Cambridge A2 Level Biology

Code (9700)

Chapter 16 and Chapter 17

Inherited change and Selection and evolution

Homologous chromosomes

Figure 16.2 shows the chromosomes from Figure 5.2 (page 95) rearranged into numbered pairs, and Figure 16.3 is a diagram of the chromosomes.

A photograph such as Figure 16.2 is called a karyogram. Karyograms are prepared by cutting out individual chromosomes from a picture-like Figure 5.2 and rearranging them.

Note the following in Figures 16.2 and 16.3.

■ ■ There are 22 matching pairs of chromosomes. T hese are called homologous chromosomes. (The word 'homologous' means similar in structure and composition.) Each pair is given a number. In the original zygote, one of each pair came from the mother, and one from the father. There is also a non-matching pair labelled X and Y. There are, therefore, two sets of 23 chromosomes – one set of 23 from the father and one set of 23 from the mother.

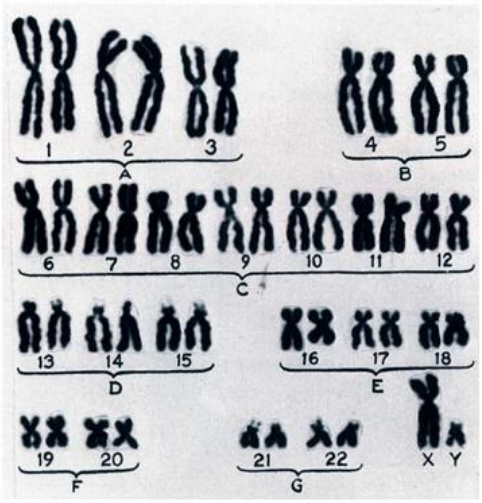


Figure 16.2 Karyogram of a human male. There are 22 homologous pairs of non-sex chromosomes (autosomes). The sex chromosomes (X, female; Y, male) are placed separately.

■ ■ The non-matching X and Y chromosomes are the sex chromosomes, which determine the sex of the individual. All the other chromosomes are called autosomes. It is conventional to position the two sex chromosomes to one side in a karyogram, so that the sex of the organism can be recognised quickly. In humans, females have two X chromosomes, and males have one X and one Y chromosome. The Y chromosome has portions missing and is therefore smaller than the X chromosome.

■■ The pairs of chromosomes can be distinguished not only by size and shape, but because each pair has a distinctive banding pattern when stained with certain stains, as shown in Figure 16.3.

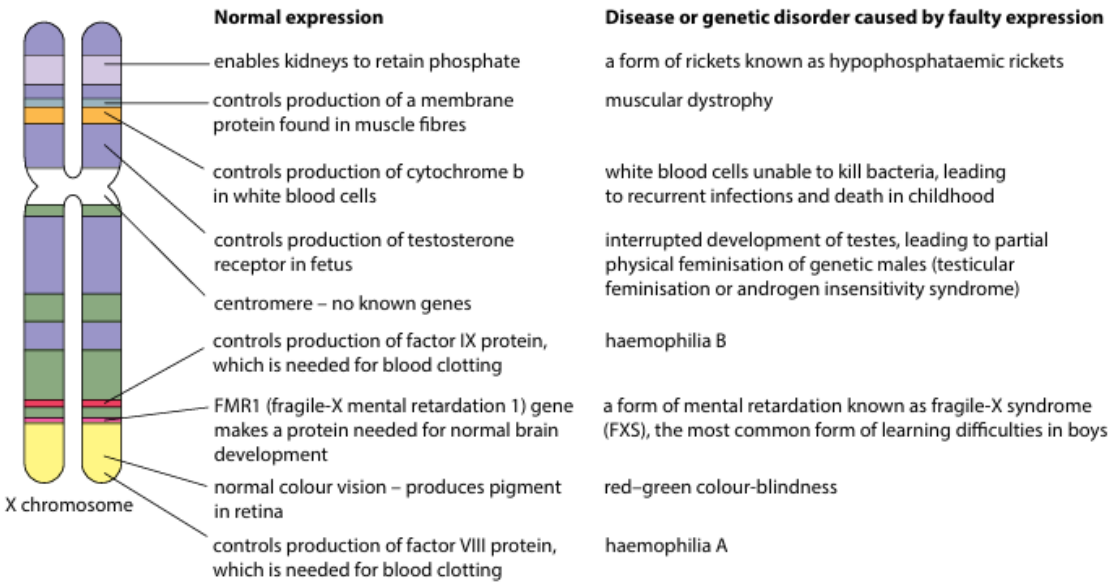


Figure 16.4 Locations of some of the genes on the human female sex chromosome (the X chromosome), showing the effects of normal and faulty expression. The chromosome has 153 million base pairs.

Haploid and diploid cells

When animals other than humans are examined, we again find that cells usually contain two sets of chromosomes. Such cells are described as diploid. This is represented as 2n, where n = the number of chromosomes in one set of chromosomes.

Two types of nuclear division

Figure 16.6 shows a brief summary of the life cycle of an animal such as a human. Two main stages are involved.

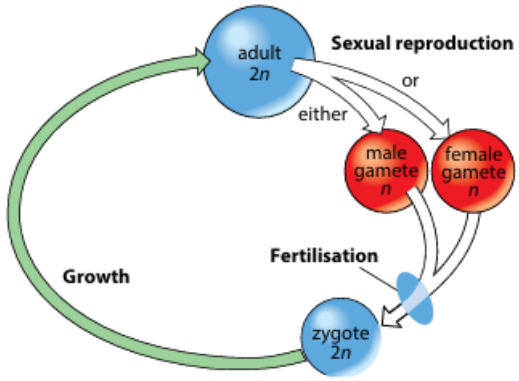


Figure 16.6 Outline of the life cycle of an animal.

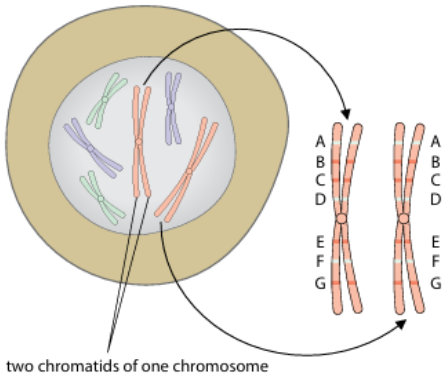


Figure 16.5 Homologous chromosomes carry the same genes at the same loci. Just seven genes, labelled A–G, are shown on this pair of chromosomes, but in reality there are often hundreds or thousands of genes on each chromosome. This is a diploid cell as there are two complete sets of chromosomes ($2n = 6$).

■ Growth. When a diploid zygote, which is one cell, grows into an adult with millions of cells, the new cells must be genetically identical, with the same number of chromosomes as the cells that divided to produce them. The type of nuclear division that achieves this is called mitosis.

■ ■ Sexual reproduction. For the life cycle to contain sexual reproduction, there must be a point before fertilisation takes place when the number of chromosomes is halved.

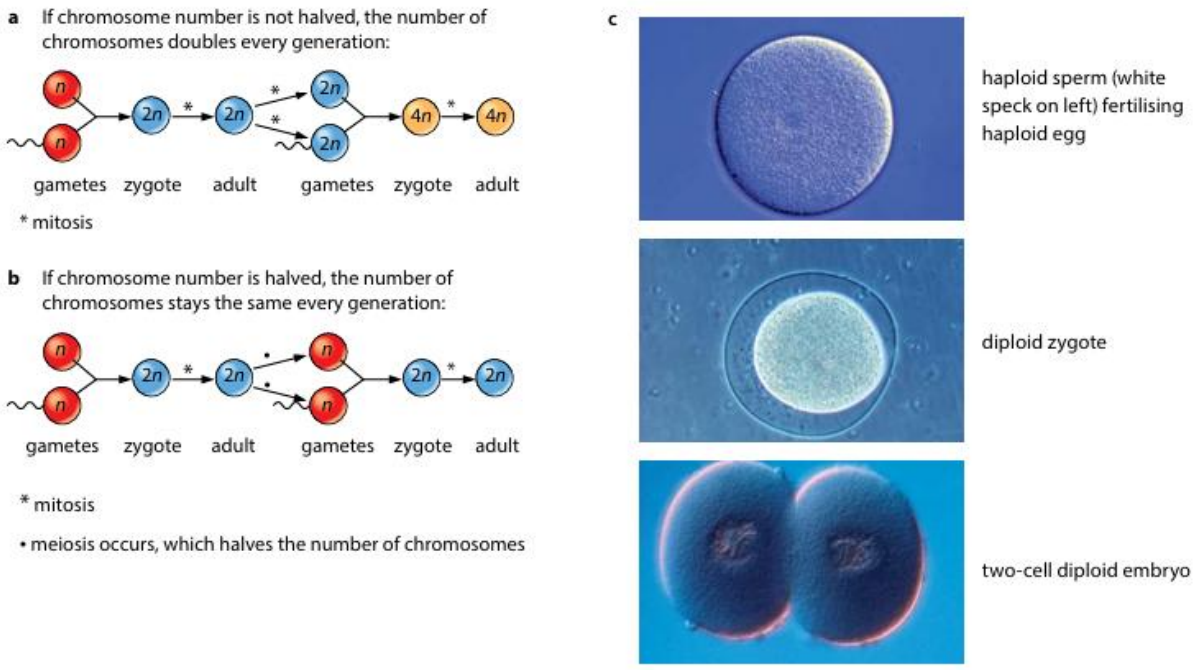


Figure 16.7 A life cycle in which the chromosome number is **a** not halved; **b** halved; **c** life cycle stages in a sea urchin .

Meiosis

Meiosis is a process in which an animal cell undergoes two divisions, meiosis I and meiosis II. Meiosis I is a reduction division, resulting in two daughter nuclei with half the parent nucleus's chromosomes. In meiosis II, chromosomes divide again, resulting in four haploid nuclei. The behavior of chromosomes in meiosis I is crucial.

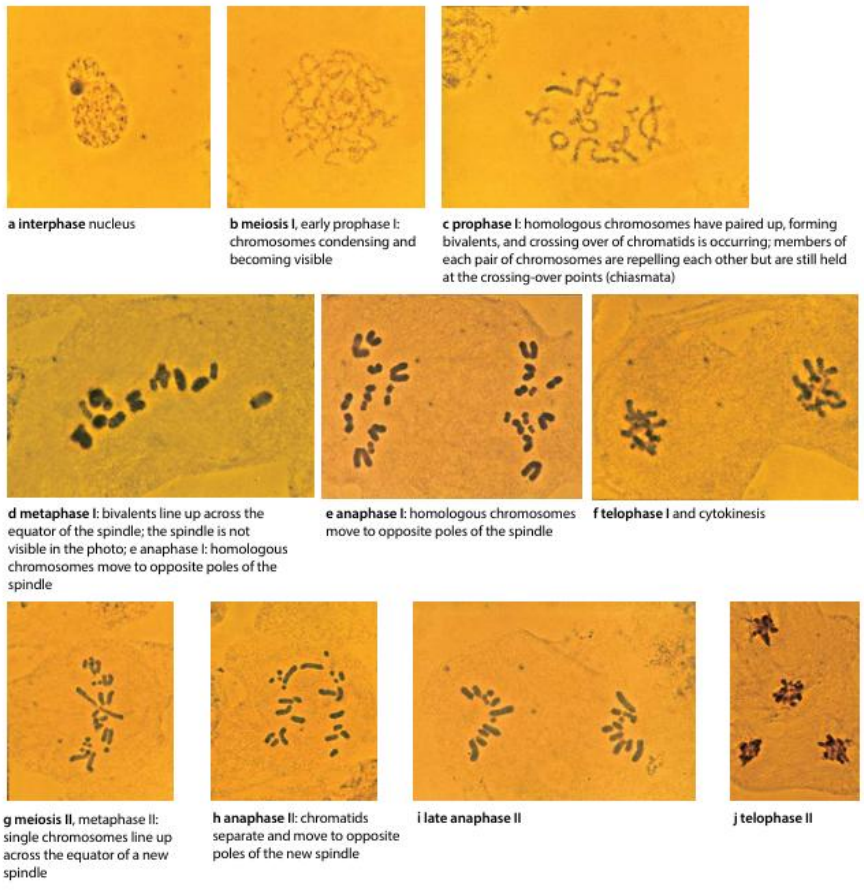


Figure 16.9 Stages of meiosis in an animal cell (locust) (×950). Interphase (not part of meiosis) is also shown.

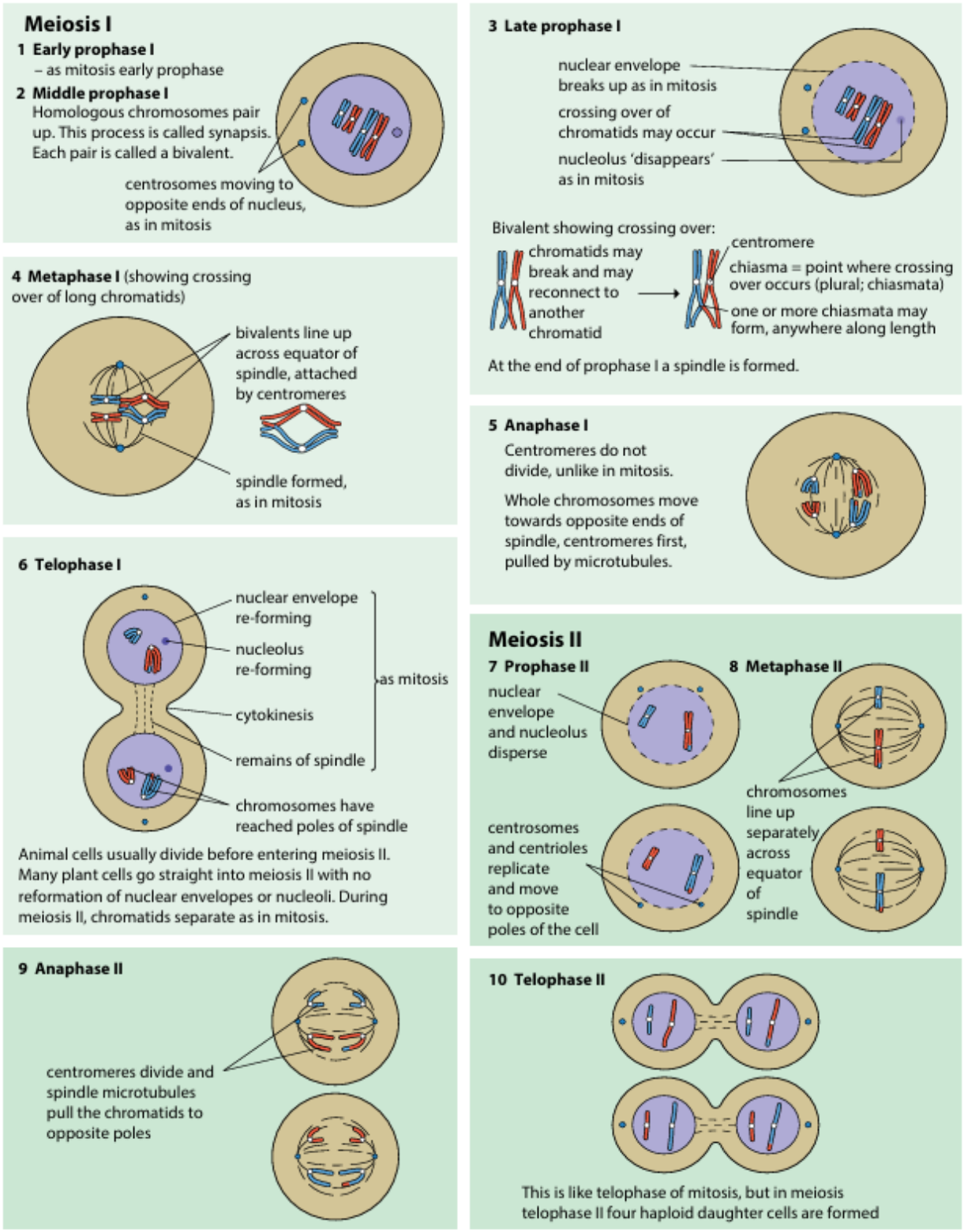


Figure 16.8 Meiosis and cytokinesis in an animal cell. Compare this process with nuclear division by mitosis, shown in Figure 5.7 (page 98).

Gametogenesis in humans

Sperm production takes place inside tubules in the testes. Here, diploid cells divide by mitosis to produce numerous diploid **spermatogonia**, which grow to form diploid **primary spermatocytes**. The first division of meiosis then takes place, forming two haploid **secondary spermatocytes**. The second division of meiosis then produces **haploid spermatids**, which mature into spermatozoa.

However, the division is uneven; one cell gets most of the cytoplasm, and becomes a **secondary oocyte**, while the other is little more than a nucleus, and is called a **polar body.**

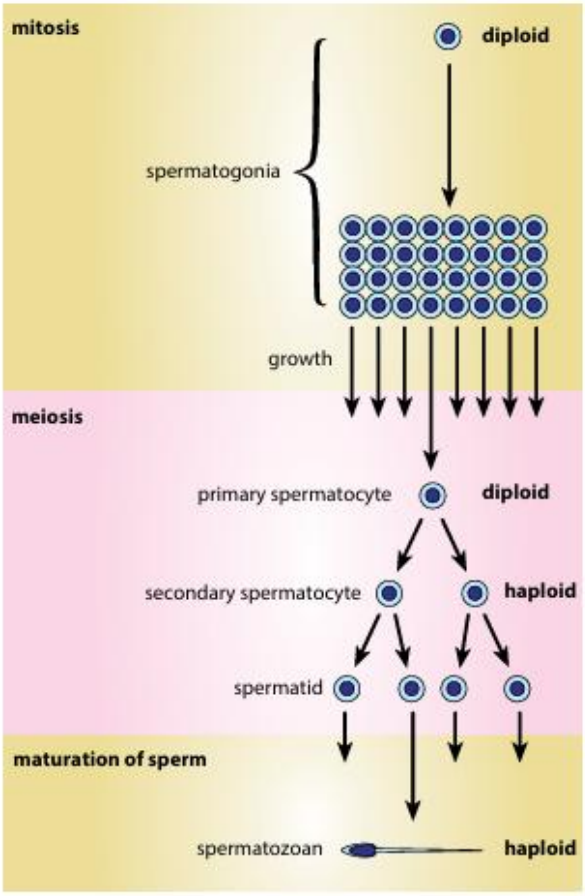


Figure 16.10 Spermatogenesis in humans.

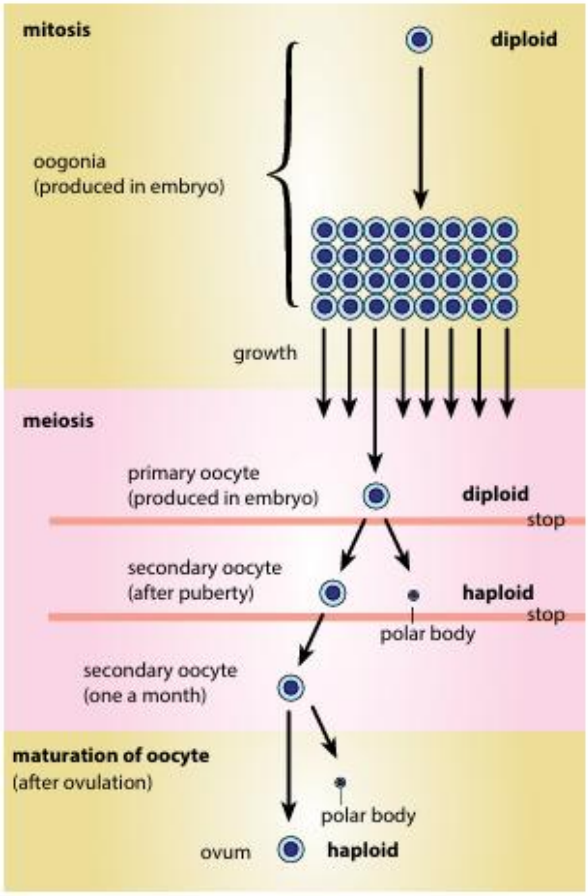


Figure 16.11 Oogenesis in humans.

Gametogenesis in flowering plants

Figure 16.12 shows the structure of a typical flower. Male gametes are produced in the anthers, and female gametes in the ovules.

Inside the anthers, diploid **pollen mother** cells divide by meiosis to form four haploid cells

These cells mature into **pollen grains**, each surrounded by a protective wall made up of a tough exine and thinner intine (Figure 16.13). One of the haploid nuclei is called the tube nucleus, and the other is the **generative nucleus**. These are the male gametes.

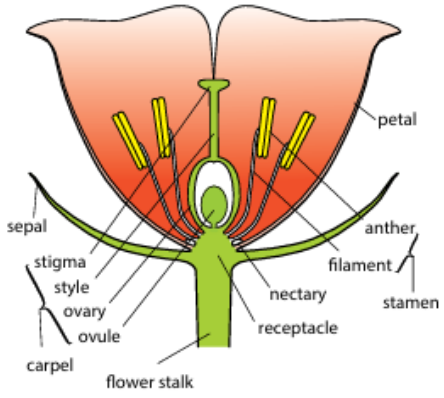


Figure 16.12 The structure of a flower.

Inside each ovule, a large, diploid, **spore mother cell** develops. This cell divides by meiosis to produce four haploid cells. All but one of these degenerates, and the one surviving haploid cell develops into an **embryo sac** (Figure 16.14)

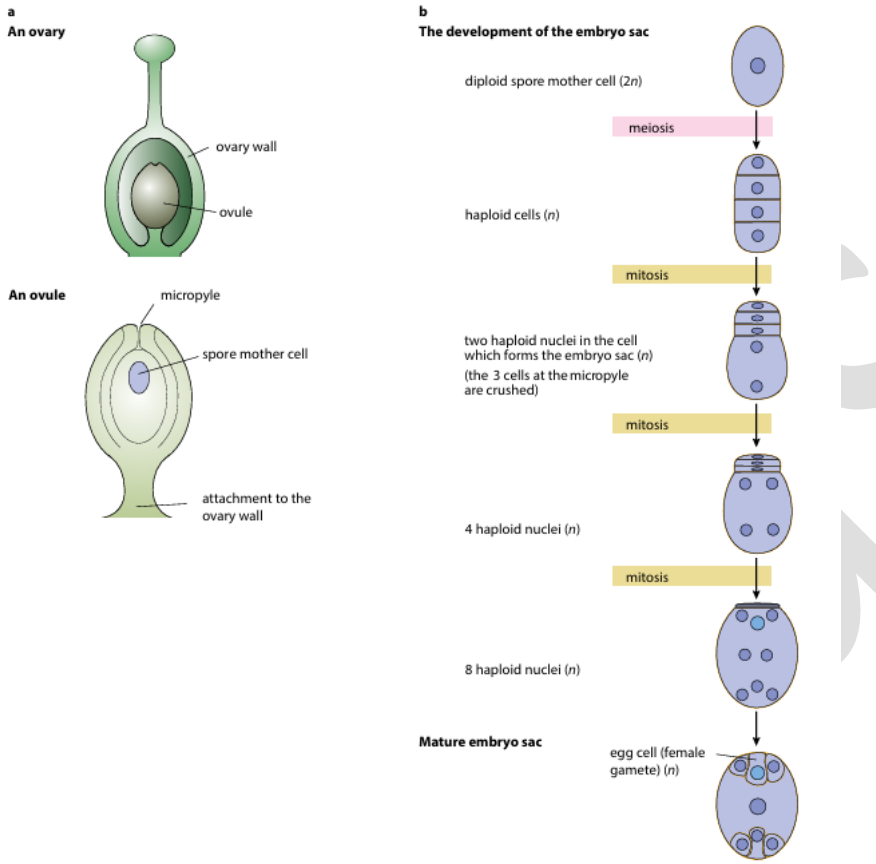


Figure 16.14 **a** Structure of an ovary and an ovule. **b** Development of the embryo sac.

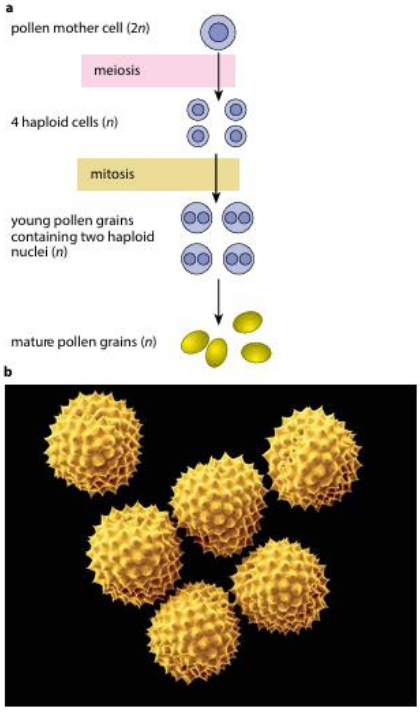


Figure 16.13 **a** The development of pollen grains from pollen mother cells. **b** Mature pollen grains (×1500).

Genetics

You will remember that a gene is a length of DNA that codes for the production of a polypeptide molecule. The code is held in the sequence of nucleotide bases in the DNA.

Alleles

The gene that codes for the production of the β-globin polypeptide of the haemoglobin molecule is on chromosome 11. Each cell contains two copies of this gene, one maternal in origin (from the mother) and one paternal (from the father).

Genotype

Most genes, including the β-globin polypeptide gene, have several different alleles.

Hb^A = the allele for the normal β-globin polypeptide
Hb^S = the allele for the sickle cell β-globin polypeptide

The letters **Hb** stand for the locus of the haemoglobin gene, whereas the superscripts ^{A} and ^{S} stand for particular alleles of the gene.

In a human cell, which is diploid, there are two copies of the β-globin polypeptide gene. The two copies might be:

Hb^AHb^A or Hb^SHb^S or Hb^AHb^S.

Genotype	Phenotype
Hb^AHb^A	normal
Hb^AHb^S	normal, but with sickle cell trait
Hb^SHb^S	sickle cell anaemia

Table 16.1 Genotypes and phenotypes for sickle cell anaemia.

An organism's **phenotype** is its characteristics, often resulting from an interaction between its genotype and its environment.

Genotype affects phenotype

Individuals with different genotypes of HbAHbA and HbAHbS have different levels of sickle cell anemia. HbAHbA individuals have two copies of the gene for normal β-globin polypeptide, while HbSHbS individuals have two copies for sickle cell β-globin polypeptide. This results in inefficient oxygen transport, leading to sickle cell anemia. HbAHbS individuals have one allele for normal β-globin and one for sickle cell β-globin, causing half of their haemoglobin to be normal and half sickle cell. This condition is dangerous and requires careful management.

Inheriting genes

In sexual reproduction, haploid gametes are made, following meiosis, from diploid body cells. Each gamete contains one of each pair of chromosomes.

Think about what happens when sperm are made in the testes of a man who has the genotype Hb^AHb^S. Each time a cell divides during meiosis, four gametes are made, two of them with the Hb^A allele and two with the Hb^S allele. Of all the millions of sperm that are made in his lifetime, half will have the genotype Hb^A and half will have the genotype Hb^S (Figure 16.15).

Similarly, a heterozygous woman will produce eggs of which half have the genotype Hb^A and half have the genotype Hb^S.

This information can be used to predict the possible genotypes of children born to a couple who are both heterozygous. Each time fertilisation occurs, either an Hb^A sperm or an Hb^S sperm may fertilise either an Hb^A egg or an Hb^S egg. The possible results can be shown like this:

Genotypes of sperm \ Genotypes of eggs	Hb^A	Hb^S
Hb^A	Hb^AHb^A normal	Hb^AHb^S sickle cell trait
Hb^S	Hb^AHb^S sickle cell trait	Hb^SHb^S sickle cell anaemia

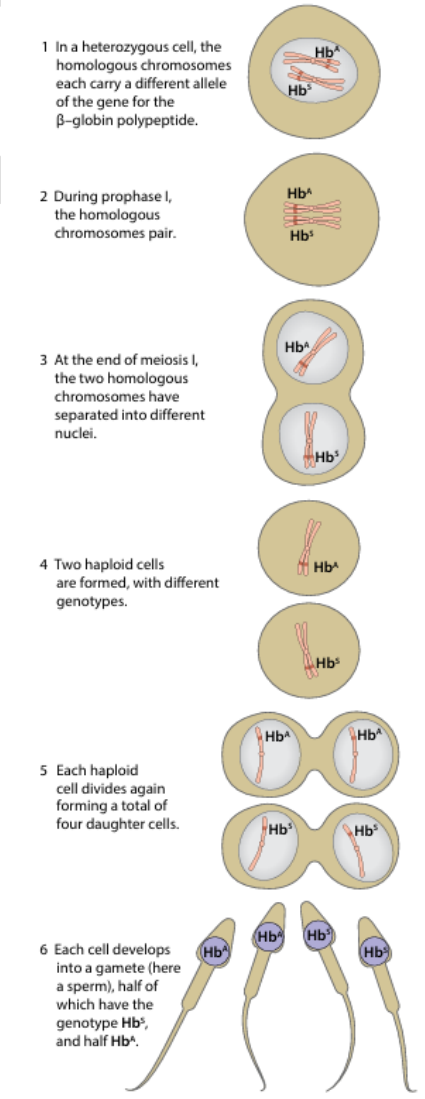


Figure 16.15 Meiosis of a heterozygous cell produces gametes of two different genotypes. Only one pair of homologous chromosomes is shown.

Genetic diagrams

A genetic diagram is the standard way of showing the genotypes of offspring that might be expected from two parents. To illustrate genetic diagrams, let us consider f lower colour in snapdragons (Antirrhinum).

One of the genes for flower colour has two alleles, namely C^R, which gives red flowers, and C^W, which gives white flowers. The phenotypes produced by each genotype are:

Genotype	Phenotype
C^RC^R	red
C^RC^W	pink
C^WC^W	white

What colour flowers would be expected in the offspring from a red and a pink snapdragon?

Parental phenotypes red × pink

Parental genotypes C^RC^R × C^RC^W

Gametes all C^R × C^R or C^W in equal proportions

Offspring genotypes and phenotypes:

Gametes from pink parent \ Gametes from red parent	C^R
C^R	C^RC^R red flowers
C^W	C^RC^W pink flowers

Thus, you would expect about half of the offspring to have red flowers and half to have pink flowers.

Dominance

In heterozygous organisms, both alleles affect the phenotype, such as normal and sickle cell haemoglobin in a person or red and white colors in a snapdragon. Codominant alleles are dominant, while recessive alleles have no effect. For example, in tomato plants, one allele produces green stems and one produces purple stems, resulting in the same shade of purple stems.

The possible genotypes and phenotypes for stem colour are:

Genotype	Phenotype
AA	purple stem
Aa	purple stem
aa	green stem

A **dominant** allele is one whose effect on the phenotype of a heterozygote is identical to its effect in a homozygote.

A **recessive** allele is one that is only expressed when no dominant allele is present.

Codominant alleles both have an effect on the phenotype of a heterozygous organism.

F1 and F2 generations

The symbols F1 and F2 may be used in genetic diagrams. These symbols have specific meanings and should not be used in other circumstances.

The **F1 generation** is the offspring resulting from a cross between an organism with a homozygous dominant genotype, and one with a homozygous recessive genotype.

The **F2 generation** is the offspring resulting from a cross between two F1 (heterozygous) organisms

Test crosses

Where alleles show dominance, it is not possible to tell the genotype of an organism showing the dominant characteristic just by looking at it. A purple stemmed tomato plant might have the genotype AA, or it might have the genotype Aa.

If the purple-stemmed tomato plant's genotype is **AA**:

Parental phenotypes	purple	green
Parental genotypes	**AA**	**aa**
Gametes	Ⓐ	ⓐ
Offspring	all **Aa** purple	

If its genotype is **Aa**:

Parental phenotypes	purple	green
Parental genotypes	**Aa**	**aa**
Gametes	Ⓐ or ⓐ	ⓐ
Offspring	**Aa** purple	**aa** green

Multiple alleles

So far, we have considered just two alleles, or varieties, of any one gene. Most genes, however, have more than two alleles. An example of this situation, known as multiple alleles, is the gene for human blood groups.

The four blood groups A, B, AB and O are all determined by a single gene. Three alleles of this gene exist, I^A, I^B, and I^o. Of these, I^A and I^B are codominant, whereas I^o is recessive to both I^A and I^B. As a diploid cell can carry only two alleles, the possible genotypes and phenotypes are as shown in Table 16.2.

Genotype	Blood group
I^AI^A	A
I^AI^B	AB
I^AI^o	A
I^BI^B	B
I^BI^o	B
I^oI^o	O

Table 16.2 Genotypes and phenotypes for blood groups.

Sex inheritance

In humans, sex is determined by one of the 23 pairs of chromosomes. These chromosomes are called the sex chromosomes.

Sex linkage

The X chromosome contains genes, including factor VIII, a protein for blood clotting. There are two alleles, H producing normal factor VIII and h causing haemophilia. The gene for haemophilia is on the X chromosome, affecting inheritance. Females have two copies of the gene, while males have one. The factor VIII gene is sex-linked, found on a part of the X chromosome not matched by the Y chromosome. Genes with sex-linked genes are represented by symbols on an X chromosome.

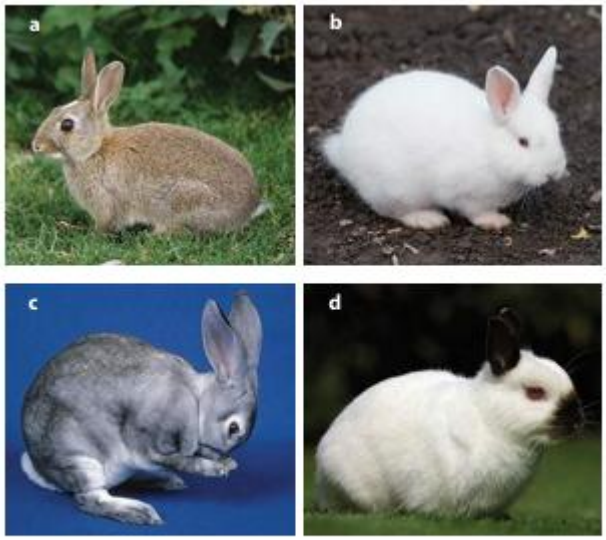


Figure 16.16 Colour variations in rabbits, caused by multiple alleles of a single gene: **a** agouti; **b** albino; **c** chinchilla; **d** Himalayan.

Parental phenotypes	normal man	carrier woman
Parental genotypes	X^HY	X^HX^h
Gametes	X^H or Y	X^H or X^h

Offspring genotypes and phenotypes:

Gametes from man \ Gametes from woman	X^H	X^h
X^H	X^HX^H normal female	X^HX^h carrier female
Y	X^HY normal male	X^hY haemophiliac male

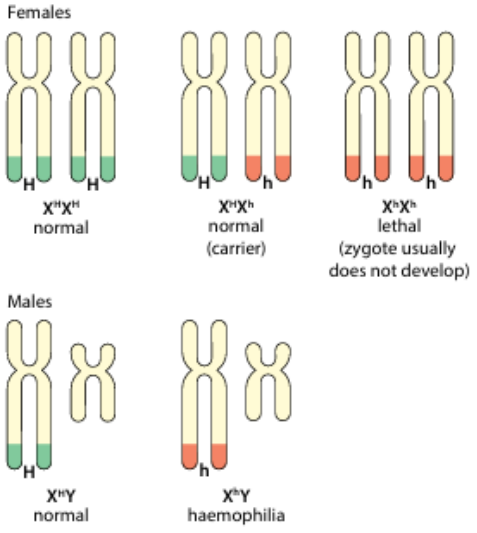


Figure 16.17 The possible genotypes and phenotypes for haemophilia.

Dihybrid crosses

So far, we have considered the inheritance of just one gene. Such examples are called monohybrid crosses. Dihybrid crosses look at the inheritance of two genes at once.

You have already seen that, in tomato plants, there is a gene that codes for stem colour. This gene has two alleles:

stem colour gene **A** = allele for purple stem
a = allele for green stem

where **A** is dominant and **a** is recessive.

A different gene, at a different locus on a different chromosome, codes for leaf shape. Again, there are two alleles:

leaf shape gene **D** = allele for cut leaves (jagged edges)
d = allele for potato leaves (smooth edges)

where **D** is dominant and **d** is recessive.

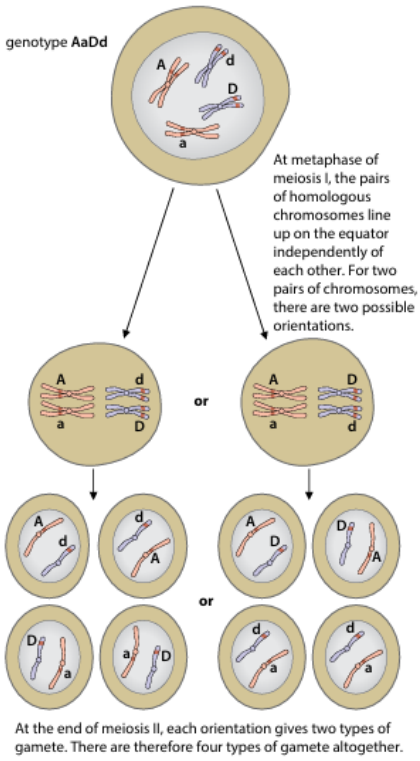


Figure 16.18 Independent assortment of homologous chromosomes during meiosis I results in a variety of genotypes in the gametes formed.

If there are many such cells undergoing meiosis, then the chromosomes in roughly half of them will probably line up one way, and the other half will line up the other way. T his is independent assortment. We can therefore predict that the gametes formed from these heterozygous cells will be of four types, AD, Ad, aD and ad, occurring in approximately equal numbers.

The plant with green stem and potato leaves must have the genotype **aadd**. Each of its gametes will contain one **a** allele and one **d** allele. All of the gametes will have the genotype **ad**.

Parental phenotypes	purple stem, cut leaves	green stem, potato leaves
Parental genotypes	**AaDd**	**aadd**
Gametes	(AD) or (Ad) or (aD) or (ad) in equal proportions	all (ad)

At fertilisation, any of the four types of gamete from the heterozygous parent may fuse with the gametes from the homozygous parent. The genotypes of the offspring will be:

		Gametes from green, potato plant (ad)
Gametes from purple, cut plant	(AD)	**AaDd** purple stem, cut leaves
	(Ad)	**Aadd** purple stem, potato leaves
	(aD)	**aaDd** green stem, cut leaves
	(ad)	**aadd** green stem, potato leaves

From this cross, therefore, we would expect approximately equal numbers of the four possible phenotypes. This 1 : 1 : 1 : 1 ratio is typical of a dihybrid cross between a heterozygous organism and a homozygous recessive organism where the alleles show complete dominance.

If **both** parents are heterozygous, then things become a little more complicated, because both of them will produce four kinds of gametes.

Parental phenotypes	purple stem, cut leaves	purple stem, cut leaves
Parental genotypes	**AaDd**	**AaDd**
Gametes	(AD) or (Ad) or (aD) or (ad) in equal proportions	(AD) or (Ad) or (aD) or (ad) in equal proportions

Offspring genotypes and phenotypes:

		Gametes from one parent (AD)	(Ad)	(aD)	(ad)
Gametes from other parent	(AD)	**AADD** purple, cut	**AADd** purple, cut	**AaDD** purple, cut	**AaDd** purple, cut
	(Ad)	**AADd** purple, cut	**AAdd** purple, potato	**AaDd** purple, cut	**Aadd** purple, potato
	(aD)	**AaDD** purple, cut	**AaDd** purple, cut	**aaDD** green, cut	**aaDd** green, cut
	(ad)	**AaDd** purple, cut	**Aadd** purple, potato	**aaDd** green, cut	**aadd** green, potato

If you sort out the numbers of each phenotype among these 16 possibilities, you will find that the offspring would be expected to occur in the following ratio:

9 purple, cut : 3 purple, potato : 3 green, cut : 1 green, potato

This 9 : 3 : 3 : 1 ratio is typical of a dihybrid cross between two heterozygous organisms where the two alleles show complete dominance and where the genes are on different chromosomes.

Interactions between loci

You have already seen interactions between alleles at the same locus, namely:

■ ■ codominant alleles in flower colour in snapdragons

■ ■ dominant and recessive alleles in tomato plant stem colour

■ ■ multiple alleles in the inheritance of the ABO blood groups.

of 9 : 3 : 4. Two loci, **A/a** and **B/b**, on different chromosomes are involved:

Genotype	Phenotype
A–B–	purple
A–bb	pink
aaB–	white
aabb	white

(– indicates that either allele of the gene may be present)

The homozyote recessive **aa** affects the **B/b** locus. Neither the dominant allele, **B**, for purple flower colour, nor the recessive allele, **b**, for pink flower colour can be expressed in the absence of a dominant **A** allele.

Parental phenotypes	white	white
Parental genotypes	IICC	iicc
Gametes	(IC)	(ic)
Offspring (F1) genotypes	all IiCc	
Offspring phenotypes	all white	

These offspring are interbred to give another generation.

Parental phenotypes white × white

Parental genotypes IiCc × IiCc

Gametes (IC) or (Ic) or (iC) or (ic) × (IC) or (Ic) or (iC) or (ic)

in equal proportions

Offspring (F2) genotypes and phenotypes:

Gametes from other parent \ Gametes from one parent	(IC)	(Ic)	(iC)	(ic)
(IC)	IICC white	IICc white	IiCC white	IiCc white
(Ic)	IICc white	IIcc white	IiCc white	Iicc white
(iC)	IiCC white	IiCc white	iiCC coloured	iiCc coloured
(ic)	IiCc white	Iicc white	iiCc coloured	iicc white

Autosomal linkage

When two or more gene loci are on the same chromosome, they do not assort independently in meiosis as they would if they were on different chromosomes. The genes are said to be linked.

Linkage is the presence of two genes on the same chromosome, so that they tend to be inherited together and do not assort independently.

The fruit fly, *Drosophila*, normally has a striped body and antennae with a feathery arista (Figure 16.19). The gene for body colour and the gene for antennal shape are close together on the same chromosome and so are linked.

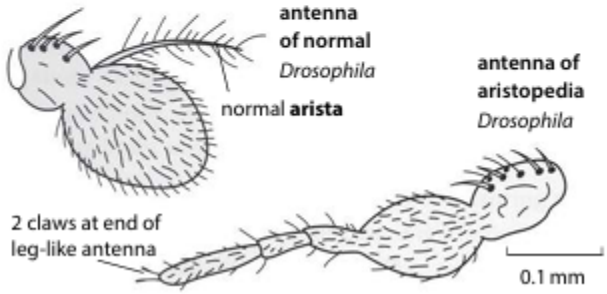


Figure 16.19 Normal and aristopedia *Drosophila* antennae.

A black body with no stripes results from a recessive allele called 'ebony'. A recessive allele for antennal shape, called 'aristopedia', gives an antenna looking rather like a *Drosophila* leg, with two claws on the end.

Body colour gene:
E = allele for striped body
e = allele for ebony body
Antennal shape gene:
A = allele for normal antennae
a = allele for aristopedia antennae

T he test cross gives a 1 : 1 ratio of the two original parental types and not the 1 : 1 : 1 : 1 ratio expected from a dihybrid cross. (If you are uncertain about these ratios, repeat the cross above but assume that the genes are not linked. This should result in 1 EeAa : 1 Eeaa : 1 eeAa : 1 eeaa.)

Crossing over

During prophase I of meiosis, a pair of homologous chromosomes (a bivalent) can be seen to be joined by chiasmata (Figure 16.20). The chromatids of a bivalent may break and reconnect to another, non-sister chromatid. T his results in an exchange of gene loci between a maternal and paternal chromatid (Figure 16.21)

Smaller numbers of **recombinant** flies are produced. These result from crossing over and 'recombine' the characteristics of the original parents into some flies that have a striped body and aristopedia antennae and others that have an ebony body with normal antennae.

striped body, normal antennae	44%	parental classes
ebony body, aristopedia antennae	44%	
striped body, aristopedia antennae	6%	recombinant classes
ebony body, normal antennae	6%	

The **cross over value** is the percentage of offspring that belong to the recombinant classes. In this case it is 6% + 6% = 12%. This is a measure of the distance apart of the two gene loci on their chromosomes. The smaller the cross over value, the closer the loci are together. The chance of a cross over taking place between two loci is directly related to their distance apart.

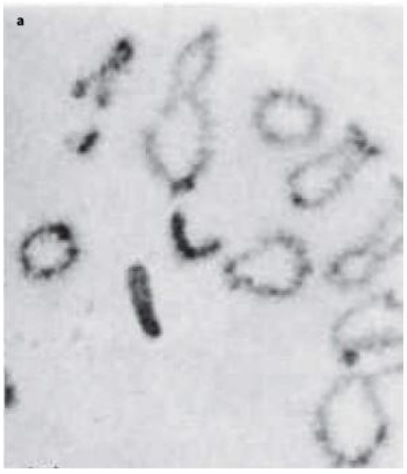
Parental phenotypes	striped body normal antennae	ebony body aristopedia antennae
Parental genotypes	(EA)(EA)	(ea)(ea)
Gametes	(EA)	(ea)
Offspring (F1) genotypes and phenotypes	all (EA)(ea) striped body normal antennae	

Male offspring were then test crossed with females homozygous for ebony body and aristopedia antennae, producing the two original parent types in equal numbers

Parental phenotypes	male striped body normal antennae	female ebony body aristopedia antennae
Parental genotypes	(EA)(ea)	(ea)(ea)
Gametes	(EA) or (ea) in equal proportions	(ea)

Offspring genotypes and phenotypes:

Gametes from male parent	Gametes from female parent: (ea)
(EA)	(EA)(ea) striped body, normal antennae
(ea)	(ea)(ea) ebony body, aristopedia antennae



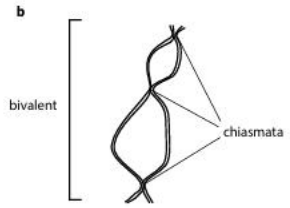
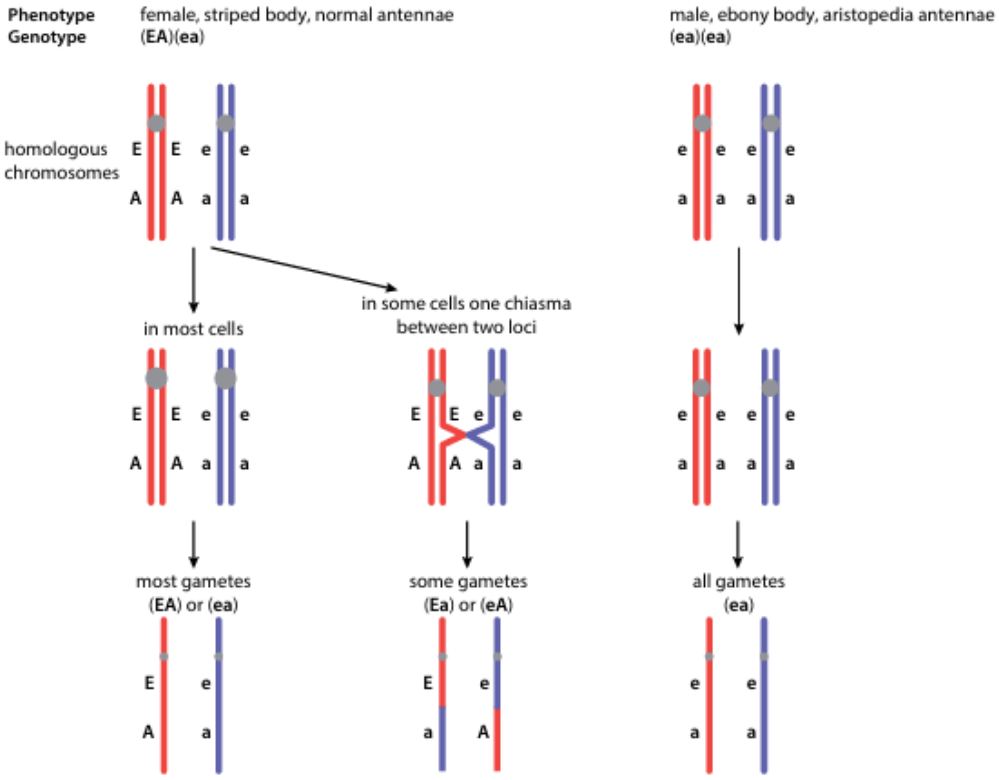


Figure 16.20 **a** Photomicrograph of bivalents in prophase I of meiosis, showing chiasmata. A chiasma shows that crossing over has occurred between two chromatids. **b** Interpretive drawing of one bivalent.



Parental phenotypes	female striped body normal antennae	male ebony body aristopedia antennae
Parental genotypes	(EA)(ea)	(ea)(ea)
Gametes	large numbers of (EA) and (ea)	(ea)
	small numbers of (Ea) and (eA)	

Offspring genotypes and phenotypes:

Gametes from female parent		Gametes from male parent: (ea)
large numbers	(EA)	(EA)(ea) striped body, normal antennae
large numbers	(ea)	(ea)(ea) ebony body, aristopedia antennae
small numbers	(Ea)	(Ea)(ea) striped body, aristopedia antennae
small numbers	(eA)	(eA)(ea) ebony body, normal antennae

Figure 16.21 Crossing over in female *Drosophila*.

The χ^2 (chi-squared) test

If you look back at the cross between the two heterozygous tomato plants on pages 380–381, you will see that we would expect to see a 9:3:3:1 ratio of phenotypes in the offspring. It is important to remember that this ratio represents the **probability** of getting these phenotypes, and we would probably be rather surprised if the numbers came out absolutely precisely to this ratio.

But just how much difference might we be happy with, before we began to worry that perhaps the situation was not quite what we had thought? For example, let

If the parents really were both heterozygous, and if the purple stem and cut leaf alleles really are dominant, and if the alleles really do assort independently, then we would expect the following numbers of each phenotype to be present in the off spring:

purple, cut $= \frac{9}{16} \times 144 = 81$
purple, potato $= \frac{3}{16} \times 144 = 27$
green, cut $= \frac{3}{16} \times 144 = 27$
green, potato $= \frac{1}{16} \times 144 = 9$

But imagine that, among these 144 offspring, the results we actually observed were as follows:

purple, cut	86	green, cut	24
purple, potato	26	green, potato	8

We then calculate the difference between each set of results, and square each difference. (Squaring gets rid of any minus signs – it is irrelevant whether the differences are negative or positive.) Then we divide each squared difference by the expected value, and add up all of these answers:

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

where: Σ = sum of
O = observed value
E = expected value

Phenotypes of plants	purple stems, cut leaves	purple stems, potato leaves	green stems, cut leaves	green stems, potato leaves
Observed number (O)	86	26	24	8
Expected ratio	9 :	3 :	3 :	1
Expected number (E)	81	27	27	9
$O - E$	+5	−1	−3	−1
$(O - E)^2$	25	1	9	1
$(O - E)^2/E$	0.31	0.04	0.33	0.11
$\chi^2 = \sum \frac{(O-E)^2}{E} = 0.79$				

The value of χ2 is determined by examining a table relating χ2 values to probabilities. The table provides probabilities that the differences between expected and observed results are due to chance. A probability of 0.05

or larger indicates that the differences are not significant. If the probability is smaller than 0.05, it is likely that the difference is significant and the assumptions about the experiment need to be reconsidered. The number of degrees of freedom in the results is also considered. The table shows a χ2 value of 7.82, representing a probability of 0.05, which is much smaller than the calculated value of 0.79. This indicates that the difference between observed and expected results is almost certainly due to chance, and there is no significant difference between what was expected and what was actually obtained.

Degrees of freedom	Probability greater than			
	0.1	0.05	0.01	0.001
1	2.71	3.84	6.64	10.83
2	4.60	5.99	9.21	13.82
3	6.25	7.82	11.34	16.27
4	7.78	9.49	13.28	18.46

Table 16.3 Table of χ^2 values.

Mutations

Genes have various alleles, each with a different sequence of bases, resulting from mutations. Mutations are unpredictable changes in an organism's genetic material, such as a change in DNA structure or chromosome aberrations. Environmental factors like ionizing radiation, ultraviolet radiation, and chemicals like mustard gas can increase the chances of mutations. A substance that increases the chances of mutation is a mutagen.

In gene mutations, there are three different ways in which the sequence of bases in a gene may be altered. These are:

- base substitution, where one base simply takes the place of another; for example, CCT GAG GAG may change to CCT GTG GAG
- base addition, where one or more extra bases are added to the sequence; for example, CCT GAG GAG may change to CCA TGA GGA G
- base deletion, where one or more bases are lost from the sequence; for example, CCT GAG GAG may change to CCG AGG AG.

Sickle cell anaemia

One example of a base substitution that has a significant effect on the phenotype is the one involved in the inherited blood disorder, sickle cell anaemia. (We have already looked at the difference between the HbA and HbS alleles on page 374 and the inheritance of this disease on pages 374–376.)

You will remember that the gene that codes for the amino acid sequence in the β-globin polypeptide is not the same in everyone. In most people, the β-globin polypeptide begins with the amino acid sequence coded from the Hb^A allele:

Val-His-Leu-Thr-Pro-Glu-Glu-Lys-

But in people with the Hb^S allele, the base sequence CTT is replaced by CAT, and the amino acid sequence becomes:

Val-His-Leu-Thr-Pro-Val-Glu-Lys-

Albinism

Albinism provides an example of the relationship between a gene, an enzyme and a human phenotype. In albinism, the dark pigment melanin is totally or partially missing from the eyes, skin and hair. In humans this results in pale blue or pink irises in the eyes and very pale skin and hair (Figure 16.22).

A mutation in the gene for the enzyme tyrosinase results in either the absence of tyrosinase or the presence of inactive tyrosinase in the cells responsible for melanin production. In these melanocytes, the first two steps of the conversion of the amino acid, tyrosine into melanin cannot take place. Tyrosine cannot be converted into DOPA and dopaquinone.

Figure 16.22 An albino boy with his classmates in South Africa.

$$\text{tyrosine} \xrightarrow{\text{tyrosinase}} \text{DOPA} \rightarrow \text{dopaquinone} \rightarrow \text{melanin}$$

Huntington's disease

So far, in the examples of the inheritance of human conditions, the mutations have been inherited as recessive alleles. Huntington's disease (HD) provides an example of a mutation that is inherited as a dominant allele. T his means that most people with the condition are heterozygous and have a 1 in 2 chance of passing on the condition to a child.

Gene control in prokaryotes

To understand how gene expression in bacteria is controlled, you must distinguish between structural genes and regulatory genes.

■ ■ Genes that code for proteins required by a cell are called structural genes. Such proteins may literally form part of a cellular structure, but they may also have some other role, such as acting as an enzyme.

■ Genes that code for proteins that regulate the expression of other genes are called regulatory genes. You must also distinguish between repressible and inducible enzymes.

■ ■ ■ The synthesis of a repressible enzyme can be prevented by binding a repressor protein to a specific site, called an operator, on a bacterium's DNA.

■ ■ The synthesis of an inducible enzyme occurs only when its substrate is present. Transcription of the gene occurs as a result of the inducer (the enzyme's substrate) interacting with the protein produced by the regulatory gene.

The lac operon

The enzyme β-galactosidase hydrolyses the disaccharide lactose to the monosaccharides glucose and galactose. In the bacterium, Escherichia coli, the number of molecules of this enzyme present in a bacterial cell varies according to the concentration of lactose in the medium in which the bacterium is growing

The *lac* operon consists of a cluster of three structural genes and a length of DNA including operator and promoter regions. The three structural genes are:

- *lacZ*, coding for β-galactosidase
- *lacY*, coding for permease (which allows lactose to enter the cell)
- *lacA*, coding for transacetylase.

Close to the promoter, but not actually part of the operon, is its regulatory gene (Figure 16.23). T he sequence of events when there is no lactose in the medium in which the bacterium is growing is as follows:

■ ■ the regulatory gene codes for a protein called a repressor

■ ■ the repressor binds to the operator region, close to the gene for β-galactosidase

■ ■ in the presence of bound repressor at the operator, RNA polymerase cannot bind to DNA at the promoter region

■ ■ no transcription of the three structural genes can take place.

When lactose is present in the medium in which the bacterium is growing:

■ ■ lactose is taken up by the bacterium

■ ■ lactose binds to the repressor protein, distorting its shape and preventing it from binding to DNA at the operator site

■ ■ transcription is no longer inhibited and messenger RNA is produced from the three structural genes. The genes have been switched on and are transcribed together (Figure 16.23).

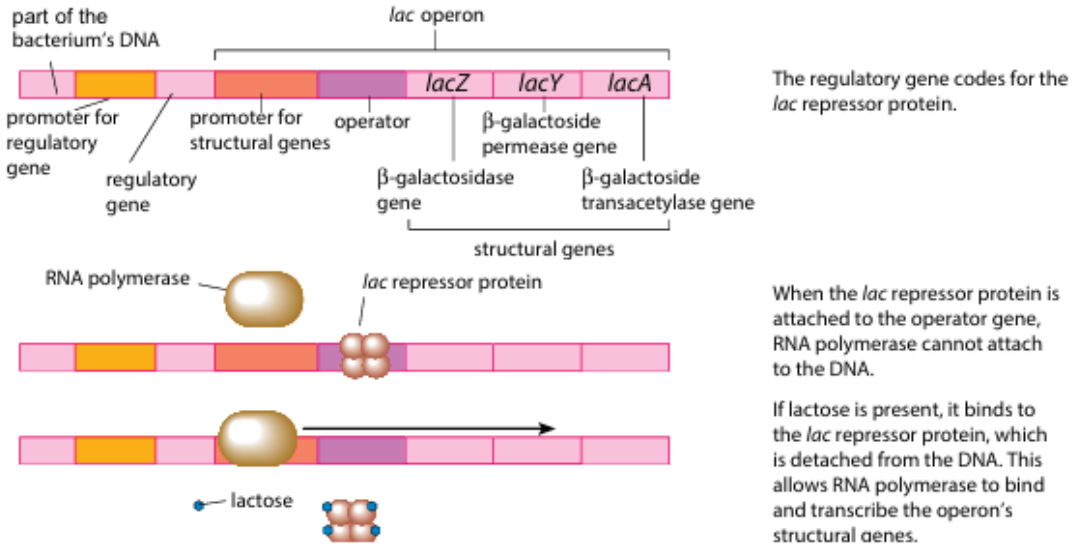


Figure 16.23 Regulation of gene expression by the *lac* operon.

Gene control in eukaryotes

In general, the number of different proteins that act as transcription factors increases with increasing size of the genome

Effects of transcription factors include the following.

■ ■ General transcription factors are necessary for transcription to occur. Th ey form part of the protein complex that binds to the promoter region of the gene concerned.

■ Other factors activate appropriate genes in sequence, allowing the correct pattern of development of body regions

■ A transcription factor is responsible for the determination of sex in mammals.

■ ■ Transcription factors allow responses to environmental stimuli, such as switching on the correct genes to respond to high environmental temperatures.

■ ■ Some transcription factors, including the products of proto-oncogenes and tumour suppressor genes, regulate the cell cycle, growth and apoptosis (programmed cell death) (Chapter 5).

■ ■ Hormones have their effect through transcription factors.

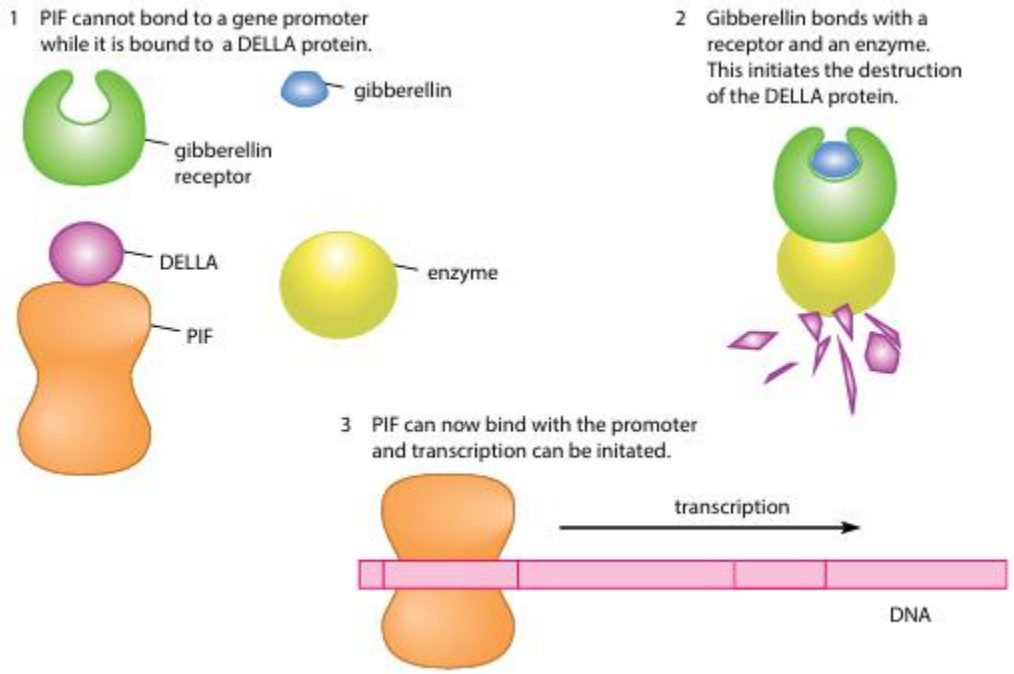


Figure 16.24 How gibberellin controls gene transcription.

Chapter 17: Selection and evolution

Variation In Chapter 16, you have seen how sexual reproduction produces genetic variation among the individuals in a population. Genetic variation is caused by:

■ ■ independent assortment of chromosomes, and therefore alleles, during meiosis

■ ■ crossing over between chromatids of homologous chromosomes during meiosis

■ ■ random mating between organisms within a species

■ ■ random fertilisation of gametes

■ ■ mutation.

The first four of these processes reshuffle existing alleles in the population. Offspring have combinations of alleles which differ from those of their parents and from each other. This genetic variation produces **phenotypic variation.**

This is probably how the sickle cell allele of the gene for the production of the β-globin polypeptide first arose. Such a change in a gene, which is quite unpredictable, is called a **gene mutation.**

Variation in phenotype is also caused by the **environment** in which organisms live.

Continuous and discontinuous variation

Qualitative differences fall into clearly distinguishable categories, with no intermediates – for example, you have one of four possible ABO blood groups: A, B, AB or O. This is **discontinuous variation**

When the heights of a large number of people are measured, there are no distinguishable height classes. Instead there is a range of heights between two extremes (Figure 17.2). This is **continuous variation.**

The genetic basis of continuous and discontinuous variation

Both qualitative and quantitative differences in phenotype may be inherited. Both may involve several different genes. However, there are important differences between them.

In discontinuous (qualitative) variation:

■ ■ different alleles at a single gene locus have large effects on the phenotype

■ ■ different genes have quite different effects on the phenotype

In continuous (quantitative) variation:

■ ■ different alleles at a single gene locus have small effects on the phenotype

■ ■ different genes have the same, often additive, effect on the phenotype

■ ■ a large number of genes may have a combined effect on a particular phenotypic trait; these genes are known as polygenes.

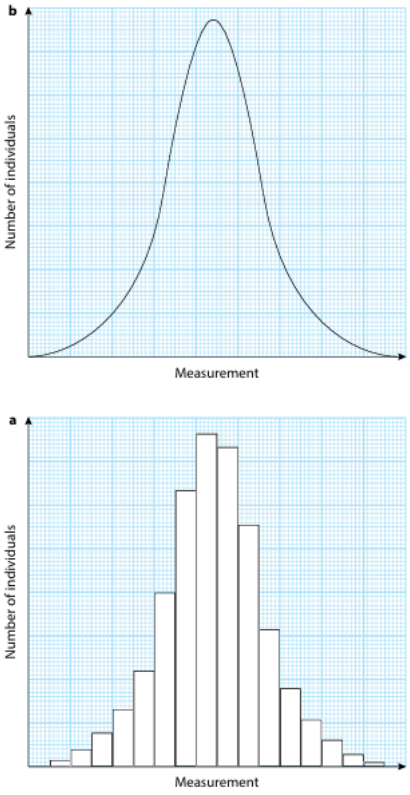


Figure 17.2 **a** Distribution curve and **b** a histogram, showing continuous variation.

Suppose that the height of an organism is controlled by two unlinked (that is, on different chromosomes) genes: A/a and B/b. The recessive alleles of both genes (a and b) each contribute x cm to the height of the organism. The dominant alleles (A and B) each add 2x cm. Since the effect of such genes is additive, the homozygote recessive (aabb) is therefore potentially 4x cm tall and the homozygote dominant (AABB) is potentially 8x cm tall. The other genotypes will fall between these extremes

Parental phenotypes	4*x* cm tall	8*x* cm tall
Parental genotypes	**aabb**	**AABB**
Gametes	(ab)	(AB)
Offspring genotypes	all **AaBb**	
Offspring phenotypes	all 6*x* cm tall	

Interbreeding these potentially 6x cm tall offspring gives all possible genotypes and phenotypes among the 16 possibilities.

Parental phenotypes	6*x* cm tall	6*x* cm tall
Parental genotypes	**AaBb**	**AaBb**
Gametes	(AB) or (Ab) or (aB) or (ab)	(AB) or (Ab) or (aB) or (ab)
	in equal proportions	

continued ...

The number of offspring and their potential heights according to their genotypes are summarised in the histogram in Figure 17.3. These results fall approximately on a normal distribution curve.

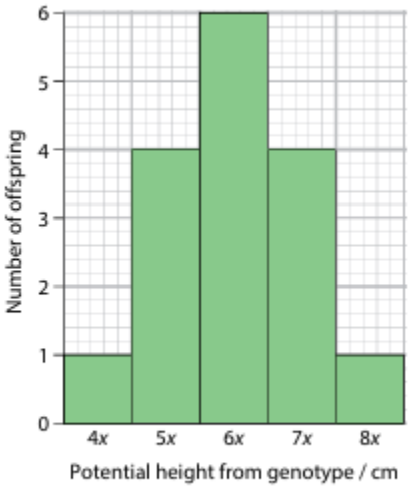


Figure 17.3 The additive effect of alleles.

Offspring genotypes and phenotypes:

Gametes from other parent	Gametes from one parent: AB	Ab	aB	ab
AB	AABB 8x cm	AABb 7x cm	AaBB 7x cm	AaBb 6x cm
Ab	AABb 7x cm	AAbb 6x cm	AaBb 6x cm	Aabb 5x cm
aB	AaBB 7x cm	AaBb 6x cm	aaBB 6x cm	aaBb 5x cm
ab	AaBb 6x cm	Aabb 5x cm	aaBb 5x cm	aabb 4x cm

Figure 17.4 In Siamese cats, dark hair develops where the skin is at a relatively low temperature.

Environmental effects on the phenotype

In our hypothetical example of continuous variation just given, the heights shown are those that would be expected from the genotype alone. If you were able to take a number of individuals, all with the same genotypic contribution to height, it would be most unlikely that their heights would be exactly the same when measured. Environmental effects may allow the full genetic potential height to be reached or may stunt it in some way.

Parental varieties were homozygous at many loci, with the first generation being genetically different but genetically identical. The second generation showed wider variation in cob length, both genetic and environmental. The t-test can compare the variation between pure-bred Black Mexican and Tom Thumb maize plants. Selective breeding should consider both genetic and environmental variation, as environmental variation is not relevant.

Cob length / cm	5	6	7	8	9	10	11	12	13	14	15	16	17	18	19	20	21
Number of Black Mexican parent cobs									3	11	12	14	26	15	10	7	2
Number of Tom Thumb parent cobs	4	21	24	8													
Number of F1 offspring cobs					1	12	12	14	17	9	4						
Number of F2 offspring cobs			1	10	19	26	47	73	68	68	39	25	15	9	1		

Table 17.1 Variation in cob length of two parental varieties of maize and of the first (F1) and second (F2) generations of a cross between them.

Natural selection

All organisms have the reproductive potential to increase their populations. Rabbits, for example, produce several young in a litter, and each female may produce several litters each year. If all the young rabbits survived to adulthood and reproduced, then the rabbit population would increase rapidly. Figure 17.5 shows what might happen.

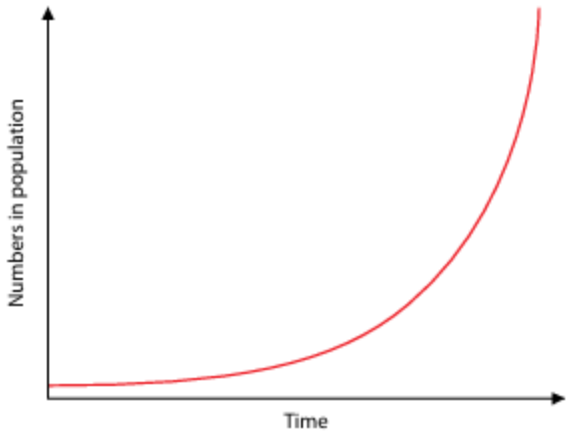


Figure 17.5 If left unchecked by environmental factors, numbers in a population may increase exponentially.

In the 19th century, rabbit populations in Australia experienced a population explosion due to the release of 12 pairs of rabbits from Britain in 1859. These rabbits, which feed on low-growing vegetation, soared due to their abundance and the few predators they ate. This led to a significant impact on sheep grazing. However, such population explosions are rare and are typically managed by environmental factors such as predation, competition, and pathogen infection. Overcrowding can also increase the risk of diseases like myxomatosis, which is transmitted by fleas.

Figure 17.6 Attempts to control the rabbit population explosion in Australia in the mid to late 19th century included 'rabbit drives', in which huge numbers were rounded up and killed. Eventually, myxomatosis brought numbers down.

T hese environmental factors act to reduce the rate of growth of the rabbit population. Of all the rabbits born, many will die from lack of food, or be killed by predators, or die from myxomatosis. Only a small proportion of young will grow to adulthood and reproduce, so population growth slows.

Environmental factors can significantly decrease population size, leading to oscillations in lemming populations. The number of young produced is greater than the number that survives to adulthood, with many dying before reaching reproductive age. Variation within a rabbit population, such as coat color, can give some an advantage in the struggle for survival. Some rabbits may be homozygous for the recessive allele that gives white coat, making them less likely to survive. Fitness refers to an organism's capacity to survive and transmit its genotype to its offspring.

Figure 17.7 Rabbits living in dense populations are more likely to get myxomatosis than those in less crowded conditions.

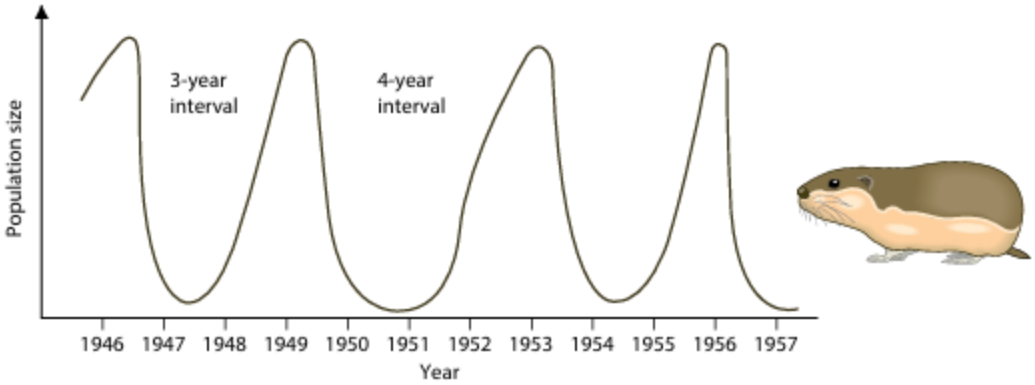


Figure 17.8 Lemming populations are famous for their large increases and decreases. In some years, populations become so large that lemmings may emigrate in one group from overcrowded areas. The reason for the oscillating population size is not known for certain, although it has been suggested that food supply or food quality may be the main cause. As the population size rises, food supplies run out, so the population size 'crashes'. Once the population size has decreased, food supplies begin to recover, and the population size rises again.

Environmental factors can decrease population size, leading to lemming populations. Variations in rabbit populations, such as coat color, can give some an advantage in survival. Fitness refers to an organism's capacity to survive and transmit its genotype.

Evolution

However, if a **new environmental factor or a new allele appears**, then allele frequencies may also change. This is called directional selection (Figure 17.9c). A third type of selection, called disruptive selection, can occur when conditions favour both extremes of a population. This type of selection maintains different phenotypes (polymorphism) in a population (Figure 17.9d).

Figure 17.10 The tuatara, *Sphenodon punctatus*, is a lizard-like reptile that lives in New Zealand. Fossils of a virtually identical animal have been found in rocks 200 million years old. Natural selection has acted to keep the features of this organism the same over all this time.

Figure 17.11 The white winter coat of a mountain hare provides excellent camouflage from predators when viewed against snow.

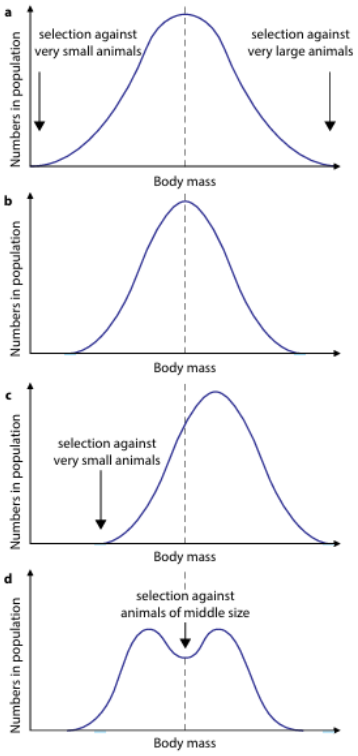


Figure 17.9 If a characteristic in a population, such as body mass, shows wide variation, selection pressures often act against the two extremes (graph **a**). Very small or very large individuals are less likely to survive and reproduce than those whose size lies nearer the centre of the range. This results in a population with a narrower range of body size (graph **b**). This type of selection, which tends to keep the variation in a characteristic centred around the same mean value, is called **stabilising selection**. Graph **c** shows what would happen if selection acted against smaller individuals but not larger ones. In this case, the range of variation shifts towards larger size. This type of selection, which results in a change in a characteristic in a particular direction, is called **directional selection**. Graph **d** shows the result of selection that favours both large and small individuals, but acts against those whose size is in the middle of the range. This is **disruptive selection**.

A new environmental factor

Imagine that we are plunged into a new Ice Age. The climate becomes much colder, so that snow covers the ground for almost all of the year. Assuming that rabbits can cope with these conditions, white rabbits now have a selective advantage during seasons when snow lies on the ground, as they are better camouflaged (like the hare in Figure 17.11).

A new allele

Because they are random events, most mutations that occur produce features that are harmful. That is, they produce organisms that are less well adapted to their environment than 'normal' organisms. Other mutations may be neutral, conferring neither an advantage nor a disadvantage on the organisms within which they occur. Occasionally, mutations may produce useful features.

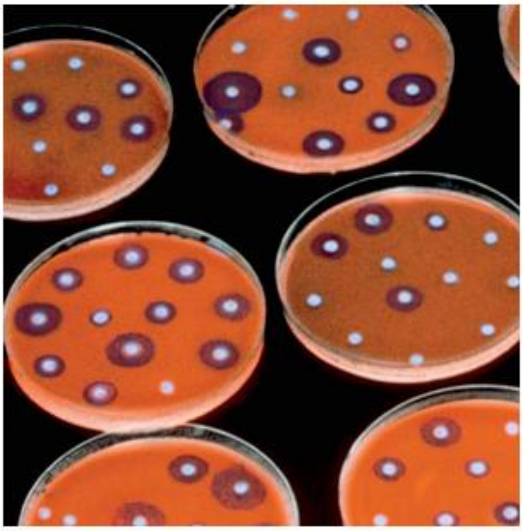


Figure 17.12 The red gel in each of these Petri dishes has been inoculated with bacteria. The small light blue circles are discs impregnated with antibiotics. Bacteria that are resistant to an antibiotic are able to grow right up to the disc containing it. See also Figure 10.16, page 215.

Antibiotic resistance

Antibiotics are chemicals produced by living organisms, which inhibit or kill bacteria but do not normally harm human tissue. Most antibiotics are produced by fungi. The first antibiotic to be discovered was penicillin, which was first used during the Second World War to treat a wide range of diseases caused by bacteria. Penicillin stops cell wall formation in bacteria, so preventing cell reproduction.

Bacteria possess a single DNA loop, allowing them to have a single copy of each gene. This results in a selective advantage, as bacteria with resistance to penicillin can survive and reproduce. This leads to antibiotic-resistant strains, which are constantly appearing due to the use of antibiotics. Alleles for antibiotic resistance often occur on plasmids, which can be transferred between bacteriums, increasing the selection pressure on bacteria to evolve resistance.

Industrial melanism

One well-documented case of the way in which changing environmental factors may produce changes in allele frequencies is that of the peppered moth, Biston betularia (Figure 17.13), in the UK and Ireland. This is a night f lying moth which spends the day resting underneath the branches of trees.

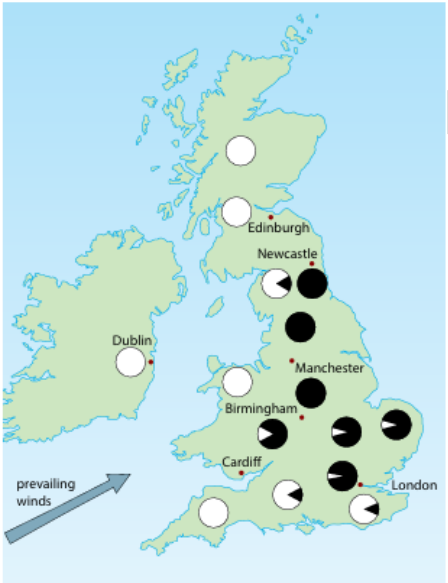


Figure 17.14 The distribution of the pale and dark forms of the peppered moth, *Biston betularia*, in the UK and Ireland during the early 1960s. The ratio of dark to pale areas in each circle shows the ratio of dark to pale moths in that part of the country.

Figure 17.13 Dark form of peppered moth on dark and pale tree bark.

The speckled and black moths are influenced by a single gene, with speckled moths producing black colors and speckled moths producing normal colors. The frequency of the speckled allele increased in industrial areas until the late 1960s due to predation by birds. In unpolluted areas, speckled moths are better camouflaged by lichens, but in polluted areas, dark moths have a selective advantage. As air pollution from industries reduces, the speckled moths' proportion will increase. Mutations to the C allele have likely always occurred in B. betularia populations, and changes in environmental factors only affect the likelihood of an allele surviving in a population.

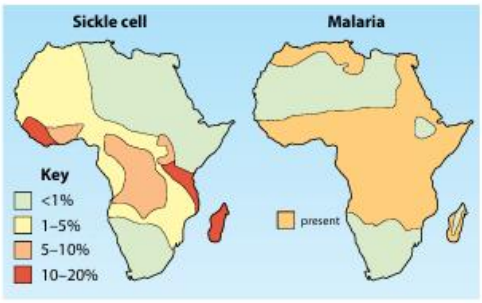


Figure 17.15 The distribution of people with at least one copy of the sickle cell allele, and the distribution of malaria, in Africa.

Sickle cell anaemia

Sickle cell anaemia is a severe form of anemia caused by an allele, HbS, of the gene that codes for the production of the β-globin polypeptide. People with this allele are less likely to survive and reproduce, and until recently, almost everyone with sickle cell anaemia died before reaching reproductive age. The frequency of this allele is high in some parts of the world, where malaria is found, causing illness and death.

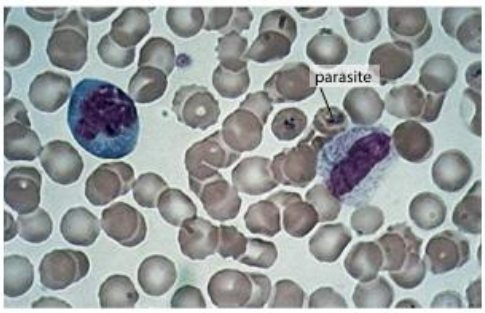


Figure 17.16 Red blood cells infected with malarial parasite. Some cells have multiple parasites.

There are, therefore, two strong selection pressures acting on these two alleles.

- Selection against people who are homozygous for the sickle cell allele, Hb^SHb^S, is very strong, because they become seriously anaemic.
- Selection against people who are homozygous Hb^AHb^A, is also very strong, because they are more likely to die from malaria.

In areas where malaria is common, heterozygotes, Hb^AHb^S, have a strong selective advantage; they do not suffer from sickle cell anaemia and are much less likely to suffer badly from malaria. So both alleles remain in populations where malaria is an important environmental factor.

In places where malaria was never present, selection against people with the genotype Hb^SHb^S has almost completely removed the Hb^S allele from the population.

The examples of natural selection given above show the effect of a **non-random** process on the allele frequencies of a population of organisms. These allele frequencies may also change thanks to a **random** process called genetic drift.

Genetic drift

Genetic drift is a change in allele frequency that occurs by chance, because only some of the organisms of each generation reproduce. It is most noticeable when a small number of individuals are separated from the rest of a large population.

The Hardy–Weinberg principle

When a particular phenotypic trait is controlled by two alleles of a single gene, A/a, the population will be made up of three genotypes: AA, Aa and aa. Calculations based on the Hardy–Weinberg principle allow the proportions of each of these genotypes in a large, randomly mating population to be calculated

We use the letter p to represent the frequency of the dominant allele, **A**, in the population and the letter q to represent the frequency of the recessive allele, **a**. Then, since there are only two alleles of this gene:

$p + q = 1$ **(Equation 1)**

- the chance of an offspring inheriting a dominant allele from both parents = $p \times p = p^2$
- the chance of an offspring inheriting a recessive allele from both parents = $q \times q = q^2$
- the chance of an offspring inheriting a dominant allele from the father and a recessive allele from the mother = $p \times q = pq$
- the chance of an offspring inheriting a dominant allele from the mother and a recessive allele from the father = $p \times q = pq$

So, $p^2 + 2pq + q^2 = 1$ **(Equation 2)**

T hese Hardy–Weinberg calculations do not apply when the population is small or when there is:

■ ■ significant selective pressure against one of the genotypes

■ ■ migration of individuals carrying one of the two alleles into, or out of, the population

■ ■ non-random mating.

Artificial selection

Selective breeding of dairy cattle Sometimes, the most important selection pressures on organisms are those applied by humans. When humans purposefully apply selection pressures to populations, the process is known as artificial selection

For thousands of years, people have sought to improve cattle by selective breeding. This involves selecting individuals with desired traits such as docility, fast growth rates, and high milk yields. These traits are passed on to their offspring, and over generations, the frequency of these traits increases. However, selective breeding presents challenges, such as large animals, long gestation periods, and small offspring production. To assess milk production, progeny testing is used to evaluate the bull's value. Selective breeders must consider the entire genotype, including background genes that adapt the organism to its environment.

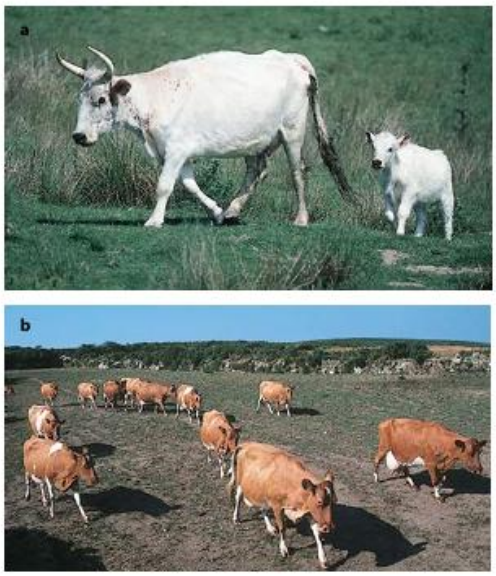


Figure 17.17 The original wild cattle from which individuals were first domesticated are thought to have looked very much like **a**, the modern Chillingham White breed. Selective breeding over many centuries has produced many different breeds, such as **b**, the Guernsey. Guernseys have been bred for the production of large quantities of fat-rich milk. Notice the large udder compared with the Chillingham.

Crop improvement

The same problem is seen when a cross is made between a cultivated plant and a related wild species. Although most species will not breed with a different species, some can be interbred to give fertile offspring. Such species are often those that do not normally come into contact with one another, because they live in different habitats or areas. The wild parent will have alleles that are not wanted and which have probably been selected out of the cultivated parent.

Wheat plants now have much shorter stems than they did only 50 years ago. This makes them easier to harvest and means they have higher yields (because they put more energy into making seeds rather than growing tall) (Figure 17.20). The shorter stems also make the plants less susceptible to being knocked flat by heavy rains, and means they produce less straw, which has little value and costs money to dispose of.

Dwarf wheat varieties carry mutant alleles of reduced height genes, which produce DELLA proteins that reduce gibberellins' effect on growth. This causes dwarfism. Rice, Oryza sativa, is also subject to selective breeding. The International Rice Research Institute and Global Rice Science Partnership aim to improve rice farmers' ability to feed growing populations. Researchers aim to produce rice varieties with resistance to bacterial and fungal diseases using selective breeding.

Figure 17.18 Wheat was first farmed in the 'fertile crescent' (shown in green) around 10 000 years ago.

Figure 17.19 Wheat breeders are attempting to produce new varieties of wheat that will be able to grow in the higher temperatures that global warming is expected to bring.

Figure 17.20 Harvesting wheat.

Inbreeding and hybridisation in maize

Maize, also known as corn, is a tall grass with broad, strap-shaped leaves. It grows best in hot summers and is grown in Central and South America, Africa, Europe, America, Australia, New Zealand, China, and Indonesia. Inbreeding causes inbreeding depression, while outbreeding produces healthier, taller, and higher yielding heterozygous plants.

The Darwin–Wallace theory of evolution by natural selection

The original theory that natural selection might be a mechanism by which evolution could occur was put forward independently by both Charles Darwin and Alfred Russel Wallace in 1856.

Figure 17.21 Maize plants in flower.

Observation 1	Organisms produce more offspring than are needed to replace the parents.
Observation 2	Natural populations tend to remain stable in size over long periods.
Deduction 1	There is competition for survival (a 'struggle for existence').
Observation 3	There is variation among the individuals of a given species.
Deduction 2	The best adapted variants will be selected for by the natural conditions operating at the time. In other words, natural selection will occur. The 'best' variants have a selective advantage; 'survival of the fittest' occurs.

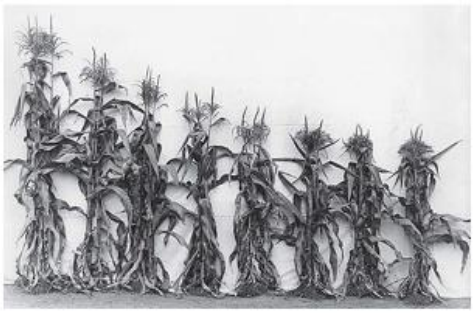


Figure 17.22 The effects of inbreeding depression in maize over eight generations.

Species and speciation

This chapter discusses the concept of natural selection, which is responsible for producing all species of organisms on Earth. However, no new species have been produced in examples of directional selection, such as antibiotic resistance in bacteria and changes in wing color in peppered moths. A species is defined as a group of organisms with similar morphological, physiological, biochemical, and behavioral features, which can interbreed to produce fertile offspring and are reproductively isolated from other species.

When deciding whether two organisms belong to the same species or two different species, biologists often rely on morphological, biochemical, physiological, and behavioral differences. However, it is difficult to determine when these features are sufficiently similar or different to define two organisms as belonging to the same or different species.

Despite these challenges, many biologists agree that the feature that determines whether two organisms belong to different species is their inability to interbreed successfully. In explaining how natural selection can produce new species, it is crucial to consider how a group of interbreeding organisms can produce another group of organisms that cannot interbreed successfully with the first group.

Reproductive isolation can take very different forms. Prezygotic (before a zygote is formed) isolating mechanisms include:

■ ■ individuals not recognising one another as potential mates or not responding to mating behaviour

■ ■ animals being physically unable to mate

■ ■ incompatibility of pollen and stigma in plants

■ ■ inability of a male gamete to fuse with a female gamete.

Postzygotic isolating mechanisms include:

■ ■ failure of cell division in the zygote

■ ■ non-viable offspring (offspring that soon die)

■ ■ viable, but sterile offspring.

Allopatric speciation

Geographical isolation has significantly influenced the evolution of many species, as seen in the unique groups of animals and plants found on islands like the Hawaiian and Galapagos. Geographical isolation requires a barrier to prevent mixing between populations of the same species, such as a stretch of water. This barrier can be physical, such as cutting down dense forests or creating small, immobile organisms. This is known as allopatric speciation, where two populations are separated geographically. However, sympatric speciation allows new species to arise without geographical barriers, demonstrating the importance of geographical isolation in species evolution.

Figure 17.23 *Hibiscus clayi* is found only on Hawaii, where it is in danger of extinction.

Sympatric speciation

Perhaps the commonest way in which sympatric speciation can occur is through polyploidy. A polyploid organism is one with more than two complete sets of chromosomes in its cells. This can happen if, for example, meiosis goes wrong when gametes are being formed, so that a gamete ends up with two sets of chromosomes instead of one set. If two such gametes fuse, then the zygote gets four complete sets of chromosomes. It is said to be tetraploid.

One well-documented instance of speciation through allopolyploidy is the cord grass Spartina anglica. This is a vigorous grass that grows in salt marshes. Before 1830, the species of Spartina that grew in these places in England was S. maritima. Then, in 1829, a different species called S. alterniflora was imported from America (Figure 17.24). S. maritima and S. alterniflora hybridised (interbred), producing a new species called S. townsendii (Figure 17.25). This is a diploid plant, with one set of chromosomes from S. maritima and one set from S. alterniflora. It is sterile, because the two sets of chromosomes from its parents cannot pair up, so it cannot undergo meiosis successfully. Nor can S. townsendii interbreed with either of its two parents, which is what makes it a different species. Although it is sterile, it has been able to spread rapidly, reproducing asexually by producing long underground stems called rhizomes, from which new plants can grow.

Figure 17.24 *Spartina alterniflora.*

Figure 17.25 *Spartina townsendii.*

Molecular comparisons between species

Molecular evidence from comparisons of the amino acid sequences of proteins and of the nucleotide sequences of mitochondrial DNA can be used to reveal similarities between related species.

Comparing amino acid sequences of proteins

As you saw in Chapter 16, changing a single amino acid in the primary structure of a protein may cause a dramatic change in its structure and function.

When the sequences of cytochrome c from humans, mice and rats were compared, it was found that:

■ ■ all three molecules consist of 104 amino acids

■ ■ the sequences of mouse and rat cytochromes c are identical

■ nine amino acids in human cytochrome c are different from the mouse or rat sequence

■ ■ most of these substitutions in human cytochrome c are of amino acids with the same type of R group (Appendix 1).

Comparing nucleotide sequences of mitochondrial DNA

Mitochondrial DNA sequences can be used to study the origin and spread of Homo sapiens. Human mitochondrial DNA is inherited through the female line and mutates faster than nuclear DNA. Differences in mitochondrial DNA sequences provide evidence for the origin of Homo sapiens in Africa and subsequent migrations. The "molecular clock" hypothesis suggests that all modern humans are descendants of one woman, Mitochondrial Eve, who lived in Africa between 150,000 and 200,000 years ago.

Extinctions

Species may become extinct, perhaps as a result of a change in climate or increased competition from a better adapted species. T he International Union for Conservation of Nature (IUCN) annually publishes a Red List of threatened species. T he 2013 list contained 21 286 species. (Visit the IUCN website at www.iucn.org and search for Red List.) T he species in the Red List are all under threat of extinction – of disappearing forever from the Earth. Of course, millions of species have become extinct in the past, sometimes huge numbers at one time in so-called mass extinctions

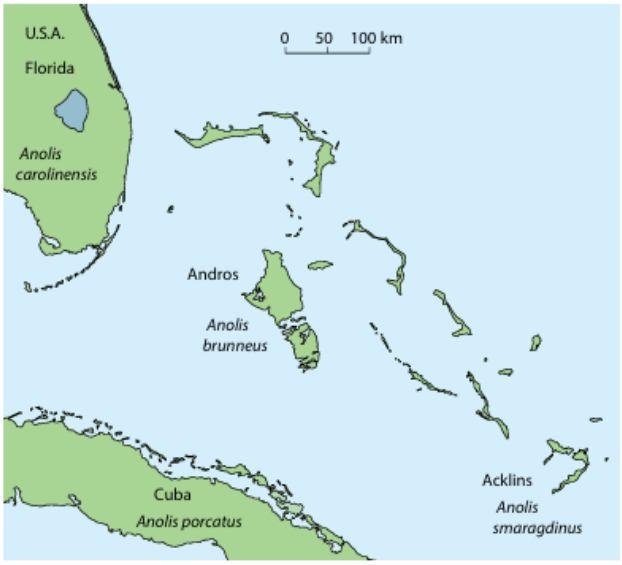


Figure 17.26 Anole lizard species in the Caribbean.

	A. brunneus			
A. brunneus		*A. smaragdinus*		
A. smaragdinus	12.1		*A. carolinensis*	
A. carolinensis	16.7	15.0		*A. porcatus*
A. porcatus	11.3	8.9	13.2	

Table 17.2 The results of comparing part of the mitochondrial DNA of four of the species of anole lizards. The smaller the number, the smaller the differences between the base sequences of the two species.

We are currently facing the likelihood of another mass extinction, this time caused by us. The main reason for this is the loss of habitats

Some endangered species have a very high profile – for example, pandas, rhinos or tigers – and you will be able to find a great deal of information about them on the internet

The global tiger population is estimated to be around 5000 individuals, with poaching being a significant issue in India, where tiger products are believed to cure various ailments. The IUCN declared the western black rhino of Africa extinct in 2011, and the last Javan rhino in Vietnam was killed by poachers. The African southern white rhino is now flourishing.

Figure 17.27 Orang-utans live only in dense tropical forest in Borneo. Deforestation is threatening their survival.

These rhino extinctions, despite years of conservation efforts, are

the result of:

■ a lack of political support for conservation

■ an increasing demand for rhino horn

■ internationally organised criminal groups targeting rhinos

Figure 17.28 Kerry slugs are found only in the south west of Ireland.

Revision questions

1.(a)(i) Distinguish asexual reproduction from sexual reproduction

(ii) State two methods of asexual reproduction which occur in flowering plants.

(iii) State two methods of asexual reproduction which occur in animals

(b)(i) Define the term 'pollination'

(ii) How is pollination usually achieved?

(iii) Why is cross pollination preferable to self pollination?

2. The drawings below show two different types of flower.

(a)(i) State two differences, visible on the drawings, between the male parts of flowers A and B.

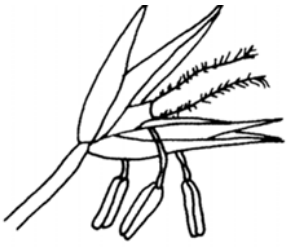
(ii) State two differences, visible on the drawings, between the female parts of flowers A and B.

b)(i) Flower A is radially symmetrical (actinomorphic). What does this mean and why is it of value to the flower?

(ii) What method of pollination is used by flower B?

(iii) State three features of flower B which help it to achieve cross pollination;

Flower A (half flower diagram)



Flower B (entire flower)

3. (a)There are 12 chromosomes in the somatic cells of the bean, Vicia.

(i) How many chromosomes does the offspring receive from the female parent via the egg cell nucleus?

(ii) How many chromosomes will be present in the primary endosperm nucleus?

(iii)A female sex chromosome is donated by X and a male sex chromosome by Y. Specify the sex chromosome content in the primary endosperm nucleus.

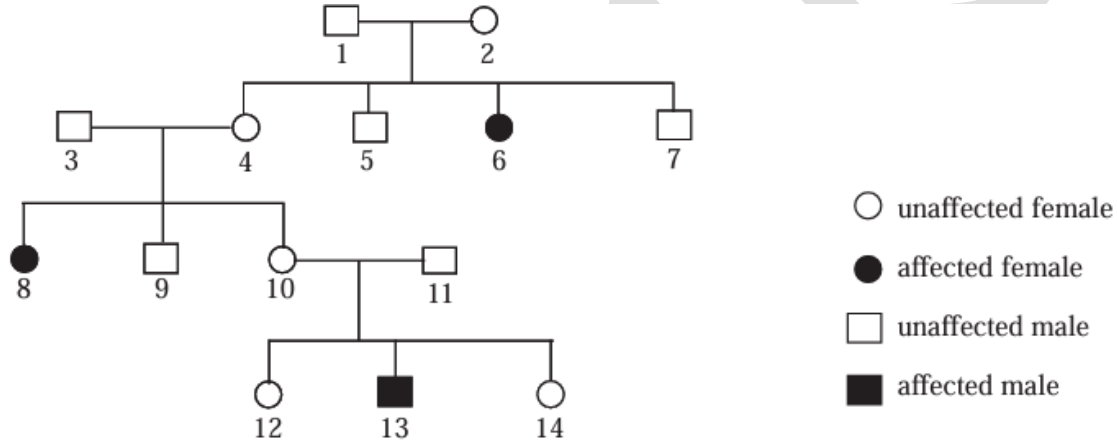
(b) The diploid chromosome number of a donkey is 66 chromosomes and that of a horse is 60 chromosomes.

(i) A mule is the offspring of a cross between a female horse and a male donkey. Calculate the number of chromosomes to be found in the somatic cells of the mule. Show your working.

(ii) Bivalent formation occurs in prophase 1 of meiosis. What is the theoretical maximum number of bivalents that could form in the mule?

(iii) Suggest why mules are infertile.

4. Alkaptonuria is a harmless, rare autosomal genetic defect in humans. The family tree below shows the pedigree of a family affected by alkaptonuria. (Individuals are numbered 1 to 14).



(a)(i)Is the condition dominant or recessive? Explain your answer.

(ii) State the numbers of all the individuals that are certain to be heterozygous for this gene.

(iii)What is the probability that individual 14 is heterozygous for this gene?

(b)Alkaptonuria occurs due to point mutation of the gene. State two other point mutation diseases of humans.

5. (a)Explain the meaning of each of the following terms.

(i) Species.

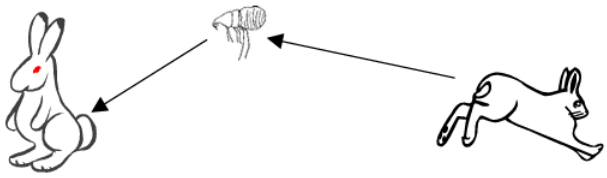
(ii) Isolating mechanism.

(b)Distinguish between each of the following pairs.

(i) Allopatric speciation and sympatric speciation.

(ii) Prezygotic reproductive isolation and postzygotic reproductive isolation

6.



Myxomatosis is a virus disease in rabbits which is transmitted from rabbit to rabbit by rabbit fleas. The virus causes blindness and then death. 12 rabbits were released in Australia in 1859. They reproduced rapidly and started to spread quickly, so that by 1886 they were advancing at about 66 miles per year. By 1905 they had spanned the continent. Rabbit proof fencing was inadequate to halt their progress.
In an attempt to control the huge rabbit population myxomatosis was introduced in 1950. It spread through the rabbit population resulting in a 99.9% mortality rate. Over the next few years, however, the rabbit population recovered although not to the pre-myxomatosis numbers. The mortality rate to myxomatosis in rabbits at present is around 40%.

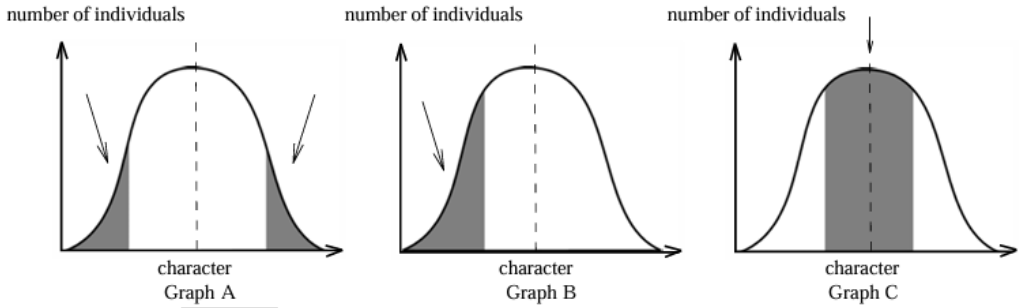
(a) Suggest two reasons for the rapid spread of rabbits across Australia between 1859 and 1905.

b.Suggest two reasons for the very high mortality rate in the rabbit population when myxomatosis was introduced in 1950.

(c)With reference to the genetic mechanisms involved explain the recovery of the rabbit population from the devastating effects of myxomatosis after its introduction in 1950.

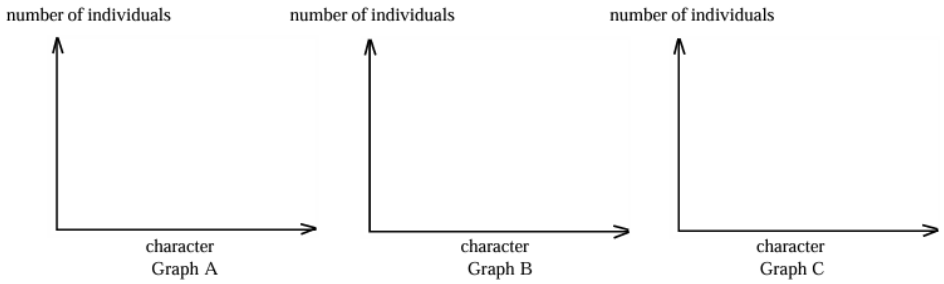
(d) Suggest why there is still a 40% mortality rate to myxomatosis in present day rabbit populations.

7. The graphs below show three types of natural selection. The shaded areas marked with arrows show the individuals in the population which are being selected against. The dotted vertical lines show the statistical means.



(a)What names are given to the types of selection shown in graphs A, B and C.

(b)Draw graphs on the three sets of axes below to show the distribution of phenotypes in A, B and C after the selection has operated for several generations. Indicate the new means with dotted lines.



(c)Describe one specific example of the type of selection shown in graph B, naming the organism and describing the character selected in your answer

8.

In the box below is a list of evolutionary terms, (i) to (x), and a jumbled list of definitions, A to J is provided underneath. Select the correct definition for each term by placing the appropriate letter in the appropriate box.

Evolutionary term		Letter
(i)	Isolation	
(ii)	Polymorphism	
(iii)	Deme	
(iv)	Adaptation	
(v)	Sympatric speciation	
(vi)	Analogy	
(vii)	Natural selection	
(viii)	Allopatric speciation	
(ix)	Industrial melanism	
(x)	Homology	

A. A local unit of population of a species in which there is random mating.

B. The restriction of interbreeding thus preventing gene flow between demes or species.

C. The production of species by isolation mechanisms operating on a gene pool in the same geographical region.

D. The presence of two or more distinct forms of a species found in the same locality at the same time.

E. The production of species by physical separation of the gene pool into different geographical regions thus restricting gene flow in the gene pool.

F. Structural similarity which results from similar specialisation in unrelated organisms.

G. Features evolve in an organism which enable it to cope better with its environment.

H. Structural similarity due to common ancestry.

I. Differential reproduction rates in nature leading to the increase in frequency of some genes and a decrease in frequency of other genes.

J. The selection of darkened phenotypes of a species due to industrial pollution.

[10]