Unit
1A

Chapter 17
DNA and inheritance

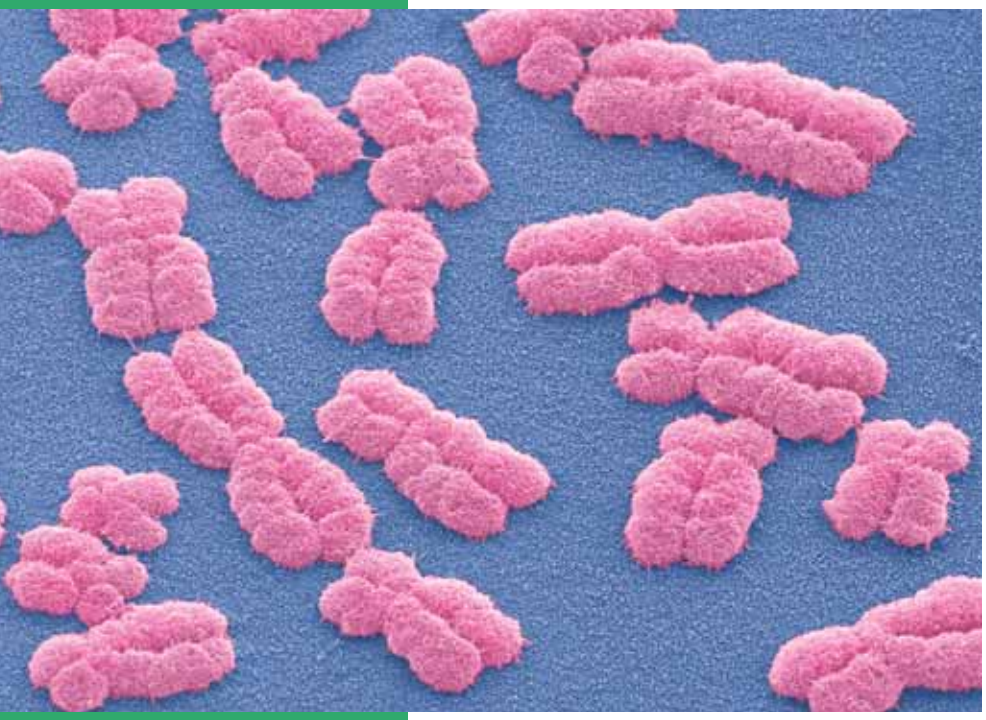


Figure 17.1 A coloured scanning electron micrograph of human chromosomes. Chromosomes are made up of deoxyribonucleic acid (DNA)

Unit content

Inheritance
DNA carries the genetic information that determines the characteristics of a person. People can be identified by their DNA.

DNA:
- and its relationship to the terms gene, chromosome and nucleus
- carries information about the individual
- is passed from one generation to the next in reproduction
- determines some features off the offspring
- can be used, for example, to:
  - *establish family links*
  - *identify people*
  - *determine certain diseases or a predisposition to certain diseases*
  - *trace ancestry*.

Your **DNA** is in the nucleus of each of your body cells. The DNA molecules contain the genes that you inherited from your parents. DNA is short for **deoxyribonucleic acid**. Each cell in the body contains a lot of DNA because DNA molecules are very large and each cell has 46 of them. In fact, if our cells were enlarged to the size of a single garden pea, our DNA would be 10 000 m long. In real terms the total length of DNA molecules in a human cell is about 2.3 m. That's a lot of DNA! Just how does it fit into a microscopic cell?

The long thread-like DNA molecules are coiled around proteins so that they will fit inside the cell's nucleus. These coils form a tangled network inside the nucleus. When a cell divides an exact copy of the DNA has to be made and one copy of each DNA molecule has to go to each of the two new cells. For cell division the DNA becomes super-coiled to form rod-like structures called **chromosomes** (see Fig. 17.2).

DNA molecules contain genes. A **gene** is a specific part of a DNA molecule that is the code for a particular characteristic of the cell. The genes make up the **genetic code** that determines the structure and activities of the cell. Unless you have an identical twin, the genes in the cells of your body are different from those of all other people in the world. Together, all the genes in your body make up the code that makes you who you are. Later on in this chapter, we will discuss how each person's unique genetic code allows DNA to be used to identify individuals and to trace their ancestry.

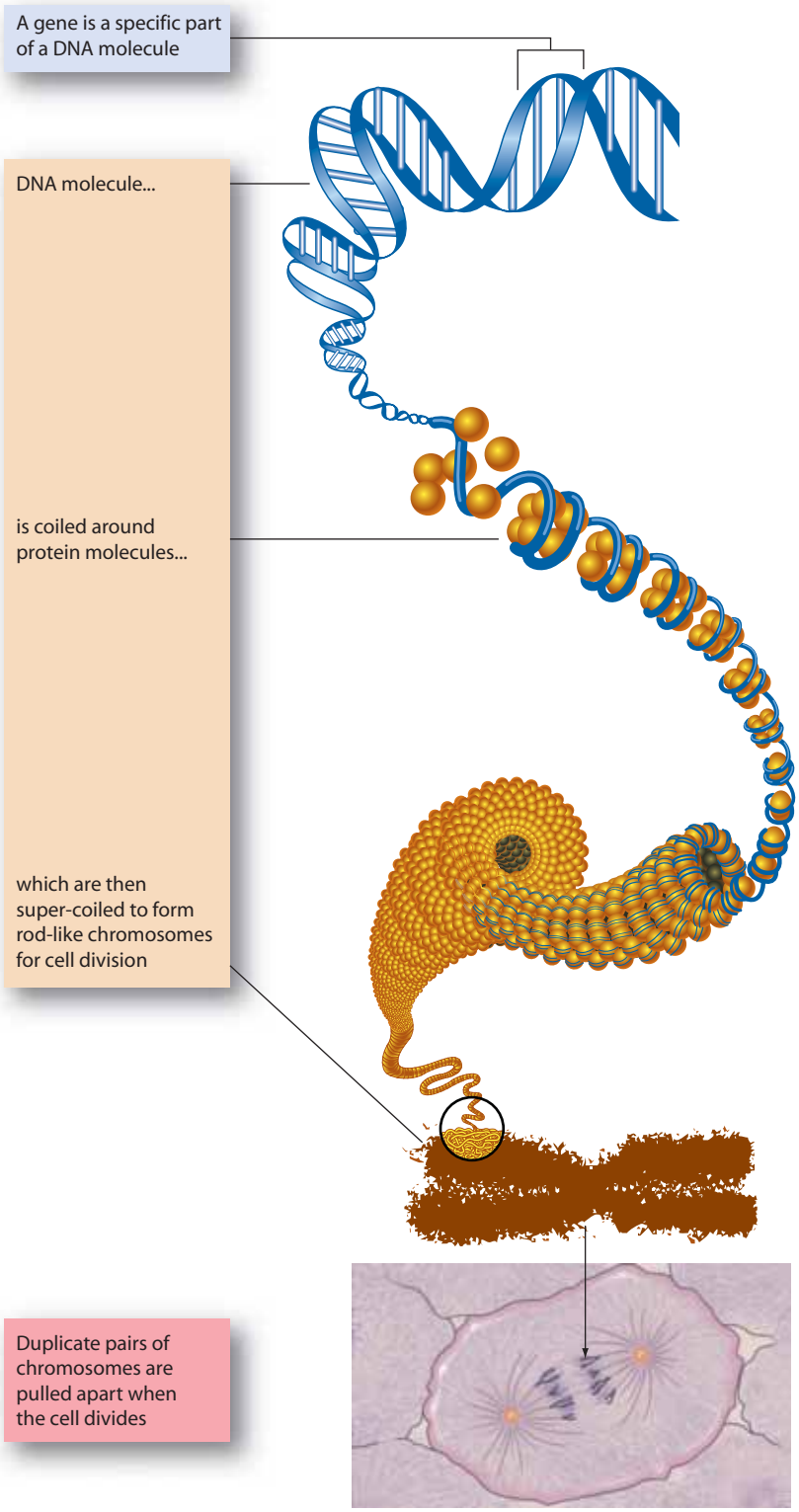


Figure 17.2 DNA is coiled around protein molecules; when a cell divides the DNA becomes super-coiled to form chromosomes

Life begins as a single cell

In the previous chapter, the joining of a sperm and an egg at fertilisation was discussed. The fertilised egg, or zygote, had chromosomes and genes from both the male parent and the female parent. You receive one set of 23 chromosomes from your father and another set of 23 chromosomes from your mother (see Fig. 17.3).

As the zygote divides by mitosis (described in Chapter 13), each new cell formed will contain the two sets of 23 chromosomes. In

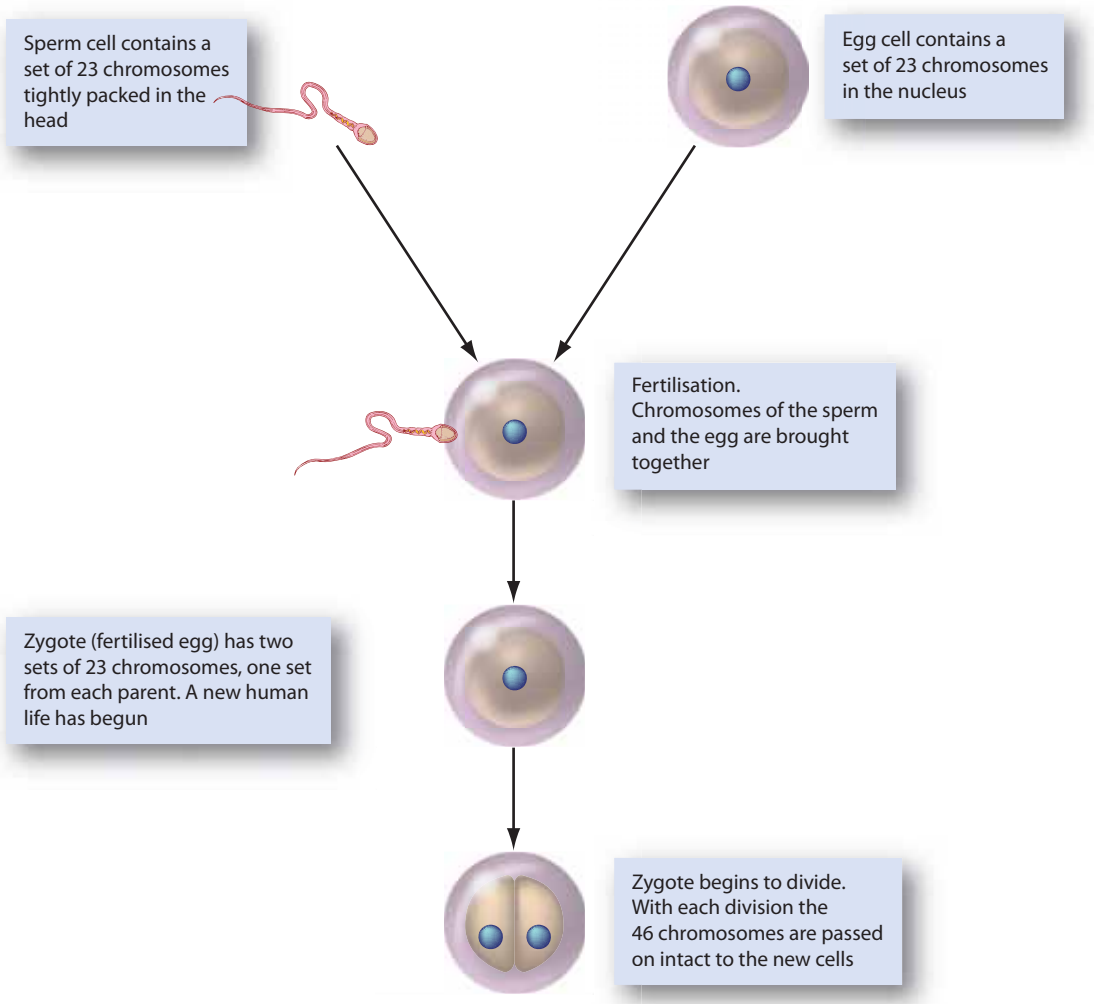


Figure 17.3 Each new human receives a set of 23 chromosomes from each parent

this way, each cell in the body has the DNA from each parent. Within each DNA molecule are hundreds of genes.

Each gene has the instructions for making a particular protein. Together the genes have all the instructions necessary to make and operate all parts of the body. Your appearance and how your body functions will depend on how your genes direct the activities of your cells.

For example, blood has red cells that transport oxygen around the body. These cells contain haemoglobin to carry the oxygen. A few of our tens of thousands of genes have the information for making the haemoglobin protein. Other genes contain instructions for making proteins in other parts of the body. Two more examples are the protein insulin that helps regulate the amount of glucose in the blood, and keratin, a protein needed for growth of hair and nails.

Table 17.1 DNA, chromosomes and genes

Structure	Location	Function
DNA: very long, complex molecules that are able to make exact copies of themselves	Within the nucleus of cells	Contains all the genetic information needed to build a functioning person from a single cell
Chromosome: rod-like structure made up of a tightly coiled DNA molecule	Formed in the nucleus when a cell is about to divide but during division is free within the cell cytoplasm	A compact form of the DNA molecule that is easily passed to the new cells at cell division
Gene: a particular section of a DNA molecule	Each DNA molecule is made up of a large number of genes	Each gene contains the information necessary for a cell to make a particular protein

DNA, the carrier of genetic information

Why do we look a bit like our parents? And why do we look a bit like our brothers or sisters? It is because DNA is the carrier of our genetic information. Each person receives a set of chromosomes from the mother and a set from the father. These chromosomes contain the genes that determine a person's characteristics. The passing of genes from parent to child is the basis of inheritance.

If we inherit our parents' genes, then why don't we look exactly like them? Children are not exactly like their parents because each child receives half their genes from their mother and the other half from their father. In this way, each child has some characteristics from each parent. Some individuals may have their mother's eye colour, but their father's hair colour; some may be the same blood group as their father but a different blood group from their mother and so on.

As we have explained (see Fig. 17.3), every human has two complete sets of 23 chromosomes. That is, their cells have 23 *pairs* of chromosomes. Each gamete (sperm or egg) has only one chromosome from each pair. The chromosomes in each pair separate randomly into the gametes when they are formed. Thus, each sperm and egg has 2^{23} possible combinations of chromosomes. That's a total of 8 388 608 possible combinations. At the time of fertilisation each parent contributes 23 chromosomes to the zygote. With 8 388 608 possible combinations of chromosomes in the egg and 8 388 608 possible combinations of chromosomes in the sperm there are 8 388 608 × 8 388 608 possible combinations of chromosomes in the fertilised egg. Can you see why no two individuals have identical sets of chromosomes and therefore identical DNA? The exception to this is identical twins, who develop from one fertilised egg and therefore have the same DNA.

DNA as a means of identification

Ancestry

As we have just seen, the DNA that is in your cells is unique. Although it has come from each of your parents, the way it has come together in your cells is different from all other people. Because it is so distinctive, a person's DNA can be used as a means of identification. In the late 1960s, scientists developed techniques to cut the long

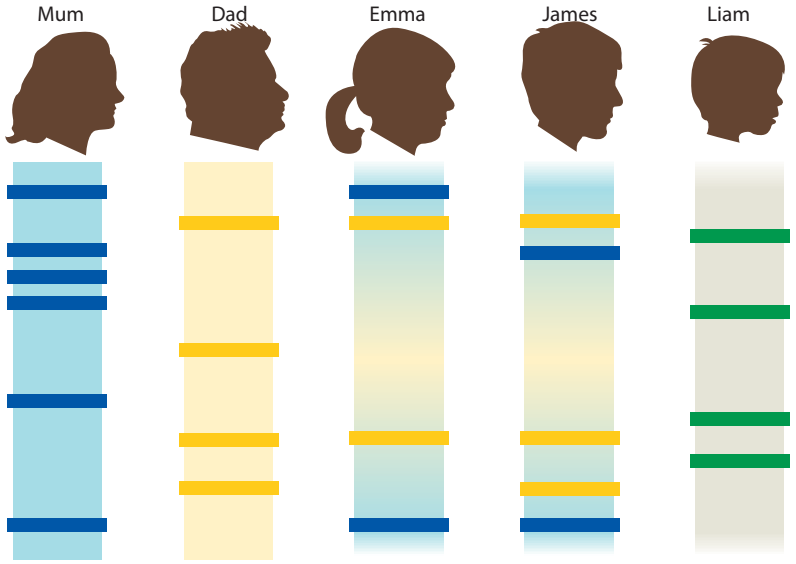
Figure 17.4 A DNA fingerprint

molecules of DNA into pieces. These pieces were then separated and placed on an X-ray film to form an image of where each piece was located. The pieces appear as different bands on the film and are often called a **DNA fingerprint** or **DNA profile** (see Fig. 17.4).

DNA fingerprints can be used to identify individuals and to trace ancestry. It has been very useful in helping some individuals identify their biological parents. As you have received half of your DNA from each of your parents, you would expect your DNA fingerprint to have some of the bands similar to those of your mother, and other bands similar to those of your father. For a person who may have been separated from their parents at birth, DNA fingerprinting can help establish the likelihood that a particular individual is one of their parents.

Figure 17.5 shows an example of how DNA fingerprinting can be used. The DNA fingerprints of a family are shown. Emma and James are biological brother and sister. They do not have the same DNA profile but there are similarities. You can also see that each of them has some bands in their profiles that are the same as bands in each of their parents' profiles. Yet neither has DNA that is identical to one parent or the other. Liam is adopted. His DNA has no bands that are the same as his adoptive parents.

Figure 17.5 DNA fingerprints of a family



In comparing DNA fingerprints, scientists make use of markers. A **marker** is a segment of DNA with known characteristics. These segments of DNA do not code for a protein, but they contain short sequences where the same pattern is repeated a number of times. Since the number of repeats in these sequences is inherited, they make useful points of comparison in genetic testing.

To determine a link between two individuals, specific markers on the DNA strand are studied. Scientists look for the number of repeats at each marker, and may examine between twenty and forty markers. If ten different markers on different chromosomes are examined, there is only a one in a million chance that two people will have the same number of repeats, except for identical twins. The more markers that match, the more likely it is that the two individuals are related (see Fig. 17.6).

Each of your parents received their DNA from each of their parents, and so on. DNA can be used to trace ancestry back through your family tree. As DNA is passed

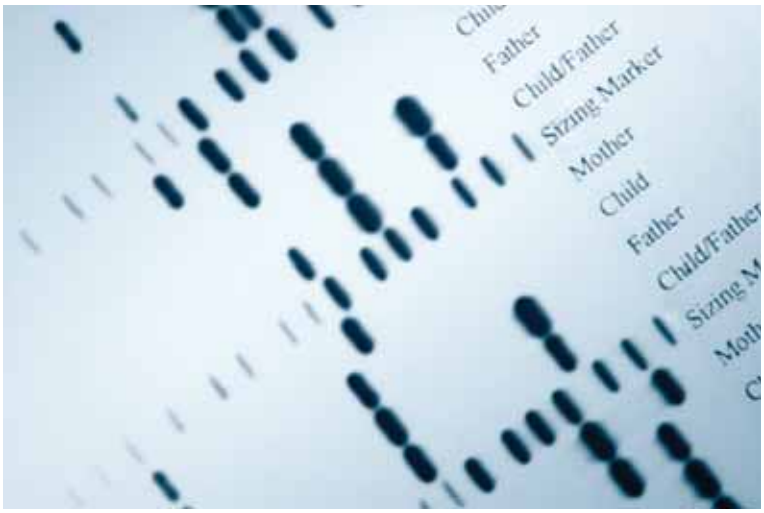


Figure 17.6 A DNA fingerprint used to help establish the father of a child: the fact that the DNA bands match does not necessarily mean that person is the father; however, a mismatch would definitely exclude that person from being the father

down from one generation to the next, some parts remain almost unchanged, while other parts change greatly. These unchanged parts of the DNA molecules provide a link between generations and they can be very useful for reconstructing family histories. However, they cannot provide a person with their entire family tree. DNA testing can only tell if two people share a common ancestor. People who marry into a family will have completely different DNA from family members who are directly related.

DNA also provides clues about ethnic origin and it has been very useful to scientists in tracing the origins of modern human populations. By using the patterns of DNA in different groups around the world it has been found that all present-day humans originated in Africa. It appears that modern humans are probably a result of migration out of Africa about 50 000 to 60 000 years ago. Recent evidence suggests that these migrating people moved very quickly, reaching places such as Australia about 50 000 years ago.

Forensic science

Police services around the world use DNA fingerprinting. It can be used to identify bodies long after other distinguishing marks have disappeared. It may also be used as evidence against a suspect in a crime. Police dramas on television frequently make use of such evidence to convict suspects. At a crime scene, police painstakingly search for objects that could be tested for DNA. Objects with blood, hairs, traces of saliva or other such evidence are carefully collected (see Fig. 17.7).

The use of DNA to help police in their investigation of crimes is relatively new. It was not until 1985 that police in the United Kingdom used DNA profiling. The first famous case was the use of DNA evidence to show that a man suspected of murder did *not* in fact commit the crime. The police then collected over 5000 samples of blood from men in the local community. DNA analysis of the samples enabled them to identify the killer.

DNA has also been used to help people who have been wrongly convicted of a crime. In 1989, Gary Dotson, jailed in the USA for rape, became the first person to have a conviction overturned on the basis of DNA evidence. In the same year

Figure 17.7 A police officer gathering evidence at the scene of a crime

You can simulate creating a DNA fingerprint at http://www.pbs.org/wgbh/nova/sheppard/analyze.html To see a video on the forensic use of DNA fingerprinting go to http://www.teachersdomain.org/resources/tdc02/sci/life/gen/sheppard/index.html and click on 'video'

Australia had its first court case involving DNA evidence. In the Australian case, blood samples from the victim's clothes were used to identify the person who committed the crime.

As a result of the increasing use of DNA profiling in police work, the Australian Federal Government provided funds for the establishment of CrimTrac. The starting point was a national DNA database, and in 2000 the CrimTrac Agency was formed to help share information between Australian police services. The following year, CrimTrac launched the National Criminal Investigation DNA Database (NCIDD) to allow police around Australia to match DNA profiles.

DNA was recently used to solve a high profile crime in the United Kingdom. The conviction of Steve Wright in England in February 2008 was due to scientists matching his DNA to that found at crime scenes. Five bodies had been found in ten days and the scientists examined hundreds of samples before they found one that contained enough DNA for a match with a profile on a DNA database. This enabled police to identify the suspect and gather further evidence. Had it not been for the use of DNA fingerprints, this killer may not have been identified quickly and may have killed others.

Disaster victims

After a disaster such as a bombing or fire, it is often difficult to identify the victims. In situations like this, scientists are called in to create DNA profiles from body parts. This occurred after the 2002 Bali bombing. Relatives of victims were asked to send objects from home, such as a toothbrush, comb or item of clothing. DNA was extracted from cells on those personal items and DNA profiles created. Using DNA profiles, fingerprints, dental records and other medical records, 182 victims were identified, eighty-eight of them Australians. In more than half the cases DNA was the deciding factor in identification.

Following the 2004 tsunami in South-East Asia, the Australian Government sent Australian forensic experts to help with the identification of the victims of that disaster. DNA profiling again proved to be invaluable in many cases.

Figure 17.8 A relative of a tsunami victim has a sample taken for DNA profiling

DNA as a predictor of disease

Some diseases are inherited—they are caused by faulty DNA. In some cases a person may have the faulty gene but they do not develop the disease. They are known as **carriers**. For that disease to occur a child must inherit the faulty gene from both parents. Testing is available so that a person may know if they are carrying a gene for a disease that they do not have. Sometimes a relative may have the disease and this alerts the person to the possibility that they may be a carrier. In other cases, certain inherited diseases are more common within some ethnic groups. Individuals from such a group could then undergo tests to check whether they carry the defective gene. Most such tests are simple blood tests to establish whether the genes for that disease are present or not.

A disease for which some people may carry the gene is cystic fibrosis. Children with this condition suffer from chest infections and problems of the digestive tract. In people of European origin it is the most common cause of death from a genetic disease. Up to 3% of people of European origin are thought to be carriers of the gene for cystic fibrosis. A saliva sample can be tested so that people with cystic fibrosis in their family can find out if they carry the gene for the disease. This information is very useful for people wishing to have children because a child can only inherit the disease if both the mother and the father carry the gene.

A simple blood test can be used to determine whether a baby has cystic fibrosis. In Australia, all babies are tested within two to three days after birth. If the baby has the disease, a special diet is given so that the child can function as normally as possible.

DNA analysis can also be used to find out if individuals will develop a particular disease later in life. Huntington disease is one such condition. It is a genetic disease that can be passed to the next generation when only one parent is affected. The symptoms of the disease do not appear until later in life. This means that a person with the condition may not be aware that they have inherited the disorder until after they have had children of their own.

DNA technology has enabled individuals with Huntington disease in their family to be genetically screened. Blood from participants is collected and checked for the presence of three established gene markers. These tests let an individual know if he or she will develop the condition. Since the condition is incurable, nothing can be done to help a person who will develop the disease. However, the information does allow that person to make a decision about having children.

Testing for genetic diseases is also done on human embryos. In the case of test tube babies the embryo can be tested before it is implanted into the uterus. Embryos that will produce a person with a disease can be destroyed. Tests can also be done on embryos that have developed naturally. If the developing baby is found to have faulty genes the parents may decide to have an abortion.

Working scientifically

Activity 17.1 Crime scene investigation

In this chapter we discussed how DNA analysis could help police to solve crimes. DNA can be used to either eliminate or establish a suspect's presence at a crime scene. However, DNA evidence may not be completely reliable and has to be treated with caution.

In this activity, you will use the following Internet sites to learn more about the use of DNA in criminal investigations.

- Is DNA evidence infallible? Read a story from the ABC's *Catalyst* program and decide for yourself: http://www.abc.net.au/catalyst/stories/s1199805.htm
- What if there is no suspect? Log on to find out how to build a picture of a suspect using DNA: http://www.abc.net.au/catalyst/stories/s720292.htm
- A shadow of doubt. More on how DNA can be misinterpreted: http://www.abc.net.au/catalyst/stories/s724179.htm

You may be able to find other sites that give you more information about the use of DNA in criminal investigations.

REVIEW QUESTIONS

1. **(a)** What is a gene?
   (b) Explain the relationship between genes, chromosomes and DNA.
2. How do genes direct the activities of cells?
3. All the cells of your body have originated from the zygote formed when a sperm from your biological father joined with an egg from your biological mother. Where did the genes that made up the zygote come from?
4. **(a)** Describe what is meant by a DNA fingerprint.
   (b) What can DNA fingerprints be used for?
5. How can police services make use of DNA fingerprints?
6. DNA has also been used to identify victims of major disasters. Describe how it was used to identify victims of the Bali bombings.
7. Describe the role of DNA in identifying people with genetic diseases.

APPLY YOUR KNOWLEDGE

1. The average length of the DNA molecules in human cells is 5 cm and there are 46 of them in each cell. Explain how 2.3 m of DNA can fit inside the nucleus of a cell.
2. Examine Figure 17.8 and compare the DNA profile for the blood on the hammer with the blood from the four suspects. Who do you think was the murderer? Give reasons for your answer.

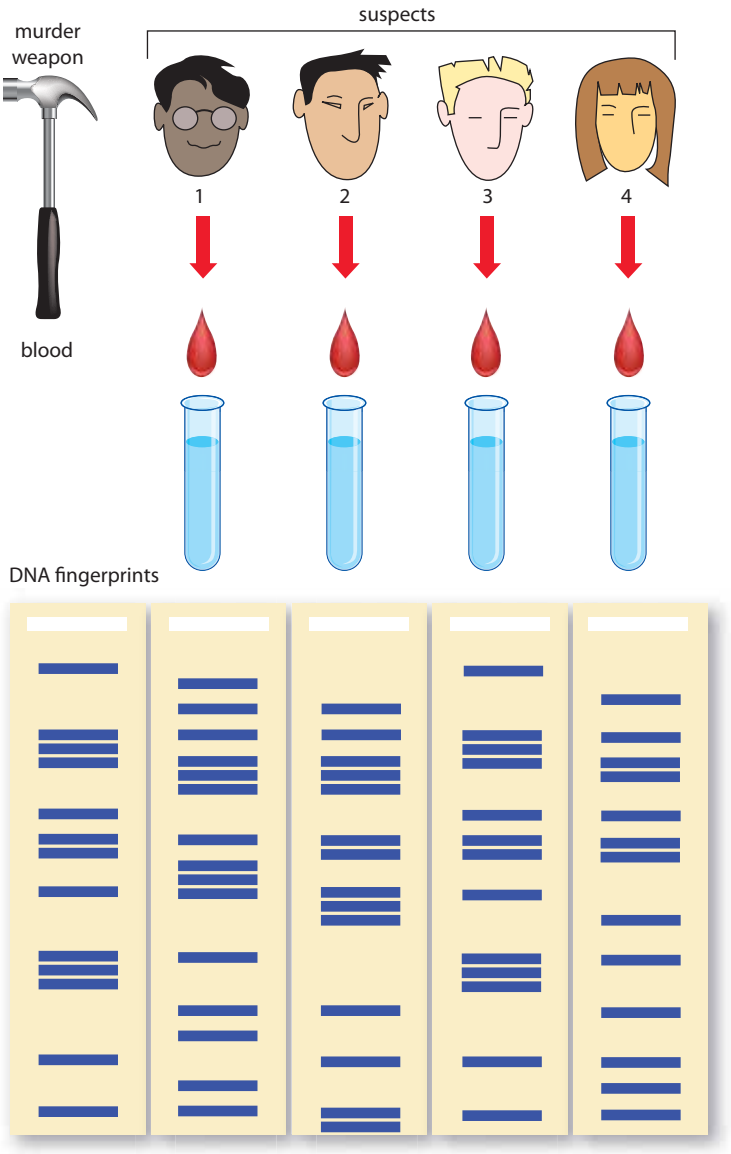


Figure 17.9 DNA fingerprints of forensic samples

3. Why is it that DNA can eliminate a person from being at a crime scene, but cannot with absolute certainty say a person was there? List some of the limitations of the use of DNA as a means of identification.
4. Australia has been involved in the identification of victims of disasters in a number of places around the world. Two of these were mentioned in this chapter. Use references to see what other identification tasks have involved Australian investigators.
5. DNA has not only been used as a means of identifying humans. Top breeders of dogs, cats and other animals use DNA to verify that they have a pedigree animal. Use the Internet to find out how important DNA has become for those breeding pedigree animals.
6. In this chapter, a number of diseases were mentioned that can be identified by DNA analysis. Use references to find at least five other diseases that are frequently the subject of DNA analysis.
7. On rare occasions newborn babies have been given to the wrong mother by the hospital where the baby was born. What could a mother do if she suspected that a baby was not her own?