Contents

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Module 7: Infectious disease

Module 8: Non-infectious disease and disorders

How to use this book

The *Pearson Biology 12 New South Wales* Skills and Assessment book takes an intuitive, self-paced approach to science education that ensures every student has opportunities to practise, apply and extend their learning through a range of supportive and challenging activities. While offering opportunities for reinforcement of key concepts, knowledge and skills, these activities enable flexibility in the approach to teaching and learning.

Explicit scaffolding makes learning objectives clear, and there are regular opportunities for student reflection and self-evaluation at the end of individual activities throughout the book. Students are also guided in self-reflection at the end of each module. There are rich opportunities to take the content further with the explicit coverage of Working scientifically skills and key knowledge in the depth studies.

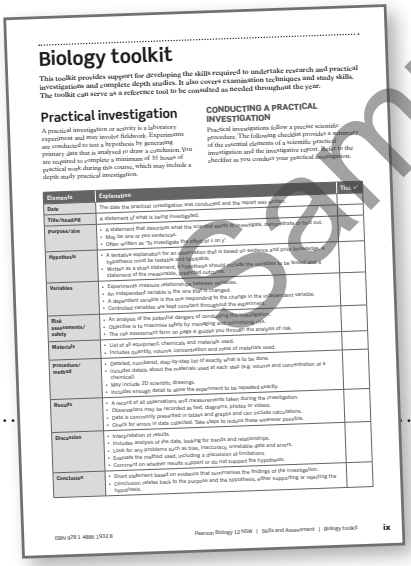
This resource has been written to fully align with the new Stage 6 Syllabus for New South Wales Biology and addresses the final four modules of the syllabus. Each module consists of five main sections:

- key knowledge
- worksheets
- practical activities
- depth study
- module review questions.

Explore how to use this book below.

Biology toolkit

The Biology toolkit supports development of the skills and techniques needed to undertake practical investigations, secondary-sourced investigations and depth studies, and covers examination techniques and study skills. It also includes checklists, models, exemplars and scaffolded steps. The toolkit can serve as a reference tool, to be consulted as needed.



Biology toolkit

This toolkit provides support for developing the skills required to undertake research and practical investigations and complete depth studies. It also covers examination techniques and study skills. The toolkit can serve as a reference tool to be consulted as needed throughout the year.

Practical investigation

A practical investigation or activity is a laboratory experiment and may involve fieldwork. Experiments are conducted to test a hypothesis by generating primary data that is analysed to draw a conclusion. You are required to complete a minimum of 35 hours of practical work during this course, which may include a depth study or practical investigation.

CONDUCTING A PRACTICAL INVESTIGATION

Practical investigations follow a precise scientific procedure. The following checklist provides a summary of the essential elements of a scientific practical investigation and the investigative cycle. Refer to the checklist as you conduct your practical investigation.

Elements	Explanation	Tick ✓
Date	The date the practical investigation was conducted and the report was written.	
Title/heading	A statement of what is being investigated.	
purpose/aim	• A statement that describes what the scientific work was to investigate, determine or find out. • May be one or two sentences. • Often written as 'To investigate the effect of x on y.'	
Hypothesis	• A tentative explanation for an observation that is based on evidence and prior knowledge. A hypothesis could be testable and falsifiable. • Written as a short statement, a hypothesis should include a prediction. It can be worded as a statement of the measurable, specified relationship.	
Variables	• Experiments measure relationships between variables. • An independent variable is the one you've changed. • A dependent variable is the one responded to the change in the independent variable. • Controlled variables are kept constant throughout the experiment.	
Risk assessments/ safety	• An analysis of the potential dangers of conducting the investigation. • Objective is to minimise safety by managing or controlling risk. • The risk assessment form on p466 is guided you through the analysis of risk.	
Materials	• List of all equipment, chemicals and materials used. • Includes quantity, volume, concentration and name of materials used.	
procedure/ method	• Detailed, numbered, step-by-step list of exactly what is to be done. • Includes details about the materials used at each step (e.g. volume and concentration of a chemical). • May include 2D scientific drawings. • Includes enough detail to allow the experiment to be repeated exactly.	
Results	• A record of all observations and measurements taken during the investigation. • Observations may be recorded as text, diagrams, photos or videos. • Data is commonly presented in tables and graphs and can include calculations. • Check for errors in data collected. Take steps to reduce these wherever possible.	
Discussion	• Interpretation of results. • Includes analysis of the data, looking for trends and relationships. • Look for any problems such as bias, inaccuracy, unreliable data and errors. • Evaluate the method used, including a discussion of limitations. • Comment on whether results support or do not support the hypothesis.	
Conclusion	• Short statement based on evidence that summarises the findings of the investigation. • Conclusion relates back to the purpose and the hypothesis, either supporting or rejecting the hypothesis.	

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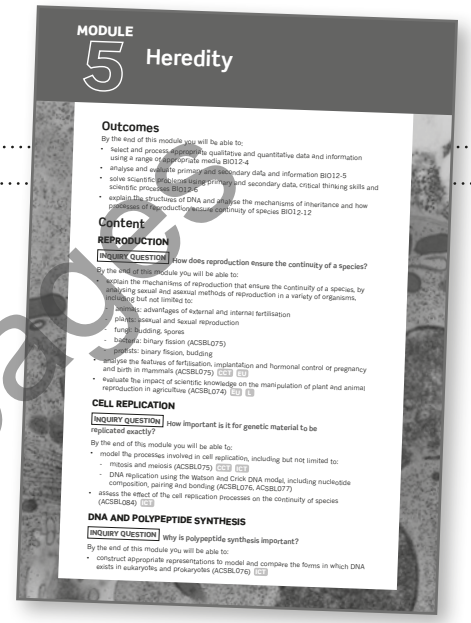
Key knowledge

Each module begins with a key knowledge section. This consists of a set of succinct summary notes that cover the key knowledge set out in each module of the syllabus. This section is highly illustrative and written in a straightforward style to assist students of all reading abilities. Key terms are in bold for ease of navigation. It also serves as a ready reference for completing the worksheets and practical activities.



MODULE 5 Heredity

Outcomes

By the end of this module you will be able to:

- select and process appropriate qualitative and quantitative data and information using a range of appropriate media BIO12-4
- analyse and evaluate primary and secondary data and information BIO12-5
- solve scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO12-6
- explain the structures of DNA and analyse the mechanisms of inheritance and how processes of reproduction ensure continuity of species BIO12-12

Content

REPRODUCTION

INQUIRY QUESTION How does reproduction ensure the continuity of a species?

By the end of this module you will be able to:

- explain the mechanisms of reproduction that ensure the continuity of a species, by analysing sexual and asexual methods of reproduction in a variety of organisms, including but not limited to:
  - genetic advantages of external and internal fertilisation
  - plants: asexual and sexual reproduction
  - fungi: budding, spores
  - bacteria: binary fission (ACSBL075)
  - protists: binary fission, budding
- analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals (ACSBL075) CS3 IT
- evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture (ACSBL074) EU L

CELL REPLICATION

INQUIRY QUESTION How important is it for genetic material to be replicated exactly?

By the end of this module you will be able to:

- model the processes involved in cell replication, including but not limited to:
  - mitosis and meiosis (ACSBL075) CS3 IT
  - DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding (ACSBL076, ACSBL077)
- assess the effect of the cell replication processes on the continuity of species (ACSBL084) IT

DNA AND POLYPEPTIDE SYNTHESIS

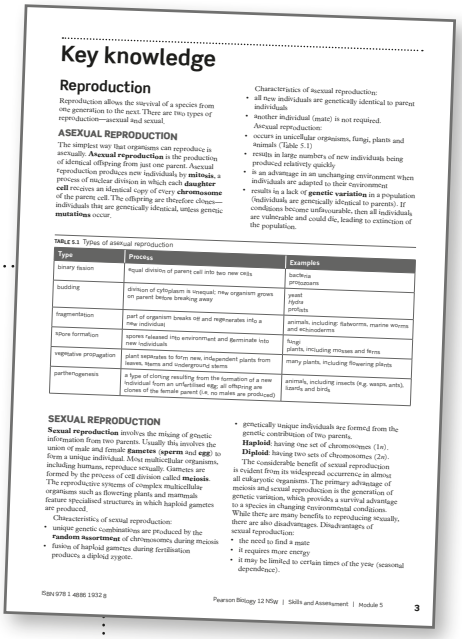
INQUIRY QUESTION Why is polypeptide synthesis important?

By the end of this module you will be able to:

- construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes (ACSBL076) IT

Module opener

Each book is divided to follow the four modules of the syllabus, with the module opener linking the module content to the syllabus.



Key knowledge

Reproduction

Reproduction allows the survival of a species from one generation to the next. There are two types of reproduction—asexual and sexual.

ASEXUAL REPRODUCTION

The simplest way that organisms can reproduce is asexually. **Asexual reproduction** is the production of identical offspring from just one parent. Asexual reproduction produces new individuals by **mitosis**, a process of nuclear division in which each **daughter cell** receives an identical copy of every **chromosome** of the parent cell. The offspring are therefore clones—individuals that are genetically identical, unless genetic **mutations** occur.

Characteristics of asexual reproduction:

- all new individuals are genetically identical to parent individuals
- another individual (mate) is not required. Asexual reproduction:
- occurs in unicellular organisms, fungi, plants and animals (Table 5.1)
- results in large numbers of new individuals being produced relatively quickly
- is an advantage in an unchanging environment when individuals are adapted to their environment
- results in a lack of **genetic variation** in a population (individuals are genetically identical to parents). If conditions become unfavourable, then all individuals are vulnerable and could die, leading to extinction of the population.

TABLE 5.1 Types of asexual reproduction

Type	Process	Examples
binary fission	equal division of parent cell into two new cells	bacteria protozoans
budding	division of cytoplasm is unequal; new organism grows on parent before breaking away	yeast *Hydra* protists
fragmentation	part of organism breaks off and regenerates into a new individual	animals, including flatworms, marine worms and echinoderms
spore formation	spores released into environment and germinate into new individuals	fungi plants, including mosses and ferns
vegetative propagation	plant separates to form new, independent plants from leaves, stems and underground stems	many plants, including flowering plants
parthenogenesis	a type of cloning resulting from the formation of a new individual from an unfertilised egg; all offspring are clones of the female parent (i.e. no males are produced)	animals, including insects (e.g. wasps, ants), lizards and birds

SEXUAL REPRODUCTION

Sexual reproduction involves the mixing of genetic information from two parents. Usually this involves the union of male and female **gametes** (**sperm** and **egg**) to form a unique individual. Most multicellular organisms, including humans, reproduce sexually. Gametes are formed by the process of cell division called **meiosis**. The reproductive systems of complex multicellular organisms such as flowering plants and mammals feature specialised structures in which haploid gametes are produced.

Characteristics of sexual reproduction:

- unique genetic combinations are produced by the **random assortment** of chromosomes during meiosis
- fusing of haploid gametes during fertilisation produces a diploid zygote.
- genetically unique individuals are formed from the genetic contribution of two parents.

Haploid: having one set of chromosomes (1*n*).

Diploid: having two sets of chromosomes (2*n*).

The considerable benefit of sexual reproduction is evident from its widespread occurrence in almost all eukaryotic organisms. The primary advantage of meiosis and sexual reproduction is the generation of genetic variation, which provides a survival advantage to a species in changing environmental conditions. While there are many benefits to reproducing sexually, there are also disadvantages. Disadvantages of sexual reproduction:

- the need to find a mate
- it requires more energy
- it may be limited to certain times of the year (seasonal dependence).

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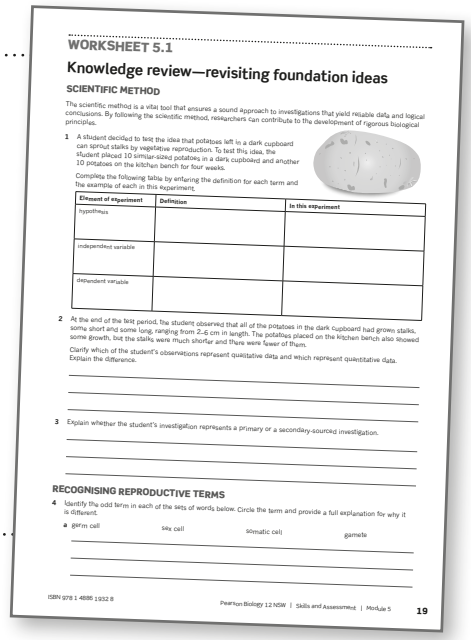
Worksheets

A diverse offering of instructive and self-contained worksheets is included in each module. Common to all modules are the initial 'Knowledge review' worksheet to activate prior knowledge, a 'Literacy review' worksheet to explicitly build understanding and application of scientific terminology, and finally a 'Thinking about my learning' worksheet, which students can use for reflection and self-assessment. Other worksheet types provide opportunities to revise, consolidate and further student understanding.

All worksheets function as formative assessment and are clearly aligned to the syllabus. A range of questions building from foundation to challenging are included within the worksheets.



WORKSHEET 5.1

Knowledge review—revisiting foundation ideas

SCIENTIFIC METHOD

The scientific method is a vital tool that ensures a sound approach to investigations that yield reliable data and logical conclusions. By following the scientific method, researchers can contribute to the development of rigorous biological principles.

1 A student decided to test the idea that potatoes left in a dark cupboard can sprout stalks by vegetative reproduction. To test this idea, the student placed 10 similar-sized potatoes in a dark cupboard and another 10 potatoes on the kitchen bench for four weeks.

Complete the following table by entering the definition for each term and the example of each in this experiment.

Element of experiment	Definition	In this experiment
hypothesis		
independent variable		
dependent variable		

2 At the end of the test period, the student observed that all of the potatoes in the dark cupboard had grown stalks, some short and some long, ranging from 2–6 cm in length. The potatoes placed on the kitchen bench also showed some growth, but the stalks were much shorter and there were fewer of them.

Clarify which of the student's observations represent qualitative data and which represent quantitative data. Explain the difference.

3 Explain whether the student's investigation represents a primary or a secondary-sourced investigation.

RECOGNISING REPRODUCTIVE TERMS

4 Identify the odd term in each of the sets of words below. Circle the term and provide a full explanation for why it is different.

a germ cell sex cell somatic cell gamete

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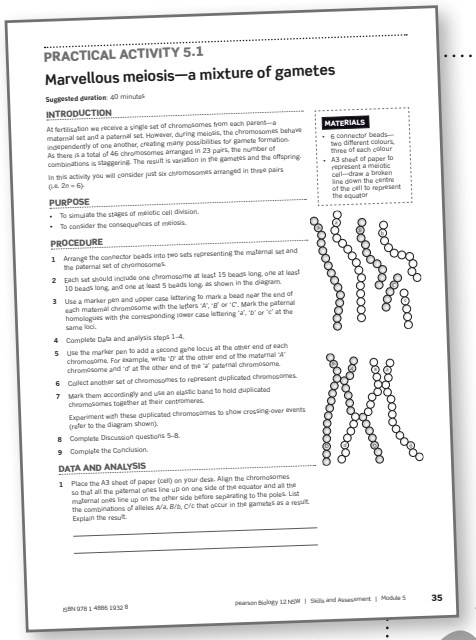

PRACTICAL ACTIVITY 5.1

Marvellous meiosis—a mixture of gametes

Suggested duration: 40 minutes

INTRODUCTION

At fertilisation we receive a single set of chromosomes from each parent—a maternal set and a paternal set. However, during meiosis, the chromosomes behave independently of one another, creating many possibilities for gamete formation. As there is a total of 46 chromosomes arranged in 23 pairs, the number of combinations is staggering. The result is variation in the gametes and the offspring.

In this activity you will consider just six chromosomes arranged in three pairs (i.e. 2n = 6).

MATERIALS

- 6 connector beads—two different colours, three of each colour
- A3 sheet of paper to represent a meiotic cell—draw a broken line down the centre of the cell to represent the equator

PURPOSE

- To simulate the stages of meiotic cell division.
- To consider the consequences of meiosis.

PROCEDURE

1 Arrange the connector beads into two sets representing the maternal set and the paternal set of chromosomes.

2 Each set should include one chromosome at least 15 beads long, one at least 10 beads long, and one at least 5 beads long, as shown in the diagram.

3 Use a marker pen and upper case lettering to mark a bead near the end of each maternal chromosome with the letters 'A', 'B' or 'C'. Mark the paternal homologues with the corresponding lower case lettering 'a', 'b' or 'c' at the same loci.

4 Complete Data and analysis steps 1–4.

5 Use the marker pen to add a second gene locus at the other end of each chromosome. For example, write 'D' at the other end of the maternal 'A' chromosome and 'd' at the other end of the 'a' paternal chromosome.

6 Collect another set of chromosomes to represent duplicated chromosomes.

7 Mark them accordingly and use an elastic band to hold duplicated chromosomes together at their centromeres.

Experiment with these duplicated chromosomes to show crossing-over events (refer to the diagram shown).

8 Complete Discussion questions 5–8.

9 Complete the Conclusion.

DATA AND ANALYSIS

1 Place the A3 sheet of paper (cell) on your desk. Align the chromosomes so that all the paternal ones line up on one side of the equator and all the maternal ones line up on the other side before separating to the poles. List the combinations of alleles A/a, B/b, C/c that occur in the gametes as a result. Explain the result.

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Practical activities

Practical activities give students the opportunity to complete practical work related to the various themes covered in the syllabus. All practical activities referenced in outcomes within the syllabus have been covered. Across the suite of practical activities, students have opportunities to design, conduct, evaluate, gather and analyse data, appropriately record results and prepare evidence-based conclusions. This can be done directly into the scaffolded practical activities. Students also have opportunities to evaluate safety and risk, and identify any potential hazards.

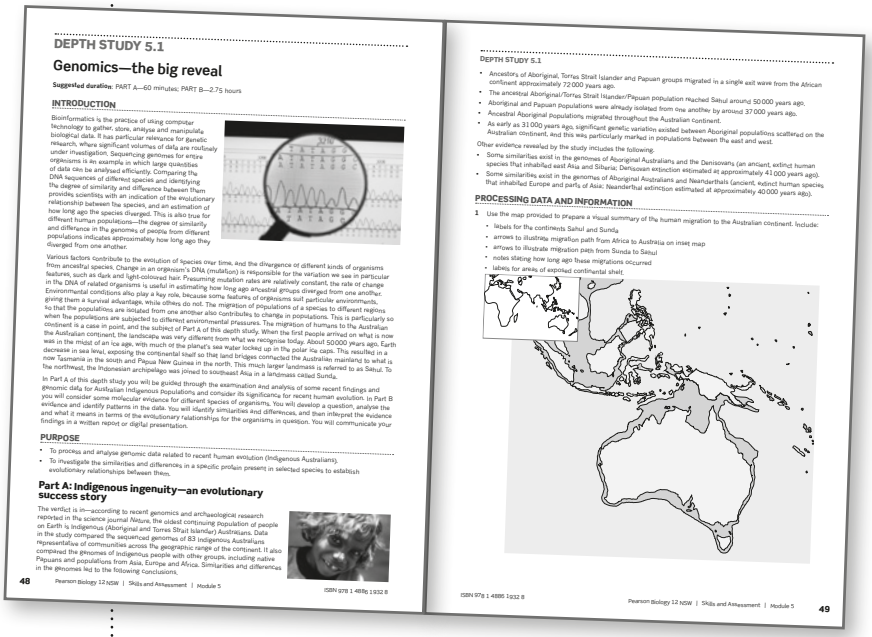
Each practical activity includes a suggested duration. Along with the depth studies, the practical activities meet the 35 hours of practical work mandated for Year 12 in the syllabus. Where there is key knowledge that will support the completion of a practical activity, students are referred back to the appropriate section.

Like the worksheets, the practical activities include a range of questions, building from foundation to challenging.

Depth study

Each module contains one suggested depth study. The depth studies allow further development of one or more concepts found within or inspired by the syllabus. They allow students to acquire a depth of understanding and take responsibility for their own learning, and promote differentiation and engagement.

Each depth study allows for the demonstration of a range of Working scientifically skills, with all depth studies assessing the Working scientifically outcomes of Questioning and predicting, and Communicating. A minimum of two additional Working scientifically skills and at least one Knowledge and understanding outcome are also assessed.



DEPTH STUDY 5.1

Genomics—the big reveal

Suggested duration: PART A—60 minutes; PART B—2.75 hours

INTRODUCTION

Bioinformatics is the practice of using computer technology to gather, store, analyse and manipulate biological data. It has particular relevance for genetic research, where significant volumes of data are routinely under investigation. Sequencing genomes for entire organisms is an example in which large quantities of data can be analysed efficiently. Comparing the DNA sequences of different species and identifying the degree of similarity and difference between them provides us with an indication of the evolutionary relationship between the species, and an estimation of how long ago the species diverged. This is also true for different human populations—the degree of similarity and difference in the genomes of people from different populations indicates approximately how long ago they diverged from one another.

Various factors contribute to the evolution of species over time. And the divergence of different kinds of organisms from ancestral species. Change in an organism's DNA (mutation) is responsible for the variation we see in particular features, such as dark and light-coloured hair. Presuming mutation rates are relatively constant, the rate of change in the DNA of related organisms is useful in estimating how long ago ancestral groups diverged from one another. Environmental conditions also play a key role, because some features of organisms suit particular environments, giving them a survival advantage, while others do not. The migration of populations of a species to different regions so that the populations are isolated from one another also contributes to change in populations. This is particularly so when the populations are subjected to different environmental pressures. The migration of humans to the Australian continent is a case in point, and the subject of Part A of this depth study. When the first people arrived on what is now the Australian continent, the landscape was very different from what we recognise today. About 50 000 years ago, Earth was in the midst of an ice age, with much of the planet's sea water locked up in the polar ice caps. This resulted in a decrease in sea level, exposing the continental shelf so that land bridges connected the Australian mainland to what is now Tasmania in the south and Papua New Guinea in the north. This much larger landmass is referred to as Sahul. To the northwest, the Indonesian archipelago was joined to southeast Asia in a landmass called Sunda.

In Part A of this depth study you will be guided through the examination and analysis of some recent findings and genomic data for Australian Indigenous populations and consider its significance for recent human evolution. In Part B you will consider some molecular evidence for different species of organisms. You will develop a question, analyse the evidence and identify patterns in the data. You will identify similarities and differences, and then interpret the evidence and what it means in terms of the evolutionary relationships for the organisms in question. You will communicate your findings in a written report or digital presentation.

PURPOSE

- To process and analyse genomic data related to recent human evolution (Indigenous Australians).
- To investigate the similarities and differences in a specific protein present in selected species to establish evolutionary relationships between them.

Part A: Indigenous ingenuity—an evolutionary success story

The verdict is in—according to recent genomics and archaeological research reported in the science journal *Nature*, the oldest continuing population of people on Earth is Indigenous (Aboriginal and Torres Strait Islander) Australians. Data in the study compared the sequenced genomes of 83 Indigenous Australians representative of communities across the geographic range of the continent. It also compared the genomes of Indigenous people with other groups, including native Papuans and populations from Asia, Europe and Africa. Similarities and differences in the genomes led to the following conclusions.

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DEPTH STUDY 5.1

- Ancestors of Aboriginal, Torres Strait Islander and Papuan groups migrated in a single exit wave from the African continent approximately 72 000 years ago.
- The ancestral Aboriginal/Torres Strait Islander/Papuan population reached Sahul around 50 000 years ago.
- Aboriginal and Papuan populations were already isolated from one another by around 37 000 years ago.
- Ancestral Aboriginal populations migrated throughout the Australian continent.
- As early as 31 000 years ago, significant genetic variation existed between Aboriginal populations scattered on the Australian continent, and this was particularly marked in populations between the east and west.

Other evidence revealed by the study includes the following.

- Some similarities exist in the genomes of Aboriginal Australians and the Denisovans (an ancient, extinct human species that inhabited east Asia and Siberia). Denisovan extinction estimated at approximately 41 000 years ago).
- Some similarities exist in the genomes of Aboriginal Australians and Neanderthals (ancient, extinct human species that inhabited Europe and parts of Asia). Neanderthal extinction estimated at approximately 40 000 years ago).

PROCESSING DATA AND INFORMATION

1 Use the map provided to prepare a visual summary of the human migration to the Australian continent. Include:

- labels for the continents Sahul and Sunda
- arrows to illustrate migration path from Africa to Australia on inset map
- arrows to illustrate migration path from Sunda to Sahul
- notes stating how long ago these migrations occurred
- labels for areas of exposed continental shelf.

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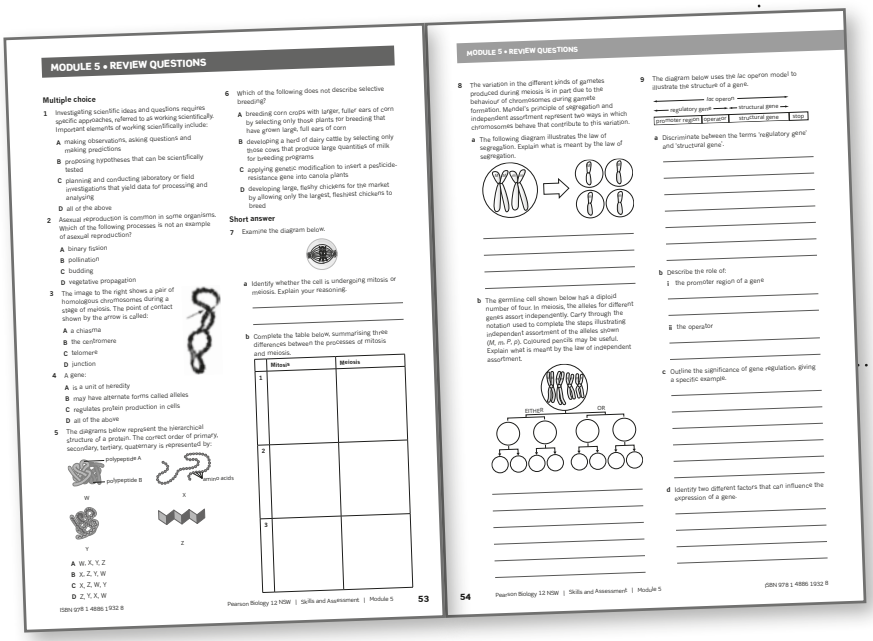
MODULE 5 • REVIEW QUESTIONS

Multiple choice

1 Investigating scientific ideas and questions requires specific approaches, referred to as working scientifically. Important elements of working scientifically include:
A making observations, asking questions and making predictions
B proposing hypotheses that can be scientifically tested
C planning and conducting laboratory or field investigations that yield data for processing and analysing
D all of the above

2 Asexual reproduction is common in some organisms. Which of the following processes is not an example of asexual reproduction?
A binary fission
B pollination
C budding
D vegetative propagation

3 The image to the right shows a pair of homologous chromosomes during a stage of meiosis. The point of contact shown by the arrow is called:
A a chiasma
B the centromere
C telomere
D junction

4 A gene:
A is a unit of heredity
B may have alternate forms called alleles
C regulates protein production in cells
D all of the above

5 The diagrams below represent the hierarchical structure of a protein. The correct order of primary, secondary, tertiary, quaternary is represented by:

polypeptide A
polypeptide B
amino acids
W
X
Y
Z

A W, X, Y, Z
B X, Z, Y, W
C X, Z, W, Y
D Z, Y, X, W

6 Which of the following does not describe selective breeding?
A breeding corn crops with larger, fuller ears of corn by selecting only those plants for breeding that have grown large, full ears of corn
B developing a herd of dairy cattle by selecting only those cows that produce large quantities of milk for breeding programs
C applying genetic modification to insert a pesticide-resistance gene into canola plants
D developing large, fleshy chickens for the market by allowing only the largest, fleshiest chickens to breed

Short answer

7 Examine the diagram below.

a Identify whether the cell is undergoing mitosis or meiosis. Explain your reasoning.

b Complete the table below, summarising three differences between the processes of mitosis and meiosis.

	Mitosis	Meiosis
1		
2		
3		

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MODULE 5 • REVIEW QUESTIONS

8 The variation in the different kinds of gametes produced during meiosis is in part due to the behaviour of chromosomes during gamete formation. Mendel's principle of segregation and independent assortment represent two ways in which chromosomes behave that contribute to this variation.

a The following diagram illustrates the law of segregation. Explain what is meant by the law of segregation.

b The germline cell shown below has a diploid number of four. In meiosis, the alleles for different genes assort independently. Carry through the notation used to complete the steps illustrating independent assortment of the alleles shown (*M*, *m*, *P*, *p*). Coloured pencils may be useful. Explain what is meant by the law of independent assortment.

EITHER
OR

9 The diagram below uses the *lac* operon model to illustrate the structure of a gene.

lac operon
regulatory gene
structural gene
promoter region
operator
structural gene
stop

a Discriminate between the terms 'regulatory gene' and 'structural gene'.

b Describe the role of:
i the promoter region of a gene
ii the operator

c Outline the significance of gene regulation, giving a specific example.

d Identify two different factors that can influence the expression of a gene.

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Module review questions

Each module finishes with a comprehensive set of questions, consisting of multiple choice and short answer, which helps students to draw together their knowledge and understanding and apply it to these styles of question.

Rating my learning

Rating my learning is an innovative tool that appears at the bottom of the final page of most worksheets and all practical activities. It provides students with the opportunity for self-reflection and self-assessment. It encourages them to look ahead to how they can continue to improve, and it helps them to identify focus areas for further skill and knowledge development.

The teacher may choose to use student responses to the 'Rating my learning' feature as a formative assessment tool. At a glance, teachers can assess which topics and which students need intervention for improvement.

RATING MY LEARNING	My understanding improved	Not confident ○ ○ ○ ○ ○ Very confident	I answered questions without help	Not confident ○ ○ ○ ○ ○ Very confident	I corrected my errors without help	Not confident ○ ○ ○ ○ ○ Very confident

Icons and features

The 2018 New South Wales Biology Stage 6 Syllabus Learning Across the Curriculum content is addressed and identified:

AHC A CC CCT DD EU ICT
IU L N PSC S WE

GO TO ➤ **GoTo icons** are used to make important links to relevant content within the book.

The **safety icon** highlights significant hazards, indicating caution is needed.

The **safety glasses icon** highlights that protective eyewear is to be worn during the practical activity.

Highlight boxes focus students' attention on important information such as key definitions, formulae and summary points.

Caution: care is required when using sharp equipment such as a scalpel, and chemicals such as hydrogen peroxide. Check any special procedures with your teacher.

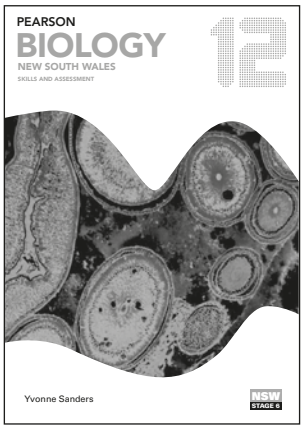
Teacher support

Comprehensive answers and fully worked solutions for all worksheets, practical activities, depth studies and module review questions are provided via the *Pearson Biology 12 New South Wales Teacher Support.* An editable suggested assessment rubric for depth studies is also provided.

Pearson Biology 12
New South Wales

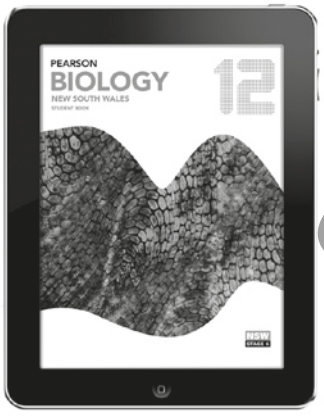
Student Book

Pearson Biology 12 New South Wales has been written to fully align with the 2018 New South Wales Biology Stage 6 Syllabus. The Student Book includes the very latest developments and applications of biology and incorporates best-practice literacy and instructional design to ensure the content and concepts are fully accessible to all students.



Skills and Assessment Book

The Skills and Assessment book gives students the edge in preparing for all forms of assessment. Key features include a toolkit, key knowledge summaries, worksheets, practical activities, suggested depth studies and module review questions. It provides guidance, assessment practice and opportunities to develop key skills.



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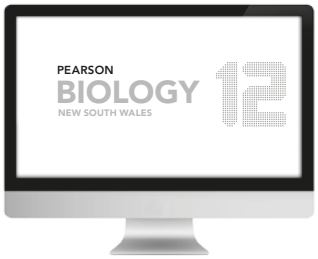

Teacher Support

Online teacher support for the series includes syllabus grids, a scope and sequence plan, and three practice exams per year level. Fully worked solutions to all Student Book questions are provided, as well as teacher notes for the chapter inquiry tasks. Skills and Assessment book resources include solutions to all worksheets, practical activities, depth studies and module review questions; teacher notes, safety notes, risk assessments and lab technician's checklists and recipes for all practical activities; and assessment rubrics and exemplar answers for depth studies.

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MODULE 5

Heredity

Outcomes

By the end of this module you will be able to:

- select and process appropriate qualitative and quantitative data and information using a range of appropriate media BIO12-4
- analyse and evaluate primary and secondary data and information BIO12-5
- solve scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO12-6
- explain the structures of DNA and analyse the mechanisms of inheritance and how processes of reproduction ensure continuity of species BIO12-12

Content

REPRODUCTION

INQUIRY QUESTION **How does reproduction ensure the continuity of a species?**

By the end of this module you will be able to:

- explain the mechanisms of reproduction that ensure the continuity of a species, by analysing sexual and asexual methods of reproduction in a variety of organisms, including but not limited to:
  - animals: advantages of external and internal fertilisation
  - plants: asexual and sexual reproduction
  - fungi: budding, spores
  - bacteria: binary fission (ACSBL075)
  - protists: binary fission, budding
- analyse the features of fertilisation, implantation and hormonal control of pregnancy and birth in mammals (ACSBL075) CCT EU
- evaluate the impact of scientific knowledge on the manipulation of plant and animal reproduction in agriculture (ACSBL074) EU L

CELL REPLICATION

INQUIRY QUESTION **How important is it for genetic material to be replicated exactly?**

By the end of this module you will be able to:

- model the processes involved in cell replication, including but not limited to:
  - mitosis and meiosis (ACSBL075) CCT ICT
  - DNA replication using the Watson and Crick DNA model, including nucleotide composition, pairing and bonding (ACSBL076, ACSBL077)
- assess the effect of the cell replication processes on the continuity of species (ACSBL084) ICT

DNA AND POLYPEPTIDE SYNTHESIS

INQUIRY QUESTION **Why is polypeptide synthesis important?**

By the end of this module you will be able to:

- construct appropriate representations to model and compare the forms in which DNA exists in eukaryotes and prokaryotes (ACSBL076) ICT

- model the process of polypeptide synthesis, including: (ACSBL079)
  - transcription and translation
  - assessing the importance of mRNA and tRNA in transcription and translation (ACSBL079)
  - analysing the function and importance of polypeptide synthesis (ACSBL080)
  - assessing how genes and environment affect phenotypic expression (ACSBL081) CCT L
- investigate the structure and function of proteins in living things L

GENETIC VARIATION

INQUIRY QUESTION **How can the genetic similarities and differences within and between species be compared?**

By the end of this module you will be able to:

- conduct practical investigations to predict variations in the genotype of offspring by modelling meiosis, including the crossing over of homologous chromosomes, fertilisation and mutations (ACSBL084)
- model the formation of new combinations of genotypes produced during meiosis, including but not limited to:
  - interpreting examples of autosomal, sex linkage, co-dominance, incomplete dominance and multiple alleles (ACSBL085) CCT
  - constructing and interpreting information and data from pedigrees and Punnett squares
- collect, record and present data to represent frequencies of characteristics in a population, in order to identify trends, patterns, relationships and limitations in data, for example: ICT N
  - examining frequency data
  - analysing single nucleotide polymorphism (SNP)

INHERITANCE PATTERNS IN A POPULATION

INQUIRY QUESTION **Can population genetic patterns be predicted with any accuracy?**

By the end of this module you will be able to:

- investigate the use of technologies to determine inheritance patterns in a population using, for example: (ACSBL064, ACSBL085) ICT
  - DNA sequencing and profiling (ACSBL086) EU
- investigate the use of data analysis from a large-scale collaborative project to identify trends, patterns and relationships, for example: (ACSBL064, ACSBL073) A CCT IU N
  - the use of population genetics data in conservation management S
  - population genetics studies used to determine the inheritance of a disease or disorder CCT ICT N
  - population genetics relating to human evolution IU

Key knowledge

Reproduction

Reproduction allows the survival of a species from one generation to the next. There are two types of reproduction—asexual and sexual.

ASEXUAL REPRODUCTION

The simplest way that organisms can reproduce is asexually. **Asexual reproduction** is the production of identical offspring from just one parent. Asexual reproduction produces new individuals by **mitosis**, a process of nuclear division in which each **daughter cell** receives an identical copy of every **chromosome** of the parent cell. The offspring are therefore clones—individuals that are genetically identical, unless genetic **mutations** occur.

Characteristics of asexual reproduction:

- all new individuals are genetically identical to parent individuals
- another individual (mate) is not required.

Asexual reproduction:

- occurs in unicellular organisms, fungi, plants and animals (Table 5.1)
- results in large numbers of new individuals being produced relatively quickly
- is an advantage in an unchanging environment when individuals are adapted to their environment
- results in a lack of **genetic variation** in a population (individuals are genetically identical to parents). If conditions become unfavourable, then all individuals are vulnerable and could die, leading to extinction of the population.

TABLE 5.1 Types of asexual reproduction

Type	Process	Examples
binary fission	equal division of parent cell into two new cells	bacteria protozoans
budding	division of cytoplasm is unequal; new organism grows on parent before breaking away	yeast *Hydra* protists
fragmentation	part of organism breaks off and regenerates into a new individual	animals, including: flatworms, marine worms and echinoderms
spore formation	spores released into environment and germinate into new individuals	fungi plants, including mosses and ferns
vegetative propagation	plant separates to form new, independent plants from leaves, stems and underground stems	many plants, including flowering plants
parthenogenesis	a type of cloning resulting from the formation of a new individual from an unfertilised egg; all offspring are clones of the female parent (i.e. no males are produced)	animals, including insects (e.g. wasps, ants), lizards and birds

SEXUAL REPRODUCTION

Sexual reproduction involves the mixing of genetic information from two parents. Usually this involves the union of male and female **gametes** (**sperm** and **egg**) to form a unique individual. Most multicellular organisms, including humans, reproduce sexually. Gametes are formed by the process of cell division called **meiosis**. The reproductive systems of complex multicellular organisms such as flowering plants and mammals feature specialised structures in which haploid gametes are produced.

Characteristics of sexual reproduction:

- unique genetic combinations are produced by the **random assortment** of chromosomes during meiosis
- fusion of haploid gametes during fertilisation produces a diploid zygote.
- genetically unique individuals are formed from the genetic contribution of two parents.

Haploid: having one set of chromosomes ($1n$).

Diploid: having two sets of chromosomes ($2n$).

The considerable benefit of sexual reproduction is evident from its widespread occurrence in almost all eukaryotic organisms. The primary advantage of meiosis and sexual reproduction is the generation of genetic variation, which provides a survival advantage to a species in changing environmental conditions. While there are many benefits to reproducing sexually, there are also disadvantages. Disadvantages of sexual reproduction:

- the need to find a mate
- it requires more energy
- it may be limited to certain times of the year (seasonal dependence).

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Fertilisation

Various structures and processes are adapted to allow haploid gametes to meet so that fertilisation can occur. The union of gametes in fertilisation may occur externally or internally, depending on the organism and its lifestyle (Figure 5.1). **External fertilisation** occurs when a male's sperm fertilises a female's egg outside the female's body. This method of reproduction is common in aquatic animals, for example amphibians, fish and sea urchins (Figure 5.1a, b). Eggs and developing young do not need to be carried inside a parent, and thousands of eggs can be fertilised at a time. However, because the developing young are exposed to the environment, many do not survive.

Internal fertilisation is when a male deposits sperm directly into a female's reproductive tract. This is a feature of many terrestrial (land) animals, including birds and mammals (Figure 5.1c, d; Figure 5.2). In mammals, fertilisation occurs in the fallopian tubes, with the **zygote** growing and dividing as it is swept towards the **uterus**. Internal fertilisation ensures gametes and developing young are protected from changes in the external environment, do not dehydrate, and are provided with adequate nutrition and a suitable environment in which to develop.

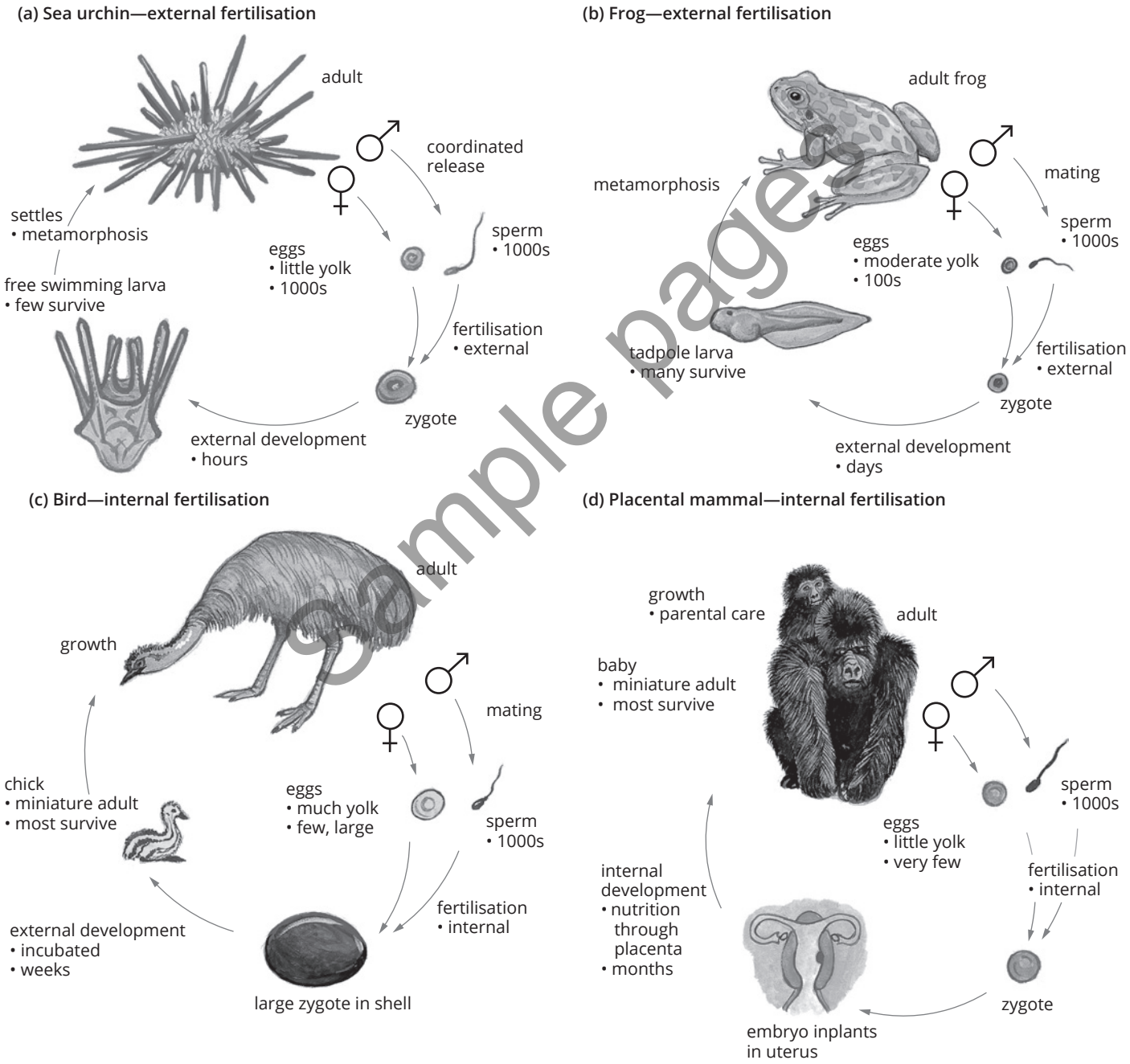

FIGURE 5.1 Life cycles of sexually reproducing organisms. External fertilisation occurs in many aquatic animals, such as (a) sea urchins and (b) frogs. Internal fertilisation is a common feature of terrestrial animals, including (c) birds and (d) mammals.

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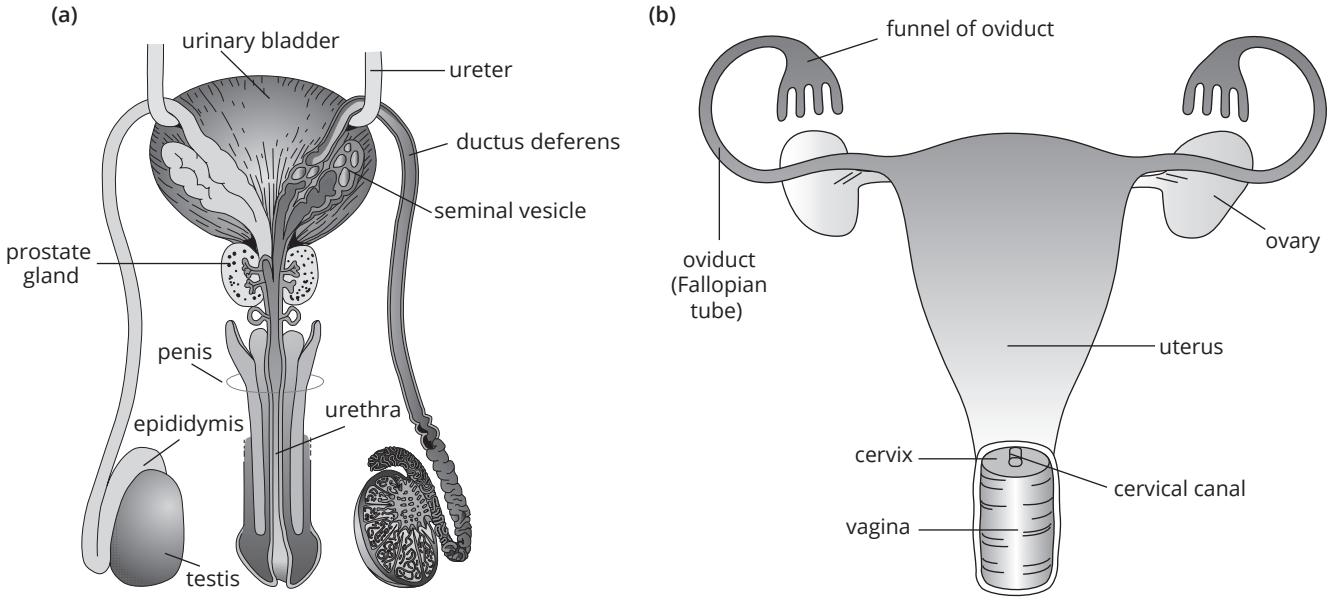

FIGURE 5.2 Reproductive systems in humans: (a) male and (b) female

Reproduction in plants can be sexual or asexual (Table 5.1). Sexual reproduction in plants requires the union of male and female gametes. As plants are sedentary, they use a range of strategies to ensure gametes come together. In flowering plants, this typically relies on insects such as bees in search of nectar; at the same time, they transfer pollen (containing the male sex cell) from the anther of one plant to the stigma of another of the same species. Pollen grains grow a tube downwards through the style towards the ovary, where fertilisation and seed (and fruit) development subsequently occur (Figure 5.3). Moths, birds and water also transfer pollen in some species of flowering plants. Grasses (which are also flowering plants) and conifers have pollen that is non-sticky and light, making wind pollination efficient.

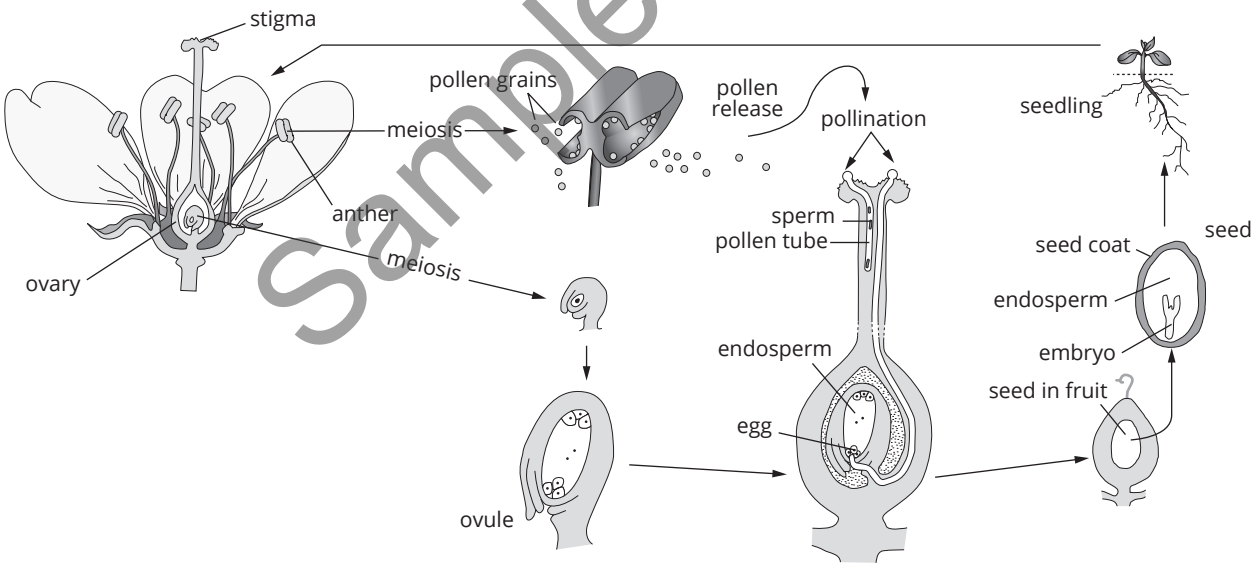

FIGURE 5.3 Male and female reproductive structures in flowering plants

Figure 5.4 provides a summary of sexual reproduction in a wide range of organisms.

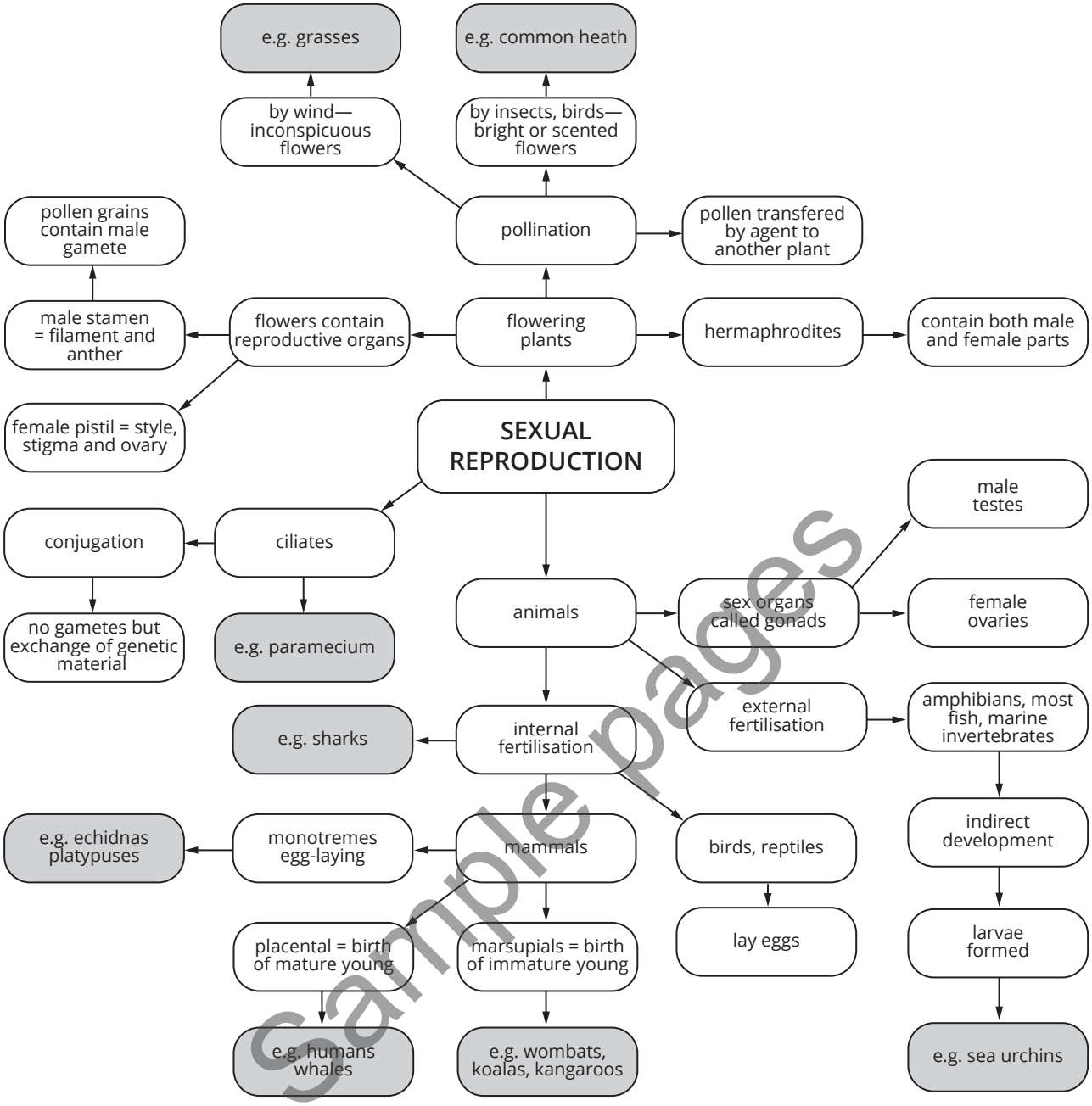

FIGURE 5.4 Sexual reproduction in animals, ciliates and flowering plants

PREGNANCY AND BIRTH IN MAMMALS

Sexual reproduction in mammals is characterised by internal fertilisation and development (with the rare exception of egg-laying monotremes—platypus and echidna). The internal environment provides optimal temperature and moisture conditions for the developing young, as well as ensuring nutrition and protection. Development of the new individual is gradual and continuous between conception and birth. However, various stages during development are recognised.

Development of the embryo and fetus in mammals

After fertilisation, rapid cell division proceeds in the development of the new individual.

The first stage of development is **cleavage**, which commences following activation of the egg by sperm penetration. Cleavage is a period of rapid cell proliferation during which the single-celled zygote is divided into many smaller cells by mitosis. By day 3–4 post-fertilisation, the zygote has become a ball of 16 undifferentiated cells, known as a **morula** (Figure 5.5). In the uterus, mitotic divisions continue, and at around five days after fertilisation the morula becomes a **blastocyst**. At approximately 8–9 days after fertilisation, the blastocyst implants into the uterine wall. The multicellular blastocyst consists of a single layer of surface cells and an inner cell mass that will later give rise to the embryo. The outer layer of cells sends out finger-like projections into a part of the uterine wall (endometrium), which develops into the **placenta** (in placental mammals only). The next stage is called

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the **gastrula**. At this stage some differentiation is evident, with clearly defined **endoderm**, **mesoderm** and **ectoderm** tissue (Figure 5.6). These three layers are destined to become specialised tissues of the various body systems.

The gastrula becomes an **embryo** at three weeks after fertilisation. The embryonic period of development is when the major organs of the body are formed from the three primary layers of the gastrula. In humans, this is completed about eight weeks after fertilisation (or 10 weeks after the last menstrual period). At the end of the embryonic stage, the developing organism has distinct features and is known as a **fetus** for the remainder of its development.

> Undifferentiated cells are called **stem cells**—they have the potential to develop into a multitude of cell types. As development continues from zygote to embryo, the potential of embryonic cells to differentiate into different kinds of cells becomes more limited.

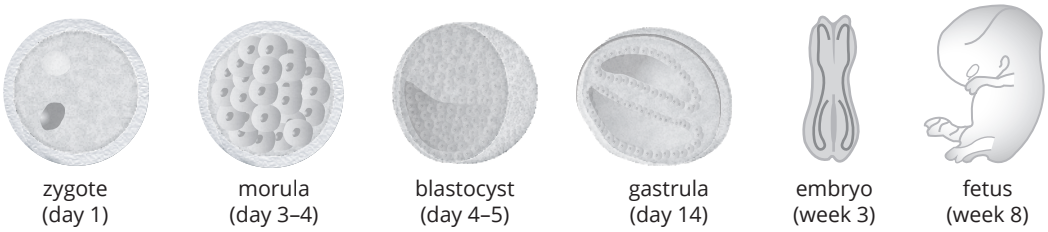

FIGURE 5.5 The development of a human zygote (fertilised egg) into a fetus

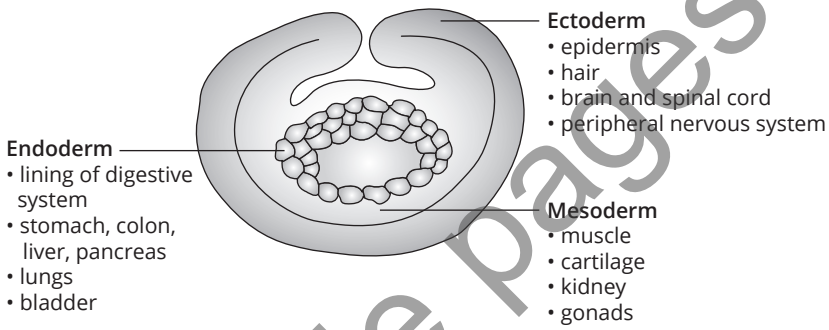

FIGURE 5.6 Gastrula with defined endoderm, mesoderm and ectoderm

MANIPULATION OF PLANT AND ANIMAL REPRODUCTION IN AGRICULTURE

The manipulation of plants and animals in breeding is as old as agriculture itself. **Selective breeding**, involving the selection of individuals with the most desirable features for reproduction, has resulted in the development of crops and stock that largely express those desirable features. For example, selective breeding has resulted in dairy cattle with large milk production capacity and watermelon with few and soft seeds.

The introduction of genetic technologies in recent times has fast-tracked the development of plants and animals in agriculture, ensuring the expression of even more specific and desirable characteristics. For example, **genetic modification** has been applied to create pest-resistant canola crops. **Cloning** and **gene editing** are also applied in agriculture to achieve desirable outcomes.

Cell replication

We know from the cell theory that all cells are derived from pre-existing cells. Prokaryotic cells replicate by a process known as **binary fission**, in which the cell and its contents are divided into two. Replication is more complex in eukaryotic cells, with nuclear division occurring during mitosis, followed by splitting the cell into two, a process called **cytokinesis**. In both cases the parent cell divides to form two identical daughter cells. When cells replicate to form identical daughter cells, the resulting cells are called **clones**. Cell replication is responsible for the production of new cells within an organism for the purposes of maintenance, growth and repair.

THE CELL CYCLE

Cells are in a constant state of activity that involves all the chemical reactions that make up the cell's metabolism, as well as growth and reproduction. Cell growth includes the replication of **DNA (deoxyribonucleic acid)** that is organised and divided for distribution to daughter cells during cell division. This cyclical activity of cells is called the **cell cycle** (Figure 5.7). Cell replication and passing on DNA to the next the generation is critical to the continuity of species.

The cell cycle has three main phases:

- interphase
- mitosis
- cytokinesis.

These phases always occur in this order, beginning with **interphase**. During interphase, the cell doubles its mass and duplicates its entire components. During mitosis the nucleus divides, and during cytokinesis the cytoplasm divides. A typical cell spends most of its life in interphase.

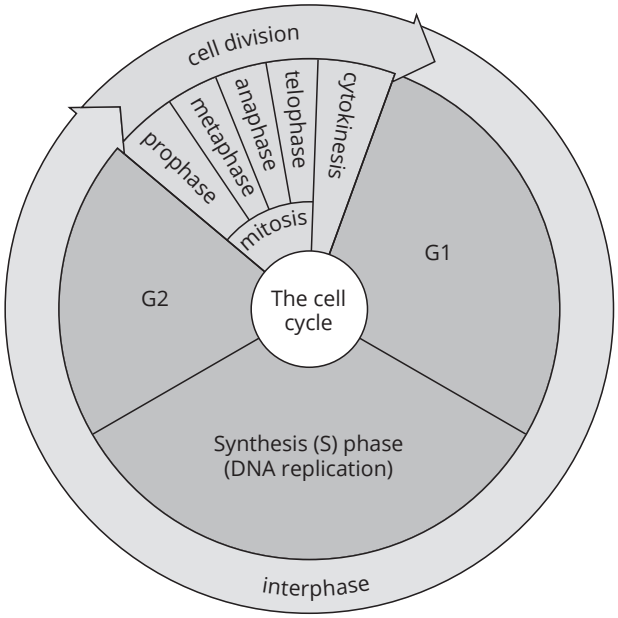

FIGURE 5.7 The cell cycle takes approximately 24 hours to complete in mammalian cells.

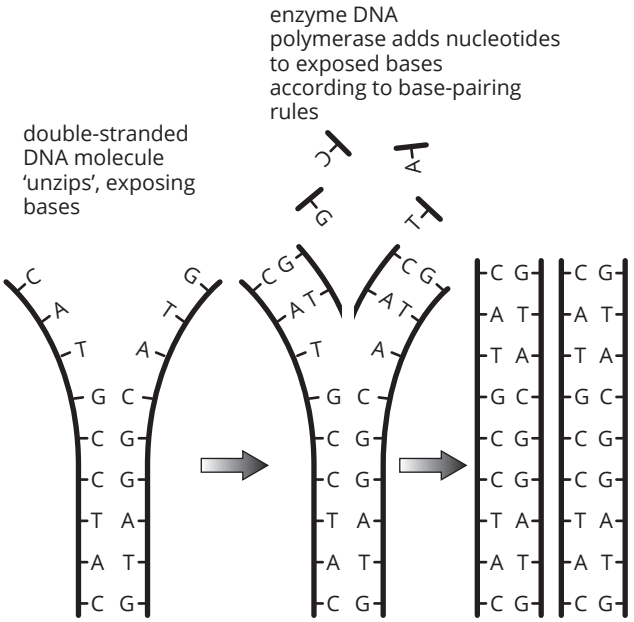

FIGURE 5.9 DNA replication—DNA polymerase adds nucleotides to build copies of the original DNA strands.

Interphase: DNA replication

The genetic information in the cells of eukaryotic organisms is packaged into threads of DNA called chromosomes (Figure 5.8). Chromosomes carry all of the information needed for cell structure and function. During cell replication, the chromosomes are organised so that the resulting daughter cells each receive precisely the same genetic material as the parent cell from which they are derived. Before this can occur, the genetic material must be duplicated. This copying process is called **DNA replication** (Figure 5.9) and occurs during interphase. During DNA replication, the two strands of DNA that make up the double helix 'unzip' or separate. The enzyme **DNA polymerase** then moves along the exposed **non-coding strands** (also known as template strands) adding **nucleotides** according to **complementary base-pairing** rules to build the new strands.

DNA replication is described as being semi-conservative because the parental strand is conserved, or retained, in the new molecule.

> **i** During interphase, the cell's DNA is replicated. DNA replication ensures daughter cells are produced that contain the appropriate type and number of chromosomes.

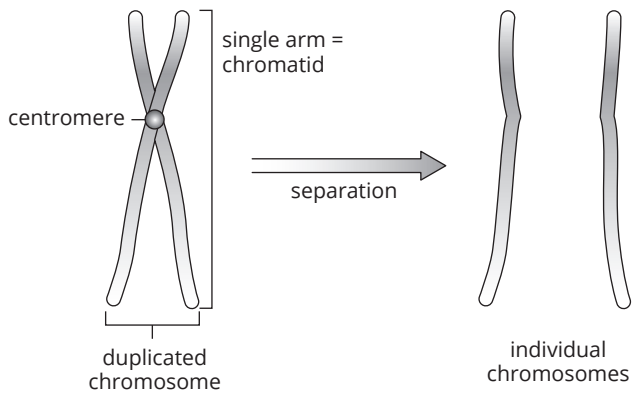

FIGURE 5.8 Structure of a chromosome

> **i** In DNA, adenine (A) always bonds with thymine (T) and guanine (G) always bonds with cytosine (C). This is called complementary base-pairing.

Mitosis

Mitosis only occurs in eukaryotic cells. It is the process by which the DNA replicated during interphase is divided into two new nuclei. Mitosis is divided into four phases: **prophase**, **metaphase**, **anaphase** and **telophase** (Figure 5.10). Cytokinesis is a separate process to mitosis and occurs after telophase, completing cell replication.

- Prophase: Chromosomes shorten and thicken, and become visible under the light microscope. The nuclear envelope dissolves and a structure called the spindle starts to form. The spindle consists of fibres that radiate across the cell from centrioles at each pole.
- Metaphase: Chromosomes line up along the equator of the cell. Each chromosome attaches to a spindle fibre by its centromere.
- Anaphase: The spindle fibres contract, causing the centromeres to split, pulling the sister chromatids towards opposite poles. (Remember, each chromosome was replicated during interphase and the two copies of each have remained joined until now.)
- Telophase: New nuclear membranes form around each of the two new groups of chromosomes.

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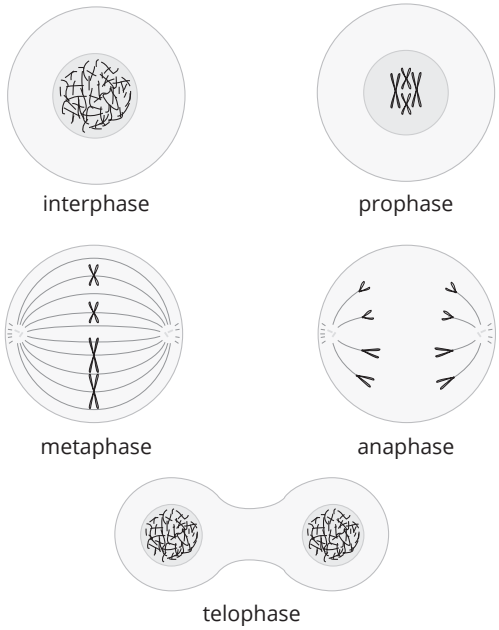

FIGURE 5.10 During mitosis, DNA replicated during interphase is divided into two new nuclei.

Meiosis

Gamete production involves a special type of nuclear division called meiosis. Unlike mitosis, meiosis produces daughter cells that are different from each other and different from the parent cell. Meiosis is required for sexual reproduction. It produces four daughter cells (gametes) that are genetically unique, creating genetic variation. Meiosis occurs only in eukaryotes and only to form the gametes.

Unlike **somatic cells**, gametes are haploid, containing only one set of chromosomes—half the full complement. Like mitosis, meiosis is divided into the phases prophase, metaphase, anaphase and telophase. However, in meiosis each of these phases occurs in two sequential rounds of division, called meiosis I and meiosis II (Figure 5.11). Meiosis I is called a **reduction division** because it reduces the number of chromosomes in the daughter cells (gametes) to half ($1n$) of that in somatic cells ($2n$). To achieve haploid daughter cells, the phases of meiosis are repeated. In metaphase I, **homologous chromosomes** (matching pairs) align together at the equator, before members of each pair move to their respective poles in anaphase I. In metaphase II, the chromosomes again align at the equator, but this time duplicated chromosomes cleave at the centromere before single chromosomes move to the poles in anaphase II.

Diploidy (having two sets of chromosomes) is restored at fertilisation when the zygote is formed.

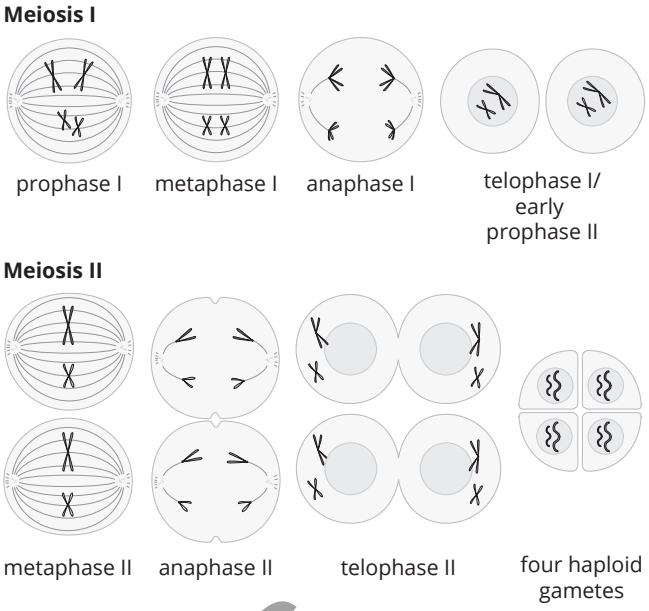

FIGURE 5.11 Meiosis produces four genetically unique gametes.

Genetic variation

Genetic variation in meiosis occurs as a result of independent assortment and crossing over.

Independent assortment: When homologous chromosomes line up together and then segregate to different poles, they do so independently of other homologous pairs.

Homologous chromosomes (Figure 5.12) are pairs of chromosomes that contain similar genetic information. That is, they contain the same **genes** (DNA sequences that code for polypeptides) but they may contain alternative forms of those genes. Alternative forms of a gene are called **alleles**. For example, earlobe shape in humans has different forms—free or attached. By convention, letters of the alphabet are assigned to represent the different alleles: '*F*' can be used to denote the allele for free lobes, while '*f*' can denote the allele for attached lobes. Similarly, the alleles *T* and *t* can be used to denote the genetic expression of 'tongue rolling' and 'non-tongue rolling'.

Crossing over: During early prophase I, when homologous chromosomes pair, they may touch at points called **chiasma** (plural chiasmata). At chiasmata, the homologous pairs may exchange chromosome segments. This results in **recombination** and is responsible for the genetic variation formed in gametes.

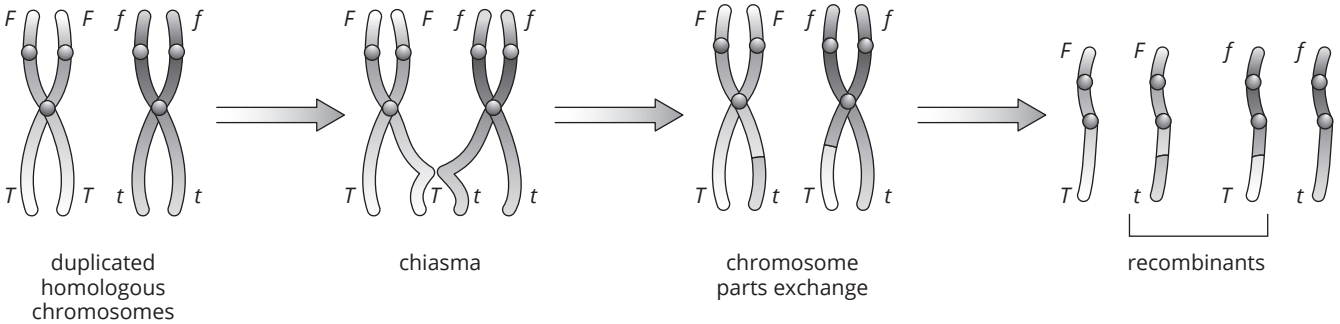

FIGURE 5.12 Homologous chromosomes are pairs of chromosomes that have the same genes, but may carry different alleles for those genes.

Cytokinesis

During cytokinesis, the cytoplasm divides and the two new daughter cells are formed.

Cytokinesis in animal cells occurs in a different way to cytokinesis in plant and fungi cells. In animal cells the cell membrane moves inwards, pinching the two daughter cells apart. In contrast, plant and fungi cells lay down a new cell membrane and cell wall between the two daughter nuclei to separate the daughter cells. Components of the new plant cell wall, called the cell plate, are initially deposited in the centre of the cell. The growth of the cell plate extends outwards until the two daughter cells are completely separated.

DNA and polypeptide synthesis

Common to all living things on Earth is the presence of the genetic material, DNA and **RNA (ribonucleic acid)**. The structure and function of these molecules are universal across all life forms.

DNA IN EUKARYOTES AND PROKARYOTES

While DNA is the genetic information common to all organisms, its organisation is different in prokaryotes and eukaryotes (Table 5.2).

Both DNA and RNA are made up of nitrogenous bases and a sugar–phosphate backbone. RNA is single-stranded and relatively short. DNA has a double-stranded helix (spiral) structure with complementary pairing of its nitrogenous bases holding the double strands together like rungs on a ladder (Figure 5.13).

TABLE 5.2 Differences in the structure and organisation of DNA in prokaryotic and eukaryotic cells

Prokaryotes	Eukaryotes
• DNA contained in a single chromosome • no membrane-bound nucleus • reproduction involves simple duplication and separation of chromosome	• DNA contained in paired chromosomes • DNA confined to membrane-bound nucleus • reproduction involves DNA replication, followed by complex steps of chromosome movements to ensure correct number of chromosomes in daughter cells

The nucleotides are named according to the nitrogenous base each includes. The bases in DNA occur in complementary pairs:

- **adenine (A)** pairs with **thymine (T)**
- **cytosine (C)** pairs with **guanine (G)**.

In RNA, **uracil (U)** replaces thymine (T) and pairs with adenine (A).

Each nucleotide is made up of a nitrogenous base (A, T, G, C or U), a pentose sugar (deoxyribose in DNA and ribose in RNA) and a phosphate group, joined by covalent bonds (Figure 5.14).

Nucleotides are chemically bonded to form polymers called **nucleic acids** (i.e. deoxyribonucleic acid and ribonucleic acid) (Figure 5.15). Nucleic acids are essentially information molecules that contain the coded instructions for **polypeptide** synthesis. Once formed, chains of polypeptides combine to form **proteins**. Proteins are biological molecules that carry out all the functions essential to life. The sequence of nucleotides in DNA is significant because of its role in protein production. A specific DNA sequence that codes for a particular polypeptide is called a gene. The **genome** is the total complement of all of the genes in an individual organism. An organism's genome is intrinsically linked to its **proteome**, which is the full complement of proteins in an individual.

The complementary strands of the DNA molecule are described as **antiparallel**, because one runs 5′ → 3′ while the other runs 3′ → 5′. Figure 5.16 shows a simplified representation of the molecule illustrating this antiparallel arrangement.

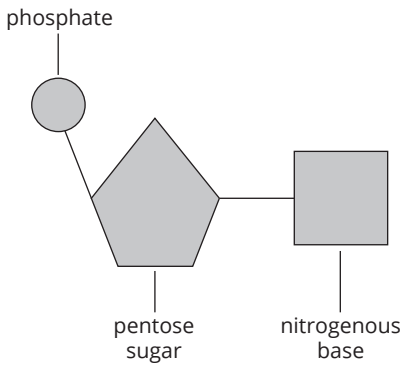

FIGURE 5.14 Nucleotide

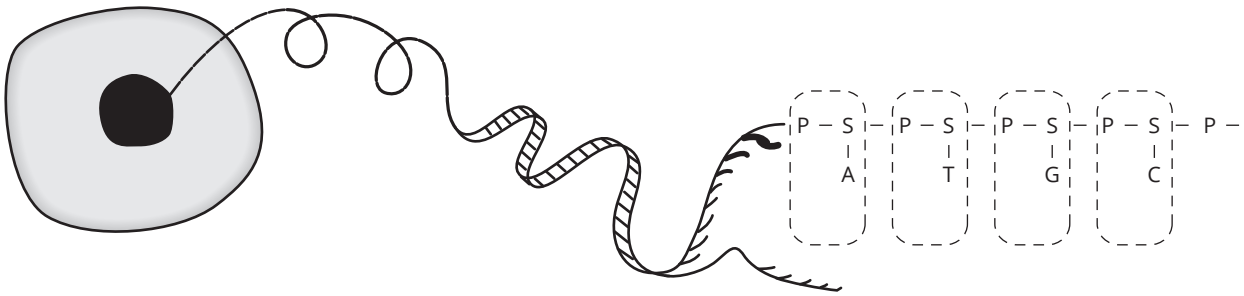

FIGURE 5.13 The DNA in the nucleus of cells unravels to reveal the double helix structure. 'P' represents a phosphate group, 'S' represents a sugar and 'A', 'T', 'G' and 'C' represent nitrogenous bases. Together these three components make up a nucleotide.

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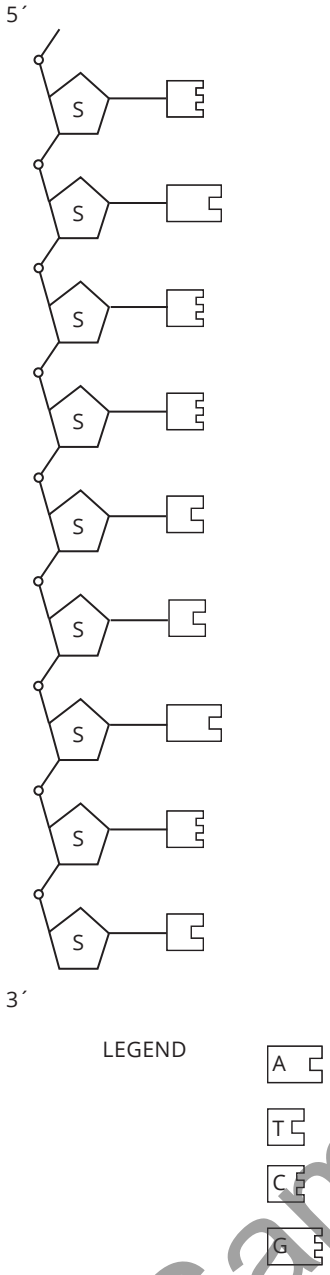

FIGURE 5.15 Nucleotide polymer—single-stranded DNA

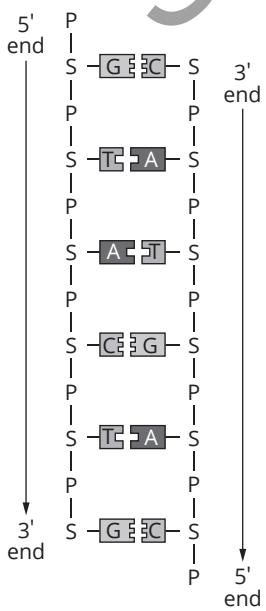

FIGURE 5.16 Nucleotides arranged in complementary pairs held together by hydrogen bonding

> In models of nucleic acids, the nucleotides are simply referred to as bases and are identified by the base letter A, T, C, G or U. This is because the sugar and phosphate units in all nucleotides are identical. The nitrogenous base is the unit that changes.

The features of DNA are summarised in Table 5.3 and illustrated in Figure 5.17.

TABLE 5.3 Features of nitrogenous bases

	Feature	Examples
purines	double-ring structure	adenine, guanine
pyrimidines	single-ring structure	cytosine, thymine

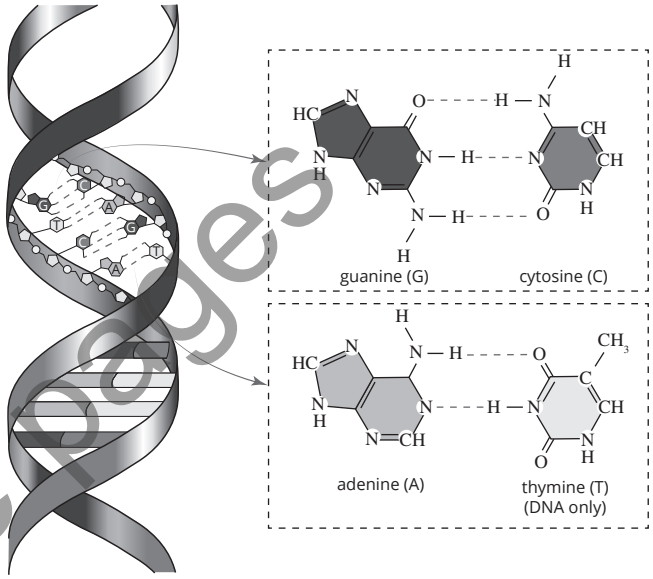

FIGURE 5.17 The helical structure of DNA. Two complementary strands form a double helix joined by base pairs guanine (G) and cytosine (C), and adenine (A) and thymine (T). Guanine (G) and adenine (A) have a double-ring structure and are known as purines, while cytosine (C) and thymine (T) have a single-ring structure and are known as pyrimidines.

There are three main forms of RNA. Each has a different role in polypeptide synthesis:

- **messenger RNA (mRNA)**: copy of the DNA template strand that takes instructions to the **ribosomes** in the cytoplasm
- **transfer RNA (tRNA)**: the molecule that brings **amino acids** to ribosomes during protein synthesis
- **ribosomal RNA (rRNA)**: is synthesised in the nucleolus and forms part of the structure of ribosomes.

POLYPEPTIDE SYNTHESIS

The sequence of nucleotide bases in genes represents the coded instructions for constructing polypeptides, the building blocks of proteins. The completed protein is the form in which the gene is expressed. Polypeptide production involves two key steps—transcription and translation.

Transcription

Transcription is the process in which the DNA template strand is copied (transcribed) to form a messenger RNA strand.

Characteristics of transcription:

- occurs in the nucleus
- mRNA is single-stranded and contains the nucleotide base uracil (U) instead of thymine (T)
- transcription begins at the **promoter**, a section of DNA that identifies the beginning of the gene
- **exons** are the coding regions of genes
- **introns** are the non-coding regions of genes
- both exons and introns are transcribed, forming a copy of the gene called pre-mRNA
- introns are subsequently cut out of the pre-mRNA, forming the final mRNA product.

The main steps in the process of transcription are shown in Figure 5.18. At the conclusion of transcription, the mRNA molecule leaves the nucleus through nuclear pores and moves to the ribosomes, where translation occurs.

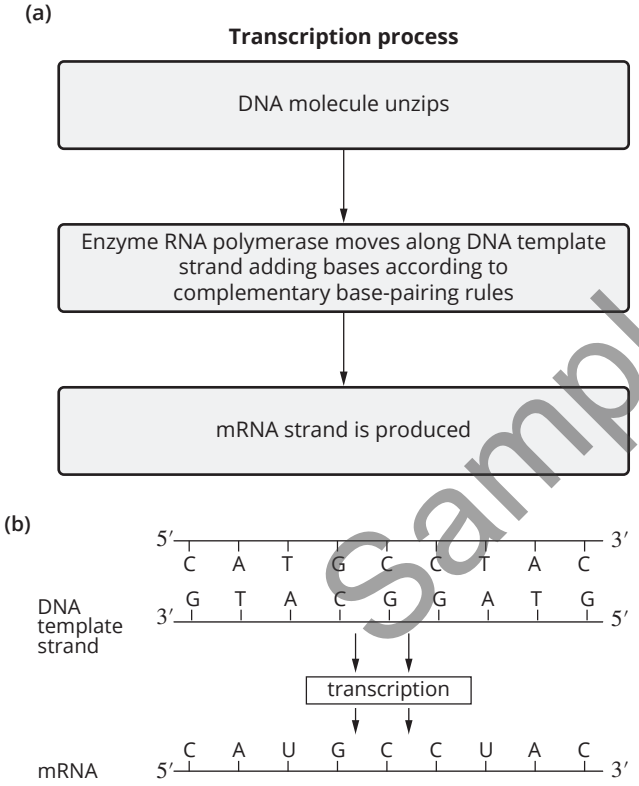

FIGURE 5.18 (a) Key steps in the process of transcription, where DNA is transcribed into messenger RNA (mRNA). (b) The single-stranded mRNA molecule contains uracil (U) instead of thymine (T).

Translation

Translation is the process in which the sequence of bases in mRNA is translated into an amino acid sequence (polypeptide).

Characteristics of translation:

- occurs in the cytoplasm at the ribosomes
- sequence of bases in mRNA is translated into amino acid sequence (polypeptide)
- each group of three bases in the mRNA codes for a particular amino acid. The groups of three bases in mRNA are called **codons**. The sequence of these codons is known as the **genetic code** (Table 5.4)
- a group of three bases complementary to the codon is called an **anticodon**. A tRNA molecule carries an anticodon and an amino acid (Figure 5.1.9)
- the amino acids are joined by peptide bonds to form a polypeptide chain.

The main steps in the process of translation are shown in Figure 5.19. Translation stops when a **stop codon** is reached. The polypeptide chain is now complete.

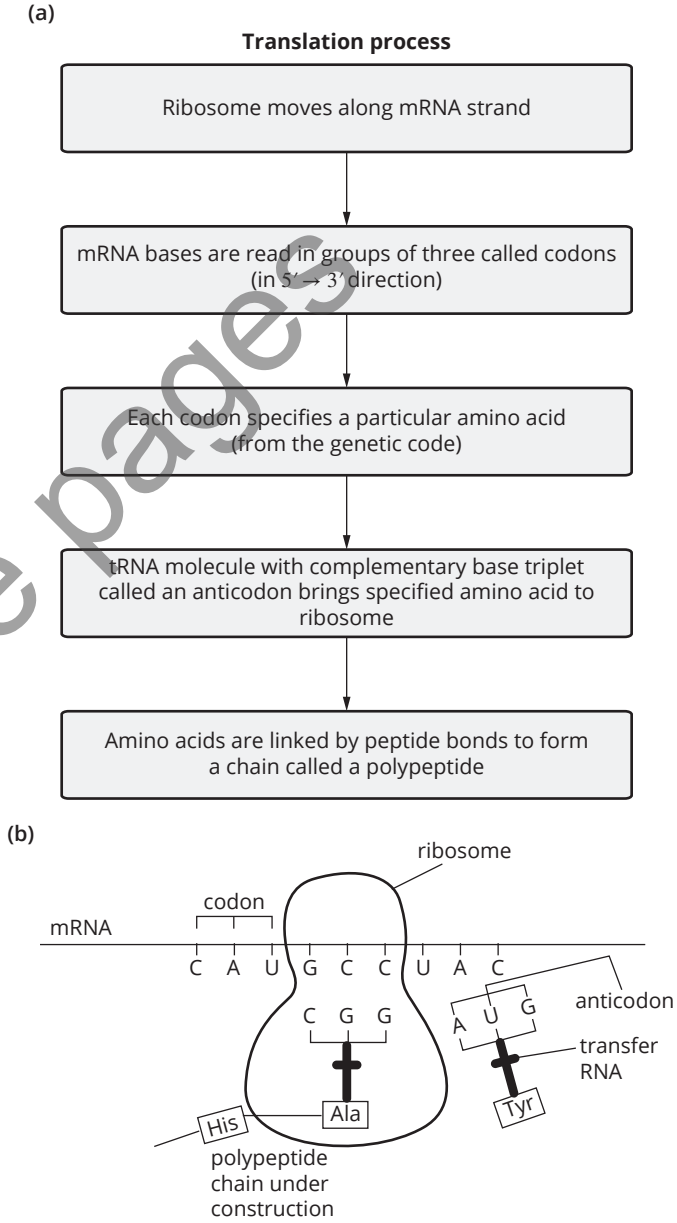

FIGURE 5.19 (a) Key steps in the process of translation, where mRNA is translated into amino acids to build a polypeptide chain. (b) Complementary anticodons brought by transfer RNA are joined in the ribosome to form a polypeptide chain.

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The genetic code is a framework that uses RNA codons to identify amino acids and their sequence in growing polypeptide chains during protein synthesis. Table 5.4 lists the 20 amino acids used in proteins and the codons that code for each amino acid. To use this table, select the first base of the codon from the first column, read across the row for the second base, and then find the third base using the last column. Abbreviations for the amino acids are listed in Table 5.5, while Figure 5.20 summarises both transcription and translation.

TABLE 5.4 The genetic code for the 20 amino acids and stop codons

First position (5′ end)	Second position				Third position (3′ end)
	U	**C**	**A**	**G**	
U	Phe Phe Leu Leu	Ser Ser Ser Ser	Tyr Tyr STOP STOP	Cys Cys STOP Trp	U C A G
C	Leu Leu Leu Leu	Pro Pro Pro Pro	His His Gln Gln	Arg Arg Arg Arg	U C A G
A	Ile Ile Ile Met (START)	Thr Thr Thr Thr	Asn Asn Lys Lys	Ser Ser Arg Arg	U C A G
G	Val Val Val Val	Ala Ala Ala Ala	Asp Asp Glu Glu	Gly Gly Gly Gly	U C A G

TABLE 5.5 Amino acid abbreviations

Ala	alanine
Arg	arginine
Asn	asparagine
Asp	aspartic acid
Cys	cysteine
Gln	glutamine
Glu	glutamic acid
Gly	glycine
His	histidine
Ile	isoleucine
Leu	leucine
Lys	lysine
Met	methionine
Phe	phenylalanine
Pro	proline
Ser	serine
Thr	threonine
Trp	tryptophan
Tyr	tyrosine
Val	valine

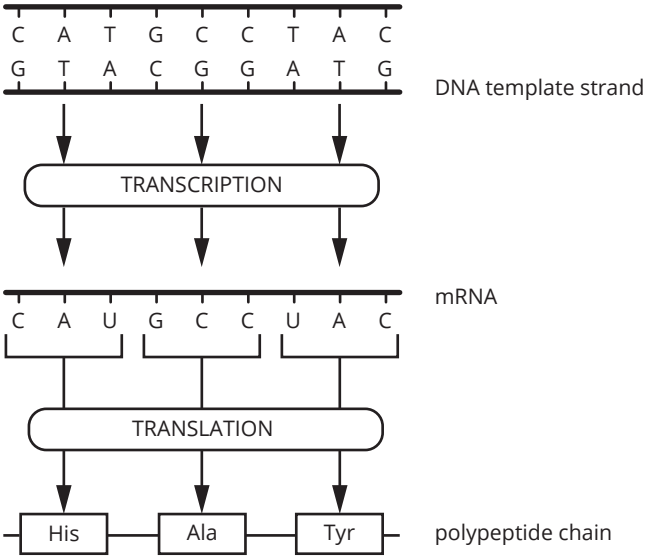

FIGURE 5.20 Summary of transcription and translation

STRUCTURE AND FUNCTION OF PROTEINS

Once polypeptide production is complete, the polypeptide chain folds to form a complex three-dimensional structure and is then referred to as a protein. Table 5.6 describes the four levels of protein structure.

TABLE 5.6 Levels of protein structure

1	polypeptide formation	primary protein structure
2	polypeptide becomes coiled or pleated	secondary structure
3	coiled polypeptide folds into 3-dimensional form	tertiary structure
4	two or more 3-dimensional polypeptide molecules bonded together	quaternary structure

Proteins are key components of cells. There are many different kinds of proteins, each with a different function, and all are vital to the normal functioning of the organism. Some examples are given in Table 5.7.

As you learnt before, the proteome is the total complement of all the proteins in an individual organism. An organism's proteome is determined by the DNA sequence of its genome. **Proteomics** (the study of proteins, including their structure and function) is an expanding field of biology that has enormous potential for improving our understanding of how organisms function, and of diseases and their treatment and management; for the development of pharmaceuticals; and for shedding light on evolutionary relationships.

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TABLE 5.7 Roles of proteins in living organisms

Role	Examples
catalysts	enzymes
structural	cell membranes muscle tissue collagen (skin) cytoskeleton cilia
communication	hormones neurotransmitters
transport	channels in cell membrane
carrier molecules	haemoglobin

GENE STRUCTURE AND REGULATION

Cells in the body have specialised structures and functions, yet they all contain the same genetic information. This occurs as a result of different genes being switched 'on' or 'off' in particular cells. For example, beta cells in the pancreas have genes for insulin production switched on (expressed), but genes related to haemoglobin production are switched off. This is called gene regulation. Gene regulation contributes to the conservation of energy and resources in cells.

Genes are typically structured so that the transcription of the coding region is carefully regulated. A **regulatory gene** (which controls transcription) is positioned before the **structural gene** (which codes for the protein). The regulatory gene is composed of a promoter region and an operator region, both upstream of the structural gene. The promoter region regulates when transcription should begin; the operator effectively switches the gene on or off by allowing transcription to begin or cease. The structural gene contains introns and exons. Once the gene is switched on, transcription proceeds. Transcription is halted by a DNA triplet that codes for a stop codon downstream of the structural gene. The *lac* **operon** (lactose operon) model in bacteria serves as a classic example of our understanding of gene function (Figure 5.21). An operon is a group of genes with a regulatory role in protein production. The *lac* operon is a group of bacterial genes responsible for the production of a lactose-digesting enzyme.

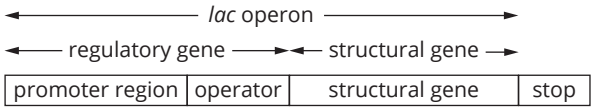

FIGURE 5.21 The *lac* operon

The expression of genes can be influenced by various environmental factors, such as temperature, light and pH.

Because all cells need to engage in life-sustaining functions, such as cellular respiration, the genes that control these processes are switched on in all cells. Such genes are referred to as 'housekeeping genes'.

GENES AND DEVELOPMENT

A group of genes called homeotic genes switch genes on and off at appropriate times during the development of organisms. For example, the genes controlling the production of fetal haemoglobin in mammals are switched on in utero, but are switched off at birth, while the genes controlling production of adult haemoglobin are switched on.

Cells are also programmed to die at different stages of development or after a period of activity. Programmed cell death is called **apoptosis**.

Genetic variation

Sexual reproduction results in offspring with a set of unique characteristics that are inherited from their parents. These characteristics vary among individual organisms.

FORMATION OF GENETIC VARIATION

Genetic variation in populations is generated as a result of mutation and sexual reproduction.

- A mutation is a change in the DNA of an individual. A **single nucleotide polymorphism (SNP)** refers to a change in a single nucleotide in a section of DNA and is responsible for alternative alleles. Mutation is the raw material for evolutionary change. Mutations introduce new alleles into populations.
- Sexual reproduction is the production of genetically unique gametes in meiosis resulting from independent assortment of chromosomes and genetic recombination in crossing over. The random union of gametes in fertilisation further mixes the genetic material, creating genetic variation.

The characteristics of **genetics** include the following.

- An organism's **genotype** is the combination of alleles that make up its genetic information.
- The **phenotype** of an organism is the observable expression of its genotype.
- An organism's phenotype is influenced by both its genotype and environmental factors.

  phenotype = genotype + environment
- A gene is a sequence of DNA that is the unit of heredity.
- An allele is an alternative form of a gene.

  The chromosomes of a homologous pair may carry the same or different alleles for a given gene.
- **Homozygous** describes an individual that carries the same alleles for a particular gene on both chromosomes of a homologous pair.
- **Heterozygous** describes an individual that carries alternative alleles for a given gene.

Example: In humans, 'handedness' is a genetic trait controlled by a gene with two alternative alleles. Right-handedness is dominant to left-handedness.

Notation: *R*: right-handed, *r*: left-handed

RR: homozygous right-handed individual

Rr: heterozygous right-handed individual

rr: homozygous left-handed individual

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Phenotypic traits can be described as dominant or recessive. **Dominant phenotypes** appear in heterozygotes and homozygotes (e.g. *Bb* and *BB*). **Recessive phenotypes** appear only in homozygotes, because two alleles are required for their expression (e.g. *bb*).

Pedigree analysis allows the patterns of inheritance of particular traits to be tracked from one generation to the next in families. The information provided in the pedigree legend, together with appropriate allelic notation, allows genotypes to be assigned to at least some individuals in the pedigree. Such an approach is useful in determining the mode of inheritance of a particular characteristic.

The pedigree in Figure 5.22 illustrates that right-handedness is inherited as an **autosomal dominant trait**. Its inheritance pattern is not linked to biological sex (so the gene is carried on an **autosome**) and the trait appears in individuals who are heterozygous (making it fit the definition of dominance).

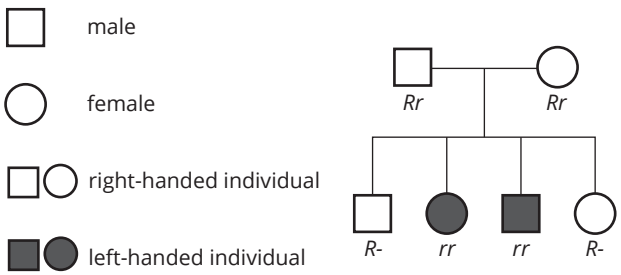

FIGURE 5.22 Right-handedness is inherited as an autosomal dominant trait. '*R*-' represents an individual that may be homozyous (*RR*) or heterozygous (*Rr*) for the trait.

Some traits do not show simple dominance or recessiveness. There are instances in which both alleles are expressed to varying degrees in the phenotype. This is called **co-dominance**. The ABO blood grouping system is an example—a single gene features multiple alleles, I^A, I^B and *i*. Individuals carrying alleles for both A antigens (a kind of protein) and B antigens express both in the phenotype and have the blood type AB, as shown in Table 5.8.

TABLE 5.8 Blood type genotypes and phenotypes

Genotype	Phenotype (blood type)
I^AI^A, I^Ai	A
I^BI^B, I^Bi	B
I^AI^B	AB
ii	O

Incomplete dominance describes a different kind of inheritance pattern in which two phenotypes are partially expressed. Flower colour in snapdragons is an example. When red-flowered snapdragons (homozygous) are crossed with white-flowered snapdragons (homozygous), they produce pink-flowered offspring. In this instance, both the alleles for red colour and white colour are partially expressed.

Genetic explanation: *R*: red, *W*: white
Parents: *RR* (red) × *WW* (white)
Offspring: all *RW* (pink)

When the pink-flowered snapdragons are crossed, they produce three different phenotypes. A **Punnett square** can be used to show this:

gametes	*R*	*W*
R	*RR*	*RW*
W	*RW*	*WW*

¼ red : ¾ pink : ¼ white

This is a 1 : 2 : 1 phenotypic ratio and is typical of the second generation in a cross involving traits that are co-dominant.

Continuous and discontinuous variation

Many characteristics are under the control of more than one gene. This is called **polygenic inheritance**.

Continuous variation: Traits are controlled by **polygenes** and characterised by a range of phenotypes; their distribution can be represented graphically by a typical bell curve. Examples include the inheritance of height, eye colour and skin colour.

Discontinuous variation: Traits are typically controlled by a single gene, usually with two allelic forms and characterised by distinct phenotypes.

For example:

- handedness—individuals are either right-handed or left-handed
- flower colour in snapdragons—two alleles result in three distinct phenotypes
- ABO blood grouping—three different alleles result in four distinct phenotypes.

Figure 5.23 shows examples of continuous and discontinuous variation.

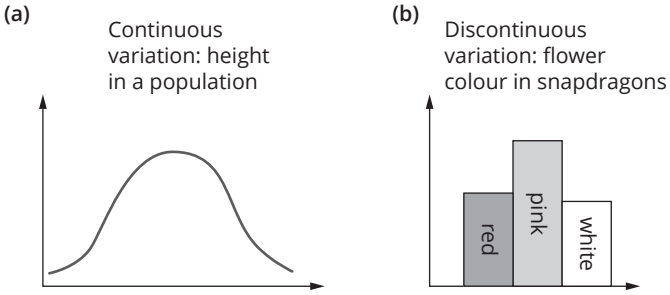

FIGURE 5.23 (a) Continuous variation (b) discontinuous variation

Environmental impact on phenotype

Chocolate-point Siamese cats demonstrate the impact of the environment on phenotype. This breed of cat carries genetic information that results in the production of dark pigment in the extremities (i.e. the tips of the ears, snout, tail and paws). In cool climates, the chocolate points are evident. However, when Siamese cats are raised in hot climates, the fur that grows at the extremities lacks the dark pigment.

Other examples of the interplay between phenotype and environment can be observed in: individuals diagnosed with phenylketonuria whose health is managed by diet, fur colour in Himalayan rabbits, and flower colour variation with soil pH.

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INHERITANCE OF GENETIC VARIATION

Gregor Mendel (1822–1884) is credited with laying the foundations of our modern understanding of genetics. He carried out breeding experiments (crosses) with garden peas to understand and interpret the patterns of inheritance that he observed.

Monohybrid crosses

A **monohybrid cross** is a cross that involves a single gene locus.

Example: Inheritance of colour in pea seeds

Yellow pea colour is dominant to green pea colour.

Notation:

Y: yellow (dominant)

y: green (recessive)

F_1: first filial generation (offspring)

F_2: second generation

Two pure-breeding plants are crossed:

Parents: *YY* (yellow) × *yy* (green)

F_1: *Yy* (all yellow)

A Punnett square is used to calculate the ratio of genotypes and phenotypes in the F_2 generation:

gametes	*Y*	*y*
Y	*YY*	*Yy*
y	*Yy*	*yy*

¾ of the offspring will be yellow; ¼ of the offspring will be green.

This is a 3 : 1 phenotypic ratio and is typical of a cross between heterozygotes in a monohybrid cross where the gene under investigation has two allelic forms.

Dihybrid crosses

A **dihybrid cross** is a cross that involves two gene loci.

Example: Inheritance of colour and shape in pea seeds

Round pea shape is dominant to wrinkled pea shape.

Yellow pea colour is dominant to green pea colour.

Notation:

R: round (dominant)

r: wrinkled (recessive)

Y: yellow (dominant)

y: green (recessive)

When pure-breeding, round, yellow pea-producing plants are crossed with pure-breeding, wrinkled, green pea-producing plants, all the offspring produce round, yellow peas.

Parents: *RRYY* (round, yellow) × *rryy* (wrinkled, green)

F_1: *RrYy* (all round, yellow)

Punnett square to calculate the F_2 ratio:

gametes	*RY*	*Ry*	*rY*	*ry*
RY	*RRYY*	*RRYy*	*RrYY*	*RrYy*
Ry	*RRYy*	*RRyy*	*RrYy*	*Rryy*
rY	*RrYY*	*RrYy*	*rrYY*	*rrYy*
ry	*RrYy*	*Rryy*	*rrYy*	*rryy*

This reveals a phenotypic ratio of 9/16 round, yellow: 3/16 round, green: 3/16 wrinkled, yellow: 1/16 wrinkled, green.

A 9 : 3 : 3 : 1 phenotypic ratio is typical of a dihybrid cross between heterozygotes where the traits under investigation are controlled by genes with two alleles.

Test crosses

A **test cross** is a cross between an individual displaying the dominant phenotype and a homozygous recessive individual. Test crosses are carried out to determine whether the individual with the dominant phenotype is homozygous or heterozygous. If offspring displaying the recessive phenotype are produced, the parent in question must be heterozygous.

Monohybrid test crosses with a heterozygous parent reveal a phenotypic ratio of 1 : 1, as shown below:

gametes	*R*	*r*
r	*Rr*	*rr*

If all offspring show the dominant phenotype, this suggests the parent in question is homozygous. The larger the number of offspring, the more reliable the results of the test cross.

gametes	*R*	*R*
r	*Rr*	*Rr*
r	*Rr*	*Rr*

Dihybrid test crosses reveal a phenotypic ratio of 1 : 1 : 1 : 1, as shown below:

gametes	*RY*	*Ry*	*rY*	*ry*
ry	*RrYy*	*Rryy*	*rrYy*	*rryy*

Chromosomes and sex determination

- **Sex chromosomes** are chromosomes that are involved in sex determination (Figure 5.24). In humans, these are the X and Y chromosomes: XX = female; XY = male.
- Autosomes are chromosomes that are not involved in sex determination.
- Diploid cells in humans contain 46 chromosomes, arranged in 23 pairs.
- There are 22 homologous pairs (the autosomes) and one pair of sex chromosomes.
- Females are **homogametic**; that is, the sex chromosomes are homologous.
- Males are **heterogametic**; that is, the sex chromosomes are not a homologous pair.
- Unlike humans, female birds are heterogametic (ZW) and males are homogametic (ZZ).

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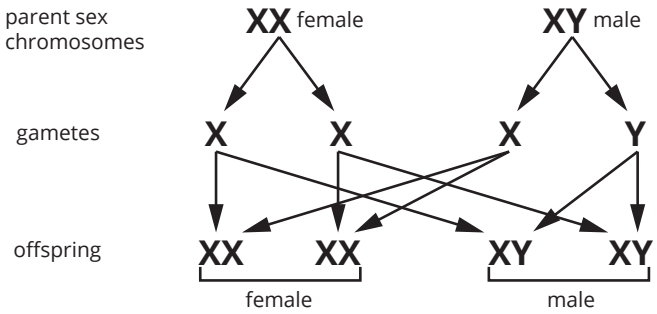

FIGURE 5.24 Sex determination in humans

Gene linkage

Linkage refers to the tendency for genes located on the same chromosome to be inherited together. Genes that are inherited together form a linkage group.

Example: Consider linked genes *P* and *Q*, represented by alleles *P*, *p* and *Q*, *q* respectively.

Notation: *PQ* denotes that alleles *P* and *Q* are located on one chromosome, and *pq* denotes alleles *p* and *q* are located on the other.

During meiosis, two kinds of gametes are expected to be produced: *PQ* and *pq*. These are called **parental types** (also called parental gametes).

The further apart the gene loci are located on the chromosome, the more likely that crossing over will occur between them. Crossing over will rearrange the genetic material, resulting in new combinations of alleles. Such gametes are called recombinants. Crossing over increases variation in the kinds of gametes produced.

Genes are considered to be linked if less than 50% of the gametes produced are recombinant. When a dihybrid test cross deviates from the expected 1 : 1 : 1 : 1 ratio, it indicates the gene loci in question are linked.

Sex linkage

Genes located on the sex chromosomes are said to be **sex-linked**. This is because the phenotype is linked to the biological sex of the individual. Tracking the pattern of inheritance of characteristics in pedigree analysis is a useful method of establishing whether or not genes are sex-linked.

Colour blindness and haemophilia in humans are sex-linked characteristics; genes controlling both characteristics are located on the X-chromosome (Figure 5.25). Traits inherited from genes on the X chromosome are called **X-linked**, while traits inherited from genes on the Y-chromosome are called **Y-linked**.

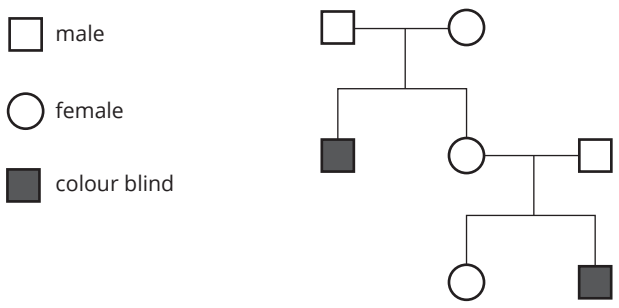

FIGURE 5.25 Colour blindness is inherited as a sex-linked, recessive trait.

X-linked dominant characteristics: An affected male will pass the trait to all his daughters but not his sons.

X-linked recessive characteristics: An affected female will pass the trait to all her sons.

Y-linked characteristics: Pattern of inheritance is always father to son.

GENETIC VARIATION IN POPULATIONS

Populations show variation in the form of traits (phenotypes). A trait that has many forms is called **polymorphic**. The **gene pool** is the genetic composition of a population. It includes the sum of all the alleles for different genes present in a population.

Selection pressures in the environment act on the phenotypes of individuals. As a result, the frequency of alleles in the population may change.

Allele frequency is the proportion of a particular allele in a population.

- Allele frequencies range between 0 and 1.
- An allele frequency of 0 means that no individuals in the population have the allele.
- An allele frequency of 1 means that all individuals in the population have the allele and are homozygous for that allele.
- The sum of the alternative alleles for a given gene adds up to 1.

Formula: p = frequency of dominant allele

q = frequency of recessive allele

$p + q = 1$

Example: In a population of 10 kookaburras, there are nine individuals with normal feather pigmentation and one albino. Of the nine individuals with normal colouring, six are homozygous dominant and three are heterozygous. The albino individual is homozygous recessive.

N = allele for pigmented feathers

n = allele for no pigment (albino)

Homozygous dominant: NN

Heterozygous = Nn

Homozygous recessive = nn

10 kookaburras = 20 alleles for feather pigment in the population (each individual carries two alleles)

Six homozygous pigmented birds ($6 \times 2N = 12N$) + three heterozygotes ($3N$) = $15N$ alleles. Therefore:

$$p = \frac{15}{20} = 0.75$$

Albino ($2n$) + three heterozygotes ($3n$) = $5n$ alleles. Therefore:

$$q = \frac{5}{20} = 0.25$$

$$p + q = 1$$

$$0.75 + 0.25 = 1$$

The equation is useful for determining the frequency of an unknown allele when the frequency of the other allele is known.

The **Hardy–Weinberg equilibrium** describes a population in which allele frequencies tend to remain constant over generations. This occurs when:

- all phenotypes have equal survival value
- no selecting agent is acting on any particular phenotype

- random mating results in viable offspring
- the population is relatively large
- there is no gene flow into or out of the population.

Inheritance patterns in a population

Understanding the pattern of inheritance of genetic traits in populations has long been the subject of human curiosity and scientific investigation. Historically, this work has been painstaking and time-consuming. However, advances in genetic and computer technologies have enormously increased the efficiency of these processes. The use of computers has meant that large volumes of data can be managed with great efficiency and speed, and used in a wide range of applications. Inheritance patterns in large populations can be analysed and predictions made with greater accuracy than ever.

GENETIC TECHNOLOGIES

A range of technological advances in genetics allow scientists to investigate, measure and manipulate the genetic information of species. These include tools used to sequence genomes, clone organisms, genetically transform or modify organisms, determine inheritance patterns and evolutionary relationships, and diagnose and treat genetic conditions. Such tools include DNA sequencing, DNA profiling, cytogenetic testing and gel electrophoresis. See Module 6 for further details about these and other genetic tools and technologies.

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POPULATION GENETICS AND BIOINFORMATICS: IDENTIFYING TRENDS, PATTERNS AND RELATIONSHIPS

Bioinformatics is the use of computers and digital databases to manage biological information. This includes gathering, processing, storing, manipulating and analysing biological data.

Bioinformatics software has been instrumental in realising the Human Genome Project and analysing the genomes of other species. Analysis of the data tells us a great deal about the genetic composition of species, including the sequence of genes and the sequence of nucleotides within genes. Comparing the sequence of nucleotides in specific genes between healthy and affected individuals is vital in the detection and diagnosis of genetic diseases.

Bioinformatics is important in conservation; for example, it allows the identification of individuals most suitable for breeding to maintain maximum genetic variation in populations. Comparison of genomes between species also provides information about the degree of relatedness of those species in evolutionary terms.

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WORKSHEET 5.1

Knowledge review—revisiting foundation ideas

SCIENTIFIC METHOD

The scientific method is a vital tool that ensures a sound approach to investigations that yield reliable data and logical conclusions. By following the scientific method, researchers can contribute to the development of rigorous biological principles.

1 A student decided to test the idea that potatoes left in a dark cupboard can sprout stalks by vegetative reproduction. To test this idea, the student placed 10 similar-sized potatoes in a dark cupboard and another 10 potatoes on the kitchen bench for four weeks.

Complete the following table by entering the definition for each term and the example of each in this experiment.

Element of experiment	Definition	In this experiment
hypothesis		
independent variable		
dependent variable		

2 At the end of the test period, the student observed that all of the potatoes in the dark cupboard had grown stalks, some short and some long, ranging from 2–6 cm in length. The potatoes placed on the kitchen bench also showed some growth, but the stalks were much shorter and there were fewer of them.

Clarify which of the student's observations represent qualitative data and which represent quantitative data. Explain the difference.

3 Explain whether the student's investigation represents a primary or a secondary-sourced investigation.

RECOGNISING REPRODUCTIVE TERMS

4 Identify the odd term in each of the sets of words below. Circle the term and provide a full explanation for why it is different.

a germ cell sex cell somatic cell gamete

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b binary fission | fertilisation | budding | fragmentation

c marsupial | placental | monotreme | amphibian

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WORKSHEET 5.2

Reproductive routines—asexual vs sexual reproduction

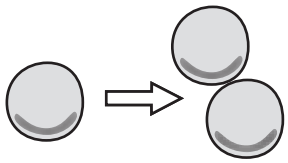
ASEXUAL REPRODUCTION

Asexual reproduction results in offspring that are genetically identical to the parent organism from which they are derived—that is, the offspring are clones of the parent. Although asexual reproduction results in identical offspring, the means of asexual reproduction varies between different kinds of organisms.

Consider the different kinds of organisms shown below.

1 Identify and describe the method of asexual reproduction involved in each case.

bacteria



Process: _____

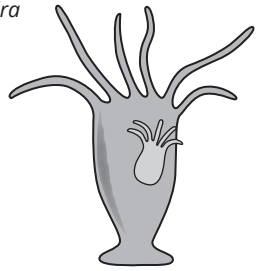
Description: _____

_____

_____

_____

Hydra



Process: _____

Description: _____

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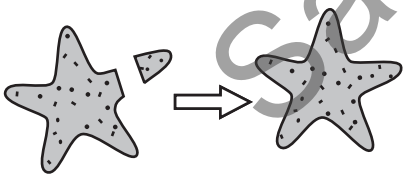
_____

_____

sea star



Process: _____

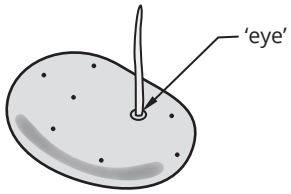
Description: _____

_____

_____

_____

potato tuber



Process: _____

Description: _____

_____

_____

_____

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2 Describe the circumstances in which asexual reproduction is advantageous to a population. Explain.

3 Describe the circumstances in which asexual reproduction can place a population at a disadvantage. Explain.

SEXUAL REPRODUCTION

Sexual reproduction results in offspring that are genetically unique. It involves genetic contributions from two parents, with offspring displaying a combination of traits from both. Offspring will be different from their parents and from each other. During sexual reproduction, haploid gametes from either parent unite in a process called fertilisation. In animals, fertilisation can be internal or external.

4 Define the following terms:

a haploid

b gamete

c fertilisation

d zygote

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5 Complete the table below, which summarises some aspects of sexual reproduction in animals.

	Fertilisation	
	External	**Internal**
Example		
Describe where fertilisation occurs and where the offspring develop.		
Outline an advantage of this kind of fertilisation.		
Outline a disadvantage of this kind of fertilisation.		

RATING MY LEARNING	My understanding improved	Not confident ◯ ◯ ◯ ◯ ◯ Very confident	I answered questions without help	Not confident ◯ ◯ ◯ ◯ ◯ Very confident	I corrected my errors without help	Not confident ◯ ◯ ◯ ◯ ◯ Very confident

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