Biology (A-level)

Inherited change (Chapter 16):

- **Homologous chromosomes** are a pair of chromosomes in a diploid cell that have the same structure as each other, with the same genes (but not necessarily the same alleles of those genes) at the same loci, and that pair together to form a bivalent during the first division of meiosis
- There are 22 matching chromosomes in humans (homologous chromosomes) – **autosomes** – and a non-matching pair labelled X and Y (**sex chromosomes**); two sets of 23 chromosomes – one set of 23 from the father and one set of 23 from the mother
- A **gene** is a length of DNA that codes for a particular protein or polypeptide
- An **allele** is a particular variety of a gene
- A **locus** is the position at which a particular gene is found on a particular chromosome; the same gene is always found at the same locus
- **Diploid cell:** one that possesses two complete sets of chromosomes; the abbreviation for diploid is 2n
- **Haploid cell:** one that possesses one complete set of chromosomes; the abbreviation for haploid is n
- Without halving the number of chromosomes into haploid gametes (meiosis – reduction division), it would double every generation

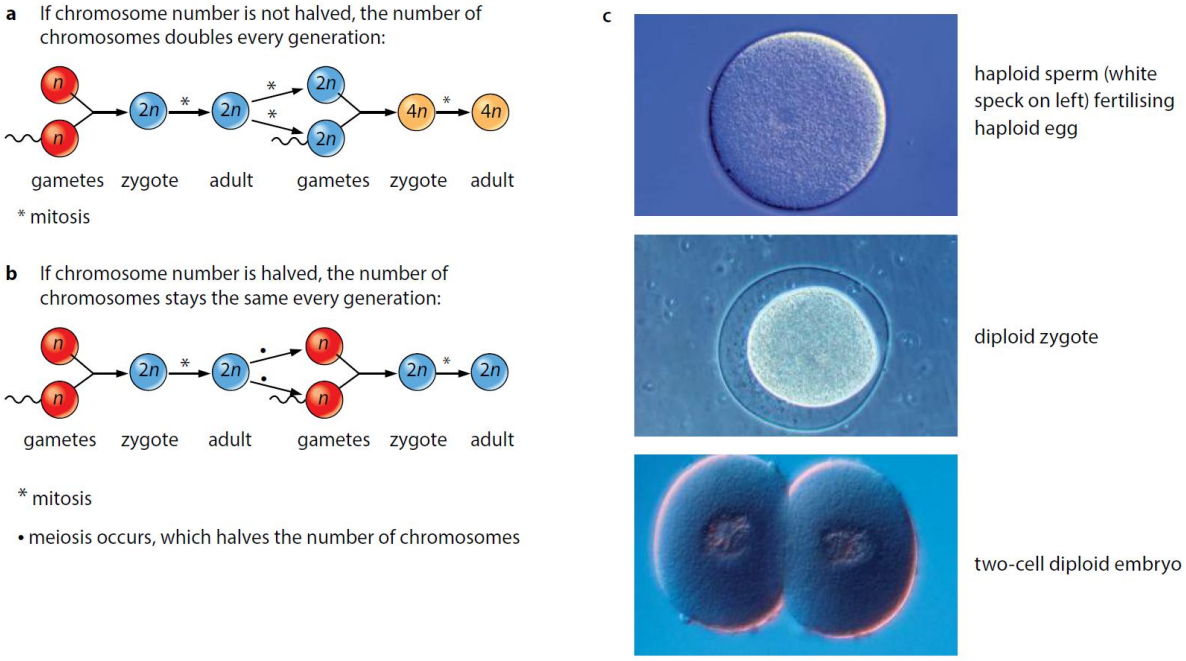


Figure 16.7 A life cycle in which the chromosome number is **a** not halved; **b** halved; **c** life cycle stages in a sea urchin .

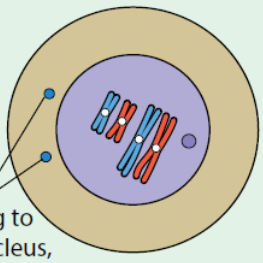
- Figure 16.8 shows an animal cell with 2*n* = 4; maternal and paternal chromosomes with different colours; remember that each centrosome contains a pair of centrioles; meiosis I is a reduction division, resulting in two daughter nuclei with half the number of chromosomes of the parent nucleus; in meiosis II, the chromosomes behave as in mitosis, so that each of the two haploid daughter nuclei divides again, resulting in a total of four haploid nuclei; these two events produce genetic variation between the daughter cells that are produced (independent assortment of homologous chromosomes and crossing over between the chromatids)

Meiosis I

1 Early prophase I
– as mitosis early prophase

2 Middle prophase I
Homologous chromosomes pair up. This process is called synapsis. Each pair is called a bivalent.

centrosomes moving to opposite ends of nucleus, as in mitosis

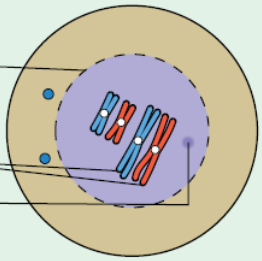


3 Late prophase I

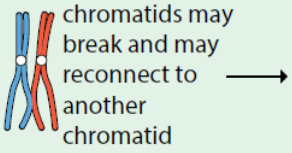
nuclear envelope breaks up as in mitosis

crossing over of chromatids may occur

nucleolus 'disappears' as in mitosis

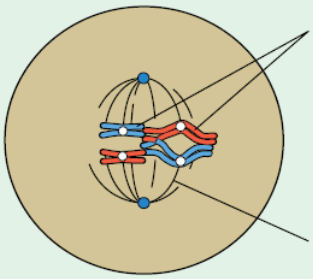


Bivalent showing crossing over:



At the end of prophase I a spindle is formed.

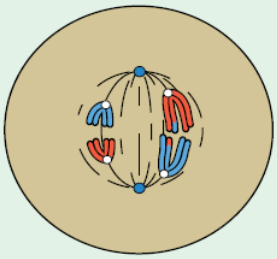
4 Metaphase I (showing crossing over of long chromatids)



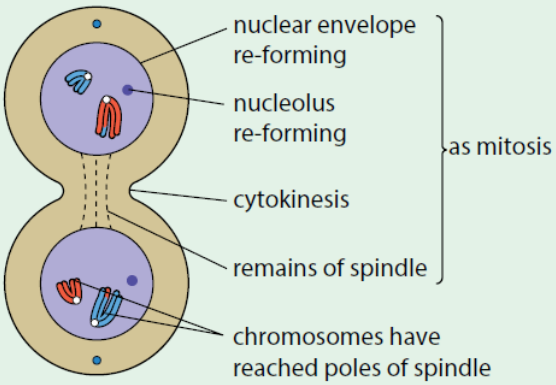
5 Anaphase I

Centromeres do not divide, unlike in mitosis.

Whole chromosomes move towards opposite ends of spindle, centromeres first, pulled by microtubules.



6 Telophase I



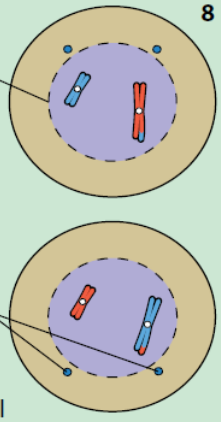
Animal cells usually divide before entering meiosis II. Many plant cells go straight into meiosis II with no reformation of nuclear envelopes or nucleoli. During meiosis II, chromatids separate as in mitosis.

Meiosis II

7 Prophase II

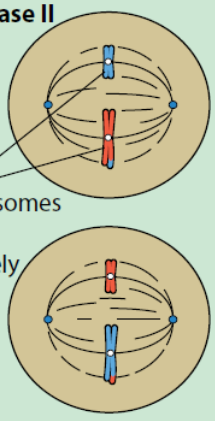
nuclear envelope and nucleolus disperse

centrosomes and centrioles replicate and move to opposite poles of the cell



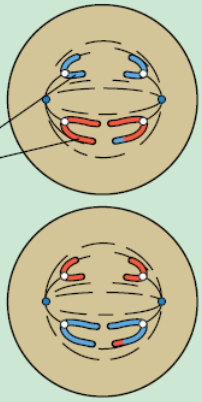
8 Metaphase II

chromosomes line up separately across equator of spindle

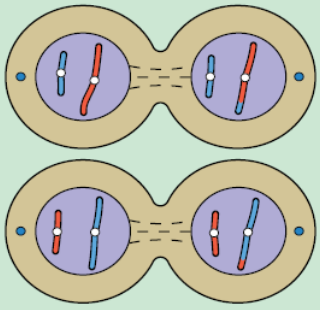


9 Anaphase II

centromeres divide and spindle microtubules pull the chromatids to opposite poles

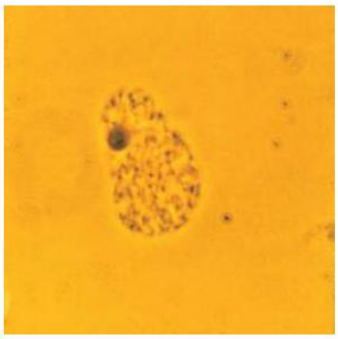


10 Telophase II

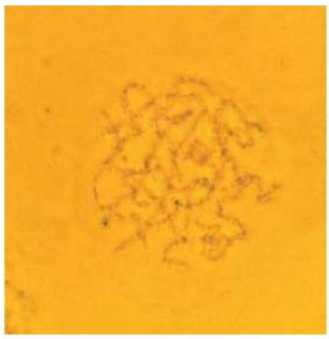


This is like telophase of mitosis, but in meiosis telophase II four haploid daughter cells are formed

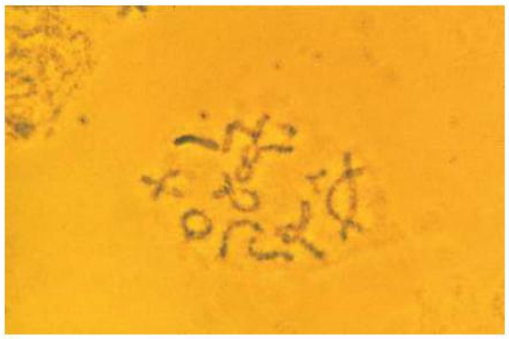
Figure 16.8 Meiosis and cytokinesis in an animal cell. Compare this process with nuclear division by mitosis, shown in Figure 5.7 (page 98).



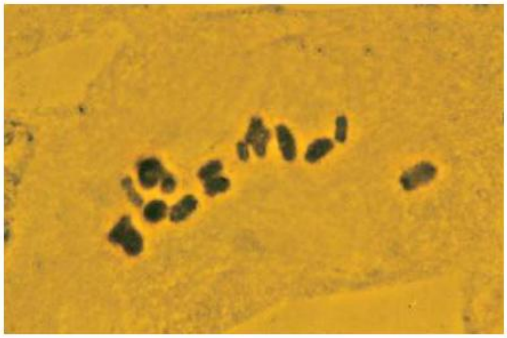
a interphase nucleus



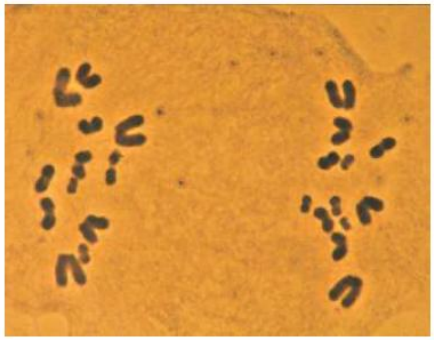
b meiosis I, early prophase I: chromosomes condensing and becoming visible



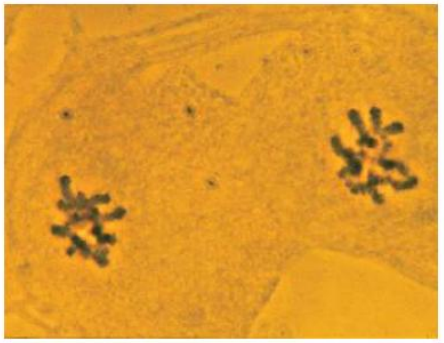
c prophase I: homologous chromosomes have paired up, forming bivalents, and crossing over of chromatids is occurring; members of each pair of chromosomes are repelling each other but are still held at the crossing-over points (chiasmata)



d metaphase I: bivalents line up across the equator of the spindle; the spindle is not visible in the photo; e anaphase I: homologous chromosomes move to opposite poles of the spindle



e anaphase I: homologous chromosomes move to opposite poles of the spindle

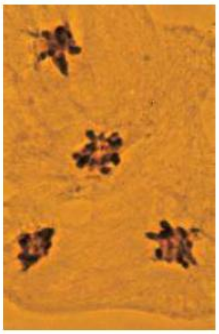


f telophase I and cytokinesis

g meiosis II, metaphase II: single chromosomes line up across the equator of a new spindle

h anaphase II: chromatids separate and move to opposite poles of the new spindle

i late anaphase II



j telophase II

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

- **Gametogenesis in humans:**
  - The formation of male gametes: **spermatogenesis** (testes)
  - The formation of female gametes: **oogenesis** (ovaries)
  - 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.
  - Ovum production takes place inside the ovaries, where diploid cells divide by mitosis to produce many **oogania** which begins to divide by meiosis but stops at

prophase I, **primary oocytes** are formed. During puberty, some of the primary oocytes proceed from prophase I to the end of the first meiotic division forming two haploid cells (**secondary oocyte** – gets most of the cytoplasm – and **polar body** – has no role in reproduction)

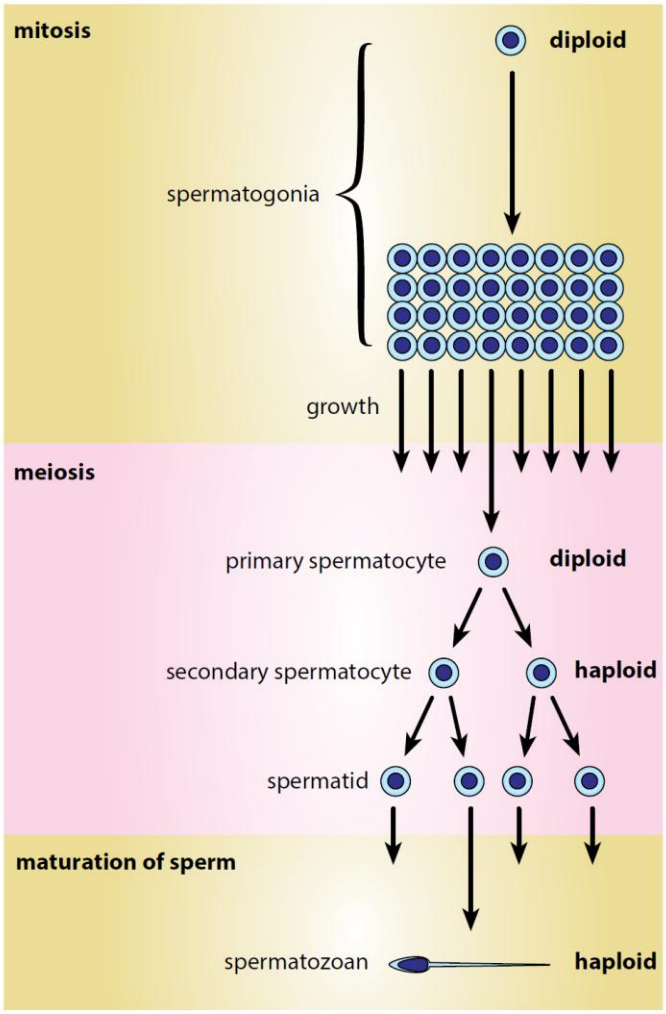


Figure 16.10 Spermatogenesis in humans.

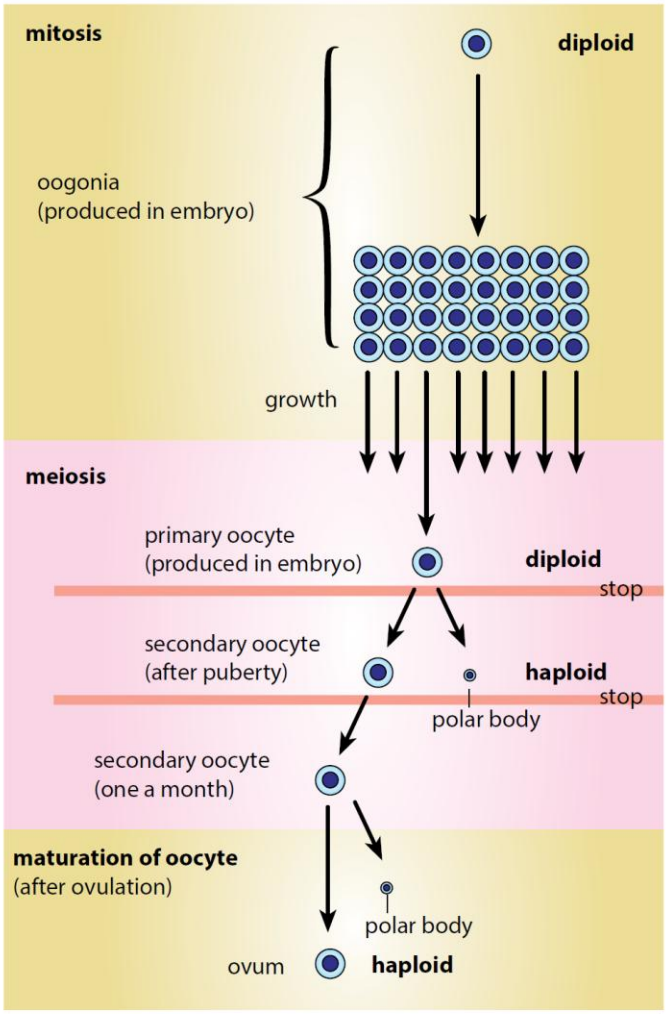


Figure 16.11 Oogenesis in humans.

- Once a month, one secondary oocyte is released into the oviduct from one of the ovaries, if it is fertilised, it continues its division by meiosis producing an **ovum**. The chromosomes of thee spermatozoan and ovum can join together to form a single diploid nucleus (zygote) which then divides by mitosis into an embryo, then a fetus

- **Gametogenesis in flowering plants:**
  - The male gametes are nuclei inside pollen grains, which are made in the anthers of a flower
  - The female gametes are nuclei inside the embryo sacs, which are made in the ovules inside the ovaries of a flower
  - Inside the anthers, **pollen mother cells** divide by meiosis to form four haploid cells, which nuclei divide by mitosis to form two haploid nuclei in each cell; matures into **pollen grains**; one of the nuclei is the male gamete nucleus which can fuse with a female nucleus to produce a diploid zygote which grows into an embryo plant

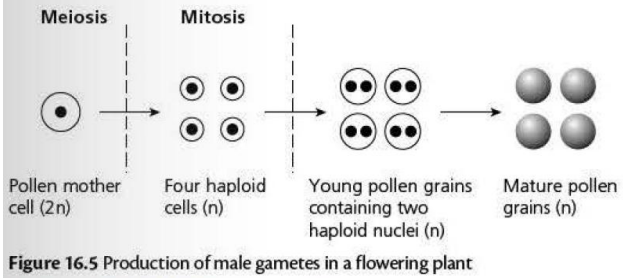


Figure 16.5 Production of male gametes in a flowering plant

- Inside each ovule, a large, diploid **spore mother cell** develops, which divides by meiosis to produce four haploid cells; all but one of these degenerates, which then develops into an **embryo sac**, which grows larger and its haploid nucleus divides by mitosis three times, forming eight haploid nucleus (one of these will become the female gamete)
- Note that in plants, unlike animals, the gametes are not formed directly by meiosis. Instead, meiosis is used in the production of pollen grains and the embryo sac and the gametes are then formed inside these structures by mitotic divisions

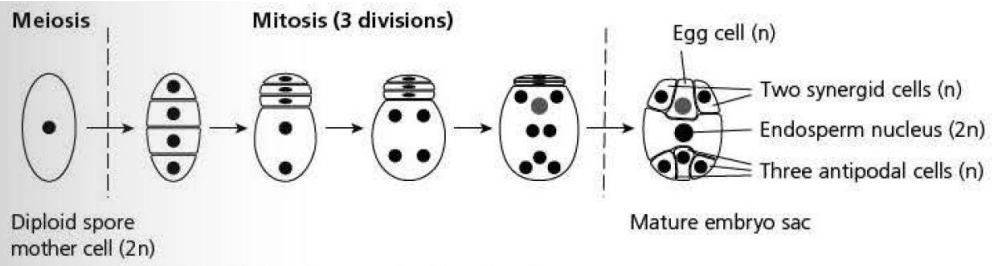


Figure 16.6 Production of a female gamete in a flowering plant

- During prophase I of meiosis, as the two homologous chromosomes lie side by side, their chromatids form links called **chiasmata** (singular: chiasma) with each other. When they move apart, a piece of chromatid from one chromosome may swap places with a piece from the other – **crossing over** – resulting in each chromosome having different combinations of alleles as it did before

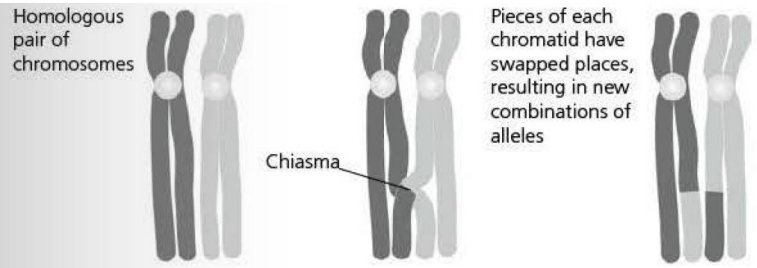


Figure 16.8 Crossing over in meiosis

- Independent assortment. At metaphase of meiosis I, the pairs of homologous chromosomes line up on the equator independently of each other. For two pairs of chromosomes, there are two possible orientations; at the end of meiosis II, each orientation gives two types of gamete. There are therefore four types of gamete altogether
- A **genotype** is the alleles possessed by an organism

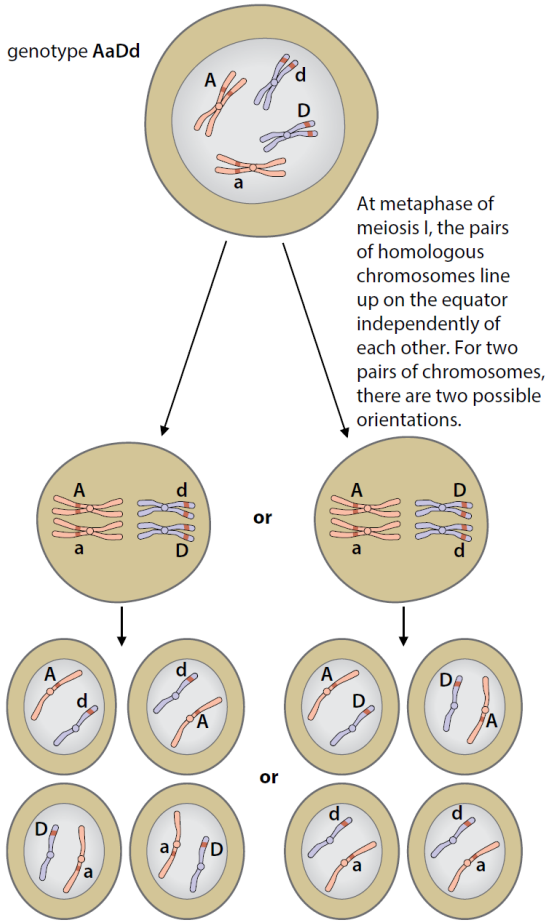


Figure 16.18 Independent assortment of homologous chromosomes during meiosis I results in a variety of genotypes in the gametes formed.

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.

The alleles that an organism has form its genotype

- **Homozygous** means having two identical alleles of a gene (e.g. Hb^AHb^A)
- **Heterozygous** means having two different alleles of a gene (e.g. Hb^AHb^S)
- Genotype affects phenotype:
  - Hb^SHb^S: coding for the production of the sickle cell β-globin polypeptide, sickle cell anaemia
  - Hb^AHb^A: coding for the normal β-globin polypeptide
  - Hb^AHb^S: Half of the person's Hb will be normal, and half will be sickle cell Hb - sickle cell trait – can be referred to as 'carriers' – they have enough normal haemoglobin to carry enough oxygen, and so will have no problems at all and immune to malaria
- An organism's **phenotype** is its characteristics, often resulting from an interaction between its genotype and its environment
- During every fertilisation, either an Hb^A sperm or an Hb^S sperm may fertilize 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

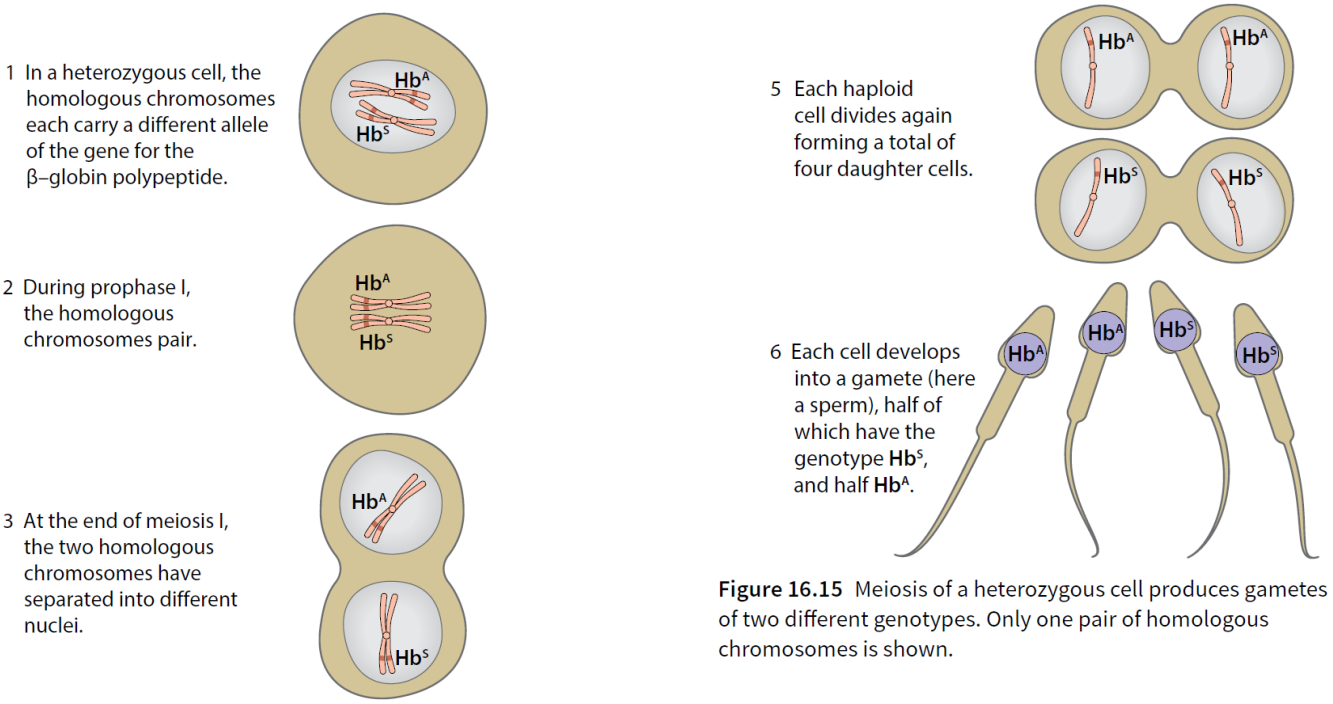


Figure 16.15 Meiosis of a heterozygous cell produces gametes of two different genotypes. Only one pair of homologous chromosomes is shown.

Monohybrid crosses

- Genetic diagram of flower colour in snapdragons (Antirrhinum) - codominant:
  - One of the genes for flower colour has two alleles, namely CR, which gives red flowers, and CW, 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

- Half of the offspring will have red flowers and half having pink flowers

- **Codominant** alleles both have an effect on the phenotype of a heterozygous organism
- 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
- Dominant and recessive example (tomato plants):

Genotype	Phenotype
AA	purple stem
Aa	purple stem
aa	green stem

- 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
- A **test cross** is a genetic cross in which an organism showing a characteristic caused by a dominant allele is crossed with an organism that is homozygous recessive; the phenotypes of the offspring can be a guide to whether the first organism is homozygous or heterozygous:
  - E.g. a purple stem tomato plant might have the genotype **AA** or **Aa**; to find out its genotype, it could be crossed with a green-stemmed tomato plant **aa**

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

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

(So from the colour of the offspring, it is possible to tell the genotype of the purple parent)

If its genotype is **Aa**:

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

- So far, only two alleles are considered of any one gene. Most genes however have **multiple alleles** (e.g. human blood groups gene)

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.

- XX: female & XY: male, where Y chromosome is much shorter than the X
- **Sex linkage**. E.g. **haemophilia** (**sex-linked gene**), in which the blood fails to clot properly due to the recessive allele **h**, resulting to the lack of factor VIII; where dominant allele, **H**, produces normal factor VIII
- The gene for haemophilia is on the X chromosome, and not on the autosome, hence affects the way that is inherited, e.g. a man does not have haemophilia while the woman is a carrier (0.25 probability: normal girl, boy, carrier girl and boy with haemophilia):

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

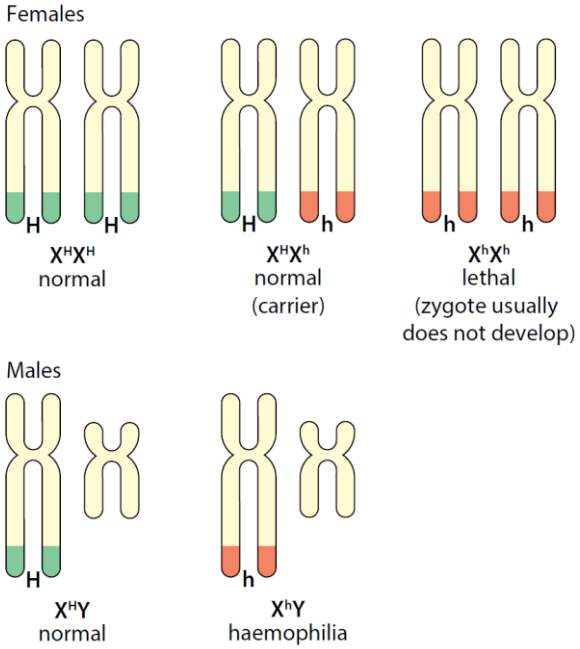


Figure 16.17 The possible genotypes and phenotypes for haemophilia.

Dihybrid crosses (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

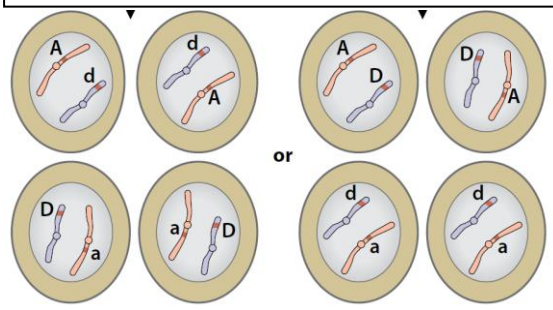
where **A** is dominant and **a** is recessive.

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.

During independent assortment of heterozygous genes, it will create four types of gametes: **AD, Ad, aD** and **ad**



- Heterozygous plant + recessive plant & Heterozygous plant + Heterozygous plant

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)

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

1 : 1 : 1 : 1 ratio of a dihybrid cross between a heterozygous organism and a homozygous recessive organism where the alleles show complete dominance

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 other parent \ Gametes from one parent	(AD)	(Ad)	(aD)	(ad)
(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

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

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.

- There are also cases where different loci interact to affect one phenotypic character (e.g. the inheritance of feather colour in chickens, between two loci: **I/i** and **C/c**
  - Individuals carrying the dominant allele, **I**, have white feathers even if they also carry the dominant allele, **C**, for coloured feathers. Birds that are homozygous recessive are also white; **IICC** + **iicc:**

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

Where 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

The usual 9 : 3 : 3 : 1 ratio expected in this generation has been modified to (9+3+1) : 3, giving 13 white : 3 coloured

- **Linkage** is the presence of two genes on the same chromosome, so that they tend to be inherited together and do not assort independently (e.g. the fruit fly, Drosophila. The gene for body colour and the gene for antennal shape are close together on the same chromosome and so are linked)

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

In this case the genotype of the striped body fly with normal antennae is written (**EA**)(**EA**) and not **EEAA**, which would indicate that the genes were not on the same chromosomes

- EA + ea: & offspring + ea:

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

The test cross gives a 1 : 1 ratio of the two original parental types and not the 1 : 1 : 1 : 1 (1 **EeAa** : 1 **Eeaa** : 1 **eeAa** : 1 **eeaa**) ratio expected from a dihybrid cross. The dihybrid cross has behaved as a monohybrid cross. The alleles that went into the cross together remained

Parental phenotypes male, striped body, normal antennae × female, ebony body, aristopedia antennae

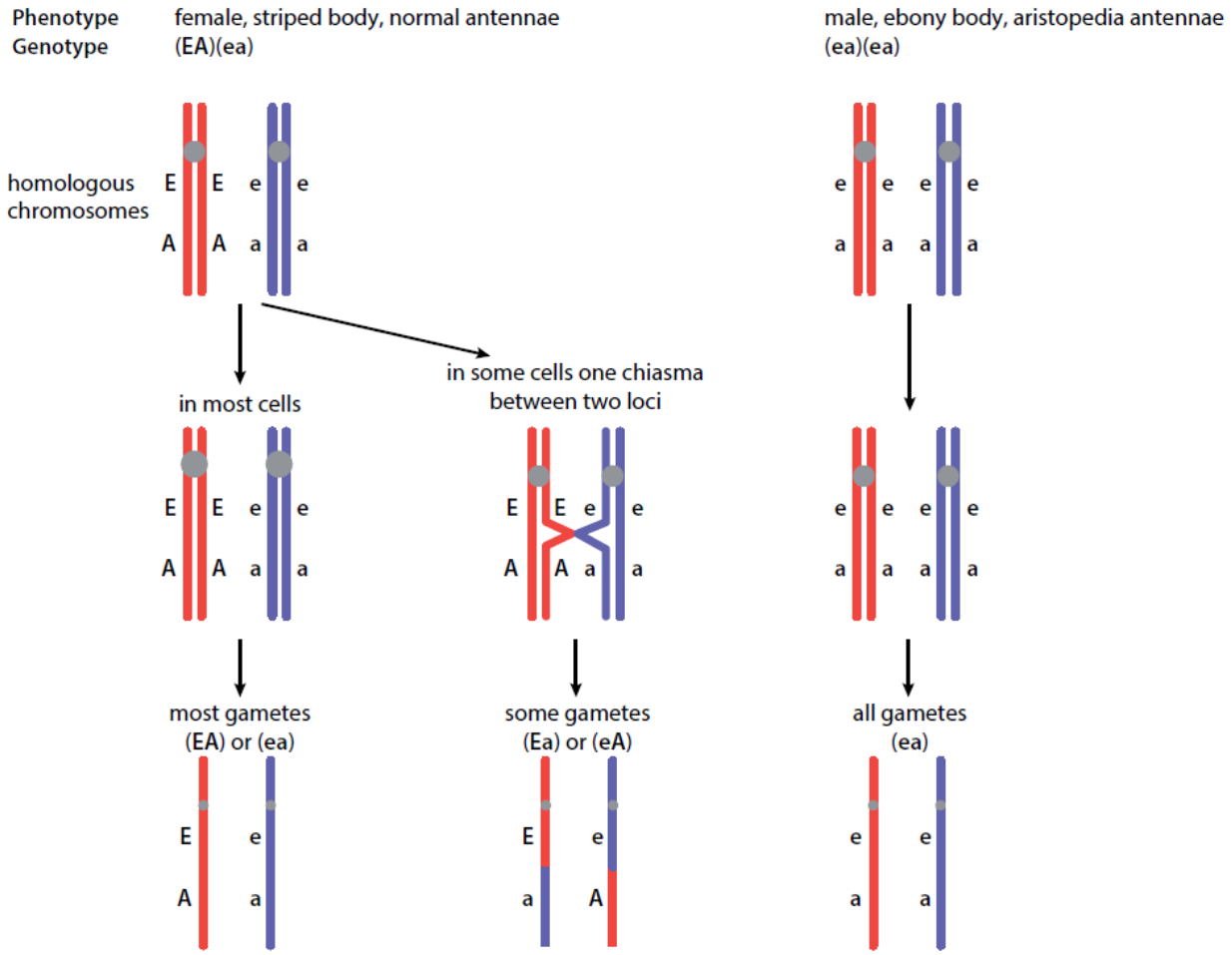
Parental genotypes (EA)(ea) × (ea)(ea)

Gametes (EA) or (ea) × (ea)
in equal proportions

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

- Recombinant (offspring caused by crossing over):
  - Cross over value is the percentage of offspring that belong to the recombinant class



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
	(ea)	(ea)(ea) ebony body, aristopedia antennae
small numbers	(Ea)	(Ea)(ea) striped body, aristopedia antennae
	(eA)	(eA)(ea) ebony body, normal antennae

Figure 16.21 Crossing over in female *Drosophila*.

In this particular cross, we would typically find:

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

- **Chi-squared (χ2) test** allows comparison between observed results and expected results, and decide whether there is a significant difference between them (e.g. two heterozygous tomato plants – 144 offspring):
  - Expected results:

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

  - Actual results:

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

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.

  - Chi-squared (χ2) test:

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

where: $\sum$ = 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

$$\chi^2 = \sum \frac{(O-E)^2}{E} = 0.79$$

    - Comparing value of χ2 with table 16.3 (using 0.05 probability)
    - Ifχ2 value represents a probability of 0.05 or larger, then we can be fairly certain that the differences between our observed and expected results are due to chance – the differences between them are **not significant**
    - Degrees of freedom: number of data classes minus 1, so in e.g. 4 -1 = 3
    - From the table 0.79 > 7.82, hence there is **no significant difference**, the difference in results are only due to chance
    - Null hypothesis: There is no significant difference between A and B
- **Gene mutation:** a change in the structure of a DNA molecule, producing a different allele of a gene
- **Mutagen:** a substance that increases the chances of mutation occurring
- Three different ways in which the sequence of bases in a gene may be altered (gene mutations):
  - Base substitution, where one base takes the place of another, e.g. CCT GAG GAG into CCT GTG GAG

  - Base addition, where one or more extra bases are added to the sequence, e.g. CCT GAG GAG into CCA TGA GGA G
  - Base deletion, where one or more bases are lost from the sequence, e.g. CCT GAG GAG into CCG AGG AG
- Base deletion and addition have significant effect on the structure, therefore function of the polypeptide that the allele codes for, and causes **frame shifts**
- Base substitution may not have any apparent effect, called **silent mutation**
- Sickle cell anaemia:
  - Base substitution
  - Normal β-globin polypeptide's **Hb^A** allele:
    - Val-His-Leu-Thr-Pro-Glu-Glu-Lys-
  - In **Hb^S** allele, CTT is replaced by CAT:
    - Val-His-Leu-Thr-Pro-Val-Glu-Lys-
  - The 'unusual' β-globin polypeptide make the Hb much less soluble to oxygen
  - The molecules tend to stick towards each other forming long fibres inside the RBC
  - RBC are pulled out of shape, into a sickle shape
  - When this occurs, RBC unable to transport oxygen
  - Severe anaemia will occur
- Albinism:
  - Causes melanin – dark pigment – to totally or partially be missing from the eyes, skin and hair; often accompanied by poor vision, rapid, jerky movement of the eyes and a tendency to avoid bright light
  - Two forms:
    - Autosomal recessive
    - Sex-linked (affects only the eyes, not the skin)
  - A mutation in the gene for the enzyme tyrosinase (transmembrane protein) results in either the absence of tyrosinase or the presence of inactive tyrosinase in melanocytes, where the membrane of the large organelle (melanosomes) can be found
  - The first two steps of the conversion of the amino acid, tyrosine into melanin cannot take place, as the inability for the conversion into DOPA and dopaquinone:

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

- Huntington's disease:
  - Inherited as a dominant allele
  - Neurological disorder resulting in involuntary movements (chorea) and progressive mental disorientation. Brain cells are lost and the ventricles become larger; age of onset is variable (most common: middle age)
  - The mutation is an unstable segment in a gene on
  - Chromosome 4 coding for a protein, huntingtin, where normally the segment is made up of a small number of repeats of the triplet of bases CAG, there is a larger number of repeats of the CAG triplet. This is called a 'stutter'. There is a rough inverse correlation between the number of times the triplet of bases is repeated and the age of onset of the condition: the more stutters, the earlier the condition appear
- ...

Selection and evolution (Chapter 17):

- 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 (excluding somatic cells – apart of cells in the reproductive organs)
- Genetic variation provides the raw material on which natural selection can act, where some individual have features that give them an advantage over other members in a population
- Phenotypic variation is also caused by the **environment**, e.g. some organisms might be larger than others due to better access of quality food while they were growing, however these variations will **not** be passed onto the offspring
- Qualitative differences fall into clearly distinguishable categories, with no intermediates – e.g. four possible ABO blood groups: A, B, AB or O. This is **discontinuous variation**
- Quantitative differences where there is a range of heights between two extremes (Figure 17.2). This is **continuous variation**

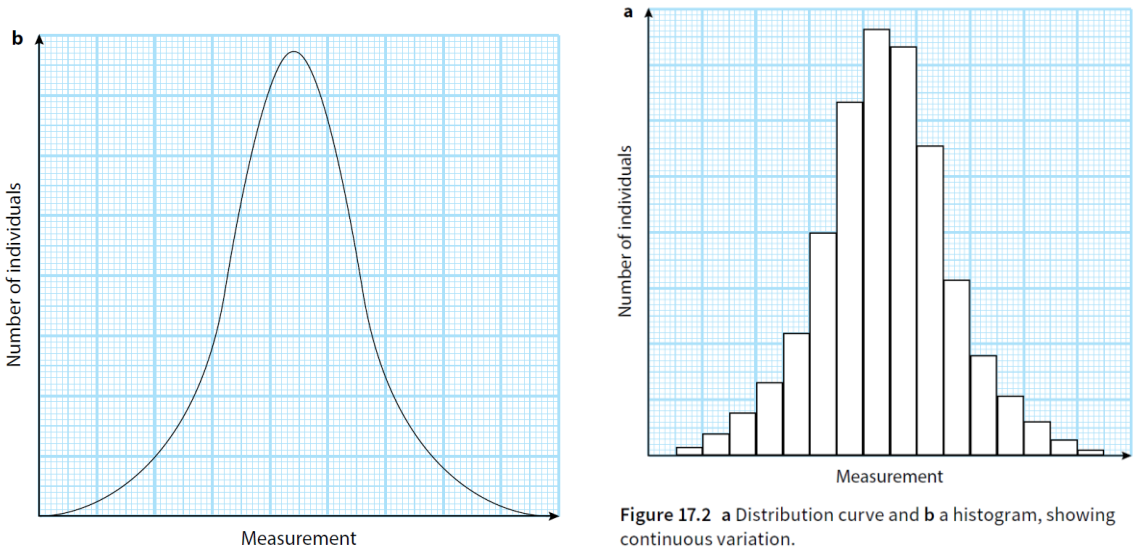


Figure 17.2 **a** Distribution curve and **b** a histogram, showing continuous variation.

- Both qualitative and quantitative differences in phenotype may be inherited; may involve several different genes; differences between them are:
  - 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 (e.g. organism's height):
    - 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

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 6*x* 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 ...

Offspring genotypes and phenotypes:

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

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.

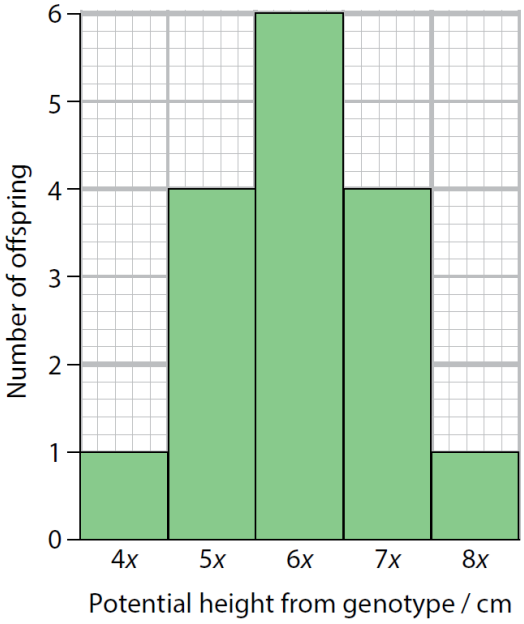


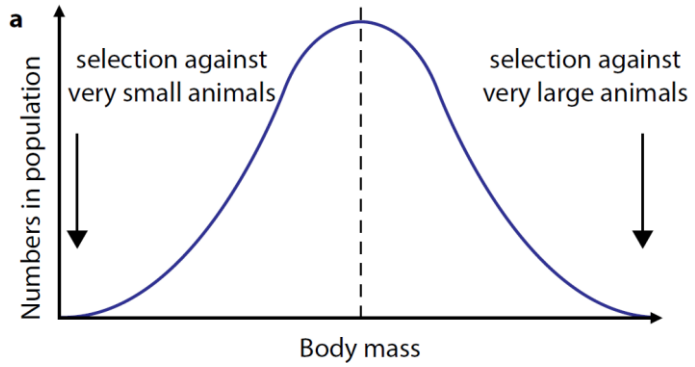
Figure 17.3 The additive effect of alleles.

- These results come from assuming that two unlinked genes, each with two alleles, contribute to the height of an organism; during polygenic (more than two alleles) continuous variation, the number of discrete height classes increases as more genes are involved and differences between these classes get less

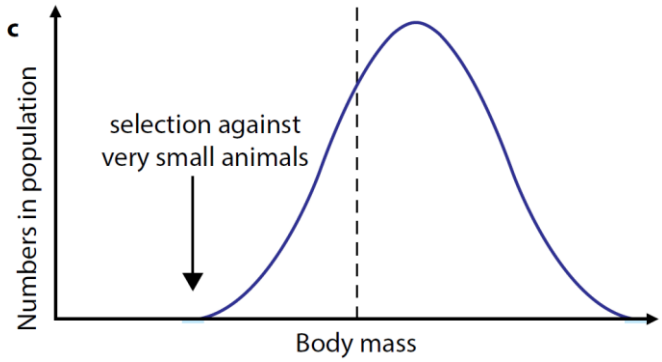
- Environmental effects may allow the full genetic potential height to be reached or may stunt it in some way:
  - One individual might have less food, or less nutritious food, than another with the same genetic contribution
  - A plant may be in a lower light intensity or in soil with fewer nutrients than another with the same genetic potential height.

- Himalayan colouring of rabbits and of Siamese and Burmese cats, colouring is caused by an allele which allows the formation of the dark pigment only at low temperature, hence the extremities are the coldest parts of the animals

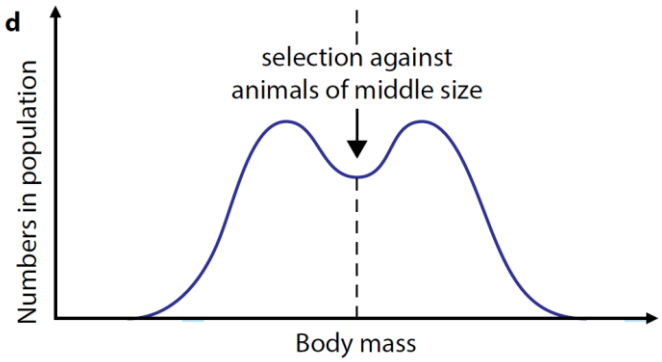
- The **t-test** is used to assess whether or not the means of two sets of data with roughly normal distributions, are significantly different from one another (p. 500)
- Various environmental factors come into play to keep down a population's number:
  - **Biotic** – caused by other living organisms such as through predation, competition for food, or infection by pathogens
  - **Abiotic** – caused by non-living components of the environment such as water supply or nutrient levels in the soil
- Once the population increases, the pressure of the environmental factors will be sufficiently great, then the population size will decrease, only when they have fallen considerably will the numbers be able to grow again; over a period of time, the population will oscillate about a mean level
- Natural selection occurs as populations have the capacity to produce many offspring that compete for resources; in the 'struggle for existence' only the individuals that are best adapted survive to breed and pass on their alleles to the next generation (selection pressure)
- **Fitness** is the capacity of an organism to survive and transmit its genotype to its offspring
- **Selection pressure** an environmental factor that gives greater chances of survival and reproduction on some individuals than on others in a population (e.g. predation and camouflage)
- Environmental factors can act as:
  - Stabilising selection:



  - Directional selection (if a **new allele** or a **new environment factor** appears causing a change in allele frequency):



- Disruptive selection (when conditions favour both extremes of a population):



- Changes in allele frequency creates the basis of **evolution**
- Antibiotic resistance:
  - There may be one or more individual bacteria with an allele giving resistance to penicillin – such as *Staphylococcus* – by producing penicillinase, which inactivates penicillin. These bacteria can survive and reproduce, while others will die (selection pressure)
- Industrial melanism:
  - Black forms of moth with allele, **C**, increases in areas near industrial cities; whereas speckled forms of moth with allele, **c**, stays constant in non-industrial areas (predation selection pressure)
  - These mutations are not caused by pollution (changes in environmental factors only affect the likelihood of an allele surviving in a population; **not** affecting the likelihood of such an allele arising by mutation
- Sickle cell anaemia:
  - Places where sickle cell allele is most common are parts of the world where malaria (caused by protoctist parasite, Plasmodium, where it enters the RBC and multiply) is found (selection pressure occurs hence selective advantage occurs)
  - There are two strong selection pressures acting on these two allele:
    - 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, as they are more likely to die from malaria
  - Heterozygous people with malaria only have about one-third the number of Plasmodium in their blood as do Hb^AHb^A homozygotes
- **Genetic drift** is a change in allele frequency that occurs by chance, as only some of the organisms of each generation reproduce (e.g. when a small number of individuals are separated (isolated) from the rest of a large population, resulting to different allele frequencies; further genetic drift will alter the allele frequencies even more and evolution will cause significant difference with the parent population) – **founder effect**
- **Hardy-Weinberg principle** allow the proportions of each of the genotypes in a large, randomly mating population to be calculated (the frequency of a genotype is its proportion to the total population; *p* represents the frequency of the dominant allele & *q* represents the frequency of the recessive allele):
  - E.g. two alleles of a single gene, **A/a**, thus three genotypes will be in the population

$$p + q = 1 \qquad \text{(Equation 1)}$$

$$\text{So, } p^2 + 2pq + q^2 = 1 \qquad \text{(Equation 2)}$$

- 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 hte two alleles into, or out of, the population
  - Non-random mating

Calculating genotype frequency

The homozygous recessives in a population can be recognised and counted. Suppose that the incidence of the **aa** genotype is 1 in 100 individuals (1%).

Then, $q^2 = 0.01$ and $q = \sqrt{0.01} = 0.1$

So, using **Equation 1**:

$p = 1 - 0.1 = 0.9$ and $p^2 = (0.9)^2 = 0.81$

That is 81% of the population are homozygous **AA**.

And, $2pq = 2 \times 0.9 \times 0.1 = 0.18$

Or, using **Equation 2**:

$2pq = 1 - (0.01 + 0.81) = 0.18$

That is, 18% of the population are heterozygous **Aa**.

- Selective breeding of dairy cattle:
  - **Artificial selection**: When humans purposely apply selection pressures to populations
  - Desired features include docility (making the animal easier to control), fast growth rates and high milk yields have been achieved by **selective breeding**
  - Individuals showing one or more of these desired features are chosen for breeding
  - Some of the alleles granting these features are passed on to the individuals' offspring
  - Over many generations, alleles granting the desired characteristics increase in frequency, while those conferring characteristics not desired by the breeder decrease in frequency
  - Background genes (the alleles of genes that adapt to its particular environment) results in offspring obtaining the same adaptations, however will not be well-adapted to a new environment (even though it will show selected traits)
- Crop improvement:
  - Gene technology is used to alter or add genes into a species in order to change it characteristics
  - Selective breeding:
    - Produced many different varieties of wheat and rice – most is grown to produce grains rich in gluten
    - Resistance towards various diseases (wheat and rice)
    - Shorter stems, for easy harvest hence higher yields (less energy used to grow tall, more to growing of seeds) – wheat and rice
  - Most of the dwarf varieties of wheat carry mutant alleles of two reduced height (Rht) genes, which code for DELLA proteins to reduce the effect of gibberellins on

growth. The mutant alleles cause dwarfism by producing more of, or more active forms of, these transcription inhibitors

- A mutant allele of a different gene, called 'Tom Thumb', has its dwarfing effect because the plant cells do not have receptors for gibberellins and so cannot respond to the hormone
- Interbreeding and hybridisation of maize for uniformity and heterozygosity:
  - If maize plants are inbred (crossed with other plants with genotypes like their own), the plants in each generation become progressively smaller and weaker – **inbreeding depression** – due to the less vigorous homozygous plants compared to heterozygous
  - Homozygous plants obtained from companies, then crossing them, producing F1 plants that all have the same genotype which have characteristics such as high yields, resistance to more pests and diseases, and good growth in nutrient-poor soils or where water is in short supply

- **Speciation:** The production of new species
- A **species** is a group of organisms with similar morphological, physiological, biochemical and behavioural features, which can breed together naturally to produce fertile offspring, and are reproductively isolated from other species
- **Reproductive isolation:** The inability of two groups of organisms of the same species to breed with one another, e.g. because of geographical separation or because of behavioural differences
- 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/separation):
  - Requires a barrier to arise between two populations of the same species, preventing them from mixing (e.g. a stretch of water, deforestation)
  - The selection pressures on these two places might be very different, resulting in different alleles being selected for
  - Over time, the two population can no longer interbreed, hence a new species had evolved
- Sympatric speciation (ecological and behavioural separation):
  - Two groups of individuals living in the same area may become unable to breed together (e.g. one group develops courtship behaviours that no longer match with the other groups, because they love in different habitats in the same area (ecological separation))
- Molecular evidence that reveals similarities between closely related organisms with reference to mitochondrial DNA and protein sequence data:
  - Mitochondria contain a single DNA molecule that is passed on down the female line; analysis of mitochondrial DNA (mtDNA) can be used to determine how closely

related two different species are; the more similar the sequence of bases in the DNA, the more closely related they are considered to be

- Amino acid sequences in proteins can be used (the protein cytochrome c, involved in the electron transport chain, is found in a very wide range of different organisms, suggesting that they all evolved from a common ancestor, where differences in the amino acid sequences in cytochrome c suggest how closely related particular species are

- Extinctions tend to be caused by:
  - Climate change, for example, global warming can result in some species inability to find adapted habitats
  - Competition, for example, a newly evolved species or an alien species, may out-compete a resident species
  - Habitat loss, for example, large deforestations
  - Direct killing by humans
- Structural genes:
  - Genes that code for proteins required by a cell
- Regulatory genes:
  - Genes that code for proteins that regulate the expression of other genes
- Difference 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 bacterium's DNA
  - The synthesis of an inducible enzyme occurs only when its substrate is present (transcription of a gene occurs as a result of the inducer (enzyme's substrate) interacting with the protein produced by the regulatory gene)
- The *lac* operon (e.g. *E. Coli*) – gene control in prokaryotes:
  - (operon is a length of DNA making up a unit of gene expression in a bacterium)

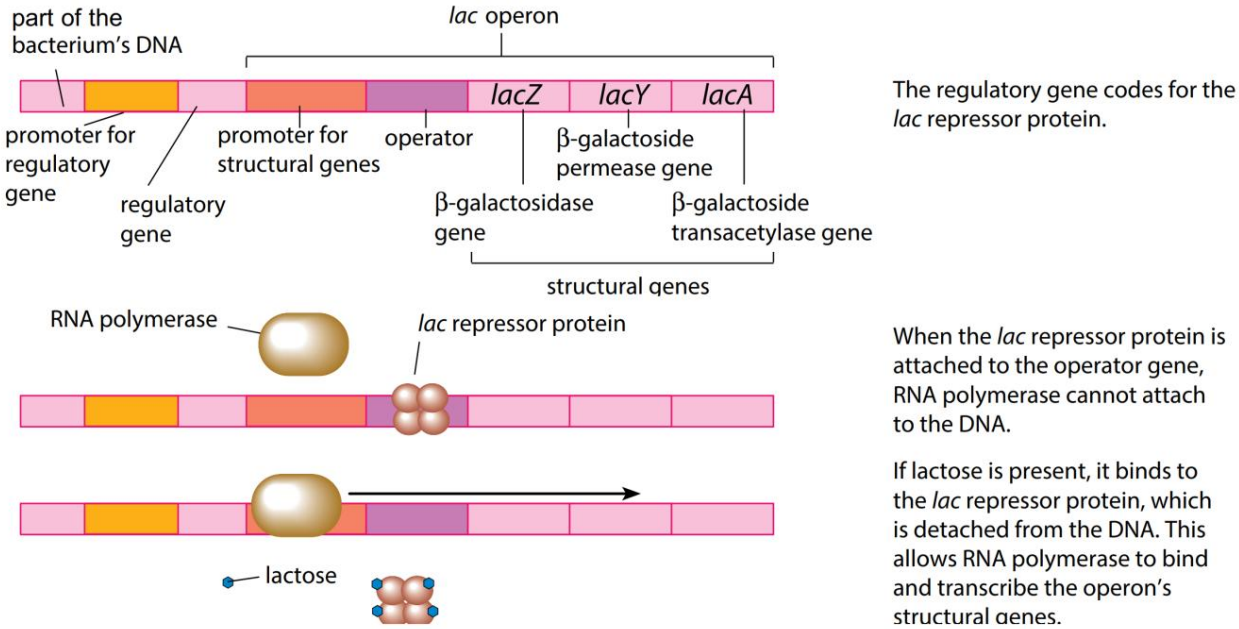


Figure 16.23 Regulation of gene expression by the *lac* operon.

- The enzyme β-galactosidase hydrolyses the disaccharide lactose to the glucose and galactose

- 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 (allows lactose to enter the cell)
  - *lacA*, coding for transacetylase
- When there is no lactose in the medium in which the bacterium is growing:
  - The regulatory gene codes for a protein called a repressor
  - The repressor binds to the operator region, close to gene *lacZ*
  - 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 take place
- The repressor protein is allosteric (two binding sites), hence when lactose binds to its site, the shape of the protein changes so that the DNA-binding site is closed
- 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
- β-galactosidase is an inducible enzyme

- Gene control in eukaryotes:
  - Transcription of a gene is controlled by transcription factors – proteins that bind to a specific DNA sequence and control the flow of information from DNA to RNA by controlling the formation of mRNA, role is to make sure that genes are expressed in the correct cell at the correct time and to the correct extend, effects:
    - Necessary for transcription to occur, form part of the protein complex that binds to the promoter region of the gene concerned
    - Activate appropriate genes in sequence
    - Responsible for the determination of sex in mammals
    - Allow responses to environmental stimuli, e.g. switching on the correct genes to respond to high environmental temperatures
    - Hormones have their effect through transcription factors
  - Gibberellin controls seed germination in plants by increasing the transcription of mRNA coding for amylase, done by breaking down of DELLA proteins (inhibits the binding of a transcription factor, such as PIF to a gene promoter), causing PIF to bind to its target promoter resulting to an increase in amylase production:

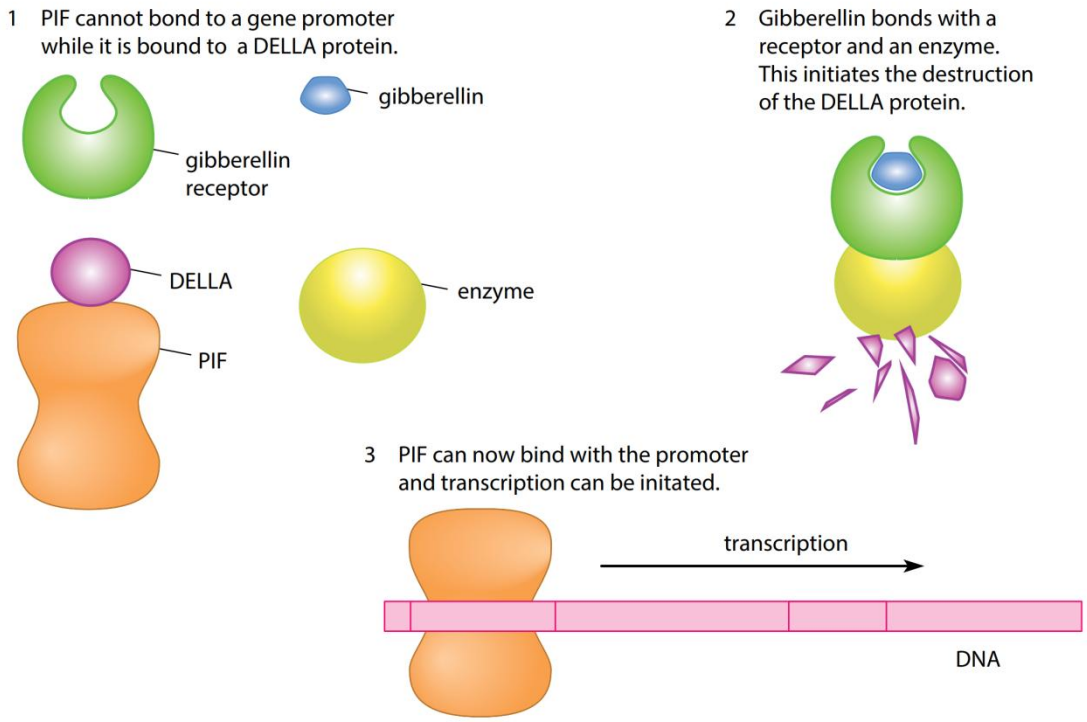


Figure 16.24 How gibberellin controls gene transcription.