

Chapter 1

Introduction

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§ 1:1 Overview of forensic DNA

With the advent of the Human Genome Project in 1990, the rate of genetic information available for use worldwide was set to increase exponentially. A short 11 years later, scientists determined the order of the three billion chemical base pairs that make up the building blocks of human DNA (deoxyribonucleic acid).¹ Since then, our understanding of the human genome has continued to grow at a blazing pace. Application of this information has been creative and crosses many fields, including medicine, biotechnology, anthropology, animal husbandry, agriculture, and, of course, law.

It has been said that the 21st century will be the century of biology.² But the opening of this new age arguably occurred during the 20th century, in 1953, when James Watson and Francis Crick, using information Rosalind Franklin

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¹A press conference marking completion of the Human Genome Project was held April 14, 2003, former President William J. Clinton presiding. The sequence was published the same day. International Consortium Completes Human Genome Project, available online at <http://www.genome.gov/11006929> (as of Jan. 23, 2015).

The entire issue of NATURE dated February 15, 2001, is dedicated to the publication of the sequence of the human genome and related commentary. It is available online at <http://www.nature.com/nature/journal/v409/n6822/index.html> (as of Jan. 23, 2015).

²John Carey, We are Now Starting the Century of Biology, BusinessWeek.com, Aug. 31, 1998, <http://www.businessweek.com/1998/35/b3593020.htm> (as of Jan. 23, 2015) (quoting J. Craig Venter, then president of The Institute for Genomic Research).

developed, first elucidated the DNA molecule's structure.³ It took another 13 years to determine how the DNA molecule actually codes information.⁴ Laboratory techniques soon followed, allowing scientists to isolate DNA from cells and to manipulate DNA fragments.⁵

In humans, about one-tenth of one percent of DNA (about three million bases) differs from person to person. In other words, the order of the bases in humans varies *on average* by one base in 1,000. As researchers began to recognize these sites of variation, others imagined using them as a type of individual identification system. In the mid-1980s, Dr. Alec Jeffreys, an English geneticist, found that certain regions of DNA contained DNA sequences that repeated over and over again next to each other. He also discovered that the number of repeated sections in a sample could differ from one person to another. He developed a method for examining variations in the lengths of these DNA repeat sequences, thus creating the ability to perform human identity tests.

Since then, forensic use of DNA as a tool of human identification has become widespread. Although initial data were somewhat crude by today's standards, DNA evidence offered courts an independent, science-based identification method. Subsequent improvements in technology have dramatically increased both the sensitivity and specificity of DNA forensics data.

Forensic use of DNA typically begins with a sample of biological material, which often is collected from a crime victim or a crime scene. Scientists identify a limited number of genetic markers in the sample by deploying small pieces of manufactured chemical sequences (primers) that seek out and bind to complementary DNA sequences of interest in the sample. A series of primers bound to a DNA sample permits amplification of the original sample to the point that the analyst can determine a DNA "profile" for the known or unidentified individual who deposited the sample.

With a DNA sample from an unknown source, the next step in the forensic identification process is comparing the

³The first description of DNA's structure appeared in a paper titled "Molecular Structure of Nucleic Acids," published in *NATURE*, Vol. 171, p. 737, 1953. For their efforts, the paper's authors, James Watson and Francis Crick, received the Nobel Prize for Physiology or Medicine.

⁴The genetic code, the mechanism by which DNA codes for proteins, was published by Holley, Khorana, and Nirenberg. For their work, they were jointly awarded the 1968 Nobel Prize in Medicine.

⁵A timeline chronicling major events in this process is available online at <http://homepage.smc.edu/HGP/history.htm#timeline> (as of Jan. 23, 2015).

sample to the known forensic DNA profile of a suspect or to the millions of DNA profiles now stored in computer databases throughout the country. Any known profile that does not match eliminates that person as the source of DNA collected from the victim or at the crime scene.

If a match is found, however, the next step is determining its significance. Even unrelated people share, on average, two or three genetic markers, so a match is of little significance absent information about the probability that it occurred solely by chance. The smaller the probability of a random match, the greater the chance that the source of the comparison sample was also the source of the DNA found at the crime scene. To reduce the chance of a random match, 13 or more DNA regions, or loci, that vary from person to person are generally used to create the DNA profile. Given the frequency with which DNA characteristics of interest appear in populations, any given “target” forensic DNA profile is going to be exceptionally rare. To quantify this chance, forensic scientists use a statistical method called the “product rule,” which involves estimating the frequency with which each genetic marker appears in the populations and multiplying the frequencies together to produce the complete profile’s frequency. This result is often also expressed as the probability that the DNA of a single person selected at random from the relevant population will match the evidentiary sample.

The use of DNA identification technology has transformed the work of those in the criminal justice system. Police, prosecutors, and defense counsel rely heavily on DNA evidence to do their jobs. Throughout the country, huge DNA databases are being compiled with DNA-based identification information of felons and arrestees that can later be tested against samples from crime scenes. Using these databases, law enforcement authorities have been able to make arrests in crimes that have gone unsolved for decades. As of December 2014, the FBI’s national DNA database, which includes genetic profiles of over 11 million offenders and 1.3 million arrestees, had produced over 270,000 DNA matches assisting in over 258,000 investigations.⁶ Of course, DNA identity evidence can aid the accused and the already-convicted as well as the accuser. As of January 2015, there had been 325 postconviction DNA exonerations in the United States.⁷

But the impact of DNA evidence in criminal trials is not

⁶Federal Bureau of Investigation, CODIS—NDIS Statistics, <http://www.fbi.gov/about-us/lab/codis/ndis-statistics> (as of Jan. 26, 2015).

⁷Innocence Project, DNA Exonerations Nationwide, <http://www.innoc>

confined simply to matters of identity. As information about variation within genes has given us a clearer understanding of the genetic component of behavior, disease, and other traits,⁸ criminal defendants have *increasingly sought* to use their own DNA to deny responsibility for and to mitigate their unlawful conduct. They hope to prove they have a genetic predisposition for violent, impulsive, or other antisocial behavior, and therefore are not culpable for their offenses.

More broadly, the impact of forensic DNA extends well beyond criminal law. For example, in personal injury cases, plaintiffs and defendants may offer genetic evidence on issues of causation and damages. In family law cases, judges may turn to genetic evidence to settle questions of paternity and to decide questions about parental rights.

In the future, advances in genetic science will inevitably raise new legal questions. As DNA testing offers us more effective disease prevention and treatment, medical professionals will have to consider new liability issues in choosing which of the hundreds of available genetic tests to give to a patient. Other legal and ethical issues will arise with breakthrough medical advances like gene therapy, stem cell research, and reproductive technologies. Advances in genetics also create risks of privacy invasion, discrimination in employment, and denial of health or life insurance. And the use of genetically modified organisms in agriculture and environmental science remains highly controversial. In all of these arenas, the potential benefits and challenges of advances in genetics may be greatly magnified as scientists move closer to creating synthetic life using man-made DNA.

Beyond genetic science, researchers are developing and refining sophisticated brain testing techniques that can shed light on both the truth of our statements and the motivations behind our actions. Researchers have already used brain imaging technology to identify simple abstract thoughts with 70% accuracy.⁹ Scans have likewise already “been used to identify brain signatures of disgust, drug cravings, unconscious racism, and suppressed sexual arousal, not to mention psychopathy and propensity to kill.”

enceproject.org/ContentDNA_Exonerations_Nationwide.php (as of Jan. 26, 2015).

⁸See generally, Broeckel and Schork, Identifying genes and genetic variation underlying human diseases and complex phenotypes via recombination mapping, 554 (Pt. 1) *J. Physiol.* 40 (Jan. 1, 2004).

⁹John Dylan-Haynes, Reading Hidden Intentions in the Human Brain (Feb. 20, 2007) 17 *Current Biology* 4, pages 323–328 in ScienceDirect, available online at <http://www.sciencedirect.com> {quick search: Reading Hidden Intentions in the Human Brain} (as of Jan. 23, 2015).

As technology evolves and advances, science and law will become more deeply entwined. Technological strides have forced people to change and expand their ways of thinking about concepts such as privacy, discrimination, and life itself. To accommodate these changes, our legal system must be prepared.¹⁰

§ 1:2 Matters covered

Chapter Two provides an overview of the DNA profile and explains how DNA testing technology identifies the individual. It begins with a history of forensic DNA testing. It summarizes DNA biology and the PCR-STR (polymerase chain reaction and short tandem repeat) testing process.

The portions of the DNA molecule used for forensic identification purposes, when considered collectively in the form of a profile, are particularly useful as an identifier because of the high degree of variability among individuals. This chapter explains how DNA's structure, its cellular organization, and its function (replication and cell division, transcription, and translation) serve the process of DNA testing for forensic DNA identification. This chapter likewise explains the analysis of short tandem repeats as the most widely used form of forensic DNA testing and details the advantages of this form of testing.

Chapter Three addresses the tasks and procedures involved in developing a DNA profile from crime scene to laboratory. It begins by discussing evidence collection. Any time evidence from a crime scene or physical examination is collected for DNA testing, it is intended to represent the biological material present at the time of a crime or assault. However, it ultimately is a representation of the biological material present at the time of collection. For this reason, careful sample collection, handling, packaging, and record keeping are essential to any crime scene investigation or examination.

The chapter explains the importance of reference samples. Reference samples are known samples taken from an individual to be used for comparison with profiles from evidence items. References may be saliva, blood, hair, or other bodily fluids or tissue. When evidence of a crime includes a perpetrator's DNA profile, a primary objective of investigators is to obtain a reference sample or samples from the suspect or other known persons for comparison purposes.

¹⁰ABA Symposium II: Public Understanding and Perceptions of the American Justice System, Panel Discussion: Changes in American Life, 62 Albany L. Rev. 1471, 1474 (1998–1999).