

This fact sheet describes the DNA testing (genetic testing) process for non-medical purposes of forensic, paternity and ancestry analysis.

### In summary

- DNA testing for forensic, paternity and ancestry looks for small differences and similarities between individuals in their DNA
- DNA fingerprinting (DNA pattern) can be used to estimate the chance that two samples are from the same person
- Informed consent from both parents, quality control of the test and availability of counselling are important for paternity testing
- Ancestry testing looks for clues about where ancestors might come from and relationships in families.

### DNA TESTING

DNA testing is the analysis of information in our chromosomes and genes that provide the instructions (the code) for making proteins. DNA testing for forensic, paternity and ancestry looks for small differences and similarities between people in their DNA. This type of testing usually involves analysis of regions between genes (non-coding DNA) and compares the findings to databases to look for similarities. DNA testing that looks for specific variations across the whole genome is now also being used.

### ABOUT DNA

DNA contains the instructions for growth and development in humans and all living things. Our DNA (**D**eoxyribo**N**ucleic **A**cid) is packaged into chromosomes that contain all our genes that provide a code for making proteins.

The DNA code is made up of very long chains of four basic building blocks (*nucleotide bases*) called **A**denine (A), **G**uanine (G), **T**hymine (T) and **C**ytosine (C) and different combinations of these chemical 'letters' makes the code. There are about 6 billion bases and over 20,000 genes found in the DNA of each person and each gene has its own specific location on a chromosome or on the mitochondrial DNA. Genes plus the region between each gene (non-coding DNA) make up a person's **genome**. Mitochondria, small units in the cells involved in energy production, have their own separate DNA.

Like genes, non-coding DNA contains large numbers of repeated chemical letters (A,G,T,C) along its length. The number of times that a sequence is repeated within a length of non-coding DNA can range from just a few to hundreds.

As we have evolved, changes have built up in our non-coding DNA as well as our genes. We all have small variations in our genetic code. That is why we are unique.

### DNA – DIFFERENCES AND SIMILARITIES IN HUMANS

Despite these variations in our non-coding DNA:

- Most humans are overwhelmingly alike in their sequence of letters in their DNA although each person's sequence is unique (except for identical twins)
- DNA samples from two people differ on average at only one letter or 'base' per thousand. Therefore, 1 per 1,000 of 6 billion bases means there are about 6 million differences in the total DNA of two unrelated people
- These variations are enough to produce all the genetic differences between these two people
- The closer two people are related to each other, the greater the chance their DNA sequence will be similar
- The small differences and similarities in the DNA sequence between people are used in DNA testing to determine the identification of individuals.

## TESTING NON-CODING DNA FOR NON- MEDICAL PURPOSES

Unlike tests on DNA for medical purposes, non-coding DNA testing looks at specific areas called **loci** (singular *locus*) in the non-coding DNA that are generally not related to our health.

These loci are found at a number of sites on each chromosome. The loci used for DNA testing are those that have different numbers of the repeated sequences in different people in the population.

All our chromosomes come in pairs, which means that each person has two partnering loci for every paired chromosome. They will have a specific number of repeats at each of these loci on both their chromosomes, and this number of repeats may be the same or different between the partnering loci.

Information from specific loci is used to create a DNA pattern or DNA fingerprint for each person.

## CREATING A DNA FINGERPRINT (OR PATTERN) FOR A PERSON

The basic principles underlying how DNA testing is done includes (*Figure 17.1*):

### Step 1:

- In the laboratory, using enzymes that act as chemical scissors, the DNA is cut into hundreds of small pieces where there are specific sequences of the DNA letters

### Step 2:

- The cut DNA is placed into a slab of jelly (a gel matrix) and an electrical current is applied so that the jelly becomes electrically charged and has a positive (+) end at the top and a negative (-) end at the bottom, just like the positive and negative ends of a battery
- As the DNA is a chemical which has a negative charge, the DNA is pulled towards the positive end of the gel or from the top to the bottom
- The pieces of DNA separate along the gel according to size: the biggest pieces move the slowest and so will remain near to the top of the gel

- The gel now contains all of the person's DNA spread from the top to the bottom of the gel.

### Step 3:

- To select out the pieces of DNA that need to be looked at, the pieces of DNA that have spread through the gel are covered with special DNA probes
- A probe is a piece of **DNA** used to detect sequences. The probes are made in the laboratory and contain a match for the DNA sequence that the test is looking for
- The probes in fact have the opposite sequence to the sequence being tested for. They match up because of the ability of the letters A and T, and C and G to pair with each other as shown in *Figure 17.2*
- If the person's DNA on the gel contains the matching sequence, the probe will combine (*hybridise*) with the person's DNA at the site of the matching sequences
- Chemicals such as fluorescent dyes are attached to the probes. When the gel is exposed to a certain type of light, the DNA on the gel that has been detected as containing a particular sequence of 'letters' will show up. The remaining DNA will not be visible
- When a number of different probes are used to detect different repeats of sequences, the gel will look like a series of bands as shown in *Figure 17.3*. Each band represents a site or locus on the non-coding DNA at which a specific repeated sequence is located
- Some of the bands appear darker than others. This is because that person has the same number of repeated sequences of letters on each of their two chromosomes where the locus is located. The band is showing the same pattern from each chromosome and so the two bands are lying on top of each other on the gel
- A person can only ever have one or two bands for each probe
- In *Figure 17.3*, Persons 1 and 2 have the same banding pattern for probe 5 and persons 2 and 3 have the same pattern for probe 7.

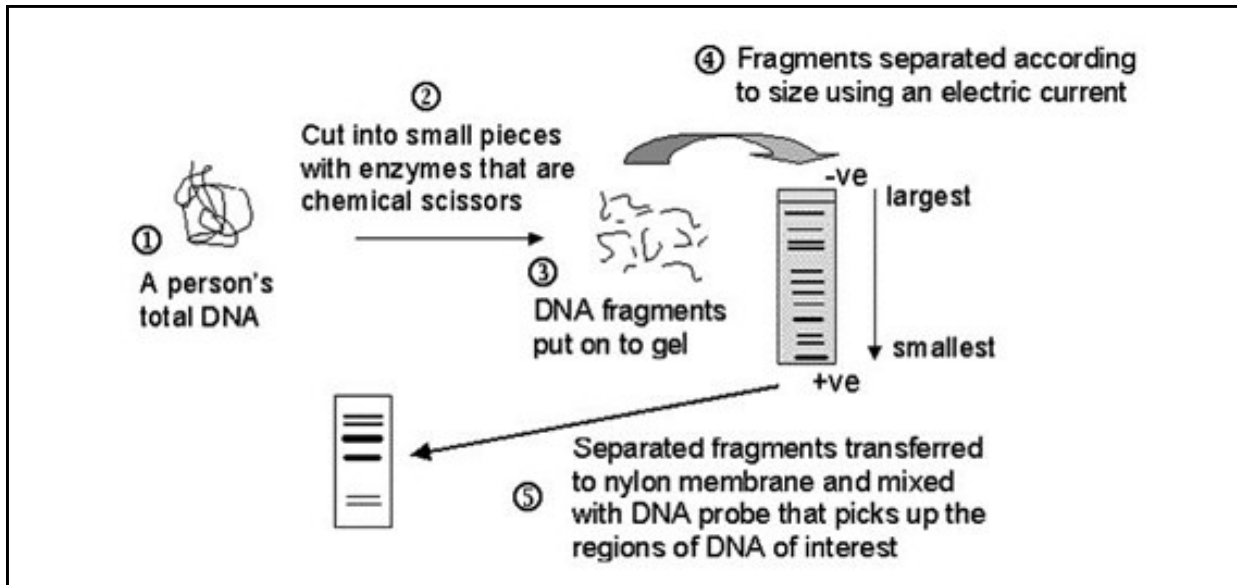


Figure 17.1: Genetic testing to create a DNA fingerprint of a person.

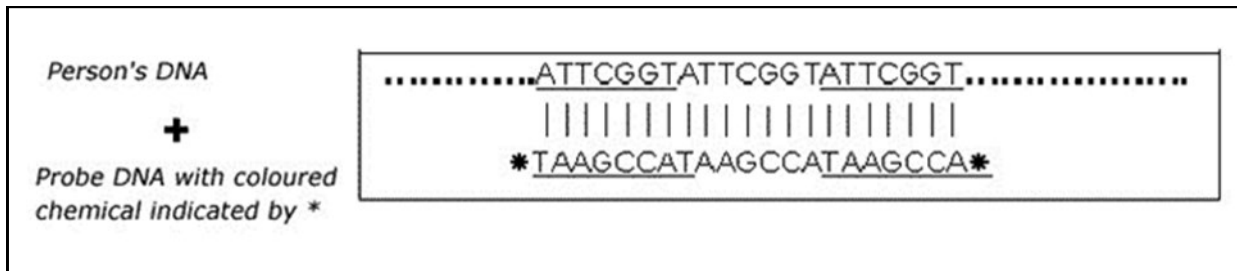


Figure 17.2: Using a probe to detect a repeated sequence in the non-coding DNA of a person.

Person 1	Person 2	Person 3	Person 4
████████	████████	████████	1
████████	████████	████████	2
████████	████████	████████	3
████████	████████	████████	4
████████	████████	████████	5
████████	████████	████████	6
████████	████████	████████	7

Figure 17.3: DNA fingerprints of three people created using seven different DNA probes manufactured in the laboratory.

- That is why it is necessary to use a number of different probes to enable differentiation between different people
- The bands at the bottom of the gel represent shorter sequences of letters than at the top. For example, a band at the top of the gel may represent a sequence of five letters repeated 30 times so it will be 150 letters or bases long. At the bottom the sequence may be made up of only four letters repeated 10 times so it will be only 40 letters or bases long.

These patterns of bands have become known as a person's **DNA fingerprint** (Figure 17.3).

### FORENSICS

DNA fingerprinting enables matching of samples with a high probability from crime scenes with those of suspects in many cases.

Increasingly DNA obtained from crime scenes is first checked to see if it matches with DNA profiles stored in databases or databanks. These databases contain the DNA profiles obtained from convicted criminals, from other crime scenes and suspects.

Forensic DNA databases have been established in many countries including the United Kingdom, the United States, Canada, New Zealand and in Australia.

### Limitations and concerns with DNA profiling and databases

DNA fingerprinting still only gives a chance or probability that two samples are from the same person.

In forensic cases, the police usually have DNA from the crime scene and are seeking the chance that there is a match with the DNA from a suspect. They are given a 'match probability' or the chance that the two samples match. The higher the probability, the greater the chance that the samples of DNA belong to the same person.

There are a number of concerns about the use of DNA material in the courtroom and the establishment of DNA databases.

These include:

- Consent for the sample
- Methods of its collection
- Procedures to ensure that it is not contaminated from the time it is taken to when it is tested
- Storage of the sample
- Its destruction after testing and
- The privacy and confidentiality of the profile generated.

An Australian Federal Government inquiry conducted jointly by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) into the protection of Human Genetic Information in Australia (<http://www.alrc.gov.au/>) examined these issues and made extensive recommendations in its final report 'Essentially Yours' (2003) on these matters.

### PATERNITY TESTING

Increasingly DNA testing is being undertaken to determine maternity, paternity or other family relationships.

Until recently, such testing was primarily paternity testing: that is, establishing whether a man had fathered a child.

Paternity testing is based on:

- A child is a combination of the DNA of its mother and father
- Using DNA profiles of all the people involved, usually mother, child and two men questioning their fatherhood, a probability can be given for one of the men being the father
- A man can be excluded as the father if he does not match with the child on at least two loci
- Inclusion as a father is usually with 99.9% probability

### Concerns associated with Paternity Testing

Paternity is often challenged in court proceedings when child maintenance, custody, and succession to property is an issue. The results of DNA profiling will only be able to be considered by the Family Court if undertaken by a laboratory accredited for paternity testing.

Paternity tests, however, are now available commercially, by mail order and through the internet.

This raises important issues for consideration including:

- The importance of informed consent by both parents for a sample to be taken from a child
- Quality control of the test undertaken
- The availability of counselling after the test result and
- Privacy of the individuals involved, particularly in relation to mail order paternity tests.

### ANCESTRY

DNA testing has recently become popular for people looking for clues about where their ancestors might come from and about relationships in families.

Genetic ancestry testing looks at the level and type of genetic relationship between individuals. As in other DNA testing, it analyses repeats of DNA sequences of letters or large numbers of variations across the whole genome and compares them to databases to look for similarities.

Ancestry testing is based on the fact that:

- Certain patterns of genetic variation are often shared among people of particular backgrounds
- The more closely related two individuals, families, or populations are, the more patterns of variation they typically share.

### Ancestry testing has a number of limitations

Ancestry testing is commonly offered as an online test through private companies. As different companies compare test results to different databases, ethnicity may not be consistent.

In addition, findings about ethnicity may be different from an individual's expectations as humans have mixed with different populations throughout history and consequently individuals may have many different variations in their DNA.

It is helpful to know what type of testing is being undertaken for ancestry to ensure testing will provide some clues to the questions being asked.

Three types of genetic ancestry testing are commonly used:

#### Y chromosome testing (YDNA testing):

This is used to explore ancestry in the direct male line. Y chromosome testing can only be done on males because females do not have a Y chromosome. As the Y chromosome passes down almost unchanged from father to son variations or **short tandem repeats** (STRs) in the Y chromosome can indicate whether families of the same surname are related.

#### Mitochondrial DNA testing (mtDNA testing):

This type of testing identifies genetic variations in mitochondrial DNA and is used to provide information about the direct female ancestral line. Both males and females have mitochondrial DNA and this type of testing can therefore be used on both sexes. However, mitochondrial DNA is only passed on by the mother and therefore tracks the female ancestral line only.

#### Autosomal DNA testing (atDNA):

These tests evaluate large numbers of variations across a person's entire genome. The results are compared with those of others who have taken the tests to provide an estimate of a person's ethnic background. This type of test may be used because Y chromosome and mitochondrial DNA test results represent only single ancestral lines, and do not capture the overall ethnic background of an individual. However results may show some variations between family members.

Larger research studies are currently being conducted by scientists to explore the history of populations as they arose, migrated, and mixed with other groups.