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 echinoderms
spore formation	spores released into environment and germinate into new individuals	fungi plants, including mosses 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 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 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.

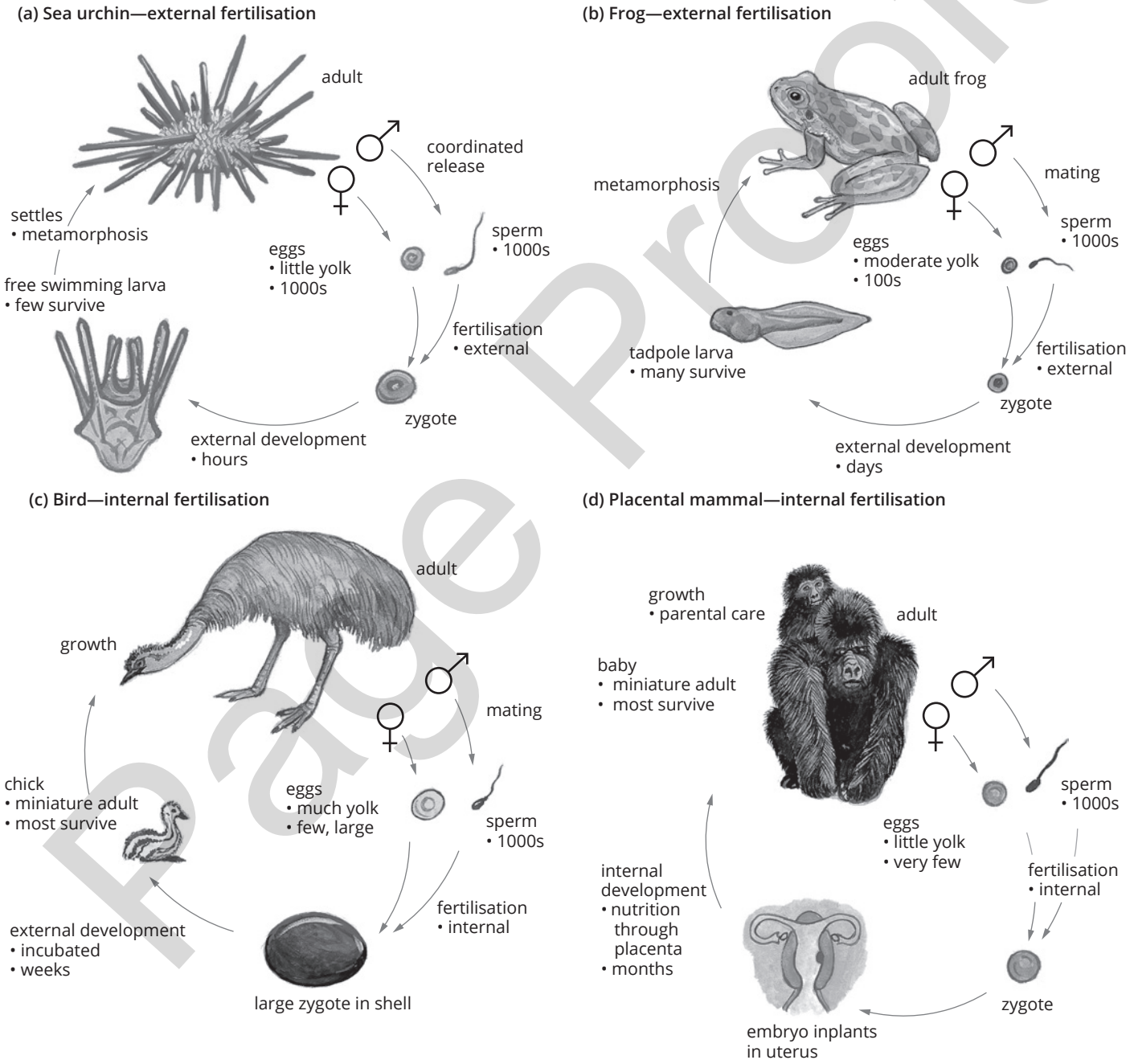

FIGURE 5.1 Life cycles of sexually reproducing organisms. External fertilisation occurs in many aquatic animals, such as (a) sea urchins and (b) frogs. Internal fertilisation is a common feature of terrestrial animals, including (c) birds and (d) mammals.

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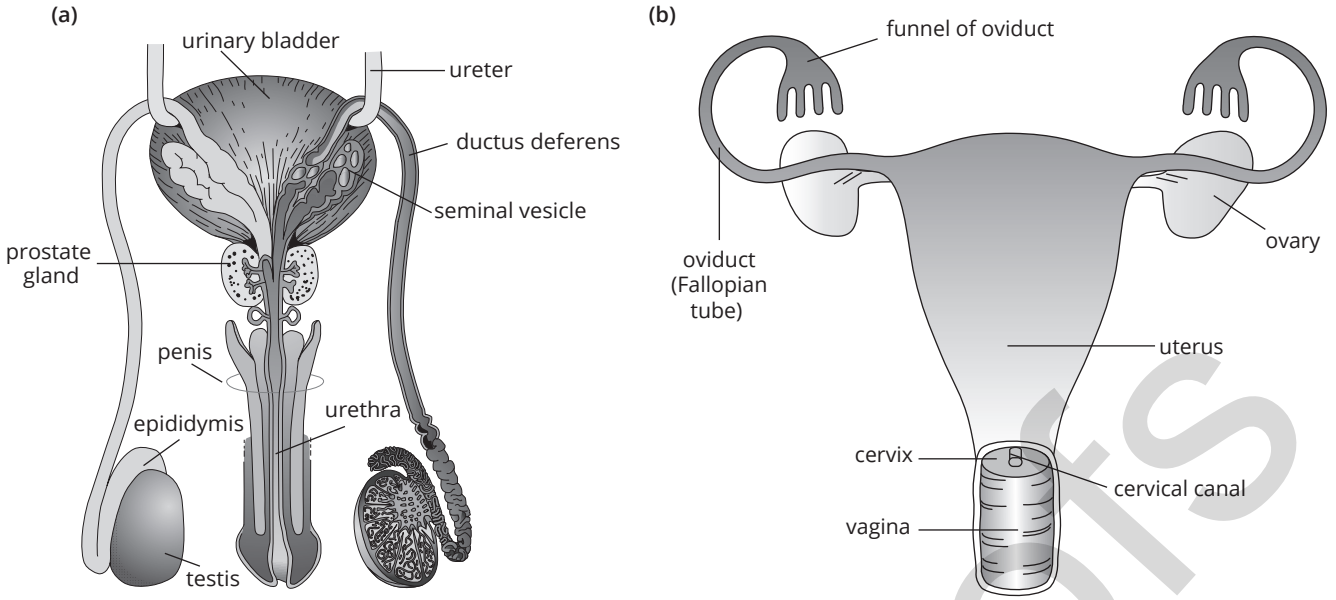

FIGURE 5.2 Reproductive systems in humans: (a) male and (b) female

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.

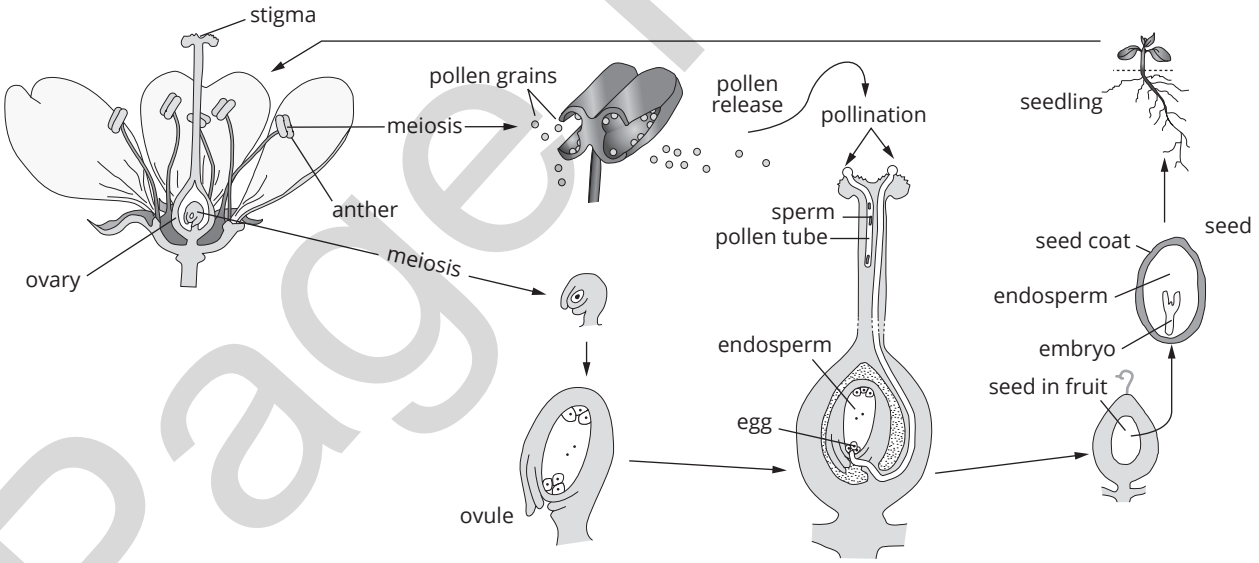

FIGURE 5.3 Male and female reproductive structures in flowering plants

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

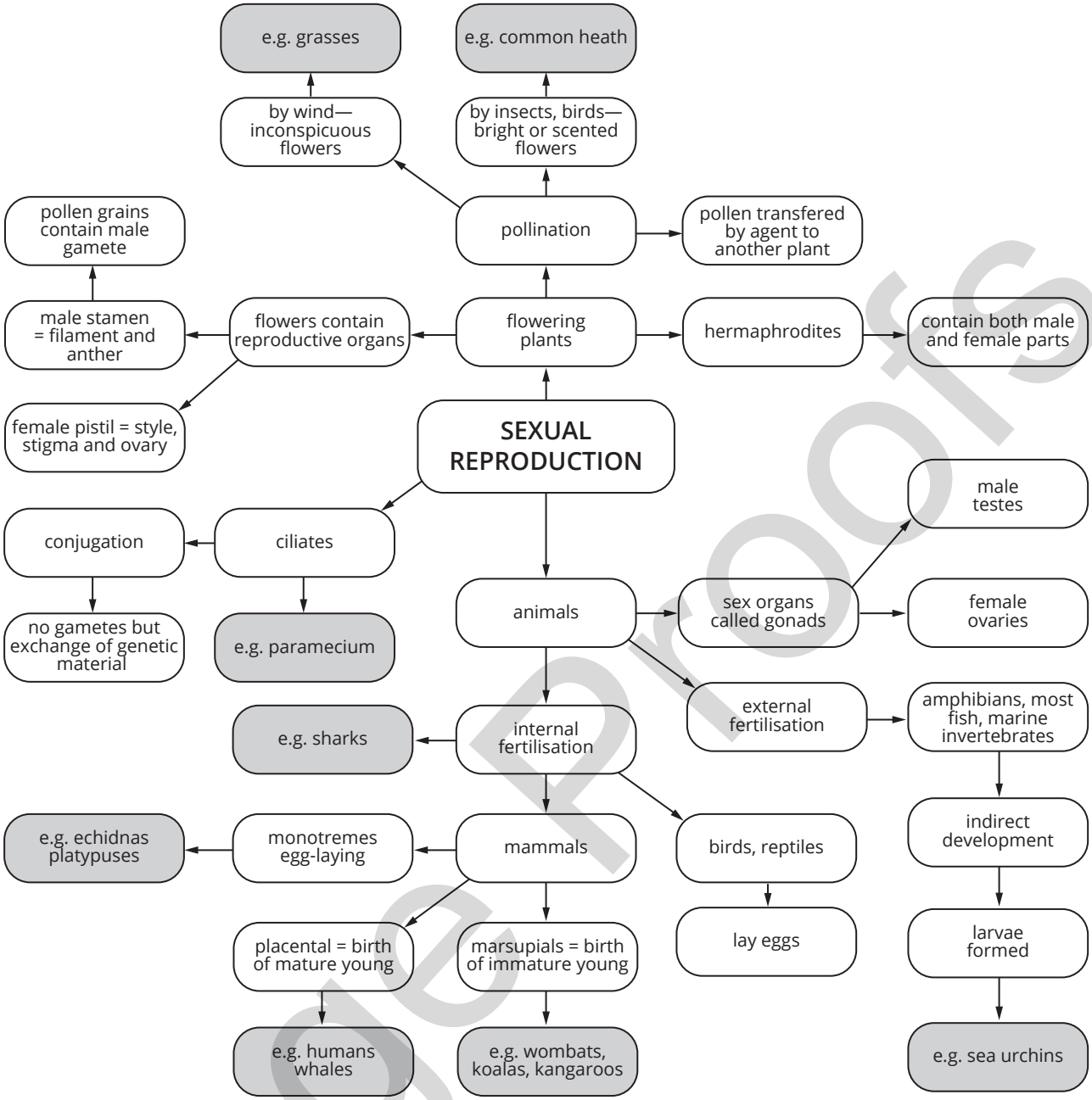

FIGURE 5.4 Sexual reproduction in animals, ciliates and flowering plants

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, we recognise various stages during development.

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.

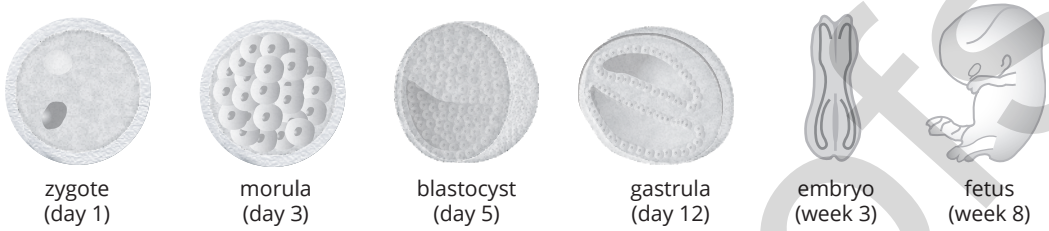

FIGURE 5.5 The development of a human zygote (fertilised egg) into a fetus

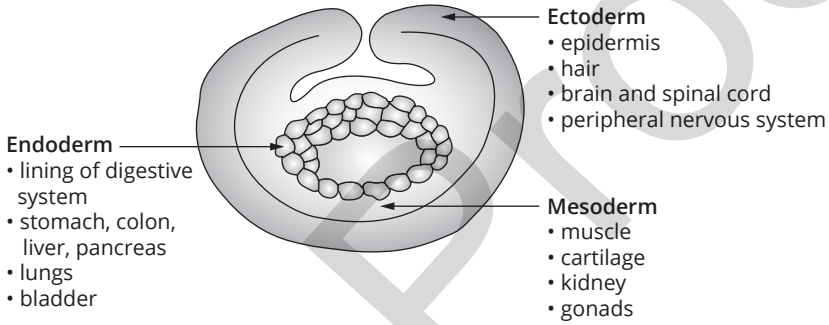

FIGURE 5.6 Gastrula with defined endoderm, mesoderm and ectoderm

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.

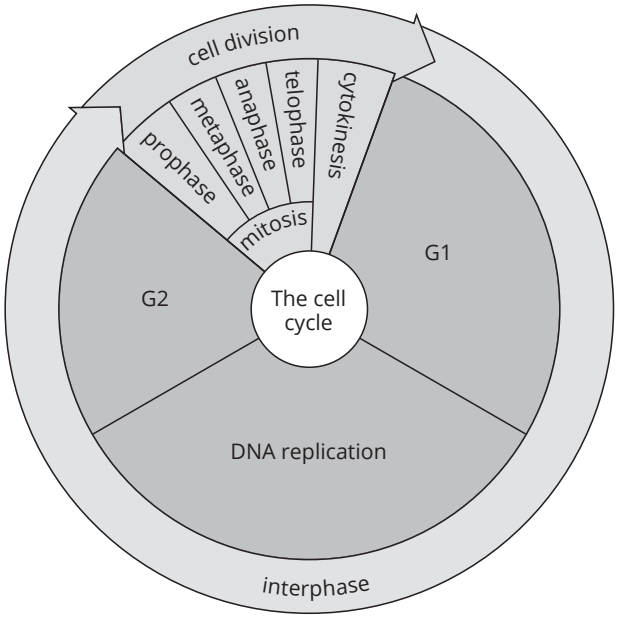

FIGURE 5.7 The cell cycle takes approximately 24 hours to complete in mammalian cells.

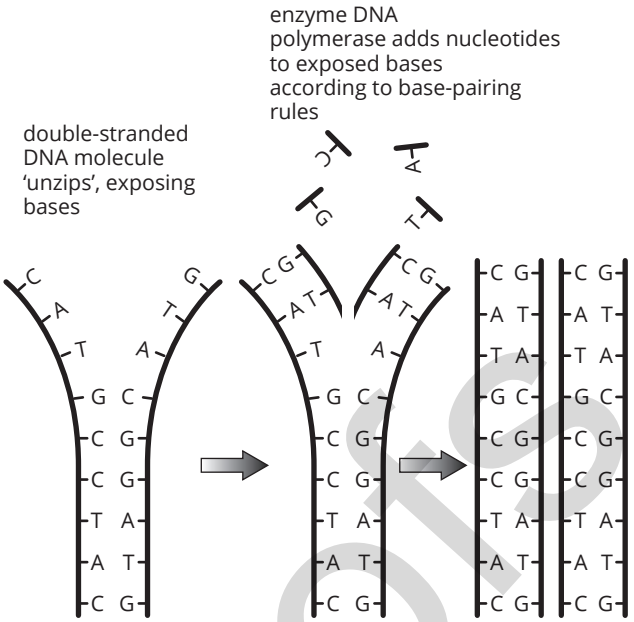

FIGURE 5.9 DNA replication—DNA polymerase adds nucleotides to build copies of the original DNA strands.

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 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.

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

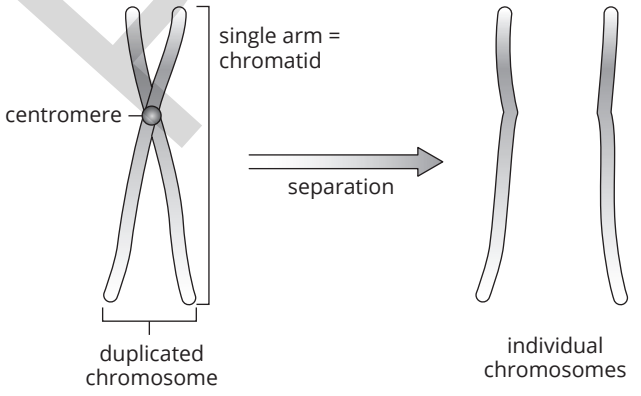

FIGURE 5.8 Structure of a chromosome

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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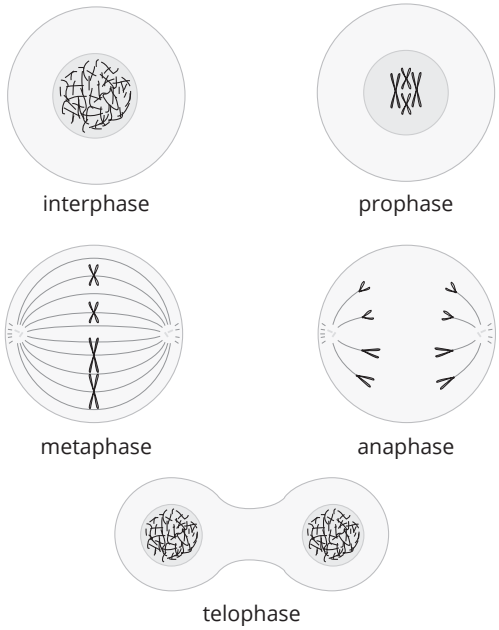

FIGURE 5.10 During mitosis, DNA replicated during interphase is divided into two new nuclei.

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.

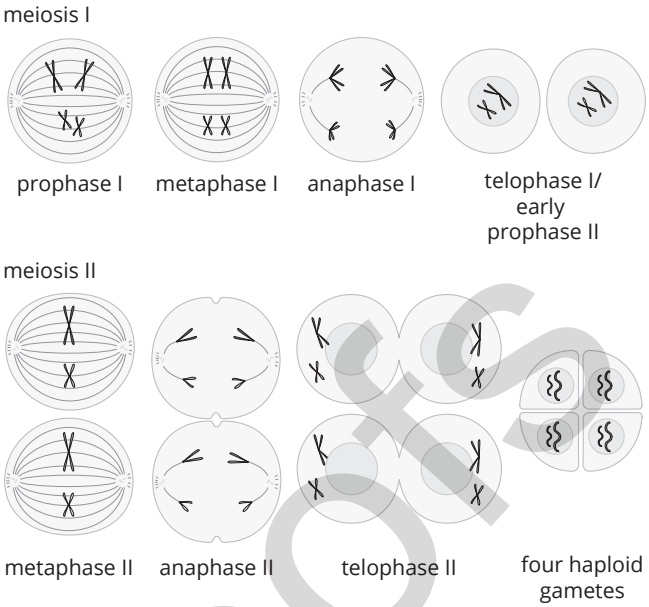

FIGURE 5.11 Meiosis produces four genetically unique gametes.

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** 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 (Figure 5.12). 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 ma y exchange chromosome segments. This results in **recombination** and is responsible for the genetic variation formed in gametes.

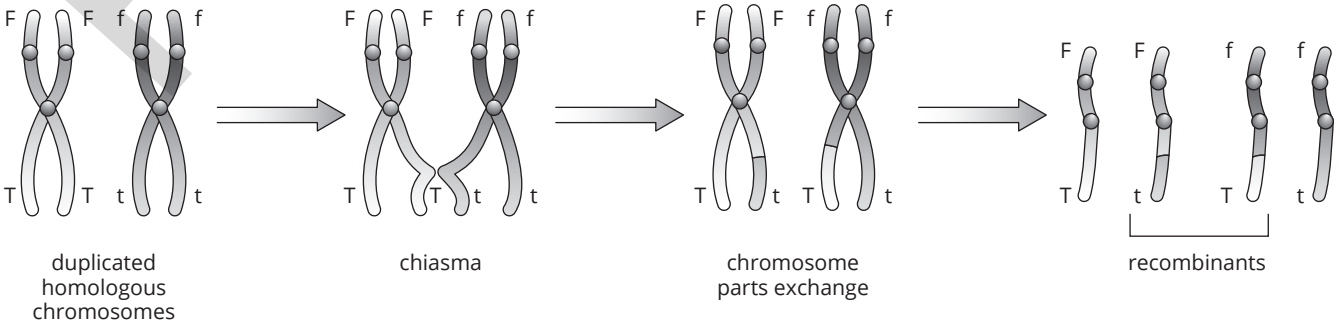

FIGURE 5.12 Homologous chromosomes are pairs of chromosomes that have the same genes, but may carry different alleles for those genes.

Cytokinesis

The division of the cytoplasm during cytokinesis marks the creation of the two new daughter cells. 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	DNA contained in paired chromosomes
no membrane-bound nucleus	DNA confined to membrane-bound nucleus
reproduction involves simple duplication and separation of chromosome	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.

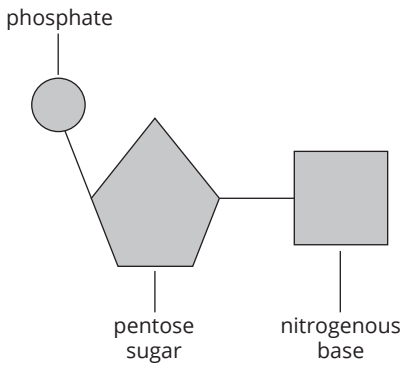

FIGURE 5.14 Nucleotide

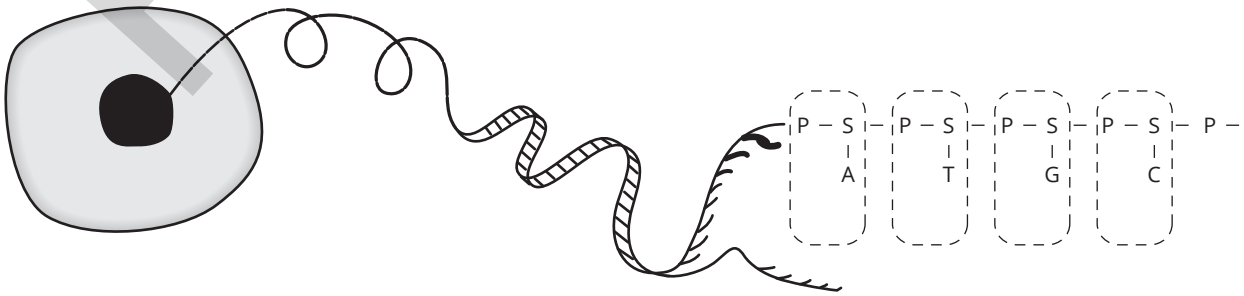

FIGURE 5.13 The DNA in the nucleus of cells unravels to reveal double helix. 'P' represents a phosphate group, 'S' represents a sugar and 'A', 'T', 'G' and 'C' represent nitrogenous bases. Together these three components make up a nucleotide.

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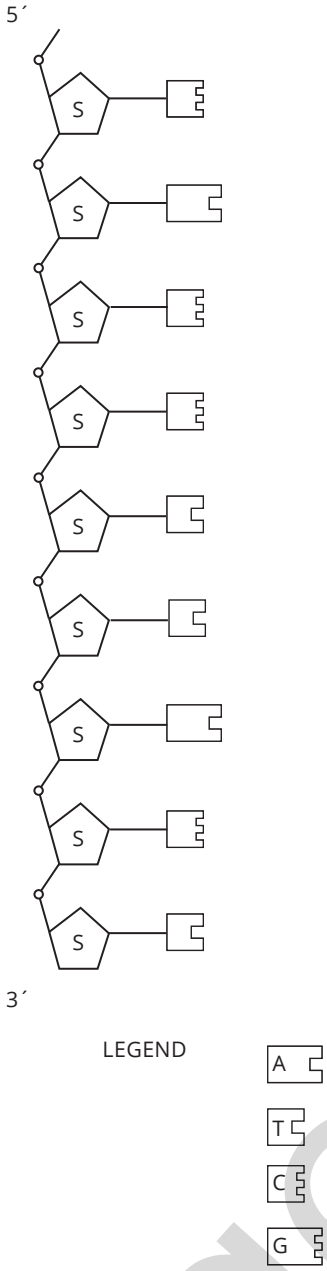

FIGURE 5.15 Nucleotide polymer—single-stranded DNA

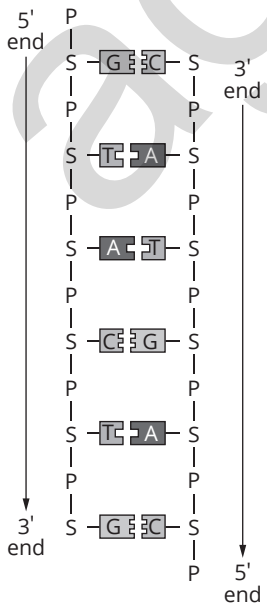

FIGURE 5.16 Nucleotides arranged in complementary pairs held together by hydrogen bonding

> 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

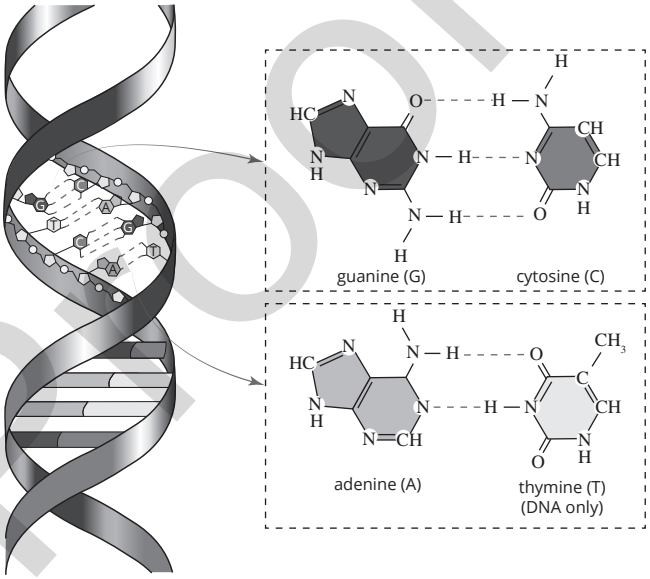

FIGURE 5.17 The helical structure of DNA. Two complementary strands form a double helix joined by base pairs guanine (G) and cytosine (C), and adenine (A) and thymine (T). Guanine (G) and adenine (A) have a double-ring structure and are known as purines, while cytosine (C) and thymine (T) have a single-ring 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 which takes **instructions** to the ribosomes in the cytoplasm
- **transfer RNA (tRNA)**: transfer RNA is the molecule that brings **amino acids** to ribosomes during protein synthesis
- **ribosomal RNA (rRNA)**: ribosomal RNA 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 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.

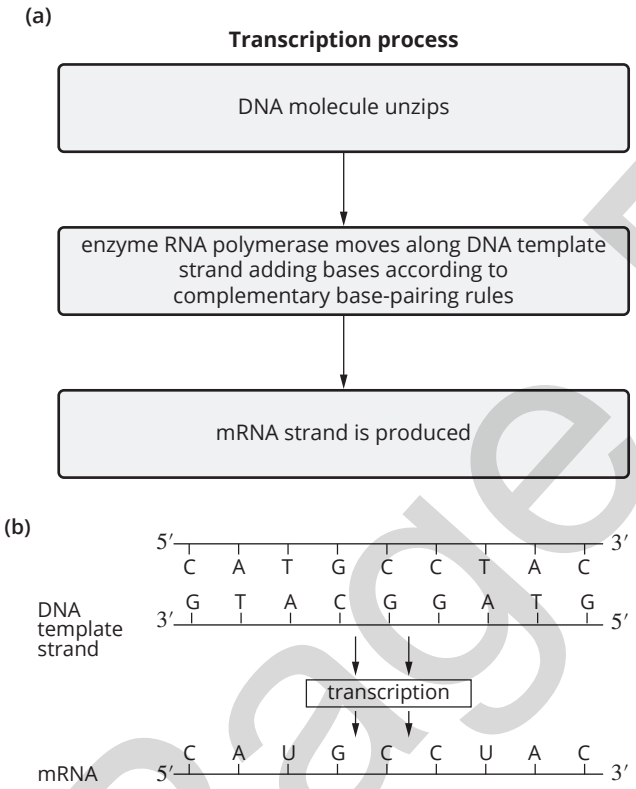

FIGURE 5.18 (a) Key steps in the process of transcription, where DNA is transcribed into messenger RNA (mRNA). (b) The single-stranded mRNA molecule contains uracil (U) instead of thymine (T).

Translation

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

Characteristics of translation:

- 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 5.3).
- 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.
- occurs in the cytoplasm at the ribosomes.

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.

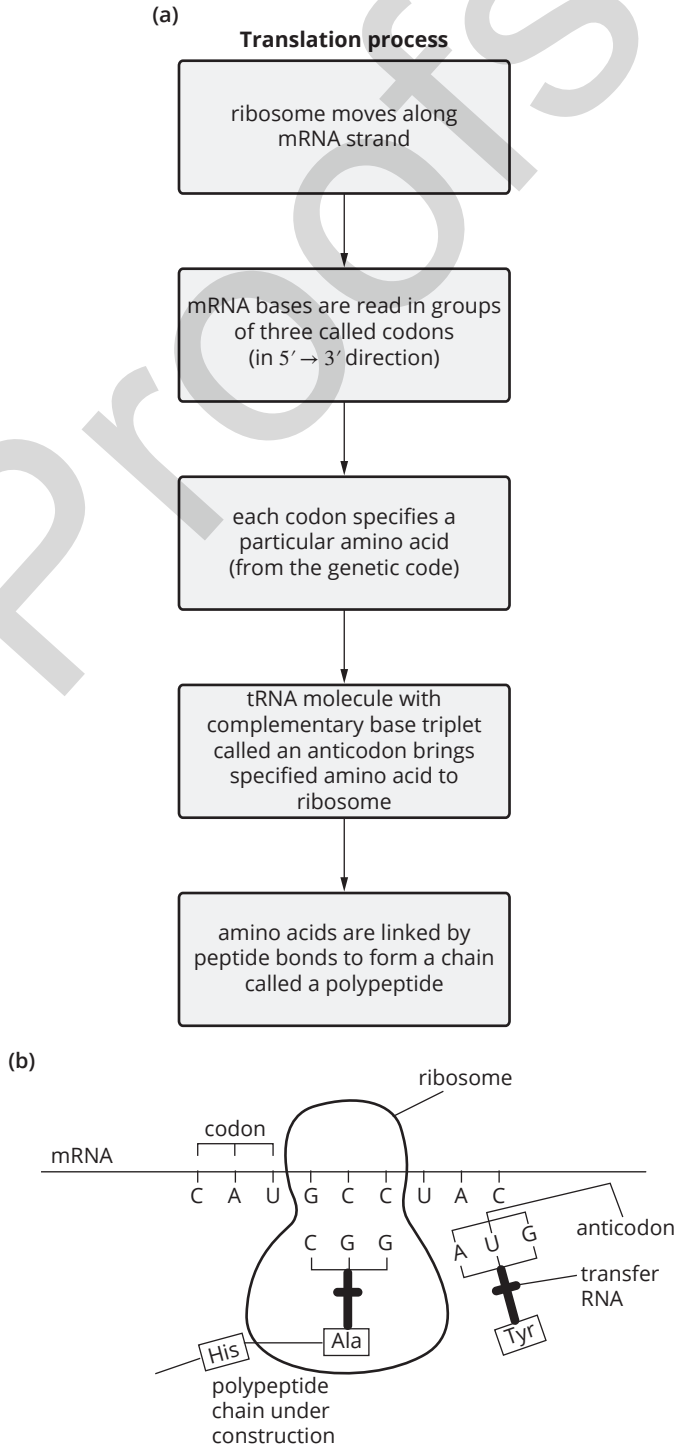

FIGURE 5.19 (a) Key steps in the process of translation, where mRNA is translated into amino acids to build a polypeptide chain. (b) Complementary anticodons brought by transfer RNA are joined in the ribosome to form a polypeptide chain.

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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

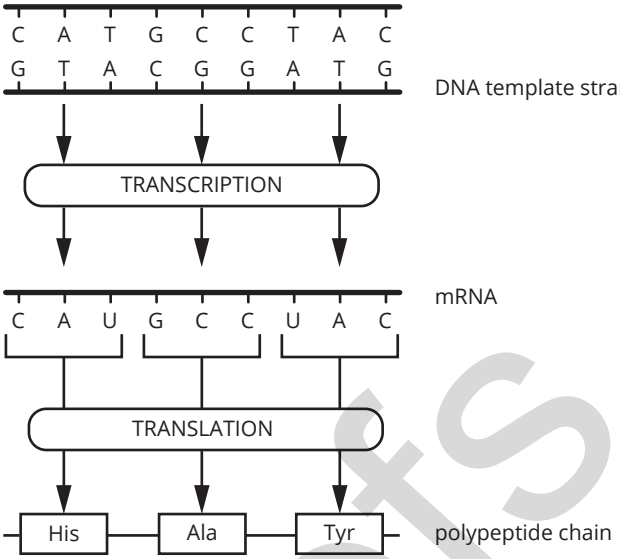

FIGURE 5.20 Summary of transcription and translation

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.

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.

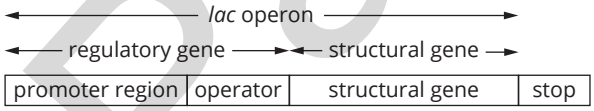

FIGURE 5.21 The *lac* operon

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** 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.

Characteristics of **genetics**:

- 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**. A trait is dominant when it appears in the phenotype of a heterozygote. Recessive traits only appear in the phenotype of homozygotes; they do not appear in the phenotype of a heterozygote.

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).

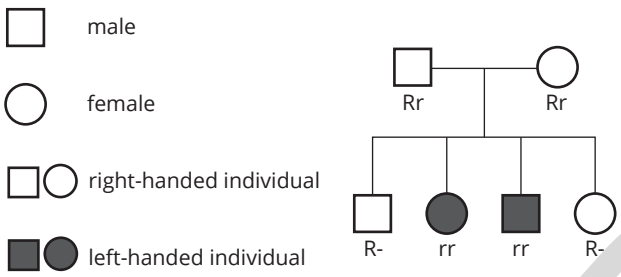

FIGURE 5.22 Right-handedness is inherited as an autosomal dominant trait. 'R-' represents an individual that may be homozyous (RR) or heterozygous (Rr) for the trait.

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 : ²⁄₄ pink : ¼ white

A 1 : 2 : 1 phenotypic ratio 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.

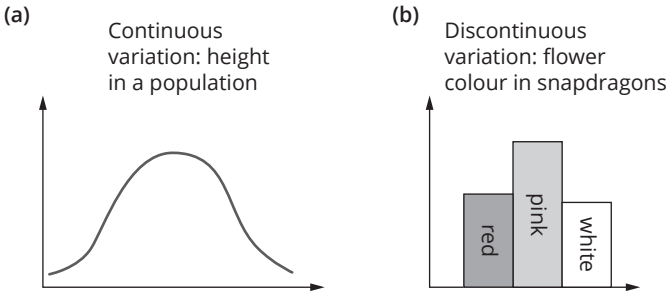

FIGURE 5.23 (a) Continuous variation (b) discontinuous variation

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.

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.

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

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.

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

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*

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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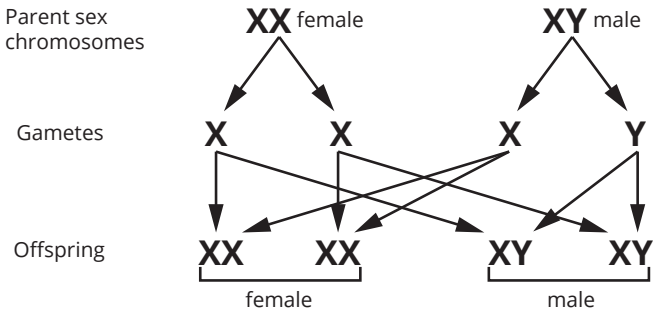

FIGURE 5.24 Sex determination in humans

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**.

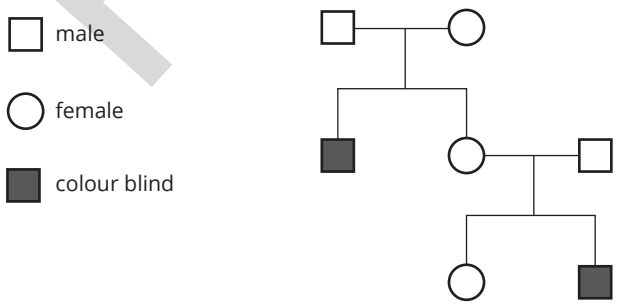

FIGURE 5.25 Colour blindness is inherited as a sex-linked, recessive trait.

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 make-up 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 make-up 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

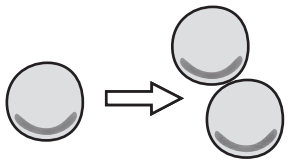
ASEXUAL REPRODUCTION

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: _____

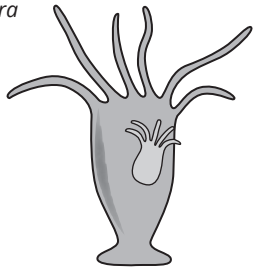
Description: _____

_____

_____

_____

Hydra



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Description: _____

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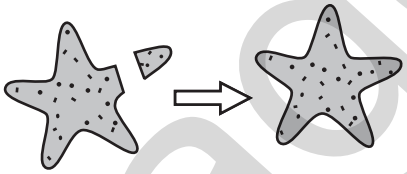
Sea star



Process: _____

Description: _____

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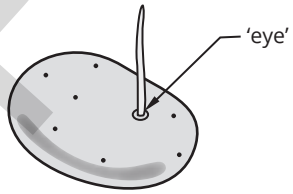
_____

_____

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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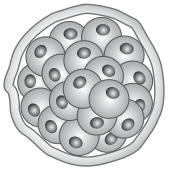
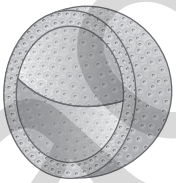
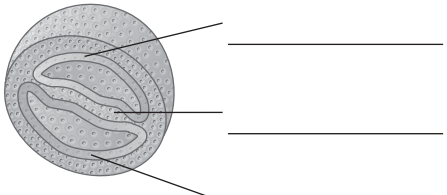
WORKSHEET 5.3

Development details—embryonic development in mammals

1 In mammals, fertilisation is followed by gradual development from zygote to embryo to fetus. To more easily follow the changes that occur, this development is divided into different stages. Select the appropriate terms from the following list to complete the table below. For each stage, describe the developmental changes that occur.

blastocyst	fetus	morula	ectoderm	embryo
gastrula	endoderm	zygote	mesoderm	implantation

Embryological development in humans		
Stage of development	**Diagram**	**Description**
_____ • fertilised egg • 1 cell	 day 1	
_____ • 16 cells	 day 3	
_____	 day 5	
_____ • endoderm • mesoderm • ectoderm	 (day 12)	

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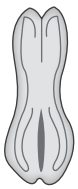
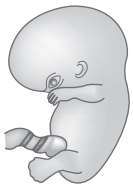
embryo	 week 3	
_____	 week 8	

2 Summarise cell growth and differentiation during the embryonic development of mammals.

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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WORKSHEET 5.4

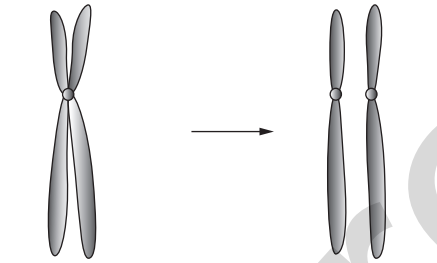
Cell cycle—chromosomes, replication and mitosis

CHROMOSOMES AND THE CELL CYCLE

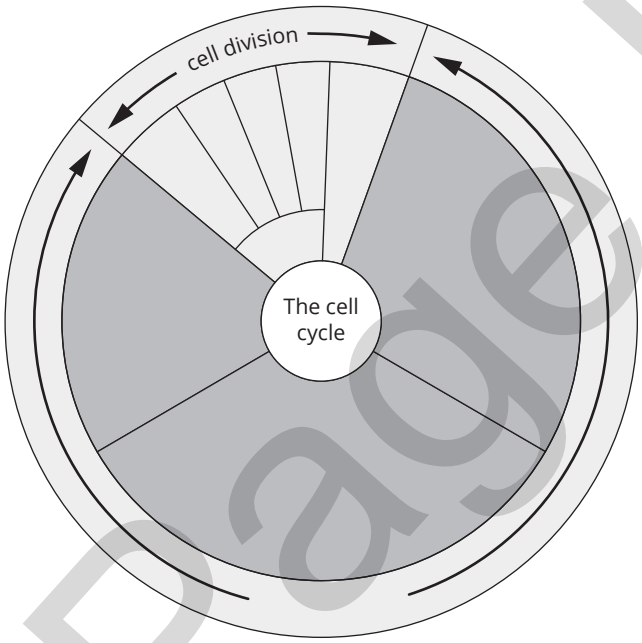
1 The genetic material in the nuclei of cells is contained in strands called chromosomes. The diagram below shows two chromosomes: one duplicated, and the other single-stranded.

a Label the centromere and a chromatid.

b Explain the difference between a chromosome and a chromatid.



2 Label the stages shown in the cell cycle below. Briefly summarise what occurs at each stage.



3 Outline the significance of cell replication. Include two key points.

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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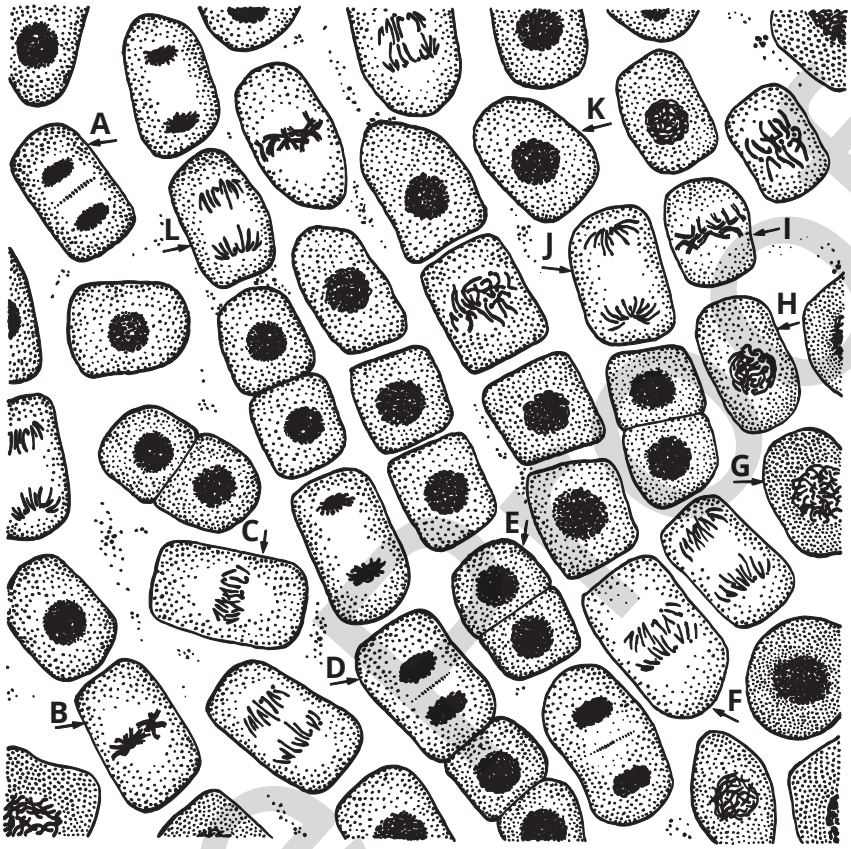
WORKSHEET 5.5

Mitosis mixer—nuclear division in somatic cells

Mitosis is the type of nuclear division that occurs in dividing somatic cells during tissue growth and repair. The illustration below represents onion root-tip cells at various stages. They have been stained to distinguish the DNA.

1 Examine the cells at the different stages of the cell cycle. Some of the cells have been labelled with a letter of the alphabet. Identify the stage of mitosis represented by these cells and record this in the table below.



Stage of mitosis	Cells in stage
interphase	
prophase	
metaphase	
anaphase (early)	
anaphase (late)	
telophase	

2 In terms of genetic make-up, describe the results of mitosis.

3 Interphase is sometimes called the 'resting phase' of the cell cycle. This is a misnomer. Outline what is happening in the nuclei of interphase cells.

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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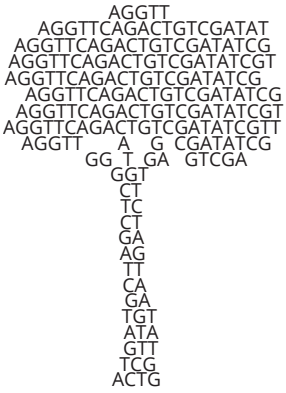
WORKSHEET 5.6

Nuclear puzzle—same pieces, different species



Millions of different species of organisms have evolved on Earth—bacteria, protists, algae, fungi, plants, animals and more. Within a single species there is also enormous diversity. And yet, we account for every individual using the same fundamental threads of genetic material—DNA. The DNA that codes for the staggering number of different organisms, and the features that make each one unique, comes in only four different forms. The pieces of the DNA puzzle, the nucleotides, are characterised by a different base molecule—adenine, thymine, cytosine or guanine. It is the infinite number of combinations that gives us such enormous variety.

1 Nucleotides are composed of the same three components. Name the molecule(s) represented by:

P _____ S _____

A, T, C and G _____

2 The nucleotide sequence in the figure to the right is part of the human β-haemoglobin gene.

Use coloured pencils to colour-code the nitrogenous bases in the legend.

Follow your code to colour the different bases along the DNA sequence.

3 Use appropriate symbols and colour coding to draw the complementary DNA sequence against this template strand.

4 Look carefully at the details of your double-stranded DNA. Describe two features of DNA that ensure complementary base-pairing occurs.

Feature 1:

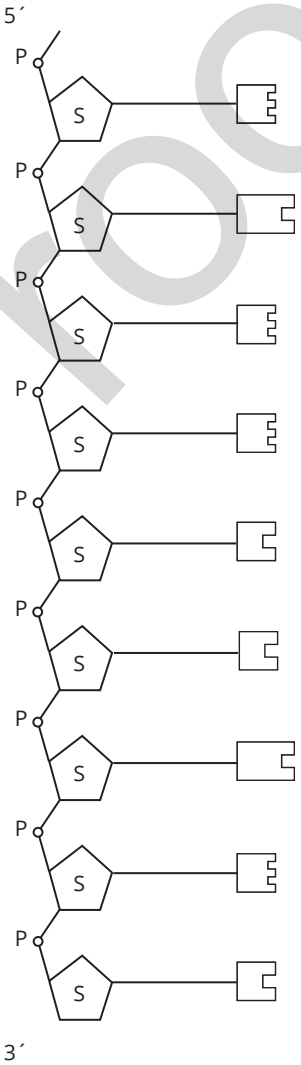
_____

_____

Feature 2:

_____

_____



LEGEND

A

T

C

G

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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WORKSHEET 5.7

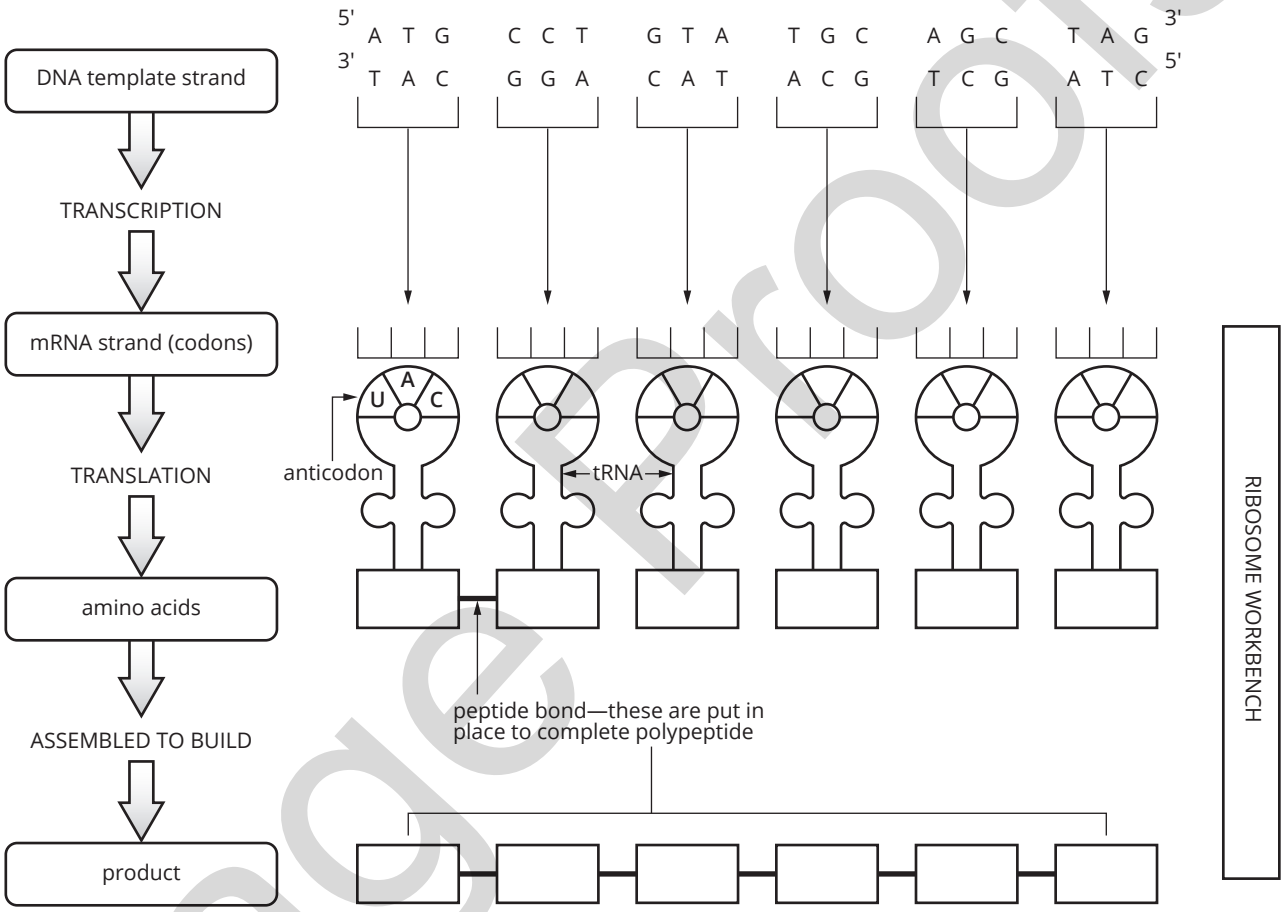
Genetic dictionary—reading the code

DNA is a biological book of words and meanings that can be likened to a dictionary where:

- the words represent the genes
- the definitions are an expression of what each word means. In this case, the expression of a gene is the protein for which it codes.

The diagram below shows each step involved in reading the DNA sequence of a gene segment. By following each step, the final meaning is revealed.

1 Use your knowledge of complementary base-pairing rules and the genetic code to add in the missing instructions in the diagram below. Refer to the genetic code in Table 5.4. GO TO ➤ p13



2 Describe what happens during:

a transcription

b translation

3 Name the first and last codons in the above sequence. Outline their significance.

4 **a** What is the name of the product that has been constructed at the end of the process?

b Outline the significance of this product, giving two specific examples.

SUMMARY

Use a single sentence to outline the fundamental role of genes.

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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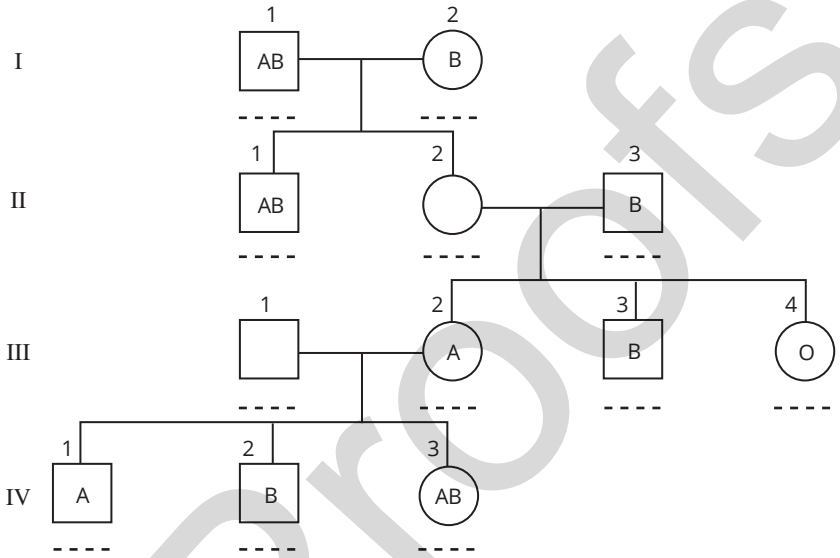
WORKSHEET 5.8

Puzzling pedigrees—analysing family histories

BLOOD RELATIVES

The ABO blood group of an individual can be determined by identifying the kinds of proteins (antigens) present on the surfaces of red blood cells. The single gene locus that codes for the production of these antigens has three alleles (I^A, I^B and i). The genotypes and phenotypes of respective individuals are shown in the table below.

ABO blood groups	
Genotype	**Phenotype**
I^AI^A	A
I^Ai	A
I^BI^B	B
I^Bi	B
I^AI^B	AB
ii	O



ABO blood group pedigree: females are represented by circles and males are represented by squares. Generations are indicated by I–IV.

1 The pedigree above indicates the blood type for some individuals. Use your understanding of inheritance and the alleles above to assign genotypes and blood types to individuals II-2 and III-1. Explain your reasoning in each case.

2 Outline the relationship between the phenotypic expression of the I^A, I^B and i alleles.

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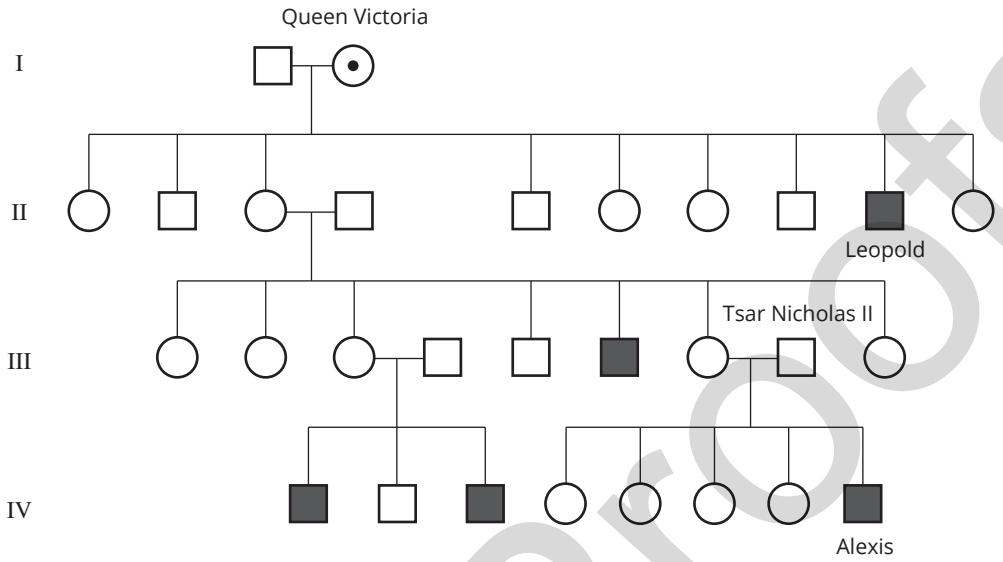
ROYAL BLOOD

The pedigree below represents part of the family tree for a European royal family. It also tracks the inheritance of haemophilia, a blood disorder that leaves sufferers without an important blood clotting factor, leading to uncontrolled bleeding after even minor injury. Single nucleotide polymorphisms (SNPs) in genes associated with clotting factors are the most common cause of haemophilia. Today, haemophiliacs are successfully treated with blood transfusions, but in the past individuals born with this disorder usually did not survive childhood.



Pattern of inheritance of the blood disorder haemophilia in a European royal family: females are represented by circles and males are represented by squares. Generations are indicated by I–IV.

3 Suggest why only males in this family tree are affected by haemophilia.

4 Queen Victoria's son, Leopold, was the first person in the family's history to have been diagnosed with the condition. The cause of the disease in this family is attributed to a mutation that occurred early in the embryological development of Queen Victoria or in a germline cell from one of her parents. Describe the evidence from this family tree that points to Queen Victoria as the origin of haemophilia in the family, and not her son Leopold.

5 In the pedigree, Queen Victoria's status as a carrier is denoted by the dot symbol. Use the same notation to identify all the other carriers of haemophilia in this family.

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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WORKSHEET 5.9

Literacy review—genetic glossary

1 Read the definitions given in the boxes on the right-hand side of the page. Choose the correct term from the list below to match each definition. Write these in the box corresponding to its definition.

genotype allele gene phenotype

	an alternative form of a gene
	a unit of hereditary information that determines the characteristics of an organism
	genetic make-up of an individual in relation to one or more genes
	physical expression of a genotype

2 The words below are grouped according to the close relationships between them. In each instance, one definition is provided for you, while you must provide the missing definitions. Hint: Use the definition provided as a guide for constructing the remaining definitions.

asexual reproduction	reproduction in which there is no mixing of DNA from different parents, resulting in offspring that are genetically identical to the parent cell or organism
sexual reproduction	
mitosis	the process of nuclear division in which duplicated chromosomes are organised and distributed so that daughter cells contain the same number of chromosomes as the parent cell, and are genetically identical to the parent cell from which they have arisen
meiosis	
homozygous	describes the status of an individual's genotype when identical alleles are present for the gene locus
heterozygous	
dominant	describes a characteristic that is observed in the phenotype of a heterozygote
recessive	
co-dominant	

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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WORKSHEET 5.10

Thinking about my learning

On completion of Module 5: Heredity, you should be able to describe, explain and apply the relevant scientific ideas. You should also be able to interpret, analyse and evaluate data.

1 The following table lists the key knowledge covered in this module. Read each and reflect on how well you understand each concept. Rate your learning by shading the circle that corresponds to your level of understanding for each concept. It may be helpful to use colour as a visual representation, for example:

- green—very confident
- orange—middle
- red—starting to develop.

Concept focus	**Rate my learning**				
	Starting to develop ◄				► Very confident
Sexual and asexual reproduction in organisms	○	○	○	○	○
Cell cycle, including DNA replication, mitosis, meiosis	○	○	○	○	○
DNA structure	○	○	○	○	○
Polypeptide synthesis—transcription and translation	○	○	○	○	○
Genetic variation and inheritance patterns	○	○	○	○	○
Impact of genes and environment on phenotype	○	○	○	○	○
Importance of DNA synthesis and cell replication	○	○	○	○	○
Importance of polypeptide synthesis for organisms	○	○	○	○	○

2 Consider points you have shaded from starting to develop to middle-level understanding. List specific ideas you can identify that were challenging.

3 Write down two different strategies that you will apply to help further your understanding of these ideas.

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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.

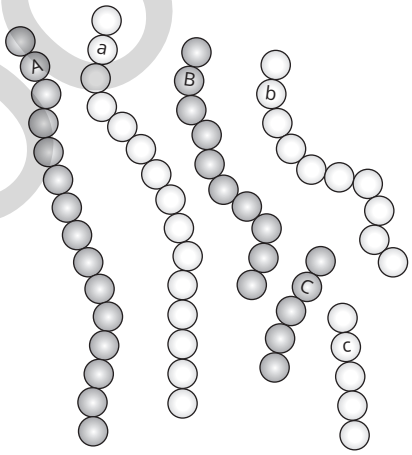
PURPOSE

- 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.

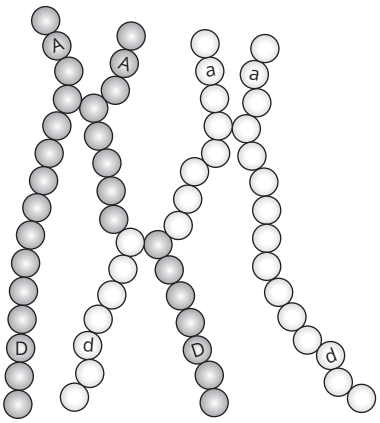
4 Complete Data and analysis steps 1–4.

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 question 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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2 Now mix up the chromosomes in your cell. Randomly move them into groups containing one set of chromosomes each, to represent possible gamete combinations. Compare your gametes with other students in your class.

Comment on the kinds of gametes produced. Are they all the same?

3 Write down all the possible combinations of chromosomes that could occur in gametes produced from a cell containing three chromosomes with alternative alleles at a given gene locus.

4 The formula for calculating the possible number of combinations, and therefore the possible number of different gametes, is 2^x, where x = number of gene loci, and the base number 2 represents the number of chromosomes in a homologous pair. This formula gives us a total of $2^3 = 8$ different kinds of gametes in step 3.

Calculate the number of possibilities when there are:

a four chromosome pairs (i.e. four gene loci)

b five chromosome pairs

c 10 chromosome pairs.

Imagine the number of possibilities for 23 chromosome pairs! Add to this the large number of gene loci on each chromosome, together with their alternative alleles.

DISCUSSION

5 What is the point of contact called for chromosomes undergoing a crossing-over event?

6 At what stage of meiosis does this event occur?

7 How does crossing over affect the kinds of chromosomes produced?

CONCLUSION

8 Outline the significance of meiosis.

9 Summarise the processes occurring during meiosis that contribute to the variety of gametes produced.

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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PRACTICAL ACTIVITY 5.2

Modelling DNA—simulating the structure

Suggested duration: 50 minutes

INTRODUCTION

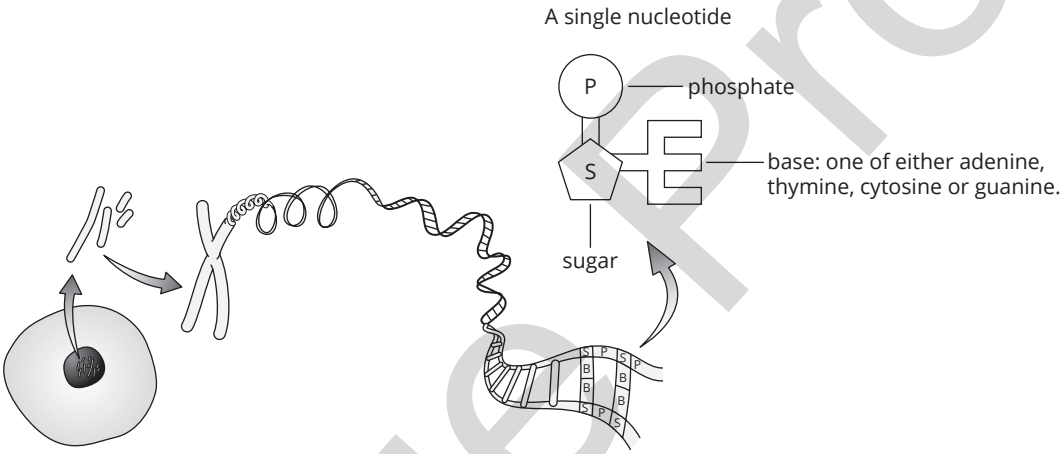
DNA, or deoxyribonucleic acid, is often referred to as the 'blueprint' for life—a universal code that provides the instructions for protein synthesis in all living organisms.

The building blocks of DNA are nucleotides composed of three parts—a deoxyribose sugar, a phosphate component and a nitrogenous base (one of either adenine, thymine, guanine or cytosine). The nucleotides link together to form two strands running in an antiparallel arrangement with complementary base-pairing between adenine (A) and thymine (T), and between guanine (G) and cytosine (C).

A single DNA molecule may measure more than a metre in length when fully unwound. To fit within the nucleus of a cell and maintain its base-pairing order, the DNA coils tightly to form a chromosome.

PURPOSE

To investigate the structure of DNA through a modelling activity.



Part A Modelling DNA at the chromosomal level

PROCEDURE

1 Using two different colours of plasticine, roll out two single-stranded chromosomes, one of each colour. Ensure their size is similar. This represents a homologous pair. The different colours represent the maternal and paternal chromosomes.

2 Use the coloured beads to mark two gene loci on your plasticine model. For one locus, show identical alleles, and for the other, show different alleles.

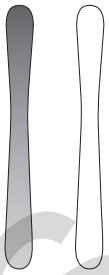
3 During DNA replication, the double helix produces an identical copy in preparation for cell division. At this point of the cell cycle, the duplicated chromosomes are held together by a centromere. Add another plasticine roll to each single chromosome to model a duplicated homologous pair.

4 Add further beads to your duplicated pair to show the alleles now present on each chromatid.

MATERIALS

- 2 different colours of plasticine or similar
- set of 2 different-coloured beads (approximately 10 of each)

DATA AND ANALYSIS



1 Describe how the chromosomes in the homologous pair are similar to and different from each other.

2 Outline the difference between a gene and an allele.

3 Explain what is meant by the terms homozygous and heterozygous.

4 Sketch a diagram of your duplicated pair of homologous chromosomes. Use colour coding and the following terms to add labels:

duplicated chromosomes chromatid centromere
gene locus homozygous heterozygous

5 Identify and describe two features of homologous chromosomes that are clearly demonstrated by your chromosome model.

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6 Share your response to step 5 with others. Are the responses of your classmates similar to or different from your suggestion? Explain.

Part B Modelling DNA at the biomolecular level

MATERIALS

- set of nucleotides photocopied from template provided (at least four of each A, T, G, C)
- A3 sheet paper
- scissors
- material to fix nucleotide 'cut-outs' to paper (e.g. tack or sticky tape)

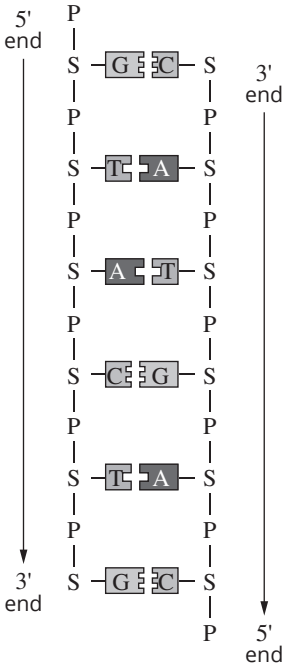
PROCEDURE

In part B you will zoom in on the DNA structure as the chromosome unwinds. When the double helix is completely unwound, the nucleotides appear in two complementary strands that resemble a ladder, with the ribose sugar and phosphate groups forming the uprights and the nitrogenous bases forming the rungs.

1 Collect a set of the photocopied nucleotide templates from your teacher. Allocate a colour for each of the four nucleotides and colour the bases accordingly. Record a legend on the A3 paper to indicate which base each colour represents. Note how each of the four kinds of nucleotides are similar to and different from each other.

2 Cut out all of the nucleotides. You should have four of each kind, making 16 individual nucleotides in total.

3 Arrange a strand of nucleotides to resemble a single strand of DNA running in the 5' to 3' direction. Use the diagram to the right as a guide. Consider the order of your nucleotides. What does the order that you selected represent?



4 Fix the nucleotides in position downwards along the length of the A3 page.

5 Construct the complementary strand against the first strand, following the base-pairing rules. Fix the nucleotides in position.

DATA AND ANALYSIS

1 Look carefully at the arrangement of the two DNA strands you have constructed. Explain why the DNA molecule is described as 'antiparallel'.

2 Describe two features of DNA that ensure complementary base-pairing is maintained between the strands.

3 One of the limitations of this modelling activity is that it does not accurately demonstrate the three-dimensional structure of the double helix. Suggest how you could modify your model to more accurately reflect the three-dimensional structure of the double helix.

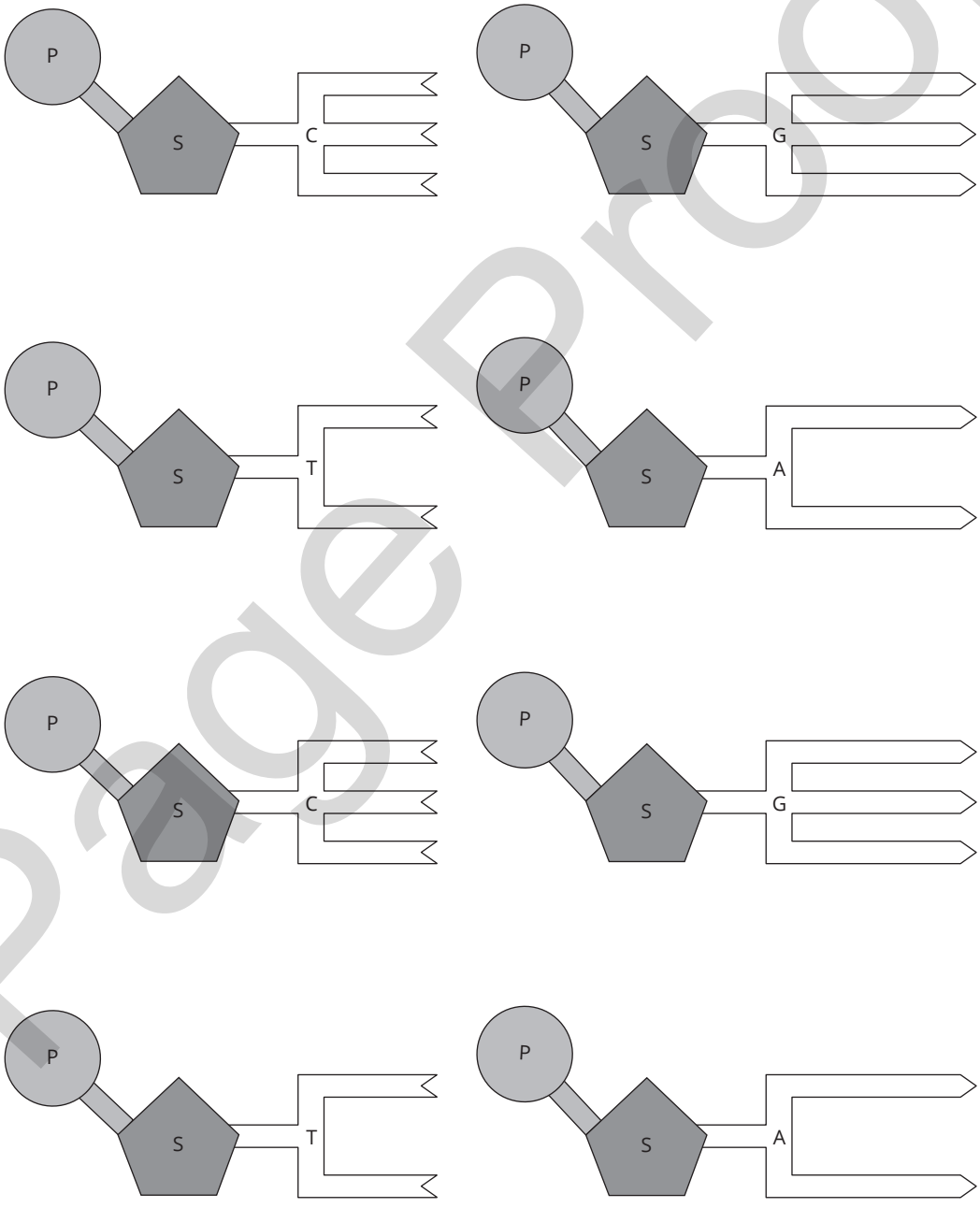
4 Identify and describe another limitation of your model in demonstrating the structure and/or function of the DNA molecule.

CONCLUSION

5 Nucleotides are the structural units of which the DNA molecule is composed. Name the three components of a single nucleotide.

6 Summarise the way nucleotides are arranged to form the double helix of the DNA molecule.

PHOTOCOPIABLE TEMPLATES



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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PRACTICAL ACTIVITY 5.3

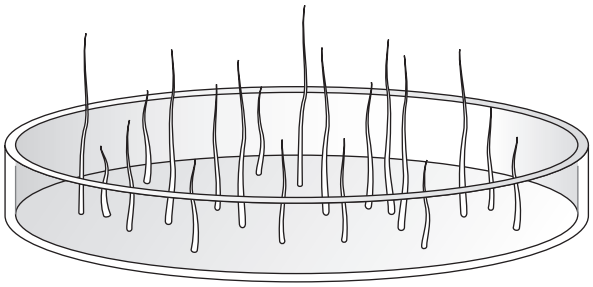
Betting on barley—a monohybrid cross

Suggested duration: 20 minutes to set up, then 60 minutes for analysis and written report approximately two weeks later

INTRODUCTION

Pigmentation in barley is controlled by a single gene with two alternative alleles. In the heterozygote, expression of green pigment masks the effect of the allele coding for no pigment (albino). The genetic barley used in this experiment is the result of a cross between plants heterozygous for the gene locus in question.

This activity recommends that a total of around 200 barley seeds be grown by the class. Your teacher will assign a number of seeds to your group, depending on the number of groups in your class.



PURPOSE

To investigate the mode of inheritance of a genetic trait in a monohybrid cross using genetic barley.

PROCEDURE

MATERIALS

- 20 seeds of genetic barley
- sheet of cotton wool
- large Petri dish
- water spray dispenser
- forceps

1. Lay a sheet of cotton wool inside the Petri dish and spray generously with water until the cotton wool is quite damp.
2. Use the forceps to arrange the barley seeds on the cotton wool so that they are evenly spaced about 1 cm apart.
3. Spray a little more water to ensure the seeds are dampened.
4. Leave the Petri dish on a bench near a window.
5. Spray the seeds twice daily to ensure they do not dry out. You should continue to do this after initial germination until the barley seedlings are at least 2 cm tall. This is likely to take about two weeks.

TWO WEEKS LATER

1. Propose a hypothesis for this investigation.

_____

_____

DATA AND ANALYSIS

2. Count the number of different-coloured seedlings and enter your results into Table 1.

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3 Collate the class data. A spreadsheet, whiteboard or overhead projector will be useful for this. Enter the class data into Table 1.

TABLE 1 Data for genetic barley practical activity

	Number of seedlings of each colour		Total
	Green	**Albino**	
Own data			
Class data			

4 Calculate the ratio of green seedlings to albino seedlings for:

a your own data _____

b the class data _____

5 **a** How does the ratio for your own data compare with the ratio for the class data?

b Which set of data is likely to be more reliable? Explain.

6 Use appropriate notation to assign genotypes to the different-coloured seedlings.

7 Homozygous green barley plants are indistinguishable from heterozygotes. In ordinary circumstances, a geneticist would carry out a test cross to determine the genotype of an individual that shows the dominant characteristic.

a Describe a test cross.

b Outline how a test cross is useful in determining the genotype of such an individual. Use a worked example to illustrate your answer.

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CONCLUSION

8 Summarise your findings in this activity. You should include the following items in your discussion.

- Comment on your hypothesis. Identify the mode of inheritance for green pigmentation in barley, drawing on the experimental evidence to support your position.
- Use a model to explain how the data supports your theory.

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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PRACTICAL ACTIVITY 5.4

Genetic roulette—people and pedigrees

Suggested duration: 50 minutes

INTRODUCTION

A look around your class illustrates many similarities between unrelated individuals—we share hair, eyes, ears, a nose, arms, legs and many other characteristics. Of course, there are many differences in the form of these characteristics between members of the group. For example, hair can be blonde, brown, red or black; straight, wavy or curly; fine in some individuals and coarse in others. There are enormous variations in eye colour. But look within a single family—there are more similarities between related individuals within a family than there are between unrelated members of your class. For example, fair skin and reddish hair are likely to be shared by individuals within one family. These characteristics are also likely to be shared by other generations within the family. Why is this? Family trees or pedigrees provide a useful way of analysing information that is inherited from one generation to the next within a family. Analysis of pedigrees gives us a clue about the way characteristics are inherited, and their pattern or mode of inheritance. They can also be used to determine the genetic make-up, or genotype, of an individual, and to predict the chances of children having particular features.

PURPOSE

- To analyse selected pedigrees to determine the mode of inheritance of genetic traits.
- To predict the possible outcomes in children born of particular partnerships in relation to inherited diseases.
- To construct pedigrees from family histories to determine modes of inheritance.

PROCEDURE

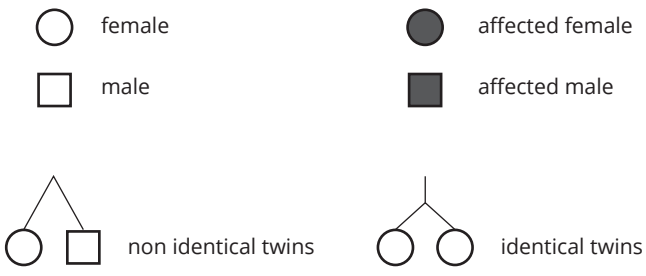
Read the information in Table 1, which outlines the key features that distinguish the modes of inheritance for different phenotypic characteristics. Use the information, along with the pedigree legend that follows, to help you answer the questions related to each of the pedigrees presented in part A and the problems raised in part B of this activity.

TABLE 1 Observed patterns for different types of traits

Pattern of inheritance	Key features
autosomal dominant	Gene loci on chromosomes other than sex chromosomes; either sex can be affected. Characteristic appears in the phenotype of a heterozygote. Affected individuals carry at least one allele for the dominant trait.
autosomal recessive	Gene loci on chromosomes other than sex chromosomes; either sex can be affected. Characteristic does not appear in the phenotype of the heterozygote. Affected individuals are homozygous recessive.
X-linked dominant	Gene loci located on X-chromosome. Affected males pass trait to all their daughters and none of their sons.
X-linked recessive	Gene loci located on X-chromosome. Affected females produce only affected sons. Expect half the sons of unaffected female carriers to be affected. Affected males produce only unaffected sons.

In pedigree analysis, symbols are used to provide specific information about individuals in a clear and simple way. Use the following legend as a guide to interpreting the pedigrees in this activity.

Legend



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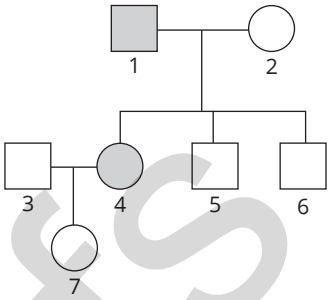
Part A Pedigree analysis

Scenario 1

The pedigrees shown in the figures to the right show the inheritance pattern of earlobe shape in two different families. 'Free lobes' are dominant to 'attached lobes', which are recessive. The gene responsible for earlobe shape has two alternative alleles represented by *E* (free lobes) and *e* (attached lobes).

1 Assign genotypes to as many individuals as possible in Figure (a).

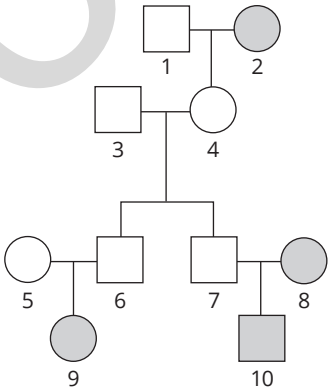
2 Describe the pattern or mode of inheritance for earlobe shape in humans.



(a) Pedigree of earlobe inheritance

3 Examine the pedigree in Figure (b). Assign genotypes to as many individuals as possible.

4 Why is it difficult to do this with confidence for individuals 1 and 3?



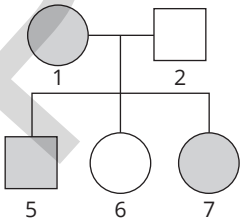
(b) Pedigree of free and attached earlobe inheritance in a family

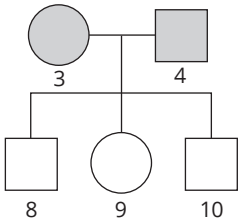
5 How can you be sure of the genotypes of individuals 6 and 7?

Scenario 2

Known family histories are also useful to geneticists in establishing the mode of inheritance for particular genetic diseases. Pedigree analysis for families that show such diseases is also important so that genetic counselling can be provided to families about the likelihood of future children being affected or carrying the allele in question. Huntington's disease is a neurological disorder that leads to gradual, permanent deterioration of nerve and muscle control, with eventual complete dependence on care. Death results after some years. Symptoms do not occur until at least the mid to late 30s.

The figure below illustrates the inheritance of Huntington's disease in two unrelated families. Individual 1 had one parent with Huntington's disease and one parent who did not have this condition.





6 Assign genotypes to each person in the first pedigree.

7 Name the mode of inheritance for Huntington's disease. Explain your choice.

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8 Explain why it is difficult to be certain of the genotypes of individuals in the second pedigree.

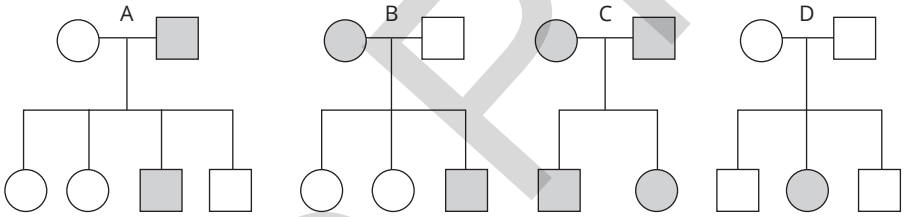
Individuals 7 and 8 are engaged to be married. Both individuals are keen to raise a family.

9 **a** Outline the chances of any children from this union developing Huntington's disease.

b Suggest options that a genetic counsellor might discuss with such a couple.

Scenario 3

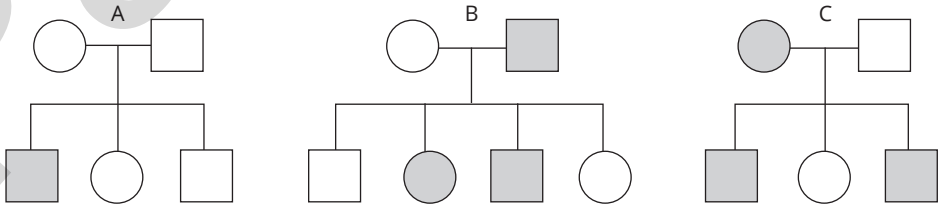
People with galactosaemia are unable to digest milk sugar (galactose). The pedigrees below (A–D) show four families with galactosaemia.



10 From the evidence in the pedigrees shown above, suggest which pedigree shows beyond doubt that galactosaemia is inherited as an autosomal recessive condition. Explain your reasoning.

Scenario 4

Red–green colour blindness is a relatively common condition, inherited as an X-linked recessive trait. The following figure shows the pedigrees of three families (A–C) in which this condition occurs.



11 Which of the three pedigrees best establishes the mode of inheritance for this trait? Explain your reasoning.

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Part B Picturing pedigrees

Prepare a pedigree chart in the space provided for each of the scenarios described below.

1 **a** An affected child is born to parents, neither of whom shows the characteristic.

b Determine the mode of inheritance. Explain.

2 **a** A man displaying a characteristic inherited as an X-linked trait marries an unaffected female. They have two affected daughters and two unaffected sons.

b Explain whether the trait is inherited as dominant or recessive.

3 **a** A woman showing a trait that has an X-linked recessive mode of inheritance has twins (a girl and a boy), followed by another girl and another two boys. Her partner is unaffected.

b Assign genotypes to all individuals in the pedigree.

CONCLUSION

1 Describe the feature of a pedigree that establishes the mode of inheritance for a particular characteristic as:

a autosomal dominant

b autosomal recessive

c X-linked dominant

d X-linked recessive

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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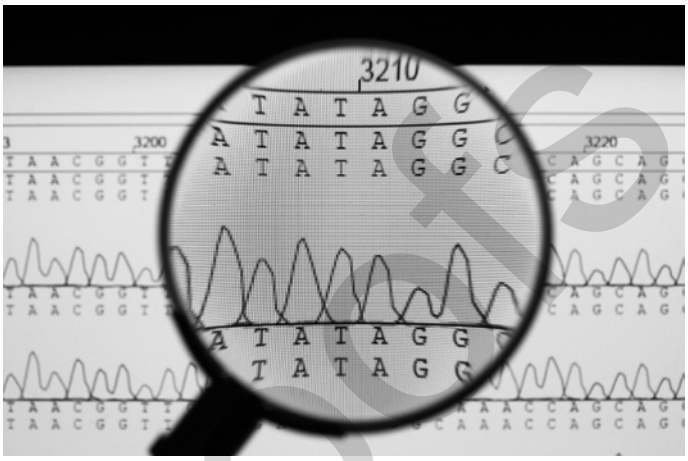
DEPTH STUDY 5.1

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 scientists 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 and 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 genomics 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:

- 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.

2 Account for the similarities in the genomes of Indigenous Australians and the Denisovans and Neanderthals.

3 On what basis have scientists arrived at the following conclusions?

a An ancestral population of Indigenous Australians and Papuans migrated from Africa around 72 000 years ago.

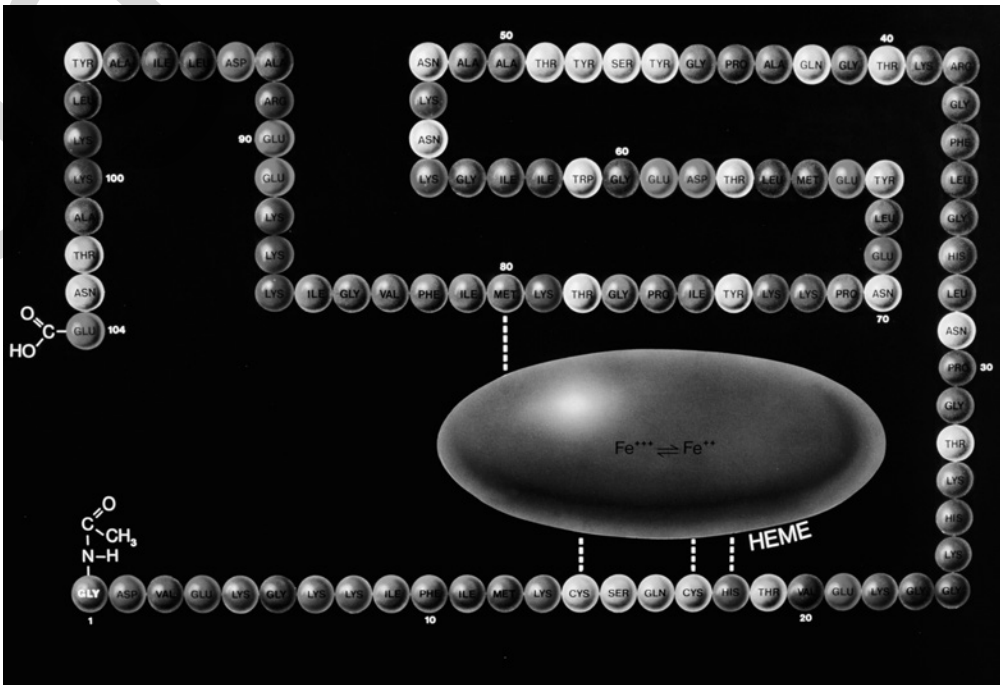
b Indigenous Australians and Papuans diverged approximately 37 000 years ago.

4 Suggest a reason to account for the significant genetic variation between Indigenous Australian populations in the east and west of the Australian continent.

Part B: Cytochrome c—a common evolutionary thread

BACKGROUND

Cytochrome c is a protein involved in ATP synthesis in the cells of organisms. ATP synthesis is a life-sustaining process in living things, so it makes sense that it is present in all organisms. As it is such a critical molecule for survival, cytochrome c is generally highly conserved. It is an excellent molecular subject for comparing the evolutionary relationships between organisms. The degree of similarity in cytochrome c molecules between different species is a useful indicator of how closely organisms are related. The greater the similarity in the cytochrome c molecule between species, the more closely related they are considered to be.



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Examine the cytochrome c data in Table 1 for three different kinds of organisms. Each of the letters corresponds to a particular amino acid in the cytochrome c molecule for each organism.

TABLE 1 Cytochrome c amino acid sequence from a section of the cytochrome c molecule

	98	60	61	62	63	64	65	66	100	101	102	103	104
Human	I	G	E	D	T	L	M	E	K	A	T	N	E
Frog	T	G	E	E	T	L	M	E	S	A	C	S	K
Rabbit	T	G	E	D	T	L	M	E	K	A	T	N	E

ANALYSING DATA AND INFORMATION

1 Use a highlighter pen to mark the points of difference the frog and rabbit display for the cytochrome c molecule compared with the humans. Record the number of similarities and difference in Table 2.

TABLE 2 Cytochrome c comparison with humans

	Same	Different
Frog		
Rabbit		

2 **a** What conclusion about evolutionary relationships between the human, frog and rabbit can you draw from the data? Explain how the data supports this.

b Represent the evolutionary relationships between the three organisms in a phylogenetic tree.

c Comment on whether the evolutionary relationship between the three organisms as suggested by the cytochrome c data is surprising or expected. Explain why you think so.

QUESTIONING AND PREDICTING

In this part of your depth study you will consider the evolutionary relationship between three different species of organisms.

Select three different species of organisms. For example:

- humans and two other primates
- humans, bacteria and another organism of your choice
- two similar species and one dissimilar, such as lion, tiger, ant or dolphin, shark, seal.

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Use resources such as the internet to find cytochrome c data for the selected species. It will be important that the data for the three species compares the same section of the cytochrome c molecule. Hint: Using carefully selected key terms will help to refine your search: for example, 'cytochrome c sequence for primates'.

3 Think about what it is you want to discover about the species. Formulate your question and write it down.

4 Think about how similar and how different the organisms appear to be. Make a statement predicting which species you expect to show the most similar cytochrome c amino acid sequence and which you expect to be the least similar. Explain why you think so.

COMMUNICATING

Prepare your final report. This will be a written report or digital presentation. Your submission should include:

a a relevant title

b an introduction—this will require a brief explanation about cytochrome c and its usefulness in this application

c identification of the species under investigation, providing both common and scientific names

d the question under investigation

e a prediction related to the expected evolutionary relationship between the three organisms

f a table clearly illustrating the cytochrome c amino acid sequence for each of the different species

g processing data and information—identify where the similarities and differences are for the different species and provide a summary of the information, for example in a table

h analysing data and information—explain what the data means in terms of the evolutionary relationships between the species

i a phylogenetic tree to illustrate the evolutionary relationships between the different species

j conclusion—revisit the question and prediction formulated in questions 3 and 4; comment on whether the evolutionary relationship between the three organisms as suggested by the cytochrome c data is surprising or expected, and explain why you think so

k other discussion items—for example, the research may have raised other questions or issues of interest. You do not need to answer all of these, but do record them; they demonstrate a deeper level of thinking and inquiry.

Guidelines

i You will communicate your findings as a written report or a digital presentation. The following information provides a guide about aspects to consider and protocols to follow.
- Keep notes of your research. Writing these in your own words demonstrates your understanding of the information.
- Use appropriate terminology—this demonstrates your familiarity with the language of the subject and the topic.
- Define new terms for your audience in your own words.
- Use footnotes to reference factual information, scientific studies and statistical data you include.
- Prepare a list of references according to expected guidelines.

ii As a guide to length, a written report should be a maximum of 800 words, not including your references. A digital presentation using media presentation software should be a maximum of 12 slides, including the title slide and references.

iii The Toolkit pages about Depth Studies will be helpful in working through this task. GO TO ➤ page xvii

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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:

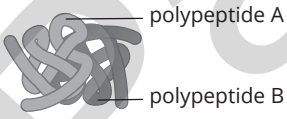
A is a unit of heredity

B may have alternate forms called alleles

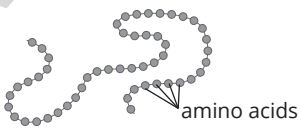
C regulates protein production in cells

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:



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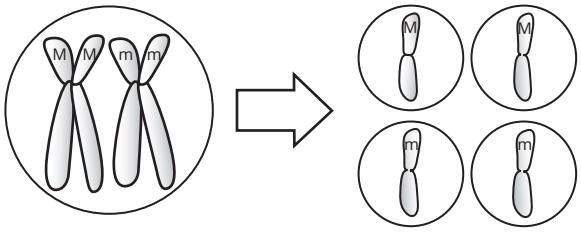
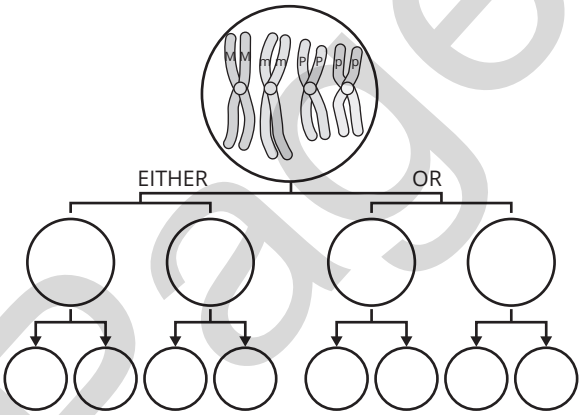
8 The variation in the different kinds of gametes produced during meiosis is in part due to the behaviour of chromosomes during gamete formation. Mendel's principle of segregation and independent assortment represent two ways in which chromosomes behave that contribute to this variation.

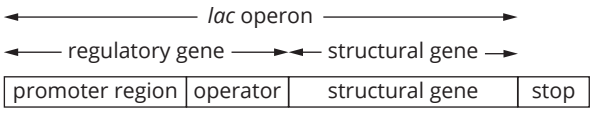
a The following diagram illustrates the law of segregation. Explain what is meant by the law of segregation.



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. Use the boxed space to explain what is meant by the law of independent assortment.



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



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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10 Newborns in Australia are routinely tested for a range of genetic diseases. One of these conditions is called phenylketonuria (PKU). This metabolic disorder is due to a faulty gene that means affected individuals do not manufacture the enzyme phenylalanine hydroxylase (PAH). PAH plays a critical role in converting the amino acid phenylalanine to tyrosine. In the absence of the enzyme, tyrosine is not formed. Instead, phenylalanine accumulates in the blood and tissues of the body. Too much phenylalanine interferes with the normal development from birth to adolescence, resulting in brain damage. Since the 1960s, all babies in Australia are screened for PKU a few days after birth using the Guthrie test, more commonly called the 'heel prick' test. Infants diagnosed with PKU are placed on a special diet free of phenylalanine, usually for the rest of their lives, and most importantly throughout childhood to puberty, when brain development is still occurring. This means a strictly vegan diet—no animal products, including meat, milk, butter, yoghurt, cheese or eggs, as these are sources of phenylalanine. PKU patients generally rely on specially prepared foods to ensure their diet contains no phenylalanine. Bioinformatics databases represent an important tool for data related to PKU and reveal the following information:

- Approximately one in 12 000 Australians are diagnosed with PKU each year.
- Since the introduction of the Guthrie test in the 1960s, affected children are identified and treated immediately, reducing detrimental effects and improving long-term health outcomes.

a Define the term 'bioinformatics'.

b Outline one advantage of the use of bioinformatics in the context of PKU.

c Discuss the significance of newborn screening programs for genetic diseases such as for PKU in Australia. Include a comment on the advantages and disadvantages of such screening programs.