Dna forensics

Science, Genetics

Before the 1980s, courts relied on testimony and eyewitness accounts as a main source of evidence. Notoriously unreliable, these techniques have since faded away to the stunning reliability of DNA forensics. In 1984, British geneticist Alec Jeffreys of the University of Leicester discovered an interesting new marker in the human genome. Most DNA information is the same in every human, but the junk code between genes is unique to every person. Junk DNA used for investigative purposes can be found in blood, saliva, perspiration, sexual fluid, skin tissue, bone marrow, dental pulp, and hair follicles (Phillips, 2008). By analyzing this junk code, Jeffreys found certain sequences of 10 to 100 base pairs repeated multiple times. These tandem repeats are also the same for all people, but the number of repetitions is highly variable. Before this discovery, a drop of blood at a crime scene could only reveal a person’s blood type, plus a few proteins unique to certain people. Now DNA forensics can expose a person’s gender, race, susceptibility to diseases, and even propensity for high aggression or drug abuse (Phillips, 2008). More importantly, the certainty of DNA evidence is extremely powerful in court. Astounded at this technology’s almost perfect accuracy, the FBI changed the name of its Serology Unit to the DNA Analysis Unit in 1988 when they began accepting requests for DNA comparisons (Lewis, 1989). There are thirteen standard tandem repeats used in modern forensics, and together these sequences create a DNA profile. Except in the case of identical twins, the probability that two people have the same genetic code at all thirteen core loci is less than one in one trillion (Crest, 2005). Investigators compare these genetic fingerprints with profiles stored in databases of previous offenders, and if they find a match, it proves that

the person was at the crime scene. DNA forensics can also narrow down suspect pools, exonerate innocent suspects, and link crimes together if the same DNA is found at both scenes. However, without existing suspects, a DNA profile cannot direct an investigation because current knowledge of genotype-phenotype relation is too vague for DNA phenotyping. For example, a profile from a first time offender that has no match in any database may give the information that the criminal is a left handed male of medium stature with red hair and freckles. It would be impossible to interview every man who fits that description. However, with available suspects, DNA forensics has many advantages over other forms of evidence. One is the longevity of DNA. Although it will deteriorate if exposed to sunlight, it can remain intact for centuries under proper conditions (Silverstein, 1996). Because DNA is so durable, investigators can reopen old cases to reexamine evidence. DNA from animals and plants can also be utilized in criminal forensics. One of the most common applications of this is the analysis of pet hair from a crime scene, which often links its owner to the crime. DNA fingerprints have also been applied to cannabis plants, and a database is being created to trace samples to their sources. This has been extremely successful so far, as this technology can distinguish between closely related, carefully bred plants (Westphal, 2003). Heather Miller Coyle of the Connecticut State Forensic Science Laboratory says, “ It links everybody together: the user, the distributor, the grower. That’s the real intent of it, to show it’s not just one guy with a little bag of marijuana, but it’s a group of people. " (Westphal, 2003). After a sample has been collected, it’s placed in a tube with ethanol and other chemicals that break the cells apart

and release their DNA. The next step is to place the tube in a microcentrifuge that uses centrifugal force to separate the solution into layers according to their weight. The tube is then incubated at 56 degrees Celsius for a few hours and spun in the centrifuge again (Butts, 2004). The heavier layers sink to the bottom, while the watery DNA layer floats above. After this layer is removed, the DNA is filtered out, and the result is usually smaller than a quarter of a drop from a medicine pipette (Butts, 2004). After isolating the DNA, scientists use a UV spectrometer to measure how much of it they have to work with. Different types of molecules absorb different wavelengths of light, so the spectrometer sends rays that DNA absorbs (Crest, 2005). With some simple calculations, the amount of DNA present can be measured based on the ratio of light that is absorbed, reflected, or passed through the sample. Since one extraction usually doesn’t produce enough DNA for analysis, scientists use the polymerase chain reaction process (PCR) to increase the amount available. First, the DNA sample is mixed with an enzyme called DNA polymerase, which produces copies of DNA under proper conditions. These conditions involve mixing the raw materials of DNA (adenine, thymine, guanine, and cytosine) and a few other chemicals. Specially designed primers are also used, which are short bits of DNA that provide instructions on where to start and stop copying the long strands in the sample (Crest, 2005). The primers are necessary because with current technology, it’s not yet possible to copy a whole DNA molecule in one reaction. PCR amplifications are generally kept under one thousand base pairs long, because the accuracy of copying decreases greatly beyond this point (Crest, 2005). Another important condition involves carefully changing

the temperature of the solution, allowing for the copies to be made. This careful regulation is done in a machine called a thermocycler. When the temperature is about 95 degrees Celsius, the paired strands of DNA from the sample untwist from one another, like a dividing zipper, in a process called denaturation (Butts, 2004). Once the strands are denatured, the temperature is lowered to allow the primers to attach to the now separate strands. After the primers attach, the DNA polymerase fastens to the primers and creates new strands of DNA by moving down the denatured strands, connecting complementary base pairs in a chain (Bianchi & LiÃ², 2007). The polymerase detaches when it reaches the stop codon, and the short strands produced link up with one another to form short double helixes. Even this does not usually produce enough DNA for analysis, so the reaction is typically repeated twenty to thirty times. For each double helix in the starting solution, two copies are produced, and in each successive reaction there would be 4 double helixes, 8, 16, and so on. Each cycle doubles the amount of double helixes in the solution, eventually giving the biologist enough DNA to examine. For most DNA samples collected from crime scenes, scientists use the polymerase chain reaction-short tandem repeat method in order to analyze them. With the PCR-STR method, the core loci from the junk code can be examined. This method is extremely accurate, letting scientists determine with a high degree of certainty whether the sample from the crime scene matches the samples of suspects. STRs, or short tandem repeats, are short segments of DNA made of three to seven repeated base sequences. Because everyone has these short tandem repeats in their genetic code, but the number of repetitions varies from

person to person, these can determine the difference between two people. For example, one person’s TH01 (a commonly used STR) sequence might be AATG-AATG-AATG, while another person’s might be AATG-AATG-AATG-AATG-AATG-AATG-AATG-AATG. The sensitive equipment used to analyze the samples easily tells the difference between short and long sequences via electrophoresis, a similar method to the one used in determining the amount of DNA in a collected sample (Bianchi & LiÃ², 2007). The amplified DNA is injected into a machine that passes a focused beam of light through it, and it displays a graph showing the number of repeats at each STR. The peak graph it gives is very useful and commonly used in court. The FBI recommends analysis of a particular thirteen core loci because these STRs are found in nearly every person in the world, but they are still highly variable from person to person (Bert-Jaap Koops, 2008). Only identical twins have the exact same number of repeats at all thirteen core loci, so samples can be matched with such certainty that it’s virtually impossible to argue that a sample of DNA could have come from more than one person. “ Nothing in science is ever considered absolutely true, but STR analysis gets pretty close to it, " says Detective Harold Thomas of the Milwaukee Police Department Violent Crimes Division. Besides its accuracy, STR analysis has many other benefits. A skilled technician can produce a profile of all thirteen core loci in about five hours from start to finish. All of the primers and raw materials of DNA can also be added in one neat reaction, reducing cost and the amount of handling time during processing (Thomas, 2011). This also reduces the chance of contamination. Since STR fragments are so short, they are very stable, much more so than fingerprint fragments and long strands

of DNA. STR evidence can be recovered from even the most badly damaged and decayed bodies. In 1994, the DNA Identification Act authorized the FBI to create a national DNA database, the Combined DNA Index System (CODIS). This originally contained only DNA profiles from crime scene evidence and convicted offenders, but in recent years there have been indexes added for arrestees and missing persons (Thomas, 2011). The profiles contained in CODIS consist of the thirteen STR loci, plus the amelogenin gene, which is found on the X and Y chromosomes and establishes the sex of unknown sample sources. Currently, CODIS contains about six million profiles (Bert-Jaap Koops, 2008). There are many advantages to having a central DNA database, but CODIS has aroused a lot of controversy over whose DNA is entered into it and how this information is used. Laws concerning DNA databases vary from country to country and sometimes between jurisdictions and states. Some countries have not yet taken a stance on the legality of DNA databases. In the US, DNA samples were originally allowed to be entered for only convicted sex offenders, but most states now allow DNA profiling of all convicted felons (Thomas, 2011). Some states collect profiles from all arrested suspects. In the United Kingdom, samples are collected from individuals arrested for all but the most minor offenses, and their database currently contains more than four million profiles (Fink, 2006). Laws also differ in the preservation of the actual DNA samples. In some US states, only the STR information is kept, and the samples themselves are destroyed after analysis. In other states and the UK, however, samples are preserved indefinitely for possible analysis with technology not yet invented (Fink, 2006). In some countries, not only are full matches kept, but partial

matches too. Partial matches may indicate that the criminal is a close relative of the person found in the database. Because DNA profiling is so effective, it has a huge amount of future potential. Many known DNA profiling techniques aren’t used yet, but volumes of successful research suggests that they will soon become routine. Right now, DNA evidence is only helpful when there are suspects for a crime, but it may soon be possible to use DNA phenotyping to provide police with clues about an unknown criminal. DNA phenotyping is the use of markers in a person’s genetic code to give information about race, appearance, and other traits. Currently, a person’s race can be found through DNA analysis, and in turn, some types of hair texture, eye and skin color, and facial features can be somewhat accurately determined (Sachs, 2003). This information could become powerful in an investigation, but its current inaccuracies forbid the use of DNA phenotyping for now. Some researchers believe that DNA phenotyping will someday be able to tell almost exactly what a person looks like by going farther than analyzing ancestry. This would mean examining other markers, such as genes that control pigmentation levels, stature, and other facial features. Studies have shown that many correlations between gene variants and physical features are fairly unreliable, but there has been some success. For example, a study from 2001 found that 90 percent of people with a particular variation of the human melanocortin 1 receptor have red hair and freckles (Sachs, 2003). Many researchers are just beginning to establish useful markers on the Y chromosome. These markers vary less between individuals than STR profiles, but they can be very useful in sexual assault cases where samples contain much more female than male DNA. By analyzing markers

found only in the male’s DNA, the profile of the perpetrator can be distinguished from the profile of the victim. CODIS does not currently contain any Y profiles, but it’s very likely that it will in the future. This would also make familial searches extremely powerful because Y chromosomes are passed along intact with only random genetic mutations from father to son. This means that all males from the same family have almost the exact same Y chromosome, and a male criminal with relatives in CODIS would be easily tracked down. Mitochondrial DNA (mtDNA) can also be very useful in identifying degraded DNA. Mitochondrial DNA is found in a cell’s mitochondria instead of its nucleus. Since each cell has hundreds of mitochondria, there are also hundreds of copies of its mtDNA, which increases the chance that it will survive for long periods of time or under harsh conditions. Right now, mtDNA is widely used in historical analysis of bones and in cells with little DNA, such as hair shafts, but it may be useful someday in criminal investigations (Phillips, 2008). Like Y chromosome analysis, mtDNA can track lineage because everyone gets their mtDNA from their mother. Another frontier in forensic genetics involves examining RNA. DNA profiles are perfect for individual identification, but they provide no information about what type of tissue is present in the sample. This is because every cell contains the same genome. On the other hand, RNA is perfect for finding the type of tissue present because different messenger RNA (mRNA) is found in different cell types. RNA profiles can also reveal the age of evidence such as a bloodstain because different RNA degrades at different rates (Phillips, 2008). If this aging analysis technique is improved, it can become very valuable to forensic scientists who currently have no other

way to determine the age of bloodstains. Currently, the main disadvantage to DNA profiling is the dead end investigators come to when there are no suspects, or samples from the crime scene have no match in CODIS (Thomas, 2011). Other problems also exist. By DNA profiling’s very personal, private nature, it has the potential for abuse by investigators who lack the integrity to perform analyses correctly. Another problem involves the possibility of mixed samples. These samples contain DNA from more than one person, such as a sample taken from a shared cigarette. About four percent of samples taken today contain DNA from more than one person (Marks, 2006). In response, the FBI has created a computer program called Pendulum that creates peak graphs based on the amount of DNA present from each person. It has a high degree of accuracy, but its information cannot be used in court, and like all computer-generated statistical estimation techniques, Pendulum is susceptible to error. Besides the controversy of CODIS and other central DNA databases, DNA profiling has many other ethical implications. Some law enforcement agencies have been involved in “ fish hunts, " where many people who have nothing to do with a crime have to donate tissue samples because they live in the vicinity of the crime scene. Those who refuse are subject to police interrogation (Thomas, 2011). Many people feel that this is harassment and an invasion of privacy. Others are concerned with databases because they’re no longer strictly criminal. In January 2006, President Bush signed a law expanding the collection of DNA samples beyond convicts to include arrestees and non-American detainees. Many people feel that using genetic markers to determine race could hurt minorities and lead to discrimination. Other

markers that give sensitive information, such as propensity for homosexuality or aggressiveness, have been put under scrutiny, as this information could also lead to prejudice. Even though DNA profiling has many limitations and ethical implications, its benefits are too great to ignore. It has solved countless crimes, and it will continue to solve infinitely more as technology improves and makes misconduct more dangerous for criminals.