

BIOLOGY

LEVELS OF LIFE

• AUSTRALIAN CURRICULUM EDITION •



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AUSTRALIAN CURRICULUM EDITION

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BIOLOGY: LEVELS OF LIFE
AUSTRALIAN CURRICULUM EDITION

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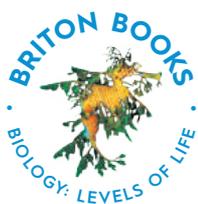
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A leafy seadragon off the coast of Yorke Peninsula, South Australia

Leafy Seadragons (*Phycodurus eques*) are endemic to Australia and are found from Lancelin WA, to Wilsons Promontory, Vic, but are mostly sighted in South Australian waters and southern WA waters. The Leafy Seadragon is the marine emblem of South Australia.

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Some images used in this book were supplied from Adobe Stock, Dreamstime, and iStock.

Brian LeCornu

Tony Diercks

Adelaide, January 2025

This e-version can be used alongside the hard copy edition, as the page integrity has been maintained. Many new links to resources have been added to the 2025 edition.

Brian LeCornu

Tony Diercks

Adelaide, January 2025

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The authors would appreciate feedback via www.britonbooks.com, including details of links that do not function correctly.

Introduction

Biology as a subject covers an enormous amount of material, and there is no way that a one-year course could possibly cover it all. Biologists generally specialise in studying a particular aspect of living things, such as what they are made of, where they live, how they work, how they evolved (and continue to evolve) and how they interact with one another.

This course attempts to give you an understanding of some of the important concepts in biology so that you will have a greater appreciation of the diversity and function of living things. You should then be able to make informed decisions about biological issues. You will be in a position to offer well-informed opinions on issues that don't have a 'right' answer. These could include questions such as: should genetic engineering be used to produce large quantities of biological substances including food? Should people who have a family history of genetic disease be permitted to have their embryos genetically altered using new biotechnological techniques? If it became possible to grow human organs in laboratory tissue culture, should this be the basis of a new 'industry'? How can we manage the growth of the human population and preserve biodiversity? These are all examples of Science as a Human Endeavour.

Like all matter, living things are made up of chemicals — atoms and molecules. These chemicals form DNA and proteins and are the basis of the simplest unit of living matter — the cell. Some living things (organisms) consist of a single cell and most of these unicellular organisms are microscopic and relatively simple. Other living things are made up of many cells, and are called multicellular. In these organisms, cells are arranged to form tissues, organs, and systems. Humans are an example of such multicellular organisms and they are quite complex in both structure and function. Organisms of the same kind form populations, populations in turn make up communities, and groups of communities form ecosystems.

So you can see that we can study living things at different levels such as: molecules, cells, organisms, and ecosystems. This idea forms the basis of this course and this book, hence: **Biology: Levels of Life.**

As you progress through the course this year you will learn of the amazingly detailed structure of microscopic cells, and how each cell is like a tiny chemical factory. But don't worry - you don't need to be a chemistry student to understand this section. You will improve your knowledge and understanding of aspects of several human systems, such as the nervous and endocrine systems and how these interact to achieve a stable internal environment - homeostasis. You will learn about the process by which populations become well-suited to their environment and how life has evolved on Earth. You will also gain a better understanding of how living things interact with each other and with their non-living surroundings.

About this Book

This book is not intended to be used exclusively in your biology studies, nor to replace your teacher. You should regularly refer to other sources of information. Your teacher will guide you through the course and provide additional explanations.

BIOLOGY: LEVELS OF LIFE, AUSTRALIAN CURRICULUM EDITION has been written to cover all of the Science Understandings of the Stage 2 SACE Board of SA Biology subject outline.

Each **Science Understanding** is indicated in the text as a **coloured heading** and each dot point is indicated by 

It is important to note that the sequence of these science understandings may not exactly match their sequence in the subject outline. This is because the authors felt that for teaching and learning purposes the sequence of ideas in the subject outline is not always suitable. The ideas are presented in chapters that stand as self-contained units of work. References to other chapters have been made where necessary. The hierarchical organisation consistent with Levels of Life has been preserved by grouping the chapters into the four topics: **DNA and Proteins, Cells as the Basis of Life, Homeostasis, and Evolution.**

While it is possible to work sequentially through the chapters in the book, it is not necessary to do so. For example, it is possible to begin with the Cells as the Basis of Life topic, and then work through DNA and Proteins, Homeostasis, and Evolution. Alternatively, a course could begin with a chapter from the topic Cells as the Basis of Life, and then continue with a chapter from the DNA and Proteins topic, and so on. Teachers and students should choose to work through the course in a way that suits their background knowledge, interests, and abilities.

Throughout the book there are numerous QR codes and websites that can be accessed for additional information or videos or animations.



tinyurl.com/n8lvs4r

Relevant Science as a Human Endeavour (SHE) examples for each topic are indicated by



One 'secret' to success in biology that we know of is: learn the meanings of new words or terms as soon as you encounter them. To help you with this, words that are considered to be important to know are indicated in bold throughout the book, and a glossary of useful terms is included before the index. Part of learning biology (or any subject for that matter) involves becoming familiar with the 'jargon' or special language of the subject.

The Study Questions at the end of each chapter are designed to assist your learning by helping you to determine whether you have understood the work. Your teacher may set these questions as the basis for discussion in class. In any case, you are strongly advised to attempt the questions as you work through the chapters, and to take immediate action if you experience any difficulty. We hope that you find this subject interesting, challenging, and enjoyable.



TOPIC



DNA and Proteins

- 1 Chromosomes and DNA
- 2 The Language of Life
- 3 Proteins
- 4 Genes and Phenotypic Expression
- 5 The Use of Genetic Information
- 6 Biotechnology (Human Manipulation of DNA)

In their endeavour to understand how biological systems work, scientists generally start by examining the component parts. To understand how a human works, physiologists examine the body systems, like the digestive system. To understand how the digestive system functions, they study the organs like the stomach and duodenum. Histologists study the function of the cells of these organs, and biochemists examine the molecules that make up the cells.

This process of reducing something down to its component parts is called reductionism and it acknowledges that biological systems have a hierarchy; that is, there are levels of life. The hierarchy runs from the biosphere, through ecosystems, communities, populations, organisms, systems, organs, tissues, cells, and molecules.

Evidence suggests that life started when conditions on primitive Earth allowed for the synthesis of the first organic molecules from simple chemicals. Once the first organic molecules joined to form the first cell, life on Earth had begun. Cells later joined to form more complex structures and then multicellular organisms. Reductionism is simply studying this process in reverse.

Many of the new technologies, like genetic engineering, depend on research on macromolecules in order to understand how to change the whole organism and even the population of an organism.

Topic one of this book looks at the first level of life — **DNA and Proteins** — two of the large chemicals that make up cells.

Science as a Human Endeavour

Throughout this topic examples that illustrate key concepts of science as a human endeavour are indicated by the symbol ▼. There are examples of communication and collaboration, development of scientific models and new technologies, influence on and by other areas of study and society, and applications and limitations of biological knowledge.

Chromosomes and DNA

1

In order to understand the chemical components of living things, including DNA and proteins, it is a good idea to review some simple chemical concepts.

- All matter is made up of **atoms**.
- Some substances, called **elements**, consist of only one kind of atom.
- There are 92 naturally occurring elements on the Earth. You will be familiar with many of the more common elements such as carbon (C), hydrogen (H), oxygen (O), nitrogen (N), sulfur (S), phosphorus (P), iron (Fe), calcium (Ca) and magnesium (Mg). The letters in brackets are the **chemical symbols** of these elements.

As there are only 92 naturally occurring elements, there can only be 92 naturally occurring types of atom. However, we know that there are literally millions of different chemical substances. For example, there are thousands of varieties of plastic, and thousands of types of mineral. How can this be, if all matter is made up of only 92 different kinds of atoms? The answer becomes clear when we realise that atoms of different kinds can join together chemically to form **compounds**.

A well recognised example of a simple compound is water. It consists of hydrogen atoms and oxygen atoms joined together in the ratio 2:1. That is, each water **molecule** consists of two hydrogen atoms joined to one oxygen atom. (See Fig. 1.1) Note that the chemical and physical properties of water are quite different from those of either hydrogen or oxygen.

When atoms of different kinds combine chemically, completely new substances are produced. It is not only the type of atoms that determine the new substance, but also their proportion. For example hydrogen peroxide, like water, consists of only hydrogen and oxygen atoms, but in the ratio 2:2, and it is highly reactive, unlike water.

Element symbols can be written together to make the **formula** for a compound. The formula for water is H_2O and the formula for hydrogen peroxide is H_2O_2 .



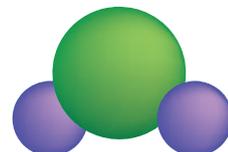
hydrogen atom (H)



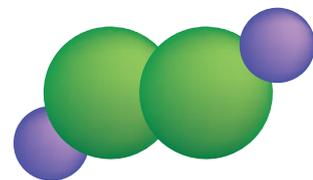
oxygen atom (O)



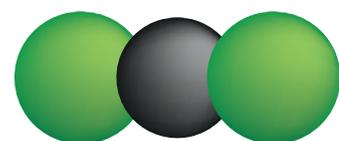
carbon atom (C)



water molecule (H_2O)



hydrogen peroxide molecule (H_2O_2)



carbon dioxide molecule (CO_2)

Fig. 1.1 Atoms and molecules

Chemical compounds are classified into two main groups — **organic** and **inorganic**.

All organic compounds:

- contain carbon
- are complex
- are produced by or associated with living things.

In most cases it is easy to tell whether or not a compound is organic. For example, water does not contain carbon, and is inorganic. Glucose ($C_6H_{12}O_6$) is organic, as it fits all three criteria. One compound that you will need to remember is carbon dioxide (CO_2) which, although it does contain carbon and it is produced by living things, is not a complex molecule, and so is not organic.

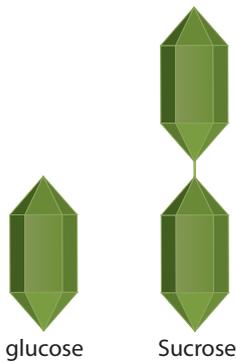


Fig. 1.2 Carbohydrates

There are four major types of organic compounds in cells and organisms: nucleic acids (DNA and RNA), proteins, carbohydrates, and lipids. Organic compounds play important roles in cells, both as structural components and in cellular reactions. Some of these compounds are relatively large and are often referred to as macromolecules. These macromolecules are made up of smaller molecules called **subunits** that join together in long chains.

DNA stores and transmits genetic information; it functions in the same way in all living things.

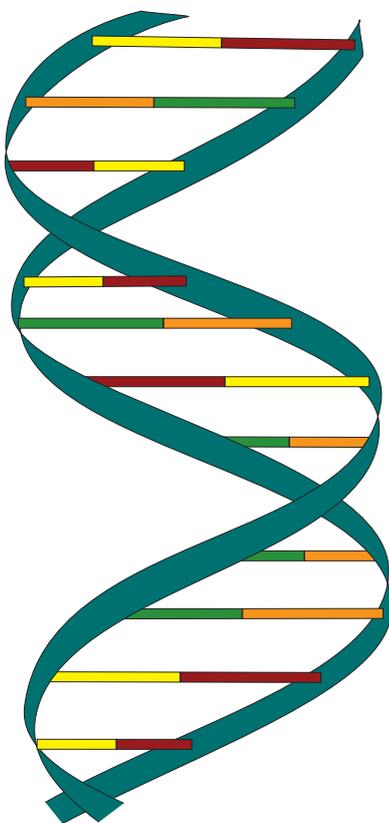


Fig. 1.3 DNA double helix

All organisms need some way of storing and transmitting the information that is necessary to direct the complex processes that they carry out. How is this information stored in cells and how is it transmitted to new cells? The answer lies in a nucleic acid macromolecule called **deoxyribonucleic acid**, or **DNA**. The discovery of the structure of DNA in 1953 by James Watson and Francis Crick at Cambridge University marked a turning point in modern molecular biology. (See Fig. 1.3)

DNA is a unique molecule because it is able to self-replicate — that is, it can make a copy of itself. In addition, its sequence of nucleotides allows it to store information that can be copied and passed on to daughter cells. Thus DNA, which is found in all known organisms, provides the link between one generation and the next.

What is surprising is that the 'language of life' or **genetic code**, as it is called, is the same for all living things. This code, which is the sequence of bases on the cell's DNA, uses three bases at a time, called codons, to direct protein synthesis - the assembling of proteins from amino acids. Thus, *DNA functions in the same way in all living things*. (Protein synthesis is discussed in detail in chapter 2).

To understand how DNA is able to perform these functions we first need to examine the structure of nucleic acids and the unique features of DNA.

Nucleic acids

We will begin our discussion of DNA by looking at some of the features that are common to all nucleic acids. Nucleic acids are long molecules (polymers) made up of subunits called nucleotides. Each nucleotide is made up of three parts: a sugar, a phosphate group, and a nitrogen base. (See Fig. 1.4) Nucleic acids contain the elements carbon, hydrogen, oxygen, nitrogen, and phosphorus.

There are two types of sugar found in nucleotides — a **ribose** sugar and a **deoxyribose** sugar. These sugars contain five carbon atoms and are very similar to one another, but they are not found in the same nucleic acid at one time. The two types of nucleic acid, deoxyribonucleic acid (DNA) and **ribonucleic acid (RNA)**, are named according to the type of sugar unit that they contain. RNA contains ribose sugars, while DNA is made using deoxyribose sugars. The nucleotides join together in long lines with the sugar of one nucleotide joining to the phosphate group of the next nucleotide. A long 'backbone' of alternating sugars and bases is formed. It is the type of nitrogen base that distinguishes different nucleotides. There are five **nitrogen bases**: adenine, cytosine, guanine, thymine, and uracil, abbreviated A, C, G, T, and U respectively. The first four are found in DNA, while in RNA uracil (U) replaces thymine (T).

Although RNA and DNA are both long chain polymers made up of nucleotides, there is a particular difference between them. RNA usually forms a single strand, whereas DNA is made into a double strand and may be over a million nucleotides long – much longer than RNA. (See Fig. 1.5) The two strands of DNA are complementary as A only pairs with T and C only pairs with G. (See Fig. 1.6) This is called complementary base pairing.

What makes DNA special?

DNA is a helical double-stranded molecule.

James Watson and Francis Crick were awarded a Nobel prize for their discovery in 1953 of the structure of DNA – the now famous 'double-helix'. (See textbox 'Discovering the double helix')

It is the double-stranded nature of DNA that lends itself to self-replication. The two strands are said to be **complementary** because each one corresponds to the other in an exact and predetermined way. Adenine (A) on one strand always pairs with thymine (T) on the other strand, and likewise cytosine (C) always pairs with guanine (G). The complementary bases on each strand are held together by weak hydrogen bonds. You may find it useful to think of a molecule

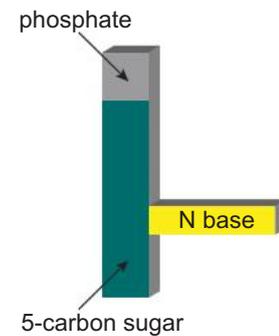


Fig. 1.4 Nucleotide

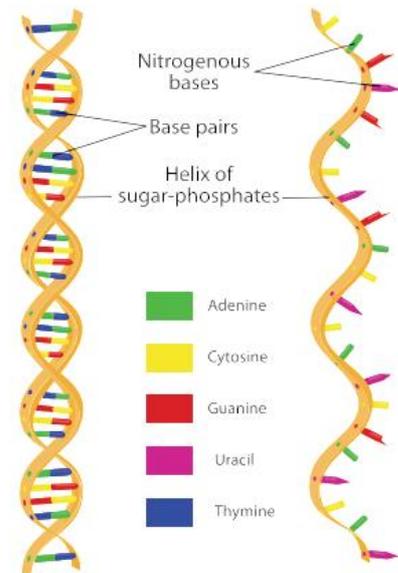


Fig. 1.5 Nucleic acids DNA and RNA

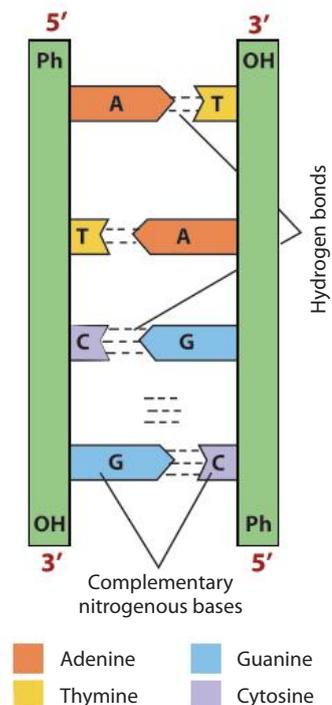


Fig. 1.6 Complementary strands of DNA

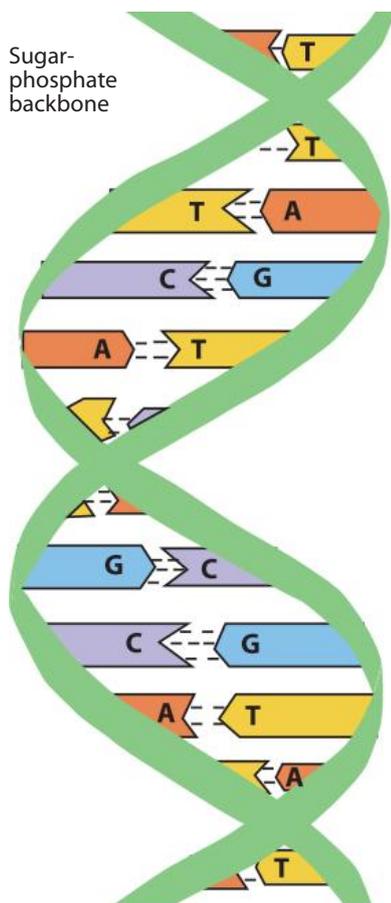


Fig. 1.7 DNA double helix

of DNA as being like a ladder. The deoxyribose sugar and phosphate sections of the nucleotides form the 'uprights' or 'sides' of the ladder, and are sometimes referred to as a 'sugar-phosphate backbone', while the nitrogen base pairs form the 'rungs'. This whole ladder-like structure is then twisted into a spiral or helix. Hence, the term **double-helix** is generally used to describe the structure of DNA. (See Fig. 1.7) If you were to read the sequence of bases along one of the strands you could work out the corresponding sequence on the other strand using the A-T, C-G rule. If part of one strand was ATCG, then the corresponding part of the other strand would be TAGC. (See Fig. 1.6)

The really incredible part of this story is that the sequence of bases along the DNA makes up a 'code' of instructions for the cell. More details of this code (called the genetic code) will be discussed Chapter 2. For now, let's just say that the genetic code stored on the DNA in a cell's nucleus, provides all the information that the cell needs in order to carry out its complex functions. This information can be copied and passed on to new cells. So you can see that DNA is important in inheritance.

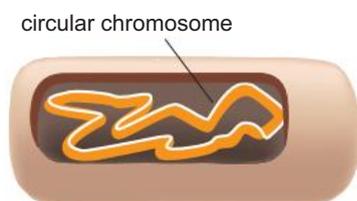


Fig. 1.8 Circular chromosome in a bacterial cell

DISCOVERING THE DOUBLE HELIX

Although Watson and Crick, who were working at Cambridge University, are the names most often quoted, two other people contributed significantly to the discovery of the structure of DNA. Rosalind Franklin and Maurice Wilkins from Kings College, London had generated the information from their research that Watson and Crick had used. In fact, Rosalind Franklin had independently postulated a double helix structure, but Watson and Crick published their ideas first. Watson, Crick and Wilkins were jointly awarded the Nobel Prize in 1962. Unfortunately, Rosalind Franklin had died in 1958, and the Nobel Prize can only be awarded to living persons.

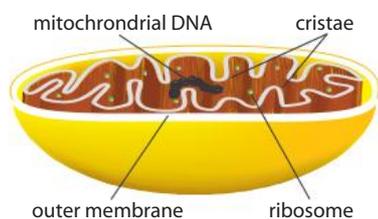


Fig. 1.9 Mitochondrion

DNA is unbound and circular in the cytosol of prokaryotes and in the mitochondria and chloroplasts of eukaryotes.

Bacterial cells (prokaryotic cells) contain a single circular chromosome, made of DNA with no protein attached to it. (See Fig. 1.8) The chromosome in a prokaryotic cell is not enclosed in a nucleus, but floats freely in the **cytosol**. The cytosol is the fluid part of the cytoplasm, not including organelles such as ribosomes. When bacterial chromosomes were studied it was found that they contain several hundred genes and that each gene has a specific location or locus on the chromosome. The role of genes is discussed in Chapter 2.

Prokaryotic cells, eukaryotic cells, mitochondria, and chloroplasts are described in detail in Chapters 7 and 8.

Mitochondria and chloroplasts have their own DNA, separate from the DNA in the nucleus. Their DNA resembles prokaryotic DNA as it is circular and it has no protein attached. (See Fig. 1.9 and Fig. 1.10)

In eukaryotes, DNA is bound to proteins (histones) in linear chromosomes, which are found in the nucleus.

Eukaryotic cells, such as human cells, contain chromosomes that are linear. It has been shown that in eukaryotic cells a particular gene also has a particular locus on a particular chromosome. (See Fig. 1.11)

In eukaryotic cells the long thread-like strands of DNA in the nucleus are coiled around proteins (**histones**) to form structures called chromosomes. (See Fig. 1.13) These chromosomes are not normally visible as discrete units unless the cell is dividing and the long strands have condensed to form shorter, thicker rod-like structures. (See Fig. 1.12) The chromosomes in prokaryotic cells do not contain histones. It is the DNA of the chromosomes that contains the vast store of information, in the form of base sequences, that is needed by the cell and the organism.

The information coded in the DNA determines what proteins are made within a cell, and hence it influences the structure and function of the cell. A segment of DNA on a chromosome that contains the complete sequence of bases required to direct the manufacture of a **polypeptide** or an RNA molecule is called a **gene**.

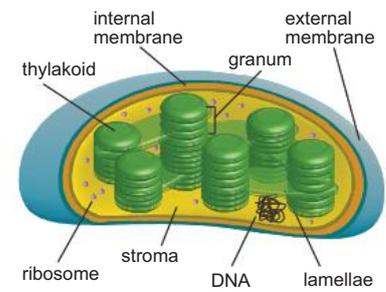


Fig. 1.10 Chloroplast

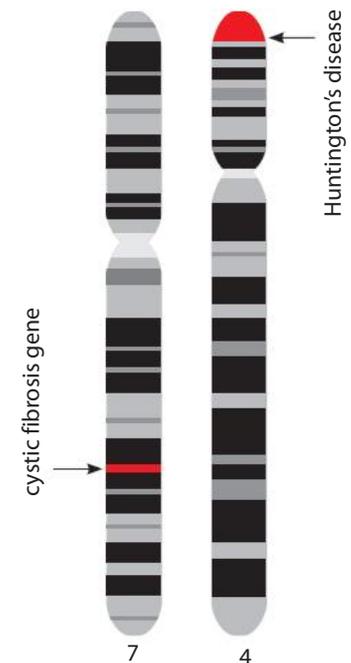


Fig. 1.11 Chromosomes 7 and 4

HISTORY OF DNA DISCOVERY

Although DNA was identified by around 1850 as the major chemical occupying a cell's nucleus, its role in storing and transmitting genetic information was not accepted by the scientific community until 1952. Up until then many scientists believed that proteins were the most likely storage form of information in cells.

The first people to recognise the importance of chromosomes in inheritance were Walter Sutton and Theodor Boveri who, in 1902, independently put forward the concept that chromosomes carry hereditary material. Their concept was largely disregarded for almost 30 years, until further experiments supported their ideas. There was still a debate over whether it was the DNA or the proteins in the chromosomes that carried the genetic information. In 1944 Oswald Avery had shown the importance of DNA in inheritance in a strain of bacterium, but many scientists were not convinced. It was not until 1952 that two biologists, Alfred Hershey and Martha Chase, performed an experiment that supported the proposal that DNA and not protein contained the inheritable material of life.

A polypeptide is a chain of amino acids linked by peptide bonds, and forms part or all of a protein.



Fig. 1.12 Chromosomes in dividing onion root tip cells



Compare chromosomes in prokaryotes and eukaryotes.

Chromosomes in Prokaryotes — bacteria	Chromosomes in Eukaryotes — plants, animals, fungi, protists
circular	linear
no histones	contain histones
located in the cytoplasm	located in the nucleus
one per cell	two or more per cell
most have no introns (non-coding DNA)	have introns (non-coding DNA)
centromere-like structure attaches to cell membrane during cell division (binary fission)	centromere attaches to spindle fibres during cell division

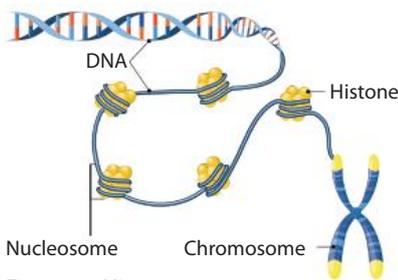


Fig. 1.13 Histones

Details of the structure and function of prokaryotic and eukaryotic cells are provided in Chapter 8.

TELOMERES – CHROMOSOME PROTECTION

Telomeres are repetitive sequences of DNA found at the end of most eukaryotic chromosomes. The telomeres in humans are single stranded DNA with several thousand repeats of the sequence TTAGGG. In 2009, Elizabeth Blackburn, an Australian scientist, Carol Greider and Jack Szostak, were awarded the Nobel Prize for their research into telomeres showing that telomeres prevent the ends of chromosomes fusing with each other, causing mutations, and that the enzyme, telomerase reverse transcriptase (TeRT) repairs telomeres.

During each DNA replication, a few nucleotides are removed from the ends of the chromosomes, so if the telomere is present, it will be shortened rather than sacrificing essential DNA. When the telomere becomes too short, the cell dies. TeRT can repair the damaged ends of telomeres and extend the life of the cell. This enzyme is found in high concentrations in both stem cells and cancer cells. (See Fig. 1.14)

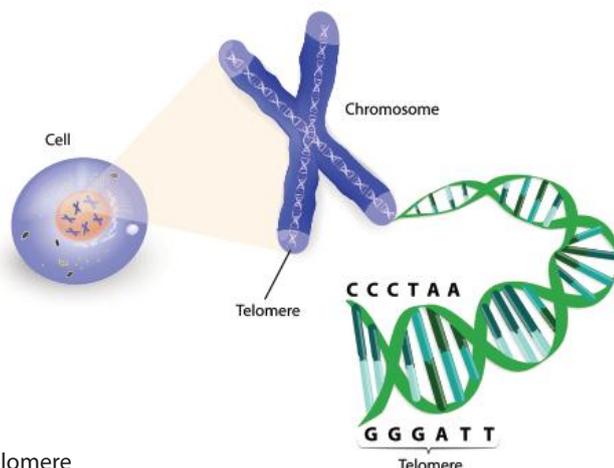


Fig. 1.14 Telomere

Study Questions

- Atoms and molecules make up all living things.
 - Explain the difference between an element and a compound.
 - What is a macromolecule?
 - What is the importance of chemical bonds in molecules?
 - Why is glucose considered an organic compound?
- What special and unique property of DNA makes it suited to the transmission of genetic information from one generation to the next?
 - How is DNA able to store genetic information?
- Complete the following table comparing DNA and RNA.

	DNA	RNA
Name of sugar present		
Names of nitrogen bases		
Number of strands in molecule		

- DNA is generally described as a 'double helix'. Draw a labelled diagram to explain what this term means when referring to the structure of DNA.
- One strand of a segment of DNA has the following base sequence: ACCGATGTG. What is the base sequence on the complementary strand of this segment of DNA?
- How does DNA in a prokaryote differ from that in eukaryotes?
- Describe the structure and function of chromosomes.
 - Describe the structure and function of a gene.
 - How is it possible to distinguish between one chromosome and another?

2

The Language of Life

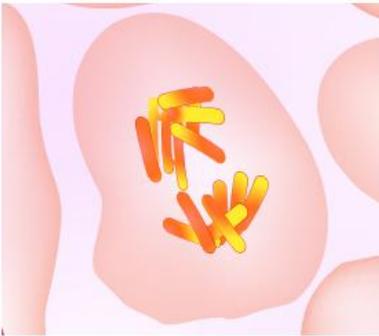


Fig. 2.1 Diagram of a cell with condensed chromosomes

Even a simple cell with one chromosome needs to make hundreds of different proteins such as enzymes. As each different protein is coded for by a different gene, there must be many genes on this single chromosome. In humans there are several thousand genes on each of the chromosomes. Thus, a chromosome is made up of many genes.

Each human body cell has 46 chromosomes. During cell division, these chromosomes condense and become visible. (See Fig. 2.1) Biologists can take photographs of these rod-shaped chromosomes and then cut and paste them to make up a composite picture called a **karyotype**. (See Fig. 2.2) The chromosomes are lined up in order of size and are numbered accordingly. There is one anomaly — the chromosomes that contain the genes determining sex (called sex chromosomes) are of two types. These are X and Y chromosomes. Females have two X chromosomes and males have one X chromosome and one Y chromosome, in addition to 44 non-sex chromosomes, called **autosomes**.

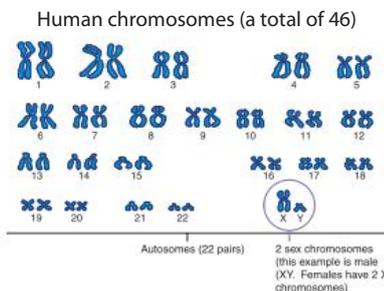


Fig. 2.2 A Human Male Karyotype

Replication of DNA allows for genetic information to be inherited.

Every time a cell divides, its DNA is copied and each new (daughter) cell receives a copy. Thus, the information on the DNA is passed from one generation of cells to the next. Similarly, when organisms reproduce, they pass genetic information on to their offspring via DNA. In the case of sexual reproduction an individual receives only half of each parent's genetic material. In humans this genetic material is carried in egg cells and sperm cells. However, this still involves copying and passing on DNA.



Describe the structural properties of the DNA molecule, including nucleotide composition and pairing, and the weak bonds between strands of DNA that allow for replication.

DNA Replication

When Walter Sutton and Theodor Boveri proposed in 1902 that chromosomes were the site of genetic information, they knew that chromosomes must be able to exactly replicate so that identical copies could be passed on to daughter cells. However, they had no idea of the mechanism involved.

Base-pairing rules and method of DNA replication are universal.



Explain the importance of complementary base pairing (A-T and C-G).

In 1953, James Watson and Francis Crick discovered that the structure of DNA — a major component of chromosomes — was a double helix, with the two strands being complementary. This was due to the specific base pairing rules: adenine (A) always forms hydrogen bonds with thymine (T), and cytosine (C) always bonds with guanine (G). Watson and Crick noted in their now famous paper on the discovery: *"It has not escaped our notice that the specific base pairing we have postulated immediately suggests a copying mechanism for the genetic material."* This statement proved to be remarkably accurate, and the process of copying the chromosomes is now called DNA replication. The base-pairing rules and method of DNA replication apply to all life on Earth – that is, they are universal. (See Fig. 2.3)

All nucleotides contain a pentose sugar containing 5 carbon atoms. The carbon atoms are numbered 1', 2', 3', 4', and 5' according to their position in the sugar. The phosphate group is attached to the 5' carbon, and the nitrogen base is attached to the 1' carbon. The phosphate group of the next nucleotide in the DNA strand attaches to the 3' carbon of the previous nucleotide in the strand. By convention, humans read DNA and RNA in the 5' to 3' direction.

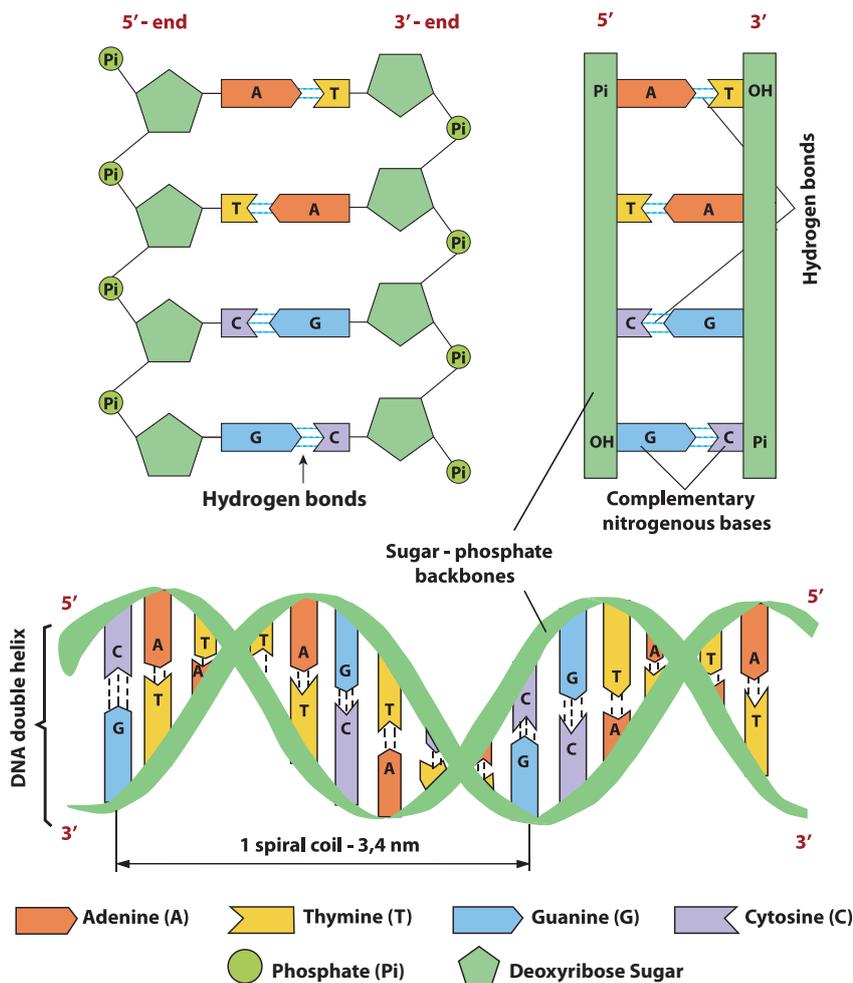


Fig. 2.3 Complementary base pairing



DNA REPLICATION



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DNA REPLICATION ENZYMES

Many different enzymes are involved in the process of DNA replication. Helicases unwind the double helix, primases initiate replication, polymerases catalyse the synthesis of new complementary DNA strands, and ligase joins DNA fragments together. Topoisomerases are involved in re-coiling of the DNA.

DNA is always synthesised in the 5' to 3' direction.



Describe and represent the process of semi-conservative replication of DNA.

The process starts with an enzyme splitting the double helix by breaking the hydrogen bonds between the nucleotide bases on each strand, leaving the bases exposed. Single free nucleotides bond to these exposed bases, according to the base pairing rules. Other enzymes join together the sugar-phosphates of the free nucleotides to form a new DNA strand. (See Fig. 2.3)

The two original strands therefore act as templates for the two new complementary strands. The result is two new DNA double helices that are identical to each other and to the original molecule. When complete, each new DNA molecule consists of one 'old' strand and one 'new' strand.

This process is therefore called semi-conservative replication. This method of DNA replication was confirmed by Meselson and Stahl in the late 1950s, using radioactively labelled nucleotides – a new technique.

After one DNA replication, all of the DNA formed contained radioactive nucleotides. The replication of DNA occurs under the influence of more than a dozen enzymes including helicases, DNA polymerases, and DNA ligases.

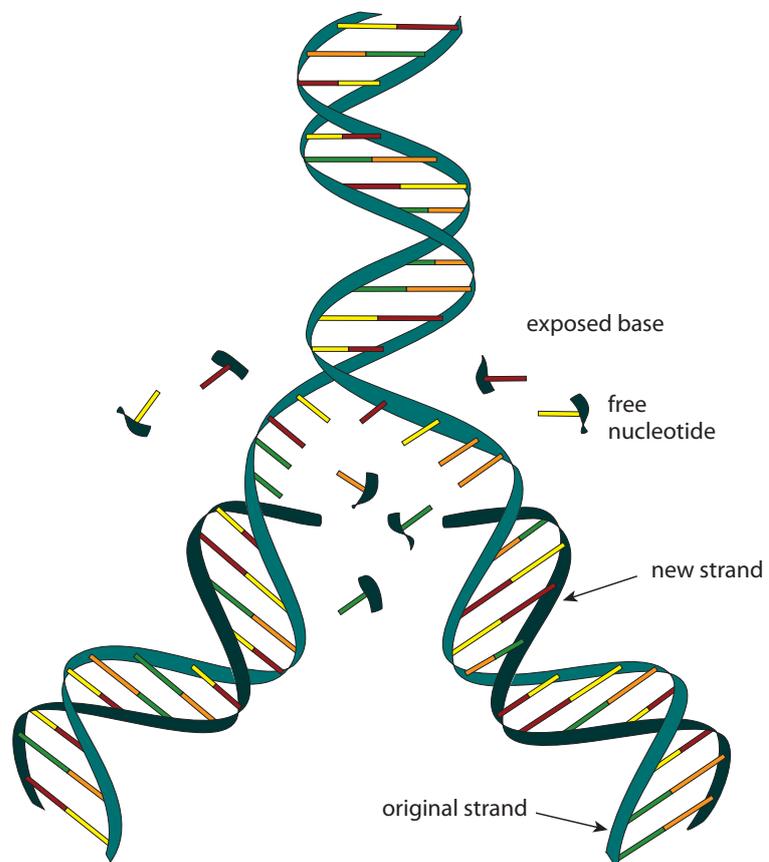


Fig. 2.4 Semi-conservative replication of DNA

The Role of DNA in Cells

All cells are made up of many different chemical compounds. Most of these compounds need to be manufactured by the cell and each manufacturing process requires many chemical reactions. Each reaction step is catalysed by a specific enzyme.

Enzymes are protein molecules made from one or more polypeptide chains and the method by which they work is discussed in Chapter 3. The genetic material in the cell (DNA) contains coded information that directs the synthesis of these enzymes. A sequence of DNA that codes for a particular polypeptide chain or RNA molecule is called a gene. So, as we have already seen, even a simple cell needs several hundred genes in order to make the range of compounds that form its structure, and to carry out the necessary chemical reactions to allow it to survive and reproduce. There are also many proteins in cells that are not enzymes, but instead are structural. Examples include components of the cytoskeleton, such as actin and tubulin.

In some cells even the membrane has more than 50 different proteins, and each of these is coded for by at least one gene. A bacterial cell such as the much-studied *Escherichia coli*, found in large numbers in the human gut, has been shown to contain at least two thousand different kinds of protein molecule that are involved in its metabolism. Each step along a **metabolic pathway** requires a specific enzyme. This means that *E. coli* must have at least two thousand genes to code for at least two thousand enzymes. (See Fig. 2.5) The amount of information stored on the *E. coli* DNA is sufficient to code for almost 4000 average-sized proteins.

Introduction to protein synthesis

Protein synthesis involves transcription of a gene into messenger RNA (mRNA), and translation of mRNA into an amino acid sequence at the ribosomes.

The Genetic Code

Although there are only four different nucleotides present in DNA, there is no restriction on the sequence of bases along the length of a DNA strand. It is this sequence of bases which is the information that directs the cell's activities.

For the information in a DNA molecule to influence the activities of a cell, it first has to be used to make RNA, and then usually translated into proteins. Some proteins function as enzymes, and it is these enzymes, present inside a cell, that will determine the chemical reaction that takes place here. To look at this in reverse,

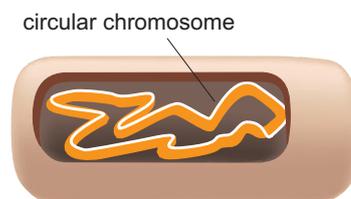


Fig. 2.5 Circular chromosome in a bacterial cell

Proteins are made up of chains of amino acids. The proteins produced by a cell, including the elements of the cytoskeleton and enzymes, determine the structure and function of the cell.

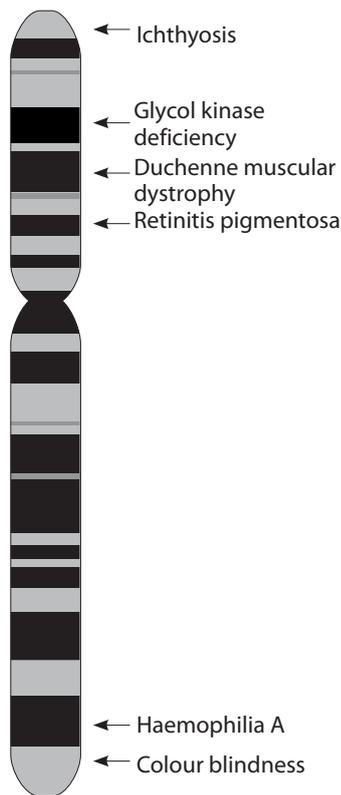


Fig. 2.6 Genes on the human X chromosome

The end products of genes include functional proteins, tRNA, rRNA and microRNA.

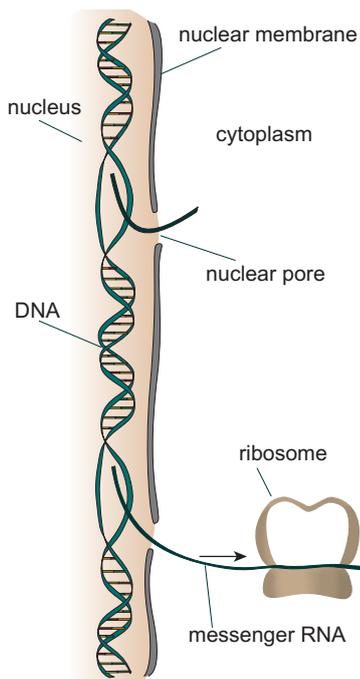


Fig. 2.7 Messenger RNA moving from nucleus to cytoplasm

the metabolism of a cell is determined by the enzymes present in the cell, and these are in turn determined by the information stored in the cell's DNA.

A gene consists of a unique sequence of nucleotides that codes for a functional protein or an RNA molecule.

A gene is a segment of a chromosome that consists of a specific sequence of bases. In fact, one gene codes for a polypeptide or an RNA molecule. (See Fig. 2.6)

In eukaryotic cells, transcription occurs in the nucleus.



Describe and illustrate the role of DNA, mRNA, transfer RNA (tRNA), and ribosomal RNA (rRNA) in transcription and translation.

Protein Synthesis

Transcription: The First Step

The nucleus is like a safe or vault within the cell as it contains the 'master copy' of genetic information in the form of DNA. Since DNA does not leave the nucleus, and the site of protein production is on the ribosomes in the cytoplasm, the information that the DNA contains must first be copied. The copy that is made from the DNA then moves out of the nucleus to the cytoplasm, where it is translated at the ribosomes. As the information in the DNA is copied as RNA, this RNA is called **messenger RNA (mRNA)**, and the process of copying is called **transcription**.

During transcription, the part of the DNA to be copied unwinds, the two strands separate, and the nucleotides that will make up the new RNA molecule begin to line up, using one of the DNA strands as a template. The base pairing rules are the same as for DNA replication, except that uracil (U) is used instead of thymine (T) in RNA. Thus uracil pairs with adenine on the DNA.

An enzyme called RNA polymerase links the new nucleotides together to form the messenger RNA molecule, which then passes through the pores in the nuclear envelope and enters the cytoplasm, where it carries the information to the ribosomes. Meanwhile, the two DNA strands rejoin and the DNA resumes its double-helix shape. (See Fig. 2.7 and Fig. 2.8)



Distinguish between coding (gene) and template strands of DNA.

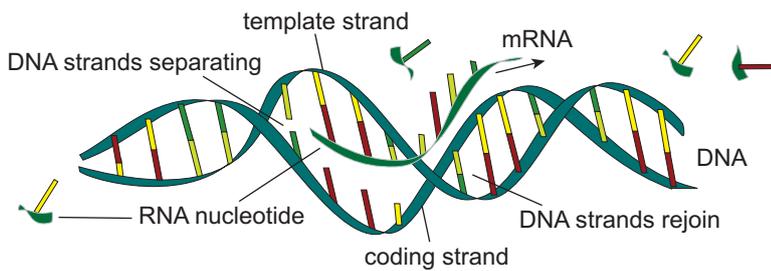


Figure 2.8 Transcription in the nucleus



Recognise that DNA strands are directional and are read 5' to 3'.

By convention, humans read DNA and RNA in the 5' to 3' direction. Also, DNA and RNA are always synthesised in the 5' to 3' direction. However, this means that the enzymes involved (DNA polymerase in DNA replication and RNA polymerase in transcription) must read the template DNA in the 3' to 5' direction. Note that ribosomes read mRNA in the 5' to 3' direction during translation.

On a chromosome, different genes may not be on the same strand of the DNA. What is it that determines which strand of the DNA will be copied? The start codon for copying the DNA is the triplet TAC, which also codes for the amino acid methionine. The DNA triplet TAC indicates where the gene begins, and all polypeptide chains begin with methionine. The two strands of the DNA run in opposite directions, so for genes on one strand, the enzyme RNA polymerase will start at a TAC and travel in one direction. On the other DNA strand, the RNA polymerase will start at a TAC and travel in the opposite direction.



Describe the relationship between DNA codons, RNA codons, anticodons, and amino acids.

Translation: From Nucleic Acid to Protein

The language of nucleotide is now converted to a language of amino acids in a process aptly named **translation**. A protein is made up of a chain of amino acids joined together. The sequence of amino acids defines the function of a particular protein. Some proteins will be structural and become part of cell membranes, while other proteins will be enzymes and thus direct the metabolism of the cell. One way or another, the proteins that a cell produces will have a profound impact on the structure and function of the cell, so it is essential that these proteins are synthesised correctly. When it comes to translation, accuracy is the name of the game.

The messenger RNA, carrying the coded instructions, attaches to a **ribosome**. Ribosomes are themselves made of RNA and protein. The RNA in ribosomes is a special type, produced in a special region of the nucleus called the **nucleolus**, and it is called



DNA IS DIRECTIONAL



tinyurl.com/44s3du3u

ANTIBIOTICS AND PROTEIN SYNTHESIS

Many antibiotics, chemicals that are anti-bacterial, work by inhibiting a step in protein synthesis in bacteria. See Chapter 11 for more information.

PEPTIDES, POLYPEPTIDES AND PROTEINS differ in their size and complexity - see glossary.

UUU	phenylalanine	UCU	serine	UAU	tyrosine	UGU	cysteine
UUC	phenylalanine	UCC	serine	UAC	tyrosine	UGC	cysteine
UUA	leucine	UCA	serine	UAA	stop	UGA	stop
UUG	leucine	UCG	serine	UAG	stop	UGG	tryptophan
CUU	leucine	CCU	proline	CAU	histidine	CGU	arginine
CUC	leucine	CCC	proline	CAC	histidine	CGC	arginine
CUA	leucine	CCA	proline	CAA	glutamine	CGA	arginine
CUG	leucine	CCG	proline	CAG	glutamine	CGG	arginine
AUU	isoleucine	ACU	threonine	AAU	asparagine	AGU	serine
AUC	isoleucine	ACC	threonine	AAC	asparagine	AGC	serine
AUA	isoleucine	ACA	threonine	AAA	lysine	AGA	arginine
AUG	start or methionine	ACG	threonine	AAG	lysine	AGG	arginine
GUU	valine	GCU	alanine	GAU	aspartic acid	GGU	glycine
GUC	valine	GCC	alanine	GAC	aspartic acid	GGC	glycine
GUA	valine	GCA	alanine	GAA	glutamic acid	GGA	glycine
GUG	valine	GCG	alanine	GAG	glutamic acid	GGG	glycine

Fig. 2.9 The mRNA codons for amino acids

ribosomal RNA (rRNA). The conversion of the coded message on the mRNA into a sequence of amino acids involves yet another type of RNA called **transfer RNA (tRNA).**

If each mRNA nucleotide (A, C, G or U) coded for one type of amino acid, then only four different amino acids could be designated, and this is clearly not enough, as there are 20 different amino acids that make up proteins. A code involving pairs of nucleotides would similarly fall short, as it would only allow 16 combinations. Triplets of the nucleotide bases would provide more than enough 'code words'. In fact, there would be 64 combinations to code for the 20 amino acids. This would allow for some 'punctuation marks' as well as having more than one base triplet per amino acid.



Fig. 2.10 tRNA with the amino acid tryptophan attached

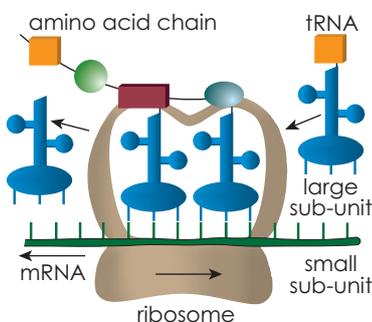


Fig 2.11 Translation at the ribosome

It has been verified that the **genetic code**, as it is called, is indeed made up of base triplets. These base triplets are called **codons**. Each of the 20 amino acids is coded for by one or more of the codons, but an individual codon only codes for one of the amino acids. In other words, several different codons may code for the same amino acid, but a particular codon always codes for the same amino acid. There are also codons for 'start' and 'stop', indicating the beginning and the end of the instruction for the synthesis of a protein. (See Fig. 2.9) The mystery continues, as we find how the message encoded in the DNA is converted into cell proteins.

There is a different tRNA molecule for each of the 20 amino acids. Each tRNA molecule is only about 80 nucleotides long, and is folded into a 'clover-leaf' shape. At one end there is an exposed triplet of bases, called an **anticodon**, while at the other end a specific amino acid is attached. (See Fig. 2.10)

The transfer RNA molecules bring their specific amino acids to the ribosome, according to the instructions coded on the mRNA. The anticodon on the tRNA matches up with the complementary codon on the mRNA. The ribosomes provide the site or 'platform' where the

tRNA and mRNA are brought together. Since the tRNA molecule is carrying a specific amino acid, the codon on the mRNA determines which amino acid arrives next in the sequence. As amino acids are brought alongside one another in this process, they are joined enzymatically with a **peptide link**, to form a growing amino acid chain and the tRNA molecules are released. (See Fig. 2.11)

Eventually, when the polypeptide chain has been completed, it breaks away from the ribosome and folds into its final protein shape. It is then ready to carry out its function within the cell, or perhaps to be secreted from the cell if it has an extracellular function. If we track the whole process of protein synthesis in reverse, we find that the order of amino acids in the polypeptide chain is determined by the order in which the tRNAs are brought to the ribosomes. This is determined by the order of codons on the mRNA, and this order is dictated by the sequence of bases on the DNA in the nucleus.

So you can see that it is ultimately the DNA that determines what proteins are made within a cell, and hence influences the structure and function of the cell. A segment of DNA on a chromosome that contains the complete sequence of bases required to direct the synthesis of a polypeptide is called a gene. It should now be clear how the sequence of bases on DNA in the nucleus is able to function as a genetic code. The whole process of protein synthesis from transcription in the nucleus to translation on the ribosomes is shown in Fig. 2.12.

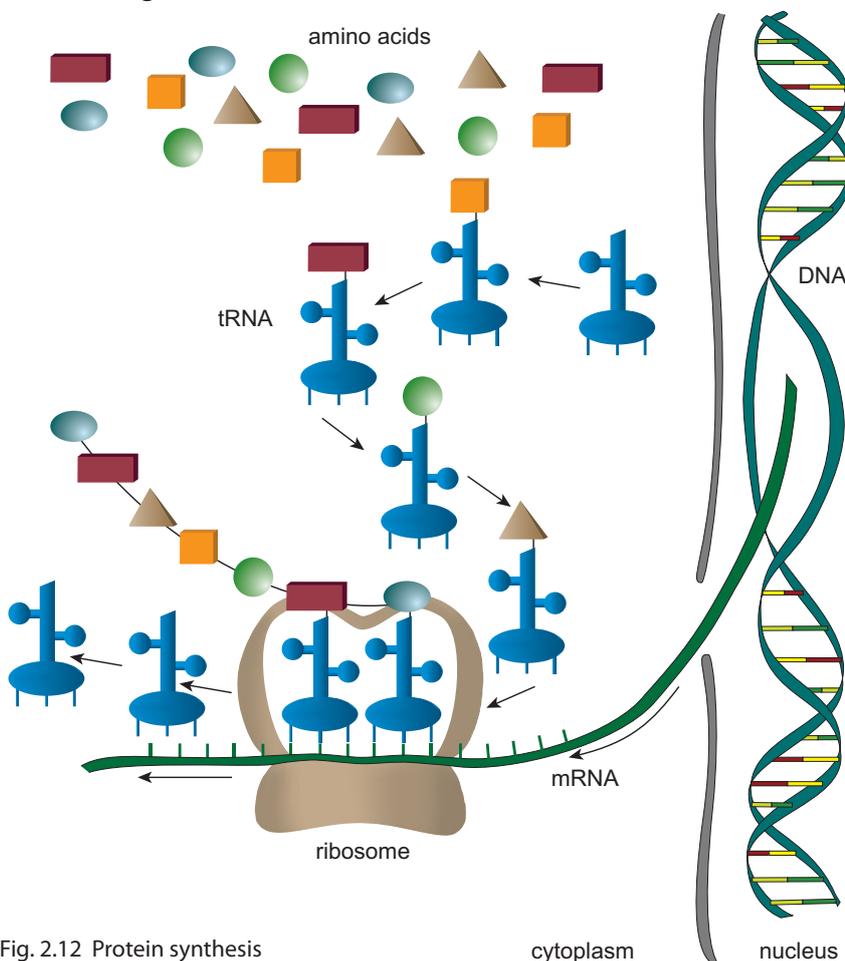


Fig. 2.12 Protein synthesis

READING DNA FROM ANOTHER SPECIES

As the DNA code is **universal**, DNA from one species can be transcribed and translated by an organism from another species. Your cells do this with DNA from the rhinovirus when you get a cold. Human DNA can be read by bacteria to make human insulin and other protein products.

PROTEIN SYNTHESIS



tinyurl.com/n8lvs4r

RIBOSOME BINDING SITES

Recent research that there are three tRNA binding sites on a ribosome.

MOST BACTERIA LACK INTRONS

Some genetic engineering processes use bacteria to produce human proteins, such as insulin or human growth hormone. The human genes that are transferred into the bacteria must first have their introns removed, as bacteria do not have the mechanism to remove introns. You can read more about this in Chapter 6.

Exons and introns



Distinguish between exons that are coding segments of DNA found in genes and introns that are non-coding segments of DNA found in genes in eukaryotes.



Describe how both exons and introns are transcribed, but only the information contained in exons is translated to form a polypeptide in eukaryotes.

REMOVING INTRONS FROM mRNA



tinyurl.com/yddlqdor



tinyurl.com/y8ow66xj

Eukaryotic cells contain introns (noncoding sequences) in their DNA. The messenger RNA produced by these cells has the introns removed before translation into protein. (See Fig. 2.13) To illustrate this, we know that in mice the enzyme dihydrofolate reductase is coded in a region of DNA 31,000 base pairs long, yet the mRNA from this is only 1,600 bases long, and only 558 of these actually code for amino acids. This indicates that only 2 percent of the original DNA is used and the remaining 98 percent of the nucleotides are non-coding sequences. The more primitive eukaryotic cells have hardly any of these introns in their DNA and probably have not changed much from the original cells. In prokaryotic cells introns are rare, and are referred to as Group II introns.

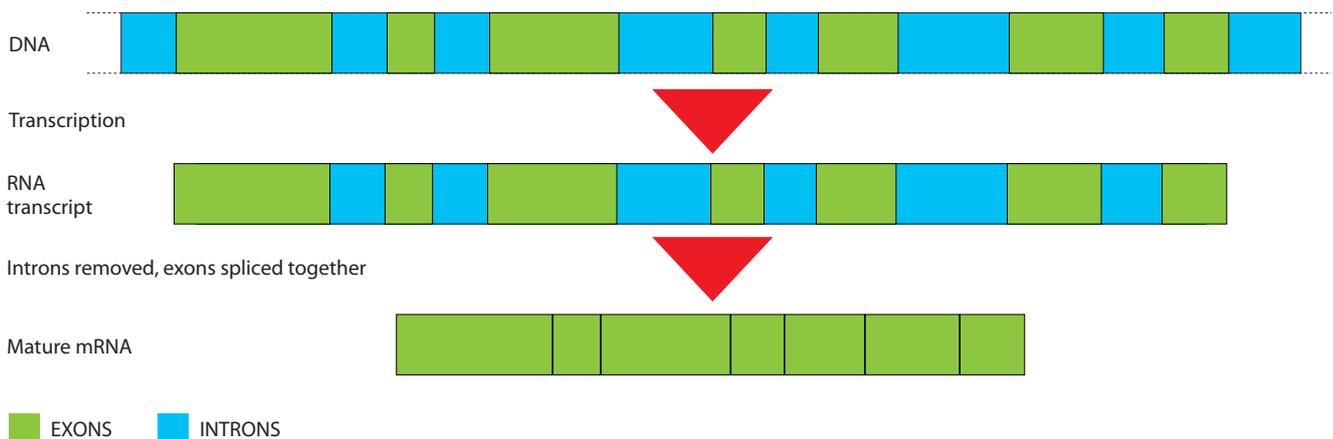


Fig 2.13 Introns and exons

The combination of different exons from a single gene can yield a variety of polypeptides due to alternative splicing. This explains why although humans have about 20,000 genes there are many more than 20,000 different proteins produced.

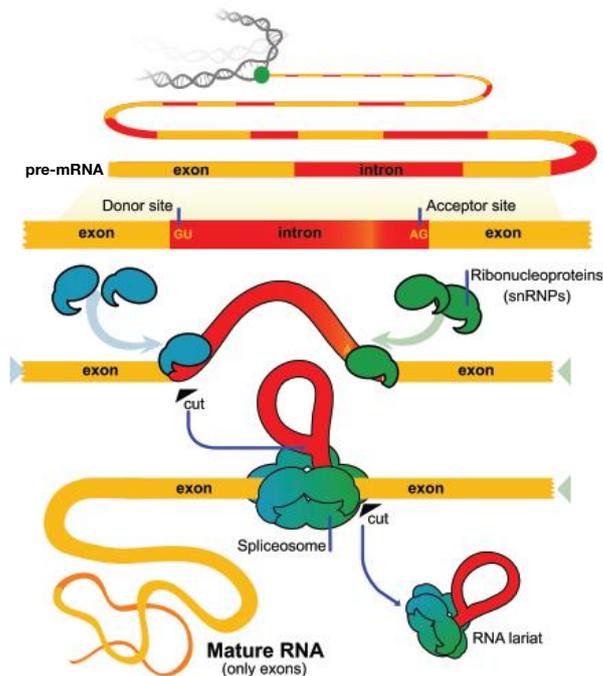


Fig 2.14 DNA to RNA

SPLICEOSOMES are a combination of special RNA and protein molecules that remove introns from pre-mRNA. A loop of RNA, called a lariat, is formed and is excised from the pre-mRNA.

1. Explain the importance of complementary base pairing in DNA molecules.
2. Describe and represent the process of semi-conservative replication of DNA. Illustrate your answer with a labelled diagram.
3. Why is it that some of the information on a DNA molecule must be 'translated into proteins' in order to direct the activities of the cell?
4. Describe and illustrate the role of DNA, mRNA, tRNA, and rRNA in transcription and translation.
5. Describe the relationship between codons, anticodons, and amino acids.
6. Explain why the genetic code must be made up of codons that are at least three bases long.
7. (a) Transcription is the first step in protein synthesis. What is transcription, where does it occur, and what is manufactured as a result of transcription?
(b) Translation is the second step in protein synthesis. Where does translation occur and what is produced as a result of this process?
8. (a) Explain the meaning of the terms codon and anticodon.
(b) What role is performed by transfer RNA (tRNA) in the synthesis of proteins?
9. Distinguish between exons and introns.
10. Describe how both exons and introns are transcribed but only exons are translated.

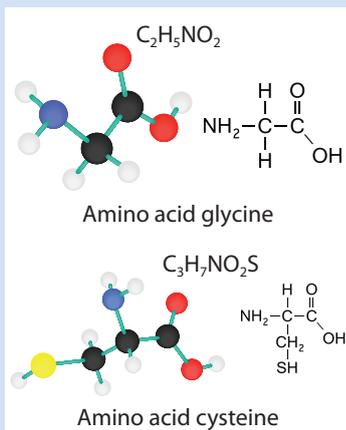
Study Questions

3

Proteins

The folding of a polypeptide to form a protein with a unique three-dimensional shape is determined by its sequence of amino acids.

All amino acids have a central carbon atom surrounded by an amine group, a hydrogen atom, a carboxyl group, and a variable group called an R group. It is the R group that may or may not contain sulfur.



THE LARGEST HUMAN PROTEIN

Titin, found in muscle cells, is the largest human protein, with a length of around 30 000 amino acids. Surprisingly, it was only named in 1979, and its complete amino acid sequence was not determined until 2001.

Proteins

Proteins are macromolecules that are extremely important for the functioning of all cells and organisms. A protein is made up of one or more polypeptide chains. Proteins contain the elements carbon, hydrogen, oxygen, nitrogen, and some contain sulfur as well. The subunits of polypeptide chains are molecules called amino acids and there are twenty different types of amino acid. The reason why some proteins contain sulfur and others do not is that only two of the twenty amino acids contain sulfur. A specific polypeptide is made up of a chain of amino acids in a unique sequence. Since the number of amino acids in a protein molecule can range from about twenty to several thousand, depending on the protein in question, the number of different proteins possible is almost limitless.

The sequence of amino acids in the protein chain determines the way it will fold up after it is made and this folding gives the protein a specific shape. It is this shape that will determine how the protein will function. Nearly all proteins carry out their function by recognising and binding to another specific molecule. For example, enzymes recognise and bind to their substrate and cell membrane receptors recognise and bind to their specific hormones. Although many proteins are globular, they each have a unique shape which matches their partner molecule. To achieve this special shape, the long chain of amino acids must fold in a precise manner. This folding is determined initially by the sequence of amino acids in the polypeptide chain.



Describe the factors that determine the primary, secondary, tertiary, and quaternary structure of proteins.

Biologists describe the structure of proteins at four levels:

- primary structure — this is the sequence of amino acids in the polypeptide chain, held together by peptide bonds

- secondary structure — this is the coiling or folding of localised sections of the polypeptide chain, involving hydrogen bonds
- tertiary structure — this is the three-dimensional shape of the entire polypeptide chain, held together by disulfide bridges
- quaternary structure — if the protein consists of two or more polypeptide chains, then this is the complex structure resulting from their bonding together

Factors that determine protein structure

The **primary structure** of a protein is determined by the sequence of bases on the mRNA that codes for its amino acid sequence. The mRNA base sequence is, in turn, determined by the sequence of bases on the DNA strand that was transcribed (that is, the template strand.) (See Fig. 3.1)

The **secondary structure** of a protein is determined by its primary structure. This determines the position of the hydrogen bonds that produce coils - called alpha-helices, or sheets - called beta-sheets. (See Fig. 3.2)

The **tertiary structure** of a protein (its overall three-dimensional shape) is determined by its primary and secondary structures. Disulfide bonds and hydrogen bonds play an important role. (See Fig. 3.3). The shape of the protein determines its function and enables molecular recognition.

Some proteins (such as haemoglobin) are made up of more than one polypeptide chain. This conglomerate is called a **quaternary structure**. (See Fig. 3.4) Haemoglobin in human red blood cells is formed by the interaction of two alpha (α) chains and two beta (β) chains.

Proteins are essential to cell structure and function.

The functions of proteins in cells include:

- structural — actin and tubulin in the cytoskeleton
- catalyse reactions — enzymes (e.g. polymerase)
- contraction — fibres in muscles (e.g. actin)
- transport — carrying oxygen (haemoglobin); transport proteins in membranes
- defence — antibodies produced by white blood cells (immunoglobulins)
- coordination — hormones (e.g. insulin; thyroxine) and receptor proteins
- storage — albumin in eggs; ferritin (stores iron).



Fig. 3.1 Primary structure of protein

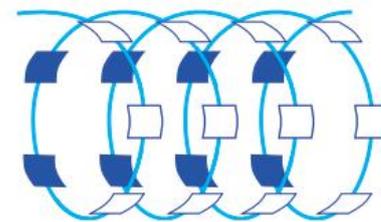


Fig. 3.2 Secondary structure of protein

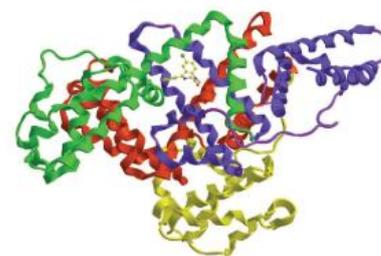


Fig.3.3 Tertiary structure of protein

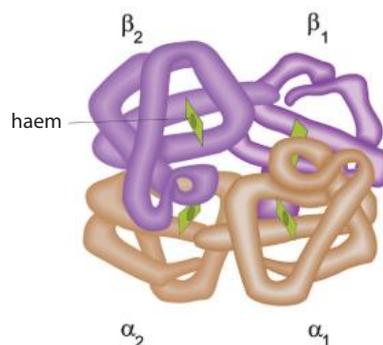


Fig.3.4 Quaternary structure of haemoglobin



tinyurl.com/mqxc5nc

SOME HUMAN HORMONES

EPO - increases production of red blood cells

Insulin - initiates uptake of glucose by cells

Growth hormone - increases muscle mass

Examples of proteins with specific shapes include enzymes, some hormones, receptor proteins, and antibodies.

How important are your senses? How would you function if you received no information from your surroundings?

For you to survive from day to day you use your senses to guide you, protect you from danger, and to make decisions that are beneficial. Your sense system works by recognising signals from the surroundings, and then responding accordingly. Often you are unaware that this is happening. On a smaller scale, your cells work in much the same way. They too have to be able to recognise signals from their surroundings and then respond appropriately. The specific shapes of molecules play an important role in cell recognition and response.

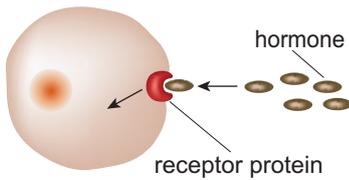


Fig. 3.5 Receptor protein in cell membrane

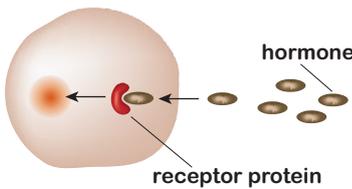


Fig. 3.6 Receptor protein in cytoplasm

Enzymes

The importance of specific shape for enzyme molecules is discussed in detail later in this chapter.

Hormones

Some hormones are made of protein. As a result of being secreted into the blood, a hormone will be transported to all parts of the body by the circulatory system. However, a particular hormone will only produce an effect in cells, tissues or organs that are 'tuned in' to that hormone by having appropriate receptors with shapes that are complementary to part of the hormone molecule. These structures are called target cells, target tissues, and target organs. For example, the target organs for follicle stimulating hormone (FSH) secreted by the anterior pituitary, are located in the testes and ovaries, and FSH will only produce an effect there, even though it will be present in the blood in all parts of the body.

Receptor molecules

Cell receptors are special protein or glycoprotein molecules and the signals that they recognise and respond to are molecular as well.

The protein receptor molecules are either embedded in the bilipid layer of the cell membrane or situated within the cell, and they have distinctive shapes. Protein hormones bind to receptor molecules embedded in the cell membrane. (see Fig. 3.5) Steroid hormones are lipid-soluble and they pass through the cell membrane binding to receptor proteins in the cytoplasm. (see Fig. 3.6) The region of a receptor molecule that has a shape that is complementary to that of a specific hormone is called the binding site. It is the **hormone-receptor complex**, whether on the cell surface or in the cytoplasm, that triggers an effect.

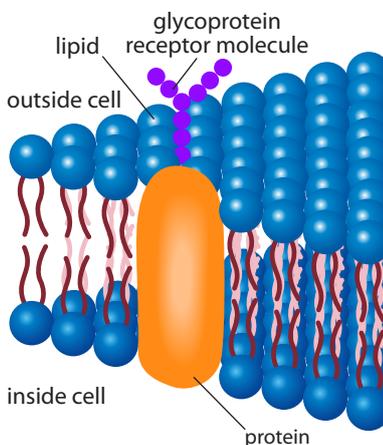


Fig. 3.7 Membrane receptor molecule

The glycoprotein receptors enable cells to recognise one another. This is a critical factor when cells differentiate to form tissues and organs. This cell recognition also forms the basis of specific immunity. (See Fig. 3.7)

Antibodies

Our defence system relies on a range of techniques to protect us from invasion by pathogens — organisms that cause disease. One of these techniques is the production of proteins called antibodies by special white blood cells. Antibodies are made of protein and are sometimes referred to as immunoglobulins, because they are globular proteins involved in immunity. All antibody molecules have the same basic structure, with two regions called antigen-binding sites having a distinctive and specific shape. You may find it useful to compare the complementary shapes of the active site of an enzyme and its substrate when considering the specificity of antigens and antibodies (or more specifically antigenic determinants and antigen-binding sites).

The antigen-binding site specifically recognises the shape of a foreign substance like a bacterium or virus, and will bind to it. (see Fig. 3.8) This neutralises the invading pathogen.

THE 'SELF' AND 'NON-SELF' CONCEPT

The immune system is able to recognise foreign molecules and cells as being different, while ignoring the body's own molecules and cells. The ability to distinguish between your own antigens and foreign antigens seems to be established before birth. Lymphocytes with receptors corresponding to your own antigens are destroyed. The most important surface antigens on the cells of an individual are blood groups on the membranes of red blood cells and a set of marker proteins on the membranes of other cells.

The major blood groups must be matched for blood transfusions. It would be desirable to exactly match the marker proteins on other cells for the purpose of tissue and organ transplants, but the enormous number of possible combinations of marker proteins makes this impossible. The only exception to this is in the case of identical twins as these markers are genetically determined, and identical twins have identical genes. For the purpose of organ and tissue transplants the donor's antigens are matched as closely as possible to the recipient's. Even so, it is necessary for the person receiving the transplant to be treated with an immunosuppressive drug such as cyclosporin. This treatment must be continued throughout the life of the recipient in order to prevent rejection of the transplant. Cyclosporin has the advantage of suppressing only part of the immune response, but even so, the body's ability to combat infection is reduced.

TRANSPORT PROTEINS

Transport proteins that are located in the cell membrane speed up the movement of a substance across the membrane. Like enzymes, they are globular proteins and each one has specific binding sites with specific shapes that bind to the substances being transported. (See Chapter 10 for more detail.)

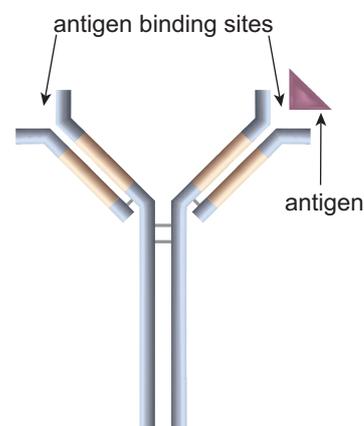


Fig.3.8 An antibody

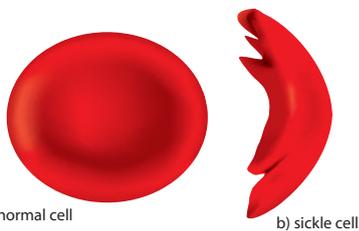
HOW AI CRACKED THE PROTEIN FOLDING CODE



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NAMING ENZYMES

Most enzymes are named using the type of reaction they catalyse or the substrate they act on. The suffix '-ase' is added to complete the name. Examples include intracellular enzymes such as ligase, hydrolase, and catalase and extracellular enzymes such as sucrase, lactase, and lipase. Exceptions to this naming system include pepsin and trypsin.



a) normal cell b) sickle cell

Fig. 3.9 Red blood cells - the faulty haemoglobin causes the cells to change shape.

A metabolic pathway is a sequence of chemical reactions that occurs in a cell.



WHAT ARE ENZYMES?



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Explain why the three-dimensional shape of a protein is critical to its function.

As we have seen, examples of proteins that work by virtue of their three-dimensional shape include enzymes, some hormones, antibodies, and receptor proteins in the cell membrane. Many human genetic diseases are due to the person's cells producing proteins which have an abnormal 3D structure.

- Tay-Sachs disease is due to a faulty enzyme - beta-hexosaminidase A. This results in neurological disorders, an enlarged head, and death in early childhood.
- Phenylketonuria (PKU) is due to a faulty enzyme - phenylalanine hydroxylase. It results in severe mental retardation.
- Type A insulin resistance syndrome is due to a faulty insulin receptor. It results in impaired blood sugar regulation and leads to diabetes mellitus (see page 155).
- Sickle cell anaemia is due to the production of haemoglobin with an incorrect shape. The red blood cells have a fragile 'sickle' shape. As they readily burst, this leads to anaemia. (See Fig. 3.9)
- Cystic fibrosis is due to a faulty channel protein in the membranes of mucus-producing cells in the respiratory system and the intestine. It results in the production of very thick and sticky mucus giving rise to lung infections and impaired digestion.

Chemical factories

One way of thinking of cells is to consider them as little chemical factories. The number and complexity of chemical reactions that take place in an average cell is enormous. Interestingly, most of these reactions would occur very slowly, or not at all, if it were not for special catalysts within the cell. A catalyst is a substance that alters (for our purposes, speeds up) the rate of a chemical reaction, without altering the products that are formed, and without being consumed in the reaction. Consequently the amount of catalyst remains constant and the catalyst needs to be present in only very small amounts because it can be re-used. The catalysts that enable the thousands of reactions in a cell to occur are called **enzymes**.

Enzymes are specific for their substrate

Enzymes are globular proteins, and each cell produces its own enzymes. One very important aspect of enzyme function is that every enzyme is specific for a particular reaction. This means that every step in a metabolic pathway is catalysed by a different enzyme. The kinds of enzymes that a cell produces will therefore determine the chemical processes that can occur within that cell. The majority of enzymes catalyse reactions within cells and are called **intracellular**. Some enzymes, although produced by cells, act outside cells and are called **extracellular** enzymes.

We learned earlier that proteins are made up of chains of amino acids and that each protein has a unique sequence of amino acids that causes it to take on a particular three-dimensional shape. It is this shape that gives rise to the specificity that enzymes display. In order to understand how enzymes work, we need to consider what happens in a typical chemical reaction within a cell. A reaction begins with reactants and ends with products.

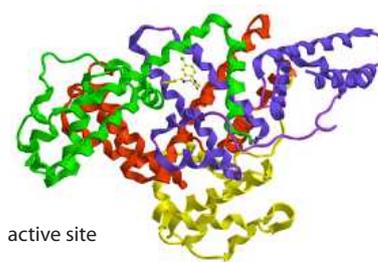
An example is given below:



For the molecules of A and B to react, they must collide with one another in a certain way, and when the correct enzyme is present this is much more likely to occur.

Each enzyme has a region on its surface with a specific shape, and this is called its **active site**. For an enzyme to function it must combine with its **substrate**, and this will only happen if the shape of the active site on the enzyme molecule is **complementary** to the shape of the substrate molecule. Even a slight change to the structure of the enzyme may prevent this recognition, resulting in the enzyme losing its ability to catalyse the reaction. (See Fig. 3.10)

For example, maltase, the enzyme that catalyses the breakdown of maltose, will not catalyse the breakdown of sucrose. This is because the shape of sucrose is not complementary to the active site of maltase. Thus, every step in a metabolic pathway is catalysed by a different specific enzyme. (See Fig. 3.11)



active site

Fig. 3.10 An enzyme is a globular protein

ENZYMES AND HOW THEY WORK



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ENZYMES



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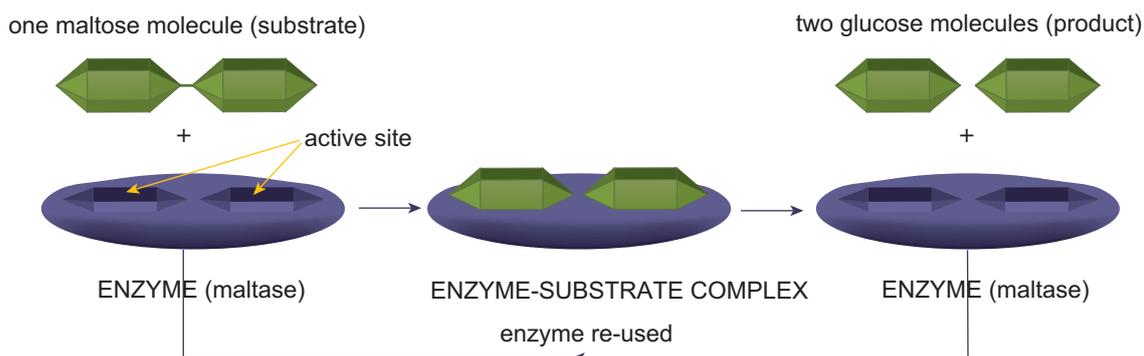


Fig. 3.11 Action of an enzyme

Describe the induced-fit model of enzyme-substrate binding.

The mode of enzyme action can be summarised as follows. First, the substrate binds to the active site of the enzyme forming an enzyme-substrate complex. When the enzyme and substrate join

INDUCED-FIT MODEL



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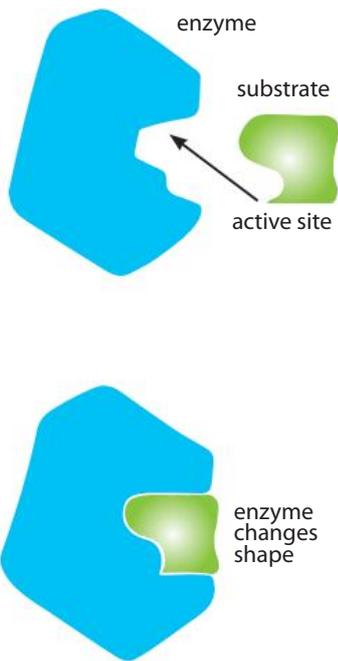


Fig. 3.12 Induced fit model

together, the active site changes shape slightly so that it fits even more exactly to the substrate. This interaction between the enzyme and its substrate is called an induced fit. (See Fig. 3.12) The enzyme-substrate complex then undergoes the reaction far more readily than the substrate would have if there were no enzyme present. The product molecule (or molecules) then breaks away from the enzyme, which is now free to combine with another substrate molecule and repeat the process.

Enzymes have specific functions and are affected by factors including:

- › temperature
- › pH
- › presence of inhibitors.

Due to their protein nature, enzymes are very sensitive to changes in their immediate environment. Their activity is affected by the temperature, the level of acidity (pH), and the presence of chemical inhibitors. Different enzymes operate best at different temperatures and pHs.

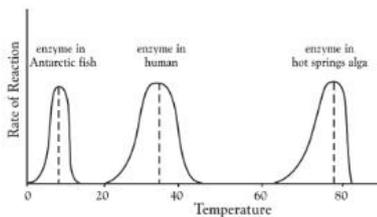


Fig. 3.13 Different enzymes operate best at different temperatures

Temperature

Most human enzymes work best at 37°C, whereas the enzymes of certain algae that live in hot springs might work best at temperatures between 60°C and 80°C. (See Fig. 3.13) These high temperatures would be sufficient to alter the shape of (denature) most proteins. Obviously, if the shape of the enzyme is altered it will cease to function, as its active site will have been altered.

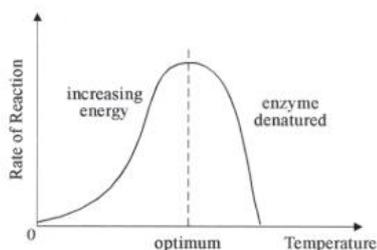


Fig. 3.14 Effect of temperature on enzyme activity

pH

A similar effect on enzyme activity occurs as a result of a change in pH. Once again, different enzymes work best at different pHs. We call the 'best' temperature or pH for an enzyme its optimum. (See Fig. 3.14 and Fig. 3.15) Salivary amylase, an enzyme which digests starch in the mouth, has an **optimum** pH of about 7 (neutral), whereas for pepsin, which digests protein in the stomach, the optimum pH is 2 (acidic).

Inhibitors

Enzymes may be inhibited by certain chemicals. A substance with a molecular shape similar to that of the substrate, for example, may bind to the active site, thus blocking it from the substrate. If the inhibitor were to bind permanently in this way, then the enzyme molecule would be rendered completely ineffective. Inhibitors that act in this way are called **competitive** inhibitors. **Non-competitive** inhibitors act in other ways. For example, a molecule may bind to the enzyme without blocking the active site, but its presence may

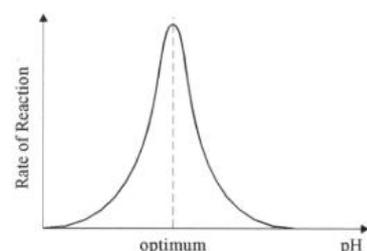


Fig. 3.15 Effect of pH on enzyme activity

distort the shape of the enzyme molecule so that the active site no longer has a shape which is complementary to that of the substrate. Many metabolic poisons work by inhibiting enzymes in one of these ways. (See Fig. 3.16)

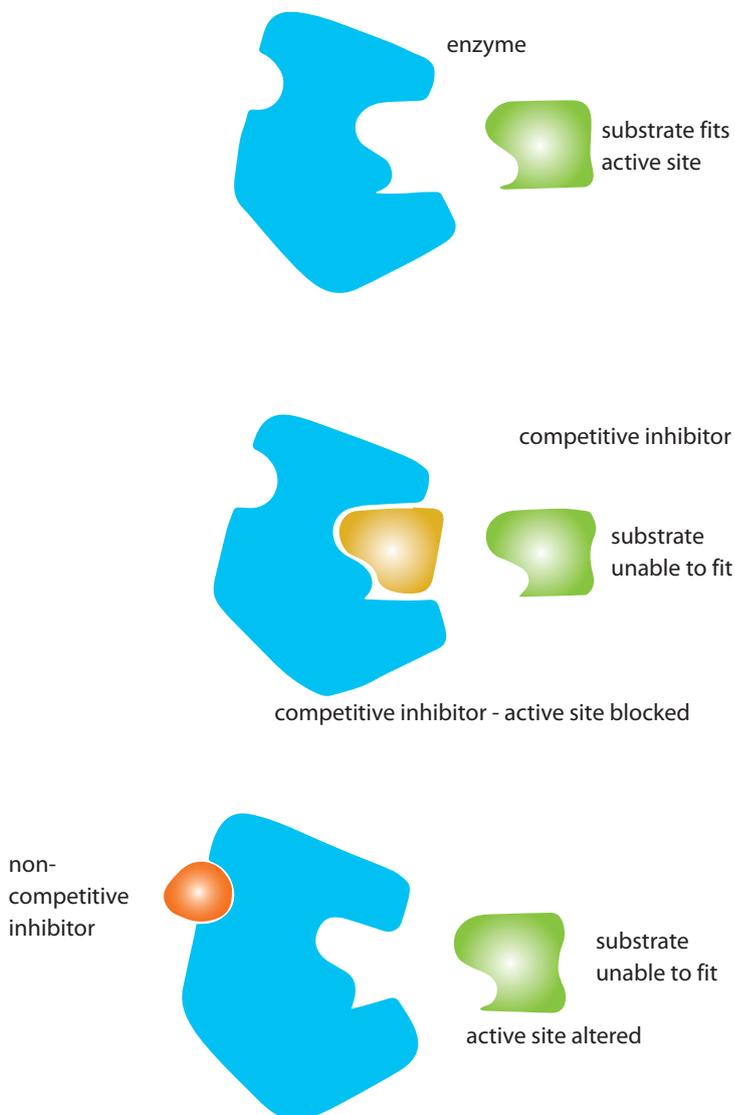


Fig. 3.16 Enzyme inhibitors

The rate of an enzyme-controlled reaction is affected by:

- > concentrations of reactants
- > concentration of the enzyme.

Concentration of reactants

For the same enzyme concentration, increasing the concentration of reactant (substrate) molecules will result in an increase in the rate of the reaction until all of the active sites on the enzyme molecules are occupied. Then the rate of the reaction will remain constant. (See Fig. 3.17)

ENZYME INHIBITORS



tinyurl.com/y9zm9dwo

Examples of chemicals that inhibit enzymes include:
 glyphosate (a herbicide)
 ritonavir (an antiviral drug)
 methotrexate (used in chemotherapy)

CYANIDE

Cyanide is a competitive inhibitor for cytochrome oxidase, an enzyme found in mitochondria. When the active site is blocked, mitochondria are unable to produce ATP, the energy source of cells. Muscle and nerve cells stop functioning and rapid death occurs.

COENZYMES AND COFACTORS

These increase enzyme activity. Coenzymes are organic molecules and cofactors are ions.

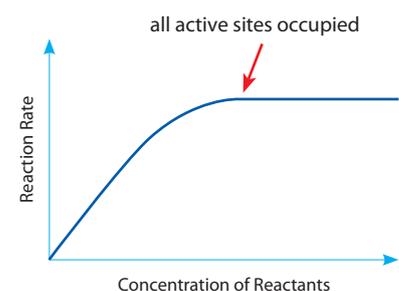


Fig. 3.17 Effect of increasing reactant concentration on reaction rate.

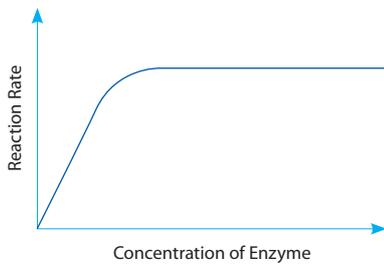


Fig. 3.18 Effect of increasing enzyme concentration on reaction rate.

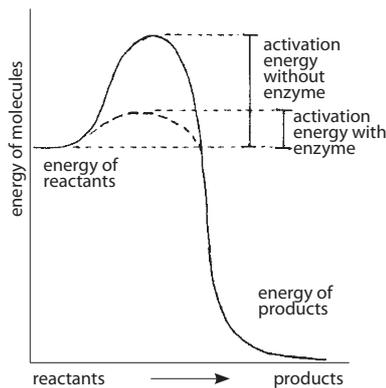


Fig. 3.19 Activation energy

Concentration of enzyme

For the same substrate concentration, increasing the concentration of enzyme molecules will result in an increase in the rate of the reaction until all of the substrate molecules have bound to active sites on the enzyme molecules. Any further increase the enzyme concentration will not increase the reaction rate. (See Fig. 3.18)

Enzymes increase reaction rates by lowering activation energy.

Generally chemical reactions will not start unless there is an input of energy. This energy is called **activation energy** because it 'activates' or initiates the reaction. This energy can be compared to the match that lights the wood of a bonfire. Although the energy from the feeble match flame is insignificant, the enormous amount of energy released from the resulting bonfire would not occur without it.

Biological enzymes have evolved to speed up the rate of many chemical reactions by lowering the activation energy needed to start the process. (See Fig. 3.19) They may do this in one of the following ways:

- by bringing reactants together in the correct orientation, so that they react more readily
- by binding to a substrate molecule in a way that puts a strain on its chemical bonds, thus making it more likely to react. The relationship between the active site of an enzyme molecule and its substrate molecule (or molecules) is explained by the induced-fit model of enzyme action earlier in this chapter.
- by making the reaction happen in several small steps, each of which requires only a small amount of activation energy. A different, specific enzyme controls each step.

Managing the large amount of heat energy released is also achieved by carrying out the overall reaction in several small steps, as each step releases only a small amount of energy. Some of this energy is stored as chemical energy, while the rest is lost as heat.

ACTIVATION ENERGY



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Study Questions

1. Describe the factors that determine the primary, secondary and tertiary structure of proteins.
2. Explain why the 3D structure of a protein is critical to its function.
3. State five functions of proteins and give an example of each.
4. (a) State the function of enzymes.
(b) What are enzymes made of and how do different enzymes differ from one another?
5. (a) Distinguish between intracellular and extracellular enzymes.
(b) Which of these would you expect to be more abundant in humans? Explain your answer.
6. Explain the terms reactant, product, and chemical reaction.
7. (a) What is meant by the term 'specific' as applied to enzymes?
(b) What structural feature of enzymes gives rise to this specificity?
8. (a) Define the term active site.
(b) Where is the active site found?
(c) What is a substrate?
(d) What is meant by the term 'complementary shape' as applied to the active site of an enzyme?
(e) Explain why a different enzyme is required for each step in a biochemical pathway?
9. Describe the induced fit model of enzyme-substrate binding.
10. Using the enzyme maltase as an example, explain how an enzyme works. In your answer refer to the terms substrate, enzyme-substrate complex, induced fit, and products.
11. (a) State three environmental factors that affect the activity of an enzyme.
(b) Explain how each of these environmental factors affects enzyme activity.
12. Represent the pH scale from 0 to 14 on a line. Label acidic, basic, and neutral on your diagram.
13. Draw a diagram to represent how the poison cyanide affects enzymes in the mitochondria.
14. Heavy metals, such as mercury, affect enzymes without attaching to their active sites. Use diagrams to describe how they inhibit enzymes and name the type of inhibition..
15. (a) What is meant by the term 'activation energy'?
(b) What effect do enzymes have on the activation energy required for biological reactions?
(c) Most biochemical reactions in cells occur in several small steps, with each step involving an enzyme. State two advantages of this method for living organisms.

4

Genes & Phenotypic Expression

REPRESENTING GENES

Mendel, the 'father' of genetics, experimented with pea plants. One of the characteristics he looked at was seed shape - either round or wrinkled.

We now call these characteristics phenotypes and the genes that control them are represented by letters. For peas, the gene for round shape is dominant and represented by 'R' while the gene for wrinkled shape is recessive and represented by 'r'.

The phenotypic expression of genes depends on factors controlling transcription and translation. These include the products of other genes, such as transcription factors, and the environment.

Phenotypic expression

The **phenotypic expression** of a gene refers to the physical, biochemical, or physiological characteristics that it produces. Eye colour and height are examples of physical characteristics. Blood group and the ability to produce insulin are examples of physiological and biochemical characteristics. Some characteristics are determined only by genes, others only by the environment, but many are determined by a combination of both.

The **phenotype** of an individual refers to its observable characteristics. Some environmental factors affect the expression of genes, which then affects the phenotype. Other environmental factors directly affect the phenotype. For example, increasing UV exposure will affect the gene that produces the pigment melanin. Loss of a limb will affect the phenotype directly, without affecting gene expression.

There are many ways in which gene products influence the phenotypic expression in an organism. Hormones are an example of a group of gene products that do this. (See table below.)

Gene product	Phenotypic expression
increased EPO	increase in red blood cells
lack of insulin	diabetes
growth hormone	increased body size and muscle mass
auxins	plant stem elongation
gibberellins	flowering, ripening of fruit
testosterone	affects embryonic development and male secondary sex characteristics
oestrogen	female secondary sex characteristics



Fig. 4.1 Red blood cells

GENOTYPE

The genes that an individual organism has for a particular characteristic are called its genotype. An example of genotype is the genes for blood group that an individual has. A child inherits one gene for blood group from their mother and one from their father. There are 3 different forms of the gene for the A, B, O blood group in humans: I^A , I^B , and i .

Gene inherited from mother	Gene inherited from father	Child's genotype
I^A	I^A	$I^A I^A$
I^A	i	$I^A i$
i	i	$i i$
I^B	I^B	$I^B I^B$
I^B	i	$I^B i$
I^A	I^B	$I^A I^B$

The A form of the gene (I^A) directs the formation of the A protein marker on the membrane of red blood cells. The B form of the gene (I^B) directs the formation of the B protein marker. The O form of the gene (i) does not direct any marker formation. A person with the genotype AO has only type A markers and has the phenotype A. A person with both type A gene and type B gene has both markers present and has the phenotype AB. (See Fig. 4.2) Thus, two people with different genotypes could have the same phenotype, as illustrated in the table below.

Genotype	Phenotype (Blood group)
$I^A I^A$	A
$I^A i$	A
$i i$	O
$I^B I^B$	B
$I^B i$	B
$I^A I^B$	AB

While the genes for blood group will be expressed continuously, the expression of some other genes may not be constant. For example, expression of the gene for growth hormone will vary throughout the lifetime of an individual.

As you know from Chapter 2, the expression of genes to form a polypeptide involves transcription followed by translation. The expression of genes to form rRNA or tRNA involves transcription only. The expression of genes can be regulated at either *transcription* or *translation*. Long-term regulation is more likely to occur at the transcription stage, whereas regulation requiring a more immediate response is likely to occur at the translation stage.

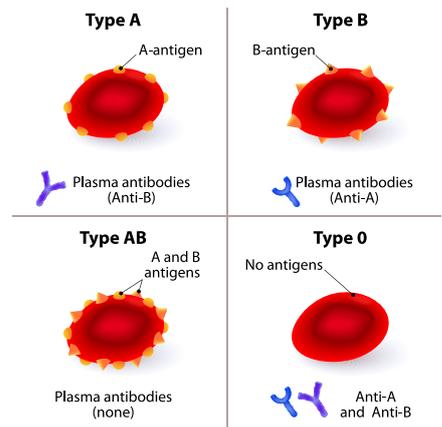


Fig. 4.2 ABO blood group

MATCHING BLOOD GROUPS

Type AB individuals produce no A or B antibodies in their blood. Type A individuals produce anti-B antibodies. Type B individuals produce anti-A antibodies. Type O individuals produce both anti-A and anti-B antibodies. This is why matching blood groups is important.

HETEROZYGOUS OR HOMOZYGOUS

Note that an individual only has two alleles for blood group. Individuals with two identical alleles are called homozygous for that characteristic. If the alleles for the characteristic are different, the individual is said to be heterozygous. (See Fig 4.2)

GENE EXPRESSION



tinyurl.com/nctsgwt

GENE REGULATION



tinyurl.com/h8shy2u

TRANSCRIPTION FACTOR
 p53 tumor suppressor is a transcription factor that promotes synthesis of proteins in cells with damaged DNA, resulting in cell death.

Genetic factors affecting transcription

Genes are not all being transcribed all the time. What determines which genes are expressed, and when? **Transcription factors** are specific regulatory proteins that control gene expression. Some transcription factors switch genes 'on', by binding to a specific site on the DNA called the promoter region. Each gene has a different promoter region, often located some distance from the gene. Other transcription factors turn genes 'off' by blocking the attachment of RNA polymerase to the DNA, preventing transcription. Transcription factors can be activated by specific hormones. This means that gene expression can be determined by the presence (or absence) of certain hormones. (See Fig. 4.3)

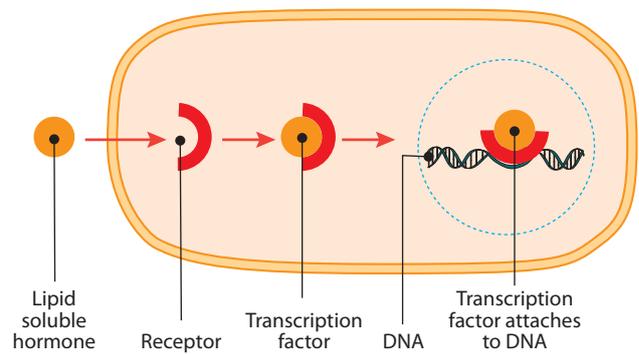


Fig. 4.3
 Transcription factor

Genetic factors affecting translation

Some proteins can prevent translation by binding to mRNA. Also, small interfering RNA (siRNA) can cut mRNA after transcription, preventing it from being translated. (See textbox 'Differentiation of Cells') Also, mRNA degradation prevents translation.

Environmental factors affect transcription and translation

Environmental factors affect protein synthesis and this changes the phenotypic expression in an organism.

Environmental factor	Phenotypic expression
lack of oxygen (e.g. due to high altitude)	increased red blood cell production
increased UV exposure	change in skin colour
lack of iodine	no metamorphosis in axolotl
lack of iodine	goitre in humans
malnutrition	reduced body size
increased light intensity	increased plant growth

In amphibians such as axolotls, (See Fig. 4.4) a lack of iodine in their diet results in the organism remaining permanently in the juvenile stage. Introduction of iodine stimulates the organism to produce a hormone which causes it to progress to the adult stage.

In mammals and birds, exposure to sunlight results in skin cells called melanocytes producing more melanin - a protein pigment which acts as a sunscreen, and makes the skin darker.

Cellular differentiation associated with tissue growth and development is controlled by gene expression.

All cells of a sexually reproducing organism, such as a human, are derived from a zygote which is a single cell that is formed from the union of a sperm and an egg. The zygote contains all the genetic information for that individual, and each time cell division occurs this information is accurately copied and passed on to the newly formed cells. This is described in detail in Chapter 12. How can such an individual contain a huge variety of cell types, when it began life as a single cell? The developing embryo contains at first two cells, then four, eight, sixteen, and so on. Even at this early stage cells are beginning to **differentiate**, that is, become different or specialised. It seems that in each cell only a small part of the genetic information is active, or 'switched on', and that this varies for different cells, depending on their position in the embryo. The result of this cell differentiation is that specialised cells, tissues and organs develop. (See Fig. 4.5)

Initially all of the cells are identical and are called 'stem cells'. As the embryo grows, the cells differentiate and become specialised cells such as muscle cells or nerve cells which join with other cells of the same type to form tissues.

Tissues are made up of cells of like form and function. Examples of tissues are muscle tissue, made of muscle cells, and nerve tissue, made of nerve cells. All cells in an individual organism each have a complete set of genes for that organism. In humans there are approximately 20 000 genes and every cell has all of these genes spread across its 23 pairs of chromosomes.

The differentiation of cells is directed by genes which are activated in the cells. For example, a muscle cell has a specialised cytoskeleton made of proteins which enables it to contract. The synthesis of these proteins is directed by particular genes on the chromosomes. On the other hand, a nerve cell has an extended cell body called an axon. The axon is held in its extended shape by proteins of the nerve cell's cytoskeleton. (See Fig. 4.6)



Fig. 4.4 Axolotl

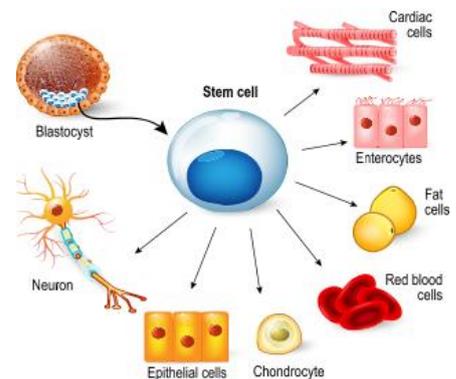


Fig. 4.5 Differentiation of cells

Gene expression can be affected by mutations in non-coding DNA (see page 171).

DIFFERENTIATION OF CELLS

One technique that cells use to differentiate is to silence many of their genes. One of the systems used to silence genes involves non-coding small interfering RNAs (siRNAs) that either bind directly to the DNA or attract other molecules to the sites of transcription or translation and these prevent the process of protein synthesis.

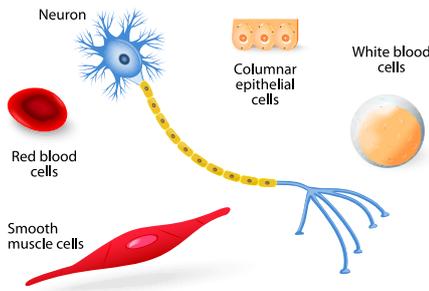


Fig. 4.6 Human cell types

All cells and their respective tissues owe their special development to the genes which are expressed in them. Once a cell has become differentiated it generally cannot change to become a different type of cell. Scientists have been working to reverse the process of differentiation and so create stem cells which could be used to repair faulty or damaged tissue such as nerve tissue in patients with spinal cord injury.

DIFFERENT CELL TYPES

Nerve cells - have specialised structures like a cell body, dendrites and axons. The axon may be very long – over a metre – and it carries nerve impulses from one part of the body to another.

Blood cells - are themselves differentiated into red blood cells and a variety of white blood cells. There are about 5000 million red blood cells per millilitre of blood, making them the most numerous type of blood cell. Each human red blood cell is shaped like a biconcave disk, lacks a nucleus, and is filled with a protein called haemoglobin. These cells, which transport oxygen, are formed from the differentiation of stem cells in the bone marrow and they lose their nuclei before entering the blood stream as mature red blood cells. Some stem cells in the bone marrow differentiate to form a variety of white blood cells with different functions. For example, some differentiated white blood cells secrete antibodies while others are phagocytic.

Epithelial cells - join together to form lining tissue and there is a variety of epithelial cell types. Each type has a specialised shape and features to fit with its function. Some of the epithelial cells lining the trachea have cilia, hair-like projections, that beat rhythmically and move mucus up towards the back of the throat. Other epithelial cells have the ability to secrete mucus. Within a single organism all of these cells contain identical genetic information, but each cell type uses a different portion of this information.

INHERITED OR ACQUIRED?

The idea that characteristics to suit the environment develop during an individual's lifetime and are then passed on to offspring was put forward in the early nineteenth century by Lamarck. However, there seems to be no evidence to support Lamarck's theory of inheritance of acquired characteristics. Imagine a young person losing a finger in an accident. Would you expect some of his or her children to be born with nine fingers?



Recognise that changes in DNA methylation and histone modification can alter gene expression.

We know that, within an individual multicellular organism, different types of cells (differentiated cells) contain the same genetic material, even though they might have quite distinct structures and functions. It is as if the cells all have the same 'library', but they are not reading the same 'books'. For many years the mechanism by which some genes are 'switched off', while others are 'switched on' was a mystery, but it has become clear that genes can become locked 'off' by the addition of a methyl group (CH_3) usually to cytosine nucleotides. (See Fig 4.7). Adenine may be methylated, particularly in prokaryotes.

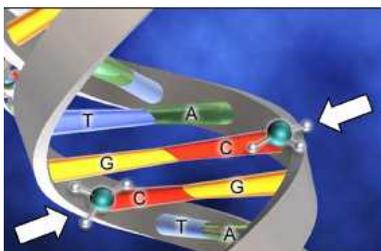


Fig. 4.7 Methylation of cytosine

This area of study has been referred to as '**epigenetics**' ('epi' meaning 'over' or 'after'). There is little doubt that epigenetic mechanisms, such as the methylation of cytosine, play an important role in the development of organisms, and the differentiation of

cells. (See Fig. 4.7) A more contentious issue revolves around the broader use of the term 'epigenetics' to suggest a more 'Lamarckian' view that characteristics acquired during an individual's lifetime can be inherited. (See textbox 'Inherited or acquired?') There is little evidence to support this idea, and any examples are extremely rare, indicating that it is not a significant driving force in evolution.

Epigenetic changes can lead to phenotypic differences between identical siblings, phenotypic differences between clones, and may cause human diseases.

Examples of human diseases caused by epigenetics (usually excessive cytosine methylation) include Fragile X, Prader-Willi, and Angelman syndromes.

Fragile X syndrome is due to methylation of cytosine in CGG repeats in the FMR1 gene. This methylation prevents the production of a protein that is needed for normal brain development. This occurs in individuals with more than 200 of these CGG repeats. This is a mutation, as normally the number of CGG repeats is between 6 and 44. Fragile X syndrome results in many symptoms, including chromosome instability and intellectual disability.

Prader-Willi and Angelman syndromes are due to faults in genes in the same region of chromosome 15. Due to epigenetic changes (methylation), normally genes on either the maternal or paternal chromosome 15 are switched off. In Prader-Willi syndrome the maternal genes in that region are switched off and there is a fault in the same region on the paternal chromosome 15. In Angelman syndrome the paternal genes are switched off and there is a fault on the maternal chromosome 15.



Explain how epigenetic modifications in genes that control cell division, such as changes in DNA methylation, can lead to cancer.

Human cells contain tumour suppressor genes that regulate cell division, and reduce the incidence of cancer. If these genes become methylated (an epigenetic change), then they will not be transcribed, their protein products will not be available to control cell division, and cancer may result.

Changes in the DNA sequence are called 'mutations'.

How could such a diversity of life that we see today have arisen from common ancestry? The most plausible answer stems from the fact that changes in the genetic material occur all the time and these can arise either spontaneously, or be induced. Such changes are called mutations.

Rett syndrome is 'epigenetic-like', in that it results from a gene being expressed that should be turned off. However the cause of this is the production of a faulty protein - the result of a mutation in another gene.



FRAGILE X SYNDROME



tinyurl.com/ymc7pbeh

HISTONE MODIFICATION

Modification of histones, such as acetylation and methylation, can affect transcription of DNA, and thus alter gene expression. Whether the histone modification increases or decreases gene expression depends on factors such as the amount of acetylation and methylation, and their location within the histone protein. For example, usually methylation of histones decreases gene expression, but if the amino acids lysine or arginine in histones are methylated an increase in gene expression can result.

Also see pages 7 and 8.

HUMAN DNA

There are about 3 billion (3×10^9) base pairs in the human genome. Only about two percent of these base pairs make up genes. For humans, there are 20 000 to 25 000 genes, distributed over our 46 chromosomes. The fact that 99.9 percent of the genome is identical for everyone explains why we are all recognisably human. The enormous number of different combinations made possible by the variation in the other 0.1 percent, or 3 million base pairs across the entire genome, (of which about 60 000 base pairs are in genes) explains why we are all unique.

Mutations in genes and chromosomes can result from errors in DNA replication or cell division, or from damage by physical or chemical factors in the environment.

Mutations can occur spontaneously, such as copying 'errors' when DNA is replicated. Cells do have a mechanism for correcting such errors, but even so, some may remain. Factors that can induce mutations are discussed below.

The Genetic Code and Gene Mutations

As we saw in Chapter 2, the genetic code which is carried on the DNA in the nucleus determines the sequence of amino acids in the cell's proteins, and that in turn determines the structure and function of each and every cell and ultimately the whole organism. Any change to the sequence of nucleotides in the DNA can produce a corresponding change in a protein within the cell. This can lead to changes in the individual's characteristics. Even small changes to the nucleotide sequence can have significant effects.

For example, in humans, sickle cell anaemia results from the production of faulty haemoglobin due to just one nucleotide change out of many hundreds in the gene for one of the polypeptide chains in the respiratory pigment haemoglobin. Recipients of two sickle cell genes have red blood cells that are very distorted, and this generally leads to premature death. However, receiving just one sickle cell gene can provide resistance to malaria — a most beneficial result in places like Africa where malaria kills millions of people every year.

Tay-Sachs disease is one in which the nervous system is damaged due to a genetic defect. A single change to a nucleotide in the gene for the enzyme hexosaminidase is virtually a death sentence for the recipient.

Cystic fibrosis is a genetic disease that affects about 1 in every 3000 live births in Australia. Most cases of cystic fibrosis are due to the inheritance of a mutated gene coding for a polypeptide that is inserted in cell membranes. The mutation is due to the deletion of three nucleotide pairs from chromosome number 7. The resulting polypeptide lacks the 508th amino acid (phenylalanine) and it folds differently from the normal molecule. This causes it to fail to function correctly, and all glands that secrete fluids then malfunction, as do the cells lining the lungs and the intestine. Approximately 1 in 25 people carry the faulty gene for cystic fibrosis. However, to suffer from the disease it is necessary to inherit a faulty gene from each parent. (See Fig. 4.8)

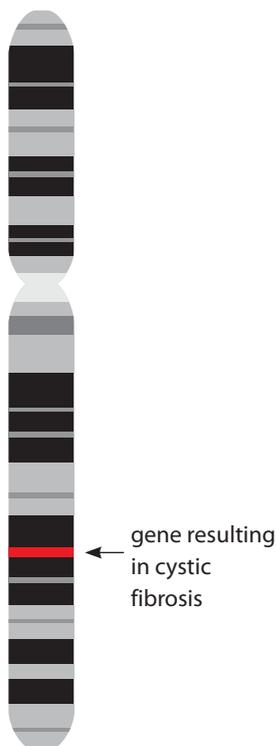


Fig. 4.8 Chromosome 7 and the cystic fibrosis gene.

Mutation rate can be increased by:

- > ionising radiation
- > mutagenic chemicals
- > viruses.

Mutations can be induced by environmental factors, such as high energy radiation (e.g. X-rays and ultra-violet), mutagenic chemicals and viruses. There are many documented examples of mutagenic chemicals and one good source of these is cigarette smoke. Many forms of cancer are the direct result of mutations. (See Fig. 4.9) Chemical agents that cause or increase the incidence of cancer are called carcinogens. This is discussed in more detail in Chapter 14 under the heading 'Uncontrolled cell division – Cancer'.

Mutations can also result from viral infection, if some of the viral DNA becomes incorporated into the host cell DNA. For example, hepatitis can result from the incorporation of viral DNA into the DNA of liver cells.

We cannot escape from random processes that can cause mutation. The soil we tread is slightly radioactive; especially in regions where the bedrock is granite. Cosmic rays are passing unseen right through us all the time. Ultraviolet light, which has photons energetic enough to damage molecules inside cells, bombards us during daylight hours. Chemical mutagens are in the air we breathe, the food we eat, and the liquids we drink. There is therefore an underlying rate of mutation in our lives, and the lives of all organisms, including bacteria.



Compare the different potential consequences of mutations in germ cells and somatic cells.

Mutations that occur in somatic (body) cells will be confined to the individual organism in which they occur. They may even be confined to a particular tissue or location. However, if mutations occur in germ cells (cells that produce gametes), then there is the potential for them to be passed on to the next generation.



Explain how inheritable mutations can lead to changes in the characteristics of the descendants.

Mutations that are inherited via gametes will appear in the zygote and then in every somatic cell and germ cell of that individual.

FACTORS CAUSING MUTATION



tinyurl.com/2p8efwyj

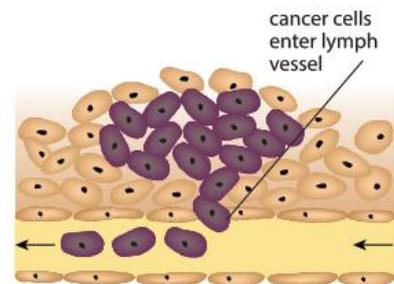


Fig. 4.9 Cancer cells.

ADDITIONAL SOURCES OF GENETIC VARIATION

If we include the effects of the following processes, the number of possible outcomes in the offspring, particularly for sexually reproducing species, is enormous.

- Possible major freakish alteration of chromosomes, such as extra chromosomes or inversions or even doubling of chromosome number in a cell.
- The changes brought about by 'jumping genes' (transposons).
- Insertion or removal of virus genes into the genotype.
- The sudden possible restoration of function of previously silent copies of a gene.

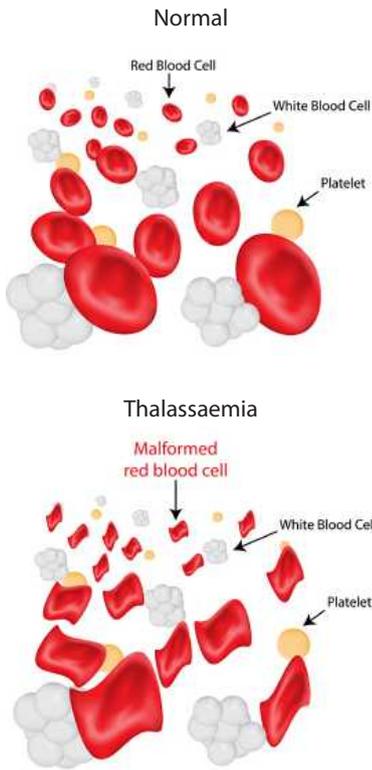


Fig. 4.10 Thalassaemia major red blood cells

Genetic Abnormalities

Some human diseases are genetic, which means that they are not infectious, but instead they are transmitted from one generation to the next via the genetic material. Children will not always inherit such a disease from an affected parent; there is usually an element of chance involved.

Examples of such genetic diseases include phenylketonuria (PKU), thalassaemia, sickle cell anaemia, and haemophilia. Phenylketonuria is a rare condition and for an individual to inherit this disease each parent must have at least one gene for PKU.

Individuals with one gene for thalassaemia have thalassaemia minor, and under most conditions they suffer no ill effects. In fact, most people with thalassaemia minor are unaware that they carry the thalassaemia gene, although it can be detected by a simple blood test. This gene is relatively common in people of Mediterranean or Asian background. People with two genes for the condition suffer from thalassaemia major, which results in deformed red blood cells that rupture easily causing anaemia. (See Fig. 4.10)

Haemophilia is a genetic disease, which has become particularly famous because a classic example occurred in the royal families of Europe in the nineteenth and early twentieth centuries. Queen Victoria of England passed the gene on to several of her children and it eventually spread to the son of the Czar of Russia and to the Spanish royal family. (See Fig. 4.11)

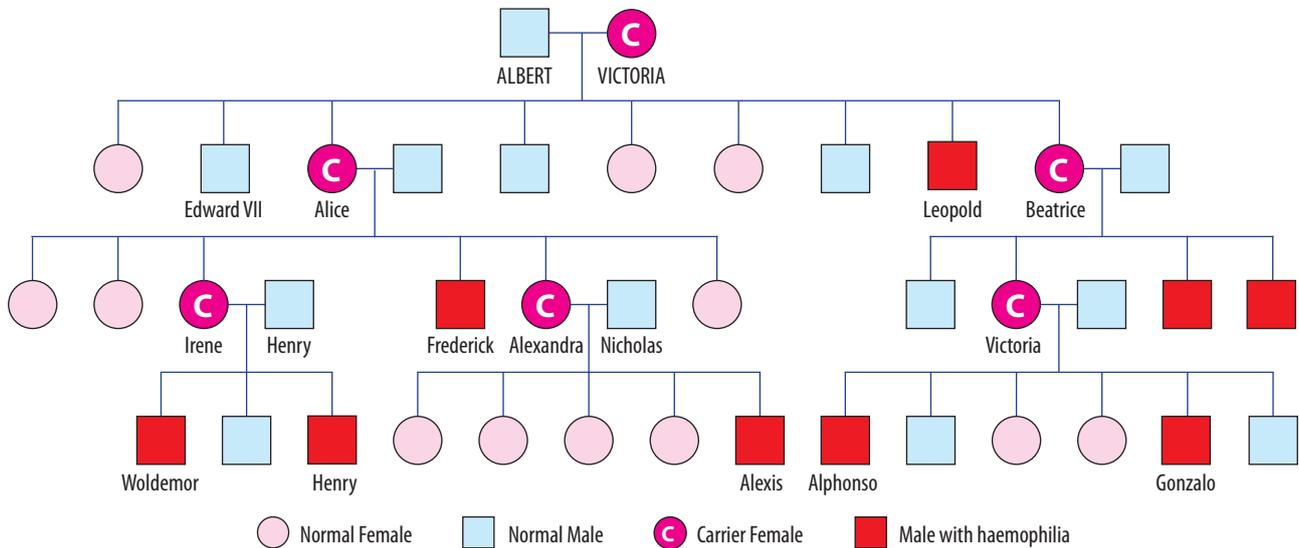


Fig. 4.11 Haemophilia in Queen Victoria's family (partly represented)

In all of the examples below except haemophilia, the disease is due to the presence of two mutated forms of the normal gene. In these cases an individual must inherit a faulty gene from each parent and therefore no normal form of the gene is present.

In the case of haemophilia, the gene is carried on the X chromosome. (See Fig. 2.6) As males have only one X chromosome, they only need to inherit one faulty gene in order to suffer haemophilia. Females have two X chromosomes, and so must inherit two faulty genes in order to have haemophilia.

There are other cases, such as the nerve disorder Huntington's disease, in which only one faulty gene needs to be inherited by either males or females. This is a dominant gene and it produces its effect even if the normal gene is present.

While it is interesting to trace the inheritance of these genetic diseases, you should realise that all genetic variations between individuals in a population are due to mutations having occurred at one time or another.

Chromosomal Abnormalities

Some mutations affect only a single gene, even a single nucleotide, and may produce a severe effect, a minor effect, or no effect at all. Other mutations involve whole chromosomes or parts of chromosomes, and these generally have profound effects on individuals. Down syndrome is an example in which individuals have an extra chromosome 21. (See Fig.4.12)

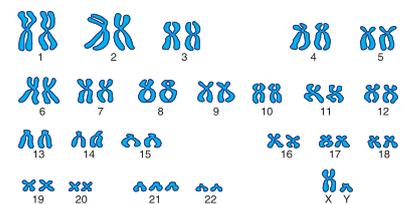


Fig.4.12 Down Syndrome Karyotype

1. Define the terms genotype and phenotype and provide examples for each.
2. Use a labelled diagram to explain how gene expression is controlled by transcription factors.
3. List five examples of environmental factors that affect gene expression and indicate the result for each.
4. Describe how methylation of cytosine nucleotides in DNA can alter gene expression contribute to cell specialisation.
5. Explain how epigenetic modifications in genes that control cell division, such as changes in DNA methylation, can lead to cancer.
6.
 - (a) What is a mutation?
 - (b) What do we mean when we say that a mutation is spontaneous?
 - (c) State three different factors which can increase the mutation rate in humans.
7. Compare the different potential consequences of mutations in germ cells and somatic cells.
8. Explain how inheritable mutations can lead to changes in characteristics of the descendants.
9. Describe two genetic diseases and how they originated.

Study Questions

5

The Use of Genetic Information



PCR FACT SHEET



tinyurl.com/y6qh4vrm



PCR



tinyurl.com/y6w63mdd



PERSONAL DNA TESTING



tinyurl.com/y78yb24u

A DNA PROFILE PROBLEM

In some rare cases, individuals have been found to have two separate DNA profiles – DNA chimerism. This is the result of a foetus absorbing some of the cells of its non-identical twin. The hybrid foetus develops into a person who has one set of DNA in some organs and another set in other organs. The DNA profile from their cheek cells can be different from that of their blood cells!

DNA can be extracted from cells.

To extract DNA from cells, the cell membrane needs to be broken to release the contents of the cells. Enzymes are used to remove histones and other proteins from the DNA. The DNA can then be isolated by treatment with ethanol, or by centrifugation.

Modern techniques can be used to analyse even small amounts of DNA.

DNA can be isolated from even a single cell and then amplified (copied) to the point where there is enough to analyse and sequence it. DNA isolated and amplified in this way can then be used in forensic science and genetic engineering.

Segments of DNA can be multiplied using the polymerase chain reaction (PCR).



Describe PCR, including the roles of:

- › heating and cooling
- › primers
- › free nucleotides
- › heat-resistant enzymes

Polymerase chain reaction

• The technique of repeatedly copying a very small amount of DNA without contamination was developed in 1983 and also earned a Nobel Prize for the inventor, Kary Mullis. The technique relies on a feature of the double helix, that it 'melts' or separates into two strands on heating, and that on cooling, the double helix reforms spontaneously by specific base pairing. But if you just start with a double strand, and allow the separated strands to rejoin when they are cooled, you will not have made any more DNA. The trick is to stop the original strands from rejoining, and have these single strands act as templates for the enzyme, DNA polymerase to complete the double strand. This is why DNA primers are used. Primers are very short stretches of DNA with complementary bases to those at the start of the DNA that is being copied. When they join to the DNA, they stop the DNA strands rejoining and act as a starting point for replication.

Enzymes are made of protein and will normally denature at the temperature needed to 'melt' the DNA. Researchers trying to solve the problem of multiplying DNA put in special DNA polymerase enzymes extracted from bacteria that live at very high temperatures. These **heat resistant enzymes** will not denature when the temperature is raised to the 'melting' point of the DNA.

Many copies of the primer are included in the mixture of **free nucleotides**, DNA polymerase, and the original DNA molecule. This increases the chance that each single strand of the original DNA molecule will bind to primer on cooling rather than to its former partner. The polymerase enzymes bring free nucleotides into place to bind to the single strands of DNA. You will now have two double strands instead of the original one. If the solution is warmed and cooled many times, the original DNA will turn into thousands of accurately copied strands, and all you have to do is keep the free nucleotides and primer in excess. (See Fig. 5.1)

The whole process is known as the polymerase chain reaction, or PCR for short, and it is the basis for much of the modern DNA technology. Nowadays the repeated heating and cooling of the DNA is automated and laboratories carry out the reaction in a machine that is about the same size and complexity as a home breadmaker. (See Fig. 5.2)

Before the invention of PCR the only way to make multiple copies of a DNA sequence in a short time was to splice DNA into a bacterial plasmid and then introduce the recombinant plasmid into a bacterial host cell. Under suitable conditions the bacterial cell divided rapidly, making new copies of the recombinant DNA with each division. This process of cloning recombinant DNA is still used if a large quantity of the protein needs to be produced from the recombinant gene. For example, the cloning recombinant DNA technique has been used to insert the gene for making human insulin into bacteria. The bacteria are then cultured to produce large quantities of human insulin. (See Fig. 5.3)

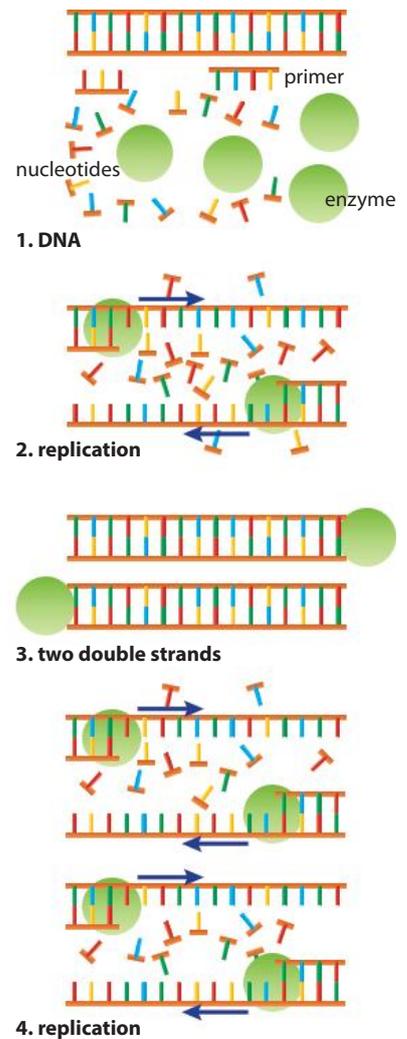


Fig.5.1 Polymerase chain reaction

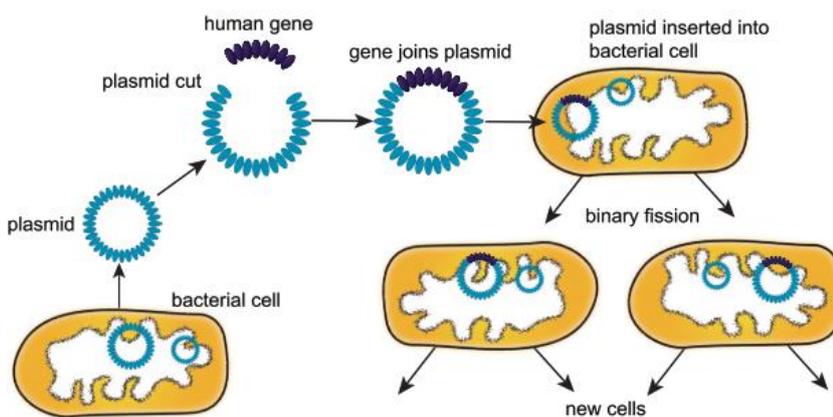


Fig. 5.3 Gene cloning



Fig. 5.2 PCR machine

Sequencing DNA

The base sequence of DNA can be determined by electrophoresis.

In the 1960s an English biologist named Fred Sanger devised a method for determining the sequence of bases on a segment of DNA. He heated the DNA so that its two strands separated. He then added DNA primers, DNA polymerase, and an excess of the four DNA nucleotides — A, C, G and T. A primer is a very short stretch of DNA that matches the start of the DNA that is being copied. The primer joins to one strand of the DNA and prevents it rejoining with its original partner.

Under normal circumstances the primers would combine with single strands and then the DNA polymerase would join the corresponding free nucleotides to the next bases on the single DNA strand until the entire double helix was completed. Sanger's technique was to add to the mixture modified nucleotides that were labelled, in addition to the normal nucleotides. The modified nucleotides prevented DNA polymerase from adding further nucleotides, so the process of DNA replication stopped as soon as the modified nucleotide was in place.

Sanger set the replication process up in four test tubes, each with a different modified nucleotide (A, C, G, and T) that stopped further attachment of nucleotides. For each test tube, Sanger knew the replication process would stop whenever a modified nucleotide was added. He then analysed the lengths of the DNA strands that were formed in each test tube using gel **electrophoresis**, and from this he was able to determine the nucleotide sequence of the DNA — that is, the order of A, C, G, and T. (See Fig. 5.4) On the right of the diagram is an electropherogram, a graphical representation of the results of electrophoresis. It is no wonder that Sanger was a Nobel Prize recipient.

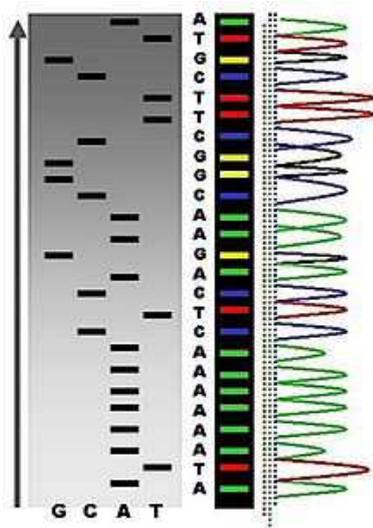
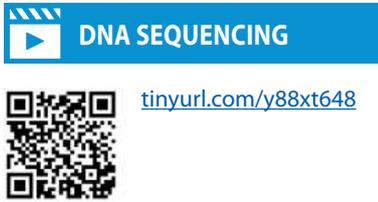


Fig. 5.4 Sequencing DNA using gel electrophoresis. On the right of the diagram is an electropherogram. The terminal nucleotide on each fragment can be identified by the colour it fluoresces.

(Abizar at English Wikipedia)

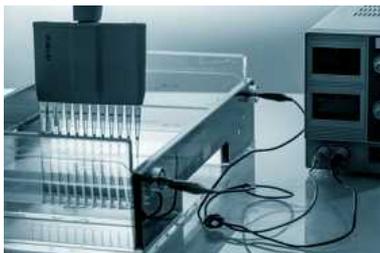


Fig. 5.5 Electrophoresis device



Describe electrophoresis.

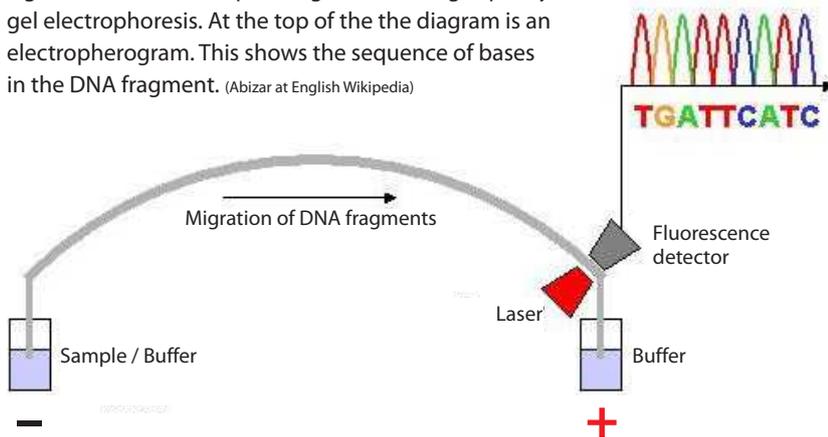
Different length DNA fragments are analysed using gel electrophoresis — a process that separates them according to their size, resulting in a banding pattern. The DNA samples are placed into wells at one end of a block of gel, such as agar, which has electrodes at each end. When the current is turned on, the DNA fragments, which are negatively charged, move towards the positive electrode, with the smaller fragments moving faster than the longer fragments. (See Fig. 5.5)

The results of electrophoresis may be displayed in an electropherogram.



Interpret electropherograms that illustrate DNA sequences.

Fig. 5.6 Automated sequencing of DNA using capillary gel electrophoresis. At the top of the the diagram is an electropherogram. This shows the sequence of bases in the DNA fragment. (Abizar at English Wikipedia)



Modern automated electrophoresis uses the same concept, but is faster and more efficient. Instead of using a block of agar, the DNA is passed through a capillary tube containing the agar. The end nucleotide of each DNA fragment is labelled with a fluorescent dye. The colour of the dye corresponds to the nucleotide. A current is applied, the DNA fragments move through the capillary tube, and a laser reads the dyed fragments as they pass. (See Fig. 5.6)

DNA profiling identifies the unique genetic makeup of individuals.

The use of DNA in forensic science began in 1985 with the work of Sir Alec Jeffreys, who developed techniques for DNA ‘fingerprinting’. He made use of an oddity of higher eukaryote DNA, the ‘junk repeat’. In the region of the centromere of a chromosome, there are long blocks of highly repetitive simple sequences that do not code for anything. For example, in the fruit fly *Drosophila*, the sequences ACAAACT, ATAAACT and ACAAAATT are repeated numerous times. These sequences, called **variable number tandem repeats** (VNTRs) can make up 25% of the total DNA. We now know that these regions of the DNA are not ‘junk’, but the function of these non-coding regions is not fully understood. The frequency of various non-coding repeats is characteristic for an individual, just as fingerprints or eye iris patterns are unique. This enables the DNA to be exactly matched to a person.

The DNA for DNA fingerprinting can be obtained from any cell with a nucleus. Blood, saliva, tears, skin, and semen all contain cells, and once the DNA has been extracted from these sources, the VNTR fragments can be removed using restriction enzymes, and then multiplied using the PCR technique.

These different length VNTR fragments are then analysed using gel electrophoresis — a process that separates them according to their size, resulting in a banding pattern. Every individual has a unique DNA banding pattern, so a suspect’s DNA can be compared to the DNA collected at the scene of the crime. (See Fig. 5.7)

SEQUENCING DNA



tinyurl.com/y3fww2zw

HOW TO CATCH A CRIMINAL



tinyurl.com/2zt7wm8d

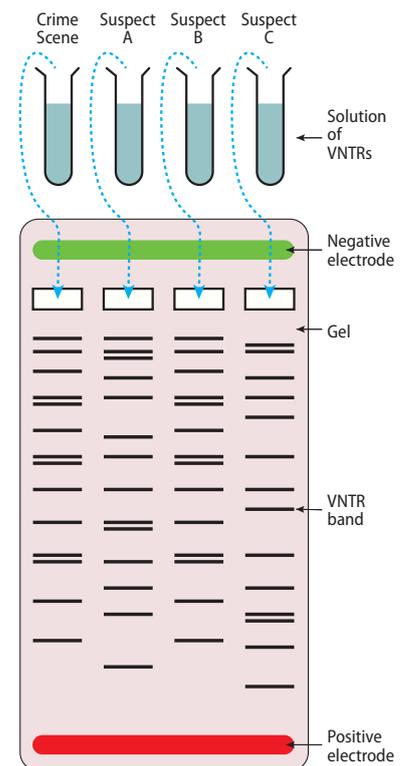


Fig. 5.7 Electrophoresis – separation of VNTRs.

DNA PROFILING AND PRIVACY

One of the concerns people have about the use of DNA profiling is that the profile will reveal information about their traits including genetic faults, potential genetic diseases, and particular characteristics they do not want revealed to others.

However, current DNA profiling only uses STRs that, apart from identifying the sex of the person, convey no other genetic information as STRs are part of the non-coding sections of DNA.

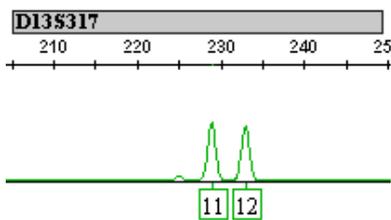


Fig. 5.8 Electropherogram - locus D13S317

 AUSTRALIAN CENTRE FOR ANCIENT DNA



tinyurl.com/yddchglj

The Australian Criminal Intelligence Commission (ACIC) uses the National Criminal Investigation DNA database (NCIDD) to match DNA profiles Australia-wide.

 CIC DNA DATABASE



tinyurl.com/amxp5kmh

VNTR analysis has been replaced by the analysis of **short tandem repeats** (STRs). These regions, scattered throughout the genome, are made up of a repeated sequence of between two and eight nucleotide bases. The analysis of STRs takes much less time than is needed for VNTRs, as automated systems are now used. Also, the results from the analysis of STRs provide greater certainty of identification.

In humans, the DNA that does not code for polypeptides can accumulate a large number of mutations, without influencing the individual's phenotype. Each individual inherits two sets of chromosomes – one set from each parent. For example, one individual might have the base sequence TATC repeated eleven times at the position (locus) D13S317 on the chromosome 13 they inherited from their father, while this sequence is repeated twelve times on the chromosome 13 they inherited from their mother. The sequences below illustrate this example.

From father:

ATCTTCTAACACATGACCGATTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTTCCATGATAGCACAT

From mother:

ATCTTCTAACACATGACCGATTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTATCTTCCATGATAGCACAT

You can see this represented in the electropherogram (See Fig. 5.8).



Explain how differences in DNA fragments, identified by DNA profiling, can be used; for example, in forensic science.

DNA profiling

On a DNA profile the result for this individual at this position (locus) would show as 11/12. Increasing the number of loci (regions of the DNA where the STRs are located) that are used increases the 'uniqueness' of a DNA profile. Analysing markers on the X and Y (sex) chromosomes will reveal the sex of the person. In forensic science, DNA profiles are used to match DNA samples from a crime scene with those from a victim or suspect.

DNA profiling can be used not only for identifying criminals, but also for custody disputes and determining genetic relationships. It is also used to detect viral DNA in blood and genetic defects in human eggs and embryos. This was difficult before PCR because the amount of DNA available is usually minute.

Genetic faults can also be identified. The gene for Huntington's disease was the first to have its position located by this method and

the mapping of the human genome relied on the use of restriction fragment length polymorphism (RFLP) techniques. Palaeontologists have even been able to study the tiny amounts of DNA left in fossils using PCR. Their results have enabled them to identify and classify ancient, long-extinct organisms, as DNA is surprisingly stable. The oldest DNA that has been analysed has lasted almost a million years.

WHAT IS DNA PROFILING USED FOR?



tinyurl.com/3nrd6bzz

The results of electrophoresis can be used to construct DNA profiles. They may be displayed in an electropherogram or in a table of data.

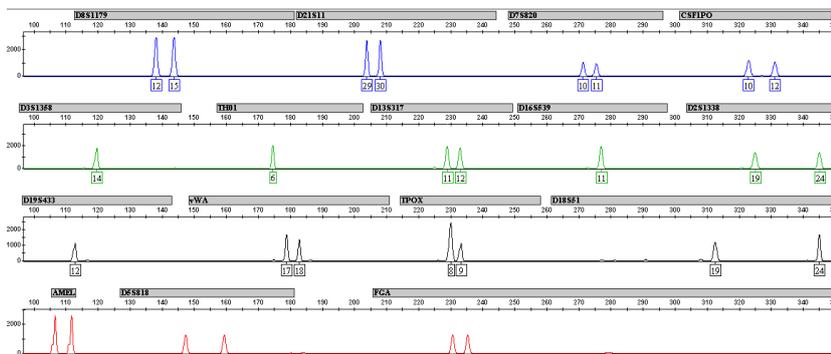


Interpret electropherograms and tables of data that illustrate DNA profiles.

Fluorescently tagged primers are used in the PCR process to amplify the STR regions of the DNA collected. When this amplified DNA is placed into a capillary electrophoresis tube, the smaller STR fragments will move faster. Each STR fragment is detected as it passes a laser beam at the end of the capillary tube. The result is displayed as a series of (paired) peaks on a graph called an **electropherogram** (see Fig. 5.9)

RFLP RESTRICTION FRAGMENT LENGTH POLYMORPHISM

At certain sites on DNA there are sections which have repetitive patterns. The length of these patterns varies from person to person- termed polymorphism. Specific restriction enzymes are used to cut the DNA at these sites. The cut lengths of DNA, called Variable Number Tandem Repeats (VNTR), from different DNA samples, can be compared by electrophoresis. The technique, called DNA fingerprinting, was invented by Sir Alec Jeffreys in 1984 and was the first of its kind to be used in forensic science. It has now largely been replaced by SNP and STR analysis.



Locus	Chromosome	STR	Allele values
D8S1179	8	TCTA	12,15
D21S11	21	TCTA	29,30
D7S820	7	GATA	10,11
CSF1PO	5	AGAT	10,12
D3S1358	3	TCTA	14,14
TH01	11	AATG	6,6
D13S317	13	TATC	11,12
D16S539	16	AGAT	11,11
D2S1338	2	TGCC	19,24
D19S433	19	AAGG	12,12
VWA	12	TCTA	17,18
TPOX	2	AATG	8,9
D18S51	18	AGAA	19,24
Amelogenin	X; Y		X,Y
D5S818	5	AGAT	10,13
FGA	4	TTTC	21,23

DNA DATABASE

Since 2017, there have been 5 more loci added to the 16 used for the Combined DNA Index System (CODIS). This system has been developed in collaboration with the FBI in the USA, and European agencies.

Fig. 5.9 A DNA profile represented by a matching electropherogram and table of data

A TIMELINE OF GENOME SEQUENCING



tinyurl.com/4ejxwyp9

DNA sequencing enables mapping of species' genomes.

Mapping a species' genome involves determining what genes it contains, and their location on its DNA. The first step in this process is to sequence the DNA, that is, work out the order of bases along each chromosome of the entire genome. Computer analysis of the base sequence enables the genes and their location to be determined – a genome map.

Examples of species whose genomes have been mapped entirely include the bacterium *E. coli*, and the yeast *Saccharomyces cerevisiae*. Although the human genome was sequenced by 2000, the mapping of the genome will take many more years.

DNA MICROARRAYS

In the early 1990s Patrick O. Brown developed a technique for identifying large numbers of DNA samples using an array of different DNA probes bound to glass slides in a 'grid' or 'array'. DNA fragments that are complementary to the probes will stick to the probes in specific parts of the grid. The microarray can be examined by a scanning microscope, and a computer can be used to analyse the results. Genetic tests that once would have taken several weeks or even months, can now be completed in a matter of hours. A slightly different technique, using synthetic DNA strands up to 20 bases long was developed at around the same time by Stephen Fodor. These are called GeneChip® arrays (Affymetrix Inc.).

THE HUMAN GENOME PROJECT

It is estimated that there are ABOUT 20 000 genes in a human cell, each with its own specific locus. Thus, a chromosome can be identified by identifying the genes that it contains.

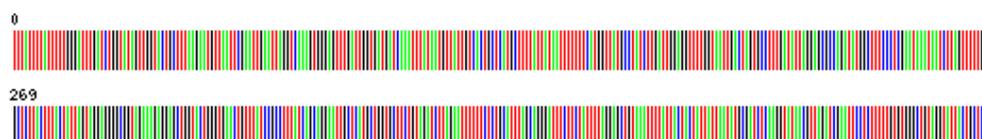
In 1988 the Human Genome Project (HUGO) was proposed. Its aim was to map the entire human genome of one man and one woman, and to store this information in computerised form. Originally it was estimated that this would take 15 years to complete, but the project which commenced in 1990 was completed in the year 2000 - well ahead of schedule due to rapid advances in biotechnology. Even so, its cost has been estimated at \$3 billion, and it is thought that it will take many more years to fully analyse the data collected!

From the information obtained, we already have a much better understanding of the cause and treatment of some genetically determined diseases. Also, we have found more clues as to how cells differentiate, how embryos develop, and we have developed a greater understanding of evolution. As with all such scientific research, there will continue to be many side benefits that were not apparent beforehand.

THE BARCODE OF LIFE DATABASE (BOLD)

The protein cytochrome oxidase is found in the mitochondria of all eukaryotes. There are slight differences in the sequence of nucleotides that code for cytochrome oxidase in each species. These unique sequences can be used to establish a database of different species. It is even possible to construct DNA 'barcodes' to catalogue these species. Uses of the database include identifying commercial fish species and illegal timber. (See Fig. 5.10)

Fig. 5.10 DNA Barcode. Each colour stripe represents a nucleotide.





Discuss the ethical, economic, and cultural issues related to the collection of genetic information.

Ethical Issues

Of concern is the way that humans might choose to use the new technology. The use of DNA profiling in forensic science, which is discussed in detail above, would probably not cause great concern to law-abiding citizens. However, if it becomes possible to inexpensively sequence part or all of an individual's DNA and screen for genetic faults — and this is very likely — should a life insurance company be able to insist on having this information before taking on a new client? Or should an employer be entitled to make it a prerequisite for employment? These tests only require a hair follicle, or even one or two cells from a lip-print on a glass, and so could be carried out without the individual's knowledge or consent.

The collection of genetic information can also be used to diagnose, prevent, and treat certain diseases. The privacy of personal genetic information remains an important issue.

While DNA profiling uses STRs that are non-coding and provide no information about a person's characteristics, **DNA phenotyping** uses coding regions of the person's DNA. It can be used to deduce certain physical characteristics such as eye colour, gender, and ethnic background. While DNA phenotyping is not currently in common usage, it has been used in some countries to identify characteristics of a suspect when a DNA profile is not on their database.

Economic Issues

It costs money to collect genetic information, and then there is the cost of storage and administration of any database that is developed. An example is the **Barcode Of Life Database (BOLD)** – see Figure 5.10. The cost of establishing and maintaining such a database needs to be weighed up against the potential gains from having access to this information. Examples include the protection of fish stocks and forests. It is possible to compare the DNA of fish that are being caught and sold, with the DNA stored in the database, to determine whether endangered species are being exploited. It is also possible to determine whether fish are being falsely labelled in order to inflate prices. In this way, consumers can be protected, and they can be confident that they are getting what they are paying for. Timber from specific forests can also be identified and tracked in this way, making it easier to protect the environment.

SHOULD WE SEQUENCE GENOMES OF BABIES



tinyurl.com/yepkjd9m

WHOLE GENOME SEQUENCING FROM BIRTH



tinyurl.com/upmrpkfd

DNA BARCODE



tinyurl.com/y651vybu

Cultural Issues

We cannot assume that people from all cultures will value or interpret genetic information in the same way. For example, due to cultural mores or religious ideology, they may have differing views on abortion and the importance of genetic disorders. People from some cultures might consider the prediction of diseases or disorders unnatural. Others may not permit invasive procedures that they consider to disturb the natural harmony of the human body. Invasive procedures, such as amniocentesis and taking samples of blood or tissues, are routinely used in the collection of genetic information. The disclosure of this kind of information may also result in certain ethnic or cultural groups being stigmatised, or even victimised, because some genetic conditions are more prevalent within them. Examples of genetic conditions that are more common in certain groups include thalassaemia and Tay-Sachs disease.

Study Questions

1. Describe how DNA can be extracted from cells.
2. The PCR technique enables us to make a huge number of copies of a specific segment of DNA.
Describe PCR, including the roles of:
 - heating and cooling
 - primers
 - free nucleotides
 - heat-resistant enzymes.
3. Electrophoresis is used to sequence DNA. Describe electrophoresis using a labelled diagram.
4. Explain what a VNTR is and illustrate with an example.
5. The results of electrophoresis of several individuals' DNA can be displayed in an electropherogram such as shown in Figure 5.7 on page 43. Explain why the VNTR sequences of DNA separate to form such patterns and why there are differences between the individuals.
6. Explain why STRs are now used to provide unique 'genetic profiles' for individuals in preference to the use of VNTRs.
7. Explain how differences in DNA fragments, identified by DNA profiling, can be used in (a) forensic science (b) medicine and (c) scientific research.
8. How is the Barcode of Life (BOLD) used to protect vulnerable species?
9. The Human Genome Project is one of the most ambitious undertakings ever started by humanity.
 - (a) State three benefits that you predict will result from knowledge of the complete human genome.
 - (b) State three problems that could arise from this knowledge.
10. Discuss the ethical, economic, and cultural issues related to the collection of genetic information.

Biotechnology (Human Manipulation of DNA)



Biotechnology can involve the use of plasmids and viruses as vectors, bacterial enzymes, and yeasts. Techniques include bacterial transformations, electroporation, and microinjection.

From the moment that the structure of DNA and its importance in inheritance was discovered it was inevitable that humans would attempt to exploit this new-found knowledge. 'Cracking' the genetic code and understanding the process of protein synthesis is all that was needed to begin the manipulation of the cell's biochemical machinery. Improvements in our understanding and advances in technology have resulted in biologists now having the ability to alter the genetic instructions in cells. This relatively new type of biotechnology is called **genetic engineering, genetic modification, or recombinant DNA technology**. This is because it often involves combining DNA from different sources, even different species.

The technique of incorporating 'foreign' DNA into organisms is now quite common. Animals and plants produced using this method are called **transgenic** organisms. The required gene is inserted into a fertilised ovum by micro-injection, viral vector, electroporation, or a gene gun, and the resulting organism then contains all of its own genes plus the 'foreign' gene. Currently, these techniques have a low success rate, but they are continuously being refined. More detail is provided later in this chapter.



Describe how particular genes can be selected using probes, and removed using restriction enzymes

Genetic Engineering

When the role of the base sequence in DNA as genetic information became known, it was only a matter of time before the technology was developed to enable DNA from different sources to be combined. Essentially, this new technology involves combining DNA in test tubes (*in vitro*), and then inserting the recombined DNA into cells where it can be expressed, via protein synthesis.

HISTORY OF BIOTECHNOLOGY USING HUMAN GENES



Genes were first transferred by scientists from one organism to another in 1973. As a result of this technology, human insulin was produced using bacteria in 1982. At about the same time, transgenic goats were used to produce human tissue plasminogen activator (tPA) in their milk. tPA is used to dissolve blood clots. It is now possible to make a range of human gene products using recombinant DNA technology.

BIOTECHNOLOGY

is the use of living things or living systems to produce useful materials. Recently, this has focused on the manipulation of genetic material.

RAPID DETECTION OF DISEASE USING ELISA



tinyurl.com/y6tkum7n

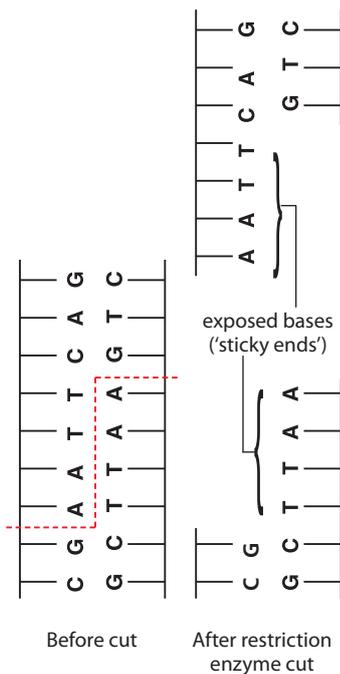


Fig. 6.1 Sticky ends of DNA resulting from a cut by EcoRI restriction enzyme.

It is possible to extract DNA from cells by breaking up the cells, separating the nuclei by centrifuging, treating these nuclei with special chemicals to remove the nuclear membrane and then isolating the DNA from the nuclear proteins. Biologists are able to pinpoint particular sections of the DNA for further study and/or manipulation. An important breakthrough in this DNA technology was the discovery in bacteria of special enzymes, called **restriction enzymes**, that cut up the very long DNA strands at set positions.

Restriction enzymes

There are many enzymes now known that cut DNA only at highly specific regions, so that if you use the same enzyme (or enzymes) on the samples from various sources, you know the fragments will always be cut at the same sequence of nucleotides. The restriction enzyme Eco R1, from *E. coli*, locates the sequence GAATTC and cuts between the G and the A. This sequence might appear every thousand bases or so. (See Fig. 6.1)

The sequence recognised by restriction enzymes is called the restriction site and it usually consists of four to six nucleotides. Some restriction enzymes cut straight across the two strands of the double helix leaving 'blunt' ends, while other restriction enzymes cut diagonally and leave some of the nucleotides from each strand unattached resulting in what have been called 'sticky ends'.

The restriction enzymes have evolved in bacteria to protect themselves against viruses and other mobile genes. Biologists have isolated large numbers of restriction enzymes from different strains of bacteria and there are more than 100 different restriction sites that can be specifically cut.

Identifying and isolating the right gene

Once the DNA has been extracted from cells and isolated from the nuclear proteins, restriction enzymes are used to cut the DNA into smaller fragments. The next task of the biologist is to identify and isolate the specific gene required. Two methods are used to achieve this; one uses a short piece of single-stranded DNA or RNA called a **probe**, and the other uses special proteins called **antibodies**. A DNA or RNA probe is used to detect the gene itself, while the antibody method is used to locate the gene product.

DNA and RNA probes

To use a probe, the biologist must know in advance a short sequence of nucleotides in the gene. If the protein for which the gene codes is known, the amino acid sequence of the protein can be used to deduce the nucleotide sequence of the gene. A short segment of single-stranded DNA or RNA with a sequence of bases that is complementary to part of the required gene is selected.

PCR CLONING

It is possible to clone a gene without using restriction enzymes by choosing appropriate primers and using PCR. This method is very efficient. One limitation is that it can be difficult to attach the replicated DNA fragments to suitable vectors, such as bacterial plasmids. Restriction enzymes that produce 'sticky ends' are still useful for this.



PCR AND CLONING



tinyurl.com/yy9bot3o

This segment is radioactively or fluorescently labelled and mixed with the double-stranded DNA fragments that contain the gene. The solution is then heated to separate the double strands. On cooling the solution, some of the probe molecules will bind to DNA fragments containing the specific complementary base sequence. The labelled probe identifies and locates the gene. (See Fig. 6.2) The gene can then be isolated and inserted into the cell of another organism.

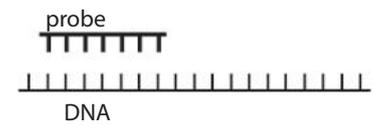


Fig. 6.2 DNA probe radioactively labelled.

Plasmids

One particularly useful application involves inserting human genes into bacterial cells. Many bacteria contain **plasmids**, which are small rings of DNA separate from the bacterial chromosome. (See Fig. 6.3) It is possible to isolate plasmids from bacterial cells and 'cut' them open with a specific restriction enzyme. Human DNA containing the desired gene is removed using the same restriction enzyme. The treated plasmids are then mixed with the human DNA which becomes incorporated into the plasmid. An enzyme called DNA ligase is then used to rejoin the plasmids into rings. These recombinant plasmids are then mixed with bacterial cells, which will take them up under appropriate conditions. The human gene has now been incorporated into the bacterial cells, and every time they reproduce, this gene will also be copied. Such a procedure is called **gene cloning**. The bacteria will then follow the instructions on the human gene to make human biochemicals, such as insulin and growth hormone. (See Fig. 6.4)

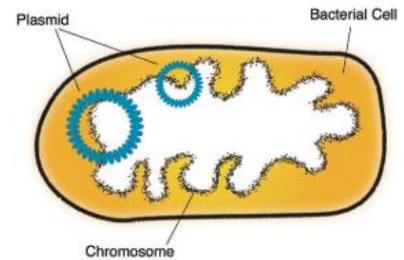


Fig. 6.3 Bacterial plasmids

Note that when transferring genes, such as the gene for human insulin, from eukaryotic cells into bacterial cells, the introns must first be removed. See textbox 'introns and bacterial cells'.

Genetic engineering can also be used in the production of vaccines, like hepatitis B vaccine. There are several ways of using this technology in vaccine production. One involves using recombinant

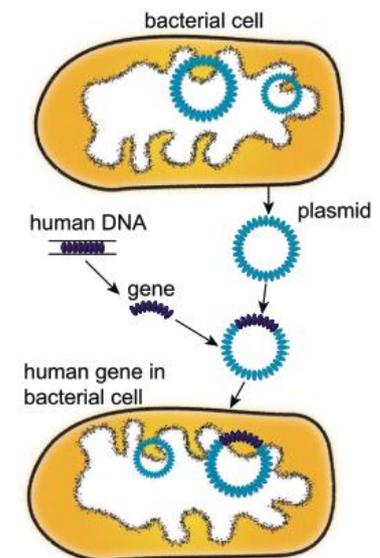


Fig 6.4 Gene cloning

INTRONS AND BACTERIAL CELLS

Eukaryotic cells contain introns (non-coding sequences) in their DNA. The messenger RNA produced by these cells has the introns removed before translation into protein. Bacterial cells usually have no introns and therefore they do not have a mechanism for removing them from mRNA. Before eukaryotic DNA is spliced into bacterial genomes, the introns must first be removed, so that the bacterial cells will be able to express the genes. This is achieved by obtaining 'mature' mRNA from eukaryotic cells, as it has already had the introns removed. The mRNA is copied into cDNA (complementary DNA) using a special enzyme called reverse transcriptase. DNA polymerase, an enzyme, is then used to convert the single-stranded cDNA into the familiar double-stranded DNA. This DNA (with no introns) is then inserted into bacterial plasmids for introduction into bacterial cells that can produce the desired protein.



tinyurl.com/4s3sx594

OTHER TECHNIQUES FOR GENE TRANSFER

ELECTROPORATION

A short electric pulse is applied to the plasma membrane of the target cell and this results in the formation of tiny holes. Particles containing the desired gene can be introduced to the cell through these holes.

LIPOSOME FUSION

Synthetic vesicles containing the selected gene are fused with the plasma membrane of the target cell resulting in the gene entering the cell's cytoplasm by endocytosis. Coating the vesicles with a special carbohydrate makes it more likely that they will fuse with the intended cells. This technique is being used in gene therapy.

GENE GUN

Microscopic particles of an inert material, such as gold or tungsten, are coated with the desired gene and 'fired' into the target cells, usually plant cells. The success rate of this technique is very low, as the gene needs to reach the nucleus and be incorporated into the genome of the cell.

DNA technology to produce a copy of the protein (antigen) from the coat of the pathogen. Another involves inserting extra DNA into the genome (see glossary) of the pathogen in order to render it harmless. In both cases the product will retain its ability to stimulate the immune system, without causing the disease.

The ability to genetically engineer crops to be higher yielding, or to be pest-resistant, is seen as a benefit to humankind as our population continues to grow. Ethical issues of the manipulation of DNA are discussed later in this chapter.



Describe how selected genes can be transferred between species.

A variety of techniques is available for transferring genes between species. These techniques, discussed below, include microinjection, and vectors (such as bacterial plasmids and viruses). There are additional techniques, such as electroporation, liposome fusion, protoplast fusion, and a 'gene gun'. Some of these are discussed in the text box on the left.

Ti plasmids

These are found in *Agrobacterium*, a bacterium that causes tumours in plants. The Ti plasmid is removed from the bacterium and the desired gene is added to it. The plasmid is returned to the bacterium, which is then used to infect the plant. The bacterium inserts the Ti plasmid (carrying the desired gene) into the plant cells. These cells divide rapidly to produce a tumour. The tumour is removed and cut into many pieces that are then treated by tissue culture techniques (See page 123) to produce many new plants whose cells contain the desired gene.

Viral vectors

Viruses are extremely small non-cellular particles made up of a nucleic acid core (DNA or RNA) and a protein coat. In addition, some viruses may have a lipid 'envelope' derived from the host cell membrane. Viruses are able to introduce their genetic material into a host cell, causing it to manufacture viral proteins, and produce new virus particles. Viruses can be modified so that they carry a desired gene, but no longer have the ability to harm the 'host' cell. When the altered viruses are used to 'infect' cells the desired gene is incorporated into the cells' DNA. (See Fig. 6.5)

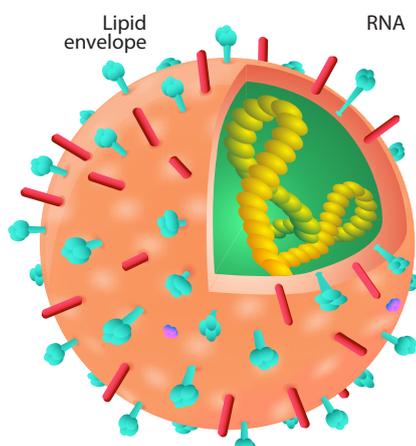


Fig 6.5 Influenza virus

VIRAL VECTORS AND PACKAGING CELLS



Using viruses as gene vectors is a technique that is being developed for gene therapy, and it involves some particularly interesting methods. First, the genes that the virus needs in order to be reproduced are removed from the viral DNA and inserted into the genome of special eukaryotic cells called **packaging cells**. The gene of interest is then spliced into the remaining viral DNA. This viral DNA, containing the gene to be used in gene therapy, is introduced into the packaging cells using a technique such as electroporation. The packaging cells then produce many copies of the complete virus particles by combining newly synthesised viral DNA and viral coat proteins. These complete virus particles then escape into the surrounding medium. Each one of the thousands of virus particles contains a copy of the gene of interest. When the newly-made virus particles are brought into contact with the target cells they will infect them, at the same time bringing the desired gene into the DNA of the target cells. Because the viruses have had their genes for reproduction removed, no more viruses will be made.

Micro-injection

In animals, the wanted gene can be inserted into a fertilised ovum by micro-injection. The ovum is held in place and a glass micropipette is passed through the plasma membrane and the nuclear membrane. The desired gene is injected via the micropipette into the nucleus. (See Fig. 6.6) The resulting animal will then contain all its own genes plus the 'foreign' gene. This technique does not have a 100 percent success rate, and is continuously being refined.

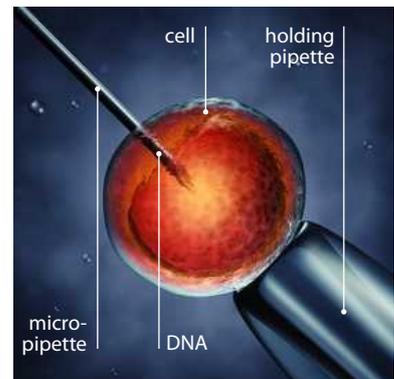


Fig. 6.6 Micro-injection

Benefits of transgenesis

It is now possible to determine how and where in the animal's body a transferred gene will be expressed. For example, transgenic goats have been developed that will produce milk containing human tissue plasminogen activator (TPA). This protein is extracted from the milk and is used by doctors to dissolve blood clots in coronary arteries and thus reduce the risk of heart attack.

In Australia, transgenic cotton plants are now widely used. These plants have had the Bt gene from a bacterium inserted into their nuclei. The Bt gene enables the cotton plants to produce a natural insecticide, thus leaving them less prone to insect attack, and reducing the need for spraying chemical insecticides.

There is a possibility that many genetic diseases may be able to be treated using gene therapy. By inserting a normal gene into cells that can then be implanted into the patient, a tissue that functions normally should be formed. Other versions of this procedure may involve using vectors such as viruses to 'infect' targeted cells in the

USE OF YEASTS IN BIOTECHNOLOGY

Saccharomyces yeasts are genetically engineered to convert plant based feed stocks into biofuels



Fig 6.7 Biotechnology

body with the desired gene. CRISPR technology can also be used to edit genes. This is explained in more detail later in this chapter.

Genetic engineering can also be used in the production of vaccines, such as hepatitis B vaccine. There are several ways of using this technology in vaccine production. One method involves using recombinant DNA technology to produce the protein (antigen) from the coat of the pathogen in order to stimulate an immune response. As the nucleic acid of the virus is not present, it cannot interfere with the cell's metabolism, and so it does not pose a danger when injected. Another method involves inserting a DNA sequence into the genome of the pathogen in order to render it harmless, while retaining its ability to stimulate the immune system. It too can be injected into a human without giving the person the disease.

The mass production of pharmaceuticals by bacteria as a result of genetic engineering is already a reality. Pure human insulin, a relatively small protein, is produced economically on a large scale by genetic engineering and this has improved the lifestyle of thousands of insulin-dependent diabetics. The previous treatment used insulin extracted from animals, and due to the slightly different amino acid sequences in this foreign DNA, there were side effects.

Ethical issues of genetic manipulation

● The ability of humans to manipulate the genetic material of organisms raises some important issues. Some of these relate to the possible effects on the environment, while others are concerned with effects on individuals and society. Because this technology is relatively new, many of its implications have yet to be fully understood, and new issues must be dealt with as they arise.

Concerns for the environment centre around fears that organisms with new combinations of DNA may develop unexpected characteristics. These individuals may threaten the survival of natural populations or species, and thus upset the ecological balance. Another argument that is raised is that the bacteria that are used in genetic engineering research may acquire characteristics that make it impossible to control them using current antibiotics. Opponents of this view claim that DNA has been altered naturally over billions of years of evolution, and that all we are doing is accelerating and directing a naturally occurring process. Even so, environmental problems caused by the development of pest-resistant crops, for example, could occur more rapidly than ever before, and be difficult to remedy.

Some food producers are using genetic engineering to produce stock and crops with desirable features, such as faster growth, improved quality, resistance to disease, and so on. Some people are worried that food produced in this way may somehow be harmful to its consumers. So far, there has been no evidence to support

this notion. In fact, it has been pointed out that plants and animals with particular desirable qualities have been selectively bred for centuries without any apparent ill-effects on consumers. It should also be remembered that the nucleic acids, proteins and other food chemicals that we ingest are broken down in the digestive system to their simpler subunits such as nucleotides and amino acids.

New DNA technology, including the use of microarrays, has greatly improved our ability to diagnose genetic diseases, but so far it has not helped with the treatment, nor has it provided a cure. It may be possible for you to find out if you will suffer from a genetic disease, but you may not be able to do anything to avoid it. This is currently the case for Huntington's disease. It is also possible to predict the likelihood of cancers, heart attacks or strokes.

Will insurance companies and employers make genetic tests a prerequisite for insurance or employment, thus discriminating against those people with a genetic disposition to particular diseases?

Another issue is whether societies should encourage modification of human genes other than those responsible for diseases. A genetically manipulated society with the elimination of undesirable traits could well be favoured by some. Who would decide what is undesirable?



Describe how CRISPR such as CRISPR-Cas9 can be used to edit and/or transfer genes.

CRISPR stands for **clustered regularly interspaced short palindromic repeats**. The technology that has been developed as a result of our understanding of CRISPR has revolutionised genetic engineering by making gene editing much faster, more affordable, and more accurate. This is how it works: some bacteria are able to protect themselves from invasion by viruses by storing part of the viral DNA in a part of the bacterial DNA called CRISPR. If the same virus attacks the bacterial cell some time later, the bacterial cell recognises this and makes a complementary RNA copy of the viral DNA segment that it had stored. This RNA is then 'loaded' into an enzyme called Cas9 (**CRISPR associated protein 9**), which is then able to cut DNA at a specific site corresponding to the viral DNA. Cas9 is an endonuclease protein that is able to cut DNA. Thus, any 'invading' viral DNA is quickly destroyed.

When biologists discovered the CRISPR-Cas9 system of cutting DNA in a highly specific way, they realised that the Cas9 protein is 'programmable'. That is, the site at which it will cut DNA is determined by the base sequence of the RNA, called **guide RNA** (gRNA), that is 'loaded' into it. That meant that the CRISPR-Cas9 system could be used to accurately cut DNA at any predetermined

FLAVR SAVR TOMATOES

In 1987, the FLAVR SAVR tomato was the first commercial crop to be genetically engineered. The GE process reduced the tomato's production of the PG enzyme which softens the fruit. With less of this enzyme the tomato ripened but remained firm for longer, and had more flavour than green tomatoes that were artificially ripened. Demand for the GE tomato remained strong from its release in 1994 until 1998, when a radiobroadcast in Britain announced that in an investigation, GE modified potatoes had caused "biological effects" on rats. Subsequently, the conclusion of the experiment shown to be incorrect, but too late - sales of FLAVR SAVR fell and it is no longer available.



CRISPR



tinyurl.com/j43kbpj



CRISPR EXPLAINED



tinyurl.com/4m6xxnz5



HOW CRISPR IS USED TO EDIT DNA



tinyurl.com/dp3bxuev

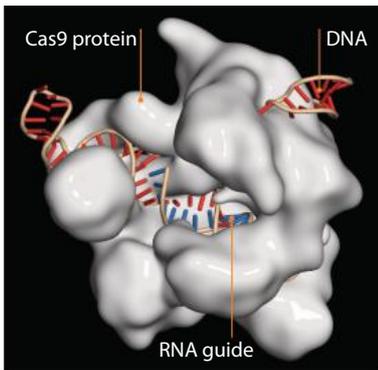


Fig 6.8 CRISPR-Cas9
 Cas9 protein - white,
 DNA - red nucleotides
 RNA guide - blue nucleotides

location. Gene editing has now become fast, simple, and inexpensive. The technique can also be used in live cells to edit genes, and to switch them on and off. (See Fig. 6.8 and Fig. 6.9) The CRISPR system can be used in any type of cell, including human cells. It is likely that genetic diseases may be treated using the CRISPR system to edit faulty genes. Even cancers may one day be cured by the appropriate use of this technology.

THE DISCOVERY OF CRISPR

This is an excellent example of how scientific discoveries often take many years to develop, and involve research in laboratories around the world. Sometimes, instead of collaboration being evident, there is 'competition', and there can be disputes that result in legal action to determine who made the discovery. The QR code and URL in the side bar provides an interesting account of this in the case of CRISPR-Cas9.

CRISPR



tinyurl.com/1t35o5v

CRISPR GENE EDITING



tinyurl.com/y8sr8q24

The first organisms to be modified using CRISPR-Cas9 technology were mushrooms in 2015. A small number of bases in the gene that codes for an enzyme that causes browning in the mushrooms was deleted, keeping mushrooms fresher longer.

Other techniques that have been used to edit genes include zinc-finger nuclease (ZFN) and transcription activator-like effector nuclease (TALEN).

SHERLOCK
 SHERLOCK is a new CRISPR system that uses Cas13a which recognises RNA instead of DNA. This may be used to diagnose certain diseases.

NONCODING DNA AND CRISPR



tinyurl.com/y5988u9w

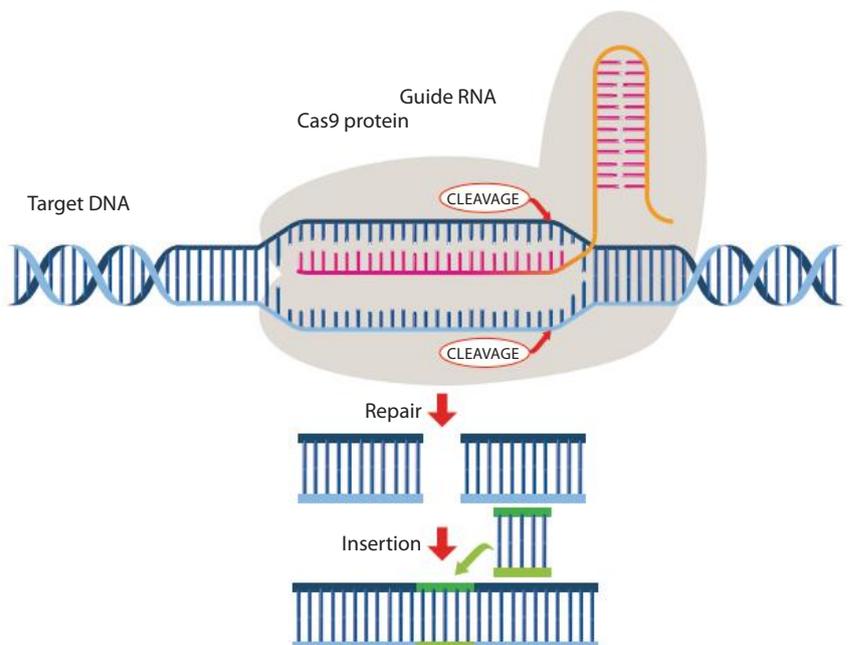


Fig 6.9 CRISPR-Cas9



Discuss the design of new proteins and their uses.

As we have seen, the function of protein molecules is determined by their shape, and their shape is determined by their amino acid sequence. A new branch of biology has developed to calculate the way in which a particular protein will fold. This involves computer analysis to determine the simplest amino acid sequence that will fold to produce a protein with the required shape. In this way, proteins with desired functions can be manufactured.

The steps involved are:

- design the required shape of the specific protein
- determine the amino acid sequence that will produce this shape
- use the genetic code to construct a DNA molecule with the base sequence that codes for the chain of amino acids
- incorporate the DNA into bacterial cells, probably with plasmids
- clone the bacteria, isolate and harvest the specific protein molecules.

Uses of designed proteins include:

- vaccines, that bind to viruses or bacteria, making them ineffective
- hollow protein 'spheres' that can deliver specific molecules (such as pharmaceuticals) to parts of the body
- channel proteins that regulate movement of specific substances across membranes enhancing their uptake
- proteins that change colour, or even glow, when they detect specific molecules

CRISPR AS A RESEARCH TOOL

One way researchers can find out the function of a gene is to "knock it out". That is, to edit it so it does not produce its normal mRNA and the corresponding protein., and then observe the effect on the cell or tissue. Scientists are currently using CRISPR technology to edit genes in embryos to understand the role of gene in the embryos' development.

There may even be non-biological uses in the future, such as thin-film solar cells and information storage on a molecular level.



PROTEIN DESIGN



tinyurl.com/y8x3wb9x



DESIGNER PROTEINS



tinyurl.com/yy4vwfrn



CURING GENETIC DISEASE



tinyurl.com/y3na7dnq

Study Questions

1. Describe how particular genes can be selected using probes, and removed using restriction enzymes.
2. Genetic engineering was made possible when DNA and then particular genes could be extracted from cells. A number of discoveries and techniques have contributed to the process. Describe the role of each of the following in genetic engineering:
 - (a) restriction enzymes
 - (b) DNA and RNA probes
 - (c) plasmids
 - (d) gene cloning.
3. Describe how selected genes can be transferred between species using bacterial plasmids, viruses, and microinjection. Use diagrams to illustrate your answers.
4. What are transgenic organisms and how are they produced?
5. Identify and explain three current uses of transgenic organisms.
6. The ability of humans to manipulate genetic material raises some important issues.
 - (a) Explain the term genetic manipulation.
 - (b) Discuss two concerns people may have for genetic manipulation of plants and animals used in agriculture.
 - (c) Discuss a concern people may have with the use of DNA technology to identify genetic diseases.
7. The CRISPR/Cas9 technology has revolutionised genetic engineering. State the biological origin of this system and describe how the organisms it was found in used it.
8. List the steps used to design specific proteins, such as Cas9 and their uses.
9. Describe how CRISPR can be used to edit and/or transfer genes.
10. Explain what is meant by the term gene therapy and state two possible uses for it.



TOPIC 2

Cells as the Basis of Life

- 7 Living Things are Made of Cells
- 8 Cell Structure and Function
- 9 Living Cells Need Energy
- 10 Movement In and Out of Cells
- 11 Cell Metabolism
- 12 New Cells from Old
- 13 Sexual Reproduction and Meiosis
- 14 Control of Cell Division

This topic is about cells — the basic units of all living things.

A cell is the simplest living structure that has independent existence and that can carry out all life processes. It was not until the first cells were formed that life on Earth commenced. Some organisms are made up of just one cell and are called unicellular organisms. Other organisms are made up of many cells, billions in some cases, and they are called multicellular organisms.

Generally, cells are microscopic and so we are not normally aware of their existence. The study of cells by biologists has helped to provide explanations of how life works and has evolved. By discovering what cells need in order to survive, we can better understand the requirements of organisms. By knowing what affects cells, we can determine the causes of some diseases and develop techniques to overcome them.

The study of cells is important to our understanding of all living things in the levels of life.

Science as a Human Endeavour

Throughout this topic examples that illustrate key concepts of science as a human endeavour are indicated by the symbol ▼. There are examples of communication and collaboration, development of scientific models and new technologies, influence on and by other areas of study and society, and applications and limitations of biological knowledge.

Living Things are Made of Cells

7

The cell theory unifies all living things

In 1665 Robert Hooke, using a simple microscope, first observed in cork tissue the small units that we now know as cells. (See Fig. 7.1) Since then it has become clear that just about every organism is made up of one or more cells. For example, the human body is made up of several billion cells, while an amoeba has only one cell.

In addition to being the structural unit of living things, cells are also the basic functional unit of life. This means that they not only make up the bodies of living things, but they also carry out all of the 'life processes'.

These ideas help to make up an important biological concept called the **cell theory**, which links all living things. It is a unifying concept. Further to the idea that all living things are made up of cells which are also functional units, the cell theory also states that every cell arises from a pre-existing cell and that cells contain hereditary material.

If we are going to understand how living things work then we need to know more about cells. Seventy or so years ago the understanding we had of cell structure was fairly limited due to the microscopic size of most cells. However, advances in techniques and the development of equipment, such as the electron microscope, have allowed us to explore the inner domain of the cell to an extent that Robert Hooke would not have even dreamt.

All cells are made up of an outer **membrane** enclosing a fluid called **cytoplasm**. This cytoplasm consists mainly of water and contains a variety of smaller structures that are collectively called **organelles** — small organs. The nature and complexity of these structures depends upon the type of cell involved.

As cells are units of life, it is useful to consider some of the features that distinguish living things from non-living things.

Living things:

- › are complex and have an organised structure
- › take in energy from their surroundings and use it
- › preserve a composition that is chemically different from that of their external environment
- › respond to stimuli
- › are able to reproduce themselves
- › grow and develop.

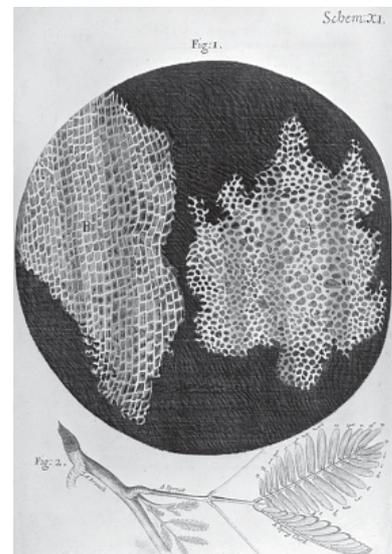


Fig. 7.1 Robert Hooke's intricate drawing made of a section of cork viewed through a microscope. He called the little box-like structures 'cells' because they reminded him of cells in a monastery.



Fig. 7.2 Light Microscope

VIRUSES

Viruses are small structures that can respond to a stimulus, cause disease and multiply, but they are not considered to be living organisms. They do not directly use energy, they cannot actively maintain their structure and they do not have all the necessary mechanisms to reproduce themselves outside of a host cell – something to consider next time you have a cold, caused by a rhinovirus.



Fig. 7.3 Diagram of a plant cell

While it is possible to think of some non-living things that meet one or two of these criteria, only living things will satisfy all of them.

The cell is the smallest unit of life that can exist independently, because it is the smallest entity that can fulfil all of the requirements of life listed above. Unicellular organisms have been doing this for billions of years, and in a laboratory human cells can exist independently for long periods in tissue culture solutions.

The cell membrane separates the cell cytoplasm from its surroundings and controls the exchange of materials, including nutrients and wastes, between the cell and its environment.

A cell can be defined as a unit of living matter (protoplasm) that is separated from its external surroundings by a membrane which regulates the passage of materials into and out of the cell. It is this membrane that defines where the cell starts and ends; it is the cell boundary. The cell membrane is sometimes called the plasma membrane. (See Fig. 7.3)

Functions of the cell membrane

The main functions of the cell membrane are to:

- › separate the contents of the cell from the external environment
- › regulate the passage of substances into and out of the cell
- › enable cells to recognise one another, and to recognise certain substances, such as hormones
- › enable attachment of the cytoskeleton.

Some of the proteins embedded in the cell membrane are enzymes.



Describe and represent the fluid mosaic model of the cell membrane

You can see that if the cell membrane allowed anything and everything to move freely through it, then the cell would very quickly lose its identity and be indistinguishable from its environment. It would seem that the cell membrane has a very important role to play in maintaining the integrity of the cell and allowing it to perform all the chemical reactions and other functions that keep it 'living'.

The structure of the cell membrane

The structure of the cell membrane has been difficult to determine as it is amazingly thin. A cell membrane is about 8 nanometres thick, whereas the thickness of the page you are now reading is about 100 000 nanometres. You would need about 12 000 layers of



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membrane to equal the thickness of this page. In 1935, long before the membrane could be examined with the electron microscope, Davson and Danielli suggested that the membrane was made up of two layers of lipid surrounded by two layers of protein. This could be thought of as a kind of 'lipid sandwich'.

The Davson-Danielli model of membrane structure was quite useful as it explained many aspects of membrane behaviour. However, with the aid of the electron microscope in the 1950s, closer and more direct observation of the membrane became possible. The latest model of membrane structure dates back to 1972, when Singer and Nicolson put forward the fluid mosaic model. (See Fig. 7.4) Whilst retaining the bilipid arrangement of the Davson-Danielli model, the **fluid mosaic model** has protein molecules penetrating through the lipid at various points. This arrangement has been somewhat poetically described as 'islands of protein floating in a sea of lipid'. The development of the fluid-mosaic model of membrane structure involved evidence from many sources and fields of study.

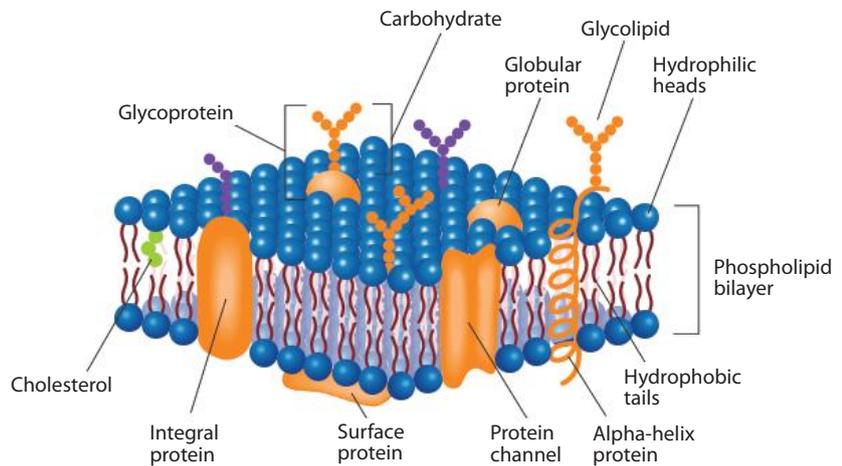


Fig. 7.4 Fluid mosaic model

LIPIDS AND PHOSPHOLIPIDS

Lipids, like carbohydrates, contain only the elements carbon, hydrogen, and oxygen, but the proportions are different. In a complex lipid molecule there are very few oxygen atoms. Lipids that are solids at room temperature are called fats and those that are liquid are called oils. A lipid molecule is made up of three fatty acids joined to a molecule of glycerol. (See Fig. 7.5) Like carbohydrates, lipids are important structural components of cells. **Phospholipids** contain two fatty acids attached to the glycerol, as well as a phosphate and an ethanolamine. (See Fig. 7.6) Huge numbers of phospholipid molecules act together as the major structural unit of all cell membranes.

Lipids also have functions of insulation and protection of certain body organs. Steroid hormones and waxes are lipid derivatives, meaning that they are produced from lipids. Cholesterol, a component of animal cell membranes, is a lipid-related compound.

CELL MEMBRANE STRUCTURE

- very thin (8nm, a page is 100 000 nm thick!!)
- fluid mosaic (latest model)
- bilipid layer of phospholipid
- protein molecules some have carbohydrates attached
- proteins act as transport channels, membrane receptors

CELL MEMBRANE 2



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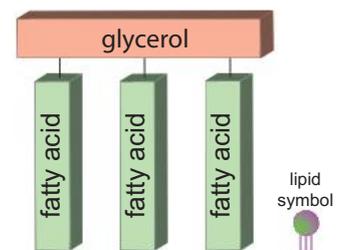


Fig. 7.5 Lipid molecule

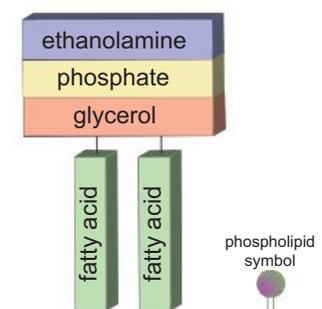


Fig. 7.6 Phospholipid molecule

As we shall see in Chapter 10, the fluid mosaic model allows an elegant explanation of the mechanisms by which substances move through the membrane, and hence, into and out of cells. The different carbohydrates on the proteins act as receptors and this explains how cells are able to recognise one another. As the term 'fluid' suggests, the membrane is not static, but is a dynamic living structure.

When we look at cells we find that they can be one of two basic forms.

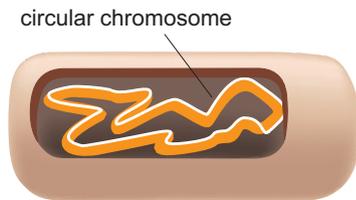


Fig. 7.7 Circular chromosome in a bacterial cell

The major types of cell are

- > prokaryotic
- > eukaryotic.



Compare prokaryotic and eukaryotic cells with respect to their:

- > size
- > internal organisation
- > shape and location of chromosomes

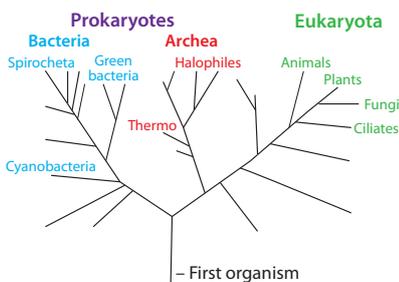


Fig. 7.8 Phylogenetic Tree of Life

Prokaryotic cells

Prokaryotic cells are very small and have their genetic material present as a circular chromosome of DNA that is not separated from the rest of the cell. (See Fig. 7.7) **Bacteria** and **archaea** (see textbox) are prokaryotic, and all other cells are eukaryotic. (See Fig. 7.8)

Eukaryotic cells – plants, animals, fungi, protists

The genetic material of **eukaryotic** cells consists of DNA associated with proteins (histones) to form linear chromosomes. These chromosomes are separated from the cytoplasm of the cell by a double-membrane nuclear envelope, giving rise to a structure called a **nucleus**. (See Fig. 7.9) Thus, a major difference between prokaryotic and eukaryotic cells is the presence of a nucleus in one but not the other.

There are other differences as well. Eukaryotic cells have a more complex internal organisation than prokaryotic cells. Eukaryotic cells contain membrane-bound organelles that prokaryotic cells lack, and are therefore 'compartmentalised'. The term **organelle** is used to describe discrete structural bodies within the cell such as the nucleus, mitochondrion and ribosome. Ribosomes do not have a membrane and the ribosomes of prokaryotic cells are smaller than those of eukaryotic cells.

In addition to their cell membrane, bacteria have an outer cell wall made of peptidoglycan. This compound is made up of

ARCHAEA

Archaea are prokaryotic unicellular organisms that are more similar to eukaryotes than they are to bacteria. Their cell walls and membranes are different in structure from those of bacteria.

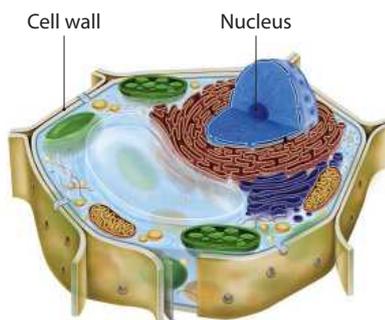


Fig. 7.9 A eukaryotic (plant) cell

polysaccharides and polypeptides combined. The cell walls of archaea are made up of different polysaccharides and polypeptides, with no peptidoglycan. Plant cells, which are eukaryotic, also have an outer **cell wall**, but this is made of a polysaccharide called cellulose. (See Fig. 7.9). Fungal cells, which are also eukaryotic, have a cell wall made mainly of complex carbohydrates called **glucan** and **chitin**, along with **glycoproteins**. (Refer to Page 73 for a comparison of plant, animal, and fungal cells.)

The table below highlights some of the differences between prokaryotic and eukaryotic cells.

Prokaryotic cells — bacteria and archaea	Eukaryotic cells — plants, animals, fungi, protists
smaller (1 - 10 µm diam)	larger (10 - 100 µm diam)
DNA circular	DNA linear
no nucleus	contains nucleus
little internal organisation	high level of internal organisation
no membrane-bound organelles	contain membrane-bound organelles
single chromosome	two or more chromosomes
bacteria have a cell wall made of peptidoglycan (polysaccharides and polypeptides)	cell wall (if present) made of cellulose (polysaccharide)

Prokaryotes only exist as single cells.

Prokaryotic organisms are either bacteria or archaea. All of these organisms are unicellular.

Eukaryotes

Eukaryotic organisms differ greatly from one another in size, shape, colour, and complexity. Although there is great diversity among eukaryotic organisms, they are made up of cells that have similar features, including membrane-bound organelles. Eukaryotic organisms range from those that are unicellular, such as *Amoeba* and *Paramecium*, to small mosses, giant gum trees, earthworms, humans, and whales.

Prokaryotic and eukaryotic cells have many features in common, which is a reflection of their common evolutionary past.

Features that prokaryotic and eukaryotic cells have in common include a phospholipid cell membrane, chromosomes made of DNA, similar protein synthesis mechanisms (including ribosomes), and the same genetic code.

FOSSIL EVIDENCE

Some of the oldest known fossils have been found in structures called stromatolites. These are made up of ancient bacterial mats in which sediment has become trapped and compressed to form rocks. Until recently, the most ancient stromatolites found were in Western Australia and they are estimated to be 3.5 billion years old. (See Fig. 7.10) In 2016, stromatolites estimated to be 3.7 billion years old were discovered in Greenland.



Fig. 7.10 Stromatolites in Western Australia

HOW MANY GENETIC CODES?

The standard genetic code is found almost universally. However the genetic code in mitochondria, chloroplasts, and some organisms, such as archaea, have minor differences. These differences are small in number, and are thought to have evolved from the standard code. Often they involve only a change in a start or stop codon.

Study Questions

- Explain the concept of cell theory.
 - Why is it a 'unifying concept'?
 - What impact has the development of microscopes had on our understanding of cells?
- List four functions of the cell membrane.
- Describe and represent the fluid mosaic model of the cell membrane using a diagram.
- Prokaryotic and eukaryotic cells are different with respect to size and structural organisation.
Complete the table below to illustrate the differences.

	Prokaryotic cells	Eukaryotic cells
Size		
Shape of Chromosome		
Presence of nucleus		
Organisation		
Presence of membrane-bound organelles		
Number of chromosomes		
Examples of organisms		

- Explain the evidence that suggests prokaryotic cells and eukaryotic cells have a common evolutionary past.

Cell Structure and Function

8

Eukaryotic cells have specialised organelles which facilitate biochemical processes.



Represent the structure and describe the function of:

- › **nucleus**
- › **nucleolus**
- › **mitochondrion**
- › **chloroplast**
- › **vacuole**
- › **Golgi body (including vesicles)**
- › **endoplasmic reticulum (rough and smooth)**
- › **ribosome**
- › **lysosome**
- › **cytoskeleton.**

Nucleus

This is usually the most prominent structure in a eukaryotic cell. If you use a microscope to look at a human cheek cell stained with methylene blue, the centrally located nucleus will be the most obvious structure. Examination with the electron microscope has shown that the boundary of the nucleus is composed of two membrane layers. This double membrane boundary, called the **nuclear envelope**, contains many **nuclear pores** lined with proteins. (See Fig. 8.1)

Experiments, including removal of the nucleus from cells, and cloning of animals, have provided evidence that the nucleus controls the activities of the cell. Put simply, the nucleus is able to determine which enzymes are made, and this in turn determines which chemical reactions can take place, and even what structure (shape etc.) the cell will have. When you look at a cell that is **not** in the process of dividing, the nucleus has a grainy appearance and its contents are referred to as **chromatin**. Chromatin is made up of DNA (deoxyribonucleic acid) and protein and it has no distinctive shape. (See Fig. 8.2) The DNA stores the cell's hereditary material and provides the information needed for the cell to function.

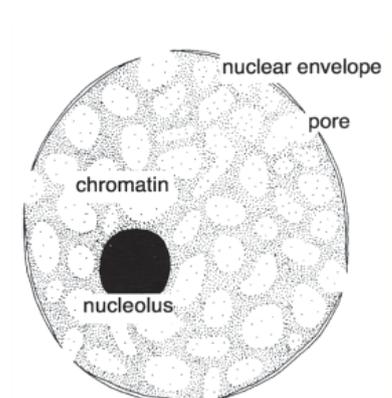


Fig. 8.1 Nucleus

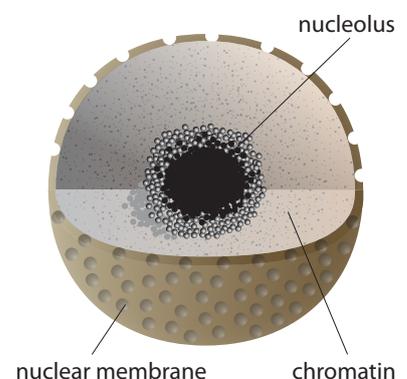


Fig. 8.2 Chromatin in the nucleus

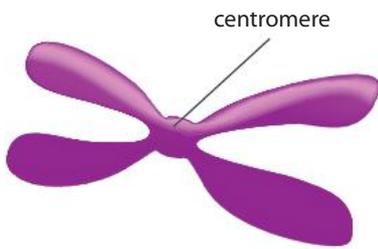


Fig. 8.3 A replicated, condensed chromosome

Before a cell can divide to produce new cells, the DNA must be copied exactly and then each daughter cell must receive a copy. In order to achieve this exact division of DNA after copying, the chromatin **condenses** to form short thick rod-like structures called **chromosomes**. (See Fig 8.3)

Nucleolus

Inside the nucleus there may be one or more circular regions that are visibly distinguishable from the rest. These are called **nucleoli**, (singular, **nucleolus**) and they are not bounded by a membrane. (See Fig. 8.1 and Fig. 8.2) They are composed of DNA and protein and are the site of ribosomal RNA (rRNA) synthesis.

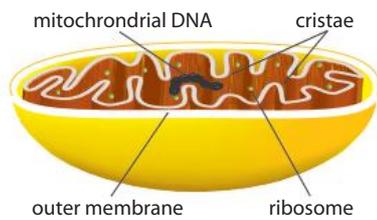


Fig. 8.4 Mitochondrion

Mitochondrion

Most eukaryotic cells contain many **mitochondria** (singular **mitochondrion**), which are sausage-shaped organelles up to about 10µm in length. Each mitochondrion has an outer membrane and an inner membrane which is folded to form structures called **cristae**. (See Fig. 8.4 and Fig. 8.5) The latter stages of aerobic respiration are carried out inside the mitochondria and a great deal of energy is released from this reaction. This is why the mitochondrion is often referred to as the 'powerhouse' of the cell, and it also explains why some cells, such as muscle cells, which require a lot of energy, have abundant mitochondria. Some cells have mitochondria congregated in particular regions where energy is needed. For example, in sperm cells, there are many mitochondria at the top of the tail. (See Fig. 8.6)



Fig. 8.5 Electron micrograph of mitochondria



Fig. 8.6 A sperm cell containing mitochondria

Chloroplast

Chloroplasts are organelles that are a type of **plastid** and they are restricted to plant cells. The other plastids which you may have heard of are leucoplasts which store starch, and chromoplasts which contain pigments. Chloroplasts have two outer membranes, while inside there is a system of membranous flattened sacs called **thylakoids**. In some places in the chloroplast these thylakoids are arranged in stacks called **grana** (singular **granum**). The fluid which surrounds the membranous sacs is called the **stroma**. (See Fig. 8.7) The thylakoid membranes contain **photosynthetic pigments** such as **chlorophyll**, and their function is to carry out photosynthesis, a series of chemical reactions which use light energy to convert carbon dioxide and water into glucose, a simple sugar. The first stage of photosynthesis requires light energy and occurs in the grana. Enzymes necessary for the final stage of photosynthesis in which sugar is manufactured are located in the stroma.

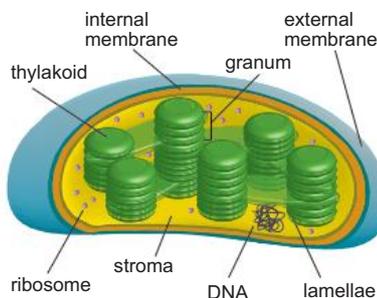


Fig. 8.7 Chloroplast

An interesting idea that is discussed in Chapter 20 relates to the possible origin of mitochondria and chloroplasts in eukaryotic cells. It has been suggested that aerobic bacteria were incorporated into larger (eukaryotic) cells and these aerobic 'invaders' became mitochondria. Also, photosynthetic bacteria may have been engulfed by larger cells to become chloroplasts. A major piece of evidence for this theory is that mitochondria and chloroplasts contain a **circular chromosome**, like prokaryotes, and that they divide independently of the cell.

Vacuole/vesicle

A vacuole is a fluid-filled space bounded by a membrane. Most mature plant cells contain a large central vacuole. In the vacuole there is a watery solution, with solutes such as salts (in the form of ions), simple sugars, and amino acids. One of the most important functions of these vacuoles is the maintenance of water and salt balance for the cell. By taking in water and enlarging, the plant cell vacuole also contributes to the growth of the cell and helps to maintain its shape. Vacuoles may also be used to store the cell's waste products, and some vacuoles contain pigments. (See Fig. 8.9)

Unlike plant cells, other eukaryotic cells do not contain large central vacuoles. Vacuoles that are found in animal cells are usually much smaller and more numerous than those of plant cells. Two examples of vacuoles that may be found in other cells are food vacuoles and contractile vacuoles. Food vacuoles are formed when a cell engulfs a particle by **phagocytosis**. The details of this process are discussed in Chapter 10.

A small vacuole is called a vesicle. These may be involved in **pinocytosis** (See page 91 for details), and with Golgi bodies in **exocytosis**. There are also specialised vesicles, such as lysosomes.

Golgi body (including vesicles)

The Golgi body (also known as the Golgi apparatus) is a stack of flattened sacs made of smooth membrane. (See Fig. 8.10) They are involved in the **packaging, modification** and **secretion** of proteins, lipids and carbohydrates manufactured by the cell. It is not surprising, therefore, to find abundant Golgi bodies in cells which are involved in secretion. Examples include gland cells such as cells of the salivary glands, which secrete saliva. The Golgi body also manufactures some carbohydrates for use outside the cell. These are packaged into vesicles that bud off from the Golgi body and migrate to the cell membrane where they fuse with it and release their contents. This secretion is an example of **exocytosis**. (See Fig. 8.11) (exo = out of, cytosis = the cell)

CONTRACTILE VACUULES

Contractile vacuoles are specialised structures found in some unicellular fresh water organisms, such as Paramecium, and they are involved in removing excess water that diffuses into the cell as a result of osmosis. (see Fig. 8.8)

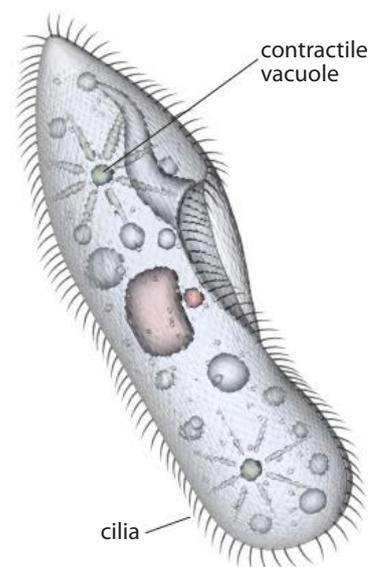


Fig. 8.8 Contractile vacuoles in a *Paramecium*

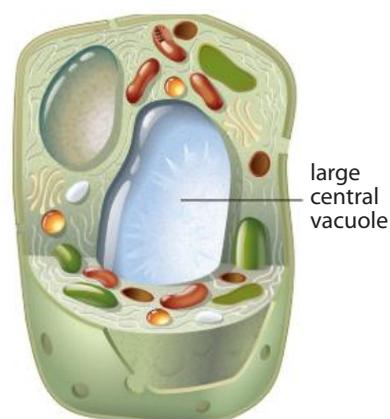


Fig. 8.9 Central vacuole in plant cell



Fig. 8.10 Golgi body with vesicles

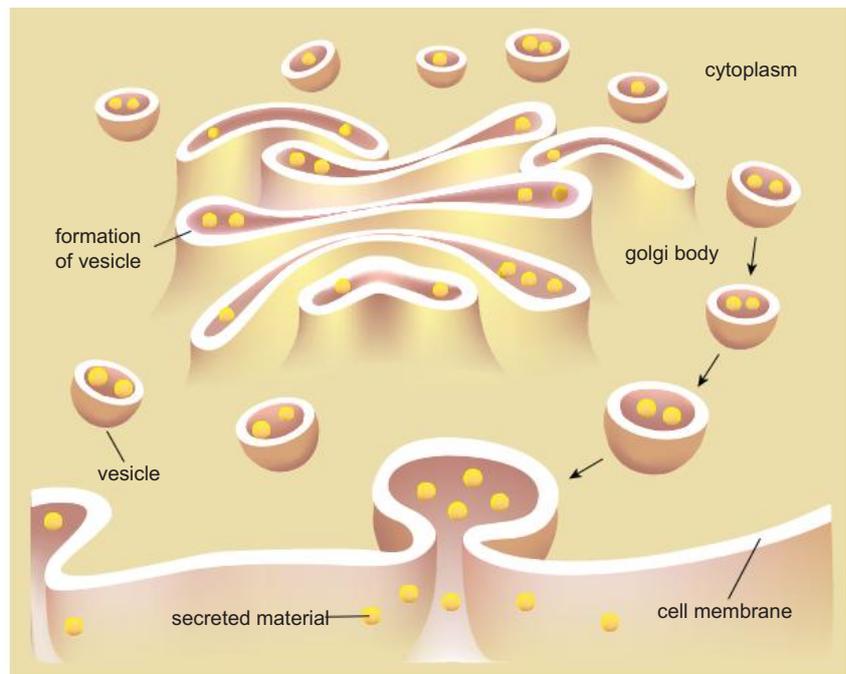


Fig. 8.11 Exocytosis

Endoplasmic reticulum

The endoplasmic reticulum (ER) is a system of membranes that extends throughout the cytoplasm from the nuclear envelope to the cell membrane. It forms an intricate network of passageways throughout the cell and is important in the transport of materials from one part of the cell to another. Some ER has ribosomes attached to it and this is referred to as **rough endoplasmic reticulum**, due to its appearance. Rough ER is the site of protein and membrane synthesis. Proteins manufactured by rough ER may have carbohydrate attached to them and are then called glycoproteins. These can be used as cell membrane receptors.

ER without ribosomes attached to it is called **smooth endoplasmic reticulum**, and it tends to be involved in metabolic processes such as lipid synthesis and carbohydrate metabolism. (See Fig. 8.12a) Smooth ER can be thought of as a kind of 'workbench' for many metabolic processes, since certain enzymes are embedded in it.

Ribosome

A ribosome is made of RNA and protein. The RNA in ribosomes is a special type, produced in a special region of the nucleus called the nucleolus, and it is called ribosomal RNA (rRNA). Ribosomes are not enclosed by a membrane. They can therefore be described as non membrane-bound organelles. Some definitions even go as far as to say that they are not organelles.

Ribosomes are the site of translation in protein synthesis. (See Fig. 8.12b)

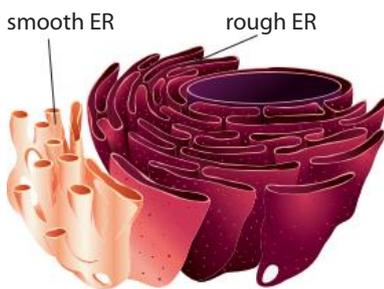


Fig. 8.12a Endoplasmic reticulum

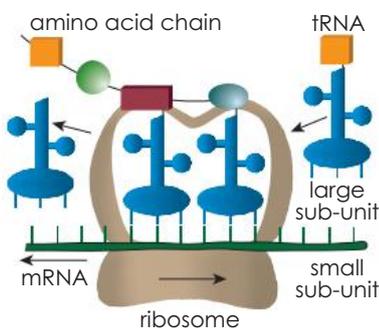


Fig. 8.12b Translation at the ribosome

Lysosome

A lysosome is a vesicle containing digestive enzymes. In some cells lysosomes fuse with 'food vacuoles' and release digestive enzymes, which break down the food. (See Fig.8.13) Examples of cells that are phagocytic and use lysosomes to digest their food include unicellular Amoebae. Certain cells of our immune system that are involved in engulfing and destroying foreign particles like bacteria also use lysosomes. Cells that carry out phagocytosis are selective, and will not engulf just any particle. For example, Amoeba cells will only engulf particles of nutritional value. Lysosomes are also involved in programmed cell death (**apoptosis**).

Phagocytosis is described in detail in Chapter 10.

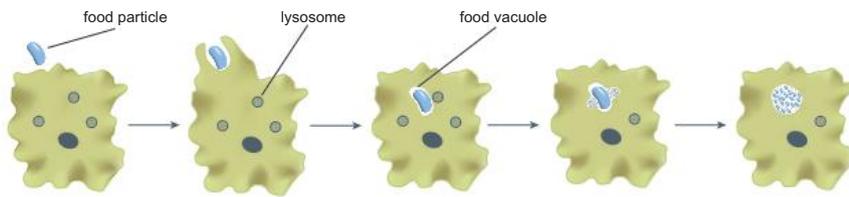


Fig. 8.13 Phagocytosis



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Cytoskeleton

All eukaryotic cells have a **cytoskeleton** which is made up of three main components: **microfilaments**, **intermediate filaments** and **microtubules**. (See Fig. 8.14) The components of the cytoskeleton are made up of protein molecules such as actin and tubulin which are globular proteins that can be assembled to form long filaments and hollow tubules. The filaments and microtubules of the cytoskeleton are made up of subunits which can be rapidly removed or inserted into the existing structure to change its shape — much like the temporary scaffolding at a building site. The cytoskeleton is therefore a dynamic structure.

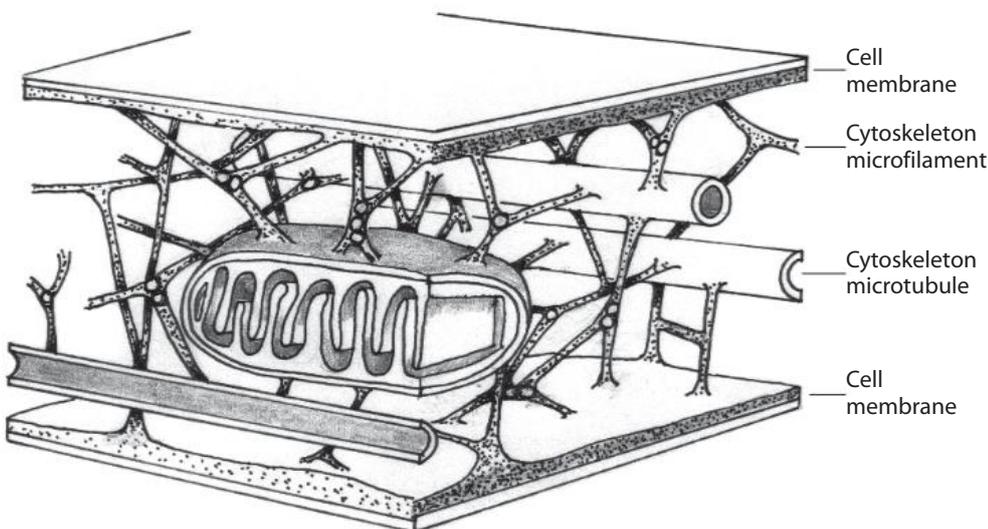


Fig. 8.14 The cytoskeleton.

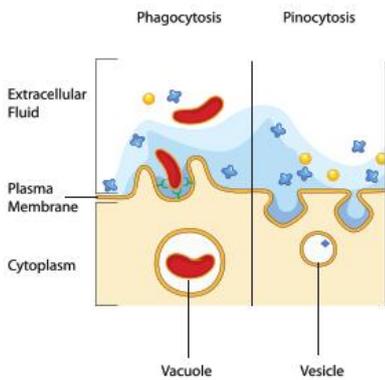


Fig. 8.15 Endocytosis

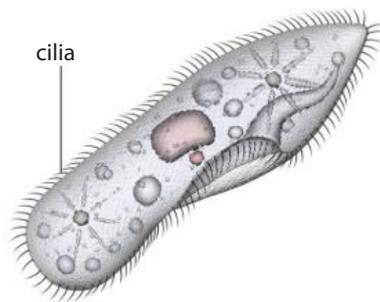


Fig. 8.16 *Paramecium* with cilia



Fig. 8.17 A human red blood cell

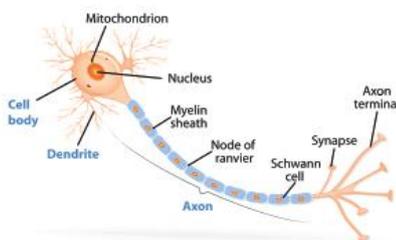


Fig. 8.18 Nerve cell

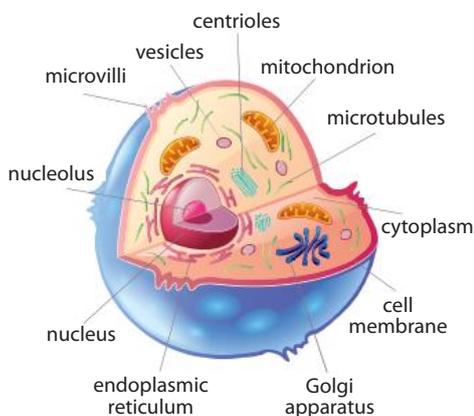


Fig. 8.19 Cell with microvilli

The main functions of the cytoskeleton are:

- > to give cells their shape
- > to be involved in cell movement
- > to hold organelles in place
- > to strengthen cells.

Microfilaments, made of the globular protein actin, are involved in intracellular movement like cytoplasmic streaming, chloroplast orientation, the pinching in of the cell membrane to form daughter cells after cell division, and the formation of a food vacuole by the process of phagocytosis. (See Fig. 8.15) The contraction of muscle is due to one group of microfilaments interacting with another. Actin proteins are also found in some prokaryotes.

Microtubules, made of the protein tubulin, are an essential part of the flagella or cilia, fine hair-like projections found on many cell membranes. The coordinated beating of these structures causes fluid movement, so that either the cell moves or fluid moves past the cell. A unicellular organism like a *Paramecium* uses cilia to move around its environment (See Fig. 8.16), while epithelial cells in the airways of our lungs use cilia to move mucus out of the lungs.

Intermediate filaments are made up of strong fibrous proteins and they are found in cells, such as skin cells, that are subject to wear and tear. The function of the intermediate filaments is to strengthen these cells and their tissues.

Cytoskeleton examples

A human red blood cell is formed into a bi-concave shape, rather than a sphere, by its cytoskeleton, and this gives it a high surface area to volume ratio, suitable for transferring oxygen by diffusion. (See Fig. 8.17) A nerve cell, with its very long axon, is another example of a cell shape that would be impossible without a cytoskeleton. (See Fig. 8.18)

The cytoskeleton also enables some cells to have highly specialised surfaces. Bundles of actin filaments support microvilli (See Fig. 8.19) on cells such as intestinal epithelium, kidney tubule epithelium, and the hair cells of the inner ear.

The organelles in the cell are not only moved by the cytoskeleton, but they are held in place by it. The chloroplasts in plant cells are held near the surface of the cell with the correct orientation to receive maximum light. When a killer T-cell of our immune system moves alongside an infected cell to destroy it, the Golgi apparatus is held in place over the infected cell by the cytoskeleton.

Movement within a cell is made possible by the cytoskeleton. During cell division, a specialised microtubule structure called the **spindle apparatus** is formed to facilitate the movement of

chromosomes. The chromosomes attach to the spindle at their centromeres. The removal of protein subunits from one end of the microtubules, thus shortening them, causes the attached chromosomes to move. Also, proteins called kinesins act like “motors” and move microtubules past one another. Cell division is discussed in more detail in Chapter 12.



Compare the structures of plant, animal, and fungal cells.

Many of the structures found in plant and animal cells are the same. For example, they both have a cell membrane, cytoplasm, and ribosomes – as do all cells. They also contain a nucleus and membrane-bound organelles – hence they are eukaryotic.

Plant cells can be distinguished from animal cells because they have a cellulose cell wall surrounding the cell membrane, and many have a large, central vacuole, and chloroplasts. (See Fig. 8.20 and Fig. 8.21) Note that not all plant cells contain chloroplasts – only those that carry out photosynthesis.

The components of fungal cells are similar to those of animal cells, except that fungal cells also contain a cell wall. Note that fungal cell walls are different in structure from bacterial cell walls and plant cell walls. (See Page 65.)

The table below summarises the main structural differences between plant and animal cells.

A comparison of plant, animal and fungal cell structure

Structure	Plant cell	Animal cell	Fungal cell
cell wall	present	absent	present
cell membrane	present	present	present
nucleus	present	present	present
nucleolus	present	present	present
mitochondria	present	present	present
chloroplasts	may be present	absent	absent
vacuoles	large, central	small	present
Golgi body	present	present	present
endoplasmic reticulum	present	present	present
ribosomes	present	present	present
lysosomes	present	present	present
cytoskeleton	present	present	present

Kinesins are used to move organelles within the cell.



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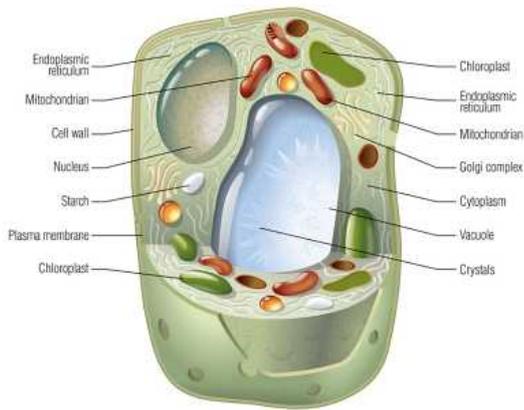


Fig. 8.20 Plant cell

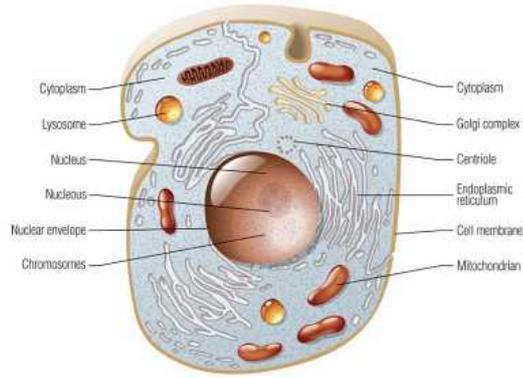


Fig. 8.21 Animal cell

Study Questions

- Eukaryotic cells have specialised organelles.
 - What is the essential role of the nucleus?
 - Describe the structure of the nuclear membrane.
 - Explain the meaning of the terms chromatin, chromosome and nucleolus.
- Describe the structure and function of a mitochondrion.
 - What cells in a human would you expect to have (i) numerous mitochondria - explain your answer. (ii) few mitochondria - explain your answer.
- What is the function of a chloroplast?
 - The following structures are found in chloroplasts - thylakoid, stroma and photosynthetic pigment. State their composition and function.
- Name four functions of plant cell vacuoles.
 - How does the vacuole contribute to cellular growth in plants?
 - What is the function of a contractile vacuole?
- List the structure and function of the Golgi body. Give examples of the cells in a human where it would be most active.
- Why are the names of rough endoplasmic reticulum and smooth endoplasmic reticulum appropriate?
- State the three main components of the cytoskeleton.
 - Name the main type of chemical compound found in each of the main components.
 - Describe a specific example of the role the cytoskeleton plays in providing a cell's shape and the type of cell movement.
 - Why is it necessary for some organelles to be able to be moved around in a cell?
- Complete the table comparing plant and animal cells.

Structure	Plant cell	Animal cell
Cell wall		
Nucleus		
Mitochondria		
Chloroplasts		
vacuoles		
cytoskeleton		
Examples of organisms		

Living Cells Need Energy

9

As we have seen, there is a large number of activities in which the cell is involved, and many of them require considerable energy. Energy is an abstract concept and as such it is difficult to define. Instead of defining what energy is we define it in terms of what it can do. Energy is therefore defined as the *capacity to do work*. The more energy an animal or plant has, the more work it can do.

All living cells need energy to carry out essential life processes such as active transport, movement of organelles or other structures, phagocytosis, pinocytosis, exocytosis, and synthesis of macromolecules. By performing these tasks cells are able to grow, repair, reproduce, and function normally.

Movement may involve the whole cell or movement within the cell. An amoeba cell needs to move about in order to locate food and a favourable environment. (See Fig. 9.1) Intracellular movement may involve structures such as chromosomes separating during cell division, or vesicles migrating to the cell membrane during secretion. There are many other examples of movement involving whole cells or parts of cells. All of them require energy from within the cell.

Cells need to synthesise a large number of compounds such as proteins, carbohydrates, lipids and nucleic acids. All synthesis reactions require energy to assemble large molecules from smaller components.

For cells to continue to function they must have a stable internal environment. Factors such as water and solute balance, pH, temperature, and oxygen and waste concentration, need to be kept within the appropriate range. In order to achieve this a range of processes like active transport need to be carried out and these require energy.

Cells require inputs of suitable forms of energy, including light energy, or chemical energy in complex molecules.

The energy that is so essential for cells to carry out their life processes can be obtained in either physical or chemical form. Some cells can use sunlight, a physical form of energy, while others must take in energy-rich compounds, a chemical form of energy. In both



Fig. 9.1 Amoeba

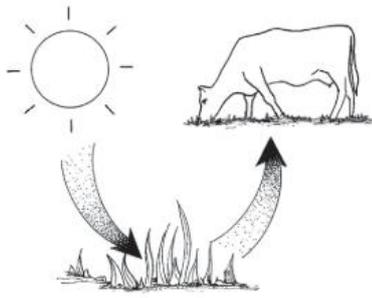


Fig. 9.2 Energy flow

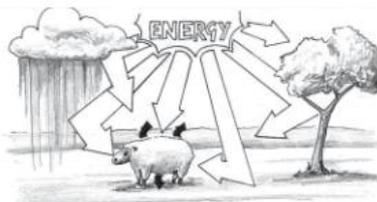


Fig. 9.3 Solar energy

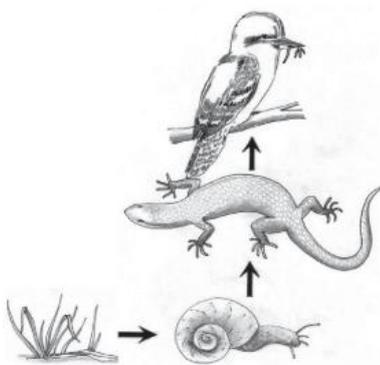


Fig. 9.4 Food chain - autotrophs to heterotrophs

cases the energy that enters the cell is then transformed so that it is useful to the cell. Examples of energy transformation that occur in cells include changes from light energy to chemical energy and from chemical energy to heat, movement, and light. Plant cells that have chloroplasts convert light energy to chemical energy and muscle cells convert chemical energy into movement. All cells produce heat as part of their chemical activities and glow worms have special cells which change chemical energy into light energy. (See Fig. 9.2)



Distinguish between autotrophs and heterotrophs.

Some organisms are able to make all the energy-rich compounds they need from simple inorganic substances, and to do this they need energy. Such organisms are called **autotrophs** and most of them use sunlight, a physical form of energy, for these synthesis reactions. The Sun is the main source of energy for life on Earth. (See Fig. 9.3) The process in which light energy is used to synthesise complex organic compounds from simple inorganic substances is called **photosynthesis**, from the Greek *photos*, meaning light. Consequently, organisms that do this are called **photosynthetic autotrophs**. Green plants make up the most common group of photosynthetic autotrophs. In addition to photosynthesis, chemosynthetic reactions are carried out by some rare single-celled organisms to make food molecules from simple raw materials.

Some organisms cannot produce all of their organic compounds from simple inorganic substances. They rely on other organisms, or their products or remains, for "food". These organisms are called **heterotrophs** or 'consumers'. (See Fig. 9.4)

The sun is the main source of energy for life.

Photosynthesis

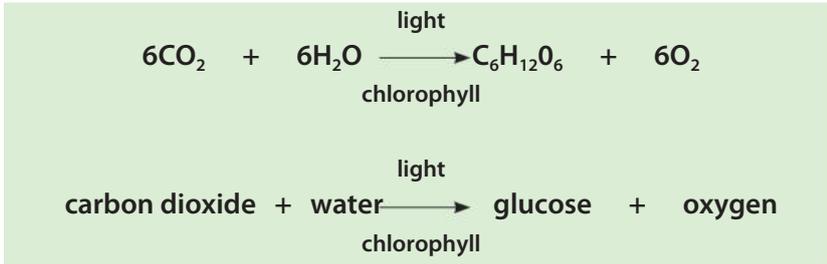
Photosynthesis is the most fundamental chemical process to life on Earth, in which solar energy is converted to chemical energy in organic molecules that can be used by cells. Photosynthetic organisms not only 'feed' themselves but are ultimately the food source of nearly all other organisms. Not only does the photosynthetic reaction provide most organisms with food, but it also takes carbon dioxide out of the atmosphere and replaces it with oxygen.

The process of photosynthesis is far from simple. For our purposes it is useful to represent photosynthesis using a summary equation, but you should remember that photosynthesis actually involves many smaller steps, each catalysed by a specific enzyme.

solar energy =
energy from the sun



Recognise that photosynthesis is important in the conversion of light energy into chemical energy, as illustrated by the following equation:



In photosynthetic eukaryotic cells (plant cells) photosynthesis occurs in specialised cell organelles called chloroplasts. (See Fig.9.5) Chlorophyll, the green pigment that absorbs light, is located in the grana. Enzymes that catalyse the many steps of photosynthesis are found in the grana and stroma. (See Chapter 8.) In photosynthetic prokaryotic cells (cyanobacteria) photosynthesis occurs in the cytoplasm which contains the chlorophyll and necessary enzymes.

Energy transformations occur within all living cells.



Recognise that energy is required to break chemical bonds and energy is released when new bonds are formed.

Transformation of energy

Chemical bonds hold atoms together and energy is needed to break these chemical bonds. Conversely, the formation of chemical bonds releases energy. When a chemical reaction occurs some chemical bonds are broken and new ones are made as new combinations of atoms are formed. If the total energy released when new bonds are made is greater than the energy required to break the original bonds then there will be a net output of energy.

The breakdown of glucose in the presence of oxygen releases energy, as the energy in the reactants is greater than the energy in the products. This is because the amount of energy required to break the bonds in the glucose and oxygen molecules (reactants) is less than the amount of energy released when the bonds in carbon dioxide and water (products) are formed. (See Fig.9.6) Some of this energy that is released can be used by the cell to do work, while the rest is lost as heat.

WATER USED, WATER PRODUCED!

The actual events of photosynthesis are more complicated than the summary equation shows. There are actually 12 water molecules used to make one molecule of glucose, but six new water molecules are produced. The summary equation for photosynthesis shows the net water use. It is interesting to note that all of the oxygen gas produced in photosynthesis comes from the water molecules, a fact demonstrated using water labelled with radioactive oxygen.

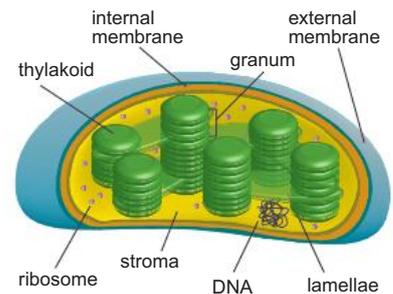


Fig. 9.5 Chloroplast

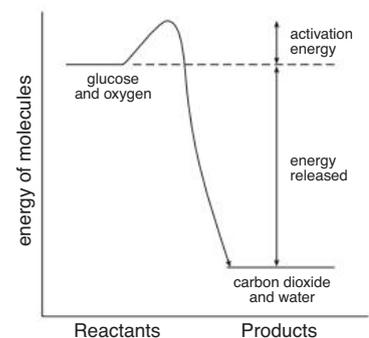
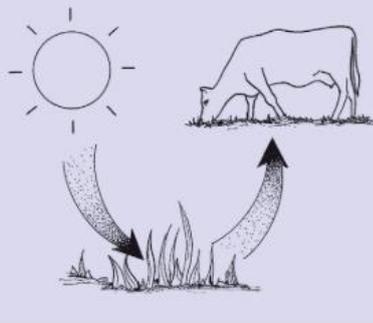


Fig. 9.6 Transformation of Energy

ENERGY FLOW

Those organisms that cannot make all of their own energy-rich compounds must obtain these from other organisms. Autotrophs make more sugar than they use themselves and the excess is stored. This stored carbohydrate is the major source of food for all other organisms. The food, or energy, is passed from one organism to another along a food chain.



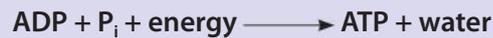
BLOCKING THE ATP CYCLE

Many poisons act by blocking the respiratory process and thus preventing the formation of the energy molecule ATP. The fact that these poisons, like cyanide, can kill in a matter of seconds indicates how vital it is that our cells carry out respiration to ensure a continuous supply of energy in the form of ATP.



Describe the formation of ATP from ADP and P_i.

One of the most important energy storage compounds in cells is called ATP. This stands for adenosine triphosphate. ATP consists of the nitrogen base adenine bonded to the sugar ribose, which in turn is bonded to three phosphate groups, called a triphosphate. The third phosphate group is held to the others by an unstable bond. The energy needed to produce ATP from ADP and P_i comes from cellular respiration.



Describe the conversion of ATP to ADP and P_i which releases energy for some metabolic reactions.

The energy that is released as a result of the conversion of ATP to ADP and P_i can be used by the cell to drive a multitude of processes that require energy, such as synthesis, cell movement, active transport, endocytosis and exocytosis. When ATP is hydrolysed, to form adenosine diphosphate (ADP) and an inorganic phosphate group which is symbolised P_i, energy is released.



Cells use ATP continuously, but they also produce it continuously. This concurrent production and breakdown of ATP is known as the ATP cycle. (See Fig. 9.7)

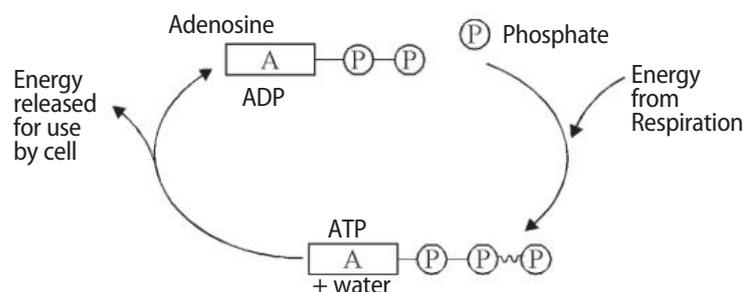


Fig. 9.7 The ATP cycle

The amount of ATP recycled by cells in this way is phenomenal. For a human this would be equal to almost the entire body weight in one day!

Cellular Respiration

All living cells need to respire in order to obtain the energy to carry out essential life processes such as synthesis, active transport, movement, growth and reproduction. Just as cells need energy, all organisms need energy in order to carry out the processes necessary to sustain life. Cellular respiration provides this energy by breaking down energy-rich organic molecules into lower energy products.

We have already seen that organisms that cannot make all of their own complex organic compounds are called **heterotrophs**. They rely on other organisms for at least some of their organic compound requirements. Ultimately all heterotrophs rely on autotrophs for the supply of organic compounds. For example, the snail obtains its organic compounds from grass and the lizard obtains some of its organic compounds from snails. The kookaburra, in turn, obtains its some of its organic compounds from lizards. In this case, the kookaburra's organic compounds have ultimately come from the grass. (See Fig. 9.8) It is important to note here that the essential difference between autotrophs and heterotrophs is the source of the organic compounds. Autotrophs make their own organic compounds, whereas heterotrophs must obtain at least some of their organic compounds by feeding.

Some heterotrophs need only to receive certain organic molecules to supply them with energy, and they make the rest – proteins, lipids, nucleic acids – themselves. We know that humans, however, have many nutritional requirements that must be supplied in order for them to continue to function normally. These include a range of organic compounds such as simple sugars, amino acids, glycerol and fatty acids, and vitamins, as well as inorganic substances such as water and mineral ions. These nutritional substances enable cells to obtain energy, to grow, and to regulate cell processes.

Types of Respiration

There are two types of chemical respiration that cells use to provide themselves with energy – **aerobic respiration** and **fermentation**. Both reactions use glucose as their starting point, but the products in each case are different.

The most efficient of the two processes is aerobic respiration. Like many other chemical processes in the cell, aerobic respiration consists of a series of small steps – a metabolic pathway. Each step in the pathway is catalysed by a specific enzyme. (See Fig. 9.9)

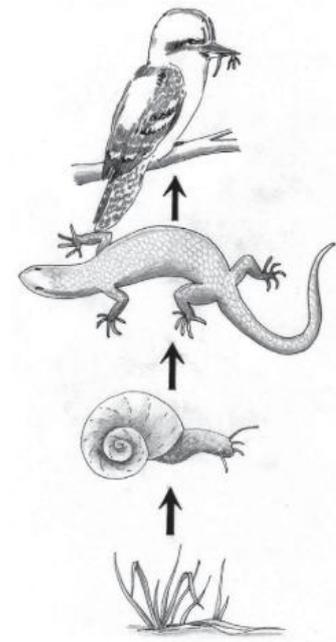


Fig. 9.8 Food chain

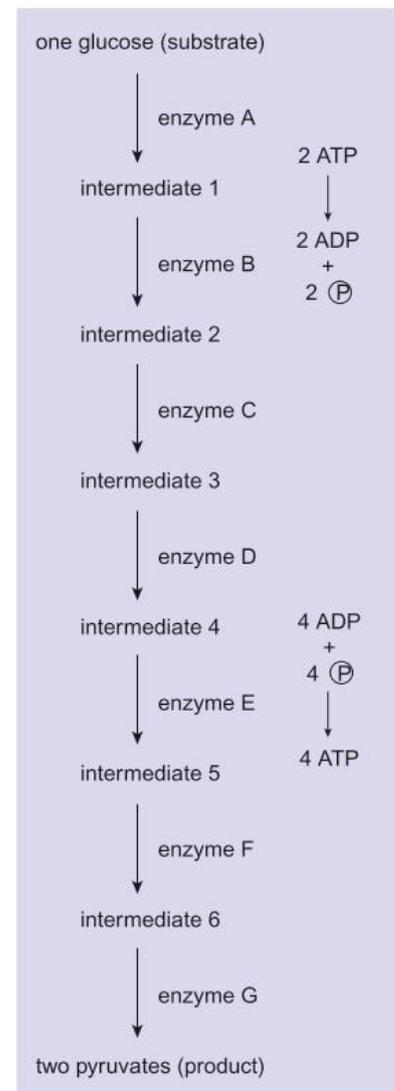


Fig. 9.9 Glycolysis – a metabolic pathway that begins respiration



Explain how most autotrophs and heterotrophs transform chemical energy for use through aerobic respiration, as illustrated by the following equation:



Energy is liberated as a result of the breakdown of energy-rich glucose, in the presence of oxygen, to carbon dioxide and water. The energy that is not lost as heat is used by the cell to make ATP, a short term energy storage compound. You might notice that the equation for aerobic respiration seems to be photosynthesis in reverse. While it is true that the reactants and products are reversed, the detailed steps for each process are completely different.

Aerobic respiration begins with the conversion of glucose to an intermediate compound called **pyruvic acid**. This metabolic pathway is called glycolysis and it occurs in the cytoplasm. (See Fig. 9.10) There are two molecules of pyruvic acid produced for each molecule of glucose broken down (pyruvic acid is a three-carbon compound) and this provides a net gain of two ATPs. Actually, four ATPs are produced in glycolysis, but two are needed to start the process off. This is how we arrive at the net gain of two.

It is remarkable that all known organisms carry out glycolysis in exactly the same way using the same enzymes to catalyse the same chemical steps. This universality is strong evidence for the concept that all living things evolved from a common ancestor.

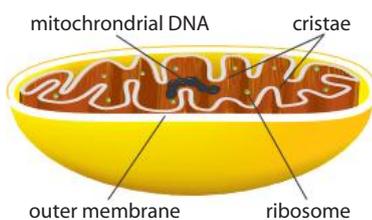
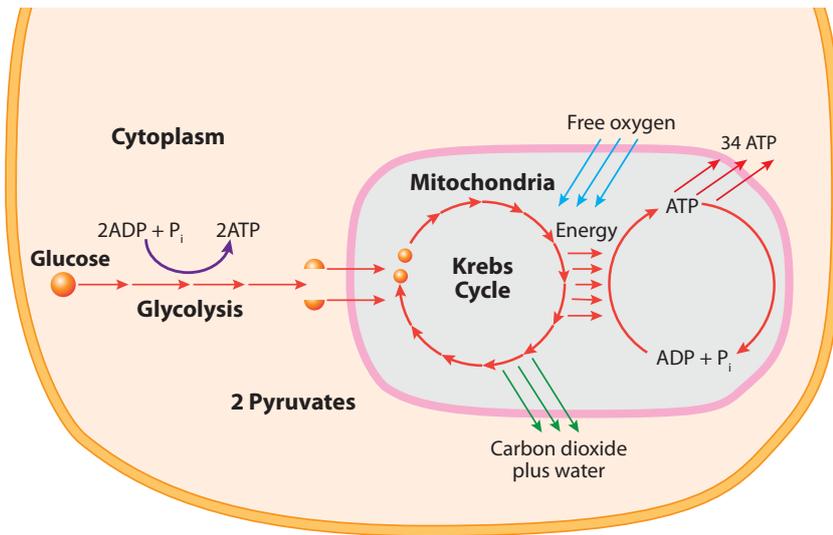


Fig. 9.10 Mitochondrion

The next two stages of aerobic respiration occur inside the mitochondria and release a much greater amount of useable energy. (See Fig. 9.10) The first of these stages is called the **Krebs cycle**, after Hans Krebs, who discovered it in the 1930s. The two molecules of pyruvic acid produced by glycolysis are converted to acetyl coenzyme A (acetyl CoA) which enters the Krebs cycle. The acetyl CoA molecules are then broken down to carbon dioxide and water. The final stage is called **phosphorylation**, and its function is to add a phosphate group to ADP, thus making the energy-rich molecule ATP. (See Fig.9.7) The energy for phosphorylation is obtained from glycolysis and the Krebs cycle in which electrons are transferred from one molecule to another.

In summary, glycolysis and the Krebs cycle break down glucose and are energy releasing processes, while phosphorylation synthesises ATP and is an energy transferring process. As a result of the completion of the two stages in the mitochondria, enough energy is released to make about 34 more ATPs. (See Fig. 9.11) Thus, about 36 ATPs are produced from the aerobic respiration of one molecule



For origin of mitochondria, see endosymbiosis page 163.

Fig. 9.11 Aerobic respiration

of glucose. Six carbon dioxide and six water molecules are released from the Krebs cycle as the only remnants of the original glucose molecule. Six oxygen molecules are consumed in the process.

The role of the mitochondria is vital in these stages. They provide four separate regions for the different reactions to occur. These regions are the outer membrane, the space between the outer and inner membrane, the inner membrane, and the space inside the inner membrane. Each of the proteins and enzymes that are necessary is located in one particular region, either embedded in one of the membranes or floating in the fluid-filled spaces. In fact, mitochondria are found in abundance in cells that have a large energy requirement, such as muscle cells or sperm cells.

NUMBER OF MITOCHONDRIA

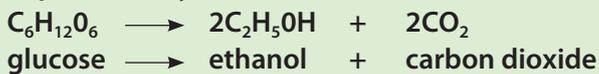
A single liver cell can have more than 2000 mitochondria whereas red blood cells have none.



Explain that fermentation is an anaerobic alternative to aerobic respiration:

in plants and yeast:

In plants and yeasts:



in animals:

In animals:



Fermentation, the process of respiration without oxygen, is also called anaerobic respiration.

MUSCLE FATIGUE

Human muscle cells are able to respire anaerobically when there is insufficient oxygen available. As a result, there is a build-up of the waste product lactic acid. It was thought that a build-up of lactic acid caused muscle fatigue. Results from recent studies suggest that this may not be the case – in fact, in some circumstances lactic acid may even improve muscle performance. A correlation between two variables does not mean that one causes the other. Lactic acid is transported in the blood to the liver, where it is converted back to pyruvic acid which is in turn broken down to carbon dioxide and water in the mitochondria of the liver cells. This process requires oxygen, and explains the body's excessive demand for oxygen following strenuous exercise – due to an 'oxygen debt'.

DID YOU KNOW?

Other fuels besides glucose can be used in respiratory metabolism, particularly aerobic respiration. Other carbohydrates, as well as protein and lipid are suitable fuels, although they must first be broken down to their constituent monomers, and amino acids must have their amine group removed in a process called deamination.

Fermentation

Many cells are also able to respire in the absence of oxygen. This process, called fermentation (or anaerobic respiration), also uses a fuel such as glucose, but the harvest of ATP is much less than for aerobic respiration, as the glucose is not completely broken down. In the case of fermentation, the products obtained depend on the type of cell involved. In plant cells and yeasts, ethanol and carbon dioxide are the products and the process is called **alcoholic fermentation**. In animal cells the sole product is lactic acid and the process is called **lactic acid fermentation**. Fermentation can be represented by summary equations (see above), although once again it must be remembered that these too are complex, multi-step processes.

Fermentation is of significant commercial importance in the wine-making and brewing industries. Under anaerobic conditions sugars are converted by yeast into ethanol (ethyl alcohol) and carbon dioxide. Yeast is also used in the baking industry – the carbon dioxide gas produced causes the product to 'rise', while the heat of the oven kills the yeast cells and evaporates the ethanol.



Compare the amount of energy released through aerobic respiration and fermentation (anaerobic respiration).

Comparing aerobic respiration and fermentation

The initial steps of aerobic respiration and fermentation are identical – both begin with glycolysis. However, in fermentation, instead of the pyruvic acid being converted to acetyl coenzyme A, it is converted to either ethanol and carbon dioxide in yeast cells and some plant cells, or to lactic acid in animal cells. This final conversion occurs in the cytoplasm. *Mitochondria are not involved in fermentation*. No oxygen is consumed and there is a net gain of only two ATP molecules per glucose molecule.

If free oxygen is present, the pyruvic acid is converted to acetyl CoA, and this is then completely broken down to carbon dioxide and water in the mitochondria. This process releases enough energy to synthesise a further 34 ATP molecules.

Note that the net gain of two ATPs from the fermentation of one molecule of glucose represents the only energy gain for the cell from this process compared to the gain of 36 ATPs from aerobic respiration. Aerobic respiration is about 18 times more effective than fermentation at producing ATP molecules.

The table below summarises the differences between aerobic respiration and fermentation.

	Aerobic respiration	Fermentation
Site in eukaryotes	cytoplasm and mitochondria	cytoplasm
Reactants	glucose and oxygen	glucose
Products	carbon dioxide and water	lactic acid in animals, ethanol and carbon dioxide in plants and yeasts
ATPs per glucose	36	2

- List the uses of energy within a cell.
- Name the physical form of energy which some cells can use.
 - Name a chemical which is energy rich and is used by cells.
 - What is an 'energy transformation'?
- Define the terms (a) autotroph and (b) photosynthesis.
- How do the photosynthesising cells of autotrophs differ from the cells of heterotrophs?
- Explain why it is that every organism in a food chain is dependent on the Sun.
- Describe two ways in which organisms use the Sun's heat energy.
- Glucose and ATP are both energy storage compounds.
 - What compound(s) result when the bonds in glucose and ATP are broken?
 - Why is ATP 'recycled' but glucose is not?
 - What is the source of the energy needed for the manufacture of ATP?
- Energy pathways such as glycolysis occur in small, regulated steps.
 - What advantages are there for living organisms to use of many small steps and not one large one?
 - Why are several different enzymes needed in the glycolysis pathway?
- Write the equation for aerobic respiration.
 - Identify the sites, products, and net ATP gained, for each stage of aerobic respiration.
 - How does aerobic respiration provide evidence for the common ancestry of all living things?
- Fermentation is an anaerobic alternative to aerobic respiration.
 - What is fermentation?
 - Write the equations for fermentation in animals, and in plants and yeasts.
 - How does fermentation differ from aerobic respiration and how are they the same?
 - State two commercial uses of fermentation.

FERMENTATION BY PROKARYOTES

Fermentation is not just confined to eukaryotes; many of our sour foods are produced by fermentation by bacteria. *Lactobacillus* convert lactose sugar in milk into lactic acid by lactic acid fermentation during production of yoghurt. Sour dough, pickles and cheeses are also produced by bacterial fermentation.

Some bacteria can further break down lactic acid into ethanol and carbon dioxide.

Clostridium bacteria can ferment glucose to form products such as acetic acid, butyric acid and hydrogen gas and *Methanogenic archaea* can further convert acetic acid into methane and carbon dioxide by fermentation.

Study Questions

10

Movement In and Out of Cells

LOCATION OF COMPOUNDS IN CELLS

Organic compounds are not scattered randomly throughout the cell. It is possible to determine their distribution within the cell by using specific dyes or reagents which either bind to the chemical concerned, or undergo a colour change. A well-known example of this is the reaction between starch and iodine/potassium iodide solution. The blue-black colour that results is a sure sign of the presence of starch within the cell. Using this test and others like it, it has been possible to make some generalisations about the distribution of different organic compounds in cells. (See Fig. 10.1) Some compounds, for example nucleic acids, are found in high concentrations inside the cell where they are needed, while there is little or no concentration of them in the extracellular environment.

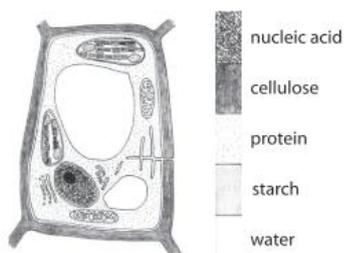


Fig. 10.1 Location of compounds in cells

In order to survive, cells require an input of matter, including gases, simple nutrients, and ions, and the removal of wastes.

All living cells must obtain materials from their surroundings and remove waste products. The materials that a cell needs depend on its function. For example, in the presence of light, a photosynthetic cell will take in carbon dioxide and release oxygen, provided that the rate of photosynthesis exceeds the rate of respiration. All heterotrophic cells require a source of energy, generally glucose, and most will require an input of oxygen in order to respire aerobically. In order to produce the range of materials needed to sustain life all cells need a supply of simple inorganic nutrients, such as ions. Examples include phosphate, chloride, nitrate, sodium, calcium, and potassium ions.

Metabolic processes in cells produce wastes that may become toxic to the cell in high concentrations, and must be removed. Depending on the type of cell and the circumstances, substances that need to be removed may include oxygen, carbon dioxide, lactic acid, ethanol, urea, and a variety of ions. Even water can be harmful to a cell if it is allowed to build up!

The cell membrane plays a vital role in allowing and/or regulating the passage of materials into and out of the cell. Now would be a good time to review the fluid mosaic model of membrane structure explained in Chapter 7.



Compare the inputs and outputs of autotrophs and heterotrophs.

Autotrophs are organisms that make all of the energy-rich organic compounds they need from simple inorganic substances, and most of them do this via photosynthesis. This means that autotrophic cells do not need to be supplied with organic compounds, such as glucose, as they can produce it from carbon dioxide and water. In eukaryotic autotrophic cells this takes place in the chloroplasts. The glucose that is produced in this way can then be used to form other organic molecules such as lipids, amino acids, nitrogen bases, and nucleotides. An input of inorganic substances containing elements such as nitrogen, phosphorus, and sulfur will also be required for this. These elements are usually obtained in the form of inorganic

substances like nitrite or nitrate ions, phosphate ions, and sulfate ions. Heterotrophic cells will also need all of these inorganic inputs, as well as an input of the organic compounds that they are unable to produce. As a result of their different metabolism, autotrophic and heterotrophic cells will have different outputs. For example, an autotrophic cell that has a rate of photosynthesis greater than its rate of respiration will have a net output of oxygen. Heterotrophic cells that are respiring aerobically will have a net output of carbon dioxide. Be careful about making generalisations – an autotrophic cell with a rate of photosynthesis lower than its rate of respiration (such as in a low light intensity) will also have a net output of carbon dioxide.

The outputs from fermentation also differ from one type of cell to another – ethanol and carbon dioxide for autotrophs, but lactic acid for heterotrophs. The tables below summarise the differences in inputs and outputs for autotrophic and heterotrophic cells.

INPUTS

Substance	Autotrophs	Heterotrophs
oxygen	for aerobic respiration when rate of respiration exceeds rate of photosynthesis	for aerobic respiration
carbon dioxide	for photosynthesis when the rate of photosynthesis exceeds the rate of respiration	not required
nitrates, nitrites	source of nitrogen for amino acid synthesis	source of nitrogen for amino acid synthesis
phosphates	source of phosphorus for nucleotide synthesis	source of phosphorus for nucleotide synthesis
calcium	a component of plant cell walls	an enzyme cofactor
other inorganic nutrients	required for synthesis reactions	required for synthesis reactions
organic compounds	not required, as they are manufactured by the cell	some are required (e.g. glucose, some amino acids, lipids), as not all can be manufactured by the cell

OUTPUTS

Substance	Autotrophs	Heterotrophs
oxygen	from photosynthesis when rate of photosynthesis exceeds rate of respiration	no output
carbon dioxide	from respiration and fermentation when their rate exceeds rate of photosynthesis	from aerobic respiration
lactic acid	not normally produced	a waste product of fermentation
ethanol	a product of fermentation	mainly yeasts
urea	not normally produced	a nitrogenous waste product from the breakdown of excess amino acids



Explain how the structure of a membrane facilitates different processes of movement through it.

DIFFUSION



tinyurl.com/mu5p8nhy

FACILITATED DIFFUSION



tinyurl.com/yc3hjdp9

OSMOSIS



tinyurl.com/y626ycyj

ACTIVE TRANSPORT



tinyurl.com/mryyjr8p

Small non-polar molecules such as oxygen and carbon dioxide are able to readily pass through the phospholipid bilayer. Protein channels allow the passage of polar molecules such as water.

As we saw in Chapter 7, the fluid mosaic model of membrane structure allows an elegant explanation of the mechanisms by which substances move through the membrane, and hence, into and out of cells. As the term 'fluid' suggests, the membrane is not static, but is a dynamic living structure. (See Fig. 10.2)

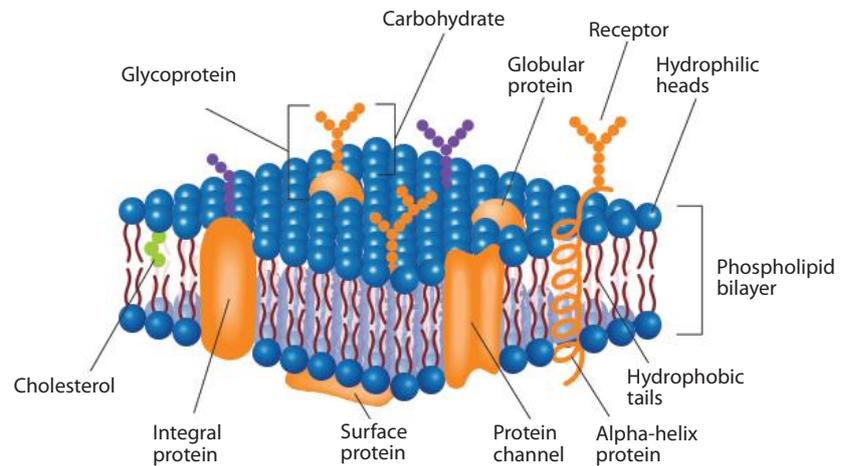


Fig. 10.2 Cell membrane - fluid mosaic model

Importance of the cell membrane

The plasma membrane restricts the passage of substances into and out of the cell. If the cell membrane allowed anything and everything to move freely through it, then the contents of the cell would diffuse out and the substances in the external environment would flow in until the concentrations became equal inside and outside the cell. A cell needs to maintain concentrations within a narrow range if it is to function normally and cell membranes have a very important role to play in ensuring that the internal environment of cells differs from their external environment. Other factors such as the level of acidity (pH) must be kept within a narrow range and this is achieved by the membrane controlling the chemical composition of the cell.

The cell membrane is not equally permeable to all substances. The size and the charge of a particle, and whether or not it is lipid-soluble are important factors. For example, if two molecules are equally lipid-soluble and have the same charge, the smaller one will pass through the membrane at a faster rate than its larger counterpart. Small uncharged molecules (such as water) are able to pass through the membrane easily, whereas even small ions (which carry a charge) find the membrane difficult to penetrate.



Explain the roles of transport proteins, including channel proteins (such as aquaporins), and carrier proteins.

Transport proteins are located in the cell membrane and their function is to speed up the movement of a substances into and out of the cell. There are two major types of transport protein: **channel proteins** and **carrier proteins**.

Channel proteins do not bind to the substances that move through them passively, with the concentration gradient. The movement is **diffusion** or **osmosis** and is a very rapid process. An aquaporin is an example of a channel protein.

Carrier proteins bind to specific molecules and some assist them to move passively across the cell membrane with the concentration gradient. This process is **facilitated diffusion**, does not require energy, and is considerably slower than movement through channel proteins. A glucose transporter protein (GLUT) is an example of a type of carrier protein. (See GLUTs textbox on page 155).

Other carrier proteins are **protein pumps** that bind to selected substances, particularly ions, and move them against the concentration gradient, using energy from the breakdown of ATP. This is **active transport**.

Ion	Concentration inside (mM)	Concentration outside (mM)
Sodium (Na ⁺)	5 - 15	145
Potassium (K ⁺)	140	5
Magnesium (Mg ⁺⁺)	30	1
Calcium (Ca ⁺⁺)	1	2.5
Chloride (Cl ⁻)	4	110

An interesting example from the table is magnesium which has a concentration inside the cell that is 30 times higher than outside the cell. Magnesium is used to activate some enzymes and to maintain the structure of protein complexes.

Substances move in and out of cells by processes such as:

- > diffusion
- > facilitated diffusion
- > osmosis
- > active transport
- > endocytosis
- > exocytosis.



Fig. 10.3 Human smooth muscle cell

WHAT IS DIFFUSION?



tinyurl.com/nptmbul

The concentration of a substance inside the cell may need to be very different from its concentration outside the cell. The table to the left of ion concentrations inside and outside a human muscle cell (see Fig. 10.3) illustrates this point.

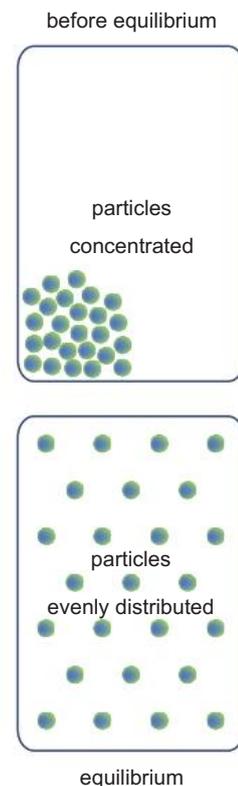


Fig. 10.4 Diffusion

Diffusion, facilitated diffusion, and osmosis are passive processes.

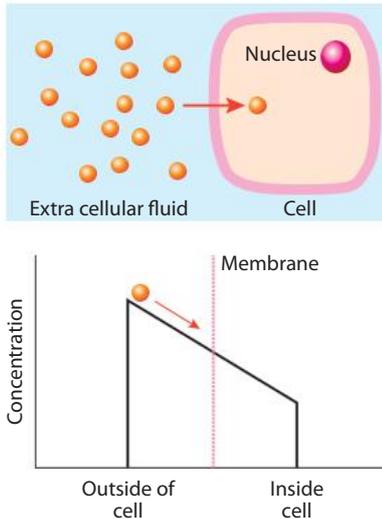


Fig. 10.5 Downhill - with the concentration gradient

Diffusion

All particles (atoms and molecules) have kinetic, or heat, energy and as a result of this they are in constant random motion. Due to this random motion there is a tendency for the particles of a substance to spread out, or diffuse, until they take up all the available space. This phenomenon is likely to be more pronounced in gases and liquids than in solids, as the particles in solids tend to be held together more tightly. This overall movement occurs due to the random movement of particles, and it will continue until the substance is equally dispersed throughout the container. We say that equilibrium is reached when this occurs. (See Fig. 10.4) Note that when equilibrium is reached the movement of particles does not cease, even though diffusion will no longer be occurring. The number of particles moving in one particular direction will then be balanced by the number moving in the opposite direction. Under these conditions the net movement is zero, and there is no concentration gradient.

A useful definition of **diffusion** is *the overall movement of a substance in a fluid from a region of high concentration of the substance towards regions of lower concentration of the substance*. The particles are moving with or following the concentration gradient. (see Fig. 10.5)

The relevance of diffusion for cells is, provided the cell membrane is permeable to a particular substance, that substance will diffuse across the membrane if a concentration gradient exists. Good examples of this include the diffusion of oxygen into cells and the diffusion of carbon dioxide out of cells. In both cases the concentration gradient is maintained by the activities of the cell. Cells continuously use up oxygen and produce carbon dioxide as a result of respiration. Diffusion is a passive process because it does not require any expenditure of energy by the cell. It occurs even in dead cells and in nonliving systems.

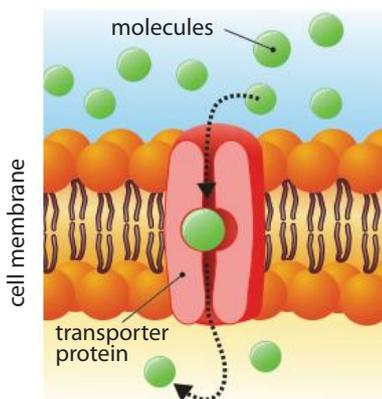


Fig. 10.6 Facilitated diffusion

Facilitated diffusion

An example of the membrane's ability to be selective is that it allows a fairly large molecule like glucose to diffuse through, while preventing the passage of other, smaller molecules. Transporter proteins in the membrane bind to certain ions or molecules and assist them across the membrane. (see fig.10.6) Other ions or molecules, even though they are smaller, have no specific protein to help them, and so cannot move across. When transport proteins assist the movement of substances such as glucose, amino acids, and ions along the concentration gradient, from a region of high concentration towards a region of lower concentration, the process is known as facilitated diffusion. No energy is required for this passive process.

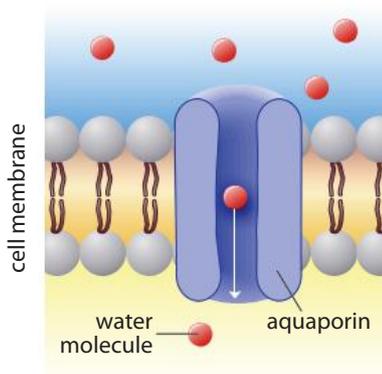


Fig. 10.7 An aquaporin - osmosis

Osmosis

Simply, osmosis refers to the *net movement (diffusion) of solvent across a semi-permeable membrane towards a region of higher solute concentration*. Cell membranes contain proteins called **aquaporins** that act as channels for the movement of water molecules. (See Fig. 10.7) In organisms the solvent is water and osmosis is just a special case of diffusion. Although it requires a semi-permeable membrane, osmosis can occur outside living cells and is a **passive** process. While it sounds strange to think in terms of the 'concentration of water', this may help you to understand that water is really just diffusing from a higher concentration of itself to a region of lower concentration of itself, or from a 'more watery' solution to a 'less watery' solution.

The role of the membrane in osmosis is to allow water to move across more easily than the solute. The significance of osmosis to cells is great, since it is the osmotic pressure that maintains the shape of an animal cell and provides support in plant cells. However, this pressure needs to be just right, and a human red blood cell will burst if it is placed in distilled water, or it will shrivel if placed in a concentrated salt solution. A solution of 0.9% sodium chloride will exactly balance the concentration of solutes in the cell's cytoplasm, resulting in no net movement of water, that is, no osmosis. Such a solution is known as 'physiological saline'. The result of excess uptake of water in plant cells is slightly less severe than in animal cells, due to the presence of the **cell wall**. When placed in distilled water, plant cells will not burst, but will completely swell with water, and are said to be fully **turgid**. If plant cells lose water, they become flaccid, or limp. Under conditions when cells become **flaccid**, the whole plant will wilt, and if the water is not replaced it will eventually die. (See Fig. 10.8)

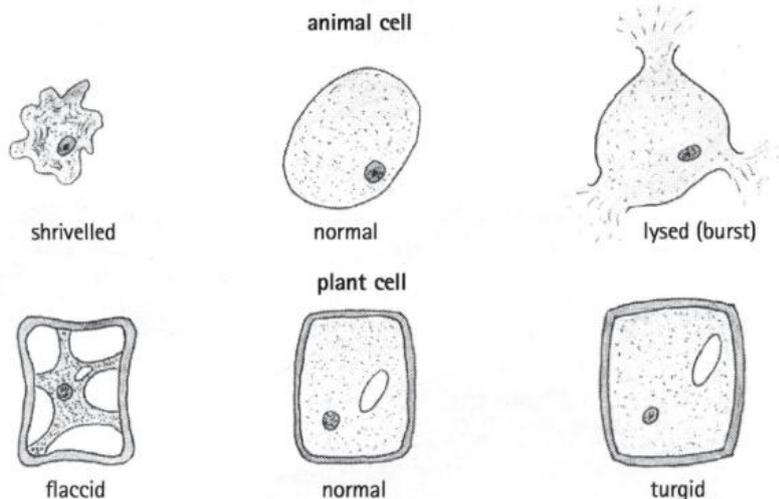


Fig. 10.8 Effects of osmosis

Note that the term 'osmosis' is only used to describe the diffusion of water across a membrane. For all other substances that diffuse the correct term is 'diffusion'.

A SIMPLE OSMOMETER

An osmometer is a tube with a semi-permeable membrane stretched across its entrance. The osmometer has a solution poured into it and it is then placed in a beaker containing pure water. Water will move across the membrane into the tube due to osmosis. The change in the level of the solution in the tube is a measure of the osmotic effect. The more concentrated the solution, the higher the level will rise. (See Fig. 10.9)

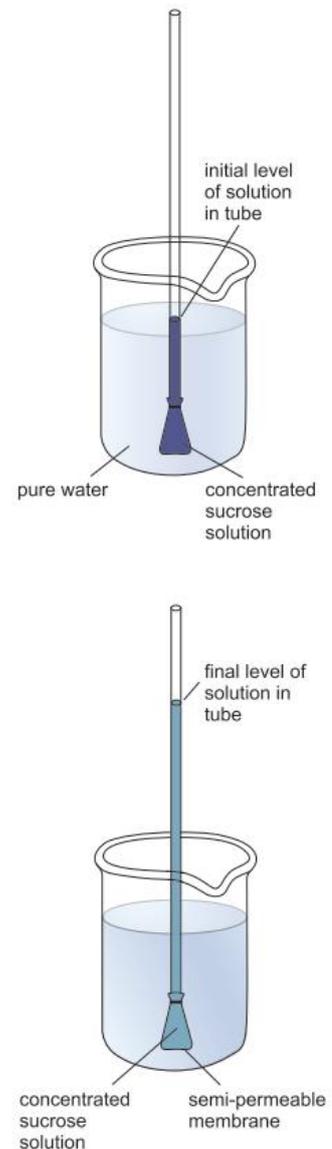


Fig.10.9 A simple osmometer

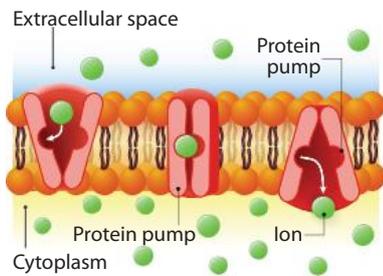


Fig.10.10 Active transport

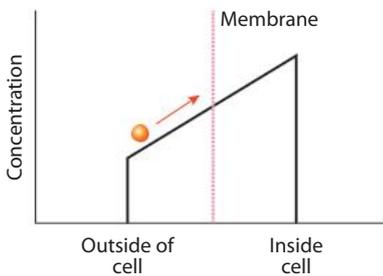
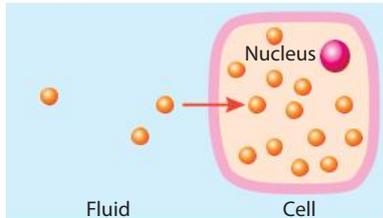
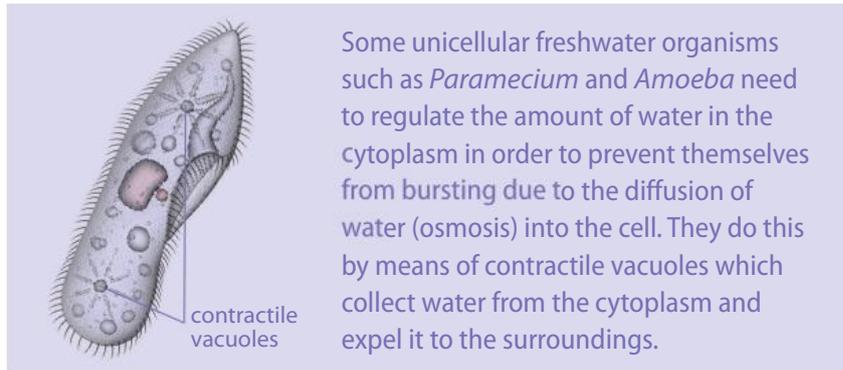


Fig. 10.11 Uphill - against the concentration gradient

MOVEMENT OF GLUCOSE

Glucose transporter proteins (GLUTs) move glucose by facilitated diffusion. Active transport of glucose is carried out by protein pumps called SGLTs.

One of the most spectacular examples of the differing proportions of chemicals in a cell's internal and external environments occurs in a marine alga in which the concentration of iodide inside the cells is some two million times higher than it is in the surrounding water. To achieve this difference the cell will expend considerable energy to move the substances against the concentration gradient.



Some unicellular freshwater organisms such as *Paramecium* and *Amoeba* need to regulate the amount of water in the cytoplasm in order to prevent themselves from bursting due to the diffusion of water (osmosis) into the cell. They do this by means of contractile vacuoles which collect water from the cytoplasm and expel it to the surroundings.

In multicellular animals, such as humans, the water content of the cells is determined by the tissue fluid that surrounds them. The composition of the tissue fluid is regulated by organs such as the kidneys. The water content of the cells is therefore regulated without the need for contractile vacuoles.

Both diffusion and osmosis are passive processes and do not require an input of energy from the cell. This is because materials are moving with the concentration gradient.

Active transport, endocytosis, and exocytosis require the expenditure of energy, usually supplied in the form of ATP.

Active transport

This process is the opposite of diffusion, since substances are moved across the membrane against the concentration gradient. A living cell is necessary, and energy, usually in the form of ATP, is required for this process. Membrane carrier proteins are involved. (See Fig. 10.10) You can think of active transport as being like pumping water uphill, (See Fig. 10.11) although you should note that in cells water is not moved by active transport. The cell is selective about what it transports in or out. In fact, a cell may be transporting one substance into the cytoplasm (potassium ions, for example), while transporting another out (such as sodium ions).

Late in pregnancy, the foetus obtains large protein molecules called antibodies from the mother's blood. To achieve this, the cells of the placenta actively transport the antibodies across their cell membranes.

Endocytosis

Some cells are able to take in particles or large molecules by enclosing them in a membranous vacuole. This process is called **endocytosis** and there are two main types, called **phagocytosis** and **pinocytosis**.

Phagocytosis involves the intake of particles and is sometimes referred to as 'cell-eating'. The membrane invaginates in the vicinity of the particle, and encloses it in a vacuole which then breaks away from the cell membrane and enters the cytoplasm. Small membrane-bound vesicles containing digestive enzymes then fuse with the

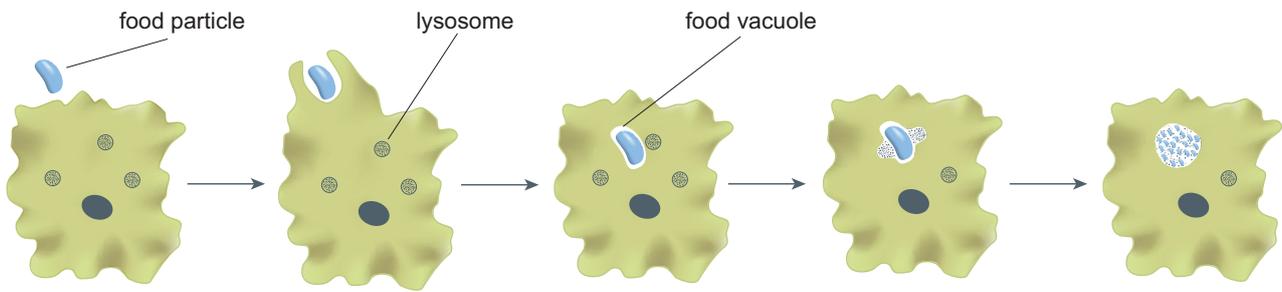


Fig. 10.12 Phagocytosis

‘food vacuole.’ These vesicles, called lysosomes, then release the digestive enzymes, which break down the particle. (See Figs. 10.12 and 10.13) Examples of cells that are phagocytic include unicellular *Amoebae* which feed in this way, and certain cells of our immune system that are involved in engulfing and destroying foreign particles like bacteria. Cells that carry out phagocytosis, are selective, and will not engulf just any particle. *Amoeba* cells will only engulf particles of nutritional value.

Pinocytosis, otherwise known as ‘cell-drinking’, is a similar process on a much smaller scale, and involves the intake of liquids or large molecules into tiny vesicles that form at the surface of the cell. Pinocytosis may be non-selective, and involve the intake of extracellular fluid, or it may be more specific, and involve the intake of certain large molecules, such as fat droplets in the small intestine. (See Fig. 10.13)

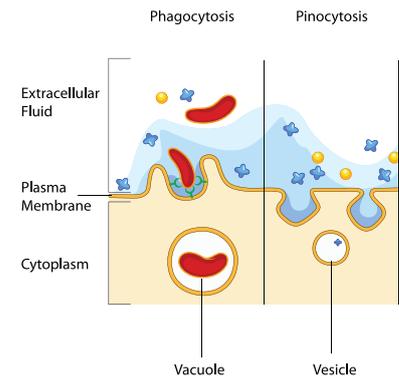


Fig. 10.13 Endocytosis

Exocytosis

Exocytosis is essentially the opposite of endocytosis. Secretion of materials produced by the cell usually involves packaging the material into a vesicle, which migrates to the plasma membrane with which it fuses, and then releases its contents to the outside. The manufacture of the material would most likely have occurred in the endoplasmic reticulum, and the packaging is a function of the Golgi body. The cells of the salivary glands, which produce and secrete saliva, are a prime example of this process, as are the cells of endocrine glands which produce and secrete hormones. (See Fig 10.14)

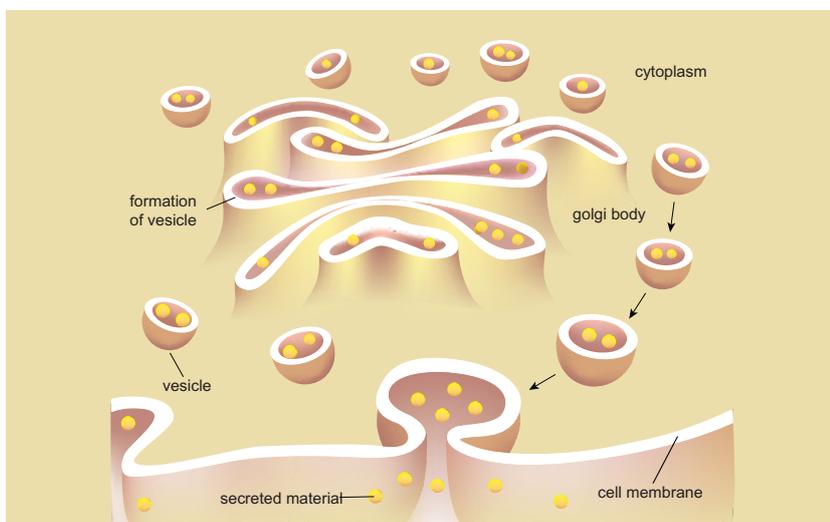


Fig. 10.14 Exocytosis

ENDOCYTOSIS



tinyurl.com/38antukr

EXOCYTOSIS



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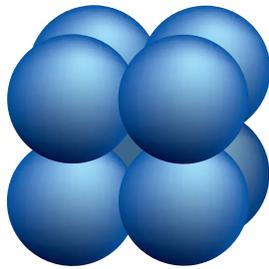


Explain how the exchange of materials across membranes is affected by factors including:

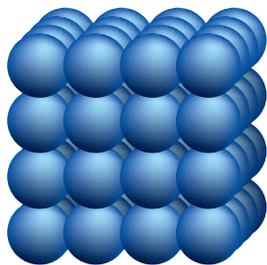
- › **surface-area-to-volume ratio of the cell**
- › **concentration gradients**
- › **the physical and chemical nature of the materials being exchanged.**



Small surface area : volume



Medium surface area : volume



Large surface area : volume

Fig. 10.15 Surface area : volume - as the block is divided up the surface area is increased, while the volume remains constant.

Surface Area Compared to Volume

The reason for microscopic size of most cells is thought to be due to the fact that as their size increases, their surface area to volume ratio decreases. (See Fig. 10.15) Cells obtain their nutrients from their surroundings by taking them in through their membrane. They also remove waste products by passing them out through the membrane. As the cell gets bigger there is proportionally less surface area, and as a result there is a decrease in the efficiency with which the cell can exchange materials with its surroundings. It may be that the ability of a cell to perform these functions efficiently limits its size. There is also the notion that the DNA, which controls the activities of the cell, may only be able to exert an influence over a finite volume. This factor would also limit the size to which most cells can grow.

Concentration gradients

As we saw in the section on diffusion earlier in this chapter, the direction of movement of a substance across a membrane when there is no input of energy depends on the difference in concentration of the substance on either side of the membrane. The particles move *with* the **concentration gradient** from a region of higher concentration of the substance towards a region of lower concentration of the substance, until equilibrium is reached. Examples of the passive movement of materials across membranes include diffusion and osmosis. In the case of cell membranes osmosis refers to the movement of water.

If particles move across membranes *against* the **concentration gradient** from a region of lower concentration of the substance towards a region of higher concentration of the substance, energy is required. Earlier we discussed examples of the active movement of particles, including active transport, endocytosis and exocytosis.

Nature of the exchange materials

Small, uncharged particles diffuse easily through cell membranes while particles that are large or charged require assistance from channel proteins, carrier proteins or even whole sections of the membrane.

Study Questions

1. In order to survive, cells require an input of matter and removal of wastes. Compare the inputs and outputs of autotrophic and heterotrophic cells in the following table, explaining why each is needed.

Substance	Input or output	Plant cell	Animal cell
oxygen	input		
oxygen	output		
carbon dioxide	input		
carbon dioxide	output		
nitrates			
phosphate			
lactic acid			
ethanol			
urea			

2. A function of the cell membrane is to control the movement of substances into and out of the cell. Why is this function vital for the proper functioning of the cell? Refer to specific examples in your answer.
3. The current model of the cell membrane is called the 'fluid mosaic' model. Why is it a 'mosaic' and why is the membrane described as a 'fluid'?
4. Explain how the structure of a membrane allows for different processes of movement through it.
5. (a) Define the term diffusion.
 (b) How does facilitated diffusion differ from diffusion?
 (c) How are these two processes similar?
6. (a) Explain the terms concentration gradient, equilibrium and passive transport.
 (b) Name two substances that move into human cells by diffusion and two substances that move out of human cells by diffusion.
 (c) State three factors which determine whether a substance will move across a membrane by diffusion.
7. In living organisms only water moves due to osmosis.
 (a) How does osmosis compare to diffusion?
 (b) Outline the special conditions which must apply in order for osmosis to occur.
8. (a) What is active transport and how is it different from diffusion and osmosis?
 (b) What is the role of a 'carrier protein' in active transport?
 (c) Name two substances that transport proteins move across membranes.
9. (a) Explain the difference between endocytosis and exocytosis.
 (b) Explain the difference between phagocytosis and pinocytosis.
 (c) Why are these processes all considered to be active, not passive?
10. (a) Explain why the size of most cells is limited by the surface area to volume ratio.
 (b) What other factor limits a cell's size?

11

Cell Metabolism

A metabolic pathway is a sequence of chemical steps that occurs in a cell

Cell metabolism is critical to the survival of cells.

To maintain life, cells need to synthesise a large number of compounds such as proteins, carbohydrates, lipids, and nucleic acids. They also need to break down compounds such as glucose to provide energy. Even a simple cell, such as a bacterium, carries out hundreds of chemical reactions. For more complex eukaryotic cells there are thousands of chemical steps. The biochemical processes that take place in a cell are referred to as **cell metabolism**.

Biochemical processes in the cell are influenced by:

- › the nature and arrangement of internal membranes
- › the presence of specific enzymes

As we saw in Chapter 3, cells are like little chemical factories. In the case of eukaryotic cells, many of the chemical processes take place in specialised structures that are often bounded by and/or contain membranes. These membranes may contain enzymes that catalyse the chemical processes.



Explain how the structure of internal membranes of mitochondria and chloroplasts facilitates some biochemical processes.

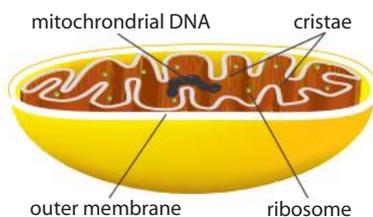


Fig. 11.1 Mitochondrion

Internal membranes of mitochondria

From Chapter 8 we know that each mitochondrion has an outer membrane and an inner membrane which is folded to form structures called **cristae**. (See Fig.11.1) The cristae provide a large surface area for the attachment of enzymes such as ATP synthase, that play an important role in aerobic respiration. The folded inner membranes of the mitochondria are the main site of ATP production.

Internal membranes of chloroplasts

Chloroplasts have two outer membranes, and an internal system of membranous flattened sacs called thylakoids. These thylakoids are arranged in stacks called grana (singular granum). The fluid which surrounds the membranous sacs is called the stroma. (See Fig. 11.2) Photosynthetic pigments such as chlorophyll are contained within the thylakoid membranes. In the first stage of photosynthesis the pigments are able to trap light energy. Thus the thylakoid membranes play an important role in the conversion of light energy into chemical energy.

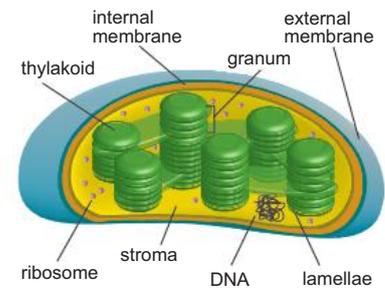


Fig.11.2 Chloroplast

The role of enzymes in metabolic pathways



Explain that in a metabolic pathway:

- › there are many regulated steps
- › each step loses some energy as heat
- › some steps produce intermediate compounds
- › specific enzymes are required at each step.

The most common source of energy for cells is glucose which is broken down in a process called **cellular respiration**. Respiration begins with the conversion of glucose to pyruvic acid (pyruvate). This metabolic pathway is called **glycolysis** and it occurs in the cytoplasm.

An energy pathway: glycolysis

Like many other chemical processes in the cell, glycolysis consists of *many regulated steps* — a **metabolic pathway**. (see Fig.11.3) Each step in the pathway is catalysed by a *specific enzyme*. In each step there is an intermediate compound produced that is converted to the next intermediate compound in the metabolic pathway. The step-wise conversion of glucose to pyruvic acid enables the cell to control the process. This ensures that the reaction does not occur too rapidly (and burn up the cell) and it maximises the amount of energy that is stored in ATP molecules during the process. Even so, some energy is inevitably lost as heat at each step. (See Fig.11.3)

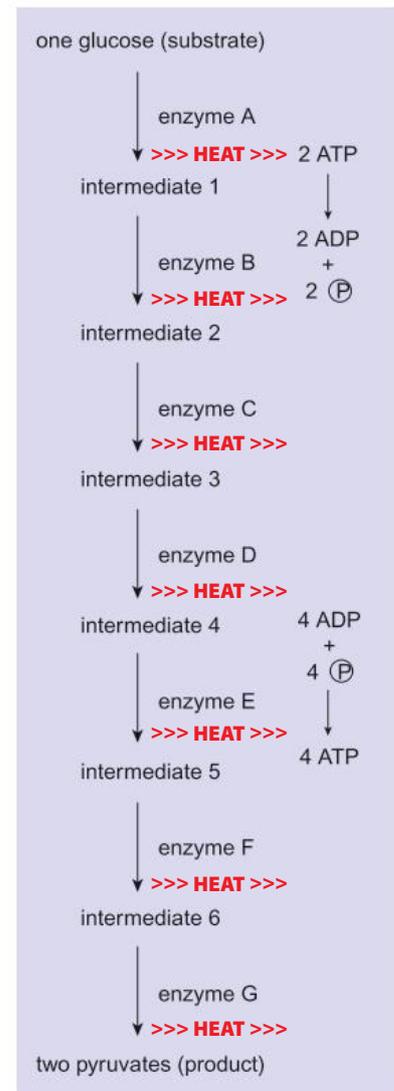


Fig. 11.3 Glycolysis – a metabolic pathway that begins respiration

Chemical reactions and energy

All chemicals contain stored energy and chemical reactions result in a change in the amount of stored energy. The change in the amount of stored energy resulting from a chemical reaction is measured by comparing

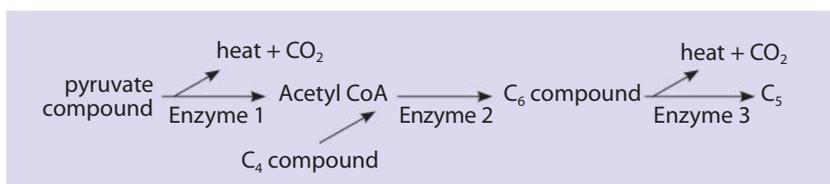


Fig. 11.4 The start of the Krebs cycle

KREBS CYCLE

When oxygen is present, the pyruvic acid molecules from glycolysis can be further broken down to carbon dioxide and water. This second stage of respiration, called the Krebs cycle (or citric acid cycle), occurs in the mitochondria, using enzymes on cristae. Like glycolysis, the Krebs cycle is a series of regulated steps, each controlled by a specific enzyme and producing an intermediate compound and heat. Details of the process of respiration are discussed in Chapter 9.

the energy of the reactants with the energy of the products. Reactions that result in products with less energy than the reactants are called **exergonic**, while those whose products have more energy than the reactants are called **endergonic**. Exergonic reactions release energy, whereas endergonic reactions consume energy.

The importance of enzymes

Many cells obtain energy by breaking down glucose to carbon dioxide and water in the presence of oxygen. However, glucose exposed to the air on a laboratory bench does not break down. How can this be explained? For life to exist, the chemical reactions that occur in cells must be controlled by specific enzymes that lower the activation energy needed. If this was not the case then chemical processes essential to life might occur too slowly, or not at all. (See Fig. 11.5) Of course, it would be just as life-threatening to have exergonic reactions, such as the breakdown of glucose to carbon dioxide and water, occur in a single step. There would be so much heat energy released that the cell would burn up!

You will remember from Chapter 3 that enzymes are globular proteins which have a region on their surface that has a specific shape. (See Fig. 11.6) This shape is complementary to the shape of the reactant or reactants of the reaction that the enzyme will catalyse. This region on the enzyme molecule is called the **active site** and the reactant or reactants, which are referred to as the enzyme's substrate (or substrates) actually undergo the reaction here. (See Fig. 11.7) The mode of action of the active site allows an explanation of one of the most important features of enzyme function — specificity.

For example, maltase, the enzyme that catalyses the breakdown of maltose, will not catalyse the breakdown of sucrose. This is because the shape of sucrose is not complementary to the active site of maltase. Thus, every step in a metabolic pathway is catalysed by a different specific enzyme. (See Fig. 11.3)

Biochemical processes in the cell are influenced by environmental factors

As we have seen, biochemical processes in cells are influenced by the presence of specific enzymes. Enzyme-controlled reactions are affected by factors including temperature, pH, the presence of

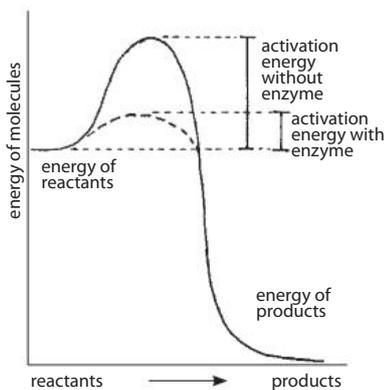


Fig.11.5 Activation energy

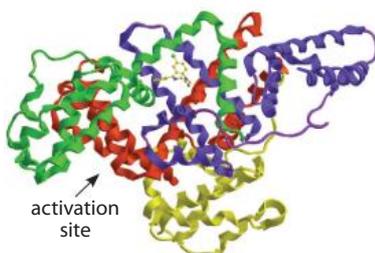


Fig. 11.6 An enzyme is a globular protein

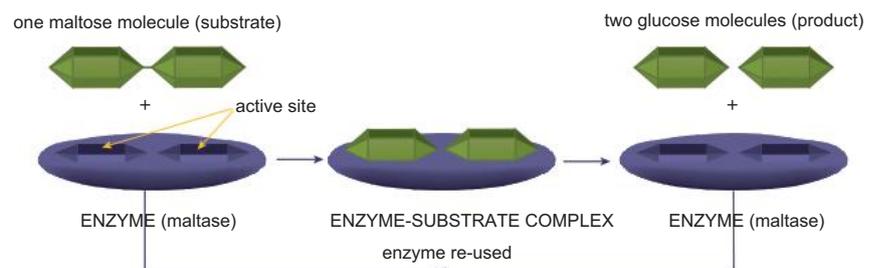


Fig. 11.7 Action of an enzyme

inhibitors, the concentration of reactants, and the concentration of the enzyme. Thus, biochemical processes are influenced by these environmental factors. Read the relevant section of Chapter 3 for a more detailed explanation.

Chemicals can interfere with cell metabolism.

When you hear that a substance is a poison, whether it is carbon monoxide, cyanide, or snake venom, you are hearing about a chemical or chemicals that can interfere in some way with cell metabolism. Carbon monoxide is deadly to mammals because it binds strongly to haemoglobin, and blocks the reaction between oxygen and haemoglobin in the red blood cells. Carbon monoxide and cyanide also disrupt the events of cellular respiration by disabling one of the enzymes in the metabolic pathway. Bottles in a chemistry laboratory are not the only source of cyanide however. Cassava root, a staple food stuff of some people, has to be treated to remove cyanide-generating compounds before it is eaten. Apricot kernels have quite high levels too. Rotten egg gas, hydrogen sulfide, is also highly toxic and like cyanide, knocks out the enzyme cytochrome oxidase. Barbiturates block another respiratory enzyme. Many more chemicals affect respiration, either by binding to enzymes, or by altering the properties of the membrane of the mitochondrion, the organelle where respiration occurs.

Chemicals that inhibit protein synthesis

The table below gives some examples of chemicals that inhibit a step in protein synthesis. Chemicals that inhibit the growth of bacteria (prokaryotes) are called antibiotics.

CHEMICAL	EFFECT ON PROKARYOTES
Tetracycline	Inhibit tRNA binding to ribosomes
Streptomycin	Prevents proper assembly of ribosomes
Chloramphenicol	Stops the growing peptide moving on from one tRNA to the next
Erythromycin	Stops growing peptide moving to new codon
Rifamycin	Prevents mRNA synthesis
EFFECT ON EUKARYOTES	
Cycloheximide	Same as chloramphenicol for prokaryotes
Amanitin	Stops mRNA synthesis
EFFECT ON BOTH	
Puromycin	Causes incomplete peptides to fall off the ribosome
Actinomycin	Prevents RNA synthesis

Antibiotics often have quite specific targets. Florey and Fleming's penicillin (see Chapter 14) attacks a step in the synthesis of bacterial cell walls. This is why mature bacterial colonies are not affected by it, as they are not making new cell walls. The antibiotic valinomycin

WHY DO METABOLIC PATHWAYS INVOLVE MANY REGULATED STEPS?

- large steps would produce unfavourable conditions, such as high temperatures and acidity
- small regulated steps release small quantities of energy that can be trapped by energy molecules like ATP
- many regulated steps provide intermediate compounds that can be used as the starting points for other reactions
- each regulated step is catalysed by a specific enzyme

METABOLIC POISONS

Enzymes may be affected by competitive or non-competitive inhibitors. See Chapter 3 for details. Many metabolic poisons work by inhibiting enzymes in one of these ways. (See Fig. 11.8)

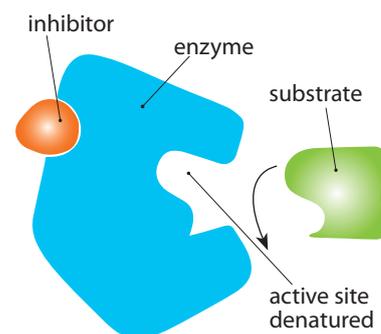


Fig. 11.8 Non-competitive inhibitor of enzymes

CONTROLLING INSECT PESTS

Nerve gases are some of the deadliest chemicals known and are the active ingredients in fly spray. The compounds in fly spray target an enzyme, acetylcholinesterase, which is involved in the breakdown of compounds that pass a nerve impulse between cells. (See Chapter 17.) When these poisons are present, the enzyme does not break down the nerve transmitter chemicals, and so the nerve signals cannot be switched off. Muscles remain permanently contracted. The insecticide malathion works this way, but it is a fine balancing act to get enough spray into the air to kill insects but not to damage humans!

High on the list of '1 gram could kill a million people' substances are bacterial toxins. Some proteins produced by bacteria such as *Clostridium botulinum*, do in fact come into that category. Less than 1 microgram is fatal to a human. But there are useful toxins too. The genes that produce an insect-killing toxin in the bacterium *Bacillus thuringiensis* (or Bt) can be incorporated into plants, which then synthesise the toxin by themselves. Not only is crop yield improved, but it also means much less insecticide needs to be used.

works by binding to potassium ions and taking them across the cell membrane and out of the cell. The bacterial cell works itself to death trying to recover its lost potassium.

Heavy metals such as lead act as non-competitive inhibitors of many enzymes in the metabolic pathways of cells. Lead poisoning is therefore due to an interference in the chemical reactions needed by the cells.



Discuss possible benefits and/or harmful effects of chemicals that human beings use.

Benefits and harmful effects of using chemicals

Humans use a vast array of chemicals, from inorganic compounds to large synthetic biochemicals. We need to consider whether the benefit we derive from them outweighs the cost. When someone benefits it may be at the expense of other humans, or it may be at the expense of other living species.

Radium is a radioactive element that occurs naturally and has had numerous medical applications. It was once thought to be healthy to bathe in natural spa waters containing radium. Radium was also used in the manufacture of luminous paint and was used on watch faces and navigation compasses. Unfortunately, the dangers of working with it were not known until many people who handled it developed cancer. In the case of users of luminous paint this often took the form of cancer of the mouth from licking their paint brushes to get a fine tip. The radiation from radium had caused mutations in their cells.

Mercury has many uses, as it is the only metal that is a liquid at room temperature. It is reasonably toxic as the element, but if it is allowed to escape into the environment and to end up in anaerobic sediments it becomes even more dangerous. Here bacteria will convert it to a particularly nasty and very poisonous compound, methyl mercury. This enters the food chain and accumulates in higher order consumers, such as humans, to the point where it can cause serious illness by disrupting metabolic pathways. Mad hatter disease is an example of such an effect.

The benefits and costs of chemicals we use in agriculture is a major area of concern. The farmer is usually using monoculture, and wants no competition for the crop. **Herbicides** are used to eliminate weeds in the young crop, and **insecticides** are used to protect the plant or the fruit. In some cases it is important that the whole crop ripens at one time. **Hormones** may be used to thin crops or produce desirable growth characteristics so that industrial scale harvesting can be used.

The beginning of the realisation that chemicals could not be sprayed without any unwanted consequences dates from a book published in 1962 by Rachel Carson. (See Fig. 11.9) Her book, *Silent Spring*, largely targeted the effect of the insecticide DDT on the environment. DDT, which acts on the nervous system, is a cumulative poison which is now banned, except in some countries, in which indoor use is approved. DDT is still one of the best ways of controlling malaria. In 2016, malaria was still endemic in 91 countries, with 50 percent of the world's population at risk. (See Fig. 11.10) So the issues involved in the use of some chemicals are complex.



Fig. 11.9 Rachel Carson

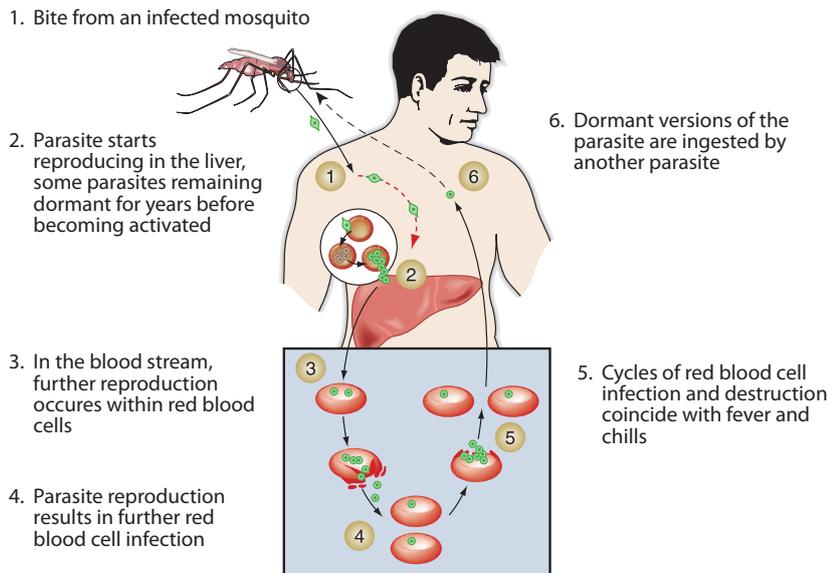


Fig. 11.10 Malaria in humans

Large quantities of our food are kept in good condition for long periods of time by using **chemical preservatives** like sulfur dioxide, vinegar or salt. In some circumstances the preservative can cause an allergic reaction in the consumer, and foods are now labelled to show the preservatives they contain. In most cases, the benefits of preservative chemicals far outweigh the detrimental effects.

The use of chemicals in medicine is a rich ground for debate on costs and benefits. One of the most famous examples is the drug *thalidomide*. It is a very effective drug for its purpose, the suppression of nausea, and it was widely used in the 1960s to treat morning sickness in pregnant women. However, children whose mothers took thalidomide during pregnancy were born with parts of their limbs missing.

Many other chemicals are used on a daily basis to maintain people's quality of life. Immunosuppressive drugs are vital for organ transplant recipients to prevent organ rejection. A variety of hormone treatments is available to combat diseases such as diabetes, to increase growth in dwarfs, to overcome hypothyroidism, and to act as contraceptives.



DDT AND MALARIA

During the mid 1950s, there was a malarial epidemic on the Pacific island of Borneo. The World Health Organisation extensively sprayed the island with the recently invented insecticide DDT to wipe out the mosquitoes that transmitted the disease. The incidence of malaria did decrease, but soon after the DDT was sprayed there was a serious outbreak of other diseases, and the villagers' huts collapsed. Investigations found that a species of caterpillar that fed on the thatched roofs of the huts was not affected by DDT, but the population of its natural predator, a wasp, was wiped out. Insect-eating lizards also died in large numbers from DDT poisoning, as did the dogs and cats that fed on the dead lizards. The rat population, normally kept in check by dogs and cats, was able to flourish, and this resulted in an outbreak of several other diseases. (see Fig 11.11)



Fig. 11.11 DDT and malaria

BOTOX

Another example of a chemical that human beings now use is botox. (see Page 140).

There are many more examples of chemicals used by humans, and some of them may have detrimental effects. Often these effects are not discovered until the chemicals have been in use for some time. Each case has to be considered on its merits and the answer is not always clear. When they were first used in the Second World War, antibiotics saved many lives, and have since been responsible for saving countless more. It is unlikely that you would know many people who have never been treated with antibiotics. But as a result of the widespread use of these chemicals over the last fifty or so years, bacteria that are resistant to all but one or two antibiotics have survived and reproduced. It will only take one so-called 'superbug', that causes a fatal human disease and that is resistant to all antibiotics, to be responsible for mass human destruction. Were the discoverers of the first antibiotics correct in putting their new-found knowledge to use and creating a new branch of the pharmaceutical industry, or should they have kept the knowledge to themselves? You be the judge!

Study Questions

1. Explain how the structure of internal membranes of mitochondria and chloroplasts facilitates some biochemical processes.
2. Define the terms cell metabolism and poison.
3. Explain why a metabolic pathway has many small regulated steps rather than one large step.
4. A number of poisons can affect the process of cellular respiration.
 - (a) What is cellular respiration and where does it take place in the cell?
 - (b) State the effects of each of the following on the process of cellular of respiration: carbon monoxide, cyanide, hydrogen sulfide, barbiturates, and antibiotics.
5. Other poisons target specific chemicals in an organism, or other cellular processes. State the chemical and/or process which is targeted by the following poisons: malathion, penicillin, valinomycin, and lead.
6. The use of chemicals has positive and negative effects. Copy and complete the following table for the chemicals listed.

Chemical	Positive effect	Negative effect
radium		
mercury		
herbicide		
DDT		
sulfur dioxide		
thalidomide		
antibiotics		

7. The use of chemicals in agriculture is a cause of concern to many people because these chemicals may end up in our food. Some plant crops are now genetically engineered to be insect resistant. State one advantage and one disadvantage to humans of these genetically engineered crops.

New Cells from Old

12

Cells arise from pre-existing cells, and cell division leads to an increase in cell number.

Cells must not only have the necessary machinery to maintain themselves, but it is also essential that they can replicate themselves. The ability to replicate is an absolute criterion for life, as living things face what is in many respects a hostile environment and cells are continually destroyed. If cells did not replicate there would be no new organisms, let alone any complex, multicellular organisms like you.

When you consider that a relatively large multicellular organism like yourself began life as a single, tiny, microscopic cell, it is obvious that there has been an enormous increase in the number of cells. All of the cells that currently make up the organs and systems of your body can be traced back to that original cell — the **zygote**. (See Fig. 12.1) Each one of the cells in your body arose from a pre-existing cell which came from another pre-existing cell and so on, back to the zygote.

The problem of cell replacement or increase in number has been solved by one cell replicating to make two descendants. The division into two is also inherent in the nature of the double helix of DNA at the heart of the cell.



Explain why the amount of DNA in a cell doubles before division.

In multicellular organisms cells need to be continually replaced, and each of the new cells must receive a copy of the genetic information stored in the DNA. For this reason the DNA in a cell is replicated (copied) before the cell divides. (See Fig. 12.2) As a result of this replication of DNA the amount of DNA in a cell always doubles before cell division. This ensures that when the cell divides each new cell will receive exactly the same amount of DNA containing identical information, provided, of course, that no mutations occur.

Your body is made up of trillions of cells, and a billion of them are dividing every second! They are continuously making more cells to replace damaged and worn out ones in structures such as skin, blood and your gut.

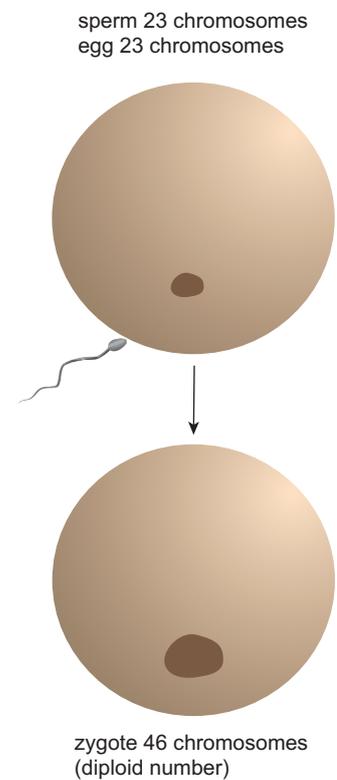


Fig. 12.1 Fertilisation restores the diploid number

Review the semi-conservative replication of DNA as explained in Chapter 2.

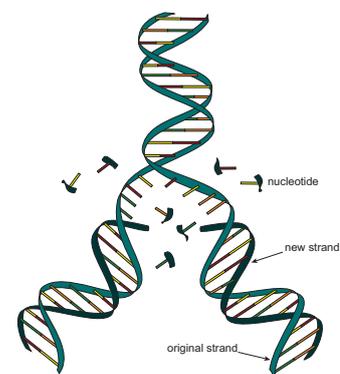


Fig. 12.2 Semi-conservative replication of DNA

CELL DIVISION



tinyurl.com/yyg919yx

Cell division in somatic cells is different from the cell division that produces gametes from germ-line cells.

The diploid number

Cells that are not sex cells are called somatic cells, or body cells. In **somatic** cells that are dividing, different chromosomes can be recognised by their characteristic sizes and shapes. This is illustrated in the karyotype in Fig. 12.3a. When the chromosomes in human somatic cells are studied carefully, it is found that each cell has two of each kind of chromosome. As explained in Chapter 2, the sex chromosomes are an exception to this rule. Two chromosomes of the same type make up an **homologous pair**. (See Fig. 12.3b). A cell that contains pairs of homologous chromosomes is called a diploid cell. We can explain the cellular events in sexual reproduction if we consider that one chromosome of each homologous pair is inherited from each parent. In human cells there are 46 chromosomes, which means 23 homologous pairs. These are called diploid cells. The diploid number in humans is 46, but this varies from species to species. However, this does not indicate the level of complexity, nor does it provide any clues to the level of intelligence, as can be seen in the following table.

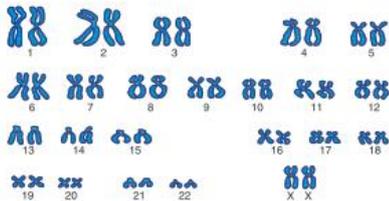


Fig. 12.3a Human female karyotype

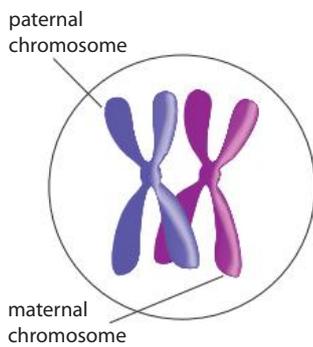


Fig. 12.3b Pair of homologous chromosomes

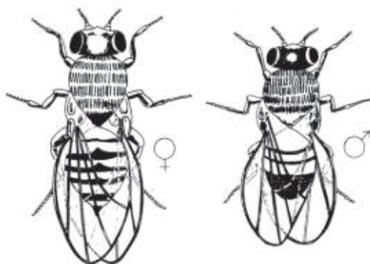


Fig.12.4 Vinegar flies

Species	Diploid number
Human	46
Vinegar fly	8
Potato	48
Mouse	40
Dog	78
Red kangaroo	20
Goldfish	94
Pea	14
Koala	16

The haploid number

The sex cells (sperm and egg cells that are derived from **germ-line cells**) have only one chromosome from each homologous pair. These cells are called **haploid**. So, two haploid cells fuse to form one diploid cell. Easy! But how are the haploid cells (sperm and egg cells) produced? Obviously, for haploid cells to be formed from diploid cells a different process must be involved.

HAPLOID AND DIPLOID

Note that the terms haploid and diploid refer to cells or organisms, but not to chromosomes.

Continuity of life requires the replication of genetic material and its transfer to the next generation through processes including binary fission, mitosis, meiosis, and fertilisation.

Each new generation must receive genetic material from the previous generation. In bacteria the process of binary fission ensures that each daughter receives an identical copy of the DNA. For asexually reproducing eukaryotes the process involves mitotic cell division. For sexually reproducing organisms, meiosis and fertilisation are involved.

In the section below we detail the processes of mitotic division and binary fission. Meiosis and fertilisation are covered in detail in Chapter 13.

The products of binary fission and mitotic division have the same number and type of chromosomes as the parent.

Although the processes of binary fission in prokaryotic cells and mitotic division in eukaryotic cells differ in their degree of complexity, in both cases the daughter cells produced receive the same number and type of chromosomes as their parent cell. This preserves the genetic information from one cell generation to the next.



Recognise, describe, and represent the process of binary fission in prokaryotic cells.

Binary fission

Although the prokaryotic cell is simple compared to a eukaryotic cell, the process of replicating the genetic material and shifting the products into daughter cells is still complex and highly coordinated. The DNA is in the form of a single, circular double helix, so it has to be cut and untwisted to allow enzymes to get to the twin strands. Enzymes with descriptive names like helicase, gyrase, swivelase and even topoisomerase are involved in this process to make replication possible. After the strands of the double helix have separated, both of the newly-formed single strands then act as templates for a new strand to form. The result is two DNA double helices (two chromosomes) in circular loops. The loops are attached separately by proteins to the cell membrane near the middle of the cell.

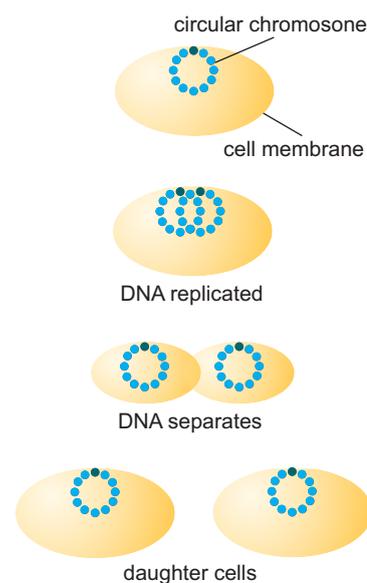


Fig. 12.5 Binary fission

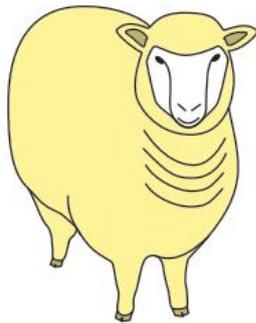


Fig. 12.6 The first mammal to be cloned was a sheep named Dolly

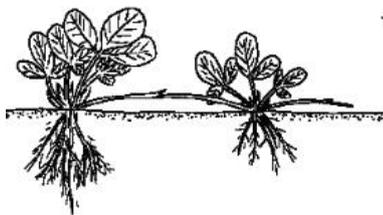


Fig.12.7 An example of vegetative reproduction

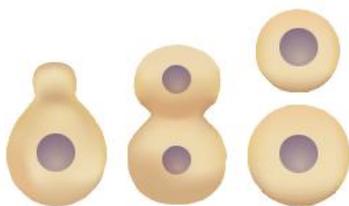


Fig. 12.8 Budding in yeast

The only source of genetic variation for the products of asexual reproduction is mutation, a spontaneous or induced change in the genetic material.

As the cell grows, the cell membrane between the attached loops expands and moves the attached loops in opposite directions. The circular chromosomes are well apart before the cell pinches across its equator to give new cells. The division into two cells is completed by the synthesis of new wall material. The process in which two prokaryotic cells are formed from one is called **binary fission** — splitting in two. (See Fig. 12.5)



Recognise, describe, represent, and name the phases of mitosis in eukaryotic cells.

In 1997 the first cloned mammal, a sheep named Dolly, was produced. (See Fig. 12.6) This cloning process involved taking the entire nuclear material from the cell of one sheep, and placing it in another sheep's ovum that had had its nucleus removed. The newly formed cell was then allowed to divide by mitosis before being implanted into the uterus of a third sheep. The result was a new individual genetically identical to the sheep that had provided the nucleus, and the process was asexual reproduction, which is not usually the case for a sheep!

The cloning of Dolly was both controversial and exciting, as it was seen by some as interfering with nature and by others as a major scientific breakthrough. However, asexual reproduction is the normal method of reproduction for many species.

Plants frequently reproduce asexually by budding, creating bulbs, or sending out runners. (See Fig. 12.7) Horticulturists make use of these processes, all of which involve mitosis, to generate large numbers of offspring from one parent with desirable features.

Budding involves the formation of a miniature offspring on the body of the parent. The new individual then breaks away from the parent and develops into a full size adult. (See Fig. 12.8)

Many animals, including seastars, sea anemones, hydras, and even some worms, reproduce asexually.

Asexual reproduction

Asexual reproduction is the process of forming offspring from a single parent without fertilisation taking place.

The role of mitosis in asexual reproduction

An important distinguishing feature of asexual reproduction is that the offspring produced in this way are usually genetically identical to their parent. The term 'parent' is deliberately used, because in asexual reproduction, there is only one parent. Not all reproduction involving one parent is asexual. (See Chapter 13)

Asexual reproduction in eukaryotic organisms involves mitotic division, since the genetic material needs to be replicated and then distributed equally to the two cells formed.

Mitosis

In eukaryotic cells, the part of the cell cycle (See Fig. 12.9) in which DNA from the original cell is equally divided between the two daughter cells is called **mitosis**. (See Fig. 12.10) Following mitosis, the contents of the cytoplasm are distributed between the two daughter cells. Other than ensuring that each daughter cell has a nucleus, this process, called cytokinesis, is much less precise.

Mitosis is divided into four stages: **prophase**, **metaphase**, **anaphase**, and **telophase**. To fully understand and appreciate the process of mitosis you need to remember why it occurs — to give rise to two new daughter nuclei with identical sets of genetic material. When a cell is not dividing, the genetic material is present in a form known as **chromatin** which consists of long, thin threads of DNA and protein, that are really only discernible under the electron microscope. You might like to think of these chromatin threads as resembling tangled spaghetti strands on a plate. During interphase, the DNA is replicated. (Imagine that each of the spaghetti strands has been doubled.) To separate these chromatin strands into two identical sets would be extremely difficult if they were to stay in this elongated and tangled form. To overcome this problem, the chromatin **condenses**, that is, the DNA strands are wound up and the chromosomes shorten and thicken and become visible under the light microscope. Each DNA strand has been replicated so that each one of these chromosomes consists of two identical **sister chromatids**, joined together at a region called the **centromere**. The first appearance of the chromosomes in the cell is an indication that prophase has commenced.

During **prophase** the nuclear envelope disintegrates, and the special regions in the nucleus called nucleoli disappear. A **spindle apparatus** begins to form. In animal cells, the centrioles divide to form two pairs, and these migrate to the regions of the cell that will become the **poles** of the spindle. Both the spindle fibres of the spindle apparatus and the **centrioles** are made of microtubules, which are made up of proteins. Plant cells do not contain centrioles. Midway between the poles is a region of the spindle called the **equator**. During **metaphase**, the chromosomes line up at the equator, also known as the metaphase plate, and attach to the spindle by their centromeres. The sister chromatids are arranged so that they face opposite poles. **Anaphase** is characterised by the V-shaped appearance of the sister chromatids as they separate from one another and move towards opposite poles. This is achieved by division of the centromeres, dynamic microtubules (spindle fibres), and the action of protein 'motors' called kinesins. The result of this carefully orchestrated process is that an identical set of genetic material has gathered around each pole.

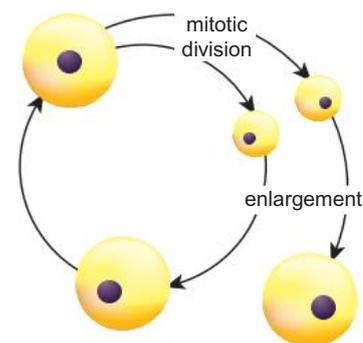


Fig. 12.9 Cell cycle

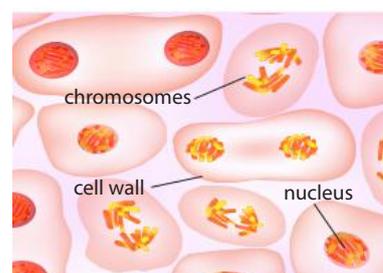


Fig. 12.10 Mitosis in onion root tip cells



tinyurl.com/3h8acnuu

Remember that this is exactly the reason for the process of mitosis. All that needs to occur to complete the process is that two new nuclear envelopes form around the two sets of chromosomes. This occurs during **telophase** when the nucleoli reappear and the short, thickened chromosomes lose their distinctive appearance and revert to long, thin strands that are only visible under the electron microscope. Mitosis is now complete, and **cytokinesis** ensures that the cytoplasmic contents are divided between the two new cells. (See Fig. 12.11)

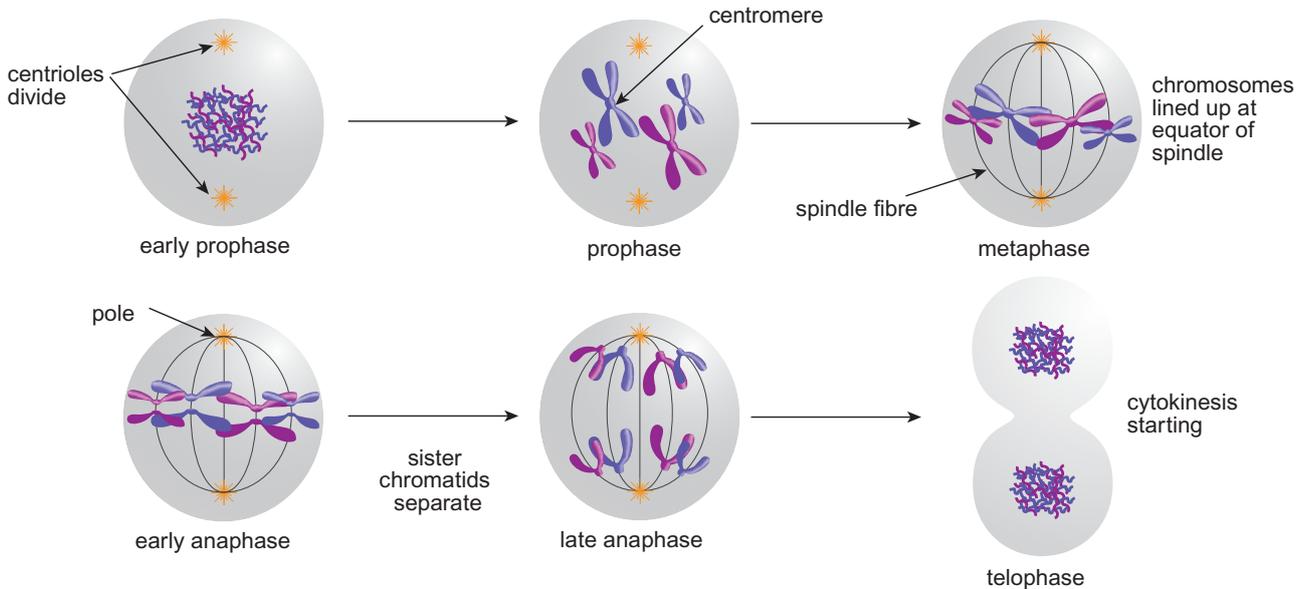


Fig. 12.11 Stages of mitosis

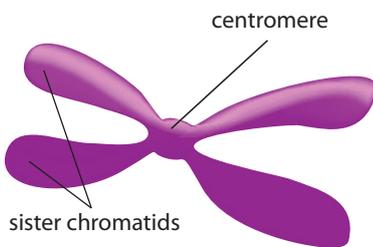


Fig. 12.12 Sister chromatids

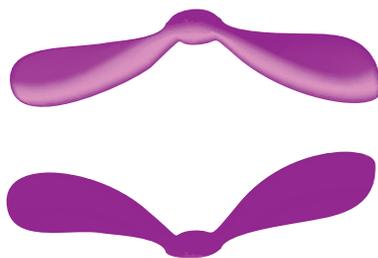


Fig. 12.13 Sister chromatids have separated – now two identical chromosomes

One of the barriers to a complete understanding of mitosis involves the term ‘chromosome’. How can a cell with four chromosomes, for example, divide in half and give rise to two cells, each with four chromosomes? Well, it depends on what you call a chromosome! When a cell is first formed, as described above, it has only one copy of each chromosome. These chromosomes are then duplicated during interphase, in preparation for the next mitotic division. When they appear in their condensed form at prophase, each chromosome is made up of two sister chromatids, joined at the centromere. (See Fig. 12.12) But, after the sister chromatids have separated at anaphase, each one can now be considered to be a ‘chromosome’. (See Fig. 12.13) So, you really can divide four chromosomes into two lots of four, provided you duplicate them first, of course!



Compare the products of binary fission and mitotic division.

The cells formed as a result of binary fission and mitotic division are genetically identical to each other and to their parent. This is because the genetic material is replicated and each daughter cell receives an identical copy. Thus the number and type of chromosomes in each daughter cell is the same as that of the parent.

Study Questions

1. Explain why the amount of DNA in a cell doubles before division.
2. Use clearly labelled diagrams to explain the terms chromosome, sister chromatid, centromere, and homologous chromosomes.
3. Explain the terms somatic cell, germ-line cell, diploid number, and haploid number.
4. Describe the process of binary fission in prokaryotic cells, and draw diagrams to represent the process.
5. For convenience, the process of mitosis has been classified into 'stages'. Use labelled diagrams to illustrate the sequence of stages during the mitotic division of a cell with four chromosomes. (two homologous pairs).
6.
 - (a) Define the term asexual reproduction.
 - (b) State the type of cell division involved in the asexual reproduction of eukaryotes.
 - (c) Why are the offspring genetically identical to each other and to the parent?
 - (d) What is mutation and why is it important for asexually reproducing organisms?
7.
 - (a) Define the terms budding and vegetative propagation.
 - (b) What is a clone? Why is the act of cloning animals considered to be controversial?
 - (c) List the types of vegetative propagation that (i) happen naturally, and (ii) require human assistance.
 - (d) What advantages might be gained by developing plants by vegetative propagation rather than from seeds?
8. Prokaryotic cells reproduce by binary fission. This process differs from mitosis in eukaryotic cells in many ways. Describe the differences between the two processes by discussing the following aspects of binary fission and mitosis:
 - (a) the way in which DNA is packed in the cell
 - (b) the nature of DNA replication
 - (c) how the sister chromatids are separated
 - (d) the way in which two new cells are finally formed and
 - (e) comparing the products of binary fission and mitotic division.

13

Sexual Reproduction and Meiosis

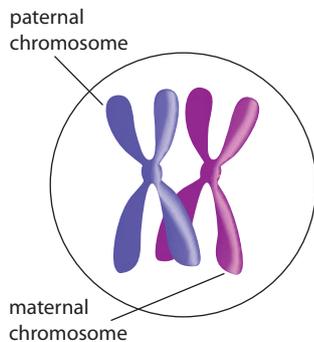


Fig. 13.1 Pair of homologous chromosomes

Note that meiosis and fertilisation occur in all sexually reproducing species.

Diploid cells contain pairs of homologous chromosomes. Haploid cells have one chromosome from each homologous pair.

In Chapter 12 we introduced the idea of the diploid number and the haploid number, as they apply to the number of chromosomes in somatic cells and sex cells. You will recall that somatic cells usually contain two of each type of chromosome – one from each parent – and that two chromosomes of the same type make up an homologous pair. (See Fig. 13.1) Gametes (sex cells) are haploid and contain one chromosome of each type.

Sexual Reproduction

In a species, the sequence of stages leading from the adults in one generation to the adults in the next generation is called the **life cycle**.

Contrary to popular belief, sexual reproduction does not necessarily involve two individual parents. Before you start drawing the wrong conclusion from this, we should define sexual reproduction. Unlike asexual reproduction, in which an offspring receives genetic information from one cell only, sexual reproduction involves the mixing (indeed, fusion) of genetic material from two cells. So, when the pollen from a flower fertilises an ovum of the same flower, or even another flower on the same plant, sexual reproduction is involved, even though there is only one parent organism. In many species, for example humans and other mammals, the male and female sex cells (called **gametes**) are produced in different individuals and therefore sexual reproduction in these organisms does involve two parents.

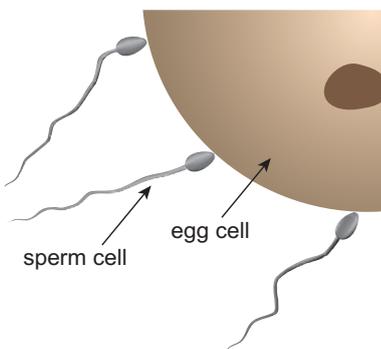


Fig. 13.2 Fertilisation

To produce gametes it is necessary for the nuclei of the cells at the 'start' of the life cycle to divide by a special process called meiosis. This results in four daughter nuclei, each with half the number of chromosomes present in the original parent nucleus. The resulting cells then differentiate to form the special cells called gametes. Gametes formed by male animals are called sperm and those formed by females are called eggs.

When a male and female gamete fuse, in a process called **fertilisation**, the result is a new cell with two sets of chromosomes. (See Fig. 13.2) This cell, called the **zygote**, is the first cell of the new organism.

You might wonder why sexual reproduction needs to occur at all; why not just break off a fragment from the parent and form a clone? One possible reason is that sexual reproduction produces offspring that show genetic variability. While it might be a good idea for many individuals to be genetically identical when things are 'going well', if the environmental conditions were to change markedly, either all would survive or all would perish. Producing populations with genetic variability would therefore seem to offer the species an evolutionary advantage and thus increase its chance of survival in an uncertain world.

Another advantage of sexual reproduction is that it masks the effects of recessive harmful genes by obtaining DNA from two sources. Either way, the species benefits.



Recognise, describe, represent, and name the phases of meiosis in eukaryotic cells.

Meiosis – producing gametes

All human body cells have 46 chromosomes. All of these cells have been formed by repeated mitotic cell divisions, and so the original cell (the zygote) must also have had 46 chromosomes, since the daughter cells of mitosis always receive an identical set of chromosomes to that of the parent cell. As the zygote was formed by the fusion of a sperm cell and an egg cell (ovum), then each of these must only contain *half* the normal number of chromosomes, that is, 23 instead of 46.

This suggests that sperm and egg cells could not be formed by mitosis, otherwise they would then have 46 chromosomes each, and the zygote resulting from their fusion would have 92! This number would then continue to increase indefinitely with each successive generation. When you consider that even one extra chromosome in an individual can produce profound abnormalities, this seems to confirm that sex cells in humans are not produced by mitosis. See examples of human chromosomal abnormalities below.

AN EXCEPTION

Red blood cells are produced with a nucleus containing 46 chromosomes, but during their development the nucleus and chromosomes are broken down leaving the cell enucleated.

SEX CHROMOSOMES

Sex chromosomes contain the genes that are responsible for determining the sex of the individual. The remaining chromosomes are called autosomes.

Humans have 44 autosomes and two sex chromosomes in their body cells.

CHROMOSOMAL ABNORMALITIES



Sometimes errors occur during meiosis and these may result in the zygote containing an abnormal complement of chromosomes. No doubt the usual fate of such a zygote is that it fails to develop further and we are not even aware of its existence. In some cases spontaneous abortion (miscarriage) may occur relatively early in the development of the foetus. Occasionally the pregnancy may proceed to full term and the baby will be born with an abnormality due to the extra or missing chromosome(s). One of the most common autosomal abnormalities results in a condition called Down syndrome. Individuals with Down syndrome have an extra chromosome number 21 – referred to as trisomy 21.

(continued on next page)

POLYPLOIDY

The presence of even one extra chromosome in animals is usually harmful, as illustrated by the examples provided. Polyploidy, the presence of extra sets of chromosomes is fatal! However, in plants, extra sets of chromosomes can be an advantage. For example, seedless watermelons, bananas, and grapes are the result of polyploidy due to the non-separation of homologous chromosomes during meiosis, followed by the fusion of cells with more than n (the haploid number). Such non-separation of homologous chromosomes is called non-disjunction. The implications of plant polyploidy in evolution are discussed in Topic 4.



Fig. 13.4 Seedless watermelon - a product of polyploidy.

Their body cells therefore have forty-seven chromosomes instead of forty-six.

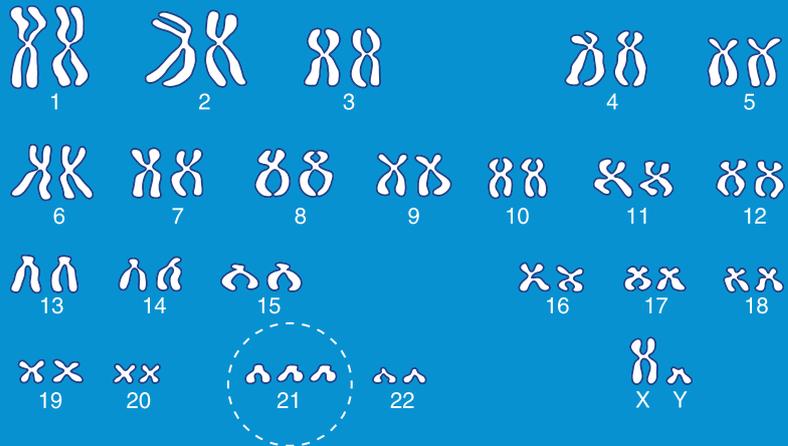


Fig. 13.3 Down syndrome karyotype

The consequences of this extra chromosome (or just an extra piece of chromosome 21) are mental retardation, a characteristic physical appearance, and a shortened life expectancy. Other conditions to result from an abnormal chromosome constitution include Turner and Klinefelter syndromes. In these cases it is the sex chromosomes that are affected. Individuals with Turner syndrome are females who have only one sex chromosome (an X chromosome). Their sex organs do not mature at adolescence, and they do not develop secondary sex characteristics. As a result they are sterile, and usually shorter than normal. Klinefelter syndrome results from a male inheriting an extra X chromosome (XXY). These males have abnormally small testes and are sterile. Often they develop enlarged breasts and other feminine characteristics.

Interestingly, the incidence of Down syndrome increases as the age of the mother increases, and pregnant women over 35 years of age are often advised to undergo amniocentesis as a precautionary measure. In this procedure a small quantity of amniotic fluid is removed by passing a fine needle through the abdominal wall. The fluid then undergoes cytological tests and the foetal cells that it contains are examined microscopically to look for any chromosomal abnormality. The use of tests such as this raises obvious ethical issues. For example, parents who discover that their unborn child is afflicted with such a disorder would then have to decide whether or not to have the pregnancy terminated. Some people with strong beliefs or opinions might decide not to have the amniocentesis in the first place.

As you have probably deduced by now, meiosis is the process by which gametes are formed. In meiosis, the chromosomes are divided in such a way that each daughter cell receives half the original number, one member of each homologous pair. For simplicity we will consider a cell that has four chromosomes, that is, two homologous pairs. We would say that it has a **diploid number** of 4 and a **haploid number** of 2. This is often abbreviated as $2n=4$ and $n=2$ respectively.

Meiosis involves two consecutive cell divisions, called **meiosis I** and **meiosis II**. As a result of these divisions four haploid daughter cells are produced, compared to the two diploid cells produced in mitosis. Like mitosis, it is convenient to divide meiosis into stages: prophase I, metaphase I, anaphase I, and telophase I, followed by prophase II, metaphase II, anaphase II, and telophase II. As you follow the behaviour of the chromosomes at each stage of meiosis in the section below, the differences between mitosis and meiosis will become apparent and this will explain the different outcomes of these processes. This is summarised in Fig.13.5.

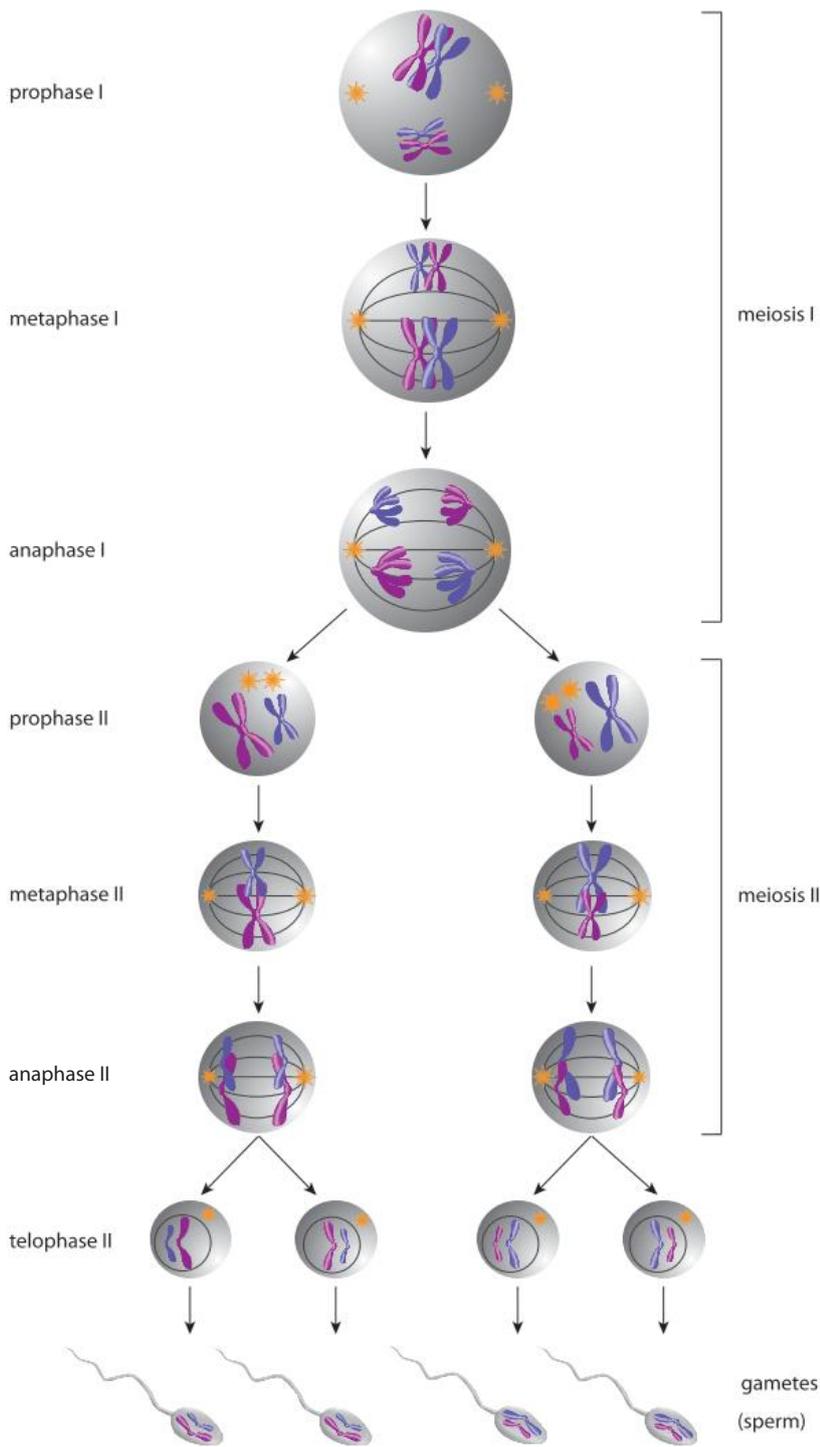


Fig.13.5 Meiosis in males (not all stages are shown) in a cell with a diploid number of four.

POLAR BODIES

In the females of some species, including humans, only one of the products of meiosis survives. In meiosis I, two cells are produced – a secondary oocyte and a much smaller ‘polar body’. Each of these is haploid. This polar body may or may not divide. Interestingly, in human females meiosis II only completes if fertilisation occurs. Then another polar body is formed. There will be one, two, or three polar bodies produced, depending on whether the first polar body divides, and whether fertilisation takes place. Human polar bodies ‘dissolve’ and play no further role in reproduction. Apparently, in some species (such as some insects) they do have a useful function.

MEIOSIS AND CROSSING OVER



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MEIOSIS AND CROSSING OVER



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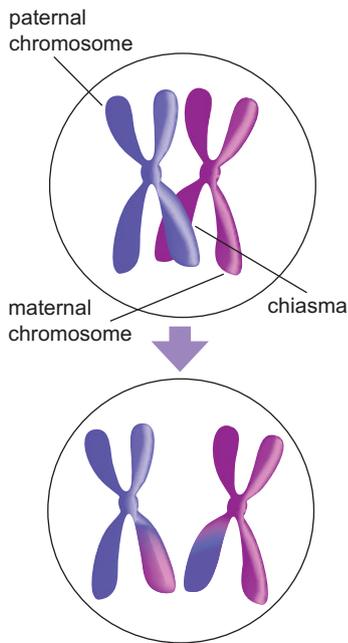


Fig. 13.6 Crossing over of homologous chromosomes during synapsis. The recombined chromosomes separate during Anaphase I

MEIOSIS - INDEPENDENT ASSORTMENT



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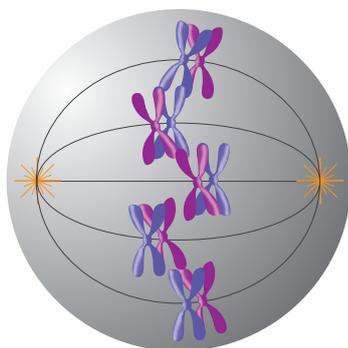


Fig. 13.7 Metaphase I – each homologous pair behaves independently of the other



Explain the importance of crossing over and independent assortment in meiosis.

Behaviour of Chromosomes in Meiosis

The chromosomes replicate before meiosis, during interphase. You will recall that this replication also precedes mitosis. However, when the condensed chromosomes appear in prophase I the homologous chromosomes do not behave separately as they do in mitosis. Instead, they pair together in a process called **synapsis**. Since each chromosome consists of two identical sister **chromatids** joined at the centromere, the structures formed by synapsis contain four **chromatids**. These structures are called **bivalents**. At this stage, corresponding sections of non-sister chromatids may touch, break, and then rejoin, such that chromosomal material is exchanged. This process is called **crossing over** and the positions at which it occurs are called **chiasmata** (singular, **chiasma**). (See Fig. 13.6) Crossing over plays an important role in increasing the genetic variability of the offspring by altering the combination of genes that is passed on from one generation to the next.

The final events in prophase I are similar to those of mitosis. In animal cells the centrioles move towards the poles, a spindle apparatus begins to form and the nucleoli and nuclear envelope disintegrate. Plant cells do not have centrioles, but the formation of the spindle apparatus, and the events that follow, are the same as for animal cells. In metaphase I, the homologous chromosome pairs (bivalents) migrate to the equator of the spindle, where they become attached to the spindle fibres so that each member of the homologous pair faces a different pole.

Each homologous pair of chromosomes behaves independently of the other pairs at metaphase I. In a zygote, one chromosome of each homologous pair has come from each gamete. We can refer to the individual chromosomes of each pair as **maternal** (from the ovum) or **paternal** (from the sperm). Thus, each body cell, derived by mitosis, contains a set of maternal chromosomes and a set of paternal chromosomes. During metaphase I of meiosis the maternal and paternal chromosomes of each homologous pair line up on the equator of the spindle independently of the other pairs. Some of the maternal chromosomes face one pole and the rest face the other pole. (See Fig. 13.7) This phenomenon, called **independent assortment** or **random assortment** greatly increases the number of possible combinations of genetic material in the gametes, by distributing the maternal and paternal chromosomes at random. This is another important source of genetic variation in sexual reproduction.



Explain why the products of meiosis are haploid cells and contain a single set of chromosomes.

Now, another important difference between meiosis and mitosis becomes evident. At anaphase I, whole chromosomes move towards opposite poles and the sister chromatids of each chromosome remain attached at the centromere. Thus, the *halving* of the diploid chromosome number occurs during meiosis I, and this is why it is sometimes referred to as the *reduction division*. By telophase I the two new haploid sets of chromosomes have gathered around opposite poles and the spindle has begun to disappear. Usually cytokinesis occurs immediately to form two daughter cells, and this is quickly followed by prophase II, in which a new spindle forms. In this case, the nuclear membrane and nucleoli do not reform between telophase I and prophase II. At metaphase II the chromosomes have migrated to the equator of the spindle and have become attached to the spindle fibres by their centromeres. This single line of chromosomes, each with its sister chromatids facing opposite poles, looks a bit like metaphase of mitosis, but remember that this time there is *only one member of each homologous pair present*, whereas in mitosis in a diploid cell, two of each type of chromosome are present.

During anaphase II the centromeres divide, the spindle fibres contract, and the sister chromatids are pulled apart towards opposite poles. At telophase II, the nuclear membrane reforms, nucleoli reappear, and cytokinesis occurs. Four daughter cells have been produced as a result of one diploid cell undergoing meiosis. Each of these four daughter cells is haploid. What is more, the events that took place during meiosis, namely crossing over during prophase I and independent assortment during metaphase I, have ensured that each of these four daughter cells has a unique combination of genetic material. (See Fig. 13.5)



Explain that fertilisation restores the diploid number.

We know that normal human diploid cells contain forty-six chromosomes. The forty-four autosomes are present as twenty-two homologous pairs. The remaining two chromosomes are the sex chromosomes, XX for females and XY for males. Normally, when gametes, or sex cells, are produced by meiosis, the chromosomes are accurately distributed such that each gamete receives one representative from each homologous pair and one sex chromosome. This will therefore be half the number of chromosomes of the diploid cells. When two gametes fuse at fertilisation, the normal diploid condition is restored. (See Fig. 13.8) The cell that is produced as a result of fertilization is called a zygote. Because fertilisation is random, that is, any male gamete can

COMPARE MITOSIS AND MEIOSIS



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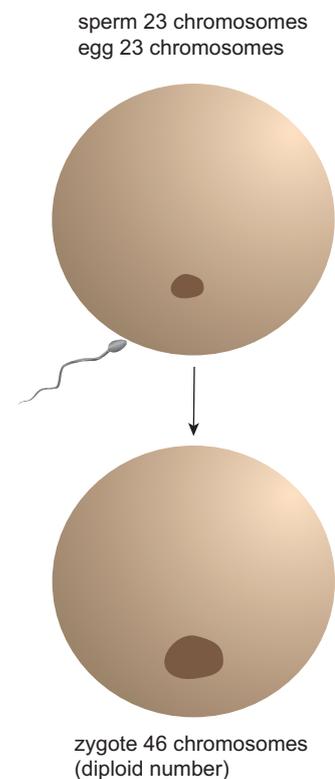


Fig.13.8 Fertilisation restores the diploid number

fuse with any female gamete of the same species, it is yet another process in **sexual reproduction** that contributes to genetic variation in the offspring. There is a constant chromosome number from one generation to the next. In humans the gametes that fuse are produced by two different individuals. In some species both male and female gametes are produced by an individual organism.



Compare the products of mitotic and meiotic cell division.

	mitotic	meiotic
number of chromosomes	diploid (2n)	haploid (n)
composition of chromosomes	identical	different
variation due to mutation only	yes	no
variation due to crossing over	no	yes
variation due to independent assortment	no	yes
type of cell	somatic	gamete
capable of further division	yes	no
capable of fusing with another cell	no	yes
number of cell products	2	4



Compare the sources and degree of genetic variation of the products of asexual and sexual reproduction.

In asexual reproduction, which involves mitotic cell division, the only source of genetic variation is **mutation**. In sexual reproduction, genetic variation results from not only mutation, but also **crossing over** and **independent** assortment during meiosis, and the **random fertilisation** of gametes. Thus, there is much greater variation in the products of sexual reproduction than in the products of asexual reproduction. The significance of this for natural selection is covered in Chapter 23.

IMPACTS AND ETHICAL CONSIDERATIONS



We saw in Chapter 4 that mutations that occur in somatic cells are confined to the individual organism in which they occur, but mutations that occur in germ-line cells may be passed on to the next generation. This has implications for the genetic manipulation of cells, where there is a perceived risk that manipulated DNA may be passed on to future generations with unknown consequences.

Study Questions

- Define sexual reproduction using the terms: gametes, fertilisation, zygote.
 - How does sexual reproduction differ from asexual reproduction?
 - What are the advantages of sexual reproduction?
- What is the difference between a gamete and a zygote?
- What are homologous chromosomes?
 - Distinguish between autosomes and sex chromosomes.
- Describe, and represent, the process of meiosis in eukaryotic cells in a sequence of labelled diagrams.
- Draw a diagram that shows a single pair of homologous chromosomes undergoing the process of crossing over. Label the sister chromatids, the centromeres and the chiasmata.
- Explain how different gametes produced by the same individual contain different mixtures of paternal and maternal chromosomes.
- Explain the significance of the process of crossing over and independent assortment in meiosis.
- How do the products of meiosis differ from the products of mitosis?
 - How does the behaviour of homologous chromosomes in prophase I of meiosis differ from their behaviour in prophase of mitosis?
- What is the difference between a haploid cell and a diploid cell?
 - Would you expect a zygote to be haploid or diploid? Explain your answer.
 - Where in the human body would you expect to find haploid cells?
- Explain why it is essential that the products of meiosis are haploid cells and contain a single set of chromosomes.
- List three ways that the products of mitotic cell division and meiotic cell division differ.
- Describe the sources and degree of genetic variation in the products of asexual, and sexual reproduction.
- Explain why it is necessary that fertilisation restores the diploid number.
- How does fertilisation contribute to genetic variability in offspring?

14

Control of Cell Division

Cell division may be regulated by internal and external factors.

In a multicellular body, the division of cells must be carefully controlled and cannot go on unchecked. The effect of cells dividing uncontrollably is seen when cancer occurs. Such 'selfish' cancer cells soon kill the organism and themselves. But organisms are also prone to wear and tear, and cells must be replaced. The undifferentiated cells in our bodies that divide rapidly are called **stem cells**. (See Fig. 14.1) A feature of the division of stem cells is that one daughter cell remains as a stem cell, and the other differentiates or at least greatly reduces its rate of division. A differentiated cell is one that has become specialised in structure and function. These cells divide either at a greatly reduced rate, or not at all. Differentiated cells tend not to be able to revert to their non-differentiated state.

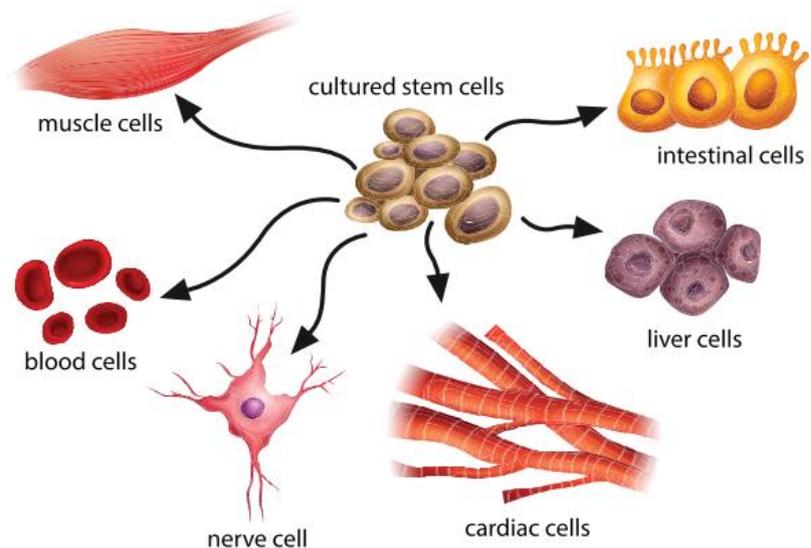


Fig. 14.1 Human Stem Cell differentiation



Describe the stages in the cell cycle (including checkpoints).

The Cell Cycle

If a cell were to divide to form two new cells, then you would expect that the size of each of the two new daughter cells would be half that of the original parent cell. If this process were to continue over and over, then all you would end up with would be smaller

and smaller cells. In order to obtain the increase in number of cells that we associate with growth in living things, there must be other processes involved as well. There must not only be an increase in cell number, but also an increase in cell size and a doubling of the amount of DNA from when a cell is first formed until it is in turn ready to divide. This alternating of cell division and enlargement is known as the **cell cycle**. (See Fig. 14.2)

After being formed as a result of cell division, a new cell enlarges by taking in water and by synthesising new cell components. This part of the cell cycle is called **interphase**, and is sometimes mistakenly referred to as a resting stage, which it is not, as not only are new organelles synthesised, but also the DNA is replicated.

Interphase is divided into three phases - G_1 , S and G_2 . The first phase is G_1 , a growth phase. DNA is synthesised (replicated) during the S phase, and G_2 is another gap, or growth phase. M represents mitosis, and it is followed by cytokinesis, when two new cells form. A cell that differentiates is said to be in G_0 . Its division apparatus is dismantled and if it is ever stimulated to divide again it does so very slowly.

In the cell cycle there are three major checkpoints — the first in G_1 , also called the restriction point, R, the second at the end of G_2 , and the third in mitosis (M). At these checkpoints the cell cycle is halted and cannot proceed until a specific signal is received by the cell cycle control mechanism. (See Fig. 14.3)

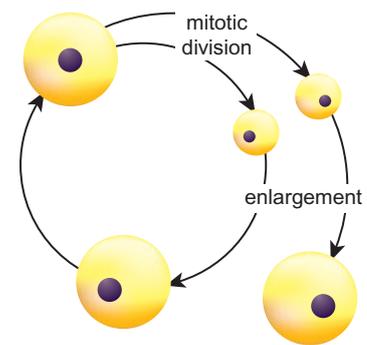


Fig. 14.2 Cell cycle



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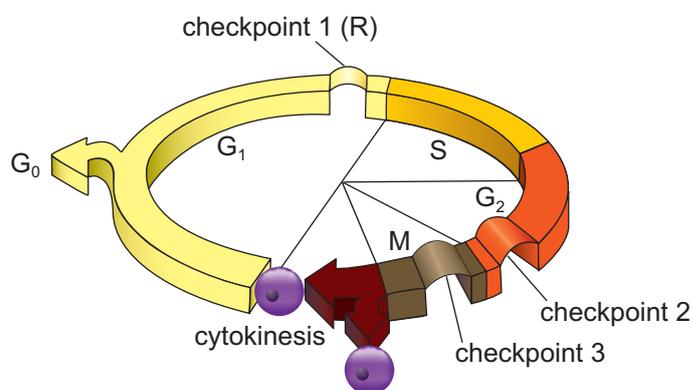


Fig.14.3 Control of the Cell Cycle

The cell produces gene products that regulate the cell cycle.

The key triggers for a cell to divide are its size and the signals it receives from the environment. If both are favourable, the cell commits itself to DNA replication and the completion of the cell cycle. The decision point of whether to divide or not to divide comes late in G_1 at the restriction point, R. During G_1 the cell has accumulated energy as ATP, glucose, and oil droplets, as copying

Review Chapter 4 to revise the link between genes and their products.

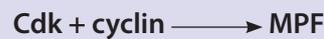
BREAKDOWN OF CYCLIN

Cyclins are proteins that control the cell cycle by combining with CDK.

The name cyclin is derived from the fact that the concentrations of cyclins in the cell rise and fall in cycles. Cyclins bring about their own destruction. They have a binding site for another protein, called ubiquitin, which is the general cell marker for damaged, distorted or erroneous proteins. Ubiquitin leads these proteins to destruction at the 'protein graveyard', the proteasome. So, during mitosis, cyclins are almost totally destroyed by the 'ubiquitin-proteasome system' (UPS).

DNA is a process that demands much energy. Once the cell has started DNA replication it must ensure that this process is complete before it moves into nuclear division, but it must also ensure that DNA is copied only once per cycle. Extra DNA or missing DNA in a daughter cell would have a devastating effect.

There is a critical second major checkpoint at the end of G₂ before the cell begins mitosis. The basis of the regulation at this point rests with two types of protein. One is the enzyme cyclin-dependent kinase (CDK) and the other is a regulatory protein called **cyclin** which activates the enzyme. When these two proteins combine they form a complex called the **maturation promoting factor** or MPF (also called mitosis promoting factor).



An increase in cyclin stimulates the start of mitosis, as the concentration of MPF will increase. (See Fig. 14.4) Under these conditions the process of mitosis will proceed through prophase and metaphase. Anaphase cannot commence until there is a decrease in MPF. (Checkpoint 3)

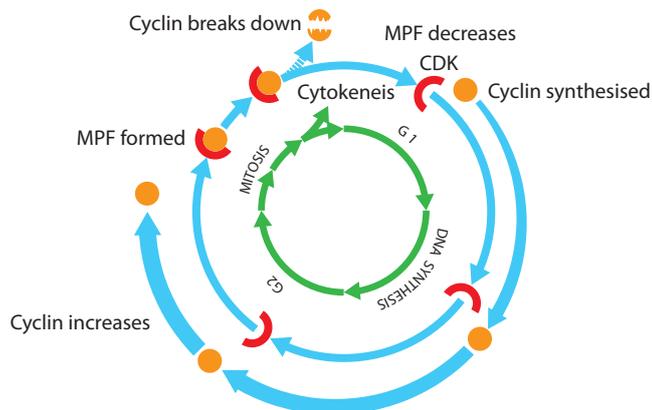


Fig. 14.4 Regulating the cell cycle

CELL CYCLE CONTROL



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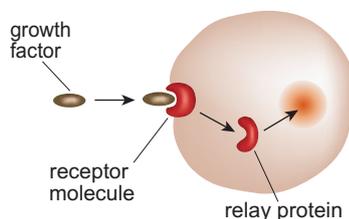


Fig.14.5 Relay protein

The external signals that stimulate cells to divide are called **growth factors**. These growth factors are proteins secreted by other cells and they bind to the receptor molecules in the cell's plasma membrane. Once this has occurred, the receptor molecules induce changes in proteins in the cytoplasm. These are called **relay proteins**, as they relay the signal from the growth factors to the cell cycle control mechanism which causes it to proceed past the restriction checkpoint. (See Fig. 14.5)

The fact that DNA replicates to form two identical copies is the reason why cells only divide in two at each division, and not more than two.

We have already seen that a cell will normally divide only if the environmental conditions are favourable. In tissue culture, when cells are grown in glass or plastic dishes in the laboratory, one of the requirements is that the cells should be in contact with a surface. If cells are growing in a dish and a line of cells is scooped out, the cells at the edge of the line will divide again until they close the gap, and touch other cells. They will then stop dividing. This is an example of a physical signal called **contact inhibition**. It is also what happens when skin is removed in an injury. The cells that are at the edge of the broken skin divide to close the gap, and then stop dividing when the gap has been closed and the cells touch.

When involved in blood clotting, platelets release a growth factor which stimulates cells to divide. As the platelets are trapped in the blood clot next to remaining undamaged cells surrounding the wound, it is these cells that divide quickly and close the wound.



Explain that hormones may regulate cell division.

Regulation of cell division by hormones

In plants three major types of hormone combine to regulate cell division in different parts of the plant. **Cytokinins** are synthesised in root tips, and **auxins** and **gibberellins** are synthesised in stem tips and other places, such as ripening seeds. These three substances are hormones, substances that are synthesised at one place and act at another place. The hormones work together so that the plant's growth is carefully coordinated. It is important that the top of the plant does not grow more than the roots can support, and vice versa.

In animals, more than fifty kinds of hormone called growth factors have been identified. Some, such as **epidermal growth factor**, target a broad range of surface cells. Others, such as **erythropoietin** (EPO) which stimulates only the production of red blood cells, are specific. These hormones are present in very low concentrations which means that cells are competing with each other for the scarce growth factor molecules. When the concentration of the hormone increases the target cells will divide more rapidly.

Another interesting example of hormonal control of cell division is the production of eggs and sperm in humans. The cells that are to form the eggs actually begin the special cell division process in the ovaries before the female is born, but the process is placed on hold at an early stage. At this point the cells are called primary oocytes. The cell division will not resume until the female reaches puberty and the hormones **LH** and **FSH** are released to restart the process. In the male, sperm production in the testes is continuous from puberty and several million sperm cells are produced each day. This production is controlled by the hormones LH, FSH and **testosterone**. The special

HUMAN GROWTH HORMONE



Human growth hormone stimulates the growth of muscles, bones, and some other organs, by increasing the rate of cell division. Excessive production of growth hormone can lead to a condition called 'gigantism' in children and 'acromegaly' in adults. Synthetic human growth hormone, produced by genetic engineering, is used to treat people with impaired growth, due to conditions such as Prader-Willi or Turner syndromes, and babies born prematurely.

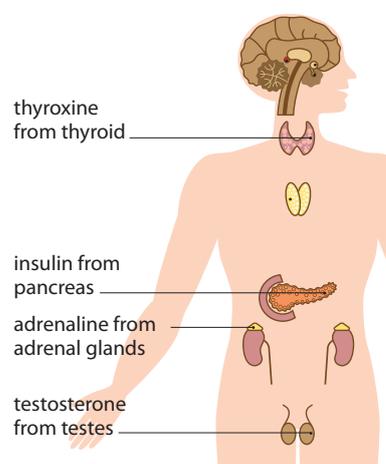


Fig. 15.3 Human endocrine system

CANCERS

Cancers result from the abnormal metabolism and growth of cells due to changes in the genetic material. These changes, or mutations, may occur spontaneously, but they can also be induced by environmental factors such as high energy radiation, including ultra-violet rays, and chemical mutagens such as those in cigarette smoke. Australia has the highest incidence of skin cancer in the world. This is due to our lifestyle, our genetic make up, and our climate. Some forms of cancer, such as colon cancer, may be diet-related.

division process involved in the formation of egg cells and sperm cells is called meiosis, and it is discussed in detail in Chapter 13.

Carcinogens upset the normal controls of cell division by causing mutations in key regulatory genes.

Uncontrolled cell division — Cancer

Roughly one person in five in developed countries will die of cancer — the uncontrolled division of cells. This proliferating cell mass may begin with a single abnormal cell. Most cancers begin when there is a change in the cell's DNA sequence, and this change is passed on to the daughter cells. If the change was not passed on to the next generation of cells then these new cells would receive unaltered DNA and be normal!

There is a strong correlation between carcinogenesis, the onset of cancer, and mutagenesis, a change in the DNA sequence. Three main classes of agent are responsible for mutagenesis:

- › chemical carcinogens causing changes to the nucleotide sequence in DNA
- › radiation, especially X-rays which produce chromosome breaks
- › viruses that can cause addition of foreign DNA sequences into the host DNA.

Evidence for chemical carcinogens has been found through some terrible effects. Workers handling the chemical 2-naphthylamine have a very high incidence of bladder cancer and many miners in the asbestos mine at Wittenoom in Western Australia developed a kind of lung cancer called mesothelioma.

Usually a single event does not cause disease, but the accumulation of several fairly rare events is needed. This is why so many cancers take between 15 and 30 years to develop. It has been estimated that each gene in your body will suffer a DNA error at the rate of one per million copies. That sounds like reasonable odds, until you learn that your body will undergo 10^{16} cell divisions. This means that there will be on average 10^{10} (10 billion) mutations of each gene during your lifetime! Exposure to mutagens will increase this number. In most cases these mutations will have no noticeable effect.

As we saw in the discussion of the cell cycle, it is normal for a dividing cell to give rise to two products with different fates; one to remain a stem cell and the other to slow its division, differentiate, and enter G_0 , where it does not divide again. When cancer occurs there may also be one of two fates for the daughter cells. Both may remain as stem cells and continue rapid division cycles, or one of the daughter cells may slow down its division but not stop dividing when it should. (See Fig. 14.6)

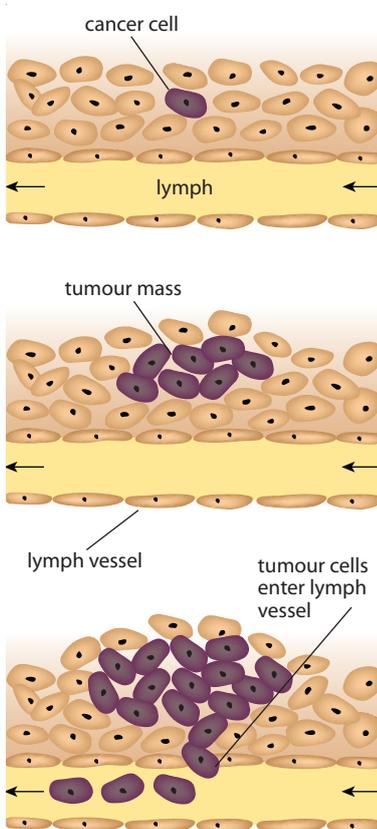


Fig. 14.6 Tumour Cells

The cells in a cancer appear to have abnormal cell cycles. In some cases it is the checkpoints that are not functioning and no growth factors are needed for the cell to move into the next phase of the cycle. In other cases, the cancer cells manufacture their own growth factors, and so their progress through the cycle is not restricted. The treatment of cancer by chemotherapy uses chemicals that interrupt the cell cycle. One class of such chemicals is called anti-mitotic drugs, as they work specifically at this part of the cell cycle.

Human beings culture cells for a variety of purposes.



Describe techniques of cell culture, and discuss the applications and limitations of contemporary examples.

What was biologist Alexander Fleming doing when he discovered penicillin? He was culturing cells of the bacterium *Staphylococcus* by growing them on a jelly-like medium. The even growth of bacterial colonies on the agar plate was interrupted at one place where the plate had been accidentally contaminated with *Penicillium* fungal spores. But the fungus did not simply overgrow the bacterium. It cleared a zone around itself by the release of the chemical penicillin, which killed the bacterial cells for some distance around the fungus. The bacterial cells had died by bursting, a process known as lysis. The penicillin story is quite complicated because the bacteria are not susceptible when the colony is mature, and in Fleming's case, the growth at low temperature also favoured the fungus. If the bacteria had been grown at the normal temperature of 37°C nothing unusual would have happened.

The ability to grow very small organisms in the laboratory has been of inestimable benefit to the advancement of science. Not only can millions be grown in a small area, but hundreds of generations can be studied in a short time. This allows mutations to be detected and the organisms carrying them to be isolated. The study of genetics relies on having visible mutants to use in breeding, and much of our understanding of biochemistry has come from growing mutant cells that lack one enzyme of a metabolic pathway.

In the case of multicellular organisms, individual cells can be removed and grown to study their properties. The most famous strain of human cells that have been cultured and used for research are called HeLa cells. These cells are from a tumour that killed a woman in 1951. (See textbox)

Treatment of cancers may include chemotherapy, which involves the administration of specific drugs that inhibit the cell cycle of rapidly dividing cancer cells. However, other cells that divide rapidly, such as blood cells and hair follicles are also affected.

HeLa CELLS

In 1951 a young American woman called Henrietta Lacks was found to have cancer of the cervix. A sample of the tumour was taken to be examined by pathologists, and they found the cells to be most unusual. Up until then, human cells had proved to be very difficult to grow as a culture, but these cells from Henrietta Lacks grew vigorously and went through each cell cycle without restraint, a feature of cancer cells.

They became the first human cells to be successfully cultured, and are now referred to as HeLa cells. Late in the 1950s cultures of HeLa cells were used to develop the vaccine against polio, and since then the cells have been used as the host cells in genetic engineering to produce a variety of proteins. The HeLa cell strain is now found in biological laboratories throughout the world and continues to be the work-horse for many human tissue experiments. An interesting ethical issue is that, apparently, neither Henrietta nor her family gave permission for her cells to be used in this way.



HENRIETTA LACKS



tinyurl.com/bddmfv36



tinyurl.com/yd4rdnph



Fig. 14.7 Robert Koch

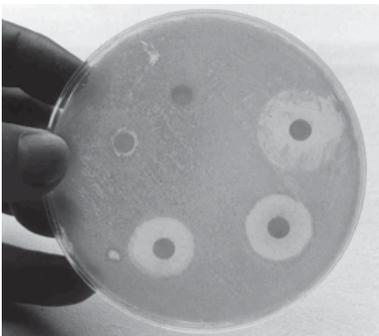


Fig. 14.8 Agar plate with bacteria showing zones of inhibition



Fig. 14.9 Blood agar with *Micrococcus luteus* bacteria

Humans have been unknowingly culturing cells for thousands of years. People brewing beer or making wine or bread have used cultures of yeast cells to break down sugars to alcohol and carbon dioxide. Likewise, the production of cheeses and yoghurt makes use of bacterial cultures. The bacteria and yeast cells were carefully grown in the correct conditions, some of the culture was used for the fermentation process, and the rest was used to begin the next culture. When it was suggested in 1835 that alcoholic fermentation was, in fact, a biological process carried out by yeasts, members of the scientific community rejected the idea. Fermentation was considered to be nothing more than a chemical reaction, and the chemists of that time argued that yeasts played no part in the process. Some even went so far as to suggest that yeasts were not living organisms, but merely substances that accumulated during fermentation.

It was not until 1860 when Louis Pasteur successfully demonstrated that yeast cells were the agents of fermentation, that most scientists were convinced.

Deliberate scientific cell culture has had a relatively short history. The early growth media were crude liquids or broths, such as blood, serum and various other concoctions of biological fluids.

The discovery that cells grow well on the surface of semi-solid media resulted in a considerable advance in convenience, and the development went from vitreous humour of the eye to gelatine, to the inert agar medium of today. 'Inert' means that the solidifying agent is not metabolised by the growing cells. Agar, a product of seaweed, is a carbohydrate polymer which was first used by a famous microbiologist Robert Koch. (See Fig. 14.7) He identified the microorganisms responsible for many severe diseases, including anthrax and tuberculosis using agar plate techniques. Agar is the common base for media that then have different ingredients added to make thousands of different formulations for growing specific microorganisms. (See Figs. 14.8, 14.9)

To initiate a culture of yeast or bacterial cells, a suitable inorganic medium must be found. As mentioned before, this medium may be either liquid or semi-solid, but it must contain all of the nutritional requirements of the cells. The medium is then 'seeded' with a pure strain of the yeast or bacterium and kept at a temperature that is suitable for the growth and division of the cells.

The culturing of bacterial cells ranges from the small-scale production on Petri dishes, as used by Alexander Fleming (See Fig. 14.10) and modern research laboratories, to recently established continuous-flow cultures of genetically-altered bacteria to produce valuable products such as insulin on an industrial scale.

An important use of cultured bacterial cells is the Ames test which is used to detect whether chemicals have mutagenic effects. A more recently developed test uses genetically engineered yeast that glows when it encounters mutagens. The yeast culture contains a protein coded for by a gene from a jellyfish. This protein emits light when the DNA has been damaged by a mutagen.

Animal Cell Culture

Animal cell cultures are also widely used in medicine to provide skin tissue and reserve cells for other organs. There are exciting prospects of growing new brain cells from undifferentiated stem cells. Animal cell cultures are now used instead of live animals for testing cosmetics and drugs, and in other scientific research. Unlike bacteria and plant cells, animal cells are unable to synthesise most of their requirements from simple compounds in the culture medium. All vitamins and other organic compounds must be supplied, and the osmotic balance, pH, and suitable temperature of the medium must be maintained. The medium must be well aerated unless the cells are growing as a monolayer.

The terms **cell culture** and **tissue culture** are almost the same, because dividing cells tend to produce clumps or layers of the same kind of cell, and that is a tissue. A major difference between the culturing of animal cells and plant cells is that plant cells are more readily able to divide than animal cells after they have differentiated; that is, when they are at stage G_0 in the cell cycle.

Plant Cell Culture

Tissue culture in plants requires firstly that a small group of cells be cut from the donor and washed with alcohol or bleaching agent to remove any contaminating microbes, such as bacteria or fungi. These washed cells are then placed in a solution containing minerals, plant growth hormones, and a supply of an energy-rich chemical such as glucose. Depending on the concentration and types of hormones used, the tissue can be made to divide or to differentiate into roots or shoots or both. This technique is widely used to produce hundreds of clones of a valuable plant such as a new orchid variety. An extension of the technique is to produce hybrids by first dissolving the cell walls, then persuading the cells of two species to fuse. (See Fig. 14.11)

In both animal and plant culture techniques it is most important to maintain sterile conditions in order to prevent opportunistic microorganisms from taking over the medium. Special apparatus such as laminar flow cabinets are used so that scientists can work on the cultures in a sterile environment, and antiseptics that kill undesirable cells are applied.



Fig. 14.10 Alexander Fleming in his laboratory

SPRAY ON SKIN

Australian scientist, Marie Stoner, and plastic surgeon, Fiona Wood, revolutionised the treatment of burns victims by developing 'spray on skin' using cell culturing techniques.



a



b

Fig. 14.11 Transferring plant cells into culture medium in a laminar flow cabinet

Study Questions

- What is the cell cycle? Illustrate your answer with a labelled diagram.
 - Describe the events that occur during interphase?
- The cell cycle has three main check points. At what stages do these occur and what is the role of each checkpoint?
- There are 3 main checkpoints in the cell cycle and some sort of signal is needed at each one before the next step proceeds. Fill in the following table stating the name(s) of the signals and how each signal is used by the cell.

	Name of signal(s)	Response to signal
Check point 1		
Check point 2		
Check point 3		

- Describe the sequence of events where cyclin and CdK control the cell cycle.
- The growth of new cells at a wound site in humans is regulated by physical and chemical factors. Describe an example of each of these.
- 'Hormones regulate cell division'. Explain the meaning of this sentence by referring to specific hormones from plants and animals.
- Define the terms carcinogenesis and mutagenesis.
 - What are the three main causes of mutagenesis?
- Cancer is a disease of the cell cycle. Explain what is meant by this statement.
- Many people who die from lung cancer have had the disease for many years. Explain this observation.
- 'Humans culture cells for a variety of purposes.'
 - Make a list of the uses, old and new, of culturing yeast, fungi and bacterial cells.
 - What other scientific uses are there for culturing cells?
- In terms of experimental design, why is it important that:
 - the cell culture medium be 'inert', and
 - the conditions of the culture are sterile?
- List some of the nutritional requirements you would expect to be added to a yeast or bacterial cell growth medium.
 - What other factors need to be kept stable in a growth medium?
- Although the culturing of HeLa cells have been used extensively in valuable scientific and medical experiments, it has also created ethical concerns? Explain the concerns.
- Animal and plant cell cultures are now used extensively.
 - What special provisions do animal cells need in their growth medium? Why are these provisions needed?
 - What special provisions do plant cells need in their growth medium? Why are these provisions needed?
 - State one advantage and one disadvantage of producing genetically identical plants by cell tissue culture.



TOPIC 3

Homeostasis

- 15 Organisms Have Tolerance Limits
- 16 Homeostasis
- 17 The Nervous System
- 18 The Endocrine System
- 19 Homeostatic Control Mechanisms

Organisms can only survive within a range of environmental conditions, such as pH, temperature, oxygen level, and water. Different organisms have different requirements, and so the ranges of suitable conditions will vary. Organisms will not survive if the conditions are outside of their tolerance limits.

Many organisms are able to survive in a wide range of conditions by maintaining a relatively stable internal environment – this is called homeostasis. The mechanisms that organisms use to maintain stability are called homeostatic control mechanisms.

In the human body this involves the nervous system and the endocrine system.

Science as a Human Endeavour

Throughout this topic examples that illustrate key concepts of science as a human endeavour are indicated by the symbol ▼. There are examples of communication and collaboration, development of scientific models and new technologies, influence on and by other areas of study and society, and applications and limitations of biological knowledge.

Organisms Have Tolerance Limits

15

Organisms survive most effectively within their tolerance limits. Factors for which organisms have tolerance limits include:

- › **body temperature**
- › **water availability**
- › **blood glucose level**
- › **carbon dioxide concentration in the blood and tissues.**

All organisms require their environmental conditions to be within a particular range in order to survive. This applies to the simplest organisms, such as bacteria, more complex eukaryotic organisms such as a Euglena or an Amoeba, and multicellular plants and animals. This particular range of environmental conditions define the organism's tolerance limits.

If the environmental conditions fall outside of the organism's tolerance limits, then it will either not survive, or it must have mechanisms to maintain its own internal environment.

Unicellular organisms are surrounded by their external environment and have limited ability to maintain their internal composition. However, some bacteria can form 'spores' in response to lack of water and may survive in this form for many years until water becomes available.

The cells of a multicellular organism are surrounded by **tissue fluid** - this is known as the **internal environment**. The composition of this fluid remains remarkably constant and enables cells to continue to function normally. If the fluid surrounding the cells has a constant composition it is easier for the cells to maintain the conditions needed for their survival. Multicellular plants maintain their solute and water balance through active transport of solutes in the roots, and by controlling water loss through the leaves by closing the pores of their stomata. (See Fig. 15.1)

Examples in humans involve maintaining stable blood glucose level, blood carbon dioxide and oxygen levels, water and solute balance, and body temperature. To achieve this stability, all of the organs of a system and all of the body systems need to work together and this requires communication between them. In humans, this communication may be by either **nerves** or **hormones**. (See Fig. 15.2 and Fig. 15.3)

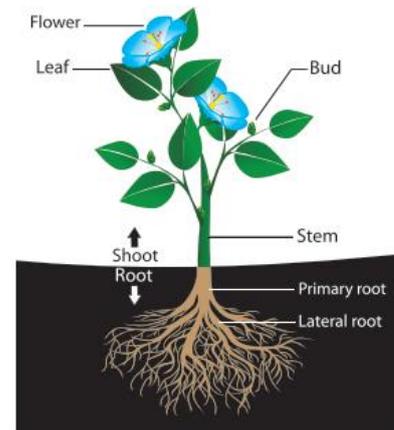


Fig. 15.1 A typical plant structure



Fig. 15.2 Human nervous system

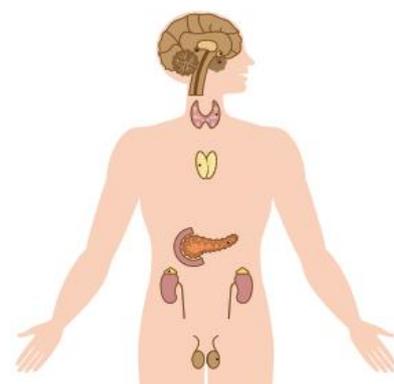


Fig. 15.3 Human endocrine system

Tolerance limits are the range of conditions required for survival of an organism.

GOYDER'S LINE

An example of a large scale boundary that designates a change in environmental conditions (tolerance limits) is the famous Goyder's line for rainfall in northern South Australia. South of the line the average rainfall is above 250mm per year, while north of the line it is below 250mm. At Goyder's line there is a transition of vegetation from mallee woodland in the south to arid scrublands of saltbush and bluebush in the north as the mallee woodland plants' tolerance limit for water availability has been reached.

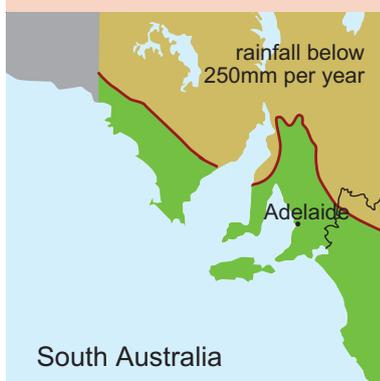


Fig. 15.4 Goyder's line

There are continuous changes in the external conditions but the organism only needs to respond to those that are possibly going to affect it. Later in this topic we will concentrate on the roles played by the nervous system and the endocrine system in humans to maintain a relatively constant internal environment – this is called **homeostasis**.

Selective responses

If the external temperature increases or decreases, this may have an effect on the activity of cells and so it is important to detect and respond to these changes. A sudden loud sound may also stimulate a response, such as rapid muscle movement, but gradual changes in sound may not be perceived as a danger and so no response is elicited. The organism is therefore selective in its response to these changes.

Some changes in the types of chemical in the air are selectively detected. A male moth may detect and respond to minute quantities of a chemical released into the air by a female moth, but humans are unable to detect this chemical even when the concentration is increased a million times. The chemical produced by the female moth has no relevance to us, we have no sensory receptors for it. However, we do respond to the detection of aromas from certain foods by salivating.

There are impacts on an organism when conditions fall outside its tolerance limits.

If you stand at a lookout point on the eastern side of the Gammon Ranges near Arkaroola in South Australia and look to the East, you can see a largely desolate landscape lined with ribbons of trees meandering in the heat haze towards Lake Frome. Why don't the trees cover the whole landscape? The trees seem to only grow on creek lines where water is draining out from the mountains onto the plains. They can compete with other vegetation only where water is more abundant. Although the trees may disperse their seeds over a wide area, only those seeds that fall near the creek will develop into mature trees. (See Fig. 15.5)



Fig. 15.5 Ribbons of trees

The amount of water is an abiotic factor that determines the distribution of plants in this community. An abiotic factor is one which is non-living. There are many other abiotic factors that affect the distribution of species within a community.

These include:

- › minerals composition
- › oxygen level
- › light availability
- › temperature
- › wind
- › pH of the environment.

Mineral composition

When wheat and barley were first planted in the sandy soils of the south east of South Australia, they grew well, but in the following years the crops became poorer and poorer. The cause of the poor performance of the crops was not a change in the rainfall, but a deficiency of phosphorus, copper and zinc. Before it was cleared by farmers, the natural bush grew well because the minerals from the soil were being recycled. However, there were insufficient minerals to support the type of exploitation that sends away produce like wheat, barley and oats to distant markets with the minerals locked inside. When farmers supplied these minerals to the soil the following remarkable improvements were obtained:

Fertiliser	Subterranean Clover Yield (kg per ha)	Lucerne Yield (kg per ha)
Untreated	187	62
Superphosphate	625	937
Super + Copper	875	2000
Super + Copper + Zinc	3250	2000

This discovery meant that previously unusable land could now be farmed. The decrease in crop yield indicated that minerals were the limiting factor and the crops were reaching their tolerance limits for the availability of this resource.

In a community, the amount of productivity achieved will often depend on a limiting factor, such as the availability of a resource. Experiments in the Sargasso Sea, the marine equivalent of a 'desert', have shown that productivity there could be greatly increased by fertilising the water with iron. In this case iron is the resource that is in short supply and limits the productivity of the community. (See textbox 'Leibig's Law')

LEIBIG'S LAW

In agriculture, the idea that the environmental factor that is in least supply will limit productivity is called Leibig's Law of the Minimum. For example, lack of light or an essential mineral will restrict the growth of a plant, even if all other requirements are at ideal levels.

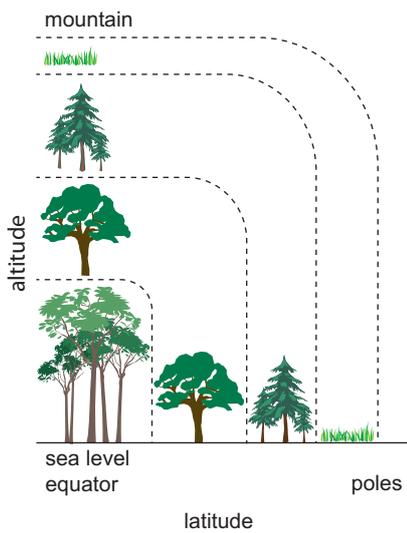


Fig. 15.6 Effect of altitude and latitude on productivity

THE SUN IS A SOURCE OF HEAT ENERGY

As well as using sunlight for photosynthesis or using the products of photosynthesis, most organisms rely on heat energy from the Sun.

Animals such as reptiles use solar energy to maintain a suitable body temperature, and plants rely on heat energy from the Sun to reach a temperature that is suitable for their metabolic processes. For all organisms the temperature of the Earth's atmosphere is critical for survival. The temperature of the atmosphere at or near sea level depends on heat energy that is radiated from the Sun. (See Fig.15.7)

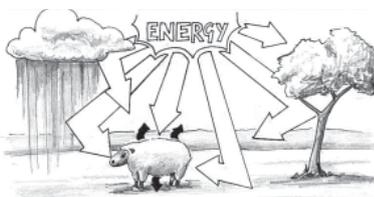


Fig. 15.7 Solar energy

Oxygen level

Lakes and rivers are quite often limited in productivity due to a lack of resources such as nitrogen and phosphorus, and problems arise when these limiting factors are overcome as a result of human activities. This has occurred on occasions in the River Murray system when toxic algal blooms have resulted from the unintentional influx of fertilisers from neighbouring properties. A body of water with a high nutrient level such as this is called **eutrophic**. In this case productivity is high, as producers are able to grow unchecked. As a result of the increase in the number of organisms there is a depletion of oxygen, particularly at deeper levels, because oxygen dissolves poorly in water. Many organisms then die due to the lack of oxygen.

Light availability

The parts of the Earth's land mass with the greatest productivity are the tropical areas near the equator, as they receive a high level of light energy all year round. On the other hand, in the polar regions there is much less energy input and productivity is very low. (See Fig. 15.6)

Life in seas and oceans is found almost exclusively concentrated in the upper few tens of metres. The reason for this is that light, the community's energy source, is rapidly absorbed by water, and the deeper reaches of the ocean are in total darkness and almost devoid of life. The producers, which are the foundation of just about every community, can only survive and photosynthesise if they have ways of staying in the top few well-lit metres.

Temperature

In terrestrial communities, temperature is also important. Mountains near the equator, such as Mount Kenya and Mount Kilimanjaro, have permanent patches of snow. If light availability was the sole factor determining the rate of photosynthesis these mountains would be the most productive habitat of all. While the plants living on them show many interesting adaptations to the cold conditions, they do not have high productivity, due to the constant low temperature.

Study Questions

1. What is meant by a tolerance limit?
2. Give two examples of the tolerance limits for:
 - (a) humans
 - (b) plants.
3. Define the terms abiotic factor and biotic factor and give two examples of each.
4. Abiotic factors determine the nature of a community that occupies a habitat.
Give examples of such abiotic factors that are the tolerance limits for the following and explain your choice of answer:
 - (a) plants on high mountains
 - (b) animals in the Australian Simpson Desert
 - (c) algae in deep water
 - (d) fish in a eutrophic lake.
5. The productivity of a community is often limited by mineral resources. Describe how productivity could be increased in these situations.

16

Homeostasis

INTERNAL ENVIRONMENT

The cells of a multi-cellular animal are surrounded by tissue fluid. This is the internal environment.

Organisms detect and respond to changes in the internal and external environment.



Explain the stimulus-response model.

Stimulus and response

To understand the mechanisms involved in detecting and responding to changes in the environment we need to clarify the terms stimulus and response. A **stimulus** can be thought of as any change in the internal or external environment that can be detected. Examples include light, sound, touch, temperature, and the concentration of certain chemicals. The next step is detection, and this is achieved by a **receptor** which then transmits a **message**. In living systems the receptor is a specialised cell or group of cells and the message that the receptor sends is in the form of a nerve impulse or a hormone. The message is received by an **effector**, which brings about a **response**.

A stimulus-response mechanism, therefore, has five elements – a stimulus, a receptor, a message, an effector, and a response. (See Fig. 16.1)

STIMULUS RESPONSE AND REFLEX



tinyurl.com/ybxc2p2b

THE SENSITIVE PLANT



tinyurl.com/yxcfdtpr

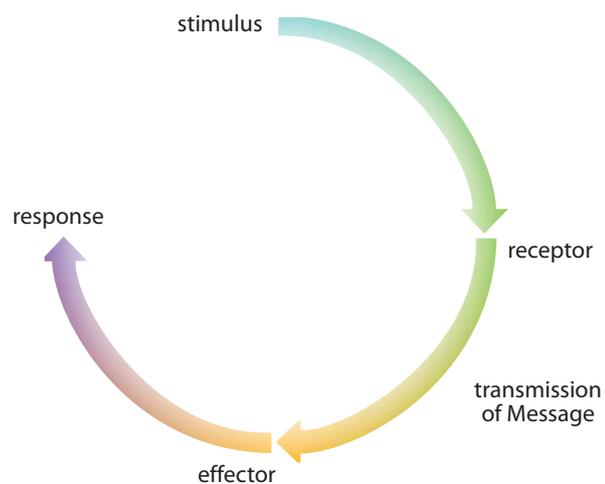


Fig. 16.1 Stimulus response model



Describe the role of sensory receptors.

Sensory receptors

Sensory receptors detect stimuli such as light, chemicals, sound, temperature and touch.

Humans have five main types of receptor that result in five senses: sight, smell, taste, hearing and touch.

The sensory receptors for seeing, tasting, smelling and hearing are located in specialised sense organs in the head. The sensory receptors for somatic sensation such as for temperature and touch, are distributed over the skin and muscles.

The receptors for each of the senses respond to the intensity, the duration, and the location of the stimulus. Once the receptors have been stimulated they send a nerve impulse to the central nervous system. There the signal is processed and interpreted. The central nervous system sends a nerve impulse along nerve cells called motor neurons which signals the effectors, such as muscles cells or glands, to respond.

If we did not have **sensory receptors**, then we would not be able to detect changes in the external or internal conditions. If we were unable to detect such changes, then we would not respond to them. This would lead to changes in our internal environment and would affect cell function, resulting in the death of tissues, organs, and the entire body.



Describe the role of effectors.

In animals an **effector** is a muscle or a gland that carries out a response, such as movement or secretion, as a result of a stimulus. For example, in humans, skeletal muscles move (shivering) in response to a decrease in body temperature, and sweat glands secrete sweat in response to an increase in body temperature.

Regulating the internal environment

The conditions of an organism's internal environment are critical to the normal functioning of its cells. Human cells need a relatively constant temperature, pH, oxygen concentration and osmotic balance in order to survive. A build-up of waste products such as carbon dioxide and urea will impair normal cell function, so these need to be removed. Organisms therefore need control mechanisms to regulate their internal environment and sensory receptors are the initiators of the control mechanisms.

RECEPTORS AND THE SENSES

Some biologists classify sensory receptors as photoreceptors, mechanoreceptors, chemoreceptors, thermoreceptors, proprioceptors, and pain receptors. Sight relies on photoreceptors, smell and taste are detected by chemoreceptors, while hearing and touch are the domain of mechanoreceptors. Temperature is monitored by thermoreceptors and proprioceptors enable you to monitor the position of your limbs and body. The role of pain receptors needs no explanation, although pain is, perhaps, difficult to define.

LIFE WITHOUT SENSORY RECEPTORS

Sensory receptors communicate information about our surroundings to our brain so it can make decisions. While you would be aware of the everyday difficulties and dangers for people who are sight or hearing impaired, there are also problems for those who lack the sense of taste or smell or touch. We use our taste and smell to detect food quality and avoid poisoning. Without heat or pain receptors we are in danger of not avoiding dangerous situations, like sharp or hot objects. Touch receptors enable us to detect objects and carry out fine manipulation. Have you ever tried to tie your shoe laces when your fingers are numb with cold?

Homeostasis is the maintenance of a relatively constant internal environment. This ensures the optimum conditions for the body to function.

Homeostatic control mechanisms are *self-regulating* ones that involve negative feedback. A simple analogy is provided by the kitchen oven. If a temperature is preset, the element (or flame) will come on until the desired temperature is reached. A further increase in temperature will cause the oven to switch off. As the oven cools the drop in temperature will cause the oven to come back on. In this way the oven temperature is kept reasonably steady because it *fluctuates* around the preset temperature. Notice that the temperature is self-regulating in that the response is a *change in temperature* which then acts as the next stimulus. (See Fig. 16.2)

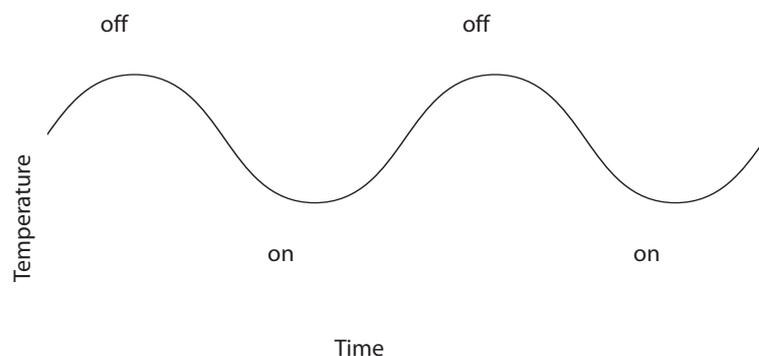


Fig. 16.2 Temperature regulation in an oven

Homeostasis involves a stimulus - response and negative feedback model.



Recognise that in negative feedback the response inhibits the initial stimulus.

Earlier we described a **stimulus-response model**. If the response were to somehow influence the stimulus, then we would have a situation called **feedback**. If the response *inhibits* the stimulus that caused it, this is *negative* feedback and this will result in control. (See Fig. 16.3)

As the stimulus increases, the response increases, and this then inhibits the stimulus. As a result, the stimulus fluctuates about a pre-set level. The maintenance of a relatively stable internal environment is called **homeostasis**, and the mechanisms that achieve this are called **homeostatic control mechanisms**. Examples of homeostatic control mechanisms are discussed in Chapter 19.

POSITIVE FEEDBACK

A response that increases the stimulus that caused it (positive feedback) will result in a spiraling effect that could get out of control. For example, if the body temperature increases above a certain level, the metabolic rate will increase, resulting in a further increase in body temperature.

This positive feedback causes an unchecked increase in temperature and leads to the death of the person. Such a situation can arise with some types of fever.

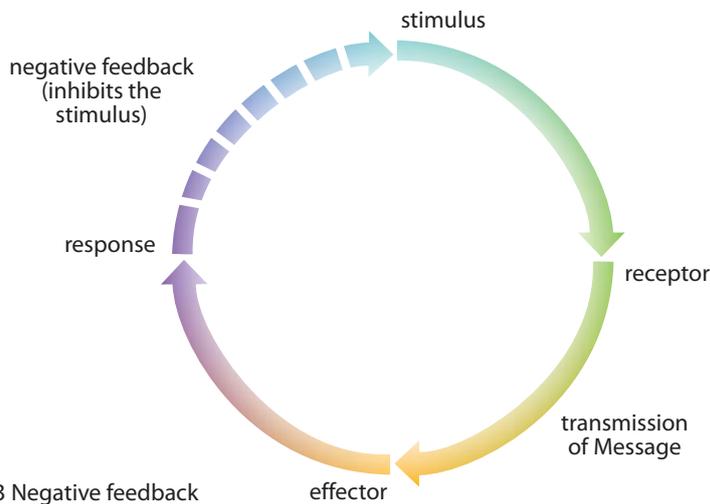


Fig. 16.3 Negative feedback

In human beings, homeostasis depends on the functioning of the nervous and endocrine systems.

The nervous system and hormones of the endocrine system both have an important role to play in the overall regulation and control. Some control mechanisms rely exclusively on only one of these communication mechanisms, while others use a combination of both. It is interesting to compare the two methods of communication.

The next two chapters address details of the nervous and endocrine systems.

1. Many homes and schools maintain a constant room temperature using an air-conditioner, which has a pre-set temperature on a control panel. If the temperature of a room rises above the pre-set level, cool air is pumped into the room. Using the above information and the stimulus-response model, state the stimulus, receptor, message, effector and response for an air-conditioned room.
2. (a) List the main types of receptors, and their role, and the five senses.
(b) Why does it make sense to list the receptors first, then the senses?
3. (a) Define the terms homeostasis and homeostatic control mechanism.
(b) What is the role of effectors in the stimulus-response model?
(c) What is the difference between negative feedback and positive feedback?
(d) Give one example of positive feedback and one example of negative feedback.

Study Questions

17

The Nervous System

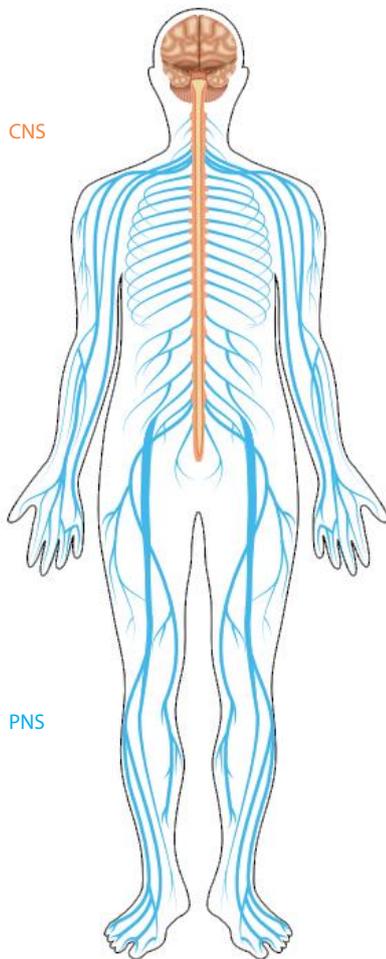


Fig. 17.1 Human nervous system

In the nervous system electrochemical pulses called **nerve impulses** travel along nerve cells, called **neurons**, from one part of the body to another specific location. For example, nerve messages travel along a specific nerve pathway from the respiratory centre in the brain to designated muscles of the chest and diaphragm to regulate breathing.

The nervous system is composed of the central nervous system and the peripheral nervous system.

The role of the nervous system is to detect stimuli, process information, and elicit a response. For convenience, the human nervous system can be divided into the central nervous system and the peripheral nervous system.

Central Nervous System (CNS)

The central nervous system consists of the brain and spinal cord. (See Fig. 17.1) Its role is to detect internal changes in the brain, receive stimuli from peripheral nerves, process information, and send nerve impulses to relevant tissues and organs to bring about a response.

Peripheral Nervous System (PNS)

The peripheral nervous system consists of nerves that lie outside the brain and spinal cord. Its role is to connect the central nervous system to all parts of the body. Peripheral nerves that originate in the brain are called cranial nerves, and those that are connected to the spinal cord are called spinal nerves. An exception is the optic nerves that are cranial, but are considered to be part of the CNS.

The peripheral nervous system contains 'voluntary' and 'involuntary' nerves. Voluntary nerves control the skeletal muscles, and make up the somatic nervous system (SNS). The autonomic nervous system (ANS) contains involuntary nerves. As their name implies, they work automatically to control things you don't have to think about, such as gut movement, heart rate, and breathing. The ANS is divided into the sympathetic and parasympathetic nervous systems. The sympathetic nervous system plays a role in the 'fight or flight' response, as we shall see in Chapter 18.

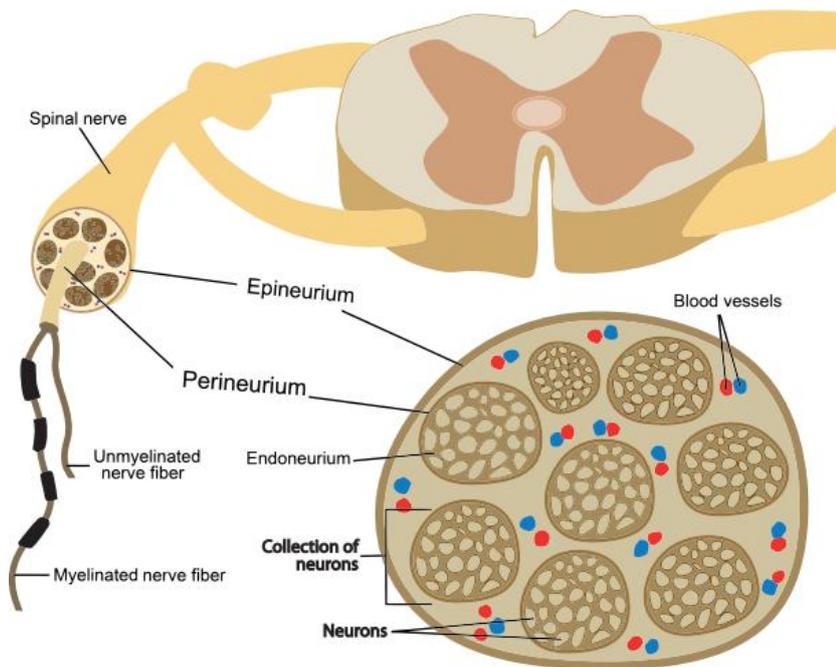


Fig. 17.2 Peripheral (spinal) nerve cross-section

A nerve is actually a 'bundle' of neurons (nerve cells), whose axons run parallel to one another. (See Fig. 17.2)



Compare the structure and function of sensory neurons, interneurons, and motor neurons.

The cells that make up the nervous system are of two main types – **neurons** and **glial cells**. Glial cells, such as Schwann cells, provide structural and metabolic support to neurons. It is the neurons that transmit impulses.

All neurons have the following structures – a cell body, an axon, and dendrites. (See Fig. 17.3)

Nerve impulses are transmitted along a **nerve pathway**, that

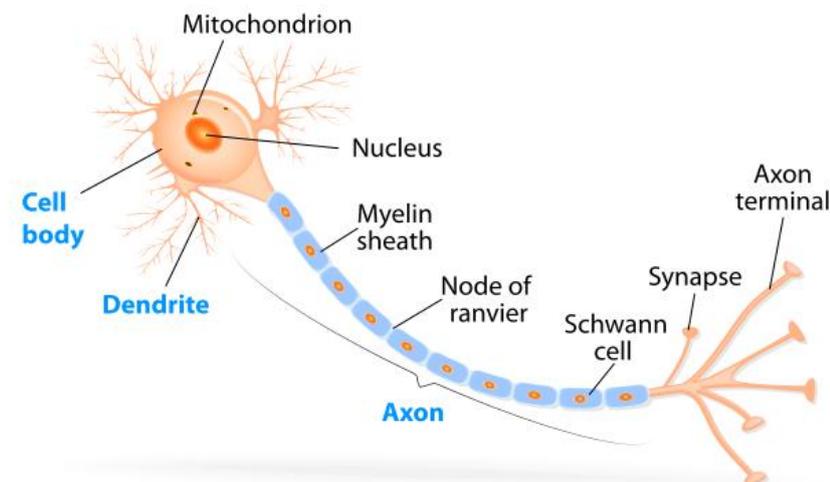


Fig. 17.3 Nerve cell (neuron)

MIXED NERVES

Spinal nerves (part of the peripheral nervous system) are called mixed nerves, as they contain both sensory neurons and motor neurons, and can carry nerve impulses to and from the CNS.

NERVOUS SYSTEM AND REFLEX ARC



tinyurl.com/245y4hkv

NEURON STRUCTURE



tinyurl.com/ybgjya5y

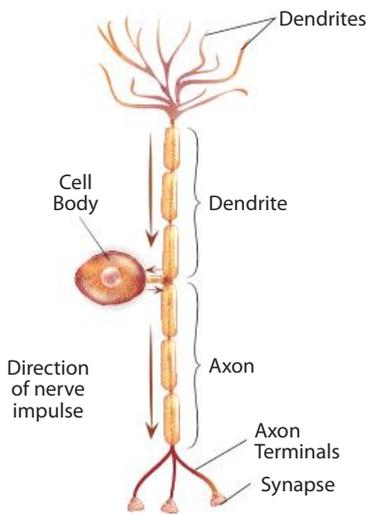


Fig. 17.4 Unipolar neuron (sensory neuron)

consists of a series of neurons with tiny gaps between them called **synapses**. The nerve impulse travels along the axon of the neuron as an electric current, but this current cannot cross the synapse. At the synapse a chemical, called a **neurotransmitter**, crosses the gap and transfers the impulse to the next neuron in the pathway. Note that a nerve impulse can only travel in one direction along a neuron, from the dendrite 'end' towards the axon terminal.

Neurons are categorised into three main types – sensory neurons, interneurons, and motor neurons.

Sensory neurons

Sensory neurons detect stimuli and carry a nerve impulse towards interneurons in the central nervous system. Stimuli, such as touch, light, and chemicals, are detected by specialised nerve endings called **receptors**, and this triggers a nerve impulse that travels along sensory neurons.

Most sensory neurons have a structure that is called **unipolar**. This is because the cell body (soma) has only one 'process' extending from it. The nerve impulse travels from the dendrites along the axon towards the axon terminals. (See Fig. 17.4)

Interneurons

Interneurons are located in the brain and spinal cord. They receive signals from sensory neurons and transmit them to motor neurons. The interneurons in the brain have longer axons, and those in the spinal cord have shorter axons.

Interneurons are **multipolar**, as they have many processes extending from the cell body. As with sensory neurons, the nerve impulse travels from dendrites along the axon to the axon terminals. (See Fig. 17.5)

Motor neurons

Motor neurons carry nerve impulses from the CNS to effectors, such as muscles and glands. This results in a response, usually movement or secretion.

Like interneurons, motor neurons are **multipolar**, as they have many processes extending from the cell body. The nerve impulse travels from dendrites along the axon to the axon terminals.

Generally, peripheral nerves carry nerve impulses to or from the central nervous system. An interesting exception, that does not necessarily involve the CNS, is the enteric nervous system – see textbox.

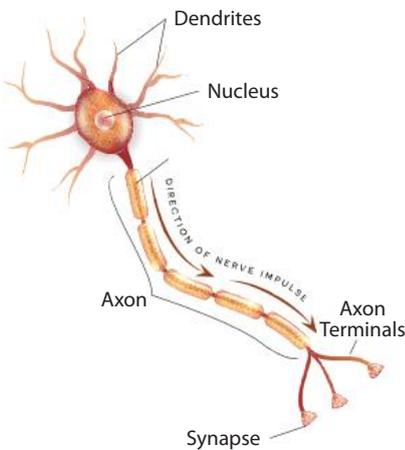


Fig. 17.5 Multipolar neuron (interneuron and motor neuron)

ENTERIC NERVOUS SYSTEM

The enteric nervous system of the gut, which can operate independently of the CNS, has its own sensory neurons, interneurons, and motor neurons. It is sometimes referred to as the body's 'second brain'.



Describe the structure of a nerve pathway from receptor to effector.

A **receptor** detects a **stimulus** and this triggers a **nerve impulse** which travels along a **sensory neuron** towards the **spinal cord** or brain in the CNS. The impulse is transmitted along nerve fibres in the spinal cord to the brain. The information is processed by the brain, which sends a nerve impulse down the spinal cord along a **motor neuron**. The motor neuron carries a nerve impulse to an **effector**, either a muscle or gland, and this results in a **response**. It is interesting to note that nerve fibres (axons) in the human body may be more than a metre long! (See Fig. 17.6)

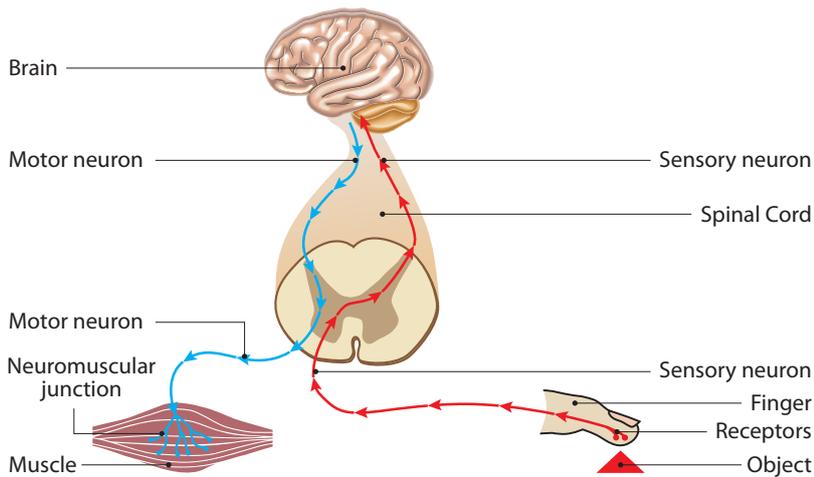


Fig. 17.6 Impulse along the spinal cord

An important exception to the nerve pathway described above is a reflex response that does not involve the brain. Details of the pathway of a reflex response are included later in this chapter.



Describe the role of synapses and neurotransmitters.

As we have seen, a nerve pathway is made up of a series of neurons that transmit an impulse from one neuron to the next. There is a junction, called a **synapse**, with a gap - called a **synaptic cleft** - between the axon of one neuron and the dendrite of the next. Transmission across the gap is by a chemical called a **neurotransmitter** that is secreted from the axon terminal of the incoming neuron. (See Fig. 17.7)

There are many different neurotransmitters, the main one being **acetylcholine**. Others include dopamine, serotonin, noradrenaline, and adrenaline. These are called *excitatory* neurotransmitters because they stimulate the next neuron in the pathway. Other neurotransmitters, such as GABA, and even the amino acid glycine, are called *inhibitory* transmitters because they block the nerve impulse. In either case neurotransmitters act by combining with receptor molecules on the membrane of the receiving cell. As we know, this involves the neurotransmitter and receptor molecules having complementary shapes.

Some drugs act by mimicking neurotransmitters at synapses.

NEUROTOXINS

Many snake venoms act as neurotoxins and cause paralysis. The production of snake antivenom by traditional means is time consuming and expensive. It involves 'milking' venom from dangerous snakes and the yields are usually very low. New technologies using recombinant DNA or nanoparticles promise more efficient methods of production. To achieve this, funding is needed from science, business, and government.

SYNTHETIC SNAKE ANTIVENOM



tinyurl.com/z7j7v5m

CSIRO SNAKE ANTIVENOM FOR DOGS



tinyurl.com/bdcue8fr

BOTOX® – AN UNEXPECTED CONSEQUENCE



A bacterium called *Clostridium botulinum*, associated with some types of food poisoning, produces a neurotoxin that reduces the release of acetylcholine at the neuromuscular junction, and this results in paralysis.

The bacterium and its toxin were first discovered in the 19th century, and it was not until the 1970s that it was first used to correct 'misalignment' of eyes. Since the 1980s the toxin (renamed 'Botox®') has also been used in cosmetic medicine to relax muscles and remove wrinkles.

Botox® is a registered trademark of Allergan, Inc.

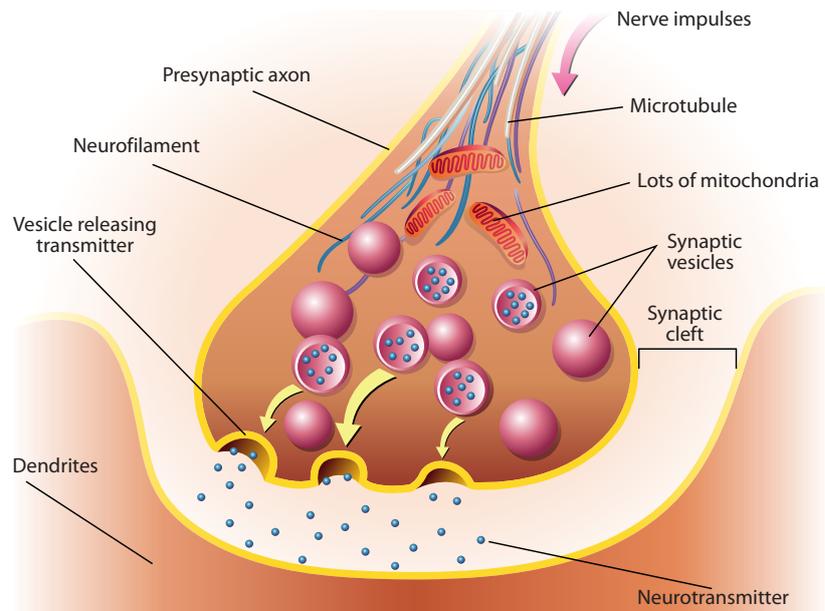


Fig. 17.7 Synapse and synaptic cleft

The site where a nerve reaches a muscle cell is called a **neuromuscular junction**. This junction is very similar to a synapse and transmission across the gap from nerve to muscle involves the neurotransmitter acetylcholine.

If a neurotransmitter, such as acetylcholine, remained in the synaptic cleft it would cause continual stimulation of the next neuron in the pathway or the effector. Following their secretion neurotransmitters are either destroyed by an enzyme, diffuse away quickly, or are absorbed by the cell that secreted them. In the case of acetylcholine, an enzyme called **acetylcholinesterase** quickly destroys the neurotransmitter molecules in the synaptic cleft.

Remember that effectors can also be glands that secrete substances. In this case the neuron secretes a neurotransmitter across the gap to stimulate or inhibit the gland cells.



Describe the role and pathway of reflex responses.

Reflex response

One of the simplest behaviours exhibited by humans is a reflex, which is an automatic response to a stimulus. An example of a reflex response is kicking your lower leg when you are tapped just below the knee – the 'knee-jerk' reflex. Another example is quickly lifting your hand away from a heat source. (See Fig. 17.8)

In both cases the brain is not directly involved as the signal from the receptor travels along a sensory neuron to the spinal cord, then along an interneuron to a motor neuron that signals the muscles – the effectors – to respond. This protects the organism by providing a rapid response to the stimulus. (See Fig. 17.9)



BOTOX®



tinyurl.com/n8v7sfv

FLY SPRAY - A NERVE TOXIN

Neurotransmitters stimulate muscles and other nerves and are then quickly broken down by enzymes, such as acetylcholinesterase, making their signal very short lived. Fly spray contains an inhibitor of acetylcholinesterase, so when a fly comes into contact with fly spray, acetylcholine does not get broken down, the nerve signals to the fly's muscles remain switched on and the muscles are permanently contracted.

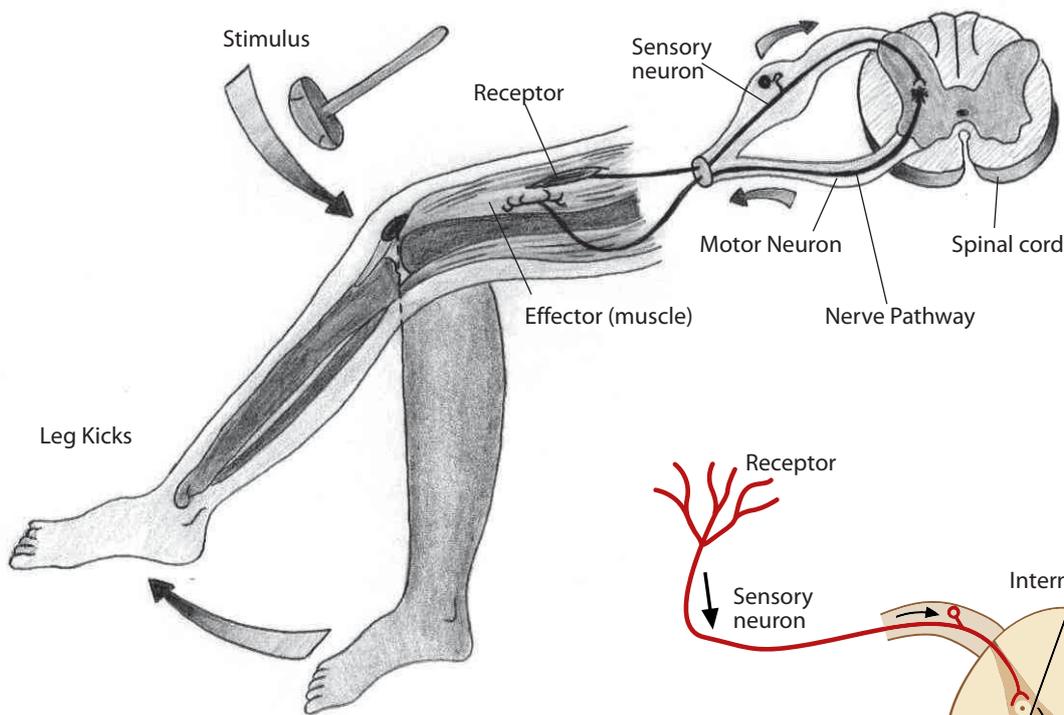


Fig. 17.8 Knee jerk reflex

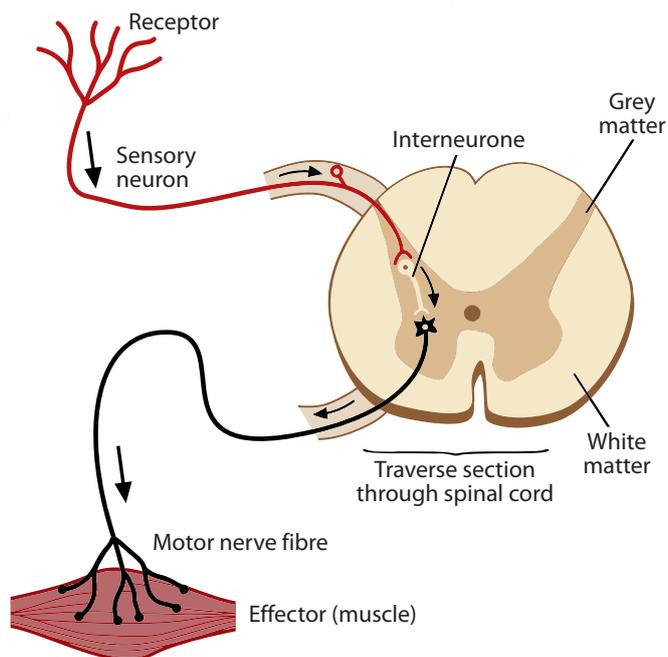


Fig. 17.9 Spinal reflex pathway

Although the brain is not directly involved, these reflexes can be modified by the brain which can increase or decrease the reflex or even totally block it. This is possible because at the same time that the nerve impulse travels from the spinal cord along a motor neuron, an impulse is also sent up the spinal cord to the brain. Other examples of reflexes are:

- the contraction of muscles in the inner ear in response to loud sound. This dampens the movement of the ear ossicles and helps prevent damage to the receptor cells by excessive noise.
- the opening and closing of the iris of the eye in response to changing light levels. The stimulus is the change in light intensity and the effectors are the muscles of the iris. (See Fig. 17.10)
- swallowing, which is a reflex triggered by the stimulation of the touch receptors of the soft palate at the back of the mouth. The response is the contraction of muscles around the top of the gullet. This closes the nasal cavity and covers the opening of the trachea, and begins the process of peristalsis at the top of the oesophagus.

DETECTION AND RESPONSE

A change in light level will cause the receptors in the eye to alter the diameter of the pupil in an attempt to keep the amount of light entering the eye constant.

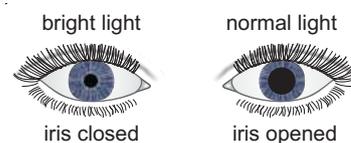


Fig. 17.10 The eye's response to light

Study Questions

1. What is the main role of the nervous system?
2. Define the following terms:
neuron
central nervous system
peripheral nervous system.
3. Name the parts of the body controlled by the voluntary nervous system.
4. Name the parts of the body controlled by the involuntary nervous system.
5. Draw a neuron and label the cell body, the nucleus, dendrites, axon, and axon terminal.
6. Compare the structure and function of sensory neurons, interneurons, and motor neurons.
7. What is the function of a receptor?
8. Name the two structures in humans that are usually effectors and describe their actions.
9. Describe the structure of a nerve pathway from receptor to effector.
10. What is the role of synapses and neurotransmitters?
11. Describe the role and pathway of reflex responses and give an example of a reflex response.
12. The brain is not usually involved in reflex responses.
 - (a) Why is this an advantage to the organism?
 - (b) Describe one reflex response from your own experience. State the stimulus, the receptor, the effector and the response.

The Endocrine System

18

Glands are made of tissue that secretes a substance or substances. Some glands secrete chemicals called hormones into the blood via extracellular fluid. They are called endocrine glands, and are referred to as 'ductless' glands. The hormones that they secrete act as chemical 'messengers'.

The endocrine glands of the body together make up the endocrine system. (See Fig. 18.1) Along with the nervous system, it is involved in coordination and control.

The endocrine system releases hormones that are amino acid derivatives, peptides, proteins, or steroids.

The following table shows the main types of hormone, where they are produced, and their effect. Only the hormones that are discussed here and in Chapter 19 are listed in the table. There are many other hormones, such as growth hormone and reproductive hormones, but their details are not required in this course.

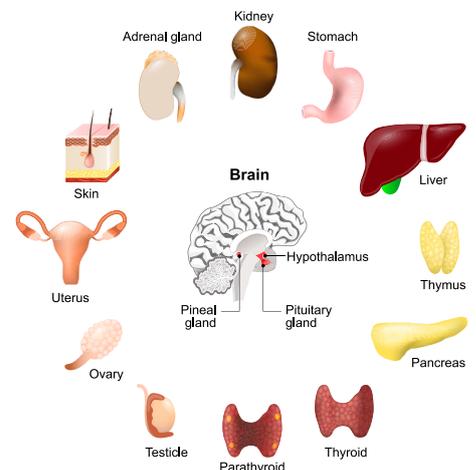


Fig. 18.1 Endocrine system

Type of hormone	Name of hormone	Secreted by	Acts on	Effect
amino acid derivative	adrenaline (also called epinephrine)	adrenal medulla	most cells, mainly muscle	increases blood sugar level, heart rate and blood pressure, blood flow to muscles, breathing rate, pupil dilation
amino acid derivative	noradrenaline (also called norepinephrine)	adrenal medulla	cardiac muscle, smooth muscle	similar effects to adrenaline - also a neurotransmitter in the cardiovascular system
amino acid derivative	thyroxine	thyroid gland	most cells	increases oxidative metabolism
peptide	antidiuretic hormone (ADH) 9 amino acids long	hypothalamus (via posterior pituitary)	renal collecting ducts	increased reabsorption of water by kidneys
polypeptide	glucagon 29 amino acids long	pancreas	most cells, particularly liver	stimulates breakdown of glycogen to glucose, increases blood sugar level
protein	insulin 51 amino acids (in 2 chains)	pancreas	mainly liver and muscle cells	lowers blood sugar level, increases glycogen storage
glycoprotein	thyroid stimulating hormone (TSH)	anterior pituitary	thyroid	stimulates production of thyroxine by thyroid gland
steroid	aldosterone	adrenal cortex	kidneys	increases sodium reabsorption and water reabsorption in kidneys, increases blood pressure

PEPTIDE, POLYPEPTIDE, OR PROTEIN?



Peptides, polypeptides, and proteins are all made up of chains of amino acids. While the numbers are arbitrary, many scientists regard a chain of 9 or fewer amino acids as a peptide, between 10 and 49 as a polypeptide, and 50 amino acids or more as a protein. This is why insulin, made up of 51 amino acids, is often referred to as a small protein.

Hormones travel to target sites via the blood.

Transport of materials

We have seen that for cells to survive they must exchange materials with their surroundings. Our cells are surrounded by tissue fluid that provides their nutrients and removes their waste products. Clearly there needs to be a transport mechanism involved, one that brings the cells' nutritional requirements from the specialised surfaces of the digestive system and the lungs, and removes the cells' waste products. In humans, internal transport is provided by the blood circulatory system, also known as the cardiovascular system. This system comprises the heart, blood vessels, and blood.

ENDOCRINE SYSTEM



tinyurl.com/429bmrjt

BLOOD CAPILLARIES

Blood is pumped from the heart into arteries and then flows from the arteries into smaller branching vessels called arterioles. These, in turn, lead to networks of microscopic vessels called capillaries. (See Fig. 18.2) Capillary walls are only one cell thick and are permeable to certain substances in the blood and tissue fluid. (See Fig. 18.3)

Many substances diffuse between the blood and tissue fluid, either through the cells of the capillary wall, or through pores between these cells and across the basement membrane. (See Fig. 18.4 and Fig. 18.5) In some cases materials may be transported across these cells by endocytosis on one side and exocytosis on the other.

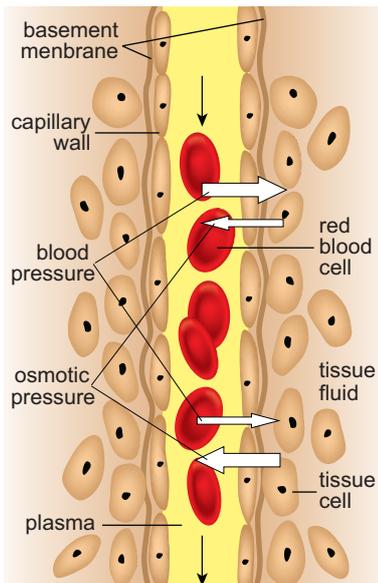


Fig. 18.3 Exchange in capillaries

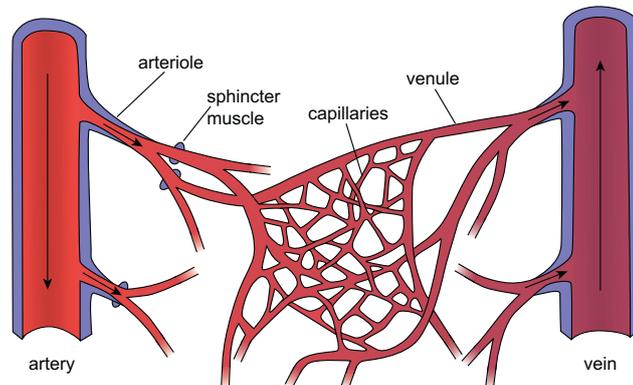


Fig. 18.2 Capillary network – the capillary network is very extensive so that all cells are close to a capillary.

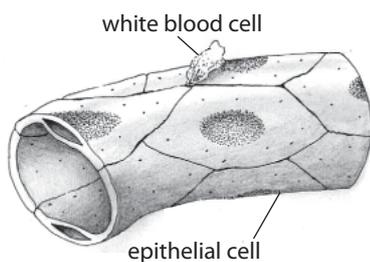


Fig. 18.4 Blood capillary

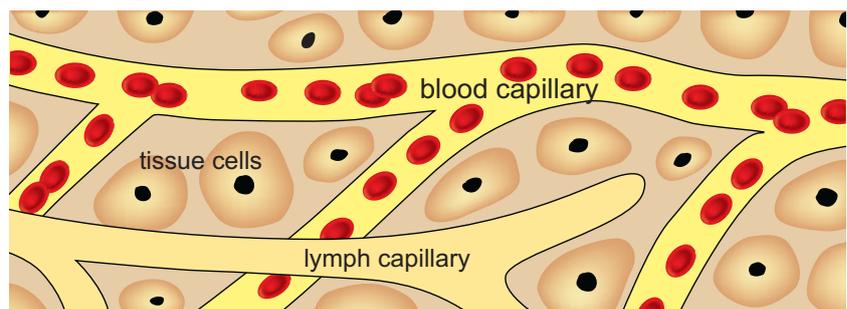


Fig. 18.5 Blood flowing through tissues

Hormones are secreted by endocrine glands into the blood, via the tissue fluid, and travel wherever blood flows throughout the body. A particular hormone will only produce an effect when it reaches **target cells** that are 'tuned in' to it. This involves molecular recognition of the hormone by receptor molecules, usually proteins, that are on the cell membrane or in the cell's cytoplasm. Generally, steroid hormones pass through the cell membrane and bind with the hormone receptor in the cytoplasm, (see Fig. 18.6) whereas other hormones that are water soluble bind to receptor molecules on the outside of the cell membrane. (See Fig. 18.7) As with all molecular recognition, complementary shapes play an important role. The binding of a hormone to its receptor molecule triggers an effect in the target cell.

Hormones can alter the metabolism of target cells, tissues, or organs.

Those cells that have the correct type of receptor for a hormone are called target cells, and they make up target tissues and target organs. For example, the target organ for thyroid stimulating hormone (TSH) secreted by the anterior pituitary, is the thyroid gland in the neck. TSH will not produce an effect anywhere else, even though it will be present in the blood in all parts of the body.

Hormones can activate enzymes in cells or alter gene expression. Their concentration in the blood is extremely low, and due to their long-lasting effects they are involved in long-term regulation such as growth and reproduction.

Hormonal responses can be stimulated by either the nervous system or other hormonal messages.

Examples of the nervous system stimulating hormone secretion include the secretion of insulin by the pancreas and the secretion of adrenaline and noradrenaline by the adrenal medulla. There are also cells in the **hypothalamus** of the brain that secrete hormones into the blood. These are called **neurosecretory** cells.

The secretion of many hormones by endocrine glands is controlled by the secretion of other hormones by the pituitary gland. For this reason, the pituitary has been referred to as the "master gland". In fact, the pituitary gland is itself controlled by the hypothalamus. (See Fig. 18.8) The pituitary gland consists of two distinct regions. The region towards the front is called the **anterior pituitary**, and the region behind it is called the **posterior pituitary**. These two regions of the pituitary play different roles.

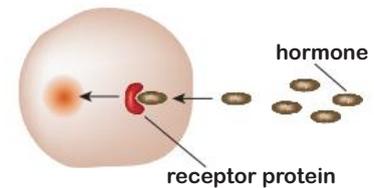


Fig.18.6 Receptor protein in cytoplasm

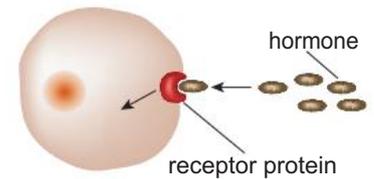


Fig.18.7 Receptor protein in cell membrane

BLOOD

The main functions of blood are transport of materials and defence. Blood consists of a fluid called plasma with different types of cells suspended in it. About 90% of plasma is water, containing many dissolved solutes. These solutes include mineral ions and plasma proteins which play an important part in controlling the osmotic pressure and pH of the blood. Also found in plasma are nutritional substances, waste products, hormones, and respiratory gases. Tissue fluid, the fluid that surrounds body cells, is very similar in composition to blood plasma, but it lacks plasma proteins.

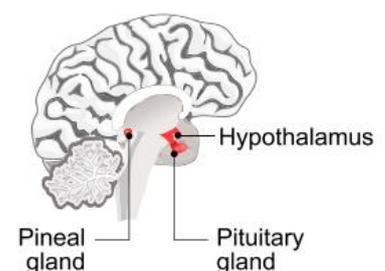


Fig. 18.8 The hypothalamus and pituitary

Other hormones released by the pituitary control sex hormone production by the ovaries and testes, and the production of cortisol by the adrenal cortex.

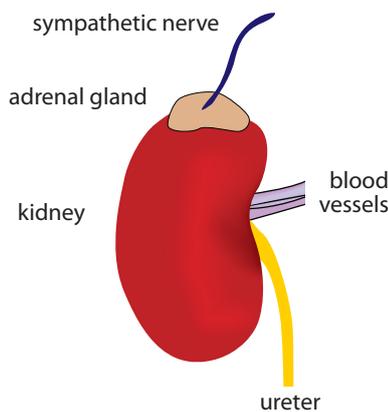


Fig. 18.9 Adrenal gland

DRUGS IN SPORT

The hormone erythropoietin (EPO) has been the subject of intense scrutiny in the sporting world, especially cycling. Some athletes endeavouring to improve their performance in long distance events have had erythropoietin injected into their bloodstream and this causes an increase in the number of red blood cells in circulation. Increasing the number of red blood cells increases the oxygen-carrying capacity of the blood. This practice can be fatal, and its use is banned in sports. The increase in the number of red blood cells causes the blood to be thicker and therefore it is more difficult for the heart to pump. The death of a cyclist in the 1996 Tour de France was attributed to the use of erythropoietin.

One example of the regulatory role of the anterior pituitary is the release of thyroid stimulating hormone (TSH). TSH acts on the thyroid gland, causing it to release thyroxine which signals cells to increase their metabolic rate and this increases body temperature.

The posterior pituitary releases **antidiuretic hormone** (ADH), which is actually produced by the hypothalamus. ADH acts on the walls of the collecting ducts in the kidneys to increase water reabsorption, and thus reduce urine output.

More details about the roles of TSH and ADH are discussed in Ch. 19.



Describe the role of adrenaline in the 'fight or flight' response.

The 'fight' or 'flight' reflex prepares the body to either defend itself -fight- or to avoid danger - flight. If you receive a fright, are stressed, or are threatened by danger, the brain sends a signal via the sympathetic nervous system to the adrenal glands (See Fig 18.9) which secrete adrenaline. This hormone acts on a wide variety of structures including smooth muscle, cardiac muscle, the brain and the pancreas.

Due to stimulation by adrenaline (also called epinephrine), smooth muscle around blood vessels of the skeletal muscles will dilate, increasing blood flow, while the smooth muscle around intestinal blood vessels will constrict, redirecting blood flow to the periphery. The heart rate and cardiac output are increased, raising blood pressure and blood flow. The smooth muscles around the bronchi relax increasing airflow to the lungs, so more oxygen is absorbed by the blood. The pancreas is also stimulated by adrenaline to increase glucagon secretion which initiates release of glucose from the liver into the blood. The increase in blood flow, oxygen and glucose level result in increased cell metabolism.

The radial muscles of the iris contract resulting in pupil dilation while the brain is put in a state of heightened awareness. These multiple responses in the presence of adrenaline ensure the body is fully prepared to act in an emergency.



Describe the role of thyroid stimulating hormone in the production of thyroxine.

The control of **thyroxine** production by **thyroid stimulating hormone** (TSH) is a classic example of negative feedback, which was explained in Chapter 16.

The hypothalamus releases **thyroid-releasing hormone** (TRH) and this causes the anterior pituitary to secrete TSH. This, in turn, triggers the production and release of thyroxine by the thyroid gland. An increase in the level of thyroxine in the blood inhibits the hormones

from the hypothalamus and the pituitary – **negative feedback**. As we shall see in the next Chapter, this system plays an important role in the control of body temperature. (see Fig 18.10)



Compare the action of the nervous and endocrine systems.

It may be easier to understand the differences between the nervous system and the endocrine system if we consider some examples. To react quickly to a moth heading towards your eye, you use the nervous system to immediately send a specific signal to your eyelid muscles to contract and then relax – that is, a rapid blink. On the other hand, to cause a range of cells to take up glucose from the blood over several hours, you need the hormone insulin to spread throughout the body in the blood, and to continue causing the effect during this time. In either case, the system used is well-suited for its intended purpose.

When sending an impulse to an effector, nerves are very selective as to which target – muscle or gland – is activated. You can move just one finger or blink one eye without having other muscles contract. If hormones were to be used to stimulate muscles they would cause many muscles to contract.

Although a hormone will only produce an effect in those cells, tissues, or organs that have the appropriate receptors, these target sites tend to be much less specific than for nerve stimulation. As we have seen, adrenalin stimulates a wide range of cells.

As well as being highly specific in its destination, a nerve impulse travels directly from one part of the body to another in a fraction of a second. Hormones, however, are not sent directly to the target tissue but travel in the blood to all parts of the body. The time taken for hormones to reach their target is therefore limited by the speed of blood flow. The response to a nerve impulse is very short-lived, so prolonged responses would require a succession of nerve impulses. This is in direct contrast to the effects of hormones that may last from minutes to months. This will be determined by the half-life of the hormone, and the frequency of its secretion. (See textbox.)

Communication	Pathway	Message	Site of Action	Speed of Action	Duration
Nervous	direct via axons of nerve cells	electro-chemical impulse	highly specific	fast	short term
Hormonal	indirect via blood	chemical	target cells - can be widespread	slow	long term

ENDOCRINOLOGY

This is the study and treatment of disorders of the endocrine system. These include diabetes (pancreas), goitre (thyroid), dwarfism and gigantism (pituitary), and Addison's disease (adrenal glands).

NERVES AND HORMONES



tinyurl.com/y96vd5al

HOW LONG DO HORMONES LAST?

Hormones circulating in the blood are eventually broken down by the liver and kidney and the resulting breakdown products are excreted in urine or faeces.

A measure of a hormone's duration of activity is its half-life - the time taken for the concentration of the hormone to be halved.

The half life of adrenalin is approximately one minute, while for thyroid hormones the half-life is several hours. Some biologists regard vitamin D as a hormone, and its half-life is several months.

Human growth hormone (HGH) is sometimes illegally used by athletes to increase muscle mass. It is, however, hard to detect as it has a half life of less than 45 minutes and testing an athletes' blood sample several hours after HGH was administered would not find an abnormally high level of the hormone.

USE OF HORMONES IN MILK PRODUCTION



A hormone called recombinant bovine somatotrophin (rBST) that increases milk yield in dairy cattle is banned in some countries, including Australia and New Zealand, but is used elsewhere, including the USA. The ban has important economic implications for Australia and New Zealand, as they are major international suppliers of dairy products, and the risks of rBST are still controversial.

The role of the hypothalamus as a 'bridge' between the nervous and endocrine systems

The hypothalamus in the brain is part of the central nervous system and it provides a 'bridge' between the nervous system and endocrine system. It receives signals via afferent nerves, sends nerve impulses via autonomic nerves, and secretes hormones and hormone-like substances that control the secretion of hormones by the pituitary gland. The hormones ADH and oxytocin are produced by the hypothalamus and travel to the posterior pituitary via a 'neural tube', where they are stored and subsequently released into the blood.

The hypothalamus also produces stimulating and inhibiting hormones that travel via the blood to the anterior pituitary. An example is TRH, which stimulates the production of TSH. (See Fig. 18.10) As part of negative feedback in the control of body temperature, the hypothalamus also responds to changes in the level of thyroxine and TSH in the blood.

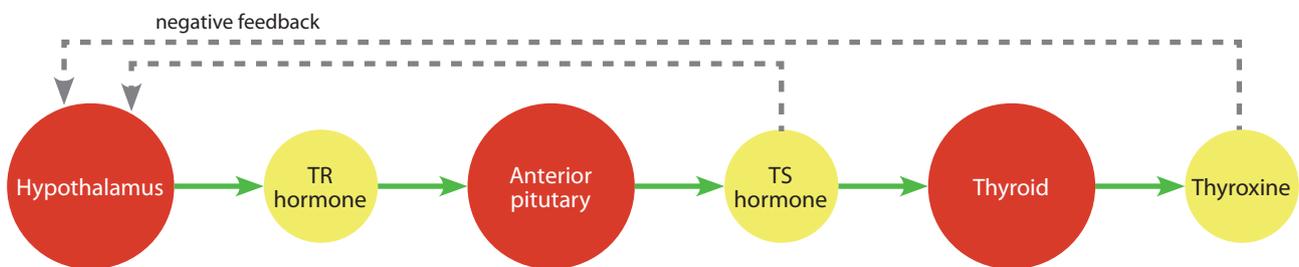


Fig. 18.10 Hormones from the hypothalamus

Study Questions

1. Explain, using an example, the meaning of the terms hormone, endocrine gland, target cell, and target organ.
2. The endocrine system is made up of the endocrine glands that secrete hormones. For the hormones, adrenaline, thyroxine, antidiuretic hormone, glucagon, insulin and aldosterone, state which gland secretes them, their target, and their effect.
3. How does a hormone's shape compare to the shape of its corresponding receptor?
4. Explain the difference between the receptor for a lipid soluble steroid hormone and the receptor for a water soluble protein hormone using labelled diagrams.
5. Endocrine glands have to be stimulated to secrete their hormones by either nerves or other hormones. Give specific examples of each method.
6. Outline the role of adrenaline in the 'flight or fight' response.
7. Describe the role of the thyroid stimulating hormone in the production of thyroxine?
8. What functions do the nervous system and the endocrine system have in common and what are the main differences?

Homeostatic Control Mechanisms

19

The nervous system and endocrine system function independently or together to achieve homeostasis.

Homeostasis is the maintenance of a steady set of internal conditions such as temperature, salt/water concentration, and blood sugar level. To achieve this control requires the body's systems to respond to the stimuli of changes in conditions and to work together in a coordinated way. Detecting stimuli involves sensory receptors. Signalling the effectors to respond involves both the nervous system and the endocrine system.



Explain how the nervous and endocrine systems work independently or together to:

- › control body temperature.
- › enable osmoregulation.
- › maintain blood sugar level.
- › monitor pH in the brain to maintain a constant carbon dioxide level in the blood.

Control of Body Temperature – involves nervous and endocrine systems

In the human body the internal temperature is normally preset to around 37°C and remains remarkably constant. Blood temperature is monitored by the **thermoregulatory centre** in the **hypothalamus** of the brain and any change from 37°C will cause several negative feedback mechanisms that may involve nerves or hormones.



Describe the action of thyroid stimulating hormone and thyroxine in metabolism.

A fall in body temperature

A fall in blood temperature below 37°C (See Fig. 19.1) stimulates the thermoregulatory centre to:

- send nerve messages to the skeletal muscle to repeatedly relax and contract. This shivering requires the muscles to respire and heat is generated this way.

HOMEOSTASIS



tinyurl.com/4sjfautz

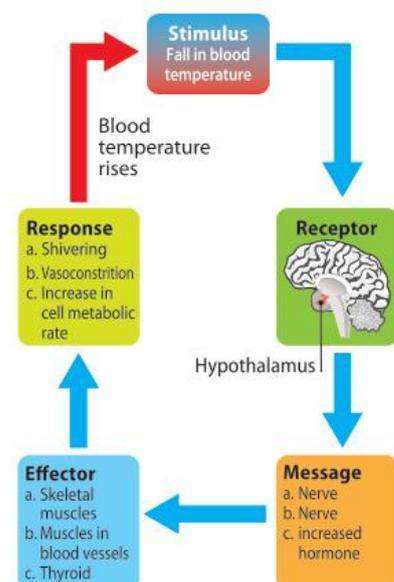


Fig. 19.1 Fall in blood temperature

FEVER AND PYROGENS

During infection, certain white blood cells release specific chemicals called pyrogens that stimulate the hypothalamus to reset the preferred temperature of the body to a higher level. This higher temperature assists the body in overcoming infections by inhibiting the growth of some pathogens, stimulating phagocytosis and speeding up the reactions of the body to repair tissues.

- b. send nerve messages to sphincter muscles around arterioles in the skin. The response is constriction of blood vessels (called *vasoconstriction*). This decreases the flow of blood to the surface of the body, directing the blood flow mainly to essential 'core' organs, such as the heart, brain, kidneys, and liver and helps to conserve heat energy.
- c. release hormones via the pituitary that stimulate the thyroid gland to secrete the hormone thyroxine. This, in turn, stimulates general cell metabolism and causes an increase in temperature. (See Fig. 19.1)

The mechanisms above either increase heat production in the body or restrict heat loss, leading to an increase in temperature – negative feedback.

A rise in body temperature

A rise in blood temperature above 37°C (See Fig. 19.2) is detected by the thermoregulatory centre and stimulates it to:

- a. send nerve impulses to the sweat glands causing them to secrete sweat onto the body's surface. Evaporation of the sweat removes heat from the skin.
- b. send fewer nerve messages to the sphincter muscles of the arterioles, causing them to relax. This is called *vasodilation* and it increases blood flow to the surface of the skin, increasing the rate of heat loss from the body.
- c. decrease the secretion of hormones to the thyroid. The thyroid lowers the amount of thyroxine released and so cell metabolism decreases.

The mechanisms above decrease heat production and increase heat loss, leading to a decrease in body temperature – negative feedback.

The thermoregulatory centre also receives nerve impulses from temperature receptors located in the skin. The stimuli received in this way seem to result in voluntary actions such as moving to a warmer or cooler place, adding or removing clothing, and so on.

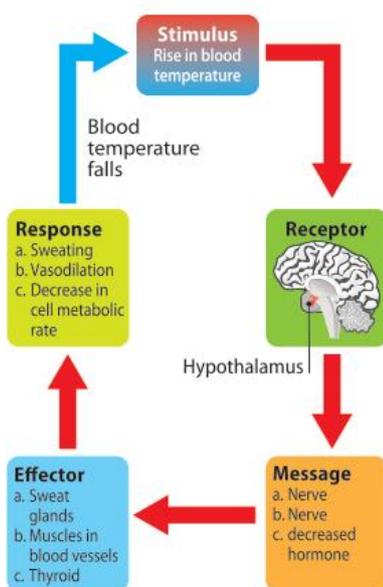


Fig. 19.2 Rise in blood temperature

Osmoregulation – involves mainly the endocrine system

Osmoregulation refers to the maintenance of water and solute balance in the body. The kidneys are the main organs involved in osmoregulation. (See Fig. 19.3)

We have seen that cells not only have nutritional requirements, but that they also produce wastes as a result of normal metabolism. An extremely important organ involved in the control of ion and solute levels and the maintenance of water balance, as well as the excretion of waste products from the body, is the kidney. The blood

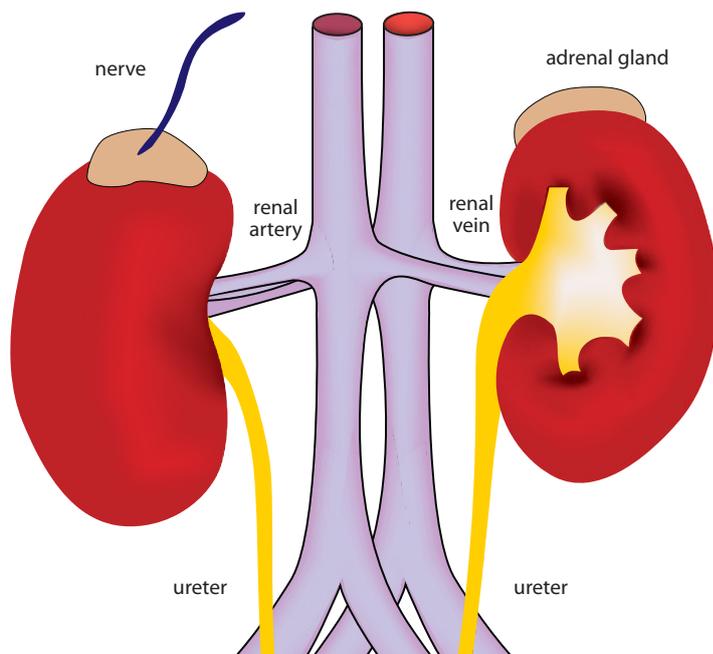


Fig. 19.3 The kidney

is continuously passing through the kidneys and as a result its composition remains remarkably constant.

Humans have two kidneys and they are located on either side of the lower abdominal cavity.

The nephron

Each kidney contains more than a million nephrons. A nephron is made up of a long, thin, partly coiled tubule closely associated with blood capillaries that deliver and remove blood. Blood arrives at a nephron via a branch of the renal artery that has become subdivided to form a tiny afferent vessel. This *afferent* vessel leads to a microscopic 'ball of capillaries' called a **glomerulus**. The glomerulus fits inside a structure called **Bowman's capsule** at one end of the nephron tubule, rather like a fist fits into a cupped hand or a baseball fits into a glove. (Remember that these structures are three dimensional.) Leaving the glomerulus is an *efferent* blood vessel that leads to a network of capillaries that are closely associated with the nephron tubule. Eventually these capillaries lead to a branch of the renal vein which returns blood from the kidney to the heart. (See Fig. 19.4b)

The method by which the nephrons regulate the composition of the blood is rather intriguing. In the glomerulus, **filtration** occurs under pressure. The pressure results from the fact that the efferent blood vessel has a slightly smaller diameter than the afferent one. The filtration occurs because pores in the capillary walls of the glomerulus allow materials to pass through on the basis of their size. These materials then pass through a *basement membrane* made of a matrix of glycoprotein fibrils (See Fig. 19.4a). Blood cells

THE KIDNEY

Each kidney is composed of easily observed regions called the cortex, the medulla, and the pelvis. A tube called the ureter leads from each kidney pelvis and connects to the urinary bladder. Each kidney is served by major blood vessels called the renal artery and the renal vein. You can picture a kidney as being a neat package of nephrons. If you understand how a single nephron works then you will understand the functioning of the kidney as a complete organ.

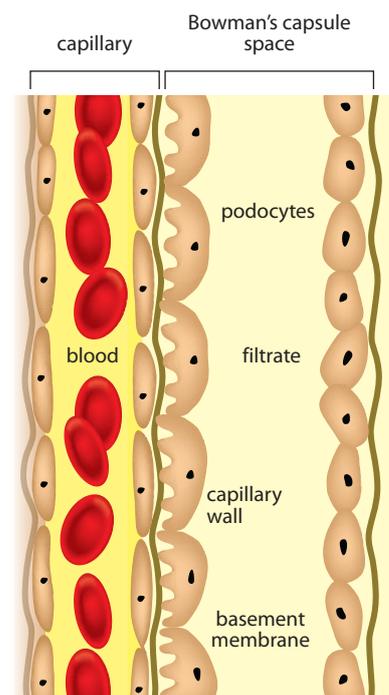


Fig. 19.4a Movement from blood to the filtrate.

For substances to move from the blood to the filtrate, they need to pass through three layers: the capillary wall, the basement membrane, and the podocyte layer.

DIURESIS

'Diuresis' is a condition in which an excessive amount of urine is produced. Anti-diuretic hormone (ADH) increases water reabsorption by the kidneys, reducing the amount of urine produced.

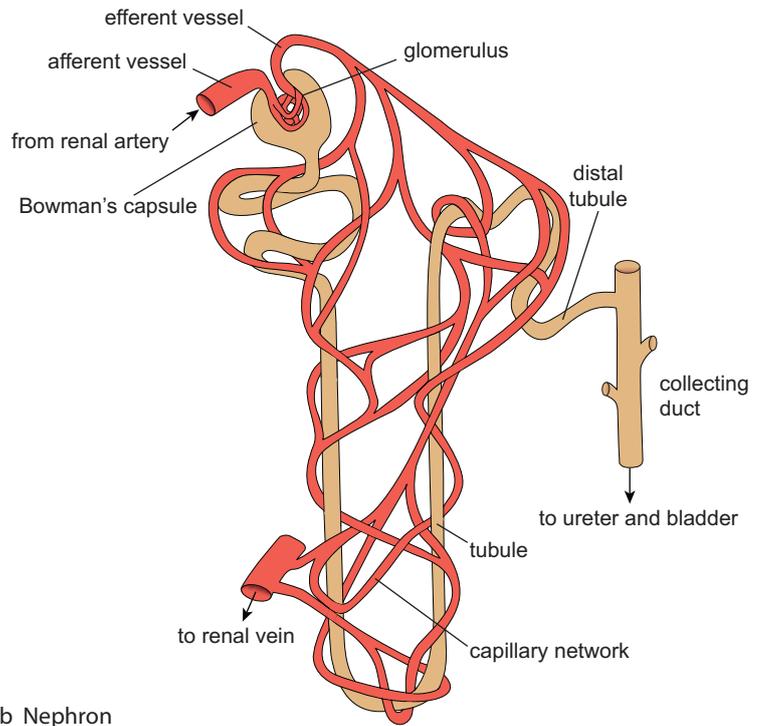


Fig. 19.4b Nephron

and plasma proteins are too large to pass through into Bowman's capsule, but water, glucose, urea, amino acids, dissolved ions, and other substances, are able to pass through the 'filter' and are collectively called **filtrate**. Note that at this stage no distinction is made between useful materials and wastes.

Once inside the nephron tubule, this filtrate is actually in the 'external' environment as there are no membranes between the lumen (inside) of the nephron tubule and the outside of the body. It is therefore essential that any useful material that has been filtered into the tubule is **reabsorbed** back into the blood and is not lost from the body. This process of reabsorption occurs along the length of the nephron tubule.

Under normal circumstances nearly all of the glucose and amino acids are reabsorbed into the blood capillaries that surround the nephron tubule. The nephron tubules have walls made of a single layer of cells. These cells have numerous *microvilli* to increase the surface area. As there are over a million nephrons in each kidney, the surface area for **filtration and reabsorption** is very large. Most of the water in the filtrate (about 99%) is reabsorbed across the nephron and the walls of the collecting ducts, but the reabsorption of water and ions in solution is regulated in a way that reflects the body's requirements at the time. Fine control of these processes involves hormones that are secreted by the pituitary and by the kidneys themselves. An important hormone from the pituitary is antidiuretic hormone (ADH), which makes the distal tubule and collecting duct walls more permeable to water and increases the reabsorption of water into the blood.

REABSORPTION

Some reabsorption of glucose, amino acids and inorganic ions is carried out by the process of active transport that it can take place against the concentration gradient. The cells lining the tubules of the nephron have large numbers of mitochondria to supply the energy for active transport.

Metabolic poisons which interfere with cellular respiration will therefore prevent the nephron tubule cells carrying out reabsorption and these vital substances will be excreted in the urine.

Water on the other hand is reabsorbed from the filtrate by the passive process of osmosis.

Urea, which is formed when cells break down amino acids, is a waste that the body must remove. Therefore, little of the urea in the filtrate is reabsorbed into the blood, and most is removed in the urine.



Describe the effect of antidiuretic hormone (ADH) on the nephron in osmoregulation

Antidiuretic hormone (ADH) – also known as vasopressin – is synthesised in the hypothalamus and stored in the posterior pituitary gland. It is secreted by the pituitary in response to an increase in the concentration of solutes in the blood detected by **osmoreceptors** in the hypothalamus. ADH is transported in the blood, and binds to receptor molecules on the cells of the distal tubules of the **nephrons**, and the **collecting ducts** in the kidneys. It makes the distal tubule and collecting duct walls more permeable to water by increasing the number of **aquaporins** present in the cell membranes on the filtrate side of the collecting ducts. (See Fig. 19.5)

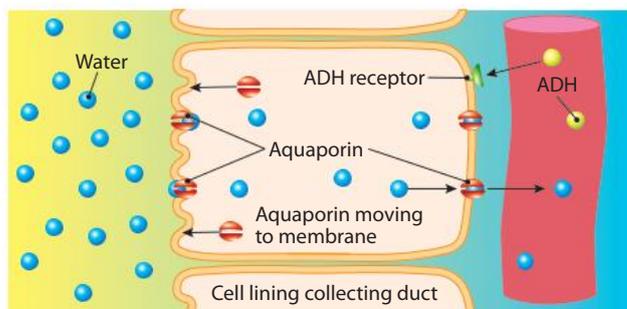


Fig. 19.5 The action of ADH on collecting ducts

In Chapter 10 we introduced aquaporins as the protein ‘channels’ that facilitate the movement of water across membranes. Aquaporins in the cell membranes on the blood side of the collecting ducts are always present. They are not affected by the presence of ADH. Thus, the effect of ADH is to increase the **reabsorption** of water into the blood by osmosis, reducing the concentration of solutes – an example of **negative feedback**.

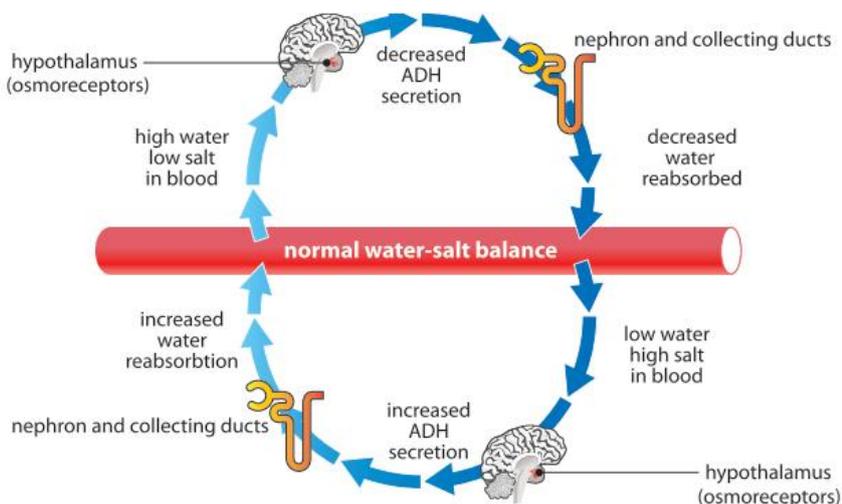


Fig. 19.6 Osmoregulation

KIDNEY FAILURE

When a person’s kidneys fail, the body is unable to maintain the correct water-salt balance in the blood and the toxins such as urea increase in concentration. This condition is fatal unless treated by renal dialysis. In this procedure the patient’s blood is pumped through fine tubes with semi permeable walls. On the outside of the tubes is a liquid with a similar composition to plasma – the dialysis fluid. Substances move across the tube walls by diffusion; urea moves out of the blood and desired salts move in. Glucose and amino acids will have the same concentration either side of the wall and there will be no net movement. The dialysis fluid is changed frequently to maintain the concentration gradients.

ADH AND AQUAPORINS



tinyurl.com/2emuk5nv

Osmoregulation, blood volume, and blood pressure are also affected by the 'renin-angiotensin-aldosterone system' which interacts with ADH, but that is beyond the scope of this course.

WHY ALCOHOL IS A DIURETIC

Alcohol inhibits the release of ADH and thus increases urine output.

mmol/L (millimoles per litre) is a unit used by chemists to refer to very small concentrations

TYPES OF DIABETES

Most people associate diabetes with sugar, but this is not always the case. Diabetes is a general term used to describe excessive urine production. Usually this is due to high blood sugar level – a condition called **diabetes mellitus**, known commonly as Type 1 or Type 2 diabetes. However, **diabetes insipidus** results from either insufficient secretion of ADH, or the inability to respond to ADH.



Discuss the links between osmoregulation, blood volume, and blood pressure.

The osmotic pressure in the blood and tissue fluid is determined by the water and solute balance. The maintenance of a constant osmotic pressure – osmoregulation – involves controlling the relative concentrations of water and solutes. Antidiuretic hormone regulates water and solute balance, and this affects blood volume and blood pressure.

An increase in the water content of the blood will increase the volume of the blood and result in an increase in blood pressure. On the other hand, a decrease in the water content of the blood will decrease the volume of the blood and result in a decrease in blood pressure.

Increasing the water content of the blood will decrease its solute concentration, and decreasing the water content of the blood will increase its solute concentration. You can see that osmoregulation, blood volume, and blood pressure all affect one another, and are closely linked.

ADH is also secreted in response to lower blood pressure or blood volume. (See Fig. 19.7) These are detected by stretch receptors in the circulatory system that send a nerve message to the hypothalamus.

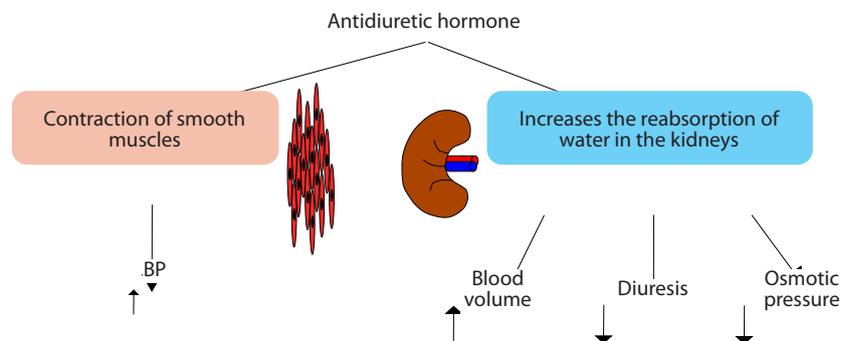


Fig. 19.7 Effects of antidiuretic hormone (ADH)

Control of blood sugar level – involves the endocrine system

The level of blood sugar (glucose) in a healthy human lies between about 4.0 and 7.8 mmol/L (millimoles per litre). The reason for the variation is that blood sugar level rises after meals, and then steadily falls. For this reason blood sugar is normally measured after fasting to allow for valid comparisons to be made. Fasting blood sugar level should be between 4.0 and 5.5 mmol/L.

The control of blood sugar level is an example of a homeostatic control mechanism. That is, it is 'self regulating' and involves negative feedback. The stimulus is either an increase or decrease in

blood sugar level, and the response is the opposite of the stimulus. Thus, an increase in blood sugar level will result in a decrease, and a decrease in blood sugar level will result in an increase. This is the principle of all homeostatic control mechanisms. To understand how this works, we need to identify the sugar (glucose) receptors that detect changes in the blood sugar level and the message that conveys this information to the effectors. We also need to identify the effectors that carry out the response.

Blood sugar (glucose) level

Cells that respond to an increase in blood glucose level are located in the pancreas. As **beta cells** in the **islets of Langerhans** in the pancreas take up glucose, for example after a meal, they secrete the hormone **insulin** into the blood. So, the beta cells can be considered as glucose receptors and the message is a hormone.

A decrease in blood glucose level causes **alpha cells** in the islets of Langerhans of the pancreas to secrete another hormone, **glucagon**.

To understand how blood sugar level is controlled we need to examine the effects of insulin and glucagon. (See Fig. 19.8)



Compare the action of insulin and glucagon in blood sugar regulation.

The effects of insulin

Many cells of the body are targets for insulin, particularly in the liver, and muscle and fat tissue. Insulin causes liver cells to convert glucose into glycogen, an insoluble storage polysaccharide, thus reducing the concentration of glucose in the cell and therefore increasing the diffusion of glucose into the cell from the blood. It causes muscle and fat cells to take in glucose from the blood. Thus, insulin causes a **decrease** in blood sugar level.

The effects of glucagon

Glucagon binds to glucagon receptors on liver cells causing the cells to convert glycogen to glucose and release glucose in to the blood. This causes an **increases** the blood sugar level.

As insulin and glucagon have opposite effects, the regulation of blood sugar level results from a balance between them.

Effectors

As we have seen, the main effectors involved in the regulation of blood sugar level are the liver, muscle cells, and fat tissue cells. They all respond to insulin by reducing blood sugar level, and liver cells respond to glucagon by increasing blood sugar level.

SUGAR LEVEL

Too much blood sugar (Hyperglycaemia) causes an osmotic problem. Not enough blood sugar (Hypoglycaemia) causes a decrease in respiration.

CONTROL OF BLOOD GLUCOSE



tinyurl.com/4wcveuce

GLUCOSE TRANSPORT PROTEINS (GLUTs)

Glucose is transported across cell membranes by glucose transport proteins called GLUTs. In mammals, 14 GLUT proteins have been identified and named GLUT1 to GLUT14. In humans, the most significant GLUTs are GLUT1, GLUT2, and GLUT4. GLUT2 allows the uptake of glucose by liver and pancreas cells. It also allows glucose to move into and out of these cells. This is particularly important in the liver, which stores excess glucose as glycogen, and releases glucose when it is required. This also enables pancreatic beta cells to accurately monitor blood glucose level. GLUT4 is found in the membranes of muscle and fat cells, and allows the uptake of glucose. It is insulin-dependent, meaning that when the insulin level is elevated more glucose is taken up and stored or metabolised by these cells. GLUT4 does not readily permit glucose movement out of cells. (See carrier proteins on page 87).

Glucose and glycogen are carbohydrates. Glucose molecules are single sugar units called monosaccharides. Glycogen is made up of many glucose units joined together and it is called a polysaccharide. (see Fig. 19.9)

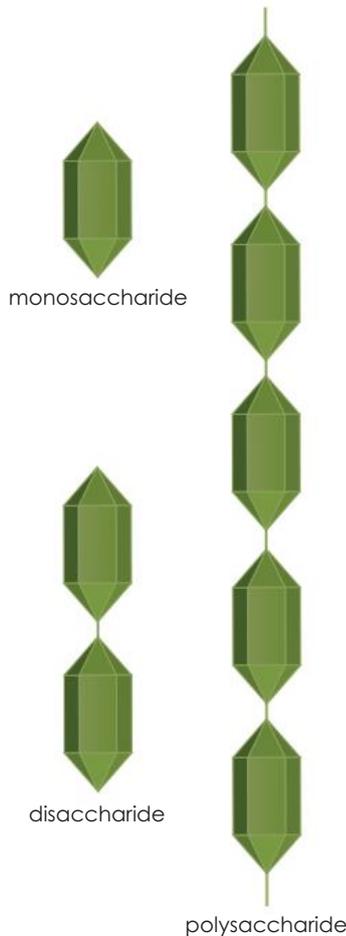


Fig. 19.9 Carbohydrates

In Chapter 5 we outlined the production of human insulin by bacteria, using genetic engineering.

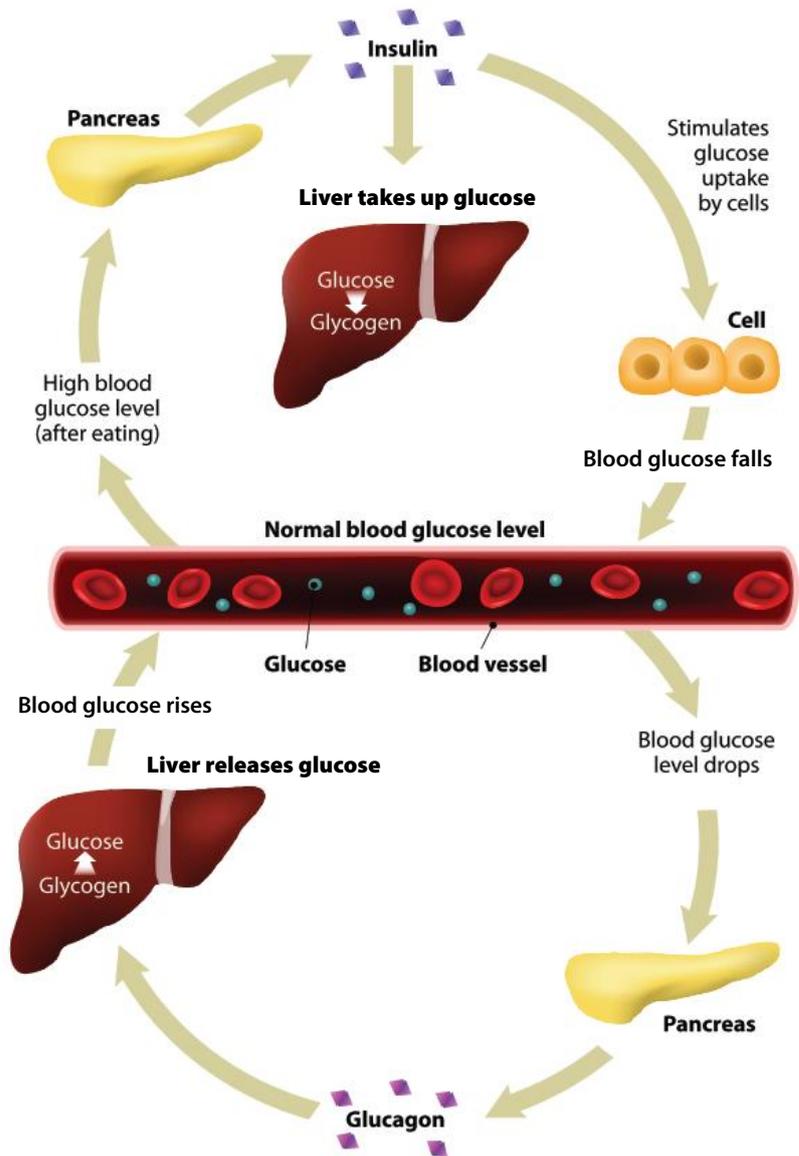


Fig. 19.8 Regulation of blood glucose



Describe how diabetes mellitus can result from a hormonal imbalance.

The term diabetes refers to the over production of urine and is usually due to the inability to maintain blood sugar level in the normal range.

Type 1 diabetes results from the inability to produce insulin due to an auto-immune disease that destroys insulin-producing cells of the pancreas.

Type 2 diabetes results from the body becoming resistant to insulin and/or being unable to make enough insulin. The current treatment for Type 2 is to control sugar intake by adopting a low calorie diet. Type A insulin resistance is discussed on page 24. There are trials of medicine that improve the body's sensitivity to insulin.

A person with Type 1 diabetes is unable to regulate their blood sugar level by producing insulin when their blood sugar level rises. Some of these people are also unable to produce glucagon if their blood glucose level falls. Thus, they run the risk of a high blood sugar level after a meal, and a low blood sugar level between meals. This can be controlled with insulin (and glucagon) injections, and by carefully monitoring food intake. Recent studies have found POMC neurons in the hypothalamus are able to detect nutrient availability and signal beta cells in the pancreas so exerting some control on blood sugar level.

Monitoring pH – involves the nervous system

Humans obtain energy by breaking down glucose to carbon dioxide and water in aerobic respiration.



Some of the carbon dioxide dissolves in the tissue fluid, but most of it reacts with water to form carbonic acid. The carbonic acid gives rise to hydrogencarbonate ions and hydrogen ions that are transported in the blood.



This increase in hydrogen ions lowers the pH of the blood.

However, the pH of the blood is not monitored directly, as hydrogen ions cannot pass from the blood to the respiratory centre in the brain due to their positive charge. However, uncharged molecules of carbon dioxide are able to pass from the blood to the cerebro-spinal fluid and form hydrogencarbonate ions and hydrogen ions, that lower the pH of the fluid. (See Fig. 19.10)

A decrease in pH, due to an increased level of carbon dioxide, will result in more nerve impulses from the brain to the respiratory muscles of the chest and diaphragm and an increase in breathing rate. This will lower the carbon dioxide concentration in the blood and increase its pH. The **increased pH** will be **detected** in the **respiratory centre** and result in fewer nerve impulses to the chest and diaphragm muscles, resulting in a decreased breathing rate. This is another classic example of a homeostatic control mechanism involving negative feedback.

CARBON DIOXIDE TRANSPORT

The transport of carbon dioxide from the tissues is interesting. It diffuses into red blood cells where some becomes attached to haemoglobin. However, most of it is converted to carbonic acid in the red blood cell's cytoplasm by the enzyme carbonic anhydrase. The carbonic acid then splits into hydrogen ions and hydrogencarbonate ions and these ions then move out into the plasma. In the lungs the reverse process occurs, with hydrogencarbonate moving back into the red blood cells to be converted to carbon dioxide which returns to the plasma. Carbon dioxide diffuses across the alveolar wall into the air space and is then exhaled.

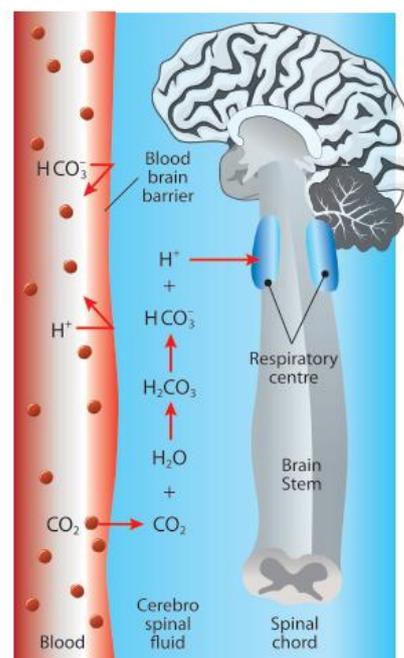


Fig. 19.10 Blood brain barrier monitoring pH

Study Questions

1. The nervous and endocrine systems work independently or together to control body temperature.
 - (a) What is the role of the thermoregulatory centre of the brain?
 - (b) Describe the stimulus-response sequence that follows a fall in body temperature.
 - (c) Describe the stimulus-response sequence that follows a rise in body temperature.
2. Explain how vasoconstriction and vasodilation assist in the regulation of body temperature.
3. Describe the action of thyroid stimulating hormone and thyroxine in metabolism and control of body temperature.
4. State two other mechanisms involved in human temperature control and explain how each of them could assist in regulating body temperature.
5. The kidney is involved in osmoregulation - the regulation of the water and solute of the blood.
Draw a diagram of the kidney nephron and label the glomerulus, Bowman's capsule, tubule, capillary network, and collecting duct. What are the special features of the nephron that enable it to carry out its function efficiently?
6. The kidneys receive a large proportion (about 20 percent) of the blood pumped with each heart beat.
 - (a) Define the terms filtration and reabsorption and state where in the nephron these processes take place.
 - (b) Describe the passage of a salt ion from the blood through the kidney to the bladder.
 - (c) Describe the passage of a water molecule from the blood through the kidney back into the blood.
 - (d) How would you expect the composition of blood in a renal vein to differ from the composition of blood in a renal artery?
 - (e) Explain the difference between urea and urine.
7. Osmoregulation involves mainly control by the endocrine system. Describe, using a stimulus-response model, the role of anti-diuretic hormone (ADH), and aquaporins, in osmoregulation.
8. Discuss the links between osmoregulation, blood volume, and blood pressure.
9. Control of blood sugar level involves the endocrine system. Compare the action of insulin and glucagon in blood sugar regulation.
10. Describe how diabetes can result from a hormonal imbalance.
11. Explain the mechanism used by the nervous system to monitor pH in the brain to maintain CO_2 level in the blood.



TOPIC

4

Evolution

- 20 How Cells Have Evolved
- 21 Defining Species
- 22 Evidence for Evolution
- 23 Gene Pools and Natural Selection
- 24 Speciation and Evolution
- 25 Human Impact

The theory of evolution is often misunderstood or misrepresented, perhaps in part due to the word 'theory'. The textbox on page 180 explains that a scientific 'theory' is actually the best explanation that we have **based on all the available evidence**. The evidence for the evolution of life on Earth, as explained by Charles Darwin in his ground-breaking 1859 publication *On the Origin of Species*, is overwhelming. The more we discover, the more scientific support there is for the theory of evolution. The discovery of the structure of DNA and the genetic code and the understanding of life that has followed, has simply added to the mountain of evidence for evolution. Comparative genomics allows us to study evolutionary relationships in this way.

Humans have invented the term **species** to define select groups of organisms. In 1745 a Swedish naturalist called Linnaeus used the convention of giving every living thing membership of a species defined by a two word name (e.g. *Macropus rufus*, a red kangaroo). This has become the basis of our modern classification system.

As well as using the idea of a species to make it easier to study the diversity of living things, we have also looked for a useful organising concept for our studies of the relationships between organisms. Biologists have categorised organisms into populations and communities, both past and present, to make this study easier.

A population is defined as a group of organisms of the same species occupying a particular space at a particular time. The individuals of a population share a common gene pool, a term that suggests the genes themselves can be selected from the pool to make up future generations. The gene pool is the sum of all the genes of all the individuals in a population.

A community refers to all the populations of different species in an area at a particular time. While a community only includes the living inhabitants, an ecosystem includes both the living inhabitants and the non-living surroundings. In an ecosystem the living things are called biotic and the nonliving factors are called abiotic.

Maintaining biodiversity and understanding the impacts that human beings have on our planet are also discussed in this topic.

Science as a Human Endeavour

Throughout this topic examples that illustrate key concepts of science as a human endeavour are indicated by the symbol ▼. There are examples of communication and collaboration, development of scientific models and new technologies, influence on and by other areas of study and society, and applications and limitations of biological knowledge.

How Cells Have Evolved

20

Evidence shows that life has existed on Earth for around 3.5 billion years, during which time it has diversified.

Evidence suggests that the Earth is at least 4.5 billion years old and that conditions when the Earth was formed were very different from what they are today. For example, it seems that the atmosphere at that time contained no oxygen but had large amounts of carbon dioxide and nitrogen. Obviously, this hostile environment would not have supported life as we know it today.

Fossil evidence like that in ancient rocks from Western Australia indicates that the first cells existed at least 3.5 billion years ago. One suggestion is that the first forms of life on Earth were prokaryotes that did not require oxygen. This idea assumes that life began in the sea, in a kind of primordial soup of organic molecules. It seems that about 2.5 billion years ago prokaryotes that were able to photosynthesise became prominent. These photosynthetic cells were very similar to the blue-green algae or cyanobacteria of today. As oxygen is a byproduct of photosynthesis, the activities of these photosynthetic bacteria began to drastically change the composition of the atmosphere by increasing the concentration of oxygen. This made it possible for some cells to use oxygen as part of their metabolism and thus become more complex. The formation of these more complex cells was an important step in the evolution of life on Earth and in this chapter we consider how cells have most likely evolved.

Existing cells are the products of evolution. There is evidence that prokaryotic cells existed before eukaryotic cells.

When compared with eukaryotic cells, prokaryotic cells are fundamentally simpler in their metabolism and structure. Eukaryotic cells would not have been able to survive in the hostile conditions provided by the Earth's early atmosphere and the best available evidence suggests that prokaryotic cells existed long before the first eukaryotic cells appeared. While prokaryotes existed at least 3.5 billion years ago, the first eukaryotic cells are thought to have been formed about 1.5 billion years ago.

Abiogenesis

The formation of living matter from non-living matter.

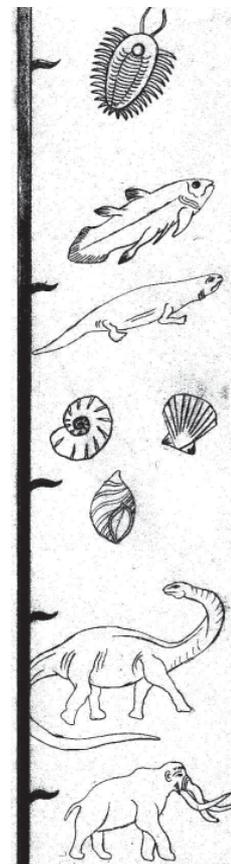


Fig. 20.1 Fossil record



Fig. 20.2 Stromatolites in Western Australia



Describe this evidence, including fossil evidence.

Some of the oldest known fossils have been found in structures called stromatolites. These are made up of ancient bacterial mats in which sediment has become trapped and compressed to form rocks. Until recently, the most ancient stromatolites found were in Western Australia and they are estimated to be 3.5 billion years old. (See Fig. 20.2) In 2016, stromatolites estimated to be 3.7 billion years old were discovered in Greenland.



Explain how the ancestry of most existing eukaryotic cells probably involved endosymbiotic events.

One of the most interesting and important events in the evolution of life on Earth involves the development of the first eukaryotic cells. It has been proposed that when all life consisted of prokaryotic cells some of the larger cells may have engulfed some of the smaller ones in a process similar to modern-day phagocytosis. In some cases the smaller engulfed cell was able to respire aerobically or was able to photosynthesise. The newly formed 'super cell' was then able to carry out more functions than either of its component cells could on their own. (See Fig. 20.3)

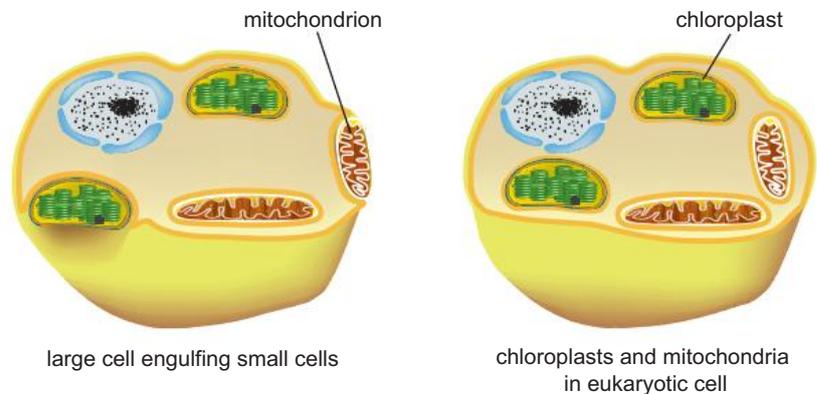


Fig. 20.3 Endosymbiosis

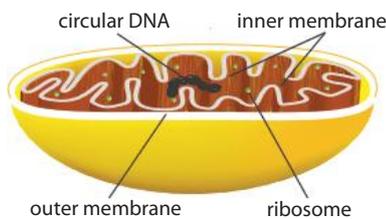


Fig. 20.4 Mitochondrion

The term **symbiosis** is used to describe the situation in which two organisms help one another and both benefit. In the case of **endosymbiosis** one cell actually lives inside another and both benefit. It is thought that mitochondria and chloroplasts may be the descendants of ancient bacterial cells that were engulfed by larger prokaryotes, and that eukaryotic cells were the product of such associations. Evidence to support this view includes:

- › chloroplasts and mitochondria have their own DNA, separate from the DNA in the nucleus. Their DNA resembles prokaryotic DNA as it is circular, it has no protein attached, and it has no non-coding sequences (introns).

- › chloroplasts and mitochondria contain their own ribosomes and these more closely resemble bacterial ribosomes than the ribosomes in the cytoplasm. These ribosomes are smaller and have a similar sequence of nucleotides to bacterial ribosomes.
- › chloroplasts and mitochondria are able to self-replicate, independently of the cell. Their replication resembles the binary fission of prokaryotes.
- › both chloroplasts and mitochondria have two membranes that are distinctly different from one another. The outer membrane is similar to the host cell's plasma membrane, while the inner membrane contains enzymes and transport proteins similar to those found in the membranes of bacteria. This suggests that the inner membrane originated from the invading prokaryote and that the outer membrane is derived from the host cell. The outer membrane was probably formed in the same way that an amoeba's food vacuole forms during phagocytosis.

Even today there are examples of endosymbiotic relationships between prokaryotic and eukaryotic cells. The epithelial cells of giant clams, bivalve molluscs, have cyanobacteria living within them. The cyanobacteria provide glucose and oxygen to the clam cells, while receiving carbon dioxide, nutrients and protection from their host.

Membranes may have formed spontaneously and the first simple cells may have used RNA as genetic information. Ribozymes may have played a role in this development.

Billions of years ago, before life on Earth began, the Earth's atmosphere was nothing like it is today. It has been shown by laboratory experiments that organic molecules could have formed under the conditions that existed at that time. Some of these organic molecules were fatty acid chains that probably gave rise to simple membranes in the form of primitive vesicles. (See Fig. 20.5) For life to exist it is not enough to just have organic molecules, even if they could gather together and form a cell-like structure. Jack Szostak, who received a Nobel Prize for his work on telomeres (see Chapter 1), went on to research the origin of life on Earth, including the formation of primitive membranes. This led him to investigate the role of RNA and ribozymes. The links in the side column are to short video clips of Szostak explaining aspects of this work.

▶ SIMPLE MEMBRANES



tinyurl.com/yd27xmjw

▶ PRIMITIVE VESICLES



tinyurl.com/ycmbcskm

▶ EVOLUTION OF VESICLES



tinyurl.com/y854o598

▶ RNA AS GENETIC MATERIAL



tinyurl.com/ybwn7d63

▶ PRIMITIVE CELL CYCLE



tinyurl.com/y89j3wyn

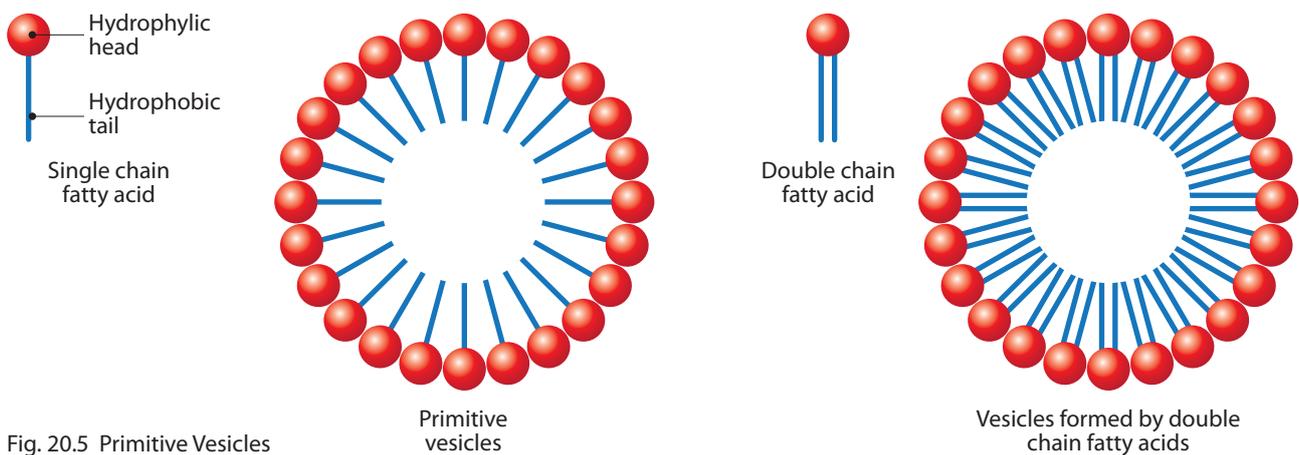


Fig. 20.5 Primitive Vesicles



Describe the possible roles of RNA and ribozymes in the first simple cells.

There needs to be a method of storing and transmitting information to enable living structures to carry out functions and reproduce. The information needs to be in a form that can be copied and then transferred to the next generation. Nucleic acids are the only molecules that appear to be able to perform both of these functions. Although all living things that we know of use DNA for this purpose, it is likely that the very first simple cells used RNA. As it is a simpler molecule than DNA, it is more likely that it formed first and that its role was eventually replaced by DNA. Interestingly, some viruses use RNA as their genetic material, but as viruses are not considered to be living, the concept that DNA is universal to all living things still holds.

Another problem of 'early life' on Earth would have been the absence of proteins, particularly those that act as catalysts. (See Chapter 3 for details about enzymes.) It has been discovered that RNA, like enzymes, can catalyse chemical reactions under certain circumstances. RNA molecules with this ability are called **ribozymes**. In 1989 Sidney Altman and Thomas Cech were awarded the Nobel Prize in Chemistry for this discovery. (See link to the left)

Some people refer to the beginning of life on Earth as the "RNA world", as it is highly likely that the first living things used RNA instead of DNA and proteins. The RNA world gave rise to what might now be called a "DNA world". You can even think of the modern ribosome as being a "leftover" ribozyme, as it is made of RNA and protein and catalyses translation in protein synthesis.

It is not surprising to find that all living things use DNA to store and transmit genetic information as its double helix structure is more stable than RNA. We know that DNA is present in the chromosomes of all eukaryotic cells when they are formed and that DNA is also present in all prokaryotic cells, as a single circular chromosome together with small loops of DNA called plasmids. So for all living things on Earth, DNA is the only carrier of genetic information known.

DISCOVERY OF RIBOZYMES



tinyurl.com/3p35dr7v

Study Questions

1. Draw a time line starting from the origin of Earth at 4.5 billion years ago to the present. On the time line mark in the following events:
 - (a) origin of the Earth
 - (b) appearance of the first prokaryotic cells
 - (c) when photosynthetic prokaryotes became prominent
 - (d) appearance of first eukaryotes
 - (e) origin of plants (about 480 million years ago)
 - (f) appearance of mammals (225 million years ago).
2. Explain why the production of oxygen was a critical event for the evolution of life on Earth.
3. Describe the evidence that prokaryotic cells existed before eukaryotic cells including fossil evidence.
4. What is the difference between symbiosis and endosymbiosis?
5. Explain how the ancestry of most existing eukaryotic cells probably involved endosymbiotic events.
6. Evidence for the evolution of eukaryotic cells focuses on chloroplasts and mitochondria.
 - (a) Outline the current theory on the origins of these organelles.
 - (b) Explain why these two organelles in particular are so critical for the evolution of life on Earth.
 - (c) Chloroplasts and mitochondria have their own DNA. How does this provide evidence for the endosymbiotic origins of eukaryotic cells?
 - (d) Apart from DNA, list other organic molecules present in chloroplasts and mitochondria that resemble those found in bacteria.
 - (e) Explain how the membranes in chloroplasts and mitochondria provide evidence of endosymbiosis.
7. Outline the possible process that formed the first membranes?
8. Describe the possible roles of RNA and ribozymes in the first simple cells.

21

Defining Species

Review the differences between a species, population, community, and an ecosystem.

If you are lucky enough to have a lawn and a garden, this is a good place to start your study of species. Your lawn probably consists of grass, clover and 'weeds' such as dandelions. Slaters, beetles and spiders will be found near the soil surface. In the soil you would expect to find earthworms. Some birds may feed on these small animals, while other birds will eat seeds from the grass and clover. These organisms belong to separate species and separate populations, but the one community.

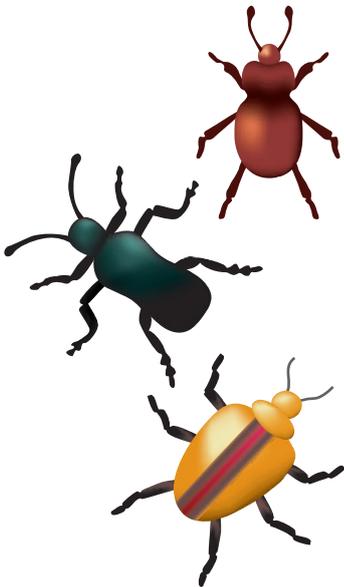


Fig. 21.1 Variation between species

Different criteria are used to define a species depending on the mode of reproduction.

Populations and Species

A population is defined as a group of organisms of the same species living in the same area. The lawn community referred to above consists of several populations. These include species of clover plants, earthworms, slaters, beetles, spiders, grasses and birds. Each one of these species can be distinguished from the others. (See Fig. 21.1)

A species that reproduces sexually can be defined by the ability of its members to actually or potentially interbreed to produce fertile offspring.

A commonly accepted definition of a species is a group of organisms that are more or less alike and can reproduce fertile offspring in their natural environment. That is not much help for asexually reproducing organisms, such as bacteria. In addition, many plants of different species readily interbreed to form hybrids.

Other criteria used to define a species include:

- › morphological similarity
- › biochemical similarity
- › sharing a common gene pool.

Morphological similarity

It is true that members of the same species generally have characteristics in common and are recognisably similar to one another. However, defining a species is surprisingly difficult if we only use features like structure, colour or behaviour, because there are variations between individuals in a species. (See Fig 21.2)

Biochemical similarity

Members of the same species have much greater biochemical similarity than members of different species. This is evident in their DNA base sequences and in the sequences of amino acids in their proteins. This idea is discussed in more detail in Chapter 22.

Common gene pool

Another definition of species is based on genetics, and states that the members of a species share a common **gene pool**. This means that a species is the largest unit of population in which genetic exchange is possible. Each species has a separate gene pool and is reproductively isolated from all other species. The concept of the gene pool is discussed in more detail in Chapter 23.

Reproductive isolating mechanisms act to maintain distinct species.



Describe pre-zygotic mechanisms (prevention of zygote formation) including:

- › temporal isolation
- › behavioural isolation
- › mechanical isolation
- › gamete isolation.



Describe post-zygotic mechanisms (prevention of fertile hybrids) including:

- › hybrid inviability
- › hybrid sterility.

Maintaining reproductive isolation

Clearly if populations are not in the same community they cannot interbreed. However, if two species are in the same community they must remain **reproductively isolated** from each other if they are to be separate species. This simply means that members of one species *cannot* interbreed with members of another species to produce fertile offspring.



Fig. 21.2 Variation within species

Reproductive barriers preventing interbreeding of different species are due to the biological features of the organisms and there are two main types of reproductive isolating mechanisms:

- › prezygotic barriers are those that prevent mating or fertilisation, and
- › postzygotic barriers are those that operate after fertilisation and prevent the development of fertile hybrids.

nocturnal – active at night
diurnal – active during the day



Fig. 21.3 Australian Green Tree Frog

Pre-zygotic barriers

Temporal isolation - isolated by time

- › The species may produce gametes in different seasons. Flowering or mating may occur at different times for the different species. This may include one species of plant flowering in autumn and another species flowering in spring
- › Species living in the same place may be nocturnal or diurnal. On a shorter time scale, one species of plant may open its flowers only at night while a similar species that has its flowers open only during the day.

Behavioural isolation

- › The mating recognition behaviour of one species may be sufficiently different as not to interest the other species. Examples include the distinct mating calls of different frog species and the courtship behaviour of various bird species.

Mechanical isolation

- › There may be anatomical differences in the genitals of animals or the floral structures of plants, so that the transfer of gametes from male to female cannot take place.

Gamete isolation

- › This prevents fertilisation even if the gametes are transferred. The male gametes may die due to inappropriate conditions, or the male and female gametes may fail to recognise each other.

Post-zygotic barriers

Hybrid inviability

- Even if mating does occur and the gametes fuse, the zygote or embryo may not develop normally as the genes from the parents are too different.

Hybrid sterility

- One of the ways that hinders cross-breeding is if the two species differ in chromosome type. Even if a hybrid offspring is formed it is sterile because the process of meiosis cannot proceed normally – homologous pairs of chromosomes cannot form. This is the case for the mule, offspring of a mating between horse and donkey. (See Fig. 21.4)

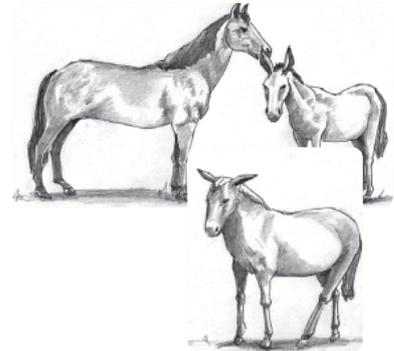


Fig. 21.4 Horse (64 chromosomes), donkey (62 chromosomes), mule (63 chromosomes)

Other barriers to interbreeding

Two species may live in the same broad area, but their habitat preferences within that area may be so different that they don't meet, so they do not interbreed.

Exceptions

Some plant hybrids, formed by interbreeding, have been known to double their chromosome number and give rise to a new species. This has occurred in the evolution of major crop plants such as wheat.

Polyploidy results from an organism having more than two sets of chromosomes.

1. (a) Define the terms community and population.
(b) Why are populations and communities said to be localised?
2. (a) What is the commonly accepted definition of a species?
(b) Describe two other methods that are commonly used for defining a species.
3. Why is the classification of some organisms, like bacteria, into species often difficult?
4. For two groups to be classified as separate species they need to be reproductively isolated from one another.
What is meant by reproductive isolation?
5. Describe the pre-zygotic mechanisms that maintain reproductive isolation.
Provide an example for each mechanism.
6. Describe the post-zygotic mechanisms that maintain reproductive isolation.
Provide an example for each mechanism.
7. If two populations (that is, separate species) of beetle in a community were found to be able to successfully interbreed, how should these populations be classified?

Study Questions

22

Evidence for Evolution



Fig. 22.1 Flinders Ranges Fossil

Evolution

If we consider the current species on Earth and examine the fossil evidence of previous populations carefully, it becomes obvious that there have been significant changes. These changes have taken place gradually over a very long period of time. Biologists use the term **evolution** to describe the changes that have occurred to life on Earth. This process of evolution is widely misunderstood, but it merely explains that the first forms of life were simple and few in number, and that changes have occurred giving rise to an incredible range of complex life forms today.

Mutation is a permanent change in the sequence of DNA nucleotides and is the ultimate source of genetic variation in a species.

Changes in the genetic material occur all the time and these changes can arise either spontaneously, or be induced. Such changes are called **mutations**.

Copying errors when DNA is replicated is an example of a spontaneous mutation. Cells do have a mechanism for correcting such errors, but even so, some may remain. Environmental factors, such as high energy radiation (e.g. X-rays and ultra-violet), mutagenic chemicals, and viruses can induce mutation in DNA. This is discussed in more detail in Chapter 4.

Mutations accumulate over time. If the mutation rate is known, it can be used as a 'clock'.

Over billions of years the cumulative effect of these mutations has been to create a diversity of DNA molecules, and this is reflected in the diversity of life forms on Earth. If it were not for these mutations, there would have been no changes, and life on Earth today would be the same as it was at the very beginning.

When life began, the DNA of the first organisms appears to have been in the form of a circular, double-stranded chromosome. As the chromosomes in these primitive cells became larger they broke up into several linear double strands, with several starting points along their length so they could be duplicated more quickly. DNA was



MOLECULAR CLOCK



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Mutation rate varies from species to species. If the mutation rate is known it can be used as a 'clock'.

also getting bigger, not only because of an increase in the number of genes, but also due to a strange increase in complexity, where large regions of a chromosome came to consist of DNA that was not coding for any product. To illustrate this, we know that in mice the enzyme dihydrofolate reductase is coded in a region of DNA that is 31,000 base pairs long, yet the RNA from this is only 1,600 bases long, and only 558 of these actually code for amino acids. This indicates that only 2 percent of the original DNA is used and the remaining 98 percent of the nucleotides are non-coding (See Fig 22.2). Biologists use the term **exons** for the DNA sequences that are translated into protein, and the sequences that are transcribed but then cut out of the mRNA are called **introns**. You can read more about this in Chapter 2.

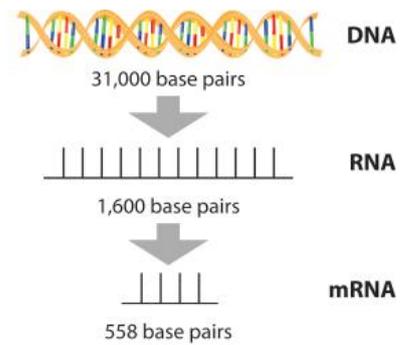


Fig. 22.2 DNA to RNA to mRNA

It has been estimated that each gene in your body will suffer a DNA error at the rate of one per million copies. That sounds like reasonable odds, until you learn that your body will undergo 10^{16} cell divisions. This means that there will be on average 10^{10} mutations of each gene during your lifetime! Exposure to mutagens will increase this number. In most cases these mutations will have no noticeable effect.

However, if the mutations occur in germ cells, they can be passed on to the next generation. Over the 3.7 billion years of life existing on earth, mutations have accumulated and resulted in genetic variation in organisms and the diversity of life on Earth.

MUTATION



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Non-coding DNA has a role in disease. (see Page 18).

In species that reproduces sexually, there are additional sources of genetic variation.



Explain the sources of genetic variation in a species that reproduces sexually

In asexual reproduction, which involves mitotic cell division, the only source of genetic variation is **mutation**. In sexual reproduction, genetic variation results from not only mutation, but also **crossing over** and **independent assortment** during meiosis, and the **random fertilisation** of gametes. (See Chapter 13 for details of these processes.) Thus, there is much greater variation in the products of sexual reproduction than in the products of asexual reproduction.

In an asexually reproducing species the increase in genetic variability in a population is relatively slow, as the direct inheritance of mutations is the only mechanism operating. Individuals that inherit harmful mutations may have reduced reproductive rates or even be unable to reproduce at all. Increase in genetic variability is dramatically sped up by sexual reproduction because new combinations of genes are produced in each generation. In sexual reproduction each gamete is unique, so each offspring that results from fertilisation has a unique combination of alleles.

COMPARATIVE GENOMICS

involves comparing the genomes of species to determine their evolutionary relationships

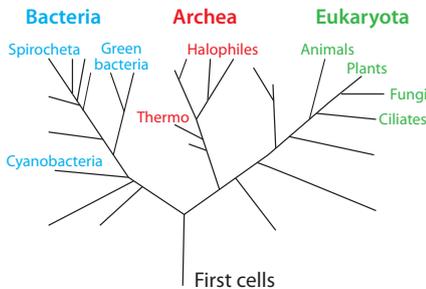


Fig. 22.3 Phylogenetic tree

LUCA



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Comparative genomics provides evidence for evolution and helps establish the likely evolutionary relationship between different species.

In Chapter 1 we introduced the idea that the ‘language of life’ or **genetic code**, as it is called, is the same for all living things. Remember that this code, which is the sequence of bases on the cell’s DNA, uses three bases at a time, called codons, to direct the assembling of proteins from amino acids. Protein synthesis is discussed in detail in chapter 2.

The fact that all cells use the same DNA system indicates that they have a common ancestry. Mutation of DNA is inevitable, and as time goes on, there will be a greater and greater difference from the original DNA due to these mutations.

If two species have evolved from a common ancestor and their separation was recent, it is likely that there will not have been enough time for more than a few new mutations in each species to have taken place. Their DNA sequences should therefore be very similar.

On the other hand, two species that have been separated for a much longer time will probably have many more differences in their nucleotide sequences as a greater number of mutations will have occurred.

Consequently, an organism’s DNA and the products of the DNA — proteins — are a record of its history.

It is the sequence of bases in an organism’s DNA that determines which proteins the organism will produce. We learnt in Chapter 2 that the information on one strand of the DNA is transcribed to messenger RNA, and how the information on the mRNA is translated on the ribosomes, producing a specific protein with a unique sequence of amino acids. Similar DNA will therefore result in similar protein.



Describe how evidence from the following techniques may be used:

- › Sequencing of common proteins (e.g. cytochromes)
- › DNA-DNA hybridisation
- › DNA sequencing, including rRNA gene sequencing in prokaryotes.

Sequencing of common proteins

Sequencing of proteins is used to compare species. One of the first molecules to be used to analyse the evolutionary relationship between species was the protein *cytochrome c*. By comparing the amino acid sequence of cytochrome c from different species we can get an idea of how closely these species are related. The fact that cytochrome c is found in all organisms that respire aerobically makes it ideal for studying evolutionary relationships. A good measure of the accuracy of these molecular tests would be the degree of agreement between conclusions based on molecular comparisons and those based on other observations. Other cytochromes, such as cytochrome oxidase, are also used. So far there has been good agreement between the results based on molecular comparisons and other techniques such as the fossil record and comparative anatomy.

DNA sequencing

As its name implies, **DNA sequencing** involves the determination of the base sequence of a segment of DNA. The sequences from two different species can then be compared directly. Details of the method of DNA sequencing and its uses are provided in Chapter 5.

rRNA gene sequencing in prokaryotes

All prokaryotes (bacteria and archaea) contain a 16S rRNA component in the small subunit of their ribosomes. As it is necessary for protein synthesis, and therefore critical to their survival, the gene that codes for it is highly conserved. This makes analysis of the 16S rRNA gene a useful tool for determining the relationship between prokaryotic species.



Describe the technique of DNA–DNA hybridisation.

Another comparison method is called **DNA–DNA hybridisation**. DNA from one species is heated to separate the complementary strands. Then single stranded DNA from the other species is mixed with the separated strands, and the mixture is cooled. Closely matched strands of DNA will bond more tightly than those that are not well matched. The newly-formed hybrid double helix — one strand from one species and the second strand from the other species — is reheated to see how readily the strands separate. Poorly matched strands separate more easily, that is at a lower temperature, than well matched ones. The DNA strands from closely related species should match well, while those from more distantly related species will be less closely matched. (See Fig. 22.4) The use of DNA–DNA hybridisation in recent years has been used mainly in the study of relationships between species of bacteria. For other organisms, DNA sequencing is generally used.

Edman degradation is a method used to determine the sequence of amino acids in a small protein.

CYTOCHROME C

Cytochrome C is an enzyme involved in respiration. As it is found in both bacteria and the mitochondria of eukaryotes it is considered the 'universal catalyst'.

See Chapter 5 for details of the Barcode Of Life Database

DNA SEQUENCING



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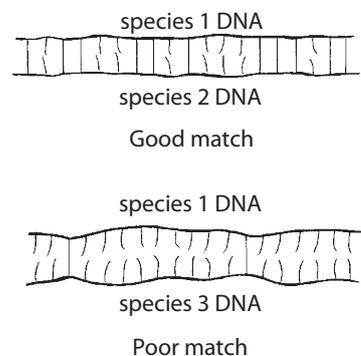


Fig. 22.4 DNA–DNA hybridisation

More closely related species have fewer differences in their DNA sequences and have separated more recently from a common ancestor than distantly related species.

Closely related species will have more similar DNA sequences, while those that are more distantly related will have less similar DNA sequences. This is because if two species have evolved from a common ancestor and their separation was recent, it is likely that there will not have been enough time for more than a few new mutations in each species to have taken place. Their DNA sequences should therefore be very similar. On the other hand, two species that have been separated for a much longer time will probably have many more differences in their nucleotide sequences as a greater number of mutations will have occurred.



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Molecular Evolution

One of the most exciting and interesting developments in modern evolutionary theory is that it is now possible to look for evolutionary clues at the **molecular level**. In Darwin's lifetime it was only possible to examine the outwardly visible characteristics of individuals in populations. Today we can describe evolution in terms of a change in the gene pool of a population. One obvious cause of this change is **mutation**, which we have previously defined as a spontaneous or induced change in the genetic material.

Biologists are able to construct evolutionary trees by comparing the sequence of nucleotides of corresponding genes of different species. The amino acid sequence of corresponding proteins can also be used. These evolutionary trees match very closely the ones made on the basis of outwardly visible characteristics. In the next section we will discuss how it is possible to detect similar sequences of DNA.

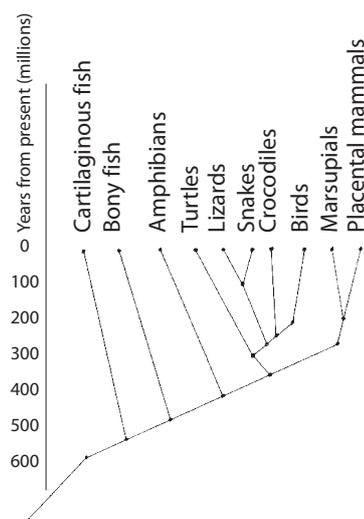


Fig. 22.5 Phylogenetic tree of vertebrates established using DNA sequencing

Phylogenetic tree diagrams represent evolutionary relationships.

Evolutionary relationships that have been inferred (for example, by comparing DNA sequences) can be represented in a diagram called a **phylogenetic tree**, which shows the divergence of species from a common ancestor over a period of time. (See Fig. 22.5), which shows the evolution of a number of vertebrate groups from a common ancestor over more than 500 million years.) The start of the main trunk represents the original ancestor, and as you move towards the tips of the branches you are moving forward in time towards the present. The points at which the branches separate (called "nodes") indicate the formation of new species from a common ancestor.



Draw and analyse simple phylogenetic tree diagrams to represent evolutionary relationships.

Fig. 22.5 shows that cartilaginous fish, like sharks, separated from the common ancestor about 500 million years ago. Marsupials and placental mammals shared a common ancestor 200 million years ago. They are more closely related to each other than to cartilaginous fish.

1. DNA has diversified over billions of years.
 - (a) Describe the changes in DNA that have resulted in its diversity.
 - (b) What evidence reflects the fact that DNA has diversified?
2. Name three factors that have caused DNA to change, and three features of DNA which indicate that it has diversified over time.
3. Explain the main cause of DNA increasing in size over time, apart from the increase in number of genes.
4. List the sources of genetic variation in species that reproduce sexually.
5. Explain how the examination of the structure of DNA and proteins has contributed to the evidence that all life on earth had the same origin.
6. Comparative genomics provides evidence for evolution and evolutionary relationships between species.
Describe each of the following techniques used for obtaining this evidence:
 - sequencing of common proteins (cytochromes)
 - DNA-DNA hybridisation
 - DNA sequencing.
7. Explain why evidence from amino acid sequencing of proteins can be used to support the evidence for evolution provided by DNA analysis.
8. When comparing the degree of similarity between the DNA of different species, what does it indicate if (i) there is a strong degree of similarity between the DNA of different species, or (ii) there is a poor degree of similarity between the DNA of different species?
9. Draw phylogenetic tree, and explain how it can be used to represent evolutionary relationships.

Study Questions

23

Gene Pools and Natural Selection

INCREASE IN NUMBERS

All living things can produce more offspring in their lives than mere replacement numbers. Charles Darwin wrestled with the mathematics of elephants' fertility, this being the slowest reproducing species he could think of. Nevertheless he was able to show that, even if we started with only two elephants and had six young from each pair, in a few generations the world would be awash with elephants.

'The elephant is reckoned to be the slowest breeder of all known animals, and I have taken some pains to estimate its probable minimum rate of increase: it will be under the mark to assume that it breeds when thirty years old, and goes on breeding till ninety years old, bringing forth three pairs of young in this interval; if this be so, at the end of the fifth century there would be alive fifteen million elephants, descended from the first pair.'

Charles Darwin
On the Origin of Species 1859.

Most people are familiar with the idea that members of the same family often share similar characteristics, such as facial features. It would seem obvious that all organisms must inherit 'information' from their parents — or parent in the case of asexual reproduction — in order to be able to carry out all the processes that we call 'living'. However we also know that, with the exception of identical twins, triplets etc., there are usually easily observable differences between brothers and sisters. It is equally obvious that each individual inherits slightly different versions of this information and that this is what gives rise to the great variation within a species such as humans.

All species have the capacity to increase in numbers from one generation to the next. Some produce so many offspring that if most of them survived it would seriously unbalance the workings of the community. Normally, not all offspring survive to reproduce. When we look for examples to illustrate the workings of natural selection, we often point to one trait that might be significant, such as camouflage colouration in *Biston betularia* moths, or running speed in antelope. The reason that there are differences between individuals is that they have inherited different genes.

A gene pool comprises all the genetic information in a population.

As we have already seen, a useful way of defining a species is to refer to the **gene pool**. This term is also used when referring to populations of individuals. Most genes have alternative forms, called **alleles**. An example of this is the ABO blood group gene described in Chapter 4. This gene has three alleles, I^A, I^B, and i. The gene pool of a population consists of all the alleles of all of the genes of all the individuals in the population.

Not all offspring will survive to reproduce

In 1798, Thomas Malthus wrote 'An Essay on the Principle of Population', a controversial paper that initiated the ground-breaking work of Charles Darwin and Alfred Wallace on natural selection. In his paper, Malthus showed that even elephants, the slowest reproducers of the animal world, could overrun the Earth in a matter of a thousand years if left to reproduce unchecked. Clearly, this has not occurred. We had two introduced species in Australia that, for a time, did

reproduce unchecked. The rabbit population reached plague proportions in the south, and the plant pest, prickly pear, (See Fig 23.1) invaded huge tracts of land in Queensland.

These two examples are unusual, as most populations remain fairly constant from one year to the next. For the moment, both the rabbit and prickly pear populations are reduced and stable.

The reason that natural populations do not increase is that not all offspring will survive to reproduce. The range of agents that culls populations includes diseases, predators, competition between members of the same species, and environmental factors such as temperature, food and water availability, and shelter.

Since offspring that are the result of sexual reproduction are genetically different, it stands to reason that some will be better suited to their environment and some will have a better chance of survival than others. They will therefore have an increased chance of reproducing and this will have an effect on the genetic makeup of the next generation. Genes that confer some kind of advantage on an organism will be more likely to be passed on to the next generation than those that are harmful. This concept forms the basis for the process of **natural selection**.

Whether or not a particular gene increases the chances of survival and reproduction of an organism depends to a certain extent on the environmental conditions at the time. For example, in many tropical regions of the world, malaria has been a serious problem. This is a blood disease that is caused by a unicellular parasite and spread by mosquitoes. Individuals in these regions who inherit a single allele for a defective red blood cell, called 'sickle cell', and one normal haemoglobin allele, seem to have increased resistance to malaria. (See text box.) Anyone who is unfortunate enough to inherit two alleles for this condition will usually die in early childhood, unless they are treated. People with two normal alleles for red blood cells are much more susceptible to malaria, which is potentially fatal. In these tropical regions, having one sickle cell allele and one normal haemoglobin gene increases an individual's chances of survival and reproduction. However, in regions where malaria is not a threat, there is no advantage in having this allele – in fact it is better to have two normal alleles. The argument about whether a genetic characteristic does increase the chances of survival and reproduction depends very much on the set of conditions present at the time.

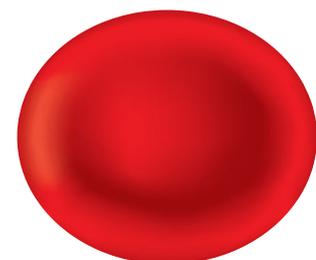
By considering the rabbit, a pest introduced into Australia, we can see how inherited characteristics improve an individual's chances of survival and reproduction. In the 1950s, the viral disease myxomatosis was introduced in attempt to reduce the plague of rabbits. Initially, the disease killed almost 99 percent of the rabbits in an area. Of the surviving one percent, some may not have come into contact with



Fig. 23.1 Prickly pear
Photo: Stan Shebs, CC BY-SA 3.0, Wikipedia

SICKLE CELL ANAEMIA

In the human populations in tropical areas there are several alleles for haemoglobin gene. The normal allele gives rise to normal haemoglobin and normal red blood cells. The abnormal alleles are recessive and tend to produce faulty haemoglobin and defective red blood cells. One of the conditions caused by the abnormal haemoglobin alleles is sickle cell anaemia. (See Fig. 23.2)



Normal Cell



Sickle Cell

Fig. 23.2 Red blood cells

CALICI VIRUS

The most recent biological control weapon against rabbits is the calici virus. This has a high kill rate, but not 100 percent. The rabbits that survive the virus probably have a natural inherited resistance and will pass this on to their offspring.

The word 'adapted' causes much confusion. Individuals that are well adapted to their environment have favourable characteristics that are inherited. An organism cannot acquire adaptations during its lifetime – it is either well adapted at the start of its life or it never will be. Populations, on the other hand, may become better adapted to their environment if the proportion of well adapted individuals increases. This process usually occurs gradually over many generations and is referred to as evolution.

the disease, while others may have been naturally resistant to the original strain of the virus. Those that had natural resistance survived to reproduce, and they tended to pass on their genes for resistance. In 1964 only 40 percent of those tested were killed. The remaining 60 percent had inherited the resistant characteristic from their resistant ancestors. Over many generations of rabbits, the proportion of resistant rabbits in the population had increased.

Natural Selection

Natural selection is a process in which organisms that are better adapted to their environment are more likely to survive and produce offspring.

Darwin used the ideas of natural selection to explain the theory of evolution as follows:

- › Within a population there is genetic variation between individuals.
- › Some individuals are therefore better suited (adapted) than others to the biotic and abiotic factors in the environment.
- › These individuals are more likely to survive longer and produce more offspring than the less well adapted ones. In particular, this applies if there are limited resources.
- › They will tend to pass genes for their favourable characteristics on to their offspring.
- › Over many generations the proportion of individuals in the population with the favourable genes will increase and eventually be maintained.

It is important to note that the changes described refer to changes in populations, not changes in individuals. Also, it should be clear that only inheritable characteristics are subject to this process of natural selection.



Recognise that a large gene pool indicates considerable genetic diversity and is found in populations that are more likely to survive selection pressures.

As you know, the gene pool of a population consists of all the alleles of all of the genes of all the individuals in the population. Thus, a population with a range of different alleles has a 'large' gene pool. Every time a mutation occurs, the gene pool increases. The gene pool of a population provides the genetic raw material for future generations.

Populations that have a limited, or *small*, gene pool will be more



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susceptible to selection pressures such as the introduction of disease, or a change in other environmental conditions. Due to the limited genetic variation in such populations, either most individuals will survive the selection pressures or most will not.

On the other hand, a population with a more diverse (large) gene pool is more likely to have at least some individuals with a genetic makeup that can survive selection pressures. The survival of these individuals will ensure the survival of the population.



Fig. 23.3 Galapagos Islands

CHARLES DARWIN — THE FOUNDER OF NATURAL SELECTION

Historically, Charles Darwin has been given much of the credit for our modern understanding of the process of organic evolution and the mechanism of natural selection. The term 'organic' is used to specify living things. The concept of evolution as a gradual process of progressive change is also used in other areas, such as the evolution of rivers, mountains, or even cultures. In 1831 Darwin sailed on the *Beagle* as the ship's naturalist. He was particularly fascinated by the distribution of finches on the Galapagos Islands (See Fig. 23.3) to the west of South America. Darwin found a number of different species of finch and he noticed that each species seemed to be restricted to one island or a small number of neighbouring islands. Differences between the species included such features as beak shape which seemed to be suited to the food available to the particular species on its island. From these observations Darwin began to formulate an idea of how the different species of Galapagos finches could have developed in such a way as to ensure that each species was well suited (adapted) to its own environmental conditions. (See Fig. 23.4)

In the 1840s Darwin proposed the process of natural selection to explain how evolution occurs. Interestingly, his book *On the Origin of Species* was not published until 1859. Apparently Darwin had been concerned that there might be adverse reaction to his theory, and he postponed its publication until another investigator, Alfred Wallace, independently arrived at almost the same conclusion as Darwin had almost two decades earlier! As you probably know, Darwin's and Wallace's views were not universally accepted. Even today, some people are reluctant to accept the elegance of modern evolutionary theory, despite the overwhelming amount of scientific evidence in support of it.

Nowadays it is possible to look for evolutionary clues at the molecular level, whereas in Darwin's lifetime it was only possible to examine the outwardly visible characteristics of individuals in populations. Today we can describe evolution in terms of a change in the gene pool of a population. One obvious cause is mutation, which we have previously defined as a spontaneous or induced change in the genetic material. If individuals with particular genes are removed from or added to the population then the frequency of those particular genes in the population will change. At the same time the frequency of other genes in the population will alter. A simple cause of a change in the gene pool of a population would be the migration of individuals into or out of the population. Natural selection may also result in a change in the gene pool of a population. This is because certain individuals with favourable genes are more likely to survive than others. As early as 1798 Thomas Malthus had recognised that populations (including human) tend to grow at a faster rate than food and other essential resources. He reasoned that as a result, not all individuals within a population would survive — in other words, there would usually be a struggle for survival.

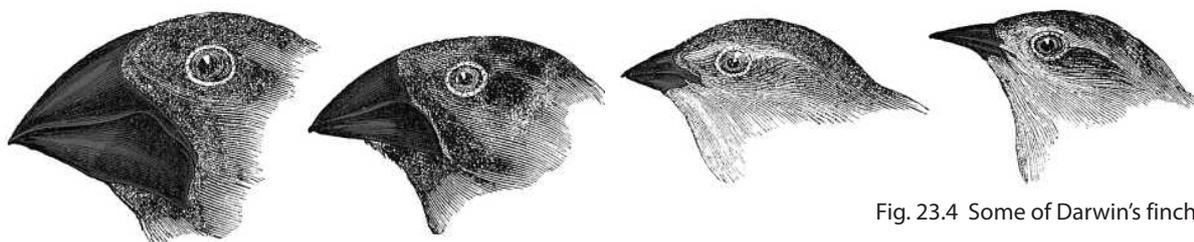


Fig. 23.4 Some of Darwin's finches

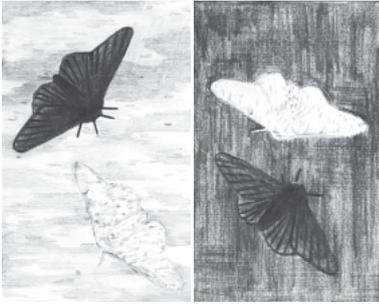


Fig. 23.5 Peppered moths, *Biston betularia*, before (left) and after the Industrial Revolution



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NATURAL SELECTION IN PEPPERED MOTHS

One of the classic examples of natural selection involves the peppered moth *Biston betularia*. This moth is found in England in two main shades. One is light with patches of darkness (hence the name 'peppered') and the other is dark in colour. The moths have a tendency to settle on tree trunks that have a covering of lichen. This provides the moths with camouflage from their predators. Before the Industrial Revolution almost all the moths were light in colour and dark ones were extremely rare. However, during the Industrial Revolution in the late nineteenth century, trees and buildings in the environment became darkened by black smoke from chimneys. As a result of this, pale moths that settled on tree trunks were easily spotted by predators such as birds, while darker moths were camouflaged. (See Fig. 23.5)

Over many generations the proportion of darker moths increased and the proportion of paler ones decreased. Since much of the pollution has recently been cleaned up there has been a reversion towards the proportions of pale and dark moths that existed before the Industrial Revolution.

A careless observer might conclude that the pollution caused the appearance of darker coloured moths. In fact nothing could be further from the truth. There was a small proportion of darker coloured moths in the population before the Industrial Revolution, and it was simply this proportion that changed. No individual moth changed colour.

A change that occurs during an individual's lifetime is called an acquired characteristic and there is no evidence to suggest that acquired characteristics can be inherited.

In the case of the peppered moth, the abiotic factor was the change in colour on the trees due to pollution, and the biotic factor was predation by birds.

HYPOTHESIS, LAW, OR THEORY?

Often these terms are used interchangeably and incorrectly.

A hypothesis is an idea or suggestion that can be tested. It may have little or no evidence to support it.

A law describes an observed phenomenon, but does not attempt to explain it or its cause. Examples are the law of conservation of momentum, the law of thermodynamics, and the law of gravity.

A theory is substantiated by observation, experiment, and evidence, usually over a long time-frame. Examples of scientific theories include atomic theory, cell theory, theory of relativity, and the theory of evolution. **A theory is not the same as a hypothesis.** When someone says "I have a theory about...", they probably mean "I have a hypothesis about...". Also, it is not true that 'theories become laws'.

A scientific theory is the best available explanation of how or why something happens, and is often based on many **supported** hypotheses. A scientific law attempts to predict what will happen, but it does not attempt to explain how or why it happens. Hypotheses, laws, and theories are subject to change or rejection on the basis of reliable and repeatable **evidence** that does not support them. This is a distinguishing feature of **scientific** knowledge.

GREGOR MENDEL — THE 'FATHER OF GENETICS'



In the 1860s an Austrian monk, Gregor Mendel investigated the inheritance of characteristics in the garden pea. He produced large numbers of pea seeds and by allowing them to grow into plants (offspring) and then analysing his results quantitatively, he was able to put forward explanations of how inheritable characteristics (traits) are passed from one generation to the next. These principles of inheritance remain valid today.

Mendel explained the inheritance of what we now call 'genes' in the following way:

- Firstly, he postulated that these inheritable factors (or genes) have alternative forms. We now know that the gene for seed shape occurs in two alternative forms, one for round seed shape and one for wrinkled. Alternative forms of a gene for a particular characteristic are called alleles.
- Secondly, he suggested that each individual has two 'factors' for each characteristic, one inherited from each parent.
- Thirdly, Mendel thought that gametes each contain only one 'factor' for each characteristic. That is, the pairs of factors are segregated (separated) during gamete formation. Then, when fertilisation occurs and two gametes fuse, the pairs will be restored in the zygote, with one member of each pair having come from each gamete (or parent).
- Finally, Mendel stated that if the two factors of a pair (which we now call 'alleles') are different, then one was expressed, and the other was masked. We call these 'dominant' and 'recessive' respectively.

Mendel's law (called the law of segregation) explains that, although each individual contains two factors (alleles) for each inheritable characteristic, the gametes will only contain one. This is because the pairs of factors separate (segregate) during gamete formation. Fortunately, this also ties in with our understanding of the cellular events that occur during gamete formation. Remember that gametes are formed by meiosis, and that each daughter cell contains only one chromosome from each homologous pair. It seems that the behaviour of inheritable 'factors' as proposed by Mendel matches closely the behaviour of chromosomes during meiosis. We now know that the reason for this correlation is that genes are located on chromosomes. Chromosomes are made of enormous lengths of DNA bound with protein, and a gene is a segment of DNA. A diploid human cell, for example, has only 46 chromosomes, but these carry information for many thousands of inheritable characteristics. Each of these is controlled by a different gene. Amazingly, the significance of Mendel's painstaking work and results was not realised until long after his death.



Explain how natural selection results in evolution by causing a change in the frequency of alleles in a population.

Consider how the selection process occurs. The environmental conditions determine which characteristics are favourable and hence which individuals will survive to reproductive age. This, in turn, determines which genes will be passed on to the next generation. Gradually, the genetic makeup of the population will change as the proportion of favourable genes in the population increases. This is what we mean when we say that evolution involves changes in the gene pool, that is, a change in the frequency of alleles.



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Evolutionary changes are affected by other factors besides selection, including:

- › sexual reproduction
- › genetic drift.

Evolution can be thought of as a change in the gene pool of a population. Natural selection is the main driver of such change. Other factors that can bring about a change in the gene pool include sexual reproduction and genetic drift, which result in changes in gene frequencies.

Sexual reproduction

We have seen how the events of meiosis ensure that each gamete is unique. The random processes of crossing over during prophase I and independent assortment during metaphase I result in new combinations of genes being formed. Furthermore, the process of fertilisation, in which two gametes fuse to produce a new offspring,

SUPERBUGS – NATURAL SELECTION IN BACTERIA

A bacterial colony is made up of millions of bacterial cells. All of these bacterial cells are the result of repeated cell division, and they have come from a single parent cell. Because these cells are formed by binary fission they should all be genetically identical. However, in any bacterial colony there will usually be several mutant cells present. There is variation between individuals. Each bacterial cell is capable of binary fission in the right circumstances, and any new mutations will be passed on to the next generation.

Even if the bacterial chromosome is not changed, the plasmids may be altered. Plasmids are small loops of DNA in bacteria that are separate from the larger chromosome. Like the chromosome, plasmids contain genetic information that can be replicated and passed on to daughter cells. In addition, plasmids can be transferred from one bacterium to another, carrying their genes with them. One class of plasmids, called R plasmids, are known to carry genes that enable the host bacterial cell to resist antibiotics. The increased use of antibiotics in agriculture and medicine has caused those populations of bacteria without R plasmids to decrease in number and those with R plasmids to increase. An unfortunate result of this is that more and more pathogenic bacteria are now resistant to antibiotics. It is becoming more and more difficult to develop new antibiotics that are effective against these resistant strains of bacteria.

The bacterium *Escherichia coli* (*E. coli*) is very susceptible to the antibiotic streptomycin, which is the product of a soil-living organism, *Streptomyces griseus*. Since *E. coli* is an inhabitant of the digestive tract of animals, *E. coli* and *S. griseus* would seldom meet and so there would seem to be little advantage to *E. coli* in having resistance genes for the antibiotic streptomycin. Very rarely, about one in a billion cell generations, a mutation arises spontaneously in *E. coli*, which makes it resistant to streptomycin. If there is no streptomycin present, this gene may be disadvantageous if it means that the bacteria with the gene reproduce more slowly than the bacteria without the gene. However, in the presence of streptomycin, the non-resistant cells will be inhibited or killed, and the next generation of *E. coli* will mainly consist of descendants of the original resistant cell. The presence of streptomycin means that resistant bacteria are selected for and non-resistant bacteria are selected against. Thus a resistant strain of *E. coli* can develop as a result of natural selection acting on variation in the population, causing a change in the gene frequency.

This is an excellent example of **selection pressure** bringing about a change in a population over several generations. Note that streptomycin does not cause the resistance. Rather, it has selected those individual bacteria with chance mutations for resistance and they have passed this genetic characteristic on to their offspring.

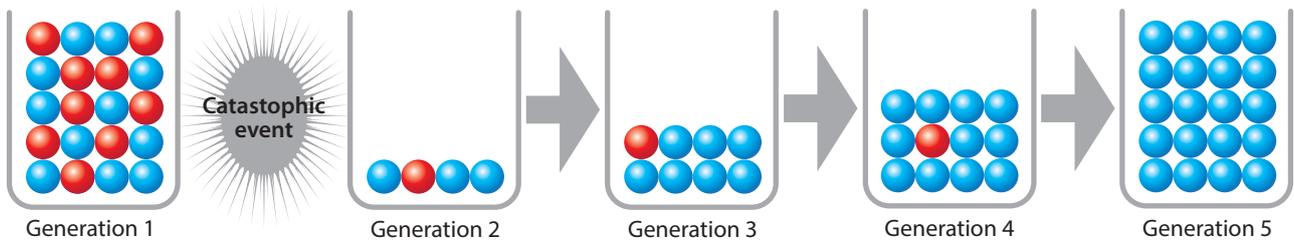


Fig. 23.6 Genetic drift

is also a random one. Chance determines which egg develops and which sperm cell will fertilise it. The offspring resulting from meiosis and fertilisation, the key cellular events in sexual reproduction, will be genetically different from one another, even if they have the same parents. The obvious exception to this is the formation of identical twins or triplets.

Genetic drift

Genetic drift (See Fig. 23.6) refers to changes in the frequencies of alleles in a population due to chance events, such as catastrophes. It is usually confined to small populations, as it is more likely that an allele can be completely removed from the gene pool by the death of a small number of individuals. This decreases the gene pool and results in evolution.



tinyurl.com/24r97tpn

1. List four factors that will limit the size of a natural population of possums in a forest.
2. The term gene pool can be used when we talk about species or populations. Define the term 'gene pool'.
With reference to both species and populations show how the term 'gene pool' is used.
3. (a) Some genes can confer some kind of 'advantage' on an organism. Explain the biological meaning of the word 'advantage' in the first sentence.
(b) The gene for sickle cell can confer an advantage in some situations but not others. Explain this observation.
4. Outline the sequence of events that occurred when a population of rabbits came into contact with the disease myxomatosis for the first time, and resulted an increase in the proportion of individuals resistant to it.
5. Explain how natural selection results in evolution by causing a change in the frequency of alleles in a population.
6. Why is a population with a large gene pool more likely to survive the introduction of a new predator or disease than a population with a small gene pool?
7. What is meant by 'genetic drift' when referring to the genes in a population?

Study Questions

24

Speciation and Evolution

Speciation may result from an accumulation of genetic changes influenced by different selection pressures or genetic drift in geographically isolated populations.



Describe the process of allopatric speciation.

We have stated earlier that a commonly accepted definition of a species is that it is a group of organisms that breed or would be able to breed if they were ever to meet, and which produce fertile offspring. Members of the same species share a common gene pool.

We tend to be very unaware of slow processes happening around us. It is only in hindsight that we realise that the decade of the 1950s was unusually cool or that of the 1970s unusually wet. Still less can we appreciate the slow movement north of the Australian landmass due to continental drift, and we cannot hope to personally experience the transition from our current climate to a deep ice age, as this will take thousands of years. Yet these changes are relatively fast in terms of evolution. The development of a new species is a process which occurs so slowly that it is also imperceptible.

We know that natural selection alters the characteristics of a population over many generations. An extreme result of the effects of natural selection would be the evolution of two or more species from one. This process is called **speciation**.

Allopatric Speciation

The process of forming new species from populations separated by geographical barriers is called **allopatric** speciation. The word 'allopatric' is derived from Latin and means 'different place'. Geographical isolation by itself does not lead to speciation. It is only when changes to the gene pools result in reproductive isolation that this occurs.

Initially, a single population of individuals is somehow split into two or more separate populations. A typical cause of this would be a newly formed geographical barrier, such as a valley, a river, a desert, a mountain or an ocean. Over thousands of years, sea levels rise and

fall, land bridges open and close, islands drift and continents grind away at each other. If the environmental conditions for the separated populations were different, then different features would be selected in each population. Provided that these populations remained geographically isolated, and there was no gene flow between them, then over many generations they could become so genetically different that even if they were brought together they would no longer be able to interbreed and produce fertile offspring. The populations would have become reproductively isolated. (See Fig. 24.1) Thus, two or more species would have developed from one. This idea of reproductive isolation was introduced in Chapter 21.

Darwin's Finches

A classic example of speciation is provided by the finches that Charles Darwin studied on the Galapagos Islands. The evidence suggests that millions of years ago the islands would have been one single landmass with only one species of finch living there. Parts of this landmass later became separated due to a rise in the sea level and continental drift, and this formed the islands. The finch populations on each island became isolated. As the finches are not strong fliers and do not travel across expanses of water there was no gene flow between the separate populations. The environmental conditions on each island became significantly different from the others, and these unique abiotic factors acted as selecting agents on the finches. Eventually the finches from each island were different from one another and the sea between the islands prevented interbreeding between the populations.

In addition to different abiotic factors influencing the gene pool, any new mutations that entered the population probably would not have occurred on the other islands, and so the populations on each island became even more genetically different.

Over time, the differences between the finches from each island became so great that, even if they did come into contact, they could no longer interbreed and produce fertile offspring. They had become separate species, derived from the one ancestral species.

In some cases, the finches were able to recolonise islands and coexist with other species, with reproductive barriers now keeping the new species distinct.

Comparison of allopatric and sympatric speciation.

Sympatric means 'same place'. Thus, **sympatric speciation** refers to the evolution of two species living in the same geographical area. It can be difficult to tell whether two species in the same geographical location have evolved in this way. In fact, some scientists still treat the idea of sympatric speciation with caution. Examples of animal



Western Rosella



Eastern Rosella

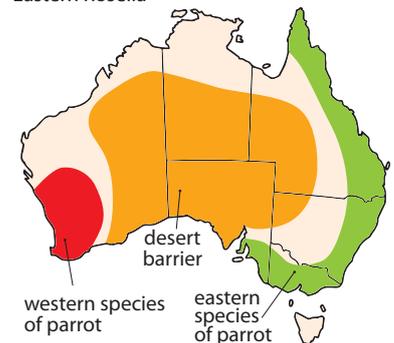


Fig. 24.1 An example of a geographical barrier – a desert. Showing the Western Rosella and the Eastern Rosella

SYMPATRIC SPECIATION

The formation of two species from a common ancestor while living in the same area.

Note: Sympatric speciation not required from 2023.

sympatric speciation are rare and controversial. It seems that sympatric speciation is more common in plants and bacteria than in animals. In the case of plants, this is because new species can form as a result of polyploidy or hybridisation.



Fig. 24.2 Wheat

Hybridisation results from the fusion of cells from two different species that gives rise to an organism with chromosomes from both species. If this occurs in animals, the offspring is more than likely sterile, so a new species has not been formed. In Chapter 21 we used the mule as an example of hybrid sterility. The chromosomes of the hybrid are not homologous, and cannot pair during meiosis. However, in plants, hybridisation can give rise to a sterile organism that may reproduce asexually, giving rise to a new species.

Alternatively, the plant hybrid may undergo polyploidy.

Polyploidy results from an organism having more than two sets of chromosomes. This can occur if chromosomes do not separate after duplication in mitosis. This can produce an individual that is fertile, as it will have homologous chromosomes that can pair during meiosis. In this way, a new sexually reproducing plant species can be formed. The polyploid cell may also undergo mitosis and produce new individuals through asexual reproduction. This is relatively common in plants, such as wheat. (See Fig 24.2)

Put simply, allopatric speciation requires a geographical barrier, whereas sympatric speciation does not. Natural selection plays a major role in allopatric speciation, but its role in sympatric speciation is questionable.

Similar selection pressures on unrelated species may lead to convergent evolution.

In Chapter 23 we saw that selection pressures can affect the survival of populations, and that populations with a large gene pool are more likely to survive selection pressures, as they are more likely to contain individuals with favourable characteristics. Now let us consider two widely separated, unrelated populations in very similar environments. If these two populations occupy the same ecological niches then the selection pressures will be very similar, and this means that in each location individuals with similar features will be selected. It would not be surprising to find that the features of these two populations come to resemble one another. The process that leads to quite unrelated populations resembling one another is called **convergent evolution**. (See Fig 24.3a)

Australian marsupials and North American placental mammals have many examples of unrelated species that have very similar appearance and characteristics. (See Fig 24.3b)



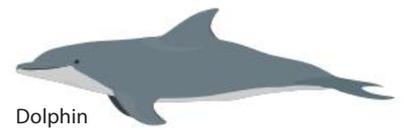
Recognise and give examples of convergent evolution.

Consider the superficial features of dolphins and sharks. Although these two organisms are not closely related, they clearly have similarities in their structure, and this can be attributed to the fact that they live under similar conditions and have been subjected to similar selection pressures. Even though both are streamlined, have a similar colouring, and have flippers and fins, the dolphin is more closely related to the hippopotamus than it is to the shark!

Other examples of convergent evolution include echidnas and hedgehogs, possums and squirrels, and platypuses and beavers. (See Fig 24.3)

Although plants of the deserts of Africa and the Americas are not closely related, they have developed similar fleshy structures for retaining water.

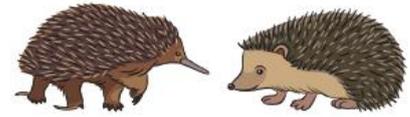
The result of convergent evolution are also apparent in many independently evolved structures that serve a similar purpose. One of the most interesting examples is the evolution of eyes. The camera-style eyes of cephalopods and vertebrates have evolved independently but are very similar. Remarkably, the cephalopod (squid, octopus) eye has no blindspot, so is even superior to our eyes! (See Fig 24.4) Another example is the wings of birds, bats, and insects that, although they have evolved separately and have different structural features, all serve the same purpose.



Dolphin



Shark



Echidna

Hedgehog



Platypus

Beaver

Fig. 24.3b Examples of convergent evolution



Fig. 24.4 Squid

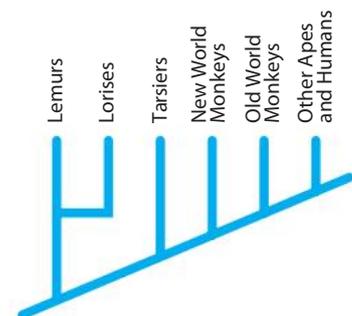
Different selection pressures may lead to divergent evolution or adaptive radiation.



Recognise and give examples of divergent evolution and adaptive radiation.

Divergent evolution involves the slow and gradual accumulation of new characteristics, culminating in the formation of new species. A common example is the evolution of primates. (See Fig. 24.4a)

Adaptive radiation refers to a special case of divergent evolution in which there is a sudden emergence of new species from a common ancestor. Note that 'sudden' means on the geological time-scale, which could mean several hundred thousand years! Compared to the age of the Earth (about 4.5 billion years), that is 'sudden'.



Early Primates

Fig. 24.4a The evolution of primates

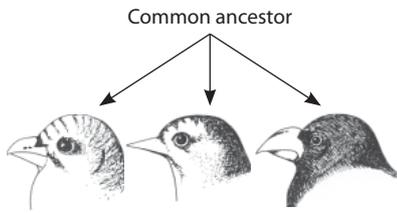


Fig. 24.5 Galapagos Island finches - each beak shape is suited to a different type of food

Examples of adaptive radiation include Darwin's finches (see Chapter 23) and the adaptive radiation of marsupials in Australia. Adaptive radiation is usually due to a change in the environment or organisms relocating to a new environment and experiencing different selection pressures. Such selection pressures could include different food sources, different predators, and different environmental conditions. The Galapagos Islands provide a clear example of this. (See Fig. 24.5)

Succession is the gradual change in the mix of species over time, following a disturbance.

Communities are continually undergoing change

We often think that our environment is constant and we do not expect to see it change much during our lifetime. While we can accept that the 1600 metre-deep Grand Canyon was formed by erosion due to flowing water, we would not anticipate that it will deepen perceptibly from year to year. Based on the idea of continental drift, we can also accept that Australia is moving northward at 7 cm per year, but we don't expect to see rainforest surrounding Adelaide just yet. However, there are major disturbances from time to time that do alter the local environment quite markedly, and so change the community that lives there.

The devastating Mount St Helens volcanic eruption in Washington State USA in 1980 wiped out huge areas of forest and left them barren for many years. (See Fig. 24.6) Several recent movies have brought home to us the possibility of an asteroid hitting our planet, and we have evidence that such major events have occurred in the past and have had a profound effect on biological communities. In Australia, natural vegetation is often subjected to large-scale bushfires which alter the landscape markedly. Although they are able to recover, the communities do undergo change as a result of these bushfires.



Fig. 24.6 Mount St Helens

Human activities also lead to changes in communities, and these are discussed in Chapter 25.



Describe the processes of primary and secondary succession.

Succession

If you travel from Adelaide to the south-east of South Australia, you pass through the town of Keith which is more than 50 kilometres from the coast. The country around Keith consists of the remnants of ancient sea dunes that were formed thousands of years ago. These dunes do not have vegetation like present day shore dunes, but are now supporting a rich and varied heathland flora. Geological events

such as the rise and fall of sea levels show us that major areas of land can become available for colonisation, and that the community that grows there after several hundred years is not the same as the one you would find after only a few years. This change in the communities in an area over time is called **succession**. (See Fig. 24.7)

Studies of succession of vegetation date from early in the 19th century. Initially it was thought that change was progressive and predictable. Now we are not so sure. We have retained some of the original terminology, such as the first plants on bare surfaces being termed colonisers or pioneers, followed by an immature phase and then by a mature or stable phase, the climax community. (See Fig. 24.8)

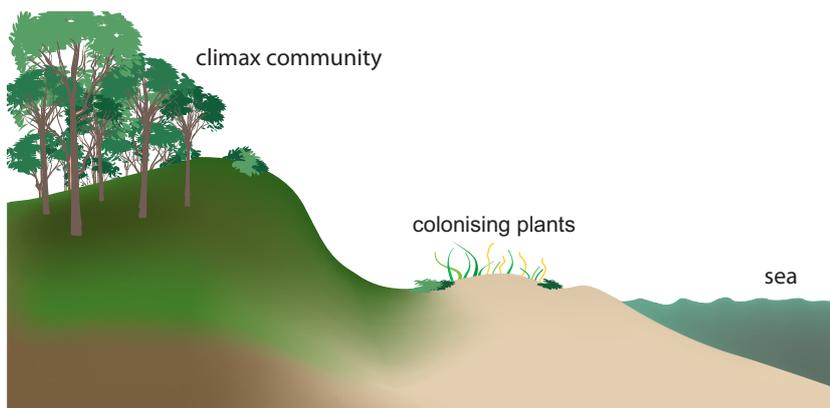


Fig. 24.8 Sand dune zonation

Certainly from completely bare ground there is a succession of vegetation. Ground may have been cleared by eruption of a volcano, the retreat of a glacier or even the retreat of a sea. The debris of a volcano can take many forms and cover large areas. Taupo, in New Zealand, is thought to have provided one of the largest-ever volcanic eruptions on Earth about 1800 years ago. It deposited 2,000 km² of compacted ash and this covered neighbouring communities. Since that time there has been a succession of communities in the region. Hawaii exists because submarine volcanoes built cones from the bottom of the ocean to above sea level. Initially they were totally barren, but now they are mostly covered with tropical vegetation. In South Australia, the mouth of the Murray river has moved north in recent years, leaving behind bare sand on the southern side to be colonised by successive communities of dune vegetation.

One of the most studied new habitats on Earth is the small volcanic island of Surtsey, which arose out of the sea off the coast of Iceland in 1963. (See Fig. 24.9) Even now plant cover on the island is patchy. The first flowering plant recorded on the island was Sea Rocket which, in 1965, gained a foothold in the decaying seaweed. Sea Rocket is now extinct on the island, but there are 25 other species of flowering plant, together with mosses, insects and small animals. Seabirds now nest on the island and they could have been the means of transport that enabled the other life forms to reach Surtsey. The excrement of the birds is an important source of nutrients for



Fig. 24.7 Sand dune plants



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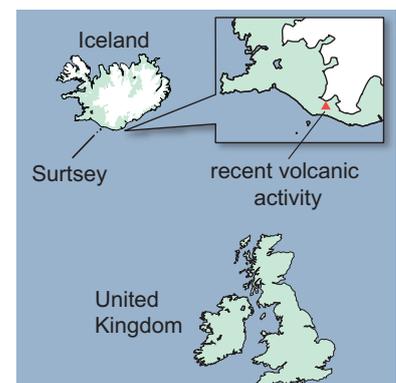


Fig. 24.9 Surtsey

the plants. What has happened is that early colonisers, like the Sea Rocket, have altered the conditions on the island, making them suitable for other organisms. These, in turn, have altered the abiotic and biotic conditions, and this process has been repeated, giving rise to the community that we see today.

An even longer-term study of new habitats has been carried out off the coast of Indonesia. (See Fig. 24.10) In 1883 the volcano Krakatoa blew up, leaving behind four smaller remnant islands on which all life had been destroyed. The islands were covered with a layer of bare, igneous rock. The most important early colonists were blue-green bacteria which formed a sticky mat, in which fern spores could germinate. The full-grown ferns in turn provided a habitat for flowering plants. The importance of these bacteria was that they could fix organic nitrogen from atmospheric nitrogen for the growth of other organisms. Now the nitrogen fixing role is in the hands (or roots) of *Casuarina* trees and legumes.



Fig. 24.10 Krakatoa

A NEARBY DISASTER

On December 26 2004, an undersea earthquake occurred in the Indian Ocean, about 250km off the coast of Sumatra, Indonesia. It measured 9.0 on the Richter scale and caused tsunamis that devastated coastal areas in several countries, and resulted in the loss of more than 250 000 human lives. In addition, much damage was caused to coastal ecosystems by the removal of flora and fauna, smothering of corals with silt, contamination of land with salt water, and eutrophication (see glossary). It will take many years for these ecosystems to re-establish, and some may never be the same.



Fig. 24.11 Krakatoa

On Surtsey we would expect the vegetation in the long term to resemble that of the nearest islands, and the vegetation on the Krakatoa group to resemble that on the mainland of Sumatra, 35 kilometres away. But the plants which will eventually inhabit these islands are not the ones that first grew there. The pioneers often have to establish on bare rock and this environment is very hazardous. The first plants face extreme and fluctuating temperatures, limited water and low nutrient supply. There is no soil to hold water or to cushion the plant against harsh conditions. Lower life forms, such as lichens, blue-green bacteria, and mosses, are the usual pioneers, and they quickly create a microclimate which holds water and dust particles. Some lichens can grow on bare rock, and weather it with acids they produce, thus beginning the long process of soil creation. A complex community soon builds up and by its very existence modifies the environment by making soil and providing dead material for decomposers and detritivores to live on. This type of succession that begins without soil is called **primary succession**.

A second form of succession, **secondary succession**, begins when a disturbance such as fire or flood destroys most or all of the original vegetation, but leaves soil rather than rock behind. Again there may be opportunistic species that can take advantage of the open environment, before a more stable community re-establishes itself. However, if the environment suffers frequent disturbance, the climax community may never be attained. It is important to remember that the community does not form automatically as a single unit, but is the result of species competing against each other for the resources. If the resources change, owing to the presence of the earlier species, then different species which are better adapted to the changed conditions may gradually oust the pioneers and the intermediate species. The types of organism that inhabit early and later stages tend to have rather different kinds of life cycles.

Species or populations that have a reduced genetic diversity have a higher risk of extinction.

As we saw in Chapter 23, populations that have a limited, or *small*, gene pool will have a reduced genetic diversity, and be more susceptible to selection pressures such as the introduction of disease, or a change in other environmental conditions. These populations will therefore be more likely to become extinct.

Genetic drift (see Chapter 23) often results from the 'bottleneck' or 'founder' effect. If only a few members of a small population survive an event, perhaps a catastrophe, they may not have the full range of alleles that was in the original population. The loss of some alleles reduces the genetic diversity of the resulting population and is known as the bottleneck effect. (See Fig 24.13) If a small number of individuals from a population establish a new population, the new population may have much less genetic diversity than the original population. This is called the founder effect.



Give examples of species with low genetic diversity.

Species with low genetic diversity include the Tasmanian devil, the cheetah, and the European bison.

Tasmanian devil

About 12 000 years ago the Tasmanian devil (*Sarcophilus harrisi*) was distributed widely across mainland Australia and Tasmania. (See Fig. 24.12) At this time Tasmania became separated from mainland Australia due to rising sea levels. By about 400 years ago, the Tasmanian devil had disappeared from mainland Australia, possibly due to competition with and predation by dingoes that were introduced about 4000 years ago, possibly even longer. The

HUMANS AND SUCCESSION



Even human beings can cause succession. The Aral Sea, once the world's fourth largest lake, in Uzbekistan has dramatically retreated due to human activities, such as irrigation for cotton, leaving an exposed sea floor. This area is the site of succession, particularly in the south. Efforts are being made to restore the lake in the north.

LICHENS AS PIONEERS

The idea of change in a community is not difficult to imagine. Consider the lichens that live on tree trunks. There is no habitat for them to live on until a tree grows. Even then there are often quite specific host requirements. A tree of one species may show a rich lichen flora while a neighbouring one of another species may remain quite bare. The habitat has to be created by other living things before lichen can appear. Other lichens in arid South Australia play a crucial role in protecting the soil with a living crust. If the hard feet of sheep break through this crust the powdery soil can blow away in the wind and other arid plants are unable to survive on the exposed subsoil. In each of these examples, one living organism creates or protects the conditions necessary for the survival of other species.



Fig. 24.12 Tasmanian devil

resulting small and isolated population in Tasmania was even further reduced in the 19th and 20th century by hunting and eradication by humans. This *greatly reduced the genetic diversity* of the population, meaning that *all Tasmanian devils are now very similar genetically*. This has particular significance because ‘facial tumour disease’, which is fatal, is prevalent in Tasmanian devils, threatening the survival of the species. When cancer cells from one individual are transferred to another by biting, these cells are not recognised as ‘foreign’ by the immune system of the bitten individual, because the individuals are so genetically similar. Thus the cells can continue to multiply in their new ‘host’ and the facial tumour disease is spread.

Cheetah

About 12 000 years ago a mass extinction event occurred, resulting in the loss of most of the world’s large mammals. At this time the size of the population of cheetahs also greatly decreased, leading to a much *reduced genetic diversity* due to the ‘bottleneck’ (See Fig 24.13) effect. In 1900, the cheetah occupied a large habitat over much of Africa, the Middle East, and Asia Minor. Since then, the habitat of the cheetah has contracted to less than a quarter of its previous size, and has become fragmented due to human activity. Today, there are about 7000 individuals in total, with the largest single population being about 1500. Because the populations are geographically isolated, there is little or no gene flow between them. As a result of inbreeding, genetic ‘defects’, such as a kinked tail, dental anomalies, and low sperm quality have become more common. These defects adversely affect the cheetahs’ ability to hunt, feed, and reproduce.

Even if the population size of cheetahs was to increase, it would take thousands of years for its genetic diversity to significantly improve.

European bison

In the 16th century there were more than 100 000 of these large mammals living in northern Europe. By the early 20th century, the population had reduced to a mere 12 individuals. There are now between 4000 and 5000 individuals, but they are descended from only 7 bison. Thus, the current population has an extremely low genetic diversity (small gene pool). In a population that lacks genetic diversity, natural selection has little effect. Any harmful mutations are likely to be widespread and therefore not easily removed, and beneficial alleles could be more easily lost by genetic drift.

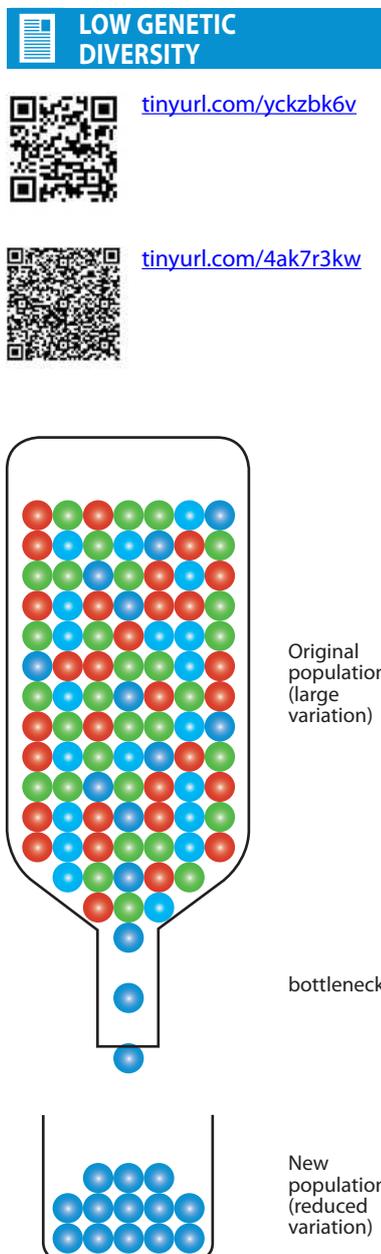


Fig. 24.13 The bottleneck effect

Study Questions

1. The evolution of two or more species from one is called speciation, and it often involves a number of steps, some of which are listed below. Using the finches of the Galapagos Islands as an example, explain each of the following terms and the contribution each makes to the process of speciation:
 - (a) geographical isolation
 - (b) gene flow
 - (c) mutations
 - (d) selection pressures – abiotic and biotic
 - (e) natural selection, and
 - (f) reproductive isolation.
2. What is meant by the term 'allopatric speciation'?
3. Compare allopatric and sympatric speciation.
4. Evolution can occur when the biotic and abiotic factors in a population's environment change.
 - (a) What are the biotic and abiotic factors that would affect the bacterium *E.coli* and contribute to the development of an antibiotic resistant strain?
 - (b) What is meant by the term selection pressure?
5. What conditions are required for convergent evolution to occur? Refer to examples of both plant and animal species in which convergent evolution has occurred.
6. What conditions are required for divergent evolution to occur?
7. Explain the term 'adaptive radiation'. What conditions are required for adaptive radiation to occur?
8. Name examples of a species in which adaptive radiation has occurred.
9.
 - (a) Define the term succession.
 - (b) Distinguish between primary succession and secondary succession.
 - (c) Which organisms usually appear first on barren rock or ground? Why are they so important to the further development of the community?
 - (d) What is a climax community and why are climax communities never seen in some areas?
10. Using an example of a species with low genetic diversity, explain why it has a higher risk of extinction than a species with high genetic diversity.

25

Human Impact

Human Impact in Australia

The arrival of the first humans in Australia about 60,000 years ago led to major changes in the existing communities. The use of fire as a hunting tool affected not just individual species, but large areas of landscape. Frequent, deliberately lit fires favoured plants with hard or minimal leaves (sclerophylls) at the expense of broad-leaved rainforest trees, and consequently the dependent populations of animals also changed.

Changes to the environment can lead to changes in communities by affecting resources such as food, habitat, predators and disease. How can we know if we are having an effect on the environment, and therefore on communities? In some cases the answer is obvious: the arrival of Europeans in the 1780's saw the clearing huge tracts of rainforest, introducing new organisms like the rabbit into Australia, or altering the course and flow of rivers. In other cases, the effects may be widespread but very subtle and we need to look more carefully in order to find evidence for change.

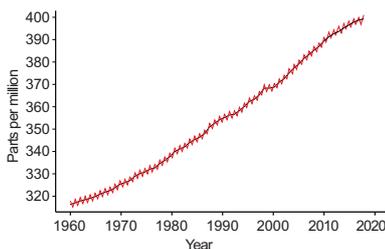


Fig. 25.1 CO₂ levels at Mauna Loa, Hawaii

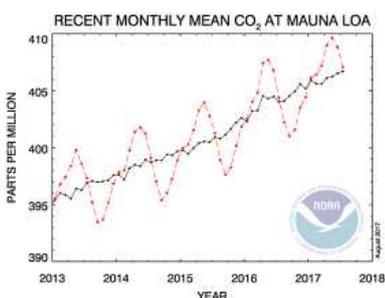


Fig. 25.2 The trend continues

Data/Image provided by NOAA ESRL Global Monitoring Division, Boulder, Colorado, USA (<http://esrl.noaa.gov/gmd/>)



Give examples of human activities that lead to climate or environmental change.

Human Impact on climate

Carbon dioxide levels in the atmosphere have been measured for many years, at sites such as Cape Grim in the south of Tasmania and Mauna Loa in Hawaii, high on an extinct volcano. (See Fig. 25.1) It appears that as a result of increased fuel usage, there is a slow but steady increase in the concentration of carbon dioxide in the atmosphere. (See Fig. 25.2) Most scientists have predicted that this increase will result in an increase in the Earth's average atmospheric temperature, due to what is called the greenhouse effect. If this increase in the atmospheric temperature does occur there will be a rise in sea levels as polar ice melts, and as water in the seas expands. This will cause considerable changes to the communities along the shoreline, as well as changing the world's weather patterns. If this happens, human practice will have had an influence on communities on a global scale.

Human Impact on the environment

At local levels there are numerous examples of human practices causing major changes to a community. In the northern hemisphere, the phenomenon of acid rain, which is the result of humans burning fuels, has caused large numbers of lakes and waterways to become sterile. The acids, primarily sulfuric and nitric, accumulate in the lakes, denaturing proteins in organisms or disrupting essential chemical reactions in cells.

Clearing of land for crops has taken place in most countries where natural communities have been replaced by crops such as wheat, rice, cotton plants, or pasture. In some instances the loss of the natural community has had widespread effects.

In the south west of Australia, the removal of large trees in the jarrah forests caused a rise in the water table in surrounding areas and a subsequent increase in salinity levels. In the tropics, removal of the rainforest has reduced the humidity and this may have had an effect on subsequent rainfall. Also, topsoil from these areas has been washed away, thus decreasing the chance of revegetation.

In coastal regions, removing the mangroves to create harbours or recreation areas has had an influence on communities out in the sea. The mangroves provide a sheltered habitat for the development of the offspring of numerous animal species before they move out to sea. A decrease in the mangroves leads to a decrease in all surrounding communities as the food chains are disrupted.

Large-scale industrial works have brought about changes in communities. The largest artificial lake in Australia, Lake Argyle in north west Western Australia, covers 740 square kilometres of previously natural vegetation, and was formed by damming the Ord River. Construction of the Three Gorges dam on the Yellow River in China is one of the the largest engineering projects in human history. It was begun in 1995 and was completed in 2003. The dam wall is 181 metres high, 2335 metres wide, and has created a new lake over 660 km long. The terrestrial communities that occupied this area have been flooded, and new aquatic communities have taken their place.

Many organisms concentrate scarce elements. An unfortunate example of this is the accumulation from atomic bomb tests of radioactive strontium by reindeer lichen.

The radioactive strontium was passed on to reindeer, rendering them unfit for consumption. After the disaster in 1986 in the Ukraine at the nuclear power plant at Chernobyl, even the sheep on farms in Wales in the United Kingdom, some 3000 km away, had to be quarantined due to the accumulation of radioactivity. Radioactive elements had been washed out of the atmosphere by rainfall and taken up by plants that the sheep then ate.

ACID RAIN

The waste gases formed by burning fossil fuels include sulfur dioxide and oxides of nitrogen. These gases dissolve in water droplets in the atmosphere forming acids such as sulfuric acid and nitric acid. Consequently, the rain that falls is acidic and is called acid rain.

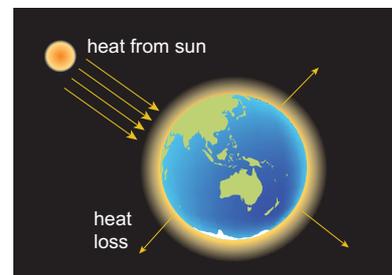


Fig. 25.3 Greenhouse gas effect
-more heat in than out

CHANGES TO THE ARAL SEA

(See textbox - Humans and Successions on page 191).

More recently, in 2011, radioactive material was released into the sea from the tsunami affected nuclear reactor in Fukushima, Japan. This will have long-term implications for the environment and is an example of the unexpected consequences of the use of scientific knowledge.

Human activities can create new and significant selection pressures on a gene pool, leading to species extinction.



Describe how these activities have caused or may threaten the extinction of species.

▼ Human activities and extinction of species

We have fossil evidence that, until recently, Australia was the home of a much greater variety of marsupials, and that some of the large forms, the megafauna, became extinct due to a combination of factors such as climate change and human activity. The megafauna overlapped with human occupation for 20 000 or more years and the hippopotamus-sized marsupial *Diprotodon* became extinct about 25 000 years ago. (See Fig. 25.4)

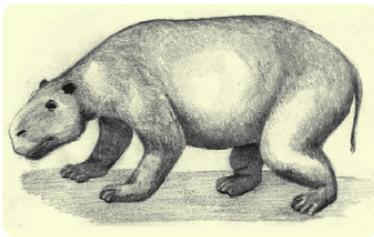


Fig. 25.4 Diprotodon

Particularly hard hit since the arrival of humans has been the carnivorous subgroup of marsupials such as the Tasmanian tiger (See Fig. 25.5), the Tasmanian devil and quolls. These animals were once present all over Australia but the 'tiger' and 'devil' became extinct on the mainland about two thousand years ago and have been confined to Tasmania since then. This demise may have been caused by the introduction of the dingo to Australia by humans about 4000 years ago, as dingoes are only found on the mainland. After Europeans arrived in Tasmania, the Tasmanian tiger was hunted to extinction, as it was considered to be a problem for sheep farmers. The last known survivor of this species died in the Hobart Zoo in 1936. Seventeen species of Australian mammal have become extinct since European settlement in 1788, and there are currently about 1600 vertebrate species in Australia of which 300 are considered to be at risk of extinction.

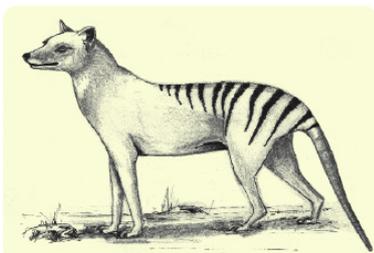


Fig. 25.5 Tasmanian tiger

In South Australia, there used to be two species of rabbit-eared bandicoots, or bilbies — the lesser bilby and the greater bilby. (See Fig. 25.6) These were quite numerous, even after European settlement, and appeared to benefit from the clearing of scrub to make way for pasture. Their demise was rapid, however, when two imported pests, the rabbit and the fox, found their way into South Australia. The rabbit competed for the bilby's food and the fox found the bilby easy prey. No live specimens of the lesser bilby have been sighted for several decades, and it is now presumed to be extinct. The other species, the greater bilby, is now confined to a few isolated colonies in central Australia.

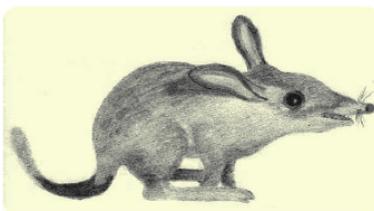


Fig. 25.6 Bilby

Lord Howe Island is a small Pacific island 700 km north east of Sydney. In the 1830s, of all the species of birds found there, ten were unique to the island. By 1870 three of these species had been exterminated as they had been either hunted for food or destroyed because they damaged crops. The introduction of rats and pigs onto the small island caused the extinction of a further five of these unique species by 1922.



Recognise that humans have an obligation to prevent species extinction.

An often-used example of extinction occurred on the island of Mauritius, where the dodo, a flightless bird, was eliminated by humans about 300 years ago. (See Fig. 25.7) In Mauritius, it was noticed that *Calvaria major* trees were also decreasing in number. The seeds in their fruit are very thick-walled, and it was thought that they needed to pass through the digestive system of a dodo before they could germinate. Although this idea has been disputed, the dodo did play an important role in the dispersal of *Calvaria* seeds. In a similar way, the cassowary is crucial for the long-term survival of rainforest trees in Queensland, and perhaps the emu for quandongs in South Australia. These examples also illustrate the importance of maintaining biodiversity, as the survival of one species often depends on the survival of another.

Maintaining biodiversity is an ethical issue with long term biological and/or environmental consequences.

Biodiversity refers to the variety of the organisms living in a region. Biologists have investigated healthy communities to find whether the main effect on producer numbers is competition among themselves or being consumed by herbivores. In most communities, carnivores keep herbivore numbers in check, so that competition between plants for resources is the main cause of plant death. There are cases of misguided human extermination of carnivores that have resulted in population explosions of herbivores. This was the case with the Kaibab deer in the USA when the extermination of mountain lions and coyotes led to plant populations collapsing due to overgrazing by the deer.

Australia also has many examples of misguided interference by humans. Introduced sheep and rabbits compete for food with kangaroos and other native grazing animals and their flocking or burrowing behaviour causes damage to the soil, leading to erosion. Dingoes may kill sheep and so they are shot as pests. We then wonder why the population of kangaroos rises until they become a nuisance and have to be culled. Some people go so far as to say that the sheep should be removed and the kangaroos used as the grazer on farms.

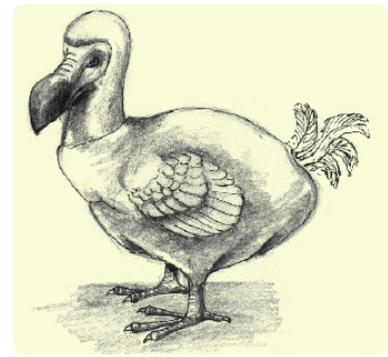


Fig. 25.7 Dodo

PALM OIL

Palm oil now has become a significant ingredient in our processed foods and each year in South East Asia, large areas of rainforest are replaced with palm oil plantations. This destruction has decreased the habitats of vulnerable species like the Orangutan and the Sumatran tiger. It is estimated that fewer than 400 Sumatran tigers are now left in the wild.



Fig. 25.8 Orangutan in Borneo. WWF status - critically endangered.

SUMATRAN TIGER



tinyurl.com/y7rgku56

When calicivirus was released in 1996 to eradicate rabbits, there was a spectacular regeneration of vegetation in some places, just as there was in the Northern Flinders Ranges when goats in the area were intensively culled. The fear was that feral cats and foxes, starved for rabbits, would turn on native animals to survive. The population of wedge-tailed eagles had built up because of the availability of rabbits. When calicivirus killed off the rabbits, eagles turned to eating roadkill.

The Importance of Biodiversity

There are more than two million identified species on Earth, and many more that have yet to be identified. The term **biodiversity** refers to the variety of species and their relative abundance.

There are at least three ways of looking at the question of why biodiversity is important.

- › One is a totally human-centred view and concentrates on what we obtain from living things. These may include foods, medicines, and raw materials. The foods that we consume come from a wide variety of plant and animal sources. Medicinal substances are often discovered in plants and then used as the basis for pharmaceuticals. The cotton, leather and wool that we use for clothing come from organisms, and timber is used in construction and paper manufacture.

Some benefits have been derived from unusual or unexpected sources. For example, the venom from some Australian snakes works by acting as an anticoagulant. This property was found to be very useful in the prevention of blood clot formation in surgical patients. In some tropical frogs, the exposed skin is protected from ultra-violet damage by a secretion. This is now being tested for application in humans. There are undoubtedly many more such features awaiting discovery. Losing any of the Earth's species could mean the loss of potential benefits to our own species.

Some very potent anti-cancer drugs have been extracted from rare animals and plants. For example, paclitaxel, an effective cancer drug, is obtained from a small, slow growing tree, the Pacific yew. Discoveries like this add to the argument that we should preserve biodiversity, especially rainforests which are the home of a wide variety of plants. Vinblastine is obtained from a small flowering plant that lives in tropical rainforests. It is also used in the treatment of cancers, as it interferes with spindle formation, disrupting cell division.

- › The second way looks at how all life on Earth, including humans, is interconnected. Upsetting the balance of nature by allowing biodiversity to decrease may have severe and unpredictable implications for us all. One intriguing theory, the so-called Gaia

hypothesis, states that all living things have evolved in ways that maintain the planet so that it is suitable for life to exist. Soil is built, minerals are cycled, and nitrogen gas is fixed. The level of free oxygen is sufficient for respiration but not so high that uncontrolled burning occurs. The temperature is regulated to some extent by cloudiness, as clouds reflect the short wave radiation directly to space, and cloudiness may be at least partly influenced by transpiration — the loss of water vapour from leaves. In short, life provides its own self-regulatory system. We should not disturb the biodiversity by our conduct until we thoroughly understand both the effects and the consequences. The argument is that all life constitutes an enormous gene pool and it shouldn't be reduced. Biodiversity should be maintained, not as conservation of single species, but as living communities.

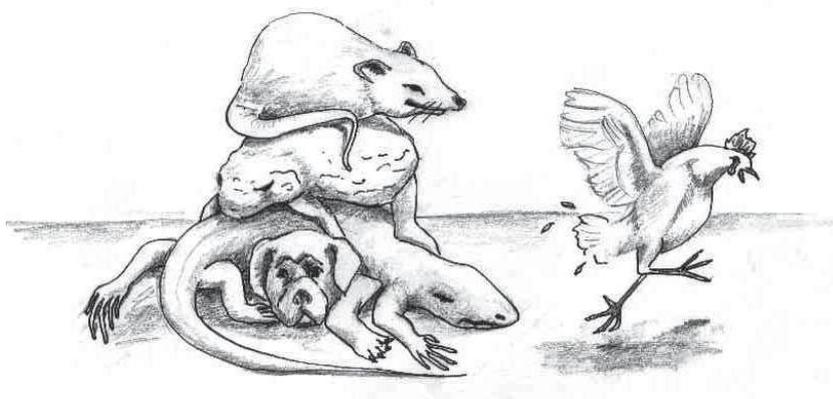


Fig. 25.9 Community interactions in Micronesia

- A third argument deals with our own morality and says that all life should be respected. The belief is that for humans to take on the decisions of life or death for other species is simply a sign of our own arrogance. Other species have just as much right to live as humans claim for themselves. Such arguments may manifest themselves as proposals to set aside certain areas as true wilderness where no humans may enter. There is also the view that the diversity of life has an aesthetic value and that we should preserve it for others to appreciate, including future generations of humans.

If the argument that genetic diversity in a species is a good thing because it means there is scope for adaptation through natural selection if the environment changes, then the same can apply to life on Earth in general.

Preserving habitat

An organism's **habitat** is its living place. It includes the medium, which may be soil, air, water or even another organism, the climate, and other organisms present.

A species never lives in isolation, but as part of a community. All species have evolved to fill a niche in their communities and

COMMUNITY INTERACTIONS

When the populations of a community are in balance, the introduction of a new population can have severe implications for the rest of the members of the community. In Micronesia, in the Pacific, rats were causing damage to sugar cane crops. (See Fig. 25.9) Large lizards with a liking for rats were brought in. Unfortunately they had an even greater liking for domestic chickens. Cane toads were brought in as an alternative food source for the lizards. The cane toads, with their poisonous skin glands, killed the lizards that ate them, and also killed cats, dogs and pigs. The rats flourished, as did imported giant snails. The snails also needed to be controlled, and there was a solution that someone knew that worked somewhere else — a flatworm that preyed on the giant snails. The flatworm was introduced but unfortunately it also wiped out most of the native snails. The end result was a new community, but not the one that the people of Micronesia intended. They were not able to predict all of the interactions between the populations involved. Are there any similar examples that you know of in your area?

KOALAS AND THEIR HABITAT

Although there are several hundred species of eucalypts in Australia, the koala can only feed on the leaves of a few of these species. The koala's distribution is limited to regions where these species of eucalypt are found. This is a good example that explains why the best way to preserve a species is to preserve its habitat. (see Fig. 25.10)



Fig. 25.10 What represents survival of a species?

SAVING A SPECIES FROM EXTINCTION

The last member of the species *Pyrenees ibex*, a type of mountain goat from Pyrenees, between Spain and France, died in 2000. Later the ibex became the first extinct animal to be successfully cloned, using skin cells that had been collected and frozen in 1999. However, the cloned ibex died shortly after birth from respiratory complications.

they have features that have been selected to ensure that they are adapted to their particular habitats. If the habitat is altered in any way, the selection pressures change and the species may be eliminated from the area. We saw earlier that it took nearly 300 years before an important function of the dodo in assisting the germination of the *Calvaria* seed was discovered. By removing the dodo from *Calvaria*'s habitat, the *Calvaria* too is in danger of extinction.

There are two generally accepted levels of conservation in preserving species, particularly animals:

- › to ensure mere survival of the largest carnivore in the community, it is considered that an area large enough to support fifty of these animals must be set aside.
- › for the species to be reasonably secure, the area needs to support 500 individuals.

In both cases, the area is a habitat that contains all of the biotic and abiotic factors necessary to support the natural community. In other words, to preserve a species it is necessary to preserve its habitat.

Does life in a zoo or game park constitute survival of a species? The genetic material may have been preserved, but if there is no habitat to which the animal can be returned, is there any point?

Even so, there are examples of plants that have been preserved for hundreds of years in unnatural surroundings, but are now planted widely for our enjoyment. The Ginkgo tree was previously thought to be extinct and was known only from fossils. In the early 1900s a few specimens were found to have been preserved in the grounds of Chinese temples, and now the Ginkgo is quite common in our suburban parks and gardens.

In North America the grizzly bear and the puma are now considered to be in danger of becoming extinct because none of the parks that is set aside for them is large enough to support a population of 500. The same reasoning applies to large endangered herbivores such as the rhino in Malaysia. In Australia, even if a few Tasmanian tigers still survive, or if a new individual can be cloned from the DNA of a preserved specimen, their numbers would be far too small to ensure the future of the species.

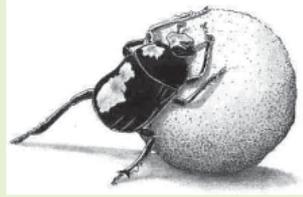
Recycling resources

Most people think of resources as being minerals, oil and gas. In biological terms, anything that is obtained from the environment and used by organisms is called a resource. These include not just mineral elements that are incorporated into organisms, but things like nesting sites for birds and suitable soil for burrowing. The resources in a community are usually in limited supply and need to be recycled from

HOW HUMAN ACTIVITIES MAY HELP:

DUNG BEETLES

Did you know that dung beetles in Australia are largely imported, just as the rabbit was? There are about 500 native species of dung beetle, but most of these prefer forests and woodland and the higher rainfall coastal regions.



Introduced cattle produce large amounts of dung on grasslands and the native species of dung beetle, adapted mainly to kangaroo dung, were unable to cope.

In some places the cowdung pats were almost carpeting the soil completely. Added to this was the fact that the pats provided food for the unpleasant buffalo fly in the subtropics and the bushfly in the rest of the country. It has been estimated that 400 million pats are dropped each day, and if each were food for 100 larvae, it is no wonder we needed corks on our hat brims.

Biologists investigating the problem believed that importing dung beetles that could deal with cow pats was a possible biological solution. Biologists investigated imported species before releasing them in the wild. Beetles were brought from tropical southern Africa for our subtropics, and from Cape Province, South Africa and southern Europe for the rest of Australia. Forty-one species were released, of which 22 have now become established. They have reduced the number of bush flies at certain times of the year by 90%, so the program is quite successful. In addition to clearing the pasture for growing grass, and decreasing the fly problem, the dung beetles have greatly increased the productivity of grassland communities by returning nutrients to the soil.

Dung beetles have three lifestyles, the dwellers, the tunnellers and the rollers. Dwellers feed in the dung and lay their eggs there. Tunnellers make breeding tunnels under the moist pat and stock them with some dung. Rollers are the glamour boys and girls of the dung beetle world, taking large spheres of dung off the pat and rolling these to a burial site and then laying their eggs in them. By doing this they reduce their exposure to competition. And competition can be fierce; 16000 beetles have been counted on a fresh elephant pat weighing 1.5 kg, and in 2 hours they had eaten, tunnelled and rolled it away completely.

There is of course a whole range of other organisms making up a food web around the dung pat. There are fungi, fungi-feeding flies and other insects, dung flies, predatory flies, wasps and beetles. For the poor dung beetle, as for the rest of us, there is no such thing as a free lunch.

However, dung beetles have had their moment of glory: ancient Egyptians revered the scarabs (the commonest dung beetle), seeing them as the link between Sun, soil and cattle.

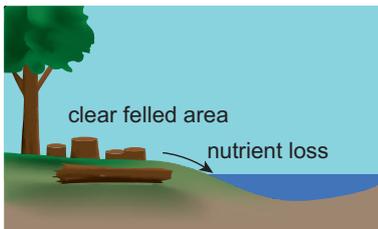


Fig. 25.11 Loss of nutrients from a disturbed area

one generation or from year to year. Nesting sites are re-used by birds from year to year, as are the burrows of hopping mice. The mineral elements that are extracted from the soil by plants also need to be recycled if the community is to live on. The minerals are not locked up in the plants forever, but are returned to the soil when parts of the plant die, or are eaten. Decomposers, such as bacteria and fungi, play a major role in this recycling process. In an undisturbed community the recycling process ensures that future populations have sufficient resources to maintain their numbers. If disturbances such as floods, fire, cropping or leaching occur, then some of these resources will be lost from the community.

Some of the best evidence that a disturbed community does lose its chemical resources comes from the Hubbard Brook Experimental Forest in the USA. Here small areas were clear-felled, but the cut timber was left on the ground. There was an immediate loss of nitrate from the soil into the stream. (See Fig. 25.11) Loss of nitrogen by leaching stayed much higher for many years than in the control areas that were left untouched. The conclusion was that the undisturbed community was retaining nearly all of its essential nitrogen by recycling it, but the disturbed community was losing this essential resource.

DINOSAUR EXTINCTION



tinyurl.com/y9stnr3y

Future evolution on Earth

Since the beginning of life on Earth around 3.5 billion years ago, many millions of species – probably billions – have evolved. Most of them (more than 99 percent) have become extinct. There have been periods of mass extinction due to catastrophic events. About 65 million years ago one such event, probably an asteroid collision, wiped out dinosaurs and most other species. Small mammals benefited from this due to reduced competition and predation. They gave rise to modern mammals – you owe your existence to this! It has been estimated that the average ‘life-span’ of a species is about 2 million years! Evidence suggests that in recent times the rate of extinction has accelerated due to human activities.

We think that there may be as many as 10 million or more species on the planet today, most of them undiscovered or unidentified. The majority are of these are probably small animals, such as insects. Up until now we have classified and named fewer than 2 million species.

Modern humans (*Homo sapiens*) have occupied the Earth for around 200 000 years. This raises some interesting questions:

- How much have humans evolved during this time?
- How is scientific and technological advancement affecting human evolution?
- What will be the predominant species on Earth in 200 000 years from now? What about 2 million years from now?

Of course, the biggest question of all is 'what influence can you and your contemporaries have on the environment and the future of humanity?'

Study Questions

1. Give three examples of human activities that lead to climate or environmental change and explain how each causes such a change.
2. Human activity can alter the physical environment of an area and the nature of the communities living there.
 - (a) List some of the impacts that humans and human activity can have on an environment.
 - (b) List some of the impacts which humans and human activities can have on populations of plants and animals living in an area.
 - (c) What is the greenhouse effect and why are coastal areas likely to be significantly affected by it?
3. Human activities can create increased selection pressures on natural populations. Describe how these activities have caused or may threaten the extinction of species.
 - (a) What do we mean when we say that a species has become extinct?
 - (b) What could cause the extinction of a species?
 - (c) What is a habitat, and why is preserving habitat essential for the survival of a species?
4. Humans have an ethical obligation to prevent species extinction. Explain the two levels of conservation suggested for preserving animal species.
5. Maintaining biodiversity is important to avoid long term biological and environmental consequences.
 - (a) What is meant by the term 'biodiversity'?
 - (b) Summarise the three main arguments for protecting biodiversity.
6. The human species has contributed to a number of biological problems, not least of which is the size of the human population.
 - (a) What problems for the environment have our great number caused?
 - (b) What current practices are there that attempt to minimise our disruptive effects on the environment?
7.
 - (a) Define the term resource and explain why recycling of resources is important in communities.
 - (b) What is a disturbed community? State two examples.
 - (c) Why is recycling more efficient in undisturbed communities?
 - (d) What resources are recycled in a natural community?
 - (e) Explain the role of decomposers in recycling.

Glossary

0 - 9

2-naphthylamine

a chemical that causes cancer of the bladder

A

abiotic

a non-living component of the environment, such as rainfall

accuracy

the degree to which a measurement is close to the true value

acid rain

rain that contains dissolved pollutants, such as sulfur dioxide, thus making it acidic

acquired characteristic

obtained during a lifetime; not inherited

actin

a globular protein that forms microfilaments in cells

activation energy

the energy required to start a chemical reaction

active

requires the input of energy – not passive

active site

a region with a particular shape on the surface of an enzyme molecule into which a substrate molecule with a complementary shape will fit

active transport

the movement of a substance across a membrane against the concentration gradient, thus requiring input of energy

adaptation

any feature of an organism that enables it to survive and/or reproduce

adaptive radiation

a special case of divergent evolution in which there is a sudden emergence of new species from a common ancestor

adenine

a nitrogen base found in nucleic acids (A)

adenosine diphosphate

see ADP

adenosine triphosphate

see ATP

adipose tissue

connective tissue made up of adipose cells that are filled with lipid

ADP

adenosine diphosphate, a molecule of adenosine with two inorganic phosphate groups attached. Use to make ATP

adrenaline

a hormone released by the adrenal glands and nerve endings. The so-called “fight or flight” hormone

adrenal glands

glands located on top of the kidneys that release the hormone *adrenaline*

aerobic respiration

a chemical process using oxygen that occurs in many cells and involves the breakdown of glucose to carbon dioxide and water, releasing energy

afferent

conducting towards; see *efferent*

AIDS

acquired immune deficiency syndrome that may result from infection with HIV

alcohol fermentation

anaerobic respiration in plant cells and yeast that forms alcohol (ethanol) and carbon dioxide

allele

an alternative form of a gene for a particular characteristic – more recently also refers to variation among non-coding DNA sequences

allopatric speciation

the process of forming new species from a single population that becomes divided by geographical barriers

alveolus

air sac (in mammalian lungs)

Ames test

a process that uses bacteria to check chemicals for carcinogenic effects

amino acid

the subunit of proteins. Of the more than 80 naturally occurring amino acids, only 20 are found in proteins

amniocentesis

a test involving removal of amniotic fluid and accompanying foetal cells through the wall of the abdominal cavity

amniotic fluid

the fluid surrounding the foetus during development *in utero*

amoeba

an aquatic single-celled organism that feeds by phagocytosis

amylase

enzyme involved the digestion of starch to maltose (a disaccharide)

anaerobic respiration

a chemical process, not requiring oxygen, that occurs in some cells and involves the partial breakdown of a fuel such as glucose, releasing some energy. In animal cells the product is lactic acid, and in plant and yeast cells the products are ethanol and carbon dioxide

anaphase

the stage in mitotic cell division in which sister chromatids separate

anaphase 1

the stage in the first meiotic cell division in which homologous chromosomes separate

anaphase 2

the stage in the second meiotic cell division in which sister chromatids separate

antibiotic

a chemical that kills bacteria or prevents their growth

antibody

a specific protein molecule that combines with the antigen that caused its production

anticodon

the region composed of three nitrogen bases on a transfer RNA molecule that is complementary to the codon on messenger RNA

antidiuretic hormone (ADH)

a hormone produced by the hypothalamus and released via the posterior pituitary; it increases water reabsorption from the collecting ducts into the blood in the kidneys, reducing urine output (diuresis); ADH is also known as vasopressin

antigen

a substance that stimulates the production of specific antibodies. Usually a protein, carbohydrate, or glycoprotein

antigen-binding site

the site on an antibody molecule with a shape that is complementary to the antigen that stimulated its production

antigenic determinant

the region on an antigen molecule with a specific shape that is complementary to the shape of the antigen binding sites on the corresponding antibody molecule

aorta

the large artery that carries blood from the left ventricle of the heart

aquaporin

a channel protein that allows the movement of water across a membrane. The process is passive and is called *osmosis*

aquatic

relating to water

arteriole

small blood vessel between capillary network and artery

artery

vessel with muscular, elastic wall that transports blood away from the heart

asexual reproduction

the production of new individuals without the mixing of genetic material

atom

smallest unit with a particular chemical characteristic. An element consists of one kind of atom.

ATP

adenosine triphosphate, a cell's short-term energy storage compound

ATP cycle

the repeated formation and breakdown of ATP to transfer energy within a cell

autosomal

not sex-linked (nor X-linked)

autosome

a non-sex chromosome (i.e. not an X or Y chromosome in humans)

autotroph

an organism that is able to manufacture *all* of its complex organic compounds from simple inorganic substances

auxin

a plant hormone that stimulates growth

axon

the long process of a nerve cell that carries nerve impulses

axon terminal

nerve impulses travel along the axon of a neuron towards the axon terminal; the axon terminal secretes a neurotransmitter that triggers an impulse in the dendrites of the next neuron in the nerve pathway, or causes a response in an effector

B**bacteria**

simple unicellular prokaryotic organisms; some cause disease

bacterial transformation

a process in which DNA is incorporated into bacteria; an example is the incorporation of recombinant plasmids into bacteria in genetic engineering

barbiturate

a chemical that has a depressant effect on the central nervous system

base triplet

see codon

basement membrane

a matrix of glycoprotein fibrils, on which epithelial cells sit

binary fission

the asexual reproduction of a prokaryotic cells involving splitting in two

biodiversity

the variety of organisms living in a region

biomass

the total mass of organic matter in a community, usually measured by dry weight

biosphere

that portion of the Earth that is inhabited by organisms

biotechnology

the use of living things or living systems to produce useful materials. Recently, this has focused on the manipulation of genetic material

biotic

the living component of an area

bivalent

paired homologous chromosomes during meiosis, consisting of four chromatids

bladder

a sac for the storage of fluid

bolus

ball of food mixed with saliva that is swallowed

Bowman's capsule

part of the nephron in the kidney

bronchiole

branch of bronchus in the lung that transports air to and from the alveoli

bronchus

one of two branches of the trachea in the lungs

budding

method of asexual reproduction in which a new individual forms on the parent and then breaks away

C**Calvin cycle**

part of photosynthesis; also known as "dark reaction"

cancer

a disease due to the proliferation of mutant cells

capillary

microscopic blood vessel involved in exchange of materials between blood and tissues

carbohydrate

organic compound containing carbon, hydrogen, and oxygen. Mono-, di-, and polysaccharides (simple and complex sugars)

carbon monoxide

a poisonous gas formed by incomplete burning of materials containing carbon. It competes with oxygen for the sites on haemoglobin.

carcinogen

a cancer-causing substance

cardiac

of the heart

cardiovascular system

system comprising heart, blood, and blood vessels

carnivore

an animal that derives its energy and nutrition by eating other animals

carrier protein

a transport protein that binds to the substance it transports. Some carrier proteins are involved in *facilitated diffusion*, a passive process. Glucose transporter proteins (GLUTs) are examples. Other carrier proteins, called *protein pumps* are involved in *active transport*, requiring energy

carrying capacity

the maximum number of organisms that a habitat can support

Cas protein

a CRISPR associated protein that is an endonuclease; it can be 'programmed' with RNA to cut DNA at a specific site - an example is Cas9. Cas13a is an endonuclease protein that can be guided to cut RNA at a specific site (see SHERLOCK)

catalyst

a chemical that assists a chemical reaction without itself being altered or used up

cell

the basic unit of living things; a unit of living matter surrounded by a membrane which regulates the passage of substances

cell culture

see tissue culture

cell cycle

the sequence of synthesis, enlargement, and division in the life of a cell

cell division

the division of a cell into two daughter cells

cell membrane

dynamic structure made of lipid and protein that separates the contents of a cell from the external environment and regulates the passage of materials into and out of the cell

cell theory

the idea that cells not only make up the bodies of living things, but they also carry out all of the 'life processes'. It is an important unifying concept for all organisms

cell wall

a structure made of cellulose that surrounds a plant cell, providing shape and support

cellulose

a polysaccharide made up of glucose subunits. It is the main component of plant cell walls

central nervous system (CNS)

this comprises the brain and spinal cord

centrifugation

a method of separating particles in a fluid on the basis of their density by spinning them at high speed using a machine called a centrifuge

centrioles

pair of structures found in animal cells that are involved in the formation of the spindle apparatus

centromere

the point at which two sister chromatids are joined

channel protein

a transport protein that **does not** bind to the substance that it transports. Substance diffuse through channel proteins. *Aquaporins* are examples of channel proteins.

chemical bond

the force that holds atoms together

chemical digestion

the breakdown of a chemical (in food) to a different, simpler substance

chemical reaction

a process in which the arrangement of atoms is altered, resulting in the formation of one or more new substances. Energy is either used up or released

chemosynthesis

synthesis of a complex organic compound from simple inorganic substances using energy from an inorganic chemical reaction

chemotherapy

the use of chemicals to treat cancer by interfering with the cell cycle

chiasma

the point at which homologous chromatids cross over during prophase I of meiosis. (plural *chiasmata*)

chlorophyll

green photosynthetic pigment found in chloroplasts

chloroplast

plant cell organelle; site of photosynthesis

cholesterol

a steroid type of lipid that is a component of animal cell membranes, but not plant cell membranes. It is an important precursor molecule for the production of other steroids

chromatin

DNA and protein located in the nucleus during interphase

chromosome

long threadlike structure made of DNA and protein, located in the nucleus; only visible with light microscope during mitosis and meiosis

cilium

a thin thread-like extension from a cell. It contains microtubules and is used for movement. Plural *cilia*

circulatory system

see cardiovascular system

climax community

a community that is stable and has ceased succession

clone

a group of genetically identical organisms produced as a result of asexual reproduction

cloning

making a set of identical copies (for example of genes or organisms)

codon

triplet of bases found on DNA or mRNA which codes for a specific amino acid in a protein

colonisers

the first plants to grow on bare surfaces. Also called *pioneers*

colony

a collection of many members of the same species. For example, bacterial colony

columnar

elongated (as in a column)

community

the sum of all the populations living in a particular place at a particular time

comparative genomics

this involves comparing the genomes of species to determine their evolutionary relationships

competitive inhibitor

an enzyme inhibitor that works by blocking the active site due to having a similar molecular shape to the substrate

complementary shapes

shapes that fit together

compound

a chemical substance made up of two or more different elements chemically combined

concentration gradient

the difference in concentration of a substance between two different regions (for example, inside and outside a cell)

congenital

a condition existing at or before birth

connective tissue

tissue that consists of a non-living matrix in which there are scattered a number of cells and protein fibres. Its function is to hold organs and other tissues together and to fill the space between them

consumer

a heterotrophic organism that feeds on other organisms or their wastes or remains

contractile vacuole

a vacuole found in some freshwater unicellular animals that maintains osmotic balance by collecting water and emptying it from the cell

controlled variables

factors that are kept constant during an experiment; see *factors held constant*

convergent evolution

the evolution of similar characteristics in species that are not closely related; this is observed in separate but similar environments. Examples include the marsupial mole and the placental mole, the platypus and the beaver, the whale and the dugong. Features such as eyes and wings have also evolved many times independently and are examples of convergent evolution

coronary arteries

arteries that branch from the aorta and supply the heart muscle

cortex

outer layer

crenate

take on a hollow appearance. This happens to red blood cells when they lose water.

CRISPR

clustered regularly interspaced short palindromic repeats. A kind of 'immune' system found in bacteria, in which they archive snippets of invading viral DNA in their genome in order to quickly identify and destroy the viral DNA on a subsequent invasion. An endonuclease (called Cas9) is 'programmed' to cut the viral DNA at a specific location. The CRISPR-Cas9 system can be used by biologists to edit DNA in cells

cristae

infoldings of inner mitochondrial membrane (singular *crista*)

crossing over

interchange between chromatids of homologous chromosomes that occurs during prophase I of meiosis see chiasma

cuticle

outer non-cellular layer, for example waxy cuticle on surface of leaf

cyanide

a competitive inhibitor for the enzyme cytochrome oxidase. When this enzyme is blocked, there is no more production of the chemical ATP, the energy source of cells. Muscles and nerve cells stop functioning and rapid death occurs.

cyanobacteria

photosynthetic bacteria, also known as "blue-green bacteria".

cyclin

a regulatory protein which activates an enzyme by combining with it to form a complex called the mitosis promoting factor or MPF. This is important in regulating cell division

cyclin dependent kinase (Cdk)

an enzyme that is only active when attached to a cyclin molecule

cyclosporin

immunosuppressive drug used by transplant recipients to prevent rejection

cystic fibrosis

a hereditary disease of the exocrine glands. The gene responsible is located on chromosome 7

cytochrome c

a universal electron carrier protein. Its amino acid sequence in different species can be compared to determine how closely they are related

cytochrome oxidase

a respiratory protein that reacts with oxygen and plays a role in energy flow within a cell

cytokinesis

division of cell contents into two daughter cells following mitosis

cytokinin

a plant hormone that stimulates cell division

cytoplasm

the fluid matrix in a cell excluding the nucleus

cytosine

a nitrogen base found in nucleic acids (C)

cytoskeleton

a network of protein fibres, consisting of microtubules, actin and intermediate filaments, found in eukaryotic cells.

cytosol

the fluid part of the cytoplasm, not including organelles such as ribosomes

dark reaction

part of photosynthesis; also known as Calvin cycle See light reaction

daughter cells

the cells produced as the result of cell division

DDT

a long-lasting insecticide with adverse ecological consequences. It was the first mass-produced insecticide and is now banned in many countries. It accumulates in higher order consumers

deamination

removal of amine group from amino acids forming urea and glucose; occurs mainly in the liver

decomposer

an organism that feeds on the dead remains and wastes of other organisms, thus recycling nutrients in the process

deconstruct

a process used to analyse a problem to facilitate finding a solution. It usually involves breaking down a complex problem into simpler components that are more able to be investigated

dendrite

the part of a nerve cell that carries a nerve impulse towards the cell body

dependent variable

the variable that is measured in a practical investigation. For example, if testing the effect of substrate concentration on the rate of an enzyme-controlled reaction, the reaction rate is the dependent variable

D**detritivore**

an organism that digests small particles of dead organic matter from any trophic level by means of an internal digestive system

diabetes

is characterised by excessive urine production. Most diabetes is glucose-related – see *diabetes mellitus*. Diabetes insipidus is related to insufficient production of, or lack of response to, ADH

diabetes mellitus

a disease in which blood glucose level is not regulated due to insufficient insulin production, or inadequate response to insulin

diabetic

a person who suffers from diabetes

differentiation

process in which an unspecialised cell develops a specialised structure and function

diffusion

overall movement of a substance in a fluid from a region of high concentration towards a region of lower concentration of the substance until equilibrium is reached

digestive system

in humans, the system of organs involved in the digestion of food materials and the absorption of nutrients

diploid cell

a cell with two of each kind of chromosome (homologous pairs); 2n

disaccharide

a carbohydrate made up of two simple sugar units (monosaccharides) joined together

disturbed community

a community in which the natural balance has been upset by, for example, the removal of a part of it

diurnal

active during the day

diuresis

excessive urine production

divergent evolution

the evolution of different characteristics from a common ancestor

dizygotic twins

twins derived from two zygotes, and therefore not genetically identical

DNA

deoxyribonucleic acid

DNA-DNA hybridisation

the technique used to compare the genomes of different species in order to establish the degree of similarity, and hence their evolutionary history

DNA fingerprinting

see *DNA profiling*

DNA ligase

an enzyme that catalyses the joining of two nucleotides

DNA polymerase

an enzyme that joins nucleotides to form a nucleic acid

DNA primer

a short length of DNA that is complementary to part of a much longer DNA strand. It acts as a starting point for DNA polymerase to replicate DNA molecules in the polymerase chain reaction or PCR

DNA probe

a short radioactively labelled segment of single-stranded DNA or RNA with a sequence of bases that is complementary to part of a gene. It is used to identify and locate the gene.

DNA profiling

a technique used in forensic science that enables individuals to be identified from samples such as blood, skin or semen on the basis of their unique DNA base sequences

DNA replication

see *semi-conservative replication*

DNA sequencing

determining the sequence of nucleotides (or sequence of nitrogen bases) in a DNA molecule

double helix

see *DNA*

Down syndrome

condition resulting from the inheritance of an extra chromosome 21; also known as trisomy 21; caused by failure of homologous chromosomes to separate during anaphase 1

E**ecosystem**

a community of organisms and its environment — an ecological system

effector

a muscle or gland that carries out a response (movement or secretion) as a result a stimulus

efferent

leading away from, see afferent

egg

see *ovum*

electropherogram

graphical results of analysis by electrophoresis

electrophoresis

the technique of separating molecules of different size using an electric field in a gel or solution

element

a chemical substance made up of one kind of atom

emphysema

a lung disease resulting from damaged alveoli; symptoms include shortness of breath

emulsification

formation of an emulsion by breaking large globules of fat into smaller globules

endergonic reaction

a chemical reaction that requires an input of energy

endo-

within

endocrine gland

“inward secreting” gland; “ductless” gland”; secretes a hormone or hormones directly into the blood

endocrine system

the system of hormone secreting (endocrine) glands

endocytosis

collective term for pinocytosis and phagocytosis; mechanism by which substances are taken into a cell by ‘infolding’ of the membrane

endonuclease

an enzyme that cuts DNA at a specific site

endoplasmic reticulum

network of interconnecting membranous passages in the cytoplasm joined to nuclear envelope and plasma membrane; involved in intra-cellular transport, storage, and lipid synthesis

endosymbiosis

an association in which one cell lives inside another and both benefit

energy

the capacity to do work

environment

the sum of all the factors that influence an organism

enzyme

organic catalyst made of protein that speeds up a specific chemical reaction without altering the products of the reaction and without being altered itself

enzyme-substrate complex

the intermediate compound formed as a result of the substrate binding to the active site of a specific enzyme molecule

epidermis

outer layer of cells; for example, upper and lower epidermis of a leaf

epigenetic

inheritable changes that are not explained by changes in the DNA sequence; one example is the methylation of cytosine, which affects gene expression, usually repressing it; this may play an important role in the switching on and off of genes during development

epiglottis

flap of tissue at the base of the tongue that covers the glottis during swallowing, preventing food from entering the trachea

epinephrine

also known as *adrenaline*

epithelium

the layer of cells that forms a surface

equilibrium species

a stable species that is well established in a community. Usually long-lived.

See *K-selected species*

erythrocyte

red blood cell

erythropoietin

a hormone that promoted the production of erythrocytes

ethical

relating to a moral judgement of whether something is ‘right’ or ‘wrong’

eukaryote

an organism that has relatively complex cells with membrane-bound organelles

eukaryotic

see *eukaryote*

eutrophication

a process in which a body of water gains an excess of nutrients, resulting in the rapid growth of algae, and depletion of the oxygen supply

evolution

process of gradual, progressive change that has resulted in the development of more complex and diverse life on earth

exchange surface

a surface that allows materials to pass across, often by diffusion

excretion

the removal of waste or excess materials from a cell or organism

excretory system

in humans, principally the urinary system consisting of the kidneys, ureters, bladder and urethra

exergonic reaction

a chemical reaction that releases energy

exo-

outside

exocrine gland

a gland that secretes a substance into a duct

exocytosis

opposite of endocytosis; a vesicle or vacuole within a cell fuses with the plasma membrane, releasing its contents

exon

a region of DNA that is expressed as a gene product

exponential growth

the geometric increase in numbers — for example 2, 4, 8, 16 etc.

extinction

the death and disappearance of all the populations of a species

extra-

outside

extracellular

outside a cell

extracellular fluid

fluid surrounding cells; also called tissue fluid

F**facilitated diffusion**

the diffusion of a substance across a membrane aided by a carrier protein – a passive process

factors held constant

in a practical investigation, these are the factors that are kept the same for all experimental groups, to allow comparison of the effect of the *independent variable* (which is deliberately altered) on the *dependent variable*; see *controlled variables*

fatty acids

long-chain molecules that are subunits of lipids

fauna

animals

feedback

process in which a response alters the stimulus that caused it; see *negative feedback* and *positive feedback*

fermentation

an anaerobic alternative to aerobic respiration - see *alcoholic fermentation* and *lactic acid fermentation*

fertilisation

fusion of a sperm cell and an ovum to form a zygote

fibroblast

a type of cell found in connective tissue

'fight or flight' response

a response to fear or stress, that prepares the body to 'fight or flee'; the hormones adrenaline, noradrenaline, and cortisol are mainly responsible

filtrate

material that has passed through a filter

filtration

the separation of particles by passing them through a filter

first order consumer

organisms that consume producers

flaccid

limp; not turgid

flagellum

a long hair-like process of a cell containing microtubules, and used for movement. Plural *flagella*

flora

plants

fluid mosaic model

a model of membrane structure that accounts for the observable behaviour of membranes. Protein channels, markers, and enzymes are embedded in a bilipid layer.

follicle stimulating hormone

FSH; pituitary hormone (gonadotrophin) that stimulates follicle development and oestrogen production in the female and sperm formation and androgen production in the male

food chain

a series of organisms, beginning with a producer, each feeding on the one before

food vacuole

a vacuole, resulting from phagocytosis, in which intracellular digestion occurs

forensic science

the use of scientific methods in legal proceedings

fructose

a monosaccharide component of the disaccharide sucrose

FSH

see *follicle stimulating hormone*

G**G0 phase**

cells that have differentiated are said to be in G0 phase

G1 phase

the first growth part of interphase, before the synthesis of new materials begins

G2 phase

the second growth stage in the cell cycle between the synthesis phase, when new materials are produced, and the onset of mitosis

gamete

a male or female haploid sex cell that fuses with another gamete to form a diploid zygote; see *germ cell*

gametogenesis

formation of gametes from primordial germ cells in the testes and ovaries

gel electrophoresis

a process that is used to separate molecules based on their size. DNA, RNA, proteins – or parts of them – are placed in a 'well' and pushed through a gel by an electric current. The smaller molecules move more quickly through the gel

gene

a segment of DNA on a chromosome that codes for a polypeptide or RNA molecule

gene cloning

the manufacture of many copies of a gene by incorporating the gene into bacterial cells using plasmids

gene flow

the movement of alleles into or out of a population due to migration or transfer of gametes

gene pool

the total of all the alleles of all the individuals in a population

genetic code

a set of codons, each comprising a three base sequence of DNA or RNA and specifying a particular amino acid in protein synthesis

genetic diversity

the range of genes present in a species

genetic drift

the disappearance of particular alleles from small populations due to certain individuals not surviving or reproducing; may be the result of a catastrophic event

genetic engineering

a process that involves the manipulation of genetic material by transferring a gene or genes from one cell to another, usually between different species

genetic modification

see *genetic engineering*

genome

the complete set of genetic material of an organism

genotype

the alleles that an organism possesses

geographical separation

the separation of two populations by a geographical barrier that may lead to speciation

germ cell

a male or female haploid sex cell that fuses with another gamete to form a diploid zygote; see *gamete*

gibberellin

a plant growth hormone

gland

a structure that secretes a substance – see endocrine gland and exocrine gland

globular protein

a protein molecule with a particular three dimensional shape

glomerulus

ball of blood capillaries that lies within the Bowman's capsule of each nephron

glucagon

hormone made of protein and secreted by the pancreas to increase blood glucose level

glucose

a monosaccharide that is an important source of energy for cells

glycerol

a component of lipids

glycogen

insoluble polysaccharide; storage form of glucose in animal cells, particularly in liver and muscle cells

glycolysis

stepwise conversion of glucose to pyruvic acid (pyruvate); common to both aerobic and anaerobic respiration; occurs in the cytoplasm

glycoprotein

protein with carbohydrate attached

goblet cells

a specialised epithelial cell that secretes mucus

Golgi body

cell organelle consisting of a stack of smooth membrane; involved in packaging and secretion; also known as Golgi apparatus

granum

a stack of thylakoid membranes in the chloroplast. The site of the light reactions of photosynthesis. Plural *grana*

greenhouse effect

the warming effect at the Earth's surface due to the atmosphere trapping heat energy, like the walls and roof of a greenhouse

gRNA

see guide RNA

growth factor

a substance that regulates growth

growth hormone

a polypeptide hormone, secreted by the anterior pituitary, that stimulates growth

guanine

a nitrogen base found in nucleic acids (G)

guard cells

pair of sausage-shaped cells in plant epidermis (particularly leaf) that line stomatal pore; degree of curvature is controlled by turgidity and determines stomatal aperture; contain chloroplasts

guide RNA

the RNA molecule (about 100 bases long) that is loaded into a Cas protein to guide it to cut DNA at a specific site

gyrase

an enzyme involved in DNA replication

H**habitat**

the place where an organism lives

haemoglobin

respiratory pigment, made of protein and iron, located in red blood cells; has a high affinity for oxygen in the lungs and a low affinity for oxygen in the tissues

haemophilia

X-linked recessive disease in which blood does not clot normally

haploid cell

a cell that contains one of each type of chromosome

HeLa cells

a line of tumour cells derived from Henrietta Lacks. These cells are particularly useful in laboratories because they divide repeatedly in cell culture

helicase

an enzyme that catalyses the unwinding of the DNA double helix during DNA replication

hepatitis

a disease of the liver. The most common forms, hepatitis A and B are caused by viruses

herbivore

an animal that feeds exclusively on plant material

heterotroph

organism that cannot produce all its complex organic compounds from simple inorganic substances and relies on other organisms or their products or remains

heterozygous

having two different alleles for a particular inherited characteristic

hierarchy

levels of organisation or complexity. Macromolecules, cells, organisms and ecosystems form a hierarchy

homeostasis

the maintenance of a relatively stable internal environment

homologous chromosomes

chromosomes with the same appearance that carry information for the same characteristics; pair up and separate during meiosis

homozygous

having two identical alleles for a particular inherited characteristic

hormone

a chemical message that is produced in one part of the body and produces an effect in another part (or parts) of the body. In animals, hormones are transported in the blood, and the regions in which they produce an effect are called target cells, tissues, or organs

human genome project (HUGO)
a project begun in 1990 and completed in 2003 with the aim of identifying all human genes and the entire base sequence of the human genome

Huntington's disease

an inherited disorder of the human nervous system caused by a dominant gene located on chromosome 4

hybrid

the offspring of two genetically dissimilar parents. For example the mule is a hybrid produced by a horse and a donkey

hybrid inviability

the inability of a hybrid to develop into a (breeding) adult; a *post-zygotic* barrier to gene flow between species

hybrid sterility

although a viable hybrid adult may form, it is sterile and cannot reproduce; a *post-zygotic* barrier to gene flow between species

hydrolase

an enzyme that catalyses the hydrolysis of a macromolecule into smaller subunits

hypothalamus

see *thermoregulatory centre*

hypothesis

an idea or suggestion that can be tested

hypothyroidism

underproduction of thyroid hormone by the thyroid gland, resulting in lethargy and reduced metabolism

I**immune system**

a organism's defence system which protects it from viruses, microorganisms, foreign tissue and cancer

immunity

resistance to infection; *passive* immunity uses introduced antibodies; *active* immunity is stimulated by foreign antigens

immunoglobulin

antibody (a *globular* protein that provides *immunity*)

immunosuppressive drug

a substance that suppresses the immune system

independent assortment

during metaphase I of meiosis, the maternal and paternal chromosomes of each homologous pair behave independently of the members of other homologous pairs. This ensures that, although the daughter cells have one member from each homologous pair, a random selection of maternal and paternal chromosomes is received. Also known as *random reassortment*

independent variable

in an investigation, the independent variable is the one that is deliberately altered to test its effect on the *dependent variable*

induced-fit model

a model of interaction between an enzyme and its substrate. When the enzyme and substrate join together the enzyme changes shape slightly so that the active site fits even more exactly to the substrate

inflammation

a non-specific defence in which blood capillaries walls increase in permeability resulting in swelling, redness, increased temperature, pain and an influx of phagocytic cells

insulin

hormone made of protein and secreted by the pancreas to decrease blood glucose level

interferon

a species-specific chemical secreted by virus infected cells to protect healthy cells from infection. Human interferon can be produced by genetically modified bacteria

intermediate compound

in a metabolic pathway, an intermediate compound is produced after the initial reactant(s), and before the final product(s). Some intermediate compounds may be linked to other metabolic pathways

intermediate filament

a component of the cytoskeleton in eukaryotic cells that is made up of protein subunits

internal environment

tissue fluid that surrounds the cells of a multicellular organism

interneuron

interneurons are located in the brain and spinal cord. They provide a connection between sensory (afferent) neurons and motor (efferent) neurons

interphase

a stage in the cell cycle *between* cell divisions; synthesis and growth occur during interphase

intra-

within

intracellular

within a cell/cells

intron

a part of a gene that does not code for a gene product, and therefore is not expressed

islets of Langerhans

tissue in the pancreas that secretes insulin and glucagon

J**junk DNA**

this term is no longer used, as it seems that non-coding DNA is most likely not 'junk'. See *non-coding DNA*

K**karyotype**

the complete set of chromosomes of an individual

Klinefelter syndrome

a condition resulting from the inheritance of an extra sex chromosome giving XXY; caused by failure of homologous X chromosomes to separate in anaphase 1 of meiosis; affected individuals are sterile males with feminine characteristics

Krebs cycle

a series of enzyme-controlled reactions that form the latter stages of aerobic respiration. A two-carbon compound is broken down to carbon dioxide and water, releasing energy that is used to make ATP. In eukaryotic cells this process occurs in the mitochondria

L**lacteal**

lymph capillary in a villus in the small intestine that transports lipids from the digestive system

lactic acid fermentation

anaerobic respiration in animal cells that forms lactic acid

leucocyte

white blood cell

LH

lutinising hormone, secreted by the anterior pituitary, stimulates ovulation and progesterone

lichen

an organism resulting from the symbiotic association between an alga and a fungus

ligase

an enzyme that catalyses the joining together of small molecules into larger ones. For example, DNA ligase

light reaction

the light-dependent stage of photosynthesis See dark reaction

limiting factor

a factor that, if varied, alters the rate or outcome of a process

lipase

an enzyme that catalyses the breakdown of lipids to fatty acids and glycerol

lipid

organic molecules containing carbon, hydrogen and oxygen, with each lipid molecule being made up of one glycerol and three fatty acid molecules. At room temperature, solid lipids are called fats and liquid lipids are called oils. Among other things, they are important energy storage compounds in cells

lipoprotein

a substance consisting of a protein combined with a lipid

locus

the particular position in which a specific gene is located on a chromosome

loop of Henle

part of the nephron tubule of the kidney

luteinising hormone

see *LH*

lymph

the transparent fluid carried from the tissues via lymph vessels; similar to tissue fluid, but with many lymphocytes; rejoins the blood and flows into the right atrium of the heart

lymph nodes

groups of cells through which lymph flows; located mainly in the neck, armpits, groin, and around major organs; contain phagocytic cells (macrophages) that remove foreign material from the lymph

lymphatic system

system composed of lymph vessels and lymph nodes that drains fluid called lymph from the tissues towards the heart; lymph nodes contain phagocytic cells that remove foreign particles

lymphocyte

a type of white blood cell; B lymphocytes are involved in humoral immunity; T lymphocytes are involved in cell mediated immunity

lyse

to break down a cell

lysosome

a membrane bound organelle located in the cytoplasm that contains hydrolytic enzymes

lysozyme

a hydrolytic enzyme found in sweat, tears, saliva, and blood; lyses bacterial cell walls

M**M phase**

the part of the eukaryotic cell cycle in which nuclear division, or mitosis, occurs

macromolecule

a large molecule, such as a nucleic acid, protein, or polysaccharide, that is made up of many smaller subunits joined together

macrophage

a cell derived from a monocyte that is able to carry out phagocytosis

madhatter disease

a disease resulting from the effects of mercury poisoning that affects the central nervous system

maternal

of or from the mother

maturation promoting factor (MPF)

a complex made up of an enzyme and a regulatory protein called cyclin. MPF plays an important role in the regulation of cell division medulla; also called mitosis promoting factor

middle part of an organ**meiosis**

two successive nuclear divisions in a diploid cell that result in the formation of four haploid cells in human males. In human females only one haploid cell is formed – see *polar bodies*; in a diploid organism gametes are formed by meiosis

membrane receptors

protein and carbohydrate molecules embedded in the bilipid layer that have distinct shapes and act as “markers” that enable cells to be recognised by each other and by molecular messages such as hormones

mesothelioma

a rare malignant tumour of the lungs resulting from exposure to asbestos

messenger RNA (mRNA)

single stranded nucleic acid transcribed from the DNA in the nucleus and transported to ribosomes in the cytoplasm for translation into a specific sequence of amino acids in protein synthesis

metabolic

relating to metabolism

metabolic pathway

a series of enzyme-controlled chemical reactions that occur in a specific sequence within a cell

metabolic rate

the rate at which chemical processes occur in a cell or organism

metabolism

all the chemical processes that occur within a cell or organism

metaphase

the stage in mitotic cell division in which chromosomes line up at the equator and attach to spindle fibres, with sister chromatids facing opposite poles

metaphase 1

the stage in the first meiotic cell division in which pairs of homologous chromosomes line up at the equator and attach to spindle fibres

metaphase 2

the stage in the second meiotic cell division in which chromosomes line up at the equator and attach to spindle fibres, with sister chromatids facing opposite poles

methylation

the addition of a methyl group (CH₃). See *epigenetic*

microarray

A grid of DNA probes attached to a glass slide, that are used to identify gene samples by complementary binding

microfilament

a component of the cytoskeleton that is made up of actin molecules

microinjection

a technique used to inject a liquid (often containing genetic material) into a cell. It is performed under a microscope using a micropipette and a micro-needle

microsatellite

see *short tandem repeat (STR)*

microscopic

unable to be seen without a microscope

microtubule

a component of the cytoskeleton made up of hollow filaments made of the protein tubulin

microvillus

microscopic finger-like extension of a cell membrane

mitochondrion

self-replicating cell organelle in which the latter stages of aerobic respiration occur

mitosis

division of the nucleus into two identical daughter nuclei

mitosis promoting factor (MPF)

see *maturation promoting factor*

molecule

an uncharged group of atoms chemically combined

monoculture

growing a single species to the exclusion of others, as is the practice in modern agriculture

monosaccharide

a simple sugar, such as glucose

monozygotic twins

twins derived from one zygote, and therefore genetically identical

morphological

to do with shape, structure, or appearance; derived from the Greek 'morph', meaning 'form'

motor neuron

a nerve cell that carries nerve impulses from the central nervous system to an effector, such as a muscle or gland

mucous membrane

moist lining of tracts such as respiratory, reproductive, and digestive

mucus

a thick secretion

multicellular

made up of many cells

mutagen

an agent that induces a mutation – that is, a change in the base sequence of DNA. Some mutagens are chemicals, such as 2-naphthylamine, bromine, and benzene; others include physical agents such as ultraviolet light, x-rays, and gamma rays. A mutagen that causes cancer is called a *carcinogen*

mutagenesis

the formation of mutations

mutation

any spontaneous or induced change in the genetic material of a cell

myxomatosis

a viral disease that was used to eradicate rabbits in Australia

N**naphthylamine**

see *2-naphthylamine* at start of glossary

natural selection

a process in which those individuals in a population who are better suited to the environment tend to survive and contribute more genes to the next generation, thus influencing the characteristics of future generations

negative feedback

a process in which a response inhibits or opposes the effect of the stimulus that caused it. This results in regulation or control, and is an important process in homeostasis

nephron

the functional unit of the kidney; there are about 1 to 2 million nephrons in a human kidney

nerve cell

a cell of the nervous system with the function of transmitting a nerve impulse

nerve impulse

a message that is carried rapidly from one end of a nerve cell to the other. It works by temporarily altering the permeability of the membrane along the nerve cell and by triggering an effect in muscles, glands or other nerve cells

nerve pathway

the pathway followed by a nerve impulse from stimulus to response. The stimulus is detected by a receptor, then the nerve pathway is usually via a sensory neuron, then an interneuron, followed by a motor neuron. The impulse from the motor neuron causes an effector (muscle or gland) to bring about a response

nervous system

a system involved in regulation and control by allowing communication between different parts of the body. In human beings it consists of the brain, spinal cord, and nerves

net primary productivity

the increase in amount of biomass resulting from photosynthesis over a period of time, minus the amount that the producers have respired for their own use

neuron

a nerve cell

neurotransmitter

a chemical secreted by the axon terminal of a neuron into the *synaptic cleft* at the *synapse* that initiates a nerve impulse in the next neuron in the nerve pathway.

Examples of neurotransmitters include acetylcholine, dopamine, noradrenaline, and even some amino acids and small peptides

niche

the unique role or 'position' of a species in a community

nitrogen base

one of the three components of a nucleotide

nocturnal

active at night

non-coding DNA

examples include: STRs (microsatellites), telomeres, introns

non-competitive inhibitor

a substance that inhibits an enzyme by combining with it in a way that alters the active site but does not compete with the substrate

nuclear envelope

a two-membrane structure enclosing the nucleus and containing nuclear pores.

nuclear pore

one of many evenly distributed openings in the nuclear envelope that allows the passage of large molecules, such as messenger RNA, between the nucleus and the cytoplasm

nuclease

an enzyme that breaks down nucleic acids to nucleotides

nucleic acid

organic polymer made up of a sequence of nucleotides; DNA is a double stranded double helix; RNA is single stranded

nucleolus

a region in the nucleus of a cell, the site of ribosomal RNA (rRNA) synthesis

nucleotide

a subunit of a nucleic acid, consisting of a five-carbon sugar, a nitrogen base, and a phosphate group

nucleus

usually the most prominent structure in a cell, containing the genetic material; controls the activities of the cell

nutrient

a substance that an organism assimilates and uses for maintenance, growth or reproduction

O**obesity**

an excess of body fat. A person whose body weight is more than 20 percent above normal is considered obese

oesophagus

the tube that transports food from the mouth to the stomach

omnivore

an organism that feeds on both plants and animals

oocyte

a cell that undergoes meiosis to form an ovum

oogenesis

ovum formation

opportunistic species

a species that has a short life span and high reproductive effort. It will increase its population numbers rapidly when conditions are favourable. See *r-selected species*

organ

a discrete structure found in an organism. It is composed of tissues and performs a specific role. For example, the heart

organ system

a group of organs that coordinate to carry out a specific function

organelle

a discrete structure found within eukaryotic cells. For example, a mitochondrion

organic compound

a compound that contains carbon. Note that carbon dioxide is not considered to be organic, due to its simplicity.

organism

a living thing

osmoregulation

the maintenance of water and solute balance; in humans the kidneys play an important role

osmosis

a natural, passive process in which there is a net movement of water across a semi-permeable membrane towards a more concentrated solution

osmotic pressure

the potential of a solution to take up water when separated from pure water by a semi-permeable membrane

osteoblast

a type of cell found in bone

ovaries (human)

organs that produce female gametes (ova) and sex hormones

ovum

female sex cell or gamete; egg cell

P**paclitaxel**

a chemical extracted from Pacific yew trees that inhibits cell growth and can be used in chemotherapy to treat cancer

pancreas

diffuse organ associated with the duodenum; secretes insulin and glucagon directly into the blood (endocrine); secretes pancreatic juice into the duodenum via the pancreatic duct (exocrine)

paramecium

an aquatic unicellular organism. Classified in the phylum Protozoa

parthenogenesis

development of a new individual from an unfertilised ovum

passive

not requiring input of energy; not active

paternal

of or from the father

pathogen

an organism that causes disease

pepsin

enzyme produced by the stomach lining in an inactive form (pepsinogen) as part of gastric juice; breaks down proteins to polypeptides

peripheral nervous system

this is made up of the nerves that transmit messages to and from the central nervous system (CNS – brain and spinal cord); it is the part of the nervous system that is outside the brain and spinal cord

pepsinogen

inactive form of pepsin

peptide

a chain of 9 or fewer amino acids

peptidoglycan

a polysaccharide used to construct bacterial cell walls

peristalsis

rhythmic wave of contraction and relaxation of the muscular wall of the digestive tract that ensures the movement of material along the oesophagus, small intestine, and large intestine

permeable membrane

a membrane that allows substances to pass through it. Compare with *semi-permeable*

pH

a measure of the acidity or alkalinity of a solution. pH 7 is neutral, below pH 7 is acidic, above pH 7 is basic.

phagocytosis

process in which a specialised cell engulfs small particles by invagination of the cell membrane and the formation of a vacuole containing the engulfed material

pharmaceutical

a medicinal drug

phenotype

any detectable feature of an organism

phenylketonuria (PKU)

an inherited autosomal recessive disorder which affects the enzyme that breaks down the amino acid phenylalanine

pheromone

a chemical used to communicate between organisms of the same species

phospholipid

a lipid-related compound that is a major component of cell membranes

photosynthesis

a chemical process in which glucose (an organic compound) is manufactured from water and carbon dioxide using light energy; oxygen is a byproduct; the necessary light energy is trapped by a photosynthetic pigment, most commonly chlorophyll

phylogenetic tree

a diagram that indicates the evolutionary relationship between different groups of organisms

physiological saline

0.9% sodium chloride solution. Has the same osmotic pressure as blood plasma

physiologist

a scientist who studies the functions of organs or organisms

phytoplankton

microscopic aquatic plants

pinocytosis

process in which a specialised cell engulfs liquids or large molecules by invagination of the cell membrane to form a pinocytic channel and the formation of vesicles containing the engulfed material

pioneers

the first plants to grow on bare surfaces. Also called *colonisers*

pituitary

endocrine gland at the base of the brain that is made up of two main parts — the anterior and posterior; the anterior pituitary produces several trophic hormones that regulate body processes such as reproduction

plasma (blood)

the non-cellular part of blood composed of water and dissolved substances such as cellular requirements and waste products, as well as plasma proteins; plasma is a straw coloured liquid

plasma membrane

cell membrane

plasmid

a small circular piece of DNA found in some bacterial cells, separate from the main chromosome; used as a vector in genetic engineering

plasmolysis

occurs in plant cells when water is lost from the cytoplasm and the cell membrane pulls away from the cell wall

pneumonia

a lung disease usually caused by viruses or bacteria and resulting in inflammation of the lung tissue

polar bodies

three very small cells produced during the meiotic development of an ovum; they break down, leaving the haploid ovum as the only product. See *meiosis*

poles

the ends of the spindle apparatus towards which chromosomes move during anaphase of mitosis and meiosis

polymerase chain reaction (PCR)

the technique of making multiple copies of a DNA sequence, by repeated heating and cooling

polypeptide

a chain of between 9 and 50 amino acids linked by peptide bonds

polyploidy

the presence of extra sets of chromosomes in an organism

polysaccharide

a macromolecule made up of thousands of simple sugar units (monomers); e.g. starch, cellulose, glycogen and peptidoglycan. Also called a complex carbohydrate

population

a group of organisms of the same species living together in the same area at the same time

positive feedback

a process in which a response enhances the stimulus that caused it

post-zygotic

after the zygote forms by fertilisation of the ovum

precision

measurements have greater precision when there is less scatter in the results

pre-zygotic

before the zygote forms by fertilisation of the ovum

primary data

data you have collected yourself; original data

primary productivity

the rate of production of organic matter from inorganic sources by autotrophs, measured as dry weight per unit area per unit time

primary succession

the initial succession in a barren area, starting with pioneer species

primer

(see *DNA primer*)

producer

an autotroph

product

a new substance resulting from a chemical reaction

productivity

the rate of production of organic matter (biomass)

prokaryote

a simple cell lacking membrane-bound organelles

prokaryotic

a unicellular organism that lacks membrane-bound organelles - Archea and bacteria

prophase

the stage in mitotic cell division in which the chromosomes first become visible, the nuclear membrane disappears, and the spindle apparatus forms

prophase 1

the stage in the first meiotic cell division in which pairs of homologous chromosomes first become visible, the nuclear membrane disappears, and the spindle apparatus forms. Crossing over may also occur

prophase 2

the stage in the second meiotic cell division in which chromosomes first become visible, the nuclear membrane disappears, and the spindle apparatus forms

protein

a macromolecule made up of more than 50 amino acid subunits joined together

protein pump

a *carrier protein* that binds to the substance that it transports. It uses energy to move a substance across a membrane against the concentration gradient

protein sequencing

determining the sequence of amino acids on a protein (or polypeptide)

protist

simple eukaryote, mainly unicellular

pulmonary

of the lungs

pure breeding

an organism that will produce offspring with the same characteristics as itself if it is self-crossed (as in pea plants) or crossed with another individual like itself

R**radiation**

electromagnetic waves, including heat, light, UV and X-rays

random error

an experimental error that occurs due to chance variation

random reassortment

see *independent assortment*

reabsorption

the process of taking up useful substances from the nephron tubules into the blood. These substances had previously been filtered out of the blood at the Bowman's capsule

reactant

a substance that is consumed in a chemical reaction

receptor

a cell or group of cells that detects a stimulus

receptor molecule

a molecule on the outside of a cell membrane that is involved in recognition or communication between cells

recessive

a genetic term used to describe a character that is normally only expressed in the homozygous condition; in a heterozygote it is masked; such characters are able to 'skip' a generation; the allele responsible is called a recessive allele

recombinant DNA

DNA that is formed by the joining of segments of DNA from different sources

recombinant DNA technology

the technique of using recombinant DNA to genetically alter a cell or organism

reflex

an automatic response to a stimulus that does not necessarily involve the brain

reflex response (see reflex)**relay protein**

a cellular protein that transfers a signal from a receptor molecule to another part of the cell

reliability

the reproducibility of a result

renal

of the kidney

renal tubule

tubule in the nephron carrying filtrate from Bowman's capsule to collecting duct. It is the site of reabsorption

repetition

carrying out an experiment on a second or subsequent occasion, in an attempt to validate the results (using a separate set of equipment and/or materials)

replication

setting up several copies of an experimental procedure in order to minimise the effect of random errors of measurement by averaging the results

reproductive effort

the amount of energy that a species puts into producing the next generation compared to the amount of energy expended in maintaining the adult organisms

reproductive isolation

a mechanism that prevents gene flow between populations

reproductive system

the system of organs that are involved in producing offspring

resolution

the resolution of a measuring instrument is the smallest increment it can measure

resolving power

the ability to distinguish two close but separate points as being separate

resource

anything from the environment that an organism uses

respiration

a chemical process that occurs in all living cells and involves the breakdown of an organic 'fuel', releasing energy to make ATP

respiratory centre

part of the brain that monitors the pH of the blood and controls breathing rate by sending nerve impulses to the muscles of the chest and diaphragm

respiratory system

the system of organs involved in the exchange of gases between the internal and external environment of an organism

response

a change in an organism due to a stimulus; usually movement by a muscle or secretion by a gland

restriction enzyme

a type of enzyme that recognises a specific base sequence of DNA and cuts the DNA at this point. Restriction enzymes were discovered in bacteria

restriction fragment length**polymorphism (RFLP)**

An individual's DNA can be broken up into fragments using restriction enzymes. The length of the fragments varies from individual to individual as they have different base sequences. These different length fragments are called Restriction Fragment Length Polymorphisms or RFLPs.

restriction point

the G1 phase checkpoint of the cell cycle. This must be passed before the cell will enter the S phase. Cells that do not pass this restriction point enter G0

restriction site

the sequence of nucleotides on a DNA molecule that is recognised and cut by a specific restriction enzyme

reverse transcriptase

an enzyme that produces a single strand of DNA using RNA as a template

rhizome

an underground stem that can be used as a means of vegetative reproduction

ribosomal RNA (rRNA)

RNA that combines with protein to form ribosomes

ribosome

cytoplasmic cell organelle that is the site of protein synthesis

ribozyme

RNA molecule that catalyses chemical reactions ('ribose enzyme')

RNA

ribose nucleic acid

S**S phase**

the synthesis phase of the cell cycle

satellite DNA

sections of the chromosome that consist of short repetitive nucleotide sequences and are non-coding

second order consumer

an organism that feeds on first order consumers

secondary data

data that someone else has produced

secondary succession

succession that occurs in an area where an existing community has been disturbed

secretion

a substance produced and released by a cell, tissue, gland or organ

segregation

the separation of homologous chromosomes during gamete formation

selection agent

a factor which acts on a population to cause natural selection

selection pressure

the action of selection agents on a population

selective exchange

the transfer of selected substances across a membrane

selectively permeable membrane

a membrane that allows only selected substances to pass through - using membrane protein channels

semen

secretion from male reproductive organs, containing sperm

semi-conservative replication

the replication of DNA that results in two new double helices, each consisting of one original strand and one new strand

semi-permeable

more permeable to some substances than to others

sensory neuron (sensory receptor)

a nerve that detects stimuli or receives a signal and passes it on to the central nervous system

sequencing DNA

a process for determining the sequence of nucleotides (and hence bases) in DNA

serum

blood plasma minus clotting proteins

sex linkage

results from a gene being carried on a sex chromosome, normally the X chromosome in humans; may then be referred to as X-linkage

sexual reproduction

reproduction involving the mixing of genetic material from two cells

SHERLOCK

is a CRISPR system that uses Cas13a which recognises RNA instead of DNA. This may be used to diagnose certain diseases

short tandem repeat

the repetition (up to 50 times) of about 2 to 6 nucleotides in a segment of (usually noncoding) DNA. As the number of repeats at a particular site (locus) can vary between individuals, STRs are useful in *DNA profiling*

sickle cell anaemia

an hereditary disease caused by a mutation on chromosome 11. The mutation causes the substitution of one amino acid in the haemoglobin chain and results in deformed and fragile red blood cells

single nucleotide polymorphism (SNP)

a single nucleotide variation in a genetic sequence

sister chromatids

two identical copies of a chromosome joined together by a centromere

skeletal system

the bones and muscles of the body

smooth muscle

muscles that lack the striations of cardiac and skeletal muscles. They are found surrounding blood vessels and the intestine and are also called involuntary muscles

solar

of the Sun

solute

a substance that is dissolved in a solvent forming a solution

somatic cell

a body cell (not a sex cell or gamete)

speciation

the formation of two or more species from one; see *allopatric speciation* and *sympatric speciation*

species

a group of organisms that are more or less alike and can reproduce fertile offspring in their natural environment; the members of a species share a common gene pool

sperm

male gamete

spermatogenesis

the production of sperm

spindle apparatus

the structure made of protein fibres that forms in the cell during mitosis and meiosis; chromosomes are arranged on the spindle apparatus, which assists their movement

squamous

flattened

starch

a polysaccharide made of glucose subunits and used as an energy store in plant cells

stem cell

an undifferentiated cell from which differentiated cells develop

steroid

a type of lipid that includes some hormones and cholesterol

sticky ends

the exposed bases at the end of a DNA fragment that has been cut by a restriction enzyme

stimulus

any change in the internal or external environment of an organism that is detected

stimulus-response model

the sequence: stimulus, receptor, message, effector, response

stolon

a long underground stem of a plant that may be involved in vegetative reproduction

STR

see *short tandem repeat*

stroma

the fluid in a chloroplast
stromatolite
a rock formed from sedimentary particles trapped by layers of photosynthetic prokaryotes. Fossils of stromatolites date back 3.5 million years

substrate

the chemical that is acted on by an enzyme

succession

the progression in area over time from pioneer species to a climax community

sucrase

an enzyme that catalyses the breakdown of the disaccharide sucrose into the monosaccharides glucose and fructose

sucrose

a disaccharide made of the monosaccharides glucose and fructose

sweat

a secretion from sweat glands in human skin, similar in composition to a dilute urine; evaporation of sweat has a cooling effect and is important in the regulation of body temperature

sweat gland

a gland in human skin that secretes sweat

swivelase

an enzyme that is involved in the process of DNA replication

sybiosis

a relationship between organisms of two different species in which both benefit

sympatric speciation

formation of two new species from a common ancestor while living in the same area without separation by a geographical barrier

synapse

the junction between an axon of one neuron and the dendrite of the next

synapsis

the process in which homologous chromosomes pair up during prophase 1 of meiosis

synaptic cleft

the gap between the axon and dendrite of consecutive neurons at the synapse

synthesis

an endergonic chemical reaction in which complex compounds are made from simpler ones

system

(see *organ system*)

T**target cell**

a cell that has membrane receptors for a particular hormone

target organ

an organ that contains target cells

target tissue

a tissue that contains target cells

Tay-Sachs disease

a lethal inherited condition due to a mutation of a gene located on chromosome 15. The disease causes the central nervous system to degenerate

telomere

a telomere is at the end of a chromosome and is made of repetitive sequences of non-coding DNA that protect the chromosome from damage. Each time a cell divides, the telomeres shorten, until eventually the cell can no longer divide

telophase

the stage in mitotic cell division in which sister chromatids have separated, two nuclear envelopes form, and the spindle apparatus is dismantled

telophase 1

the stage in the first meiotic cell division in which homologous chromosomes have separated, two nuclear envelopes form, and the spindle apparatus is dismantled

telophase 2

the stage in the second meiotic cell division in which sister chromatids have separated, two nuclear envelopes form, and the spindle apparatus is dismantled

template strand

the strand of DNA that is 'copied' by RNA polymerase to form RNA. The DNA strand that is complementary to the template strand is called the *gene*

temporal

means 'time'; a temporal reproductive barrier refers to species that cannot interbreed due to being fertile or active at different times, e.g. different seasons or different times of the day

testes

male sex organs that produce sperm and male hormones

testosterone

male sex hormone (androgen); it stimulates sperm production and the development of male secondary sex characteristics

thalassaemia

an inherited autosomal disease; individuals who are homozygous for the condition suffer from thalassaemia major (which is fatal if not treated), and those who are heterozygous have thalassaemia minor (which is symptomless under normal conditions)

thalidomide

a pharmaceutical that reduces nausea but also causes foetal deformity when taken by pregnant women

thermoregulatory centre

located in the hypothalamus in the brain; detects and responds to changes in body temperature

thylakoid

flattened membranous sacs that make up the grana in chloroplasts. The site of the light reactions of photosynthesis

thymine

a nitrogen base found in DNA (T)

thyroid gland

an endocrine gland that releases the hormone thyroxine to control the metabolic rate of cells

thyroid stimulating hormone (TSH)

also known as thyrotropin; a hormone released by the pituitary gland to stimulate the thyroid gland to secrete thyroxine

thyrotropin-releasing hormone (TRH)

also known as *thyroid releasing hormone*;

thyroxine

a hormone produced by the thyroid, stimulates general cell metabolism

tissue

a group of similar cells that perform a specific function. For example, muscle tissue

tissue culture

a laboratory technique for growing cells and tissues in a nutrient medium

tissue fluid

the fluid that surrounds the cells of the tissues

tolerance limits

the range of conditions required for survival of an organism

topoisomerase

an enzyme that alters the degree of coiling of DNA

toxic

poisonous

trachea

the "windpipe" or tube that transports air between the mouth and the bronchi

transcription

the process in which one strand of the DNA in the nucleus of a cell is used as a template to manufacture mRNA. The mRNA then travels into the cytoplasm and attaches to a ribosome for translation into a protein

transfer RNA (tRNA)

each tRNA molecule brings a specific amino acid to the ribosome according to its anticodon and the codon on the mRNA

transformation

see bacterial transformation

transgenesis

the transfer of genes from one species to another

transgenic organism

an organism formed by combining DNA from different species

translation

the part of protein synthesis that occurs on ribosomes; the codons on the mRNA determine the sequence of amino acids in the protein

transport protein

a membrane protein that allows the movement of selected substances across the membrane. See *carrier protein* and *channel protein*

transposon

a gene that can relocate or duplicate itself in another part of the DNA

trophic level

a feeding level of a food chain. For example, the first trophic level consists of producers

trypsin

an enzyme secreted by the pancreas that catalyses the breakdown of protein into polypeptides

tubule

a small tube

tumour

a mass of cells resulting from the uncontrolled growth of malignant cells

turgid

the term used to define a plant cell that cannot take in more water and is fully distended

Turner syndrome

a condition resulting from the inheritance of only one sex chromosome (an X chromosome) due to the failure of X chromosomes to separate in anaphase 1; affected individuals are females whose sex organs do not mature at adolescence, and they do not develop secondary sex characteristics. As a result they are sterile, and usually shorter than normal

U**ultrastructure**

the fine (detailed) structure

unicellular

single-celled

uracil

a nitrogen base found in RNA

urea

a nitrogenous waste product resulting from the breakdown of amino acids (deamination); primary production site is the liver; excreted by the kidneys

ureter

tube that transports urine from the kidney to the bladder

urethra

tube connecting the bladder to the external environment; in males it transports urine and semen; in females it transports urine only

V**vaccination**

a method of disease prevention that involves inoculating an individual with antigen (often an altered form of the pathogen), to stimulate the immune response, including the production of memory cells. Upon subsequent exposure to the same antigen the memory cells are able to respond faster, with a larger and longer lasting response

vaccine

the material that is inoculated for the purpose of vaccination

vacuole

a membrane-bound structure in a cell, containing water with dissolved materials; in animal cells vacuoles tend to be small and numerous, if present; in plant cells there is usually a large central vacuole that plays an important role in controlling water and solute balance

valid (validity)

results are valid if they are credible and the experimental design is sound because it tests the hypothesis

valve

a structure that allows flow in only one direction; located in the heart, veins, and lymph vessels

variable

the factors that are changed (independent variable), measured (dependent variable) or kept constant in an experiment (controlled variable)

vaso-constriction

the narrowing of a blood vessel, usually an artery or arteriole, to reduce blood flow

vaso-dilation

the widening of a blood vessel, usually an artery or arteriole, to increase blood flow

vasopressin

see *antidiuretic hormone*

vector

an organism or structure that carries material from one individual to another

vegetative propagation

an asexual method of reproduction in plants

vein

a major blood vessel with an inelastic wall that transports blood towards the heart

vena cava

the major vein that carries deoxygenated blood from the body to the right ventricle of the heart

venom

a poisonous substance secreted by an animal such as a spider or snake

ventilation

a process in which gases are moved across the respiratory surface; in humans ventilation is achieved by breathing

ventricle

a heart chamber that receives blood from the atrium and pumps it to an artery

venule

a small vein that transports blood from a vein to a capillary network

vesicle

a small membrane-bound sac filled with fluid

villus (plural, villi)

a fingerlike projection lining the wall of the small intestine that increases the surface area for absorption of the products of digestion

virus

a non-cellular structure made of nucleic acid and protein that is able to infect a cell and alter the cell's metabolism

Y**Y****Y chromosome**

one of the sex chromosomes in a male human diploid cell. Males have one X and one Y chromosome

yeast

a unicellular, eukaryotic organism, classified as a fungus. Used for fermentation in brewing, baking, and winemaking

Z**zero population growth (ZPG)**

a theory that states that to maintain ecological balance on Earth the human population should remain at a constant size, with each person replacing only himself or herself

zooplankton

microscopic aquatic animals

zygote

the diploid cell that is formed when two gametes fuse

X**X chromosome**

one of the sex chromosomes in a human diploid cell. Females have two X chromosomes

X-linked

an inherited condition caused by a gene located on the X chromosome

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