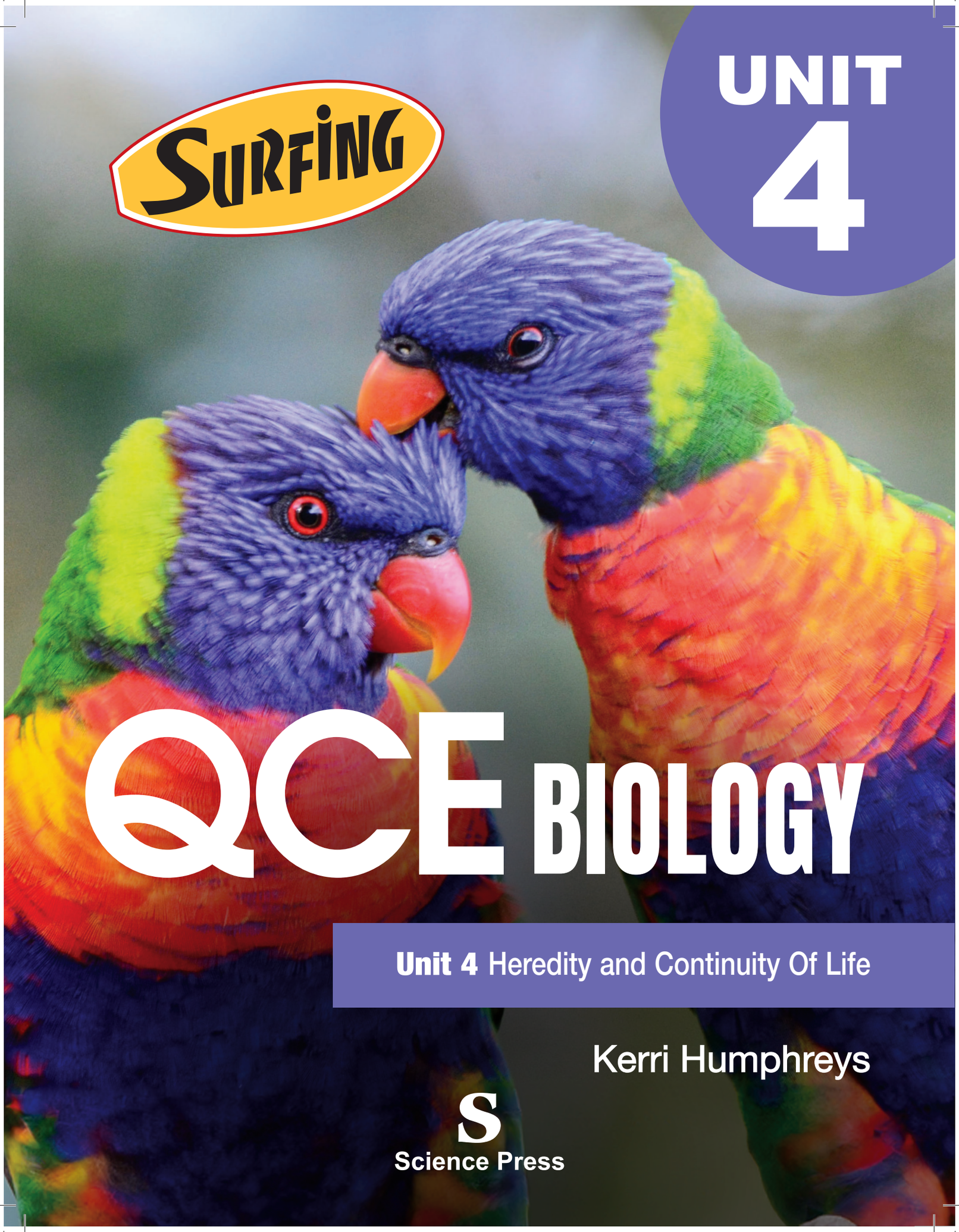
SURFING
UNIT
4
QCE BIOLOGY
Unit 4 Heredity and Continuity Of Life
Kerri Humphreys
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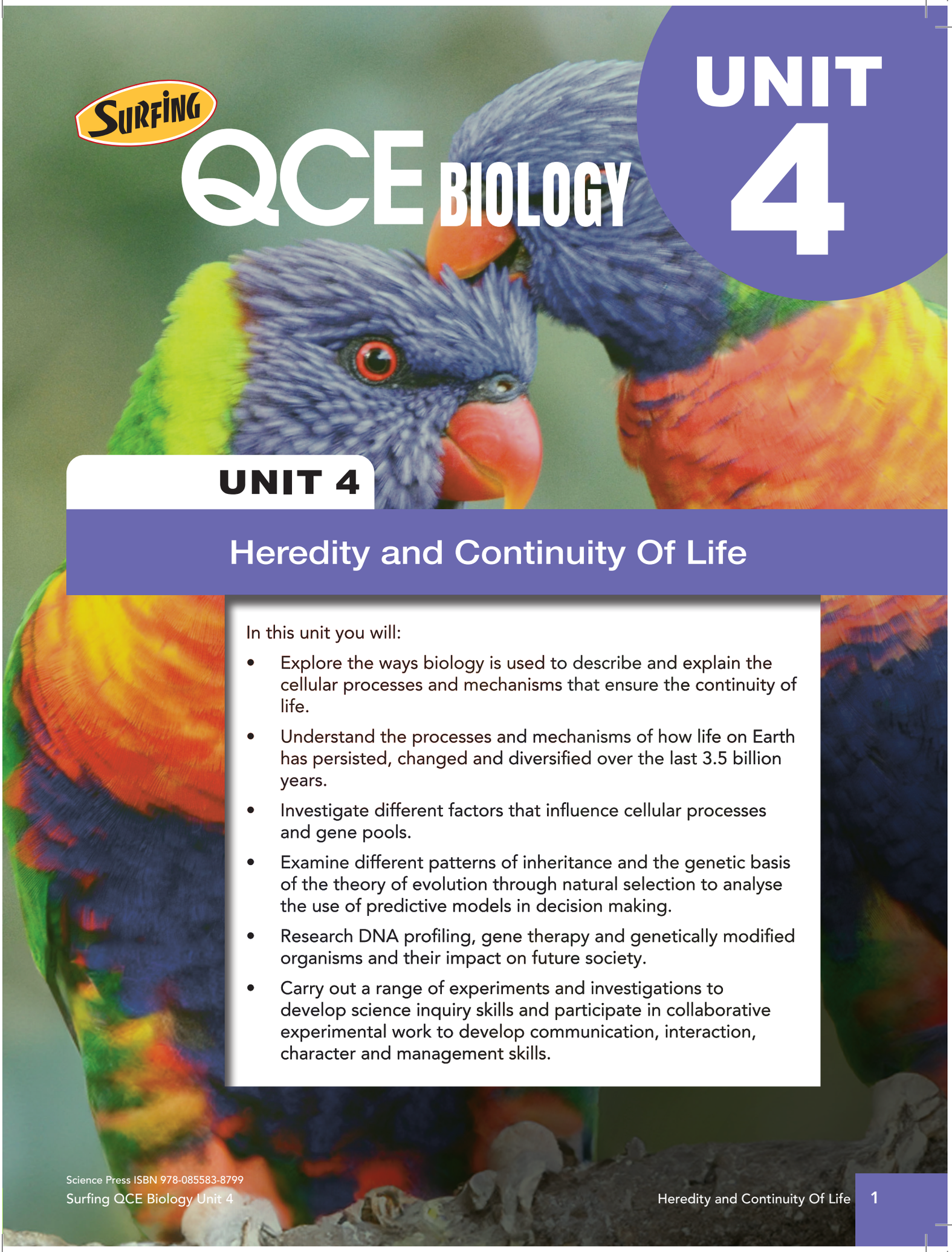

SURFING

QCE BIOLOGY

UNIT 4

UNIT 4

Heredity and Continuity Of Life

In this unit you will:

- Explore the ways biology is used to describe and explain the cellular processes and mechanisms that ensure the continuity of life.
- Understand the processes and mechanisms of how life on Earth has persisted, changed and diversified over the last 3.5 billion years.
- Investigate different factors that influence cellular processes and gene pools.
- Examine different patterns of inheritance and the genetic basis of the theory of evolution through natural selection to analyse the use of predictive models in decision making.
- Research DNA profiling, gene therapy and genetically modified organisms and their impact on future society.
- Carry out a range of experiments and investigations to develop science inquiry skills and participate in collaborative experimental work to develop communication, interaction, character and management skills.

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SURFING

QCE BIOLOGY

UNIT 4

TOPIC 4.1

Genetics and Heredity

In this topic you will:

- Describe and explain DNA structure and replication.
- Identify the processes in meiosis and explain errors in meiosis.
- Explain how variation occurs and use human karyotypes to predict genetic disorders.
- Explain the process of protein synthesis.
- Explain how mutations occur and alter genotype.
- Explain how gene expression is regulated in response to environmental signals.
- Explain how genes from the HOX transcription factor regulate morphology.
- Use probability models for inheritance problems.
- Infer patterns of inheritance and predict genotype frequencies.
- Explain the polymerase chain reaction and gel electrophoresis and their use in practical applications.

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1 Assumed Knowledge Topic 4.1

QUESTIONS

1. Define mitosis.
2. The diagram shows the last division of meiosis in the anther of a flower.
   (a) What is meiosis?
   (b) What would be produced, in this diagram?

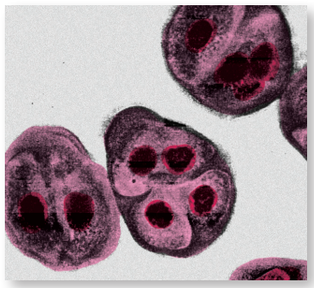

Figure 1.1 Meiosis in an anther.

3. The diagram shows a type of human cell.

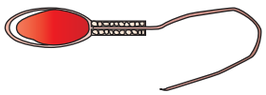

Figure 1.2 Type of human cell.

What is the name of this cell and what is its function?

4. Define genome.
5. What is a chromosome?
6. What is meant by genotype?
7. Define fertilisation.
8. Why is it important for gametes to have half the number of chromosomes of the species?
9. How is information transferred when cells reproduce themselves?
10. What does DNA stand for?

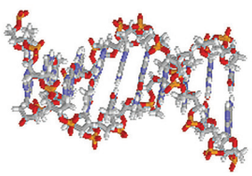

Figure 1.3 DNA.

11. Name the basic unit of DNA.
12. Where is DNA located in cells?
13. Outline the structure of the DNA molecule.
14. What is the relationship between genes and DNA?
15. Explain the advantages of DNA replicating exactly.
16. Why is Gregor Mendel often referred to as the 'father of genetics'?
17. Identify the factors that determine the features of an organism.
18. Use an example to show how environment influences the appearance of an organism.
19. Use an example to show how genes determine the features of an organism.
20. What is the 'Watson-Crick' model of DNA?
21. Distinguish between autosomes and sex chromosomes.
22. The diagram shows the karyogram for person A.

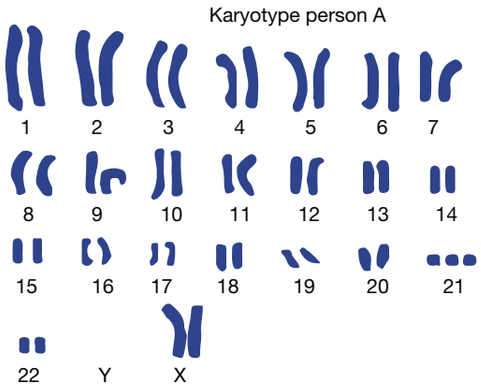

Figure 1.4 Karyogram.

   (a) Is this person male or female?
   (b) Define trisomy.
   (c) Person A has a trisomy disorder. Name this disorder.

23. What is meant by the genome sequence?
24. Define a gene pool.
25. What is meant by allele frequency?
26. Define polyploidy.
27. What is a mutation?
28. What is comparative genomics?
29. What is a mutagen?
30. The diagram shows the electromagnetic spectrum.

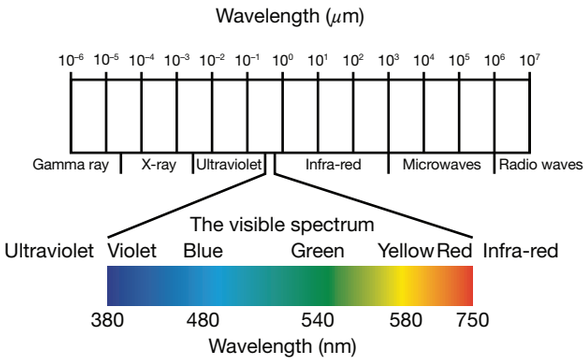

Figure 1.5 Electromagnetic spectrum.

Which parts of the electromagnetic spectrum involve ionising radiation that causes mutation?

31. Define biotechnology.
32. Distinguish between a somatic cell and a germ line cell.
33. Distinguish between introns and exons.
34. Identify four causes of genetic variation.

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2 Prokaryote and Eukaryote DNA

The genome of prokaryotes is structurally different to the genome of eukaryotes with the amount of DNA in a prokaryote being considerably less than the amount of DNA in an eukaryote. The process of DNA replication is fundamentally the same in prokaryotes and eukaryotes.

Prokaryote DNA

Most prokaryotic DNA is in circular ring in the nucleoid region of the cytoplasm. They do not have the histone protein framework of eukaryote cells. Many prokaryotes also have **plasmids** which a smaller rings of DNA that contain a limited number of genes.

Prokaryote ribosomes are slightly smaller than eukaryote ribosomes and have different protein and RNA.

In DNA replication the process starts at the origin of replication site and continues from the two forks at each end of the replication bubble. There are no telomeres as prokaryote DNA is circular with no ends that can be eroded with each replication process.

When the DNA code is used in protein synthesis the process of translation starts while transcription is still in progress. When the protein has been synthesised it quickly diffuses to its site of function.

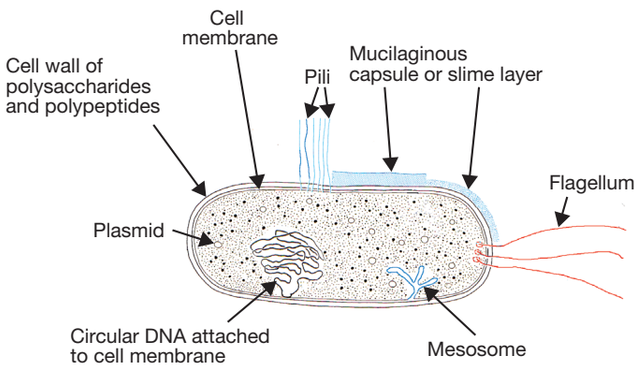

Figure 2.1 Typical prokaryote cell.

Eukaryote DNA

The chromosomes containing DNA of eukaryotes are found inside the nuclear membrane in the nucleus and are packaged with large amounts of protein with about one-half of the protein present being histone. The DNA-protein complex is called **chromatin**. Plasmids are found in some eukaryotes, e.g. yeast.

In eukaryotes DNA replication is initiated at many points along the chromosomes. There is also a framework of fibres throughout the nucleus that anchor the parental strands of DNA during the DNA replication process.

Telomeres are protective structures with repetitive codes that are found at the ends of eukaryotic chromosomes. The nucleotide sequence in telomeres is not a gene code but multiple repetitions of a short sequence that is eroded with each replication. Since eukaryotic DNA is in linear chromosomes and DNA polymerase can only add to the 3′ end of an existing chain, each replication involves the erosion of the 5′ ends of the daughter DNA strand. Telomeres act as a buffer and increase the time before genes near the ends of DNA molecules start to be eroded.

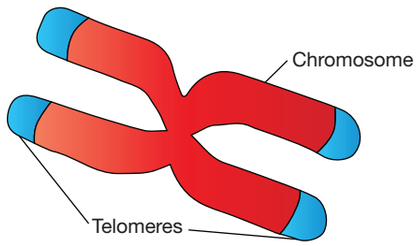

Figure 2.2 Telomere.

When the DNA code is used in protein synthesis transcription occurs in the nucleus where the code is transcribed onto mRNA. The mRNA then has to leave the nucleus and go the cytoplasm where translation occurs. While in the nucleus there is extensive RNA processing with RNA splicing removing **introns** which are long noncoding stretches of nucleotides.

QUESTIONS

1. Construct a table to compare the DNA of prokaryotes and eukaryotes.
2. What is a plasmid?
3. (a) What is a telomere?
   (b) Why are telomeres important?
   (c) Why are telomeres needed in eukaryotes and are not needed in prokaryotes?
4. Define chromatin.
5. In which type of cells would you *least* expect to find plasmids?
   (A) Cyanobacteria.
   (B) Methanogens.
   (C) Yeast.
   (D) Animal cells.
6. Which of the following correctly associates telomeres with their DNA code and type of cell in which they are found?

	DNA code	Type of cell
(A)	Short repeating nucleotide sequence	Eukaryote
(B)	Short repeating nucleotide sequence	Prokaryote
(C)	Gene code for a polypeptide production	Eukaryote
(D)	Gene code for a polypeptide production	Prokaryote

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5 Activity – Making a Model Of DNA

In eukaryotic cells a chromosome is one extremely long linear DNA molecule. In prokaryotic cells there is a circular DNA molecule.

There are many ways you can construct a model to show the structure of DNA. Models can be made of paper, cardboard, pipe cleaners, plastic sticks, coloured balls or created using multimedia graphics. The model needs to be able to show the triplet code of DNA which is the basic instruction of the genetic code.

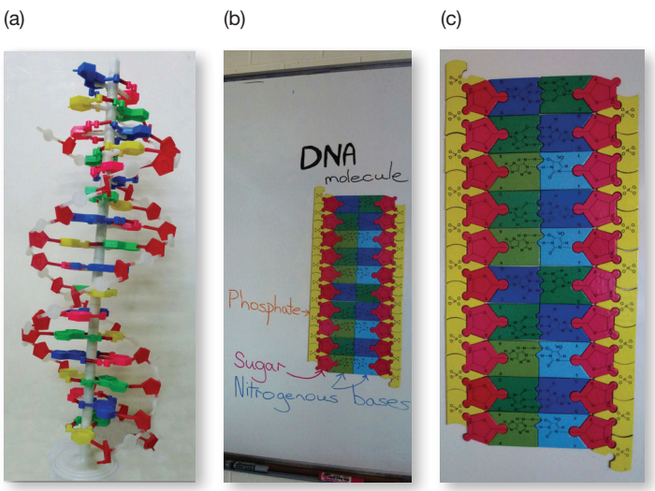

Figure 5.1 (a) Three-dimensional model of the DNA molecule. (b) Building a two-dimensional model of DNA on a classroom whiteboard using cardboard cut-outs that have magnetic strips which stick to the whiteboard. (c) Two-dimensional jigsaw model of DNA using a purchased DNA kit.

The **triplet code** is three bases next to each other in a molecule of DNA and specifies a particular amino acid. Reading the sequence of nucleotides in the triplet code in DNA means that you are reading the sequence of amino acids which will be joined together to form a polypeptide. There are three codes that are 'stop' signals which stop the process of gene expression and the translation of the code into a polypeptide. The DNA codes ATT, ATC and ACT signal 'stop'. The DNA code TAC is the code for the amino acid methionine but also acts as a 'start' signal for translation. Note that these start and stop codes are usually given as the mRNA code (e.g. stop codes UAA, UAG and UGA) as it is the **mRNA code** that is used in definitions and used to identify different amino acids. The mRNA code is complementary to the DNA code with uracil substituting for thymine. In gene expression the mRNA code is read in the cytoplasm when the code is translated to produce a polypeptide.

In gene expression when the DNA code is translated from the mRNA code into a polypeptide it is important to read the three nucleotide bases of an amino acid in the correct **reading frame**. Any alteration of the code will affect the ability to make a particular polypeptide.

Thus it is important when making a model of DNA to know what the code means when it is translated to produce a polypeptide.

Three-dimensional models

You can use molecular model kits with sticks and balls and make a model similar to the one built by James Watson and Francis Crick. A model using plastic sticks, coloured spheres and/or pipe cleaners has the benefit that it gives a three-dimensional vision of DNA to show the double helix structure.

Another way of showing this three-dimensional structure is to make an 'edible' DNA, e.g. using different coloured jellybeans to represent the nitrogenous bases, the phosphate and the sugar. The jellybeans can be joined with toothpicks to represent the bonds holding the nucleotides together.

Two-dimensional models

Many models of DNA use jigsaw cut-outs of coloured paper or cardboard and align the pieces to give a two-dimensional view of the ladder and rung structure of DNA. There are DNA jigsaw kits that can be purchased to use in class or you can create your own jigsaw. The jigsaw pieces can also be used to show the process of DNA replication and protein synthesis.

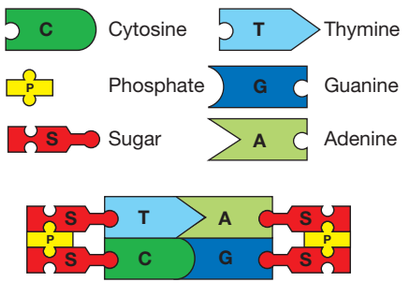

Figure 5.2 DNA jigsaw.

Evaluating a model of DNA

When evaluating a model the model must be consistent with known information. For example, one nucleotide of DNA must contain one phosphate, one deoxyribose sugar and one nitrogenous base, e.g. adenine (A), thymine (T), cytosine (C) or guanine (G). The nitrogenous bases must also be in complementary pairs, e.g. A-T and C-G.

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7 Discovering Telomeres

The discovery of DNA, its structural properties and its functionality involved the collaboration of people from a range of disciplines and continues to develop with the discovery of new evidence.

Telomeres are structures made of DNA sequences at the end of a chromosome that maintain chromosome stability. The length of telomeres varies in different types of human cells, e.g. longest in muscle and shortest in leucocytes.

Elizabeth Helen Blackburn, Jack William Szostak and Carol Widney Greider

Elizabeth Blackburn (1948-) was born in Tasmania and after receiving her PhD at the University of Cambridge in 1975 in biochemistry, she began researching the protozoan *Tetrahymena thermophilia*. In 1980 she noticed a repeating codon at the end of a linear rDNA that varied in size – she had discovered telomeres. The number of repeats can vary.

With her colleague Jack Szostak (1952-), she proved in 1982 that the DNA telomeric repeats protected the chromosome from degrading. This is important as when the DNA of linear chromosomes replicates, there is an **end replication problem** due to the nature of the polymerases and one strand is not fully replicated. This means a DNA fragment is lost and the chromosome shortens. The telomere cap protects gene information that is needed for the cell to survive from degrading.

Continued study with her PhD student Carol Greider (1961-) showed that an enzyme, they named **telomerase**, adds base pairs to the 3' end of DNA conserving the code lost by cellular division and prevents chromosomes shortening over time. This limits the cellular ageing of cells.

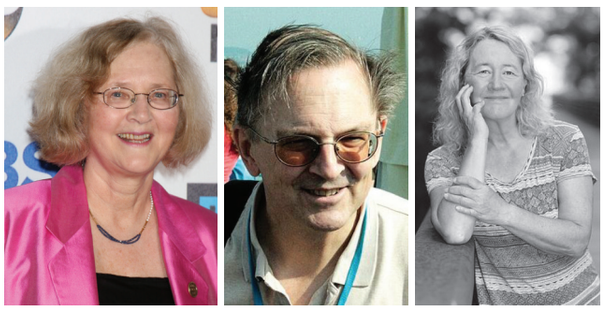

Figure 7.1 Elizabeth Blackburn (left), Jack Szostak (middle), Carol Greider (right).

Jack Szostak was born in London, UK and grew up in Canada. He completed his PhD in biochemistry at Cornell University. He collaborated with Elizabeth Blackburn at Havard Medical School in experiments on ciliates and yeast and helped prove that telomeres' DNA prevents chromosomes from being broken down.

Carol Greider was born in California and received her PhD from Berkeley in 1987 under Elizabeth Blackburn. In 1984, with Elizabeth Blackburn she discovered telomerase which produces the telomeres' DNA.

In 2009 Elizabeth Blackburn, Carol Greider and Jack Szostak were awarded the Nobel Prize in Physiology or Medicine. For their work on telomeres, telomerase and its impact on the understanding of cellular division. Elizabeth Blackburn was the first Australian female to become a Nobel laureate.

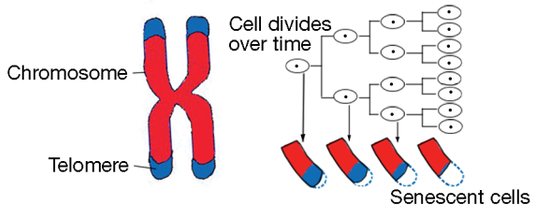

Figure 7.2 Telomeres shorten with each cell division

James Watson and Francis Crick

In 1953, James Watson (1928-2025) and Francis Crick (1916-2004) suggested the double helix structure of DNA with two phosphate sugar strands winding around the outside of nitrogenous bases with the pairs A-T and C-G. Watson and Crick based this structure on X-ray diffraction photographs of DNA taken by Rosalind Franklin. Maurice Wilkins was Franklin's colleague and showed her X-ray photographs to Watson. From the photographs, Watson and Crick were able to work out the distance between atoms, the angles between bonds and the size of atoms. They could then construct a three-dimensional model of DNA.

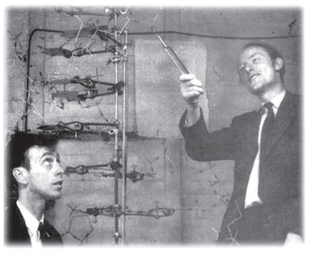

Figure 7.3 Watson and Crick.

QUESTIONS

1. (a) What is a telomere?
   (b) Who discovered telomeres?
   (c) Why are telomeres sometimes compared to an aglet – the small sheath at the end of a shoelace?
2. (a) What was discovered by Elizabeth Blackburn working in collaboration with Jack Szostak?
   (b) What is the significance of this discovery for chromosome stability?

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10 DNA Replication and Continuity Of a Species

The coding in the DNA molecule provides all the necessary information to build structural and functional cell components. DNA replication allows the coding to be passed from one generation to the next. The replication is **semi-conservative** with each new DNA molecule consisting of one DNA strand from the template and one newly synthesised DNA strand.

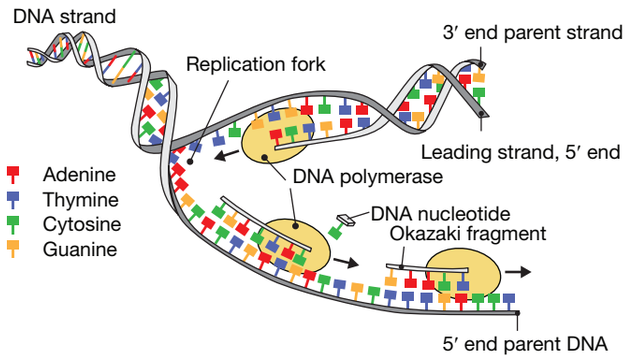

Figure 10.1 DNA replication.

Universality of DNA replication

Many features of DNA replication are universal, e.g. the nitrogenous base pair coding not only holds the genetic information that will determine the features of a particular species but the adenine-thymine and cytosine-guanine pairing is found across all species.

Since the 'backbone' of the DNA molecule consists of covalently bonded 3′ carbon of one sugar to the 5′ phosphate of the adjacent sugar it is possible to have molecules of indefinite length. Though there are some differences between prokaryotic DNA which is a circular molecule and eukaryotic DNA which is linear with a large amount of associated protein and some RNA.

Table 10.1 Diploid number of different species.

Species	Scientific name	Chromosomes (2*n*)
Fruit fly	*Drosophila melanogaster*	8
Red kangaroo	*Macropus rufus*	20
Cane toad	*Rhinella marina*	22
Wollemi pine	*Wollemia nobilis*	26
Bread wheat	*Triticum aestivum*	42
Human	*Homo sapiens*	46
Chimpanzee	*Pan troglodytes*	48
Orangutan	*Pongo pygmaeus*	48
Western gorilla	*Gorilla gorilla*	48
Dog	*Canis lupus familiaris*	78
Great white shark	*Carcharodon carcharias*	82

Continuity and evolution

The complementary base pairing allows DNA replication to produce another generation with the continuity of life and continuity of a species. The exact replication of the code is needed in binary fission, e.g. for unicellular organisms and for meiosis in eukaryotes for gamete formation.

In DNA replication there is also the possibility of mutation, e.g. base substitution, deletion, insertion, duplication, translocation so that variations can arise leading to evolution and speciation. For example, there is some evidence that human chromosome 2 formed by the fusion of two ancestral chromosomes telomere to telomere leading to humans having 46 chromosomes while the great apes have 48 chromosomes. Though some people propose that chromosome 2 split into two in the great apes making them 48 chromosomes from 46.

QUESTIONS

1. What information is stored in the DNA code?
2. Explain why DNA replication is semi-conservative.
3. Identify a feature of DNA replication that is universal.
4. Explain why the length of a DNA is not limited.
5. The diagram shows a section of DNA replication.

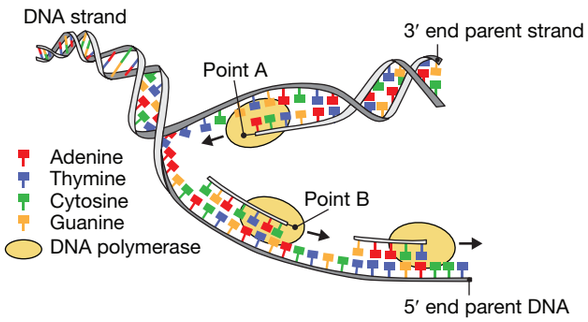

Figure 10.2 Section of DNA replicating.

   (a) Name the nucleotide that will be added at point A and the nucleotide that will be added at point B.
   (b) On the diagram identify which is the leading strand and which is the lagging strand in DNA replication.

6. How is prokaryotic DNA different to eukaryotic DNA?
7. Use some examples to show that different species have different numbers of chromosomes.
8. Use an example to show that different species can have the same number of chromosomes.
9. Explain why DNA is important for the continuity of a species.
10. Explain how DNA replication is involved in evolution and speciation.
11. Use an example where evolution has involved a change in chromosome number.

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QUESTIONS

1. Define meiosis.
2. How many daughter cells are produced in meiosis?
3. Where does meiosis occur?
4. Describe the daughter cells produced by meiosis.
5. What are homologous chromosomes?
6. State the law of independent assortment.
7. What are linked genes?
8. Identify how linked genes can be inherited independently of each other and become part of different gametes.
9. State the law of segregation.
10. Draw a flow chart to show the process of meiosis.
11. The diagram shows a cross section of a plant anther using low power of a light microscope.

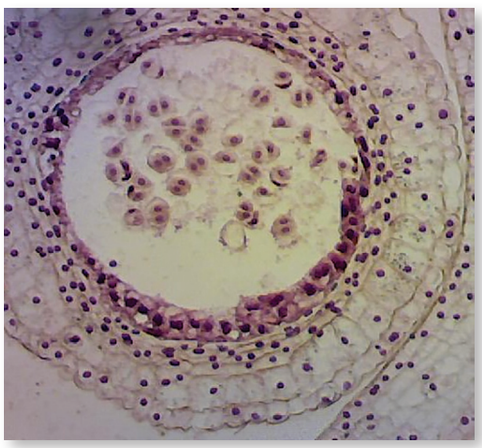
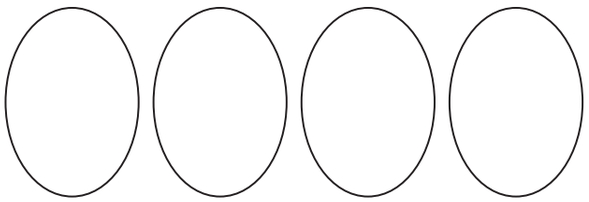


Figure 14.2 LP Plant anther.

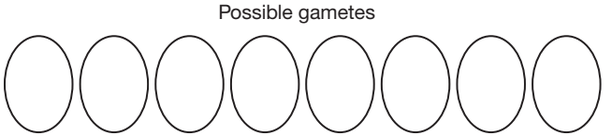
Explain why an anther was chosen to study the stages of meiosis and identify the stage in the diagram.

12. The diagram shows a cell with $2n = 6$. Copy the diagram and fill in the possible cell alignments for metaphase I and the possible gametes (no crossing over).

Possible alignments in metaphase I



Possible gametes



13. The diagrams show sections of anther with different stages of meiosis.

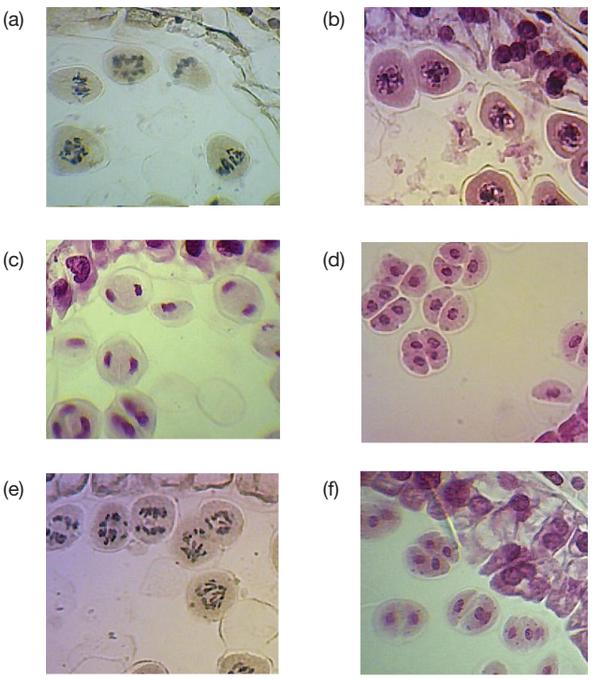
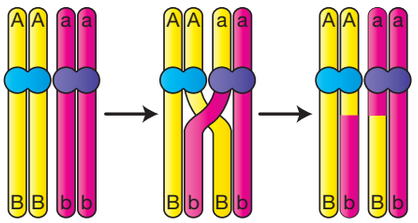


Figure 14.3 Different stages of meiosis.

Identify what is happening in each diagram.

14. The diagram shows crossing over with a pair of homologous chromosomes. Alleles A, a, B and b are indicated on each chromosome.



What are the genotypes of the daughter cells?

(A) AA, aa, BB, bb
(B) AB, Ab, aB, ab
(C) AB, AB, ab, ab
(D) AA, AB, bb, ab

15. In which stage of meiosis would you expect to see the homologous chromosomes separated and moving to opposite poles?

(A) Prophase I.
(B) Anaphase I.
(C) Prophase II.
(D) Anaphase II.

16. Which of the following occurs in meiosis but not in mitosis?

(A) Homologous chromosomes align.
(B) Chromatids line up at the equator.
(C) Spindle fibres move chromosomes to the poles.
(D) Chromatids joined at centromere.

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There is a 'lock and key' recognition of the protein molecules on the end of the tip of the acrosome and the cell membrane of the egg.

Fertilisation in plants

In the **seedless plants**, e.g. mosses, liverworts and ferns a flagellated sperm is released and it needs to swim to the egg cell. This is why these groups of plants need to live in moist areas.

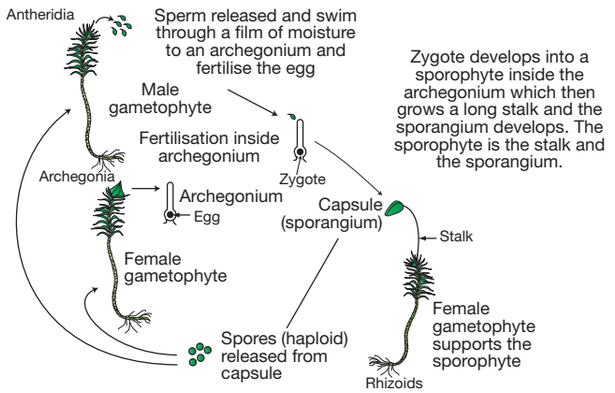

Figure 18.4 Fertilisation in moss.

In **seed plants**, e.g. gymnosperms and angiosperms pollen grains are released and can travel long distances by wind or pollinators to reach the egg. The sperm of seed plants do not need mobility and do not need water to reach the egg. Without the need for water for fertilisation the seed plants can live in a broader range of conditions.

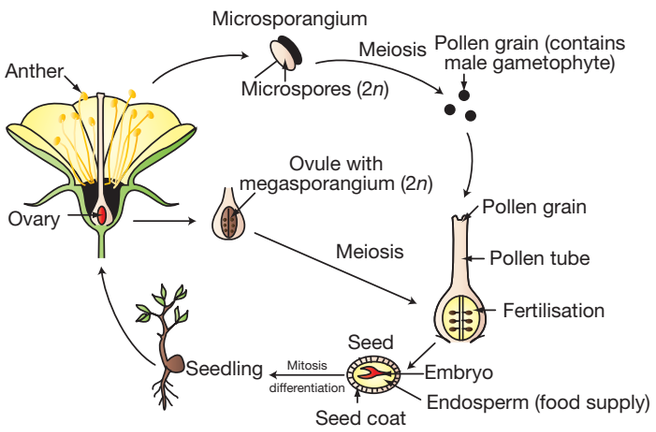

Figure 18.5 Fertilisation in flowers.

The **gymnosperms** have 'naked seeds' that are not in ovaries. The seeds are exposed on modified leaves, e.g. in conifers the leaves form cones, e.g. Tasmanian cedars (*Athrotaxis* spp), cypress pines (*Callitris* spp), kauri pines (*Agathis* spp), and plum pines (*Podocarpus* spp). When pollen grains land on an ovule they enter the ovule through the micropyle. A **micropyle** is a small opening in the ovule of a seed plant through which the pollen enters. Fertilisation occurs within the ovule.

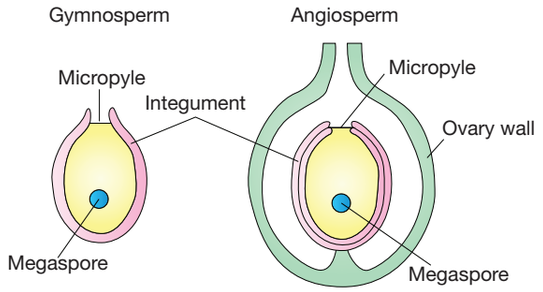

Figure 18.6 Micropyle in seed plants.

The **angiosperms** are the flowering plants. The flower is a specialised structure for sexual reproduction. When pollen lands on the stigma a pollen tube grows down the style to the ovary and enters the ovule through the micropyle. The pollen tube releases two sperm cells into the embryo sac of the ovule and there is **double fertilisation**. One sperm fertilises the egg and the other sperm fuses with the large central cell to become the **endosperm** which has the food reserves for the developing embryo.

QUESTIONS

1. Define fertilisation.
2. What is a gamete?
3. Define zygote.
4. Explain why fertilisation maintains the diploid number of a species.
5. Distinguish between internal fertilisation and external fertilisation.
6. Explain why fertilisation produces variation.
7. Explain why variation is needed in a population.
8. In the vertebrates identify which groups use external fertilisation and which groups use internal fertilisation.
9. Define copulation.
10. What is an intromittent organ?
11. (a) What is the acrosome?
    (b) What is the acrosomal reaction?
12. (a) What are the seedless plant?
    (b) How does fertilisation occur in the seedless plants?
13. What are the seed plants?
14. Explain why seed plants are found in a broader range of environments than seedless plants.
15. Name some Australian conifers.
16. What is the micropyle?
17. What is a flower?
18. How does the pollen of an angiosperm reach the stigma?
19. Explain why there is double fertilisation in angiosperms.
20. Explain why internal fertilisation is more advantageous than external fertilisation?
21. Many aquatic animals, e.g. fish have spawning grounds where females release eggs and males release sperm. Outline the advantage of this strategy.

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23 Chromosome Abnormalities

There are several types of chromosome abnormalities. Some individuals can have extra or missing chromosomes and some individuals have a different number of whole sets of chromosomes.

Polyploidy is the possession of more than two sets of chromosomes per nucleus, e.g. hexaploid (6*n*).

Monoploidy is the loss of an entire set of chromosomes. It is the haploid number of chromosomes.

Aneuploidy is the addition of all or part of a chromosome, e.g. in humans there could be 45 or 47 chromosomes instead of the normal 46 chromosomes in the nucleus.

Monosomy is the lack of one chromosome from the normal number of chromosomes.

Trisomy means there are three copies instead of the normal two copies of a particular chromosome.

Non-disjunction

The change in the number of chromosomes can be due to non-disjunction during cell division. Non-disjunction is an error during mitosis or meiosis when both members of a pair of homologous chromosomes or both sister chromatids fail to separate properly.

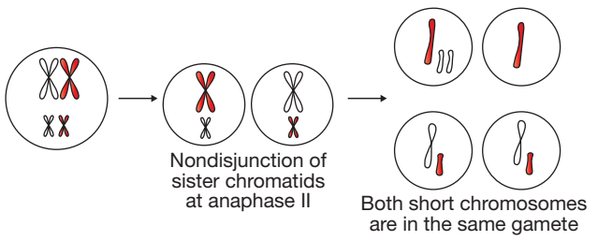

Figure 23.1 Non-disjunction in anaphase II in meiosis.

Non-disjunction leads to an abnormal number of chromosomes in a gamete. If it occurs in anaphase II there will be two affected daughter cells – one with two copies of the chromosome and the other with no copies of that chromosome.

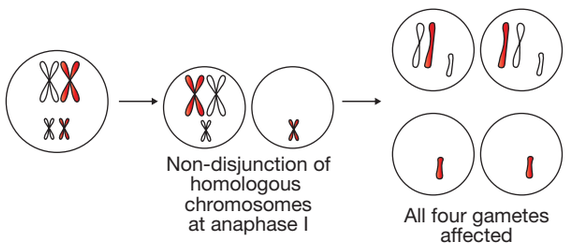

Figure 23.2 Non-disjunction in anaphase I in meiosis.

If the homologous chromosomes do not separate during anaphase I then all four daughter cells are affected – two daughter cells will have an additional chromosome and two cells will be missing one chromosome.

During fertilisation the fusion of an abnormal gamete with a normal gamete will create a zygote with an incorrect number of chromosomes, e.g. missing a chromosome or having an additional chromosome. Many cases of trisomy 21 (Down syndrome) are due to non-disjunction.

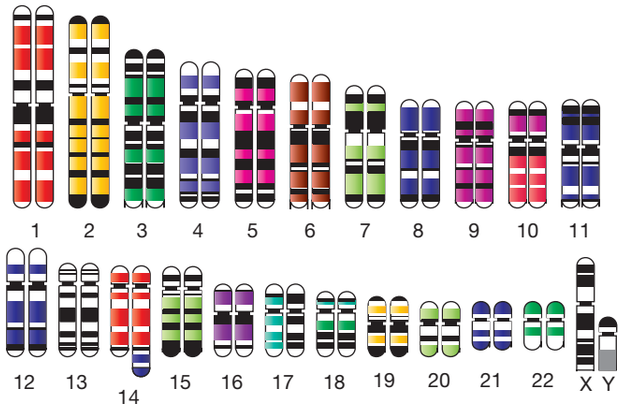

Figure 23.3 Trisomy 21 with translocation to chromosome 14.

Non-disjunction can also occur during mitosis. If the error occurs during early embryonic development then a large number of cells will have the aneuploidy condition. In mosaic Down syndrome the person has some cells with trisomy 21 and some cells with the normal 46 chromosomes in a cell.

Unequal crossing over in meiosis

Chromosome abnormalities can occur when crossing over between adjacent non-sister chromatids in the tetrad in prophase I if meiosis is unequal. In the resulting gametes one cell may have a chromosome with a gene deletion and another cell will have a chromosome with a gene duplication. Transposable elements in the chromosome provides the sites for crossing over in non-sister chromatids.

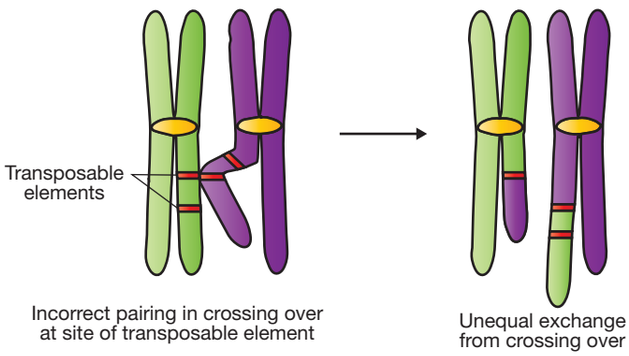

Figure 23.4 Unequal crossing over.

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26 Polypeptide Synthesis – Transcription

DNA stores genetic information. The sequence of bases determines the sequence of amino acids in protein molecules. The smallest unit that can code for amino acids is a **codon**, or triplet of bases. For example, the DNA code CCA codes for the amino acid glycine. Protein synthesis involves the **transcription** of the code from DNA to messenger ribonucleic acid (mRNA) and then the **translation** of the code from the mRNA to sequencing the amino acids to form a protein.

Scientific knowledge about the process of gene expression and polypeptide synthesis has rapidly increased as new research methods and developments in DNA technology have enabled biologists to investigate the structures and functions of molecules in cells.

The new technologies have shown how signals from outside the cell, e.g. hormones or other signalling molecules can start the process of polypeptide synthesis and gene expression.

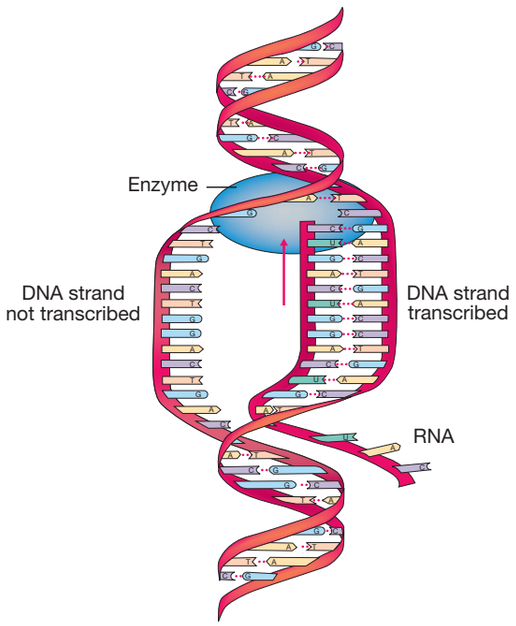

Figure 26.1 Transcription.

Pre-mRNA

The first step of polypeptide synthesis involves the unwinding of the DNA and one side, the **template strand**, is copied to form mRNA. RNA is similar to DNA except that it contains a **ribose** sugar instead of deoxyribose sugar, it is always single stranded and it is complementary with base pairs to the template strand except that it contains the nitrogenous base **uracil** instead of thymine. The genetic code is just about universal for living organisms from unicellular bacteria to multicellular plants and animals.

In eukaryotic cells the RNA coding transcript from the DNA coding gene is modified in RNA processing before the functional code leaves the nucleus. The initial transcript forms **pre-mRNA** (precursor messenger ribonucleic acid).

Prokaryotic cells do not have a nucleus so when transcription begins translation will start before transcription is completed. There is no RNA processing of bacterial mRNA before translation begins.

Transcription occurs in the 5′ → 3′ direction from the template strand and starts at a specific **promoter** base sequence of nucleotides that signal the beginning of the gene. Different genes have slightly different promoter sequences which means that different genes can be transcribed at the same time. The enzyme **RNA polymerase** causes the two strands of DNA to separate and then links the RNA nucleotides together to form mRNA.

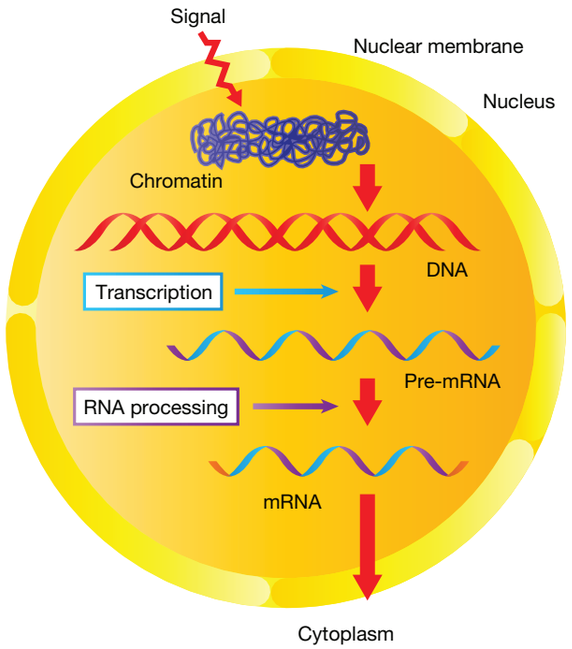

Figure 26.2 The first steps in gene expression.

RNA processing

RNA processing occurs in the nucleus of eukaryotes and involves several actions. Each end of the pre-mRNA is modified in a particular way to help protect the mRNA from being degraded by enzymes and to aid the movement of the functional mRNA to the cytoplasm.

The 5′ end of the mRNA chain is **capped** and at the 3′ end an enzyme adds a **poly-A tail** (polyadenylated tail). A poly-A tail is the addition of 50 to 250 adenine nucleotides to the end of the mRNA.

RNA processing also includes RNA splicing. During **RNA splicing** the noncoding introns are removed from the pre-mRNA and the exons are joined together.

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27 Discovering Introns and Exons

The discovery of DNA, its structural properties and its functionality involved the collaboration of people from a range of disciplines and continues to develop with the discovery of new evidence.

Introns and **exons** are parts of eukaryotic genes.

Introns are the non-coding sequences of DNA or RNA while **exons** are sequences of DNA or RNA that code for proteins.

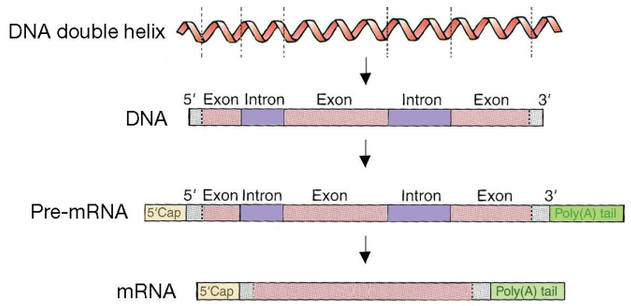

Figure 27.1 Linear eukaryotic DNA consists of introns and exons.

Richard J Roberts

Richard John Roberts (1943-) was born in England and after receiving his PhD at the University of Sheffield in 1969 in chemistry, he moved to the USA to become a research fellow at Harvard University. In 1972 he became a senior staff investigator at the Cold Spring Harbor laboratory in New York and then moved to New England Biolabs, MA, USA where he showed how RNA can be divided into introns and exons, after which the exons are joined together. As the process can occur in different ways, there is the potential for a gene to form a number of different proteins.

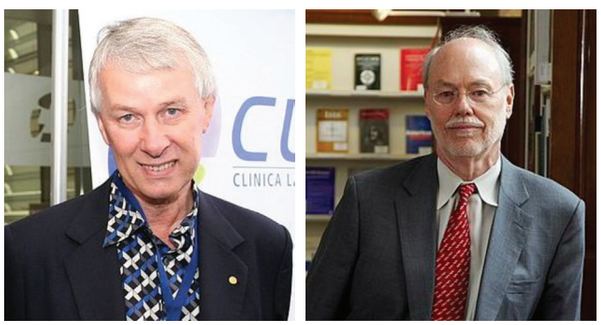

Figure 27.2 Richard Roberts (left) and Phillip Sharp (right).

Phillip A Sharp

Phillip Allen Sharp (1944-) was born in Kentucky, USA and completed his PhD in chemistry at the University of Illinois in 1969. In 1974 he moved to the Massachusetts Institute of Technology (MIT) where he noticed that there was not any long nuclear RNA. He speculated that nuclear RNA was processed into shorter mRNAs and carried out experiments that showed the mRNA of an adenovirus had four separate, discontinuous segments of DNA (now called exons).

Richard Roberts and Phillip Sharp shared the 1993 Nobel Prize in Physiology or Medicine for their independent discovery of split genes. Each announced his research results in 1977.

Further developments

The work of Roberts and Sharp laid the foundation for further investigation into gene expression and the function of introns.

The presence of several introns in a gene, means there can be alternative splicing in RNA processing, allowing one gene to code for several transcripts and be involved in several complex cellular functions. Introns also separate genes in DNA and have segments that act as transcription promoters and enhance gene expression.

QUESTIONS

1. (a) Distinguish between introns and exons.
   (b) Where are introns and exons found?
2. (a) What was discovered by Richard Roberts?
   (b) What was discovered by Phillip Sharp?
   (c) How were they rewarded?
   (d) What was the collaboration between Roberts and Sharp?
3. How has our knowledge of introns and exon developed since the initial discovery by Roberts and Sharp?
4. The diagram shows the initiation of transcription. The promoter is upstream from the transcription initiation site.

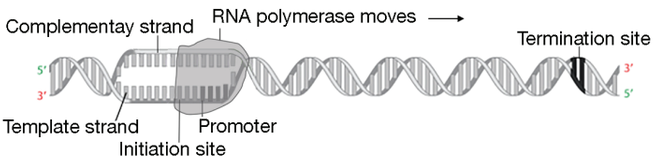

Figure 27.3 Initiation of transcription.

Which sections of the DNA code will be present in the mRNA when it leaves the nucleus?

(A) Introns. (B) Exons.
(C) Promoter. (D) Regulatory region.

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QUESTIONS

1. Define a genome.
2. Define a gene.
3. Distinguish between introns and exons.
4. What is the centromere?
5. (a) What is a telomere?
   (b) Explain why eukaryotic chromosomes need telomeres.
6. Explain why the term 'junk DNA' should not be used.
7. What is meant by gene expression?
8. (a) What are the two steps in protein synthesis and gene expression?
   (b) Where do these steps occur in eukaryotic cells?
9. What is the triplet code?
10. How do the base pairing rules apply to RNA?
11. The diagram shows the relationship between a gene and a chromosome.

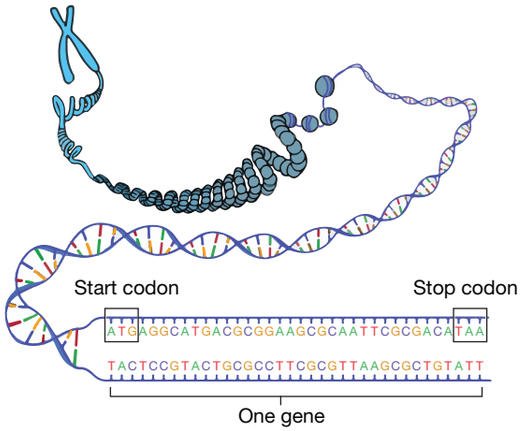

Figure 28.3 A gene and a chromosome.

From the diagram what is found at the beginning and at the end of a gene?

12. What is RNA processing?
13. Distinguish between pre-mRNA and mRNA.
14. Outline what happens in RNA processing.
15. The diagram compares DNA and RNA.

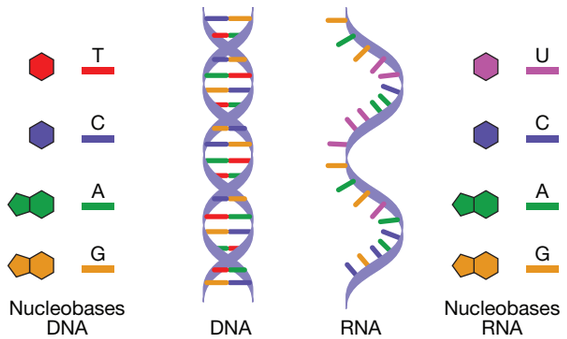

Figure 28.4 DNA and RNA.

Construct a table to compare DNA and RNA looking at the number of strands, nitrogenous bases and types.

16. The diagram shows a process that occurs in some types of cells.

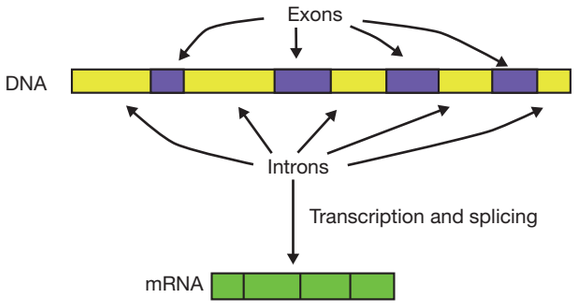

Figure 28.5 Exons and introns.

Where does this process occur?
(A) In prokaryote cells.
(B) In the nucleus of eukaryote cells.
(C) In the cytoplasm of eukaryote cells.
(D) In mitochondria.

17. What is the name of the sections of DNA that are not part of the functional code that is sent to the cytoplasm?
(A) Introns (B) Exons
(C) Extrons (D) Intrudons

18. The diagram shows a chromosome.

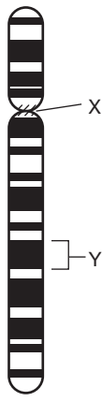

Figure 28.6 Chromosome.

Identify structures X and Y.

	Structure X	Structure Y
(A)	Telomere	Centromere
(B)	Centromere	Telomere
(C)	Centromere	Gene
(D)	Telomere	Gene

19. Which of the following is likely to have non-coding DNA?
(A) Insulin, myosin, haemoglobin.
(B) Haemoglobin, centromeres, introns.
(C) Telomeres, myosin, functional RNA.
(D) Functional RNA, centromeres, telomeres.

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30 Polypeptide Synthesis – Translation

Translation is a cellular process that uses the genetic code on mRNA to synthesise a polypeptide. The sequence of nucleotides is translated into a sequence of amino acids which are joined together to form the polypeptide.

Translation occurs in the cytoplasm on ribosomes. The translator is **tRNA** (transfer ribonucleic acid).

Translation occurs in three main stages – initiation, elongation and termination. In **initiation** the mRNA, tRNA with the first amino acid of the polypeptide that is to be formed and the two subunits of a ribosome are brought together to form the initiation complex. In **elongation** the amino acids are brought to the mRNA and added to the growing polypeptide chain. In **termination** a stop codon (e.g. UAG, UAA, UGA) in the mRNA is recognised by a protein release factor causing the polypeptide to be completed and released and the translation assembly comes apart.

UUU Phe	UCU Ser	UAU Tyr	UGU Cys
UUC Phe	UCC Ser	UAC Tyr	UGC Cys
UUA Leu	UCA Ser	UAA stop	UGA stop
UUG Leu	UCG Ser	UAG stop	UGG Trp
CUU Leu	CCU Pro	CAU His	CGU Arg
CUC Leu	CCC Pro	CAC His	CGC Arg
CUA Leu	CCA Pro	CAA Gln	CGA Arg
CUG Leu	CCG Pro	CAG Gln	CGG Arg
AUU He	ACU Thr	AAU Asn	AGU Ser
AUC He	ACC Thr	AAC Asn	AGC Ser
AUA He	ACA Thr	AAA Lys	AGA Arg
AUG Met	ACG Thr	AAG Lys	AGG Arg
GUU Val	GCU Ala	GAU Asp	GGU Gly
GUC Val	GCC Ala	GAC Asp	GGC Gly
GUA Val	GCA Ala	GAA Glu	GGA Gly
GUG Val	GCG Ala	GAG Glu	GGG Gly

U – uracil (thymine)	Cys – cysteine	Met – methionine
C – cytosine	Gln – glutamine	Phe – phenylalanine
A – adenine	Glu – glutamine	Pro – proline
G – guanine	Gly – glycine	Ser – serine
Ala – alanine	His – histidine	Thr – threonine
Arg – arginine	He – isoleucine	Trp – tryptophan
Asn – asparagine	Leu – leucine	Tyr – tyrosine
Asp – aspartic acid	Lys – lysine	Val – valine

Figure 30.1 The triplet genetic code found in mRNA.

tRNA

Each tRNA can translate a particular mRNA codon into a particular amino acid. One end of the tRNA has a nucleotide triplet called the **anticodon** which is complementary to the mRNA code. The other end of the tRNA carries the amino acid and takes it to the ribosome for translation. This gives the tRNA its distinctive shape which looks like a 3-leaf clover leaf when flattened to draw in two dimensions. In eukaryotes tRNA is made in the nucleus and moves to the cytoplasm.

The cytoplasm of a cell contains a supply of amino acids that are either synthesised from other compounds or brought into the cell for protein synthesis. The 3′ end of tRNA is the attachment site for the amino acid. There are 20 different amino acids and there are 20 different **synthetase** enzymes that link the correct amino acid to the tRNA with the correct anticodon. Each tRNA can be used many times – to pick up an amino acid, to deposit the amino acid onto the polypeptide chain at the ribosome and then to move away to pick up another amino acid.

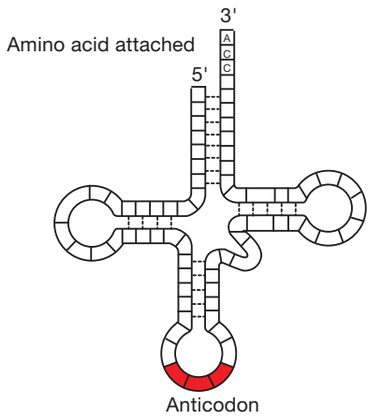

Figure 30.2 tRNA.

At the ribosome

In the cytoplasm the mRNA moves to a ribosome where it binds on to the ribosome at the end with the 'start' codon. The sequence of codons is translated into a sequence of amino acids that make up a particular polypeptide.

A transfer RNA (tRNA) in the cytoplasm moves to the codon on the mRNA which is at the ribosome. At one end of the tRNA there is an anticodon to complement the codon on the mRNA and on the other end of the tRNA is the amino acid.

Ribosomes are made up of two subunits which consist of proteins and ribosomal RNA (rRNA) with about two thirds of the mass of a ribosome consisting of rRNA. The large subunit binds with tRNA and the smaller subunit binds with mRNA.

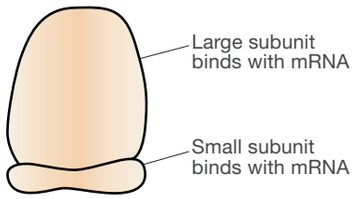

Figure 30.3 Structure of a ribosome.

A ribosome has *three* binding sites for tRNA. The **P site** holds the tRNA with the growing chain. The **A site** holds the tRNA carrying the amino acid about to be added to the chain. The **E site** has the exiting tRNA that has delivered its amino acid and is about to leave the ribosome.

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57 DNA Polymerase

DNA polymerases are enzymes that catalyse the elongation of new DNA at a replication fork by the addition of nucleotides to the existing chain. The DNA polymerase adds nucleotides one by one and the rate of elongation depends on the species, e.g. in human cells the rate is around 50 nucleotides per second while in bacteria it can be 500 nucleotides per second.

There are different DNA polymerases and different species have different numbers of DNA polymerases. For example, *E. coli* has two different DNA polymerases involved in replication – DNA polymerase III and DNA polymerase I. However, the structure of DNA polymerase is **highly conserved** so that the subunits and the functions they perform are similar from one species to another.

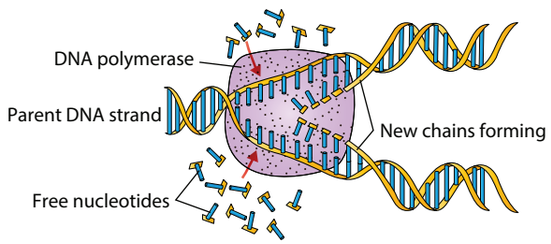


Figure 57.1 DNA polymerase is involved in DNA replication.

How it works

DNA polymerase 'reads' the existing DNA as a template code and adds free nucleotides to the 3′ end of a newly formed strand. It cannot begin the formation of a new chain and needs a **primer** to initiate replication. DNA polymerase usually works in pairs to create the two new DNA strands. It can only work in the 5′ → 3′ direction which means the two strands of the replication fork are copied in different ways. The **leading strand** is copied continuously in a direction towards the replication fork while the **lagging strand** is synthesised in Okazaki fragments which are short sections in a direction away from the replication fork.

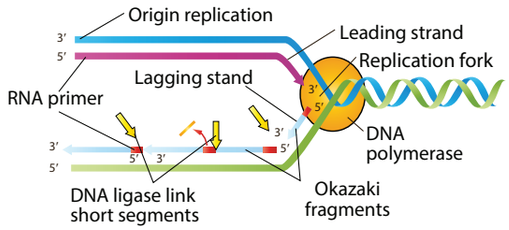


Figure 57.2 The two strands elongate in 5′ → 3′ direction.

As soon as a nucleotide is added to a strand, DNA polymerase will 'proofread' the code against the template checking for a mistake so that misplaced pairs can be corrected. If a mistake is found the enzyme will remove the nucleotide and then continue synthesis.

Because DNA polymerase only adds to the 3′ end of a pre-existing polynucleotide it means it cannot complete the 5′ ends of daughter strands where the primer was attached. This means that DNA in daughter cells gets shorter each time the DNA replicates and the cell divides.

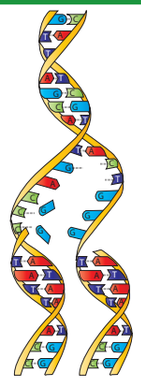
Diagram of DNA replication	What is happening at each stage
	DNA double helix.
	Double helix unwinds.
	Double helix unzips and two strands separate catalysed by the enzyme helicase.
	Free nucleotides from the cytoplasm join at the free sites as the helix unwinds. Adenine always pairs with thymine and guanine pairs with cytosine to maintain the sequence of nucleotides.
	DNA polymerase catalyses the formation of bonds to join the nucleotides to the DNA strand.
	Two identical strands of DNA are formed from the original strand.

Figure 57.3 DNA replication.

QUESTIONS

1. Define DNA polymerase.
2. How can the rate of elongation due to DNA polymerase vary?
3. DNA polymerases are highly conserved. What does this mean?
4. Outline how DNA polymerases work.
5. Explain why there has to be a leading strand and a lagging strand.
6. Explain why a primer is needed in DNA replication.
7. The diagram shows a problem with DNA polymerase and DNA replication.

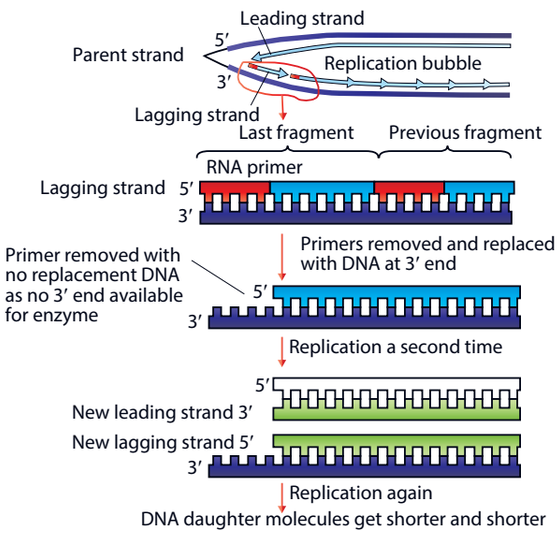


Figure 57.4 DNA polymerase and replication.

Outline the problem that is shown in this diagram.

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58 Recombinant DNA Technology

Recombinant DNA (rDNA) is DNA that has been artificially created from two or more different sources. Recombinant DNA technology uses **restriction enzymes** which cut DNA into short segments. Hundreds of different restriction enzymes have been identified with each restriction enzyme acting on a specific short sequence of DNA called the **restriction site**. The restriction enzyme cuts both strands of DNA at particular points on both DNA strands. In many instances the coding of the restriction site is repeated many times in the DNA molecule and the restriction enzyme will act on all these sites cutting the DNA molecule into many **restriction fragments**. The double stranded restriction fragments have at least one single stranded end called a **sticky end**.

Recombinant DNA techniques include – gene splicing using restriction enzymes and ligases to produce recombinant DNA; polymerase chain reactions to amplify or modify DNA sequences; use of DNA vectors and microinjection for carrying genes into nuclear DNA in the production of transgenic multicellular organisms.

How to make recombinant DNA

The basic steps involved in making rDNA involve:

1. The DNA from both sources is treated with the same restriction endonuclease to cut the same site on both molecules so that the ends have an overhanging piece of DNA.
2. The DNA preparations are mixed so that the complementary sticky ends can pair with each other using DNA ligase to link the two to form rDNA.

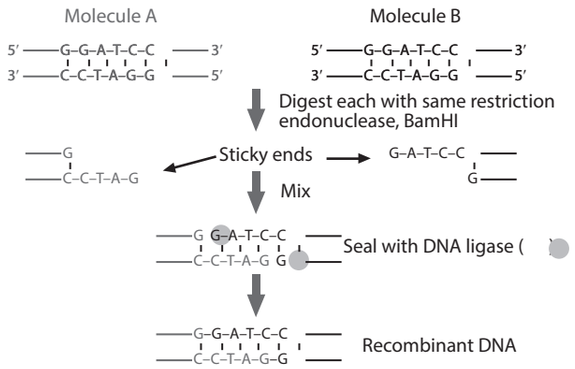

Figure 58.1 Making rDNA.

Different recombinant DNA technologies

Once the rDNA has been formed, it needs to be cloned so there are many copies, e.g. for sequencing, analysis, and insertion into cells. For example, a required gene is removed from its source cell and inserted into a bacterium plasmid. This recombinant bacterium is allowed to reproduce and thus make many copies of the inserted gene.

Polymerase chain reaction

Polymerase chain reaction (PCR) was developed by Kary Mullis (1944-) in 1983 and has been used since 1985 for cloning in vitro and many other applications. In 1993 Mullis and Michael Smith (1932-2000) were awarded the Nobel Prize for Chemistry – Mullis for his work on PCR and Smith for his work on site directed mutagenesis.

PCR takes a small amount of DNA and copies it many times. In PCR, the DNA is heated, e.g. to 60°C to separate the two strands, mixed with **primers** and then **DNA polymerase** extends the nucleotide sequence. The DNA polymerase comes from hot springs bacteria and is resistant to heat.

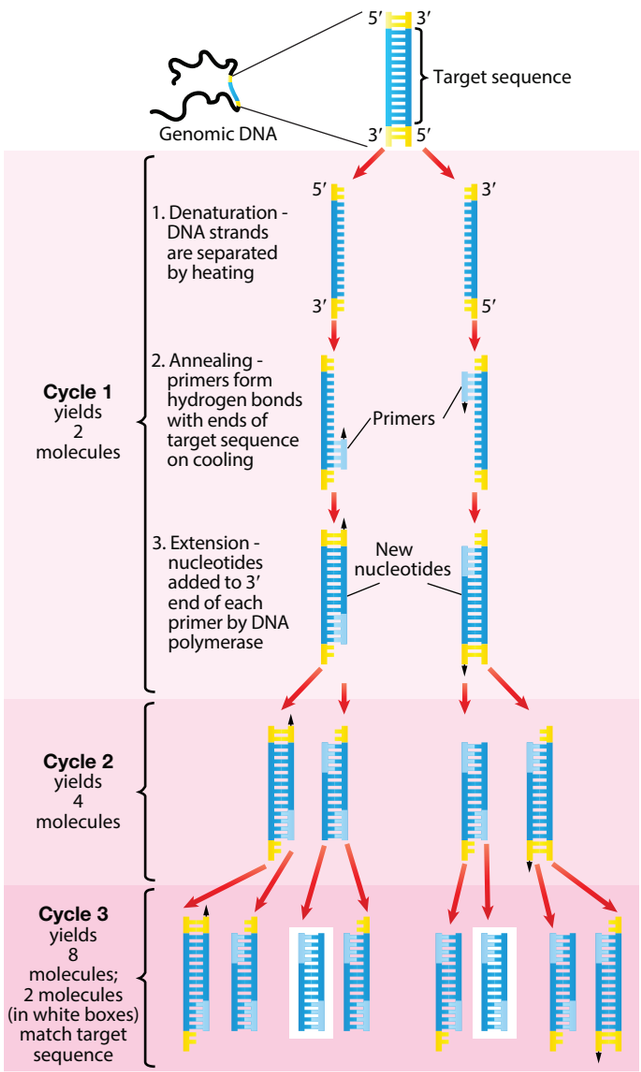

Figure 58.2 Polymerase chain reaction – a target sequence is amplified and copied many times in a test tube.

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64 Polymerase Chain Reaction

The **polymerase chain reaction** (PCR) is a technique for amplifying DNA in vitro by incubating nucleotides with special primers and DNA polymerase. It allows any chosen segment of DNA to be copied many times in a test tube in a relatively short time span.

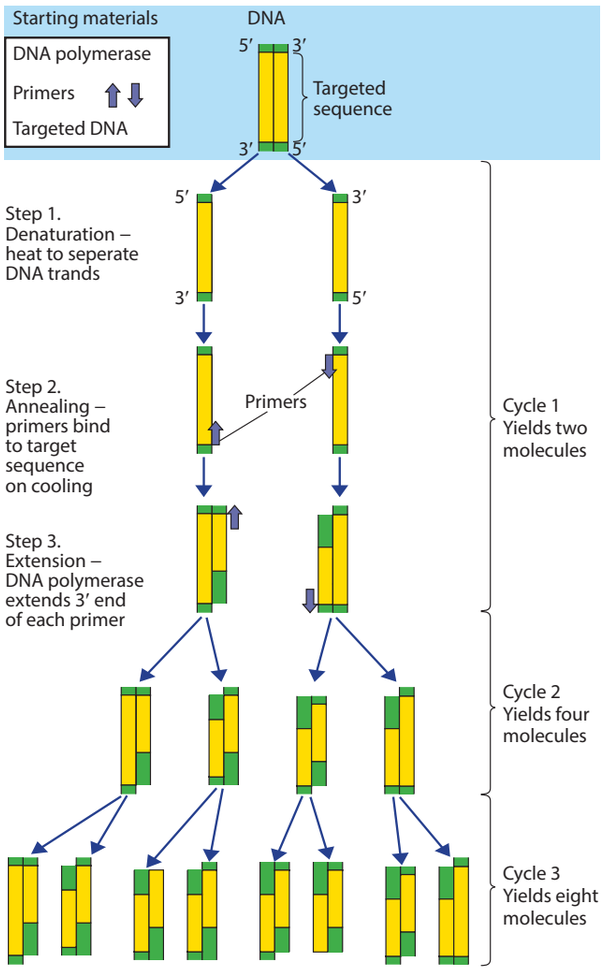

Figure 64.1 DNA polymerase is involved in DNA replication.

How it works

There are three basic steps in PCR which will exponentially increase the number of molecules each cycle.

Step 1 – **Denaturation** – the DNA strands are separated by heating.

Step 2 – **Annealing** – the two primers – each respectively complementary to the end of one of the strands of the target sequence bind by hydrogen bonding to its target DNA sequence during cooling.

Step 3 – **Extension** – a heat stable DNA polymerase extends the primers in the 5′ → 3′ direction.

The ability to amplify very small amounts of DNA has been invaluable for researchers, e.g. when studying the DNA sequences from archaeological remains and forensic evidence as these samples are often in very small amounts and can be partially degraded.

QUESTIONS

1. What is the polymerase chain reaction?
2. Outline why PCR is important.
3. What are the three main steps in PCR?
4. Taq DNA polymerase is named after the thermophilic bacterium *Thermus aquaticus* from which it was extracted. This bacteria lives in hot springs. Discuss why the discovery of Taq DNA polymerase greatly aided research investigations.
5. The diagram shows a type of DNA manipulation.

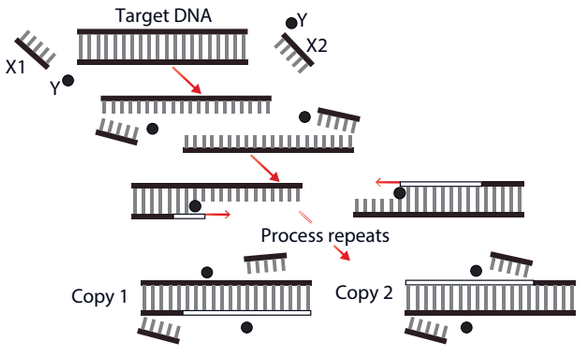

Figure 64.2 DNA manipulation.

   (a) Identify X1, X2 and Y.
   (b) Explain what is happening in this diagram.
6. Explain why PCR is invaluable when studying DNA samples that come from crime scenes or archaeological remains.
7. Explain why PCR is important in prenatal diagnosis.
8. What is the correct order of the polymerase chain reaction?
   (A) Extension, annealing, denaturation.
   (B) Annealing, extension, denaturation.
   (C) Annealing, denaturation, extension.
   (D) Denaturation, annealing, extension.
9. Which of the following DNA manipulation techniques amplifies a DNA sample?
   (A) Polymerase chain reaction.
   (B) DNA ligation.
   (C) Gel electrophoresis.
   (D) Southern blotting.
10. In the polymerase chain reaction, what is the function of DNA polymerase?
   (A) Cause denaturing of parent DNA strand.
   (B) Attach to end of target DNA to initiate process.
   (C) Extend primers in 5′ → 3′ direction.
   (D) Extend new DNA in 3′ → 5′ direction.

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75 Technology and Disease

Advances in our understanding of gene structure and function is changing the way we diagnose, prevent and/or treat disease.

An analysis of body proteins shows that there are many more proteins produced by a human cell than genes in the genome. **Alternative gene splicing** allows a given gene to produce more than one protein by editing the mRNA after transcription, e.g. by converting exons into introns or introns into exons. Epigenetic marks, e.g. acetylation and methylation influence gene activity and control gene expression.

Using gene editing, e.g. CRISPR-Cas9, the epigenetics of many diseases can be investigated to increase our understanding of conditions that lead to disease states, e.g. the silencing or abnormal behaviour of particular genes.

Disease diagnosis

DNA samples are cut using restriction enzymes, amplified using PCR and then the fragments are separated on size using gel electrophoresis. The DNA fragments are added to a nylon membrane and a labelled DNA probe added. **DNA probes** are used to identify specific genetic disorders by testing for specific mutated alleles. The probe has a specific sequence of nucleotides complementary to the target allele. **Hybridisation** occurs when the probe binds to the allele. The probe can be tagged with a fluorescent or radioactive marker which can then be detected using UV light or X-ray film (autoradiography) respectively.

A **DNA microarray** is a glass slide with microscopic spots of DNA probes attached in rows. A human fluorescently labelled DNA sample is washed over the array with complementary base pairing occurring if the sample contains target alleles. UV light will show any DNA that is bound to a probe identifying any of the targeted mutated alleles.

Disease diagnosis using DNA probes can identify specific mutations allowing personalised medicine using specific treatments, e.g. a specific type of mutation of the HER2 proto-oncogene that causes breast cancer can be effectively treated with Herceptin, while other types of mutation of this gene do not respond well to this drug.

Disease diagnosis identifies genetic health risks and individuals can adjust lifestyle choices to help reduce the development of that particular disease. For a disease like Huntington's disease which develops after the age of 30 years old, an individual found to have the allele with genetic screening, can prepare for the disease and also make reproductive choices, e.g. use IVF to screen embryo choice.

Genetic counsellors can be consulted about choices and treatment options after genetic screening.

Treatment

Monoclonal antibody therapies. A library of antibodies is being compiled as the specific genes that code for specific antibodies are determined. This aids in the diagnosis and treatments of diseases. Antibodies (immunoglobulins) are proteins that bind to a particular antigen and are produced by B lymphocytes. Monoclonal antibodies can be produced from hybridomas which are lymphocyte cells, e.g. from rat spleens that have been fused with human cancerous myeloma cells. Chinese hamster ovary (CHO) cells are also used using recombinant techniques to engineer cells that produce antibodies with a human stalk.

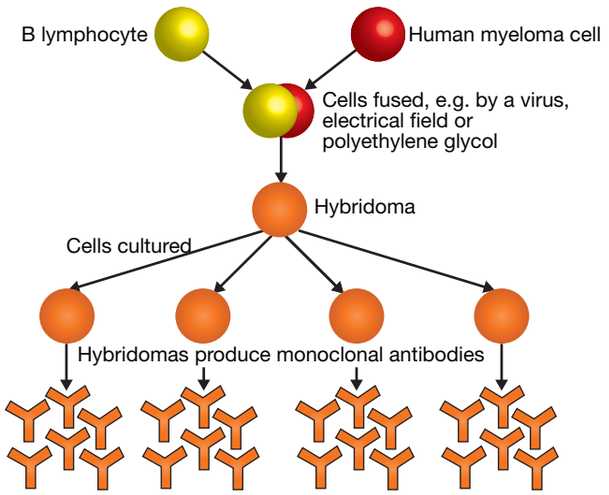

Figure 75.1 Hybridomas produce monoclonal antibodies.

Monoclonal antibodies are used in the treatment of autoimmune diseases, e.g. myasthenia gravis, a neuromuscular disease that causes weakness in skeletal muscles, breathing and moving parts of the body.

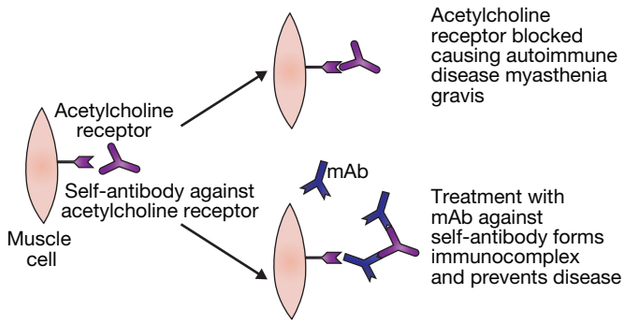

Figure 75.2 Treatment with monoclonal antibodies.

In the treatment of some cancers, e.g. some kidney cancers dendritic cells have been removed from the patient's blood and mixed with kidney cancer cells in the laboratory to form a dendritic cell vaccine. These cells are then re-injected into the patient to act as an antigen to start an immune response, e.g. against the kidney disease.

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9. How does DNA appear in eukaryotes and in prokaryotes?

	Eukaryote	Prokaryote
(A)	Unbound, circular DNA	Bound to proteins, circular DNA
(B)	Double stranded DNA bound to protein	Unbound circular DNA
(C)	Unbound, double stranded DNA	Double stranded, circular DNA
(D)	Double stranded circular DNA	Unbound linear DNA

10. Highly variable gene sequences can be identified using gene mapping. When are these gene sequences unable to identify individuals?
    (A) When blood groups are not compatible.
    (B) When the individuals involved are twins.
    (C) When the individuals belong to the same family.
    (D) When generational pedigrees are unavailable for the required trait.
11. If N is normal leaf shape and n is wrinkled leaf shape, which of the following genotypes represents plants with different genotypes but the same phenotype?
    (A) Nn and nn
    (B) Nn and Nn
    (C) NN and nn
    (D) NN and Nn

Use the following diagram for the next THREE questions.

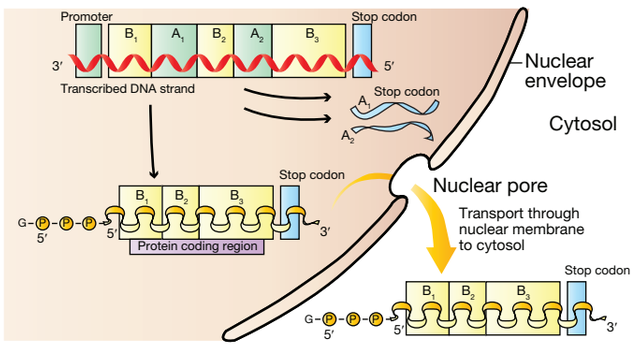

Figure TT4.1.3 Process involving DNA.

12. In this process what is represented by parts A_1 and A_2 and parts B_1, B_2, B_3?

	A_1 A_2	B_1 B_2 B_3
(A)	Exons	Introns
(B)	Introns	Exons
(C)	DNA	RNA
(D)	Histones	Nucleotides

13. What is shown in this process?
    (A) Translocation.
    (B) Differentiation.
    (C) Translation.
    (D) Transcription and RNA processing.
14. During this process the following section was added to the strand.

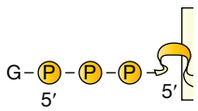

Figure TT4.1.4 Section added to strand.

What happens in this process?
    (A) A cap of a modified base is added to the 5' end of mRNA chain.
    (B) A cap of a modified base is added to the 3' end of mRNA chain.
    (C) A poly-A tail is added to the 3' end.
    (D) A poly-A tail is added to the 5' end.
15. In humans, thick lips are dominant over thin lips.

Figure TT4.1.5 Thick lips and thin lips.

If a man with thick lips has a child with thin lips, what conclusion can be drawn about the mother?
    (A) She must have thin lips.
    (B) She must have thick lips.
    (C) She could have thin lips or thick lips.
    (D) She must be homozygous for this trait.
16. The diagram shows part of an experiment carried out by Thomas Hunt Morgan with red eyed and white eyed fruit flies.

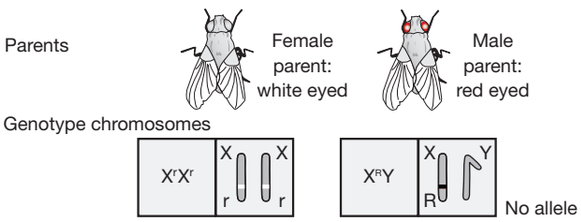

Figure TT4.1.6 Part of an experiment by TH Morgan.

What gametes would be produced in this cross?

	Female parent	Male parent
(A)	$\frac{1}{2}X^R$ $\frac{1}{2}X^r$	$\frac{1}{2}X^R$ $\frac{1}{2}Y$
(B)	All X^r	$\frac{1}{2}X^R$ $\frac{1}{2}Y$
(C)	$\frac{1}{2}X^R$ $\frac{1}{2}Y$	All X^R
(D)	$\frac{1}{2}X^R$ $\frac{1}{2}X^r$	$\frac{1}{2}X^r$ $\frac{1}{2}Y$

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SURFING

QCE BIOLOGY

UNIT 4

TOPIC 4.2

Continuity Of Life On Earth

In this topic you will:

- Distinguish between microevolution and macroevolution.
- Explain natural selection, gene flow and genetic drift.
- Investigate evolutionary radiation and mass extinctions.
- Analyse genotypic changes due to selective pressures on gene pools.
- Describe speciation and different mechanisms of isolation.
- Infer species relatedness from cladograms, phylograms and molecular sequence data.
- Link reduced genetic diversity with increased risk of extinction.

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77 Assumed Knowledge Topic 4.2

QUESTIONS

1. Define evolution.
2. Distinguish between macroevolution and microevolution.
3. Define speciation.
4. The diagram shows a cane toad.

Figure 89.1 Cane toad.

Outline why there is concern in Queensland and the Northern Territory about the spread of cane toads across the land and selective pressures on other species.

5. Identify some areas of evidence for evolution.
6. Define palaeontology.
7. Define biochemistry.
8. Draw a flow chart to show the evolution of the vertebrates from the ancestral fish.
9. What is comparative genomics?
10. What is a fossil?
11. What is an index fossil?
12. The diagram shows when fossil animals called trilobites existed on Earth.

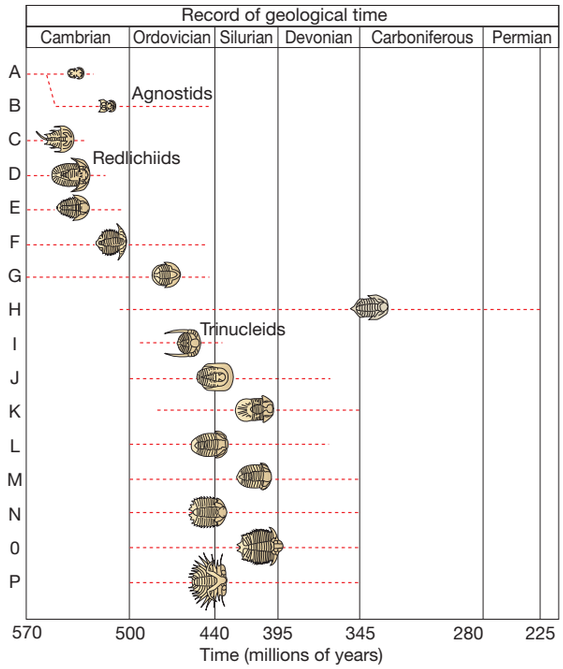

Figure 77.2 When trilobites lived.

Explain why trilobite D is useful as an index fossil but trilobite H is not particularly suitable to be used as an index fossil.

13. Define natural selection.
14. State the law of superposition.
15. Use an example to show how present day organisms have developed from different organisms in the distant past.
16. When does evolution occur?
17. (a) What is a clade?
    (b) What is a cladogram?
18. What is the approximate time scale of life on Earth?
19. How does the genetic diversity of a population affect the risk of extinction?
20. The table shows the changes and diversification of plants on Earth since the Cambrian.

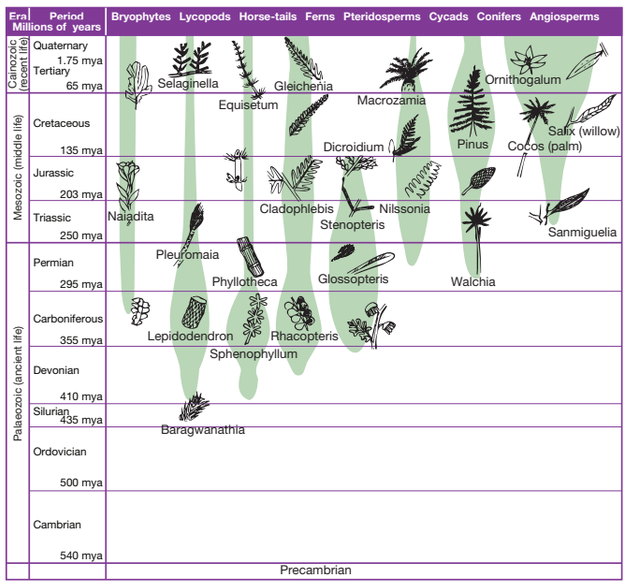

Figure 77.3 Plant life since the Cambrian period.

From this diagram:

(a) In which period and era did Eucalypts appear?

(b) Lycopods are seedless vascular plants that include the club mosses, spike mosses and quillworts. What is the oldest lycopod?

21. The graph shows the percentages of extinctions that have occurred since the Cambrian.

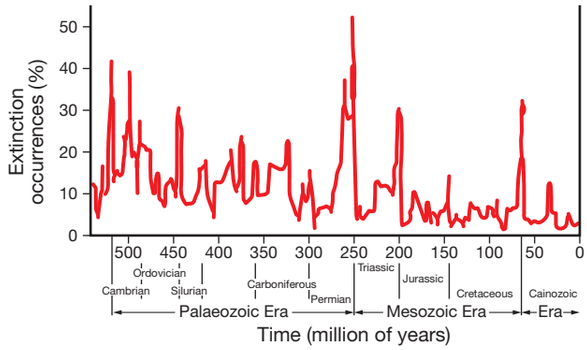

Figure 77.4 Plant life since the Cambrian period.

From this graph when was the highest percentage of extinctions of life on Earth since the Cambrian period?

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80 Genetic Drift

Genetic drift occurs when chance events cause unpredictable changes in allele frequencies from one generation to the next. Genetic drift occurs more easily in small populations than in large populations. Genetic drift is due to random events and since natural selection is rarely involved it does not necessarily lead to adaptations to specific environmental conditions.

Genetic drift is most noticeable if a population stays small over many generations. An allele may be eliminated by chance from a population even if it has an adaptive advantage.

Chance events that can affect survival and reproduction leading to genetic drift include the following.

- Natural disasters, e.g. an area may be flooded in a flash flood and those that make it to high ground survive.
- Some are accidentally killed by another species that is not a predator, competitor or pathogen.
- Chance events in fertilisation, e.g. by chance most offspring die and the only survivors have a particular genotype.
- Soils in one area become waterlogged leading to only some plants and animals surviving due to their chance location.

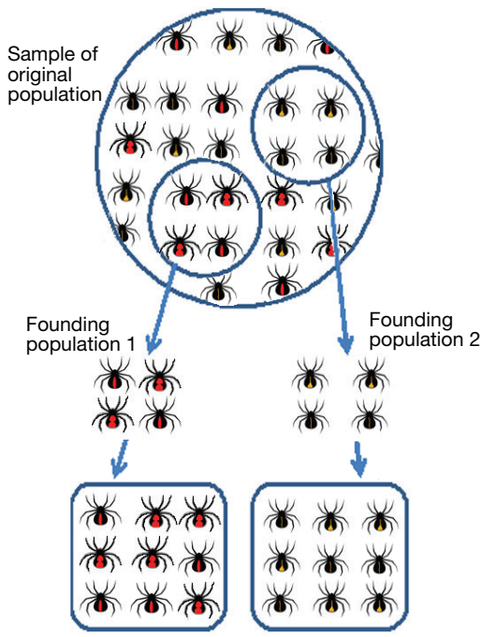

Figure 80.1 Founder effect.

Founder effect

The founder effect occurs when a few individuals become isolated from a larger population and form a new population whose gene pool composition is not reflective of the original source population.

In isolated human populations the founder effect can account for the high frequency of certain genetic disorders in these populations. On the island of Tristan da Cunha in the Atlantic Ocean there is a high frequency of the recessive allele that causes the degenerative eye disease retinitis pigmentosa. One of the early colonists had this recessive allele and over many generations many descendants have the allele.

Bottleneck effect

The bottleneck effect occurs when a large population is reduced in size, e.g. by a natural disaster or human activities and the gene pool of the surviving population is much smaller than the original gene pool. It is random which alleles are present in the survivors and which alleles are absent. With a small population there is a greater likelihood of further genetic drift which will continue until the population is large enough that chance events have less impact.

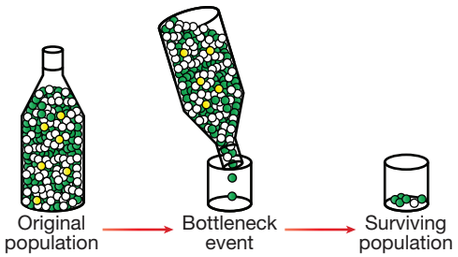

Figure 80.2 Bottleneck effect.

Around 10 000 years ago a bottleneck occurred with the cheetah population leading to the current low genetic variability of cheetahs.

QUESTIONS

1. Define genetic drift.
2. In what way is genetic drift different to natural selection?
3. Identify some chance events that can lead to genetic drift.
4. In the Old Order Amish of Lancaster, Pennsylvania which was established in the early 1770s there is a high proportion of the recessive allele that leads to a combination of dwarfism and polydactylism (extra fingers). Suggest how this high allele frequency could have occurred.
5. Explain why a population that stays small and isolated from other populations is likely to show genetic drift.
6. When studying ABO blood groups (I^A, I^B and i) it was found that Aboriginal Australians lacked the I^B allele. This means that the Aboriginal Australians do not have blood groups A or AB unless partners have come from other groups. Suggest how the lack of allele I^B could have occurred.
7. Explain why genetic drift can decrease genetic variation within a species and increase the amount of variation between local populations.

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QUESTIONS

1. Define microevolution.
2. List the four main processes that lead to microevolution.
3. The fossil history of the horse shows the increase in size, decrease in the number of toes and changes in tooth structure. Charles Darwin proposed that there is a slight change in the population each generation so that over time these changes lead to the evolution of a new species. Explain why the model of natural selection proposed by Darwin is often called 'gradualism'.

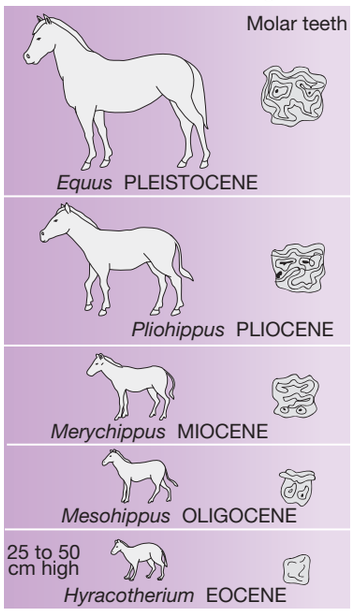

Figure 81.3 Evolution of the horse.

4. The diagram shows adaptive radiation in Australian marsupials.

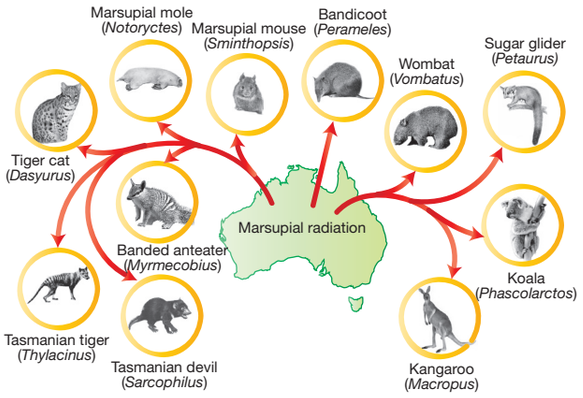

Figure 81.4 Adaptive radiation in Australian marsupials.

Explain why natural selection is important in adaptive radiation.

5. Stripe rust of wheat (*Puccina striiformis*) is a plant pathogen that shows a single founder event in Australia in 1979. The form that entered Australia was similar to a form found in Europe. The stripe rust has recently become a problem in Australia as it infects both winter wheat and spring wheat. The taxonomy of *Puccina striiformis* was revised in 2010 and the stripe rusts on wheat and grasses is now separated into four species based on molecular and morphological data.
   (a) Where was the originating source of the *Puccina striiformis* that entered Australia?
   (b) What could have led to the presence of the different forms of *Puccina striiformis* currently now found in Australia?
6. *Cryptasterina hystria* is an Australian starfish found in the mangroves and intertidal zone in only a few locations in south-east Queensland. It is an hermaphrodite that can produce both sperm and eggs simultaneously and self-fertilises within its own body cavity. Studies show that *C. hystria* can self-fertilise and that there is very low genetic variation in the species.
   (a) Suggest why there is low genetic variation in *C. hystria*.
   (b) Outline the importance of self-fertilisation for *C. hystria*.
7. Explain why hybridisation can at times be an advantage for one group but a disadvantage for another group.
8. Use an example to explain why many bacteria are now resistant to antibiotics.
9. During the ice ages sea level dropped and land bridges formed between mainland Australia, Tasmania, New Guinea, and the Indo-Malaysian peninsula. How would these land bridges have affected gene flow?
10. A new drug was developed that reduced a throat infection that had symptoms of pain when swallowing, runny nose and headache. After one week a few of the patients once again had the same symptoms. What can be concluded from this data?
    (A) The infection was caused by a virus.
    (B) Some of the pathogens present before treatment with the drug had a natural resistance to the drug and natural selection increased their numbers.
    (C) During the treatment some of the pathogens changed so they became resistant to the drug and they then increased in numbers.
    (D) There was gene flow between the pathogen colonies causing resistance and it took a while for the frequency of the resistance allele to increase.
11. A far distant ancestor of the mammals had a single gene for detecting odours. Gene duplication has led to different mammals having different numbers of olfactory receptor genes and different abilities to detect smell. Why is duplication important?
    (A) Gene duplication is a source of variation.
    (B) Duplication causes genetic drift.
    (C) Duplication controls the ability of mammals to detect different odours.
    (D) Gene duplication shows the mutation rates for all genotypes.

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QUESTIONS

1. Identify the main features possessed by mammals.
2. Outline the difference between monotreme, marsupial and placental mammals.
3. Outline where present day native monotreme and marsupial mammals can be found.
4. Construct a table to compare monotremes, marsupials and placental mammals.
5. Explain why there were few species of placental mammals on Australia until European settlement.
6. The diagram shows the coastline of Australia 65 000 years ago during an ice age.

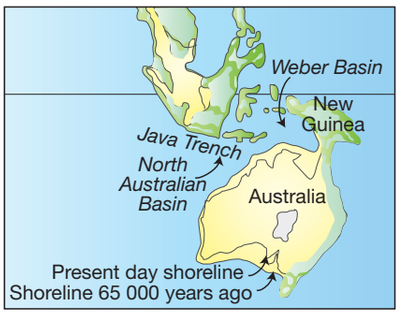


Figure 106.3 Australian coastline 65 000 years ago.

Discuss how the changed coastline would have affected animal life on Australia.

7. The table below shows features of three animals – L, M, and N.

Feature	Species L	Species M	Species N
Development of embryo	In egg	In mother	Free-swimming larvae
Body temperature	Ectotherm	Endotherm	Ectotherm
Cloaca	Present	Not present	Not present
Habitat of adult	Terrestrial	Marine	Terrestrial

For this information, which species is likely to be a mammal?

(A) M only. (B) L and N.
(C) L and M. (D) M and N.

8. *Lystrosaurus* is a genus of mammal-like reptiles which has been found in Antarctica, South Africa, South America and India. It was terrestrial, egg laying and lived about 240 million years ago. The fossils of *Lystrosaurus* have been used to show the existence of an ancient landmass. What was the name of this landmass?

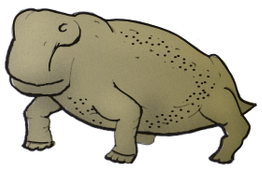
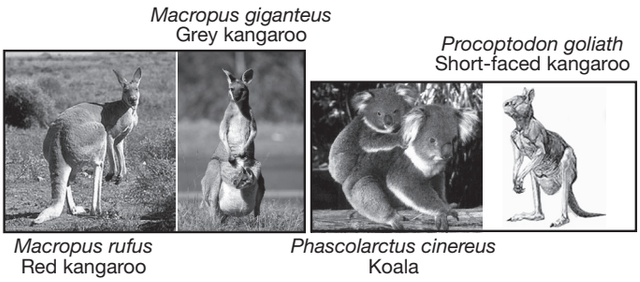


Figure 106.4 *Lystrosaurus*.

(A) Pangaea (B) Gondwana
(C) Laurasia (D) Indarctica

9. The Wallace line is a boundary between Asia and Australia named by Alfred Russell when he was observing biological differences between the two areas. What is a main consideration when determining the position of this line?
   (A) Climate differences between the areas.
   (B) Mountain ranges formed by folding and faulting.
   (C) Volcanic activity and associated tsunamis.
   (D) Continental shelves and ancient sea levels.
10. What is the only marsupial found in north America?
   (A) Carolina shrew possum.
   (B) Washington possum.
   (C) Virginia opossum.
   (D) Mississippi marsupial mouse.
11. Which of the following was the top marsupial predator in Australia before the arrival of European settlers?
   (A) Dingo. (B) Numbat.
   (C) Tasmanian tiger. (D) Wombat.
12. The Chicxulub impact crater in central Mexico is believed to have been caused by a meteorite that could have been up to 16 km across and was the size of Mount Everest. It struck the Earth about 65 million years ago and its arrival coincided with the end of the Cretaceous period. How did life on Earth, including life in Australia, change at this time?
   (A) Extinction of the anaerobes.
   (B) Mass proliferation of the trilobites.
   (C) Extinction of the dinosaurs.
   (D) Extinction of the woolly mammoth.
13. Many palaeontologists use the relative age of fossils to construct timelines. What is meant by relative age?
   (A) The age of the fossil is considered older than the layer above it.
   (B) The age of a fossil is considered older than the layer below it.
   (C) Age is relative to other fossils of that species.
   (D) Age depends on Einstein's theory of relativity.
14. The diagram shows four Australian animals.



Which of these species are extant?

   (A) *Macropus rufus* and *Macropus giganteus*.
   (B) *Macropus rufus*, *Macropus giganteus* and *Phascolarctus cinereus*.
   (C) *Phascolarctus cinereus* and *Procoptodon goliath*.
   (D) Only *Procoptodon goliath*.

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18. The population of a small rodent extends across a landmass that encompasses a variety of ecosystems in different climate zones.

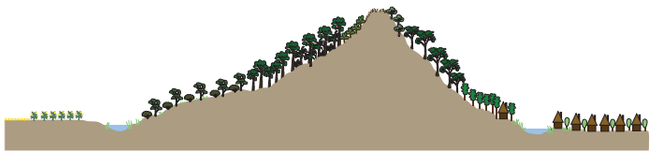

Figure TT4.2.6 Transect across a large landmass.

Which of the following genetically determined characteristics would be *least* likely to have regional differences?

(A) Body proportions. (B) Fur colour.
(C) Mating calls. (D) Blood disease.

19. The map shows the entrance point A of Aboriginal peoples into Australia.

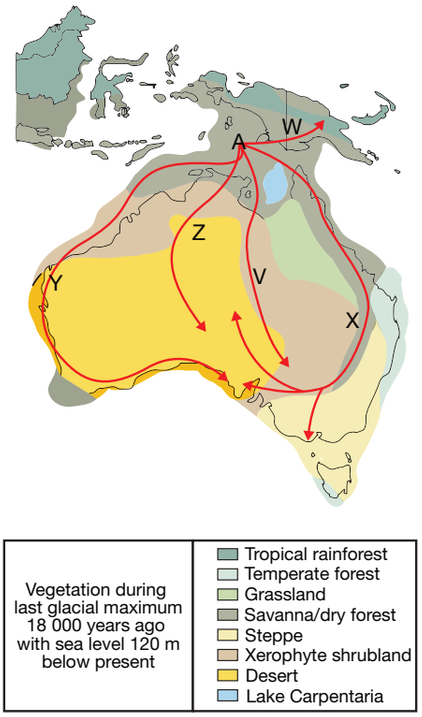

Figure TT4.2.7 Aboriginal migration patterns.

After the rapid migration each group stayed in their own area gaining strong attachment to their land and geneticists have identified 54 unique Aboriginal mtDNA haplogroups. Why is this important genetic information?

(A) It shows how gene flow increases genetic diversity in each population.
(B) It shows genetic drift in separated populations.
(C) It shows mutation did not occur after colonisation.
(D) It shows phenotypic change to suit the environment.

20. The diagram shows changes in a fish population in a lake over time.

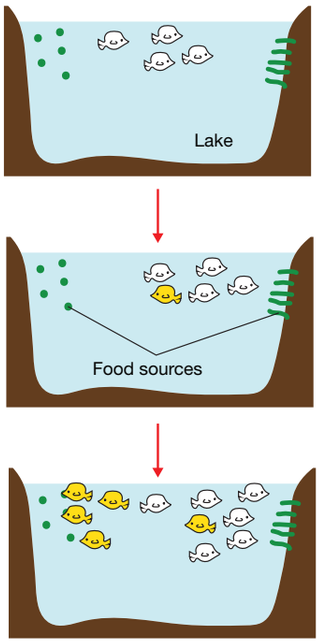

Figure TT4.2.8 Lake population.

What type of change is shown in the diagram?

(A) Allopatric speciation.
(B) Parapatric speciation.
(C) Sympatric speciation.
(D) Peripatric speciation.

Section B – Written Response (30 marks)

21. Describe the mechanism of evolution. (3 marks)
22. Use a named example to show how an advance in technology has changed scientific thinking about evolutionary relationships. (3 marks)
23. During the Pleistocene there were periods with abundant rain and high humidity and periods of low humidity with ice ages forming ice sheets, e.g. in Antarctica and Greenland. During this time there were many instances in Australia of forms adapted to wet and semi-arid conditions becoming fragmented and isolated in refuges around the periphery of the continent.
    (a) Explain why this is an example of allopatric speciation in Australia during the Pleistocene epoch. (2 marks)
    (b) Research has suggested that the Lake Eyre Basin and the Flinders Ranges formed the 'Eyrean Barrier' and can help to account for the divergence of southern Australian birds, e.g. populations of the mulga parrot, *Psephotus varius* and the red rumped parrot *Psephotus haematonotus* which are found in arid scrublands and grasslands in inland southern Australia.

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