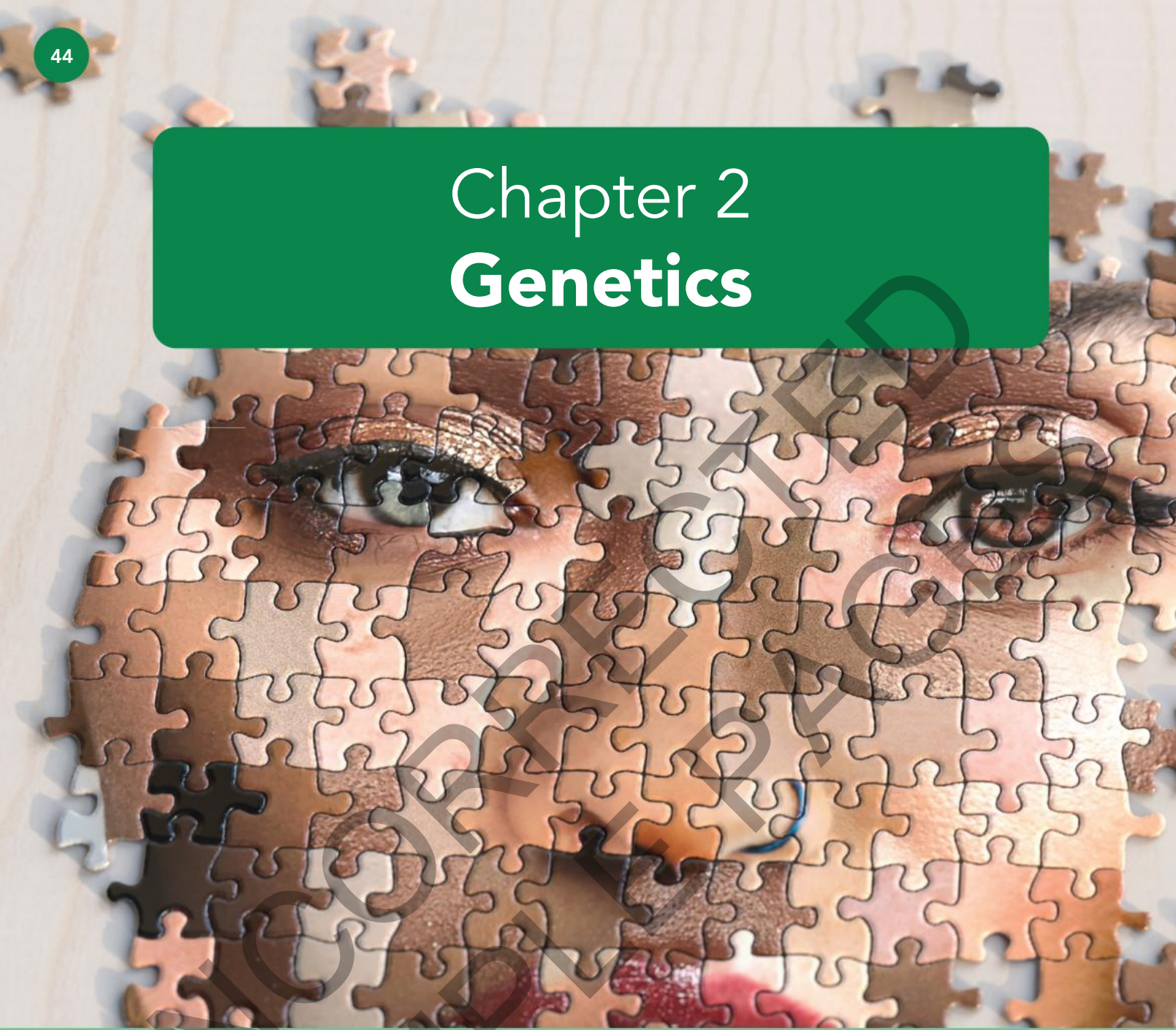
Chapter 2
Genetics



Chapter introduction

Everyone is unique, but you may look similar to your parents. This is thanks to the genes that they passed on to you. Genes carry the information that determines your traits and are made of DNA. This chapter explores the structure and function of DNA and genes in heredity. You will consider how variation is increased in offspring and predict patterns of inheritance. You will also examine what happens when there are problems with DNA and how these may be caused by the environment.

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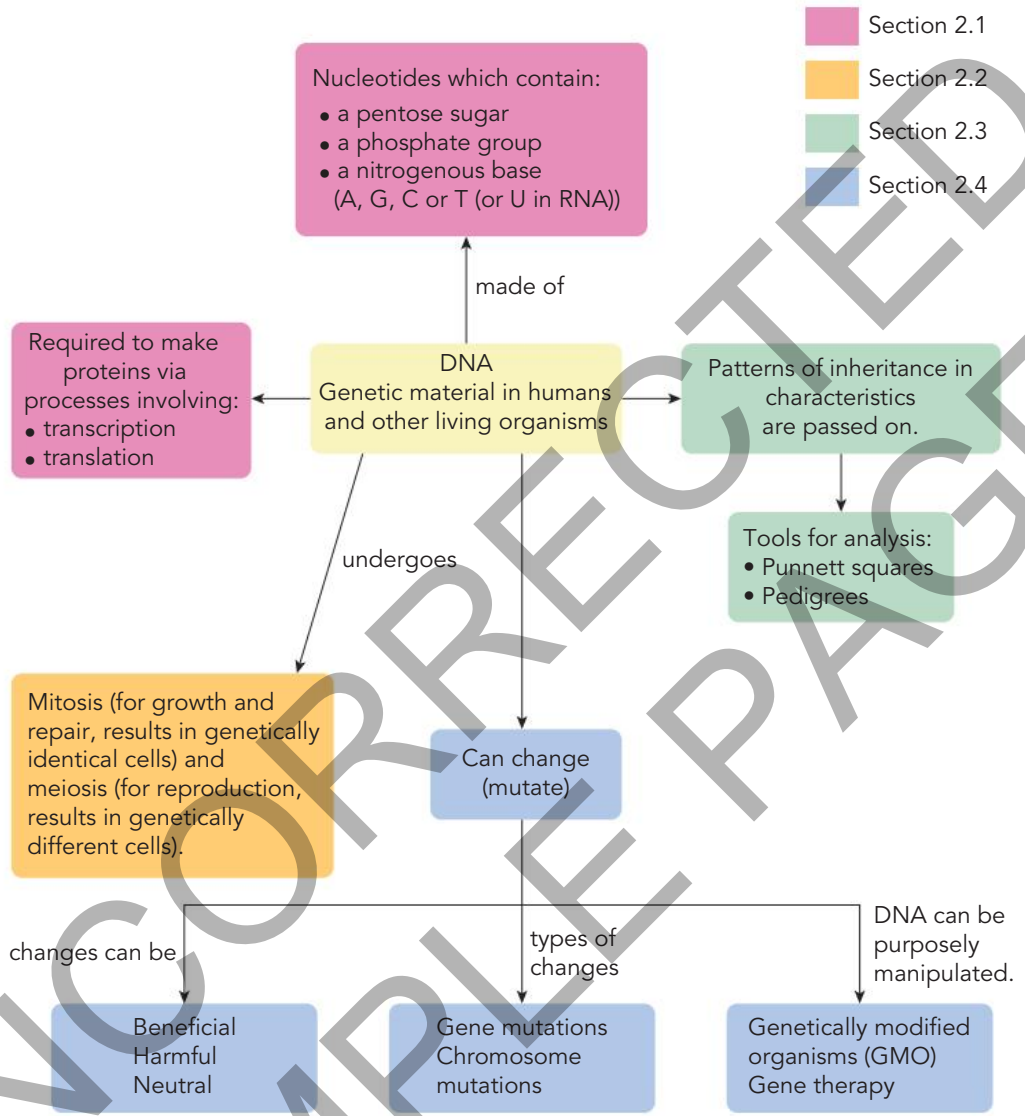
Chapter map



Curriculum content

Biological sciences

Cell division processes of meiosis and mitosis produce new cells with chromosome numbers specific to their role; chromosomes contain genes that are composed of DNA (deoxyribonucleic acid)

• comparing the role and number of chromosomes in the cells produced by mitosis and meiosis	2.2
• modelling the structure of DNA	2.1

Patterns of monohybrid inheritance, including autosomal dominant/recessive and sex-linked recessive inheritance, can be predicted using pedigrees and Punnett square crosses

• interpreting pedigrees and predicting offspring for autosomal dominant/recessive alleles, such as hair colour in guinea pigs, leaf colour in barley, seed shape and colour and plant height in peas	2.3
• interpreting pedigrees and predicting offspring for sex-linked recessive alleles, such as red-green colour blindness and haemophilia	2.3

Collaborating and applying

Illustrate how advances in scientific understanding often rely on developments in technologies and engineering and technological and engineering advances are often linked to scientific discoveries

• exploring how the development of fast computers has made the analysis of DNA sequencing possible	2.4
• examining how the work of Rosalind Franklin, James Watson, Francis Crick and Maurice Wilkins contributed to the development of the double helix structure of DNA	2.1

Illustrate how proposed scientific responses to contemporary issues may impact on society

• examining karyotypes and applications of biotechnologies, such as DNA profiling, gene therapy and genetic engineering	2.2, 2.4
• investigating why agricultural practices have changed to include widespread use of genetically engineered crops	2.4

Glossary terms

Allele
Aneuploidy
Autosome
Base triplet
Carrier
Centromere
Chromatin
Chromosome
Chromosome mutation
Codominance
Codon
Complementary base pairing
Crossing over
Deletion
Diploid (2*n*)
DNA
Dominant
Embryo
Fertilisation
Filial
Gametes
Gene
Gene mutation
Gene therapy
Genetic engineering
Genetic screening
Genetically modified organism
Genetics
Genome
Genotype
Germline mutation
Gonads
Haploid (*n*)
Heredity
Heterozygous
Homologous chromosomes
Homozygous
Hydrogen bonds
Incomplete dominance
Induced mutation
Insertion
Inversion
Karyotype
Locus
Meiosis
Mitosis
Monogenic
Monohybrid cross
Monomer
Mutagenic
Mutation
Non-disjunction
Non-homologous chromosomes
Nucleotide
Ova
Pedigree
Phenotype
Point mutation
Polymer
Polypeptide
Punnett square
Recessive
Recombination
Reduction division
Sex chromosomes
Sexual reproduction
Sister chromatids
Somatic cells
Somatic mutation
Spontaneous mutation
Stem cell
Substitution
Telomeres
Test cross
Transcription
Transgenic organism
Translation
Trisomy
Zygote

2.1 Introduction to genetics and DNA

WORKSHEET
DNA structure

VIDEO
Structure of DNA

Learning goals

At the end of this section, I will be able to:

1. Recall the components that make up the structure of DNA and RNA molecules.
2. Describe the relationship between DNA, genes and chromosomes.
3. Describe the steps in which DNA is used to synthesise proteins.

Genetics

Genetics is the study of how certain traits (characteristics) can be inherited from a previous generation. It involves the study of **genes**, genetic variation and **heredity**. Genes are considered the basic unit of inheritance. They are made of **d**eoxyribo**n**ucleic **a**cid, or **DNA** for short. This hereditary material is passed from parents to offspring and contains the information needed to specify traits.

genetics
the study of genes, genetic variation and heredity

gene
a length of chromosome made of DNA, the basic unit of inheritance

heredity
the genetic passing on of traits from one generation to the next

DNA
deoxyribonucleic acid; the molecular unit of heredity, containing the genetic information responsible for the development and function of an organism

polymer
a molecule made from many repeating subunits called monomers

monomer
a single subunit that when joined together repeatedly makes a polymer

nucleotide
a monomer subunit of a nucleic acid, consisting of a phosphate group bound to a five-carbon sugar, which in turn is bound to a nitrogenous base

hydrogen bonds
chemical bonds that hold the two DNA strands together

Structure of DNA

DNA is a double-stranded molecule that forms a 'double helix' shape, like a twisted ladder. DNA is a **polymer** as it is made up of many **monomer** subunits called **nucleotides**.

The nucleotides that make up DNA have three major components:

- a five-carbon (pentose) deoxyribose sugar
- a nitrogenous base – one of adenine (A), guanine (G), cytosine (C) or thymine (T)
- a negatively charged phosphate group.

The nucleotides bind together to form two long strands that wind around each other. Each strand has a backbone made of alternating deoxyribose sugar and phosphate groups.

The nitrogenous bases from one strand bind to complementary bases on the other strand using **hydrogen bonds**.

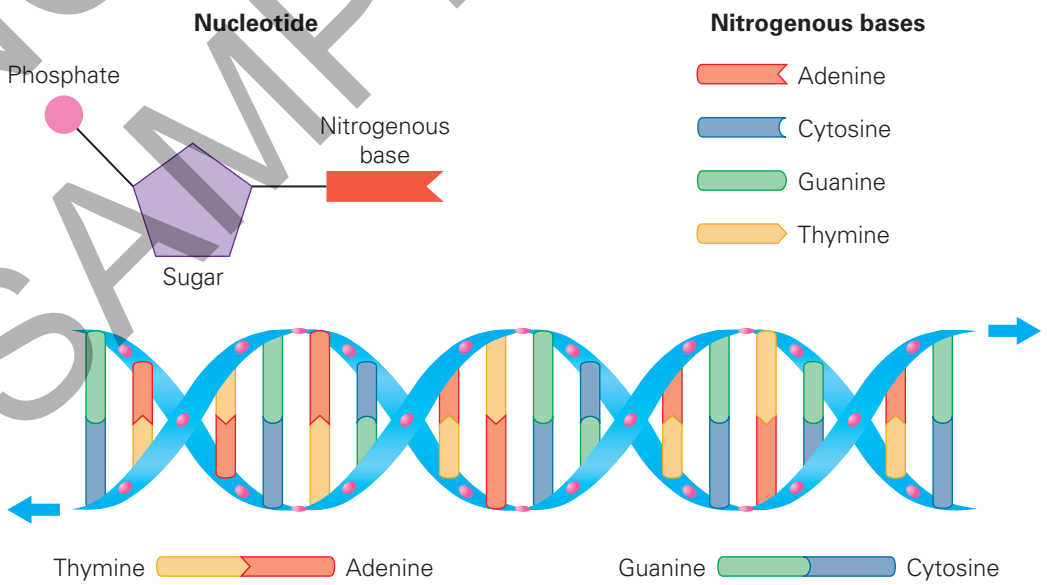


Figure 2.1 The double helix structure of DNA: the subunits called nucleotides all join up to form the double-stranded DNA. Note the sugar–phosphate backbone on the edge of the DNA molecule and the nitrogenous bases joining in the centre.

Figure 2.2 Traits are passed on from generation to generation.

Adenine (A) always pairs with thymine (T), and guanine (G) always pairs with cytosine (C). One way to remember the A–T and G–C pairings is 'Apple in the Tree, Car in the Garage'. This **complementary base pairing** occurs because of the shape of the nitrogenous bases and the number of hydrogen bonds they can form (Figure 2.3).

Guanine and adenine have a double-ring structure, which means they belong to a group of chemical molecules called purines. A helpful way to remember that adenine and guanine are purines is 'Pure As Gold'. Cytosine and thymine have a single-ring structure and belong to the group called pyrimidines. Purines always pair with pyrimidines, but the number of hydrogen bonds between them depends on the structure. Guanine binds with cytosine with three hydrogen bonds, while adenine and thymine form two hydrogen bonds.

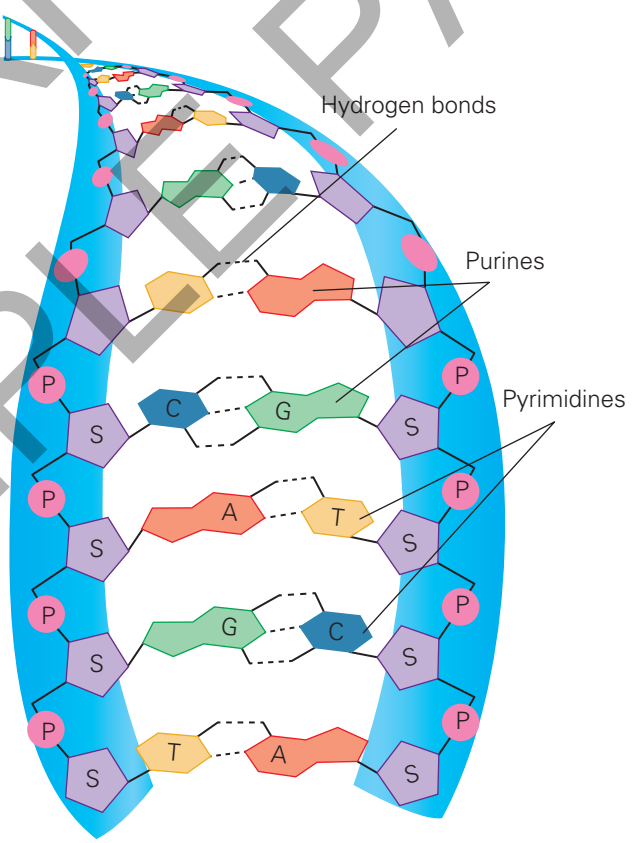


Figure 2.3 A closer look at the structure of DNA. The sugar–phosphate backbone forms the outer edge of the helix and the nitrogenous bases form complementary pairs in the centre. Hydrogen bonds hold the two strands together.

complementary base pairing adenine only binds with thymine and cytosine only binds with guanine

Try this 2.1

Modelling DNA using lollies

Be careful
Do not to consume lollies in the laboratory. Beware of allergies.

Materials

- liquorice ribbons or sour strips
- a handful of jelly babies
- toothpicks

Method

1. Sort the jelly babies into four groups of colours.
2. Pair up the jelly babies so that one particular colour always goes with another particular colour, for example, red with yellow and orange with green.
3. Place a pair of jelly babies onto each toothpick as if you were making lolly kebabs. Ensure that the pairs are always of matched colours.
4. Attach your lolly kebabs to the long strips of liquorice.
5. Keep doing this until you have about 5–7 horizontal toothpicks attached and it starts to look a bit like a ladder.
6. The paired coloured sweets represent the base pairs, while the liquorice is the sugar–phosphate backbone.
7. Pick up your lolly ladder and twist it to represent the double helix shape of DNA.

Practical 2.1

Extracting DNA from cells

Be careful
Wear safety glasses and a lab coat. Do not consume food items.

Time period
Approximately 1 hour

Aim
To investigate and extract DNA from strawberries.

Materials

- plastic sandwich bag (or other material to contain strawberry and liquid)
- strawberry
- DNA extraction solution provided (10 mL)
- filter funnel and gauze (or other gauze-style filter)
- cold ethanol solution
- test tube (or small beaker)
- stirring rod
- plastic pipette

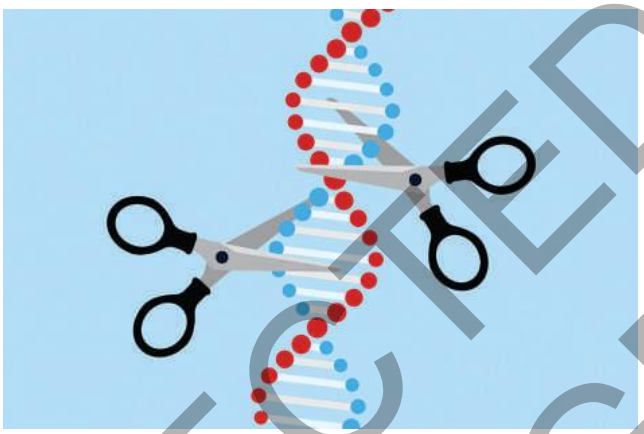
DNA extraction solution:

- dishwashing liquid or shampoo (5 mL)
- table salt (0.75 g)
- water (45 mL)

continued …

Method

1. Wash the strawberry with tap water and remove the green leaves. Add the strawberry to the plastic sandwich bag.
2. Add the DNA extraction solution (10 mL) and close the sandwich bag, removing the excess air.
3. Squash the strawberry into the liquid using your hands until the strawberry is roughly crushed.
4. Pour the strawberry mixture through a filter funnel lined with gauze into a test tube.
5. Discard the gauze and strawberry remains into a bin.
6. Add an equal volume of cold ethanol to the test tube using a plastic pipette.
7. Slowly mix the two layers of liquid with a stirring rod.
8. Collect the white solid that forms on the stirring rod. This is DNA.



Results

Write observations for what occurs during each step of the method.

Discussion

1. Describe whether the white solid could contain all of the DNA of the strawberry.
2. Visually compare the amount of solid you collected to that of other groups in the classroom. Were results consistent? Explain your answer.
3. Describe the variables that were not kept consistent between groups in the classroom.
4. Suggest any changes that could be made to the method to improve the quality of the recorded data in future experiments. Justify your suggestions by explaining how each change would improve the data quality.

Conclusion

1. Draw a conclusion regarding the efficiency of this method at extracting all the DNA from a strawberry, based on your and other groups' observations.

Did you know? 2.1

How long is DNA?

The DNA found in only one cell, if fully unravelled, would be around two or more metres long! The length of a piece of double-stranded DNA is commonly expressed as the number of complementary nitrogenous base pairs it contains. The DNA in an average human cell has an estimated 63 000 000 base pairs.

Quick check 2.1

1. **Recall** the three key components that make up a nucleotide.
2. **Describe** the overall shape of DNA.
3. If a strand of DNA contained the nitrogenous bases seen below, **state** the bases in its complementary strand. DNA strand: ATATAGATAGATCAGACA.

Explore! 2.1

The discovery of DNA

Friedrich Miescher was a Swiss biochemist who first observed DNA in 1869. However, it took almost a century for scientists to understand the structure of DNA and the mechanisms by which it carries genetic information.

Research the following scientists who, among many others, contributed to our understanding of DNA:

- Friedrich Miescher
- Erwin Chargaff
- Francis Crick and James Watson
- Rosalind Franklin and Raymond Gosling
- Maurice Wilkins.

Summarise their contributions and place them on a timeline.

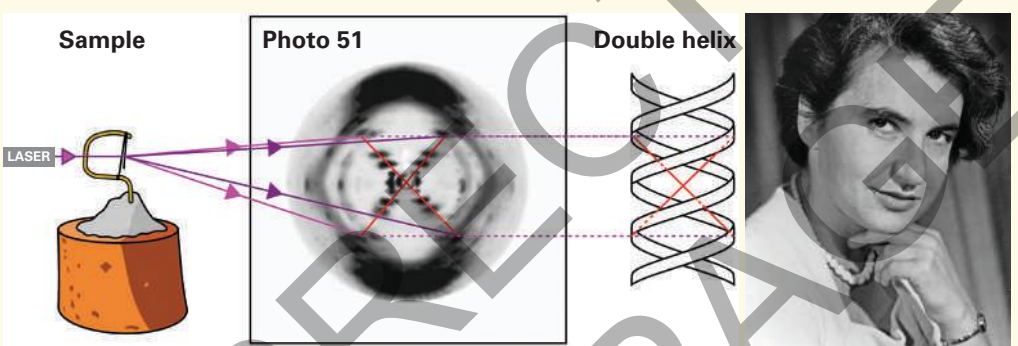


Figure 2.4 Dr Rosalind Franklin (right) and Raymond Gosling fired X-rays at a DNA strand stretched across a paperclip in 1952. The resulting picture was critical to the discovery of the double helix structure of DNA, providing key information that enabled James Watson and Francis Crick to build the first correct model of the structure of DNA.

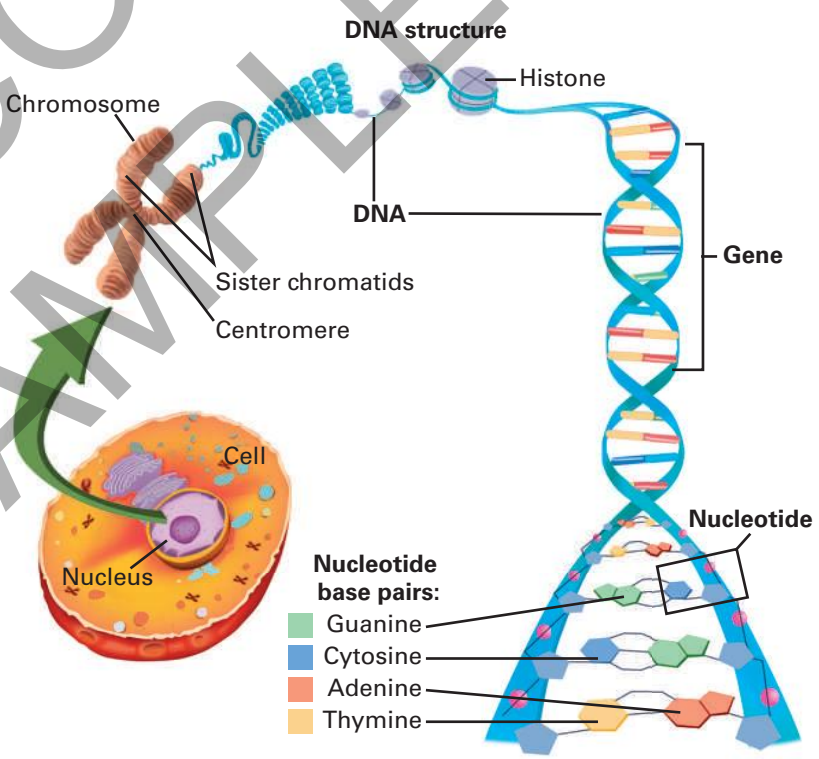


Figure 2.5 The relationship between DNA and chromosomes

What is a chromosome?

The DNA found in the nucleus of a cell is usually in a form called **chromatin**. This is a highly organised complex of DNA and associated proteins such as histones. By wrapping around histones, the long strands of DNA can fit inside the nucleus. When it is time for the cell to replicate, chromatin condenses into structures called **chromosomes**. Each molecule of DNA forms one chromosome. Along the chromosome are genes, which are the units of inheritance. Genes are sections of DNA that hold the specific instructions required for making every protein in our bodies.

The familiar X-shape of a chromosome is only seen after DNA replication has occurred. Shortly before replication, the chromosome appears as a single condensed DNA molecule. DNA replication produces two identical **sister chromatids** that are joined together by **centromeres** to form the distinct X-shape (Figure 2.6). At the ends of each chromosome are protective structures made of DNA and protein called **telomeres**. These protect the chromosomes, but every time DNA replicates, the telomeres shorten. Eventually, the telomeres shorten to the extent that replication can no longer occur.

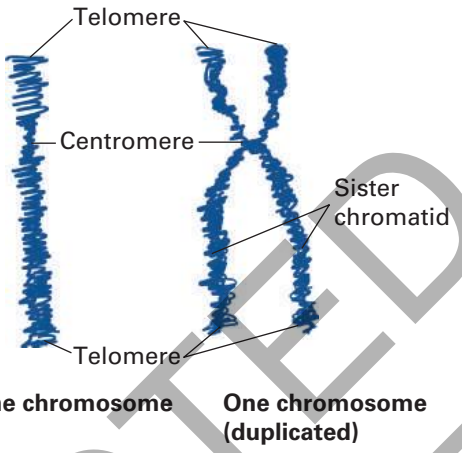


Figure 2.6 Before replication, a chromosome is a single molecule (left). After DNA replication, each chromosome consists of two molecules called sister chromatids (right).

VIDEO
DNA and RNA

chromatin
a mixture of DNA and proteins that form chromosomes

chromosome
a thread-like structure of tightly wound DNA and proteins

sister chromatids
two copies of the same chromosome, connected at a centromere

centromere
a structure that holds two sister chromatids together

telomeres
structures made from DNA and proteins that protect the ends of chromosomes

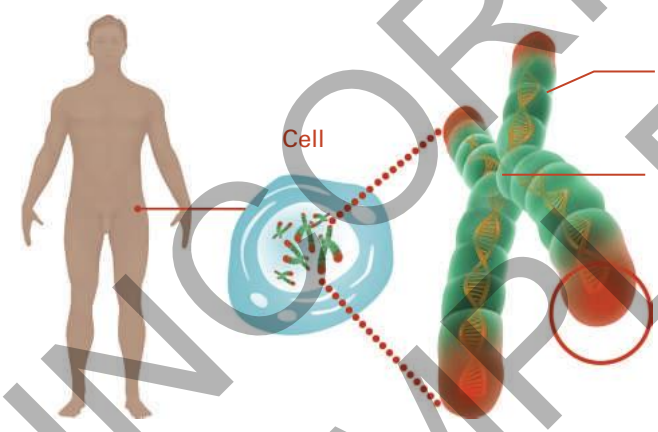


Figure 2.7 The distinct X-shaped chromosome, showing the two sister chromatids and the centromere

Figure 2.8 A scanning electron micrograph of a duplicated human chromosome

Making thinking visible 2.1

Compass points: Telomeres and immortality

Telomeres contain thousands of repeated copies of the base sequence TTAGGG. When we are infants, our telomeres are approximately 10 000 bases long, but each time the cell divides, they slowly get shorter. A cell will die when its telomeres become too short. By the time we are in our eighties, our telomeres may only be 5000 bases long, but this varies from tissue to tissue and from person to person. A person in their thirties may well have telomere lengths of a person in their nineties and vice versa.

continued …

This means that telomeres are a key component of ageing, and there are efforts to use them to slow down or reverse the ageing process. Some companies say they can measure your telomere length, suggesting that it will indicate your true, biological age. Despite limited evidence, there are also companies that sell products that they claim can maintain or lengthen telomeres – in effect, making you younger or possibly allowing immortality.

Complete the Compass points activity about manipulating telomeres to prevent ageing:

1. E = Enthusiastic: What generates enthusiasm within you regarding this concept? What are the potential benefits?
2. W = Worries: What causes concern about this idea? What are the potential drawbacks?
3. N = Need for Information: What other details or facts are necessary to evaluate this idea? What additional knowledge would aid in assessment?
4. S = Suggestion for Progress: What is your current viewpoint or proposal regarding this idea? How can you continue to assess this concept?

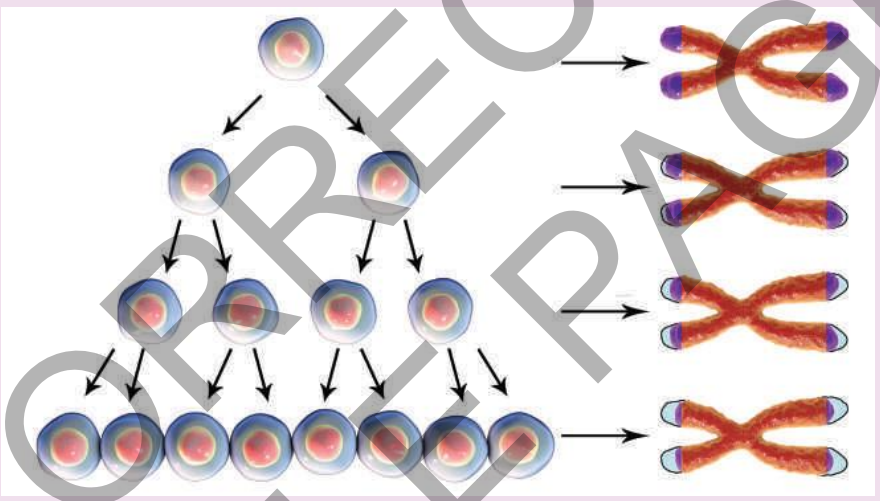


Figure 2.9 Telomeres shorten every time a cell divides.

The *Compass points* thinking routine was developed by Project Zero, a research centre at the Harvard Graduate School of Education.

	DNA	RNA
Number of strands	Two	One
Type of sugar	Deoxyribose	Ribose
Nitrogenous bases	Guanine Cytosine Adenine Thymine	Guanine Cytosine Adenine Uracil
Where is the molecule found?	Nucleus, mitochondria and chloroplasts (in eukaryotes) Cytoplasm (in prokaryotes)	Cytoplasm Nucleus Associated with ribosomes
Different forms	DNA Mitochondrial DNA (mtDNA) Chloroplast DNA (cpDNA)	Messenger RNA (mRNA) Transfer RNA (tRNA) Ribosomal RNA (rRNA)

Table 2.1 The main differences between the two nucleic acids, DNA and RNA

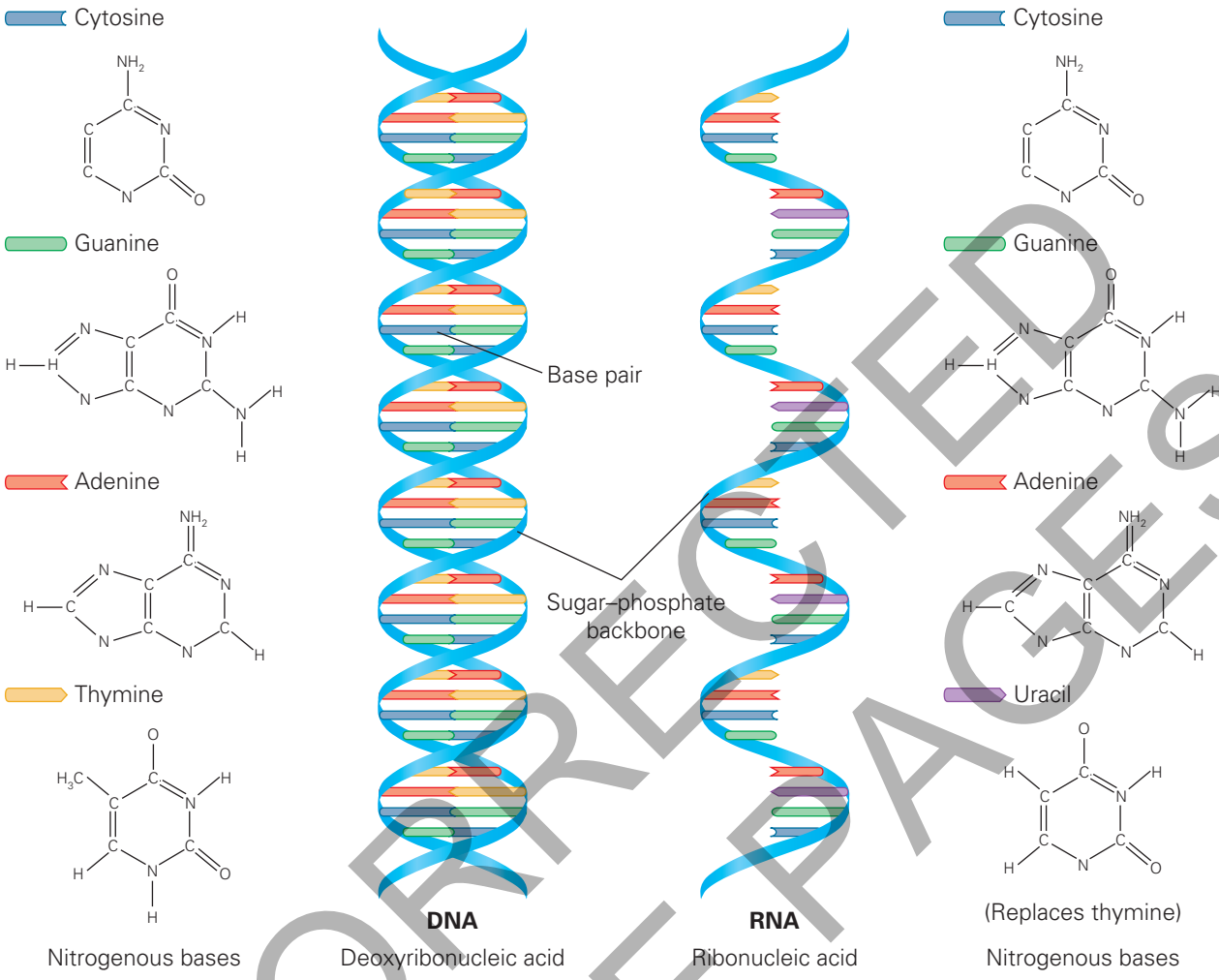


Figure 2.10 DNA and RNA

Different forms of nucleic acids

There are four types of biological macromolecules: proteins, carbohydrates, lipids (fats and oils) and nucleic acids. Nucleic acids are found in two forms: DNA and ribonucleic acid (RNA). The two types of nucleic acids differ in several key ways, which are outlined in Table 2.1 and shown in Figure 2.10.

Quick check 2.2

1. **Describe** the relationship between chromosomes, genes, DNA and base pairs.
2. **Contrast** the structures of DNA and RNA.

The purpose of DNA: Making proteins

Why do we need proteins?

Proteins are complex molecules that play a critical role in many body functions. We use approximately one billion proteins every day, just to function normally. Proteins help provide structure to cells and build and repair tissue. Protein molecules play many other varied roles in the body; they include haemoglobin molecules that carry oxygen around our body and the hormones that regulate our glucose levels and development. Collagen and keratin are structural proteins, making up the structural components of organs. Enzymes such as amylase and lipase are catalytic proteins that help control the rates of reactions. Other proteins help us fight pathogens, transport molecules and move our muscles.

WORKSHEET
Protein synthesis

Explore! 2.2

Protein functions

The many roles of protein include structural, contractile, transport, catalytic, regulatory and immunological functions. Research the following proteins and determine their function.

Protein	Function
Oxytocin	
Actin	
Ferritin	
Insulin	

The link between DNA and proteins

Genes are sections of DNA that hold the specific instructions required for making each of the proteins in our bodies. The sequence of the nitrogenous bases in a gene provides the instructions (or code) to produce a protein. A group of three nitrogenous bases is called a **base triplet/codon**, and these code for a particular amino acid. Amino acids are the monomers of proteins. The order of nitrogenous bases specifies the sequence of amino acids that forms a particular protein.

base triplet
a set of three nitrogenous bases that code for an amino acid

transcription
the first stage of protein synthesis in which the base sequence of DNA is copied into mRNA

translation
the second stage of protein synthesis in which a sequence of mRNA is translated into a sequence of amino acids

Quick check 2.3

1. **Recall** why proteins are important.
2. **Describe** the relationship between DNA, amino acids and proteins.

Protein synthesis

To synthesise (or build) proteins using the DNA code in genes, the gene must be expressed; that is, the instruction in the DNA is converted into a protein. This means two processes need to occur: **transcription** and **translation**.

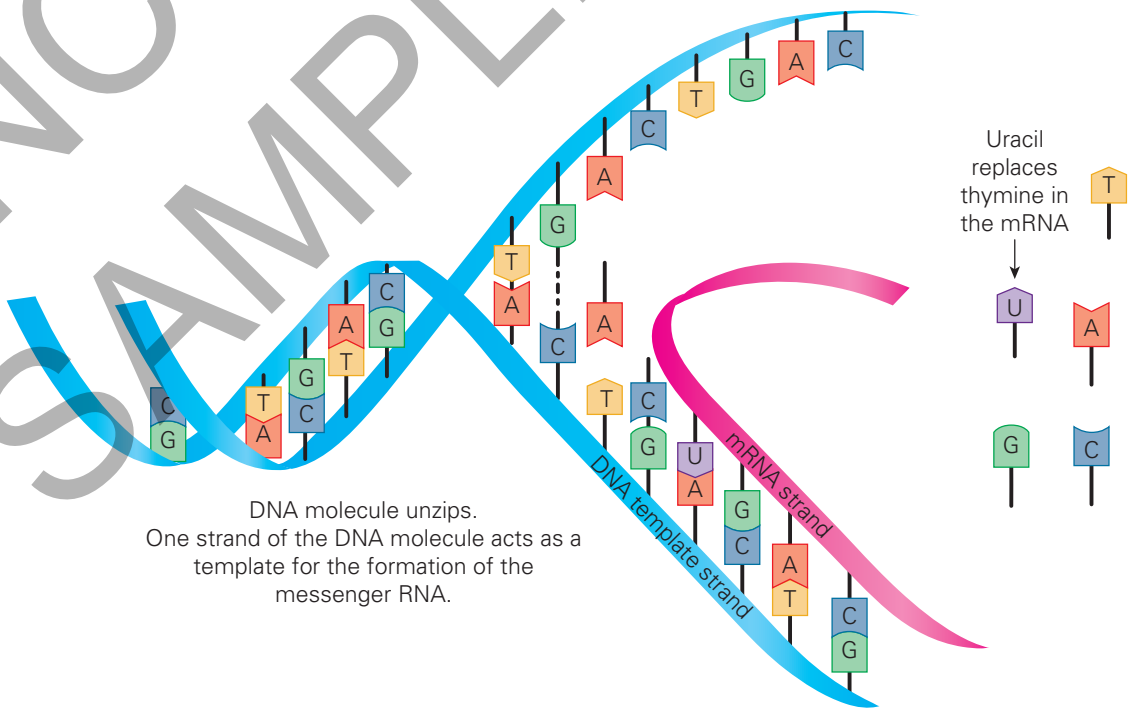


Figure 2.11 When the DNA double helix unwinds and unzips, one strand of the DNA acts as the template on which messenger RNA is formed.

To transcribe means to put into written form, so in this process the DNA code for a specific protein is 'written down' in the form of mRNA (messenger RNA).

To translate means to convert into another form, so in this process the code (written down as mRNA) is converted into a chain of amino acids known as a polypeptide chain. This can then be folded to become a protein.

Transcription

Transcription happens in the nucleus of the cell. The transcription process requires getting access to the DNA code in the genes, otherwise the code cannot be copied. Transcription begins when an enzyme (RNA polymerase) binds to the gene being copied. This causes the DNA double helix to unwind, allowing the enzyme to read the code on one of the DNA strands (called the DNA template strand). As the enzyme reads the code, it builds a complementary copy of the gene using RNA nucleotides: C, G, A and U. RNA follows the same complementary pairing rules as DNA, with one exception. Where DNA has the nitrogenous base thymine (T), RNA has uracil (U). The newly formed single-stranded RNA is called messenger RNA (mRNA). When transcribing DNA into mRNA, only one strand of DNA is used as a template. If both strands were transcribed, it would result in two different RNA sequences coding for different proteins, leading to confusion and potentially incorrect protein synthesis. When the mRNA peels away from the DNA template strand, the DNA double strands rejoin and wind back up into a helix. The mRNA then leaves the nucleus and goes to the site of protein production, the ribosomes.

Try this 2.2

Base pairings

Complete the base sequence of the DNA complementary strand and then the mRNA strand formed from the DNA template strand given below.

Complementary DNA	
DNA template	T A C C C G A A A G T G
mRNA	

Translation

Once mRNA reaches the ribosomes in the cytoplasm, it must be 'decoded' or translated to make the necessary protein. The nitrogenous bases in the mRNA are decoded in groups of three called **codons** (base triplets in the original DNA template strand). Each codon in the mRNA specifies which amino acid is added to the **polypeptide** chain.

WIDGET
Building a protein

Figure 2.12 simplifies the process of translation. In reality, translation requires several enzymes and another type of RNA called transfer RNA (tRNA). It occurs at the ribosomes, which may be freely floating in the cytoplasm or attached to the endoplasmic reticulum (ER), creating the rough ER. Each tRNA molecule transports one specific amino acid towards the ribosome for delivery.

codon
three nucleotides (base triplet) on mRNA that code for an amino acid

polypeptide
a chain of amino acids, forming part or the whole of a protein molecule

Table 2.2 shows the 20 different amino acids that form the building blocks for every protein. Using this table, you can identify the amino acid that each codon codes for. For example, if the first codon of your mRNA strand is AUG, you locate the A in the left column and the U in the top row and find where they intersect. At that spot is a box containing four different codons. You then find where the G in the right column of the table intersects and you will see your codon

AUG. Next to the codon it says Met, which means AUG codes for the amino acid methionine in eukaryotes. The codon AUG is unique as it is also a START codon. This means that it always is the first codon in the transcribed mRNA that undergoes translation. There are also three STOP codons which signal for the process of translation to stop rather than coding for another amino acid.

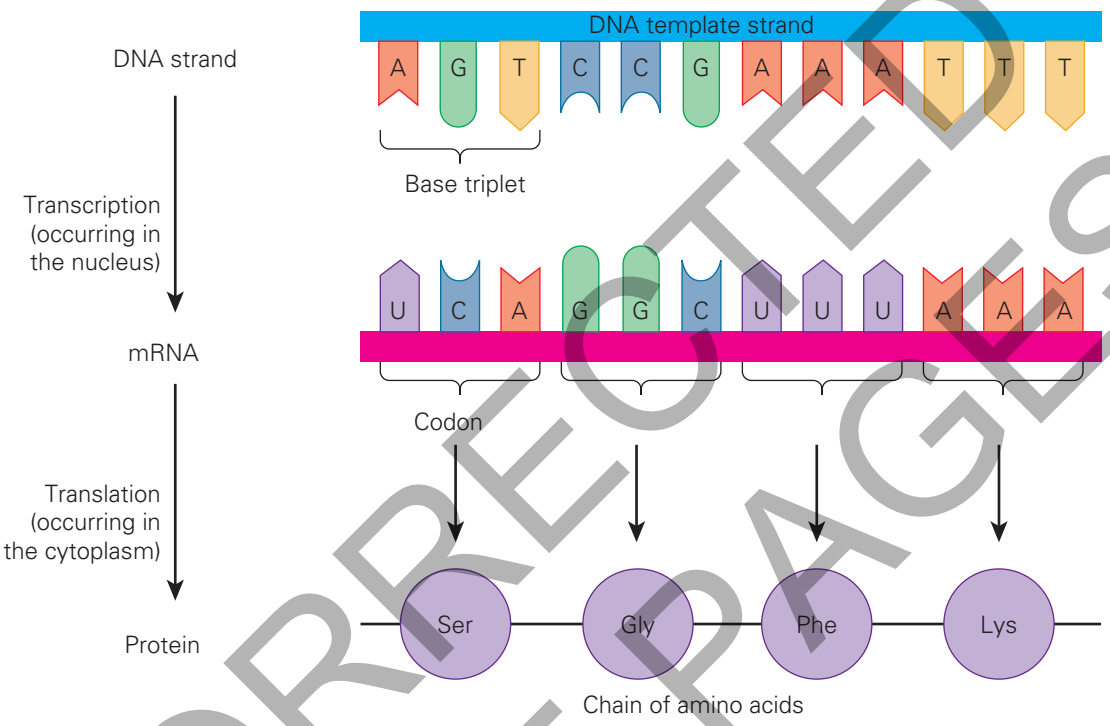


Figure 2.12 The process of transcription and translation from a DNA molecule to an amino acid chain

Second letter in the codon

First letter in the codon	U		C		A		G		Third letter in the codon
U	UUU	Phe (F)	UCU	Ser (S)	UAU	Tyr (Y)	UGU	Cys (C)	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu (L)	UCA		UAA	STOP	UGA	STOP	A
	UUG		UCG		UAG	STOP	UGG	Trp (W)	G
C	CUU	Leu (L)	CCU	Pro (P)	CAU	His (H)	CGU	Arg (R)	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln (Q)	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile (I)	ACU	Thr (T)	AAU	Asn (N)	AGU	Ser (S)	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys (K)	AGA	Arg (R)	A
	AUG	Met (M) START	ACG		AAG		AGG		G
G	GUU	Val (V)	GCU	Ala (A)	GAU	Asp (D)	GGU	Gly (G)	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu (E)	GGA		A
	GUG		GCG		GAG		GGG		G

Table 2.2 Table of amino acids showing that the genetic code is degenerate, which means that more than one codon can code for a single amino acid. This redundancy means that 61 of the 64 codons code for the 20 amino acids (the other three are stop codons).

Explore! 2.3

Using biotechnology to produce therapeutic proteins

Traditionally, vaccines contain a weakened virus, or a protein of the virus, to trigger the body's immune response and provide immunity.

Two of the common COVID-19 vaccines work in that way. AstraZeneca (a viral vector vaccine) contains material from the SARS-CoV-2 virus within the shell of another virus. Novavax (a protein subunit vaccine) contains part of the coronavirus spike protein which is injected directly.

mRNA vaccines have been in development for decades but had been progressing slowly. Financial backing from governments and commercial organisations is critical for research, and the significant funding provided worldwide to address the COVID-19 pandemic accelerated mRNA vaccine biotechnology.

mRNA vaccines work differently to traditional vaccines. Instead of delivering a viral protein, the vaccines deliver mRNA. COVID-19 mRNA vaccines contain instructions for making the spike protein from the SARS-CoV-2 virus. The Pfizer and Moderna vaccines package the mRNA into a lipid nanoparticle that can pass through the cell membrane.

Vaccine mRNA is modified to be more efficient at protein synthesis and resistant to degradation. This means it can be used as a template many times. Manufacturers use computer-controlled techniques for generating the required strands of RNA. This method is cheaper and safer than manufacturing proteins or growing viruses. mRNA vaccines have huge potential for preventing and controlling future epidemics and pandemics.

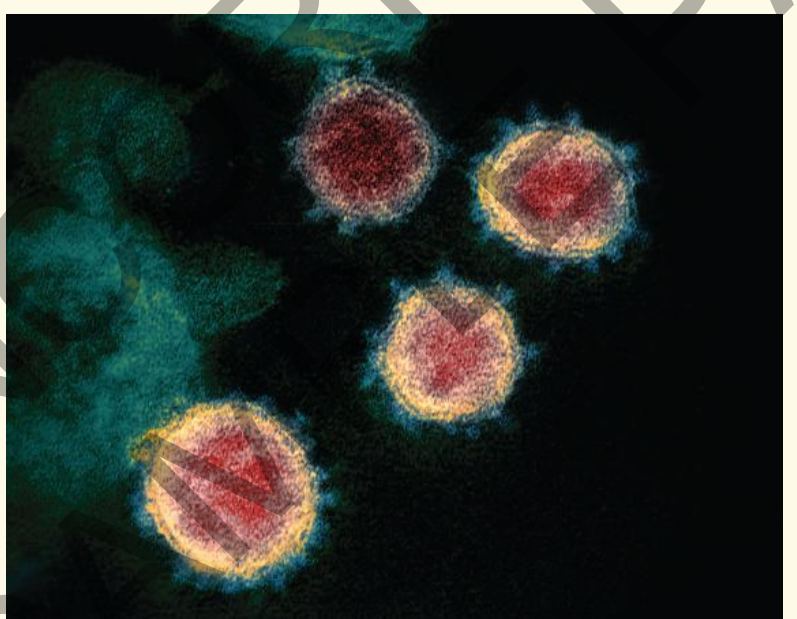


Figure 2.13 A colourised electron microscope image of SARS-CoV-2 virus particles. The spike proteins around the edge give coronaviruses their name, as *corona* is Latin for crown.

Quick check 2.4

1. The following codons are found in a strand of DNA: TAA TTA TCG ACT ACT AGC.
   State the complementary mRNA strand.
2. **Contrast** transcription and translation.
3. **Explain** why you do not need to read the code on both strands of DNA when transcribing DNA into mRNA.

Go online to access the interactive section review and more!

Section 2.1 review

Online quiz 

Section questions 

Teachers can assign tasks and track results 

Section 2.1 questions

Remembering

1. **Name** the basic building blocks or monomers of DNA.
2. **Recall** where proteins are created (synthesised) in the cell.
3. **Recall** the name for the shape of DNA.
4. **Illustrate** and label a nucleotide.
5. **Recall** the four bases found in DNA and the complementary base pair rule.
6. **Recall** why all mRNA strands start with the codon AUG.

Understanding

7. A template strand of DNA is found to contain the following bases: TAC GGA TCA TCG TGG GAA GCA GGC ATT.
   a) **State** the complementary DNA strand.
   b) Using the above template strand of DNA, **state** the bases on the mRNA strand.
   c) Using Table 2.2, **identify** the amino acids that this strand of DNA would code for.

Applying

8. **Describe** the purpose of DNA.
9. **Describe** the relationship between DNA, genes and chromosomes.
10. **Explain** how the body produces so many different proteins with only four different nitrogenous bases.
11. **Explain** how the genetic information found in the chromosomes within the nucleus of a cell reaches the ribosomes for protein synthesis.
12. Make a flow chart to **summarise** the key steps in transcription and translation.

Analysing

13. **Contrast** the structures of mRNA and DNA.
14. **Differentiate** between the functions of a structural protein and a catalytic protein, providing examples of each.

Evaluating

15. A template strand of DNA is found to contain the following bases: TCC TGA TGA TGG GGG GCA AAA CGC GTA.
    Something went wrong during the transcription of this template strand, and the mRNA strand contains the following bases:
    AGG ACU ACU ACC CUC CGU UUU GCG CAU.
    a) **Determine** the mistake in the mRNA strand above.
    b) **Propose** the outcome of this mistake on the protein produced.
16. **Propose** what may occur if there was a problem with a certain protein in your body.

2.2 Passing on genetic information

Learning goals

At the end of this section, I will be able to:

1. Recall the steps involved in DNA replication.
2. Describe the stages of cell division in mitosis and meiosis.
3. Use a karyotype to determine information about an individual.

WORKSHEET DNA replication and reproduction

VIDEO Sexual reproduction

Two different forms of reproduction occur among organisms: asexual and **sexual reproduction.** Sexual reproduction requires two organisms to each contribute a **gamete**; these are the sex cells that combine to produce a unique offspring. Gametes are formed in the **gonads** of the male (testes) and the female (ovaries). Each gamete contains half the genetic information needed to form a new organism of the same species. When the gametes – sperm and **ova** (eggs) – meet and fertilise, they form a **zygote** with a full set of genetic information. The zygote divides and as the number of cells increases, cells begin to take on special functions. Eventually, the zygote becomes an **embryo**.

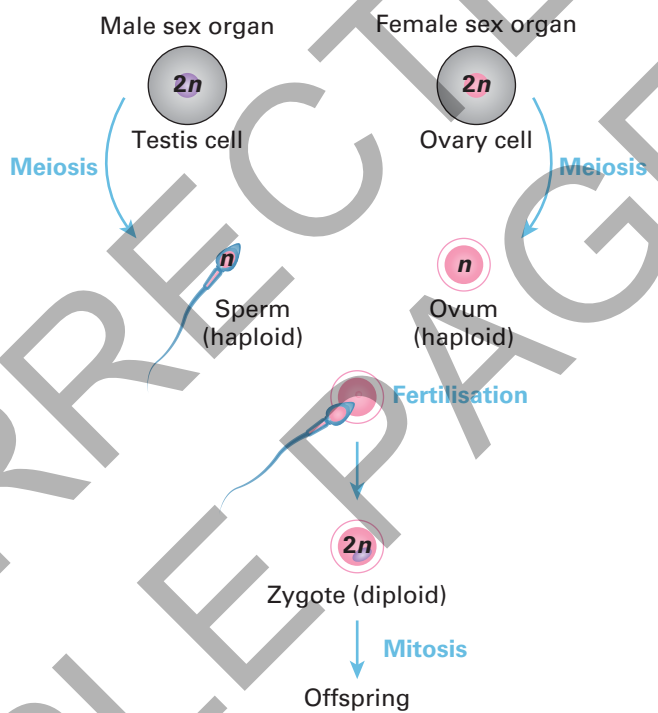


Figure 2.14 Sexual reproduction involves a gamete from a male and a female combining to form a zygote.

You will notice that Figure 2.14 has some new terms:

- Cells that contain only one set of chromosomes are known as **haploid (*n*)**. Gametes are haploid.
- Cells that contain two sets of chromosomes are known as **diploid (2*n*). Somatic cells** (body cells) and the zygote are all diploid.
- **Meiosis** is the name of the process by which the gonads make the haploid gametes.
- **Mitosis** is the name of the process by which diploid somatic cells make identical diploid copies of themselves for growth and repair.

In humans, the haploid number (*n*) is 23. This means that gametes contain 23 single unpaired chromosomes. When the egg and sperm meet and fertilise, the two sets of chromosomes come together and form the diploid zygote (2*n*). The zygote contains 23 pairs of chromosomes, meaning the diploid number (2*n*) in humans is 46. The haploid and diploid numbers vary between species, but always remain the same for all the organisms within that species. For example, the platypus has a diploid number (2*n*) of 52, which means it has 26 pairs of chromosomes in its somatic cells. This also means the egg and sperm of the platypus contain half this amount of genetic information, so its haploid number (*n*) is 26 chromosomes.

sexual reproduction form of reproduction that involves the fusion of gametes from two parents and introduces variation to the offspring

gametes sex cells (sperm and ova) with half the usual number of chromosomes

gonads the sexual organs: testes in males and ovaries in females

ova (singular: **ovum**) mature female reproductive cells

zygote a fertilised egg produced by the fusion of male (sperm) and female (ovum) gametes

embryo the initial stage of early development in multicellular

haploid (*n*) a cell containing only one set of unpaired chromosomes

diploid (2*n*) a cell containing two sets of chromosomes

somatic cells the body cells of an organism

meiosis the process by which the gonads make the haploid gametes

mitosis the process by which diploid somatic cells make identical diploid copies of themselves for growth and repair organisms

Did you know? 2.2

Different diploid numbers

The diploid number of chromosomes in a species is not related to whether an organism is bigger or more complicated. For example, a koala, a tammar wallaby and garlic all have a diploid number of 16 but are very different!

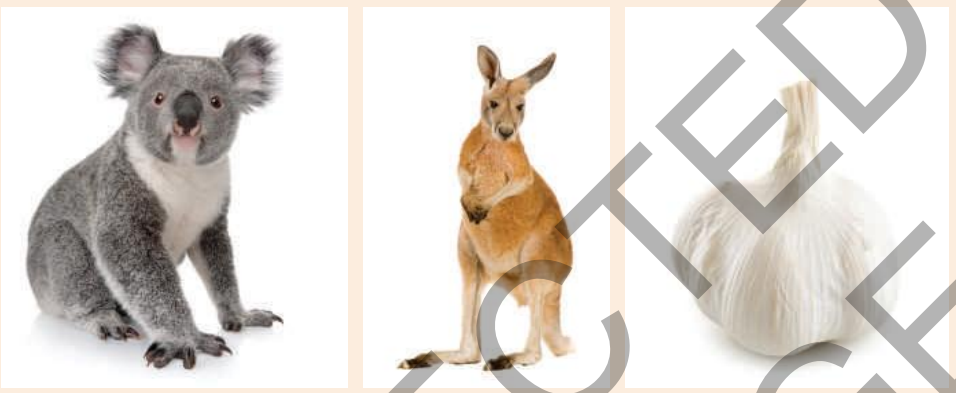


Figure 2.15 Some very different organisms contain the same number of diploid chromosomes.

Quick check 2.5

1. **Differentiate** between the terms 'haploid cell' and 'diploid cell', providing an example of each.
2. If a sheep's body (somatic) cell contained 54 chromosomes, **calculate** how many chromosomes would be found in its gametes.

How does DNA copy itself?

Before a cell divides, the DNA must be replicated to provide two copies of each chromosome, one for each new cell. This occurs before the processes of mitosis and meiosis. DNA replication must be precise, otherwise errors can affect the code in the DNA and lead to **mutations** (permanent changes in the DNA). The steps involved in DNA replication are outlined below and summarised in Figure 2.16.

mutation
a change in the genetic code of a cell

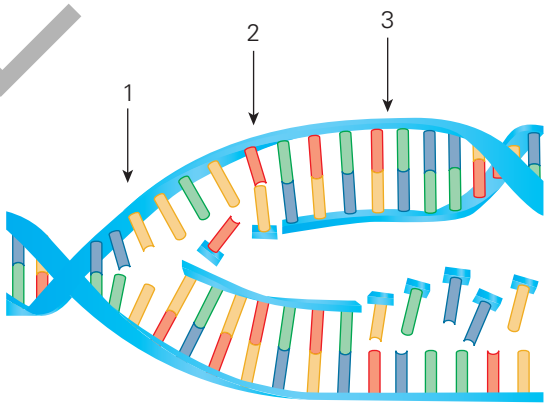


Figure 2.16 DNA replication: (1) the DNA must first unwind and unzip and then (2) the new DNA nucleotides can be added following complementary base pairing rules. (3) This results in two identical strands of DNA.

1. The DNA molecule unwinds and 'unzips' (breaks the hydrogen bonds) between the nitrogenous bases. This happens with the help of an enzyme called DNA helicase.
2. Another enzyme called DNA polymerase attaches new nucleotides to the exposed nitrogenous bases. The enzyme follows the complementary pairing rule, where adenine can only join with thymine, and cytosine can only join with guanine. The two new strands of DNA will be identical to the original parent strand.
3. The newly added nucleotides are connected with new hydrogen bonds, which results in two identical strands of double-stranded DNA being formed. Each new strand is one-half of the original strand and one-half that was newly built. For this reason, replication is sometimes described as being semi-conservative because half of the original DNA molecule is conserved in each of the two new molecules.

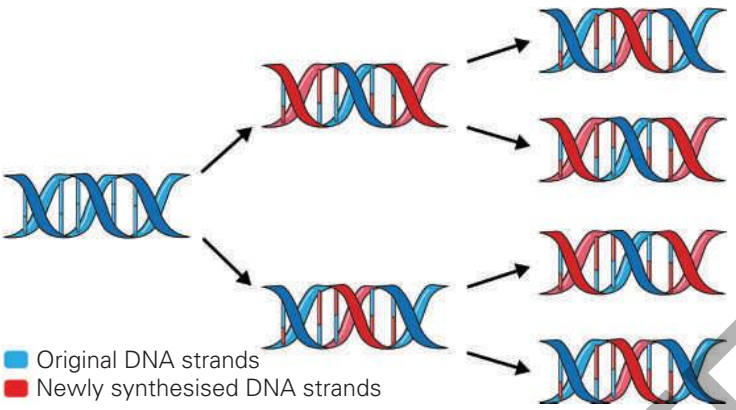


Figure 2.17 DNA replication is described as semi-conservative because the two new strands of DNA each contain one parent strand and one newly synthesised strand.

Quick check 2.6

1. **Explain** the purpose of DNA replication.
2. **Summarise** the steps of DNA replication.

Mitosis

Growth and repair of damaged tissues requires a form of cell division called mitosis. Mitosis is also important in asexual reproduction to make identical copies of cells. The diploid ($2n$) parent cell divides to form two diploid cells that are genetically identical, called daughter cells. The sequence of steps in mitosis is shown in Table 2.3. Mitosis produces every cell in the human body, except gametes, which are produced by meiosis and result in haploid (n) cells.

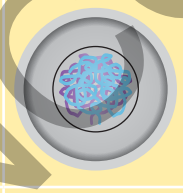
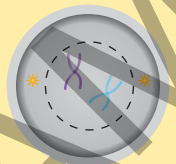
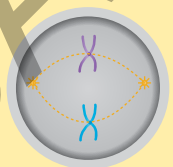
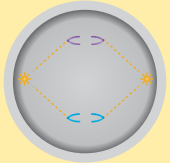
The stages of mitosis		
Interphase $2n$ (before mitosis)		• Parent cell is diploid ($2n$). • DNA replication occurs. • Chromosomes are not visible. • The cell gets bigger. • Organelles replicate.
Prophase		• The nuclear membrane breaks down. • DNA condenses and appears as distinct chromosomes. • Spindle fibres begin to form.
Metaphase		• Chromosomes are arranged along the centre of the cell. • Each chromosome is attached to a spindle fibre by the centromere.
Anaphase		• The sister chromatids split at the centromere and are pulled to either end of the cell by the spindle, centromere first. • Spindle fibres then begin to disappear.

Table 2.3 The stages of mitosis

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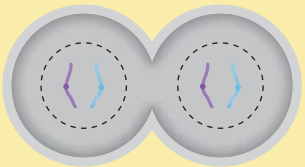
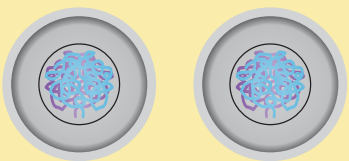
The stages of mitosis		
Telophase		• The nuclear membrane re-forms around the two sets of chromosomes, forming two new nuclei. • The chromosomes decondense and are no longer visible.
Cytokinesis 2 × 2*n* (after mitosis)		• Division of cytoplasm starts. • The cell pinches in half and divides into two genetically identical diploid daughter cells. • In plant cells, a structure called the cell plate is formed that transforms into a new cell wall.

Table 2.3 *(continued)*

Quick check 2.7

1. **State** which cells undergo mitosis.
2. **Summarise** the steps of mitosis.

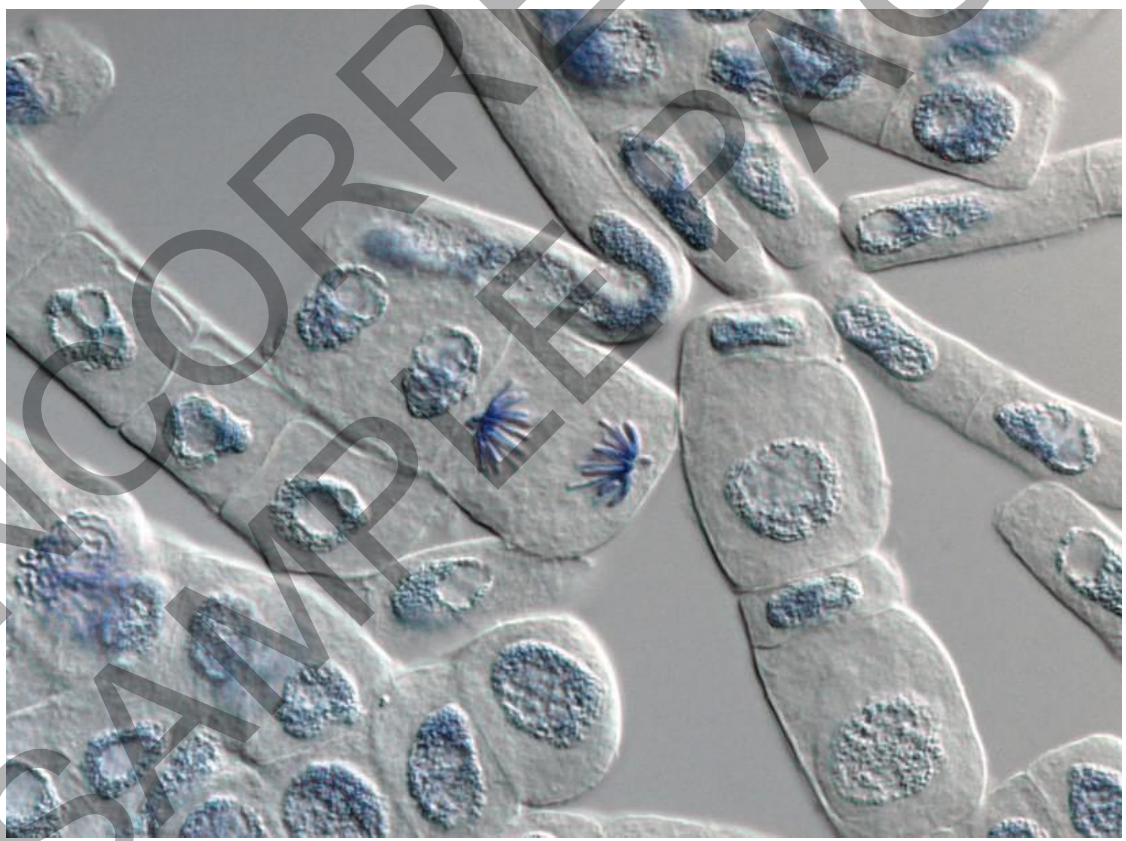


Figure 2.18 Plant cells in the process of mitosis viewed under a light microscope. The central cell shows the chromosomes moving to each end of the cell in anaphase. Can you identify some of the other stages?

Practical 2.2

Observing cells in a dividing root tip

Be careful

Ensure you carry the microscope appropriately: hold the arm with one hand and place one hand under the base. Ensure you don't make big changes in magnification, so as not to damage the glass slide.

Aim

To observe cells carrying out mitosis in a growing onion root tip.

Materials

- prepared, stained slide of the growing section of an onion root tip caught at different stages of cell division
- microscope

Method

1. Using the lowest magnification, place a prepared slide of a growing onion root tip on the stage of the microscope.
2. Position the slide so that the pointed narrow end of the root tip is clearly visible.
3. Look for a cluster of rapidly growing cells near this region.
4. Observe several different cells at various stages of cell division under the highest magnification of the microscope.

Results

Carefully choose one cell with a clearly outlined cell wall and showing chromosomes in a certain stage of cell division. Sketch a copy of what you observed, labelling the cell wall and the chromosomes. Remember to use a sharp pencil and to document the magnification the sketch is taken at.

Data processing

1. The mitotic index is a quantitative expression of the amount of cell division that a particular tissue is undergoing. It is the ratio of the number of cells undergoing mitosis to the number of cells that are not undergoing mitosis. It can be calculated using the following equation:

$$\text{Mitotic index} = \frac{\text{number of cells in the field of view undergoing division}}{\text{total number of cells in the field of view}}$$

   Calculate the mitotic index for the tissue sample you have observed in your microscope.
2. Using the field of view in your microscope, complete the table and calculate the percentage of cells in each cell cycle stage.

Cell cycle stage	Number of cells in that stage	Percentage of cells in that stage
Interphase		
Prophase		
Metaphase		
Anaphase		
Telophase		
Total		

3. Use the calculated percentages to predict which cell cycle stage is the longest and which is the shortest in your sample of cells. Explain your answer.

Discussion

1. Explain how you could tell which cells were dividing.
2. Describe the main features of each stage of cell division. Could you use these to identify the stage of mitosis for a particular cell?
3. List the structures in the cells you could observe. Which structures were present that you could not observe? Why might this be the case?

Meiosis

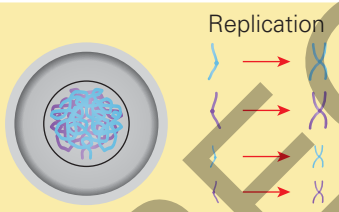
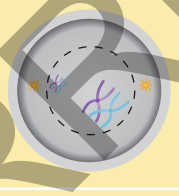
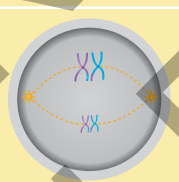
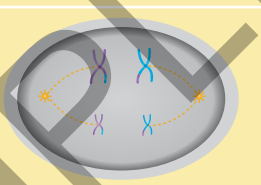
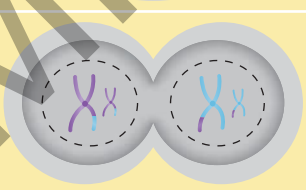
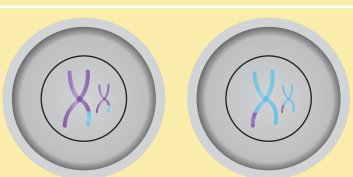
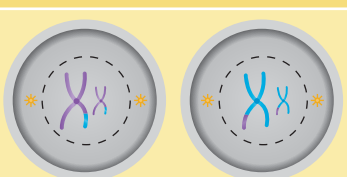
Meiosis is the process by which animals and plants produce gametes for sexual reproduction. It starts with a diploid ($2n$) parent cell, which contains two sets of chromosomes—one from each parent. To ensure that offspring receive the correct number of chromosomes, meiosis reduces the chromosome number by half, producing four haploid (n) daughter cells (gametes). This reduction in genetic material is why meiosis is called a **reduction division**.

reduction division cell division which results in a reduction of genetic material in the daughter cells

homologous chromosomes a matching pair of chromosomes, with one having been inherited from each parent; they have the same gene sequence and loci, length and centromere position

crossing over the exchange of genetic material between homologous chromosomes during meiosis, leading to genetic diversity in gametes

Meiosis occurs in two stages: meiosis I and meiosis II. Meiosis I separates homologous chromosome pairs, reducing the chromosome number from diploid to haploid. Meiosis II then divides the sister chromatids of each chromosome, similar to mitosis, ensuring that each gamete has a complete set of single chromosomes. These two divisions are essential for maintaining chromosome number across generations and increasing genetic diversity.

Meiosis I		
Interphase $2n$ (before meiosis)		• Parent cell is diploid ($2n$). • DNA replication occurs. • Chromosomes are not visible. • The cell gets bigger. • Organelles replicate.
Prophase I		• The nuclear membrane breaks down, the DNA condenses and chromosomes become visible. • **Homologous chromosomes** pair up and **crossing over** can occur.
Metaphase I		• Homologous chromosomes align at the equator.
Anaphase I		• Paired homologous chromosomes separate from each other and are pulled to opposite ends of the cell by the spindle fibres.
Telophase I		• The cell membrane pinches in. • The nuclear membranes re-form. • Two separate nuclei form.
Cytokinesis (after meiosis I)		• Division of cytoplasm starts. • The cell pinches completely into two haploid (n) daughter cells.
Meiosis II		
Prophase II		• Nuclear membranes disappear. • Spindle fibres re-form in both cells.

continued ...

Table 2.4 The stages of meiosis I and meiosis II

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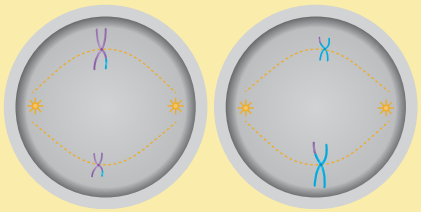
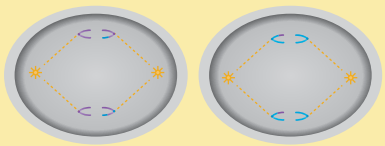
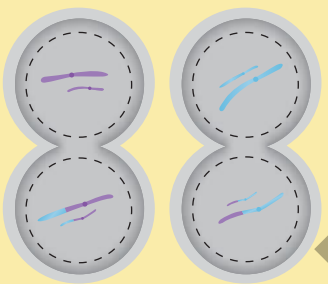
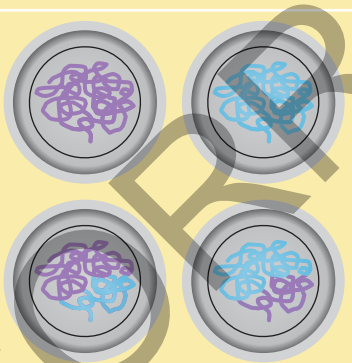
Metaphase II		• Chromosomes line up at the equator.
Anaphase II		• The chromosomes are pulled apart, separating the sister chromatids, which are pulled to opposite sides of the cell. • The spindle fibres disappear.
Telophase II		• The nuclear membrane re-forms. • The chromosomes start to become less visible.
Cytokinesis 4 × *n* (after meiosis II)		• Division of cytoplasm starts. • The cells produce four genetically different haploid daughter cells (*n*), each containing half the amount of DNA that was in the parent cell.

Table 2.4 *(continued)*

Did you know? 2.3

Creating variation

Crossing over can occur in prophase I of meiosis I when homologous chromosomes get so close together that some of their genetic material gets exchanged. It results in the chromosomes recombining to form a new combination of **alleles**, increasing the diversity of a species and therefore increasing the ability to respond to changing environments over time. This is how meiosis can increase genetic variation.

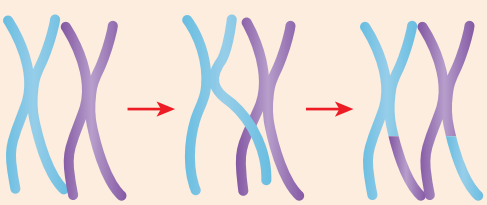


Figure 2.19 Recombination occurs when homologous chromosomes undergo crossing over in prophase I.

allele
different form of the same gene

recombination
the rearrangement of genetic material, especially by crossing over

Quick check 2.8

1. **Describe** why the process of meiosis is also known as 'reduction' cell division.
2. **Recall** where meiosis occurs in humans.
3. **Explain** why crossing over is an important process during meiosis.

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Fertilisation

fertilisation the fusion of male and female gametes to form a zygote

stem cells undifferentiated cells with the ability to develop into various specialised cell types

In humans, **fertilisation** occurs when two haploid gametes fuse to form a diploid zygote. For fertilisation to occur, the sperm must complete the acrosome reaction. The acrosome is a structure, located in the head of the sperm, that is filled with enzymes. These enzymes are released to digest their way through the corona radiata and zona pellucida, the outer layers of the ovum. Once the sperm penetrate these layers, the cell membranes of one of the sperm cells and the ovum fuse together, and the sperm nucleus can enter the ovum. The fusing of cell membranes also triggers a reaction that causes the zona pellucida to harden. This is called the cortical reaction, and ensures that only one sperm fertilises the egg, preventing polyspermy, which would lead to an abnormal number of chromosomes and non-viable embryos.

Stem cells

The zygote will develop into an embryo, which initially consists of **stem cells**. These cells are undifferentiated and have the potential to become any cell type if removed from the embryo. As the embryo grows, the stem cells can differentiate into specialised cells that form various tissues and organs. These embryonic stem cells, derived from early embryos, can differentiate into a wide range of cell types but are challenging to obtain.

In contrast, adult stem cells, found in specific body tissues like bone marrow and skin, have a more limited differentiation potential but are more accessible. Stem cells hold significant promise for medical treatments, including replacing damaged cells in conditions like type 1 diabetes and spinal cord injuries.

Explore! 2.4

HeLa cells

HeLa cells are a line of human cancer stem cells that have been widely used in scientific research since their discovery in 1951. These cells originated from a tissue sample taken from Henrietta Lacks, an African–American woman diagnosed with cervical cancer. Without her knowledge or consent, cells from her tumour were cultured and found to have the ability to survive and proliferate indefinitely.

Figure 2.20 A statue of Henrietta Lacks in Bristol, England, created by artist Helen Wilson-Roe. The statue was unveiled on the 70th anniversary of Lacks' death.

Find some reliable sources online to complete the following:

1. Investigate the history of HeLa cells, including the story of Henrietta Lacks and the ethical controversies surrounding the use of her cells without consent.
2. Explore various genetic technologies that utilise HeLa cells, such as cancer research, drug development and vaccine production.
3. Choose an ethical framework to guide your analysis (e.g. utilitarianism, deontology, virtue ethics, rights-based, care-based).
4. Reflect on the ethical concerns related to consent, ownership and the use of biological materials.
5. Construct a well-organised written argument that incorporates evidence from your research.
6. Clearly outline your chosen ethical framework and how it applies to the use of HeLa cells.
7. Discuss the ethical implications of continuing to use HeLa cells in genetic research, considering both the benefits and the controversies.

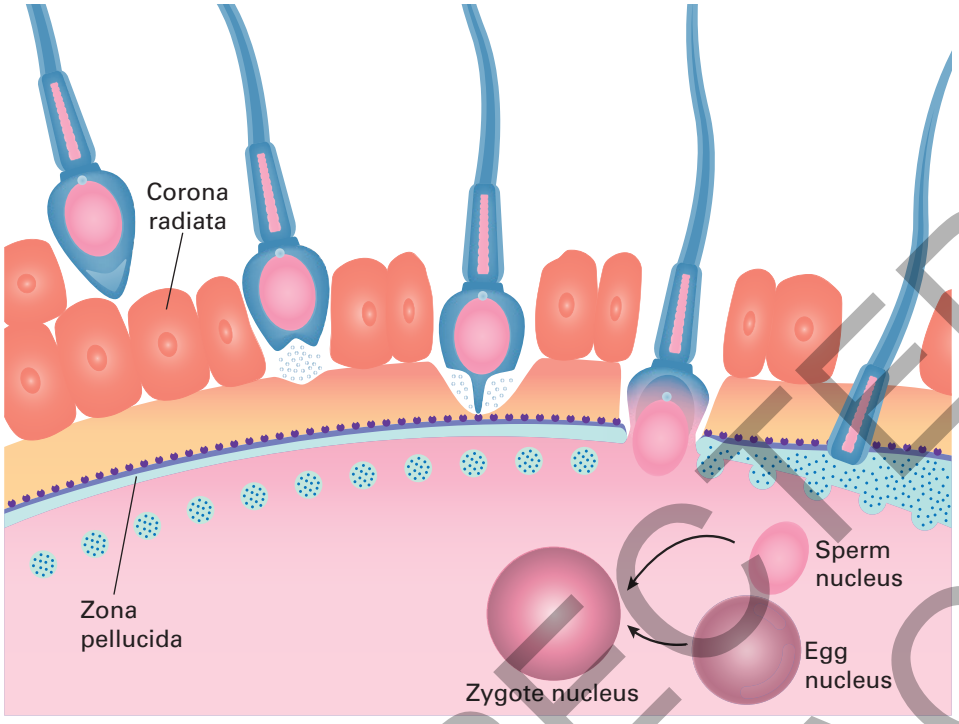


Figure 2.21 Once a sperm fertilises the egg, the nuclei of the sperm and the egg fuse.

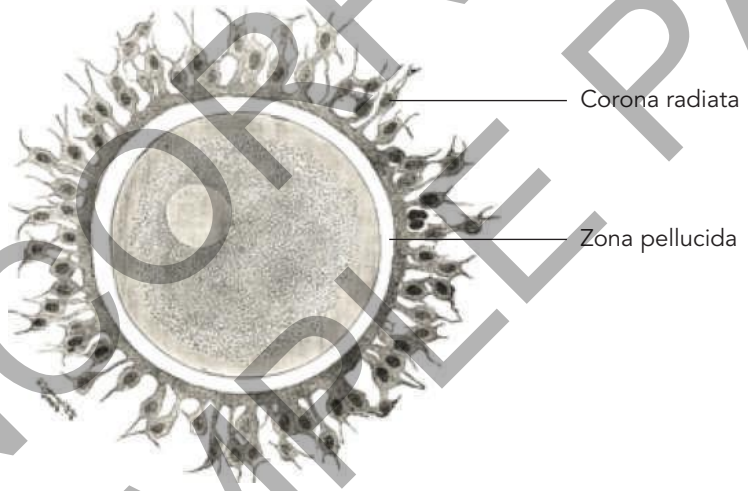


Figure 2.22 A human ovum

Quick check 2.9

1. Copy the table below and **define** the terms.

Term	Definition
Meiosis	
Fertilisation	
Zygote	
Haploid	
Diploid	

2. **Identify** the missing words in the following sentence.
Male gametes are called _____, whereas female gametes are called _____.

Explore! 2.5

Types of twins

You, or someone you know, might have a twin sister or brother. There are two main types of twins: identical (monozygotic) and fraternal (dizygotic). However, there is another, very rare form of twins known as semi-identical (sesquizygotic). Only two cases have ever been recorded, one in the US and one in South East Queensland. Conduct some research and answer the following questions:

1. Describe how monozygotic and dizygotic twins are formed.
2. Draw a diagram to show how these different types of twins are formed.
3. Discuss why sesquizygotic twins are so rare.

VIDEO
Mitosis and meiosis

Meiosis versus mitosis

Although the processes of meiosis and mitosis have similarities, there are some differences:

- In meiosis, chromosomes recombine by crossing over, producing chromosomes with new combinations of genes. In mitosis, chromosomes are replicated to make identical copies of themselves.
- Meiosis involves two divisions that produce four haploid (n) daughter cells containing unique genetic material that is different to the parent cells. Mitosis involves one division that produces two diploid ($2n$) daughter cells with genetic material identical to the parent cell.

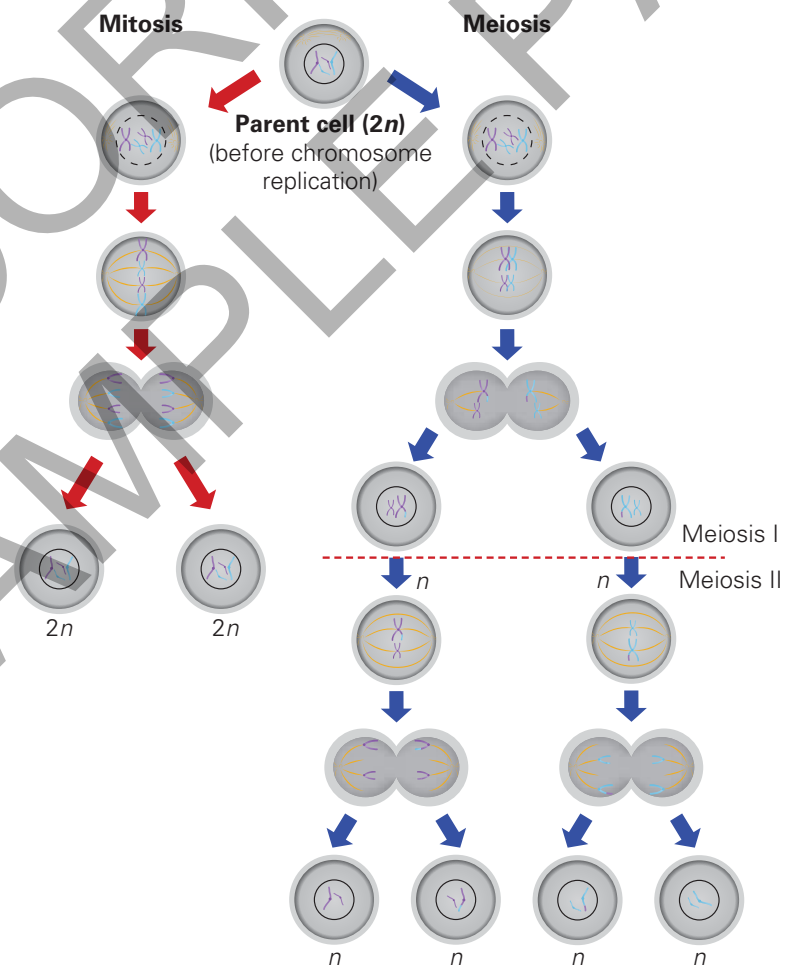


Figure 2.23 A summary of the processes of mitosis and meiosis

Karyotypes

The number of chromosomes varies between species, but in diploid organisms, chromosomes in body cells are always present in pairs. For example, humans have a total of 46 chromosomes, consisting of 22 pairs of homologous chromosomes and one pair of sex chromosomes in the nucleus of each body cell. However, gametes contain only 23 chromosomes – 22 autosomes and one sex chromosome – ensuring that the correct chromosome number is restored during fertilisation.

Homologous chromosomes are present in body cells because we receive one from each parent. Homologous chromosomes have the same length, centromere position and gene band patterns and can be displayed in a **karyotype** (Figure 2.24). Chromosomes that do not match (chromosomes from different pairs) are called **non-homologous chromosomes**.

karyotype the chromosomes of an individual, often displayed according to size and gene band patterns

non-homologous chromosomes chromosomes that do not belong to the same pair

autosome any chromosome that is not a sex chromosome. In humans, these are chromosome pairs 1 to 22

sex chromosomes chromosomes that determine the sex of an organism

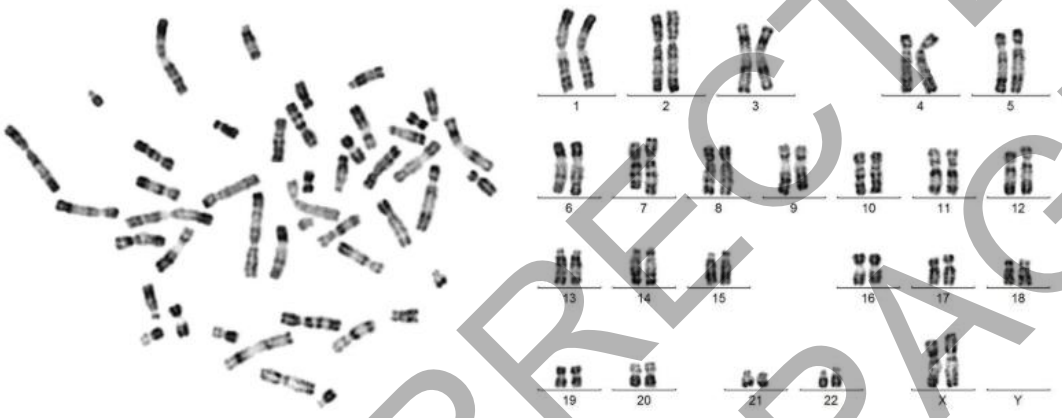


Figure 2.24 The homologous chromosomes seen under a microscope (left) can be arranged according to size in a karyotype (right). Pairs 1–22 are autosomes, while pair 23 is the sex chromosomes. The presence of two X chromosomes indicates this is a karyotype from a female.

Autosomes

Of the 23 pairs of chromosomes in humans, pairs 1 to 22 are known as **autosomes**. These 44 chromosomes are found in both males and females.

Sex chromosomes

In humans, the chromosomes making up the 23rd pair are the **sex chromosomes**. They contain genes that determine the sex of the individual. Females receive two X chromosomes (one from each parent), whereas males receive an X chromosome from the mother and a Y chromosome from the father (Figure 2.25).

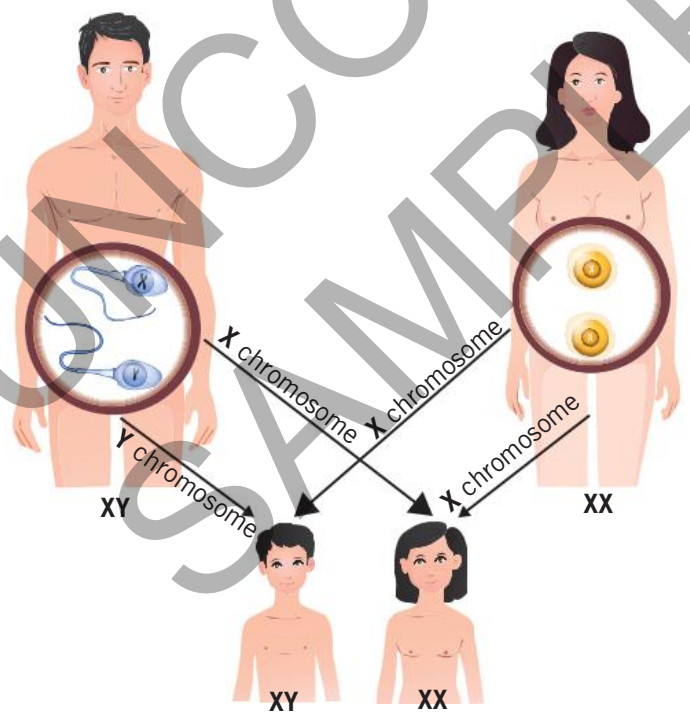


Figure 2.25 If an egg is fertilised by a sperm carrying an X chromosome, the child will be female. If an egg is fertilised by a sperm carrying a Y chromosome, the child will be male. Therefore, it is the male's sperm that determines the sex of the child.

Practical 2.3

What are the chances?

Aim

To investigate the chances of conceiving a male or female at fertilisation.

Useful formulas

$$\text{Theoretical probability (\%)} = \frac{\text{number of a particular outcome (e.g. heads)}}{\text{total number of all possible outcomes (e.g. heads and tails)}} \times 100$$

$$\text{Experimental probability (\%)} = \frac{\text{number of a particular outcome (e.g. heads)}}{\text{total number of all experimental trials (e.g. heads and tails)}} \times 100$$

$$\text{Percentage error (\%)} = \frac{(\text{experimental probability} - \text{theoretical probability})}{\text{theoretical probability}} \times 100$$

Materials

- one coin

Method

1. Copy the results table into your science journal.
2. Calculate the theoretical probability of landing heads during a toss.
3. You will now calculate the experimental probability of your own experiment by tossing the coin 50 times, recording the number of heads and tails in the table.

Results

1. Calculate the experimental probability of receiving a head and a tail based on both the experimental data for your own and the class results.
2. Calculate the percentage error, comparing the difference between the theoretical and experimental probabilities for both your own data and the class data.
3. Optional: Graph the relationships between the number of trials and percentage error.

	Individual results	Class results
Number of heads		
Number of tails		
Total number of trials (throws)		
Experimental probability		
Percentage error		

Discussion

1. Compare the experimental probabilities of your own and the class results.
2. If a head represents the Y chromosome and a tail represents the X chromosome, explain how accurately this models the process of fertilisation. What would the coin represent?
3. Make a claim about the chances of conceiving a male or female based on this activity.
4. Discuss whether increasing the number of trials affects the percentage error of the experiment.

Did you know? 2.4

Sex chromosomes in the animal kingdom

In some animals, such as birds, females have two different sex chromosomes (ZW) while males have two of the same (ZZ). In some reptiles, the temperature of the incubating environment determines the sex of the embryo. In some insects, females are XX and males have one X chromosome (written XO).

Monotremes have a complex sex chromosome system. Female platypuses have five pairs of X chromosomes and males five X and five Y chromosomes. Female echidnas have five pairs of X chromosomes and males five X and four Y chromosomes.

Figure 2.26 Female platypuses have five pairs of X chromosomes.

Try this 2.3

Can you sort chromosomes into a karyotype?

Go online and find a karyotype activity. There are many interactives that allow you to sort chromosomes into order and ones that allow you to analyse karyotypes.

Science inquiry 2.1

Genetic testing

When a person is pregnant, their doctor may conduct several tests to screen for genetic and chromosomal abnormalities in the foetus. Creating a karyotype lets the doctor check for abnormalities in the number or length of chromosomes. If anything appears abnormal, the doctor can undertake further genetic tests to check for specific disorders. One such test is chromosomal microarray (CMA) testing, which is much more detailed genetic testing than karyotyping. Chromosomal microarray testing detects around one abnormality in every 70 foetal samples with a normal karyotype. Scientists think that CMA testing will replace karyotyping for prenatal testing.

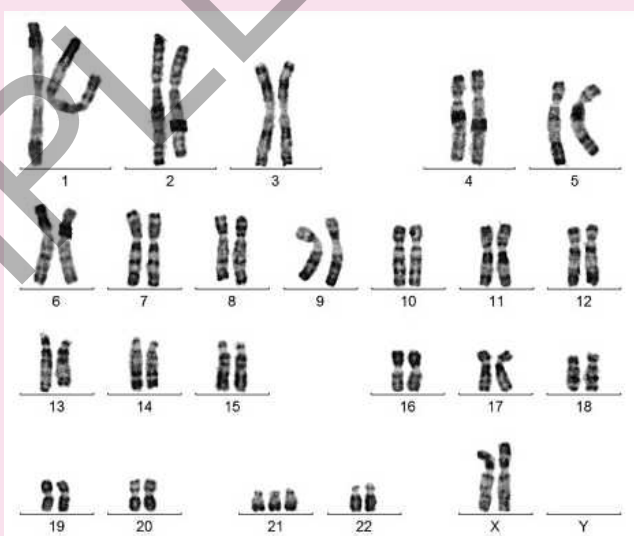


Figure 2.27 A well-known chromosomal abnormality is trisomy 21 or Down syndrome, where the foetus has three copies (instead of two) of chromosome 21.

continued ...

Genetic testing will identify carriers of genetic mutations and help people make informed decisions, such as in embryo selection. Genetic counsellors work with people who are concerned about having, or have been diagnosed with, a genetic condition.

Many people are also having genetic testing to discover more information about their ancestry or to connect with unknown relatives. Although it is against the law to be discriminated against because of genetic information, there is some concern regarding its use, either personally or by organisations such as insurance companies or medical facilities.

Have a class discussion regarding the ethical issues of non-therapeutic genetic testing performed by commercial companies. You may want to consider privacy concerns, the possible emotional impact, potential lack of medical knowledge and possible exploitation of vulnerable individuals.

Asexual reproduction

Asexual reproduction is a mode of reproduction in which a single parent produces offspring that are genetically identical to itself, meaning they are clones of the parent organism. Unlike sexual reproduction, which requires the combination of genetic material from two parents, asexual reproduction doesn't involve gametes or fertilisation.

There are various forms of asexual reproduction, each suited to different organisms:

- Binary fission: Common in single-celled organisms like bacteria, where the cell divides into two, each becoming an identical organism.
- Budding: Seen in organisms like yeast and hydra, where a new individual grows off the parent organism and eventually detaches to live independently.
- Fragmentation: Certain animals, like starfish and some plants, can grow into a complete organism from a fragment of the parent body.
- Vegetative propagation: In plants, new individuals grow from stems, roots, or leaves. Examples include strawberry runners and potato tubers.
- Spore formation: Many fungi, algae and some plants reproduce through spores, small structures that develop into new organisms without fertilisation.
- Parthenogenesis: Found in some reptiles, insects, and fish, where an egg cell develops into a new organism without fertilisation, producing offspring genetically similar to the mother.

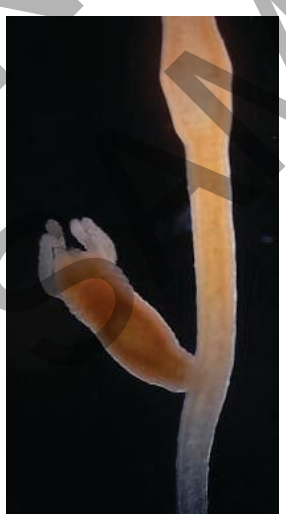


Figure 2.28 Budding hydra **Figure 2.29** Vegetative propagation

A key advantage of asexual reproduction is its efficiency; organisms can reproduce quickly and in stable environments, allowing populations to grow rapidly. However, because offspring are genetically identical to the parent, there is less genetic variation, which can make the population vulnerable to diseases and changing environmental conditions.

Quick check 2.10

1. **Compare** mitosis and meiosis using a Venn diagram.
2. **Explain** what karyotypes can show and why this is useful.

Section 2.2 review

Online quiz 

Section questions 

Teachers can assign tasks and track results 

Go online to access the interactive section review and more!

Section 2.2 questions

Remembering

1. **State** one key difference between mitosis and meiosis.
2. **Recall** the phase of meiosis when pairs of matching chromosomes separate.
3. **State** the name of the syndrome that arises from trisomy 21.
4. **State** what the term 'homologous chromosomes' means.

Understanding

5. **Explain** why gametes need to be haploid.
6. **Explain** what is meant by the term 'reduction division'.
7. **Explain** why a chromosome missing a part may lead to a disorder or syndrome.
8. **Explain** why the cells produced during meiosis are genetically unique.

Applying

9. Complete the following table to **differentiate** between mitosis and meiosis.

	Where it occurs	Purpose	Features of daughter cells
Mitosis			
Meiosis			

10. **Contrast** the daughter cells produced in mitosis versus meiosis.
11. **Illustrate** a diagram to represent the steps of mitosis for a cell with two pairs of chromosomes. Be sure to use a different colour for each pair of chromosomes.

Analysing

12. **Contrast** anaphase I in meiosis and anaphase in mitosis.
13. **Contrast** prophase I and prophase II in meiosis.

Evaluating

14. **Critique** the following claim: It is the male that determines the sex of the child.
15. **Predict** the chances of a woman giving birth to a girl if she has already given birth to three boys, giving reasons for your response.

2.3 Patterns of inheritance

Learning goals

At the end of this section, I will be able to:

1. Define the terms 'homozygous', 'heterozygous', 'dominant' and 'recessive' with respect to genetic inheritance.
2. Use and interpret Punnett squares to identify possible genotypes and phenotypes of offspring.
3. Use pedigree diagrams to determine the type of inheritance that is occurring.
4. Explain codominance, incomplete dominance and sex linkage.

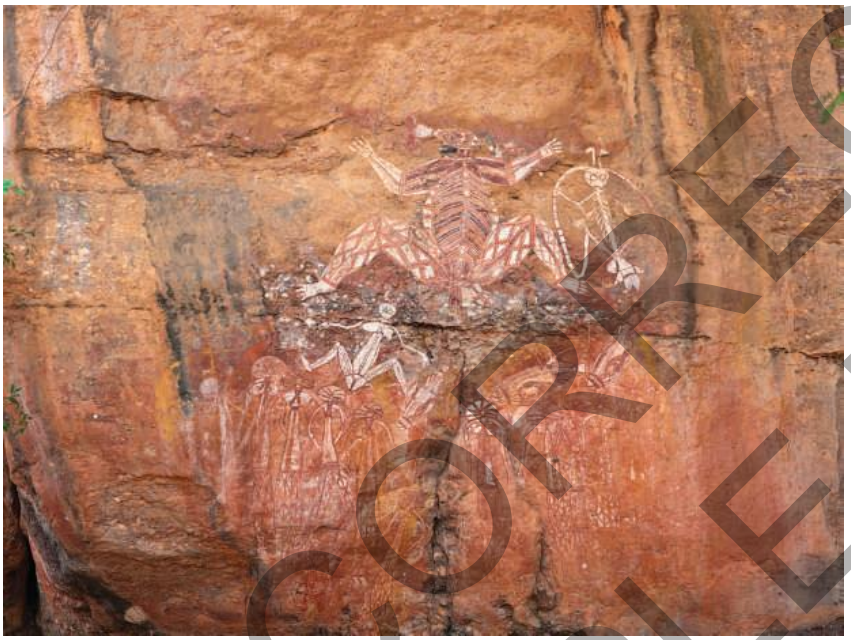


Figure 2.30 This artwork from Kakadu National Park shows Namondjok, a Creation ancestor who broke kinship laws.

Aboriginal and Torres Strait Islander communities have understood inheritance patterns for a long time. They recognise how traits can be passed down to the next generation and how negative traits and illnesses often appear in the children of closely related parents. Because of this deep knowledge, communities created complex and sophisticated kinship systems which allow individuals to understand who they are and how they fit into the social structure and even the universe. These kinship systems are strictly adhered to and dictate who can or cannot marry through marriage laws. Kinship and family structures also determine how individuals behave towards others and the responsibilities each person has towards others, natural resources and the land. For example, a person may be obliged to care for their sibling's offspring. These children usually address their aunt or uncle as 'mother' or 'father' and their cousins as 'brothers' or 'sisters'. Although they are aware of their biological parents, the societal (kinship) laws assign equal importance to other relatives. When conversing with one another, Aboriginal and Torres Strait Islander people usually speak Aboriginal English and frequently use the terms 'brother' or 'sister', which are derived from the kinship terminology and connections.

Inheriting traits from our parents

This cultural understanding of inheritance is reflected in modern genetics. Inheriting traits from our parents is now known to be governed by genes – specific sections of DNA passed from parent to offspring. Each characteristic or trait that we inherit is controlled by one or more pairs of genes. Members of each gene pair are found at the same location on their respective homologous chromosomes. This is called the **locus** of a gene.

locus (plural: loci) the location of a gene on a chromosome

Genes are sections of DNA that code for a particular protein that contributes to our characteristics, such as a gene for eye colour. However, not all forms of a gene are the same; for example, different people can have different eye colours. We use the term 'allele' to describe the different forms of a gene.

We get one copy of each chromosome from each parent, resulting in us inheriting one copy of each gene from each parent. The combination of alleles an organism has for a particular gene is known as the organism's **genotype**. If two identical alleles are present in a person's genotype, they are known as **homozygous** for that particular trait. However, if there are two different alleles present for the same trait, then the person is **heterozygous** for that trait.

For example, consider the trait of freckles. This trait is controlled by a single gene that is represented using two alleles. The allele of the dominant trait is the presence of freckles (we assign it the capital letter 'F' to show it is dominant), and the allele for the recessive trait is the lack of freckles (assigned a lower case 'f' to show it represents the recessive trait). A trait or characteristic is described as **dominant** if you need only one allele for it to be expressed (FF or Ff). For a **recessive** characteristic, the allele needs to be inherited from both parents for it to be expressed (ff).

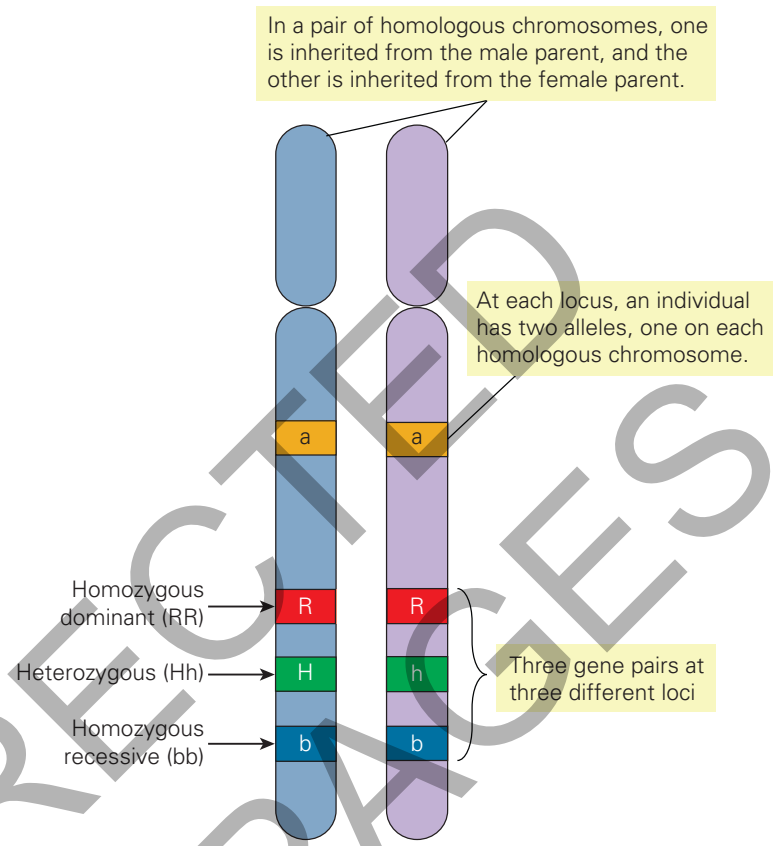


Figure 2.31 Gene loci on homologous chromosomes

Did you know? 2.5

The genome

The **genome** refers to all the genetic material in the chromosomes of an organism, including its genes and DNA sequences. The human genome is made of approximately 3.2 billion nitrogenous base pairs, which make up about 20 000 genes on 23 pairs of chromosomes. Other organisms have different genome sizes. Surprisingly, only a small percentage of the genome, around 1-2%, contains genes that code for proteins; the vast majority of the genome is made up of non-coding DNA which does not directly code for proteins, although it can still play important roles in gene regulation and other cellular functions.

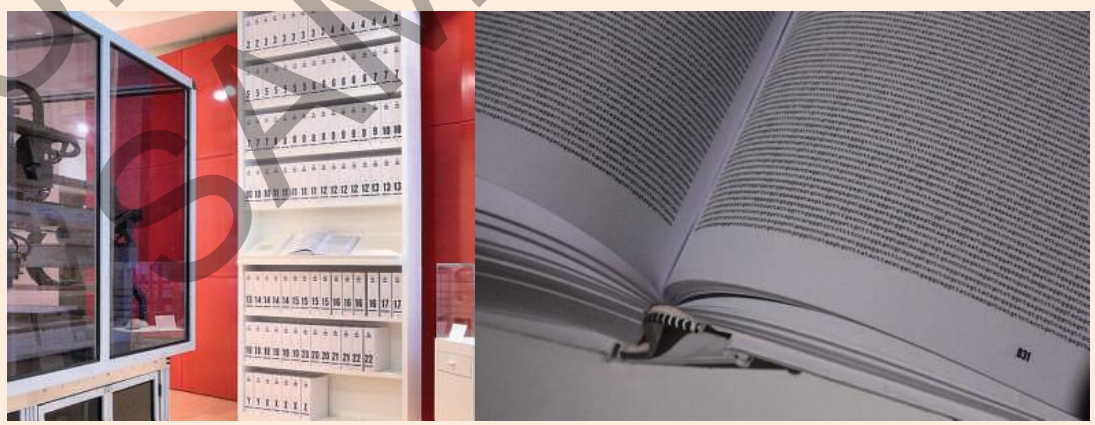


Figure 2.32 The first publication of the human genome was printed as 109 books in 23 volumes. The around three billion units of human DNA code are printed in a small font, and each book is a thousand pages long.

genotype
the combination of alleles an organism has for a particular gene

homozygous
having two identical alleles at a particular gene locus

heterozygous
having two different alleles at a particular gene locus

dominant
a characteristic in which the allele responsible is expressed in the phenotype, even in those with heterozygous genotypes

recessive
a characteristic in which the allele responsible is only expressed in the phenotype if there is no dominant allele present

genome
the complete set of genetic material in an organism

Figure 2.33 The presence of freckles is an example of a dominant trait.

If you inherit the freckles allele from both parents, you are homozygous dominant for that trait and your genotype will be FF. If you inherit the no-freckles allele from both parents, you are homozygous recessive for that trait and your genotype will be ff. If you inherit a freckles allele from one parent and a no-freckles allele from the other parent, you are heterozygous for that trait and your genotype will be Ff.

The interaction of an organism's genotype with the environment results in its **phenotype**, or its physical appearance. Typically, multiple genes and multiple environmental factors interact in the development of most traits. In the example in Figure 2.33, the presence of freckles is the person's phenotype for that trait. The freckles are present due to the person's genotype and their environmental exposure to UV light.

phenotype the observable characteristics of an organism, resulting from both genotype and the environment

To determine whether an allele represents a trait that is dominant or recessive, scientists look at the phenotype of a heterozygous organism. If the trait is expressed in this phenotype, it must be the dominant trait.

Quick check 2.11

1. Copy the table below and **define** the terms.

Term	Definition
Locus	
Allele	
Genotype	
Phenotype	
Dominant	
Recessive	
Heterozygous	
Homozygous dominant	
Homozygous recessive	

2. **Explain** why a genotype can indicate that a person will have freckles, but there are not many on their face.

Practical 2.4

Inherited features

Aim

To use data to determine whether some key characteristics are dominant or recessive traits.

Materials

- mirror

Optional extension: Collect information from other students or family members for further validity analysis.

Method

Part 1: Prepare the results table

1. Copy the results table into your science journal.

continued …

Part 2: Collect data on the variables

1. Identify whether you possess each of the inheritable features described below.
2. Record your results in the table.
3. Find out how many members of your class share the same features as you do. Record your results in the table.

   Optional: Find out how many members of your family display the traits.

   a) Attached ear lobes: are your earlobes attached or unattached to the side of your head?

   b) Widow's peak: do you have a V-shaped peak in the centre of your hairline as shown in Figure 2.34?

   c) Tongue roll: can you roll your tongue as shown in Figure 2.35?

   d) Front tooth gap: is there a definite gap between your two top front teeth as shown in Figure 2.36?

   e) Mid-digital hair: is there any hair on the second joint of at least one of your fingers?

   f) Long second toe: is your second toe longer than your big toe as shown in Figure 2.37?

Figure 2.34 This girl has a widow's peak (V-shaped peak) in the centre of her hairline.

Figure 2.35 This woman is able to roll her tongue.

Figure 2.36 This man has an obvious gap between his top front teeth.

Figure 2.37 The second toe is longer than the big toe on this foot.

Results

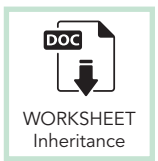
1. Calculate the total number of people sampled in this survey.
2. Calculate the percentage presence of the trait in your family and within the class.
3. Draw an appropriate graph (or graphs) to compare the percentage presence of each trait within your family with the percentage presence in the whole class.

Feature	Trait present in you? (Y/N)	Number of family members with trait	Presence of trait in family (%)	Number of class members with trait	Presence of trait in class (%)	Presence of trait in total sample (%)
Attached ear lobes						
Widow's peak						
Tongue roll						
Dimples						
Mid-digital hair						
Long second toe						
Sample size (*Total number of people surveyed*)	1					

continued →

Discussion

1. Describe which traits were commonly found in your family.
2. Describe which traits were commonly found in the class.
3. Describe which traits were commonly found in the whole sample.
4. Do your findings support the idea that a feature such as tongue-rolling is inherited? Explain your answer.
5. Predict from the data which traits are likely to be recessive and dominant. Justify your answer with data.
6. If you repeated the survey on the same people, would you get a similar result? Support your answer relating back to the theory behind inheritance of traits.
7. Can reliable conclusions and predictions be drawn from the results?
8. Compare the values from your family with those from the class. Were there any significant differences with some traits? Justify your answer with data.
9. Compare the values from your family with results from several other student families. Were there any significant differences with some traits? Justify your answer with data.
10. Can you draw valid conclusions as to which traits are dominant and recessive from this data? Justify your response.
11. Sample size is a factor that commonly affects data in small surveys. Discuss how this relates to the activity.
12. Can you identify possible limitations of this method? Justify your answer.
13. Suggest any changes that could be made to the method to improve the quality of the data in future surveys. Justify your suggestions by explaining how each change will improve the data quality.



WORKSHEET
Inheritance

Determining chances of inheritance

Gregor Mendel

Gregor Mendel is known as the father of genetics. Born in 1822, he was an Austrian monk who carried out experiments in his garden using pea plants and discovered some of the fundamental laws of heredity we still rely on today.

Mendel looked at the features of pea plants, which could be tall or short (dwarf). He assigned the following letters to represent these alternative characteristics (known today as alleles): T = tall and t = dwarf. Mendel bred the plants and observed the height of the plants in the next generation. He discovered that tall pea plants could be either pure breeding (TT or homozygous dominant) or hybrid (Tt or heterozygous). The dwarf plants could only be tt (homozygous recessive). Today, we call this the plant's genotype.

Mendel also used the following notation to represent different levels of generations:

- P = parent generation
- F_1 = first **filial** generation (first generation of offspring)
- F_2 = second filial generation (second generation of offspring).

filial
offspring of a cross

An easy way to predict the outcome of **monogenic** traits is to use a **Punnett square**, a specialised grid invented by Reginald Punnett. A Punnett square is a visual representation of a **monohybrid cross**, which is a breeding experiment involving two individuals that differ in a trait of interest. In a monohybrid cross, the parents are typically represented by letters to denote their respective alleles for the trait being studied.

Figure 2.38 Gregor Johann Mendel was the founder of the science of genetics, using pea plants to show dominant and recessive traits. He produced large amounts of data which all supported his theory, but later statistical analysis suggests that he may have falsified his data – it was simply too good to be true! Large data sets and statistical analyses are vital for validating scientific findings.

monogenic
a trait that is controlled by a single gene

Punnett square
a specialised grid to show genetic crosses

monohybrid cross
a type of genetic cross or breeding experiment that considers the inheritance of a single trait or characteristic, typically controlled by a single gene with two alleles

To use a Punnett square, you need to know the genotype of the parents to work out what genetic information they could pass on to their offspring via the gametes. For example, we know a heterozygous, tall pea plant has the genotype Tt. This means that when the plant makes its gametes, they could contain either a T or a t, as gametes only carry one of the alleles from the parent. In contrast, a homozygous tall pea plant has the genotype TT, so its gametes will all contain a T.

Worked example 2.1

Cross between a pure-breeding tall plant and a pure-breeding dwarf plant

Parents: TT × tt

gametes	T	T
t	Tt	Tt
t	Tt	Tt

You will notice that all the predicted F_1 offspring (shaded) have the genotype Tt. We can write this as:

F_1 genotype: 100% Tt

We can also see that offspring with Tt would all physically appear tall, as T stands for the tall allele which is dominant over the dwarf allele (t). So, we can write this as:

F_1 phenotype: 100% tall plants

Worked example 2.2

Cross between two of the hybrid F_1 plants from Worked example 2.1

F_1: Tt × Tt

gametes	T	t
T	TT	Tt
t	Tt	tt

F_2 genotype: 25% TT; 50% Tt; 25% tt

F_2 phenotype: 75% tall plants; 25% dwarf plants

We can also write the predicted outcomes as ratios. In this case, when two heterozygous tall pea plants were crossed, there was a genotypic ratio of 1TT : 2Tt : 1tt and a phenotypic ratio of 3 tall : 1 dwarf.

Choosing letters for genetic notation

When selecting letters to represent alleles, it is important to choose ones that are easily distinguishable in both uppercase and lowercase forms. Avoid letters that look similar in both cases, as this can cause confusion when reading genotypes.

Letters to avoid using include c, s, w, o, v and x.

Huntington's disease

Huntington's disease is inherited as a dominant trait on an autosome. It usually appears in an affected person as neurological symptoms that develop around 30–50 years of age resulting from an abnormal protein being made. The HH genotype is lethal because it disrupts early embryonic development, preventing survival to birth. This means that all living individuals affected by Huntington's disease will be heterozygous (Hh).

If we let H = abnormal Huntington's protein allele (Huntington's disease) and h = 'normal' protein allele (unaffected), then we can use these notations to help work out probable outcomes in the offspring.

Worked example 2.3

Cross between two individuals unaffected by Huntington's disease

Parents: hh × hh

gametes	h	h
h	hh	hh
h	hh	hh

F_1 genotype: 100% hh

F_1 phenotype: 100% unaffected

Worked example 2.4

Cross between an individual with Huntington's disease and an unaffected individual

Parents: Hh × hh (Someone with two H alleles will not survive to birth so the individual with Huntington's disease must be Hh).

gametes	H	h
h	Hh	hh
h	Hh	hh

F_1 genotype: 50% Hh; 50% hh

F_1 phenotype: 50% with Huntington's disease; 50% unaffected

Try this 2.4

Punnett squares

1. A black mouse mates with a brown mouse. There is a large number of offspring and all of them are black.
   a) Which trait is dominant?
   b) Draw a Punnett square and determine the genotypes of the offspring.
2. A recessive gene causes a condition called cystic fibrosis. If a homozygous recessive individual has a child with a heterozygous individual, what are their chances of producing offspring with cystic fibrosis?

Quick check 2.12

1. **Contrast** genotype and phenotype.
2. Brown eyes are dominant over blue eyes.
   a) **Select** letters for the brown eye and blue eye alleles.
   b) **Create** a Punnett square for a couple who are both heterozygous.
   c) **State** the genotypic and phenotypic ratios of the offspring.
   d) **State** the chances of this couple producing a blue-eyed child.

Test cross

A **test cross** can be used to discover whether an organism showing a dominant trait is homozygous dominant or heterozygous. It involves observing the offspring of the individual in question if it mates with a homozygous recessive individual (for example, aa). This means the individual of unknown genotype mates with an individual of known genotype. By looking at the offspring ratio, we can deduce the unknown genotype of the individual in question.

test cross a genetic cross between a homozygous recessive individual and an individual of unknown genotype showing the dominant trait to determine the unknown genotype

For example, in guinea pigs, coat colour may be black (B) or white (b). You can find out if a black guinea pig is homozygous black (BB) or heterozygous black (Bb) by doing a test cross between the black guinea pig and a white guinea pig (bb).

If the unknown black guinea pig is BB, the cross would look like this:

Parents: BB × bb

gametes	B	B
b	Bb	Bb
b	Bb	Bb

F_1 genotype: 100% Bb

F_1 phenotype: 100% black

This means if all the offspring are black, the unknown black guinea pig is probably BB (or homozygous dominant).

If the unknown black guinea pig is Bb, the cross would look like this:

Parents: Bb × bb

gametes	B	b
b	Bb	bb
b	Bb	bb

F_1 genotype: 50% Bb; 50% bb

F_1 phenotype: 50% black; 50% white

This means if roughly half the offspring are black and half are white, the unknown black guinea pig is probably Bb (or heterozygous). Note that these expectations are only true when based on large numbers of offspring because they are probabilities. However, if even one white offspring is born, the black guinea pig must be heterozygous in order to have passed on a recessive b gene.

Worked example 2.5

Determining the genotype of a dominant parent

A breeder is investigating the inheritance of a dominant trait in rabbits, controlled by a single gene. The allele for black fur (B) is dominant over the allele for white fur (b). The breeder crosses a black-furred rabbit with a white-furred rabbit and records the following offspring:

Fur colour	Number of offspring
Black	24
White	26

Based on this data, determine whether the black-furred parent is homozygous dominant (BB) or heterozygous (Bb).

Step 1: Predict offspring ratios

Working

If the parent is homozygous dominant (BB):

Cross: BB × bb

gametes	B	B
b	Bb	Bb
b	Bb	Bb

F1 phenotype: 100% black-furred

Explanation

Since the white-furred trait is recessive, we already know that the white-furred parent is homozygous (bb). If the black-furred parent is homozygous dominant (BB), all offspring would inherit one B allele, making them 100% black-furred (Bb).

Working

If the parent is heterozygous (Bb):

Cross: Bb × bb

gametes	B	b
b	Bb	bb
b	Bb	bb

F_1 phenotype: 50% black-furred; 50% white-furred

Explanation

If the black-furred parent is heterozygous (Bb), offspring would be in a 1:1 ratio of black-furred (Bb) to white-furred (bb).

Step 2. Compare with observed data

Black-furred parent genotype: heterozygous (Bb).

The breeder observed:
24 black-furred rabbits
26 white-furred rabbits
This is very close to a 1:1 ratio, which matches the expected outcome of a heterozygous (Bb) × homozygous recessive (bb) cross. Since a homozygous dominant (BB) parent would not produce white-furred offspring, the data strongly suggests that the black-furred parent is heterozygous (Bb).

Quick check 2.13

1. **Explain** the importance of a test cross.
2. Silky feathers in a bird species are caused by a gene with a trait recessive to normal feathers. If you had a normal-feathered bird, **describe** a way to determine whether it is heterozygous or homozygous. Use Punnett squares to help with your answer.

Try this 2.5

Positive or negative

One vital gene located on chromosome 1 in humans is the gene controlling for Rhesus (Rh) blood type. Rh positive blood is dominant to Rh negative blood, so we can assign the alleles D (Rh positive) and d (Rh negative). Therefore, an Rh positive person can be either DD or Dd with respect to this gene, but an Rh negative blood type can only be homozygous, dd.

Instructions

In this activity, we will use coloured counters and felt-tipped pens to model the passing of Rh alleles from heterozygous individuals to their offspring.

1. Copy the results table into your science journal.
2. Choose one coloured counter. Print D on one side and d on the other to represent the alleles of the chromosomes. This counter represents an individual who is Dd for Rh blood type, that is, able to produce gametes (eggs or sperm) of the type D and d in equal proportions.
3. Find a partner in the room with a different coloured counter.
4. Make an unbiased toss of your two counters to produce an offspring. The labels facing up on the counters will represent the alleles of the gametes produced by the parents. Record the genotype of the offspring in the Family 1 row of the following table. Repeat this step three more times to produce a total of four children.
5. Generate a total of four families (by pairing up with three different students) with each having produced four children.
6. Record the number of each type of genotype found in the children of each family in the table following table. Pool your total results with those of the class.

Results

	Genotype of children		
	DD	**Dd**	**dd**
Family 1			
Family 2			
Family 3			
Family 4			
Total numbers across the class			

Analysing

1. Using the class data, calculate the probability of obtaining the offspring genotypes:
   a) DD
   b) Dd
   c) dd
2. What are the proportions of the different genotypes in your group of 16 children compared to those generated across the whole class? Discuss whether this is expected.
3. A woman with Rh positive blood insisted that she was the daughter of a rich, elderly couple who both had Rh negative blood. Use a Punnett square to show your working out to explain whether this woman's statement could be valid.

Did you know? 2.6

Multifactorial inheritance

Most traits in organisms are shaped through interactions between multiple genes and a range of environmental factors. This concept, known as multifactorial inheritance, recognises that genes don't work in isolation. Instead, different genes may each contribute small effects that, combined, influence traits like height, skin colour, and even the risk of developing certain diseases. These genetic contributions are then influenced by various environmental factors – such as diet, lifestyle, climate and exposure to toxins – which can further modify trait expression.

For example, height is determined by numerous genes that each play a role in growth, bone structure, and metabolism. However, factors like nutrition during childhood and adolescent development also significantly impact height, showing that genes set a potential range, while the environment helps determine where an individual falls within that range.

Sex linkage

As we have seen, in humans the sex chromosomes are the 23rd pair. Human females are homogametic for the X chromosome while males are heterogametic with one X and one Y chromosome.

The X chromosome also contains many genes not related to sex determination (in contrast, the Y chromosome has very few). Characteristics that are not related to determining the sex of the individual but are coded for by X chromosome genes are termed X-linked. When we predict X-linked traits using a Punnett square, we must use the sex chromosomes and superscripted letters above the X chromosomes to represent the trait.

carrier
an individual with a recessive allele for a disease but who does not have the disease due to being heterozygous

For example, Duchenne muscular dystrophy (DMD), a disorder where the muscles progressively degenerate, is inherited as an X-linked recessive trait. The genotype for an unaffected female would be X^DX^D, a **carrier** female (who has a recessive allele for the disease but is heterozygous so does not have the disease) would be X^DX^d, and a female with DMD would be X^dX^d. On the other hand, an unaffected male would be X^DY and a male with DMD would be X^dY. As there is no gene for this condition found on the Y chromosome, we do not write ^{D} or ^{d} next to the Y. Because males have only a single copy of the X chromosome, they are more likely to be affected by sex-linked disorders. A single allele of a mutated gene on an X chromosome will cause the disease in a male but not in a female (who would need two DMD alleles to be affected).

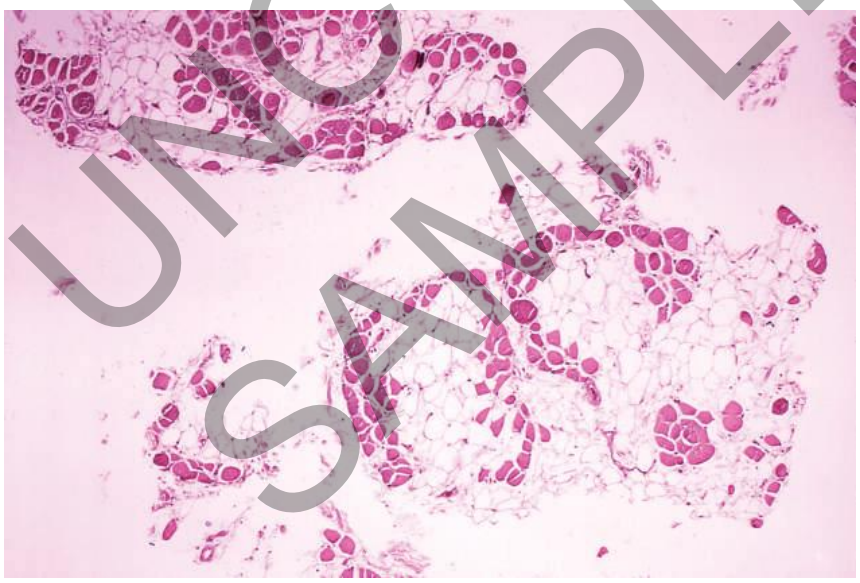


Figure 2.39 In Duchenne muscular dystrophy, muscle fibres (red) are replaced by fat cells (white).

Worked example 2.6

Cross between a heterozygous normal-vision female (carrier) and a normal-vision male

Colour blindness is inherited as an X-linked recessive trait. The genotype of a heterozygous normal-vision (carrier) female is X^BX^b and of a colour-blind male is X^bY.

Parents: $X^BX^b \times X^BY$

gametes	X^B	X^b
X^B	X^BX^B	X^BX^b
Y	X^BY	X^bY

F_1 genotype: 25% X^BX^B; 25% X^BY; 25% X^BX^b; 25% X^bY

F_1 phenotype: 25% normal-vision female; 25% normal-vision male; 25% carrier normal-vision female; 25% colour-blind male

Try this 2.6

Sex-linked inheritance

Now it is your turn to practise. Remember that the gametes need to include the sex chromosomes, and the genotype and phenotype both need to include the sex chromosomes as well.

1. Using a Punnett square, cross a normal-vision female (homozygote) with a colour-blind male.
2. Using a Punnett square, cross a colour-blind female with a normal-vision male.
3. Explain why more males than females have colour blindness.

Codominance

When there are two equally dominant alleles for a trait, they will both be expressed in the phenotype of a heterozygote. This is **codominance**. Look at the picture of the chickens in Figure 2.40. The colours black and white are equally dominant (their alleles are given two different capital letters), so when you cross a pure black chicken with a pure white chicken, you get a heterozygote. In the heterozygote, both colours are expressed equally, so you get a speckled chicken with black feathers and white feathers!

VIDEO
An example of codominance

codominance
both alleles are expressed equally in the phenotype

Genotype	WW	BB	BW
Phenotype	White	Black	Speckled

Figure 2.40 White and black are equally dominant alleles and are said to be codominant. The evidence of this is their heterozygote offspring that has a mix of black and white feathers.

VIDEO
Blood types

Blood types

In humans, our blood type is a codominant trait. Our red blood cells have proteins on the surface called antigens, and these determine whether our blood is A, B, AB or O. The I gene is often used in genetic notation for the ABO system. The I stands for the isoagglutinogen gene, which determines the presence of blood group antigens.

Each person has two copies of the isoagglutinogen gene, one from their mother and one from their father. This gene is found on chromosome 9, and there are three possible alleles:

- I^A = production of antigen A (type A blood) is dominant.
- I^B = production of antigen B (type B blood) is dominant.
- i = production of neither antigen (type O blood) is recessive.

As we have two copies of chromosome 9, there can be six different allele combinations:

- Type A blood can have genotypes $I^A I^A$ or I^A i.
- Type B blood can have genotypes $I^B I^B$ or I^B i.
- Type AB blood can only have the genotype $I^A I^B$.
- Type O blood can only have the genotype ii.

I^A an I^B are both dominant over i, but are codominant to each other.

Worked example 2.7

Cross between heterozygous blood type A and heterozygous blood type B

Parents: $I^Ai \times I^Bi$

gametes	I^A	i
I^B	I^AI^B	I^Bi
i	I^Ai	ii

F_1 genotype: 25% $I^A I^B$; 25% I^B i; 25% I^A i; 25% ii

F_1 phenotype: 25% AB blood type; 25% B blood type; 25% A blood type; 25% O blood type

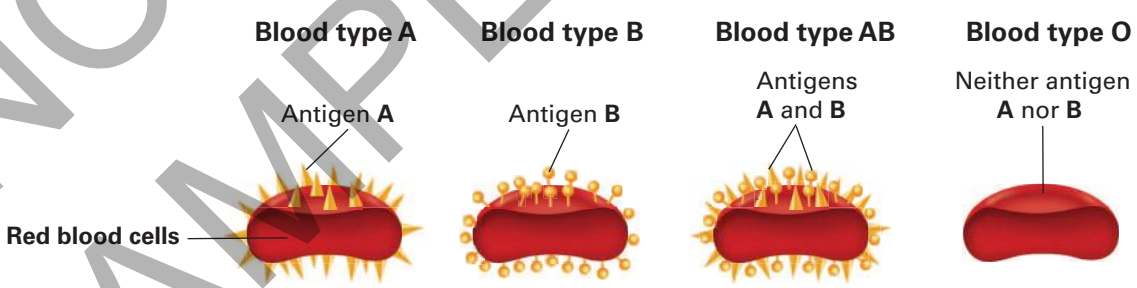


Figure 2.41 Illustration of the different antigens on the surface of red blood cells

Explore! 2.6

Incomplete dominance

incomplete dominance a form of inheritance in which both alleles are partially expressed, producing a third intermediate phenotype

Incomplete dominance occurs when neither of the alleles is dominant over the other, resulting in a third phenotype which is an intermediate form of both alleles. Wavy hair is an example of incomplete dominance. A person with two curly hair alleles (CC) will have curly hair. A person with two straight hair alleles (SS) will have straight hair. However, a person with one curly and one straight allele (CS) will have wavy hair. To distinguish between different versions of the

continued …

same gene, alleles are often written with the same letter but in superscript. For example, H^CH^C could represent two curly hair alleles, and H^SH^S could represent two straight hair alleles. In this way, H^CH^S would indicate the heterozygous condition, resulting in wavy hair as an intermediate phenotype.

1. The flower colour of snapdragons is an example of incomplete dominance. Draw an example of a cross for a white flower (C^WC^W) and a red flower (C^RC^R).
2. Eggplant colour is another example of incomplete dominance. Explain how the three colours of eggplant shown in Figure 2.43 are possible.

Figure 2.42 Wavy hair is an example of incomplete dominance.

Figure 2.43 Deep purple (left), violet (middle) and light green (right) eggplants

Quick check 2.14

1. **Explain** why males are more likely to be affected by recessive X-linked disorders than are females.
2. **Describe** codominance.

Pedigrees

One way of finding out how a trait is inherited is to follow the inheritance pattern over two or more generations. This technique is called pedigree analysis. To study patterns of inheritance in humans, geneticists investigate the frequency and occurrence of a particular gene over many generations to determine whether the traits are dominant or recessive. The chart formed is called a **pedigree** and it is like a family tree.

WORKSHEET
Pedigrees

pedigree
a chart that shows relationships between family members and indicates which individuals have certain genetic traits

Each generation is numbered in Roman numerals and forms a row on the pedigree. Individuals are numbered using Arabic numerals (1, 2, 3 etc.) from left to right on each row. The symbols used are:

- Unaffected female
- Unaffected male
- Female with the trait being investigated
- Male with the trait being investigated
- Carrier of trait or
- Mating is represented by a horizontal line
- A vertical line connects offspring to parents

Reading a pedigree

There are some points to keep in mind when reading a pedigree and determining inheritance:

- A trait that is common in a population is not necessarily dominant.
- When asked to determine a genotype in a pedigree, you will need to establish the mode of inheritance. You can use Punnett squares to help you.

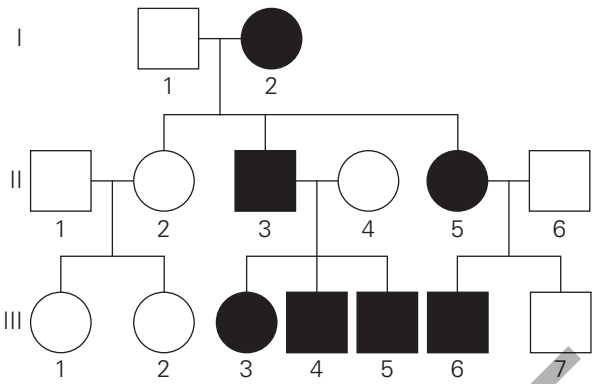


Figure 2.44 An example of a pedigree showing three generations of a family. Individuals affected by a particular trait are shown in black. Note how male I-1 and female I-2 had three children, and their eldest is an unaffected female (II-2).

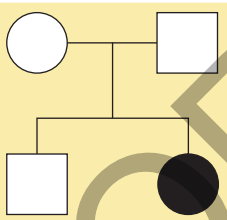
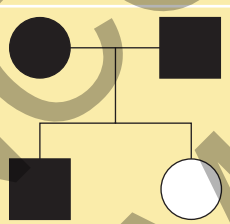
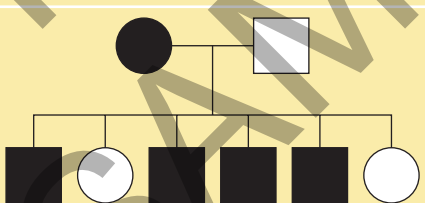
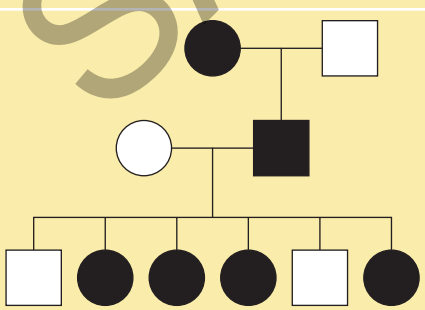
Mode of inheritance	Pedigree chart	What to look for	Examples
Autosomal recessive		Two unaffected parents can have an affected child. Males and females are equally affected. The trait may disappear from a branch of pedigree and then reappear in later generations. Two affected recessive parents must have all affected recessive children.	Albinism, cystic fibrosis, sickle cell anaemia and thalassaemia
Autosomal dominant		Two affected parents can have an unaffected child. Males and females can be equally affected, yet all of the affected individuals must have at least one parent affected.	Huntington's disease and haemochromatosis
X-linked recessive		Affected mothers produce affected sons. Affected fathers *cannot* pass the trait to their sons.	Colour blindness, haemophilia and Duchenne muscular dystrophy
X-linked dominant		Affected fathers produce daughters that are all affected. Affected fathers *cannot* pass the trait to their sons, but affected homozygous females will pass the trait on to all of their daughters and sons.	X-linked hypophosphatemic rickets

Table 2.5 Different modes of inheritance and how to identify them in a pedigree chart

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Did you know? 2.7

Famous people have genetic disorders too!

Megan Fox has a condition called brachydactyly. This is an autosomal dominant condition that causes shortened fingers or toes. Jessie J has Wolff–Parkinson–White Syndrome, an autosomal dominant condition that causes a very fast heart rate.

Figure 2.45 Megan Fox and Jessie J both have genetic disorders.

Quick check 2.15

1. Study the pedigree chart in Figure 2.46. It illustrates the inheritance of achondroplasia (dwarfism) in humans.

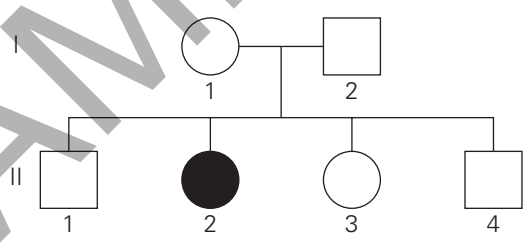


Figure 2.46 Pedigree chart

a) **Identify** a piece of evidence that suggests that this condition is recessive and not dominant.
b) **Identify** the genotype of individuals I-1, I-2, II-1 and II-2.
c) Suppose individual II-2 has children with a man who does not have achondroplasia but is a carrier for the condition. **Determine** the chance of having a child with achondroplasia and the chance of having a child without achondroplasia. Use a Punnett square to show how you arrived at your answer.

Go online to access the interactive section review and more!

Section 2.3 review

Online quiz

Section questions

Teachers can assign tasks and track results

Section 2.3 questions

Remembering

1. **State** how many copies of each gene we have and where they come from.
2. **Recall** the name of the person known for being the father of genetics.
3. **Name** an example of codominance and of incomplete dominance.

Understanding

4. **Describe** what pedigrees are useful for.
5. **Explain** how sex linkage is different from other types of inheritance.
6. **Explain** why, for a particular characteristic, two homozygous recessive individuals can only produce homozygous recessive children.
7. **Explain** what is meant by the term 'carrier'.
8. **Explain** why a test cross is useful.

Applying

9. Dimples are dominant to no dimples.
   a) **Select** appropriate letters for these alleles.
   b) **Identify** the genotype and phenotype of a person who is:
      i) homozygous recessive for dimples
      ii) heterozygous for dimples
      iii) homozygous dominant for dimples.

Analysing

10. The pedigree chart in Figure 2.47 shows the inheritance pattern of a disease that is caused by a single gene.
    a) **Identify** the sex of the person labelled II-5.
    b) **State** whether the disease is dominant or recessive, giving reasons for your answer.
    c) **State** the genotype of individual II-5, giving reasons for your response.

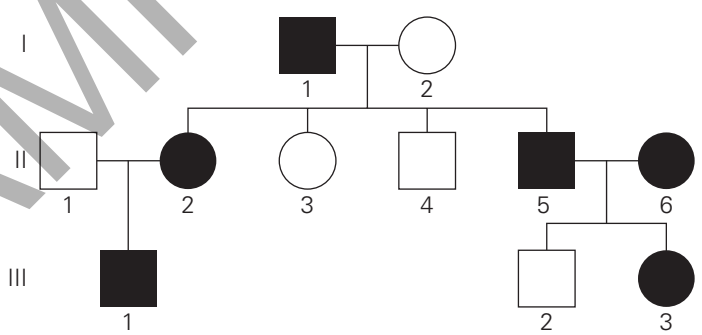


Figure 2.47 Pedigree chart

11. **Contrast** codominance and incomplete dominance.
12. The pedigree chart in Figure 2.48 shows the inheritance patterns for haemophilia. Haemophilia is an X-linked recessive disorder.
    Use evidence from the pedigree to **justify** that haemophilia is recessive.

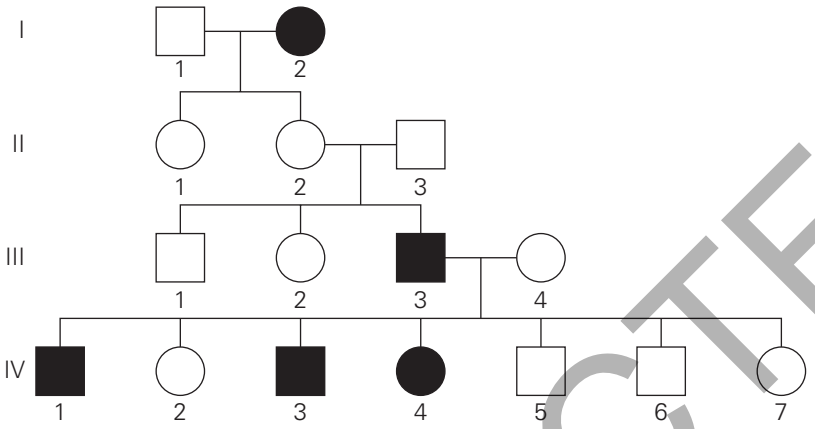
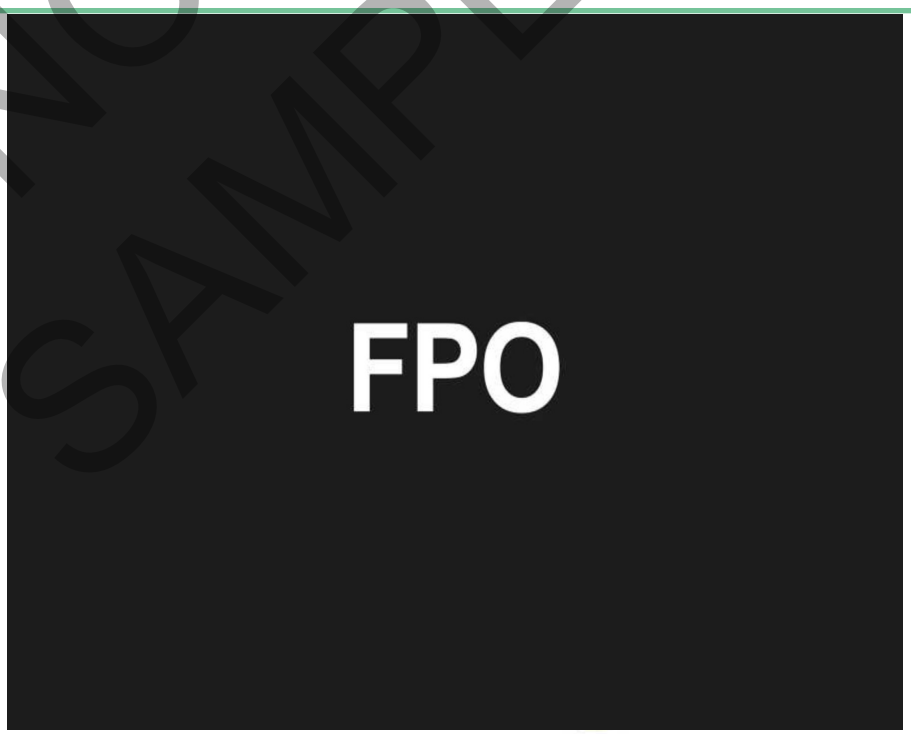


Figure 2.48 A haemophilia pedigree

Evaluating

13. In fruit flies, eye colour is sex linked. The eye colour allele is located on the X chromosome. The red eye allele is dominant over the white eye allele.
    a) **Select** appropriate letters for the alleles.
    b) Using a Punnett diagram, **determine** the offspring a male heterozygous fly that mates with a female homozygous recessive fly will produce.
    c) **Decide** the genotypic and phenotypic ratios of the offspring.
14. In mice, black hair is dominant to brown hair. Mice with black hair can be either homozygous or heterozygous.
    Propose how you would find out if a mouse with black hair was homozygous or heterozygous. Support your response with a Punnett square.
15. A brown-eyed male and a blue-eyed female produced all blue-eyed children. **Discuss** whether that means blue eyes is the dominant trait.



2.4 Changes in DNA

WORKSHEET Changes in DNA

Learning goals

At the end of this section, I will be able to:

1. Distinguish germline and somatic mutations.
2. Recall types of point and chromosome mutations.
3. Explore the applications of DNA manipulation.

point mutation a mutation in which a single nucleotide is changed

chromosome mutation a mutation involving large segments of DNA

spontaneous mutation a naturally occurring mutation

induced mutation a mutation produced by environmental factors

mutagenic causing mutations in DNA

germline mutation a mutation of DNA in gametes which can be inherited

somatic mutation a mutation that occurs in somatic (body) cells which cannot be inherited

Sometimes, the process of DNA replication makes errors, which cause changes to the genetic code. Often these errors are repaired, but when they are not, they become permanent changes in the genome of the cell. This is known as a mutation. Mutations may be small, such as a change in a single nucleotide (**point mutation**), or large, involving entire segments of DNA within a chromosome (**chromosome mutation**). Mutation is the only way that new alleles can be created within a population.

A mutation can be described as **spontaneous** (naturally occurring) or **induced**. Induced mutations are caused by exposure to **mutagenic** agents in the environment like radiation (X-rays, ultraviolet rays or nuclear radiation) or chemicals. Mutations that occur within the gametes are called **germline mutations**. These can be inherited and will influence the next generation. Mutations that occur within somatic cells will only affect the individual and will not be passed down to future generations. This form of mutation is known as a **somatic mutation**.

Explore! 2.7

Mutagens

Many factors in the environment are known as 'mutagens' – that is, they cause mutations.

1. Research some environmental mutagens.
2. From the list created in Question **1**, divide your mutagens into categories according to their source, such as chemical mutagens and radiation.

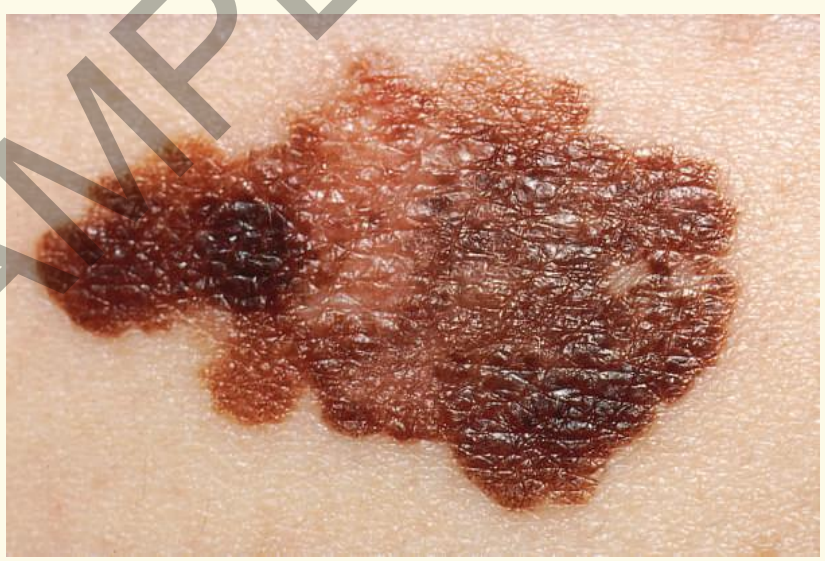


Figure 2.49 Melanoma is linked to UV light exposure as well as gene changes. A mole may be a sign of skin cancer if it has irregular borders, an asymmetrical shape or if it changes in colour, shape, size or height.

Did you know? 2.8

The Black Death left a genetic mark

Many individuals with European ancestry carry genetic mutations that protected their ancestors from the bubonic plague, a disease caused by *Yersinia pestis*, a species of bacterium spread by fleas. When plague struck Europe in 1348 and again in 1665, millions of people died. However, some people had mutations that made them more likely to survive.

By examining the DNA of people who died before, during and after the Black Death, researchers have found that carrying two copies of a mutated ERAP2 gene improved a person's survival by 40%. The allele codes for a protein that is involved in the immune response, and the mutation produced a heightened response. This meant that the survivors of bubonic plague passed on this beneficial trait to their offspring. However, this mutation is also problematic. Individuals who carry the mutated ERAP2 gene are at an increased risk of autoimmune diseases.

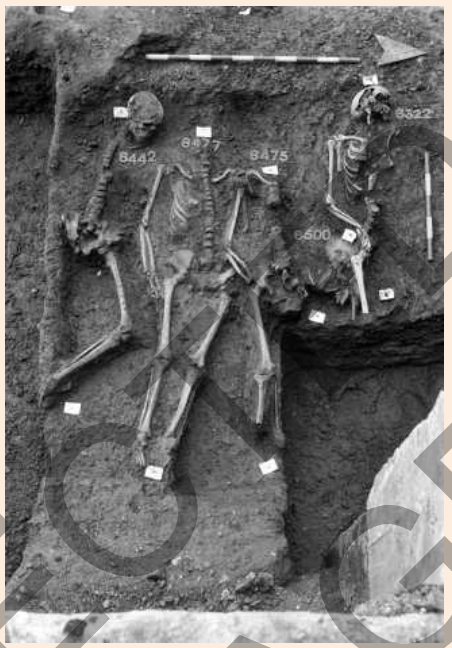


Figure 2.50 A small mass burial pit of plague victims excavated in East Smithfield, London. The significant number of deaths during plague outbreaks resulted in many similar mass burials across Europe.

Beneficial, harmful, and neutral mutations

Mutations have three possible consequences:

- **No effect – neutral**. Neutral mutations are non-lethal and make no difference to the organism's ability to survive and reproduce in its environment.
- **Negative effect – harmful**. Harmful mutations produce proteins that malfunction or cause proteins not to be produced at all. They can cause genetic disorders or cancer and result in organisms that are less likely to survive, reducing the likelihood that the genetic material will be passed on to the next generation.
- **Positive effect – beneficial**. Beneficial or advantageous mutations give rise to new versions of proteins that increase the chance that the organism will survive and reproduce to pass its genetic material to the next generation. Beneficial mutations increase genetic variation in a population and are essential for evolution to occur.

Quick check 2.16

1. If a mutation is to be passed on to the next generation, **recall** where it must occur.
2. **Summarise** the possible effects of mutations that occur in somatic cells.

Gene mutations

Gene mutations refer to changes in the DNA sequence that makes up a gene. This involves changes to any of the four nitrogenous bases in the nucleotides that make up the genes.

gene mutation a permanent alteration in the DNA sequence that makes up a gene

There are several types of gene mutations, including point mutations, insertions, deletions and inversions.

substitution where one nucleotide is swapped for another

insertion where one or more extra nucleotides are inserted into the DNA

deletion where a nucleotide is deleted from the sequence

inversion where two nucleotides reverse their order

Changing the nucleotides could change the message carried by the gene, which may change the order of amino acids making up the protein.

- A point mutation is the most common type of gene mutation, and involves the **substitution** of one nucleotide for another (for example, ATG becomes ACG). This mutation only changes the DNA code for a single amino acid. If the new sequence codes for the same amino acid, it is called a silent mutation. An example of a disease caused by a substitution mutation is sickle cell anaemia.
- An **insertion** mutation occurs when an extra nucleotide (or more than one) is inserted into the DNA sequence (for example, ATG becomes ATCG). This type of mutation changes the DNA code for all amino acids that follow and is called a frameshift mutation. An example of a disease caused by an insertion mutation is fragile X syndrome.
- A **deletion** mutation occurs when a nucleotide is deleted from the sequence (for example, ATG becomes AG). This type of mutation changes the DNA code for all amino acids that follow and is also a frameshift mutation. An example of a disease caused by a deletion mutation is Duchenne muscular dystrophy.
- An **inversion** mutation occurs when two nucleotides reverse their order in the DNA (for example, ATG becomes AGT). This mutation only changes the DNA code for a single amino acid, and it could code for the same amino acid (silent mutation). An example of a disease caused by an inversion mutation is haemophilia.

Try this 2.7

Remembering mutations

Find a way to remember the different types of mutations using normal words.

For example:

- Normal code – WING
- Deletion – WIG
- Substitution – WIND
- Insertion – WRING

Chromosomal changes

Chromosome mutations

If a mutation occurs in a gene on one of the pair of homologous chromosomes, there will still be a normal copy of the gene on the other chromosome. The same applies with chromosome mutations. As chromosomes occur as homologous pairs, if one chromosome is abnormal, the other is still likely to be normal. The different types of chromosome mutations are illustrated in Figure 2.51.

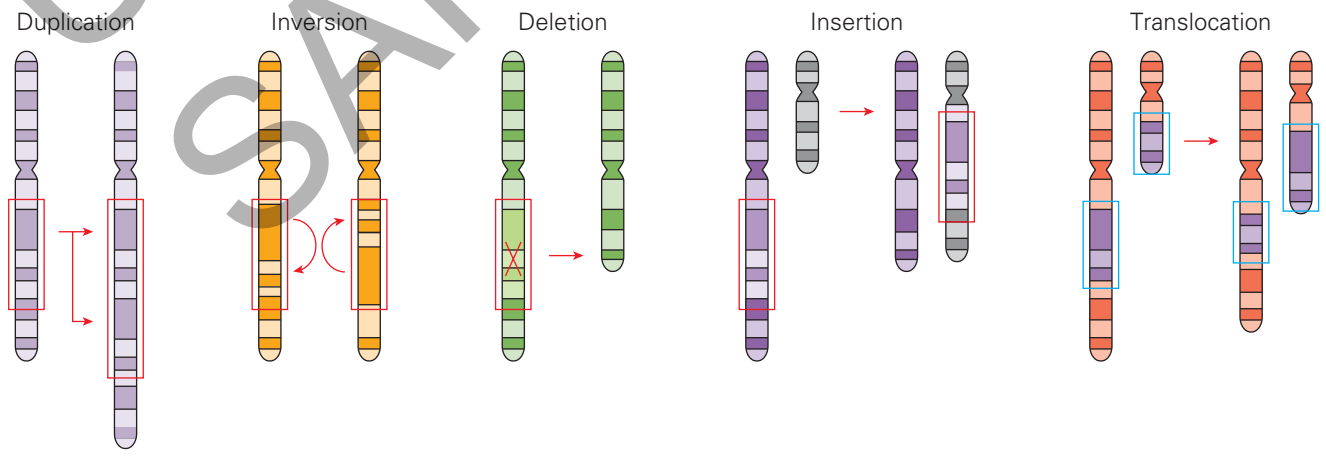


Figure 2.51 The five different types of chromosome mutation. Can you identify what is changing in each type?

Chromosome number abnormalities

Certain individuals may have one more or one fewer chromosome because of **non-disjunction** during meiosis. Non-disjunction means that the chromosomes failed to separate correctly when making gametes, so the gametes end up with an abnormal number of chromosomes (**aneuploidy**).

Trisomy occurs when an organism has a third copy of a chromosome that should only be present in two copies. Trisomy is therefore an example of aneuploidy. The most common trisomy among embryos that survive to birth is Down syndrome, or trisomy 21. People with this inherited disorder have distinct facial characteristics and experience developmental delays.

non-disjunction the failure of homologous chromosomes or sister chromatids to separate correctly in meiosis

aneuploidy the presence of an abnormal number of chromosomes in a cell

trisomy when an organism has a third copy of a chromosome

Did you know? 2.9

Sex chromosome abnormalities

Sex chromosome abnormalities occur when a person is missing a whole sex chromosome or has an extra sex chromosome due to non-disjunction. Problems can also occur when a person is missing part of a sex chromosome. Some disorders of sex chromosome are Klinefelter syndrome, XX male, XYY male, Turner syndrome, XXX female and XY female. Some of these disorders are surprisingly common. For example, many men with Klinefelter syndrome (who have two X chromosomes and one Y) have no signs or symptoms, and it is estimated that only one in six are ever diagnosed.

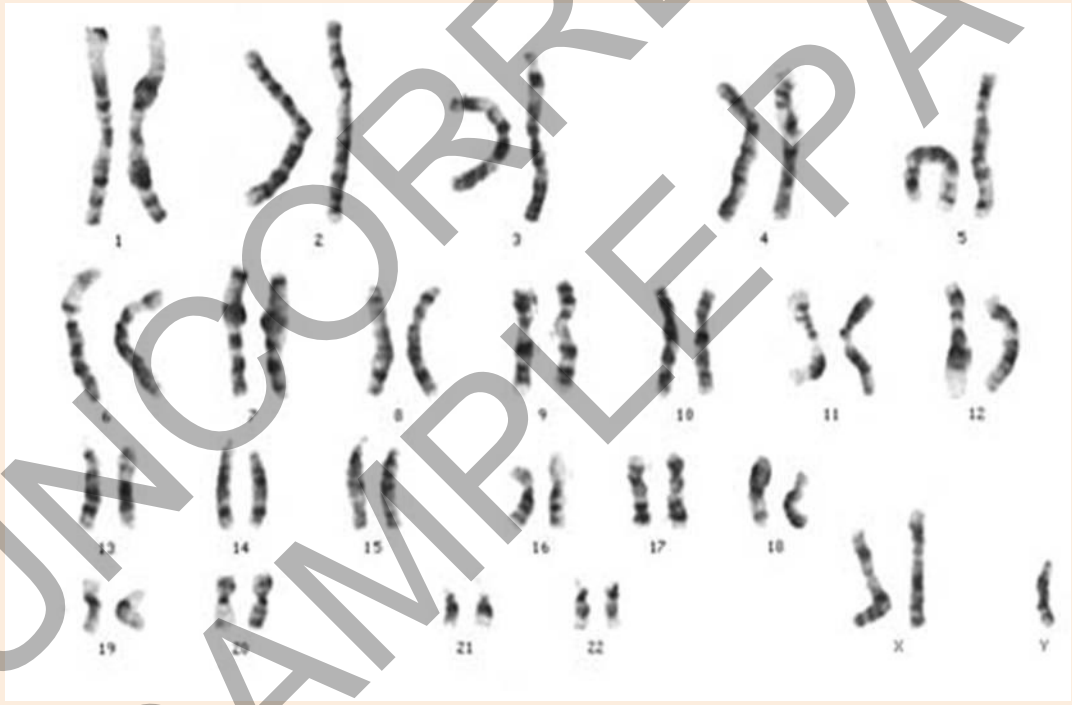


Figure 2.52 A karyotype of a male with Klinefelter syndrome

Quick check 2.17

1. **Describe** a substitution mutation.
2. **Recall** what the term 'aneuploidy' means.

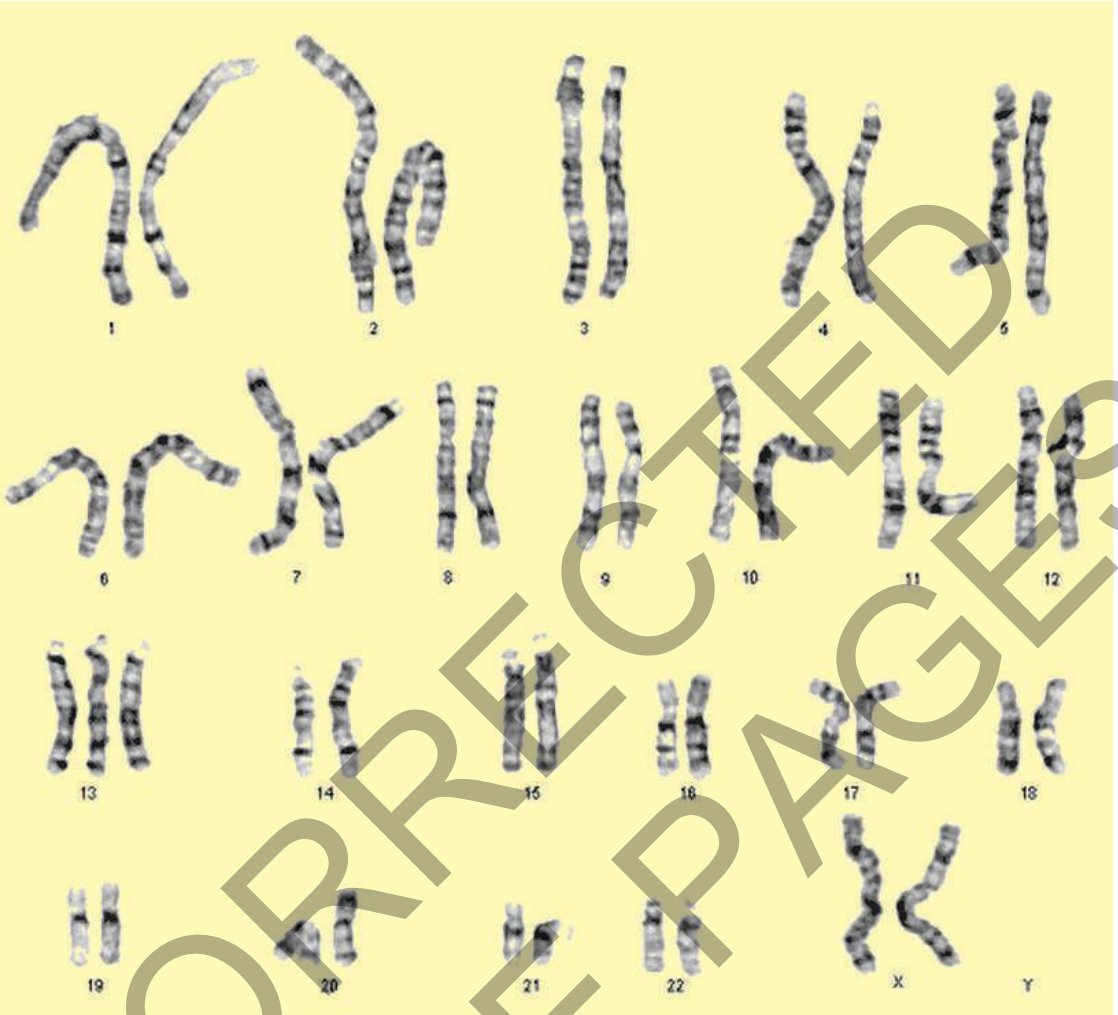


Figure 2.53 There are other types of trisomy apart from trisomy 21. Trisomy 13 and 18 result in more severe conditions. Which trisomy is depicted in this karyotype?

Did you know? 2.10

Madeline Stuart

Madeline Stuart is the first professional model with Down syndrome, and she is Australian! Although the fashion industry has a long way to go in terms of diversity, Madeline is one of the women who are changing the game. As well as being a model, she has also represented Queensland at the Special Olympics in cricket and basketball.

Figure 2.54 Madeline Stuart models at the New York Fashion Week.

DNA testing

A person's DNA can provide information relating to their ancestry or parentage, confirm if they have a hereditary disease, and inform whether they are a carrier for genetic disorders that may affect their children.

genetic screening genetic tests carried out across the population to identify people at risk of genetic disorders

Genetic screening is a type of genetic testing, but instead of identifying a genetic condition, it is used to evaluate an individual's risk of developing a certain disease. These tests are routinely done for anyone in the population, even if they have not shown a family history of the disease. Genetic screening is usually done during pregnancy and at birth. In Australia, about 99% of babies are screened every year. Of the babies screened, around one in every thousand has a condition that would otherwise have gone undetected. Genetic screening provides an opportunity for preventive measures and early treatment.

Figure 2.55 In Western Australia, the newborn bloodspot screening test screens for conditions such as cystic fibrosis, phenylketonuria, hypothyroidism, galactosaemia and congenital adrenal hyperplasia.

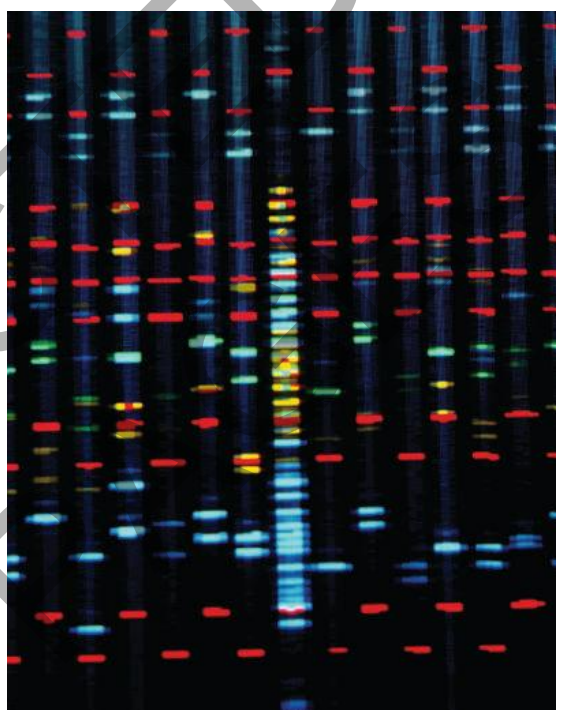


Figure 2.56 A DNA sequence or 'genetic fingerprint' on a computer monitor. The development of fast computers made the analysis of DNA sequences possible.

Making thinking visible 2.2

Think, feel, care: Genetic screening

In your class, divide into teams and engage in a discussion about perspectives within a scenario about genetic testing. You may wish to consider the following scenario, although your teacher may provide you with an alternative.

A woman who is 12 weeks pregnant attends a clinic and expresses her concern about her husband's family history of Huntington's disease (HD). Her sister-in-law tested positive last year, although she is not yet showing symptoms of the disease, but the woman's husband refuses to be tested. Due to this, the woman is asking for her unborn baby to be screened to determine whether it carries the HD gene. If the test comes back positive, which means that her husband carries the HD gene, the woman will discuss terminating the pregnancy with her husband.

Step inside each person's point of view in this scenario. As you think about what you know, consider what each person might understand, respond to and value in the issues raised. Don't forget to consider the legal and ethical issues.

The *Think, feel, care* thinking routine was developed by Project Zero, a research centre at the Harvard Graduate School of Education.

Science inquiry 2.2

Precision medicine

Pharmacogenomic tests look for genetic variants that are associated with an individual's response to specific medicines, by testing genes that affect drug metabolism. Its goal is to help doctors to select the drugs and doses that are best suited to each person.

Figure 2.57 Research has shown that people with red hair tolerate pain differently to people with other hair colours. Due to a mutation on the MC1R gene, they require more anaesthesia but need lower doses of opioids as they are more responsive to those painkillers.

Quick check 2.18

1. **Explain** why an individual might choose to be screened for genetic diseases.
2. **Identify** some ethical issues with genetic testing.

VIDEO
CRISPR

Manipulating DNA

Genetic engineering technology allows scientists to manipulate genetic material and transfer genes between different organisms. One of the most exciting developments of the last decade is CRISPR, a technology that can edit genetic material quickly and cheaply. Developed by Jennifer Doudna and Emmanuelle Charpentier, the CRISPR tool lets scientists change or delete DNA bases and insert entirely new genes (Figure 2.58). The gene editing technology has almost endless potential, including editing crops so they're more nutritious or able to survive in different conditions, curing genetic diseases, producing powerful new drugs or even resurrecting extinct species. In 2022, a New Zealand woman became the first to receive CRISPR treatment to treat heart disease by permanently lowering her cholesterol.

genetic engineering deliberately modifying genetic material to manipulate the characteristics of an organism

genetically modified organism an organism that has had its genome altered using genetic engineering techniques

Genetically modified organisms

A **genetically modified organism** (GMO) is an organism whose genome has been altered by humans. Commonly, they are crop species that have been modified to improve growth rates, yield, quality and nutritional value. Genetic modification can decrease pest and disease susceptibility, reducing the need for pesticides.

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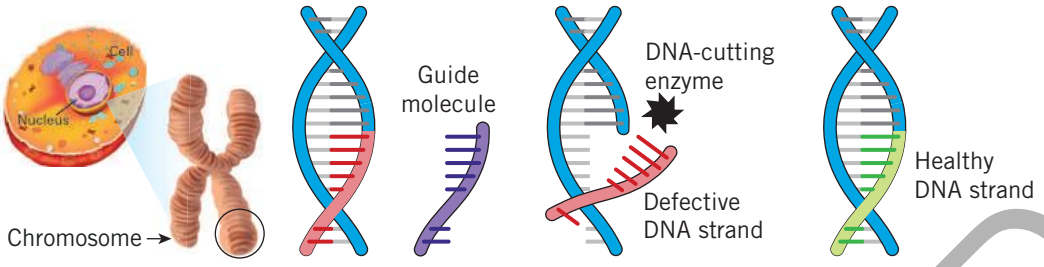

A cell is transfected with an enzyme complex containing: a guide molecule, DNA-cutting enzyme and a copy of healthy DNA.

The guide molecule finds the target DNA strand.

The enzyme cuts out the target DNA strand.

The healthy DNA copy replaces the defective DNA.

Figure 2.58 CRISPR gene editing technology allows DNA to be manipulated in a highly precise way.

Agricultural practices have changed to include the widespread use of genetically engineered crops. Crop plants are modified to have a greater tolerance to environmental stressors such as drought or heavy metals, allowing plants to grow in new areas. For example, some plants have been genetically engineered to grow with less water so that they are more resistant to climate change. Until recently, only three GM crops were approved to be grown in every mainland Australian state: canola (oilseed rape), cotton and safflower. After a ban was lifted, a wider range of GM crops can now be grown in every mainland state.

Try this 2.8

GM food

A great debate continues to rage within society regarding the health risks associated with eating GM food. Research two advantages and two disadvantages of the use of GM food and be prepared to debate with your classmates.

Figure 2.59 Cassava root has been genetically modified to have increased levels of beta carotene (right). This decreases post-harvest deterioration and also improves the nutritional value of the crop.

Did you know? 2.11

De-extinction

'De-extincting' is the process of bringing an extinct species back to life through genetic engineering by using fragments of DNA from preserved specimens. De-extincting species is a controversial idea, and many scientists argue that funding would be better spent conserving existing species and their habitats. A start-up biotechnology company called Colossal Biosciences quickly raised millions of dollars after announcing plans to de-extinct the woolly mammoth, the thylacine and the dodo. In 2025, the company claimed to have revived the dire wolf, last seen over 10 000 years ago. A white wolf featured on the cover of *Time* magazine, named as one of three 'dire wolf' puppies – Romulus, Remus and Khaleesi.

However, scientists soon clarified that these were genetically edited grey wolves. With dire wolf DNA too fragmented to clone, researchers had inserted selected gene sequences into grey wolf embryos to mimic some traits.

Figure 2.60 Colossus Biosciences has produced three genetically engineered dire wolf puppies by using modern grey wolves and ancient DNA retrieved from the remains of extinct dire wolf ancestors. The DNA was inserted using CRISPR technology.

Figure 2.61 Golden rice and normal rice

Figure 2.62 Mosquitoes have been genetically modified to slow the growth of malaria-causing parasites in their gut, preventing transmission of the disease to humans.

Genetic engineering is also helping to alleviate global malnutrition and disease. A type of rice called golden rice has been engineered to prevent blindness in children caused by vitamin A deficiency (Figure 2.61). Another type of GM rice has been developed that has four times more iron than normal rice.

transgenic organism an organism that possesses a foreign gene or segment of foreign DNA in its genome because of human experimentation

Transgenic organisms (TGOs) are the result of a type of genetic modification where a 'foreign' gene, or a segment of 'foreign' DNA is inserted into an organism's genome such as in the mosquitoes in Figure 2.62. A foreign gene is one that is taken from another species. All transgenic organisms are GMOs, but not all GMOs are transgenic.

By inserting human genes into mice, human diseases can be mimicked, particularly as the genome and physiology of mice are so similar to humans. Transgenic mice are being used to study conditions such as obesity, heart disease, diabetes and Alzheimer's disease. Transgenic pigs are

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being investigated as a source of organs for transplants, which could address severe donor organ shortages. CRISPR technology has greatly reduced the complexity of the creation of transgenic animals, making the process faster and cheaper.

Explore! 2.8

Genetically modified tomatoes

Many foods come from genetically modified organisms, and we call them GM foods. The Flavr Savr tomato was the first commercially grown GM food that was granted a licence for human consumption. Released in 1994, the tomato had an improved shelf life and was developed for long-distance shipping. Recently, a GM tomato has been developed that contains higher levels of anthocyanins, a pigment that occurs naturally in purple or blue fruits and vegetables such as blueberries or red cabbage. By transferring a snapdragon gene into a tomato's genome, they have produced a purple tomato that has the same levels of anthocyanins as blueberries.

1. Research why the team chose to genetically modify a tomato.
2. The high levels of anthocyanins in the purple tomatoes are beneficial for several reasons. Research the effect of anthocyanins on:
   a) shelf life
   b) plant reproduction and survival
   c) human health.

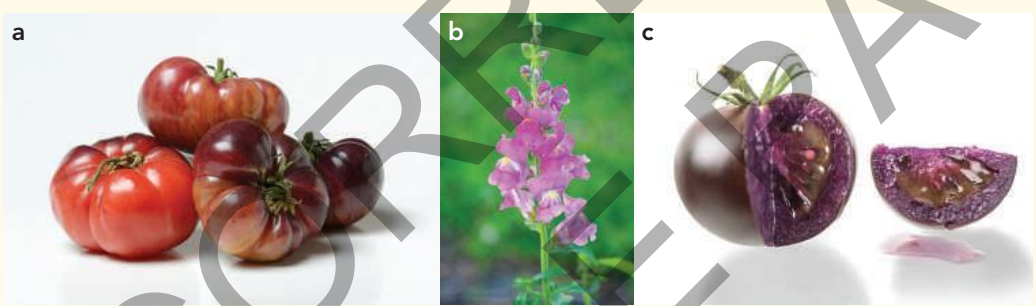


Figure 2.63 The process by which the John Innes Centre and the Sainsbury Laboratory created a GM purple tomato, which was approved for growing and breeding in the US in 2022. **a)** Existing purple-skinned tomatoes do not contain useful levels of anthocyanins. **b)** Genes from purple snapdragon flowers were added to the tomato. The snapdragon genes act like 'on switches', causing the purple pigment to be expressed throughout the tomato, instead of just in the skin. **c)** The resulting purple tomato is high in anthocyanins, which have a range of benefits for human health.

Human genes have been engineered into other mammals to supply the protein products of those genes, usually for medical purposes. For example, transgenic goats have been genetically engineered to produce milk that contains human antithrombin III. This is a protein that is used to prevent clots in patients with hereditary antithrombin deficiency when they are in surgery or giving birth. A herd of approximately 80 goats could supply enough human antithrombin III for all of Europe.

Figure 2.64 Transgenic C5 plums contain a gene that makes them resistant to plum pox virus.

Figure 2.65 A CSIRO study found that genetically modified sheep grow bigger and faster and can produce more milk and wool. However, they required more care than non-GM sheep.

Making thinking visible 2.3

I used to think ... now I think ... : Xenotransplantation

Xenotransplantation is the transplantation of living cells, tissues or organs from one species to another. Transgenic pigs are promising xenotransplantation donor organisms as they share many anatomical and physiological characteristics with humans. In 2022, the first pig-to-human heart transplant took place. The operation was successful and, due to genetic modification, the heart was not rejected by the patient's immune system. Unfortunately, he died 60 days later; scientists are still investigating why this happened. Scientists have also been successful in transplanting genetically modified pig kidneys into a brain-dead patient, with them functioning normally throughout the study period.

Think about what you have learned about transgenic organisms and xenotransplantation and complete the following sentence stems.

In the past, my viewpoint was …

Currently, my viewpoint is …

The *I used to think ... now I think ...* thinking routine was developed by Project Zero, a research centre at the Harvard Graduate School of Education.

Gene therapy

gene therapy a process by which a copy of a functional gene is introduced into an organism

Gene therapy is a process by which a copy of a functional gene is introduced into an organism. The gene is then switched on to produce the functional protein that is missing. This technique aims to treat inherited disorders like cystic fibrosis by directly targeting the genotype (unlike treatments that target the symptoms). Technical difficulties may arise when trying to specifically target the cells of

affected tissue, and when targeting one specific gene without interfering with the function of other essential genes. Ethical issues also arise. Some people argue that gene therapy should be restricted to somatic tissue only, so the introduced gene is not transmitted through to the next generation. What do you think about this issue?

Science inquiry 2.3

DNA data storage

Digital technology currently works on the binary system, which only uses the digits 0 and 1. Any information that is input digitally is converted to this system, and digital devices will change those values to the necessary form of information. It's predicted that by 2035 the total volume of digital data generated by humans is expected to reach 2142 zettabytes (a zettabyte is equivalent to 1 000 000 000 000 000 000 000 bytes). Currently, most data is stored in the 'cloud', but the cloud is actually huge data centres stored in warehouses that require large amounts of energy to run.

However, by translating binary (0 or 1) values into one of the four DNA bases, it is possible for DNA to be reliably used to store huge amounts of digital data. Some scientists say that the entire world's data could be stored in DNA that would fill a shoebox.

Rather than storing 0s and 1s individually, DNA allows them to be stored in pairs such as this:

- 00 A
- 01 T
- 10 C
- 11 G.

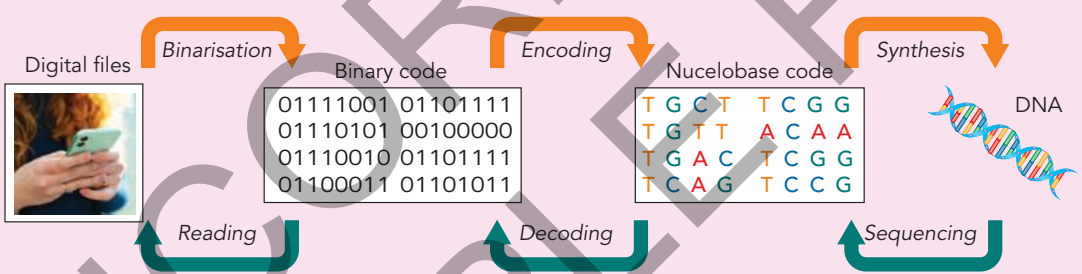


Figure 2.66 Encoding data into DNA code might be the future of efficient data storage.

After storing data in DNA, it can then be read using a DNA sequencer. This reads all of the bases and then converts it to binary. Improvements in sequencing techniques allow for billions of DNA sequences to be easily read at once and, when kept in suitable conditions, DNA can be preserved for hundreds of thousands of years without any energy input.

New sequencing techniques are also allowing sections of DNA to be used as identification tags. This DNA barcoding is accelerating the pace of research in fields such as chemical engineering, materials science and nanotechnology.

Quick check 2.19

1. **Recall** what GMO stands for.
2. **Describe** the process of gene therapy.
3. **State** an example of the application of genetic screening.
4. **Discuss** the reasons for the adoption of genetic screening by groups in society.
5. **State** five conditions that are screened for in Western Australia through the newborn bloodspot screening tests.

Go online to access the interactive section review and more!

Section 2.4 review

Online quiz | Section questions | Teachers can assign tasks and track results

Section 2.4 questions

Remembering

1. If a mutation is to be passed on to the next generation, **state** where it must have occurred.
2. **Define** the term 'mutagen'.
3. **Recall** the definition of a genetically modified organism.

Understanding

4. Using the table below, answer the following questions.

Second letter in the codon

First letter in the codon	U		C		A		G		Third letter in the codon
U	UUU	Phe (F)	UCU	Ser (S)	UAU	Tyr (Y)	UGU	Cys (C)	U
	UUC		UCC		UAC		UGC		C
	UUA	Leu (L)	UCA		UAA	STOP	UGA	STOP	A
	UUG		UCG		UAG	STOP	UGG	Trp (W)	G
C	CUU	Leu (L)	CCU	Pro (P)	CAU	His (H)	CGU	Arg (R)	U
	CUC		CCC		CAC		CGC		C
	CUA		CCA		CAA	Gln (Q)	CGA		A
	CUG		CCG		CAG		CGG		G
A	AUU	Ile (I)	ACU	Thr (T)	AAU	Asn (N)	AGU	Ser (S)	U
	AUC		ACC		AAC		AGC		C
	AUA		ACA		AAA	Lys (K)	AGA	Arg (R)	A
	AUG	Met (M) START	ACG		AAG		AGG		G
G	GUU	Val (V)	GCU	Ala (A)	GAU	Asp (D)	GGU	Gly (G)	U
	GUC		GCC		GAC		GGC		C
	GUA		GCA		GAA	Glu (E)	GGA		A
	GUG		GCG		GAG		GGG		G

The original DNA strand is GTC GGG ATA CGG CTC.
A gene mutation occurs, whereby the C in the first codon is replaced with a T, resulting in GTT GGG ATA CGG CTC.

a) **State** the name of this gene mutation.
b) **State** the mRNA strand that was copied from the original DNA strand.
c) **State** the new mRNA strand produced from the mutated DNA strand.
d) **State** whether the amino acids stay the same or change.
e) Another mutation occurs, whereby a C was added to the beginning of the above original DNA strand, giving CGTC GGG ATT CGG CTC.
State the name of this mutation and describe how it would affect the subsequent amino acids that it codes for.
f) Write out the mRNA code for this mutation and **identify** the corresponding amino acids.

Applying

5. **Explain** what will happen if a nucleotide is deleted from the sequence of nucleotides within a gene.
6. **Explain** what usually results when mutations occur in non-germ (somatic) cells.
7. **Explain** whether or not all mutations are detrimental (harmful).
8. **Explain** how a mutated gene can lead to a genetic disorder.
9. **Illustrate** a diagram to outline how non-disjunction may occur and cause aneuploidy.

Analysing

10. **Contrast** a genetically modified organism and a transgenic organism.
11. **Contrast** a spontaneous mutation and an induced mutation.

Evaluating

12. Using the DNA sequence ACAATTGGTAGCTGAGTTGGCCCGTA, **create** an example of the following mutations.
    a) substitution
    b) inversion
    c) deletion
    d) insertion.
13. **Discuss** why the substitution of one nucleotide may not be as critical to the functioning of the protein as the deletion or insertion of a nucleotide.
14. **Propose** an explanation for the relationship between increased rates of skin cancer and the thinning of the ozone layer.
15. **Construct** a mind map that shows the different types of mutations that may occur.

Chapter review

Chapter checklist

Success criteria		Linked questions
2.1	I can recall the components that make up the structure of DNA and RNA molecules.	6, 13
2.1	I can describe the relationship between genes, DNA and chromosomes.	1
2.1	I can describe the steps in which DNA is used to synthesise proteins.	7, 18
2.2	I can recall the steps involved in DNA replication.	3, 4, 15
2.2	I can describe the stages of cell division in mitosis.	17
2.2	I can describe the stages of cell division in meiosis.	17
2.2	I can use a karyotype to determine information about an individual.	10
2.3	I can define the terms 'homozygous', 'heterozygous', 'dominant' and 'recessive' with respect to genetic inheritance.	5
2.3	I can use and interpret a Punnett square to identify possible genotypes and phenotypes of offspring.	20, 23
2.3	I can explain sex linkage.	11, 19
2.3	I can distinguish between codominance and incomplete dominance.	22
2.3	I can use a pedigree diagram to determine the type of inheritance that is occurring.	19
2.4	I can explain the different types of mutations, including their causes and effects.	16
2.4	I can describe some ways in which DNA can be manipulated.	25

Go online to access the interactive chapter review

Scorcher competition

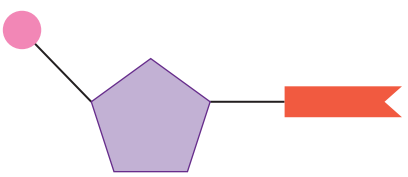
Review questions

Data questions

Review questions

Remembering

1. **State** the name of the position on a chromosome where a particular gene is located.
2. **Recall** two autosomal recessive inheritable traits.
3. **Recall** the name of the enzyme that unwinds the DNA double helix during DNA replication.
4. **Name** the structure formed when DNA condenses and becomes visible before cell division.
5. **Define** the terms 'homozygous', 'heterozygous', 'dominant' and 'recessive'.
6. **Identify** the parts of the nucleotide in the following figure.
7. **State** the location in the cell where transcription and translation occur.
8. **State** how many chromosomes you would expect to find in a:
   a) human somatic cell
   b) human gamete
   c) human somatic cell with Down syndrome.

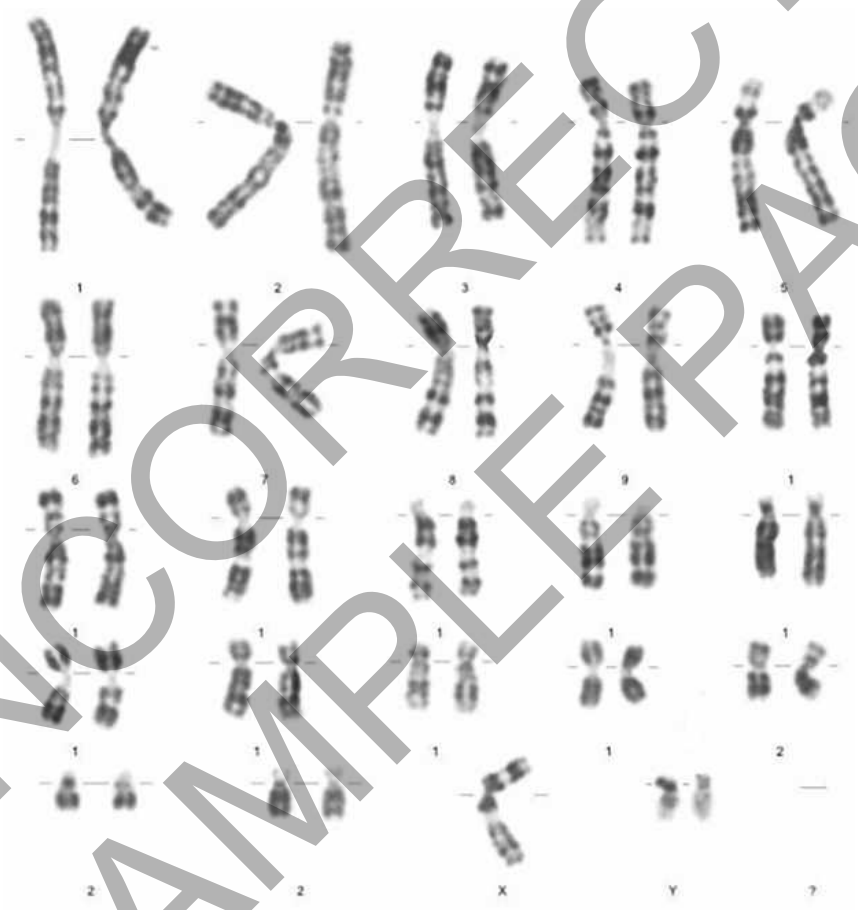


Understanding

9. **Explain** why males have more chance than females of inheriting an X-linked recessive trait.
10. Assuming eye colour in humans is controlled by a single pair of genes, and that brown eyes are dominant to blue eyes, **determine** the genotype of a brown-eyed man if he and a woman with blue eyes produce a baby with blue eyes.
11. **Illustrate** a diagram to show how two nucleotides join together with hydrogen bonds.
12. **Explain** why chromosomes are not always visible.
13. When cells reproduce themselves, the DNA is replicated. **Explain** why this is necessary.
14. **Explain** how aneuploidy occurs.
15. **Contrast** meiosis and mitosis.
16. **Illustrate** a diagram to show the processes of transcription and translation.

Applying

17. **Identify** what information can be gained from the following karyotype.



18. Using a Punnett square, **determine** the genotypes and phenotypes of the children if a normal-vision female carrier of colour blindness has children with a normal-vision male.

Parents: $X^BX^b \times X^BX$

gametes	X^B	X^b
X^B	X^BY^B	X^BY^b
Y	X^BY	X^bY

19. **Construct** a pedigree with the following information.
A woman with Huntington's disease marries a man who does not have Huntington's disease. They have three children. The first child is a male, the second child is a female, and both do not have the disease. The third child, a male, does develop the disease. Remember that Huntington's disease is an autosomal dominant genetic disorder.

Analysing

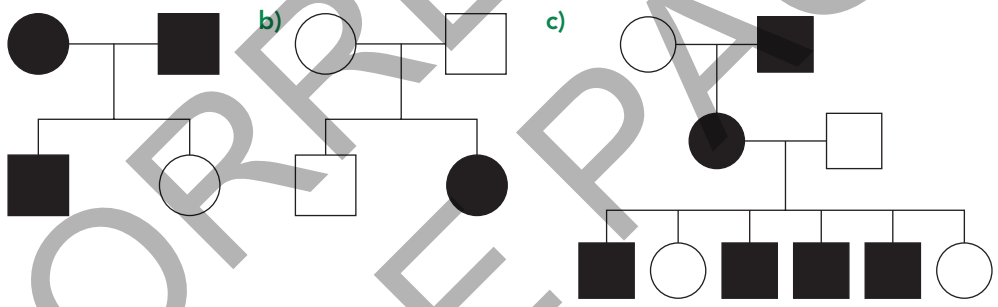
20. The table below shows several different organisms with their diploid chromosome number.

Organism	Chromosome number (2*n*)
Chimpanzee	48
Human	46
Horse	64
Onion	16
House fly	12
Worm	20

a) **Identify** the organism with the highest number of chromosomes in its gametes.
b) **State** the haploid number of the onion.

21. Use information from the following pedigrees to **infer** the inheritance pattern shown.

a) **b)** **c)**



22. An abandoned baby was handed in at the police station. Later, two women claimed to be the baby's mother. Blood studies revealed that woman 1 had blood type A, and that woman 2 was blood type AB. The baby was blood type O. **Deduce** which woman could possibly be the baby's mother and which woman can be ruled out completely. Show your working.

23. Chondrodystrophy is an autosomal recessive condition that is governed by a single gene with two alleles. In turkeys, affected embryos die approximately 16 days after fertilisation, so they do not survive long enough to hatch. Two turkeys that are heterozygous for the condition are crossed. Show your working to **determine** the phenotypic and genotypic ratios of the offspring.
Let C = normal allele and c = allele with chondrodystrophy.

Evaluating

24. Personal genome screening can be readily ordered over the internet. **Evaluate** the pros and cons of having DNA testing so readily available at an affordable cost.

25. You had genome screening and found out that your genes conferred a 25% chance of developing a disease. However, you also know that environmental and lifestyle factors play a large role in whether you develop the disease.
a) **Evaluate** how this information would affect your life.
b) **Discuss** how this scenario is different from knowing about a disease that is not influenced by lifestyle factors.

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Data questions

A student prepared an onion root tip slide and examined the cells using a light microscope. She recorded the number of cells in each stage of the cell cycle.

The table shows her results.

Stage of cell cycle	Number of cells recorded
Interphase	340
Prophase	13
Metaphase	4
Anaphase	3
Telophase	6

Table 2.6 Number of onion cells in each stage of the cell cycle

She then calculated the mitotic index for different areas of the root tip. The graph shows her results.

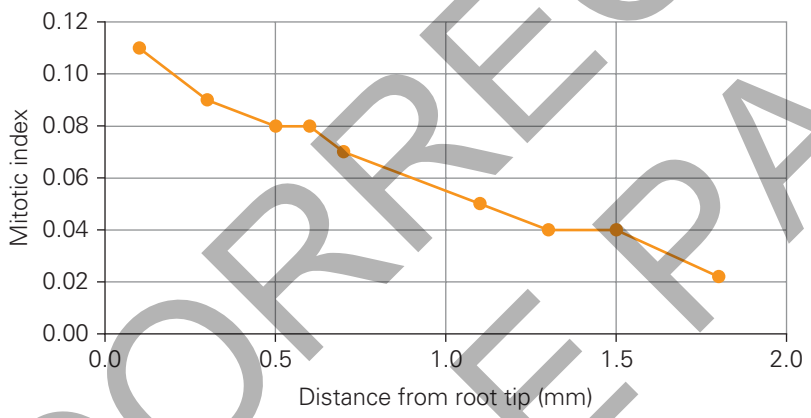

Figure 2.67 Mitotic index plotted against distance from root tip

1. The mitotic index of dividing tissue is calculated with the following formula:

$$\text{Mitotic index} = \frac{\text{number of cells seen that were undergoing mitosis}}{\text{total number of cells seen}}$$

   Calculate the mitotic index of the tissue shown in the table.
2. The duration of any phase of the cell cycle can be calculated with the following formula:

$$\text{Phase duration (minutes)} = \frac{\text{number of cells seen in that phase}}{\text{total number of cells seen}} \times \text{cell cycle duration of the organism.}$$

   The cell cycle duration for the onion is 24 hours.
   Calculate the duration of metaphase.
3. **Identify** the trend shown in the graph.
4. **Extrapolate** the data in the graph to estimate the mitotic index of the cells found 2 mm from the root tip.

STEM activity: Designing and prototyping an assistive device for individuals with a genetic disease

Background information

When a mutation occurs that is not beneficial, complications occur. An example of a disease with genetic causes is scoliosis. Scoliosis is a sideways curvature of the spine that usually occurs before puberty. Genetic diseases such as cerebral palsy and muscular dystrophy can cause scoliosis, but the cause of most scoliosis is unknown.

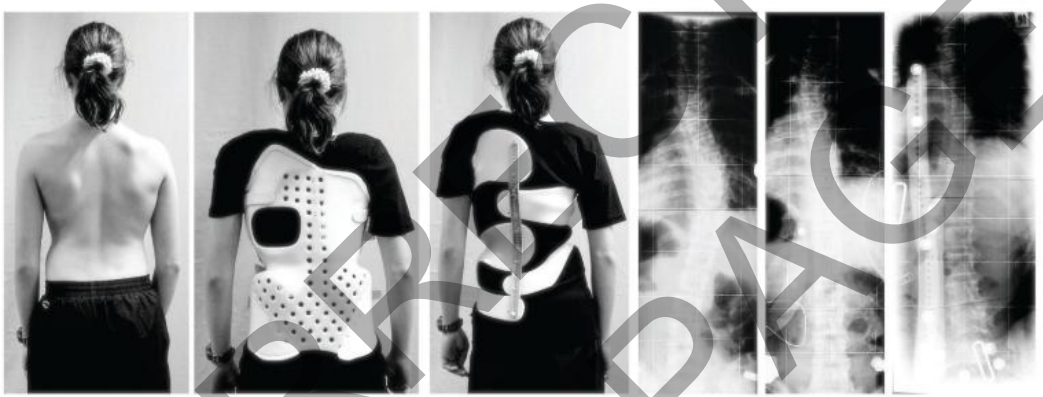

Figure 2.68 Various brace designs can be used for the treatment of scoliosis.

DESIGN BRIEF

Research a genetic disease. Design and build an apparatus that can help to improve the quality of life for people with the selected disease.

Activity instructions

In groups of three or four, conduct basic research on a genetic disease. Then design an apparatus that will improve the quality of life for people with that condition. Consider how the product would be built and marketed. Each team member needs to have a clear role but must be able to contribute to all aspects of the project.

Suggested materials

- computer
- pencil
- paper
- ruler
- balsa wood
- plaster
- papier mâché
- chicken wire
- 3D printer
- cardboard
- poster paper

Research and feasibility

1. Research genetic diseases and as a group decide which genetic disease will be the focus.
2. Create a table of the causes and effects of the disease.

Genetic disease cause	Effects on part of body	Ideas to help
e.g. Muscular dystrophy Muscle loss	Reduced joint movement	Brace and orthotics for feet
e.g. Haemophilia Blood doesn't clot effectively	Whole body can be bruised easily	1. Creation of children's play suit 2. Creation of some type of buffing edging that could be applied to furniture

3. Find pictures or diagrams of current equipment used, and annotate them with features and characteristics that relate to assisting people living with this disease.

Design and sustainability

4. Sketch potential design solutions (at least two) and annotate the purpose of the apparatus and what it is made of. Describe how it will improve the quality of life for people with this disease. How will it improve on current aids or tools (if any)?
5. Reflect on the materials you would use in real-world construction and comment on the durability and sustainability of the materials.

Create

6. Build a prototype of your design using available construction materials.

Evaluate and modify

7. Reflect on the prototype you have created and its effectiveness to help people with your chosen genetic disease.
8. Discuss as a group the modifications you would make in your solution to increase the effectiveness of design.
9. Present your prototype to the class. Outline the effectiveness of the prototype and demonstrate supporting ideas that show how the prototype would improve the quality of life of a person with this genetic disease.