

DNA FINGERPRINTING

An Interactive Qualifying Project Report

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ABSTRACT

The aim of this Interactive Qualifying Project was to research the different DNA fingerprinting technologies and study their effect on today's society. Chapter one and two discussed the two main methods for obtaining DNA fingerprints, and the way DNA samples are collected and stored. Next, landmark DNA court cases were analyzed as examples for allowing complex technology into U.S. courts. The analysis showed that these cases provided a rigorous critique of the technology and enacted several legal precedences. To remind the reader of the affect that DNA fingerprinting has had on solving crimes, some sensational DNA court cases were described. Chapter four described the purpose of DNA databases, and discussed privacy rights that accompany each type. Lastly, the authors determined their own conclusions about this new technology including whose DNA should be entered into databases.

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PROJECT OBJECTIVES

The purpose of this Interactive Qualifying Project is to analyze the new technology of DNA fingerprinting, and determine its impact on society. Chapter one defines the different methods for performing fingerprints. Chapter two explains proper procedures for collecting and storing DNA samples. Chapter three analyzes several U.S. landmark DNA court cases, and reveals how courts rigorously examined the technology and enacted several standards for allowing complex technical information in courts. Chapter four describes the main types of DNA databases, and discusses their purpose and privacy rights associated with each. Lastly, based on the research done, the authors provide their own conclusions about new technology of DNA fingerprinting.

Chapter-1: DNA Fingerprinting, Description and Types

Abdulaziz Alamer

DNA fingerprinting, also known as DNA profiling, is a method used to distinguish individuals from each other. The distinction is based on comparing the types of differences (termed polymorphisms) in specific locations on DNAs. All human DNAs are about 99.8% identical to each other, which makes us human. The other 0.2% difference is what distinguishes each individual from the other, except for identical twins. These DNA patterns are taken from biological samples such as hair, blood, sperm, and others. This chapter will introduce DNA and its components, define some important terminology used in the DNA profiling procedures, summarize the two main DNA fingerprinting types, and list some DNA applications.

DNA fingerprinting was first discovered and used by Sir Alec John Jeffrey, a British professor of genetics at the University of Leicester (Jeffreys et al., 1985a). In 1985, this method was used for the first time in the case of a Ghanaian immigrant boy who needed evidence to verify his biological relationship with his mother to avoid being deported (Jeffreys et al., 1985b). To perform this profiling, Alec John Jeffrey compared sequences of DNA called mini-satellites, which contain repeating patterns (polymorphisms). One half of the pattern matched the mother and the other half matched the father, proving that the mother is biologically related to the boy, and he was not deported. In addition to paternity testing, DNA profiling also has applications for criminal forensics, identifying unknown human remains, and for molecular archaeology.

DNA Introduction

Deoxyribonucleic acid (DNA) is the basic building block of all living organisms. It contains the vital information about a species, in this case human, used to control cellular functions, behavior, and development. DNA is found in the nucleus inside cells. Red blood cells are the only non-nucleated cell, so they contain no nuclear DNA. All cells of the body contain DNA within their mitochondria, which are organelles used to produce energy. Both nuclear and mitochondrial DNA typing can be performed. DNA was discovered in 1868 by a Swiss biologist, Friedrich Meischer, who isolated DNA from white blood cells found in used bandages (Dahm, 2005). Meischer claimed that the nucleic acids, now called DNA, contain genetic information, but he was not able to prove it. In 1943, Oswald Avery and colleagues at Rockefeller University proved that Friedrich's claim was true, by showing that a previously seen “transforming substance” that is capable of giving *Pneumococcus* bacteria new properties is in fact composed of DNA (Avery et al., 1944; Arnold, 2009).

The DNA structure was elucidated in 1953 (Watson and Crick, 1953). DNA is formed from two strands (blue in the diagram) twisted together like a ladder, the double helix (**Figure 1.1**). Each strand's backbone is composed of a polymer containing phosphate residues and a deoxyribose sugar. Phosphate and deoxyribose are bound to a nitrogenous base to make the fundamental unit of DNA structure, the *nucleotide*. Four bases (colored rungs in the diagram) are present in DNA: adenine (A), cytosine (C), guanine (G), thymine (T). The bases point inward on the twisted strands, and pairs make hydrogen bonds with bases on the other strand. A pairs with T, and C pairs with G. The type and order of nucleotides found within a strand of DNA specifies its genetic traits. This genetic sequence dictates all the main properties of an organism.

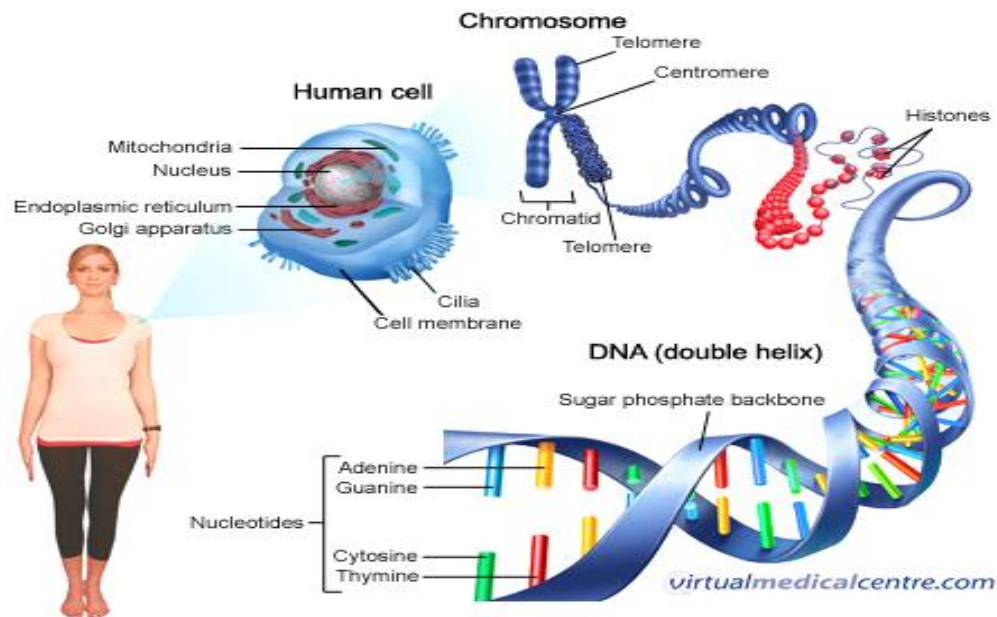


Figure 1.1: Diagram of a Cell, Nucleus, Chromosome, and DNA. Nuclei are found in every eukaryotic cell, except for red blood cells which contain no nuclei. The nucleus manages all cell activities, and separates DNA from the cytoplasm by a nuclear envelope. The nuclear envelope is a double membrane that isolates the DNA from the cytoplasm and protects it from damage and controls passing materials. (Virtual Medical Center, 2008)

Chromosomes

Chromosomes are “X”-shaped structures located in the nucleus of a cell at specific stages of the cell cycle. In Mitosis, two cells with identical genomes are created, the DNA condenses from a highly diffuse state (where it is replicated and expressed) into tightly wound structures called chromosomes (**Figure 1.1**, upper center, and **Figure 1.2**). Humans have 23 pairs of chromosomes (i.e. 46 chromosomes all together). The point where the chromosome pairs join is called centromere (**Figure 1.2**, red structure). The centromere contains DNA with highly repetitive DNA sequences, called mini-satellites. These mini-satellites are often used for DNA

fingerprinting analysis because they vary between individuals. They are also used for DNA genetic markers in linkage analysis and for population studies.

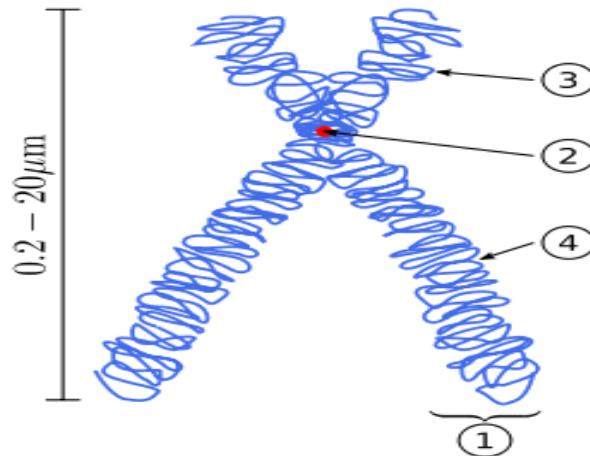


Figure 1.2: Diagram of a Chromosome. Shown is the main X shape of a typical human chromosome, containing two chromatids bound to each other in the center by the centromere (red on the diagram). DNA exists in this shape during mitosis when each chromatid separates and moves into each daughter cell. (Wikipedia, 2008)

DNA Loci and Repeating DNAs

A locus (singular of loci) is a specific physical location of a gene or other DNA sequence on a chromosome, like a genetic street address. Most of the DNA sequence of an individual does not vary much. As already stated, humans have about 99.8% of DNA sequences in common. These sequences cannot vary much or they become non-functional. However, within the 0.2% of our DNA that varies between individuals lies repeating DNA sequences. These repeating sequences often are not functional, so they can vary considerably without altering function. In DNA profiling, two types of repeating DNA sequences are used to perform identifications: variable number tandem repeats (VNTRs) and short tandem repeats (STRs). These repeat

sequences differ from each other in their overall length, the sequence of the repeat, and the length of the repeating unit itself. With VNTRs, the repeat sequence can be as short as two bases long, but is usually 8-10 bases long (Chantler, 2004). The overall VNTR length can extend up to forty repeats, and varies between individuals. In addition, one person can inherit a given number of repeats from the mother and a different number from the father (Chantler, 2004).

Short tandem repeats (STRs) as their name implies are shorter than VNTRs, with a repeating element of two to seven nucleotides (Butler and Reeder, 2004). Due to their short length, STRs are easily amplifiable by PCR (discussed below), while VNTRs are not. Because STRs are easy to analyze by PCR, the STR-PCR type of DNA analysis has become standard in the industry. **Figure 1.3** shows the standard 13 core STR loci currently analyzed for DNA profile entries into the FBI's CODIS database (discussed in Chapter-5).

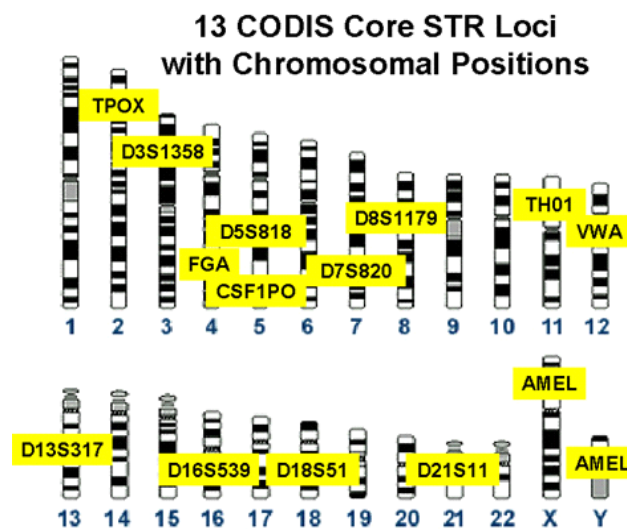


Figure 1.3: Diagram of the 13 Core Loci Analyzed in CODIS Entries. Note that the STRs (yellow in the diagram) are found on a variety of chromosomes, and have a variety of chromosomal positions. The STR analysis has become the standard in the industry. (National Institute of Standards, 2011).

DNA Analysis Techniques

Over the years, scientists have developed better ways to make the DNA profile more reliable and accurate. There are two main ways of analyzing DNA fingerprinting, which are amplifying and non-amplifying, and each has its own advantages and disadvantages.

Non-Amplifying DNA Fingerprints

The original method of preparing a DNA fingerprint was a restriction fragment length polymorphism (RFLP) protocol (**Figure 1.4**). Restriction fragments are segments of DNA cleaved from the main strain by cutting with restriction nucleases. These enzymes recognize specific DNA sequences and cut it. The fragments between restriction sites vary in length depending on the number of repeating sequences located on the fragment. RFLP analysis detects a particular fragment and determines its length (Davidson, 2006; Lerner et al., 2006). A restriction enzyme is used to cut the DNA into fragments, and then the DNA is separated by size by electrophoresis through a gel substance using electric current. The pattern of DNA fragments is then blotted to a membrane, and the membrane is hybridized to a labeled single stranded DNA probe which is complementary to a VNTR or RFLP of interest. Using a piece of x-ray film, the labeled fragment will be noticeable. RFLPs are not strongly affected by contamination, but they require a relatively large amount of DNA (RFLPs are typically too long to be amplified by PCR) and take a 1-2 weeks to perform. RFLPs require about 25 hairs, or a nickel-sized spot of fluid (Freeman, 2001).

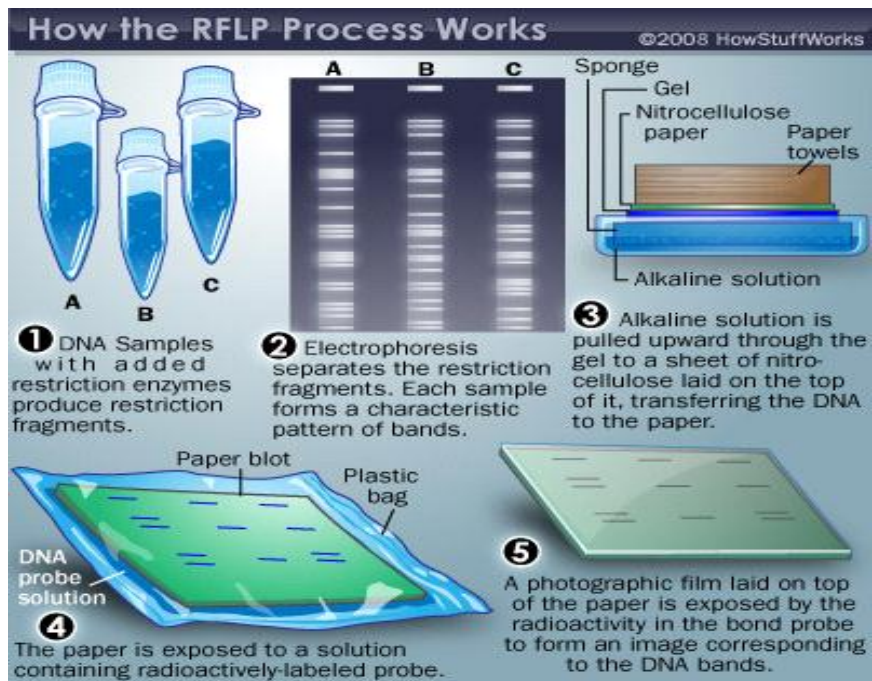


Figure 1.4: Diagram of the RFLP Process. The DNA sample is cut with restriction enzymes (upper left) then separated by size using electrophoresis (upper center). The DNA fragments are blotted to a membrane (upper right) and hybridized to a labeled probe (lower right) to visualize the RFLP or VNTR of interest (lower left). (Freeman, 2001)

To better understand how an RFLP analysis might look, **Figure 1.5** shows the results of a paternity case where an RFLP was used to determine whether Jill’s child Payle (third lane) is Jack’s son. Note that Payle inherited a 12.4 kb fragment (band on the left side) from both Jill and Jack, so that band could have come from either the mother or father, and cannot provide useful information here. However, Payle also has a band around 6 kb which came from neither Jack nor Jill, so it must have come from Payle’s biological father, who is likely not Jack.

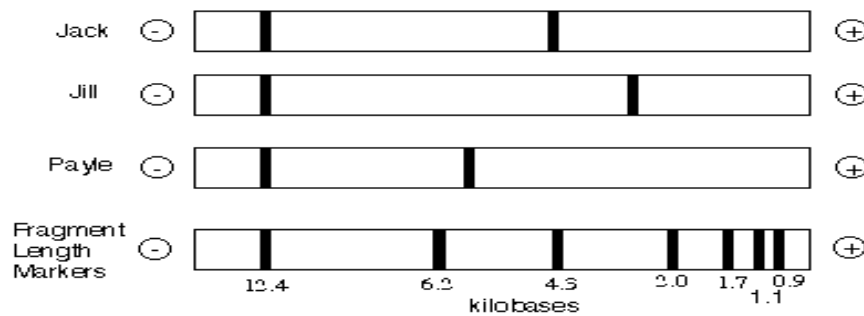


Figure 1.5: Example of an RFLP Analysis for a Paternity Case. Jill is the known mother of Payle, and the analysis was run to determine whether Jack is the father. Note that Payle has a band at about 6 kb that is not derived from either Jill or Jack, so it must have come from the real father of Payle, which is not Jack. (Davidson, 2006)

Amplifying-Type DNA Fingerprints

The second main method for DNA identification analysis is the PCR-STR technique. As discussed previously, STRs are short enough to be amplified by PCR. Because PCR is so sensitive and rapid, this technique is now used the most often. PCR was discovered in 1986 by Kary B. Mullis (Mullis et al., 1986) who later earned the 1993 Nobel Prize in Chemistry (Rice, 2006). A thermocycler is used to control the temperature of a reaction tube through a series of cycles to amplify the DNA (**Figure 1.6**). The reaction tube contains template DNA (for example isolated from crime scene evidence), nucleotides as DNA precursors, two primers (sense and anti-sense) that flank an STR of interest, and a special type of DNA polymerase (Taq polymerase) that can withstand multiple rounds of near boiling temperatures. The first step is denaturation, done at a temperature of 94°C which melts open the double-stranded DNA to make it single-stranded and capable of hybridizing to DNA primers. Next, the thermocycler cools the temperature to around 55°C to allow the primers to anneal to their complementary sites to the left and right of the STR. Then, the thermocycler raises the temperature to 72°C, the optimum

temperature for Taq polymerase to allow the synthesis of new DNA strands using the primers as start sites and the denatured DNA as template. This creates two new strands of DNA, each containing one old and one new fragment. The PCR cycle of denaturation, annealing, and synthesis is repeated by the thermocycler from 30-40 times to create millions of copies of the STR of interest. The PCR process can be completed within a few hours. After performing the PCR, the products of the reaction are analyzed by either gel electrophoresis or capillary electrophoresis to determine the length of the amplified STR, which provides a measure of the number of repeats at that location. PCR is sensitive enough to be able to amplify the DNA from a single cell. But its sensitivity can also be a problem when contamination is present, as the contaminated DNA will also become amplified.

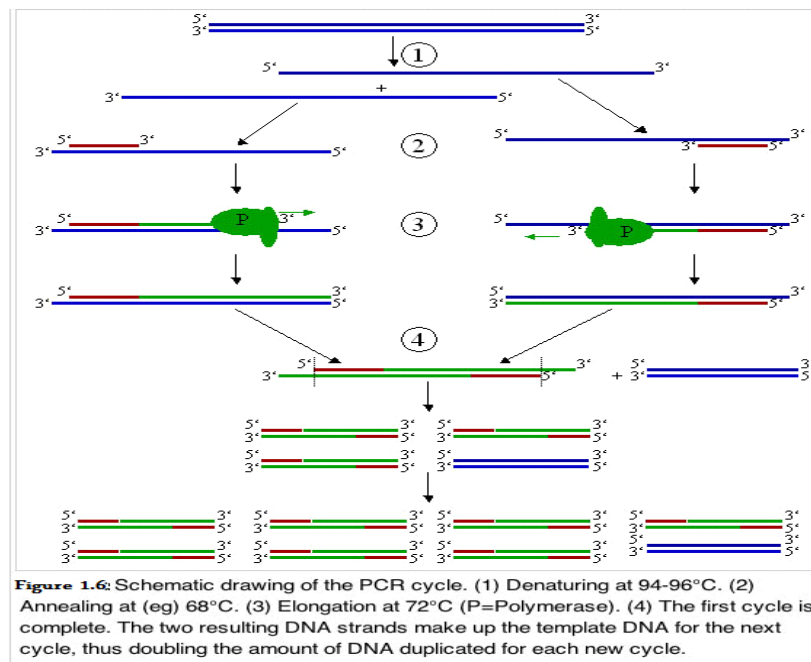


Figure 1.6: Diagram of the PCR Process. Shown are the key steps of PCR, including denaturation of the DNA template (step-1), annealing of the two primers upstream and downstream of the STR of interest (step-2), synthesis of new DNA by Taq polymerase (step-3), and the repetition of the cycle (step-4) to create millions of copies of the STR of interest. (Rice, 2012)

DNA Fingerprinting Applications

DNA fingerprinting has a variety of applications, from paternity testing, to criminal forensics, to identifying unknown human remains, to predicting genetic diseases, to aiding wildlife management, to identifying human historical migrations in archaeology. Paternity testing was the very first use of DNA fingerprinting technology (Jeffreys et al., 1985b), and this application is now the most frequent use of the technology, being used hundreds of thousands times per day (Butler, 2005). The test is not just used to prove paternity, but all types of familial relations (mother/daughter, mother/son, father/son, and father/daughter). **Figure 1.7** shows two examples of a paternity test. In the left panel (paternity exclusion), the pattern of the child does not contain any bands derived from the biological father, so the male is excluded as a father. While in the right panel (paternity inclusion), the upper band of the child is derived from the biological father and the lower band from the mother, so they are the real parents.

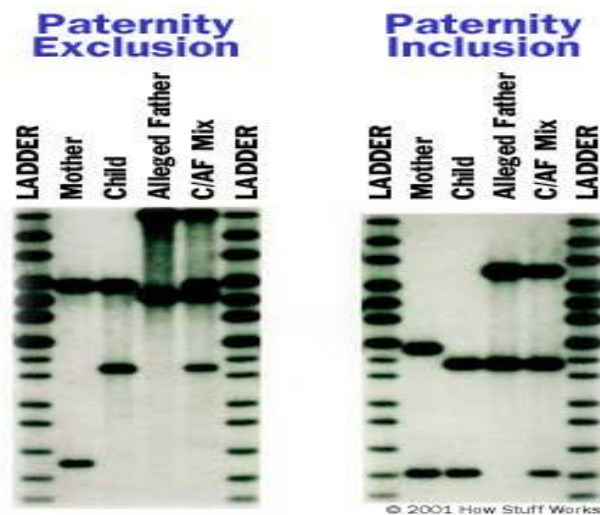


Figure 1.7: Example Data For A Paternity Test. The figure shows the paternity exclusion (left panel) and paternity inclusion (right panel). (Harris, 2010)

The second most frequent use of DNA testing is in criminal forensics. DNA profiles obtained from crime scene evidence are compared to profiles stored in the FBI's CODIS database that contains profiles from other crime scenes and from previous offenders. Criminal DNA databases allow law enforcement personnel to determine whether several crimes might be related to each other, or to determine who was present at a crime scene (if the profile is in the database). Since 1997, the US FBI mandates 13 core STR loci (previously shown in Figure 1.3) to be part of the information entered into CODIS, the United States national database. When no match is found between the crime scene sample and a database entry (exclusion), the suspect did not match a previous offender. However, if there is a match (inclusion), the question immediately arises as to under what probability would anyone's DNA make a similar match at that locus. This is determined by knowing the estimated allele frequency of that specific STR repeat in the general population or within his ethnic group. For example, if an 11 repeat STR at locus-1 is found in about 10% of the population, that 10% is multiplied by the frequencies of the other 12 loci also analyzed to obtain the overall likelihood of the match. When all 13 loci are analyzed, the probability of a match occurring randomly is one in several billion (Norrgard, 2008).

Molecular archeology is another form of DNA fingerprinting application. In this case, the DNAs from ancient samples are analyzed and compared to present-day DNA profiles. For example, DNA analysis was used to show that the 5,000 year old mummy known as Otzi the Tyrolean Iceman, likely originated in a small town in the northern Italian Alps (Handt et al., 1994). This hunter was likely killed by an arrow, and he fell into a crevice and remained covered with ice for 5,000 years until the ice thawed and he was found. DNA was taken from his frozen

stomach cells and indicated he originated from the north of Italy. Being totally covered with snow, and protected in the crevice from moving glaciers helped keep his body intact.

Chapter-1 Bibliography

Arnold P (2009) Oswald Avery and a Big Moment in the History of Genetics.
<<http://www.brighthub.com/science/genetics/articles/14588.aspx>>

Butler JM (2005) Forensic DNA Analysis.
<<http://www.dna.gov/basics/analysishistory/>>

Butler JM, Reeder DJ (2004) A Brief Introduction to STRs.
<<http://www.cstl.nist.gov/biotech/strbase/intro.htm>>

Chantler, Peter (2004) Variable Numbers of Tandem Repeats (VNTRs).
<http://www.rvc.ac.uk/review/DNA_1/4_VNTRs.cfm>

Dahm R (2005) Friedrich Miescher and the Discovery of DNA. *Developmental Biology*, 278: 274–288.

Davidson College (2006) Department of Biology. RFLP Method - Restriction Fragment Length Polymorphism. <<http://www.bio.davidson.edu/courses/genomics/method/RFLP.html>>

Freeman S (2001) Creating a DNA Profile.
<<http://science.howstuffworks.com/dna-profiling1.htm>>

Greg M (2010) Familial DNA Testing Scores A Win in Serial Killer Case. *Science*, 329: 262.
<<http://www.sciencemag.org/content/329/5989/262>>

Harris, William (2010) HowStuffWorks. DNA Analysis: Traditional Techniques.
<<http://science.howstuffworks.com/environmental/life/genetic/dna-evidence2.htm>>.

Jeffreys AJ, Wilson V, Thein SL (1985a) Individual-specific ‘fingerprints’ of human DNA. *Nature*, 316: 76-79.

Jeffreys AJ, Brookfield J, Semeonoff R (1985b) Positive identification of an immigration test-case using human DNA fingerprints. *Nature*, 317: 818-819.

Joan E (2012) National Human Genome Research Institute. Locus.
<<http://www.genome.gov/glossary/index.cfm?id=116&textonly=true>>

Lerner L, Lerner B, Gale C (2006) RFLP (Restriction Fragment Length Polymorphism). World of Forensic Science.

<<http://www.enotes.com/forensic-science/rflp-restriction-fragment-length-polymorphism>>

Mullis K, Faloona F, Scharf S, Saiki R, Horn G, Erlich H (1986) Specific enzymatic amplification of DNA *in vitro*: the polymerase chain reaction. *Cold Spring Harb Symp Quant Biol.* 51 Pt 1: 263-273.

National Institute of Standards (2011). <<http://www.cstl.nist.gov/strbase/fbicore.htm>>

Norrsgard K (2008) Forensics, DNA Fingerprinting, and CODIS.

<<http://www.nature.com/scitable/topicpage/forensics-dna-fingerprinting-and-codis-736>>

Phillips T (2012) DNA Fingerprinting

<<http://biotech.about.com/od/glossary/g/DNAfinger.htm>>

Vierstnetze, Andy (1999) Principle of the PCR.

<<http://users.ugent.be/~avierstr/principles/pcr.html>>

Rice, George (2012) Polymerase Chain Reaction (PCR).

<http://serc.carleton.edu/microbelife/research_methods/genomics/pcr.html>

Rosner D (2007) How does DNA Fingerprinting Work?

<<http://trendyscience.blogspot.com/2007/08/how-does-dna-fingerprinting-work.html>>

Sudbery P (1998) Human Molecular Genetics. Cell and Molecular Biology in Action series.

<http://grupo.us.es/gfml/dna/genetic_ingeniering/repetitive_dna.htm>

The Biology Project (2000) University of Arizona. STR Polymorphisms.

<http://www.biology.arizona.edu/human_bio/activities/blackett2/str_description.html>

The Economist Newspaper, CASE HISTORY: DNA's Detective Story.

<http://www.economist.com/node/2477036?story_id=2477036>

The National Health Museum (2012) Use of DNA in Identification.

<http://www.accessexcellence.org/RC/AB/BA/Use_of_DNA_Identification.php>

Virtual Medical Center (2008) Introduction to DNA.

<<http://www.virtualmedicalcentre.com/anatomy/dna-deoxyribonucleic-acid/37>>

VNTR (2011) U.S. National Library of Medicine. National Institutes of Health.

<<http://www.nlm.nih.gov/visibleproofs/education/dna/vntr.pdf>>

University of Leicester, What Sir Alec did next.

<<http://www2.le.ac.uk/departments/genetics/jeffreys/whatnext>>

Watson JD, and Crick FHC (1953) A Structure for Deoxyribose Nucleic Acid. *Nature*, 171: 737-738.

Wikipedia (2008) Chromosome <<http://en.wikipedia.org/wiki/File:Chromosome.svg>>

Chapter-2: DNA Forensics

Sabrina Sanchez

State-of-the-art analysis of well-chosen DNA loci by DNA fingerprinting technology is completely useless if the DNA sample itself has been contaminated, degraded, or compromised. The more scientists work with DNA, the more accurate their protocols have become for collecting, handling, shipping, and storing DNA evidence. The purpose of this chapter is to discuss these techniques which enable the DNA sample to be properly used for analysis.

Key Individuals at a Crime Scene

Present at a serious crime scene are usually police officers, a CSI unit, a district attorney, specialists, detectives, and sometimes a medical examiner. A warrant allowing the location to be searched may be required; if so it must be approved by a district attorney and judge. If not, the investigation may proceed. An example of a case requiring no warrant would be a crime committed in a victim's apartment in which only the victim resided. Police officers are typically the first to arrive at a crime scene. It is their duty to arrest the perpetrator if he/she is still present, and call for an ambulance if someone needs to be attended to. They are also responsible for properly protecting the evidence by securing the crime scene. The CSI unit documents details and collects physical evidence. The district attorney is present to determine if and what type of warrant may be needed to proceed in an investigation. The medical examiner will attempt to determine a probable cause of death, and other specialists are called in for expert analysis. Detectives will consult with CSI or will interview witnesses, and will then investigate the crime using all information obtained from the crime scene (Layton, 2004).

Securing a Crime Scene

To restrict outside access by unauthorized individuals, and prevent any destruction of evidence, a barrier or rope or tape must be set up. Once secured, a safe area can be designated for setting up equipment or taking breaks for professionals. The first officer at the scene usually does this. A now standard protocol in critical incident management refers to a three layer/perimeter that should be used to secure a crime scene (Byrd, 2000). The outer layer keeps onlookers and non-essential people away and safe, and is typically larger than the crime scene itself. The inner layer can include a command post and comfort area just outside of crime scene, and the last layer is the crime scene itself.

Collecting DNA Evidence

Once a crime scene has been secured, an investigator or evidence technician will come in as a forensic specialist to organize a step-by-step approach to use for recognizing, documenting, and recovering physical evidence (Byrd, 2000). At any crime scene it is important to take special precautions when dealing with evidence that may contain DNA. The fewer the individuals handle the evidence, the lower the risk of DNA contamination.

DNA in tissues is contained in the nucleus of the cell, or in smaller amounts in mitochondria. The types of tissue that can contain DNA are numerous. “Blood, semen, skin cells, tissue, organs, muscle, brain cells, bone, teeth, hair, saliva, mucus, perspiration, fingernails, urine, and feces are just a few” (President’s DNA Initiative, 1999). Tissues contain various amounts of DNA. When comparing tissues obtained from different locations in the body, scientists have done experiments to quantify the amount of DNA that can be found in typical samples. The larger the sample size, the larger amount of DNA that can be extracted from that sample. **Figure-2.1** shows the typical amount of DNA that can be present in some common

evidence samples and their PCR success rates (Kaye and Sensabaugh, 2000). Note that semen is especially rich in DNA and has a high PCR success rate. Also note that skin cells left on a door knob or the root end of a shed hair have low PCR success rates.

Type of Sample	DNA Content	PCR Success Rate
Blood	20,000–40,000 ng/mL	
stain 1 cm x 1 cm	ca. 200 ng	> 95%
stain 1 mm x 1 mm	ca. 2 ng	
Semen	150,000–300,000 ng/mL	
on post-coital vaginal swab	0–3000 ng	>95%
Saliva	1000–10,000 ng/mL	
on a cigarette butt	0–25ng	50–70%
Hair		
root end of pulled hair	1–750 ng	>90%
root end of shed hair	1–12 ng	<20%
hair shaft	0.001–0.040 ng/cm	
Urine	1–20 ng/mL	
Skin cells		
from socks, gloves, or clothing repeatedly used		30–60%
from handled objects (e.g., a doorknob)		<20%

ng = nanogram, or 1/1,000,000,000th of a gram; mL = milliliter; cm = centimeter; mm = millimeter

Figure-2.1: Typical DNA Content of Various Tissues and their PCR Success Rates.
(Kaye and Sensabaugh, 2000).

Other tissues not shown in the figure have the ability to preserve DNA for long periods of time, although the yield may be low. These tissues include bone, teeth, and fossils. At old crime scenes, these latter types of evidence are better to use for DNA extraction and the DNA is better preserved (Tibor et al., 2000). As an example of extracting DNA from ancient samples, Michael Balter discussed the discovery of a 40,000 year old Siberian ancient finger found to contain an attainable mitochondrial DNA sequence (Balter, 2010).

Individuals collecting DNA samples must be cautious about preventing degradation and contamination. Contamination occurs when DNA from another source is mixed with the DNA relevant to the crime. This can happen by many ways, including sneezing, coughing, or if someone touches their face and then touches the evidence. As discussed in Chapter-1, polymerase chain reaction (PCR) is a procedure used to make copies of DNA *in vitro*. PCR is extremely sensitive, and can even amplify DNA from a single cell. So if contamination occurs, PCR can become problematic because it will not distinguish between a suspect's DNA and DNA from another source. Anything can be contaminated, whether the sample is big, small, or delicate, so preventing contamination must be taken into serious consideration. Using disposable gloves and changing them often can be very helpful when handling evidence. Using disposable instruments and thoroughly cleaning other non-disposable instruments will also help prevent contamination. It is also crucial to avoid touching any area where DNA may exist.

Any sample that is wet should be air-dried, and evidence should be put in paper bags or envelopes. Plastic bags or staples should be avoided to prevent contamination or degradation, as this would prevent moisture from evaporating and could lead to DNA degradation (President's DNA Initiative, 1999). DNA degradation refers to the process of the nucleotide sequence breaking down and being unavailable for analysis. This can occur after the cell containing the DNA is broken open, and the DNA is exposed to a harmful environment. Repeated freezing and thawing the sample or excessive heat can physically degrade or damage the DNA. Direct sunlight and warm conditions may also damage DNA, so officers try to keep the majority of materials at room temperature (President's DNA Initiative 1999).

Extracting DNA from Crime Scene Evidence

Before DNA can be analyzed by STR-PCR or RFLP techniques, it must first be extracted from its biological source material. Typical extraction methods include cell lysis, DNA chemical purification, and DNA precipitation. Some tissues require pre-processing, such as blood requiring the removal of red blood cells (that contain no nuclei) and concentrating white blood cells (that contain nuclei). According to Life Technology Protocols, one method that can be used involves allowing the blood sample to first clot at room temperature undisturbed for 15-30 minutes (Life Technologies, 2007). The clot is then removed by refrigerated centrifugation at 1,000-2,000 x g for 10 minutes. The supernatant that results is the serum. The serum is then transferred into a clean polypropylene tube and kept at 2-8°C. Precipitation of the DNA can be facilitated by the addition of a salt (like sodium chloride). Salt, “is added as the negatively charged DNA molecules bind Na^+ and this encourages the free strands of DNA to aggregate. When the sample is vortexed in the presence of phenol-chloroform, and then centrifuged the serum proteins will remain in the organic phase (or at the interface) and can be drawn off carefully. The DNA will then be located in the aqueous phase. SDS is often included to help remove lipid membranes. To avoid degradation, protease is sometimes avoided, but many labs still use this enzyme when extracting DNA. The DNA in the aqueous phase is then precipitated by adding cold ethanol or isopropanol, and then centrifuging. The DNA is not soluble in ethanol and will precipitate. The DNA pellet can be washed with ethanol to remove any salt that may have been previously used. The resulting washed DNA pellet can be dried, and then resuspended in a buffer such as Tris-EDTA (TE) (Rice, 2012).

When it comes to evidence liquids, many laboratories use kits that do most of the work. All of the kits use enzymes, specific lysis buffer/extraction buffers, and chemicals to obtain DNA

from cells. They are helpful in preventing contamination, but highly concentrated enzymatic solutions can damage DNA. The Hirt DNA Extraction Kit separates high molecular weight nuclear DNA and low molecular weight mitochondrial DNA. Another kit often used to extract DNA from saliva is the Oragene Kit (Schwartz and Pilgrim, 2006). In this procedure, saliva is collected in a container and transferred to a microcentrifuge tube containing Oragene resin. The suspension is mixed by vortexing for a few seconds, and the sample is incubated on ice for ten minutes. The suspension is centrifuged for 5 minutes at room temperature at 13,000 rpm to pellet the resin bound to cell debris and proteins. The resulting clear supernatant containing DNA is transferred to a fresh microcentrifuge tube, and then 95-100% of room temperature ethanol is added. Gentle inversion and incubation for 10 minutes precipitates the DNA. The sample is centrifuged at room temperature for 2 minutes at 13,000 rpm to pellet the DNA. The supernatant is discarded, avoiding the DNA pellet. The DNA pellet is dried, and then dissolved in TE buffer.

With respect to extracting DNA from hard surfaces like bone, the bone can be pulverized to make a powder. The outer surface of the bone is first bleached and rinsed with distilled water. Sometimes the bone outer surface is sanded by a dental drill, then irradiated with ultraviolet (UV) light to help eliminate DNA contamination and expose soft bone tissue for extraction. The cleaned bone is then pulverized by freezing it in liquid nitrogen then grinding it (SPEX CertiPrep Group, 2012). The bone powder containing DNA is then put into an extraction buffer, and then DNA is isolated by standard procedures. Sometimes a non-pulverizing procedure is used that induces cell lysis by using cycles of high and low pressures in a Barocycler instrument (Pressure BioSciences, 2012).

Another method of DNA extraction uses zinc. Zinc has been found to bind DNA and produce aggregates so large that centrifugation is not required to isolate it. After a few hours

incubation with $ZnCl_2$, gravity itself allows the DNA to sediment. The $ZnCl_2$ concentration needed depends on the sample's volume and pH. Less zinc is required for larger samples or alkaline samples. In some cases, phosphates are also added to facilitate DNA aggregation. The DNA pellet is resuspended in TE buffer (Eduard and Jaroslav, 1997).

When isolating DNA from hair, it is better if the hair has been pulled from the scalp because it is more likely to contain the hair follicle, the white bulb at the end of a hair strand. The hair can be pulled by forceps, then placed into a vial filled halfway with silica desiccant. A few hours later, if the desiccant is blue the sample is dry and in good shape, so can be stored at room temperature. This silica desiccant method can also be used with feces after the feces are left to air dry. Another potential source of DNA is urine, which can be placed in a polypropylene screw-cap vial and keep refrigerated without adding any extra liquids or chemicals (Schwartz and Pilgrim, 2006).

DNA Transport and Storage

The evidence collected from a crime scene should be collected into clean and unused containers, bags, or envelopes. Moist or wet biological evidence should be collected into clean, unused plastic containers. If biological material is sealed in plastic, it must be transported to a lab in less than 2 hours to prevent microorganisms from growing, which can lead to DNA degradation (Schiro, 2001). Once at the lab, wet evidence is left to dry, and may be repackaged into new, clean containers. Items should be stored separately to avoid cross contamination, and should be securely closed during transportation. Each container should have the initials of the person who collected the sample, the date and time it was collected, a complete description of its original location, and the name and file number of the investigating agency. Before transporting the sample, the investigator should check the evidence to keep track of what was found and to

make sure things all items were collected correctly. With proper storage, DNA can even be extracted from old tissues. But if the samples are improperly stored, naturally occurring enzymes found in animal cells can degrade DNA, affecting any future analysis. Some of these enzymes need an aqueous environment to function correctly, so drying or freezing the sample can be performed to help prevent degradation. Repeatedly freezing and thawing samples or excessive heat can physically degrade the DNA (Schwartz and Pilgrim, 2006).

With respect to storing solid or thick tissues, silica desiccants can be used, but other methods include freezing the sample or storing it in 95-100% ethanol. Solid tissues can be placed by forceps in a previously cleaned polypropylene screw cap vial, then the vial can be halfway filled with silica indicator or ethanol. If ethanol is chosen, almost any temperature is acceptable, from room temperature to -80°C. Some samples can be transported in a cooler with ice packs or dry ice. When a frozen tissue is used and sampled, the remainder should be returned to the freezer to avoid repeated freezing and thawing. Blood can be drawn into a labeled purple top EDTA tube or red top blood collection vial, and then frozen. Blood can also be collected onto Whatman FTA Micro-cards, then air dried and placed in bags with desiccant. It is ideal to collect as much blood as possible in case repeated analyses are required (Schwartz and Pilgrim, 2006).

Evidence “Chain of Custody”

An evidence chain of custody is a chronologically ordered record used to control access to physical possession of crime scene evidence. Before allowing evidence into court, the trial judge must be convinced that the evidence was properly collected and has not been tampered with by unauthorized individuals. Evidence custodians should document and keep track of any transfer of original evidence using an Evidence Tag Log and Form-13437 (the National Forensic

Laboratory Request Form) that documents all authorized agents and transfers. These forms will have a note section containing specific information about the evidence itself. The authorized individual is required to record no more than one day's worth of access entry. Each form is attached the outside of an evidence container. This is done to establish a chain of custody from the initial evidence obtainment throughout the entire judicial proceedings. If any evidence requires additional examination, an internal chain of custody must be created by that forensic examiner. Maintaining the chain of custody is crucial if laboratory analysis discovers DNA evidence is contaminated to help rule out authorized individuals. In all cases, the fewer the people handle the evidence the lower the risk of contamination. In addition to the chain of custody, an Evidence Access Control Log Form is sometimes used to record controlled area access where evidence is stored (IRS, 2012).

Chapter-2 Bibliography

Balter, Michael (2010) Ancient DNA From Siberian Fingers a Possible New Human Lineage. *Science* 327: 1566-1567.

Byrd, Mike (2000) Duty Description for the Crime Scene Investigator.
<http://www.crime-scene-investigator.net/dutydescription.html>

Eduard K, Jaroslav K (1997) DNA Extraction by Zinc.
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC146667/pdf/251870.pdf>

IRS.gov (2012) Chapter-4: Investigative Techniques.
http://www.irs.gov/irm/part9/irm_09-004-009.html

Kaye DH, Sensabaugh Jr GF (2000) Reference Guide on DNA Evidence.
[http://www.fjc.gov/public/pdf.nsf/lookup/sciman00.pdf/\\$file/sciman00.pdf](http://www.fjc.gov/public/pdf.nsf/lookup/sciman00.pdf/$file/sciman00.pdf)

Layton, Julia (2004) How Crime Scene Investigation Works. How Stuff Works.
<http://science.howstuffworks.com/csi.htm>

Life Technologies (2007) Plasma and Serum Preparation.
<http://www.invitrogen.com/site/us/en/home/References/protocols/cell-and-tissue-analysis/elisa-protocol/ELISA-Sample-Preparation-Protocols/Plasma-and-Serum-Preparation.html>

President's DNA Initiative (1999) What Every Law Enforcement Officer Should Know about DNA Evidence (1999) National Institute of Justice.
<http://www.dna.gov/audiences/investigators/know>

PressureBiosciences.com (2012) DNA Extraction from Bone Fragments without Pulverization.
http://www.pressurebiosciences.com/downloads/app_notes/AN-10003.v1.pdf

Rice, George (2006) DNA Extraction.
http://serc.carleton.edu/microbelife/research_methods/genomics/dnaext.html-George rice 2012

Schiro, George (2001) Collection and Preservation of Blood Evidence from Crime Scenes.
<http://www.crime-scene-investigator.net/blood.html>

Schwartz MK, and Pilgrim K (2006) Protocols for Collecting and Storing DNA Samples.
http://www.rmrs.nau.edu/wildlife/genetics/pdfs/SchwartzPilgrimDNA_tissue_storage_12_12_06.pdf

SPEX CertiPrep Group (2012) <http://www.spexcertiprep.com/>

Tibor K, Csanád ZB, Antónia M and Raskó I (2000) A Simple and Efficient Method for PCR Amplifiable DNA Extraction from Ancient Bones.
<http://www.ncbi.nlm.nih.gov/pmc/articles/PMC102752/>

Chapter-3: Landmark DNA Court Cases

Sabrina Sanchez

Over the years, technology has become more and more advanced. Although some technologies may appear to benefit mankind in an obvious way, their acceptance in the courtroom has not always been straightforward. In the U.S. legal system, landmark trials set legal precedence and standards for accepting complex information in court. For DNA fingerprinting, the early standards were based on non-DNA trials, but later, specific DNA trials were used to thoroughly critique the technology and further refine the standards. This chapter highlights some important landmark cases that have contributed to the acceptance of DNA fingerprinting evidence in courts.

Frye v. United States, 1923

In Washington DC, on November 25th, 1920, physician Dr. Robert W. Brown was murdered in his office. The murder was witnessed by one of Dr. Brown's co-workers who chased an African-American man out of the office. The police had no leads on the suspect, but months later James Alphonzo Frye was arrested for armed robbery, and eventually confessed to the robbery and the murder. Frye based his defense on a then new systolic blood pressure deception test (lie detector) which a defense expert witness claimed the defendant took and passed. In the 1920's, the lie detector test measured a suspect's blood pressure during a series of questions and answers, and was said to show when a subject was lying. However, the District of Columbia court judge ruled that the expert testimony was inadmissible because the technology lacked a *general acceptance* in the scientific community. After 4 days of trial, Frye was given a

life sentence for second-degree murder (the lie detector debate reduced the debate from first degree murder) (*Frye v US*, 1923).

Because of the refusal to accept his “expert witness”, Frye appealed his case to the Court of Appeals in the District of Columbia. The three-judge appellate panel ruled for the United States in a “short opinion that became one of the most notorious opinions written by a federal appeals court.” In the opinion, the court explained how the expert testimony would not be allowed in this case because the technology was not *generally accepted* in the scientific community, and had not been extensively reviewed by several experts. This trial set a precedent that later became known as the Frye Standard, that any new technology accepted in court must be generally scientifically accepted (*Frye v. United States – Significance* 2010). Currently, many states continue to rely on the Frye Standard when admitting evidence into court. Those states that argue against using the Frye Standard believe it is hard to prove a technique’s general acceptance, which favors the defense when using new technology. Instead, these states usually use the later 1975 Federal Rules of Evidence standard of stressing a technique’s helpfulness, reliability, and relevance (*Genelex Corporation*, 2012).

Sarbah v. Home Office, 1985

In 1983, Andrew Sarbah arrived in London’s Heathrow airport after visiting his mother’s estranged husband in Ghana. But when Andrew arrived, he was held at Heathrow by immigration officials claiming his passport had been forged. His mother, Christiana Sarbah, and Andrew struggled to prove to the England Home Office which governs immigration that they were mother and son. It took an intervention from Parliament Member Martin Stevens to allow Andrew to stay at his mother’s house in London (*Tripod.com*, 2012).

Workers at the Hammersmith Law Centre obtained various forms of evidence, photographs, and statements from family members to support the Sarbah family claim. Blood tests were performed, but did not prove that Andrew was her son instead of only a relative. At an immigration hearing, the family's evidence was rejected. However, Andrew's deportation was delayed due the family's appeal. The family had seen a newspaper article that introduced Alec Jeffreys at Leicester University and his then new DNA fingerprinting technology that could prove genetic relations (Jeffreys et al., 1985). Centre workers believed the situation was an ideal case to test Jeffrey's new technology.

Jeffreys extracted blood samples from Christiana, Andrew, an unrelated individual, and Christiana's three undisputed children. DNA fingerprints were made using the RFLP-type test discussed in Chapter-1. The probe was a multi-locus probe complementary to the tandem repeat sequences found in hyper-variable mini-satellites, and produced a complex pattern of bands. Although the father's DNA was not available, Jefferys reconstructed his fingerprints by identifying bands that were present in the other three children but not in the mother's profile. When the father's bands were compared to Andrew's and his mother's bands, the results showed that the likelihood of Andrew not being the son was "one in a trillion". The Home office was forced to accept the DNA evidence and went further to announce "that it would not contest future immigration cases if similar DNA evidence were available" (Tripod.com, 2012). Andrew was allowed to stay in in England with his family, and the case became the world's first court case to use DNA fingerprinting.

Andrews v. State of Florida, 1988

Rapist, Tommy Lee Andrews, was the first person in the U.S. to be found guilty of a crime using DNA evidence (Andrews v. State of Florida 1988). In 1986, Andrews was thought to

be a suspect in more than 20 assaults in Orlando, Florida, but a 1987 case proved his downfall. On February 21, 1987, the victim was awakened and threatened by a strong black male. The victim fought back, and because of her efforts she was cut various places by the intruder, followed by forced vaginal intercourse. After the attack, a physical examination of the victim “revealed the presence of semen in the victim's vagina” (Orfinger et al., 1988). Both victim and her attacker were blood type O; the attacker was a “secretor” where his blood type was secreted into body fluids, but that could not prove Andrews was the attacker. The only conclusion that could be made was that the defendant was part of a large population with blood type O; other evidence would be needed to identify the criminal.

On November 3, 1987, a scientist from Lifecodes Corporation and an MIT biologist performed DNA fingerprint analysis on Andrew’s DNA and compared it to the profile of the DNA from the rape victim. The profiles were found to match, and on November 6, 1987, a guilty verdict was announced. Andrews was sentenced to 22 years in prison. This case attracted much attention by the press, and initially created a “media blitz favorable to the new technology” (Genelex Corporation, 1995). But the positive media blitz only lasted one year until the technology was thoroughly critiqued in the Castro case.

People v. Castro, 1989

One of the most significant U.S. court cases to examine DNA fingerprinting technology is People vs. Castro, 1989. In this case, defendant Joseph Castro was charged with the murder of his neighbor, a 20 year-old pregnant woman Vilma Ponce and her two-year old daughter. Scientists from Lifecodes Corporation analyzed bloodstains found on the defendant’s wrist watch, and determined the blood belonged to Vilma Ponce (People v. Castro, 1989). The

prosecution wanted the DNA evidence entered into trial, but the defense argued the data was flawed. Several expert witnesses battled back and forth in court critiquing DNA technology.

In August 1989, Judge Gerald Sheindlin established a new Castro standard that mandated pre-trial hearings to determine the admissibility of DNA evidence before a jury sees it, and created a three-pronged test that determined whether DNA evidence should be admitted in court. The first prong questioned (as with the Frye Standard) whether there was a *generally accepted* scientific theory that stated DNA testing could be *reliable*. The second prong questioned whether techniques exist that are capable of producing reliable DNA results. The third prong questioned whether the testing lab performed accepted DNA tests in this particular trial (People v. Castro, 1989). The judge ruled that prongs-1 and 2 were satisfied, but that prong-3 was not, and the test results performed by Lifecodes lacked key controls, so he decided to not admit Castro's DNA evidence. However, the judge's ruling proved to be moot, as the case never went to trial. Castro confessed to the murders in 1989.

Following this landmark case, it was determined that DNA technology needed to be more standardized, so the TWGDAM (Technical Working Group on DNA Analysis Methods) was created to help establish universal procedures for DNA testing. The case is now known as one of "the most rigorous testing of DNA evidence ever performed" (Crime Laboratory, 1991), and recognized that even the best testing labs needed controls for their experiments. Using an extensive pretrial hearing to determine how the tests were performed in each trial case, the court would decide whether the evidence was reliable enough to use in that specific trial.

People v. Miles, 1991

In the state of Illinois, Reggie E. Miles was accused of two counts each of home invasion and residual burglary, five counts of aggravated criminal sexual assault, and one count each of criminal sexual assault, aggravated unlawful restraint, and armed robbery (People v. Miles, 1991). DNA evidence was obtained from the sexual assaults, and testing was performed by Cellmark Labs on Miles' DNA and the DNA collected from the crimes. The profiles matched, but the defense wanted the DNA evidence excluded on the basis that Cellmark Labs was previously shown to have problems with their DNA methodology (Castro prong-3). However, the judge ruled that Cellmark used protocols approved by the TWGDAM standards, so he allowed the DNA evidence at trial. In June 1990, the jury convicted Miles on all accounts, and in July 1990 the judge sentenced Miles to 120 years in prison.

Miles appealed the verdict in July 1990, on the basis that the judge allowed unreliable DNA evidence and allowed controversial expert testimony about statistical likelihoods. In August of 1991, the Fourth District Appellate Court of Illinois affirmed the trial court's ruling (with a corrected sentencing judgment) (Appellate Court of Illinois, 2011). The original guilty verdict was also upheld in subsequent appeals in 2001, 2005, 2008, and 2011 (Appellate Court of Illinois, 2011).

The case of Miles v. Illinois evaluated and strongly supported the new TWGDAM guidelines for DNA testing, and shifted the tide in favor of DNA testing in general (People v Miles, 1991).

People v Paul Eugene Robinson, 2000

On August 25, 1994, a 24-year-old woman named Deborah L, awakened in her Sacramento apartment to a man she had never seen before who wore gloves and held a kitchen knife. She started to scream, but he threatened her and proceeded to rape her. When he was done, he escaped, and the police were left with no leads (People v. Paul Eugene Robinson, 2000). A rape kit was prepared for the victim, but the DNA was not immediately analyzed.

Six years later, in August of 2000, as the statute of limitations was about to expire for the rape, Detective Peter Willover contacted Anne Marie Schubert, a sexual assault prosecutor and DNA expert who had information about keeping old cases open. Det. Willover determined that Schubert had filed a “John Doe Warrant” against a criminal she was following that identified her subject by DNA profile but not by name, address, or physical characteristics, as would normally be the case with a traditional warrant. Detective Willover thought this John Doe Warrant approach might work for his 1994 open rape case to stop the statute of limitations clock, so he had DNA analysis performed on 13 loci on the semen DNA from the rape kit, and filed a John Doe warrant based on that DNA profile (Findlaw.com, 2012). The John Doe profile was found to match that of 31-year old Paul Eugene Robinson, whose DNA profile had previously been entered into CODIS on November of 1998 for parole violation on an unrelated spousal battery crime. For verification purposes, Robinson’s DNA profile was determined from a fresh blood sample, and the profile again was found to match the 1994 rape (People v. Robinson, 2010).

At trial, the John Doe Warrant was declared valid for stopping the statute of limitations clock, the DNA evidence was allowed, and Robinson was convicted of two counts each of forcible oral copulation and rape, and one count of penetration with a foreign object (People v. Paul Eugene Robinson, 2000). Robinson was sentenced to 65 years in state prison by Judge

Peter Mering. Robinson appealed his case, but the original guilty verdict was upheld by the Third District Court of Appeal (Ofgang, 2010; Findlaw.com, 2012).

The Robinson trial became the first conviction of someone using only DNA evidence, and opened the door for using John Doe Warrants to stop the statute of limitations for other sex crimes when only a DNA profile of a suspect is known (People v Robinson, 2010; Ofgang, 2010).

Chapter-3 Conclusion

Although it was not always the case, DNA fingerprinting is now well known and widely used in U.S. courts. Several landmark trials have helped critique and standardize the technology, and showed how to use pre-trial hearings to validate DNA evidence for each trial. The new standards have played a major role in helping incorporate DNA fingerprinting evidence in U.S. trials, helping to convict the guilty or exonerate the innocent. Each case brought a greater awareness of the power of the technology when it is properly performed. These landmark cases were crucial for increasing the admissibility of DNA evidence in courts.

Chapter-3 Bibliography

Andrews v. State of Florida (1988) District Court of Appeal of Florida, Fifth District, 533, Southern Series, 2d, pp. 841.

Appellate Court of Illinois (2011) The People of the State of Illinois v. Reggie E. Miles. http://www.state.il.us/court/R23_Orders/AppellateCourt/2011/4thDistrict/October/4100634_R23.pdf

Crime Laboratory Digest (1991) TWGDAM Guidelines for a Quality Assurance Program for DNA Analysis, Vol. 22, pages 21-43. <http://www.cstl.nist.gov/div831/strbase/twgdam1.htm>

Findlaw.com (2012) People v. Robinson, January 25, 2010. <http://caselaw.findlaw.com/ca-supreme-court/1498121.html>

Frye v. United States (1923) Supreme Court of Appeals of District of Columbia, 293 F. 1013; 1923 U.S. App. LEXIS 1712; 54 App. D.C. 46; 34. A.L.R. 145, December 3, 1923, Decided. http://www.patrickcrusade.com/FRYE_V_US.html

Frye v. United States – Significance (2010) JRank. <http://law.jrank.org/pages/23627/Frye-v-United-States-Significance.html>

Genelex Corporation (2012) DNA in the Courtroom: Chapter 1. <http://genelex.com/dna-learning/book-1-science-law.html>

Jeffreys AJ, Brookfield JF, Semeonoff R (1985) Positive identification of an immigration test-case using human DNA fingerprints. *Nature*, 317: 818-819.

Orfinger J, Dauksch D, Daniel J (1988) TOMMIE LEE ANDREWS v. STATE FLORIDA (10/20/88) COURT OF APPEAL OF FLORIDA, FIFTH DISTRICT. http://fl.findacase.com/research/wfrmDocViewer.aspx/xq/fac.19881020_0045464.FL.htm/qx

Ofgang, Kenneth (2010) Supreme Court: DNA-Based Prosecution of ‘Doe’ Was Timely. *Metropolitan News*. January 26, 2010. <http://www.metnews.com/articles/2010/robi012610.htm>

People v. Castro (1989) Supreme Court, Bronx County, New York, Criminal Term, Part 28, 545 New York State Series, 2d, page 985. <http://jolt.law.harvard.edu/articles/pdf/v03/03HarvJLTech223.pdf>

People v. Miles (1991) Appellate Court of Illinois, Fourth District, Vol. 577 Northeastern Series, 2d ed., pp. 477.

People v. Paul Eugene Robinson (2000) Court of Appeal of the State of California, Third Appellate District (Sacramento), C044703 (Super. Ct. No. 00F06871). http://www.denverda.org/DNA_Documents/Robinson-CA.pdf

People v. Robinson (2010) The PEOPLE, Plaintiff and Respondent, v. Paul Eugene ROBINSON, Defendant and Appellant. <http://caselaw.findlaw.com/ca-supreme-court/1498121.html>

Tripod.com (2012) DNA Fingerprinting, The First DNA Test. <http://dnafingerprinting19.tripod.com/id14.html>

Chapter-4: DNA Database Ethics

Abdulaziz Alamer

As mentioned in Chapter-1, DNA databases are an assembly of DNA profiles on computers for the purpose of DNA analysis. DNA databases such as the FBI's Combined DNA Index System (CODIS) are used to help solve crimes (The FBI's...2000; Adams, 2002). While databases like the Icelandic Genetic Database can be used to try to find genes that map to specific genetic diseases (Hlodén, 2000). The development of databases containing DNA profiles has significantly improved our ability to solve crimes, and made it easier to find criminals by matching the DNA found at crime scenes to previous offender profiles in the database. But only previous offender profiles are in the database. The ability to solve crimes would be further enhanced if everyone's profile was in the database. But privacy groups worry about the potential leak of genetic information. So, whose DNA profiles should be included in criminal databases? This chapter will discuss both types of DNA databases, and investigate some of the privacy issues associated with each type.

The CODIS Database

The FBI's Combined DNA Index System (CODIS) actually consists of software that runs and maintains several linked criminal forensic DNA databases. CODIS is one of the largest DNA databases in the world, and it first appeared in 1990 linking twelve states. As of March 2002, CODIS has assisted in over 4,719 investigations in 32 states (Adams, 2002). As of 2010, CODIS contained over 8,646,417 offender profiles, 328,067 forensic profiles, and aided 119,764 investigations (CODIS Brochure, 2011) (**Table 4.1**). Sometimes a DNA profile from a crime scene is searched against the Convicted Offender Index. Other times, the entries in the Forensic

Index (containing profiles from various crime scenes) will be searched against themselves to identify linked crimes. The public is often concerned with the potential linkage of information from DNA databases, so with respect to CODIS it is important to note that no medical information exists within this database. As previously mentioned, CODIS entries consist only of the number of repeats for each of 13 core loci carefully chosen to not contain any genetic medical information. Therefore, medical information cannot be hacked from CODIS.

	Offender Profiles	Forensic Profiles	Investigations Aided	Forensic Hits	National Offender Hits	State Offender Hits	Total Offender Hits
2000	460,365	22,484	1,573	507	26	705	731
2001	750,929	27,897	3,635	1,031	167	2,204	2,371
2002	1,247,163	46,177	6,670	1,832	638	4,394	5,032
2003	1,493,536	70,931	11,220	3,004	1,151	7,118	8,269
2004	2,038,514	93,956	20,788	5,147	1,864	11,991	13,855
2005	2,826,505	126,315	30,455	7,071	2,855	18,664	21,519
2006	3,977,433	160,582	43,156	9,529	4,276	28,163	32,439
2007	5,372,773	203,401	62,059	11,750	6,508	43,305	49,813
2008	6,539,919	248,943	80,948	14,122	8,479	58,304	66,783
2009	7,688,286	298,369	101,766	17,636	10,969	75,186	86,155
2010*	8,646,417	328,067	119,764	19,940	12,791	89,598	102,389

* Through July 2010

Table 4.1: CODIS DNA Database Statistics. This table shows the number of offender and forensic profiles in the CODIS database, the number of investigations it has aided, and the number of “hits” that occurred between 2000 and 2010 (CODIS Brochure, 2011)

Probability of a CODIS Match

In order to determine the likelihood that a particular DNA match could also occur randomly, scientists must have a rough idea of how frequent a given STR repeat is present in a given population. This information is termed the *allele frequency*, where the allele in this case refers to the type of STR an individual has at that location. As discussed in Chapter-1, at a given location, individuals can have two alleles, one inherited from each parent. So for example, at one STR locus, an individual could have 2 and 4 repeats, while another individual might have 3 and 7 repeats. Scientists must know approximately how often a 2,4 pattern occurs in the general population. The allele frequencies at all 13 locations are multiplied together to obtain the overall likelihood of a match. When all 13 core loci are analyzed, the likelihood of a random match occurring is one in several billion (Norrsgard, 2008). The higher the number of entries in CODIS, the more accurate scientists can determine allele frequencies, and the more likely the data will get accepted into the courtroom. The more loci analyzed for a given sample, the more accurate the analysis. Therefore, in addition to helping solve crimes, databases are also needed to help assign accurate allele frequencies to individual loci. An allele frequency based only on 100 samples is far less accurate than one based on 500,000 samples.

Medical Genetic Databases

In contrast to CODIS, medical genetic databases can contain entire human genomes. An example of a genetic database is the one in Iceland (Hloden, 2000). Iceland's National Health Database includes medical and family history records, which is information far in excess of the CODIS 13 core loci. The purpose of Iceland's database is to help map gene mutations to specific diseases. deCODE, a biomedical company, was granted permission to research genes associated

with over 30 diseases using Iceland's national health records. Iceland is a good place to have such a genetic database. Because of its small population of about 275,000 people, and detailed individual medical records that have been maintained by public health services since 1915, this makes obtaining donor consent easier to obtain. In addition, its relatively genetically homogeneous population makes it easier to find genetic mutations (Hloden, 2000). In 2012, scientists used the Icelandic database to identify a mutation in the gene encoding the amyloid precursor protein (APP) that prevents individuals from accumulating the A β toxin in their brains, and prevents them from getting Alzheimer's disease (Jonsson et al., 2012).

Due to the type of information within medical databases, they have privacy issues far in excess of a criminal database. Although any Icelander can opt out the database at any time, the data already entered will not be removed, raising privacy rights issues. And any leak of information from the database could lead to an abuse of information if any individuals are subsequently denied health care coverage. In this database, an individual's information is encrypted, however codes can be broken. Most experts who reviewed the project's privacy measures have determined that the information in the database is personally identifiable (Hloden, 2000). Because of this, it is very important that individuals provide informed consent when contributing their DNA to a medical database.

DNA Database Ethics

With respect to whose DNA should be entered into CODIS database, in the U.S. individual states make this decision. **Table 4.2** shows the 50 states and their requirements as of 2010 for entering DNA into CODIS. From a crime solving point of view, *everyone's* DNA should be included in CODIS. This could be achieved in the future by taking a cotton swab of

the cheek at time of birth. But no state is currently considering this. The state of Massachusetts requires all *convicted* felons and some *convicted* juveniles to enter their profiles. Only 15 states require *arrestees* to submit their DNA samples, and only 9 states ask people who are not guilty because of mental illness to provide their DNA samples.

State	All Felonies	Some Juveniles	Some Misdemeanors	Some Arrestees	Not Guilty By Mental Defect or GBMI	Other
Alabama	X					
Alaska	X	X		X -- Violent felonies.		
Arizona	X	X		X -- Many serious felonies.		Includes residential and criminal burglary.
Arkansas	X	X -- Violent crimes only.	X -- Some sexual offenses.		X	
California	X	X		X -- Expansion to all felon arrestees starts in 2009.		Includes those convicted of terrorist activity in violation of weapons of mass destruction provisions; and those convicted of a qualifying offense in another state.
Colorado	X	X				Includes any person who has a duty to register as a sex offender, including probationers, habitual offenders as condition of parole, and those released without parole supervision.
Connecticut	X				X	Includes persons on probation or parole prior to discharge from supervision.
Delaware	X		X -- Certain child endangerment or abandonment crimes.			
Florida	X	X			X	Includes persons on probation, parole, release or supervision following conviction of certain offenses.
Georgia	X	X				Includes probationers convicted of qualifying offense.
Hawaii	X	X			X	Includes qualifying persons in prison, on probation or parole, parole violators.
Idaho		X				Most felons are included.
Illinois	X	X	X -- Any person required to register as a sex offender, includes some misdemeanors.			Includes people held under civil commitment law, those found guilty but mentally ill for a sex offense, persons seeking transfer to state under interstate compact, stalking and residential burglary.
Indiana	X					Includes qualifying offenders on probation or parole.
Iowa	X	X	X Any person required to register as a sex offender. Any criminal offenses against minors included.		X	Includes qualifying parolees and offenders on work release and offenders receiving a deferred judgment of felony.

Kansas	X	X		X -- Felony or drug grid level 1 or 2; expands after June 30, 2008 to include all persons arrested for a felony.		
Kentucky		X				Includes those convicted of unlawful transaction with a minor, promoting sexual performance of a minor, Burglary I and II and Class A and B felonies involving death or serious injury to the victim.
Louisiana	X	X		X --If funds authorized.		
Maine	X	X	(May include a lesser included offense if a qualifying offense was originally charged.)			Includes all Class A, B, C serious crimes and Class D and E convictions if the person had prior felony conviction for which DNA not collected.
Maryland	X	X	X	X -- Violent crimes, burglary and breaking and entering of a motor vehicle.		
Massachusetts	X	X				
Michigan	X	X		X -- Violent felonies.		
Minnesota	X	X	(May include offenses "arising out of same set of circumstances.")	X -- Specified serious crimes upon judicial finding of probable cause.		
Mississippi	X					
Missouri	X					
Montana	X	X				
Nebraska						
Nevada	X		X -- Failure to register as a convicted person.			
New Hampshire		X				Includes violent crimes.
New Jersey	X	X	X -- Any crime for which a sentence of imprisonment of 6 months or more is imposed.		X	
New Mexico	X	X		X -- Specific violent felonies.		
New York	X		X -- Many misdemeanors.			
North Carolina	X				X	Includes persons on community supervision.
North Dakota	X			X -- All felonies -- effective 01/09.		Many serious felonies, including burglary.
Ohio	X	X	X -- Certain child victim offenses.			

Oklahoma	X					2001 law requires planning to incrementally add qualifying felonies to the database, to include all felony offenses by 2006.
Oregon	X	X				
Pennsylvania		X				Includes violent and sexual offenders.
Rhode Island	X					
South Carolina	X	X	(May be required by court order for any offense.)	X -- Violent felonies punishable by more than 5 years in prison.		Includes qualifying offenders on community supervision.
South Dakota	X	X		X -- Violent felonies punishable by more than 5 years in prison.		
Tennessee	X	X		X -- Violent felonies, upon finding of probable cause.		Includes those persons seeking transfer to the state under interstate compact who have committed qualifying offense.
Texas	X	X	(May be required by court order for any offense.)	X -- Post-indictment only in certain sex crimes.		Expanding to all felons contingent upon federal funds.
Utah	X	X	X -- Class A misdemeanors. Others may qualify if convicted on lower degree of qualifying offense.		X	Includes persons convicted in another state of a qualifying offense.
Vermont	X		(Only if as part of a plea agreement.)			
Virginia	X	X		X -- Violent felonies, including attempts.		
Washington	X	X				Includes those who have been convicted out of state or under federal law of a violent offense.
West Virginia	X					
Wisconsin	X	X	X -- Some misdemeanors for which sex offender registration is required.		X	
Wyoming	X	X				Includes all persons required to register as a sex offender.

Table 4.2: List of US States and Their 2010 Requirements for CODIS Entries. (National Conference of State Legislature, 2010)

DNA Database Privacy Rights

The more DNA samples CODIS contains, the more accurate we can assign allele frequencies, and the more likely it is to find a DNA match to someone. Thus, having everyone's DNA profile in the system (not just previous offenders) would make it easier to solve crimes. However, the public is concerned that DNA databases violate an individual's right to privacy. The American Civil Liberties Union is concerned with having DNA taken from innocent people (ACLU, 2004). Convicted felons have some privacy rights, but the moment they commit a violent crime, felons have fewer rights. Therefore, felons may still have the right to be housed in semi-private facility, but not to withhold their DNA from analysis by the state that could help solve a crime. When debating when to include a convicted felon profile to the CODIS database, one needs to think about the privacy rights of the victim. For example, in a case of murder, the same way you take away someone else's right to live, you surrender your right to privacy. People's right to privacy without apparent cause and search warrant is protected by the Fourth Amendment of the U.S. Constitution, which states:

“The right of the people to be secure in their persons, houses, papers, and effects, against unreasonable searches and seizures, shall not be violated, and no Warrants shall issue, but upon probable cause, supported by Oath or affirmation, and particularly describing the place to be searched, and the persons or things to be seized” (U.S. Constitution, 2008)

The reason many people are against contributing their DNA profiles to databases is they are afraid of their genetic information becoming public through illegal hacking. In the public's point of view, publicizing such information would allow insurance companies or prospective employers to gain medical predisposition information on individuals from the database, which could lead to denial of health and life insurance, discrimination by employers, and discrimination

by coworkers, etc. However, as mentioned in chapter-1, the only information that goes into CODIS is how many repeats you have at 13 core STR loci. None of the information the 13 core loci contain medical disposition data. One cannot get more information out of a database than what it was initially entered into it. Despite the fact that medical predisposition information cannot be extracted from CODIS, one could obtain such data from the *original* DNA sample stored in the freezer. Therefore, this could be prevented by destroying the original DNA sample after obtaining its CODIS information. However, some people think the samples should not be destroyed, because they think that the samples could be used later if there is a problem with original CODIS entry or a DNA matching techniques are discovered. On the other hand, perhaps one could "always draw a fresh sample from the suspected individual and test it" (Mayer-Schoenberger, 2003).

Medical genetic databases are a totally different story with respect to ethics, as these databases indeed contain medical predisposition data which could be leaked to the public if the database is hacked and a link is made from an entry to an individual. So, individuals contributing their DNAs to this type of database should do so only with informed consent.

Chapter-4 Conclusions

Regardless of the majority of people who fear DNA databases, there is a good amount of people who support having a universal DNA database that includes the entire earth population. This database is a forensic database, which means when analyzing DNA profiles it only tests 13 specific core loci. In other words, it does not include any medical predisposition information so people should not fear insurance denial or discrimination by employers. A way to prevent access to the original DNA samples that are often stored in freezers is to destroy them after obtaining

forensic information needed from it. The author of this IQP believes that everyone's DNA should be included in the database for only criminal justice purposes and without any medical predisposition information. The process of collecting every baby born DNA sample will require time and effort, but in one generation we could have a universal DNA database.

Chapter-4 Bibliography

ACLU (2004) ACLU Challenges California Law Permitting Government Seizure of DNA Samples from Innocent People. American Civil Liberties Union.
<<http://www.aclu.org/technology-and-liberty/aclu-challenges-california-law-permitting-government-seizure-dna-samples-inno>>

Adams, Dwight E (2002) Congressional Statement: The FBI's CODIS Program. Federal Bureau of Investigation, U.S. Department of Justice.
<<http://www.fbi.gov/congress/congress02/adams051402.htm>>

Bereano, Phillip (2000) Does Genetic Research Threaten Our Civil Liberties?
<<http://www.actionbioscience.org/>>

Brenner, Charles (2004) Forensic Mathematics of DNA Matching. DNA Profile Probability.
<<http://dna-view.com/profile.htm>>

CODIS Brochure (2011) FBI.gov. http://www.fbi.gov/about-us/lab/codis/codis_brochure/

Hloden, Oksana (2000) For Sale: Iceland's Genetic History.
<<http://www.actionbioscience.org/genomic/hlodan.html>>

Mayer-Schonberger, V (2003) Strands of Privacy: DNA Databases and Informational Privacy and the OECD Guidelines.
<<http://www.ksg.harvard.edu/dnabook/>>

National Conference of State Legislature (2010) State Laws on DNA Data Banks Qualifying Offenses, Others Who Must Provide Sample.
<<http://www.ncsl.org/programs/cj/dnadatabanks.htm>>

Norrsgard K (2008) Forensics, DNA Fingerprinting, and CODIS.
<<http://www.nature.com/scitable/topicpage/forensics-dna-fingerprinting-and-codis-736>>

Pierce, Kent (2009) DNA Clears Man of Murder, Rape.
<http://www.wtnh.com/dpp/news/crime/news_wtnh_new_haven_dna_clears_man_charges_to_be_dropped_200908190544>

The FBI's Combined DNA Index System Program (2000) FBI, U.S. Department of Justice.
<<http://www.fbi.gov/hq/lab/codis/brochure.pdf>>

U.S. Constitution - Fourth Amendment. FindLaw.
<<http://caselaw.lp.findlaw.com/data/constitution/amendment04>>

Zetter K (2010) DNA Sample from Son Led to Arrest of Accused 'Grim Sleeper'
<<http://www.wired.com/threatlevel/2010/07/dna-database/#ixzz0wwYG4QfS>>

PROJECT CONCLUSIONS

This project was done in an attempt to help educate the public on the history, scientific basis, and technology involved in DNA fingerprinting. Chapter 1 introduced the reader to the general topic of DNA fingerprinting, also known as DNA profiling. This technology has been used since 1985 to distinguish individuals from each other using polymorphisms, which are differences found within DNA at specific locations (loci). The chemical components of DNA were discussed, and two main procedures for its analysis were explained: VNTR-RFLP type analysis, and STR-PCR type analysis. VNTR type analysis can take weeks to perform and requires relatively large amounts of DNA, while STR-PCR analysis can be done in only days and is far more sensitive. However, STR-PCR analysis is sensitive to contamination, so when sufficient DNA is available for analysis both procedures should be performed. The chapter concluded by summarizing the main uses for DNA testing.

The second chapter of this project introduced how DNA from crime scenes is collected, transported, tracked (chain of custody), and stored. It discussed the importance and the methods used to prevent contamination and degradation when handling evidence. It is important that each type of DNA evidence be stored correctly for that type to prevent degradation. It is also important that contamination from other DNA sources is prevented. All of these conditions are crucial when attempting to use DNA evidence in court. In all cases, limiting the people allowed to handle the evidence lowers the risk of contamination.

Chapter 3 introduced the reader to several landmark DNA court cases in which technology was introduced and critiqued. This series of cases established legal precedence for admitting complex technology into courts, and mandates the use of pre-trial hearings for each

trial to validate the specific evidence to be presented. The landmark trials also mandated the standardization of the technology, and raised the bar technically for any lab wishing to analyze DNA for courts.

The last chapter introduced the reader to the topic of DNA databases and their ethics. Many people fear DNA databases because they are afraid if their information is compromised (and released to the public or to insurance companies), it might lead to insurance denial or discrimination. However, many people are not aware of the differences between forensic DNA databases (such as CODIS) and medical genetic databases (such as the Iceland Genetic Database). CODIS entries only contain information related to 13 core loci, and these sites do not contain any medical predisposition information. So, even if the CODIS database was hacked, no medical information resides in it. Such is not the case for the Icelandic Genetic Database, which includes information on entire genomes, and which requires higher levels of pre-consent, and protection of privacy. For CODIS entries, the authors of this report agree with the current laws of the state of Massachusetts that all convicted (not arrested) persons of sex crimes and violent crimes should be required to donate DNA to CODIS. However, one author believes that the entries should be expanded to include all individuals *convicted* of *any* crime (if the conviction is solid), while the other author believes that from this time forward DNA should be taken from all newborns to greatly expand the database for helping solve crimes even for *first* offenders.

In conclusion, DNA fingerprinting has come a long way from 1985 when it was first used in England to settle a paternity case involving immigration. Over time, the methods for extracting, transporting, and storing DNA have advanced to help prevent DNA contamination and degradation. DNA can even be analyzed from ancient samples under some circumstances. DNA databases have allowed thousands of crimes to be solved from hits to previous offenders.

In the future, everyone's DNA may reside within CODIS by taking cheek swabs at time of birth, as is currently being debated by some U.S. states.