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DNA FINGERPRINTING AND ITS USE AS EVIDENCE IN LANDMARK COURT CASES

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ABSTRACT

Forensic DNA fingerprinting evidence has been a source of debate ever since it was first used in an English courtroom in 1987. This Interactive Qualifying Project examines many of the landmark court cases that have pushed for the acceptance and refinement of this type of evidence. This project also explores specific DNA fingerprinting techniques, forensics, and population genetics that all serve as a basis for the evidence.

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Chapter 1

Background

"DNA identification is the latest in a long line of controversial forensic science methods (fingerprints, lie detectors, voiceprint). More than the others, however, this advance stimulates the imagination in ways akin to the mythical theft of fire from the gods. DNA-based techniques address growing fears about violent crime. They also respond to even more basic hungers; they seem capable of vanquishing the age-old foe of uncertainty itself (Billings, 1992)."

DNA Structure and Function

Life as we know it is based on the fundamental building block known as the cell. The human body contains approximately 10 trillion cells. Countless volumes of genetic information are present in the nuclei of these cells in the form of deoxyribonucleic acid, more commonly known as DNA. "And, except in the case of identical twins, no two humans share the same DNA sequence (US Congress, 1990)."

While Gregor Mendel deduced the classic principles of genetics in 1865, it was not until 1953 that the real structure of DNA was presented (figure 1-1). James Watson and Francis Crick pulled together information from such sources as Linus Pauling's description of hydrogen bonding and the α helix, and X-ray crystallography data. Data and measurements revealed by the X-ray crystallography allowed them to propose a double helix structure with a sugar-phosphate backbone on the outside (Cooper, 1997).

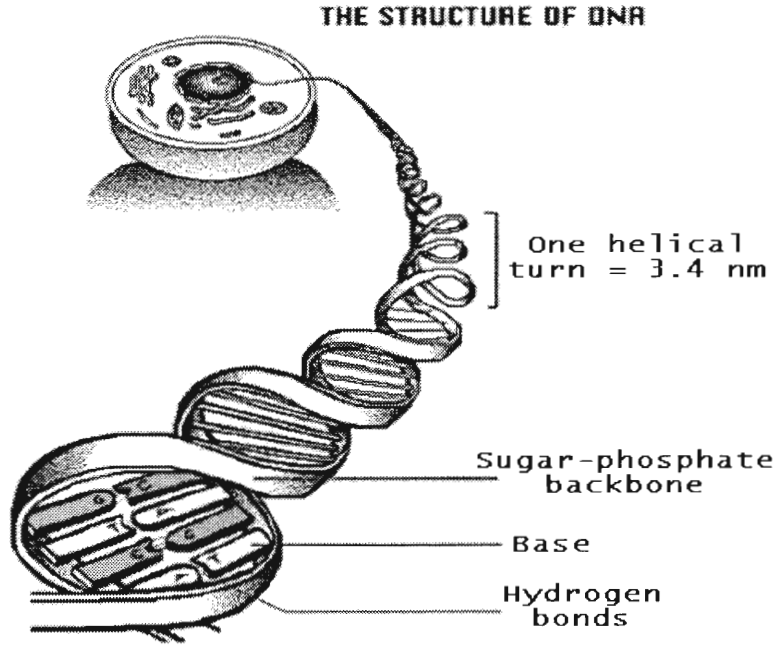


Figure 1-1
The Structure of DNA
Source: Access Excellence (Genentech, 1998).

This model of DNA presents a system of four nucleic acid bases. The bases fall under two categories, purines (adenine [A] and guanine [G]) and pyrimidines (cytosine [C] and thymine [T]). Hydrogen bonds form between purines and pyrimidines, with A only pairing with T and G only pairing with C. This specificity makes the two strands of the DNA molecule complementary. Each strand has the information to specify the sequence of the other strand (Cooper, 1997).

DNA Fingerprinting

Every cell in the human body, with the exception of red blood cells, contains DNA. The same DNA sequences are present in all of these cells. The human genome is made up of approximately 3.3 billion base pairs. Only around 3 million bases differ

between any two people. In reality, based on these numbers, any two people are 99.9% identical. It is the 0.1% difference that makes DNA fingerprinting a useful tool. The fact that DNA is identical everywhere in the body gives many sources for samples to be used in the DNA tests.

DNA fingerprinting, also known as DNA typing, was pioneered in 1984 by Alec Jeffreys at the University of Leicester, England. The process focuses on the sections of DNA that are different from individual to individual. Specifically, the process deals with nucleotide sequences, three to twenty base pairs long, repeated anywhere from just a few times to over one hundred times. These groups of repeated sequences are called minisatellites or VNTRs (Variable Number of Tandem Repeats). All people have these repeated sequences, but the number of repeats at certain loci varies from person to person. An "analysis of multiple VNTR loci constitutes a nearly unique genetic profile for every individual (Elwell, 1995)."

Restriction Fragment Length Polymorphism (RFLP) is one of the most common techniques used to measure the lengths of the sections of DNA containing the VNTRs. This technique uses gel electrophoresis to separate the DNA fragments by size. The specific methods by which RFLP and other DNA fingerprinting technologies are employed will be discussed in detail in the Chapter 3. Appendix D contains a set of illustrations showing the RFLP technique for producing a DNA fingerprint.

Chapter 2

Forensics

Before DNA evidence can be admitted into a courtroom, it is usually subjected to an admissibility hearing. In this hearing, the evidence can be attacked from many different angles. One of the most damaging is an attack based on poor forensic technique. This can range anywhere from the collection of the sample, to its transportation and storage, to the types of tests used on the samples. Therefore, it is important that certain procedures be followed along every step of the way.

DNA can be extracted from many types of tissue. The best specimen is definitely a 1-mL sample of whole blood. 15 mg of skin is also quite adequate. Table 2-1 shows some other sources of DNA and their approximate yields (Kirby, 1990). Buccal epithelial cells from mouth swabs, and hair follicles are two other common sources for DNA.

Table 2-1 DNA Content of Tissues

Source	**DNA content (approximate)**
Amniotic fluid	65 ng/mL (1 x 10^4 cells/mL at 16 weeks gestation)
Blood (mammalian)	40 µg/mL (1 µL = 4 x 10^3 to 11 x 10^3 WBC, 1 WBC = 6.6 pg DNA)
CVS	8 µg/mg
Fibroblast culture	6.5 µg/T_{25} flask (approximately 1 x 10^6 cells)
Hair roots	250 ng/plucked hair root
Liver	15 µg/mg
Muscle	3 µg/mg
Skin (whale)	3 µg/mg
Sperm	3.3 pg/cell

Samples must be clearly labeled to prevent confusion. They should be labeled with the source, tissue type, and date collected. Possible contamination sources, storage temperature, and any other relevant information should also be recorded separately. Photographs of the crime scene showing where samples were taken from should also be taken if possible. While this all may seem like a lot of extra work, "an incorrectly or insufficiently labeled specimen may be as worthless as an unlabeled specimen (Kirby, 1990)." Also, it is important that a record be kept as to who has had custody of the samples at each step along the way. This may be important information to show that the evidence has not been tampered with.

Collection and storage techniques differ depending on the type of specimen present. Tissue specimens can be placed into a lysis buffer and refrigerated. This helps prevent the degradation of the DNA. Dried stains can be placed into clean envelopes, and kept cool and dry. In the case of stains, the best stain is at least the size of a dime. Autopsy tissue should be wrapped in aluminum foil, placed in a clean plastic bag, and frozen immediately (Kirby, 1990).

It is important that the extraction of the DNA be completed as soon as possible after collecting the specimen. The amount of usable DNA that can be extracted decreases with time. Freezing will greatly increase the stability of any sample and help preserve the DNA for future extraction. If a sample has to sit for more than a couple of days before DNA extraction, freezing is recommended. It is to be noted however that repeated freezing and thawing presents the chance of damaging the DNA. The samples should be

transported to the testing laboratory by one or two-day courier service to ensure quick delivery and the ability to trace packages quickly (Kirby, 1990).

Once the specimens are collected and stabilized, the testing laboratory begins the process of isolating the DNA. It is of the utmost importance that the testing facility follows certain established procedures. For DNA evidence to be admitted into a court of law, it must be shown that the laboratory followed generally accepted practices in the testing process. In 1989, the FBI's Technical Working Group on DNA Analysis Methods (TWGDAM) published a set of guidelines for forensic RFLP analysis. These guidelines include "considerations for personnel education and training, proper documentation of pertinent records, evidence handling, validation of analytical procedures, technical controls and standards, data analysis and reporting, proficiency testing, and independent auditing (US Congress, 1990)." These guidelines provide a framework from which testing organizations can base their policies and practices. More recently, the FBI's DNA Advisory Board introduced a set of "Quality Assurance Standards for Forensic DNA Testing Laboratories" (see Appendix C). These guidelines, which went into effect October 1, 1998, serve as a basis for accreditation for DNA testing laboratories.

One of the most important steps in the forensic process is, of course, the DNA test itself. Scientists have many types of tests available to them. Each test is best suited to certain applications, so it is the job of the scientists to decide which testing procedure is appropriate for a given case. While there are tests that can be done on samples in the picogram range, the tests lose a lot of their discriminatory power, which can lead to unreadable, unusable test results (Weedn, 1996). Also, when choosing which test is

appropriate for the circumstances present, the analyst must also decide which types of controls to use to ensure there has not been any contamination. The use of specific controls can also provide information as to the sex of the person that the sample is from.

Careful evidence collection, handling, and testing can be some of the most important parts of an investigation. As attorneys continue to find more ways to attack DNA evidence, forensics experts must stay one step ahead. Chains of custody and testing procedures must be well established to be able to back up the evidence. Following procedures such as those setup by the FBI's TWGDAM, being certified by the DNA Advisory Board standards, and keeping careful documentation can greatly increase the chance that DNA evidence will be accepted by the court.

Chapter 3

DNA Fingerprinting Techniques

Restriction Fragment Length Polymorphism (RFLP) Analysis

Restriction Fragment Length Polymorphism (RFLP) Analysis is the most common DNA test used today. It is based on the differences present in every human's genome. Certain base pair sequences are repeated over and over again in the genome. The number of repeats varies from person to person. Hence the name Variable Number of Tandem Repeats. The number of repeats is measured by RFLP analysis. When examined at multiple loci, this creates a unique genetic map for each person.

For RFLP to be successful, it is necessary to have large (high molecular weight), intact DNA. RFLP analysis is a six-step process. First the DNA is isolated from the specimen. It is then cut using a restriction enzyme. The different size pieces of DNA are separated using gel electrophoresis. The DNA is then transferred to a nylon membrane. A DNA probe is then hybridized to the membrane. Lastly, a process known as autoradiography visualizes the location of the probe's hybridization (US Congress, 1990).

Depending on what the DNA is being extracted from, the procedure changes slightly. If the sample is whole blood, then the following steps must be completed. Since red blood cells do not contain DNA, they are lysed and removed along with the plasma. The white blood cells that remain are pelleted in a centrifuge, washed, and then they too are lysed. Protein is removed by phenol extraction. Chloroform is added to remove the phenol. DNA is then precipitated with alcohol from a salt solution that contains a

moderate concentration of monovalent cations. Other DNA sources (semen stains, tissue cells) are lysed and the protein digested by proteinase K. The rest of the process proceeds as with whole blood. This process can be done manually or with a machine (Kirby, 1990).

Restriction enzymes are enzymes that cleave DNA at very specific locations based on the sequence of base pairs. It is important that particular enzymes are used. Scientists have available to them numerous enzymes that will cleave sites on either end of fragments containing the VNTRs. Enzymes must be used under specific environmental conditions to achieve optimum reactivity (Kirby, 1990).

Gel electrophoresis is used to separate the DNA by fragment size. Linear, double-stranded DNA moves at a rate that is inversely proportional to the log of the molecular weight. It also varies directly with the voltage that is applied. Different concentrations of gel are valuable for examining different size ranges. The gel sits, submerged in a buffer, in a buffer tank. The DNA is injected into wells on the gel. A current is then applied to force the DNA fragments to migrate down the gel, with the smallest pieces moving the fastest. With full size (maxi) gels, a voltage of 30-33 Volts is applied overnight. This gives increased separation efficiency. After the electrophoresis is complete, the gel can be dyed and viewed under ultraviolet light (Kirby, 1990).

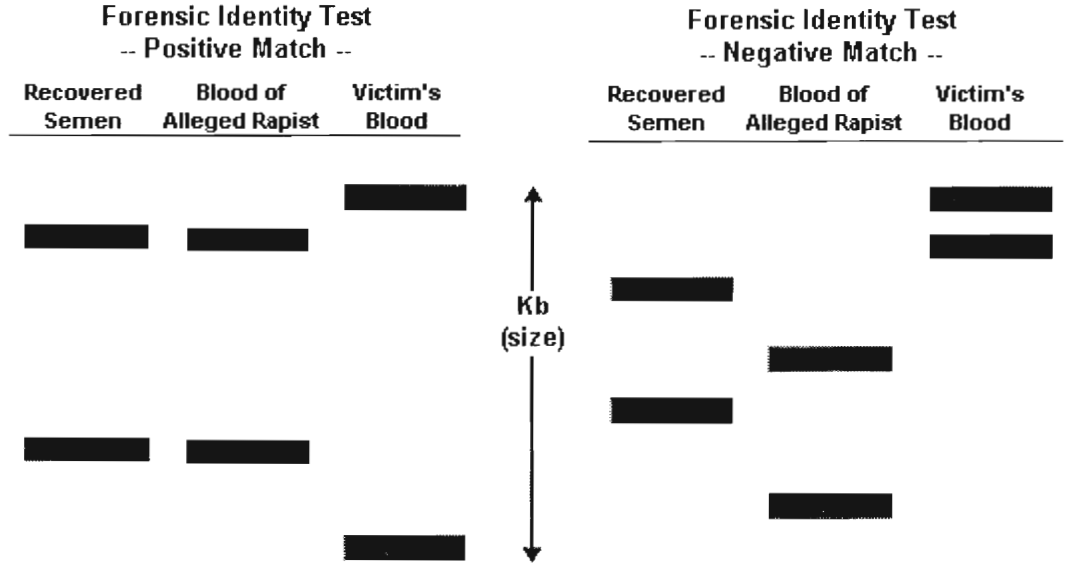
The next step in the RFLP process is to perform a Southern blot to transfer the DNA to a nylon membrane. The nylon membranes are much more stable and less fragile then the gels. The DNA is transferred to the membrane through capillary action. The DNA must first be denatured with a denaturing solution (1.5M NaCl, 0.5M NaOH or

0.4M NaOH for alkaline conditions). The blotting apparatus is set up as shown in Appendix D, with layers of blotting pads on either side of the gel and membrane. This is put in a container with the transfer solution, which slowly works its way up to the top blotting pad through capillary action. As this occurs, the DNA is pulled from the gel up to the membrane. The membrane should be wrapped in Saran Wrap and placed in a -20°C freezer to prevent DNA degradation (Kirby, 1990).

The hybridization step of RFLP involves applying labeled DNA probes. Probes are single stranded pieces of DNA around 50 bp, which recognize a specific region in the DNA (the VNTRs). The probes are normally labeled with ^{32}P, a radioactive form of phosphate that emits β particles. Probes can be of two varieties, single or multilocus. Forensic analysis usually involves several single-locus probes, which give better resolution of the bands. The hybridization is done simply by placing the probe and the membrane in a buffered salt solution at a specific temperature. After hybridization is complete, the membrane is once again wrapped in plastic wrap. It is then placed next to a piece of x-ray film for anywhere from several hours to several days. This is the process of autoradiography. The developed film is termed an autoradiograph, or autorad for short (Baird, 1996).

The autorad is then interpreted either visually or by a computer. Initially, the test results are inspected to see if there is just the same general pattern between the sample DNA and the DNA from the suspect. This inspection is quite powerful for excluding or including samples as similar. If the samples appear to be somewhat similar, a computer is used to get a completely objective determination of the comparison. In the RFLP animation in Appendix D there is an autorad from the DNA analysis in a rape case. Five probes were used in that case. It is fairly evident that the specimen recovered matches the sample given by the suspect. Figure 3-1 clearly illustrates a positive and a negative match.

Figure 3-1



Positive and negative matches in a forensic identity test
Source: DNA Identity Testing, compiled by Lifecodes, Corp. (1991).

Polymerase Chain Reaction (PCR)

Oftentimes, not enough evidence from a crime scene can be recovered to do traditional RFLP analysis. The Polymerase Chain Reaction helps compensate for this. PCR has the ability to amplify picogram starting amounts to microgram levels. Single-copy genomic sequences over 2 Kb have been amplified more than 10 millionfold in just a few hours.

Each cycle of the Polymerase Chain Reaction starts with the heat denaturation of the DNA. An oligonucleotide primer is added to each DNA strand. Thermal stable Taq Polymerase is then used to facilitate the extension of the primers. The reaction is done in the presence of a buffer, and the nucleotides dATP, dCTP, dGTP, and dTTP. These are the bases that get added to the strands by Taq Polymerase. This process must be completed in an environment capable of thermal cycling. There are machines that precisely control the temperature at every step along the way. The process is then done over and over again, each time producing twice the number of DNA molecules it started with (US Congress, 1990). Figure 3-2 illustrates this process.

PCR has many benefits. It allows the testing laboratory to reproduce enough DNA to use in testing. PCR can be used to build up highly degraded samples. Also, DNA from sources other than human DNA will not amplify if the primers that are used are designed specifically for human nucleotide sequences. The benefits seem endless. Unfortunately, there are drawbacks to PCR. There is a chance that PCR can change the nucleotide sequence slightly. Normally it is not enough to change the test results. It is also extremely easy to introduce contamination due to the very nature of the reaction. If

even a small section of human DNA gets into the reaction, it will be greatly amplified. (Kirby, 1990)

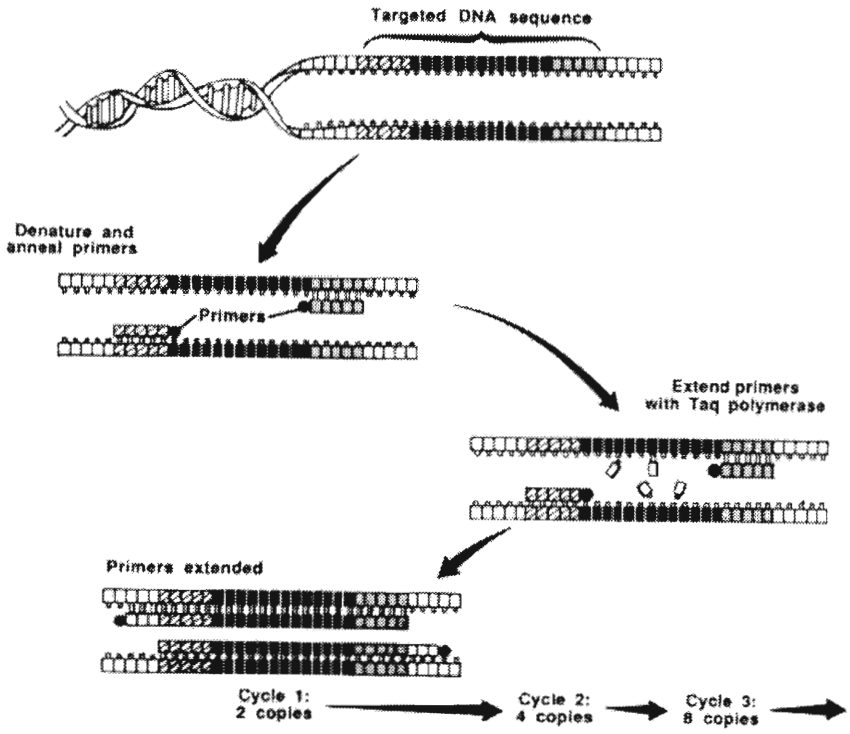


Figure 3-2
Amplification of DNA using PCR
Source: DNA Fingerprinting: An Introduction (Kirby, 1990).

Amplified Fragment Length Polymorphism (AMP-FLP)

When RFLP analysis is performed on PCR amplified sequences instead of restriction enzyme cut sequences, it is given the term Amplified Fragment Length Polymorphism (AMP-FLP). These tests are done on silver stained polyacrylamide gels. The AMP-FLP approach is quite useful when little genetic material is available for testing. Results from this test can be achieved within one day, as opposed to the possible

weeks needed for conventional RFLP analysis. As well as being quite rapid, this is a good high-resolution technique (Kirby, 1990).

Sequencing

It is possible to determine the actual sequence of nucleotides in a section of DNA. This type of testing is generally done on mitochondrial DNA, in areas such as the D-loop. Sequencing could theoretically give a degree of identification that is not dependent on any statistics, since two sequences could be directly compared for identification. This technique is also helpful for finding information about lineage, since mitochondrial DNA is maternally inherited. If sequencing were to be used in forensics, a large number of regions in the DNA would have to be tested (Baird, 1996).

An added benefit of this technology is that it can be performed on very small samples from a crime scene. While a cell only has one nucleus, it has many mitochondria, and hence many sources of mitochondrial DNA.

While these are some of the most important DNA fingerprinting techniques, they are not the only ones in existence. Different circumstances may lead a lab to use another, lesser-used test. And, as in any science, new techniques are always being discovered and existing techniques are being constantly refined.

Chapter 4

Population Genetics

Once the necessary DNA tests are complete and a match is found, the suspect is not automatically guilty. The match only means that the sample from the suspect is consistent with the sample from the crime scene. The sample from the crime scene could be from the suspect or from someone else in the population whose profile matches the suspect when certain loci are tested. It is now the job of the population geneticist to calculate the probability that the two samples could match purely by chance in the given population (Massachusetts Bar Association, 1995).

The standard equation utilized by population geneticists is the Hardy-Weinberg equation. This equation expresses the frequency of occurrence of certain combinations of genes, assuming random mating, lack of significant selection, or mutation. The simplest form of the equation is $p^2 + 2pq + q^2 = 1$, where p is the frequency of allele (gene) A and q is the frequency of allele B. A population that satisfies the above equation is said to be in Hardy-Weinberg equilibrium (Baird, 1996).

Scientists use population databases to make their calculations. These databases contain genetic information such as the frequency of certain alleles for various racial/ethnic groups. Once it is found if the population being dealt with is in Hardy-Weinberg equilibrium, the analyst must examine the population database for the alleles that have been tested. Assuming the loci are not linked in some way, the analyst can simply use the multiplication rule for probability calculations. This involves multiplying

the probabilities of each of the alleles existing in the given population. This is valid since each "event" is random and independent of the others (Kirby, 1990).

One concern that many geneticists have is whether the population and its various racial subpopulations are truly in Hardy-Weinberg equilibrium. There was also question as to whether the various loci were actually independent of each other. Based on these concerns, the National Research Council recommended the use of a ceiling principle for such calculations. This resulted in an underestimation of the actual frequency of a given DNA profile. Based on all empirical data available, there is no scientific basis for these arguments. This led the National Research Council to recently reject the ceiling principle. They only suggest now using the Hardy-Weinberg equation with some additional weighting factors if necessary for certain subpopulations (Baird, 1996).

All of these calculations can quickly lead to statistical probabilities in the range of 1 in 10 million individuals or more. Jurors are easily overwhelmed by numbers such as these. It is important that they are instructed to only give the statistics their due weight. Also, the prosecution must have other evidence to back up their statistical findings.

Chapter 5

Landmark Court Cases

DNA testing was first used in a criminal case in the mid nineteen-eighties in the small English county of Leicestershire. This technique was used to help find the man wanted in the rapes and murders of two fifteen-year-old girls. These crimes occurred in 1983 and 1986. Colin Pitchfork, convicted in 1987 is now serving two life sentences for the murders and two ten-year sentences for the rapes. Since this first case, DNA evidence has been used thousands of times to both convict and prove defendants innocent (US Congress, 1990).

Certain court cases stand out in the history of the United States judicial system as landmark cases. These cases presented important and oftentimes groundbreaking arguments that have helped form the foundations of numerous other cases. They are referenced time and again by attorneys across the country. Some advance the acceptance of new types of evidence. Others help to protect defendants from possible prejudice and errors in laboratory procedures while testing evidence. All of the cases that follow present important steps in some facet of justice. A listing of the first court case in each state to utilize DNA evidence is included in Appendix B.

FRYE v. UNITED STATES
No. 3968
293 F. 1013

Court of Appeals of District of Columbia
Submitted November 7, 1923
Decided December 3, 1923

James Alphonzo Frye was convicted of murder in the second degree. He subsequently appealed to the Supreme Court of the District of Columbia. The only point that was being argued was the result of a deception test made on the defendant. The test, a systolic blood pressure deception test, is based upon the idea that rises in the systolic blood pressure are brought about by nervous impulses in the autonomic nervous system. It is claimed that fear, rage, pain, conscious deception, concealment of facts, guilt of crime and the accompanying fear of detection will all raise the systolic blood pressure. The rise caused by giving false testimony can be distinguished from mere fear of the examination.

Prior to the trial, Frye was given this deception test and his counsel attempted to have the scientists who conducted the test testify as an expert witness. Counsel for the government objected and the objection was sustained. Counsel for the defendant then suggested that the proffered witness conduct a test for the jury. This was again denied.

In their brief to the court, the counsel for the defendant presented the following summation of the basis of their case:

> "The rule is that the opinions of experts or skilled witnesses are admissible in evidence in those cases in which the matter of inquiry is such that inexperienced persons are unlikely to prove capable of forming a correct judgement upon it, for the reason that the subject-matter so far partakes of a science, art, or trade as to require a previous habit or experience or study in it, in order to acquire a

> knowledge of it. When the question involved does not lie within the range of common experience or special knowledge, then the opinions of witnesses skilled in that particular science, art, or trade to which the question relates are admissible in evidence (293 F. 1013)."

In their ruling, the court stated that they "will go a long way in admitting expert testimony deduced from a well-recognized scientific principle or discovery, [but] the thing from which the deduction is made...[must] have gained general acceptance..." (293 F. 1013). It was the court's decision that the systolic blood pressure test had yet to achieve this status. The judgement was affirmed.

The implications of this case are felt even today. The Frye Standard of Admissibility is used quite extensively. This standard states that the scientific community must generally accept a test before it is accepted in a court of law. It is often times the lone test used in deciding whether to admit evidence into a case. More recently, however, more stringent tests have come into use.

UNITED STATES of America v. John W. DOWNING
No. 82-1766
753 F2d 1224

United States Court of Appeals, 3rd Circuit
Argued January 27, 1984
Decided January 25, 1985

John W. Downing was convicted of mail fraud, wire fraud, and interstate transportation of stolen property in US District Court for the Eastern District of Pennsylvania. Downing appealed the conviction.

Circuit Court Judge Becker stated that Federal Rule of evidence 702, which pertains to expert testimony, allows a defendant to use an expert in human perception and memory as a means of determining the reliability of eyewitness identifications. He went on to say that the district court erred in the decision to not allow the testimony of a defense psychologist. The district court believed that the psychologist's testimony could not meet the "helpfulness" standard, Federal Rule 702. This was deemed harmful to the defendant since he was convicted solely on the grounds of eyewitness testimony.

Federal Rule of Evidence 702 defines expert testimony as follows:

> "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 (US Congress, 1990)."

This rule also regulates the types of theories, techniques, and principles from which the expert may rely on. It also states that the testimony does not have to be beyond the level of comprehension of a lay person. The testimony can just be in an area where the expert can be of assistance.

The case was vacated and remanded to the district court to decide whether to admit this testimony in the case. Then, the case would be retried and the testimony of the psychologist once again put up for consideration.

The implications of this case are not as evident as in the Frye case, but they are nonetheless important. This case shows the courts that they need to look more at what tests need to be used when, and in what capacity, in deciding what evidence is allowed into the courtroom. This is especially true in such cases where the inclusion or exclusion of a single piece of evidence can have such a profound effect.

The PEOPLE of the State of New York, v. Joseph CASTRO, Defendant
545 N.Y.S.2d 985

Supreme Court, Bronx County, Criminal Term, Part 28
August 14, 1989

The defendant stood accused of two counts of second-degree murder. It was alleged that on February 5, 1987 he stabbed to death Vilma Ponce, twenty-one years old and seven months pregnant, and her two-year old daughter. The main evidence in the case was a wristwatch belonging to Castro. There were bloodstains on the watch that the prosecution was attempting to prove belonged to the victims. The defendant claimed it was his blood on the watch. The prosecution sought to introduce DNA evidence to identify the blood.

What followed has been referred to as the most comprehensive and extensive legal examination of DNA testing to date in the United States. The pre-trial Frye hearing took place over a twelve-week period, producing a transcript of approximately five thousand pages. It was an intense and technical look at DNA testing as a whole, as well as the methods employed by Lifecodes, Corp., the forensic DNA testing facility used in this case.

The court devised a three-prong test for DNA evidence to aid them in evaluating the evidence presented.

1. Is there a theory, which is generally accepted in the scientific community, which supports the conclusion that DNA forensic testing can produce reliable results?
2. Are there techniques or experiments that currently exist that are capable of producing reliable results in DNA identification and which are generally accepted in the scientific community?

3. Did the testing laboratory perform the accepted scientific techniques in analyzing the forensic samples in this particular case? (545 N.Y.S.2d 985)

Judge Gerald Sheindlin held that when the generally accepted tests are performed, DNA testing evidence is admissible. However, he stated that the testing laboratory failed in several respects to use generally accepted techniques for obtaining reliable results with regards to the DNA evidence of inclusion.

This case set up a new set of DNA evidence inclusion rules that are often used along with the typical Frye test. This is another step in ensuring that the defendant gets a fair and just trial. This case is also important due to the depth in which the evidence was examined. The fact that the pre-trial hearing alone took twelve weeks shows that the courts were not about to let this issue go through without examining every available piece of information.

UNITED STATES of America, Appellee, v. Matthew Sylvester TWO BULLS, a/k/a Matthew Sylvester Two Bulls, Jr., Appellant
No. 90-5040
918 F.2d 56

United States Court of Appeals, Eighth Circuit
Submitted August 14, 1990
Decided October 31, 1990

The defendant was convicted of aggravated sexual abuse and sexual abuse of a minor in the US District Court for the District of South Dakota. He was convicted of the rape of a fourteen-year-old girl on the Pine Ridge Indian Reservation in South Dakota. DNA evidence showed a very high probability that the semen found on the girl's underwear was that of Two Bulls. Two Bulls attempted to have this evidence suppressed. The district judge, after hearing just one government witness, decided that DNA evidence

is sufficiently accepted by the scientific community, and so the evidence could be presented. The defendant was sentenced to 108 months in prison and two years of supervised release. Two Bulls appealed the conviction.

During his appeal, Two Bulls argued that the trial court used Federal Rule of Evidence 702, when it should have used the more rigorous Frye Standard. Based on this, he argued the district court violated his due process by having an incomplete pre-trial suppression hearing. He also felt that the three-step test devised in People v. Castro (545 N.Y.S.2d 985) should have been used. The three steps are:

1. Decide whether the scientific community generally accepts the theory that DNA tests produce reliable results.
2. Decide whether techniques currently exist that are generally accepted by the scientific community and are capable of producing reliable results.
3. Decide whether the laboratory used these techniques in this case.

The court also presented the following statement in response to Two Bulls' argument for the use of Frye over Rule 702:

> Regardless of which rule may be followed, we feel Rule 702 and Frye both require the same general approach to the admissibility of new scientific evidence. Neither rule should permit speculative and conjectural testing which fails normal foundational requirements necessary for the admissibility of scientific testimony or opinion.

In court, it was held that DNA evidence is still subject to attack based on prejudice, relevancy, and laboratory procedures. Because of this fact, it would seem logical that both sides of the question should be heard before a ruling is passed on the admissibility of questionable evidence. The trial court ruled after only hearing one government witness.

Chief Judge Lay found that the court erred in only hearing one witness, as well as in determining the admissibility of the DNA evidence without determining if the testing procedures used by the FBI crime lab were conducted properly. Upon review, the case was vacated and remanded with the instructions to hold an expanded pre-trial hearing on the admission of DNA evidence. The trial court was to decide the following:

1. Does the scientific community generally accept DNA evidence?
2. Were the testing procedures used generally accepted as reliable if performed properly?
3. Were the tests performed properly in this case?
4. Is the evidence more prejudicial than probative in this case?
5. Are the statistics used to determine the probability of someone else having the same genetic characteristics more probative than prejudicial under Rule 403? Cf. Schwartz, 447 N.W.2d at 428 (limiting the use of population statistics since "juries in criminal cases may give undue weight and deference to presented statistical evidence and we are reluctant to take that risk.")

The implications of this case are far reaching. It did more than bring up a few new questions to ask based on how DNA evidence was tested and handled. It brought forward the issue of prejudice as it applies to DNA evidence and directly approaches it in admissibility hearings. The jury must now decide whether the evidence presented is more probative than prejudicial. This case further fights for *any* defendant's right to a fair trial.

STATE of South Dakota, Plaintiff and Appellee v. Will D. WIMBERLY, Defendant and Appellant
No. 16987
467 N.W.2d 499

Supreme Court of South Dakota
Argued November 26, 1990
Decided March 20, 1991

After being charged with rape in the first degree, rape in the second degree, and sexual contact with a child under the age of sixteen, the defendant was convicted of first degree rape in Circuit Court, Eighth Judicial Circuit, Meade County, South Dakota, Judge Scott C. Moses presiding. Wimberly appealed his conviction on several grounds. He claimed there was a speedy trial violation, an improper chain of custody of his blood, improper testimony from one of the witnesses, and he challenged the admittance of the DNA evidence.

Wimberly attempted a feeble attack on the admittance of the DNA evidence based on the their lack of reliability. A Frye hearing was held in which an FBI agent testified as to the reliability of the processes used in the DNA test. Wimberly could produce no evidence to refute the agent's statements. This part of the appeal failed.

All articles of the appeal were eventually denied except for the improper testimony of one of the witnesses. Judge Wuest felt that the witness should not have been allowed to testify that is was the opinion of another suspect in the case that it was the defendant who raped the victim.

Although the admission of the DNA evidence was found to be acceptable, based on the possible attacks due to prejudice, relevancy, and laboratory procedures, the judge felt that the admission criteria set up in United States v. Two Bulls (918 F.2d 56) should

have been used. He also held that the burden of proof with respect to the validity of DNA evidence should lie with the state, not the defendant. The case was reversed and remanded for a new trial.

There is one large issue that makes this a landmark case. Even though the admissibility evidence presented by the FBI agent in the Frye hearing was accepted, that was not enough for the judge in the case. Judge Wuest held that even though the evidence as it was presented was admissible, the procedures set up in *Two Bulls* should have been followed. The Frye test leaves too many points open for attack. This seems to present a general movement of the judicial system towards more stringent admission standards. This case, along with other cases of this nature, continues to increase the protection for the defendants – protection from errors and wrongful discrimination.

STATE of South Carolina, Respondent v. Stacy FORD, Appellant
No. 23209
392 S.E.2d 781

Supreme Court of South Carolina
Heard October 4, 1989
Decided May 7, 1990

The details of the crime and this case are scarcely relevant to this being a landmark case. A ruling passed down by the Supreme Court justice has become the focus of this case. It was ruled that in South Carolina, Frye hearings would no longer be necessary. The reliability and acceptability of DNA evidence had been proven sufficiently. DNA analysis was now free to be admitted in the same manner as any other scientific evidence that is routinely used in court proceedings, such as fingerprint analysis

and A/B/O blood tests. However, issues pertaining to prejudice or relevancy may still be brought up to challenge the admission of the DNA evidence.

This is clearly a groundbreaking ruling. The Supreme Court in South Carolina felt so strongly about the capabilities of DNA testing that it has brought it on par with other forms of evidence. Besides streamlining part of the judicial system, this makes a clear statement that the implications of DNA evidence are finally being given their due worth and trust.

The STATE of Ohio, Appellee, v. PIERCE, Appellant
No. 90-1898
597 N.E.2d 107

Supreme Court of Ohio
Submitted June 2, 1992
Decided September 2, 1992

In the Court of Common Pleas, Pierce was convicted of two counts of rape and one count of kidnapping. Pierce appealed the conviction, but it was affirmed by the Court of Appeals. Next he brought his case to the Supreme Court of Ohio. It was argued that he committed the first rape of a girl on the way to school on January 4, 1988. The next rape was of a girl sunbathing in a park on May 2, 1988. Then, on June 6, 1988 it was argued that Pierce approached a girl while brandishing a handgun. The victim was able to get away by striking the assailant with her keys.

Pierce's DNA was compared with samples from the two rape victims. Counsel for the defendant attempted to have the DNA evidence thrown out on the grounds that it is not accepted by the scientific community yet, and is still unreliable. The Court of Appeals upheld the decision of the Court of Common Pleas. The case then went on to the State Supreme Court. Once again, the Supreme Court upheld the previous conviction.

The importance of this case is found in the ruling passed down by the Supreme Court. The court held that the DNA evidence is admissible. In response to the unreliability of the tests, the court stated that any questions concerning the reliability of that evidence goes only to the weight placed on the evidence. This decision points towards a growing acceptance of DNA based evidence. The court understands the implications of this type of evidence and is affording it its due worth.

---- v. Richard J. SCHMIDT
No. ----
(Science Vol. 282 p851-2)

Louisiana Court System
Convicted October 1998

The defendant, a Louisiana doctor was convicted in the attempted murder of a former lover, Janet Allen. He injected her with HIV and Hepatitis C tainted blood from two of his patients. The prosecution introduced DNA analysis of HIV strains as evidence. This was the first case in the United States in which such evidence was used. The analysis connected the HIV infection in Allen to the infection in one of Schmidt's patients.

Counsel for the defendant attempted to have the DNA evidence thrown out on the grounds that the laboratory work was sloppy. They also argued that without an epidemiological study of other possible routes of infection, the DNA analysis was meaningless. Both the Court of Appeals and the District Court ruled that the evidence could be used. Schmidt was eventually convicted. He is now planning on appealing his conviction.

This is a landmark case in two ways. This was the first time in the United States in which DNA analysis of HIV strains was used in criminal court. Also, it is a stepping point for what will, no doubt, be a barrage of lawsuits of this nature. It is conceivable that many people who were infected with HIV by a sexual partner will bring them to court. It could also be used in cases of food poisoning, or even biological warfare. Both

sides in this case agreed that there needs to be an explicit set of rules formulated for the use of this type of evidence.

Chapter 6

Exceptional Cases

Certain cases exist that may not be landmark cases in the strictest sense, but they are nonetheless important. They have brought attention to the use of DNA fingerprinting. Many have also caught the eyes of the general public and were discussed over countless breakfast tables. These cases are given the title "Exceptional Cases."

The Grandmothers of the Placo de Mayo

In 1975 the military overthrew the government of Isabel Peron in Argentina. Thousands and thousands of people began to disappear. Many of these were young children. By the late nineteen-seventies and into the nineteen-eighties a group of mostly older women, calling themselves the Grandmothers of the Placo de Mayo began to march and protest in the main square in Buenos Aires. Through various routes, these people began to find out that military families were raising their missing children.

After the fall of the military government in 1983 from the Falklands War, the Grandmothers contacted the American Association for the Advancement of Science. They wanted help in proving that these children were in fact theirs. They wanted it to be proven using genetics. With the help of Mary Claire King, they began to get court orders for genetic testing. This testing provided proof of the children's rightful parents (Genentech, 1998).

So far, over fifty children have been identified and returned to their parents. Many of the missing children are now reaching adulthood and are able to search on their own for their families. Now that some of the grandmothers are dying, they are setting up a DNA databank so that the children will be able to find out who their real families are even after the grandmothers die (Genentech, 1998).

Anastasia and The Romanov Family

For over 300 years, the Romanovs were the ruling family in Russia. On July 17, 1918, Tsar Nicholas II became the last tsar of Russia as he and his family were brutally murdered by the Bolshevik army, led by Vladimir Lenin. The people of Russia were told that only the tsar was killed and the rest of his family had been brought to safety. After the civil war ended, many Romanov imposters began to surface. Most were disproven rather quickly. One that could not be discredited so easily was a woman claiming to be Anastasia, one of Nicholas' daughters. She was quite convincing, in that she could provide certain details about the family and she bore a striking similarity to pictures of Anastasia. For nearly 30 years, she fought to be recognized as the real Anastasia. In 1970, a German court ruled that she was not in fact Anastasia. She gave up and moved to the United States and died in 1984.

In 1989, it was announced that the bodies of the royal family had been found in a mass grave in the Koptyaki Forest. Tests concluded that it was the Tsar's family, but there were two skeletons missing – those of Alexei and Anastasia. Numerous people called for the DNA comparison of the supposed Anastasia with that of the Romanov

family. A lab sample of her tissue was tracked down and the DNA was compared. They did not match. She is actually believed to be Franzisca Schanzkowska, a Polish woman who had disappeared from a Berlin boarding house. The mystery had finally been solved. However, the whereabouts of the two missing skeletons are still not known (Constitutional Rights Foundation, 1997).

The O.J. Simpson Murder Case

Dubbed the "Trial of the Decade" by many, the O.J. Simpson murder case was one of the most high profile criminal cases that the United States had seen in recent years. Beloved celebrity and football player Orenthal James Simpson stood accused of brutally murdering his wife, Nicole Brown Simpson, and her friend Ronald Goldman, on June 12, 1994. The evidence against him seemed insurmountable. Some of the evidence included: bloodstains from 13 locations, a sock at the foot of O.J.'s bed, stained with Nicole's blood, bloodstained leather gloves, and bloodstains in his Bronco.

DNA testing identified Nicole's blood on the sock, as well as O.J.'s blood at the scene. Unfortunately, due to poor techniques for sample collection, handling, and testing coupled with claims of a racial conspiracy, the defense was able to discredit the results of the DNA tests. In the end, the prosecution could not prove their case beyond a reasonable doubt and Simpson was acquitted. This case will no doubt prompt investigators to establish much tighter rules for the collection and handling of evidence (Lachter, 1997).

APPENDIX A

Glossary of Terms

Information taken from
"The Science of DNA Analysis: DNA Profiling," and
"DNA Identity Testing", articles compiled by Lifecodes, Corporation.

AGAROSE: The neutral gelling fraction of agar commonly used in gel electrophoresis.

ALLELE: One of several forms of a gene occupying a given locus on the chromosome.

AMINO ACID: The building blocks of proteins. There are 20 common amino acids; they are joined together in a strictly ordered "string" that determines the character of each.

AMPLIFICATION: The production of many copies of a region of DNA.

ANNEAL: The process by which the complementary base pairs in the strands of DNA combine.

ANTIGEN: Any molecule whose entry into an organism provokes synthesis of an antibody (immunoglobulin).

AUTORADIOGRAPHY: A technique used for detecting radioactively labeled molecules (by their effect of creating an image on photographic films) in a cell or tissue.

AUTOSOMAL DOMINANT: Genetic trait (or a gene) carried on one of the autosomes that produces an observable phenotype, even if present in only one copy.

AUTOSOMAL RECESSIVE: Genetic trait (or gene) carried on one of the autosomes that must be present in two copies (both of the alleles present must be of the same type) in order for the gene to be expressed and the trait seen in the phenotype.

AUTOSOME: Any chromosome other than the sex chromosome.

BACK MUTATION: Reverses the effect of a mutation that had inactivated a gene; thus, it restores wild phenotype.

BACTERIOPHAGE (PHAGE): A virus that infects a bacterial cell. Phage consist of a core of genetic material (DNA or RNA) carrying the particle's genetic information that is surrounded by a protein coat or capsule. When a phage infects a host cell, the cell machinery that manufactures protein in response to genetically encoded instructions is commandeered by the phage and used to produce offspring phage. The lambda bacteriophage is frequently used as a vector in recombinant gene experiments.

BANDING: A technique of staining chromosomes in characteristic patterns of cross bands.

G BANDS: The dark and light cross-bands seen in chromosomes after treatment with trypsin and Giemsa stain.

Q BANDS: The pattern of bright and dim cross-bands seen on chromosomes under fluorescent light after quinacrine staining.

BASE PAIR (bp): In nucleic acids, adenine must always pair with thymine (or, in RNA, with uracil) and guanine with cytosine. The specificity of base pairing is fundamental to DNA replication and to its transcription into RNA.

BIOTECHNOLOGY: The collection of industrial processes that involve the use of biological systems. For some industries, these procedures invoke the use of genetically engineered microorganisms.

BREAKAGE and REUNION: A mode of genetic recombination in which two DNA duplex molecules are broken at corresponding points and then rejoined crosswise.

CAPSID: The external protein coat of virus particles.

CELL: The basic unit of life. In humans the cell is normally composed of a nucleus and cytoplasm. There are about 10 trillion cells in the human body.

CENTIMORGANS (cM): A functional measurement of the distance between two linked loci, which is determined by the frequency of recombination between the loci. A map distance of one centimorgan is equivalent to a recombination frequency of one percent.

CHIMERA: An individual composed of cells derived from different zygotes.

CHORIONIC VILLOUS SAMPLING (CVS): A procedure used for prenatal diagnosis at nine to twelve weeks gestation. Fetal tissue for analysis is aspirated by catheter through the cervix from the villous area of the chorion under ultrasonic guidance.

CHROMOSOME: The location of hereditary (genetic) material within the cell. This hereditary material is packaged in the form of a very long, double-stranded molecule of DNA surrounded by and complexed with several different forms of protein. Genes are found arranged in a linear sequence along chromosomes, as is also a large amount of DNA of unknown function.

CLASSICAL GENETICS: The body of knowledge that deals with the laws of genes such as determined by appropriate test matings.

CLONE: A group of genetically identical cells or organisms asexually descended from a common ancestor. All cells in the clone have the same genetic material and are exact copies of the original.

CODON: A triplet of three bases in a DNA or RNA molecule, specifying a single amino acid.

COLONY HYBRIDIZATION: A technique for using *in situ* hybridization to identify bacteria carrying chimeric vectors whose inserted DNA is homologous with some particular sequence.

COMPLEMENTARY DNA (cDNA): DNA synthesized from a messenger RNA template rather than the usual DNA template. cDNA is often used as a DNA probe to help locate a specific gene in an organism.

CONJUGATION: The one-way transfer of DNA between bacteria in cellular contact.

CONTROLS: Samples of predetermined concentrations (known or unknown to the analyst) treated as unknowns in an assay. Controls are included as part of quality control for each test.

COSMIDS: Plasmids into which phage lambda cos sites have been inserted: as a result, the plasmid DNA can be packaged in vitro in the phage coat.

CROSSING-OVER: The reciprocal exchange of material between chromosomes that occurs during meiosis and is responsible for genetic recombination.

CUT: A break that occurs in both strands of a DNA molecule opposite one another.

DELETIONS: Result from the removal of a sequence of DNA, the regions on either side being joined together.

DENATURATION: Conversion of DNA from the double-stranded to the single-stranded state, usually accomplished by heating to destroy chemical bonds involved in base pairing

DENATURATION OF PROTEIN: Describes its conversion from the physiological conformation to some other (inactive) conformation.

DNA (Deoxyribonucleic Acid): The molecule containing hereditary information in all but the most primitive organisms (some viruses that use RNA). The molecule is double-stranded, with an external "backbone" formed by a chain of alternating phosphate and sugar (deoxyribose) units and an internal ladder-like structure formed by nucleotide base-pairs held together by hydrogen bonds. The nucleotide base-pales consist of the bases adenine (A), cytosine (C), guanine (G), and thymine (T) whose structures are such that A can hydrogen bond only with T, and C only with G. The sequence of each individual strand can be deduced by knowing that of its partners. This complementarity is the key to the information transmitting capabilities of DNA.

DNA POLYMERASE CHAIN REACTION: A technique for amplifying a selected portion of DNA by synthesizing oligonucleotides complementary to it.

DNA PROBE: A molecule (usually a nucleic acid) of known structure and/or function that has been tagged with some tracer substance (a radioactive isotope or specific dye-

absorbing compound) that is used to locate and identify a specific gene or region of a chromosome or portion of the genome.

DOMAIN: A region of the amino acid sequence of a protein that can be equated with a particular function, or a corresponding segment of a gene.

DUPLICATION: Presence of a segment of a chromosome in duplicate. Duplication may involve whole genes, series of genes, or only part of a gene.

ELECTROPHORESIS A technique for separating different molecules based on their differential movement in an electric field.

END LABELING: Describes the addition of a radioactively labeled group to one end (5' or 3') of a DNA strand.

ENDONUCLEASE: An enzyme that cleaves bonds within a DNA or RNA strand.

ENZYME: A functional protein that catalyzes a chemical reaction. Enzymes control the rate of metabolic processes in an organism.

EUKARYOTE: A higher, compartmentalized cell characterized by its extensive internal structure and the presence of a nucleus containing the DNA. All multicellular organisms are eukaryotic. The simpler cells, the prokaryotes, have much less compartmentalization and internal structure; bacteria are prokaryotes.

EXON: Any segment of an interrupted gene that is represented in the mature messenger RNA product and, thus, codes for protein.

EXONUCLEASE: An enzyme that removes bases sequentially from the ends of a linear DNA molecule; may be specific for either the 5' or 3' end of DNA or RNA.

EXPRESSION: The process by which the blueprint contained in DNA is converted into the structures and biochemical mechanisms present and operating in a cell.

EXTRANUCLEAR GENES: Reside in organelles such us mitochondria and chloroplasts outside the nucleus.

FILTER HYBRIDIZATION: Performed by incubating a denatured DNA preparation immobilized on a nitrocellulose filter with a solution of radioactively labeled DNA or RNA.

FLANKING SEQUENCE: A region of a gene preceding or following the transcribed region.

GENE: The portion of a DNA molecule that comprises the basic, functional hereditary unit; a sequence of DNA that produces a specific product.

GENE AMPLIFICATION: Increase, within a cell, of the number of the same gene. Amplification may be spontaneous or induced.

GENE MAPPING: Determining the relative locations of different genes on a given chromosome.

GENETIC CODE: The biochemical basis of heredity consisting of codons (base triplets along the DNA sequence) that determine the specific amino acid sequence in proteins.

GENETIC ENGINEERING: A technique used to modify the genetic information in a living cell, reprogramming it for a desired purpose (such as the production of a substance it would not naturally produce).

GENETIC LETHAL: A mutation that renders the affected individual incapable of reproduction, either through infertility or through lack of production of viable offspring and, thus, unable to transmit the mutation to the next generation.

GENETIC MARKER: Any character that acts as a signpost or signal of the presence or location of a gene, chromosome, or hereditary characteristic in an individual, a population, chromosome, or a DNA molecule.

GENOME: The total genetic information contained in an organism's genes. Also described as the total content of all chromosomes in an organism.

GENOTYPE: The total genetic information contained in the chromosomes of an organism; the genetic makeup of an organism.

GERM PLASM: The total genetic variability available to an organism, represented by the pool of germ cells.

HOTSPOT: A site at which the frequency of mutations (or recombination) is very much increased.

HYBRID CELL: A cell formed by fusion of two cells of different origin in which the two nuclei have merged into one. Can be cloned to produce hybrid cell lines.

HYBRIDIZATION: In molecular genetics, pairing of an RNA and DNA strand or of two different DNA strands.

HYBRIDOMA: Hybrid cell resulting from the fusion of a myeloma cell ("immortal") and a lymphocyte (cell producing an antibody).
IMMUNITY: The ability of a prophage to prevent another phage of the same type from infecting a cell or the ability of a plasmid to prevent another of the same type from becoming established in a cell. Different mechanisms are involved in the two types of immunity.

IMMUNOTOXIN: Molecule toxic to the cell, attached to an antibody.

INDIRECT LABELING: A technique for examining the organism of DNA by making a cut at a specific site and isolating all fragments containing the sequence adjacent to one side of the cut: it reveals the distance from the cut to the next break(s) in DNA.

INSERTION: A structural chromosomal aberration in which part of an arm of one chromosome is inserted into the arm of a non-homologous chromosome.

IN SITU HYBRIDIZATION: Performed by denaturing the DNA of cells squashed on a microscope slide so that reaction is possible with an added single-stranded RNA or DNA; the added preparation is radioactively labeled and its hybridization is followed by autoradiography.

INTRON: A segment of a gene that is initially transcribed but is then removed from within the primary transcript by splicing together the sequence on either side of it.

KARYOTYPE: The characteristic arrangement of an individual's chromosomes, arranged in pairs.

KILOBASE (Kb): A unit of 1,000 bases in DNA or RNA.

LEADING STRAND: Strand of DNA synthesized continuously in the 5' to 3' direction.

LETHALITY: A measure of a mutation's effect on the reproductive capability of the affected individual, unrelated to length of life.

LIBRARY: A set of cloned fragments together representing the entire genome.

LIGASE: Enzyme that catalyzes the repair of nicks in DNA.

LIGATION: The formation of a phosphodiester bond to link two adjacent bases separated by a nick in one strand of a double helix or DNA (The term can also be applied to blunt-end ligation and enjoining of RNA).

LINKAGE: Describes the tendency of genes to be inherited together as result of their location on the same chromosome; measured by percent recombination between loci.

LINKAGE GROUP: Includes all loci that can be connected (directly or indirectly) by linkage relationships; equivalent to a chromosome.

LIPOSOME: A structure with a lipid membrane like that of a cell that can be filled with specific substances and then used as a delivery vehicle to transport those substances to the interior of a target cell by fusion with the cell's own membrane.

LIQUID HYBRIDIZATION: See SOLUTION HYBRIDIZATION.

LOCUS: The position of a chromosome at which the gene for a particular trait resides; locus maybe occupied by any one of the alleles for the gene.

LOD SCORE: From "Logarithm of Odds," a computation of the likelihood of linkage between two loci at a specific recombinational frequency, theta (θ). A lod score of 3 for a θ of 0.30 means that the likelihood of linkage between the two loci at a distance of 30 centimeters is 1,000 times more likely than non-linkage.

MAP UNIT: A measure of distance between two loci on a chromosome bases on the percentage frequency of recombination between them; one map unit is equivalent to one centimorgan or one percent recombination.

MARKER (DNA): A fragment of known size used to calibrate an electrophoretic gel.

MARKER (GENETIC): Any allele of interest in an experiment.

MELTING TEMPERATURE (T_m)**:** The midpoint of the temperature range over which DNA is denatured.

MENDELIAN: Referring to a trait that is controlled by a single gene and which, therefore, shows a simple pattern of inheritance (dominant or recessive).

MESSENGER RNA (mRNA): An RNA transcribed from the DNA of a gene, forming the template for which a protein is translated.

MITOCHONDRIAL DNA: The DNA in the circular chromosome of the mitochondria, cytoplasmic organelles that possess their own unique DNA. Mitochondrial DNA is present in many copies per cell, is maternally inherited, and evolves five to ten times as rapidly as genomic DNA.

MOLECULAR GENETICS: The study of the nature and biochemistry of the genetic material. Includes the technologies of genetic engineering that involve the directed manipulation of the genetic material itself

MONOCLONAL ANTIBODIES: Antibodies derived from a single source or clone of cells that recognize only one kind of antigen.

NICK: A break in one strand of a DNA molecule in which no bases are removed.

NICK TRANSLATION: Describes the ability of E. coli DNA polymerase I to use a nick as a starting point from which one strand of a duplex DNA can be degraded and replaced by resynthesis of new material; is used to introduce radioactively labeled nucleotides into DNA in vitro.

NONSENSE CODON: Any one of three triplets (UAG, UAA, UGA) that cause termination of protein synthesis. Also called a Termination Codon.

NONSENSE MUTATION: Any change in DNA that causes a nonsense (termination) codon to replace a codon representing an amino acid.

NUCLEIC ACID: A polymer composed of DNA or RNA subunits.

NUCLEOTIDES: The fundamental units of nucleic acids. They consist of one of the four bases - adenine, guanine, cytosine, and thymine (uracil in the case of RNA) - and its attached sugar-phosphate group.

OLIGONUCLEOTIDE Nucleic acid molecules formed by the joining of a small number of nucleotide bases (generally fewer than 10 or 20). A short sequence of DNA or RNA.

PENETRANCE: Refers to the frequency with which the effects of a gene (whether dominant or recessive) known to be present are seen in the individuals carrying it.

PHAGE: See BACTERIOPHAGE.

PHENOTYPE: The observable properties of an organism resulting from the interaction between its genotype and the environment.

PHILADELPHIA CHROMOSOME (Ph^1): The structurally abnormal chromosome 22 typically occurring in a proportion of the bone marrow cells in most patients with chronic myelogenous leukemia. The abnormality is a reciprocal translocation between the distal portion of the long arm of chromosome 22 and the distal portion of the long arm chromosome 9.

PLASMID: Hereditary material that is not part of the chromosome. Plasmids are circular and self-replicating. Because they are generally small and relatively simple, they are used in recombinant DNA experiments as acceptors of foreign DNA.

PLEIOTROPY: If a single gene or gene pair produces multiple effects, it is said to exhibit pleiotropy (or have pleiotropic effects).

PLOIDY: The status of the chromosome set in the karyotype; also used as a suffix denoting the degree of multiplication of chromosome sets, as aneuploidy, diploidy, haploidy, etc.

POINT MUTATION: Substitutions of single base pairs within a DNA molecule.

POLYGENIC: Determined by many genes as different loci with small additive effects.

POLYMORPHISM: A gene or unexpressed DNA variant that occurs in a population with a frequency too great to be explained by mutation.

POLYPEPTIDE: A chain of amino acids, held together by peptide bonds between the amino group of one and the carboxyl group of an adjoining one.

PRIMARY TRANSCRIPT: The original unmodified RNA product corresponding to a transcription unit.

PROKARYOTE: Cell or organism without a nucleus separated by a membrane. The chromosomes are bathed directly in the cytoplasm. The best examples are the bacteria.

PROMOTER: Sequence of DNA giving the starting signal for transcription.

PROTEIN: A linear polymer of amino acids; proteins are the products of gene expression and are the functional and structural components of cells.

REASSOCIATION OF DNA: The pairing of complementary single strands to form a double helix.

RecA: The product of the recA locus of E. coli, a protein with dual activities, acting as protease and also able to exchange single strands of DNA molecules.

RECOMBINANT DNA: DNA molecules that have been assembled with the use of restriction enzymes, usually but not always by splicing together fragments from different species.

RENATURATION: The reassociation of denatured complementary strands of a DNA double helix.

REPLICATION: The formation of two new strands of DNA from existing DNA, permitting the reproduction of an identical new cell as the result of the division.

RESTRICTION ENDONUCLEASE: See RESTRICTION ENZYME.

RESTRICTION ENZYME: A nuclease that recognizes specific sequences in DNA and cleaves the DNA strand at those points; used in recombinant DNA technology.

RESTRICTION ENZYME SITES: The specific sequences in the DNA at which one of the restriction enzymes recognizes and cuts the DNA.

RESTRICTION FRAGMENT LENGTH POLYMORPHISM (RFLP): A polymorphism in DNA sequence that can be detected on the basis of differences in the length of DNA fragments produced by digestion with a specific restriction enzyme.

RESTRICTION MAPPING: A linear array of sites on DNA cleaved by various restriction enzymes.

REVERSE TRANSCRIPTASE: An enzyme that catalyzes the synthesis of DNA on an RNA template.

REVERSE TRANSCRIPTION: Synthesis of DNA on a template of RNA accomplished by Reverse Transcriptase.

RIBONUCLEIC ACID (RNA): A polynucleotide consisting of a backbone of alternating phosphate and sugar (ribose) molecules to which are attached the nucleotide bases adenine (A), thymine (T), guanine (G), and uracil (U, which replaces the cytosine, C, of DNA). Three are several classes of RNA that serve different purposes, including messenger RNA (mRNA), transfer RNA (tRNA), and ribosomal RNA (rRNA).

RIBOSOME: Cytoplasmic organelles composed of ribosomal RNA and proteins, on which polypeptide synthesis from messenger RNA occurs.

RNA-DRIVEN HYBRIDIZATION REACTIONS: Reactions that use an excess of RNA to react with all complementary sequences in a single-stranded preparation of DNA

SANDWICH HYBRIDIZATION: A technique designed to simplify pretreatment of sample DNA and increase the rate of hybridization of the DNA probe with its target sequence in the clinical sample by allowing annealing to occur in solution rather than on a filter. The term is derived from the nature of reaction, which involves a three-component hybridization system. Two of the components are non-overlapping DNA fragments complementary to the target DNA in the sample, which is the third component. One fragment, the labeled probe DNA, hybridizes with the sample DNA in solution. This hybrid is then "captured" by the second complementary DNA fragment, which is immobilized on the filter.

SATURATION HYBRIDIZATION EXPERIMENT: A large excess of one component, causing all complementary sequences in the other component to enter a duplex form.

"SHOTGUN" METHOD: A technique for obtaining the desired gene that involves chopping up the entire genetic complement of a cell using restriction enzymes, then attaching each DNA fragment to a vector and transferring it into a bacterium and, finally, screening the bacteria to locate those producing the desired product.

SINGLE GENE DISORDER (MENDELIAN DISORDER): A genetic disease caused by a single gene that shows a simple pattern of inheritance (e.g., dominant or recessive, autosomal or X-linked).

SINGLE LOCUS PROBE: This type of probe recognizes a single region on a specific chromosome. Because there are pairs of chromosomes, most patterns from a SLP will produce a two-band pattern.

SIZE MARKER: DNA fragments of known molecular weight and base pair length, such as plasmid and phages digested with restriction enzymes, run on electrophoresis gels for the determination of DNA sample fragment sizes.

SOUTHERN BLOT: A technique for transferring DNA fragments separated by agarose gel electrophoresis to a nitrocellulose filter, on which specific DNA fragments can then be detected by their hybridization to radioactive probes.

STICKY ENDS: Complementary single strands of DNA that protrude from opposite ends of a duplex or from ends of different duplex molecules; can be generated by staggered cuts in duplex DNA.

STRINGENCY: The buffer salt concentration and temperature used in the DNA blot wash post-hybridization process. As these parameters are changed, the degree of binding of probe to target DNA changes.

SYNTENY: Presence together on the same chromosome of two or more gene loci, whether or not they are close enough together for linkage to be demonstrated. Adjective: Syntenic.

TANDEM REPEATS: Multiple copies of the same DNA sequence arranged in direct succession along a chromosome.

TERMINATION CODON: One of the three codons (UAG, UAA, or UGA) that cause termination of protein synthesis. Also called a NONSENSE CODON.

TRANSCRIPTION: The synthesis of RNA on a DNA template.

TRANSDUCTION: The process by which foreign DNA becomes incorporated into the genetic complement of the host cell.

TRANSFORMATION: The transfer of genetic information by DNA separated from the cell.

TRANSLATION: The synthesis of a polypeptide with an amino acid sequence derived from the codon sequence of a corresponding messenger RNA.

TRANSLOCATION: The transfer of a segment of one chromosome to another chromosome. If two non-homologous chromosomes exchange pieces, the translation is reciprocal.

TRANSPORTATION IMMUNITY: Refers to the ability of certain transposons to prevent others of the same type from transposing to the same DNA molecule.

TRANSPOSON: A DNA sequence able to replicate and insert one copy at a new location in the genome.

TRIPLET: In molecular genetics, a unit of three DNA or RNA bases, coding for a specific amino acid; a CODON.

VARIABLE NUMBER OF TANDEM REPEATS (VNTR): The variable number of repeat core base pair sequences found at specific loci in the genome. The variation in length of the alleles formed from the repeats provides the basis for unique individual identification.

VECTOR: A transmission agent; a DNA vector is a self replicating DNA molecule that transfers a piece of DNA from one host to another.

VIRUS: An infectious agent that requires a host cell in order for it to replicate. It is composed of either RNA or DNA wrapped in a protein coat.

WASH: The process of removing non-bound or loosely bound probe from blots after hybridization. This process reduces background interference.

APPENDIX B
The First DNA Decision and Appeal Per State in the United States

Information taken from
New Frontiers in Evidence: Daubert, Lanigan, and DNA Testing,
prepared by the Massachusetts Bar Association, 1995.

Alabama

First Case: *People v. Perry,* guilty of rape, Scottsboro, 1988
Appellate Decision: Accepted DNA, remanded Perry to trial court on basis of statistics and three-pronged Frye standard, 1991, trial court found general acceptance of statistical analysis, 1992.
Data Bank established: 1994
Legislated Admissibility passed: 1994

Alaska

First Case: DNA evidence accepted in criminal trial in Kodiak, 1990.
Data Bank established: no
Legislated Admissibility passed: no

Arizona

First case: People v. *Bible,* Superior Court judge ruled to allow the introduction of DNA evidence in the murder trial of Richard Bible. Bible was charged with murder, kidnapping, and child molestation in the disappearance and death of a 9-year-old girl. Cocinino County, 1989.
Appellate Decision: *People v. Bible,* 1993. DNA evidence admitted, but statistical estimates excluded
Data Bank Established: 1990
Legislated Admissibility passed: no

Arkansas

DNA evidence was admitted in a Little Rock rape case following a pretrial hearing in November 1989.
Appellate Decision: *Prater v. Arkansas,* 1991. Affirmed DNA admissibility.
Data Bank established: no
Legislated Admissibility passed: no

California

First case: *People v. Axell,* guilty of murder, Ventura County Superior Court, 1989.
Appellate Decision: *People v.* Soto, 1994. Affirmed admission of evidence and statistics.
Data Bank established: 1990
Legislated Admissibility passed: no

Colorado

First case: *People v. Gallagher,* guilty of sexual assault, El Paso County, 1988.
Appellate Decision: *Fishback v. State of Colorado,* 1993. Admitted DNA evidence and left statistical frequency questions to trial courts.
Data Bank established: 1990
Legislated Admissibility passed: no

Connecticut

First case: *People v. Williams,* PCR test conducted fails to show link between defendant and woman stabbed to death in a shopping center parking lot. Stamford, 1989.
Appellate Decision: *State v. Silvri,* 1994. Judgment reversed and case remanded with recommendation to conform with NRC report.
Data Bank established: 1994
Legislated Admissibility passed: 1994

District of Columbia

First case**:** *Green v. District of Columbia.* After being convicted in June 1989, Green pursued DNA testing, which revealed that he was not the individual whose semen had stained the victim's clothing. Rape, Kidnapping and sodomy charges dropped in 1990.
Appellate Decision: *U.S.* v. *Porter,* 1994, Affirmed admissibility and use of ceiling principle.
Data Bank established: no
Legislated Admissibility passed: no

Florida

First case: *People v. Andrews,* guilty of rape, 1987
Appellate Decision: *Toranzo v. Florida,* 1992. Affirmed admissibility and frequency estimate.
Data Bank established: 1990
Legislated Admissibility passed: no

Georgia

First Case: *People v. Redding,* DNA evidence admitted after pretrial hearing in rape case. Defendant pleaded guilty. Decatur, 1988.
Appellate Decision: *Morris v. State,* 1994. Affirmed use of evidence and statistics
Data Bank established: 1992
Legislated Admissibility passed: no

Hawaii

First case: *People v. Manning,* guilty of assault and burglary, Wailuku, 1989.
Appellate Decision: *State v. Montalbo,* 1992. Admitted DNA evidence and found objections regarding statistical calculations go to weight rather than admissibility
Data Bank established: 1992
Legislated Admissibility passed: no

Idaho

First case: *Peoples v. Horsley,* guilty of rape, Sandpoint, 1988.
Data Bank established: no
Legislated Admissibility passed: no

Illinois

First case: Case of Gary Dotson. Cook County judge vacates 1979 rape conviction after DNA test exculpates defendant. Dotson had been convicted in a 1977 rape, for which he spent 6 years in jail. Alleged victim recanted story in 1985, and Governor James Thompson granted clemency but rape conviction remained on his record. Dotson had requested that the case be reopened for new trial after PCR tests excluded him. Cook County, 1989.
Appellate Decision: *People v. Liscomb,* 1991. Accepted DNA tests and statistical calculations.
Data Bank established: 1990
Legislated Admissibility passed: no

Indiana

First case: *Peoples. Hopkins,* guilty of rape/sodomy/murder, Fort Wayne, 1989.
Appellate Decision: *McElroy v.* State, 1992. Court upheld DNA admissibility and frequency calculations.
Data Bank established: 1990.
Legislated Admissibility passed: 1991

Iowa

First case: *People v. Vargason,* guilty of sexual abuse, Johnson County, 1989.
Appellate Decision: *State v. Ripperger,* 1994. Affirmed use of evidence and statistics.
Data Bank established: 1990
Legislated Admissibility passed: no

Kansas

First case: *People s. Pioletti,* guilty of murder, Wichita, 1988.
Appellate Decision: *Stare v. Wilson,* 1991. Ruled that procedural questions related to weight of evidence rather than admissibility.
Data Bank established: 1991
Legislated Admissibility passed: no

Kentucky

Appellate Decision: *Harris* v. *Commonwealth,* 1992. Affirmed the use of DNA evidence and statistics.
Data Bank established: 1992
Legislated Admissibility passed: no.

Louisiana

First case: *People v. Quatrevingt,* guilty of murder/rape, New Orleans, 1990.
Appellate Decision: State v. Quatrevignt, 1992. Ruled DNA evidence is pro se admissible under Louisiana statute.
Data Bank established: no
Legislated Admissibility passed: 1990

Maine

First case: *People v. McLeod.* The prosecution, in sexual molestation case, withdrew DNA evidence during a preliminary hearing on the reliability of the data, Portland, 1989.
Data Bank established: no
Legislated Admissibility passed: no

Maryland

First case: *People v. Tasker.* Defendant pleads guilty to second-degree rape and draws 5-year prison sentence in case where DNA evidence introduced. Anne Arundel County, 1988.
Data Bank established: 1994
Legislated Admissibility passed: 1991

Massachusetts

First case: *People v. Curnin,* guilty of rape, Worcester, 1989.
Appellate Decision: *Commonwealth v. Daggett,* 1993. The Supreme Court, unable to reach a consensus on the admissibility of DNA evidence, ruled that even if the evidence was erroneously admitted, it was a harmless error.
Data Bank established: no
Legislated Admissibility passed: no

Michigan

First case: *People v. Fagan,* guilty of rape, Flint, 1988.
Data Bank established: 1994
Legislated Admissibility passed: no

Minnesota

First case: *People v. Nielson,* guilty of murder, Ramsey County, 1989.
Appellate Decision: *State v. Johnson,* 1993. Upheld admissibility of DNA evidence including frequency statistics for each individual locus and not the product of all loci.
Data Bank established: 1990
Legislated Admissibility patted: 1989

Mississippi

First case: *People* v. *Weaver,* guilty of rape, Hinds County 1989.
Appellate Decision: *Polk v. State,* 1992. Court found 3-prong Frye standard satisfied and evidence properly admitted.
Data Bank established: from and after January 6, 1996
Legislated Admissibility passed: no

Missouri

First case: *People v. Thomas,* guilty of rape, St. Louis, 1989.
Data Bank established: 1990
Legislated Admissibility passed: no

Montana

First case: *People v. Drummond.* Sexual intercourse without consent allegedly committed by a State institution attendant against a developmentally disabled patient. The victim gave birth and the DNA comparisons were done by Lifecodes. Defendant pleaded guilty. Jefferson County. 1989.
Data Bank established: no
Legislated Admissibility passed: no

Nevada

Data Bank established: 1990
Legislated Admissibility passed: 1989

New Hampshire

First case: *People v. Barnaby.* DNA analysis admitted, according to State Attorney General's Office. Hillsborough County, 1989.
Appellate Decision: Stares'. *Vandebogart,* 1992. Reversed admission of DNA evidence and remanded case to trial court to determine if ceiling principle was acceptable, which the trial court did.
Data Bank established: no
Legislated Admissibility passed: no

New Jersey

First case: *People v. Beard.* Nearly 3 months after being charged with murder, the defendant was released after a judge ruled that authorities arrested the wrong man. The primary suspect in a 1975 Georgia murder disappeared after the crime. Mistakenly, his cousin (the defendant in this case) was arrested. DNA tests conducted by Lifecodes proved the jailed man was not the father of a man known to be the son of the suspect. Union County, 1989.
Appellate decision: *State v. Williams,* 1991. Affirmed the use of PCR.
Data Bank established: no
Legislated Admissibility passed: no

New Mexico

First case: *People v. Collins.* A man once charged with killing his stepdaughter was released from prison 1 October 1989 pending the outcome of DNA testing.
Open murder charges against the defendant were dismissed in the interim. Santa Fe, 1989.
Appellate Decision: *State v. Anderson,* 1994. Reversed Court of Appeals ruling that DNA evidence, admitted by trial court, was inadmissible due to statistical methodology.
Data Bank established: no
Legislated Admissibility passed: no

New York

First case: *People v. Zambrana,* guilty of murder, New City, 1987.
Appellate Decision: *Peoples. Wesley,* 1994. Affirmed admissibility and statistical estimates.
Data Bank established: 1994
Legislated Admissibility passed: no

North Carolina

First case: *People v. Mills,* guilty of murder, Salisbury, 1989.
Appellate decision: *Stare v. Pennington,* 1990. Affirmed use of DNA evidence and statistics.
Data Bank established: 1993.
Legislated Admissibility passed: no

North Dakota

Data Bank established: no
Legislated Admissibility passed: no

Ohio

First case: *People v. Dascenzo,* guilty of aggravated murder, Montgomer County, 1988.
Appellate Decision: Ohio *v. Penton,* 1993. Affirmed PCR and reiterated relevancy as admissibility standard rather than *Frye* standard.
Data Bank established: no
Legislated Admissibility passed: no

Oklahoma

First case: *People v. Hunt,* first time Lifecodes testifies regarding DNA evidence in criminal case, defendant acquitted of murder, Norman, 1987.
Data Bank established: 1991
Legislated Admissibility passed: no

Oregon

First case: *People v. Gutch,* DNA test results offered for admission at 7-month pretrial hearing, Clatsop County, 1989.
Appellate Decision: *State v. Lyons,* 1993. Affirmed use of DNA evidence and statistics.
Data Bank established: 1991
Legislated Admissibility passed: no

Pennsylvania

First case: *People v. Trubia,* guilty of murder/rape, Lackawanna County, 1988.
Appellate Decision: *Commonwealth v. Rogers,* 1992. Affirmed DNA admissibility under Frye.
Data Bank established: no
Legislated Admissibility passed: no

Rhode Island

First case: In re: Case involving rape of a nursing home resident. Juvenile admitted sufficient facts to establish the charge against him; sentenced to 3 years at a juvenile facility. 1989.
Data Bank established: no
Legislated Admissibility passed: no

South Carolina

First case: *People* v. *Evans,* guilty of rape/burglary, Charleston County, 1989.
Appellate Decision:
Data Bank established: no
Legislated Admissibility passed: no

South Dakota

First case: *People v. Wimberly,* guilty of rape, Meade County, 1989.
Appellate Decision: *State v. Wimberly,* Affirmed use of evidence and statistics.
Data Bank established: 1990
Legislated Admissibility passed: no

Tennessee

First case: FBI testimony in rape case in Blountville results in admission of DNA evidence, 1989, Trial pending.
Appellate Decision: *People v. Harris,* 1992. Affirmed admissibility and frequency estimate under both Frye and relevancy test.
Data Bank established: 1991
Legislated Admissibility passed: 1991

Texas

First case: *People v. Trimboli,* A DNA test that triple-murder defendant Ronald Trimboli had hoped would clear his name instead gave prosecutors additional evidence against him. Test concluded that semen found on the bedspread where one of the three victims was raped matched a sample Trimboli had given for the test. Trimboli's two earlier trials for the three murders both ended in mistrials, first because of jury misconduct and later because a jury deadlocked, 6 to 6. The third trial resulted in a conviction on all three counts of murder. Tarrant County, test in 1988, convicted in 1989.
Appellate Decision: *Kelly v. Texas,* 1992. Affirmed DNA admissibility and concluded Frye was no longer part of Texas law.
Data Bank established: no
Legislated Admissibility passed: no

Utah

First case: *People v. Bickmore,* DNA evidence admitted, mistrial declared on other grounds, Ogden, 1989.
Data Bank established: 1994
Legislated Admissibility passed: no

Vermont

First case: *United States v. Jakobtz.* Admissibility hearing pending in rape case. Defense attorney has filed request that genetic evidence not be used in court. In August 1989, judge ruled that hair, blood, and saliva samples could be taken from defendant for testing. U.S. District Court, Burlington, 1989.
Data Dank established: no
Legislated Admissibility passed: no

Virginia

First case: *People v. Reynolds,* DNA evidence admitted in murder case, Henrico County, 1988.
Appellate Decision: *Spencer v. Commonwealth,* 1990. Affirmed use of DNA evidence.
Data Bank established: 1990
Legislated Admissibility passed: 1990

Washington

First case: *People v. Young,* DNA tests exclude Young, who had been identified by the victim as the rapist. Charges dropped, Snohomish County, 1989.
Appellate Decision: *State v. Cauthron,* 1993. Reversed trial courts admission of DNA match without population statistics and remanded.
Data Bank established: 1990
Legislated Admissibility passed: no

West Virginia

First case: *People v. Ferrell,* guilty of murder, Petersburg, 1989.
Data Bank established: 1993
Legislated Admissibility passed: no

Wisconsin

First case: *People v. Banks,* guilty of rape, Kenosha County 1989.
Data Bank established: 1993
Legislated Admissibility passed: 1993

Wyoming

First case:
Appellate Decision: *Springfield v. State,* 1993. Affirmed use of DNA evidence and statistical calculations.
Data Bank established: no
Legislated Admissibility passed: no

Military

First case: *United States v.* Scott. Rape case. Military judge approved request for DNA tests, but DNA sample too degraded to perform the testing. U.S. Marine Corp. 1988.

APPENDIX C

Information taken from http://www.fbi.gov/lab/report/dnatest.htm

Quality Assurance Standards for Forensic DNA Testing Laboratories

Preface

Throughout its deliberation concerning these quality standards, the DNA Advisory Board recognized the need for a mechanism to ensure compliance with the standards. An underlying premise for these discussions was that accreditation would be required to demonstrate compliance with the standards and therefore assure quality control and a quality program. Accordingly, the Board recommends that forensic laboratories performing DNA analysis seek such accreditation with all deliberate speed. Additionally, the Board strongly encourages the accrediting bodies to begin positioning themselves to accommodate the increasing demand for accreditation.

Proposed Mechanism to Recommend Changes to Standards

Once the Director of the FBI has issued standards for quality assurance for forensic DNA testing, the DNA Advisory Board may recommend revisions to such standards to the FBI Director, as necessary. In the event that the duration of the DNA Advisory Board is extended beyond March 10, 2000 by the FBI Director, the Board may continue to recommend revisions to such standards to the FBI Director. In the event that the DNA Advisory Board is not extended by the FBI Director after March 10, 2000, the Technical Working Group on DNA Analysis Methods [TWGDAM] may recommend revisions to such standards to the FBI Director, as necessary.

Effective Date

These standards shall take effect October 1, 1998.

3. QUALITY ASSURANCE PROGRAM

Standard 3.1 The laboratory shall establish and maintain a documented quality system that is appropriate to the testing activities.

3.1.1 The quality manual shall address at a minimum:

(a) Goals and objectives
(b) Organization and management
(c) Personnel qualifications and training
(d) Facilities
(e) Evidence control
(f) Validation
(g) Analytical procedures
(h) Calibration and maintenance
(i) Proficiency testing
(j) Corrective action
(k) Reports
(l) Review
(m) Safety
(n) Audits

4. ORGANIZATION AND MANAGEMENT

Standard 4.1 The laboratory shall:

4.1.1 (a) have a managerial staff with the authority and resources needed to discharge their duties and meet the requirements of the standards in this document.

(b) have a technical manager or leader who is accountable for the technical operations.

(c) specify and document the responsibility, authority, and interrelation of all personnel who manage, perform, or verify work affecting the validity of the DNA analysis.

5. PERSONNEL

Standard 5.1 Laboratory personnel shall have the education, training, and experience commensurate with the examination and testimony provided. The laboratory shall:

5.1.1 have a written job description for personnel to include responsibilities, duties, and skills.

5.1.2 have a documented training program for qualifying all technical laboratory personnel.

5.1.3 have a documented program to ensure technical qualifications are maintained through continuing education.

5.1.3.1 Continuing education -the technical manager or leader and examiner/analyst(s) must stay abreast of developments within the field of DNA typing by reading current scientific literature and by attending seminars, courses, professional meetings, or documented training sessions/ classes in relevant subject areas at least once a year.

5.1.4 maintain records on the relevant qualifications, training, skills, and experience of the technical personnel.

Standard 5.2 The technical manager or leader shall have the following:

5.2.1 **Degree requirements:** The technical manager or leader of a laboratory shall have at a minimum a Master's degree in biology-, chemistry-, or forensic science-related area and successfully completed a minimum of 12 semester or equivalent credit hours of a combination of undergraduate and graduate course work covering the subject areas of biochemistry, genetics, and molecular biology (molecular genetics, recombinant DNA technology), or other subjects which provide a basic understanding of the foundation of forensic DNA analysis as well as statistics and/or population genetics as it applies to forensic DNA analysis.

5.2.1.1 The degree requirements of section 5.2.1 maybe waived by the American Society of Crime Laboratory Directors (ASCLD) or other organization designated by the Director of the FBI in accordance with criteria approved by the Director of the FBI. This waiver shall be available for a period of two years from the effective date of these standards. The waiver shall be permanent and portable.

5.2.2 **Experience requirements:** A technical manager or leader of a laboratory must have a minimum of three years of forensic DNA laboratory experience.

5.2.3 **Duty requirements:**

5.2.3.1 **General:** manages the technical operations of the laboratory.

5.2.3.2 Specific duties:

(a) is responsible for evaluating all methods used by the laboratory and for proposing new or modified analytical procedures to be used by examiners.

(b) is responsible for technical problem solving of analytical methods and for the oversight of training, quality assurance, safety, and proficiency testing in the laboratory.

5.2.3.3 The technical manager or leader shall be accessible to the laboratory to provide onsite, telephone, or electronic consultation as needed.

Standard 5.3 Examiner/Analyst shall have:

5.3.1 a minimum of a BA/BS degree or its equivalent degree in biology-, chemistry, or forensic science- related area and must have successfully completed college course work (graduate or undergraduate level) covering the subject areas of biochemistry, genetics, and molecular biology (molecular genetics, recombinant DNA technology) or other subjects which provide a basic understanding of the foundation of forensic DNA analysis, as well as course work and/or training in statistics and population genetics an it applies to forensic DNA analysis.

5.3.2 a minimum of six months of forensic DNA laboratory experience, including the successful analysis of a range of samples typically encountered in forensic case work prior to independent case work analysis using DNA technology.

5.3.3 successfully completed a qualifying test before beginning independent casework responsibilities.

Standard 5.4 Technician shall have:

5.4.1 on-the-job training specific to their job function(s).

5.4.2 successfully completed a qualifying test before participating in forensic DNA typing responsibilities.

Standard 5.5 Laboratory support personnel shall have:

5.5.1 training, education, and experience commensurate with their responsibilities an outlined in their job description.

6. FACILITIES

Standard 6.1 The laboratory shall have a facility that is designed to provide adequate security and minimize contamination.

The laboratory shall ensure that:

6.1.1 access to the laboratory is controlled and limited.

6.1.2 prior to PCR amplification, evidence examinations, DNA extractions, and PCR setup are conducted at separate times or in separate spaces.

6.1.3 amplified DNA product is generated, processed, and maintained in a room(s) separate from the evidence examination, DNA extractions, and PCR setup areas.

6.1.4 the laboratory follows written procedures fur monitoring, cleaning, and decontaminating facilities and equipment.

7. EVIDENCE CONTROL

Standard 7.1 The laboratory shall have and follow a documented evidence control system to ensure the integrity of physical evidence.
This system shall ensure that:

- 7.1.1 evidence is marked for identification.
- 7.1.2 chain-of-custody for all evidence is maintained.
- 7.1.3 the laboratory follows documented procedures that minimize toss, contamination, and/or deleterious change of evidence.
- 7.1.4 the laboratory has secure areas for evidence storage.

Standard 7.2 Where possible, the laboratory shall retain or return a portion of the evidence sample or extract.

- 7.2.1 The laboratory shall have a procedure requiring that evidence sample/extract(s) are stored in a manner that minimizes degradation.

8. VALIDATION

Standard 8.1 The laboratory shall use validated methods and procedures for forensic casework analyses.

- 8.1.1 Developmental validation that is conducted shall be appropriately documented.
- 8.1.2 Novel forensic DNA methodologies shall undergo developmental validation to ensure the accuracy, precision, and reproducibility of the procedure. The developmental validation shall include the following:
  - 8.1.2.1 Documentation exists and is available which defines and characterizes the locus.
  - 8.1.2.2 Species specificity, sensitivity, stability, and mixture studies are conducted.
  - 8.1.2.3 Population distribution data are documented and available.
    - 8.1.2.3.1 The population distribution data would include the allele and genotype distributions for the locus or loci obtained from relevant populations. Where appropriate, databases should be tested for independence expectations.
- 8.1.3 Internal validation shall be performed and documented by the laboratory.
  - 8.1.3.1 The procedure shall be tested using known and non-probative evidence samples. The laboratory shall monitor and document the reproducibility and precision of the procedure using human DNA control(s).
  - 8.1.3.2 The laboratory shall establish and document match criteria based on empirical data.
  - 8.1.3.3 Before the introduction of a procedure into forensic casework, the analyst or examination team shall successfully complete a qualifying test.

8.1.3.4 Material modifications made to analytical procedures shall be documented and subject to validation testing.

8.1.4 Where methods are not specified, the laboratory shall, wherever possible, select methods that have been published by reputable technical organizations or in relevant scientific texts or journals or have been appropriately evaluated for a specific or unique application.

9. ANALYTICAL PROCEDURES

Standard 9.1 The laboratory shall have and follow written analytical procedures approved by the laboratory management/technical manager.

9.1.1 The laboratory shall have a standard operating protocol for each analytical technique used.

9.1.2 The procedures shall include reagents, sample preparation, extraction, equipment, and controls, which are standard for DNA analysis and data interpretation.

9.1.3 The laboratory shall have a procedure for differential extraction of stains that potentially contain semen.

Standard 9.2 The laboratory shall use reagents that are suitable for the methods employed.

9.2.1 The laboratory shall have written procedures for documenting commercial supplies and for the formulation of reagents.

9.2.2 Reagents shall be labeled with the identity of the reagent, the date of preparation or expiration, and the identity of the individual preparing the reagent.

9.2.3 The laboratory shall identify critical reagents and evaluate them prior to use in casework. These critical reagents include but are not limited to:

(a) Restriction enzyme
(b) Commercial kits for performing genetic typing
(c) Agarose for analytical RFLP gels
(d) Membranes for Southern blotting
(e) K562 DNA or other human DNA controls
(f) Molecular weight markers used as RFLP sizing standards
(g) Primer sets
(h) Thermostable DNA polymerase

Standard 9.3 The laboratory shall have and follow a procedure for evaluating the quantity of the human DNA in the sample where possible.

9.3.1 For casework RFLP samples, the presence of high molecular weight DNA should be determined.

Standard 9.4 The laboratory shall monitor the analytical procedures using appropriate controls and standards.

9.4.1 The following controls shall be used in RFLP casework analysis:

9.4.1.1 Quantitation standards for estimating the amount of DNA recovered by extraction.

9.4.1.2 K562 as a human DNA control. (In monitoring sizing data, a statistical quality control method for K562 cell line shall be maintained.)

9.4.1.3 Molecular weight size markers to bracket known and evidence samples.

9.4.1.4 Procedure to monitor the completeness of restriction enzyme digestion.

9.4.2 The following controls shall be used for PCR casework analysis:

9.4.2.1 Quantitation standards which estimate the amount of human nuclear DNA recovered by extraction.

9.4.2.2 Positive and negative amplification controls.

9.4.2.3 Reagent blanks.

9.4.2.4 Allelic ladders and/or internal size makers for variable number tandem repeat sequence PCR-based systems.

Standard 9.5 The laboratory shall check its DNA procedures annually or whenever substantial changes are made to the protocol(s) against an appropriate and available NIST standard reference material or standard traceable to a NIST standard.

Standard 9.6 The laboratory shall have and follow written general guidelines for the interpretation of data.

9.6.1 The laboratory shall verity that all control results are within established tolerance limits.

9.6.2 Where appropriate, visual matches shall be supported by a numerical match criterion.

9.6.3 For a given population(s) and/or hypothesis of relatedness, the statistical interpretation shall be made following the recommendations 4.1,4.2, or 4.3 as deemed applicable of the National Research Council report entitled "The Evaluation of Forensic DNA Evidence' (1996) and/or court-directed method. These calculations shall be derived from a documented population database appropriate for the calculation.

10. EQUIPMENT CALIBRATION AND MAINTENANCE

Standard 10.1 The laboratory shall use equipment suitable for the methods employed.

Standard 10.2 The laboratory shall have a documented program for calibration of instruments and equipment.

10.2.1 Where available and appropriate, standards traceable to national or international standards shall be used fur the calibration.

10.2.1.1 Where traceability to national standards of measurement is not applicable, the laboratory shall provide satisfactory evidence of correlation of results.

10.2.2 The frequency of the calibration shall be documented for each instrument requiring calibration. Such documentation shall be retained in accordance with applicable Federal or state law.

Standard 10.3 The laboratory shall have and follow a documented program to ensure that instruments and equipment are properly maintained.

10.3.1 New instruments and equipment, or instruments and equipment that have undergone repair or maintenance, shall be calibrated before being used in casework analysis.

10.3.2 Written records or logs shall be maintained for maintenance service performed on instruments and equipment. Such documentation shall be retained in accordance with applicable Federal or state law.

11. REPORTS

Standard 11.1 The laboratory shall have and follow written procedures for taking and maintaining case notes to support the conclusions drawn in laboratory reports.

11.1.1 The laboratory shall maintain, ins case record, all documentation generated by examiners related to case analyses.

11.1.2 Reports according to written guidelines shall include:

(a) Case identifier
(b) Description of evidence examined
(c) A description of the methodology
(d) Locus
(e) Results and/or conclusions
(f) An interpretative statement (either quantitative or qualitative)
(g) Date issued
(h) Disposition of evidence
(i) A signature and title, or equivalent identification, of the person(s) accepting responsibility for the content of the report

11.1.3 The laboratory shall have written procedures for the release of case report information.

12. REVIEW

Standard 12.1 The laboratory shall conduct administrative and technical reviews of all case files and reports to ensure conclusions and supporting data are reasonable and within the constraints of scientific knowledge.

12.1.1 The laboratory shall have a mechanism in place to address unresolved discrepant conclusions between analysts and reviewer(s).

Standard 12.2 The laboratory shall have and follow a program that documents the annual monitoring of the testimony of each examiner.

13. PROFICIENCY TESTING

Standard 13.1 Examiners and other personnel designated by the technical manager or leader who are actively engaged in DNA analysis shall undergo, at regular intervals of not to exceed 180 days, external proficiency testing In **accordance with these standards. Such external proficiency testing shall be an open proficiency testing program.**

13.1.1 The laboratory shall maintain the following records for proficiency tests:

(a) The test set identifier
(b) Identity of the examiner
(c) Date of analysis and completion
(d) Copies of all data and notes supporting the conclusions
(e) The proficiency test results
(f) Any discrepancies noted
(g) Corrective actions taken

Such documentation shall be retained in accordance with applicable Federal or state law.

13.1.2 The laboratory shall establish at a minimum the following criteria for evaluation of proficiency tests:

(a) All reported inclusions are correct or incorrect.
(b) All reported exclusions are correct or incorrect.
(c) All reported genotypes and/or phenotypes are correct or incorrect according to consensus genotypes/phenotypes or within established empirically determined ranges.
(d) All results reported as inconclusive or uninterpretable are consistent with written laboratory guidelines. The basis for inconclusive interpretations in proficiency tests must be documented.
(e) All discrepancies/errors and subsequent corrective actions must be documented.

(f) All final reports are graded as satisfactory or unsatisfactory. A satisfactory grade is attained when there are no analytical errors for the DNA profile typing data Administrative errors shall be documented and corrective actions taken to minimize the errors in the future.
(g) All proficiency test participants shall be informed of the final test results.

14. CORRECTIVE ACTION

Standard 14.1 The laboratory shall establish and follow procedures for corrective action whenever proficiency testing discrepancies and/or casework errors are detected.

14.1.1 The laboratory shall maintain documentation for the corrective action. Such documentation shall be retained in accordance with applicable Federal or state law.

15. AUDITS

Standard 15.1 The laboratory shall conduct audits annually in accordance with the standards outlined herein.

15.1.1 Audit procedures shall address at a minimum:
(a) Quality assurance program
(b) Organization and management
(c) Personnel
(d) Facilities
(e) Evidence control
(f) Validation
(g) Analytical procedures
(h) Calibration and maintenance
(I) Proficiency testing
(j) Corrective action
(k) Reports
(1) Review
(m) Safety
(n) Previous audits

15.1.2 The laboratory shall retain all documentation pertaining to audits in accordance with relevant legal and agency requirements.

Standard 15.2 Once every two years, a second agency shall participate in the annual audit.

16. SAFETY

Standard 16.1 The laboratory shall have and follow a documented environmental health and safety program.

17. SUBCONTRACTOR OF ANALYTICAL TESTING FOR WHICH VALIDATED PROCEDURES EXIST

Standard 17.1 A laboratory operating under the scope of these standards will require certification of compliance with these standards when a subcontractor performs forensic DNA analyses for the laboratory.

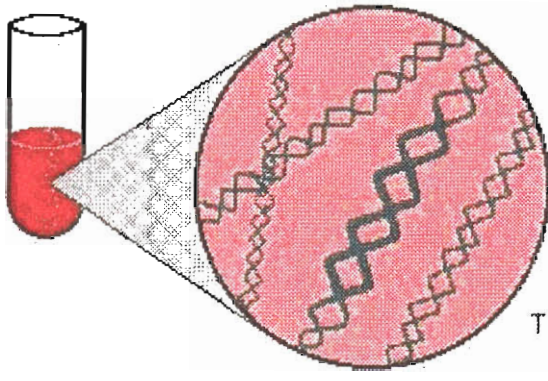
17.1.1 The laboratory will establish and use appropriate review procedures to verify the integrity of the data received from the subcontractor.

APPENDIX D

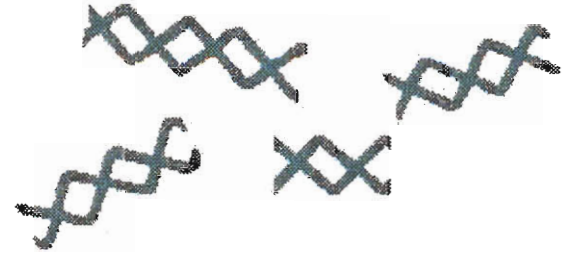
"Making a DNA Fingerprint"

Screen captures taken from http://vector.cshl.org/resources/southernblotting.html

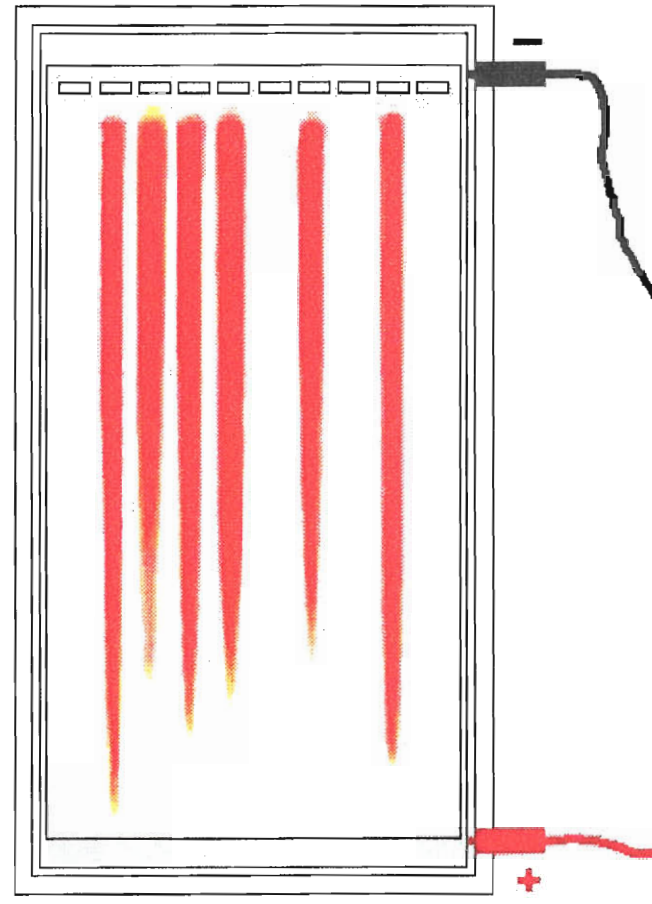
DNA is extracted from samples of blood, semen, saliva, or hair roots.



Then the DNA is cut up into a precise set of fragments with restriction enzymes.



The DNA fragments are electrophoresed on an agarose gel, which acts as a molecular sieve to separate the fragments by size. After staining, the DNA appears as a smear in each lane, indicating the presence of thousands of restriction fragments ranging in size from very long (at the top) to very short (at the bottom).



The gel is placed on a blotting apparatus consisting of:

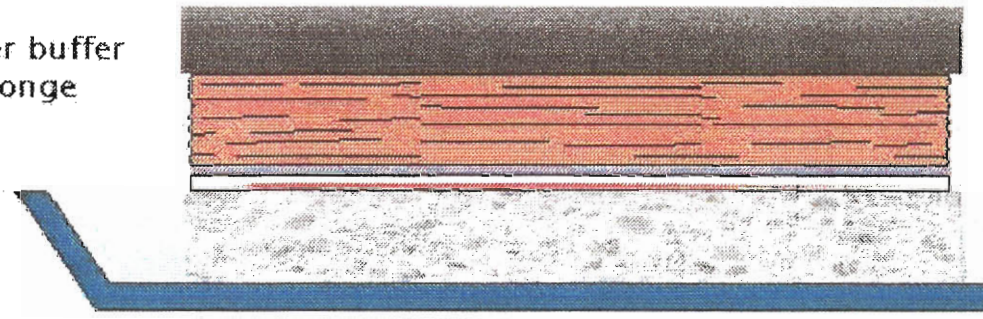
Weight

Paper Towels

Nylon Membrane

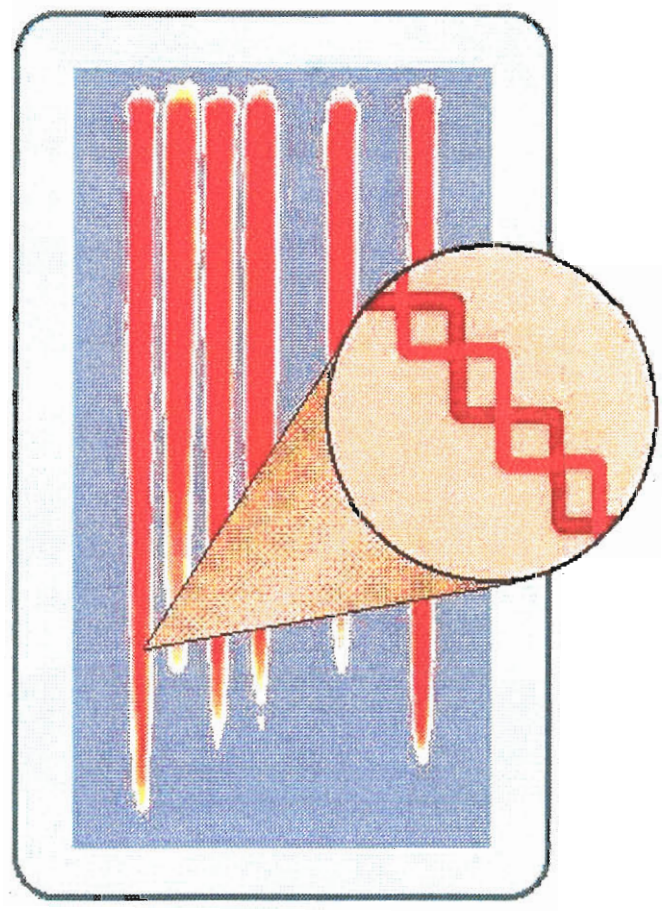
Gel

Transfer buffer with sponge



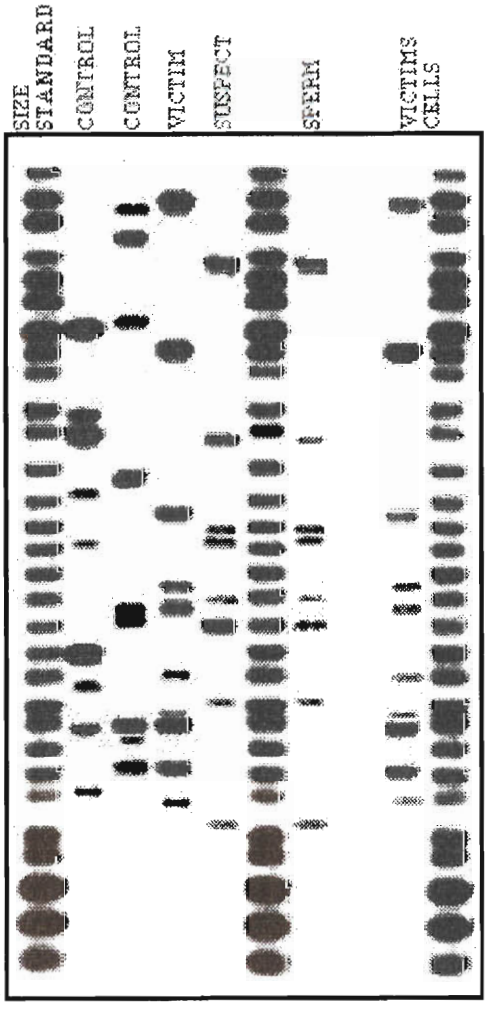
The DNA is transferred to the nylon membrane when the transfer buffer is absorbed by the paper towels. The buffer is alkaline so the DNA denatures into single strands during the transfer step.

The nylon membrane is incubated in the presence of a single-stranded DNA probe that has been radioactively labeled. The DNA probe binds to its matching DNA sequence on the membrane.



Excess probe is washed away, and the membrane is sandwiched to x-ray film. Radioactivity emitted from the probe exposes the film, showing the locations where it has bound to its complementary sequence.

In this rape case, a series of five probes was used to create the DNA fingerprints of the evidence, victim, and suspect.



Probes 1-5

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