Unit
1A

Chapter 18
Inherited differences

Figure 18.1 A family group with several generations

Unit content

Variation and evolution
Variations occur between individuals. Families can show different genetic traits.

Variation:
- offspring resemble their parents in many ways
- genetically identical twins can be used to study differences between nature and nurture (influence of genes and environment)
- characteristics run through families:
  - physical features
  - predisposition to diseases.

Have you ever heard the relatives of a newborn baby talking about the baby's appearance? They will say things like 'she looks just like her mother', 'he has his father's nose', 'her eyes are just like her grandmother's' or 'his mouth looks just the same as aunt so and so's'. Some of our characteristics are similar to our relatives because some of our genes are the same as theirs.

Children most resemble their parents, and their parents resemble their grandparents. They also show physical characteristics that are often very similar to their brothers and sisters, and to other close relatives. Features such as eye and hair colour, nose shape, body build and others seem to occur in generation after generation (see Fig. 18.1).

In the previous chapter, we learnt that DNA is the carrier of our genetic information. Each of us receives a set of chromosomes from each of our parents, and these chromosomes contain the genes that determine our characteristics. This passing of genes from one generation to the next is the basis of inheritance. In this chapter we will examine how some characteristics run through families—but even within a family there are differences in characteristics.

Variation between individuals

Although individuals belonging to the same species have much in common—for example, all dogs are recognisable as dogs—individuals also show differences. These differences in characteristics within a species are known as **variation**. It is now known that variation is partly inherited and partly due to the influence of the environment. With the exception of identical twins, humans possess different genes and therefore tend to develop different characteristics. However, even identical twins, if brought up in different environments, can show differences.

Inherited differences include a wide range of characteristics. Individuals differ in:

- external appearance
- internal body structure
- body chemistry
- internal functioning
- patterns of behaviour.

When we think of environment we usually think of things like climate or where a person lives. However, environment has a much broader meaning. **Environment** is all of the non-inherited influences on a person. It will include climate and where the person lives, but will also include what they eat, diseases they have suffered, sports they have played, accidents they have been involved in and so on.

How do we decide which of a person's characteristics are due to the genes they have inherited and which are due to their environment? One of the ways of investigating this is through the study of twins. Identical twins have the same genes so any differences between them must be due to environmental influences. A problem in studying twins is that most identical twins are brought up together so that the two of them will be exposed to very similar environmental influences.

Even though twins usually receive similar treatments as they grow up, their environments cannot be exactly the same. There will always be differences in eating habits, the type of exercise undertaken, how much sleep they get and many other factors. Even before they are born, identical twins have differing environments because they occupy different parts of the uterus.

(a)

(b)

Figure 18.2 The shape of the earlobes is one inherited characteristic: a person may have **(a)** attached earlobes or **(b)** free earlobes

Figure 18.3 Identical twins that have been brought up together: what differences can you see?

Figure 18.4 Identical twins that have been brought up separately: what differences can you see between these twins?

In one study, the standing height of fifty pairs of identical twins was measured. The individuals of each identical twin pair were not exactly the same height. On average, the members of a pair differed by 1.7 cm. As identical twins have inherited the same genes, the source of this variation must be the environment.

Sometimes, and for a whole variety of reasons, identical twins are separated when they are very young and brought up in different homes. Study of identical twins under these circumstances provides another way of examining environmental influences on their characteristics. Since their genes are identical, any differences must be due to the different environments. In one investigation of twins reared apart, nineteen pairs of identical twins who had been separated and brought up in different homes were examined. In some cases the individuals did not even know they had a twin until the study was done. The average difference in height between pairs of identical twins reared apart was 1.8 cm. This result is almost exactly the same as that for identical twins raised together. This result suggests that the effects of the environment on height for twins raised together or raised apart is not that much different. It appears therefore that inheritance is the major influence on the height of an individual.

Twins who are not identical are usually subject to the same kinds of environmental factors because they are born to the same parents at the same time. However, they have inherited different genes because each develops from a different egg and a different sperm. If the genes for height were different in non-identical twins, then we would expect the average height difference of a pair of non-identical twins to be larger than that between identical twins. In fact, it is. The study found that it was over twice as large (on average 4.4 cm), confirming that inheritance is the most important factor in body height.

Table 18.1 Twins

Identical	Non-identical
Formed from the same sperm and the same egg	Formed from different sperm and different eggs
Identical genes	Different genes
Must be the same sex	Need not be the same sex
Identical in appearance	No more alike than any brother or sister

Figure 18.5 A set of non-identical twins

As the environment for a pair of non-identical twins is relatively similar, you would expect such twins to look more alike than ordinary brothers and sisters. Brothers and sisters are similar to non-identical twins in that each has resulted from a different egg and a different sperm. However, the environmental influences for ordinary brothers and sisters are often quite different. Factors such as a difference in age often result in different family situations. Therefore you would expect greater differences between normal brothers and sisters than between non-identical twins. However, this was not the case. The average difference in height between pairs of ordinary brothers and sisters, measured at the same age, was 4.5 cm, almost exactly the same as that for non-identical twins. Therefore, the differences between the environments of ordinary brothers and sisters, and the environments of non-identical twins, do not seem to be important for body height.

The results of investigations on the height of twins and other brothers and sisters are summarised in Table 18.2.

While the various twin studies show that inheritance is the major factor in determining height we only inherit a potential to grow to a certain height. Sometimes environmental factors limit that potential. Young people in South Korea are about 12 cm taller than people of the same age in North Korea. Both groups would have about the same potential for growth but North Koreans do not reach their potential because of poor diet and other environmental factors.

There are many other examples of human characteristics that result from an interaction between heredity and environment. Weight is an obvious one. Identical twins that had totally different diets would tend to have different body shapes, especially if one twin ate a higher proportion of foods containing fats. This would lead to one twin weighing more than the other. In this situation the influence of the environment appears to be far greater than the genes that have been inherited.

Table 18.2 Results of investigations into the heights of twins and other brothers and sisters show that inheritance is the main factor in determining a person's height

Investigation	Genes	Environment	Average difference in height (at the same age)
Identical twins	Same genes	Brought up together	1.7 cm
Identical twins	Same genes	Brought up apart	1.8 cm
Non-identical twins	Different genes	Brought up together	4.4 cm
Ordinary brothers and sisters (not twins)	Different genes	Brought up together	4.5 cm

Another example where the environment appears to have a big influence is intellectual development. Each of us inherits from our parents the potential for a certain level of intellectual development. Whether the potential is realised will depend on the environment. Babies who lack the stimulation of parents and others playing with them and talking to them are unlikely to develop their full intellectual potential. In a similar study to the one related to height, the intellectual development of identical twins reared together was compared with that of identical twins reared apart. The results indicated that the difference in intellectual development was slightly greater for those twins reared apart.

For more information about twin studies and the Swedish Twin Registry go to http://www.abc.net.au/rn/talks/8.30/helthrpt/stories/s892403.htm

Australia has a registry of twins that aids scientists in studying the effects of the interaction between heredity and environment. However, the largest twin registry in the world is in Sweden and it dates back to 1886. The Swedish register has over 140 000 twins and they are in about equal thirds of identical, same sex non-identical and unlike sex pairs. Such a large database has allowed scientists to study the contribution genes make compared to the influence of the environment for a wide variety of characteristics: for example, the effect the environment may have on certain diseases, exposure to toxic chemicals, taking certain medications or diet. In addition, by studying identical twins who have been brought up apart, the effects of a shared environment in the home when growing up can also be determined.

Studies of such twin data have found that a person's genes affect the chances of developing a cancer. Genes account for about 30% of the chance of getting some forms of cancer. On the other hand, for some cancers, genes don't appear to have any effect at all.

Characteristics in families

Many physical characteristics appear to be common in certain families. In your own family a particular physical feature may be common. Characteristics such as eye colour, shape of the nose, hair colour, length of the face and height may be evident across a number of generations. Examine Figure 18.1. What physical features appear to be common in this family?

Table 18.3 Some inherited physical characteristics

Characteristic	Various forms of the characteristic
Ear lobes	Free or attached
Lips	Broad or thin
Eyelashes	Long or short
Nostrils	Broad or narrow
Natural hair colour	Dark, red or blonde
Body hair	Little body hair or a lot
Hair type	Curly or straight
Eye colour	Brown, hazel, green, blue or grey
Nose shape	Roman or straight
Cheeks	Dimples or no dimples
Chin	Cleft in chin or no cleft
Hairline shape	Widow's peak or straight
Thumb extension	Hitchhiker's thumb or straight thumb
Ability to roll the tongue	Tongue roller or unable to roll tongue
Hand clasping	Cross left thumb over right or cross right thumb over left

(a)

(b)

(c)

Figure 18.6 Some inherited physical characteristics: **(a)** dimple cheeks; **(b)** cleft chin; **(c)** hitchhiker's thumb

Figure 18.7 A person who can roll their tongue

(a)

(b)

Figure 18.8 Hand clasping: **(a)** cross left thumb over right; **(b)** cross right thumb over left

Table 18.3 shows some physical characteristics that may appear in a family. There are many others. Do some of these characteristics occur in your family? For example, can every one in your family roll the tongue (see Fig. 18.7)? What happens when they clasp their hands together? Without thinking about it, fold your hands together by interlocking your fingers (see Fig. 18.8). Which thumb is on top—your left or your right? This is another characteristic that appears to run in families.

Inheritance of certain diseases

Certain diseases can be inherited. Cystic fibrosis and Huntington disease, discussed in Chapter 17, are examples of diseases that run in families. These diseases are caused by faulty DNA. They are inherited because the faulty DNA is accurately copied before being passed to the offspring from a parent. There are many other diseases that can be passed on from one generation to another.

In some cases, a child must inherit two copies of the faulty gene to show the disease. In these circumstances, called **recessive inheritance**, the parents do not show the condition and are said to be carriers (see Chapter 17). This is shown diagrammatically in Figure 18.9. Cystic fibrosis is a disease of this type.

Another disease to be inherited in this way is **thalassaemia**. Thalassaemia is a disease in which a defect in the formation of the red blood pigment, haemoglobin, results in anaemia. Anaemia is when the blood is not able to carry sufficient oxygen to the cells. People with thalassaemia require frequent blood transfusions throughout their lives and special drugs to remove iron that tends to build up in the body.

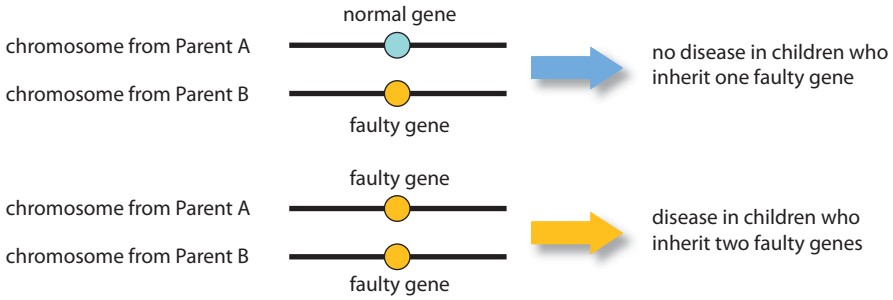

Figure 18.9 Recessive inheritance: two copies of the faulty gene need to be inherited for a child to be affected by the disease

With some diseases, only one parent has to pass on the faulty gene for the disease to occur. This is **dominant inheritance**. Huntington disease is inherited in this way. This is shown diagrammatically in Figure 18.10. Another disease that is inherited in this dominant way is **achondroplasia**. This disease is a form of dwarfism. People with the disease have short limbs, a prominent head and normal intelligence.

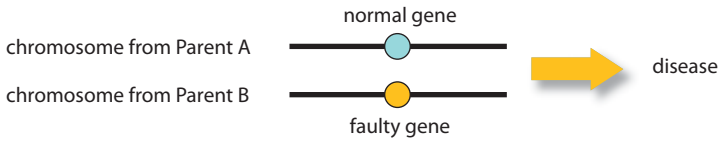

Figure 18.10 Dominant inheritance: only one copy of a faulty gene needs to be passed on for a child to inherit the disease

In the situation with Huntington disease and achondroplasia, only one parent needs to have the disease for it to be passed on. On the other hand, diseases such as cystic fibrosis and thalassaemia need both parents to be carrying the faulty gene for it to be passed on. All these diseases have clear genetic links. In others, such as some forms of cancer, the genetic links are not so clear. In these cases a combination of genes and environment is involved. A person is not born with the disease. They only inherit an increased chance of developing the disease. This increased chance of getting a disease is called a **predisposition** to that disease. It does not mean the person *will* get the disease. Certain environmental or lifestyle factors need to be present to cause the disease to develop. For example, a person may have a gene that increases the chance of developing lung cancer. If that person was also a smoker, they would be very likely to develop the disease.

For a video and simulated test for the inherited disease haemochromatosis go to http://www.koshland-science-museum.org/exhibitdna/inh01.jsp#

Breast cancer is another disease where there are genetic links in some families. However, only 5% to 10% of all breast cancers are *caused* by inherited genetic factors. Inherited breast cancers are due to two particular faulty genes. Out of every 100 women who inherit one of these faulty genes, as many as sixty will develop breast cancer by the age of 50. By the age of 70, approximately eighty will have developed breast cancer.

With breast cancers that are not inherited there is strong evidence of a link with the female hormone oestrogen. Studies show that oestrogen stimulates the cells of the breast to form cancerous cells. The longer a woman is exposed to high levels

of oestrogen in the body the more likely she is to develop breast cancer. Women who take birth control pills or have used oestrogen replacement therapy after menopause are therefore at a greater risk. On the other hand, the same studies indicate that the less time a woman is exposed to oestrogen, the lower her risk of developing breast cancer. Pregnancy, breastfeeding, physical activity, and a low fat, high fibre diet all help to reduce the amount of oestrogen flowing through the body.

For breast cancers that are not inherited, a family history of breast cancer indicates a greater risk of developing the disease. If a woman's mother, sister or aunt had breast cancer, there is an increased chance of the woman getting the disease. Women who have not had children, or women who did not have their first child until after the age of 30, also have a two-to-five-times greater risk. Other factors that can increase the chance of getting breast cancer include: being overweight, high fat intake, excessive alcohol consumption and dietary deficiencies.

Heart disease is another condition where genes appear to play a part. For example one of the main causes of heart disease is clogging of arteries through a build-up of fatty deposits. These block the flow of blood. Scientists have discovered a gene that increases the chances of a person getting clogged arteries. This gene does not cause the fatty deposits in the arteries. It works with other factors, such as diet and possibly other genes, to put those individuals who carry it at a greater risk of getting heart disease.

Working scientifically

Activity 18.1 Family characteristics

Certain physical characteristics are common in families. In this activity, you will select a number of easily recognisable physical characteristics and determine which members of your family have them.

What to do

1. Draw up a table similar to Table 18.4.
2. Observe as many of your relatives as possible including parents, brothers and sisters, grandparents and cousins. You can choose aunts and uncles but only if they are genetically related to you; that is, do not choose an aunt or an uncle who has married into your family. Replace the numbers at the top of the columns with the people's names. Using the highlighted letter, list the characteristics for all your family members. Add extra columns if you need to.
3. List your own characteristics in the last column.

Studying your data

1. List those characteristics that appear to be common to most of the members of your family.
2. Were there any characteristics that did not appear in your family at all?
3. Compare your data with other members of your class. How different are their family characteristics from yours?

Table 18.4 Family characteristics

Characteristic	Relative								
	1	2	3	4	5	6	7	8	9
Ear lobes (**A**ttached or **F**ree)									
Lips (**B**road or **T**hin)									
Eyelashes (**L**ong or **S**hort)									
Nostrils (**B**road or **N**arrow)									
Natural hair colour (**D**ark, **R**ed or **B**lond)									
Body hair (**Y**es or **N**o)									
Hair type (**C**urly or **S**traight)									
Eye colour (List colour)									
Nose shape (**R**oman or **S**traight)									
Cheeks (**D**imples or **N**o dimples)									
Chin (**C**left or **N**o cleft)									
Hairline shape (**W**idow's peak or **S**traight)									
Thumb extension (**H**itchhiker or **S**traight)									
Ability to roll the tongue (**R**oller or **N**on-roller)									
Hand clasping (**R**ight thumb or **L**eft thumb)									

REVIEW QUESTIONS

1. Why do offspring resemble their parents? In your answer, explain the role that genes play in inheritance.
2. How has the study of identical twins helped to work out the effect of the environment on human characteristics?
3. Give one example where inheritance appears to be the major influence on a characteristic, and one where the environment is more important.
4. List the differences between identical and non-identical twins.
5. Explain the way diseases such as cystic fibrosis and thalassaemia are passed from one generation to the next. How is this different from the way diseases such as Huntington disease and achondroplasia are inherited?
6. Explain what is meant by the phrase 'a predisposition to a disease'?
7. List examples of diseases where there may be a predisposition in some families. What other factors need to be present for these diseases to develop?

APPLY YOUR KNOWLEDGE

1. In early 2008, Australia had thirty-six sets of identical triplets. Identical twins are formed when the fertilised egg splits into two cells each of which develops into an embryo. Find out how identical triplets are formed.
2. **(a)** Young North Koreans who grew up in the 1990s when there was severe famine in the country are, on average, much shorter than their parents. Suggest a reason for this.
   (b) Will the heights of the children of North Koreans who grew up in the 1990s be similar to their parents or their grandparents?
3. Breast cancer may be caused in many ways. Some forms have a definite inherited cause. Others appear to be due to environmental influences. Find out how a woman can reduce her chances of developing breast cancer.
4. Prostate cancer is of increasing concern as male members of the population grow older. What is the major influence on this disease—genetics or environment?
5. In a family with a predisposition to lung cancer being exposed to cigarette smoke may be enough to trigger the development of disease. Use the Internet to research the effect of passive smoking on the development of lung cancer.
6. Some inherited characteristics help people to survive in their usual environment. For example, the Inuit, who live in the Arctic, tend to be short and heavily built. On the other hand, Australian Aboriginal people from Central Australia are usually tall and slim. Suggest how these inherited characteristics would help the Inuit and the Aborigines to survive in their natural environment.