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FORENSIC DNA EVIDENCE
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Introduction

DNA evidence, and in fact any type of scientific evidence, strikes fear in the heart of most members of the criminal justice community. Nevertheless, the use of DNA has become inseparable with the investigation of violent and sexual crimes. In fact, recent advances in technology and laboratory throughput have made DNA analysis a tool in the investigation of less serious crimes as well. For example, DNA profiles can now be detected on swabs of steering wheels, and be used to solve crimes such as automobile theft. Recent television shows have made amateur forensic scientists out of potential jurors, who have now come to expect the presentation of scientific evidence at trial.

The increased use of DNA evidence has created a need for training of law enforcement personnel, prosecutors, defense attorneys, and judges alike. DNA evidence must be presented in a way that is understandable to the trier of fact, and effective defense against this evidence requires more than a cursory knowledge of the science. This article will describe the basic science of DNA and how it is analyzed in crime laboratories.

This is not meant to be a scientific article per se, and as such the use of jargon will be minimized where possible. The incessant footnoting that usually accompanies references to scientific discoveries and facts will be omitted to increase readability.

‘DNA 101’

DNA stands for Deoxyribonucleic Acid. It is a chemical entity that is found in all living things. DNA is known as the ‘genetic blueprint’ of life. It is an instruction manual that enables human beings to carry out all of the necessary life processes. DNA is also the genetic material. Humans inherit half of their DNA from their mother and half of their DNA from their father. In addition to serving as life’s instruction manual, DNA is capable of copying itself so that all new cells in the body have identical DNA content.
With the exception of identical twins, no two human beings will have the exact same DNA content. Most of the DNA found in humans is the same, however since it dictates our ‘humaness.’ Human DNA specifies that if all goes well, we will have one head, two arms, etc. Of interest to forensic science are the few areas of DNA that differ greatly between individuals.

Because the DNA content is consistent throughout the body, different types of samples from the same individual will give the same result upon DNA testing. For example, if the DNA from your blood is tested and compared to DNA from your saliva, the results will match. Furthermore, your DNA type does not change with time. So, if blood was shed by a suspect at a crime scene many years in the past, a new sample drawn from the suspect at time of arrest can be used for comparison to the old crime scene sample. DNA is also relatively sturdy and can remain testable even when subjected to harsh conditions (e.g. exposure to heat and moisture). DNA profiles are routinely being obtained from biological evidence collected decades ago.

DNA is found within structures in the human body known as cells. Some examples are white blood cells (red blood cells actually do not contain DNA), sperm cells, cells lining oral and vaginal cavities, and skin cells. The most common type of DNA analysis performed looks at the DNA contained within the nucleus of the cell. The nucleus is the ‘brain’ of the cell. Within the nucleus, the DNA is packaged in structures known as chromosomes. Chromosomes are pieces of DNA wound around a ‘scaffolding,’ in a manner analogous to thread on a spool. Humans have 23 pairs of chromosomes in the nucleus of their cells. The reason that chromosomes are found in pairs is that each parent contributes one chromosome to the pair. The X and Y chromosomes are perhaps the best known of the chromosomes since they determine the gender of the individual. Females have two X chromosomes (XX) and males have and X and a Y chromosome (XY). Within the chromosome pair (XX or XY), one chromosome is inherited from each parent. Since a female can only pass on an X chromosome, the chromosome passed on (X or Y) by the father determines the gender of the offspring. Since there is equal probability of the father passing on an X or a Y chromosome, the chance of the offspring being male is 50% and the chance of the offspring being female is 50%.

In addition to the DNA found in the nucleus of the cell (nuclear DNA), DNA can be found outside of the nucleus in structures known as mitochondria. The analysis of mitochondrial DNA (mtDNA) has limited application and is too expensive, specialized, and time-consuming for most crime laboratories to perform. Because there are many copies of mtDNA within a cell (thousands compared to a single copy of nuclear DNA), mtDNA is more likely to survive in samples that have undergone extremely harsh treatment. A perfect example of the utility of mitochondrial DNA typing is in the identification of victims of the World Trade Center tragedy on September 11, 2001. Because of the extreme conditions that human remains were exposed to, conventional nuclear DNA typing was not effective on many of the samples. Mitochondrial DNA typing was successfully used on these types of samples. Mitochondrial DNA typing is also useful in the analysis of hair shafts. Hairs that have been forcibly removed from the scalp tend to have tissue attached to the root that contains nuclear DNA. However, shed hairs do not have the necessary root tissue, and therefore only mtDNA is present.

The analysis of mtDNA is considered a last resort if all else fails. Mitochondrial DNA is a relatively poor discriminator of individuals when compared to the nuclear DNA. Another issue is that mitochondrial DNA is maternally inherited, so all maternal relatives will have the same mitochondrial DNA profile. This is a double-edged sword. On one hand, if mtDNA from a hair at a crime scene matches a male suspect, it could be argued that the hair is actually from his mother, brother, cousin, etc. On the other hand, the maternal inheritance is very useful in the identification of missing persons, because a family member can provide a reference sample of mtDNA. If, for example, skeletal remains are found that are believed to be the missing person, the mitochondrial DNA profile of the remains should match the reference sample of the family member.
Analysis of DNA in Crime Laboratories

For this discussion we will consider only nuclear DNA testing, which is the most common type. A simple example of a DNA case would be a residential burglary, during which the perpetrator cuts himself and bleeds while breaking a window. Samples for analysis in this type of case might include a sample of the blood left at the crime scene along with reference blood samples taken from a male suspect and anyone living in the house. In the laboratory, the testing of the crime scene sample and the reference blood samples would be conducted with identical protocols. Reference samples would, however be tested separately from crime scene samples, so as to eliminate any possibility of contamination of the crime scene samples.

In the first step of analysis, the DNA must be removed from the cells and separated from other components of the cells. This is accomplished by taking a small cutting of the bloodstain, placing it in a test tube, adding certain chemicals, and heating up the test tube. During this process, known as DNA extraction, the cells are ruptured releasing the chromosomes. The DNA is unwound off of the chromosome structure and is separated from other cellular components.

Next it must be determined how much, if any, testable human DNA was recovered from the sample. This is a critical step, because a particular amount of DNA is required in the next steps in the typing process. If too much or too little DNA is used in the typing process, ambiguous results could occur. Also, all of the human DNA recovered from the sample may not be necessarily testable. For example, if the sample was exposed to conditions such as moisture and heat, bacterial growth may have caused the DNA to become degraded (chopped into pieces). Depending on the degree of degradation, DNA testing results may be incomplete or non-attainable. Another issue occurs when biological stains are deposited on certain substrates such as black denim jeans. When a portion of the jean material containing the stain is cut out and analyzed, the dye present in the jean material can interfere with the DNA typing process. Using a computer-driven process known as real-time PCR, DNA analysts can get an excellent estimate of the amount of testable human DNA present in the sample after the extraction process.

After the DNA is extracted from the bloodstain, and it has been determined that there is a sufficient amount of testable human DNA present, the DNA is prepared for typing using the PCR (Polymerase Chain Reaction) process. During the PCR process, certain areas of interest on the DNA are targeted, labeled for detection later in the typing process, and copied numerous times. Human DNA can be thought of as a volume of encyclopedias. The PCR process ‘bookmarks’ and copies the most useful information in this set of encyclopedias.

There are currently 14 areas of DNA that are commonly targeted for forensic typing. One of these areas resides on the X and Y chromosomes, and is known as Amelogenin. DNA testing at this area can determine if the bloodstain is from a male or female. The other 13 areas commonly tested are known as STR’s (Short Tandem Repeats). DNA’s structure contains a code that is made up of 4 chemicals (abbreviated A, T, C, and G). If we were to look at all of the DNA within a cell, we would see that the code consists of some 3 billion of these chemicals in various arrangements. STR’s refer to a certain way that the DNA code is arranged. In certain areas, a specific order of 4 of these chemicals is repeated over and over. For example, at a particular DNA location, the code letters ‘ATTA’ might be repeated (e.g. ATTA-ATTA-ATTA-ATTA). STR’s are useful in forensic science, because different individuals may have different numbers of these repeats at particular areas.

DNA typing results are obtained by looking at the number of repeats found on the chromosome inherited from the mother and the number of repeats found on the chromosome inherited from the father. So, for example, if we performed DNA typing on an individual’s reference blood sample, we might detect 12 repeats on one chromosome and 16 repeats on the other chromosome. In this scenario, the result would be reported as a ‘12, 16’. The greater the number of repeats present, the larger the piece of DNA. It is analogous to a train in that the more boxcars there are, the longer the train. Thus, the DNA can be measured in order to determine the number of repeats present. DNA is typically measured using a scientific instrument that performs a process known as capillary electrophoresis.
To summarize the analysis procedure so far:

-the cells were broken apart and the DNA was purified from the other contents
-the amount of testable human DNA recovered was determined
-the PCR process was conducted in order to isolate 14 areas of DNA, mark them for detection, and make numerous copies
-one of the areas determines gender, the other 13 areas are STR’s
-STR’s are areas on the DNA at which individuals may have different lengths of a particular pattern of A’s, G’s, T’s, and C’s
-the lengths of these areas are determined using capillary electrophoresis

Once this long process has been completed, the results obtained for the crime scene sample are compared to the results obtained for the reference blood samples. In our example of the residential burglary, let’s assume that DNA testing revealed that the bloodstain left at the crime scene came from a male. Immediately, we can exclude any female living in the house as a source of that bloodstain. We are now left with the male suspect and any male living in the house, from whom blood was taken and tested. By looking at the typing results of the 13 STR locations, we will be able to distinguish between the suspect and other tested males. If the suspect’s DNA profile matches the DNA profile of the crime scene bloodstain, then he is included as a possible contributor of that blood. Currently, using these 13 STR DNA locations, forensic scientists are able to make statements such as “the bloodstain from the crime scene, which matches the male suspect, is expected to occur in 1 in a quadrillion (1,000,000,000,000,000) individuals.” Considering that there are only about 6 billion (6,000,000,000) individuals on Earth currently, this would be overwhelming evidence that the crime scene blood came from the male suspect.

**Conclusion**

While the introduction of DNA evidence has been tremendous for both proving guilt and innocence, it has also brought about the need for training. The justice system must be educated about DNA in order for it to reach its full potential in the investigation of crime.