

# Chapter 19

## Characteristics of offspring

## Unit 2B



### Unit content

#### Inheritance

Genetic counselling uses information from pedigrees, genetic testing to provide an analysis of the risk associated with some of these mutations.

Pedigrees:

- construction and interpretation of pedigrees for autosomal and sex-linked conditions
- probabilities of producing affected offspring for autosomal and sex linked inheritance.

Genetic testing of parents and offspring for:

- gene and chromosomal abnormalities.

Human Genome Project:

- information provided by the Human Genome Project
- range of possible uses for this information.

#### Approaches to investigating and communicating human biology

Investigate, for example:

- probabilities of genetic inheritance.

**Figure 19.1** Five generations of one family. Family characteristics are passed from one generation to the next through inheritance

The discussion of inheritance in Chapter 18 was based on simple Mendelian genetics. In humans, the development of all body organs is regulated by a large number of genes. In addition, there is wide variation in the age at which a particular genetically controlled characteristic appears. Many of our characteristics develop well before we are born, but others, such as hair and eye colour, may not appear until shortly after birth. Still others, such as growth of body hair, become evident during childhood and some characteristics, such as breast development, are not expressed until maturity. In all these cases more than one gene may have an influence on the development of the trait, so that for human characteristics it is often difficult to give clear indications of dominance and recessiveness.

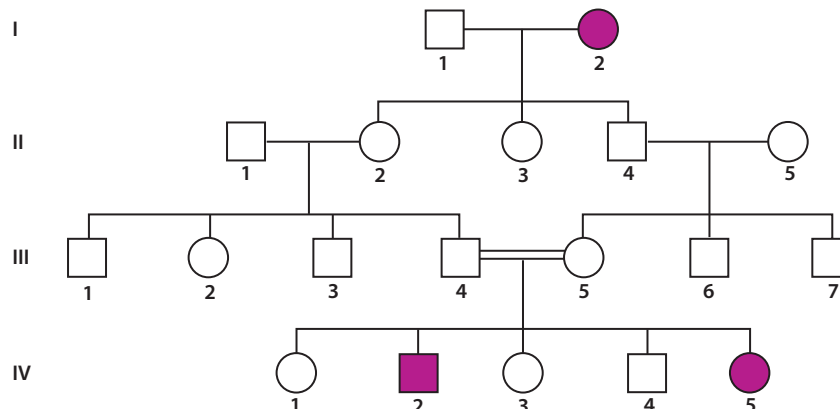
## Constructing pedigrees

One of the easiest ways of observing the pattern of inheritance within a family is by the construction of a **pedigree** or family tree. A number of conventional symbols are used in the construction of such trees. Males are represented by squares ( $\square$ ) and females by circles ( $\circ$ ). Those with the particular characteristic under study are shaded ( $\blacksquare$  or  $\bullet$ ). Marriage is represented by a horizontal line joining the symbols ( $\square$ — $\circ$ ). A marriage between two close relatives, usually cousins, called a **consanguineous** marriage, is shown by double horizontal lines ( $\square$ == $\circ$ ). A vertical line extending down from the horizontal one connects to the children produced by that marriage. If there are a number of children from a particular marriage, each is connected by a vertical line to a horizontal one, which in turn links up with the line joining the two parents. This sounds complex when described in words, but when you look at an actual pedigree (Fig. 19.2) you will see that it is really quite simple.

One human characteristic that is obvious to any observer is skin colour. This trait depends mainly on the amount of **melanin**, a yellow-black pigment produced by special skin cells called **melanocytes**. The production of melanin by the melanocytes is dependent on particular enzymes. In a very small number of cases, humans fail to synthesise one of the enzymes and pigmentation does not occur. These individuals are called **albinos** and they have white skin, white hair and pink eyes (due to the reflection from blood vessels in the eye). Two alleles are involved in pigmentation. The dominant allele controls normal enzyme production and thus the presence of melanin. The recessive allele causes abnormal enzyme production. Genetically, albinos are homozygous for the recessive allele and therefore do not produce any pigment.

Production of melanin is a good example of how simple dominance operates in humans. Genes controlling the normal production of melanin are dominant to the defective gene responsible for albinism. Figure 19.2 illustrates four generations of a family in which one of the original parents (the shaded circle) was an albino. Albinism

**Figure 19.2** A pedigree with one original parent an albino



did not occur again in this family until the cousins in the third generation married and produced children. Notice that the generation numbers are shown by Roman numerals on the left-hand side, and individuals in each generation are numbered from left to right using normal numbers.

We can use this pedigree to work out the genotypes, with respect to skin pigmentation, of all the individuals in the family. We will represent the dominant allele for normal pigmentation by  $A$  and the recessive allele for albinism by  $a$ .

- As the characteristic has skipped a number of generations it *must* be recessive ( $aa$ ).
- As the cousins in the third generation produced two children with albinism, a recessive trait, the cousins must each have been heterozygous ( $Aa$ ).
- It follows, therefore, that at least *one* of each of the cousin's parents (in generation II) must have been heterozygous as well, otherwise the cousins could not have inherited the recessive allele for albinism.
- Since the female in generation I is an albino, we know that the parents of the cousins, who are brother and sister, must be heterozygous.

Figure 19.3 shows what we have worked out so far.

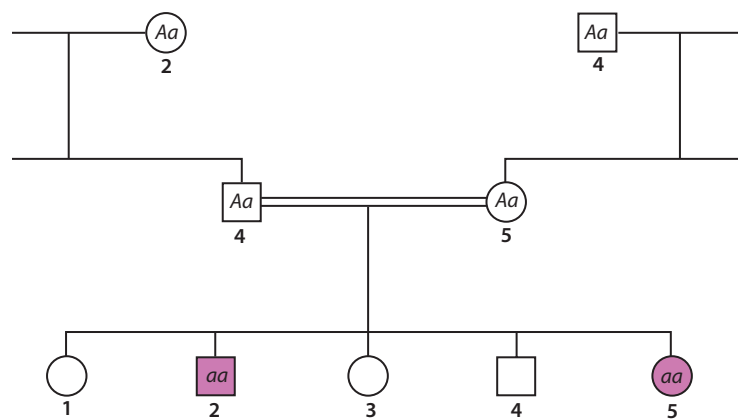
As each individual with normal pigmentation must have at least one dominant characteristic, we can now complete the pedigree above to the stage shown in Figure 19.4.

Without further information it is difficult to determine the genotypes of some of the individuals. We do not know if those marked  $A?$  are  $AA$  or  $Aa$ . More than likely, those marked as  $A?$  in the first and second generations were  $AA$ , otherwise albinos could have been expected in the third generation. However, we cannot be sure without further information.

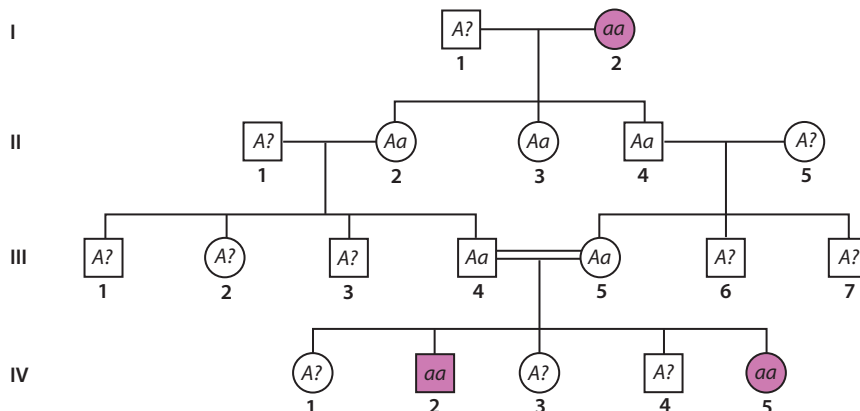
Another human characteristic that can be traced through a family is whether the earlobes are attached or free (Fig. 19.5). Generally speaking, those with free earlobes carry a dominant allele  $F$ , and such individuals may be of two genotypes:  $FF$  or  $Ff$ . Attached earlobes are a recessive trait and, therefore, have the genotype  $ff$ . The pedigree in Figure 19.6 shows three generations of a family. Those with free earlobes are shown as open symbols; those with attached earlobes are shaded.

Once again, all the individuals shown shaded must be homozygous recessive ( $ff$ ), whereas the

**Figure 19.3** Genotypes of a family with albino alleles

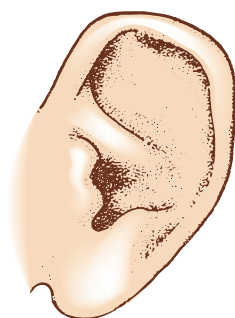


**Figure 19.4** Complete pedigree of a family with albino alleles



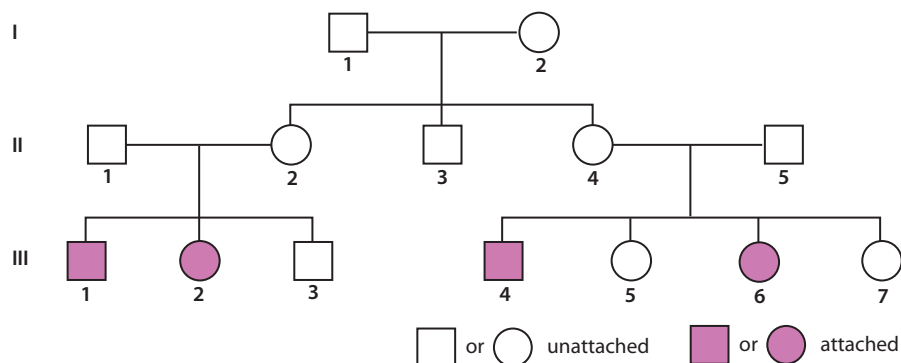


(a)

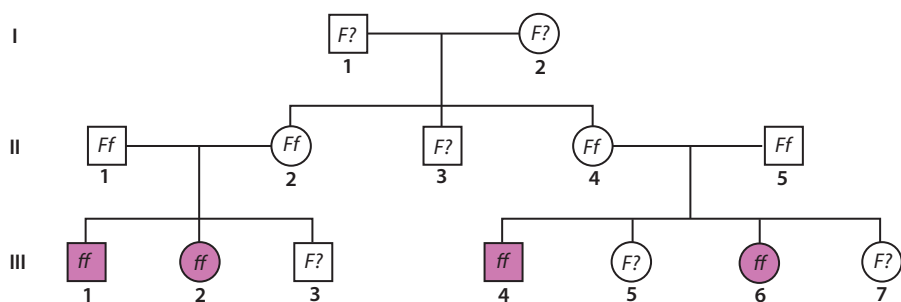


(b)

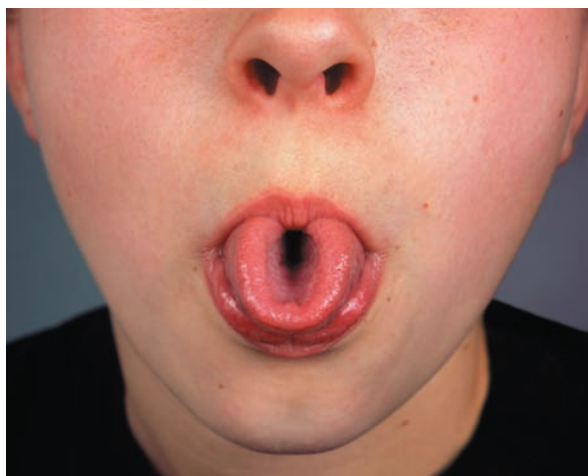
**Figure 19.5** (a) Attached and (b) free earlobes



**Figure 19.6** Pedigree for a family with attached earlobes appearing in the third generation



**Figure 19.7** Genotypes for a family with attached earlobes appearing in the third generation



**Figure 19.8** A tongue roller

parents of the second generation must all be heterozygous ( $Ff$ ). Apart from knowing that all other individuals have at least one  $F$  allele, it is difficult to determine their exact genotypes. At least one of the original parents must have been heterozygous to produce children who were heterozygous. Figure 19.7 summarises what we are able to determine from the information we have.

People are able either to roll their tongues or not (Fig. 19.8) and this is an inherited characteristic. If we consider non-rolling as a recessive characteristic ( $r$ ), then the pedigree in Figure 19.9, with two individuals in the first generation who cannot roll their tongues, illustrates how a recessive trait can be a frequently observed characteristic in a particular family.

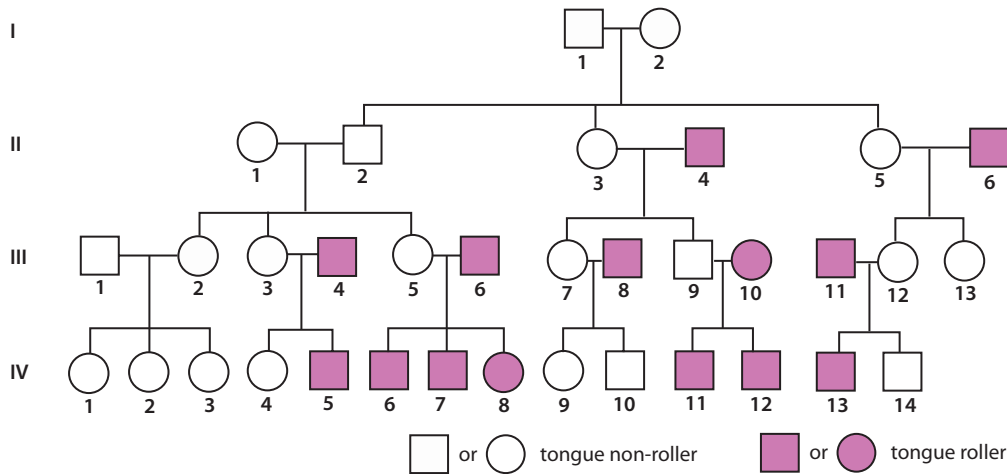
The inability to roll the tongue for this family, although a recessive trait, occurs frequently. (It is a common misconception that recessive characteristics will appear less frequently in a family or a population.) All the individuals shown unshaded

have the recessive characteristic and thus have the homozygous recessive genotype ( $rr$ ). All other individuals possess at least one  $R$  allele. Using this knowledge, can you work out the genotypes of all persons shown in the pedigree? Try it before referring to Figure 19.10, which shows the probable genotypes.

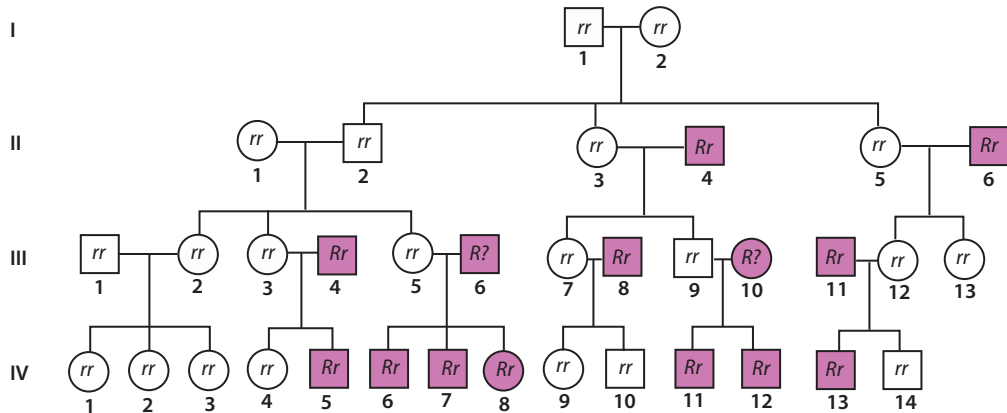
As you can see, there are only two individuals for whom we are not certain whether they are homozygous or heterozygous for tongue rolling.

In Chapter 18 X-linked characteristics were discussed. You will recall that these were characteristics that were controlled by genes on the X chromosome. Genes carried on the X chromosome have a distinctive pattern of inheritance. As males have only one





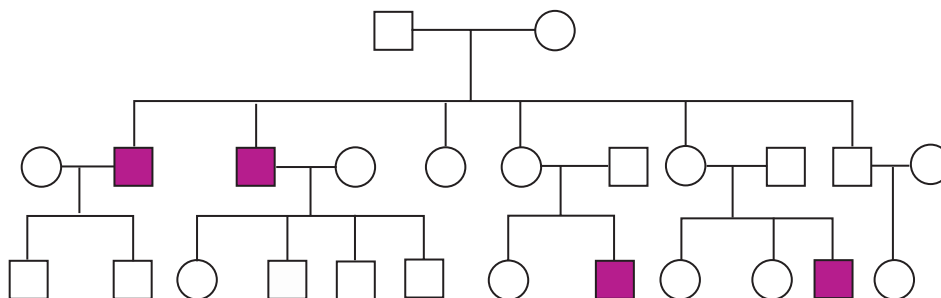
**Figure 19.9** Pedigree for a family showing tongue rollers and non-rollers



**Figure 19.10** Genotypes of a family showing tongue rollers and non-rollers

copy of the X chromosome (they are *hemizygous*) recessive characteristics are expressed in the phenotype of males. A typical pedigree (see Fig. 19.11) will show clusters of affected males connected through unaffected carrier females. There will be *no* cases of direct male to male transmission because males transmit their X chromosomes to their daughters and not to their sons. Each son of a carrier mother will have a 50% chance of being affected. A predominance of male members with the condition does *not* necessarily mean it is X-linked. You need to examine the pedigree carefully to ensure that the only explanation for the pattern of inheritance displayed is that it is X linked.

Table 19.1 outlines how to determine the patterns of inheritance in pedigrees.



**Figure 19.11** A typical pedigree for a family showing a characteristic linked to the X chromosome

**Table 19.1** Determining patterns of inheritance in pedigrees

- Every person who shows a **dominant** characteristic need only have *one* allele for that characteristic, and every person possessing such an allele *must* show the characteristic.
- A person with a **dominant characteristic** *must* have at least one parent with the characteristic. It *cannot* skip a generation.
- Two people *with* a **dominant characteristic** can have a child *without* that characteristic.
- Two people who do not possess a **dominant characteristic** *cannot* have a child with such a characteristic.
- Every person who shows a **recessive characteristic** *must* have two alleles for that characteristic.
- A person with a **recessive characteristic** *does not* have to have a parent with the characteristic. It *can* skip a generation.
- Two people *without* a **recessive characteristic** can have a child *with* the characteristic.
- Two people *with* a **recessive characteristic** *cannot* have a child *without* the characteristic.
- A characteristic linked to the **X chromosome** *cannot* be passed from a father to a son. Fathers can only pass such a characteristic to their daughters.
- Two people *without* an X-linked characteristic can have a *son with* the characteristic but *not a daughter*.

## The probability of inheriting a particular condition

Probability is the chance that something will happen. It is usually expressed as a fraction ( $\frac{1}{8}$ ,  $\frac{1}{4}$ ,  $\frac{1}{2}$ ), a decimal (0.13, 0.25, 0.50) or a percentage (13%, 25%, 50%). When a probability is zero, it means that there is no chance of an event occurring. On the other hand, a probability of one, or 100%, indicates that an event must occur.

In Chapter 18 the way the sex of a child is determined was discussed. In that discussion, it was said that girls and boys are born in approximately equal numbers. Expressed as a probability we would say that the chance of a couple having a female child (or male, for that matter) is a half. This could be expressed as a fraction ( $\frac{1}{2}$ ), a decimal (0.5) or a percentage (50%).

In the family of tongue rollers shown in Figure 19.10, the parents in the first generation could only produce offspring with the same genotype. All their children were tongue non-rollers. Therefore, the probability of these parents producing a non-roller was one, and of producing a roller was zero. However, if we examine the offspring of their daughters, both of whom married tongue rollers, the probabilities are quite different. Their children have a 50% chance (or  $\frac{1}{2}$ , or 0.5) of being a tongue roller and a 50% chance (or  $\frac{1}{2}$ , or 0.5) of being a non-roller. (You may wish to construct a Punnett square to check this for yourself.)

If both parents were heterozygous for tongue rolling we could show the probabilities in the following way:

Parents' genotypes:	$Rr \times Rr$
Gamete genotypes and probabilities:	$(\frac{1}{2} R + \frac{1}{2} r) \times (\frac{1}{2} R + \frac{1}{2} r)$
Offspring genotypes and probabilities:	$\frac{1}{4} RR + \frac{1}{4} Rr + \frac{1}{4} rR + \frac{1}{4} rr$
	Or $\frac{1}{4} RR + \frac{1}{2} Rr + \frac{1}{4} rr$
Offspring phenotypes and probabilities:	$\frac{3}{4}$ rollers + $\frac{1}{4}$ non-rollers

(You can also check this out with a Punnett square.)

## Making predictions from a pedigree

One of the most frequent reasons for constructing a pedigree for a family is to investigate the pattern of inheritance of a genetic disorder. **Single-gene disorders** are disorders caused by the inheritance of a single defective gene, or disorders that result from the mutation of the structure of a single gene. The pattern of inheritance of single-gene disorders follows the basic laws of heredity already described. However, the severity of the disorder is often variable and difficult to predict. Over 4000 different disorders of this type have been identified in humans. Most of them are dominant traits, some are recessive, and a lesser number are linked to one of the sex chromosomes.

You can use the criteria listed in Table 19.1 to make predictions about the chances of inheriting a single gene disorder.

### Dominant inheritance

Dominant alleles that cause severe defects in people are rarely passed on because people with such alleles frequently die before they have the opportunity to reproduce. One exception is the condition called **Huntington disease**, formerly Huntington's chorea. The symptoms of this hereditary disorder seldom appear before 40 years of age. It is characterised by occasional involuntary flailing movements of the arms and legs. In addition, the person often has difficulty making voluntary movements of the limbs. Other symptoms include writhing movements of the hands, head, trunk and feet, and a progressive loss of the ability to think clearly, referred to as **dementia**.

As Huntington disease is controlled by a dominant allele, the condition is very likely to be passed on from one generation to the next. Since the condition does not become apparent until later in life, the children of a person with the condition may not be aware that they themselves may have inherited the disorder until after they have had children of their own.

Genetic screening is now available for those with a parent who either has Huntington disease or died from it. Blood from participants and their close relatives is collected and analysed for the presence of three established gene markers. These tests establish an individual's risk of developing the condition. Since the condition is incurable, nothing can be done to help people diagnosed as having the disease, but those people can then make decisions about whether or not to have children.

Some other genetic disorders caused by dominant alleles on the non-sex chromosomes are:

- **Achondroplasia** is a form of dwarfism, characterised by short limbs, a prominent head, normal intelligence and a waddling gait.
- **Facioscapulohumeral muscular dystrophy** is a rare form of muscular dystrophy affecting the facial muscles (facioscapulohumeral means face, shoulder and arm). Other muscles are gradually affected, making it difficult to raise the arms above the shoulders, to lift objects or to walk normally.
- **Neurofibromatosis** is inherited as a dominant characteristic and affected individuals exhibit numerous tumours along the peripheral nerves. The tumours are composed of a dense proliferation of nervous and fibrous tissue, and cause abnormalities of the skin and flesh as well as distortions of bone structure.

### Recessive inheritance

A person who carries a recessive allele but does not show the recessive phenotype is known as a **carrier** for that characteristic. If both parents are carriers, some of their children may receive one defective allele from each parent. This results in the children being homozygous for the recessive allele, and then being affected by the condition.

When both parents carry a recessive allele for the same abnormal characteristic, each of their children has a 25% chance of inheriting the disease concerned. However, there is also a 25% chance that each child will not inherit the abnormal gene from either parent, and a 50% chance of receiving only one abnormal allele and thus being a carrier for the condition, like the parents.

If  $N$  is the normal allele and  $n$  the allele that causes an abnormality, this can be shown as illustrated in Figure 19.12:

**Figure 19.12**

<b>Parents</b>		$Nn$	$\times$	$Nn$
<b>Gametes</b>		<b>Female</b>		
		$\frac{1}{2}N$	$\frac{1}{2}n$	
<b>Male</b>	$\frac{1}{2}N$	$\frac{1}{4}NN$	$\frac{1}{4}Nn$	
	$\frac{1}{2}n$	$\frac{1}{4}Nn$	$\frac{1}{4}nn$	

**Offspring:** 25% normal + 50% carrier + 25% affected

The incidence in the population of severe recessive disorders that are not linked to a sex chromosome is low because it is unlikely that a carrier from one family will mate with another carrier of the same recessive condition. So, although a particular condition may be an inherited disorder, it could suddenly appear where there has been no previous family history of the disease. In a consanguineous marriage, because the couple are close relatives, the chance of them both being a carrier for a recessive allele is greater. Consanguineous marriages often occur for cultural or for geographical reasons in certain populations and are generally between cousins. In these cases there may be a high incidence of a particular genetic disorder, as the related parents have received some of their genes from a common ancestor and, therefore, have a greater chance of being carriers of a gene for the same abnormal condition. However, this is not often the case, and the chance of cousins marrying and having an abnormal child are little higher than if two complete strangers were to marry. Both Albert Einstein and Charles Darwin married their first cousins and for both all their children were normal.

As explained in Chapter 18, consanguineous marriages were once common among the people inhabiting countries around the Mediterranean Sea. In these people, the incidence of **thalassaemia**, a recessive disease in which anaemia results from defects in the formation of haemoglobin, is relatively high.

Many recessive disorders produce serious abnormalities. **Phenylketonuria (PKU)** is a good example of a disease of this type. The gene concerned controls the production of an enzyme called phenylalanine hydroxylase. This enzyme converts the amino acid phenylalanine to tyrosine. If this enzyme is not present, then phenylalanine will accumulate in the bloodstream and become toxic. This toxicity results in damage to the growing brain, and thus extreme mental retardation, as well as a tendency towards epileptic seizures and a failure to produce normal skin pigmentation.

Phenylalanine is classed as an essential amino acid and so must be present in the diet. Enough of this amino acid must be present so that a child may grow. For those individuals born homozygous recessive for PKU, excessive amounts of phenylalanine in the diet can be dangerous. Fortunately, the disease can be identified almost immediately after birth. A blood sample is usually taken by pricking the baby's heel (called a



heel stick) within two to three days after birth. Special diets restricting the intake of phenylalanine and replacing it with substitutes can, if begun early in the child's life, largely, if not entirely, correct the symptoms.

**Cystic fibrosis** is another disorder controlled by one recessive allele. Children with the condition suffer from chest infections, a lack of digestive enzymes and an increased salt loss. It is the most common lethal genetic disease in people of European origin, in whom up to 3% of the population are thought to be carriers. Once again, a blood sample is usually taken from the baby's heel within two to three days after birth. When a child is identified as having the disease, special diets are given with low fat, high carbohydrate and high protein. The diet is supplemented with pancreatic extract and large doses of vitamins A, D and K. This does not cure the disease but it does enable the child to function as normally as possible.

Couples who are concerned that they may be carriers for cystic fibrosis can be tested via a laboratory test done on a sample of blood or saliva. If results show that both prospective parents are carriers, genetic counselling should be considered before starting a family.

## Genetic counselling

Today most women in Australian society are aware of the risks of producing a baby with a birth defect. During pregnancy most women take special care of their health and avoid alcohol, cigarettes and other drugs to ensure the baby is as healthy as possible. However, if a couple has already produced a child with a birth defect, they are naturally very anxious about the possibility of the same thing happening in later pregnancies. In other cases, one partner may have a birth defect and the couple is concerned that the problem could occur in their children. Other couples may have close relatives who have inherited diseases or have given birth to children with such disorders. In all these cases, the people concerned may seek advice on the risk of the inherited disorder occurring in their children. The advice given in these situations is called **genetic counselling**. By examining the incidence of a disorder in the family tree, the probability that a particular problem will occur can sometimes be determined. The couple can then decide whether to risk having a baby with the inherited disorder.

Suppose a couple, with no history of genetic disorders in either family, had a child with an autosomal recessive condition such as thalassaemia. What would be the probability that their next child would inherit the same condition? A genetic counsellor would tell them that there was a one in four chance of each of their subsequent children having thalassaemia. Should they decide to have another child under these circumstances? There are now available procedures that can detect the presence of many genetic disorders in the foetus before birth (see Chapter 15). If a genetic disorder were diagnosed, the couple would then have the option of terminating the pregnancy. Although genetic counselling and modern diagnostic techniques can alleviate much suffering, the responsibility for decision making must still lie with the individuals concerned. Decisions to risk having an abnormal child or to terminate a pregnancy are not made lightly.

## The Human Genome Project

The Human Genome Project was an international research effort, launched in 1990, which grew out of the rapid advances that were being made in chromosome mapping. Scientists decided to map the genes in all 46 chromosomes in the human genome. The **genome** is the complete set of genetic information of an organism. For humans it is the complete sequence of the nucleotides that make up the 20 000–25 000 genes

You can find out more about the human genome project at <http://www.biotechnologyonline.gov.au/human/genomeproject.cfm>

For detailed information on genome mapping go to <http://www.ncbi.nlm.nih.gov/About/primer/mapping.html>

in our chromosomes. The project was completed in 2003 but analysis of the data will continue for many years. Once all the information is available scientists will be able to find and study the genes involved in human diseases much more efficiently and rapidly than ever before.

As more information becomes available from the Human Genome Project, genetic counsellors will be better able to predict the future likelihood of some diseases. For example, if the gene responsible for Huntington disease is present, it is almost certain that the symptoms will eventually appear, although the time of onset cannot be accurately predicted. Information from the genome project also helps counsellors predict which individuals have an increased susceptibility to disorders such as heart disease, cancer or diabetes, which result from complex interactions between the genes a person carries, the environment in which they live and the lifestyle they follow. In these cases there is no certainty that symptoms will occur, but the risk is greater for individuals with specific genotypes than for others in the population.



## EXTENSION

In Australia, genetic counselling has been effective in providing information to couples at risk of having a son with muscular dystrophy. In Western Australia, the incidence of the condition in the population in 1961 was 1 in 20 000. By 1976 this had been reduced to 1 in 55 000. As mutations occur spontaneously, the disease will continue to occur in the population at about this level.

- Western Australia has been a world leader for many years in muscular dystrophy research. Use reference material to determine the latest research that is taking place.
- Find out how genetic counselling has contributed to the decrease in the incidence of muscular dystrophy.



## Working scientifically

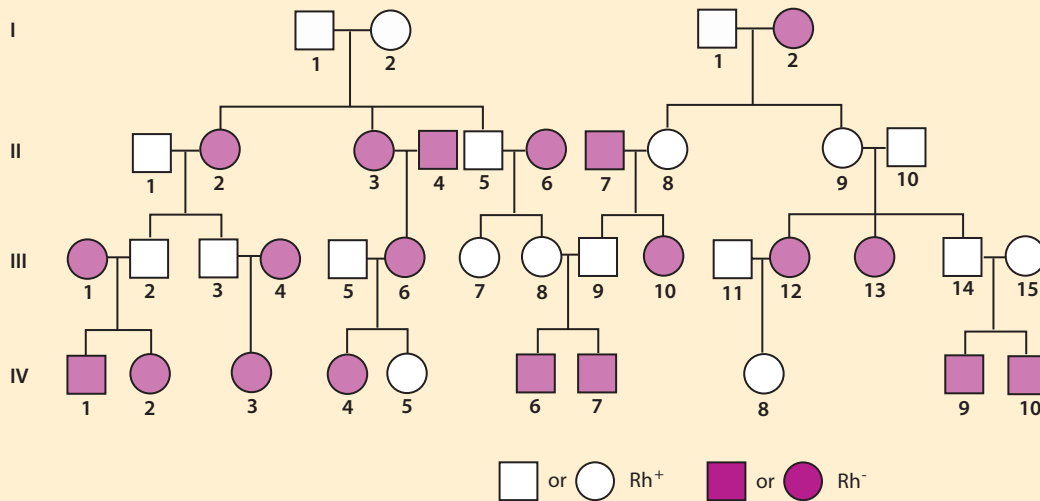
### Activity 19.1 Examining pedigrees

#### Pedigree 1

The inheritance of Rh blood groups in humans follows the laws of simple dominance. The letters 'Rh' were used because it was from experiments in 1939 with the blood of rhesus monkeys that the blood groups were first identified. There are two types: Rhesus-positive ( $Rh^+$ ) and Rhesus-negative ( $Rh^-$ ). In Australia, about 85% of the population is  $Rh^+$  and 15%  $Rh^-$ .

#### What to do

Study the pedigree in Figure 19.13. The  $Rh^+$  trait is determined by a dominant allele and persons who are  $Rh^+$  are shown by open symbols. Shaded symbols represent  $Rh^-$  people. Determine the genotypes for all the individuals shown.



**Figure 19.13** Pedigree for a family in which some members are Rh<sup>-</sup>

### Interpreting your results

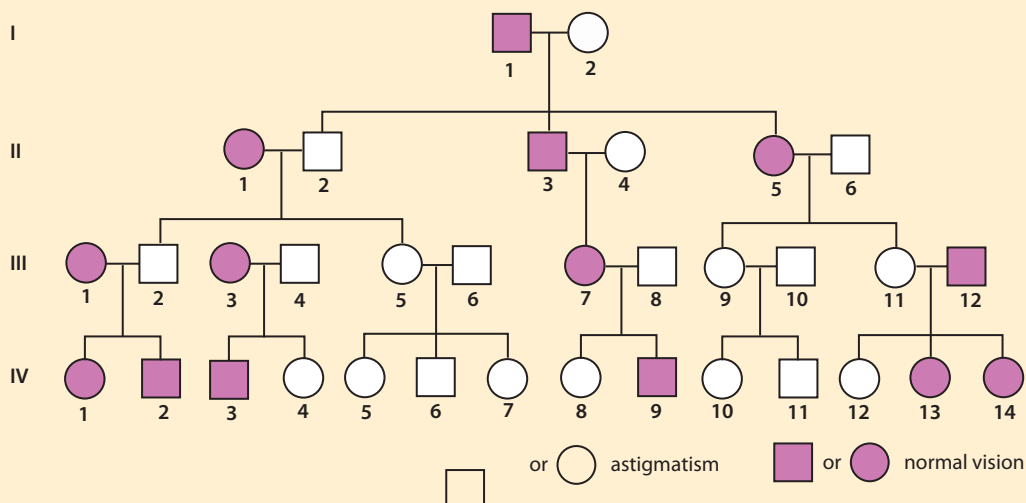
1. Can you be absolutely certain about the genotypes for all individuals in the first generation? Give reasons for your answer.
2. What are the genotypes of the male (III 5) and the female (III 6) in the third generation who married and produced two daughters? Describe the genotypes using words and using appropriate letters. Explain why you can be certain of their genotypes.
3. Do the two Rh<sup>+</sup> females in the fourth generation (IV 5 and IV 8) have the same genotypes? Give an explanation for your answer.
4. Is the Rh blood group controlled by a gene on an autosomal or an X chromosome?

### Pedree 2

In many families the visual defect astigmatism is an inherited condition. The pedigree in Figure 19.14 shows one such family.

### What to do

Carefully examine the pedigree and work out the genotypes for all individuals shown. Is there any member of the family about whose genotype you are uncertain?



**Figure 19.14** Pedigree for a family in which some members have astigmatism

### Interpreting your results

Is astigmatism in the family illustrated dominant or recessive? Give reasons for your answer.

### Activity 19.2 A family with Huntington disease

In this chapter Huntington disease was described as an inherited disorder that results in lack of control over muscles and progressive mental deterioration to the point where sufferers are unable to look after themselves. The symptoms rarely appear before 40 years of age and by that time individuals with the disorder may have passed the gene for the condition to their children. Huntington disease is transmitted by a dominant gene.

The paragraph below describes a family in which Huntington disease has occurred.

Jennifer is 45 years of age and has just developed the symptoms of Huntington disease. Her father, James, is 70 years old and is hospitalised with the disorder, but her mother, Anne, two years younger than her father, does not have the condition. Jennifer's husband, John, also 45 years of age, does not have Huntington disease, and there is no history of the condition in his family. Jennifer's older brother, Malcolm, does not have the disease.

Jennifer and John have two children, Andrew (25 years old) and Michele (21 years old). Michele is married to Tony, who is the same age as her brother, and she has just given birth to their first child called Darren. There is no history of Huntington disease in Tony's family.

### What to do

Construct a pedigree to show all the individuals in the family. Indicate the individuals who have Huntington disease by shading the relevant circles or squares.

### Interpreting the family tree

1. Write down the possible genotypes of James, Anne, Jennifer and John. Explain the symbols you are using.
2. What is the probability that Michele has inherited Huntington disease? Using a Punnett square, set out the cross between Michele's parents in full.
3. Is there any possibility that Darren has inherited the disease? Explain, using a Punnett square to set out the cross between his parents in full.
4. Is Huntington disease controlled by a gene on an autosomal or an X chromosome?



### REVIEW QUESTIONS

1. List five rules that must be observed when constructing a pedigree.
2. (a) What is probability?  
(b) If two people, each with the recessive allele for albinism, have children, what is the probability of them having an albino child?
3. Describe the pattern of inheritance of the following disorders:  
(a) cystic fibrosis  
(b) colour blindness



- (c) achondroplasia
- (d) phenylketonuria.
- 4. People with Huntington disease often have children even though their children will have a 50% chance of inheriting the disease. With such a high probability of passing the disease on, why do such people continue to produce children?
- 5. Briefly outline the symptoms caused by the allele for phenylketonuria, how it can be identified in newborn infants, and the treatment that is given.
- 6. Explain why a father with an X linked condition is not able to pass the characteristic to his sons.
- 7. Why do couples who are first cousins have a slightly higher risk of having a child with an inherited disorder compared with unrelated couples?
- 8. Discuss what is meant by genetic counselling and how it may assist people in deciding whether to have a child or to continue with a pregnancy.
- 9. Why was the Human Genome Project set up? What are the anticipated outcomes for this project?

### APPLY YOUR KNOWLEDGE

1. When constructing a pedigree, suggest how you should show the parentage of a child born as a result of artificial insemination by a donor (AID).
2. Using the pedigree for the royal families of Europe illustrated in Figure 21.7, what was the probability that Queen Victoria would produce a daughter who was a carrier for haemophilia?
3. The first child born to a married couple with normal vision is a male with red-green colour blindness. Calculate the probability that their second child will also be colourblind. Give a clear explanation for your answer. Remember, red-green colour blindness is X linked.
4. In the United States about 6 in every 100 children whose parents are first cousins die before the age of 10 years. Where the parents are unrelated, the figure is about 2.5 in every 100. Can you suggest reasons for this big difference in mortality for the first 10 years of life?
5. Using the Internet, research the current progress being made with the data from the Human Genome Project.
6. (a) Chloe is  $Rh^+$  but her brother Jason is  $Rh^-$ . Both of Chloe's parents are  $Rh^+$ . What is the probability that Chloe is a carrier for the recessive Rh allele?
- (b) Chloe married Mitchell and they had a daughter, Emma. What is the probability that Emma is  $Rh^-$  if:
  - (i) Chloe is a carrier for the recessive allele?
  - (ii) Chloe is not a carrier for the recessive allele?

