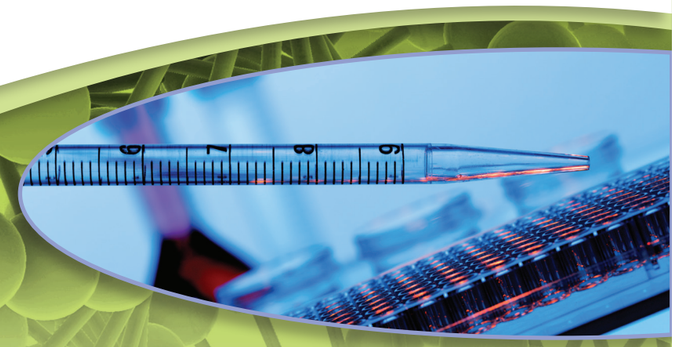
PUBLIC UNDERSTANDING OF BIOTECHNOLOGY



DNA profiling

What is DNA profiling?

One thing that all humans have in common is that each person is unique - just like everyone else!

The basis of this individuality lies in the genetic information encoded by each organism in its DNA (deoxyribonucleic acid). Each individual carries a unique DNA sequence. Thus, while any group of organisms will have a general DNA sequence that will identify it as, for example, human or a particular species of bird, individuals within each group will have their own exclusive sequences.

Spectacular advances made during the 20th and 21st centuries in the science of molecular biology have made it feasible, in terms of time and money, to identify these differences and to reveal them as a list of letters and numbers that can be used to distinguish between any two individuals. This alpha-numeric sequence can be used as a personal identifier and is referred to as a "DNA profile" (previously called a "**DNA fingerprint**").

DNA profiling has been so popularised by the media in TV series such as CSI, that the public are generally aware of its application in identifying suspects in linked murders, rape cases and violent crimes, or in exonerating the wrongfully accused, and in establishing an identity for corpses, skeletons or victims of mass disasters.

So sensitive is the technology that, although blood, saliva or semen are the main sources of DNA, a profile can be obtained from fragments of bones or even a single cell left on touched objects such as a steering wheel, a licked stamp, the handle of a gun, the inside of a glove, a scarf, a hat, a bite wound, a cup or cool drink can, or a cigarette butt. Furthermore, DNA itself and the technologies used are so sturdy that a DNA profile can even be established years after an event.

DNA profiling - a short history

In 1984, Alec Jeffreys, a scientist at Leicester University in England – who was later knighted for his contributions to forensic science – realised the exciting possibility that his research could be used to identify individuals from differences in their DNA sequence. Two years later his idea was validated when British police used DNA fingerprinting (as it was then known) in three world "firsts" – firstly, undertaking mass DNA screening of males in an area, and then using this information to exonerate one man and to convict another of the rapes and murders of two high school pupils in the small village of Narborough in Britain.

Concomitantly, in 1986, American scientist Kary Mullis developed the PCR (polymerase chain reaction) which added another dimension to DNA profiling by making it possible to copy specific target regions of a single molecule of DNA to make millions of copies of the nucleotide sequences in a matter of hours.

PCR makes it possible to make multiple copies of extremely small quantities of DNA. As a result, sample size is no longer a limiting factor in characterising DNA recovered from a crime scene and it is possible to produce a DNA profile from evidence not visible to the naked eye as well as from extremely degraded samples. (For more information on the PCR process, visit **http://en.wikipedia.org/wiki//Polymerase.chain.reaction).**

Together, these technologies opened a new era of forensic DNA profiling; in 1995, the UK set up the world's first DNA database, the USA followed in 1998 and, in same year, South Africa entered its first DNA profiles into what is now called the National DNA Database of South Africa (NDD). In 2006, the world's first fully automated system for high volume forensic DNA analysis and profiling became operational in the South African Police Service (SAPS) Forensic Science Laboratory (FSL), Tshwane. Financed by a European Union initiative to build capacity in SA, this 37 m long, 4 m wide, system can run 22 hours per day 365 days a year, handling 8 000 samples a day for DNA extraction and further processing. In 2012 another state of the art forensic facility was opened in Plattekloof in the Western Cape.

How does DNA profiling work?

Genetic information is stored in cells as DNA, a long molecular chain, on which the precise linear order of stretches of four chemicals (called nucleotides) constitute individual genes. In turn, these genes encode the specific protein products that are needed for the cells of an organism to

A second fear relates to the accuracy of the profiling and the chances of error resulting in wrongful conviction. Many arguments have been presented by both sides but it is remarkable to note that to date 302 people in the USA have been found innocent of crimes they did not commit through the work undertaken by a public policy organisation called The Innocence Project. These include the exoneration in 2007 of Jerry Miller after he had served 25 years for a rape that he did not commit. Interestingly, work is currently underway to establish a similar Innocence Project in South Africa.

Ethically, the discussion centres on a balance between the rights of the individual and the rights of society. Williamson and Duncan (2002) argue there are only two fair possibilities for DNA profiling: everyone or no one. If one purpose of a DNA database is to deter crime, why not profile everyone? If so, when? At birth, at the age of consent? The cost of testing the entire population however, also needs to be taken into consideration. If samples are taken only from convicted offenders, there are concerns that some groups will be over-represented in the criminal intelligence database (social and racial inequality). Interrogating the database is likely to score more hits within these groups perpetuating the situation. Furthermore, if providing a sample for DNA analysis is voluntary but a suspect refuses to give a sample, the assumption is the person has something to hide.

What is the future of DNA profiling?

Research is underway to develop a portable handheld DNA analyser, which can be used at the crime scene. The Y chromosome is inherited, like a surname in many cultures, through the male line of descent. Thus, it is has been proposed that Y chromosome profiling and interrogation of a Y chromosome DNA database will allow prediction of the surname of male suspects or victims of crime from DNA alone. More importantly, in South Africa where gang rape is prolific, markers on the Y chromosome may assist in the identification of all the perpetrators. Similarly, markers on mitochondrial DNA are used to trace an individual's ancestry through the female line of descent. There is intense interest in developing "DNA Photofits" where, solely through DNA analysis, a physical profile (Identikit) of a suspect/victim can be drawn.

However, currently, there are few absolute tests for identifying features such as hair, eye or skin colour, because not only are these characters the result of variations in many different genes and environmental factors, but there are also ethical concerns. Another possible development is DNA profiling of the myriad of microbes that co-habit our skin and bodies. It is believed this will make it feasible to distinguish between identical twins, whose own DNA profiles are identical, as every individual will harbour their own zoo or garden of microbes.

One thing is certain – there will be amazing advances – to quote Sir Alec Jeffreys "if you had told me (in 1984) that 20 years later this technology would directly touch the lives of 10 million people worldwide, I would have thought 'fantasy, no way' – I am amazed".

This fact sheet has been reviewed by independent experts and has followed SAASTA's Scientific Editorial Process.
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grow and function. In humans, the DNA in every cell is split between 23 pairs of chromosomes. The coding sequences of DNA that make up the genes are interrupted by long stretches of DNA that do not code for proteins and which are consequently called "non-coding DNA" or more loosely referred to as "junk DNA". In this "junk DNA", there are numerous chromosomal locations that contain short stretches of DNA where a particular sequence of 2 - 8 nucleotides is repeated in tandem a number of times. These repeat units, known as Short Tandem Repeats (STRs), or microsatellites, always occur at the same chromosomal location, called "locus" and, although they are inherited stably from parent to child, they vary substantially between individuals. The biotechnology that allows this variation to be captured and recorded forms the basis of DNA profiling as it allows scientists to discriminate between individuals.

Basically, specific markers are targeted on different chromosomes and by adapting nature's own system of copying DNA by using PCR, only the DNA region of interest is amplified, instead of the whole chromosome. Therefore, each marker region or "locus" is specifically replicated. The size of the copied product will vary depending on the number of repeat units in the STR. These size differences can be measured using automated technology and the analyses recorded as a series of letters and numbers on a computer printout. In practice, several markers are analysed simultaneously to generate an alpha-numeric sequence which is individual-specific. It is therefore very unlikely that two people will have identical DNA profiles. Comparison of the profile generated will produce a match if two DNA samples were derived from the same individual. Furthermore, in any individual, one member of each paired chromosome is inherited from an individual's mother and the other member from the father, and in turn only one of each pair will be passed down to the following generation. This allows family relationships to be established, which forms the basis not only of paternity testing but also helps in identifying unknown corpses and skeletons by comparison with close relatives.

How many markers are needed to generate a unique DNA profile?

An important consideration is the number of markers needed to generate a unique genetic profile. Currently in South Africa, the SAPS FSL uses a set of nine markers and a tenth marker which allows gender discrimination. Statistically, this means that the probability of a false positive match between two people (other than identical twins) is approximately one in a billion. However, in 2004, Sir Alec Jeffreys suggested that as the UK has such a large DNA database, 15-16 markers should be used to reduce the chances of two people, in a given population, having the same profile to one in a trillion. The greater the number of loci analysed, the less chance there is of two people sharing the same forensic DNA profile.

Legislation and regulation of forensic DNA profiling

United Kingdom

In 1995, the first criminal DNA database was established in the UK, where laws specify compulsory DNA sampling and indefinite storage of DNA profiles of persons suspected, reported, charged, convicted, or cautioned for any recordable offence. Currently, the UK combined criminal and crime scene databases contain over five and a half million profiles.

In South Africa

Currently, the collection and retention of DNA profiles for criminal intelligence purposes is governed by the Criminal Procedures Act (CPA) of 1977 which was promulgated long before the advent of DNA profiling was used for crime detection. The existing South African NDD currently contains approximately 130 000 DNA profiles the majority of which are collected from crime scenes (called crime stains) and the remainder from DNA samples of persons suspected of a crime (reference profiles).

Due to the potential value of DNA as a law enforcement tool, legislating policies and procedures to regulate a national DNA database for criminal intelligence purposes has become a matter of urgency. To this end, The Criminal Law (Forensic Procedures) Amendment Bill B2-2009 was drafted and adopted by Cabinet in December 2008. The DNA Bill, currently still under review by Parliament seeks to address gaps in our current legislation dealing with the collection, storage and use of DNA evidence and to provide for the expansion and administration of a national DNA database which would be called the National Forensic DNA Database of South Africa (NFDD).

The new law, if passed, will provide for the establishment of a comprehensive criminal intelligence database that will be used by the SAPS for speculative searching against reference indexes. Five index subsets will contain DNA profiles of convicted offenders, crime scene stains, volunteers, arrestees and an elimination group composed of persons working with the collection and analysis of crime scene samples. The latter is necessary as the exquisite sensitivity of PCR technology may result in contamination of a crime scene sample by replication of DNA from a single cell accidently derived from investigators or DNA analysts working in the SA FSL. Crime scene stains will be kept indefinitely, whereas reference profiles will be kept for restricted time periods. It is important to note that the actual DNA sample obtained from any individual will be destroyed once a full forensic DNA profile has been obtained and loaded onto the database. The law will also allow DNA profiles to be established retrospectively from convicted offenders by a cheek swab, which can be administered by a specially trained police officer – unlike the existing legal requirement for a blood sample taken by a medical doctor. South African profiles will be generated with 10 markers (one of which will allow gender discrimination) in the SA FSL. The FSL follows international standards using a set of markers, developed commercially under strict specifications, which create a unique genetic profile. It is important to note that no genetic disposition or other distinguishing feature can be read from this profile which will be generated for criminal intelligence purposes only.

Who does DNA profiling in South Africa?

All forensic cases, i.e. crime scene, missing persons' remains etc., are handled by the SAPS FSL. Many private laboratories and state laboratories throughout South Africa undertake paternity testing, while individuals at universities undertake research on a wide variety of organisms using DNA profiling as a tool. At the University of the Western Cape, identification of human remains following exhumation of apartheid activists' graves has been performed in partnership with the National Prosecuting Authority stemming from the work of the Truth and Reconciliation Commission.

DNA profiling – a South African case stories

In 2002, DNA profiling exonerated six persons accused by the community of the rape of a nine-month old baby girl, named Tshepang; at the same time, profiling identified the actual perpetrator. It has also been used to help unravel the mystery of Happy Sindane, and find the mother and identify the father of twin babies found apparently abandoned in a taxi.

The potential success of a criminal intelligence database is highlighted by the conviction of a serial rapist, Shavani Phophi in June 2011. Phophi, known as the Muldersdrift rapist, was found guilty of six rapes, three robberies with aggravating circumstances, and two cases of theft. Five of the rape victims were adult women to whom Phophi offered work and then lured to Nooitgedacht to rape them in the veld. The Investigative Psychology Unit of the SAPS used a number of strategies, including DNA, to successfully link all the adult rape cases and to locate the suspect in his shack in Kyasands. After arrest the suspect was then also linked through the DNA database to the rape of a 10-year-old girl in 2005. Without the database, the case of the little girl would not otherwise have come to the attention of the police, because the other victims were adult females raped between June 2009 and May 2010. Phophi subsequently stood trial for all these cases of sexual assault and received a combined sentence of two life sentences and a further 95 years behind bars.

Can DNA profiling be used in other organisms?

Genetic variation occurs in all other forms of life and the power of DNA profiling has been expanded into animal and plant, viral and bacterial profiling. It has applications in conservation, poaching and animal smuggling, authenticating consumer products, in tracing pollution outbreaks, in forensic investigations and in infectious disease research.

Researchers at several South African universities are investigating the application of DNA profiling to animal and plant identification, for example, in conservation management and in preventing poaching of such varied species as abalone, rhino, elephant, parrots, blue crane and cycads. DNA profiling can identify a particular animal or part, for example, meat or blood on an axe that may have been found under suspicious circumstances.

Internationally, canine and feline DNA databases have been established and, in 1996, evidence based on a DNA profile match between cat hairs found on jacket at the scene and a cat owned by a murder suspect lead to his conviction. In South Africa, DNA profiling is used to authenticate the cultivars used in wine making, and has been used in identifying different strains of sweet potato in bio-banks. Research based on DNA profiling of the organisms that cause HIV/AIDS and TB is used by South African scientists to gain an understanding of the factors driving the spread of these deadly epidemics.

What societal issues are associated with DNA profiling?

Concerns expressed by society are that DNA profiling may violate an individual's genetic privacy, revealing knowledge – such as disease susceptibilities, behavioral traits, and paternity – that the person may not want to know, or have others, such as relatives, employers, insurance companies, know. However, a DNA profile, derived for forensic purposes, reveals nothing about an individual's genetic makeup and following the adoption of the new DNA Bill, all DNA samples derived from people (not crime stains) will be destroyed after a profile has been obtained and loaded onto the database. .