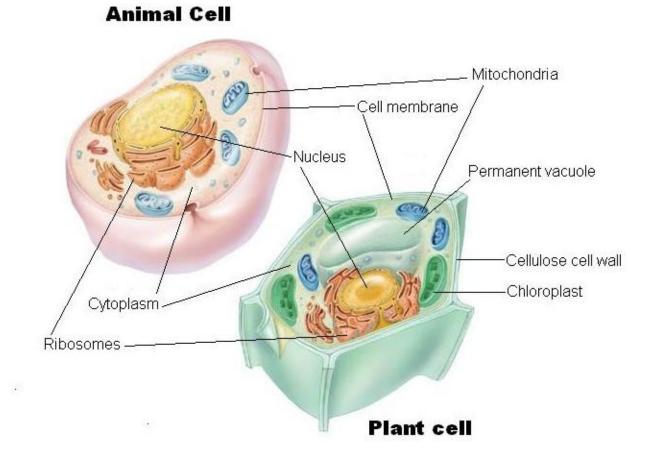


# AQA GCSE Biology Unit 2 Summary Notes

# 12.1 Animal and Plant Cells

# <u>Cells</u>

- Cells are the smallest unit of life.
- All living things are made of cells.
- Most human cells, like most other animal cells, have the following parts:
  - o nucleus
  - o cytoplasm
  - o cell membrane
  - o mitochondria
  - $\circ$  ribosomes
- Plant cells also have:
  - o cell wall
  - o chloroplasts
  - o permanent vacuole

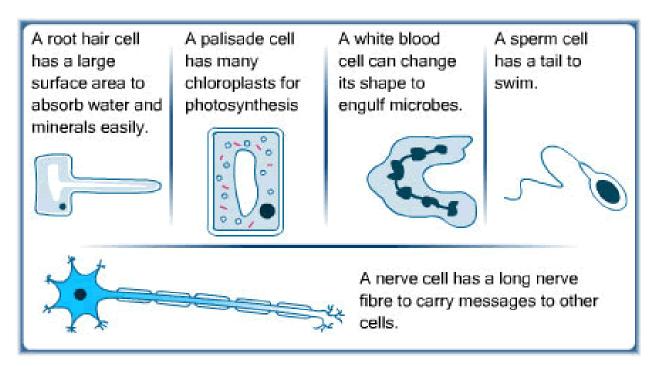


#### What do these structures do?

- Nucleus controls the activities of the cell.
- Cytoplasm where most of the chemical reactions take place.
- Cell membrane controls the passage of substances in and out of the cell.
- Mitochondria where most energy is released in respiration.
- Ribosomes where protein synthesis occurs.
- Cell wall strengthens plant cells.
- Chloroplasts absorb light energy to make food in plant cells.
- Permanent vacuole filled with cell sap in plant cells.

# Other facts:

- The chemical reactions inside cells are controlled by enzymes. Examples:
  - Enzymes for respiration are in the mitochondria.
  - $\circ~$  Enzymes for photosynthesis are in the chloroplasts.
  - $\circ~$  Enzymes for protein synthesis are on the ribosomes.
- Cells may be specialised to carry out a particular function. Examples:



# Tissues, organs and systems

- A tissue is a group of similar cells carrying out a particular function.
- An organ is a group of different tissues carrying out a particular function.
- A system is a group of different organs carrying out a particular function.

# Examples:

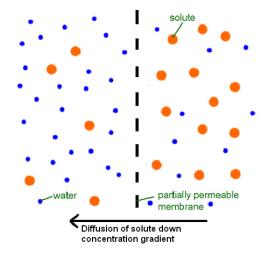
Cell	Tissue	Organ	Organ System
Nerve	Nervous	Spinal cord	Nervous
Ciliated	Epithelium	Trachea	Breathing
Root hair	Epidermal	Lateral root	Root
Palisade	Mesophyll	Leaf	Shoot

# 12.2 Movement into and out of cells

- To get into or out of cells, dissolved substances have to cross the cell membranes.
- Solutes = particles in solution eg glucose, sodium ions, chloride ions.
- Solvent = liquid in which the particles are dissolved eg water.
- Solute and solvent molecules move around randomly.
- Solutes can move into and out of cells by diffusion and osmosis.

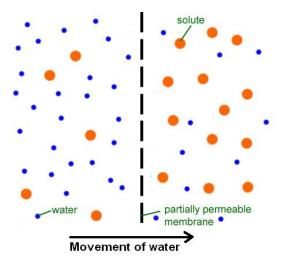
# **Diffusion**

- Diffusion is the spreading of the particles of a gas, or of any substance in solution, resulting in a net movement from a region where they are of a higher concentration.
- Oxygen required for respiration passes through cell membranes by diffusion.
- The greater the difference in concentration, the faster the rate of diffusion.



# <u>Osmosis</u>

- Water often moves across boundaries by osmosis.
- Osmosis is the diffusion of water from a dilute to a more concentrated solution through a partially permeable membrane that allows the passage of water molecules.
- Differences in the concentrations of the solutions inside and outside a cell cause water to move into or out of the cell by osmosis.
- If there is a higher solute concentration on one side of a membrane, water will move in that direction.

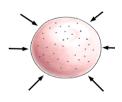


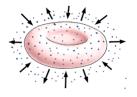
## Osmosis in animal cells:



If animal cells are placed in a solution that has a higher solute concentration than the cytoplasm, then water will leave the cell by osmosis, until it shrinks and dies.

If animal cells are placed in a solution that has a lower solute concentration than the cytoplasm, then water will enter the cell by osmosis until it bursts.

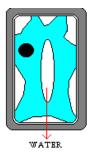




This is why it is vital that we maintain the concentration of our body fluids at an equal solute concentration to our cells' cytoplasm.

#### Osmosis in plant cells:

If plant cells are placed in a solution that has a higher solute concentration than the cytoplasm, then water will leave the cell by osmosis, and the cell membrane separates from the cell wall (plasmolysis). This will cause a plant to wilt.





If plant cells are placed in a solution that has a lower solute concentration than the cytoplasm, then water will enter the cell by osmosis until it is fully turgid, and the cell wall prevents any more water entering. This is important in enabling plants to remain upright.

# 12.3 Photosynthesis

# **Photosynthesis**

- Photo = light
- Synthesis = making of (glucose)
- Photosynthesis = making glucose using light

#### **Complex definition:**

• A process in green plants that produces complex organic chemicals (glucose) from simple inorganic chemicals (carbon dioxide and water) using light energy.

#### More detail:

- Light energy is absorbed by a green substance called chlorophyll.
- Plants look green because chlorophyll reflects green wavelengths of light.
- The plant then uses this energy for photosynthesis.

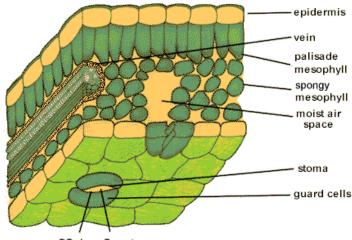
- Light energy is used to convert carbon dioxide and water into sugar (glucose).
- Oxygen is released as a by-product.

#### Word equation:

Carbon Dioxide + Water  $\xrightarrow{\text{chlorophyll}}$  Glucose + Oxygen Light Chemical equation (do not need to learn!):  $6CO_2 + 6H_2O \xrightarrow{} C_6H_{12}O_6 + 6O_2$ 

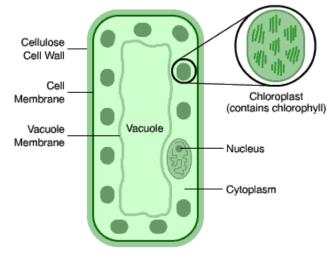
#### Where does photosynthesis happen?

- Leaves are the main site of photosynthesis.
- Photosynthesis mainly in occurs in the mesophyll cells.
- These cells contain lots of chloroplasts.
- Chloroplasts contain chlorophyll.
- Two types of mesophyll cells:
  - Palisade cells absorb most light.
  - Spongy cells absorb the rest.





# A palisade mesophyll cell:



#### Factors that limit the rate of photosynthesis

# 1) Temperature

A low temperature will limit the rate as the molecules will move less and therefore the reaction happens slower

# 2) Carbon dioxide

A shortage of CO<sub>2</sub> will limit the rate as fewer molecules will be available for the reaction.

# 3) Light intensity

A shortage of light means there is less energy to power the reaction.

# Limiting factors explained:

- If one of these factors is closest to its minimum value it will limit the rate.
- Increasing this factor will increase the rate.
- The rate will continue to increase until another factor becomes limiting.
- Any further increase in the original factor will now not increase the rate.
- With no limiting factors, increasing a factor above a certain level will not increase the rate. All chlorophyll molecules are being used.

# Farming practices

- Farmers artificially manipulate the environment in which they grow plants.
- They grow plants in greenhouses or in polythene tunnels.
- They can control the temperature in greenhouses using heaters and ventilation.
- They can artificially increase the carbon dioxide levels.
- They can control the light using fluorescent lamps.
- By doing all of this, their plants grow faster and certain plants can be grown in this country out of their natural growth season. Eg tomatoes can be grown all year round.
- Therefore, they increase their profits.

# How do plants use glucose?

- The glucose produced in photosynthesis may be converted into insoluble starch for storage
- Plant cells use some of the glucose produced during photosynthesis for respiration.

# Why do plants need minerals?

- Plant roots absorb mineral salts including nitrates needed for healthy growth.
- For healthy growth plants need mineral ions including:
  - o nitrate for producing amino acids which are then used to form proteins.
  - magnesium which is needed for chlorophyll production.
- The symptoms shown by plants growing in conditions where mineral ions are deficient include:
  - <sup>o</sup> Stunted growth if nitrate ions are deficient.
  - o yellow leaves if magnesium ions are deficient.

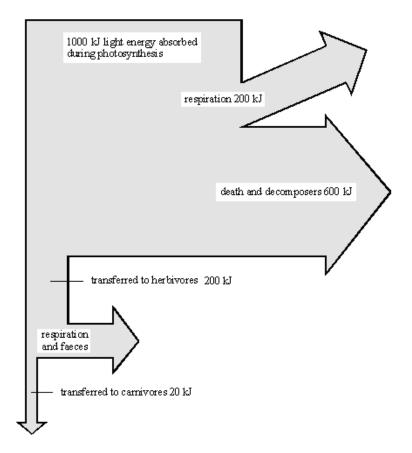
#### 12.4 Energy flow in ecosystems

# Food Chains

- Radiation from the Sun is the source of energy for most communities of living organisms.
- Green plants capture a small part of the solar energy which reaches them.
- This energy is stored in the substances which make up the cells of the plants.
- **Food chain** = a diagram to represent feeding relationships between organisms, showing direction of energy flow.
- **Food web** = a diagram that represents all of the feeding relationships between organisms in an ecosystem.

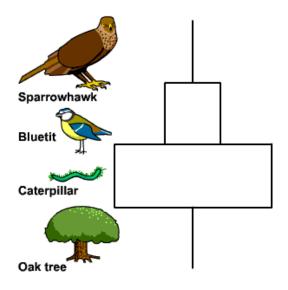
#### Energy transfer in food chains

- At each stage in a food chain, less material and less energy are contained in the biomass of the organisms.
- The amounts of material and energy contained in the biomass of organisms is reduced at each successive stage in a food chain because:
  - Some materials and energy are always lost in the organisms' waste materials.
  - Respiration supplies all the energy needs for living processes, including movement.
  - Much of this energy is eventually lost as heat to the surroundings.
- These losses are especially large in mammals and birds whose bodies must be kept at a constant temperature which is usually higher than that of their surroundings.



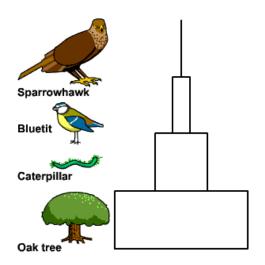
## Pyramids of numbers

- Diagrams that display the numbers of organisms at each trophic level in a food chain
- Each block is drawn to scale.
- These do not necessarily represent amount of biomass:



# Pyramids of biomass

- The biomass at each stage can be drawn to scale and shown as a pyramid of biomass.
- Biomass = mass of living material



- All pyramids of biomass are pyramid shaped.
- The mass of living material (biomass) at each stage in a food chain is less than it was at the previous stage.

#### Improving Energy Transfer in farming

- The efficiency of food production can be improved by reducing the number of stages in food chains.
- Less energy is lost along the food chain between the producer and the human.
- This requires more people to convert to a vegetarian diet.

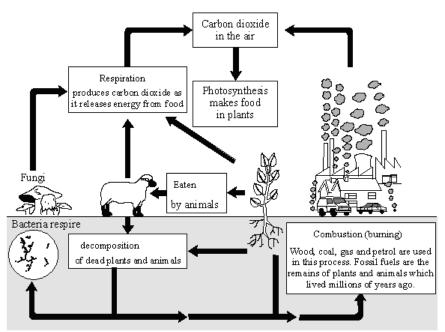
- This may be necessary if people in developing countries are to sustain their population growth.
- The efficiency of food production can also be improved by restricting energy loss from food animals by:
  - Limiting their movement.
  - Controlling the temperature of their surroundings.
- This occurs in battery farming, for which there are many ethical concerns:
  - Animals' movement is limited.
  - They suffer pain and discomfort.
  - They do not live in their natural environment.

# 12.5 Recycling of waste material

#### Recycling in ecosystems

- Living things remove materials from the environment for growth and other processes.
- These materials are returned to the environment either in waste materials or when living things die and decay.
- Materials decay because they are broken down (digested) by microorganisms (decomposers).
- Microorganisms digest materials faster in warm, moist conditions.
- Many microorganisms are also more active when there is plenty of oxygen.
- The decay process releases substances which plants need to grow.
- In a stable community, the processes which remove materials are balanced by processes which return materials.
- The materials are constantly cycled.

# Carbon Cycle



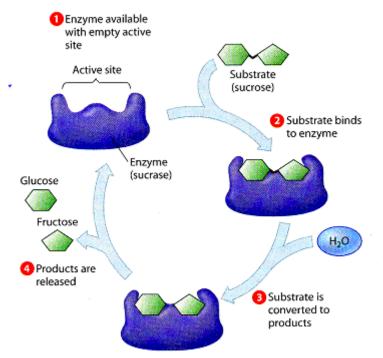
- The carbon cycle is the constant cycling of carbon in ecosystems.
- Carbon dioxide is removed from the environment by green plants for photosynthesis.
- The carbon from the carbon dioxide is used to make carbohydrates, fats and proteins which make up the body of plants.
- Some of the carbon dioxide is returned to the atmosphere when green plants respire.

- When green plants are eaten by animals and these animals are eaten by other animals, some of the carbon becomes part of the fats and proteins which make up their bodies.
- When animals respire some of this carbon becomes carbon dioxide and is released into the atmosphere.
- When plants and animals die, some animals and microorganisms feed on their bodies.
- Carbon is released into the atmosphere as carbon dioxide when these organisms respire.
- By the time the microorganisms and detritus feeders have broken down the waste products and dead bodies of organisms in ecosystems and cycled the materials as plant nutrients, all the energy originally captured by green plants has been transferred.

# 12.6 Enzymes

#### Enzymes structure and function

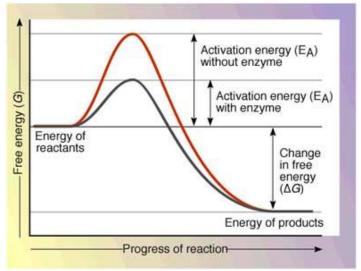
- Enzymes are biological catalysts.
- Catalysts increase the rate of chemical reactions.
- Enzymes are protein molecules made up of long chains of amino acids.
- These long chains are folded to produce a special shape which enables other molecules to fit into the enzyme.
- This shape is vital for the enzymes function.
- Normally only one type of molecule (the substrate) will fit into the enzyme.
- The active site is the part of the enzyme which the substrate fits into.



# **Activation Energy**

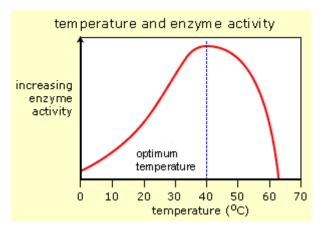
- In order for a chemical reaction to take place, energy is required.
- This is called the activation energy.
- Enzymes reduce the activation energy of a reaction.

AQA GCSE Biology - Unit 2 summary notes



#### Effect of temperature on enzymes

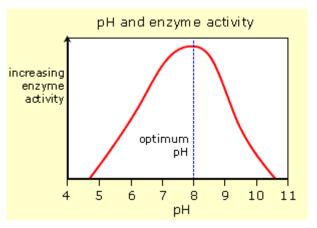
- Like most chemical reactions, the rate of enzyme-controlled reactions increases as the temperature increases.
- The enzyme and substrates move around faster so they collide more often.
- The temperature when the enzyme is working fastest is called the optimum.
- This is true up to approximately 40°C, higher than this and the structure of the enzyme changes.
- As a result, the active site becomes a different shape and the substrate no longer fits.
- It is then described as denatured.



# The effect of pH on enzymes

- pH can also affect the shape of the active site.
- It does this by affecting the forces that hold the enzyme molecule together.
- A change in pH can stop the enzyme completely.
- Different enzymes work best at different pH values.
- Eg. Stomach enzymes work best in acidic conditions.
- Mouth enzymes work best in neutral conditions.

#### AQA GCSE Biology - Unit 2 summary notes



#### Intracellular enzymes

Enzymes inside living cells catalyse processes such as:

- Respiration
- Photosynthesis
- Protein synthesis

# **Respiration**

- Definition: The process of transferring energy from food molecules in every living cell.
- Aerobic respiration uses oxygen
- Anaerobic respiration uses no oxygen

#### Aerobic respiration

• Glucose reacts with oxygen, producing carbon dioxide and water as waste products.

# Word equation:

Glucose + Oxygen Carbon dioxide + Water + Energy

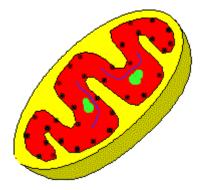
Chemical equation (do not learn!):

 $C_6H_{12}O_6 + 6O_2 \longrightarrow 6CO_2 + 6H_2O$ 

- Respiration actually involves a series of many small reactions.
- Each reaction is controlled by an enzyme.

# <u>Mitochondria</u>

- Most of the reactions in respiration happen in the mitochondria.
- The inner surface of the mitochondria is highly folded to increase the surface area for enzymes.



# Energy use

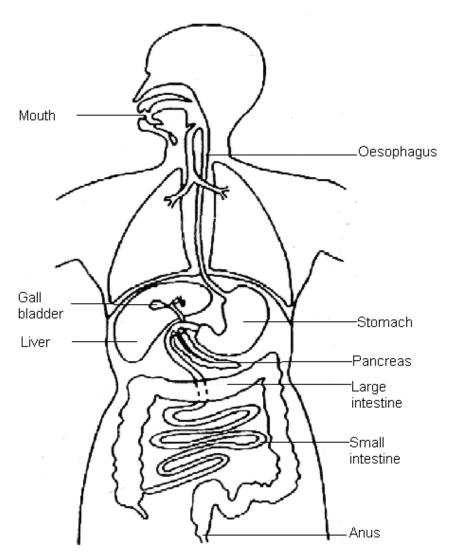
The energy that is released during respiration is used:

- To build up larger molecules using smaller ones.
- In animals, to enable muscles to contract.
- In mammals and birds, to maintain a steady body temperature in colder surroundings.
- In plants, to build up sugars, nitrates and other nutrients into amino acids which are then built up into proteins.

# Digestion

- Some enzymes work outside the body cells.
- These are called extracellular enzymes.
- The digestive enzymes are produced by specialised cells in glands and in the lining of the gut.
- The enzymes then pass out of the cells into the gut where they come into contact with food molecules.
- They catalyse the breakdown of large molecules into smaller molecules.
- Digestion is the process where food is broken down into substances the body can absorb.
- Nutrition is the process of taking in and using food.

#### The Human Digestive System



#### Digestion in the mouth

- Food is chewed to create a larger surface area for the action of enzymes.
- Saliva is released which contains amylase.
- Amylase digests starch into smaller sugars (maltose).
- Further chewing enables swallowing.
- The food enters the oesophagus.

# **Digestion in the stomach**

- Food enters the stomach from the oesophagus.
- The walls of our stomach produce juice.
- This juice contains:
  - A protease enzyme called pepsin.
    - This digests proteins into amino acids.
  - Hydrochloric acid this kills bacteria in our food. It creates pH3.
  - Mucus this protects the wall of our stomach from acid and pepsin.
- The wall of our stomach is muscular, and churns our food.
- The food remains in our stomach for a few hours. The proteins are digested.

• Food leaves our stomach in small squirts into the small intestine.

#### Digestion and absorption in the small intestine

The small intestine has 2 main jobs:

- To complete the digestion of the food
- To absorb the soluble products of digestion into the blood (this is covered in unit 3).

#### **Digestion in the small intestine**

3 juices are released:

#### 1. Bile

- Produced by the liver.
- Stored in the gall bladder.
- Released into the small intestine.
- 2 main things in bile:
  - Alkali to neutralise the stomach acid
  - Bile salts which convert large fat droplets to small fat droplets for a large surface area for the enzymes to act on.

#### THERE ARE NO ENZYMES IN BILE!

#### 2. Pancreatic juice and

#### 3. intestinal juice

- Both are released into the small intestine.
- Both contain 3 main enzymes:
  - Amylase to complete the digestion of starch into sugars.
  - Protease to complete the digestion of proteins into amino acids.
  - o Lipase to break down fats into fatty acids and glycerol.

#### Making use of enzymes

- Some microorganisms produce enzymes which pass out of the cells.
- These enzymes have many uses in the home and in industry.
- In the home, biological detergents may contain protein-digesting and fat-digesting enzymes (proteases and lipases).

Advantages	Disadvantages
These are more efficient at removing stains from clothes.	If the clothes are not fully rinsed, protease enzymes may remain in the clothes, which digests protein in the skin.
Lower washing temperatures can be used which saves energy.	This can lead to irritation, allergies and dermatitis.

- In industry:
  - Proteases are used to pre-digest the protein in some baby foods.
    - This reduces how much the baby needs to digest the food.
  - Carbohydrases are used to convert starch into sugar syrup.
    - This is cheaper than extracting sugar from sugar cane.
  - Isomerase is used to convert glucose syrup into fructose syrup:
    - This is much sweeter

• It therefore can be used in smaller quantities in slimming foods.

Advantages	Disadvantages
Enzymes enable industrial reactions to take	Enzymes are sensitive to temperature and
place at lower temperatures.	pH changes.
Supplying heat is expensive. When	Temperature and pH need to be carefully
enzymes are used, industrial procesess can	monitored and controlled, which can be
be cheaper.	expensive.

## 12.7 Homeostasis

- Homeostasis = Controlling internal conditions.
- Waste products which have to be removed from the body include:
  - Carbon dioxide produced by respiration most of this leaves the body via the lungs when we breathe out
  - Urea produced in the liver by the breakdown of excess amino acids this is removed by the kidneys in the urine, which is temporarily stored in the bladder.
  - Internal conditions which are controlled include:
    - $\circ$   $\;$  The water content of the body
    - The ion content of the body
    - o Temperature
    - Blood sugar levels

#### **Osmoregulation**

- If the water or ion content of the body is wrong, too much water may move into or out of the cells and damage them.
- Water and ions enter the body when we eat and drink.
- Sweating helps to cool the body.
- More water is lost when it is hot, and more water has to be taken as drink or in food to balance this loss.
- The kidneys control the water content of the blood.

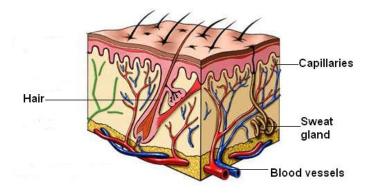
# Maintenance of body temperature – Thermoregulation

- All enzymes work best at 37°C
- The metabolism of cells releases heat which warms the animal's body.
- Metabolism is most efficient If internal temperature is maintained when the external temperature changes,

# Thermoregulatory centre

- Body temperature is monitored and controlled by the thermoregulatory centre in the brain.
- This centre has receptors sensitive to the temperature of blood flowing through the brain.
- Also temperature receptors in the skin send impulses to the centre giving information about skin temperature.

<u>Skin</u>



- Skin contains sweat glands
- Skin has network of capillaries just under the surface

If the core body temperature is too high:

- Blood vessels supplying the skin capillaries dilate (get wider):
  - More blood flows through the capillaries.
  - More heat is lost from the skin.
- Sweat glands release more sweat:
  - This cools the body as it evaporates.

If the core body temperature is too low

- Blood vessels supplying the skin capillaries constrict (get narrower):
  - This reduces the flow of blood through the capillaries.
  - This reduces heat loss from the skin.
- Muscles may shiver.
  - Their contraction needs respiration, which releases some energy as heat.

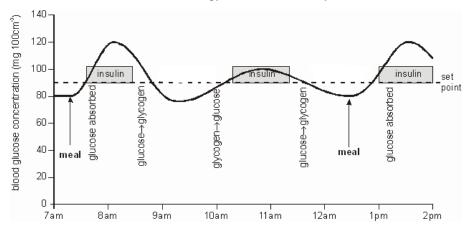
# Regulation of blood glucose

- The blood glucose concentration of the body is monitored and controlled by the pancreas.
- The pancreas produces the hormone insulin, which allows glucose to move from the blood into the cells.
  - 1. Food eaten
  - 2. Carbohydrates digested into glucose
  - 3. Glucose absorbed into blood
  - 4. Blood glucose levels rise
  - 5. Detected by the pancreas
  - 6. Insulin released into blood
  - 7. Insulin reaches the liver
  - 8. Glucose converted in glycogen
  - 9. Glycogen stored

10. Blood glucose levels return to normal

- This process ensures that the blood glucose never rise too high immediately after a meal.
- The glycogen can be broken down to restore blood glucose levels later in between meals.

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# **Diabetes**

- Diabetes is a disease in which a person's pancreas does not produce enough of the hormone insulin.
- After a meal, blood glucose concentration may rise to a very high level.
- This can be fatal if it is not controlled.

# The discovery of insulin

- Banting and Best were scientists in the 1920s who were investigating a cure for diabetes.
- They removed parts of the pancreases of dogs to make them diabetic.
- They extracted insulin from the pancreases of other dogs.
- They injected the insulin into the diabetic dogs.
- This controlled the blood sugar of the diabetic dogs.
- They then injected the insulin into a boy suffering from diabetes.
- His blood sugar levels were controlled.

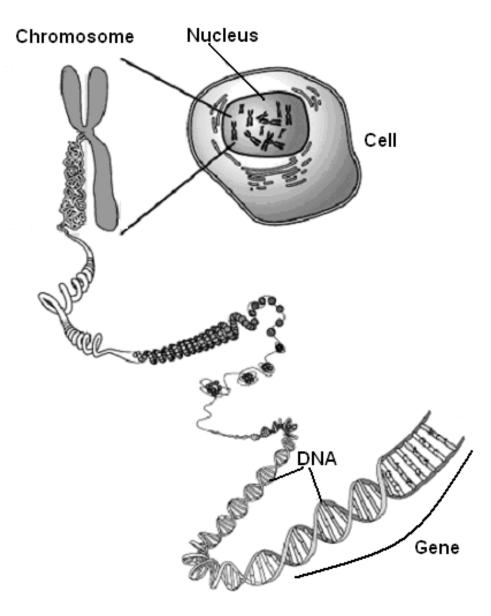
# Treating diabetes

- Diabetes may be treated by careful attention to diet and by injecting insulin into the blood.
- Insulin from pigs and cows was used to treat diabetic people for many years.
- Insulin is now produced using micro organisms that have been genetically engineered to contain the human insulin gene.
  - This modern insulin is chemically identical to human insulin.
- Pancreas transplants can also be used to treat diabetes in some people.
  - However, there are not enough dead donors to supply all the people who need them.
  - Also, organ transplants are not always successful.
  - In the future, stem cells may be used to cure diabetes.
    - However, this requires the use of human embryos, which many people disagree with.

#### 12.8 Inheritance

# **Genetic material**

- In the nucleus of a typical human body cell there are 23 pairs of chromosomes.
- We inherit one set of 23 from each of our parents.
- Chromosomes are made from a large molecule called **DNA** (deoxyribose nucleic acid).
- DNA has 2 main roles:
  - 1. It can replicate prior to cell division (mitosis or meiosis).
  - 2. Its code is used to synthesise proteins.
- A gene is a small section of DNA.
- Each gene codes for a particular combination of amino acids which make a specific **protein**.
- These proteins determine our characteristics.
- Some characteristics are controlled by a single gene.
- Each gene may have different forms called alleles.
- Eg. Eye colour has a gene. Everyone has 2 copies of this gene. Alleles for eye colour may be blue, brown, green etc.
- Therefore each person may have 2 different alleles for eye colour.

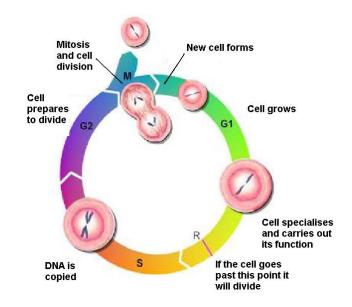


# **Genetic Fingerprinting**

- Each person (apart from identical twins) has unique DNA.
- Samples of DNA can be found in blood, semen and saliva.
- Special techniques are used to cut the DNA and then separate them according to length across a gel.
- Repeating sequences within the DNA can be identified.
- Each individual's DNA produces a specific pattern.
- This can be used:
  - o To identify criminals
  - $\circ$  To decide whether someone is the biological father of a child.

# **Cell Division and Growth**

- New body cells are produced:
  - When the animal is growing.
  - To repair damaged tissues.
  - To replace worn out tissues.
- Mitosis occurs:
  - All of the chromosomes are replicated.
  - The nucleus is divided in 2.
- This results in two genetically identical nuclei.
- The cell divides in 2 to form 2 genetically identical cells.
- Some cells undergo cell division again and again.
- Some cells carry out their function then die.



# Cell differentiation

- Differentiation results when some genes are turned on, some are turned off.
- Once the cells are specialised they carry out their role.

# Asexual reproduction

- The cells of the offspring produced by asexual reproduction are produced by mitosis from the parental cells.
- They contain the same genes as the parents.

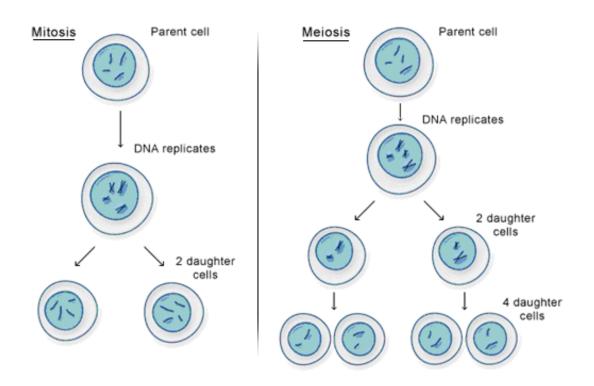
# Stem cells

- Most types of animal cells differentiate at an early stage.
- Many plant cells retain the ability to differentiate throughout life.
- In mature animals, cell division is mainly restricted to repair and replacement.
- Stem cells can be made to differentiate into many different types of cells e.g. nerve cells.
- There are very few stem cells in an adult.
- There is currently a lot of research involving the use of stem cells to treat various diseases and injuries.
- Cells can be taken from human embryos, adult bone marrow and umbilical cords.
- Treatment with these cells may help cure conditions such as paralysis.
- However, many people are concerned about the use of human embryos to treat diseases.
- They feel that all embryos have got the potential to become a baby, and that they should not be used in this way.

# Sexual reproduction

# Gamete formation

- Gametes are sex cells (sperm and egg cells).
- They are formed in testes and ovaries.
- A cell containing a full set of chromosomes (chromosomes in pairs) divides to form cells with half the number set of chromosomes (a single set):
  - $\circ$   $\,$  Meiosis occurs:
    - Copies of the chromosomes are made.
    - The nucleus divides twice to form 4 nuclei.
  - Then the cell divides twice to form four gametes.
    - Each has a single set of chromosomes.



# **Fertilisation**

- When gametes join at fertilisation, a single body cell with new pairs of chromosomes is formed.
- A new individual then develops by this cell repeatedly dividing by mitosis.

# **Variation**

#### Asexual reproduction leads to very little variation:

- Organisms that reproduce asexually, create new individuals by mitosis.
- They are genetically identical to the parent.
- They may develop differently due to differences in their environment.

#### Sexual reproduction leads to much more variation:

- Meiosis ensures that all gametes contain the same genes, but have a different selection of alleles.
- Also, it is random which sperm fertilises which egg.
- Therefore all individuals (except for identical twins) produced sexually are genetically different.

#### Mendel's discoveries

- Mendel worked out the main principles of inheritance in the 19<sup>th</sup> century.
- He studied inheritance in pea plants.
- He noticed that certain characteristics that were shown by 2 pea plants were not always shown in their offspring.
- However, when he crossed these offspring together, the characteristics sometimes reappeared in the next generation.
- He carried out thousands of crosses with pea plants, and he found that many characteristics were inherited in predictable patterns.
  - He proposed the idea of separately inherited factors:
    - Each individual inherits a set of factors from each of their parents.
      - It is the combination of these characters that determines the characteristics of an individual.
- Mendel's discoveries were not recognised until after his death Why?
  - He published his work in an obscure journal
  - DNA, chromosomes and genes were not yet discovered
  - People could not accept the link between plants and humans.

# Inheritance

- When we are conceived, we receive one copy of each gene from both parents.
- Therefore we have two copies of every gene, but they may be 2 different alleles.
- Different combinations of alleles may lead to differences in the characteristic.
- An allele, which controls the development of a characteristic when it is present on only one of the chromosomes, is a **dominant** allele.
- An allele, which controls the development of characteristics only if the dominant allele is not present, is a **recessive** allele.

Eg tongue rolling

- There is a gene that control our ability to roll our tongues.
- There are 2 alleles for this gene:
  - The allele that allows us to roll our tongues is dominant (R)
  - The allele that prevents tongue rolling is recessive (r)

#### Possible parent combination 1:

Parents characteristics:	tongue roller	Х	non-tongue roller
Parents alleles:	RR	Х	rr
Possible alleles in gametes:	R and R	Х	r and r

Possible alleles in gametes: R and R

At fertilisation:

	R	R
r	Rr	Rr
r	Rr	Rr

(This is called a Punnett square)

Offspring: All tongue rollers (that carry the non-tongue rolling allele).

#### Possible parent combination 2:

Parents characteristics:	tongue roller (both carrying th	X e non-tongue	tongue roller rolling allele)
Parents alleles:	Rr	Х	Rr
Possible alleles in gametes:	R and r	Х	R and r

At fertilisation:

	R	r
R	RR	Rr
r	Rr	rr

Offspring:

3 tongue rollers : 1 non-tongue roller

- These are approximate ratios
- There is a 75% **chance** of a tongue roller being born in this family.
- There is a 25% chance of a non-tongue roller being bornin this family.

#### Possible parent combination 3:

Parents characteristics:tongue rollerXnon-tongue roller(carrying the non-tongue rolling allele)RrXrrParents alleles:RrXrrPossible alleles in gametes:R and rXr and rAt fertilisation:XXX

RrrRrrrrRrrrrRrrr

Offspring:

1 tongue roller : 1 non-tongue roller

- Again, these are approximate ratios
- There is a 50% chance of a tongue roller being born in this family.
- There is a 50% chance of a non-tongue roller being born in this family.

#### **Inherited Conditions in Humans**

#### Huntington's disease:

- A disorder of the nervous system.
- It causes nervous tissue to break down when the individual is in their late twenties or thirties.
- It is fatal.
- It is caused by a dominant allele of a gene.
- If someone inherits one copy of this allele, they will develop the disease.
- It can therefore be passed on by only one parent who has the disorder.
- Unfortunately, the parent who passes on the gene may not know they have the disorder if they have children before the disease is expressed.

#### **Cystic fibrosis**

- A disorder of cell membranes.
- It causes thick, sticky mucus to accumulate in the lungs and the digestive system.
- This causes:
  - Lung infections
  - Problems with breathing
  - Problems with digestion and absorption.
- It is caused by a recessive allele of a gene.
- The parents may be carriers of the disorder without actually having the disorder themselves.
- It can therefore be passed on by parents, neither of whom has the disorder.
- To develop the disease, the allele must be inherited from both parents.

# Embryo screening

- People in families that have had certain genetic disorders can have a genetic test to see if they carry the allele for the disease.
- If they do carry the allele, their embryos can be screened to see if it is affected.
- They can then decide whether to have an abortion.
- This is very controversial.
- Also, many people are concerned about this because in the future it may enable people to choose other characteristics in their children.

#### Sex determination

- In human body cells, one of the 23 pairs of chromosomes carries the genes which determine sex.
- In females the sex chromosomes are the same (XX).
- In males the sex chromosomes are different (XY).

Parents sex:	male	Х	female
Parents sex chromosomes:	XY	Х	XX
Possible chromosomes in gametes:	X and Y	Х	X and X

At fertilisation:

	Х	Y
х	XX	XY
х	XX	XY

- There is a 50% possibility of being a girl or a boy.
- As a foetus, we all start off with female characteristics.
- Presence of Y chromosome causes male development.
- Absence of Y chromosome continues female development.
- The sex of the baby is determined by the sex chromosome inherited from the father.