BIOLOGY

LEVELS OF LIFE

• AUSTRALIAN CURRICULUM EDITION •

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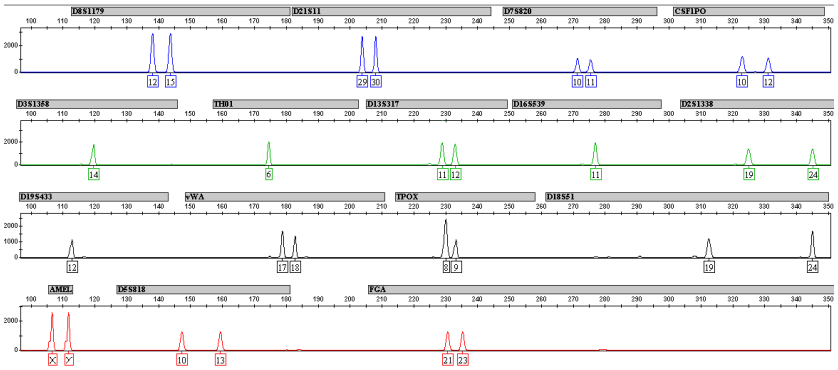
the mapping of the human genome relied on the use of restriction fragment length polymorphism (RFLP) techniques. Palaeontologists have even been able to study the tiny amounts of DNA left in fossils using PCR. Their results have enabled them to identify and classify ancient, long-extinct organisms, as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

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)



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

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

DNA FINGERPRINTS AND PROFILES

tinyurl.com/ydbld8tw

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 It has now largely been replaced by SNP and STR analysis.

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 devloped in collaboration with the FBI in the USA, and European agencies.

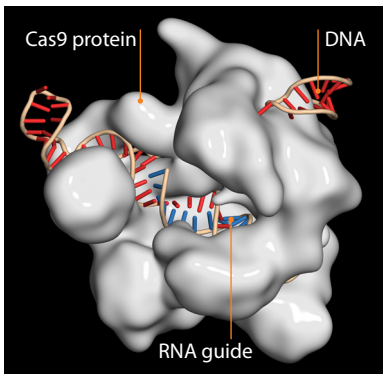


Fig 6.8 CRISPR-Cas9
Cas9 protein - white,
DNA - red nucleotides
RNA guide - blue nucleotides

THE DISCOVERY OF CRISPR

tinyurl.com/hwxvcqf

CRISPR

tinyurl.com/1t35o5v

CRISPR GENE EDITING

tinyurl.com/y8sr8q24

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

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.

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).

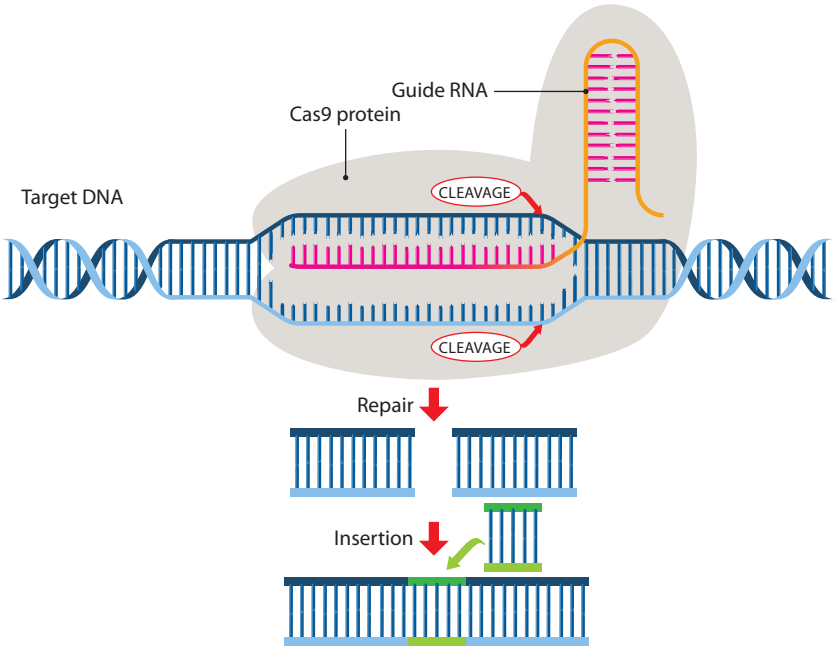


Fig 6.9 CRISPR-Cas9

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.

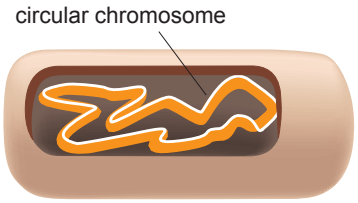


Fig. 7.7 Circular chromosome in a bacterial cell

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

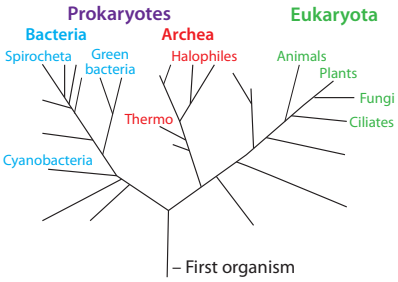


Fig. 7.8 Phylogenetic Tree of Life

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.

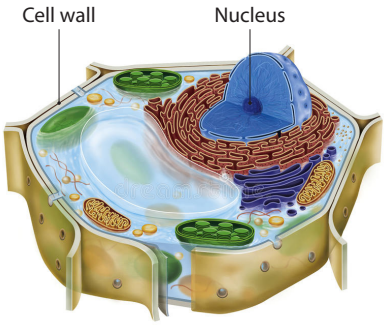


Fig. 7.9 A eukaryotic (plant) cell

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

Diffusion, facilitated diffusion, and osmosis are passive processes.

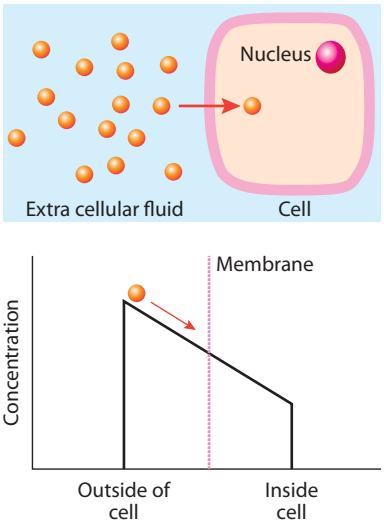


Fig. 10.5 Downhill - with the concentration gradient

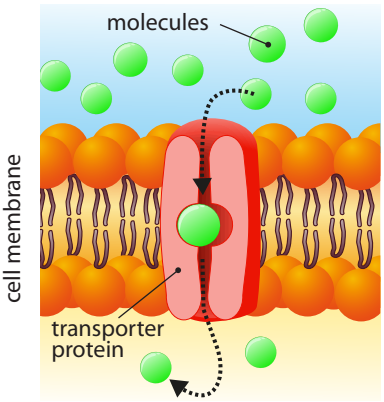


Fig. 10.6 Facilitated diffusion

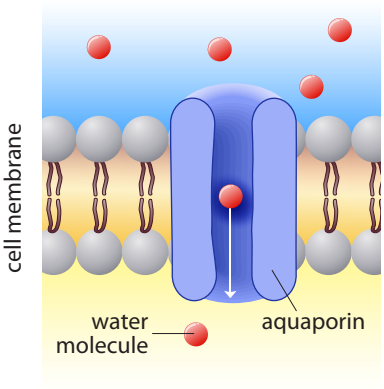


Fig. 10.7 An aquaporin - osmosis

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.

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.

BOTOX® – AN UNEXPECTED CONSEQUENCE

A bacterium called *Clostridium botulinum*, associated with some types of food poisoning, produces a neurotoxin that reduces the release of actylcholine 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.

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.

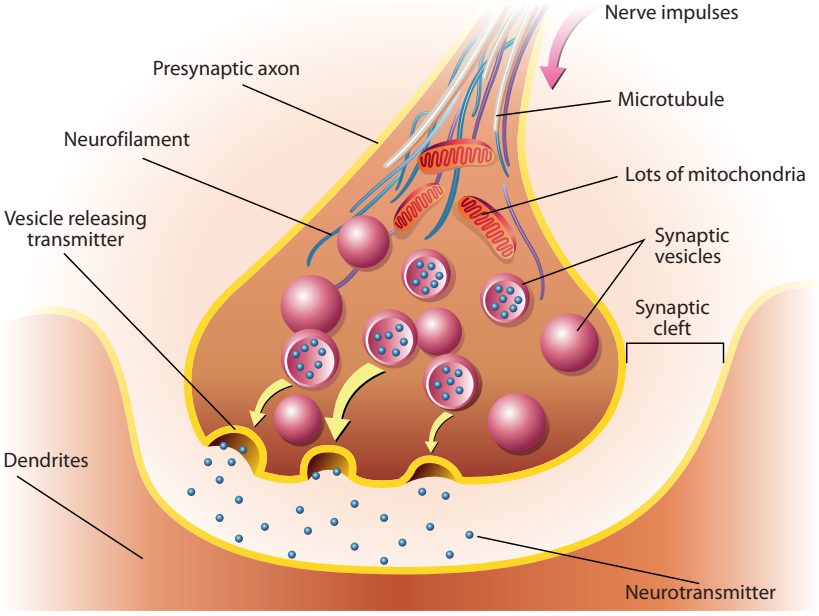


Fig. 17.7 Synapse and synaptic cleft

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 at 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)

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 role of antidiuretic hormone (ADH) 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 **collecting ducts** in the kidneys. It makes the 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)

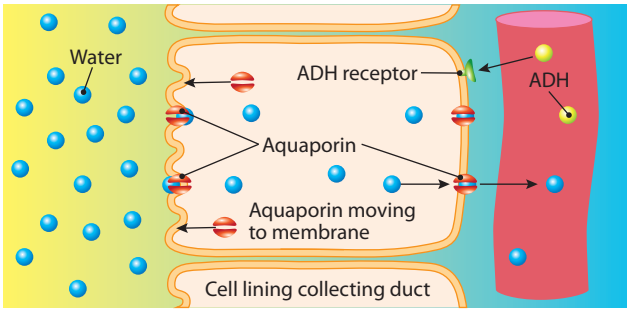


Fig. 19.5 The action of ADH on collecting ducts

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**.

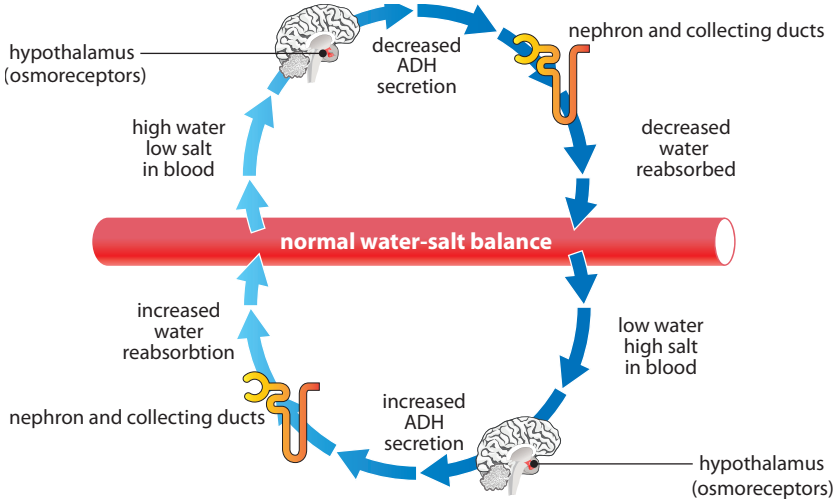


Fig. 19.6 Osmoregulation

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/yynvnnr5

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 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)

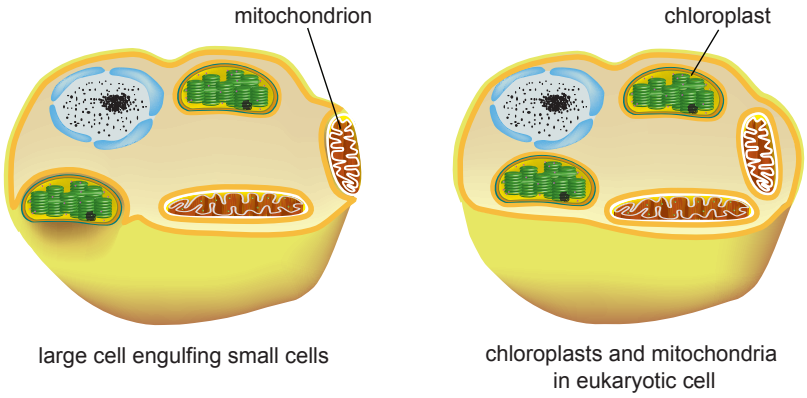


Fig. 20.3 Endosymbiosis

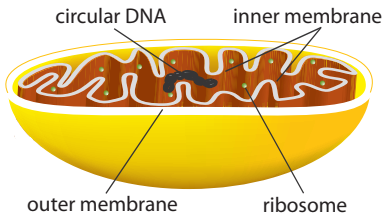


Fig. 20.4 Mitochondrion

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).

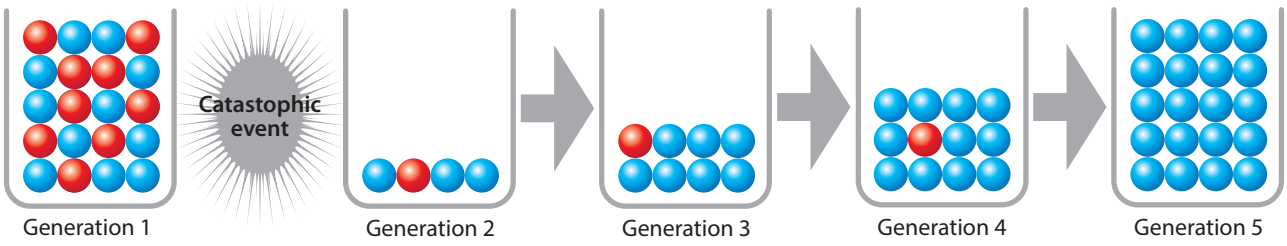


Fig. 23.6 Genetic drift

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.

Study Questions

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?

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