

DNA FINGERPRINTING: THE CODE TO IDENTIFICATION

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DNA (deoxyribonucleic acid) is the genetic material of almost all organisms, including humans. It has a very simple chemical composition, which includes four different nucleotides or bases called adenine, thymine, cytosine and guanine which are commonly known by the letters A, T, C and G. The human genome (the entirety of human DNA) is made up of approximately 3.2 billion of these bases that are organised into 23 pairs of chromosomes. DNA is found in almost every cell in the human body, from blood cells, to skin cells, to liver cells, and the order of the bases is the same in all the cells from one individual. DNA is a code—the genetic make-up of an individual that specifies information for a seemingly limitless number of outcomes for that individual, including hair and eye colour and susceptibility to certain diseases.

The close genetic relationship between all humans means that the sequence of bases in the DNA of any two people is very similar (approximately 99.9 percent similar). Every individual inherits 50 percent of his or her genome from each parent. However, no two people (apart from identical siblings, such as identical twins) have exactly the same sequence of bases. DNA fingerprinting (or profiling) is based on that fact. Some of the differences are in single bases in the code. These are like spelling differences, and are called SNPs (Single Nucleotide Polymorphisms, pronounced 'snips'). Other differences result from short repeats of individual bases, somewhat like a stutter in the code. These are usually referred to as Short Tandem Repeats (STRs).

DNA fingerprinting has several purposes. In forensic analysis it can be used to determine if a biological

sample found at a crime scene matches that of a particular individual. Determining whether there is a match would be straightforward if the DNA sequences of the entire genomes could be compared. However, although DNA sequencing technology is becoming simpler, it is still difficult (and expensive) to determine the precise sequence of the 3.2 billion bases of a complete human genome. Biological samples from crime scenes (such as blood, semen, even individual hairs) are typically small, making it extremely difficult to isolate enough DNA to sequence the entire genome. DNA fingerprinting therefore relies on the comparison of regions where STRs and/or SNPs are found.

In a typical DNA fingerprinting analysis, if DNA is isolated from a crime scene sample, the pattern of STRs (the number of repeats) at defined regions can be determined. An advantage of DNA fingerprinting is that the results are easily filed in a computer and so can be stored until a suspect is found. The results can then be compared to the DNA patterns in a sample from a suspect (often from cheek cells), or to a database of suspects. When the STR or SNP pattern from a crime scene sample matches that of the sample from a suspect, the forensic scientist can estimate the likelihood of that match occurring by chance by examining data about specific STR/SNP occurrences in the DNA of the general population.

One of the most common methods of DNA fingerprinting analyses STRs at thirteen different places in the genome. Each variant of an STR is shared by only a small proportion of the population. Thirteen matching STRs mean it is highly unlikely that two samples could

come from different sources. However, it is possible to misinterpret the statistics. A statement that there is a tiny probability of finding a match if the accused is innocent is not the same as stating that the probability of the accused being innocent is just as minute. For example, if the probability of a match is one in 100,000 and the population size is 1,000,000 (similar to the size of Dublin), then there are likely to be ten people in the population with the same DNA fingerprint. In the absence of any other evidence, the probability that the sample did not come from the accused is still very high (nine in ten). In practice, the probability of finding a match is usually much lower than one in 100,000 (and can be as small as one in 100,000,000,000,000). Even so, law officials need to be very careful when comparing crime scene samples to DNA databases of individuals with no other known connection with a crime.

One of the biggest advantages of DNA fingerprinting—that it can be carried out with very small samples of genetic material—can also lead to problems. Advances in technology mean that it is possible to develop DNA fingerprints from tiny samples (such as skin cells on a pen, or a single hair on a piece of clothing), and from very old samples, such as blood spots. However, it is vital to control against contamination when using small samples. Sometimes it is not possible to test several STRs if the DNA from the sample is degraded, and this affects the statistical measurements of a match.

As stated above, individuals inherit their DNA from their parents, and as a result DNA fingerprints are also inherited. On average, a child shares 50 percent of his or her fingerprint with each parent and 50 percent with any siblings. DNA fingerprinting is therefore a very powerful tool for testing paternity and family relationships. This genetic inheritance must be taken into account, however, when dealing with forensic samples; even first cousins share 12.5 percent of their DNA fingerprint.

There are a few specialised applications of DNA fingerprinting that are used for analysing relationships. Mitochondria are specialised structures within cells that have their own DNA, quite separate from the DNA in the nucleus of the cell. Mitochondria (and therefore mitochondrial DNA) are passed from the mother to all of her children. Mitochondrial DNA fingerprinting is therefore useful for identifying family groups. For example, in the early 1990s, mitochondrial fingerprinting was used to identify

bones found in a shared grave in Russia. The victims turned out to be members of the royal family, the Romanovs, who had been executed in 1918. Because the Y chromosome, part of the nuclear genome, is passed largely intact from father to son for many generations, DNA fingerprinting of the Y chromosome can be used to trace male lineages.

Human DNA fingerprinting tends to be associated with guilt. A Bill was published by the Irish government in 2010 which would allow the collection of DNA samples from every person arrested in Ireland, and use them to generate a database of DNA profiles. The profiles could then be compared with those obtained from any crime scene samples. It must be remembered, however, that DNA fingerprinting is a tool, which rarely proves guilt on its own. Genetic fingerprinting can show that semen came from an accused man, for example, but it cannot determine if sex was consensual. Of course, DNA fingerprinting can also be important for proving innocence. The Innocence Project in the US (<http://www.innocenceproject.org/>) has used DNA testing to exonerate over 250 people, many of whom had been wrongly convicted on the basis of incorrect eyewitness reports.

It should be noted that DNA fingerprint analysis is not restricted to humans. It is an effective tool in the pedigree analysis of animals. It is also used in species and animal identification, for example, to identify whale meat which it is illegal to sell in many countries, and to trace the beef in Irish supermarkets back to the farm of origin. DNA fingerprinting can also be used to track the source of bacterial infections and outbreaks of food poisoning. For example, suspicions were raised in Germany in 2011 when three deaths were associated with *E. coli* strains with the same DNA fingerprint. This strain subsequently caused the deaths of at least 46 people in Germany, and several more in France in a second outbreak. The original source of the bacteria may never be known, but one possible origin is sprouted fenugreek seeds imported from Egypt.

In conclusion, DNA fingerprinting is a very powerful tool, in forensics, in paternity tests of people and animals, in epidemiology (detecting the source of epidemics), and in agriculture. We can look forward to future refinements that will allow testing of ever-smaller samples, and also to the development of cheap and easy sequencing of whole genomes, which may ultimately render fingerprinting redundant.

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