Unit
# 1B

# Chapter 19
# Human variation

**Figure 19.1** Variation in humans

## Unit content

**Inheritance**
Genes are the fundamental units of heredity.

The gene concept:
- a gene is a section of DNA
- genes influence characteristics
- in sexual reproduction, offspring contain chromosomes from both parents.

**Variation and evolution**
Phenotype is affected by the environment and genotype.

Relationship between genotype and phenotype:
- genes alone or environment alone or genes and environment together contribute to phenotype
- phenotype can be used to deduce genotype
- examples of characteristics determined by genetics only, environment only and both
- the phenotype of offspring is limited by genes of the offspring's parents.

**Cells, metabolism and regulation**
The body detects and responds to changes in its internal environment that are outside its tolerance limits. Dysfunctions are caused when tolerance limits are exceeded.

Tolerance limits:
- conditions resulting from exceeding tolerance limits
- individual difference related to tolerance limits *e.g. diet, alcohol*.

Some people are able to roll their tongue, others are not. There is a photograph of a tongue roller on page 221. Amy cannot roll her tongue but her mother and her father are both tongue rollers. Her brother Scott and sister Holly are also rollers. How is it that, in a family of tongue rollers, Amy has missed out on the ability to roll her tongue?

Tongue rolling is an inherited characteristic. Inherited characteristics are controlled by the genes that we inherit from our parents. In this chapter we will look at how the genes interact to produce the particular characteristics of a person.

A gene is a segment of DNA that contains the code for making a particular protein. Chromosomes are made of DNA so there is a large number of genes grouped together in a chromosome. Each person has two sets of chromosomes. One set came from the mother through the egg and one set came from the father through the sperm. As the cells of the new individual divide, copies of the chromosomes are passed on to the new cells. In this way every cell (except eggs and sperm) in an individual's body contains two copies of each chromosome and therefore two copies of each gene.

Most genes have two or more forms. These alternative forms of the gene are called **alleles**. Figure 18.2 on page 217 shows how a person may have earlobes that are either attached or free. These are the alternative forms, or expressions, of the gene that determines the shape of a person's earlobes. In a similar way the gene for tongue rolling has two alleles. One form of the gene gives the ability to roll the tongue, the other form does not.

## Inheritance

An individual may inherit two identical or two different alleles from their parents. When two different alleles are present they interact in specific ways. In many cases, one of the alleles will mask the effect of the other.

Alleles that mask the effect of another are referred to as **dominant**. Tongue rolling is caused by a dominant allele. This allele masks the effect of the allele for non-rolling (being unable to roll the tongue). An allele for a characteristic that is masked by another allele is said to be **recessive**. Two recessive alleles need to be present for a person to have the characteristic.

Therefore, in the case of the inheritance of ability to roll the tongue, there are three possibilities:

- two dominant tongue rolling alleles—the person will be able to roll the tongue
- one dominant tongue rolling allele and one recessive non-rolling allele—the person will be able to roll the tongue
- two recessive non-rolling alleles—the person will be unable to roll the tongue.

It is much easier to refer to these alleles by a letter than by a name. For example, for the gene determining tongue rolling, the allele for rolling could be represented by *R* and the allele for non-rolling by *r*. Notice that the dominant characteristic is represented by a capital letter, and the recessive characteristic by a lowercase letter. Using these letters we could represent:

- two dominant tongue rolling alleles by *RR*
- one dominant tongue rolling allele and one recessive allele by *Rr*
- two recessive non-rolling alleles by *rr*.

Each parent contributes one copy of each gene to the fertilised egg, the zygote. Each copy of the gene will have alternatives: in the case of tongue rolling, two alternatives—the dominant allele for rolling or the recessive allele for non-rolling. The combination of these two alleles in the zygote is called the genotype.

The **genotype** is the genetic make-up of an individual. It is determined by the alleles for the characteristic that is being considered. In the case of tongue rolling we have three genotypes: *RR*, *Rr* and *rr*. In two of these three gene combinations, or genotypes, the alleles are the same (*RR* and *rr*). When the alleles are the same they are said to be **homozygous**. When they are different, *Rr*, they are said to be **heterozygous**.

A person who has at least one dominant allele for tongue rolling will be a roller. Two recessive alleles have to be inherited for a person to be a non-roller. What the person actually looks like, or what they can do, is called the **phenotype**. There are two phenotypes: ability to roll the tongue (roller) and inability to roll the tongue (non-roller).

What we have discussed so far is summarised in Table 19.1.

**Table 19.1** A summary of some of the terms and symbols used in describing inheritance

| Possible combinations of the alleles for tongue rolling | Genotype | | Phenotype |
|---|---|---|---|
| | In words | In symbols | |
| Two dominant | Homozygous dominant | *RR* | Roller |
| One dominant and one recessive | Heterozygous | *Rr* | Roller |
| Two recessive | Homozygous recessive | *rr* | Non-roller |

Cystic fibrosis (see Chapter 17) is a disease that is determined by a recessive allele. Two recessive alleles need to be present for a person to develop the disease. If a person has only one allele for the condition, that allele is masked by the dominant normal allele. To examine this situation, we will use *N* to represent the dominant normal allele and *n* to represent the recessive allele for cystic fibrosis. Therefore we have three possible genotypes:

- *NN*: two dominant normal alleles; homozygous dominant
- *Nn*: one dominant normal allele and one recessive allele for cystic fibrosis; heterozygous
- *nn*: two recessive alleles for cystic fibrosis; homozygous recessive.

Two recessive alleles need to be inherited for a person to have cystic fibrosis. A person who inherits the *NN* genotype will be normal. If a person inherits the *Nn* genotype they will not have the disease but will be a **carrier** of the allele. Carriers are able to pass the affected allele on to their children.

Using these terms we can look at another example. Dimples in the cheeks are due to a single gene, with dimples the dominant characteristic and a lack of dimples the recessive one. If we use the letter *D* to represent the dominant allele for dimples, then *d* will represent the recessive allele for no dimples. Again we will have three different genotypes:

- *DD*: homozygous dominant for dimples
- *Dd*: heterozygous
- *dd*: homozygous recessive for no dimples.

Because dimples are dominant, the genotypes *DD* and *Dd* will both have the same phenotype—both will result in the person having dimples in their cheeks. The genotype *dd* will have a different phenotype. A person with this combination of alleles will not have any dimples.

**Table 19.2** Terms relating to inheritance

| Term | Meaning |
|---|---|
| Gene | The factor that determines an inherited characteristic; located in the chromosomes |
| Allele | An alternative form of a gene (e.g. the gene for dimpled cheeks has two alleles, dimples and lack of dimples) |
| Dominant | An allele that masks the effect of another allele (e.g. a person with alleles for dimples and no dimples will have dimples in their cheeks because dimples is dominant to no dimples) |
| Recessive | An allele that is masked by the presence of an alternative allele (e.g. the allele for no dimples is masked by the allele for dimples in the cheeks) |
| Homozygous | The situation where an individual has the same alleles for a particular characteristic (e.g. a person with two alleles for dimples in the cheeks is homozygous) |
| Heterozygous | The situation where an individual possesses different alleles for a particular characteristic (e.g. a person with one allele for dimples and one allele for no dimples is heterozygous) |
| Phenotype | The actual characteristics of an individual; it is determined by the alleles the person has for that characteristic (e.g. a person with one allele for dimples and one for no dimples will have the phenotype dimples in the cheeks) |
| Genotype | The genetic make-up of an individual; the combination of alleles that a person has for a characteristic (e.g. a person with one allele for dimples and one for no dimples will have the heterozygous genotype) |

## Characteristics that are determined by genes alone

The human characteristics that we have considered so far are all determined by genes alone. You can either roll your tongue or you cannot; you either have attached ear lobes or you do not. These characteristics are determined by the genes you received from your parents. You cannot change these inherited characteristics. Examples of such characteristics are listed in Table 18.3 on page 220.

As we have seen, a number of diseases are determined genetically. Huntington disease and achondroplasia both occur when a dominant allele is passed from one of the parents to the zygote. Because it is a dominant allele the child will show that characteristic. On the other hand, cystic fibrosis and thalassaemia are caused by recessive alleles. A person must inherit two alleles to have the disease. Both parents

**Figure 19.2** A woman with albinism

need to be carriers for the disease and both must pass on their recessive allele to the zygote. The baby will then be homozygous recessive and will have the disease.

One obvious human characteristic is skin colour. Skin colour depends mainly on the amount of **melanin**, a yellow-black pigment that is produced by special cells in the skin. There are a number of genes that control the amount of melanin produced in the skin cells. This is why there is such a wide range of different skin colours. However the *ability* to produce melanin is controlled by just one gene with two alleles. The dominant allele controls the normal production of melanin, while the recessive allele does not. A very small number of people are unable to produce pigment in the skin or any other tissues. These individuals have **albinism** and they have white skin, white hair and pink eyes (due to the reflection from blood vessels in the eye). People with albinism are homozygous for the recessive allele and therefore do not produce any pigment. Most people are able to produce melanin because they are either heterozygous or homozygous dominant.

## Characteristics that are determined only by the environment

There are some characteristics that are determined by the environment alone. One example is the effect of iodine deficiency in the diet of growing children. In some parts of the world where people consume mostly one type of food, like rice, there can be a deficiency of iodine. Seafood contains a lot of iodine and such places are often remote from the sea.

Iodine is very important for the normal functioning of the thyroid gland. The thyroid gland is located in the neck just below the larynx (voice box). It consists of two parts that lie on either side of the windpipe (see Fig. 19.3). The thyroid gland produces a hormone that is important for the control of a number of the chemical reactions that occur in body cells.

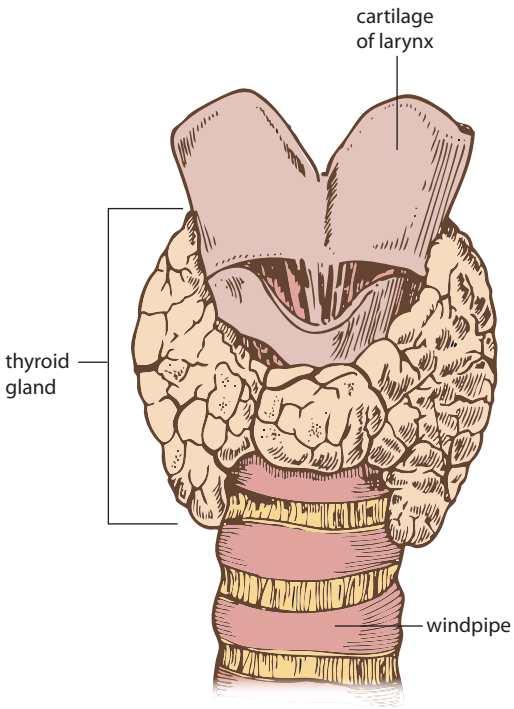


**Figure 19.3** Location of the thyroid gland

The hormone contains iodine so in people who do not have enough iodine in the diet, the thyroid gland cannot make enough hormone for the cells to work normally. This affects children as they are developing. The most severe result of iodine deficiency is **cretinism**. Children with cretinism may suffer from poor mental development, deafness, inability to speak and stunted physical growth. Affected children are often not able to stand or walk normally.

Adults who suffer from a lack of iodine develop goitre. **Goitre** is when the thyroid gland grows larger as it tries to make enough hormone to enable the cells to work normally (see Fig. 19.4).

In areas where iodine is lacking in food, it is frequently added to table salt or other foods such as flour, water and milk. It has been estimated that approximately two billion people worldwide are affected by iodine deficiency. Addition to table salt is the most economical way to ensure that people in the poorer parts of Asia and Africa get enough of this important nutrient. Iodised table salt is readily available in supermarkets in Australia (see Fig. 19.5).

**Figure 19.4** A person with goitre

Infectious diseases are also an environmental influence that can affect a person's characteristics. If micro-organisms infect the membranes that cover the brain and spinal cord, the membranes can become swollen. These membranes are called the meninges (see Fig. 8.5 on page 92) and swelling of the meninges is called **meningitis**. Meningitis can be serious due to the closeness of the swelling to the brain and spinal cord. Without immediate medical attention, serious damage to the brain, or even death, may occur.

If babies in their first month of life get the disease they are at great risk of becoming deaf, losing future intellectual ability, and having learning disorders that will affect their performance at school. Even older children can have brain damage following meningitis. For the elderly, meningitis usually results in other medical problems that may result in coma and death.

In Australia, **amoebic meningitis** sometimes occurs in summer. The infecting organism is a single-celled animal called an amoeba, which may reproduce in the warm water of swimming pools, lakes and ponds during the hotter months. It gains entry to the body when water is sniffed up the nose while swimming or playing in infected water. Symptoms of meningitis include severe headache, fever and stiffness of the neck.


**Figure 19.5** Iodised table salt

Another infection that can have serious consequences is rheumatic fever. **Rheumatic fever** is a disease that affects the heart, joints, skin and central nervous system. It mostly occurs in children between the ages of 5 and 15 years and develops following an infection by bacteria called streptococci.

Rheumatic fever in Australia is not as common as it was before antibiotics became readily available. The symptoms of rheumatic fever usually occur about two weeks after an ear, nose or throat infection. They include fever, widespread joint pain and swelling, particularly of the arms and legs. Heart failure may develop, causing a child to feel tired and short of breath, with nausea, vomiting, stomach ache or a hacking cough. Most people with rheumatic fever recover, but the heart can be permanently damaged in some cases.

Complications from rheumatic fever that may occur are damage to the heart muscle and heart valves. The valve between the left atrium and ventricle (see Fig. 6.4 on page 68) is often damaged. The damaged valve may allow blood to leak back into the atrium from the ventricle. Later in life this may result in heart failure.

Disease and poor nutrition are just two of the environmental factors that may cause a change in a person's characteristics. Some others include:

- immunity to an infectious disease through immunisation or through having had the disease
- increased numbers of red blood cells through living at high altitude
- changes in the lungs due to smoking
- permanent injuries due to accidents
- changes in behaviour caused by drug use.

## The interaction of genes and the environment

Many inherited characteristics are modified by the environment. In the previous chapter, we discussed how characteristics controlled by the inherited genes for body weight and intellectual development could be changed by the environment. Another obvious characteristic that is modified by the environment, especially for those people of European descent living in Australia, is skin colour. In summer, most light-skinned people whose skin is overexposed to sunlight develop darker skin. During the winter months, the skin lightens again as the sunlight is less intense.

Exercise and other training programs can modify many of our physical characteristics. The genes that determine basic body build and size of muscles are inherited. These inherited characteristics can be altered using exercise and training programs. People involved in the highest levels of sport train regularly to build up particular muscle groups. Swimmers at national and Olympic level frequently have very broad shoulders (see Fig. 19.5) and an enlarged lung capacity from their specialised training programs. On the other hand, body builders train to build up all their muscle groups (see Fig. 19.6). Most people involved in intensive training programs are also very careful about their diet. The food we take in will also influence many of our inherited characteristics.

Chapter 18 explained how certain types of cancer are more common in some families than in others. Some people inherit a predisposition to a form of cancer. Factors such as exposure to certain chemicals, or to sunlight, affect whether a person will develop a particular cancer. Diet now appears to be a factor as well but in the opposite way. There is growing evidence that certain foods may reduce cancer risk even in people with a predisposition to cancer.

The environment can also play a dramatic role in the effects of certain inherited diseases. Phenylketonuria (PKU) is a genetic disease that prevents the body breaking

**Figure 19.6** Swimmers train to develop shoulder muscles and to increase lung capacity: notice how broad this swimmer's shoulders are

**Figure 19.7** Body builders train to develop all muscle groups

down a particular amino acid. That amino acid then builds up in the blood causing severe symptoms of progressive mental disability and fits. It is possible to prevent the gene from having this effect if a baby with PKU is kept on a strict diet that avoids the amino acid. Later, when the development of the child's nervous system is complete, the amino acid in the diet will not have such a serious effect. In the treatment of PKU an environmental factor, the special diet, has prevented the gene from causing a serious disability.

## Variation in tolerance limits

### *Diet*

The genes that we inherit determine not only the physical characteristics that we can see, but also our body chemistry. The chemical reactions in our cells determine how the various parts of the body operate. We have just seen that PKU is an inherited condition that affects the way a particular amino acid is broken down. Earlier in the chapter, the effect of a lack of iodine in the diet was discussed. These are just two instances of how diet may affect body functioning.

Many of the foods in our diet are essential but if we consume too much or too little of a particular nutrient things may go wrong. In other words there are tolerance limits for some nutrients. **Malnutrition** is poor health due to eating a diet that is unbalanced. It may be due to either over nutrition, too much of a certain food, or under nutrition, too little of a food. Over nutrition is when the intake of food is greater than what is needed. If too much energy is consumed it results in a person becoming overweight and obese. This in turn may lead to high blood pressure, heart disease and diabetes.

Under eating can suppress the development of the brain and other body functions. **Kwashiorkor** is a condition that usually affects children aged 1–4 years, although it also occurs in older children and adults. It is caused by insufficient protein in the diet and results in the arms and legs becoming extremely thin due to a lack of muscle development. In addition, the stomach swells and there is reduced immunity to disease. Most children who are left untreated have permanent mental and physical disabilities. In severe cases, the child may die.

In Chapter 21, deficiencies of vitamins and minerals are discussed. For some vitamins and minerals, too much can be a problem. Too little vitamin A can result in blindness and an increase in digestive and respiratory infections. On the other hand, large amounts of vitamin A are highly poisonous. Too much vitamin A can cause headaches, pain and weakness of the bones, and an enlarged liver. Excessive intake of iron can cause vomiting and diarrhoea, and if it is continual may result in damge to the arteries of the heart.

People's tolerance limits to nutrients vary. Some people suffer from **lactose intolerance**. Lactose is a sugar found in milk and other dairy products. It is broken down to simple sugars in the small intestine but some people lack the enzyme needed for the breakdown. In people who are intolerant to lactose, consuming milk or milk products can cause stomach cramps, diarrhoea and other symptoms.

In a small number of people lactose intolerance is due to the inheritance of two recessive alleles, one from the mother and one from the father. Many other people are able to break down lactose when they are young but become lactose intolerant as they

get older. This also seems to be inherited and it is much more common in some ethnic groups than others. People of European origin rarely develop lactose intolerance but it is common among Asians, Africans and Australian Aboriginal people.

## Alcohol

Alcohol is not essential to body functioning so a lack of alcohol has no adverse affects. On the other hand, too much alcohol in the diet can have undesirable effects on body functioning. Individuals inherit a particular tolerance to alcohol that will vary from person to person. Like most drugs, the more alcohol that is consumed, the greater the tolerance becomes. However, there are some people who find it difficult to tolerate alcohol at all. These people's bodies lack the chemicals required to break alcohol down into carbon dioxide and water.

The amount of the chemicals in the body for alcohol breakdown also appears to vary. The more effective the chemical breakdown, the higher the tolerance to alcohol. Conversely, people who break down the alcohol at a slower than normal rate tend to have a lower tolerance to alcohol. These individuals become affected by alcohol much more quickly. This is evident in some ethnic groups who lack the chemicals for alcohol breakdown. Such a genetic predisposition probably evolved over thousands of years because these groups did not have alcoholic drinks in their particular culture.

Another factor in the ability to tolerate alcohol without adverse behavioural effects is body build. Alcohol dissolves in water. People who have more body fat have a smaller proportion of water in their bodies. This means that people with more body fat will be more affected by alcohol than people with less fat. This is the reason why women are less able to tolerate alcohol than men. The proportion of body fat varies a lot but on average women have 25% fat and men 15%.

## REVIEW QUESTIONS

1. Using examples, explain the difference between:
   **(a)** an allele and a gene
   **(b)** genotype and phenotype
   **(c)** heterozygous and homozygous.
2. Give examples of dominant human characteristics and for each one state the recessive characteristic.
3. Some people may be carriers for a particular condition. Explain what is meant by the term 'carrier'.
4. List three characteristics that are thought to be determined by genes alone.
5. What is albinism? Explain how a person with two parents who have normal pigment production could have albinism.
6. Describe the effects that iodine deficiency may have on a growing child.
7. Some infectious diseases can have a major influence on a person's phenotype. Give an example of one of these diseases and explain the effect it may have on a person.
8. Describe how exercise and other training programs can modify the effect of the genes a person has inherited.
9. What is malnutrition? Describe what can occur if a person:
   **(a)** consumes too much food
   **(b)** does not consume enough food.

## APPLY YOUR KNOWLEDGE

1. **(a)** For a gene with two alleles there are three possible combinations of alleles in the fertilised egg. Explain how there could be any one of three combinations of alleles in the zygote with just two alleles for a gene.
   **(b)** How many combinations of alleles would be possible in the zygote if a gene had three alleles?
2. If we assume that free ear lobes (*F*) are dominant over attached earlobes (*f*), list the genotypes for the following allele combinations:
   **(a)** homozygous dominant
   **(b)** heterozygous
   **(c)** homozygous recessive.
3. In what situations would you be able to deduce a person's genotype by looking at their phenotype? Give an example to illustrate your answer.
4. In many families, a Roman-shaped nose is dominant to a straight nose. If a man from a family who have had Roman noses for many generations married a woman from a family with straight noses for many generations, what would their children probably look like? What would be their probable genotype?
5. Figure 19.2 shows a person with albinism. Her parents had normal skin and eye colouring. With respect to the genes for melanin production, what would be the genotypes of that person's father and mother?
6. Besides iodine, there are other minerals that are important in the diet. Find out what may happen if a person is in an environment where there is a lack of calcium in their food.
7. As a class or school project, approach one of the organisations in your community concerned with a particular genetic disorder. They may be willing to provide a speaker to inform you about the latest information available on their particular disease and its treatment.