance of each procedure are described. The chapter introduces the reader to several important issues related to the interpretation of test data. Some background knowledge of biochemistry and the molecular biology of DNA is necessary to understand the specific details of these technical procedures. It is helpful to have some knowledge of basic statistics and the laws of probability to appreciate the significance of the test results.

The first part of Chapter 4 discusses human genetics, population genetics, and statistics. Mendel's laws of heredity and the Hardy-Weinberg equilibrium are explained. Population genetics provides the scientific foundation for using DNA testing to develop genetic profiles whose frequencies of occurrence are so rare that each can be considered unique to an individual.

In certain cases, where the quality or quantity of nuclear DNA is limited, mitochondrial DNA testing is conducted. In the case of sexual assault, Y-chromosomal STR DNA testing can be advantageous. These tests as well as single-nucleotide polymorphism analysis are also discussed in Chapter 4.

A discussion of the importance of having a national database of digitized DNA profiles, CODIS, to help solve crimes where biological evidence has been found but where no suspects have been identified by eyewitness or police investigation is also included in Chapter 4. The CODIS database has been especially helpful when serial criminals perpetrate their crimes across state lines. A discussion of several statistical considerations in genetic testing, including the power of discrimination, probability of exclusion, and the likelihood ratio, are also included. There is also a discussion of how to treat evidence that is composed of a mixture of DNA from a number of sources. Regarding the use of DNA in civil law, we provide a discussion of how paternity is currently established by accredited testing laboratories.

The fourth chapter also discusses the need for quality control and quality assurance in the forensic laboratory. Quality assurance is demonstrated by the laboratory's accreditation, certification of its personnel, and proficiency testing. The chapter closes with a review of four DNA reports, the first dealing with RFLP, the second with PCR-based testing including HLA DQA1, Polymarker, and D1S80, the third, describing results of a PCR-STR analysis, and the fourth a paternity report based on DNA testing.

The fifth chapter will provide those who work in the criminal justice system, but who have little or no science background, the ability to understand and interpret DNA evidence with respect to past and present law. This chapter bridges the disciplines of science and the law by focusing on the admissibility of scientific evidence. It shows how the law applies to the evidence that has been collected and analyzed, and the findings and subsequent summary report issued by the laboratory. The chapter includes a discussion of the Federal Rules of Evidence, the *Daubert, Joiner*, and *Kumho Tire* decisions of the U.S. Supreme Court, as well as the judicial gatekeeping function of judges. It outlines what types of evidence are or are not admissible and relevant cases are discussed. The various aspects of a legal proceeding are detailed, including arraignment, grand jury, discovery, preparation for trial, jury selection and voir dire, opening statements, direct examination, cross-examination, and closing arguments.

The sixth chapter of this book introduces the reader to the concepts and procedures of challenging or defending DNA evidence. It opens with the importance of developing a strategy of how to have DNA evidence admitted at trial. It also explains how to make the best use of an expert witness by thoroughly and properly preparing him or her to testify. There is a discussion of how to introduce the expert and his or her credentials to the court to be deemed qualified as an expert witness. There is a list of issues that should be brought out during direct examination of the expert, allowing the jury to learn about the DNA testing, experimental observations, and, finally, the expert's conclusions.

The chapter then addresses the defense effort to mitigate the effects of DNA evidence being introduced by the prosecution. There are a number of potential routes to attack the admissibility of all or part of the DNA evidence including (a) expert not qualified, (b) expert not certified, (c) laboratory not accredited, (d) lack of discovery,

and (e) improperly obtained evidence. There may be a challenge to the statistics, to the database, or to "insufficient" quality control or quality assurance used in the laboratory, or to a perceived break in the chain of evidence. These arguments may be helpful to the attorney who has never before litigated a DNA trial and can be beneficial for the experienced attorney by its comprehensive review of important issues to be brought out in testimony. It concludes with a discussion of the type of summation that might be effective in trials of this sort.

The seventh and final chapter of this book discusses postconviction appeals based on analysis of existing DNA evidence. It discusses the role of the prosecutor and defense counsel in achieving a just solution for the innocent convict. It also discusses the legal standards governing postconviction testing. The Innocence Project has been highlighted as the first and most successful organized effort to exonerate innocent convicts. The chapter ends with a brief discussion about the future of DNA technology. It attempts to explain how new and improved technology will make analysis of evidence at the crime scene possible. Testing will be far more rapid, more economical, easier to perform, less labor intensive, and even more reliable. Today's technology sometimes fails to identify the source of evidence that had become seriously degraded or corrupted as a result of environmental insult. The same evidence could produce results using the technology of the future. In addition to the currently performed STR analysis, sequencing analysis and SNP detection technology are both likely to become more and more utilized by the forensic analyst. The impact of these changes on the criminal investigatory process and on existing national, state, and local DNA database collections is also explored.

This book is unprecedented in its merger of law with the science of DNA. Now for the first time in a single volume lawyers, judges, scientists, professors, students, and experts can find everything that they need in order to understand the forensic and legal applications of DNA. The science of DNA and its potential in and outside of the courtroom is unlimited. It has already changed the face of jurisprudence as perhaps the single most important development in science and in the law in the past half century. The evolution of the merger has just begun. The book takes the reader on a guided tour of what the future holds in store for all of us, but instead of being mere passive observers, each of us can now be active participants in the ever changing world of science and the law.

LAWRENCE KOBILINSKY THOMAS F. LIOTTI JAMEL OESER-SWEAT

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