Chapter 18
Principles of inheritance

Unit
2A

Unit content

Inheritance
Principles of Mendelian genetics are used to predict variations in offspring.

Inheritance:

- dominant, recessive, co-dominant, autosomal and sex linked inheritance
- sex determination
- monohybrid crosses using Punnett squares and simple probabilities.

The relevance of human biology to everyday life
Individual differences influence the technologies used to inform the diagnoses of different medical conditions.

Individual differences:

- genetic disorders linked to particular populations e.g. Tay-Sachs, sickle cell anaemia and thalassaemia.

Figure 18.1 Gregor Mendel

From very early times, farmers realised that many characteristics of domesticated plants and animals are passed from parent to offspring. Exactly how these characteristics, or **traits**, were transmitted was not clear. It was thought by many that offspring were simply a blend of the characteristics of the two parents. However, this was not the case. The first clear explanation of patterns of inheritance was provided by an Austrian monk, Gregor Mendel, in 1865. After spending two years at the University of Vienna, he returned to his monastery as a schoolteacher. Here he was able to link his two loves, nature and mathematics, through a careful study of the reproductive behaviour of pea plants. After 10 years of research, Mendel put forward two principles relating to inheritance:

1. that the various hereditary characteristics were controlled by factors (that we now call **genes**) and that these occurred in pairs
2. that during the formation of the gametes (in humans, the eggs and the sperm), the pairs of factors separate. Each gamete receives only one set of factors, or genes, the other set going to another gamete. Gametes unite at fertilisation, allowing different combinations of genes to come together.

Mendel's findings went unnoticed for 35 years before their significance was fully appreciated. At the same time as his work was being rediscovered, scientists were making considerable advances in **cytology**, the study of cells. A young American graduate student, Walter Sutton, was able to link the work of Mendel to that of the cytologists. His observations of the behaviour of chromosomes during meiosis (the type of cell division that takes place to produce gametes, see Chapter 5), and Mendel's speculation on the separation of the hereditary factors during the formation of gametes, led Sutton to suggest that the hereditary factors, or genes, were located in the chromosomes. This important hypothesis, contained in a research paper he published in 1903, led to the **chromosome theory of heredity**.

After the rediscovery of Mendel's work by scientists in 1900, it became apparent that the same principles used to explain inheritance in plants also applied to human traits (Table 18.1). Early studies usually concerned the inheritance of readily identifiable and fairly conspicuous characteristics in individual families. From these, pedigrees were established. A **pedigree** is a family tree that shows the members of the family who have a particular characteristic. Using pedigrees, several hundred human traits were shown to be inherited by single pairs of genes.

Table 18.1 Some inherited traits in humans

Dominant	Recessive
Free earlobes	Attached earlobes
Broad lips	Thin lips
Long eyelashes	Short eyelashes
Broad nostrils	Narrow nostrils
Abundant body hair	Little body hair
Curly hair	Straight hair
Mongolian eye fold	No eye fold
Astigmatism	Normal vision
Roman nose	Straight nose
Huntington disease	No disorder
Achondroplasia	Normal build
Normal enzyme production	Phenylketonuria
Normal pigmentation	Albinism

Mendel's discoveries

To understand how the characteristics in Table 18.1 are passed from one generation to another it is necessary to review the early work of Mendel. Although he worked with plants, his discoveries apply equally well to animals.

Mendel conducted breeding experiments with the edible garden pea, *Pisum*, and was impressed by the fact that it possessed a number of characteristics, or traits, that were expressed in contrasting forms. He studied seven pairs of contrasting characteristics in which the alternatives were easily identifiable (Fig. 18.2).

Before beginning an experiment, Mendel made sure his plants were **pure-breeding** for the characteristic he wished to study. Pure-breeding plants are those that produce the same characteristic in each succeeding generation when bred among themselves. He then crossed, or interbred, the pairs of contrasting traits. For example, plants pure-breeding for yellow-coloured seeds were crossed with plants pure-breeding for green seeds. He found that the offspring (or **progeny**) resembled only *one* of the parents. In the example given, the offspring were all plants that produced yellow seeds. These offspring are referred to as **hybrids** because they have genetic information for green seed colour as well as genetic information for yellow seed colour even though they are all yellow. Thus, only *one* of the pair of contrasting characteristics appeared in the offspring. Mendel referred to the characteristic shown by the hybrid as the **dominant trait** because it masked the appearance of the other characteristic, which he called the **recessive trait**.

For more information on the life of Gregor Mendel go to http://www.augnet.org/default.asp?ipageid=777

Characteristic studied	Dominant character	Recessive character
Seed shape	round	wrinkled
Seed colour	yellow	green
Seed-coat colour	coloured	white
Pod shape	inflated	constricted
Pod colour	green	yellow
Flower position	axial	terminal
Stem length	long	short

Figure 18.2 The seven pairs of contrasting characteristics in garden peas studied by Mendel

When Mendel allowed the hybrid plants to self-pollinate, a second generation of plants was produced. In this generation the characteristics reappeared in the ratio of about three with the dominant trait for every one with the recessive trait (3:1). From these results Mendel concluded that the hereditary factors, or genes, were unchanged as they passed from one generation to the next. He further reasoned that each pea plant had two hereditary factors for each characteristic under study. During the formation of gametes these factors are separated (or, as Mendel called it, segregated), each gamete receiving only one factor, or gene, for each trait. This is known as the **principle of segregation**. As offspring are formed by the union of a male and female gamete, each offspring receives one gene for each characteristic from each parent.

Monohybrid crosses

A **cross** is the mating of two organisms. In a **monohybrid cross** only one pair of contrasting characteristics is studied. For example, yellow and green pod colour in peas or tongue rolling and non-rolling in humans. It is much easier to refer to the genes by a letter than by name. For a particular characteristic, the genes for an individual are, therefore, represented by two letters, one for the gene that originated from the female parent, and one for the gene that originated from the male parent. If the gene is a dominant one it is shown as a capital letter; if it is for a recessive characteristic a lower case letter is used. For the garden pea, green pod colour is dominant to yellow, so the gene for green pod colour would be represented with a capital *G* and the recessive gene for yellow pod colour with a lower case *g*.

For pure-breeding plants of green pod colour the symbols used would be *GG*; pure-breeding plants of yellow pod colour would be *gg*. Hybrids, with one of each gene type, would have the symbols *Gg*. The alternative forms of the gene for seed colour, in this case *G* and *g*, are called **alleles**. For a pair of contrasting characteristics the two alleles may occur in one of three possible combinations: *GG*, *gg* or *Gg*. In two of these three gene combinations, or **genotypes**, the alleles are the same (*GG* and *gg*). These are described as **homozygous**, whereas the hybrid, *Gg*, with one of each allele, is termed **heterozygous**. The three genotypes listed produce only two types of pod colour in garden peas—green for *GG* and *Gg*, and yellow for *gg*. This physical appearance, or what the seeds *look* like, is called the **phenotype**. The terms used to describe inheritance are given in Table 18.2.

Using these terms, we can now look at some examples. Let us examine another of Mendel's crosses. He interbred pure-breeding, long-stemmed pea plants with pure-breeding, short-stemmed plants. The homozygous long-stemmed plants can be represented by the letters *HH*; the homozygous short-stemmed plants by *hh*. During the formation of gametes by meiosis, the pairs of chromosomes separate, with one of each pair going to each gamete. This means that the pairs of alleles segregate with only one allele for a characteristic carried by each gamete. In this case, all the gametes of the long-stemmed plants will have the allele *H*, and all the gametes of the short-stemmed plants will have the allele *h*. Therefore, the offspring will all be the same, *Hh*. As the allele for long stem is dominant, the offspring will all appear as long-stemmed plants. The allele for short stem is masked as it is a recessive characteristic. The offspring are referred to as the **first filial generation**, denoted by the symbol F_1. This cross is shown, diagrammatically, in Figure 18.3.

If the first filial generation is self-pollinated, a second set of offspring are produced. This is the **second filial generation**, or F_2. In this case, the hybrid F_1 plants produce gametes with half containing the allele for long stem and half containing the allele for short stem. Every male gamete has an equal chance of meeting a female gamete, so that the chance of getting particular genotypes in the second generation can be shown mathematically, as in Figure 18.4.

Table 18.2 Terms relating to Mendelian genetics

Term	Meaning
Gene	The factor that determines an inherited characteristic; located in the chromosomes
Allele	The alternative forms of a gene (e.g. the gene for pod colour in peas has two alleles, green and yellow); an individual normally has only two alleles of each gene
Dominant	An allele that masks the effect of another allele (e.g. a pea plant with alleles for green and yellow pods will produce green pods because green is dominant to yellow)
Recessive	An allele that is masked by the effect of an alternative allele (e.g. the allele for yellow pod colour is masked by the allele for green colour)
Homozygous	The situation where an individual has the same alleles for a particular characteristic; also called pure-breeding (e.g. a pea plant with two alleles for green pod colour is homozygous green)
Heterozygous	The situation where an individual possess different alleles for a particular characteristic; also called hybrid (e.g. a pea plant with alleles for both green and yellow-coloured pods is heterozygous)
Phenotype	The physical appearance of an individual as determined by the expression of the alleles for that characteristic (e.g. a pea plant with alleles for green-coloured and for yellow-coloured pods will have the phenotype green pods)
Genotype	The genetic make-up of an individual as determined by the alleles for the characteristic being considered (e.g. a pea plant with an allele for green-coloured pods and one for yellow-coloured pods will have the heterozygous genotype)

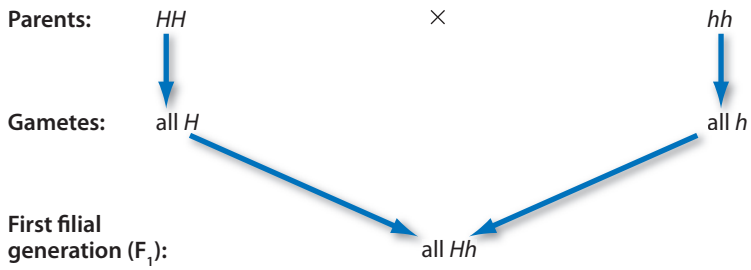


Figure 18.3 The first filial generation

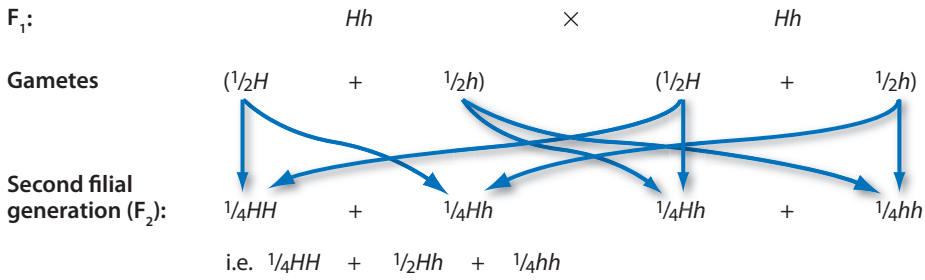


Figure 18.4 The second filial generation

From the final equation in Figure 18.4 you will notice that one-quarter of the F_2 are homozygous for long stems (*HH*), one-half are heterozygous (*Hh*), and one-quarter are homozygous for short stems (*hh*). However, the homozygous long-stemmed plants and the heterozygous plants would *appear* the same; that is, three-quarters of the second generation would appear long-stemmed and one-quarter short-stemmed.

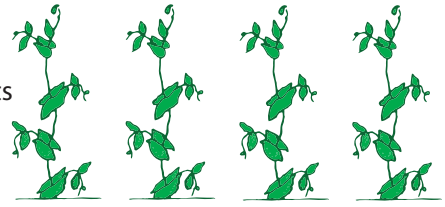
Some students find it easier to use **Punnett squares** to work out genetics problems. The Punnett square is named after RC Punnett, a British geneticist of the early twentieth century, who devised the square to use in his work on heredity. The Punnett square method for the example above is as follows.

Parents *HH* × *hh*

Gametes		Female	
		½*H*	½*H*
Male	½*h*	¼*Hh*	¼*Hh*
	½*h*	¼*Hh*	¼*Hh*

F_1 genotype all *Hh*

F_1 phenotype all long stemmed plants



The F_2 is produced by self-pollinating the F_1 (i.e. *Hh* × *Hh*):

Gametes		Female	
		½*H*	½*h*
Male	½*H*	¼*HH*	¼*Hh*
	½*h*	¼*Hh*	¼*hh*

Each 'box' within the inner 'square' represents the proportion of that genotype that will occur in the offspring. Adding the number of boxes within the square, we obtain:

F_2 genotype ¼*HH* + ½*Hh* + ¼*hh*

F_2 phenotype ¾ long-stemmed and ¼ short-stemmed

Sex determination

An examination of birth records for Australia this century would indicate that girls and boys are born in approximately equal numbers. However, a given family does not necessarily contain the same number of boys as girls. For centuries people have tried to explain how the sex of a child is determined, and it was not until scientists began to examine the nuclei of cells that they realised the chromosome sets in the nuclei of cells of men and women were slightly different. In women, the forty-six chromosomes in the nucleus of each cell were in twenty-three matched pairs, whereas in men the forty-six chromosomes were in only twenty-two matched pairs, the twenty-third pair consisting of two unmatched chromosomes (Fig. 18.5).

Examination of the twenty-third pair of chromosomes in males indicated that one of the pair was similar to the chromosomes of the twenty-third pair in females, but the other was much smaller. The large chromosome became known as the ***X* chromosome** and the smaller the ***Y* chromosome**. Females, therefore, had two *X* chromosomes, and males one *X* and one *Y*. These are called **sex chromosomes**. The chromosomal basis for sex determination is shown schematically in Figure 18.6. The twenty-two pairs of non-sex chromosomes, called **autosomes**, are not normally involved in sex determination and are not shown. All the eggs produced by a female

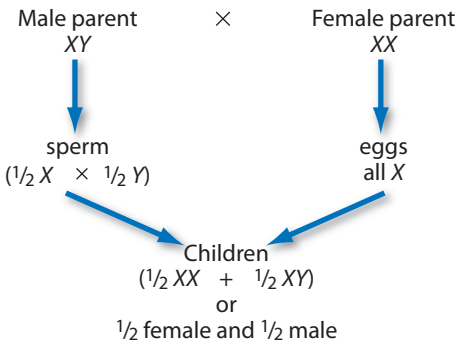


Figure 18.6 Chromosomal basis for sex determination

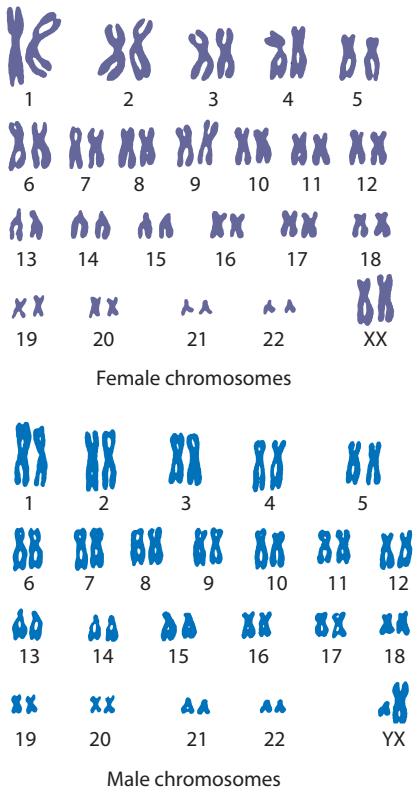


Figure 18.5 Human chromosomes: as chromosomes become visible only during cell division, each appears as a double strand ready for division; the strands are joined at one point, so that each double strand is referred to as a single chromosome

are of one type with respect to the sex chromosomes: they all possess one *X* chromosome. On the other hand, the male's sperm are of two types: about half contain an *X* chromosome and about half a *Y* chromosome. From this information it should be clear that it is the chromosomal complement of the father's sperm that determines the sex of the child. If an *X*-bearing sperm fertilises the egg, the zygote (fertilised egg) will develop into a female; if a *Y*-bearing sperm fertilises the egg, the zygote will develop into a male.

Sex-linked characteristics

Examine the *X* and *Y* chromosomes illustrated in Figure 18.5 again. Notice how the *Y* chromosome is very small compared to the *X*. Obviously, the *Y* chromosome could not have the same number of genes in it as the *X* and, therefore, most of the genes in the *X* chromosome will lack matching alleles in males. For females, however, the normal pairing of alleles will exist. When characteristics located on the *X* chromosome are studied, it is found that the pattern of inheritance is different in the two sexes. Characteristics that show different patterns in the two sexes are called **sex-linked** or ***X*-linked traits**. Two common traits of this type are red-green colour blindness and haemophilia.

The ability to discriminate between the colours red and green is controlled by a gene located in the *X* chromosome. Individuals who are unable to distinguish between the two colours possess the recessive allele of this gene. As the gene is located in the *X* chromosome, this form of colour blindness is found more frequently in males than in females.

In the case of the children of a colourblind man and a woman with normal vision, all would have normal vision. However, the daughters could produce sons who were colourblind. The daughters are carriers for colour blindness because, although they are not colourblind themselves, they can have children who are colourblind. This is shown in the Punnett squares below.

Parents X^bY × X^BX^B

		Female	
Gametes		$\frac{1}{2}X^B$	$\frac{1}{2}X^B$
Male	$\frac{1}{2}X^b$	$\frac{1}{4}X^BX^b$	$\frac{1}{4}X^BX^b$
	$\frac{1}{2}Y$	$\frac{1}{4}X^BY$	$\frac{1}{4}X^BY$

Children: $\frac{1}{4}X^BY + \frac{1}{4}X^BY + \frac{1}{4}X^BX^b + \frac{1}{4}X^BX^b$

½ normal visioned males + ½ normal visioned females but carriers

Note how in crosses for sex-linked characteristics the symbols include the sex chromosome (either *X* or *Y*) and the symbol for the allele being studied. The colourblind man is **hemizygous** for the recessive allele. (As there is no allelic counterpart for males with sex-linked traits, the term 'hemizygous' is used instead of homozygous or heterozygous.) His wife is homozygous for the normal allele (X^BX^B), while all the daughters are heterozygous for the normal allele (X^BX^b), as they inherit the recessive allele from their father. Such people who carry a recessive allele, but do not show the recessive phenotype, are known as **carriers**. The son is normal, as he inherits a normal gene from his mother and the *Y* chromosome with no allelic counterpart from his father.

If a carrier daughter had children with a man with normal vision the pattern of inheritance for their children would result in daughters who may be carriers and sons who have a 50% chance of having the recessive allele.

Parents X^BY × X^BX^b

Gametes		Female	
		½X^B	½X^b
Male	½X^B	¼X^BX^B	¼X^BX^b
	½Y	¼X^BY	¼X^bY

Children: ¼X^BY (normal male) + ¼X^bY (colourblind male) + ¼X^BX^B (normal female) + ¼X^BX^b (normal female but a carrier)

Is it possible for females to be red-green colourblind? If a male with red-green colour blindness has children with a woman who carried the recessive allele, then they have a 50% chance that their daughters will be colourblind and a 50% chance that their sons will also have the trait:

Parents X^bY × X^BX^b

Gametes		Female	
		½X^B	½X^b
Male	½X^b	¼X^BX^b	¼X^bX^b
	½Y	¼X^BY	¼X^bY

Children: ¼X^BY (normal male) + ¼X^bY (colourblind male) + ¼X^BX^b (normal female but a carrier) + ¼X^bX^b (colourblind female)

Haemophilia is another sex-linked characteristic. It is a relatively rare disease in which the blood clots slowly or not at all. The defective allele is recessive to that controlling normal clotting of the blood and is carried on the *X* chromosome. Males, therefore, can be either normal or haemophiliacs, as they have only one *X* chromosome. Females can be homozygous normal; heterozygous, and, therefore, carriers of the condition; or haemophiliacs. This last case is extremely rare.

The pattern of inheritance for haemophilia is similar to that already studied for red-green colour blindness. Haemophiliac fathers pass the recessive gene to their daughters. Carrier mothers may pass a defective gene to their sons, who will be haemophiliacs, or to their daughters, who will then also carry the gene. The most famous family pedigree for haemophilia is that of the European royal families descended from Queen Victoria.

EXTENSION

Queen Victoria was a carrier of the gene for haemophilia and passed the condition on to some of her children.

Find out:

- which of her children were carriers of the condition and if any of her children were haemophiliacs
- how Queen Victoria came to be a carrier for haemophilia given that none of her ancestors showed the trait.

Besides haemophilia and red-green colour blindness, there are a number of disorders in humans that are inherited through recessive genes in the *X* chromosome. These conditions include diabetes insipidus, in which the kidneys are unable to concentrate urine, and Duchenne muscular dystrophy, a progressive wasting disease of the voluntary muscles.

Diabetes insipidus is a sex-linked disorder in which the affected individual passes very large quantities of urine and gradually becomes dehydrated. Death may result unless water is available to replace that lost. The **Duchenne form of muscular dystrophy** is a wasting disease of the leg muscles and later the arms, shoulders and chest. At times it may be due to a mutation, either in a woman to make her a carrier or in a boy to give him the disease. Duchenne muscular dystrophy usually becomes apparent around the age of 3 to 5 years, when muscle weakness becomes evident. As the years pass, more and more muscle tissue wastes away and is replaced by fatty substances. By around 12 to 14 years of age the child is confined to a wheelchair and later is bedridden. With respiratory failure, death becomes inevitable. Generally speaking, boys with the Duchenne form of muscular dystrophy have little chance of living beyond 20 to 25 years.

Inheritance of mitochondrial DNA

Human eggs and sperm both have mitochondria, but while an egg has many hundreds, a sperm has only about 100—just enough to provide the energy for the sperm to swim to the egg. After a sperm has penetrated the egg at fertilisation the mitochondria in the sperm are rapidly destroyed. This means that our nuclear DNA comes from the nucleus of the egg and the sperm but the mitochondrial DNA comes from the egg. In other words, we inherit nuclear DNA from both parents but we inherit mitochondrial DNA only from our mothers. You got your mitochondrial DNA from your mother; she got it from her mother, who got it from hers, and so on.

Since mitochondrial DNA is inherited only through the maternal line it has been very useful in determining relationships between individuals and groups within the human species. It has also been used to demonstrate the evolutionary relationships between humans and closely related species. Mitochondrial DNA has been extracted

from Neanderthal fossils and seems to confirm that modern humans are not descended from Neanderthals and that modern humans did not interbreed with Neanderthals.

EXTENSION

Mitochondrial Eve is a name that has been given to the woman who, when traced through the female line, is the most recent common ancestor for all living humans. The mitochondrial DNA in all humans alive today is derived from her.

Find out:

- how the matrilineal line is traced back to Mitochondrial Eve
- how long ago Mitochondrial Eve is believed to have lived
- in what part of the world she lived
- if the mitochondrial DNA of all humans is derived from Mitochondrial Eve, does that mean she was the only human female alive at the time
- how it is possible that one woman could be the matrilineal ancestor of us all.

Codominance

There are many situations in genetics where the alleles of a particular gene are neither dominant nor recessive. One case is the inheritance of flower colour in sweet pea plants. It was found that when homozygous red-flowered plants were crossed with homozygous white-flowered plants the offspring all had pink flowers. Neither red nor white was dominant: both colours were expressed in the heterozygous offspring as pink. It appeared as though the red and white colours had blended together to form a flower colour in between the two parental colours. As neither parental colour was dominant, and as a form of blending appeared to have taken place, the term **codominance** was used to describe the situation. This pattern of inheritance can also be called **incomplete dominance** although some people do make a fine distinction between the two.

It was found that, when two of these heterozygous pink-flowered plants were crossed, the offspring produced were in the ratio of one-quarter red-flowered plants, one-half pink-flowered plants and one-quarter white-flowered plants. These results indicated that the alleles for red and white had not mixed in the pink offspring: each allele had retained its identity. Let us consider this situation using F^R to represent the red allele and F^W to represent the white allele, as shown in Figure 18.7.

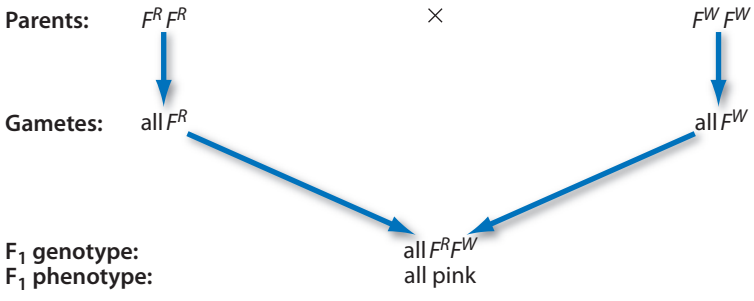


Figure 18.7 Codominance

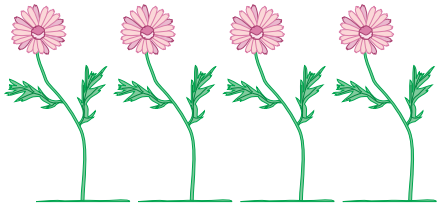
The Punnett square method can be used for this example:

Parents F^RF^R × F^WF^W

Gametes		Female	
		½F^W	½F^W
Male	½F^R	¼F^RF^W	¼F^RF^W
	½F^R	¼F^RF^W	¼F^RF^W

F_1 genotype all F^RF^W

F_1 phenotype all pink

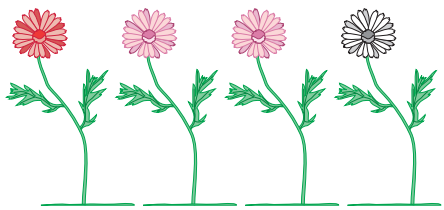


Crossing F_1 individuals produces the following F_2:

Gametes		Female	
		½F^R	½F^W
Male	½F^R	¼F^RF^R	¼F^RF^W
	½F^W	¼F^RF^W	¼F^WF^W

F_1 genotype ¼F^RF^R + ½F^RF^W + ¼F^WF^W

F_1 phenotype ¼ red + ½ pink + ¼ white



This situation is not limited to plants. Humans also show codominance; for example, the B^M and B^N alleles for the M and N antigens in human blood. Neither allele is dominant to the other, and so three phenotypes are possible: type M blood, type N blood and type MN blood. People with type M blood have the genotype B^MB^M, those with blood type N have the genotype B^NB^N and those with blood type MN have the heterozygous B^MB^N genotype. Again, during crosses between various blood types, the identity of the separate alleles is maintained. However, as neither allele is dominant to the other, heterozygous individuals have *both* characters because they have both antigens.

The conventions used for naming alleles are outlined in Table 18.3.

Table 18.3 Conventions for naming alleles

• **DOMINANT** alleles are represented by **UPPER CASE** letters
• **recessive** alleles are represented by **lower case** letters
• **CODOMINANT** alleles are represented by **UPPER CASE** letters
• Use the **same letter** for each allele of a gene; use **superscripts** for codominant alleles (e.g. I^A, I^B)
• **Do not** use letters where upper and lower case are not easily distinguished (e.g. *Ww*, *Ss*, *Cc*)
• **For X-linkage** the *X* and *Y* chromosomes **must** be shown; use superscripts for the alleles (e.g. X^H, Y^h)

EXTENSION

Many human characteristics are determined by multiple alleles. For example, the ABO blood group system is based on the fact that an individual can possess any two of three alternative alleles.

Find out:

- how the pattern of inheritance is determined in the case of the ABO blood groups
- other examples of multiple alleles that determine human characteristics.

Genetic diseases within populations

The incidence in the population of severe recessive disorders that are not linked to a sex chromosome is low, as it is highly unlikely that a carrier from one family will mate with another carrier of the same recessive condition. However, in some regions of the world where populations have been geographically or culturally isolated, marriages between close relatives are common. The probability of having a child with a genetic disease then increases. Marriages of this type are generally between cousins. In these cases there is often a high incidence of a particular genetic disease, as the related parents have received some of their genes from a common ancestor and, therefore, have a greater chance of being carriers of an allele for the same abnormal condition.

Marriages between cousins were once common among the people inhabiting the 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. As thalassaemia is most frequent in countries along the Mediterranean coast, in Australia it occurs in those of Mediterranean origin, especially immigrants from Italy and Greece, or their children. People with thalassaemia require frequent blood transfusions throughout their life and special drugs to remove the excess iron that tends to build up in the body.

Another inherited condition is **sickle-cell anaemia**. It occurs mainly in black Africans, or in people of black African ancestry. In the tropical zone of Africa, up to 40% of the population of some tribes carry the allele for sickle-cell anaemia. The disease occurs when a person inherits the allele from both parents. It results in the red blood cells being a crescent-like, or sickle, shape (Fig. 18.8). The disease is usually fatal as the sickle-shaped cells do not carry as much oxygen as normal red blood cells. They also stick together and block small blood vessels. Heterozygotes normally show no ill

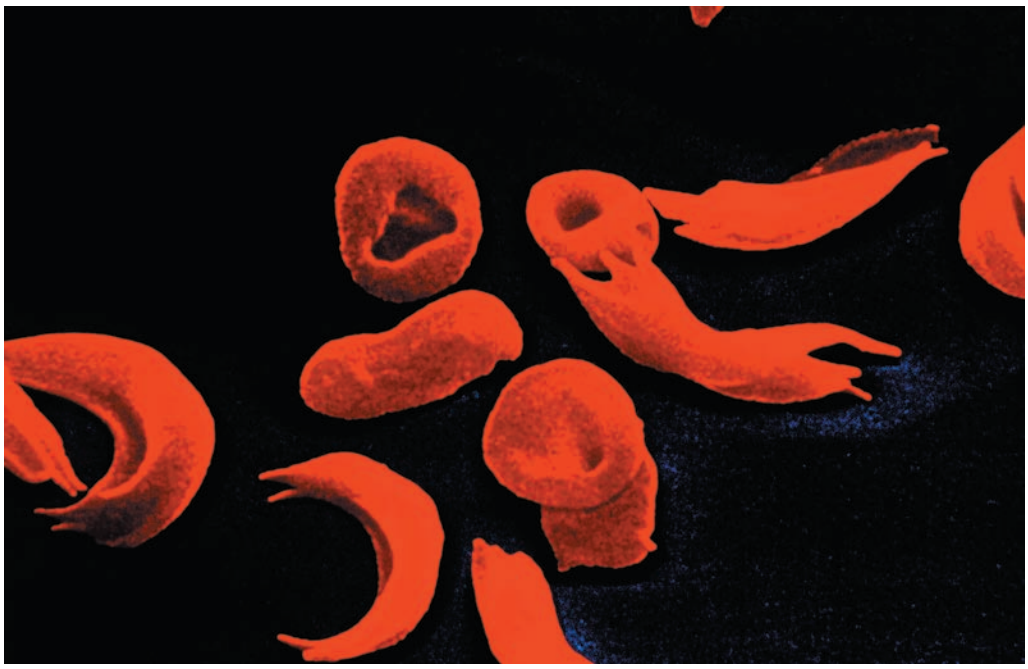


Figure 18.8 Blood of a person with sickle-cell disease. Instead of the usual biconcave disks the red blood cells are often shaped like sickles

effects unless oxygen is in short supply. When this occurs their red blood cells show mild 'sickling'. These individuals are carriers and suffer from **sickle-cell trait**. (Thus, sickle cell anaemia is an example of codominance.) The sickle-cell trait gives certain advantages to those who have it. It provides a degree of immunity to malaria, a disease that is prevalent in those parts of the world where the sickle-cell gene is found.

A third genetic disorder, inherited in an autosomal recessive pattern, is **Tay-Sachs disease (TSD)**. This hereditary disorder of lipid metabolism occurs most frequently in individuals of Jewish descent from eastern Europe (the Ashkenazi Jewish population). It is a fatal disorder caused by a missing enzyme that results in the accumulation of a fatty substance in the nervous system. A baby who has Tay-Sachs develops normally for the first few months and then deterioration causing mental and physical disabilities begins. Death usually occurs in early childhood.

The same mutation that causes Tay-Sachs in Ashkenazi Jews also occurs in the Cajun population of southern Louisiana in the United States. Cajuns are an ethnic group who have been reproductively isolated for several hundred years because of language differences. It has been suggested that the mutation may have entered the Cajun population when a Jewish family assimilated into Cajun society.

Working scientifically

Activity 18.1 Marsians

Marsians are an imaginary group of people from the red planet, Mars. Their skin colour is determined by two alleles—one for red skin colour and one for white skin colour. Red is dominant to white on Mars. In this activity we will investigate whether Marsians follow the principles of Mendelian genetics by simulating a cross between two heterozygous Marsians.

You will need

For each pair:

- 2 containers—2 L ice-cream containers work well
- 20 red beads or counters to simulate the dominant red allele (*R*) in each gamete
- 20 white beads or counters to simulate the recessive white allele (*r*) in each gamete
- felt pen, tally sheet, pencil

What to do

1. Label one container 'Male Parent' and the other 'Female Parent'.
2. Place 10 of the red beads (gamete with *R* allele) in each container, then 10 of the white beads (gamete with *r* allele).
3. Prepare a tally sheet similar to the one below.

	Genotypes in the Marsian offspring			
	RR	*Rr*	*rR*	*rr*
1				
2				
3				
4				
5				
6				
7				
8				
9				
10				

4. Shake the containers well. Draw out one bead (gamete) from each container in turn and place a tick in the relevant box on the tally sheet to show the combination of alleles in the offspring.
5. When you have completed 10 draws, place the beads back into the container. Your partner should repeat steps 1 to 4 above. Together you should now have two completed tally sheets.

Studying your data

1. Because the first three columns all contain the dominant allele (*R*), the individuals with these genotypes will all appear red. Tally up the number of red offspring.
2. Individuals with the genotype *rr* will appear white. How many white offspring do you have?
3. What is the ratio of the phenotypes, red to white?
4. Combine your data with the other groups in the class to obtain a bigger sample. What is the ratio now?

Interpreting your data

1. Has this activity shown that inheritance of skin colour in Marsians follows the principles of Mendelian genetics?
2. How close were your results to the expected result of 3:1?

3. Was the ratio calculated by combining all groups in the class closer to the expected? Explain why this was the case.
4. If the red and white alleles had been incompletely dominant, with the heterozygous colour being pink, what ratio would you now obtain? What ratio would you have expected?

REVIEW QUESTIONS

1. Define the following terms:
   (a) pure-breeding
   (b) progeny
   (c) hybrid
   (d) dominant
   (e) recessive.
2. Briefly describe what is meant by the principle of segregation.
3. Using examples, distinguish between:
   (a) homozygous and heterozygous
   (b) phenotype and genotype
   (c) allele and gene
4. **(a)** What is the first filial generation?
   (b) Distinguish between the first filial generation and the second filial generation.
5. **(a)** What is a monohybrid? Give an example to explain your answer.
   (b) What is codominance? Give an example in which codominance influences the characteristics of offspring.
6. Explain how the sex of a child is determined at the time of fertilisation.
7. Describe the difference in appearance between the X and Y chromosomes.
8. **(a)** What are autosomes?
   (b) How many autosomes occur in (i) each normal human cell (ii) each sperm or egg?
9. **(a)** What are sex-linked (or X-linked) characteristics?
   (b) Give examples of such characteristics.
10. How can mitochondrial DNA be used in researching the ancestry of people?
11. **(a)** What is sickle-cell anaemia? Describe why it is usually lethal.
    (b) What is sickle-cell trait? Does sickle-cell trait have any advantages for people who have it?

APPLY YOUR KNOWLEDGE

1. In garden peas, round seed shape is dominant to wrinkled seed shape. Pure-breeding round seed plants were crossed with pure-breeding wrinkled seed plants. Determine the expected genotypes and phenotypes of the F_1 and F_2, and the expected proportions.
2. In humans, normal melanin production is dominant to albino, which produces white hair and pink eyes. The first child born to a married couple with normal pigmentation is an albino. Calculate the chances that the second child will also be an albino. Give a clear explanation for your results.
3. In guinea pigs, black fur colour is dominant over white fur colour. How could an animal breeder test whether a black guinea pig is homozygous or heterozygous?

4. In humans, free earlobes are dominant over attached earlobes. A woman heterozygous for free earlobes marries a man with attached earlobes. Use a Punnett square to determine the chance of producing children with attached earlobes.
5. In many families, a Roman-shaped nose is dominant to a straight nose. If a man from a family pure-breeding for a Roman nose has children with a woman from a family pure-breeding for a straight nose, what would they look like? If one of the children has children with a person from a family with a long history of straight noses, what types of noses would you expect the grandchildren to possess and in what proportions?
6. When plants that are pure-breeding for wrinkled seeds are crossed with plants that are pure-breeding for round seeds, the ratio of genotypes in the F_2 is 1:2:1 and the ratio of phenotypes is 3:1. Explain what causes the genotypic ratio to differ from the phenotypic ratio.
7. How is the inheritance of flower colour in sweet pea plants an exception to Mendel's principle of dominance?
8. If a human male with blood group M has children with a female with blood group N, what blood groups would they possess? If one of the children has children with a person with blood group M, what blood groups could the grandchildren possess? Construct the crosses for each of these matings. List the genotypes and phenotypes that would be expected and the probability of obtaining each genotype and phenotype.
9. A woman from a family with no history of haemophilia marries a man who is a haemophiliac. What is the probability that they will produce
   (a) sons with normal blood clotting,
   (b) sons with haemophilia,
   (c) daughters who are carriers of haemophilia,
   (d) daughters who will be haemophiliacs?
10. Red-green colour blindness is a sex-linked characteristic. Under what circumstances would a couple produce daughters who all had normal vision and sons who were all colourblind? Describe the genotypes of both parents and all the children.
11. A woman has a brother with Duchenne muscular dystrophy. What information could be given to the woman about the risk of her having a child with Duchenne muscular dystrophy?
12. Explain why certain inherited diseases are more common in some populations than in others. For example, thalassaemia is more common among people of Mediterranean origin and Tay-Sachs more common among Ashkenazi Jews.
13. The Ashkenazi Jews were a reproductively isolated population in Europe for about 1000 years. There was little inter-marriage with other groups including other Jews. Since the mid 20th century many Ashkenazi Jews have intermarried with other Jews and with people of other faiths. In the future, what would you expect to happen to the prevalence of Tay-Sachs disease in people descended from Ashkenazi Jews?
14. As a class 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 of their role in the community and to give you the latest information available about their particular disease and its treatment.