Fact sheet **17**

FORENSIC, PATERNITY AND ANCESTRY
DNA TESTING

This fact sheet talks about DNA testing for non-health-related reasons.

IN SUMMARY

- DNA testing for the following looks for small differences and similarities between people in their DNA
  - forensics (to do with solving crimes)
  - paternity and maternity (checking who your biological parent is)
  - ancestry (looking for likely ethnic and/or cultural background)
- DNA profiling (DNA patterns) 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 study of information in our chromosomes and genes that provide the instructions (the code) for making proteins. DNA testing for forensics, paternity and ancestry looks for small differences and similarities between people in their DNA. This type of testing usually involves looking at sections between genes (non-coding DNA) and looking for similarities and differences. DNA testing that looks for variations across the whole genome (all of the DNA code) is now also being used.

ABOUT DNA

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

There are about 3 billion base pairs spelling out the DNA code. There are over 20,000 genes found in the DNA of each person, and each gene has its own specific location or address. Genes plus the region between each gene (**non-coding DNA**) make up a person's **genome**.

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

- Although each person's sequence is unique, we are mostly alike in the sequence of letters in our DNA
- DNA samples from two people differ on average almost one letter or 'base' per thousand. Therefore, there are several million differences in the DNA between two unrelated people
- The closer two people are related to each other, the higher 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 work out identity.

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TESTING NON-CODING DNA FOR NON- MEDICAL REASONS

Unlike tests on DNA for medical reasons, 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 may for example have different numbers of the repeated sequences in different people in the population. These sections of DNA have a higher chance of being unique between people compared with other sections of DNA, and may be used for profiling.

Our chromosomes come in pairs, which means that we each have two partnering loci for every paired chromosome. There will be a specific number of repeats at each of these loci on both chromosomes in each pair. This number of repeats may be the same or different between the partnering loci.

Information from specific loci may be used to make a DNA pattern or DNA profile for each person.

CREATING A DNA PROFILE (OR PATTERN) FOR A PERSON

An approach to DNA profiling may include: (*Figure 17.1, 17.2*):

Step 1:

- In the laboratory, polymerase chain reaction (PCR) produces many copies of DNA with short tandem repeats (STRs). These are specific short sequences of the DNA letters of different sizes.

Step 2:

- The copied DNA has fluorescent labels and may be applied to a narrow tube (capillary) that behaves like a sieve. An electric current may then be used to separate the DNA fragments according to size.

  The biggest pieces move the slowest and so will come out of the capillary last and smaller pieces come out first.

- Fluorescent signals will be read by an instrument every time a DNA fragment comes off the capillary, and the timing of this will be according to size

- The unique readout (profile) for a person will be a series of 'peaks' of different colours that may be compared between individuals.

Figure 17.1:
Genetic testing to create a DNA profile of a person. Image from yourgenome, Genome Research Limited at www.yourgenome.org/facts/what-is-a-dna-fingerprint

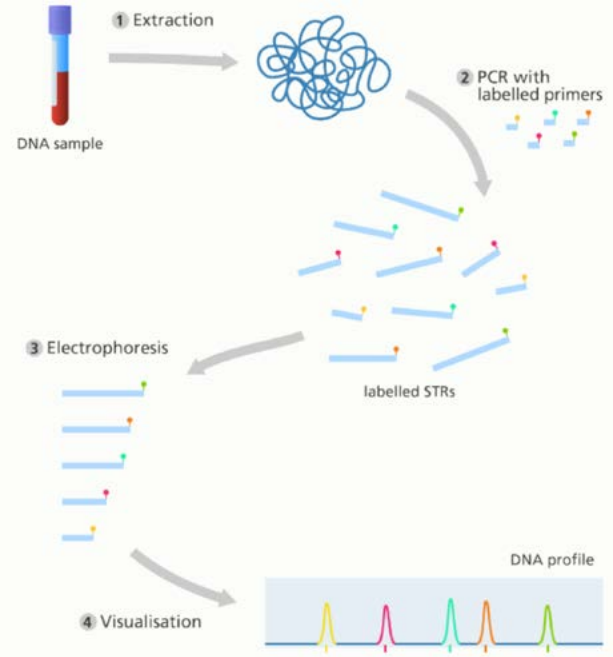
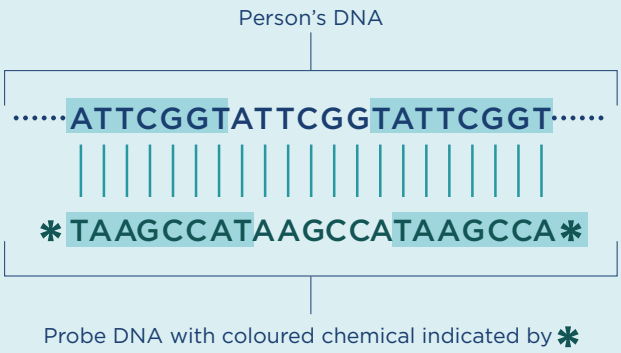


Illustration showing the steps in DNA profiling. Image credit: Genome Research Limited

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



FORENSICS

DNA profiling helps match samples from crime scenes with those of suspects with high probability.

Increasingly, DNA 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 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 Australia.

LIMITATIONS AND CONCERNS WITH DNA PROFILING AND DATABASES

DNA profiling 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 'match probability, the greater the chance that the samples of DNA belong to the same person.

There are some concerns about using DNA material in the courtroom and the storing of information on DNA databases.

These include:

- Consent for getting the sample
- How it is collected
- Making sure that the sample is not mixed with someone else's DNA from the time it is taken, to when it is tested
- How the sample is stored
- Whether the sample is destroyed after testing and
- The privacy and confidentiality of the profile made.

PATERNITY TESTING

DNA may be tested to work out maternity, paternity or other family relationships.

Until recently, such testing was mainly paternity testing: that is, working out whether a man had fathered a child.

Paternity testing is based on:

- A child is a combination of the DNA from their mother and father
- Using DNA profiles of all the people involved. This is usually the mother, child and two men questioning their fatherhood, a probability can be given for each man 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 may be challenged in court proceedings when child maintenance, custody, and succession to property is an issue.

The results of DNA profiling will only be considered by the Family Court if done by a laboratory approved for paternity testing.

Paternity tests, however, are 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 people 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 people. 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 DNA patterns 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 someone's expectations as humans have mixed with different populations throughout history. This means that we may have many different variations in our DNA.

It is helpful to know what type of testing is being organised for ancestry to make sure it will provide the right answers to the questions being asked.

The following are genetic ancestry testing that may be 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 show whether they may be from the same family via the male line.

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. Mitochondrial DNA is, however, only passed on by the mother and therefore it does not track through the male line.

Autosomal DNA testing (atDNA):

These tests look at 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 the Y chromosome and mitochondrial DNA test results represent only single ancestral lines. They do not capture the overall ethnic background of a person. Results may, however, show some variations between family members.

Larger research studies aim to look at the history of populations as they arose, migrated, and mixed with other groups.