THE VOICE OF THE GENOME

Microscopes

+ Advantages of the light microscope

- Living plants and animals or parts of them can be seen directly.
- Light microscopes are relatively cheap and available.

− Disadvantages of the light microscope

- Artefacts might be produced during preparation, so what is seen may be the result of preparation rather than real.
- Light microscopes have limited powers of resolution and magnification.

+ Advantage of the electron microscope

- They have huge powers of magnification and resolution.

− Disadvantages of the electron microscope

- All specimens are examined in a vacuum
- Specimens undergo severe treatment that is likely to result in artefacts.
- Electron microscopes are extremely expensive and we don't have one. (LOL)

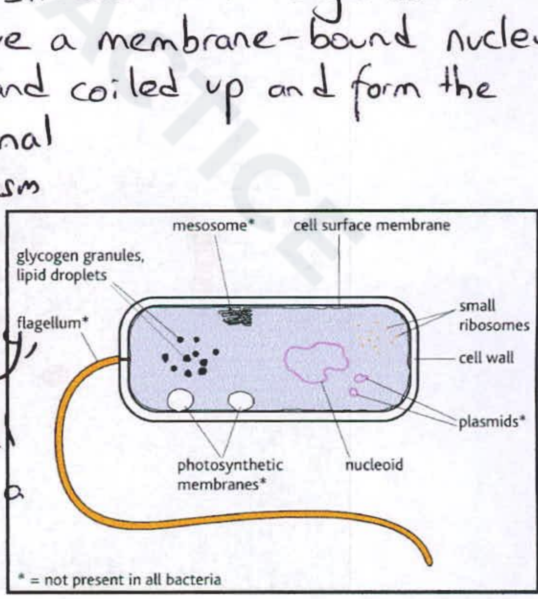
Eukaryotic and prokaryotic cells

Eukaryote – is a cell or organism where the nucleus is surrounded by a nuclear membrane. or eukaryotic cell – a cell with a nucleus and membrane-bound organelles in its cytoplasm.

Prokaryote – a cell that does not have a nucleus or membrane-bound organelles. Bacteria are prokaryotic.

Structure of prokaryotes

Prokaryotic cells lack much of the structure and organisation of eukaryotic cells. They do not have a membrane-bound nucleus. The genetic material is a single strand coiled up and form the nucleoid. Sometimes there are additional bits called plasmids. The cytoplasm contains enzymes, ribosomes and food-storage granules but lacks endoplasmic reticulum, Golgi body, mitochondria and chloroplasts. Respiration takes place on a special piece of the cell membrane called a mesosome.



The ultrastructure of eukaryotic cells

The eukaryotic cell is seen as a bag of organelles, most of which are made up of membranes. Organelle: A structure within the cytoplasm of eukaryotic cells that performs a discrete function. With the exception of ribosomes, organelles are surrounded by at least one layer of membrane. (membrane-bound)

* The fluid around the organelles is an aqueous solution of chemicals called cytosol.

+ The typical animal cell

→ cytoplasm + nucleus = protoplasm
→ protoplasm + cell surface membrane = cell

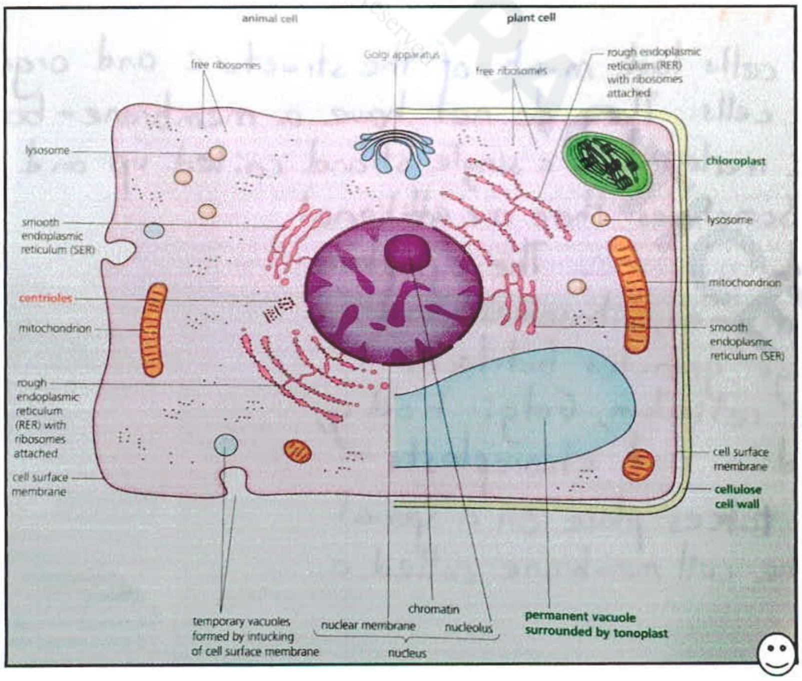
* A typical animal cell has several organelles.

- The nucleus

The nucleus is usually the largest organelle, which is usually spherical in shape, and is surrounded by a double nuclear membrane containing nuclear pores. Inside the nuclear membrane are two main substances nucleic acids and proteins. In the nucleus there is at least one nucleolus - an extra-dense area of almost pure DNA and protein. The nucleolus is involved in the production of ribosomes. Nucleolus is thought to play a part in the control of cell growth and division too.

- Diagram showing the ultrastructure of a typical animal and plant cell:

* mitochondrion:



- Mitochondria

Mitochondrion - organelle which is the site of aerobic cellular respiration. In mitochondria, a series of complicated biochemical reactions take place, energy is released from food by respiration using oxygen. This energy is in the form of ATP.
* Any cell with an energy-demanding function, eg muscle cells or cells that carry active transport, will contain large numbers of mitochondria.

Mitochondria are surrounded by an outer and inner membrane. They also contain their own genetic material. The inner membrane is folded to form cristae surrounded by a fluid matrix.

- The centrioles

Centrioles - structures in an animal cell that are involved in spindle formation during cell division.

- The cytoskeleton

Cytoskeleton is a feature of all eukaryotic cells. Cytoskeleton - 3D web-like structure that fills the cytoplasm, made up of microfilaments and microtubules. This organelle gives the cytoplasm structure and keeps the organelles in place as well as being closely linked with cell movements and transport within cells.

- Vacuoles

Vacuoles are not permanent feature in animal cells. Vacuole - membrane-lined enclosure in a cell formed when needed in animal cells, e.g. digestive vacuoles, contractile vacuoles.

- Endoplasmic reticulum (ER)

Endoplasmic reticulum (ER) - A 3D network of membrane-bound cavities spreading through the cytoplasm. It plays a major role in (protein) transport within the cell.

There are two different ERs:

- Rough endoplasmic reticulum (RER): Endoplasmic reticulum covered in ribosomes involved in synthesis and transport of proteins. The function of ribosomes is to make proteins and once they are made the RER isolates and transports these proteins. Often proteins are transferred from the RER to the Golgi apparatus. Sometimes this occurs by direct contact; sometimes it occurs via vesicles. Vesicle: A small sac of cytoplasm enclosed by membrane. Although they are much smaller than vacuoles, there is no difference between them.

* The RER has a large surface area for the synthesis of all the proteins. Cells that secrete materials have a large amount of RER.

• Smooth endoplasmic reticulum (SER): Endoplasmic reticulum without ribosomes, involved in the synthesis and transport of lipids. SER is separate from the RER and is not usually found near a cell's nucleus. Lots of SER is found in the testes and in the liver.

Tip: The amount and type of endoplasmic reticulum in a cell give an idea of the type of job the cell does.

– The Golgi apparatus

Golgi body is made up of stacks of parallel, flattened membrane pockets called cisternae, formed by vesicles from the endoplasmic reticulum fusing together. The Golgi body has a close link with, but is not joined to, the rough endoplasmic reticulum.

+Steps

- Proteins are brought to the Golgi body in vesicles which have pinched off from the RER where they were made.
- Vesicles fuse with the membrane sacs of the Golgi body and the protein enters the Golgi stacks.
- Proteins are modified in the Golgi body.
- These may be enclosed in vesicles to form an organelle known as a lysosome.
- Alternatively enzymes may be transported through the Golgi body and then in vesicles to the cell surface membrane where the vesicles fuse with the membrane to release extracellular enzymes.

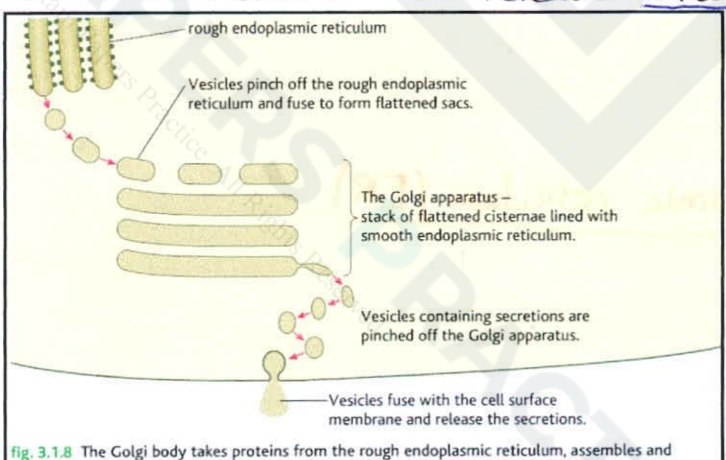


fig. 3.1.8 The Golgi body takes proteins from the rough endoplasmic reticulum, assembles and packages them and transports them to where they are needed. This may be the surface of the cell or different regions inside it.

– Lysosomes

Lysosomes are small spherical vesicles bound by a single membrane. They contain a mixture of hydrolytic enzymes, which are produced by the RER and modified in the Golgi apparatus. Lysosomes are involved in the breakdown of important food vacuoles, old organelles and harmful bacteria that have invaded the body and been engulfed by a phagocyte. A lysosome may also fuse with the outer cell membrane to release extracellular enzymes. Lysosomes can also self destruct; it digests itself in a process called apoptosis.

+ The typical plant cell

Plant cells contain cytoplasm and a nucleus. You will find rough and smooth endoplasmic reticulum spreading throughout the cytoplasm, along with active Golgi bodies. Mitochondria produce ATP which is as vital to the working of the plant cell as it is to the animal cell.

Plant cells contain some structures/organelles that animals do not.

- The plant cell wall

Plant cells tend to be more regular and uniform in their appearance because each cell is bonded by a cell wall. The cell wall is an important feature which gives plants their strength and support. It is made up of largely insoluble cellulose.

• Cellulose

Cellulose is a polysaccharide made up of β-glucose units. Other complex carbohydrates are polymers of α-glucose units. The only difference between α and β-glucose molecules is the way in which the -H and -OH groups are bonded to carbon-1.

* In cellulose β-glucose units are held together by 1,4-glycosidic bonds, where one of the monomer units has to be turned round (inverted) so the bonding can take place. This linking of β-glucose molecules means that the hydroxyl groups stick out on both sides of the molecule. This means hydrogen bonds can form between partially positively charged hydrogen atoms of the hydroxyl groups and the partially negatively charged oxygen atoms. This is known as cross-linking.

* Starch vs cellulose

Hydrogen bonds give cellulose considerable strength. Cellulose molecules remain as very long, straight chains unlike starch molecules which are compact and globular. Starch is an important source of energy in the diet of animals, whereas cellulose is indigestible for most of them.

In the cell wall, groups of cellulose molecles form microfibrils. These cellulose fibrils are laid down in layers held together by a matrix of hemicelluloses and other short-chain carbohydrates.

* In normal circumstances the cell wall is freely permeable to substances. However, the cell wall can become impregnated with suberin in cork tissues, or with lignin to produce wood and become less permeable.

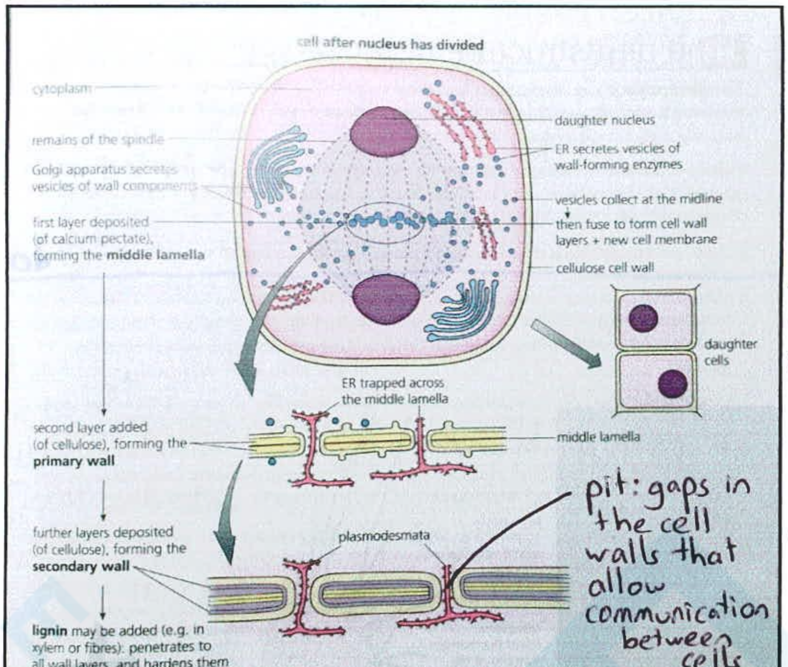


Figure 4.23 Plasmodesmata develop as new cell walls form between plant cells

- The plant cell consist of several layers. The first boundary between new cells is a gel-like layer of calcium pectate called the middle lamella. Some of the endoplasmic reticulum of the parent cell becomes trapped in the gaps in this middle lamella. This trapped reticulum persist when the cellulose wall is laid down, forming plasmodesmata.

- Plasmodesmata:
Cytoplasmic connections between plant cells through gaps in their cell walls. They are part of the symplast pathway through which, for example, inorganic ions are able to pass from cell to cell without having to pass through cell walls or cell surface membranes.

* The cellulose microfibrils and the matrix build up on either side of the middle lamella. To begin with these walls are very flexible, with the cellulose microfibrils all orientated in a similar direction. They are known as primary cell walls. Then forms the secondary cell wall which is inflexible, rigid plant cell wall built up as cellulose microfibrils are laid densely at different angles to each other. This makes the composite material much more rigid. Hemicelluloses harden it further.

-Vacuole

A vacuole is any fluid-filled space inside the cytoplasm surrounded by a membrane. In most plant cells the vacuole is a permanent structure with an important role. Tonoplast - the membrane surrounding plant vacuoles which controls the movement of substances into and out of the vacuole. Vacuole is filled with cell sap that maintains an osmotic gradient.

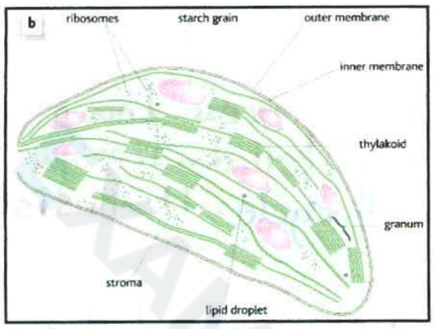
-Functions:

- Maintaining cell shape
- Storage of a number of different substances such as pigments and proteins.

- Chloroplasts

Chloroplast is the organelle in some plant cells which contains chlorophyll and is the site of photosynthesis. Almost all plant cells contain the genetic information to make chloroplasts and so in some circumstances different areas of a plant will become green and start to photosynthesise.

* Chloroplasts contain chlorophyll, the green pigment that is largely responsible for trapping the energy from light, making it available for plant use.
* Chloroplasts are formed from a stem cell known as a leucoplast.



- Thylakoids: The folded inner membranes of a chloroplast.
- Granum: The stacks of thylakoids embedded in the stroma of a chloroplast.
- Stroma: The matrix of a chloroplast, in which grana are embedded.

- Amyloplasts

Amyloplasts are another specialised plant organelle and, like chloroplasts, they develop from leucoplasts. They are colourless and are used to store amylopectin, a form of starch. Amyloplasts are found in large numbers in areas of a plant that store starch.

The organisation of cells

Multicellular organisms are made up of specialised cells. The specialised cells are organised into groups of cells known as tissues. Many tissues are further organised into organs.

• Tissues

Tissues are groups of similar cells that all develop from the same kind of cell. There are only four main tissue types in the human body – epithelial tissue, connective tissue, muscle tissue and nervous tissue.

– Epithelia

→ Squamous epithelium: lining of blood vessels
→ Cuboidal and columnar epithelium: lining of many other tubes
→ Ciliated epithelium: mucus-secreting tissue
→ Compound epithelium: skin, where the surface is scratched

- Organs

An organ is made up of a group of tissues that are grouped into a structure so that they can work effectively together.

- Systems

In animals, in many cases a number of organs work together as a system to carry out large-scale functions in the body.
Most of the cells in tissues, organs and systems have differentiated during development so that they are capable of carrying out their specific function.

Cell division

In prokaryotic cells, cell division occurs by a process known as binary fission, whereas in eukaryotic cells, cell division is part of a regulated process called the cell cycle. It consists of some stages.

The cell cycle in unicellular organisms: asexual reproduction

Unicellular eukaryotic organisms grow quickly under favourable conditions. They then divide into two. Since this division results in the production of new organisms, it is a form of reproduction. As no formation or fusion of gametes is involved, it is called asexual reproduction.

→ Asexual reproduction

(+) → Does not rely on finding a mate
→ Can give rise to large numbers of offspring very rapidly

(−) → The offspring are genetically identical to the parent organism. Changes can cause the total destruction of a group of genetically identical organisms.

Reproduction strategies:

- Binary fission: involves mitosis followed by the splitting of an individual. Seen in simplest groups of organisms.
- Sporulation: involves mitosis and the production of spores which are capable of growing into new individuals. Seen in fungi and plants.
- Fragmentation: occurs when organisms replace parts of their body.
- Budding: Form of asexual reproduction involving an outgrowth from the parent plant which develops into a smaller, genetically identical individual.

The cell cycle in multicellular organisms: growth and repair

Mitosis isn't just about asexual reproduction. It plays a vital role in growth as well. Growth is a permanent increase in the number of cells, or the mass or size of an organism. Dry mass is the most accurate way of measuring growth.

There are three distinct aspects of growth. They are cell division, assimilation and cell expansion.

In multicellular organisms, the life cycle of individual cells is more complex. Here, life begins as a single cell that grows and divides, forming many cells. These new cells allow the organism to grow, eventually making up the adult organism. Only certain of these cells, however, retain the ability to grow and divide throughout life. Even when growth has stopped, these cells are able to replace old or damaged cells providing repair.

* Chromosomes

A chromosome is made up of a coiled threads of DNA and proteins. Chromosomes have the following features.

① The shape of a chromosome is characteristic.

→ Each chromosome has a particular, fixed length. They have a narrow region called the centromere. Centromere – The region where the chromatids are joined together in pairs before separating during cell division.

② Chromosomes occur in homologous pairs.

→ They are called homologous because the two chromosomes have the same shape, and, more importantly, have the same genes in the same order.

③ For each species, the number of chromosomes is fixed.

→ Humans have 46 chromosomes and that makes 23 pairs.

④ Chromosomes are copied prior to division, and chromosomes condense

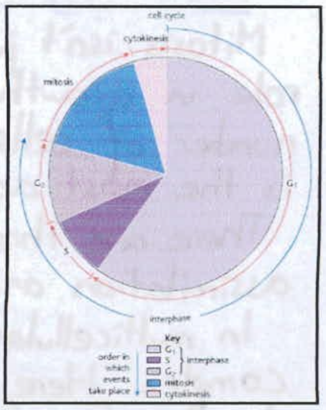
→ When the cell starts to actively divide, the chromosomes condense. This is achieved with the help of positively charged proteins called histones. The DNA winds around the histones to form dense clusters known as nucleosomes. These coil further and give supercoils. In supercoiled areas the genes are not available to be copied to make proteins.

→ Chromosomes duplicate by semi-conservative replication of DNA prior to cell division. Both original and new chromosomes, then, condense.

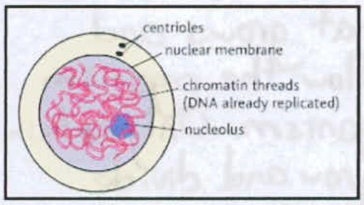
Cell cycle

Cell cycle is the sequence of events making up cell division.
One cell cycle of a eukaryotic cell can be described as:

- Interphase (G_1, S, G_2)
- Mitosis
- Cytokinesis



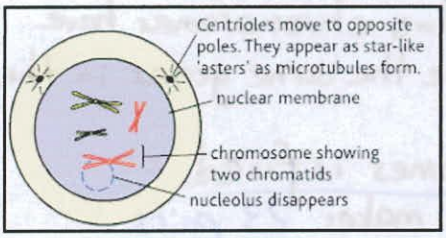
Interphase



Interphase is the period of non-division in the cell cycle and is further distinguished into three stages:

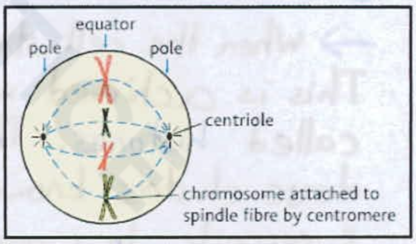
G_1 – the 'first growth' phase	In the nucleus, some genes are 'switched on' and their base sequence is transcribed to pre-mRNA molecules. Editing of pre-mRNA to mature mRNA is also occurring. The cytoplasm increases in volume (grows) by producing new proteins and cell organelles, including mitochondria and endoplasmic reticulum.
S – synthesis	In the nucleus, the semi-conservative replication of DNA occurs. New histones are synthesised and attach to the replicated DNA in the nucleus. Each chromosome becomes two chromatids attached at the centromere Growth of the cell continues.
G_2 – the 'second growth' phase	In the nucleus, replicated DNA is 'double checked' for errors and corrected if any errors are found. If correction is not possible, the cell cycle is normally halted at this G2 phase. Cell growth continues by further synthesis of proteins and cell organelles.

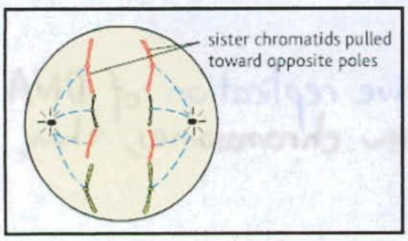
Mitosis (PMAT)



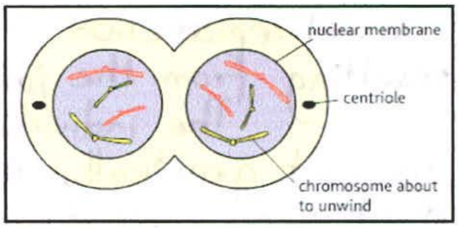
Prophase: The chromosomes coil and condense by a process of supercoiling. Towards the end of prophase, it is possible to see that they consist of two chromatids held together at the centromere. The nucleolus gradually disappears and the nuclear membrane breaks down. The centrioles move to opposite poles and begin to form the spindle.

Metaphase: Spindles have now pulled the chromosomes into the centre of the cell, where they line up on the equator line.





Anaphase: The centromere of each chromosome divides. The spindle fibres attached them shorten, resulting in two chromatids being pulled by their centromeres to opposite poles of the spindle. Once separated chromatids are referred to as chromosomes back again.



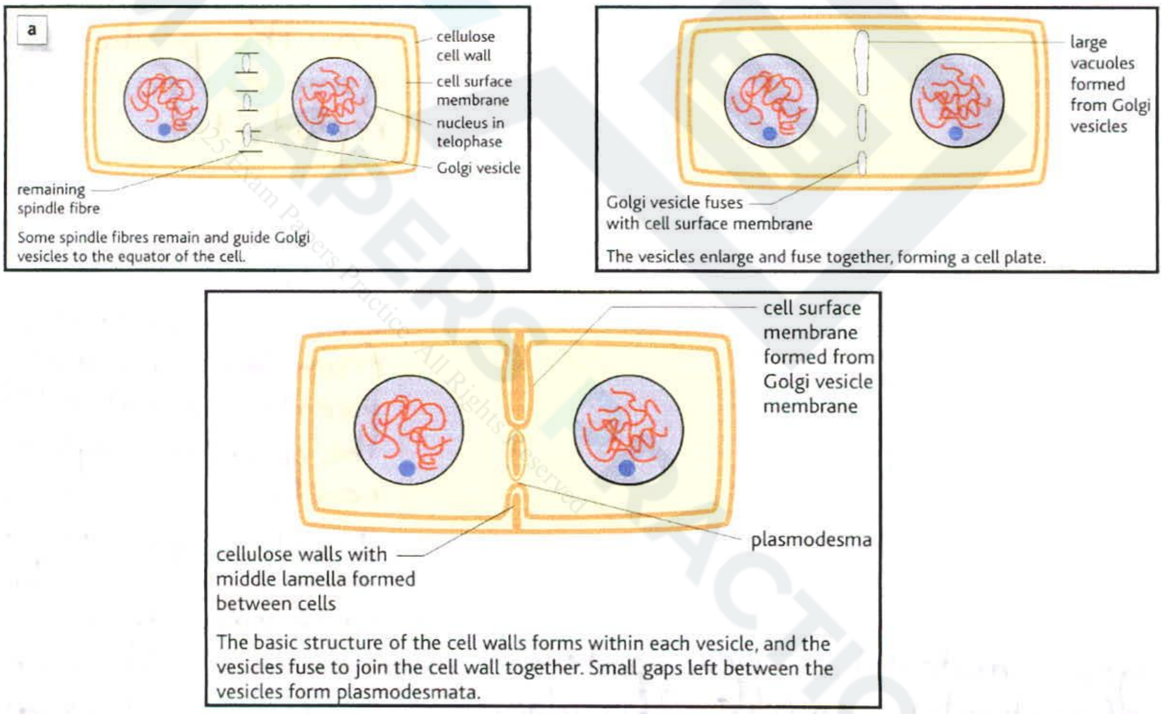
Telophase: A nuclear membrane reforms around both groups of chromosomes at the opposite ends of the cell. The chromosomes decondense by uncoiling, becoming chromatin again. One or more nucleoli reform in each nucleus.

Cytokinesis

Cytokinesis is the division of the cytoplasm to form two daughter cells during cell division. Vesicles from the Golgi apparatus are involved in cytokinesis of both animal and plant cells.

In animal cells, a cleavage furrow (contractile fibres) develops in the middle of the cell. Contraction of this cleavage pinches the cytoplasm in half. As this happens, cell organelles become distributed between the two developing cells.

In plant cells,



Some spindle fibres remain and guide Golgi vesicles to the equator of the cell.

The vesicles enlarge and fuse together, forming a cell plate.

The basic structure of the cell walls forms within each vesicle, and the vesicles fuse to join the cell wall together. Small gaps left between the vesicles form plasmodesmata.

Sexual reproduction

Sexual reproduction is the production of a new individual resulting from the joining of two specialised cells known as gametes. The individuals that result from sexual reproduction are not genetically the same as either of their parents. In a changing environment, this gives a greater chance that one or more of the offspring will have a combination of genes that improves their chance of surviving and going on to reproduce.

Meiosis and the formation of gametes

Meiosis

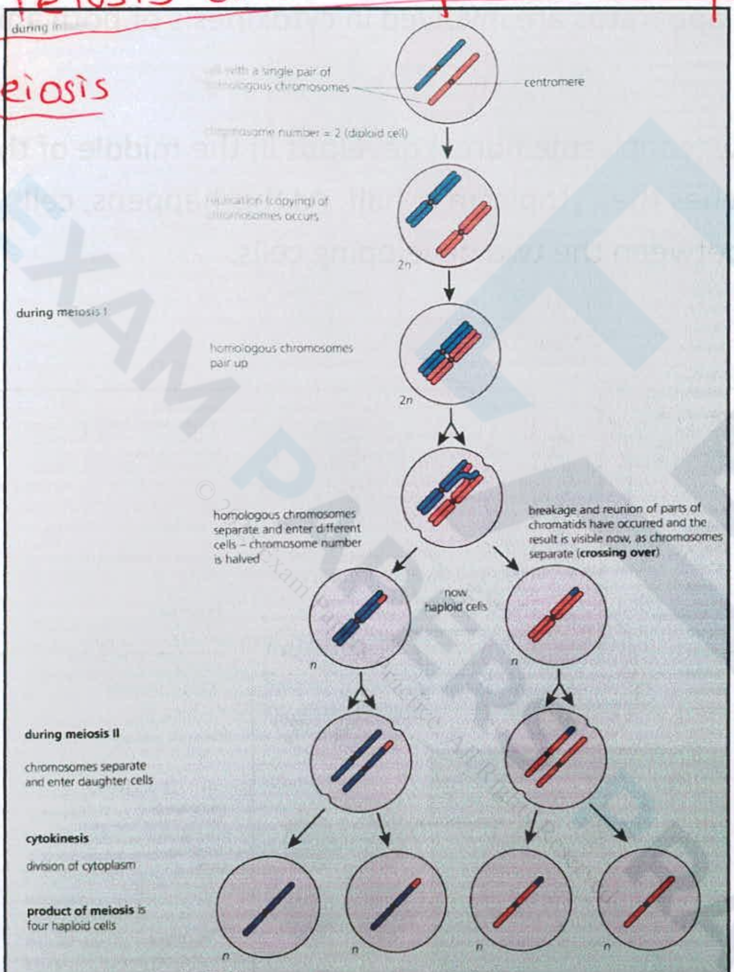


Figure 5.8 An overview of meiosis

Meiosis: Cell division which results in the production of four daughter cells each with half of the chromosome number of the original cell. Meiosis occurs only in sex organs and is involved in the production of gametes for sexual reproduction.

The importance of meiosis

- Independent assortment: the chromosomes that came from the individual's two parents are distibuted into the gametes completely at random. In each new gamete any number from none to all 23 chromosomes could come from either your maternal or your paternal chromosomes. There are millions of combinations possible. This alone guarantees great variation.
- Crossing over: this process takes place when large, multienzyme complexes cut and stitch bits of the maternal and paternal chromatids together. The points where the chromatids break are called chiasmata. These are important in two ways. First, the exchange of genetic material leads to added genetic variation. Second, errors in the process lead to mutation and this is a further way of introducing new combinations into the genetic make-up of a species.

Gametogenesis

The gametes that make sexual reproduction possible are formed in a process called gametogenesis.

Gametes

Gametes are haploid sex cells. Haploid - cell containing a single set of chromosomes (n). Diploid – having two sets of chromosomes in the cell (2n).

Gametes are formed in special sex organs. Many organisms often have temporary sex organs. In more complex animals the sex organs are usually more permanent structures that are called the gonads.

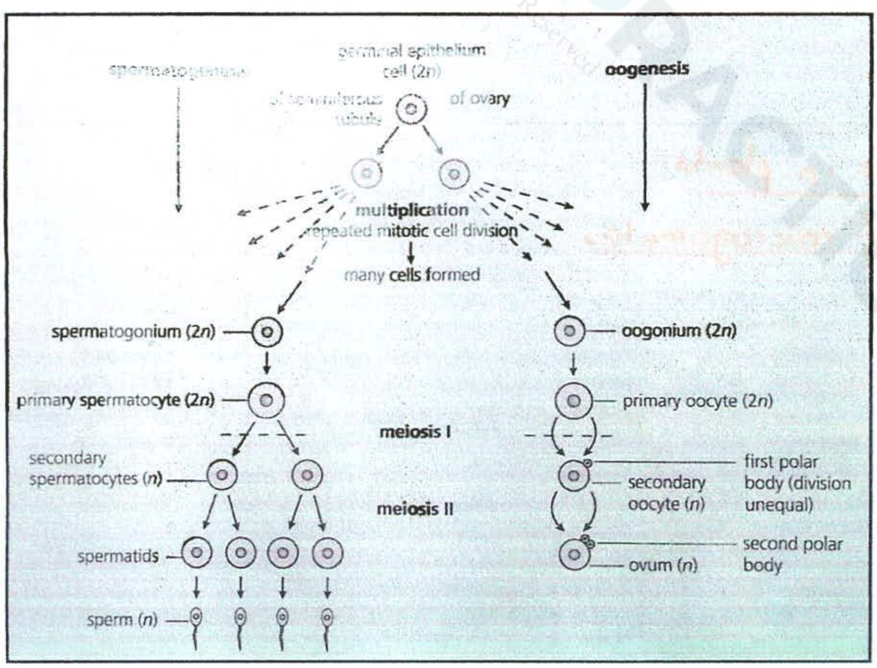
		(male)		(female)
Sex organs →	flowering plants →	anthers	—	ovaries
Sex organs →	animals →	testes	—	ovaries

Gamete production in mammals

Sperm cells in the male mammal are produced in the testes. Ova in the female are produced in the ovaries.

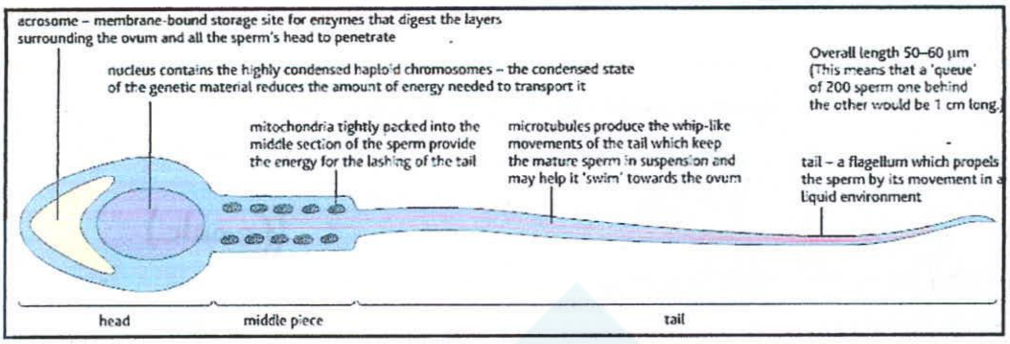
Spermatogenesis is the formation of spermatozoa. Each primordial germ cell in the testes results in large numbers of spermatozoa and there are enormous numbers of primordial germ cells.

Oogenesis is the formation of ova. Each primordial germ cell in an ovary results in only one ovum.



Spermatozoa: many, mini, motile

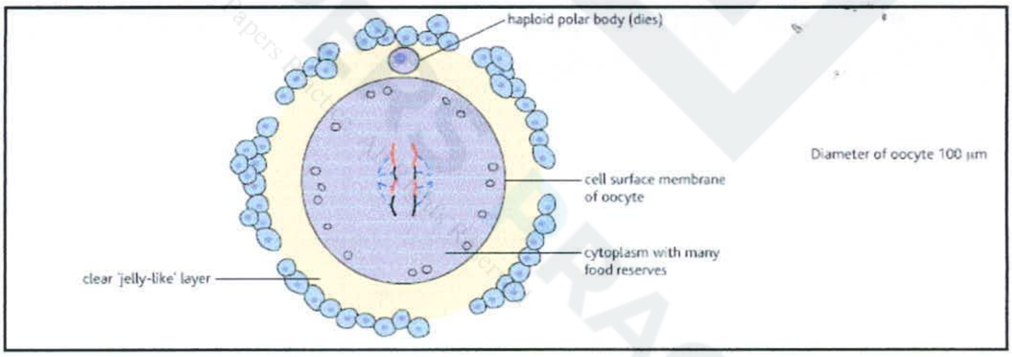
Spermatozoa have several tasks to fulfil. They must remain in suspension in the semen so they can be transported through the female reproductive tract, and they must be able to penetrate the protective barrier around the ovum and deliver the male haploid genome safely inside.



Ova: few, fat, fixed

Although spermatozoa of most animals are very similar in size, the same cannot be said for ova.

Eggs do not move on their own, so they do not need contractile proteins but they usually contain food for the developing embryo. The main difference between eggs of various species is the quantity of stored food they contain.



Gametogenesis in plants

- Formation of microgametes

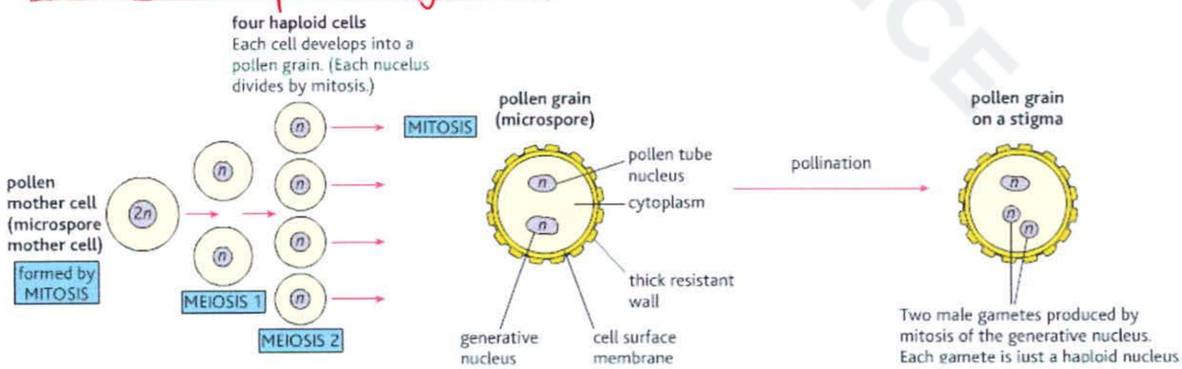


fig. 3.2.8 Microgametogenesis: the formation of the pollen grain.

• Formation of megagametes

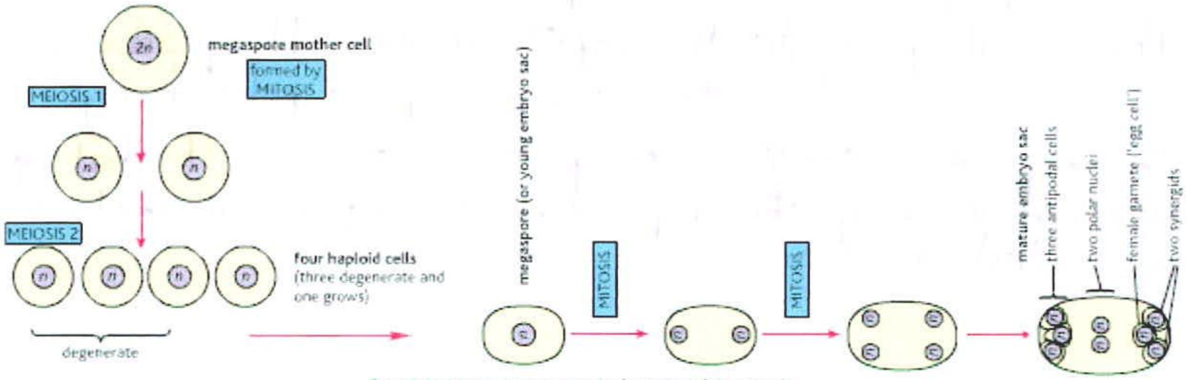


fig. 3.2.10 Megagametogenesis: the formation of the egg cell.

Getting gametes together

For sexual reproduction to be successful, the gametes must meet. In plants, some flowers attract other organisms to transfer the pollen from one plant to another. Others rely on the wind. In animals a wide variety of strategies are used to make sure the gametes meet. They fall into two categories:

- External fertilisation: occurs outside the body, with the gametes shed directly into the environment. This is common only in aquatic species, because spermatozoa and ova dry in the air.
- Internal fertilisation: involves the transfer of the male gametes directly to the female.

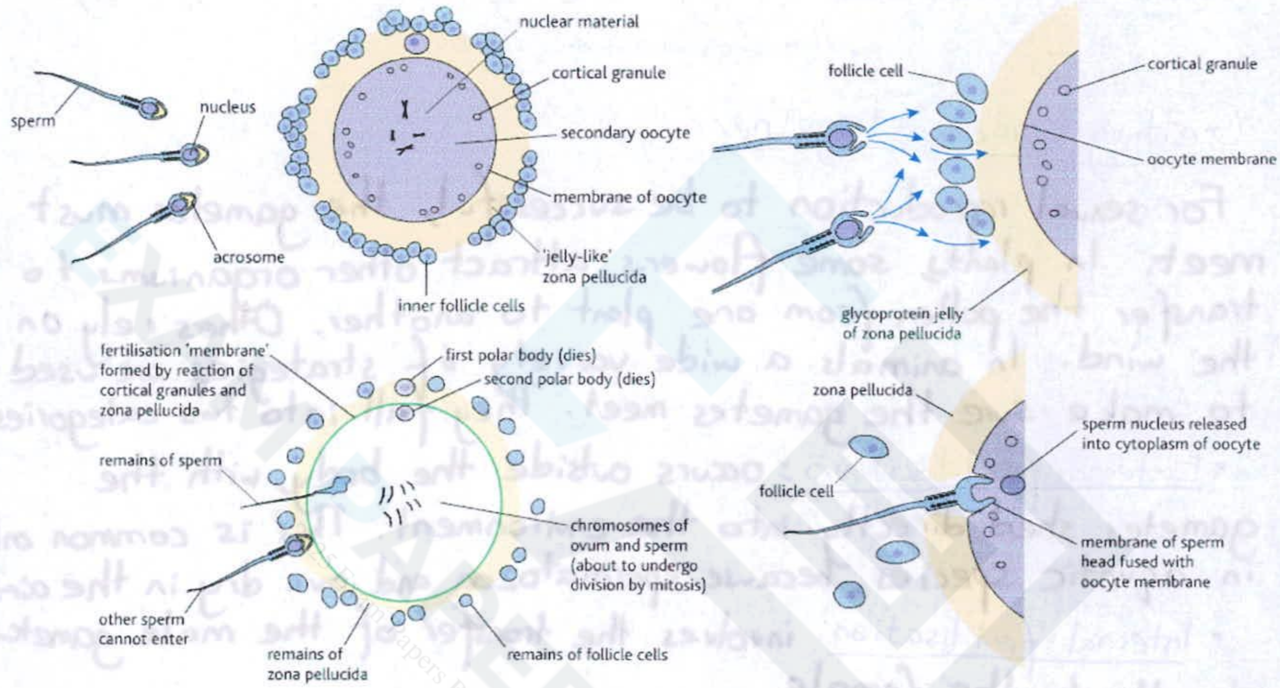
Fertilisation

Fertilisation is the fusion of male and female gamete's nuclei.

• Fertilisation in humans (mammals)

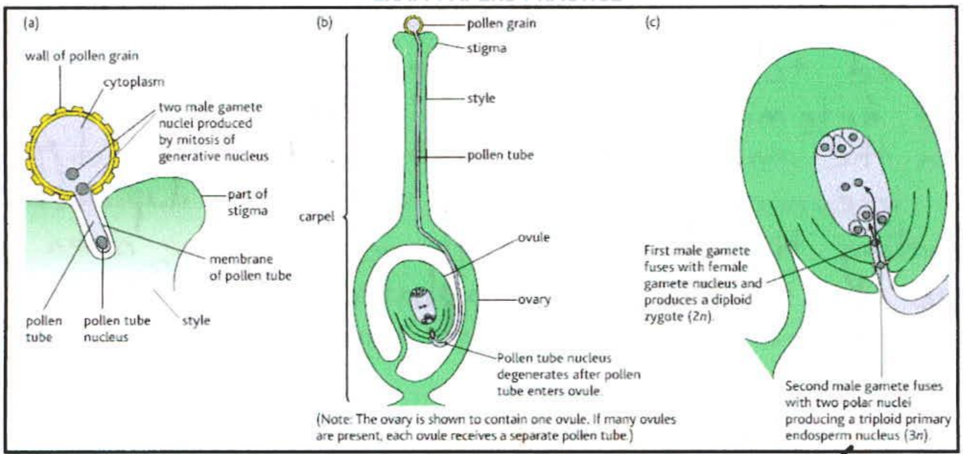
The ovum released at ovulation has not fully completed meiosis and is called a secondary oocyte. It is surrounded by a protective jelly-like layer known as the zona pellucida and also by some of the follicle cells. One or more spermatozoa reach the secondary oocyte and the acrosome reaction occurs. During this reaction, the acrosome swells and fuses wit the cell surface membrane of the sperm, releasing its hydrolytic enzymes. Enzymes released from the acrosomes hydrolyse the glycoprotein from which zona pellucida is made. This allows the passage of the spermatozoa to the surface membrane of the oocyte. Microvilli on the surface of the oocyte then engulf the sperm head. The cortical reaction is initiated, in which

the cortical granules are released from the oocyte. This causes zona pellucida to harden, preventing polyspermy - the entry of more than one sperm in the oocyte.
The secondary oocyte to complete meiosis II, producing a haploid ovum and another smaller haploid polar body.
The haploid nucleus from the sperm now fuses with that of the ovum, forming the diploid nucleus of a cell that is now called a zygote.



• Fertilisation in plants

The pollen grain lands on the surface of the stigma of the flower during pollination. If they recognise each other, pollen grain germinates and a pollen of the same species from a different plant penetrates the carpel.
A pollen tube begins to grow out from the tube cell of the pollen grain through the stigma into the hollow style. It continues to grow down towards the ovary, and the generative cell containing the generative nucleus travels down it. The generative nucleus divides by mitosis to give two generative nuclei. Once the tube has entered the micropyle, the two male nuclei are passed into the ovule so that fertilisation can occur. Flowering plants undergo double fertilisation. One male nucleus fuses with the nuclei of the two polar bodies to form the endosperm nucleus, which is triploid. The other male nucleus fuses with the egg cell to form the diploid zygote.



Embryo development and cell differentiation

In humans the fertilised egg cell or zygote has the potential to form all the 216 different cell types needed for an entire person. It is said to be totipotent. Totipotent - cell with the potential to form all known cell types within an organism.

The first stage of the process is known as cleavage. Cleavage is a special form of mitosis which involves rapid cell division without interphase pauses for assimilation and cell expansion. The result of cleavage is a mass of small, identical and undifferentiated cells forming a hollow sphere known as a blastocyst. One large zygote cell forms a large number of small cells in the early embryo. The tiny cells of the human embryo are known as stem cells. Stem cells are undifferentiated cells, but have the potential to develop into many different types of specialised cells from the instructions in their DNA. The very earliest cells in an embryo are totipotent like the zygote. In the blastocyst the outer layer of cells goes on to form the placenta and the inner layer of cells have already lost some of their ability to differentiate. They are pluripotent. Pluripotent - cells with the potential to form most of the cell types needed in an organism.

blastocyst:

The formation of different cell types

Only after a few days of conception, cells are already predestined to become one type of tissue or another. This cell determination is closely linked to the position of the cells in the embryo. The hypothesis that this differentiation occurs irreversibly at an early stage was disproved by transplanting tiny patches of tissue from one area of an early embryo to another. The transplanted cells differentiated to form the tissue linked to their new position, not their original position.

Cause of cell differentiation

Undifferentiated cells become differentiated cells when they develop a particular structure. This is due to differential gene expression.

- Correct stimulus is given to the unspecialised cells.
  - Internal stimulus: Signal or change from inside the body or cell.
  - External stimulus: Signal or change from outside the body or cell.
- Some genes are switched on and become active; other genes are switched off.
  - The most common way of controlling gene expression is by switching on and off the transcription of certain genes. One way in which this is brought about is by supercoiling parts of chromosomes, preventing the genes there from being transcribed, and uncoiling other areas, opening them up for transcription so that new proteins are made.
- Messenger RNA (mRNA) is made from the active genes only.
- The mRNA moves to the ribosomes; the ribosomes read the mRNA and the appropriate protein is made.
- The protein can permanently alter the structure and function of the cells.

Stem cells and beyond

Embryonic stem cells have the potential to become any cell in the body. Stem cells are:

- undifferentiated (unspecialised) cells
- which can keep dividing
- and that can give rise to other cell types

The earliest embryonic stem cells are totipotent. By ~~around~~ the blastocyst stage, the inner cells of this ball are pluripotent. Pluripotent cells become more specialised as the embryo develops. By around three months of pregnancy the cells have become sufficiently specialised that when they divide they only form more of the same type of cell.

→ James Thomson and his team managed to culture human embryonic stem cells that were still pluripotent. They originally obtained the cells from spare embryos which have been produced during IVF.

Umbilical cord stem cells: The blood that drains from the placenta and umbilical cord after birth is a rich source of pluripotent stem cells. If this blood is frozen and stored, those stem cells will be available throughout the life of a child and be used, should the child need treatment. Drawbacks include large space needed for storing of blood and genetic disorders are present in the stem cells.

Adult stem cells: Adult stem cells exist in the form of undifferentiated cells found among the normal differentiated cells in a tissue or organ. They differentiate when needed. These somatic stem cells are multipotent because they can give a limited range of cells. Adult stem cells have been found in many different organs and tissues. These could be extracted from a patient and treated so that they develop into cells that the patient needs. Using patient's own adult stem cells avoids the risk of rejection of new tissue.

Therapeutic stem cell cloning: Healthy cloned cells are produced from the patient. The cloned human embryo is used as a source of stem cells with DNA that matches the patient perfectly. Stem cells will be harvested from the embryo, which is destroyed in the process. The embryonic stem cells will be cultured in a suitable environment so that they differentiate into the required tissue. These tissue cells will than be transferred to the patient where they can do their job without the immune system rejecting them.

Pitfalls and potential benefits of stem cell therapy

- Scientist are still trying to determine the triggers that control cell differentiation, and scientists need to be able to control this properly to produce the cells needed for a patient.
- There are concerns that stem cells could cause the development of cancers in the body.

\+ There are no cures for many of the conditions that stem cell therapy might solve

→ Organ transplants have a problem. Glycoproteins on the surface of your cell membranes act as part of your cell recognition system. Your immune system recognises your own cells and different cells - and destroys the non-self cells. This is great if you pick up an infection, but potentially lethal if your immune system attacks a transplanted organ. After a transplant people have to take immunosuppresant drugs for the rest of their lives and this puts them at greater risk of infectious diseases.

\+ One major advantage of embryonic stem cell therapy is that it could avoid this risk of rejection.

The ethics of using stem cells

Against:

- Amounts to killing an unborn child / abortion / murder;
- Interfering in nature / may have unforeseen consequences

  Example: may have genetic consequences for future generations / may lead to wider abuses in future / may get out of control / technology may be used in eugenics;

  Example: pressure to harvest human eggs purely for medical use;
- The money could be better used for other types of research

For:

- Spare embryos from IVF would be destroyed anyway;
- The potential to relieve human suffering is too great to ignore; unethical not to allow the research;
- You can't undiscover something / it's going to happen anyway;
- The research could be regulated.

Expressing the genome

The phenotype is the outward expression of a cell or organism due to the interaction of the:

- genotype
- environment.

→ Your genetic make-up obviously plays a very large part in determining your phenotype or apperance. Studies involving plants can be used to investigate the effect of the genotype and environment on the phenotype of organisms.

1 Genotype

- Seeds taken from mature plants so they are genetically different.
- Supply seeds with same environmental conditions.
- Allow to grow into plants.
- Observe any differences.

→ Any differences in height, etc must be due to genotype.

2 Environment

- Plant clones are used. No genetic difference in plants.
- All environmental conditions keep the same except one. (Altering only one environmental factor)
- Allow to grow into plants.
- Observe any differences.

→ Any differences must be due to that altered environmental factor.

* It is more difficult to investigate the impact of environment on the phenotype with animals because of the difficulty of producing large numbers of cloned, genetically identical organisms, and the ethical aspects of examination.

Interaction of genes

Some of your characteristics are very clearly the result of the genes you have inherited. A very few depend on the information carried on a single gene that has just two alternative alleles. Many depend on a single gene that has multiple genes, or are polygenic traits in which phenotypic features result from the interaction of a number of different genes.

Epistasis: In some polygenic traits one gene affects or alters the expression of another. This is known as epistasis.

* Don't confuse epistasis with dominance. Dominance occurs between two alleles of the same gene. Epistasis happens between two genes coding for quite separate phenotypic features, when one locus **masks** the expression of another.

Example of epistasis:

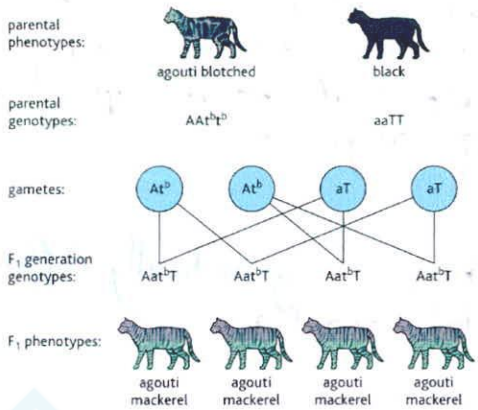


fig. 3.4.1 Coat colour in cats – an epistatic gene in action.

The tabby coat pattern gene has three alleles: **T**, $\mathbf{T^B}$ and $\mathbf{t^b}$. **TT** or $\mathbf{Tt^b}$ gives a mackerel-tabby coat pattern with vertical curving black stripes, whilst $\mathbf{t^bt^b}$ is recessive and gives the classic blotched-tabby coat with swirls of black. $\mathbf{T^BT}$, $\mathbf{T^BT^B}$ or $\mathbf{T^Bt^b}$ gives a ticked or freckled appearance. However, without at least one dominant agouti allele on the agouti locus none of the tabby patterns will show up in the phenotype because the cat will have a solid coat colour.

Variation

In any population of organisms there are two types of variation.
- Discontinuous variation
- Continuous variation

Discontinuous variation: It is shown by features that are either present or not, such as sex. These features are generally determined by one or at most a very few genes, and the environment does not usually have an effect.

Continuous variation: Some phenotypes are affected by alleles at many loci (**polygenic inheritance**) as well as the **environment** and this can give rise to phenotypes that show **continuous variation**. Characteristics that show continuous variation include weight and height in an animal species.

Studying continuous variation

When studying continuous variation in a population you need to take **large samples**. You also need to collect your large sample **randomly** from as much of the organism's habitat as possible. By doing these you can get an accurate picture of the whole species. Data like these can be displayed using a graph or histogram, to show the frequency distribution of the characteristic clearly.

* **Polygenic inheritance:** More than one gene is involved in influencing the phenotype. The genes will be at different **loci**.

Normal distributions and standard errors (HSW)

If a feature of an animal or plant shows continuous variation, then the frequency will be a normal distribution curve. Considering heights of human female population,
→ The mean height is exactly at the centre of the normal distribution curve, and all the heights are distributed equally about the mean.
It forms a bell-shaped curve, which will be found studying many kinds of variation.
Standard deviation: The standard deviation is a statistical measure of the amount of difference from the mean in the sample. About 68% of the sample will be within 1 s.d. of the mean, 95% will be within 2 s.d. ~~within~~
* Smaller standard deviation, the more reliable results are.

Studying variation in humans

There are several difficulties in studying the interaction of genotype and environment in human beings. It is very important during any experiment that all the organisms are subjected to same conditions. Then as far as possible any differences between them can be seen as the results of genetic differences alone. But in human beings, imposing conditions like these is impossible.

Twin studies

One strategy is to consider genetically identical individuals to answer questions about the interaction of nature and nurture. Identical twins are human clones. Non-identical twins are normal siblings which have a similar environment. Ordinary siblings are useful as a control group.
In twin studies, the differences between identical twins are studied and compared with the differences between non-identical twins and normal siblings.
Identical twins who are separated at birth provide a rare and useful resource. If twins who have been reared apart show strong similarities in a trait, this suggest that the influence of the genotype on the characteristic is very strong.
If twins reared together are quite similar for a trait, but twins reared apart show a greater difference, it suggest that the environment has a stronger influence on that characteristic.

Genes and environment in human features (Examples)

- Monoamine oxidase A

Monoamine oxidase A is an enzyme found in the nervous system, the liver and the gut, where it breaks down monoamines. Many of the chemicals involved in transmitting nerve impulses across the synapses (neurotransmitters) are also monoamines.

If too much or too little of the enzyme is formed, this can lead to a number of different illnesses and problems.

→ Low levels of MAOA seem to be linked not just to depression but also to addictive behaviour and in some cases criminal behaviour.

→ High levels of the enzyme have been linked to risk-taking and aggressive behaviour.

- Cancers

Cancer cells do not respond to the normal mechanisms that control cell growth and the cell cycle. They divide rapidly to form a mass of abnormally growing cells (a tumour) which invades the surrounding tissues and other parts of the body in a splitting process called metastasis. A such tumour is known as a malignant tumour.

* The causes of cancer are many, but they include both genetic and environmental factors.

- Genetic ↦ The mutations that result in cancer affect the control of the cell cycle, usually by disrupting the chemical systems that control the stages of the cycle.

→ Mutations interfering with the accurate replication of DNA.

→ Protooncogenes mutate to give oncogene which produces uncontrolled amounts of proteins which stimulate the cell cycle so the cell cycle is constantly stimulated causing cancer.

→ Tumour suppressor genes give breaks to the stages of cell cycle.

- Environment ↦ Environmental factors play an important part in cancer by increasing the likelihood of these cancer-causing mutations occuring. So factors such as the tar in cigarette smoke, the chemicals in alcoholic drinks, asbestos and ionising radiation all increase the likelihood of a mutation in your DNA which in turn causes cancer.

e.g: Melanoma → cancer caused by UV rays from the Sun.

BIODIVERSITY AND NATURAL RESOURCES

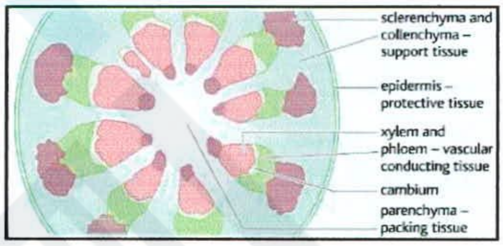
The structure of plant stems

The primary function of a stem is support, to hold the leaves in the best position for obtaining sunlight for photosynthesis and flowers for pollination. The stem provides flexibility and has the strength to stay upright.

The other major function of stems is the transport of materials about the plant. Most stems are green — that is, they contain chlorophyll but photosynthesis is not their major function.

- The tissues that make up the stem:

- Epidermis: The outer layer of the stem is the epidermis, which protects the cells beneath it. Epidermal cells secrete cutin, a waxy substance which helps to prevent water loss from the stem and protects against the entry of pathogens.

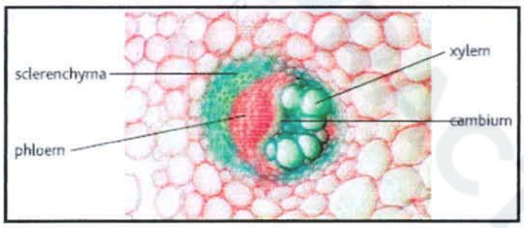


- Parenchyma: These are unspecialised cells which are the most common type of plant cells. Some of the parenchyma in the stem is modified into collenchyma and sclerenchyma.
- Collenchyma: Collenchyma cells have thick primary cell walls which give the tissue its strength. These cells are found around the outside of the stem, just inside the epidermis, and they give plenty of support but remain living, so stretch as the plant grows.
- Sclerenchyma: Strengthening and supporting tissue found in plants, made up of heavily lignified, thick walled cells. It develops as the plant gets bigger, to support the increasing weight of the upper part of the plant. Sclerenchyma tissue is found around the vascular bundles in older stems and in leaves. All sclerenchyma cells have secondary cell walls made of cellulose microfibrils laid down at right angles. Lignin is deposited on the cell walls of these fibres and this makes the fibres strong yet flexible. The strength of the fibres depends on their length and how much they are lignified. Once the fibre is lignified the cell contents die because lignin is impermeable to water, and so the fibres are hollow tubes. Sclerenchyma cells can also become completely impregnated with lignin, when they form sclereids. Sclereids are plant cells with thick, lignified walls.

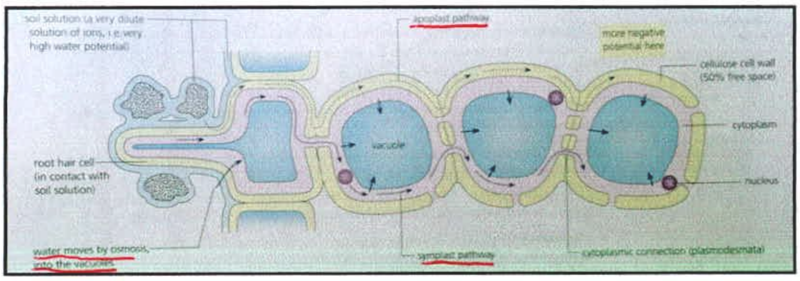
Transport in plants

Vascular bundles are found through the plant, including the stem, and they contain the transport tissues xylem and phloem.

- **Xylem**: Tissue carries water and dissolved minerals (transport) up the transpiration stream and provides support to woody plants.
  → Xylem starts off as living tissue. The first xylem form is called protoxylem. As the vessels grow, a complex carbohydrate polymer called lignin is laid down in the spaces between the cellulose fibres of the cell wall. This makes them much stronger and supportive but also impermeable, so the cytoplasm dies. The end walls between the cells break down so the xylem forms hollow tubes ideal for water transport.
  → In order to allow water and ions to be transferred to all parts of the plant, vessels have pits. These areas of the cell wall are where there is no lignin and water can move laterally as well as up the stem.
  → The lignified xylem vessels are very strong and play a very important supportive role in the stems of particularly larger plants. In smaller plants, support mainly comes from the turgid parenchyma cells in the centre, and the sclerenchyma and collenchyma.
- **Phloem**: is living tissue which transports the dissolved product of photosynthesis both up and down the plant to the site where they are needed.
- **Cambium**: is a layer of unspecialised cells which differentiate to cells that form both the xylem and the phloem.



The uptake of water by plants



The three routes of water movement through plant cells and tissues are shown in the figure. These are as follows:

- Mass flow: through the interconnected free spaces between the cellulose fibres of the plant cell walls. This pathway does not pass through membranes or the living contents of the cell and is known as the apoplast pathway. It is the major route of water transport.
- Diffusion: through the cytoplasm of the cells using the channels through cell walls called plasmodesmata. This is called the symplast pathway. Organelles and molecules slow diffusion, so this is a much restricted pathway.
- Osmosis: from vacuole to vacuole of plant cells using their partially permeable membranes. This is called the vacuolar pathway. This is driven by the gradient of water potential. Root hair cells take up water from the soil in this way as their contents have a much more negative water potential than the very dilute solution of ions found in soil water.

Movement of water up xylem vessels

The movement of water in the xylem depends on transpiration, which is the loss of water vapour from the surface of the plant, mainly from the leaves.

→ The evaporation of water from the aerial parts of the plant means that a tension is applied to the water column in the xylem vessels. This pulls water molecules up, but is insufficient on its own.

→ Water molecules are attracted to each other by hydrogen bonds, giving water a property called cohesion. Water therefore has great tensile strength and can be drawn upwards to a great height without breaking.

→ Water molecules are also attracted to molecules in vessel walls because of adhesion. This also assists in their movement up the xylem without the column breaking.

The importance of inorganic ions to plants

- Nitrogen: Nitrogen ions are used to make amino acids and therefore proteins. Nitrates are also needed for the plant to make DNA and many hormones, as well as a range of other compounds in plant cells.

+ When plants lack nitrates the older leaves turn yellow and die, and growth is stunted.

• Calcium: Calcium ions in the middle lamella of plant cells combine with pectin to form the calcium pectate which holds the plant cells together. Calcium ions also play a role in the permeability of membranes.

+ When plants lack calcium the growing points die back and the **young leaves** are yellow and crinkly.

• Magnesium: Magnesium ions are needed to produce the green pigment chlorophyll. Magnesium is also needed for the activation of some plant enzymes and the synthesis of nucleic acids.

+ Without magnesium yellow areas develop on the older leaves and growth slows down.

* To get minerals into the cells they often have to be moved up a concentration gradient by active transport. Any minerals dissolved in water absorbed from the soil are carried in the apoplast pathway.

Plants as natural sources

- Microfibrils

Cellulose microfibrils are arranged differently in primary and secondary cell walls:

• Primary cell wall: microfibrils laid down in a criss-cross manner, embedded in a glue of hemicelluloses and pectins. Lignin is never present in these cell walls.

• Secondary cell wall: microfibrils laid down in sheets with each sheet having the cellulose microfibrils running in one direction only. These are embedded in hemicelluloses and pectins. Lignin is also present in these cell walls.

- Functions of starch, cellulose and cellulose microfibrils and plant fibres in plant cells

• Starch is the energy storage molecule. It is made up of many α-glucose units which can be used in respiration.
• Cellulose is a major component of plant cell walls formed of many β-glucose units.
• Cellulose microfibrils have many hydrogen bonds that together make it strong. In addition, the arrangement of cellulose microfibrils and the glue that binds them give strength and flexibility to plant cell walls whilst allowing them to be fully permeable to water.
• Fibres (schlerenchyma and xylem) give support and some allow the transport of water and mineral ions.

Useful properties of cellulose and plant fibres

• Fibres

The fibres are very long schlerenchyma cells and xylem tissue and are usually very tough. Cellulose and lignified cellulose are not easily broken down either by chemicals or enzymes. On the other hand, the matrix of pectates and other compounds around the fibres can usually be dissolved or removed.

Plant fibres have great tensile strength - they cannot easily be broken down by pulling. This, along with their flexibility, makes them very useful.

• Wood

Wood is a composite material, made of lignified cellulose fibres embedded in hemicelluloses and lignin. The cellulose fibres make the wood very resistant to compression (compressive strength). Wood also retains some of the flexibility of the matrix. Lignified plant fibres are very resistant to chemical and enzyme breakdown. This property makes plant fibres, as wood, a good choice as a building material.

→ Wood also locks up carbon dioxide and is a sustainable resource if it is managed carefully with replanting programmes. Even if it is burned, wood can be carbon neutral.

Plant fibres, starch and sustainability

- The use of fossil fuels is not sustainable as they are non-renewable resources.
- Both starch and plant fibres are renewable sources as they come from plants.
- Both can be burned to release heat energy.
- Starch can be processed into bioplastic to replace oil-based plastics whilst plant fibres can be used for rope.

Bioplastics

Plastics are synthetic polymers - long-chain molecules that are made up of repeating units of small monomer molecules. Most plastics are polymers made from petrochemicals, originating from oil which is a non-renewable resource. These plastics are non-biodegradable.

Scientists are increasingly looking at the possibilities of bioplastics - plastics based on biological polymers such as starch and cellulose.

Bioplastics have two large potential benefits:

- They are a sustainable resource. Plants can be grown easily to supply the needs of the bioplastics industry
- Bioplastics are biodegradable. Because they are based on biological molecules, bacteria and fungi can usually break them down, even if the process can be very slow.

+Will bioplastics take over from oil-based plastics?

→ The science and technology needed to produce them are becoming increasingly available. However, the plastics made from petrochemicals have extremely useful properties. Economics and ethical considerations are also important.

Bioplastics are still much more expensive than the oil-based alternatives. The technology is still new and economies of scale plays a role.

Ethically, there aren't enough crops to feed the world's rapidly increasing population. In the face of limited crops, who decides whether they are used for food to satisfy the immediate hunger of people around the world, or for biofuels or bioplastics to try and work towards a sustainable future for everyone?

Plant pharmacies

Plant produce a vast range of chemicals, some with the function of deterring animals that try to eat the plant or of destroying microorganisms that might cause disease. These chemicals can be extracted and used in the production of drugs.

One major advantage of extracting and purifying the beneficial drugs found in plants is that it is possible to give known, repeatable doses of the active ingredient.

-Once the active ingredient is detected in a plant, it is produced artificially for mass production.

Both for individuals and for society the impact of these drugs is far-reaching. People are ill less often, they are less severely ill and they are living longer.

William Withering's digitalis soup

- Withering, a doctor, learned of a remedy for treating a heart condition.
- He isolated the active ingredient from the 20 or so ingredients in the remedy. It was digitalis from foxglove.
- He trialled different doses on patients to find the most effective treatment.
- He carefully recorded all his findings.

Modern drug development

Every medicine that comes onto the market today is the result of years of research and development. A new medicine has to be:
- effective
- safe
- stable
- easily taken into and removed from the body
- capable of being made on a large scale

→ Researchers often use computer models to fit new structures into the active site of enzymes or receptors that are thought to play a significant role in disease processes. A compound is the produced.

→ The new compound is first tested on cell structures, tissue cultures and whole organs in the lab.

→ Animal experimentation follows this where mammals are used which are as similar as possible to humans. Animal testing is very expensive and time consuming and is the centre of much ethical debate.

→ Then follows the clinical trials.

Contemporary drug testing protocols

If the animal testing has been successful, the very first human trials follow.

Three-phase testing

Phase 1	Phase 2	Phase 3
•Details -A few healthy people are used -Range of doses •Function -To check it does not have any unexpected side effects -That it behaves in the manner predicted from animal tests.	•Details -c. 100-500 patients with the condition used •Function -Finding out more about the ideal dose -Identifying side-effects -To check that it works on the condition as anticipated	•Details -c. 5000 people are used -Normally involves a double blind trial •Function -To collect as much data as possible including effectiveness compared to placebo and standard treatment and frequency of any side effects

Placebo

Name given to tablet/treatment that appears identical in all ways to the drug except that it is chemically inactive.

Double blind trial

- Patients are randomly divided into two groups.
- One group recieves drug/treatment and the other group recieves the placebo or standard treatment.
- Neither the patients nor those recording any changes in the patients know who has received the drug and who has received the placebo or standard treatment. This reduces the chances of bias.

Modern protocols vs William Withering's digitalis soup

Similarities:

- Both isolated a possible drug/treatment.
- Both initially tested on a small number number of patients and then a larger group of patients.

Differences:

- Only modern protocols test on animals before phase 1.
- Only modern protocols have phase 1 testing where drug is tested on healthy people.

→ Some consider phase 1 testing unethical, because most commonly less well off people volunteer. One way to resolve this might be by not offering money to volunteers.

- Only modern protocols have double blind trials, including using a placebo, undertaken to collect data for statistical analysis.

Species and evolution

- What is a species?

Species is a group of closely related organisms that are all potentially capable of interbreeding to produce fertile offspring.

→ Scientists make decisions about which organisms belong in the same species, and how they are related, in a number of ways. Originally scientists just looked closely at the outer and sometimes inner appearance or morphology of the organism. However, the appearance of an organism can be affected by many different things, and there can be a huge amount of variation within a group of closely related organisms.

→ Today there are more sophisticated ways of comparing organisms. As many different characteristics as possible are used in order to define and identify a species. The main characteristics are:

- morphology and anatomy (external and internal structure)
- cell structure (whether cells are eukaryotic or prokaryotic)
- physiology (blood composition) and chemical composition

(comparisons of nucleic acids and proteins, and the similarities in proteins between organisms)

Molecular phylogeny: Analysis of different chemicals and genes in different organisms to identify interrelationships.

* The evidence from biochemical analysis may support or conflict with relationships based on morphology. For example, molecular phylogeny has revealed that there are in fact three domains.

A new taxonomic grouping

All life was thought to be split into prokaryotes and eukaryotes. Then came the discovery of species of bacteria that survive and prosper in extremely hostile environments. These are called extremophiles.

These microorganisms of extreme habitats all have cells that we can identify as prokaryotic. However, the larger RNA molecules present in the ribosomes of extremophiles were discovered to be different from those of previously known bacteria. Further analyses of the biochemistry of extremophiles, by means of molecular phylogeny, in comparison with that of other groups, suggested new evolutionary relationships and led on to a new scheme of classification.

* Molecular phylogeny compares the structure of a particular molecule from different organisms to discover their degree of evolutionary relatedness.

As a result, we now recognise three major forms of life, called domains. The organisms of each domain share a distinctive, unique pattern of ribosomal RNA and there are other differences which establish their evolutionary relationships. These domains are:

- the Archaea (the extremophile prokaryotes)
- the Eubacteria (the true bacteria)
- the Eukaryota (all eukaryotic cells)

Characteristic	Bacteria	Archaea	Eukaryota
Membrane-enclosed nucleus	Absent	Absent	Present
Membrane-enclosed organelles	Absent	Absent	Present
Peptidoglycan in cell wall	Present	Absent	Absent
Membrane lipids	Ester-linked, unbranched	Ester-linked branched	Ester-linked unbranched
Ribosomes	70S	70S	80S
Initiator tRNA	Formylmethionine	Methionine	Methionine
Operons	Yes	Yes	No
Plasmids	Yes	Yes	Rare
RNA polymerases	1	1	3
Ribosomes sensitive to chloramphenicol and streptomycin	Yes	No	No
Ribosomes sensitive to diphtheria toxin	No	Yes	Yes
Some are methanogens	No	Yes	No
Some fix nitrogen	Yes	Yes	No
Some conduct chlorophyll-based photosynthesis	Yes	No	Yes

Evalvation by the scientific community

The idea of three domains illustrates the scientific process and the important role of critical evaluation of new data by the scientific community.

1 Scientific journals

All scientists publish full details of their investigations in well-known scientific journals. Their reports must contain full details of their methodology, the original data and an analysis of their findings, following some strict rules.

2 Peer review

Other scientists check the details of the method, the data collected and the validity of conclusions. Peer reviewes often ask for more details or a revision of conlusions before approving its publication.

3 Conferences

Universities and other institutions often host meetings of scientists from around the world specialising in one particular area of research. The most important function of these meetings is to allow individuals to share ideas, discuss common problems and argue their case where different models are proposed.

4 Evidence from other scientists

The evidence will help to support the suggestion, to reject the suggestion or lead to a modification of the suggestion.

Ecology and adaptation

- Ecology: The study of the relationships between living organisms and their environment.
- Ecosystem: Environment including all the living organisms interacting, the cycling of nutrients and the physical and chemical environment in which the organisms are living.
- Habitats: Place where an organism lives.
- Community: All of the populations of living organisms which live in a habitat at any one time.
- Population: Group of organisms of the same species, living and breeding together in a particular niche in a habitat.
- Niche: The role of an organism within an ecological community. Ecological niche describes not only where a species lives but all of its activities, its predators and how it interacts with the non-living environment around it.

Adaptation to niches

A successful species is adapted well to its niche, meaning that individuals in that species have characteristics that increase their chance of survival and reproduction, and therefore of passing those characteristics on to the next generation. There are 3 main types of adaptation that improve the changes of survival. Most organisms show more than one type.

Type of adaptation	Description	Examples
Anatomical adaptation	A physical/structural adaptation – it may be external or internal	Kidneys of some desert mammals have long loops of Henle to reduce water loss by producing very concentrated urine.
Behavioural adaptation	A change in the behaviour of an organism to increase its survival chances.	Some ectothermic (cold-blooded) organisms such as lizards orientate themselves to maximise their absorption of heat from the Sun until they reach their active temperature.
Physiological adaptation	These tend to be changes in the internal biochemical functioning of the organism in response to an altered environmental stimulus.	People who move from sea level to high up a mountain slowly increase their oxygen-carrying capacity by producing more erythrocytes.

Natural selection

Natural selection is the process by which the organisms best suited to a particular environment are most likely to survive and pass on their advantageous genetic characteristics to their offspring.

* Natural selection can lead to adaption and evolution.

The evidence and arguments for natural selection are as follows:

① The individuals in a species are not identical, but show variations in their characteristics. Gene pool – All the different versions of genes found in a population. Genetic diversity and a gene pool arise via:

- random assortment: of paternal and maternal chromosomes in meiosis.
- crossing over: of segments of individual maternal and paternal homologous chromosomes that results in new combinations of genes on the chromosomes of the haploid gametes produced by meiosis.
- the random fusion of male and female gametes in sexual reproduction.

* Additionally, mutations can cause small changes in genes and they are a source of variation on which natural selection acts. Mutations can increase the size of the gene pool of a population, by increasing the number of different alleles.

Allele frequency - The relative frequency with which a particular allele is found in a population.

(2) Environmental conditions change.

(3) Natural selection removes some individuals with alleles that are not as advantageous

(4) The remaining individuals grow and reproduce, passing on the advantageous alleles.

(5) Adaption / evolution takes place

- Evolution: Evolution is the change in frequency of certain alleles in a gene pool over time due to natural selection.

• Summary:
- Genetic variation in population
- Mutation as a source of genetic variation
- Selection pressure due to changing conditions
- Selection
- Survival of the individuals with advantageous alleles
- Beneficial alleles passed on
- Change in allele frequency over populations

Selection for change

• Directional selection: occurs anywhere that environmental pressure is applied to a population. It is natural selection showing a change from one phenotypic property to a new one more advantageous in the circumstances.

• Diversifying selection: causes an increase in the diversity of the population rather than a trend in one particular direction. It occurs when conditions are very diverse and small subpopulations evolve different phenotypes suited to their very particular surroundings.

• Balancing selection: takes place, when as a result of natural selection, variety is maintained by keeping an allele within the population even though it might seem to be disadvantageous.

Increasing biodiversity

Changes in niches can result in different selection pressures and in change within a species. If that change is great enough, we may consider eventually that a new species has evolved from the old one.

→ **Isolating mechanisms**

For different species to evolve from one original species, different populations of the species usually have to become isolated from each other, so that mating, and therefore gene flow, between them is restricted.

- Geographical isolation
- Ecological isolation
- Seasonal isolation
- Behavioural isolation
- Mechanical isolation

* Allopatric speciation takes place when populations are physically separated in some way.
* Sympatric speciation takes place when two populations are geographically still close together.

Endemism

Endemism describes the situation where a species is found in only one particular area.

→ It should come as no surprise that isolated islands frequently contain endemic species. The availability of niches to species that first colonise the island, the different selection pressures of those niches compared with the home environment and the founder effect of a limited gene pool all combine to result in the evolution of new species that occur only in that small area.

- Founder effect: Process by which any unusual alleles become relatively common in a population if only a small number of individuals set up a population and one of the founder members has that unusual allele.

→ Places where endemism is common often have a rich biodiversity in terms of species numbers but relatively low genetic diversity. This is one reason why areas with many endemic populations are very vulnerable to the introduction of disease.

Species and biodiversity

Biodiversity is the variety of different organisms within a habitat. Two aspects to consider are:

- **Species richness** — the number of different species within an area of known size at a particular time. A species is a group of organisms with so many features in common that they can mate and produce sexually viable offspring.
- **Genetic diversity** — the genetic variation within a species.

It is important to conserve the global biodiversity:

→ All the organisms in an ecosystem are interdependent, and they affect the physical conditions around them. These ecosystems are also interlinked on a larger scale across the Earth. If biodiversity is reduced in one area, the natural balance may be destroyed elsewhere.

- **Keystone species**: A species which has a major effect on its environment. A large number of other species depend on a keystone species for their survival.
- **Biodiversity hotspot**: Area which is particularly rich in different species.

Measuring biodiversity

1 Measuring species richness

To estimate the species diversity, random sampling can be carried out. A number of quadrats of known size are randomly placed in an area. All the species found in each quadrat are recorded and used to estimate the total species richness.

Another method is capture/recapture technique — method of estimating the population of a particular species in an area, which involves capturing individuals, marking them and releasing them, and then capturing some of them again.

2 Measuring genetic diversity

Find the number of different alleles in a gene pool.

- different: Each gene may have one to many different alleles
- alleles in a gene pool: The total number of genes a species has

Conservation and genetic diversity

There are numerous reasons why it is important that we conserve endangered species and two ways to do so are:

- zoos
- seed banks

The roles of zoos in conserving endangered animals

1) Education

Informing all age groups who visit zoos of various conservation issues including;

- the illegal trade in certain animal products
- the need to maintain biodiversity
- captive-breeding programmes including their success stories
- illegal poaching of animals or excessive hunting

2) Scientific research

Zoos, universities can work together on projects that are of benefit to the conservation of animals, including:

- control of diseases that are reducing populations
- behavioural studies to further appreciate the needs of animals in captivity
- development of techniques to further improve breeding success

3) Captive-breeding programmes

These are schemes designed to encourage endangered species to breed so that

- their numbers increase, reducing the risk of extinction
- subsequently some individuals can be released into the wild or protected areas, to maintain or re-establish wild breeding populations.
- the genetic diversity of the species is maintained.

4) Re-introduction programmes

These programmes endeavour to release captive-bred individuals back into the wild so that natural breeding populations can be maintained or re-established.

The importance of maintaining genetic diversity during captive breeding programmes

The problem is that some zoos can only have a small number of individuals, so interbreeding is likely to occur. This leads to:

- reduced genetic diversity and, therefore, a reduced chance of adapting to environmental change.
- increased risk of a genetic condition becoming more common in the breeding population.

* Some of the techniques used to reduce inbreeding and maintain genetic diversity:

→ Do not allow the organisms to repeatedly breed with the same partner, possibly by isolating partners

→ Select partner, possibly by adding a potential partner to a cage, IVF and inter-zoo swapping.

→ Keep a record/database of individuals in captivity and their breeding history, so that choice of partners is controlled.

The role of seed banks in conserving endangered plants

Seeds from a variety of endangered plants can be stored in a dormant state in seed banks. Seeds rather than living plants are stored because:

- less space is required so more species can be held in the available space
- most plants produce large numbers of seeds so collecting small samples is unlikely to damage the wild population.
- easier to store because dormant
- more cost effective.

How a seed bank can be used to conserve a rare plant species

Seeds collected from a number of individual plants

Seeds from several plants are likely to have a higher genetic diversity than seeds from a single plant.

↓

Seeds X-rayed to check for fully formed embryos

This allows only viable seeds (ones that could germinate) to be selected for storage.

↓

Seeds dried to remove water

Reducing water content increases the length of time a seed can be stored and remain viable e.g a 1-2% decrease in water content doubles storage time.

↓

Seeds stored in the cold e.g −20°C

Reducing the temperature also increases the length of time a seed can be stored and remain viable. A 5°C drop in temperature doubles storage time.

↓

Some seeds periodically germinated to check viability

Some seeds are planted to check that they will still germinate and grow.

↓

If less than 75% germinate, those that did germinate are allowed to grow into mature plants that produce their own seeds which are then stored (→ back to: Seeds collected from a number of individual plants)

If 75% or more germinate, the remaining seeds are retained in cold storage and will be checked again for viability