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# DNA Fingerprinting

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# DNA FINGERPRINTING

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By:


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# ABSTRACT

This project investigated the impact of the new technology of DNA fingerprinting on society, especially the legal system and database ethics. Our conclusions propose an expansion of DNA databases to include individuals convicted of any felony, not just violent crimes. The benefit to society of such DNA databases to identify unknown corpses, determine paternity, place a suspect at the scene of a crime, develop leads where otherwise there were none, and link crimes to identify serial criminals outweighs any privacy issues especially for convicted felons.

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# EXECUTIVE SUMMARY

Five decades ago, Watson and Crick discovered the secrets of DNA structure. DNA Fingerprinting, or DNA profiling, was first adopted in 1984 by Oxford University educated Alec Jeffreys. Jeffrey's discovery opened a whole new world that, once proven and perfected, would unlock markers to visualize each person's unique identity. Inside each human being, as well as plants, animals, and microorganisms, lies a unique DNA structure. Today, DNA Fingerprinting is "rapidly becoming the primary method for identifying and distinguishing individual human beings" (Betsch, 1994). Some of the applications of DNA fingerprinting techniques include: murder cases, rape cases, paternity testing, diagnosis of inherited disorders, military identification, and molecular archaeology.

Forensic DNA analysis has been admitted into United States courtrooms since 1987. DNA profiling does not claim to be an absolute identification, but may be very strong evidence and is considered to be just one part of the entire case. DNA profiling is used primarily in sexual assault cases. Prior to DNA testing, labs were limited regarding the amount of genetic information that could be obtained from evidence samples. Now with DNA testing, the genetic content of the evidence sample itself can be examined. In the case of a rape investigation, four profiles are formed: the first is a DNA profile from the blood of the victim, the second DNA profile is formed from the blood of the defendant, thirdly a vaginal swab is taken, and the female and male fractions are separated and profiled separately. The victim's blood profile and the vaginal swab profile should match alleles. The defendant's blood sample profile should match the male fraction profile in order to consider him as a suspect. It is important to remember that DNA profiling does not incriminate a suspect with absolute certainty, but if the profiles don't

match it can exclude with absolute certainty. In the case of likely matches, statistics are used to determine the frequency that this DNA profile likely appears in the general human population.

"Since the discovery of DNA fingerprinting at the turn of the 20th century, science has assumed an increasingly important and powerful role in the decision making process of our judicial branch" (Biancamano, 1996). Many Landmark cases, which set the standards for admitting DNA into the courtroom, as well as the reliability and acceptability of DNA techniques, will be discussed to prove the methods used today are valid and reliable when performed properly. Some of the earlier cases that will be talked about did not actually involve DNA, but what they did accomplish was to set precedence by establishing new rules for admittance of technical evidence, and to set new generally accepted scientific standards in the court of law. Some of these landmark cases include: Frye v. United States in 1923, Federal Rules of Evidence 702 (Rule 702) in 1975, Colin Pitchfork in 1986, Andrews v Florida in 1988, People v Castro in 1989, Two Bulls v US in 1990, and the case of Paul Eugene Robinson in 2003 where the DNA evidence was the primary basis of the conviction of sexual assault.

The world's largest DNA database is a U.S. system called the Combined DNA Index System, or CODIS. "CODIS blends computer and DNA technologies into an effective tool for fighting violent crimes" (Brown and Niezgoda, 1995). Some people feel that these DNA databases are a great help to society, giving new hope to forensic cases that would otherwise have no leads. Others feel they are an invasion of privacy, and that these databases should consist only of samples collected from violent crime offenders. Still others do not even understand what they are. In this project we explain what DNA databases are, and what they are used for. Based on the research performed for this project, it is our conclusion that database samples should be expanded to include persons convicted of any felony, not just violent felons.

We feel that the potential benefit to society of these databases far outweigh an individuals right to privacy, especially for a convicted felon who we feel gave up such rights (to not give a blood sample) when the crime was committed. We also feel that the original DNA sample should be destroyed after information on the 13 core forensic loci are entered in CODIS. No known medical information exists in the 13 core loci, but such information could be obtained in the future from the original DNA sample. Destruction of the sample should eliminate the public's fears that medical information can be obtained from the database.

Amidst the controversy over the use of DNA databasing as a means of crime solving, one fact remains clear: DNA evidence has been critical in the arrests and convictions of hundreds of thousands of criminals who would have otherwise gone unprosecuted.

# PROJECT OBJECTIVE

DNA fingerprinting is a powerful new technology, which is used to assist in convicting the guilty and exonerating the innocent. The topic of DNA fingerprinting however remains controversial in the courtroom regarding technical issues, and also has legal, cultural and political consequences. This controversy is mainly derived from the lack of knowledge regarding the procedures of DNA fingerprinting, how it is obtained, analyzed, and reported. The purpose of this project is to help to eliminate the public's doubt, concerning DNA fingerprinting and help them to convert their reservations about the science into support for the use of DNA fingerprinting in the courtroom. The focus of this paper will be directed toward the layperson in order to target the prospective population of jurors. Jurors are members of our community who are required to evaluate the relevance of all evidence including DNA evidence. Thus, once they are on a jury, and DNA evidence is brought in, they will be able to comprehend the data and make an informed verdict. This paper will cover the primary areas of the public's concerns, as well as provide history of DNA analysis, and future advances of the science for admission into the courtroom. Description of the lab report, as well as the actual data used to formulate the lab report will help the public develop an appreciation for the formulation of the results of the actual testing. The objective of this paper was accomplished by first, outlining the science of the main techniques of DNA fingerprinting in laymen's terms. Secondly the project describes the processes of collection, storage, and prevention of contamination of the DNA evidence. Thirdly, landmark DNA court cases that established legal precedents for admitting DNA in U.S. courts will be revisited and analyzed. Lastly, many of the controversies regarding the DNA databases will be analyzed. This project not only informs the reader about the facts of the history of DNA

fingerprinting, but it will also entice the reader to encourage and support the use of DNA evidence in the courtroom. The overall goal of this paper is for the reader to walk away with an understanding of DNA fingerprinting's positive impact on society.

# CHAPTER 1: DNA FINGERPRINTING TECHNIQUES

Brian Steele and Morgan Reynolds

## WHAT ARE DNA FINGERPRINTS?

DNA Fingerprinting, or DNA profiling, was first adopted in 1984 by Oxford University educated Alec Jeffreys (Keegan, 2004). Jeffreys stumbled upon this theory while working on myoglobin, the gene that codes for an iron-containing protein in muscles (Biographies, 2004). Jeffrey's discovery opened a whole new world that, once proven and perfected, would unlock each person's unique identity. Inside each and every human being, as well as plants, animals, and microorganisms, lies a unique DNA structure (Antler, 2004). Conventional fingerprints occur only on the fingertips, and are capable of being altered, but a DNA fingerprint is the same for "every cell, tissue, and organ of a person" (Betsch, 1994). DNA is incapable of being altered, and is "rapidly becoming the primary method for identifying and distinguishing among individual human beings" (Betsch, 1994). In science, DNA Fingerprinting does not point to a unique individual (Keegan, 2004). However, it provides a profile, and then the probability that there are others who also match the profile is determined leading to the match or conviction (Keegan, 2004). So what is DNA fingerprinting used for? Some of the applications of DNA fingerprinting techniques include: murder cases, rape cases, paternity testing, diagnosis of inherited disorders, military identification, and molecular archaeology.

In this chapter, we will explore each application, as well as simplify DNA for the average person, show how to run a fingerprint, trace its growth and impact on society, and look toward future uses such as national DNA databases and DNA fingerprints as one day becoming our National Identification Cards.

## DNA EXPLAINED IN SIMPLE TERMS

Deoxyribonucleic acid, more commonly known as DNA, is the complex chemical structure that uniquely identifies each and every organism. An organism's complete set of DNA is known as a genome. DNA is the fundamental building block of the genome (An Introduction to DNA, 2002). DNA is located inside an organism's chromosomes. A chromosome is a structure found in the cell nucleus that contains genes, which are the functional and physical unit of heredity passed from parent to offspring. Chromosomes are composed of DNA as well as proteins. Each parent contributes one chromosome to each pair of a child's chromosomes, so children get half of their chromosomes from their mothers and half from their fathers. A human has 46 chromosomes, or 23 pairs of chromosomes. Organisms differ in the number of chromosomes that they embody. For example, dogs contain 39 pairs, a puffer fish contains 21 pairs, and a sun flower has 17 pairs. "Chromosomes are merely the containers of DNA" (Rohloff, 2000). An illustration of an organism's DNA is shown in Figure 1.

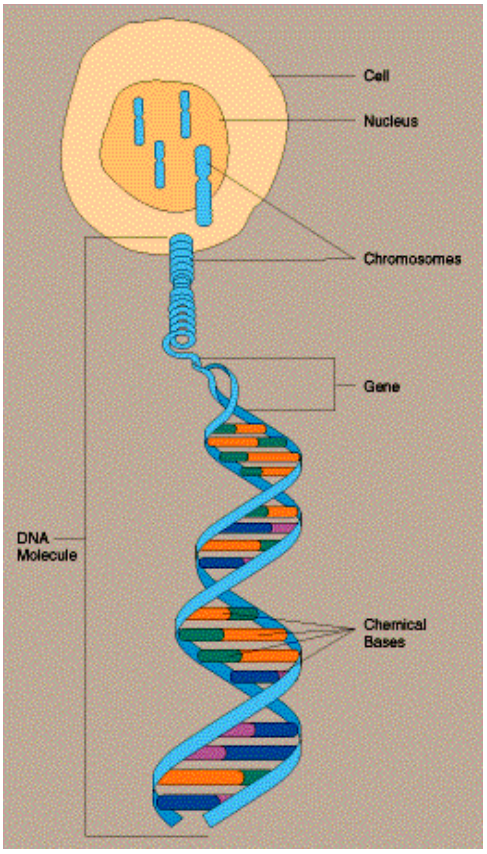



**Figure 1 – DNA is packaged in units of chromosomes, housed in the cell's nucleus. (Understanding Gene Testing, 1994-2004)**

Now that we know where DNA is located, we can identify what it is, and how it works. "DNA itself is a double-helix polymer. A polymer is simply a large molecule that is produced by

several smaller units linking together and forming a chain" (Rohloff, 2000). The term double-helix comes from the discoveries of Francis Crick and James Watson. "On Feb. 28, 1953, Francis Crick walked into the Eagle pub in Cambridge, England, and, as James Watson later recalled, announced that 'we had found the secret of life.' Actually, they had. That morning, Watson and Crick had figured out the structure of DNA" (Wright, 1999). Think of the DNA structure as a twisted ladder, as shown in Figure 2. The rungs of the ladder contain complementary pairs of nitrogen bases - Adenine always paired with Thymine, and Guanine always paired with Cytosine. The twisted uprights of the ladder are formed by alternating deoxyribose, or sugar, and phosphate molecules, which together with one base pair make nucleotides (DNA from the Beginning, 2002). The human genome is made up of 3 billion nucleotides (Natural Toxins Research Center, 2004). "Encoded in this DNA is the information for about 100,000 genes. About 95% of the DNA is considered "junk", that is, it may not have a known function. About 5% of the DNA has a known function and is considered the coding regions of the DNA (Natural Toxins Research Center, 2004).

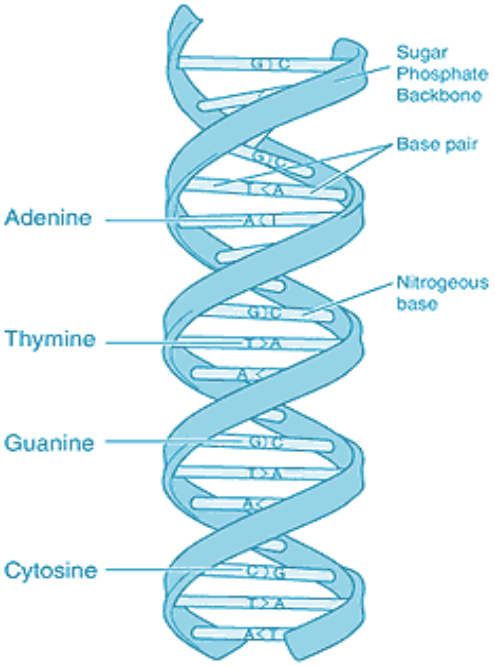



**Figure 2 – Watson and Crick's discovery of the DNA "Double Helix" (Devitt, 2002)**

So now we know that every organism is made up of cells, which encompass chromosomes, which harbor our DNA. The specific position on a chromosome of a gene where DNA is located is known as a locus. The locus, which is a stretch of DNA, is what is analyzed for variability using different methods of testing.

**How is DNA Used in the Human Body?**

In order for an individual to grow, cells must be produced. In order for cells to be produced they need to be copied so that the new cell can be a replicate of the existing cell. DNA provides this process of replication of genetic information. Therefore the DNA itself needs to be replicated because each cell needs a complete DNA strand in order to dictate the formation of proteins from that cell. With the exception of mitochondrial DNA, nuclear DNA never leaves the nucleus. Copies of the genes within the DNA are sent out of the nucleus, which in turn provides the instructions for the formation of specific proteins assigned to that specific gene. The copies of the DNA are known as RNA. There are three forms of RNA: messenger RNA (mRNA), ribosomal RNA (rRNA), and transfer RNA (tRNA). The process of replication is very precise. An enzyme, helicase "unzips" the DNA helix for a length of approximately 1,000 nucleotides resulting in two individual strands of DNA (Watson, 2003). The enzyme DNA polymerase, travels up one end of the DNA strand placing complimentary bases down the other strand forming complementary base pairs. A third enzyme known as DNA ligase, attaches one newly formed strand to the previously replicated strand, and the process repeats itself continuing down the DNA strand.

Now that the DNA is replicated, it needs to dictate the formation of the proteins. The process of producing proteins is called gene expression. Gene expression occurs in two stages; transcription and translation. In the process of transcription, the mRNA is formed from a gene within the DNA. Translation is the process of mRNA directing the production of specific proteins (Johnson, 2003).

The process of transcription takes place within the cells nucleus. The protein called RNA polymerase binds to one strand of the DNA and moves along the strand. As it moves along, the polymerase pairs each nucleotide with its complimentary RNA version. This form of RNA includes exons and introns (Watson, 2003). The exons are the coding regions of the DNA and the introns are the non-coding regions of the DNA. The introns are cut out of the RNA transcript and the coding regions (exons) are joined together. A three-nucleotide sequence on an mRNA is called a codon, and this corresponds to a specific amino acid. The result of the transcription process produces an mRNA copy of a gene. The mRNA then moves out of the nucleus into the cytoplasm of the cell and the process of translation begins (Johnson, 2003).

In translation (see Figure-3) , ribosomes use the mRNA to actually produce the protein. The ribosomal subunits bind to the mRNA. The tRNA molecules then become attached to their specific amino acid through the action of enzymes. Amino acids are then brought to the ribosome in a specific order that has been directed by the mRNA. Peptide bonds form between the amino acids as the mRNA moves along the ribosome sites. The chain of amino acids then forms the protein. Each specific code from the mRNA forms a specific protein (Watson, 2003).

**The Steps of Translation:**

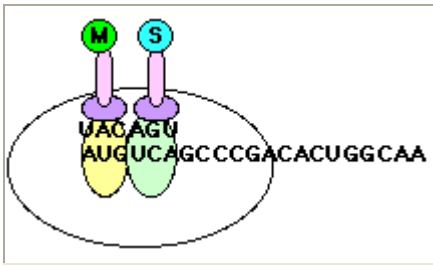
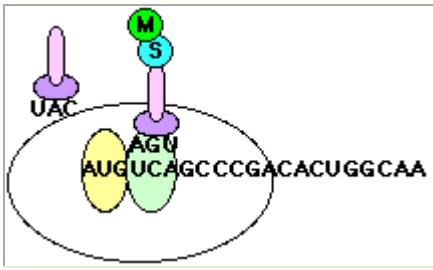
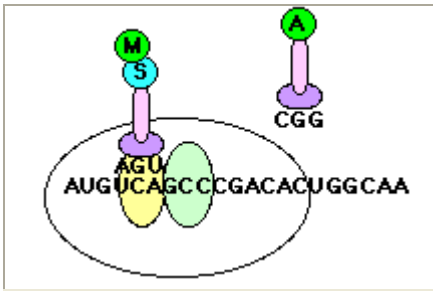
| | |
|---|---|
|  | Initial steps: Messenger RNA is bound to ribosome with the start codon (AUG) at the P site. A transfer RNA molecule with the amino acid methionine (M) and the anticodon UAC has bound to the exposed start codon. The codon UCA is exposed at the A site. |
|  | A second transfer RNA molecule, with the anticodon AGU and the amino acid serine (S) has bound to the A site. The 2 amino acids are close enough to form a peptide bond between them. |
|  | A peptide bond has formed between M and S and the peptide is bound to the A site. The methionine transfer RNA leaves, and the P site is exposed. |
|  | The ribosome has moved along the messenger RNA one codon, bringing the peptide to the P site. This exposes the A site and the next transfer RNA, carrying alanine (A) is about to bind. |

**Figure 3:** This describes the process of translation, which is the second stage in the formation of amino acids and then eventually proteins. (The Steps of Translation, 2004)

**What is the Process of Making a DNA Profile?**

The key to DNA profiling is to make a comparison of unique loci of the DNA left at the crime scene with a suspect's DNA. The portion of the genome where there is a lot of diversity among individuals is called polymorphic regions. The polymorphic regions used for forensics are the non-coding regions. These are the regions of the DNA that do not code for proteins and they make-up 95% of our genetic DNA. These regions are therefore called the "junk" portion of the DNA. Although these "junk" regions do not generate proteins, they can regulate gene expression, they aid in the reading of other genes that do formulate the proteins, and they are a large portion of the chromosome structure (How DNA Evidence Works, 2004).

The non-coding DNA regions are made-up of length polymorphisms, which are variations in the physical length of the DNA molecule. The DNA profile analyzes the length polymorphisms in the non-coding areas. These polymorphisms are identical repeat sequences of DNA base pairs. The number of tandem repeats at specific loci on the chromosome varies between individuals. For any specific loci, there will be a certain number of repeats. These repeat regions are classified into groups depending on the size of the repeat region. Variable number of tandem repeats (VNTRs) have repeats of 9-80 base pairs. Short tandem repeats (STRs) contain 2-5 base pair repeats (Brief Introduction to STRs, 2004).

For each of our 23 pairs of chromosomes, we inherit one copy of each chromosome from our mother and the other from our father. "This means that you have two copies of each VNTR locus, just like you have two copies of real genes (Figure-4). If you have the same number of sequence repeats at a particular VNTR site, you are called homozygous at that site; if you have a different number of repeats, you are said to be heterozygous" (How DNA Evidence Works, 2001).

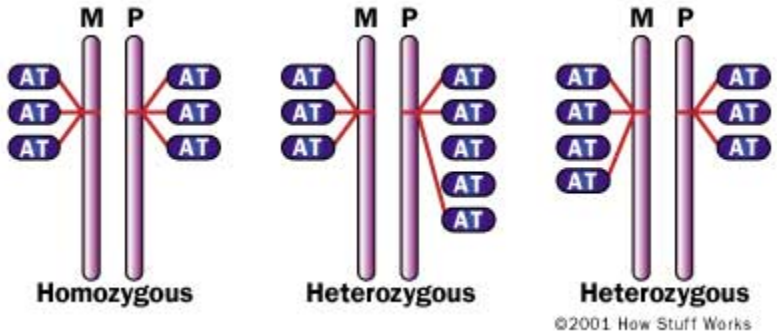



**Figure 4:** The diagram above displays three possible VNTR combinations.

DNA analysis is a laboratory procedure that requires a number of steps. There are a number of techniques used by different laboratories, however in this paper two techniques will be reviewed: Restriction Fragment Length Polymorphism (RFLP), and Polymerase Chain Reaction (PCR) using Short Tandem Repeats (STRs).

The first step in both procedures involves the extraction and purification of the DNA. Before a DNA sample can be analyzed, the DNA needs to be isolated from the other organic and non-organic portions of the sample. The type of sample will determine the technique used to isolate the DNA. The sample may be boiled with a detergent that breaks down the proteins and other cellular material but does not affect the DNA. Enzymes may be added to break down proteins and other cellular material. Organic solvents may be used to separate the DNA from the other organic and non-organic material. The DNA is then separated from the proteins and other cellular material (DNA Forensics, Problem Set 1, 2004).

## THE TWO MAIN DNA TESTS: RFLP AND PCR/STR

*RFLP's*

The two main DNA tests are restriction fragment length polymorphism (RFLP) and polymerase chain reaction (PCR) amplification of short tandem repeats (STRs). Other testing methods exist, but they lack accuracy and precision.

The RFLP is considered to be more accurate than the PCR, mainly because of the size of the sample being analyzed, the need for a fresh sample of the DNA, and its non-amplification of contaminating DNA (Biology 4A, 2004). The RFLP, however, is more costly, and requires a longer time period to complete the analysis. In Figure 5, an RFLP is shown from start to finish.

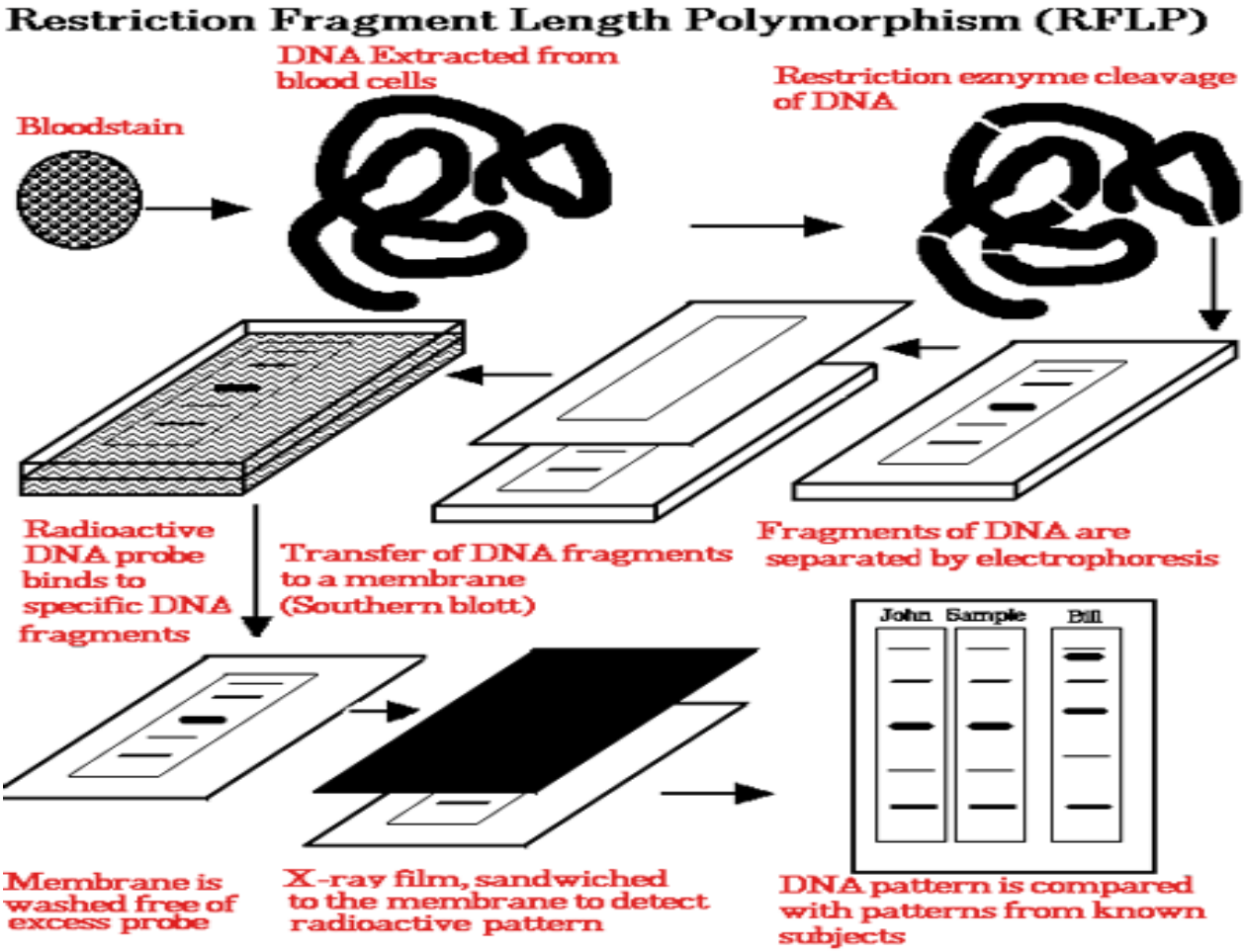



**Figure 5 - One test used to create DNA fingerprints is known as Restriction Fragment Length Polymorphism, or RFLP. Here it is shown from the first step to the finished product. (On the Human genome Project, 2004)**

The first step to this RFLP test is to recover a sample of the DNA of which will be tested. For an RFLP, the sample size needs to be large enough, and undamaged. In Figure 6, RFLP sample sizes are shown for specific substances that are most commonly tested. Once the required sample size is recovered, it is extracted from the sample, and then cut into fragments by restriction enzymes. The DNA fragments have a negative charge and can be separated by a technique called gel electrophoresis, which separates the pieces of DNA based on their size.

| Sample Size for RFLP Fingerprinting | |
|---|---|
| Blood | 15 ul |
| Semen | 5 ul |
| Skin | 5 mg |

**Figure 6 – Sample size of substances needed to conduct an RFLP test. (Micro 7, 2004)**

The next step is known as a Southern Blot, or Southern Hybridization, named after E.M. Southern who invented it while analyzing viral DNA. This is a "procedure that allows detection of specific DNA gene sequences from a mixture of DNA fragments, and is one of the most widely used procedures in molecular biology" (On the Human Genome Project, 2004). This procedure consists of the transfer, or blotting, of the separated DNA fragments onto a nitrocellulose membrane. The membrane is then hybridized, or bound, with a labeled probe that is specific for one VNTR region. "If an X-ray photograph is taken of the Southern Blot after a radioactive probe has been allowed to bond with the denatured DNA on the paper, only the areas where the radioactive probe binds [thick black bands in Figure-5] will show up on the film. This allows researchers to identify, in a particular person's DNA, the occurrence and frequency of the particular genetic pattern matched by the probe" (Brinton and Lieberman, 1994). These bands, once compared to other known samples, are the final output of DNA Fingerprinting..

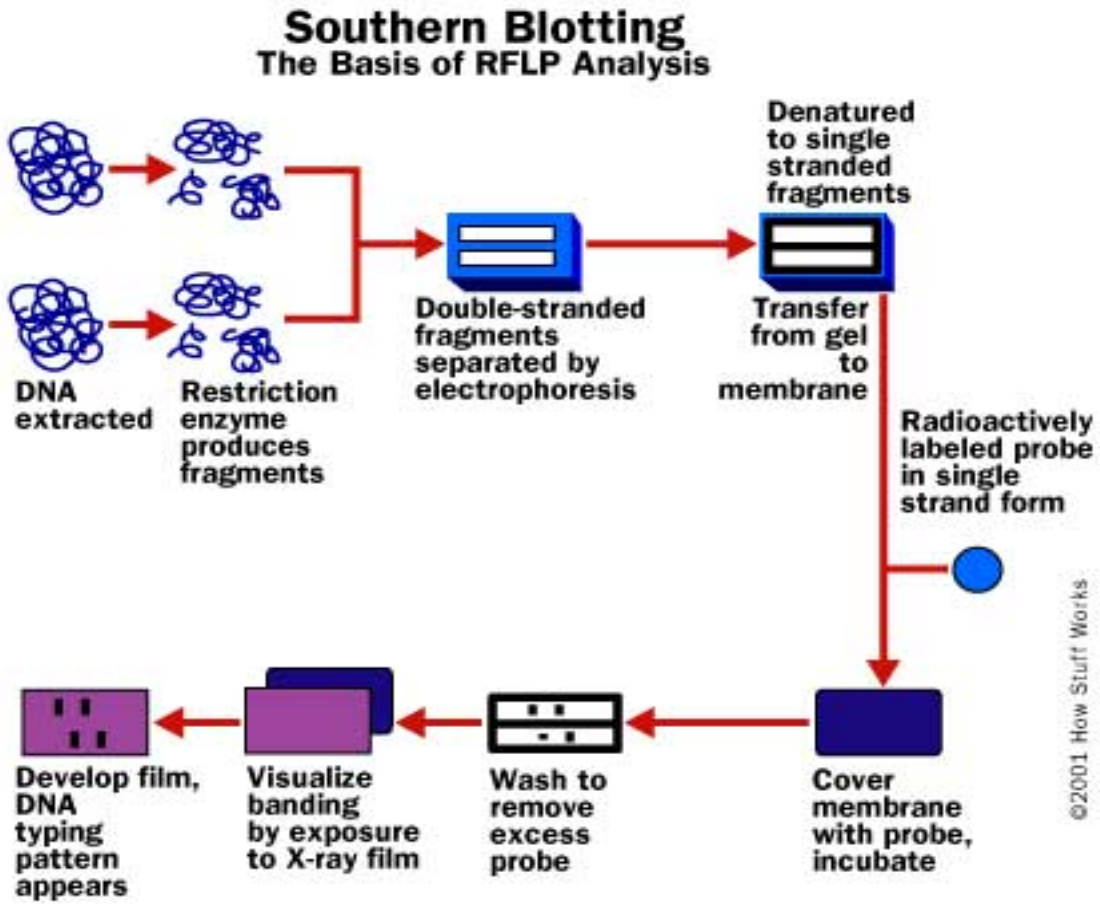



**Figure 7:** Description of Southern Blot Procedure. (How DNA Evidence Works, 2001)

**Polymerase Chain Reaction (PCR)**

PCR is a new technique that was developed in 1986 by Kary Mullis and was applied to forensics in 1991 (Forensic Fact Files DNA Profiling, 2004). PCR has the ability to replicate genetic material, and even proofread and make corrections on the copies (Forensic Fact Files DNA Profiling, 2004). It involves the repeated copying of specified areas of DNA molecules. These areas are the alleles and are specific sequences of base pairs. These target areas are the variable regions. At either end of these target areas are "flanker" bases that are the non-variable regions. The flankers occur at the same locations on the chromosomes of all people. The basis of PCR is to reproduce thousands of copies of a particular variable region of the DNA that is of interest. This results in DNA fragments that vary in length due to the variability in the number of repeated regions. The process is used to amplify a small amount of DNA taken from a sample of blood, hair, etc. With each cycle of copying, the quantity of the target allele is doubled resulting in an exponential amplification of the gene in the PCR. After 30 copying cycles, there will be one billion times more copies of the target alleles than at the start of the amplification process (Principle of the PCR, 1999).

Analysis of the alleles uses length variation of the amplified alleles. The alleles used are repeated units of the same sequence of bases. The length variation is the number of times that a particular base sequence is repeated between flankers. PCR techniques are divided into different categories depending on the number of base pairs in the repeated units. Variable Number of Tandem Repeats (VNTR) uses between 9 and 40 bases per unit; Amplified Fragment Length Polymorphism (AmpFLP) uses between 8 and 16 bases per unit; and Short Tandem Repeats (STR) uses between 4 and 6 bases per unit (Schumm, 1996). STRs will be focused on in this paper because it is the technique of choice for most forensics laboratories presently.

During forensic examination, an STR with a known repeat sequence is extracted and separated by electrophoresis. The distance that the STR migrated is examined. However, capillary columns are now used rather than the gel electrophoresis that was used in the RFLP analysis. Two short pieces of synthetic DNA called primers are specially designed to attach to a conserved common non-variable region of DNA, which flanks the variable target region of the DNA. A mixture of chemicals including the primers, the individual bases and a copying enzyme (polymerase) is added to the solution with the original DNA strand that is to be copied. The primers are short chains of the bases that make up the single strand of DNA. Primers are complementary to the conserved regions flanking the targeted STR (black arrows in Figure-6). The primer used in the PCR amplification process attaches a fluorescent tag to the target alleles enabling the distance traveled by the DNA fragments during capillary electrophoresis to be detected using a fluorescent scanner (Blackett Family DNA Activity 2, 2004).

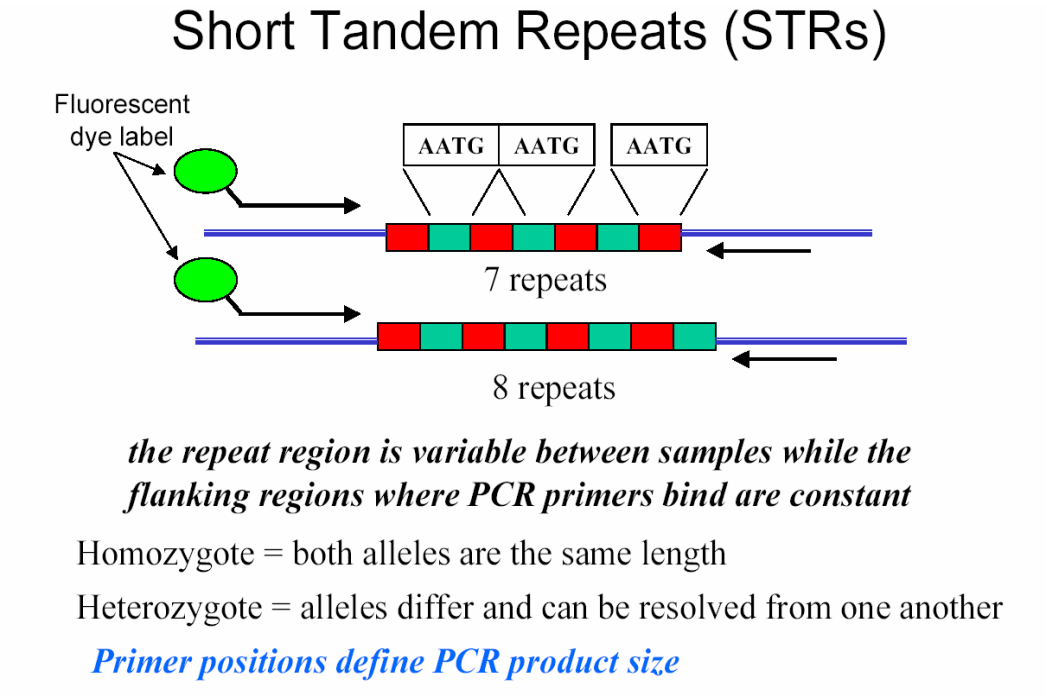



**Figure 8** (Kline, 2001)

There are three steps in the PCR process (see Figure-7):

1) Denaturation: This process takes place at 94 degrees Celcius. This stage separates the DNA double helix, and forms DNA single strands. This also forces every enzymatic reaction to come to a halt.

2) Annealing: This usually takes place at 54 degrees Celsius. The primers bind to their complementary bases on the now single-stranded DNA.

3) Extension: This takes place at 72 degrees Celsius. This step involves the synthesis of DNA by polymerase. Starting from the primer, the polymerase reads the template strand and matches it with complementary bases. The result is two new helixes, each composed of one of the original strands plus its newly assembled complementary strand (Principle of the PCR, 1999).

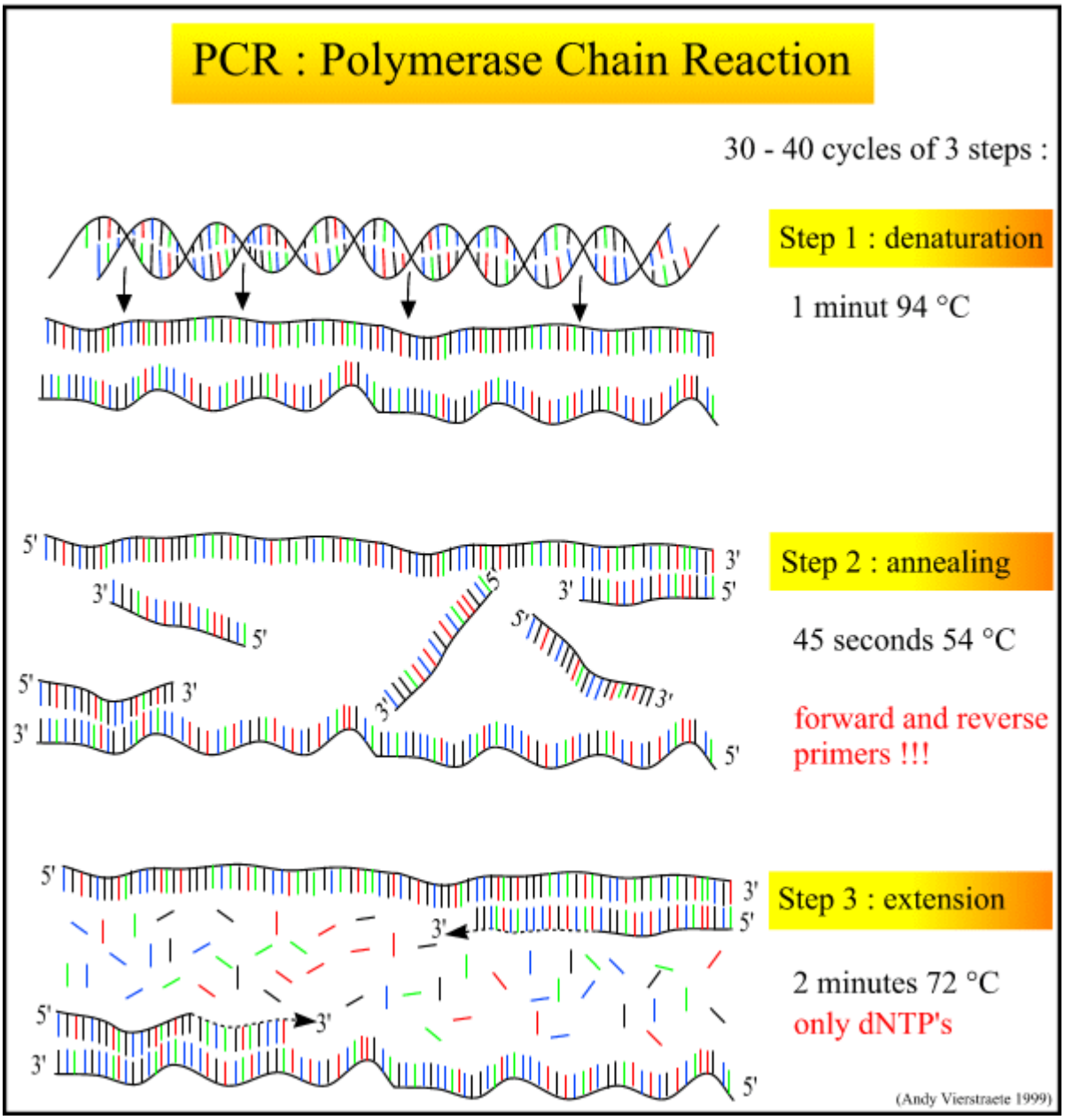



**Figure 9**: Steps in PCR. (Principle of PCR, 2004)

In order to obtain more copies of DNA, the three step process is repeated. Each phase only takes 1-3 minutes, thus if you completed the process for 45 minutes, millions of copies could be generated because every time the process is repeated the amount of DNA doubles (exponentially) (Figure-8).

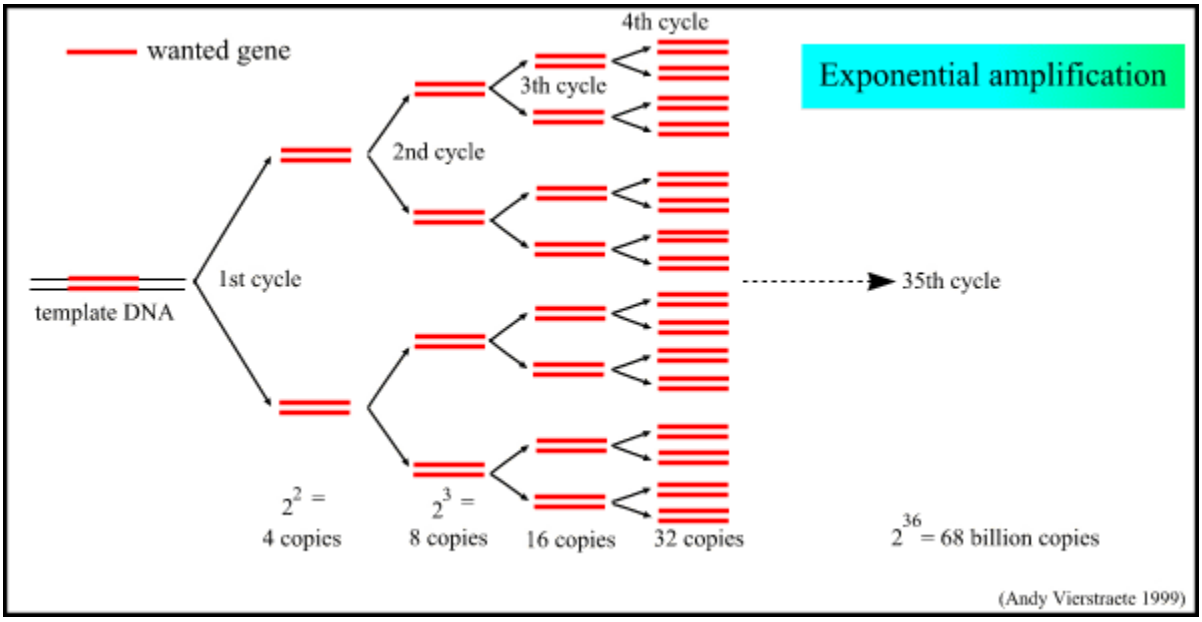



**Figure 10:** Diagram of Exponential Growth in the Number of DNA Copies During PCR. (Principles of the PCR, 1999)

Following the PCR reaction, internal DNA length standards are added to the reaction mixture, and the DNAs are separated by length in a capillary gel electrophoresis machine.

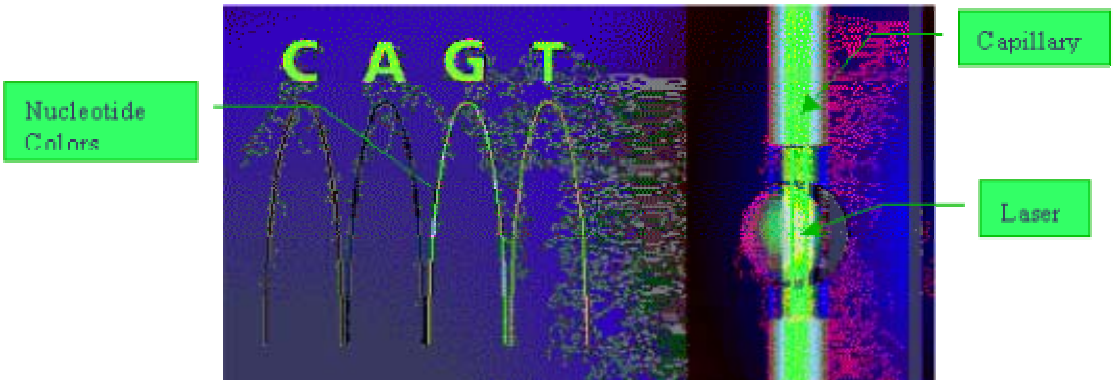



**Figure 11:** Capillary Gel Electrophoresis (How DNA Sequencer Works, 2004)

As the DNA moves through the gel, a laser excites the fluorescent tags and a pattern of bands representing the lengths of the STR is made.

The lengths of the amplified DNA are shown on a scale at the top of the graph (see Figure-10), which is referred to as the Profiler Ladder. The target STR loci are shown by blue boxes below the size ladder. Below the blue boxes are the multiple peaks indicating known size standards for each STR locus (blue peaks in the figure). The lower panel shows the allele numbers for the sample DNA, Norma in this example. One number comes from the father and the other number comes from the mother. If the numbers of alleles are the same as each parent, the person is homozygous for that allele, and if they are different the person is heterozygous for that allele.

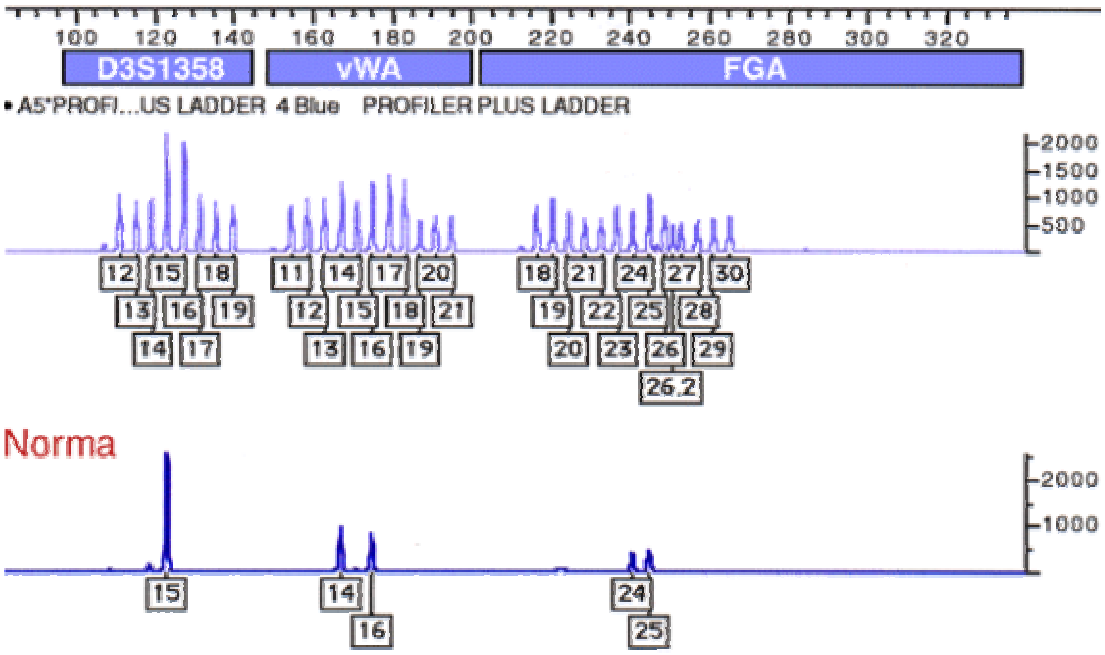



**Figure 12 :** Diagram of fluorescent tags from Norma's sample compared with that of the standard ladder. (Blackett Family DNA Activity 2, 2004)

To extend the number of different loci that can be analyzed in a single PCR reaction, multiple sets of primers with different color fluorescent labels are used. The final DNA profile is built by using several STR loci (5-10 or more) simultaneously. The distances of the primers are carefully

adjusted from the target sequence to prevent the products from the different loci from overlapping during gel electrophoresis (Blackett Family DNA Activity 2, 2004).

The STR loci were initially typed in a manner that allowed for several loci to be run simultaneously through automated capillary electrophoresis and then typed by a computer analysis, which used fluorescent-based laser detection systems. When this technology first came available in 1998, many crime labs began typing three STR loci (THO1, TPOX and CSF) and a sex determination gene (Amelogenin) using "Green I" typing kit (Burt, 2004).

The newer more complicated STR typing kit is called the Profiler Plus which types 9 STR loci (D3S1358, vWa, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D7S820) as well as a separate kit called the Cofiler Kit which types the Green I loci, two of the Profiler Plus loci (D35S1358, D7S820) and a new locus (D16S539). The Profiler Plus identifies and labels fragments of DNA that contain STRs. The STRs are analyzed with three sets of colored primers. A tenth marker is used to distinguish a male (X, Y) from a female (X, X) and is called AMEL. The Cofiler Plus is used to amplify the additional 4 STR loci. The combination of the two analyzers provides 13 STR loci needed for the CODIS database (which will be described later).

The analyzer that completes test is an ABI Prism 310 Genetic Analyzer (Figure-11).

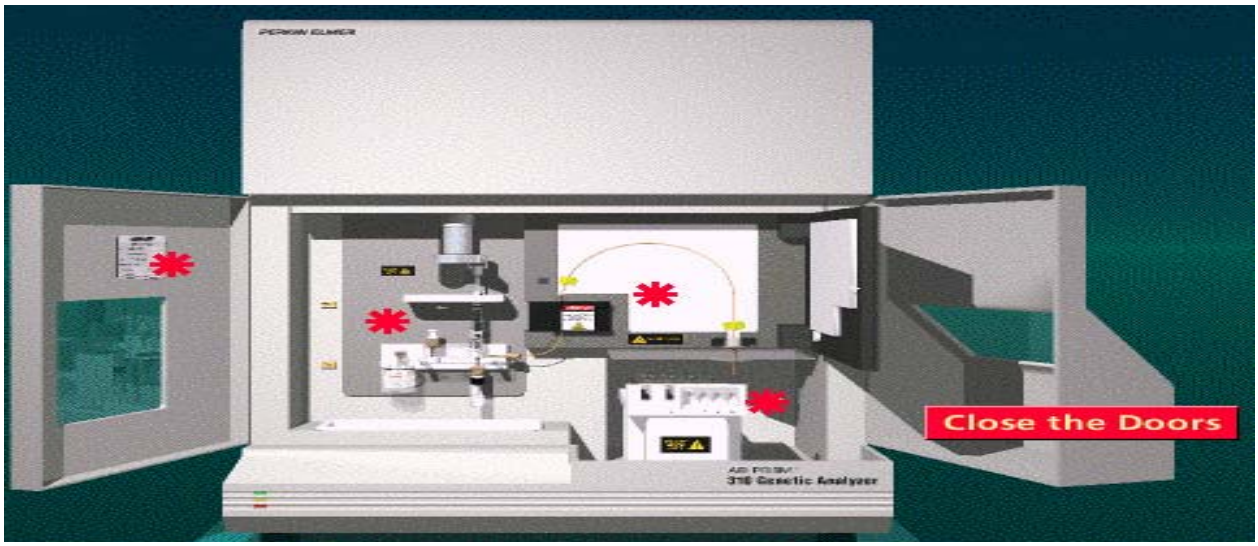



**Figure 13: ABI Prism 310 Genetic Analyzer** (How DNA Sequencer Works, 2004).

The Genetic Analyzer measures the fragment lengths and therefore determines which alleles are present. This analyzer uses new primers, the PCR stage has a new reaction mix, electrophoresis is done in a new polymer medium instead of the gel, and a capillary tube is used instead of the poured slab, a laser forms fluorescence which uses a prism, and lastly, in order to interpret the results, two computer programs are used.

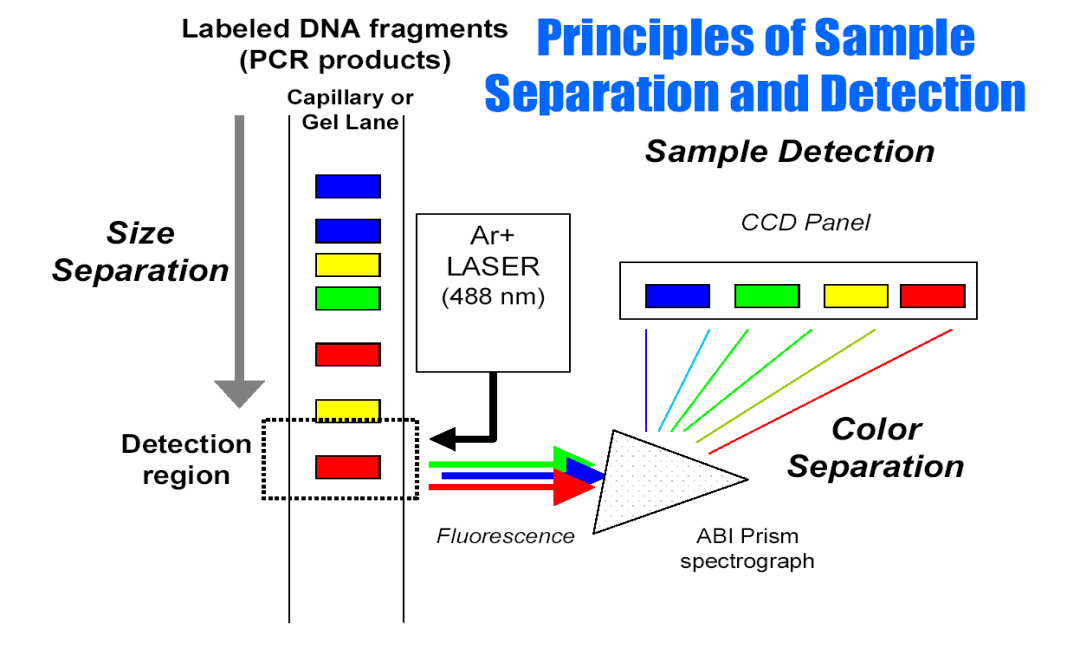



**Figure 14. Fluorescent Color Detection in STR Analysis.** (Kline, 2001)

**What are the Advantages of PCR?**

PCR analysis requires a smaller sample than RFLP, which allows DNA from only 50 cells to be analyzed at a crime scene. Also PCR allows the analysis of DNA fragments that may only differ by a single base pair, even degraded DNA molecules can be used for PCR while RFLP requires intact molecules. Also, PCR fragments because they are shorter can be more accurately sized than RFLP fragments. The disadvantage of PCR is that it is more susceptible to contamination by foreign DNA (Benecke, 1997).

**Lab Report**

The results of the DNA testing are compiled in a lab report that is prepared to be presented as part of the criminal case. The lab report consists mainly of the samples that were tested, types of DNA tests that were conducted; it also states which subjects can be excluded and

which suspects could not be excluded from the case. Lastly, a "table of alleles" (Figure-13) displays the DNA profile of each individual sample.

| | D3S1358 | VWA | FGA | Amel | D8S1179 | D21S11 | D18S51 | D5S818 | D13S317 | D7S820 |
|---|---|---|---|---|---|---|---|---|---|---|
| Blood Stain | 15, 16 | 15, 15 | 25, 26 | XY | 12,13 | 27,30 | 13,14 | 10,11 | 9,12 | 10,12 |
| Suspect 1 | 16, 18 | 15, 16 | 21, 24 | XY | 12,14 | 27,28 | 13,17 | 11,12 | 8,11 | 8,12 |
| Suspect 2 | 15, 15 | 18, 18 | 19, 23.2 | XY | 13,15 | 29,30 | 17 | 11 | 8,9 | 9,10 |
| Suspect 3 | 15, 16 | 15, 15 | 25, 26 | XY | 12,13 | 27,30 | 13,14 | 10,11 | 9,12 | 10,12 |
| Suspect 4 | 16, 16 | 16, 17 | 19, 24 | XY | 14 | 30,30 | 13,16 | 9,11 | 10,11 | 9,10 |

**Figure 15:** Example of Table of Alleles. Only suspect-3 has a DNA profile that matches the DNA profiles observed in the blood sample." (Understanding the Lab Report, 2004)

Listed across the top of the chart are the names of the targeted loci, and the left column shows the samples that were tested (including the sample taken from the crime scene and the DNA profiles of the individual suspects). The numbers within the table represent the alleles that are contained within each locus for each sample. Each allele is an STR site. It is important to remember that offspring gain one allele from each parent, therefore, most of the time two numbers are displayed at each locus site (one allele number from the mother and one from the father). There may be one number at a loci site if the same allele was inherited from the mother and the father making that site homozygous. The technician then compares the suspects alleles to the sample obtained at the crime scene. Following the comparison of the above example, the technician is able to exclude suspects 1, 2, and 4 because they contain different alleles at more than one locus. Suspect 3 has the exact same alleles at each of the loci compared with the sample from the crime scene. The AMEL locus is used to determine the sex of the suspect. A female would be XX and a male would be XY. In this report, all of the suspects are male. "In a

case like this, the lab report will typically say that Suspects 1, 2 and 4 are excluded as possible sources of the blood, and Suspect 3 matches or is included as a possible donor" (Understanding the Lab Report, 2004).

Statistical information is also found in the lab report. The crime labs determine the frequency of each individual allele within given populations and then the individual frequencies of the alleles are multiplied together. For example, "if 10% (1 in 10) of Caucasian Americans are known to exhibit the 14 allele at the first locus (D3S1358) and 20% (1 in 5) are known to have the 15 allele, then the frequency of the pair of alleles would be estimated at 2 X 0.10 X 0.20 = 0.04, or 4% among Caucasian Americans" (Understanding the Lab Report, 2004). This statistic is used mainly to exclude individuals as possible suspects, however if the statistics yield a one in 260 billion frequency (FBI laboratory standard), it is stated in the report as reaching a "scientific certainty". This means that the lab is quite certain that this suspect matches the DNA in question (Blackett Family DNA Activity 2, 2004).

Lawyers frequently take the lab reports and accept their results without searching the actual data from the lab results any deeper. A computer does not analyze the DNA. The technician needs to interpret and make educated decisions about the matching of the profiles whenever there are discrepancies or close calls when reading the graphic representations of the DNA test results (Budowle, 2000).

The electropherogram in Figure-14 displays 4 suspects, one bloodstain, and 3 of the Profiler Plus loci. Each locus is labeled with a colored dye, and the Genetic Analyzer measures the length of the DNA segments by electrical separation in the capillary tube. The laser light illuminates the fluorescent colors, which are captured by the computer camera. The peaks represent the intensities of these lights. "Based on the color of the light, and the time it took the

DNA to pass through the capillary, a series of computer programs determines which alleles are present at each locus. The position of the peaks on the graph indicates how long it took the allele to pass through the capillary, which indicates the length of the underlying DNA fragment. From this data, the computer program infers which allele is represented and generates the appropriate label" (Understanding the Lab Report, 2004). The height of the peaks in the electropherogram is equivalent to the amount of DNA present at that position. RFU (relative fluorescent units) are the units utilized in measuring the height of the peaks, and reflects the intensity of the fluorescent light, which was found by the camera.

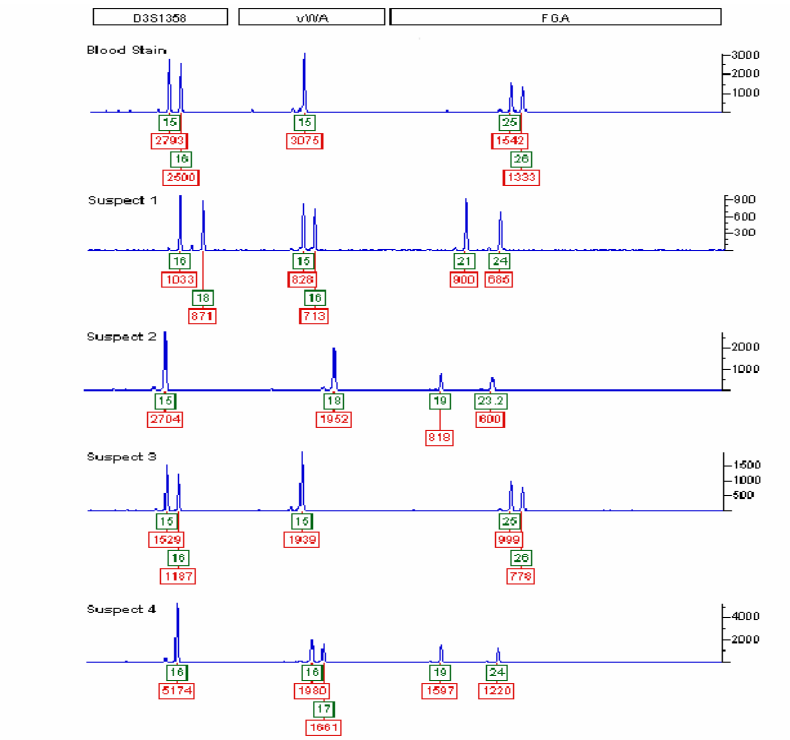



**Figure 16:** "Electropherograms Showing the Results of Profiler Plus Analysis of Five Samples at Three Loci (D3S1358, vWa and FGA). Which suspect is a possible source of the blood? Green boxes immediately below the peaks label the name of the alleles present, while red boxes below indicate their heights in RFUs" (Understand the Lab Report, 2004)

When alleles are compared, if they have the same height and location, they are assumed to originate from the same individual. However, peak height imbalances can occur, causing misinterpretation of data. If a sample contains more than one individuals DNA, it is called a mixture (Sullivan, Personal Interview, 2004). Sometimes the semen and vaginal fluids will be mixed causing discrepancies in the results. The ability to distinguish between fluids will vary between cases because it is dependent upon the quality and quantity of the sample. A mixture can cause more than two alleles to appear at a single locus. Many labs go under the assumption that the taller peaks (larger quantities of DNA) represent the primary contributor, however this can be misleading and may be only an assumption. The technician needs to determine which

allele belongs to which individual, however their interpretation is not always correct (Understanding Lab Report, 2004).

Degradation of the sample is also a contributing factor to misinterpretation. The next chapter will discuss how to prevent degradation; however if it does occur, the height of the peaks on the electropherogram decrease in a downward slope across the graph, as seen in the example in Figure-15.

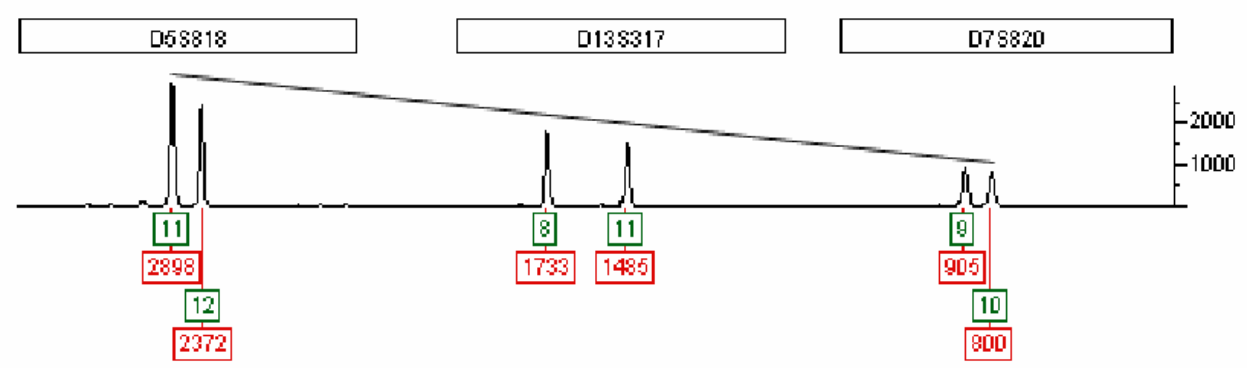



**Figure 17:** The progressively smaller peak heights in this sample from left to right are indicative of partial DNA degradation. (Understanding the Lab Report, 2004)

This occurs because degradation has more of an affect on the longer sequences of DNA, which are located on the right side of the graph. Some of the peaks will be too short to analyze causing the technician to simply guess about the presence of the authenticity of the peak.

Allelic dropout is another source of misinterpretation (Figure-16). If the sample is of low quality, or the sample is degraded, one of the alleles may not appear on the graph as a peak high enough for the analyzer to detect its presence. If everything else in the profile matches the sample from the crime scene, the technician could report possible dropout linking the sample to the crime scene. If the technician does not report a dropout, the lawyer could argue that the sample does not match with any certainty.

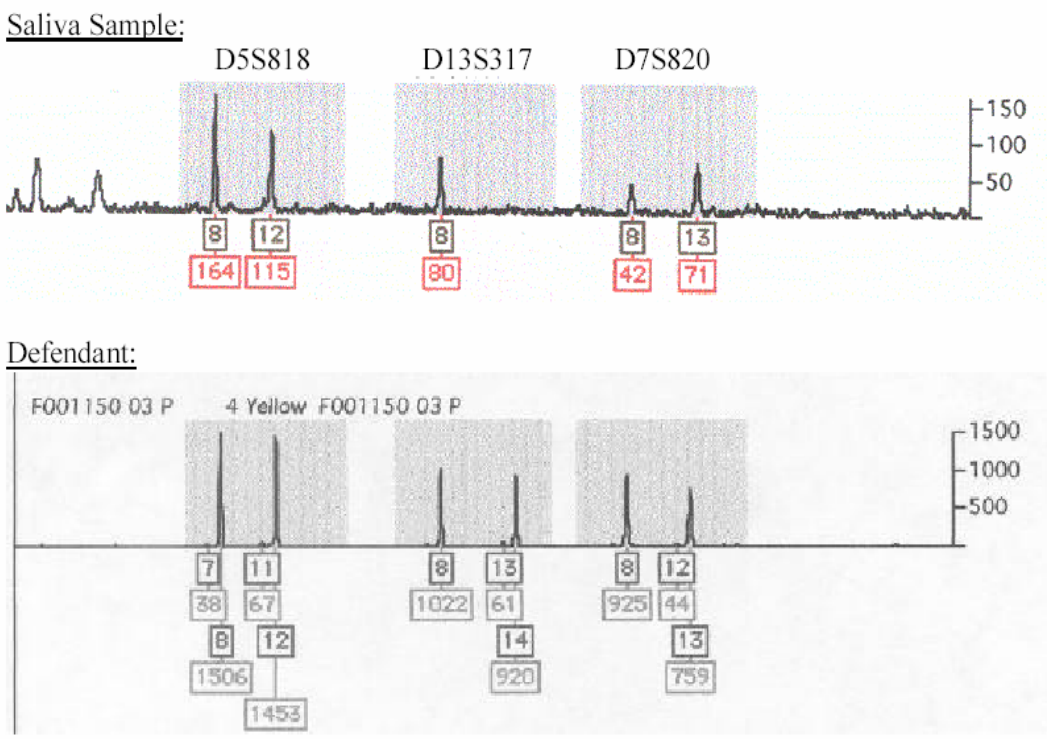



**Figure 18:** Allelic dropout or the wrong man? Note the absence of the second allele at locus D13S317. (Understanding the Lab Report, 2004)

There are times that peaks occur where they don't belong on the graph. "Stutter peaks" usually appear before an actual peak and are usually very small. It is sometimes difficult to interpret a stutter peak from a peak that is present due to a mixture in the sample. "Noise" can form "flashes" on the graph, which can be large enough to be confused with a real peak. Air bubbles, or contamination of the sample usually causes noise. The technician uses their best guess to determine whether it is a legitimate peak or just "noise" in the sample. "Pull-ups" are caused by a malfunction in the equipment. The equipment mixes up the detection of the dye color. For example, a locus labeled with blue dye could be interpreted as a yellow signal, thus causing false peaks at the yellow loci. Again, the technician needs to determine if the peak is valid. "Spikes" are very narrow peaks and they can occur because of air bubbles in the sample or changes in the

voltage when the electric current is applied to the sample. They can be identified because they usually occur in the same position in all four colors. “Blobs” are false peaks mainly the result from the colored dye detaching from the DNA fragment. These peaks are usually wider than real peaks and are usually seen in only one color. Individual labs do have certain thresholds for the heights of their peaks to be considered “real”. However, these thresholds can vary between labs and even vary between individual technicians and cases causing discrepancies and misinterpretation of the data. Figure-17 below shows examples of some false peaks:

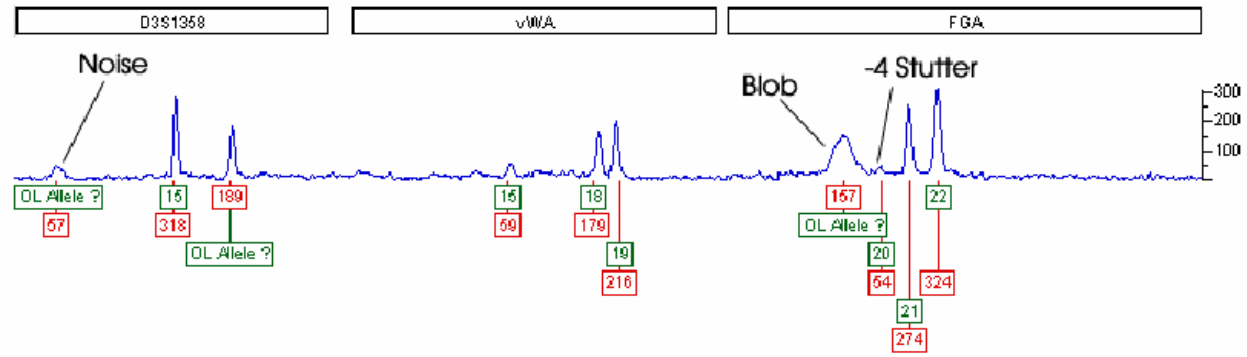



**Figure 19:** Blobs and others false peaks may hide the presence of true alleles. (Understanding Lab Report, 2004).

When analyzing the results of a DNA profile, there can be one of 3 conclusions drawn; inclusion, exclusion or inconclusive. Inclusion is when the suspect’s DNA profile forms a match to the profile formed from the evidence gathered at the crime scene. The validly of the conclusion results is determined from the number of loci examined, and the probability rate of that profile being replicated in the entire population. Exclusion occurs when the profile from the suspect does not coincide with the profile formed from the crime scene evidence. By excluding an individual, this does not indicate they are innocent, it simply adds to the entire caseload of evidence. For example, a rapist could have used a condom during the crime and he would be excluded in the DNA test because his semen was not found. However, other evidence could

point to him as the perpetrator. Inconclusive report means that there could have been a low quantity of DNA being tested, or the sample was a mixture leading to faulty results. Thus, the suspect could not be included or excluded (Understanding DNA Evidence, 2004).

## APPLICATIONS OF DNA FINGERPRINTING

*Murder or Rape Applications*

DNA Fingerprinting has evolved to take part in a plethora of different applications. The first, and most well known application is forensics, either murder or rape cases. Suppose an officer arrived at a crime scene of a murder, and the only evidence that could be found was a bloodstain on the carpet from the suspect. If you were put on a jury and told, based on scientific evidence using DNA Fingerprinting, that this suspect was the murderer, would you convict him? In the beginning stages of its existence, courts would not always allow the evidence to be admitted, based on the fact it hadn't been completely proven to be accurate. As you'll see in a following chapter, many court cases and laws continued to set precedence, and DNA Fingerprinting eventually became more and more accessible and less controversial in the court of law. The FBI began to establish DNA Fingerprinting as means of connection to crimes early on. Between the years of 1989 and 1996, they used the technique in about 10,000 sexual assault cases (Establishing Innocence, 2004). In those cases, 2,000 prime suspects were proven innocent based on the results of the DNA profiling

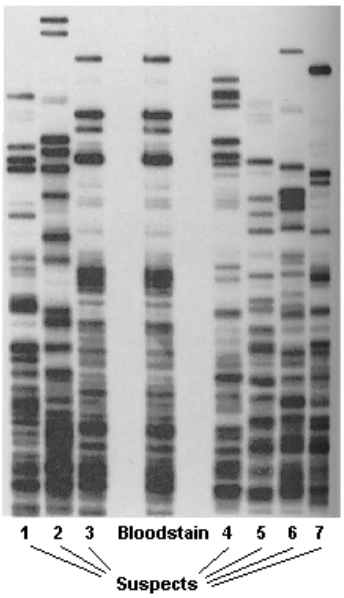



**Figure 20 – Evidence that is now used thanks to DNA Fingerprinting techniques. (Huskey, 1995)**

(Establishing Innocence, 2004). In Figure 8, you can see an example of an RFLP fingerprint analysis of 7 suspects relative to a crime scene bloodstain. It seems very clear that Suspect 3 was present at the crime scene. It is also true to say that without this method of testing, those 2,000 innocent prime suspects could most likely have faced conviction (Establishing Innocence, 2004). Many cases from decades ago have been recently reopened and their verdicts changed by admitting new DNA evidence. Many of these cases will be explained in depth in a later chapter.

*Paternity Testing*

Paternity testing has also become a very commonly used application of DNA Fingerprinting. When a child is born, the child inherits 23 chromosomes from the mother, and 23 chromosomes from the father. Therefore, when a DNA test is done, "the visible band pattern of the child is unique. Half matches the mother and half matches the father". A DNA paternity test is the most accurate form of testing possible to determine parentage. If the patterns do not match on two or more probes, then the father is 100% excluded, which means he is without question not the father. If a match is present on every DNA probe, the probability of paternity is 99.9% or greater. In a Court of Law, 99% is accepted as proof of paternity (DNA Diagnostic Center, 2004). In Figure 9, you can see that the mother (blue bands) and father (orange bands) both have separate DNA patterns. Now look at daughter 1. You can see that some of the mothers same blue bands are inherited as well as some of the fathers orange bands. Daughter 2 has the mothers blue bands, but not the orange band, instead

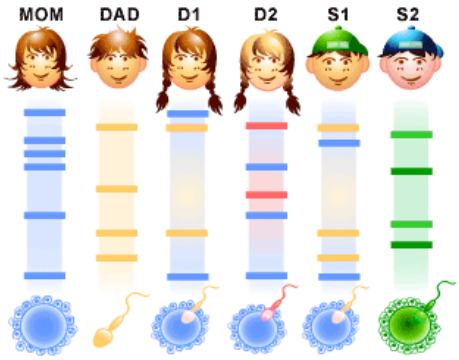



**Figure 21 – A DNA Paternity Test showing results after being tested using DNA Fingerprints. (Antler, 2004)**

red bands. This proves that the father of daughter 1 is not the father of daughter 2, but a father from the mother's previous marriage. Son 1 is a child of both of the shown parents as well. Son 2, however, is adopted because he doesn't have any of the parents DNA.

One of the most famous paternity cases involved Thomas Jefferson, the third President of the United States, who in 1802, was accused of impregnating Sally Hemings, a slave at the Jefferson estate. No verification or conviction was ever brought about however. The story was sustained throughout the decades, and in 1998, Dr. Eugene Foster and a team of geneticists, conducted DNA tests to prove the accuracy of these accusations. The test results proved that it was indeed a Jefferson who fathered Sally Heming's son, Eston. At the time of the child's birth, there where approximately twenty-five Jefferson's living in Virginia, all of who carried the chromosome that would match the child's. The verdict proved that it was probable, yet not conclusive, that Thomas Jefferson was the father of Eston Hemings (The Plantation, 2004).

*MIA Soldiers*

Another application of DNA Fingerprinting is for historical clarifications and identifications. Over the last decade, hundreds of missing in action (MIA) soldiers, that were never identified, have been identified through the use of DNA Fingerprinting. How is this possible? Relatives of unidentified soldiers who are willing to use a sample kit to submit a blood and saliva sample to the laboratory, where "forensic anthropologists, forensic dentists and equipment recovery specialists who work at the U.S. Army Central Identification Laboratory" (Tippy, 2004) analyze the sample, and are able to match DNA with MIAs, with as little as a bone fragment recovered from a killed soldier (Tippy, 2004).

"The most famous American memorial for unidentified soldiers killed in combat is located at Arlington National Cemetery in Arlington, Virginia" (Unknown Soldier). It has inherited the name as the Tomb of the Unknown Soldier (Unknown Soldier, 2004). In 1998, the remains of soldiers killed during the Vietnam War, which occurred between 1959 and 1975, were removed for hope of identification. After testing was completed, the remains were identified as First Lieutenant Michael Blassie, an Air Force pilot who was killed when his helicopter was shot down in Vietnam in 1972. After the positive identification, his family buried the remains in Saint Louis, near Blassie's childhood home. In 1999, Pentagon officials made the decision that based on technological advances in this DNA profiling and the ability to identify MIA's, no other soldier from the Vietnam War would be buried in the Tomb of the Unknown Soldier (Unknown Soldier, 2004).

*Inherited Disorder Testing*

DNA Fingerprinting is also capable of determining inherited disorders in adults, children, and unborn babies. A historical example to prove this involves our sixteenth president and the issue of Marfan's Syndrome (Betsch, 1994), a disease which is characterized by unusually long and nimble limbs. As you know, President Lincoln was abnormally tall during his lifetime which lasted from 1809 to 1865. Could DNA evidence from Lincoln be recovered that would prove this disease existed? According to David F. Betsch, "the technology is so powerful that even the blood-stained clothing from Abraham Lincoln has been analyzed for evidence of a genetic disorder called Marfan's Syndrome" (Betsch, 1994). Although many attempts have been made to clearly identify this disorder in Abraham Lincoln, no conclusive result has been reached due to damaging of historical artifacts. This genetic testing allows people to "test for some

anomaly that flags a disease or disorder" (Inherited Disorders, 2002). "Much of the current excitement in gene testing, however, centers on predictive gene testing: tests that identify people who are at risk of getting a disease, before any symptoms appear" (Inherited Disorders, 2002).

*Personal Identification*

DNA fingerprinting has seen tremendous growth over the last couple of decades, but is there more on the horizon for new applications? One of the main challenges in the field is to create an ideal system of personal identification that would be "indelible, unalterable and –unlike an ID card –part of the individual" (Marx, 1998). The scientific goal is to generate this as a national standard, where every citizen is included within the database, and to prove that there are no drawbacks, such as misuse of medical information during the identification process, or any cautious steps that are taken before DNA fingerprinting becomes the standard (Marx, 1998). In 2002, Alec Jeffreys, the inventor of DNA fingerprinting, put forth his theory on the DNA databases. He believed that "every citizen's genetic information should be stored on the UK national register" (O'Brien, 2002). Jeffreys went on to say that "if we're all on the database, we're all in exactly the same boat – the issue of discrimination disappears" (O'Brien, 2002). The advantages and disadvantages of DNA databases, as well as its progress in the modern day society, will be explained in more depth in a following chapter.

Another challenge in this growing field is to simplify and hasten the procedure of running a DNA fingerprint which will revolutionize forensics. In Australia, researchers have claimed that they have a technique that is twice as fast as conventional DNA technology and is capable of cranking out 1,000 samples a day, where most laboratories manage only a few dozen (Dayton, 2002). "Moreover, profiles are derived from DNA contained in a single cell — existing

procedures require at least 500 cells" (Dayton, 2002). Team leader, Ian Findlay, of the Australian Genome Research Facility, said of his team's recent discoveries, that "it's such a breakthrough we're waiting for the rest of the world to catch up." (Dayton, 2002)

So this is where the debate begins. In the chapters to follow, forensics of DNA, landmark DNA court cases, and DNA databases will be examined. Also issues such as the validity of these tests, and the use within the courtroom and why some courts still will not completely trust DNA evidence in the courtroom.

# CHAPTER 2: DNA FORENSICS

Morgan Reynolds

Forensic DNA analysis has been admitted into United States courtrooms since 1987. The common question being answered is "Who is the source of this biological material?" (Inman and Rudin, 1997). DNA profiling was first used in the United States courtroom in the case of Tommie Lee Andrews. (He was suspected of serial rape, therefore, the investigators wanted to link his DNA to the DNA of the 23 victims. Because of the DNA analysis technology and the CODIS system, Andrews was linked to the series of rapes, and sentenced to 115 years (Ramsland, 2004). Profiling has helped to acquit or convict suspects of rape and/or murder however it is used in less than one percent of all criminal cases (Genetic Science Learning Center, 2004). DNA profiling does not claim to be an absolute identification, but may be very strong evidence and is considered to be just one part of the entire case. DNA profiling is used primarily in sexual assault cases. Prior to DNA testing, labs were limited regarding the amount of genetic information that could be obtained from semen. Now with DNA testing, the genetic content of the sperm itself can be examined. The RFLP method of analysis needs a considerable amount of material for testing and may be unable to determine anything from a sample that has any degradation. The PCR analysis needs only a minute amount of sample because it replicates the DNA and can work even with degraded samples (Sullivan Personal Interview 2004).

In the case of a rape investigation four profiles are formed (Figure-10): the first is a DNA profile from the blood of the victim, the second DNA profile is formed from the blood of the defendant, thirdly a vaginal swab is taken, and the female and male fractions are separated and profiled separately. The victim's blood profile and the vaginal swab profile should match alleles. The defendant's blood sample profile should match the male fraction profile in order to consider him as a suspect. It is important to remember that DNA profiling does not incriminate a suspect it can only exclude suspects.

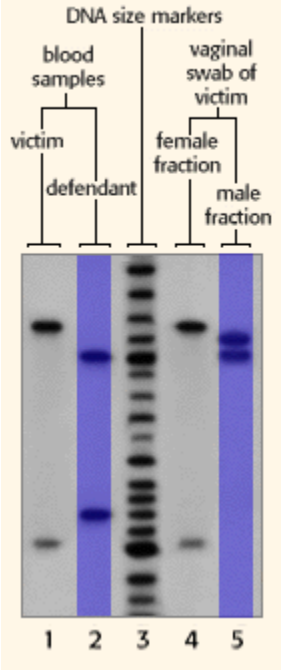



**Figure 22: Example of Rape Investigation.** Exclusion of male suspect, because lane 5 (male fraction) does not match with lane 2 (Defendant). (DNA Forensics Problem Set 1, 2004)

Once a suspect's blood is matched with the male fraction, statistics come into play. The technician then attempts to determine the frequency that this DNA profile could appear in the general human population. It is important to note that DNA profiles will be identical for identical twins. Also if a Black suspect is being profiled, they are more likely to be a closer match to other Black people's profile than to a person of a different race. The individual loci have their own frequency statistics. For example, if the technician is investigating a 5-locus DNA profile and the frequency of each individual loci were .01, .02, .06, .10, .03, these frequencies would be multiplied together to obtain $3.6 \times 10^{-8}$ = one in 36 billion. The technician would then report that 1 in every 36 billion people could have this specific DNA profile. The

jury would use this probability statistic to determine if this match occurred by chance. Not every case will result in a low probability of the profile being duplicated. It depends upon the number of loci tested; the more loci tested the lower the probability rate would be.

Another helpful tool in incrimination is a system known as CODIS (Combined DNA Index System). This system intertwines computer technology with science. CODIS is a database where convicted felons' DNA profiles are entered. In the state of Massachusetts it was recently determined that all felons would be entered into the CODIS system after just one conviction. Prior to this ruling, the criminal was included in the system only after being convicted of a violent felony (Sullivan, Personal Interview 2004). The system has two indexes. The forensic index contains DNA profiles from biological evidence left at the crime scenes, and the offender index contains the DNA profiles of individuals convicted of violent crimes. This enables federal, state, and local crime labs to exchange information electronically to help link crimes. Once a felon's profile is completed, it can be run through the CODIS system to determine if the individual can be matched to other crimes with other victims anywhere in the United States. To demonstrate the power of DNA profiling in forensics, the following example is presented:

> July 1999: The FBI Laboratory's DNA Analysis through its Unknown Subject Sexual Assault Program with the Washington, D.C Police Department received ten sexual assault cases that the National DNA Index System matched to three sexual assaults in Jacksonville FL. The Jacksonville cases occurred in March, April and September of 1998. CODIS had previously linked the Jacksonville cases to one another in May 1999. DNA evidence was crucial in these three cases because none of the victims were able to describe the offender, and no other physical evidence was left at the crime scenes. Five of the ten Washington, D.C cases had already been linked together using CODIS before the national hit with Florida. The other D.C. cases were identified at later dates. In early July 1999, Leon Dundas, who is now deceased, was identified through DNA analysis as the perpetrator of thirteen assaults (The FBI's DNA Databasing Initiatives, 2000).

Five decades ago, Watson and Crick discovered the secrets of DNA structure. The study of genetics assists scientists and doctors to diagnose and treat diseases, develop new plants and vegetables and even to produce clones of adult sheep. The use of DNA evidence in forensics is also a remarkable technological advance however it is just one of many types of evidence used to help solve a crime. It is important to examine other clues such as motive, eyewitness description, fingerprints, weapon, or additional evidence linking a suspect to the crime scene. Forensic scientists currently scan 13 DNA regions (Figure-11) that vary from person to person and use this analysis to create a DNA profile of that individual.

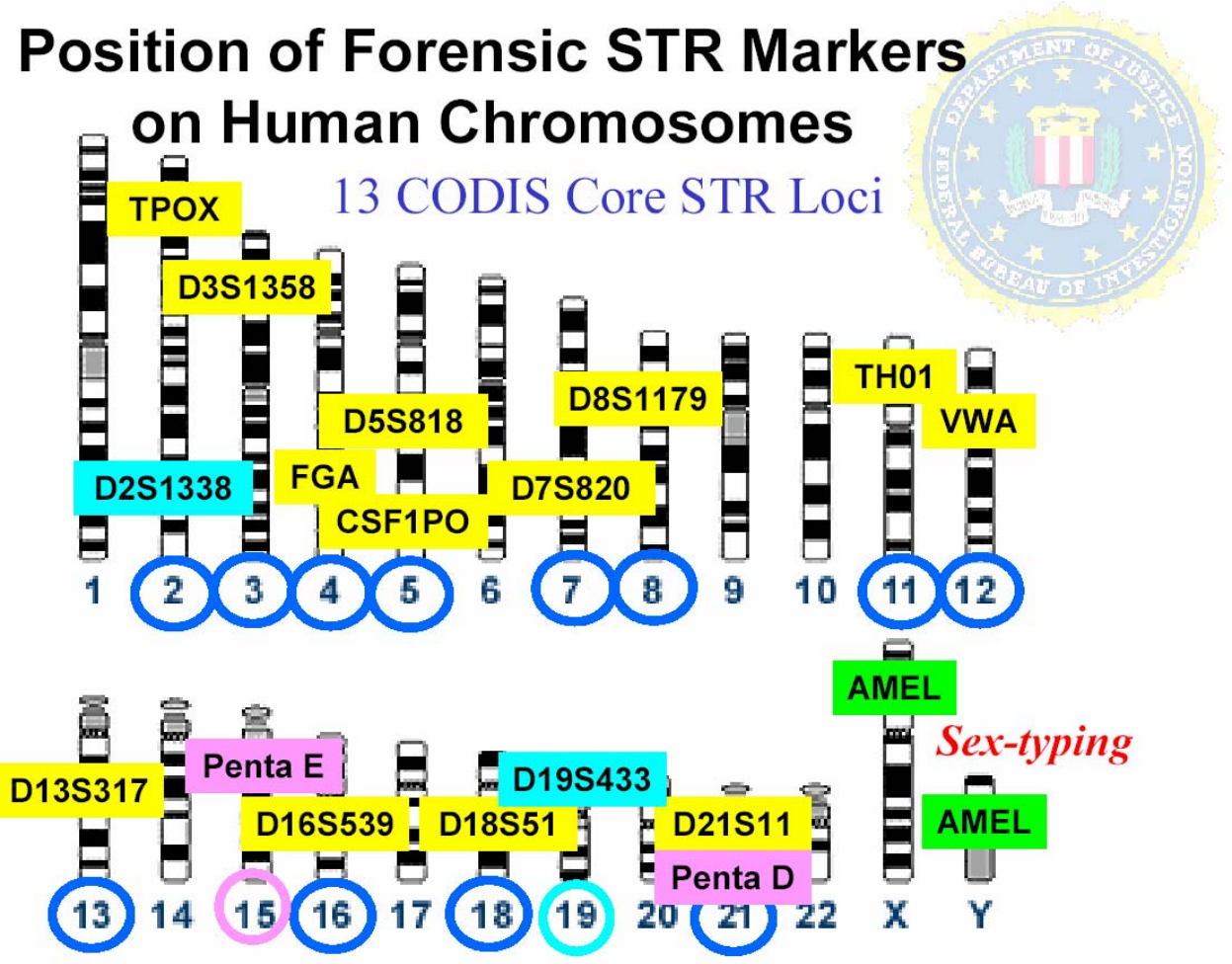



**Figure 23:** 13 Core Loci
(Kline, 2001)

Some examples of DNA uses for forensic identification include:

- Identify potential suspects whose DNA may match evidence left at crime scenes
- Exonerate persons wrongly accused of crimes
- Identify crime and catastrophe victims
- Establish paternity and other family relationships
- Identify endangered and protected species as an aid to wildlife officials (could be used for prosecuting poachers)
- Detect bacteria and other organisms that may pollute air, water, soil and food
- Match organ donors with recipients in transplant programs
- Determine pedigree for see or livestock breeds
- Authenticate consumables such as caviar and wine

(The FBI's DNA Databasing Initiatives, 2000)

DNA profiling was brought into the public eye in 1985 (How DNA Evidence Works, 2004). Since that time DNA profiling's power and prestige has escalated. At its break-through, there were several misconceptions about its use and validity. From 1985 to 1995, VNTRs were the main focus of the analysis. However, PCR process was developed shortly after which changed everything. STRs then played a large role with PCR and this was a great improvement because VNTRs were too large to be amplified. Most recently, the FBI decided on 13 specific loci to be tested when forming a profile. DNA profiling was not looked upon favorable at first because the New York Times was raising many questions and skepticism poorly representing the first publication of the study in 1992. The report wanted an "interim ceiling principle" which had two main objectives: first, they wanted to be conservative in terms of favoring the defendant,

and secondly they wanted to show the necessity to classify people by ethnic groups. In 1996 another study was completed by the National Academy of Sciences and had a much more favorable outlook and was able to abandon the ceiling principle. However they were questioning the statistical portion of the DNA profiling. The 1996 report was also emphasizing VNTRs instead of STRs because the STR analysis was still very new in development. Now 50 states have databases. Most have changed to STR analysis. The numbers indicate that about 190,000 felons are entered in the CODIS database and 9,000 in the forensic profile database (Crow, 2004). In a few years the numbers should increase considerably as the process of testing becomes more automated.

Based on the advances thus far in DNA forensics, it is plausible to formulate predictions for future advancements. The testing techniques will improve by allowing more STR loci to be analyzed at once and the testing will become more automated with less subjective interpretation. The capillary electrophoresis technique will require less material providing faster results and smaller analyzers. Some of the analysis may actually take place at the crime scene. PCR analyzers will be miniaturized onto chip-size devices. Not only will these devices be available at the crime scene, they will also be available in physician offices to get results in the office of DNA analysis. This will aid in the diagnosis of infectious or genetic disorders or detect an inherited predisposition to cancer or heart disease in the physician's office. More loci will be tested and therefore there will be more distribution of alleles allowing for even better statistical information. Other technologies may improve regarding separation of suspects DNA from other material in the sample.

Before DNA evidence can be admitted into court as evidence it needs to go through a pretrial hearing. The new technology needs to pass certain tests of acceptability in the scientific

community. The DNA analysis had to prove to be a scientifically sound in method, theory, and interpretation. As time went on and the DNA procedures become more reliable and scientifically accepted, DNA gained increasing acceptance in the courts, however the main challenges were directed at the way samples were interpreted or at the poor handling of the specimen evidence, such as happened in the O.J. Simpson trial. Most of the evidence appeared to incriminate Simpson including; the victims blood in his truck, his shoe prints next to the blood trail, a suicide note written by Simpson when he realized he would be arrested, and a black glove which contained traces of fiber from Goldman's jeans. In addition to all of this seemingly incriminating evidence, the blood found at the crime scene matched Simpson's blood. Numerous labs completed the testing and yielded identical results. However, the blood was not packaged properly, and the blood sample was said to have degraded while it was stored in the lab truck. The defense attorneys basically stated that the blood samples were handled poorly thus the DNA profile should not be trusted in this case. The jury agreed with the defense and rendered a verdict of "Not Guilty" (Ramsland, 2004).

Today, there are numerous procedures that every reliable crime lab use to elevate any possible discrepancies. Christine Sullivan, investigative DNA analyst of the Massachusetts State Crime Lab, described their procedures for handling the samples. Each sample is sealed with a bar code to identify the specific source of the sample. Each DNA technician has an identification badge. When a technician needs to use the sample for a portion of the testing, they "swipe" their badge which identifies which sample they have in their possession. When they return the sample to storage, their badge is "swiped" again to indicate that they returned the sample. The times are also recorded so there is never any lapse of time when the sample is not accounted for.

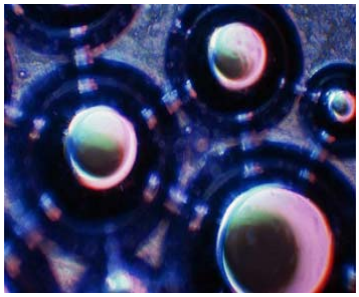
The process of DNA analysis begins at the crime scene. The police officer or investigator needs to visually inspect the scene to determine the items that will be important to collect as evidence to the crime. The most obvious items would be weapons used in the crime. However some of the most powerful DNA evidence may not be as obvious, yet DNA evidence can be collected from almost anything. It is possible that the criminal was very careful not leave any obvious clues or weapons however an experienced and informed investigator would be looking for the less obvious items. Even though a stain cannot be seen, there may be enough cells from the criminal left on various articles to be used for DNA profiling. The following is a list of possible locations of DNA evidence and the source of DNA from the crime scene:

| EVIDENCE | POSSIBLE LOCATION OF DNA ON THE EVIDENCE | SOURCE OF DNA |
|---|---|---|
| **Baseball bat or similar weapon** | **Handle, end** | **Sweat, skin, blood, tissue** |
| **Hat, bandanna, or mask** | **Inside** | **Sweat, hair, dandruff** |
| **Eyeglasses** | **Nose or ear pieces, lens** | **Sweat, skin** |
| **Facial tissue, cotton swab** | **Surface area** | **Mucus, blood, sweat, semen, ear wax** |
| **Dirty laundry** | **Surface area** | **Blood, sweat, semen** |
| **Toothpick** | **Tips** | **Saliva** |
| **Used cigarette** | **Cigarette butt** | **Saliva** |
| **Stamp or envelope** | **Licked area** | **Saliva** |
| **Tape or Ligature** | **Inside/outside surface** | **Skin, Sweat** |
| **Bottle, can, or glass** | **Sides, mouthpiece** | **Saliva, sweat** |
| **Used Condom** | **Inside/outside surface** | **Semen, vaginal or rectal cells** |
| **Blanket, pillow, sheet** | **Surface area** | **Sweat, hair, semen, urine, saliva** |
| **"Through and through" bullet** | **Outside surface** | **Blood, tissue** |
| **Bite mark** | **Person's skin or clothing** | **Saliva** |
| **Fingernail, partial fingernail** | **Scrapings** | **Blood, sweat, tissue** |

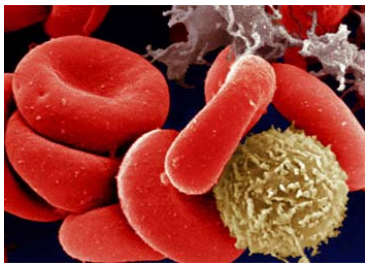
("What Every Law Enforcement Officer Should Know About DNA Evidence",)

The items lists above are examples of items that would possibly contain body fluids (blood, semen, saliva, vaginal fluid, hair with follicle tissue at the root, soft bone, deep muscle

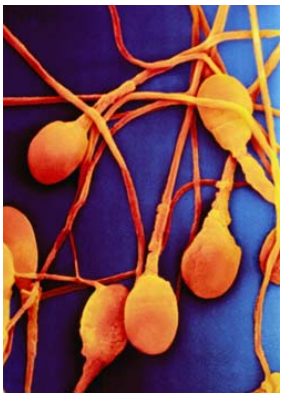
tissue or urine) (some are also shown in Figure 12 below) and thus may be used for the DNA extraction and analysis.



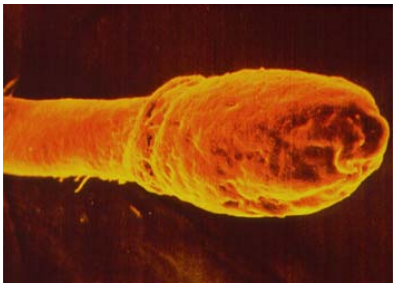
**Saliva**



**Blood**



**Sperm**



**Hair Root**

**Figure 24:** Above pictures are magnified samples biological evidence. (You're on the Case, 2004)

The evidence will be of no use in a court if it becomes totally degraded (the breaking down of DNA into smaller fragments by chemical or physical process) because it was not handled properly or becomes contaminated (the undesirable transfer of material to physical evidence from another source). Therefore, the collection, packaging, storing and shipping of the DNA evidence is critical. An investigator should always use latex gloves when collecting samples, and change gloves when handling different items. Each item must be packaged individually. All stains (blood, semen, etc) must be air-dried or dried with a hair dryer on the

coolest setting. If a large amount of liquid is present, a fan can be utilized. All stains must be sealed in paper envelopes or paper bags. If condoms are located at a sexual assault crime scene, they should be placed in a sterile tube. If a sterile tube is not available, the condom should be air dried and then placed in a layer of paper bags for packaging. It is imperative that the evidence's location and custody of the package is known and accounted for at all times. Thus all packages must be labeled with the case number, item number and date. Also, the initials of the person handling the evidence must be on the seal of the bag. If a stain is located on a surface which cannot be moved (i.e. road or sidewalk) then a photograph must be taken with a ruler to help show the precise location of the stain. Then take the sterile cotton swab and moisten it with distilled water. Take the swab and rub it all over the stain until the stain is completely on the swab. If the stain is quite large, more than one swab can be used to gather the stain. As a control, two additional swabs must be used. The first control swab will be used to swab the area adjacent to the stain, and the second control swab will contain solely the swab itself and the water used on the other swabs. All the swabs must be air dried and then placed in properly labeled and marked envelopes or paper bags. It is imperative that evidence never be placed in plastic bags because they contain moisture, which could lead to degradation. Paper bags allow moisture to escape, helping to prevent degradation. You should never scrape dried stains unless it is on a smooth surface and you are sure you won't lose any of the sample. For example, if a stain was on a wall, a piece of folded paper could be placed underneath the stain and a sterile blade could scrape the stain onto the paper. The paper would then be folded, marked with evidence tape and initialed. Some evidence will be unable to dry such as liquid urine, vomit, organ tissue, bone, etc, thus these substances should be placed in an airtight container. Identification and labeling is the same as previously described. It is important to note that

formaldehyde should never be used in the process of preserving biological evidence because these chemicals degrade the DNA. Blood samples from known living individuals should be extracted and placed in tubes with purple tops, properly identified, placed in a paper container, sealed with evidence tape, stored in a refrigerator. If the sample contains HIV or hepatitis, it must be labeled clearly on the outside of the package. A dead individual's known blood standard should be transferred by syringe into a tube with a purple top. The same procedure for custody and identification is followed in this case as it was above. Once the DNA is collected, it is important that it is stored properly. Cold and dryness are ideal conditions for storage of the biological sample because it reduces the risk of bacterial degradation. Stains, which are dried, should be frozen (-20 degrees Celsius) or refrigerated (4 degrees Celsius) in their respective paper bags. Undried tissues such as bone, liquid urine, etc should be kept in the above conditions. Glass containers should never be used to store samples because glass breaks when frozen. Liquid blood samples should not be frozen but kept at 4 degrees Celsius in their original glass tubes (Successfully Investigating Acquaintance Sexual Assault).

An aged crime scene will have limited amounts of useable DNA evidence. However, when faced with this situation, investigators will utilize dental tissue. Dental tissues have the strength to withstand harsh environmental conditions, which cause degradation and prevent conventional dental identification. Once the DNA is extracted from the dental tissues, it can be compared to the known sample of either stored blood, clothing, cervical smear, etc (Forensic Dentistry Online, 2004).

If the evidence needs to be shipped, the shipping needs to be completed on a business day not on a weekend or holiday to avoid sample storage over a weekend. A case submission letter should be attached to the exterior and addressed to the forensics lab. A dried stain should be cut

out of the garment and shipped on an overnight service and packaged individually in a properly sealed box or envelope and clearly marked. Undried tissue should be placed in a sealed plastic container on dry ice in a Styrofoam container. All liquid or known whole blood samples should be individually packaged in a Styrofoam tube package or separately wrapped in bubble wrap with tape. The individual tubes should be put in cardboard boxes, which contain chips of Styrofoam for extra protection. Blood standards should be shipped at room temperature and blood containing HIV or hepatitis should be shipped with "class 6.2/95 CAN /8-2saf-T-PAK" (Successfully Investigating Acquaintance Sexual Assault).

Evidence can be subjected to a variety of environmental hazards before arriving at the lab of analysis. Most samples are not affected by contaminants; however some samples may show reduced DNA activity, leading to no DNA profile, an incomplete profile, or inconclusive results. Therefore the evidence should be collected and packaged using the procedures that prevent the contamination of the DNA sample. DNA evidence can be contaminated when DNA from another source gets mixed with DNA relevant to the case. This can happen when someone sneezes or coughs over the evidence or touches his/her mouth, nose or other part of the face and then touches the sample to be tested. If the DNA is contaminated with additional DNA, this will get copied in the PCR process and the results will be inconclusive. Investigators and lab personnel always wear gloves, use clean instruments, and avoid touching other objects when handling the evidence. Heat and humidity can also accelerate the degradation of DNA, which is why plastic bags are not used to store the sample.

If possible, it is ideal to freeze samples for long-term storage. This slows the degradation process, however it does not prevent it. It is possible to store a sample at room temperature for over 25 years, however the STR profile of that sample will display some degradation. Dry

biological samples might benefit from being stored in sealed plastic bags in the freezer. If the sample were very small, it would benefit from freezing rather than being maintained at room temperature.

DNA profiling is a very powerful tool when handled and processed correctly. It can be used quickly to eliminate a suspect, and can reduce the chances of a wrong conviction. The evidence however needs to be handled in the manner outlined above, an unbiased forensic laboratory should do the analysis, and lastly, the jurors and judges must receive an accurate and effective report of the evidence (What Every Law Enforcement Officer Should Know About DNA Evidence). Five decades ago, Watson and Crick discovered the secrets of DNA. The study of genetics assists scientists and doctors to diagnose and treat diseases, develop new plants and vegetables and even to produce clones of adult sheep.

# CHAPTER 3: LANDMARK DNA CASES

Brian Steele

"Since the discovery of DNA fingerprinting at the turn of the 20th century, science has assumed an increasingly important and powerful role in the decision making process of our judicial branch" (Biancamano, 1996). In many cases, that role proves to be the deciding factor for the outcome of the trial in both civil and criminal cases. However, "without government standards and a uniform procedure, are these methods valid, reliable, and admissible in court?" (Biancamano, 1996) The technical evidence that is available through DNA fingerprinting has not yet achieved its full potential dominance in the courtroom, and still seeks to become the standard for highly technical evidence.

In this chapter, landmark cases, which set the standards for admitting DNA into the courtroom, as well as the reliability and acceptability of DNA techniques, will be discussed to prove the methods used today are valid and reliable when performed properly. Some of the earlier cases that will be talked about did not actually involve DNA, but what they did accomplish was to set precedence by establishing new rules for admittance of technical evidence, and to set new generally accepted scientific standards in the court of law.

### *Frye v. United States, 1923: General Acceptance of Evidence*

Evidence is very important when the court needs to determine the outcome of a case. But how do you determine whether the evidence is reliable and effective? In 1923, the landmark case of Frye v. United States established "that somewhere in this twilight zone the evidential force of the principle must be recognized, and while courts will go a long way in admitting expert

testimony deduced from a well-recognized scientific principle or discovery, the thing from which the deduction is made must be sufficiently established to have gained general acceptance in the particular field in which it belongs" (Frye v. United States, 1923). In other word's, the “Frye Standard” states that scientific evidence that a court uses to aid the case must have general acceptance in the scientific community to be admitted into the court room.

In this 1923 case, James Alphonzo Frye was arrested for murder in the second degree in Washington, D.C. Frye denied the accusations and the "counsel for the defendant offered an expert witness to testify to the result of a deception test made upon the defendant" (Frye v. United States, 1923), which would prove Frye's innocence. This deception test, otherwise known as a polygraph, or lie detector, had recently been invented. The defense asked the inventor, William Moulton Marston, of Harvard University, to administer the test to Frye. The test found Frye to be “telling the truth” which they thought would prove his innocence. The defense felt that Marston’s reputation had enough standing for this test to be admissible, but the court ruled against this new technique by stating "we (the court) think the systolic blood pressure deception test has not yet gained such standing and scientific recognition among physiological and psychological authorities as would justify the courts in admitting expert testimony deduced from the discovery, development, and experiments thus far made" (Frye v. United States, 1923).

This major landmark case set the standards for what evidence can be used, and what evidence lacks general acceptance in the court of law. Over the next several decades, many cases refer back to this Frye Standard when they face the issue of admitting new scientific techniques and technical evidence into the court. The Frye Standard will eventually play a major role when DNA Fingerprinting finds itself as a key piece of evidence (see below).

***Federal Rules of Evidence 702 (Rule 702), 1975: Expert Witnesses and Reliable Techniques***

In 1975, Congress enacted the Federal Rules of Evidence, or Rule 702, which would provide a more stable law to replace the rather vague and stringent Frye Standard. Rule 702, with regard to the admissibility of scientific evidence, states:

> "If scientific, technical, or other specialized knowledge will assist the trier of fact to understand the evidence or to determine a fact in issue, a witness qualified as an expert by knowledge, skill, experience, training, or education, may testify thereto in the form of an opinion or otherwise, if (1) the testimony is based upon sufficient facts or data, (2) the testimony is the product of reliable principles and methods, and (3) the witness has applied the principles and methods reliably to the facts of the case" (New Technologies, 2004)

"In other words, if a jury would find the testimony of one with specialized knowledge to be helpful, a court may admit it, even if the technique does not have a well proven general acceptance in the scientific community. Following the enactment of Rule 702, many courts rejected the *Frye* standard of "general acceptance" in favor of the more liberal "helpfulness" standard of Rule 702" (New Technologies, 2004).

Rule 702, as you will soon find out, will be a key standard for the rise of DNA fingerprinting techniques in the mid 1980s because if the Frye Standard had remained, DNA fingerprinting would not have been allowed in the courtroom since its use was not generally accepted at that time, nor would it have evolved to what it is today.

***Colin Pitchfork, 1986: The Black Pad Killer***

Following Alec Jeffreys discovery of the technique now known as DNA fingerprinting in 1984, many cases have been overturned or solved due to the DNA evidence. The world's first DNA fingerprinting conviction took place following the death of two rape victims, one in 1983, and another in 1987, in a small village of Narborough, located in England (Autopsy, 2004). In 1983, Lynda Mann, "just fifteen years old, was discovered along a shady footpath, savagely raped and strangled" (Batt, 1999). The police were unable to gain any leads, and of the 150 men in the town, none were deemed the prime suspect. There was, however, a very important piece of evidence that was recovered. A minute sample of semen from the rapist was uplifted and stored, and would later come in handy. In 1987, another rape took place in the same region and again, the victim was a fifteen year old named Dawn Ashforth. She had also "been brutally raped and strangled" (Batt, 1999). Police then realized that it must have been the work of the same man. One of the policemen in the town had read an article on DNA fingerprinting, a new forensic technique at that time. Police had a possible suspect in custody named John Buckland, a seventeen-year-old dishwasher, who had admitted to the murder of Dawn, but proclaimed he did not rape and kill Linda. Police and investigators decided to test the new technique, and in doing so, found that Buckland's DNA did not match the DNA of the semen found at either crime scene. Buckland was pronounced innocent of both murders, while police were left with no suspect. If this new forensic technique could prove innocence, they decided it could certainly prove guilt. DNA Fingerprints were done on every male between the age of 13 and 30 in the region (Batt, 1999). The problem was, no match came back positive and the technique was criticized immediately. However, a short time later, the case, as well as the pivotal start of DNA Fingerprinting occurred. "A woman who worked in a local bakery told investigators that while

drinking in a pub with some co-workers, one of them claimed he'd taken the blood test for another man" (Autopsy, 2004). That was it! The man taking the test was Ian Kelly, and when questioned by police, turned over the name of Colin Pitchfork. Pitchfork avoided the test, and almost got away with the murders. "On January 22, 1988, Pitchfork plead guilty, the case never went to court, and he became the first murderer caught using DNA evidence" (Autopsy, 2004). Pitchfork was known as "The Black Pad Killer" because the two victims were found near a footpath known as the Black Pad (Autopsy, 2004). The case was not only a breakthrough for proving someone's guilt, but also it was the first case in which a suspect was cleared due to the advances in the field of DNA fingerprinting.

**Figure 25:** Colin Pitchfork, the first man to be convicted of a murder using DNA Fingerprinting (Autopsy, 2004).

***Andrews v Florida, 1988: DNA in the U.S. Court Room!***

Now that DNA Fingerprinting was gaining power and validity overseas in England, it was time to put it to the test in America. In 1986, in Orlando, Florida, following a large number of rapes, Tommy Lee Andrews was caught, charged, and convicted with one of those victims rape and sentenced to twenty-two years in prison (Andrews v. State of Florida, 1988). The question was, how can Andrews be related to the other cases when there is a lack of valuable evidence? Investigators wanted to try a DNA match with DNA samples from all of the victims and compare them to Andrews. Once the samples were taken and tested at Lifecodes lab, results proved the rapes were committed by one man, so he was charged every single rape. Now, how could the prosecution use this evidence to pin all the rape charges on Andrews? DNA testing, being a relatively new scientific technique was susceptible to validity accusations as well as its

accuracy. The court required a pretrial hearing which would determine if the evidence could be admitted into the case, which if it where, would make the case, but if not, Andrews would get away on just one count of rape. The pretrial was "long and complex, but finally the judge allowed the evidence into the case" (Ramsland, 2003). This was an enormous step for DNA Fingerprinting setting the legal precedence that such evidence can be admitted in U.S. courts. Now that the evidence was admitted, Andrews was doomed because the new case now found him guilty and increased his prison sentence to one hundred and fifteen years (Ramsland, 2003).

The Andrew's case was the first of many U.S. cases solved using DNA Fingerprinting techniques, but as you will see in the next case that it would still take time for the advancement to be completely agreed upon throughout the justice system.

***Castro v New York, 1989: The Three Pronged Test***

Despite the success of the admittance of DNA fingerprinting evidence in the Andrew's case, the technique was still in its early stages and its position in the courtroom was still undetermined. In the case of People v. Castro of New York, the technology was put under heavy scrutiny. Joseph Castro was charged with murdering his neighbor as well as her two-year-old daughter (Ramsland, 2003). The New York Supreme Court was interested in dissecting the technique of DNA fingerprinting to establish current protocols, as well as learn the detailed steps in which these tests are done to ensure complete satisfaction and competence. They came up with a three-prong test that they felt would clearly identify acceptable evidence for admittance.

The test included:

1. Is there a generally accepted scientific theory stating that DNA testing can be reliable?

2. Do techniques exist that can produce reliable DNA results?

3. Did the testing lab perform these accepted DNA tests in this trial?

In the Castro Case, the evidence put into question was a bloodstain on Castro's watch which would be analyzed to match the victim. However, the court ruled that DNA tests could only prove the blood was his own, not that from the victim. The case ended when Castro confessed to the murders in late 1989. DNA evidence would not have been allowed into this case because the third criterion was not met because Lifecodes failed to adhere to current practices during its testing of the sample (People v Castro, 1989).

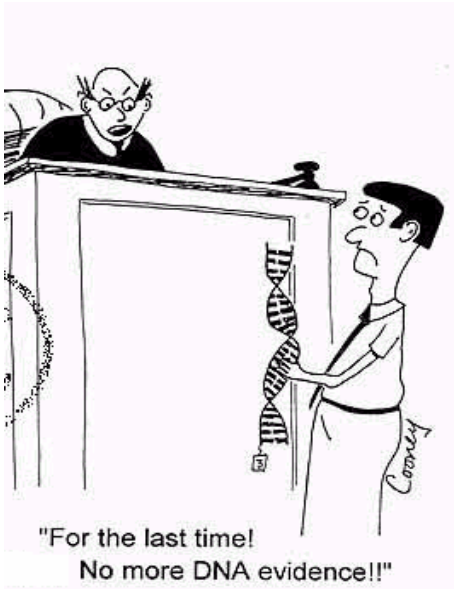



**Figure 26:** In the early stages, DNA Fingerprinting was not completely trusted in the court room. The first case to critically evaluate DNA testing was People v. Castro. (DNA Cartoons, 2004)

Following this case, the FBI stepped in and developed a standard known as the Technical Working Group on DNA Analysis Methods, or TWGDAM (Miller, 2004). "TWGDAM is comprised of scientists from industry, forensic laboratories, and the academic community, who meet several times each year. In its effort to build consensus, and to define guidelines for DNA laboratories, quality assurance guidelines for forensic DNA testing and guidelines for DNA proficiency testing were subsequently published by TWGDAM. Adherence to these guidelines is now often considered by courts to be a major factor in determining the admissibility of DNA test results as forensic evidence. And, in April 1991, TWGDAM revised and expanded these guidelines in anticipation of the next generation of DNA

technology" (Miller, 2004). People v. Castro was the first case where DNA practices were questioned, and methods were standardized.

***Two Bulls v US, 1990: The Five Pronged Test***

Following the Castro case, rules and regulations became the roadblock for the growth of admittance of DNA fingerprinting evidence. However, in 1990, Two Bulls v US would take many of the previous case rulings and pull them all together to make a clearer and more definitive ruling on this growing topic.

Two Bulls v US, which involved a Sioux Indian by the name of Lynette Two Bulls (Two Bulls v. United States, 1990), took many of the preceding cases and interpreted them in such a way that would create a standard and landmark for which all future cases involving DNA could refer to.

While configuring this new standard, the Castro three-prong test was considered too strict, while the Frye Standard as well as Rule 702 was considered correct. The new test formed by this case was now a five-prong test that would be performed at a pre-trial hearing whenever evidence includes DNA. The test is as follows:

1. Is DNA testing generally accepted?
2. Is the testing procedure used here generally accepted?
3. Was the test performed correctly here?
4. Is the evidence more prejudicial than probative, and if so, disallow it.
5. Is the statistics of the DNA match more prejudicial than probative?

   If so disallow it.

The first prong originated from the Frye test, and still questions the general acceptance of DNA theory, which at this time has gained general acceptance. Prongs two and three come from

the People v. Castro case, but in this situation, are not as strict, and simply question the acceptance and testing techniques to solidify the testing as scientifically correct in this specific case. The fourth and fifth prongs were brought about to prove that evidence and test statistics are not prejudicial, or biased, but rather that they are probative and able to prove something useful to the court.

***Miles v Illinois, 1991: TWGDAM Upheld***

In 1991, Reggie Miles, an Afro-American, was charged with rape. Investigators had recovered DNA evidence from a bed sheet at the scene of the crime, and sent it to be tested at Cellmark. Right from the start, the evidence was a long shot because Cellmark had previously, on many occasions, failed to comply with testing standards required by TWGDAM leading to rejection in cases.

In this case however, it was concluded that Cellmark had correctly performed the testing, and produced accurate statistics to uphold the conviction and swing the tide back in favor of DNA fingerprinting. The test results showed that the probability of someone other than Miles at the crime scene was one in 300,000 (Agabin, 2000).

The ruling in this case strengthened TWGDAM as well as the five-prong test set up in Two Bulls v. US. This case also changed the court's view on DNA evidence proving it to be reliable and competent which gave it a stronger edge for acceptance for future cases.

***Paul Eugene Robinson, 2003: ONLY DNA Evidence Used!***

"District Attorney Jan Scully announced today that Paul Eugene Robinson was sentenced to the maximum term of 65 years in state prison for five counts of sexual assault occurring in August 1994" (Scully, 2003). This press release occurred on June 26, 2003, but its significance goes far beyond a routine sexual assault case. It was the first case where a suspect was convicted solely on DNA evidence.

In 1993 and 1994, a series of sexual assaults occurred in the Cal Expo area of California. Following six years of investigating, a "John Doe" warrant was filed on "the individual" belonging to the victim's semen samples. California had a criminal database set up, and with a "cold hit" to the original rape evidence, Robinson's DNA was positively matched. In 2003, Robinson was convicted on five counts of sexual assault (Scully, 2003). "This is all new territory, but hopefully in 10 years, it will be an everyday thing" (Associated Press, 2000). This was said by Sacramento Police Detective Peter Willover following the capture of the rapist known only as the "Second Floor Rapist." If the investigators did not issue the DNA warrant, Robinson would have never been caught, and the rape case would have been closed due to the statute of limitations law, which allows a case to stay open only for six years (Associated Press, 2000). In 2000, a "$50 million grant from the state Office of Criminal Justice Planning was distributed to police departments around the state to do DNA testing on old rape cases" (Associated Press, 2000).

Altogether, these landmark cases prove that DNA Fingerprinting has taken enormous steps forward in not only the scientific community, but also in the judicial sector of our society. These cases all involved setting precedence's for admitting complex technical evidence in the courtroom, however the public is likely not aware of any of them.

# CHAPTER 4: DNA DATABASES

Kerri Mongelli

The existence of DNA databases is a subject of much contention. Some people feel that these databases are a great help to society giving new hope to forensic cases that would otherwise have no leads. Others feel they are an invasion of privacy and that these databases should consist only of samples collected from violent crime offenders. Still others do not even understand what they are. This chapter will examine what DNA databases are, why they are needed, and why the public is often against them.

## The CODIS Database

The world's largest DNA database is a U.S. system called the Combined DNA Index System, or CODIS. "CODIS blends computer and DNA technologies into an effective tool for fighting violent crimes" (Brown and Niezgoda, 1995). Currently CODIS consists of two separate indexes to help create leads in crimes where forensic evidence has been collected. One of the indexes, the Convicted Offender Index, contains the forensic profiles of people convicted of felony sex acts and other violent crimes. The other index, the Forensic Index, contains profiles taken from crime scenes. This latter index is used primarily to link crimes. There is also a population file, a database of anonymous profiles used to determine the statistical significance of a match.

The CODIS database system is a three-tiered system comprised of local, state, and national levels (CODIS Combined DNA Index System). The local system, also known as LDIS (Local DNA Index System), is installed at individual crime laboratories and is usually run by

police departments, sheriff's offices, or state police agencies. Local DNA forensic examiners use CODIS software to enter DNA profiles and submit them to one of the indexes (The Convicted Offender or Forensic indexes) or to the population file. The title of custodian is bestowed upon the person in charge of the sharing of the DNA data with other tiers of the CODIS program. All profiles are created at the local level and then have the ability to be shared with the state and, in turn, the national indexes. Once profiles are submitted, they can be searched against other subject profiles in the index or can be forwarded by the custodian to the state level where it can be shared by all the local CODIS programs within the state.

At the second tier of the CODIS system is the "State DNA Index System", or SDIS. The state index is operated and overseen by "the agency responsible for implementing the state's convicted offender statute". This is the level at which statewide inter-laboratory searching occurs. Any profile that is submitted to the state level by the local custodian can be checked against all others submitted by other labs in the state. The custodian at the state level can also choose to send DNA profiles to the national level in order to share the information with the entire CODIS system coast to coast.

Finally, at the top rung of CODIS is the National DNA Index System, or NDIS. The FBI operates this level of the DNA catalog. The DNA Identification Act of 1994 (CODIS, 2004) made the establishment of the CODIS database by the FBI possible by formalizing their authority to create a national index for law enforcement purposes. The NDIS is able to search, share, and exchange forensic profiles, as is done at the state and local level, but being at the highest tier it also has the ability to institute guidelines and standards that both the state and local index systems must follow in order to remain accredited. Currently all states except Mississippi participate in the National DNA Index System. As of April 2004, the total number of profiles in

the index was 1,762,005 composed of 80,302 profiles from The Forensic Index, and 1,681,703 from The Convicted Offender Index (CODIS, 2004).

## Database Uses

Now that we have looked at what DNA databases are, we will begin to examine their uses. First, it is important to comprehend the calculations done to determine the probability of a match in order to appreciate the need for larger DNA databases.

### *Match Probabilities*

Although forensic evidence is becoming increasingly popular as a means by which to either confirm or refute claims made in the courtroom, a DNA match is only as persuasive as the statistics that substantiate it. Evidence brought into a courtroom in the form of a match is ineffectual in swaying a case if the jury disregards it due to a lack of concrete support. If a prosecutor or defender desires to utilize such forensic evidence, it is in their favor to understand the significance of a match before they bring it under scrutiny.

When the validity of a match is brought into question, one must examine the characteristics of the DNA profile itself and the possibility of the match occurring simply by chance. "If the DNA profile consists of a combination of traits that figure to be extremely rare," such as a highly unusual pair of alleles at a certain locus, then "the evidence is very strong that the suspect is the contributor" (Brenner, 2004). Conversely, if the profile is common, "it is easier to imagine that the suspect might be unrelated to the crime and that he matches the crime scene DNA only by chance." Although a qualitative analysis can provide an initial impression of how significant the evidence is, ultimately the argument will come down to equations and final

figures. The explanation of the equations used to determine match probabilities is easier to understand when presented along with the aid of a visual example.

| DNA Profile | | Allele frequency from database | | | | Genotype frequency for locus | |
|---|---|---|---|---|---|---|---|
| Locus | Alleles | times allele observed | size of database | Frequency | | formula | number |
| CSF1PO | 10 | 109 | 432 | $p$= | 0.25 | $2pq$ | 0.16 |
| | 11 | 134 | | $q$= | 0.31 | | |
| TPOX | 8 | 229 | 432 | $p$= | 0.53 | $p^2$ | 0.28 |
| | 8 | | | | | | |
| THO1 | 6 | 102 | 428 | $p$= | 0.24 | $2pq$ | 0.07 |
| | 7 | 64 | | $q$= | 0.15 | | |
| vWA | 16 | 91 | 428 | $p$= | 0.21 | $p2$ | 0.05 |
| | 16 | | | | | | |
| | | | **profile frequency=** | | | | **0.00014** |

**Table 1**: Example of Four Loci and Their Frequencies (Brenner, 2004).

First, we look at the alleles found at each locus examined in a particular DNA profile. In the example in Table 1, four loci are analyzed: CSF1PO, TPOX, THO1, and vWA. For example, the alleles found at locus CSF1PO are types 10 and 11 (second column in the table). In a database of 432 allele entries, allele 10 was observed being at this locus 109 times, and 11 was

observed 134 times. So the frequency of allele 10 (here known as p) is 109/432 (0.25), and the frequency of allele 11 (here known as q) is 134/432 (0.31). If one considers that an allele came from each parent then it is possible that either the 10 came from the mother and the 11 from the father (which would calculate to a frequency of pq) or that the 10 came from the father and the 11 from the mother (another pq). Therefore, the frequency that a random person out of the 432 tested alleles would have the alleles 10 and 11 at the locus CSF1PO is 2pq, or 0.16. The aforementioned calculation was performed at only one locus whereas when calculating a match probability it is repeated through all the loci tested for that sample to obtain the frequency of each. To arrive at the final frequency each of the loci's frequencies are multiplied together as can be seen by the final column in the chart above. In the above example with the four loci, the frequency of a match for this profile comes out to be 0.00014 or about 1 in 7000. This means that "the chance is 1/7000 that some (particular) person other than the suspect" may have left the matching evidence. "In 1997, the FBI announced the selection of 13 STR Loci to constitute the core of the United States national database" so that today, most forensic DNA analyses use the standard 13 core loci and the odds become considerably less (The Biology Project, 2000).

Now that the equations used to determine the statistical significance of a match have been explored, one may ask what all of these numbers have to do with the importance of DNA databases. The answer lies in the very basics of probability and statistics. As an example, consider a coin toss. One may flip a quarter four times and have it land on heads three times and on tails only once, leading to the assumption that coins fall on heads three fourths of the time. Because a sample is considered to represent the whole, the data gathered from the four trials should be able to predict how many times a coin will fall on a certain side no matter how many times it is thrown. In this case if the coin is thrown 100 times this sample says it should fall on

heads 75 times. Knowing anything of probability of course the actually ratio of a head to tails landing is one out of two. Why is the first assumption inaccurate? The sample size is just too small. The more times you flip a coin the closer it will come to being one out of two because the larger the sample the easier it is to observe average tendency. Sample size is important to DNA databases in the same way. In the sample calculation that was done with TABLE 1 the database sizes were 432 and 428. If the database had been much larger and more extensive the frequencies would be more accurate.

*Example of Cold Case Hits*

In addition to helping define match probabilities, databases are also used for "cold hits". In this case, DNA profiles gathered from crime scene evidence are scanned against a large database of previous criminal offenders (the Offender Index). Such a match is termed a "cold hit" because it provides the police with an investigative lead that would not otherwise have been developed" (Brown and Niezgoda, 1995). One such "hit" was made in 1994 bringing closure to rape victim Debbie Smith's case. In 1989 Debbie had been abducted from her home and raped. Evidence was collected and a suspect was identified. Initial serology exams, however, excluded this suspect. The evidence from Debbie's case was preserved, and in 1994 when the county she lived in experienced an outbreak of sexual assault and rape crimes, the evidence was resubmitted in order to try to link the crimes. No link was found, but now there was a forensic profile in the database for Debbie's rapist. In the years following, the state collected more profiles by obtaining DNA samples from all of their convicted felons. The profiles were periodically searched against unsolved crimes. In 1995 Debbie's rapist was finally identified by a match in the Offender Index. His profile had been entered into the index when he was convicted of

abduction and robbery. Although at the time of the match he was already serving a 161 year sentence, the District Attorney decided to prosecute the case. "When informed that the man who raped her had been identified, Debbie Smith said, 'I feel like a weight has been lifted from my shoulders'"(Brown and Niezgoda, 1995). One hope is that expanding CODIS's scope by including more DNA profiles will increase the amount of cold case hits, and consequently reduce the number of unsolved cases, free criminals, and tattered lives.

*Example of Crime Linking*

As can be seen by the example above, DNA hits do not only find criminals to put in jail, but can also keep repeat offenders there. In the case of Debbie Smith, her rape was the offender's third strike. If convicted he would likely be sentenced to more years in prison, and any chance of parole would be postponed if not eliminated. Convictions made by linking crimes can keep dangerous criminals from victimizing again. In Missouri in 1996, for example, two young girls were abducted from bus stops and raped. The cases occurred at opposite ends of the city but the police were able to link the cases using DNA found at the scenes. They were unable to identify the suspect, however, until a development in 2000 when the DNA was reanalyzed and linked to a rape that had occurred in 1999 to a suspect identified as Dominic Moore. Moore confessed to the charges along with two other rapes occurring in 1999. The DNA from these 1999 rapes was linked to the rapes in 1996 through CODIS, and Moore was therefore identified as the perpetrator for all of the crimes (Success Stories, 2004).

**Whose DNA?? The Recent Massachusetts Legislation**

How do we decide whose profiles go into the database? On November 12, 2003 Governor Mitt Romney (R, MA) signed into law the Commonwealth of Massachusetts's new policy regarding DNA databasing (Senate No., 187, 2003). Up until February 10, 2004, when the new bill was passed, Massachusetts law only permitted authorities to obtain DNA samples from persons convicted of one of thirty-three kinds of enumerated felonies. These crimes spanned a number of offenses that included rape, assault of a child, burglary, prostitution and even murder. The new legislation sought to expand the criminal DNA database to include all convicted felons.

Many public safety organizations did not feel that this was extensive enough. The Massachusetts District Attorneys Association (MDAA, 2004) was one of many citizens, public policy groups, and public safety organizations that lobbied for a change in the state law. As expressed in their legislative priorities for the 2003-2004 session, the MDAA noted that by the time a defendant "commits one of those thirty-three designated offenses, he has already committed, on average, thirty-four other crimes." (2003-2004 Legislative Priorities)

In passing this legislation, Massachusetts joined twenty-eight other states that have enacted similar laws that expanded the scope of criminals included in DNA databases. In 1998, as few as five states had passed laws requiring that some or all felons were to be tested. In 2000, that number grew to a mere seven states. However, in the past four years, all fifty states have enacted one version or another of the law to force the collection of DNA samples from inmates convicted of sex crimes (Prisontalk, 2004), and twenty-nine states have passed legislation that require some or all felons to be included in a criminal DNA database.

Massachusetts has been in the forefront of the nationwide push to require all convicted felons to provide samples to a state-run database. Other states, such as Louisiana and Colorado have gone as far as to obtain a DNA sample from every individual that is arrested (Porteus, 2003). Known as "arrestee testing," a sample would be collected at the time of arrest and would be compared against past unsolved crime scenes. New York also tried to implement "arrestee testing" but was unsuccessful. They were, however, able to expand their current law. "The agreed upon legislation will add close to 100 new crimes that require a convicted criminal to provide a DNA sample to the Statewide DNA databank"(NY State, 2004).

On a national level, Attorney General John Ashcroft recently proposed spending $1 billion over the next few years to analyze "approximately 350,000 unexamined DNA samples taken from crime scenes, to pay for the backlog of specimens drawn from convicted sex offenders and inmates, and to develop technologies that compare samples in a fraction of the time it takes now" (Prison Talk, 2004). As one can see, the United States, on both the state and a nation level, has used legislative means to continue to implement and modernize the usage of DNA databases in the ongoing fight against crime.

**Civil Rights Violations?**

While many advocates believe that the use of genetic code is a powerful tool in law enforcement, others see it as an infringement on the civil liberties of felons. It is widely believed that once a DNA sample is collected that it can potentially be misused. The American Civil Liberties Union (ACLU) is in the forefront of the organizations opposed to universal DNA samplings from all individuals arrested (Willing, 2003). Barry Steinhardt, Associate Director of the ACLU, notes that police departments tend to keep "half of the biological material from the

swab" taken during a DNA sampling and could potentially analyze that material to reveal private information about the criminals in question (Prison Talk, 2004).

"However the authors of this IQP argue that the individual rights of a convicted offender (to not give blood) are sacrificed at the time the crime is committed, so convicted offenders should be required to submit samples" (Adams, 2004). Also, as pointed out by Danielle Fisher (2004) on a website on DNA databasing," Technicians extract the DNA profiles… from "non-coding" DNA regions—areas consisting of patterns useful for identification of a person, but otherwise containing no genes. As a result, profile specimens say nothing about genes which encode an individual's predisposition to develop particular diseases and traits." Therefore, while the profile kept in a database can continue to be useful in solving cases, as long as access to the remaining biological material is restricted or the material is destroyed, there is no need to worry about the profiles. DNA profiling is becoming more popular and as it grows, so will the demand for the genetic information. Because of this, we argue for the destruction of the sample remainders in order to forego the legal battles that would ensue over who should be allowed access.

Amidst the controversy over the use of DNA databasing as a means of crime solving, one fact remains clear: DNA evidence has been critical in the arrests and convictions of hundreds of thousands of criminals who would have otherwise gone unprosecuted.

# CHAPTER 5: CONCLUSION

Kerri Mongelli

This project has been an effort to bring to the public a greater understanding of DNA fingerprinting and its uses. To begin our examination we looked at DNA structure and the process of creating a DNA profile. To continue with the paper we explored the stringent steps that must be taken to collect and preserve forensic evidence to help its acceptance in the courtroom. It logically followed that we should inspect the historical significance of the precedence set by landmark court cases concerning the admissibility of technical evidence including DNA profiles. Finally we looked towards the future with the establishment and ongoing legislative modifications of the CODIS system. We chose these subjects to focus on because it is important to inform the general public of them in order to dispel any myths regarding the usage of DNA in forensics. We felt that this topic provided a perfect example of a new technology with great impact on society, worthy of an IQP.

The advances in forensic technology have allowed the sophistication and standardization of the process by which forensic evidence is collected and preserved in order to increase the chances of its acceptance in the courtroom. We have found that due to the nature of most forensic evidence, heat and moisture can cause severe degradation to samples. Because of this it is necessary to avoid packaging DNA evidence in plastic containers because plastic does not allow moisture to escape, and the DNA degrades. Also, because contamination, especially in the case of PCR analysis can render the resulting profile virtually useless, it is essential that only clean instruments are used in the collection, and that care is taken to collect elimination control samples. Chain of custody must also be established to avoid any argument of tampering. The importance of the treatment of this genetic material cannot be overstressed as the world painfully

learned in the OJ Simpson trial. Without the rigid guidelines that are now in place, it would be difficult to argue the credibility of the evidence itself.

The landmark court cases described in this project were selected to depict the changing ideas about the acceptance of technical evidence in U.S. courts. In 1975, Rule 702 granted that evidence could be considered admissible if it is based on fact and reliable methods. Previously the more stringent Frye Standard had been in place, declaring that only if the evidence was collected using scientifically sound and *generally accepted* practices could it be used in the courtroom. This Frye standard, had it been kept in practice, would never have allowed DNA profiles due to the initial questionability of the practice. However as DNA profiles became increasingly popular as a means by which to either substantiate or refute claims made in the courtroom, the methods used in collection, preservation, and presentation have become more standardized and better known, and have therefore afforded the evidence greater acceptance when executed correctly.

The final subject of this paper is one of much contention, the existence and expansion of DNA databases. The nation's database of forensic profiles is termed the Combined DNA Index System, or CODIS. The growth of this database is essential to the future of solving cases through the use of genetic profiling. Which DNA samples are entered into the database is a subject of controversy, and varies from state to state. We agree with the recent Massachusetts legislation requiring all persons convicted of felonies (and certain misdemeanor sex crimes) to submit samples to CODIS, not just those convicted of violent felonies. Increasing the number of DNA profiles on record in the national database makes possible a number of potential benefits. Larger databases, along with increasing the number of loci analyzed to 13, allow us to better assign the probabilities of a match. Also with boosting the number of people in the index comes a greater

possibility of a cold case hit, or a crime link. Cold case hits can wipe out cases that would otherwise remain unsolved, while crime linking can strengthen convictions and provide closure to cases otherwise left open. The reason some are opposed to the inclusion of many individuals in the index is that they fear that genetic information stored in samples used for DNA fingerprinting will be used improperly. While the profiles contained in the current standard 13 core loci themselves cannot be used to determine genetic disorders, race, or disease predispositions, the original DNA sample taken from the individual can. Because of this, we feel that the best way to move forward with databases is to first control the fear of misuse by retaining only the forensic profiles (with no medical information) while destroying the original samples they came from. By doing this, the inclusion of profiles other than those of previously convicted criminals can be collected without causing the legal issues that arise from determining who would have rights to view the information.

It is important that the knowledge contained here is not confined to those who specialize in DNA forensics such as the FBI. The knowledge must be expanded to all members of law enforcement and even the public as to where to find evidence and what procedures need to take place in order to obtain reliable end products. They can benefit from knowing how to handle a stain or other evidence at a crime scene. The more informed people are about possible contamination and proper packaging of forensic evidence, the more likely is that the first person at the scene of a crime will know how to handle it. DNA evidence can be brought to its full potency when both the standardization of methods and the growth of databases come together to create a system that is widely accepted in the courtroom and also accurate in its findings. The appropriate measures can mean the difference between bringing the case to conclusion or letting it remain unsolved.

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