## <u>A2 Biology – Revision Notes</u> Unit 4 – Energy, Control And Continuity

## **Biochemistry**

- 1. ATP (adenosine triphosphate) is required for endothermic processes, but can be re-synthesised when coupled to exothermic processes ATP  $\rightleftharpoons$  ADP +  $P_i$ .
- 2. ATP is synthesised across the inner membranes of the mitochondria and chloroplasts, hence they are adapted to give the maximum surface area. ATP-ase enzymes are powered by a proton gradient (set up by an electron transfer chain) that provides the energy for ATP synthesis.
- 3. NADH (NADPH in photosynthesis) and FADH<sub>2</sub> are reduced coenzymes that are used to carry electrons to a different part of the organelle.
- 4. The absorption spectrum for chlorophyll shows which wavelengths of light are absorbed (mostly blue and red, hence green light is reflected). The action spectrum shows what the rate of photosynthesis is at different wavelengths (i.e. how well the light is used).
- 5. Photosynthesis occurs in two stages:
  - a. Light Dependant Reactions (on the thylakoid membrane):
    - i. A photon hitting a chlorophyll molecule in photosystem II (PSII) excites an electron, triggering the photolysis of H<sub>2</sub>O into oxygen, protons and electrons.
    - ii. The released electrons pass through a series of electron carriers (plastoquinone, b<sub>6</sub>-f (a proton pump), then plastocyanin) before reaching PS I.
    - iii. At PS I, the electrons are excited again by incident photons. They then pass through the ferredoxin electron carrier.
    - iv. The electrons can either travel back to plastoquinone, powering the proton pump (cyclic photophosphorylation), or reach the NADP reductase enzyme (non-cyclic photophosphorylation), whereby NADP<sup>+</sup> ions are reduced to NADPH.
    - v. The proton gradient (from b<sub>6</sub>-f) powers the production of ATP from ATP-ase enzymes in the thylakoid membrane.
  - b. Light Independent Reactions (in the stroma) the Calvin-Benson cycle:
    - i. The enzyme rubisco (ribulose biphosphate carboxylase) catalyses the fixing of a CO<sub>2</sub> molecule to the 5-carbon ribulose biphosphate.
    - ii. This decays into two 3-carbon molecules of phosphoglycerate.
    - iii. ATP from the LDR is used to form diphosphoglycerate.
    - iv. NADPH is used to reduce this to glyceraldehyde phosphate (GALP).
    - v. One molecule of GALP is removed per 3 molecules of  ${\rm CO_2}$ , and the rest go on to be modified into ribulose phosphate then ribulose biphosphate.
- 6. Aerobic respiration occurs in four stages:
  - a. Glycolysis (in the cytoplasm):
    - i. The glucose reacts with 2 ATP molecules (a primer).
    - This then breaks down into two molecules of pyruvic acid (3-carbon), producing 4 ATP molecules.
    - iii. Overall 2 ATP is produced by glycolysis.
  - b. The link reaction (in the matrix) Pyruvic acid reacts with coenzyme A to form acetylcoenzyme A (effectively 2-carbon) and release CO<sub>2</sub>.
  - c. The Krebs Cycle (in the matrix):
    - i. Oxaloacetic acid (4C) reacts with acetylcoenzyme A to form citric acid (6C).
    - ii. Citric acid is oxidised to  $\alpha$ -ketoglutaric acid (5C), releasing  $CO_2$ .
    - iii. α-Ketoglutaric acid is oxidised to succinic acid (4C), releasing CO<sub>2</sub>.
    - iv. Succinic acid is modified to malic acid, then Oxaloacetic acid.
    - v. The NADH and FADH<sub>2</sub> formed move to the electron transport chain.
  - d. The electron transport chain (on the cristae):
    - i. A series of cytochromes on the inner mitochondrial membrane accept the electrons from NADH and FADH<sub>2</sub>, and they pass along the chain.
    - ii. Each of the cytochromes acts as a proton pump, using the energy from the electrons to actively transport  $H^{\scriptscriptstyle +}$  ions across the membrane.
    - iii. The electrons reduce O<sub>2</sub> into H<sub>2</sub>O at the end of the chain.
    - iv. The protons move back across the membrane through ATP-ase enzymes, hence synthesising ATP.

## **Physiology**

1. The body is controlled by two major systems:

- a. The nervous system electrical stimuli, specific action, controls most movement (voluntary / involuntary), fast responses.
- b. The endocrine system hormonal control, more general effects, longer-term responses.
- c. The hypothalamus links the nervous and endocrine systems together.
- 2. A reflex arc is the initiation of an instant (conditioned) reaction, not involving the higher centres of the brain. There are three neurons involved:
  - a. Sensory neurone links the receptor with the spinal cord.
  - b. Relay neurone connects the incoming and outgoing neurones.
  - c. Motor neurone transmits impulses to the effector.
- 3. A postural reflex is one that maintains position and body control without us having to constantly think about fine adjustments for example the knee jerk reflex (this is initiated by stretch receptors in the patellar tendon).
- 4. Hormones are peptides, proteins or lipids produced by endocrine glands, which fit into specific receptor molecules on target cells, to trigger a change in their intracellular activity.
- 5. Homeostasis is the maintenance of a constant internal body environment. There are two nervous systems involved in homeostasis (of the autonomic division):
  - a. Sympathetic nervous system generally stimulates.
  - b. Parasympathetic nervous system generally inhibits.
- 6. Negative feedback is the principle that if a factor changes, the homeostatic mechanism acts to reverse that change and restore the factor to a resting level i.e. it keeps levels constant.
- 7. Positive feedback is the principle that if a factor changes, the mechanism will act to increase the change in that factor and bring it further away from a resting level i.e. it brings about change.
- 8. There are a number of receptors to different stimuli in the dermis:
  - a. Thermoreceptors there are separate hot and cold thermoreceptors.
  - b. Pacinian corpuscles receptors for deep pressure.
  - c. Meissner's corpuscles touch receptors (on hairless skin only).
  - d. Free nerve endings touch and pain receptors (all over the body).
- 9. There are two types of organism with respect to thermoregulation:
  - a. Endotherms produce and maintain their own body temperature (humans = 36.9°C).
  - b. Ectotherms rely on external environment for body temperature.
- 10. Thermoregulation is maintained by the hypothalamus central receptors detect the blood temperature, and peripheral thermoreceptors on the skin detect that of the external environment.
- 11. When the temperature is too high:
  - a. Sweating takes place water has a high latent heat of vaporisation, therefore removes a lot of heat energy through evaporation.
  - b. Vasodilation of capillaries takes place, and arteriovenous shunt vessels shut down blood moves to the surface so heat can be lost through radiation.
  - c. Erector pili muscles relax hairs goes down so heat isn't trapped.
  - d. The body becomes inactive muscles aren't used as often, to avoid heat generation.
- 12. When the temperature is too low:
  - a. Sweating stops, and shivering takes place muscles vibrate to generate heat.
  - b. Vasoconstriction of surface capillaries takes place, and shunt vessels dilate, to direct blood away from the skin surface.
  - c. Erector pili muscles contract hairs are raised to trap air and conserve heat.
  - d. Blood migrates away from the skin to the liver, where it is stored as a blood reservoir.
- 13. The thyroid gland controls the rate of metabolism, thus when the temperature drops, its secretions are raised to increase the metabolic rate. Iodine is essential for correct function.
- 14. The parathyroid glands are four small glands embedded in the thyroid that regulate the level of calcium ions in the blood.
- 15. The blood glucose level is controlled by the islets of Langerhans in the pancreas:
  - a. Hyperglycaemia (too much blood glucose):
    - i. The  $\beta$ -cells in the pancreas secrete insulin into the bloodstream.
    - ii. Insulin promotes cellular respiration, hence more glucose is used.
    - iii. The uptake of glucose by the liver and muscles is promoted, and glycogenesis takes place (production of glycogen).
    - iv. The conversion of glucose into fat is accelerated in the adipose tissue.
  - b. Hypoglycaemia (too little blood glucose):
    - i. The  $\alpha$ -cells in the pancreas secrete glucagon into the bloodstream.
    - ii. Glucagon stimulates glycogenolysis (the conversion of glycogen into glucose) and the conversion of amino acids to glucose in the liver.

- iii. In times of starvation, gluconeogenesis (production of glucose from lipid or protein sources) takes place.
- 16. There are three major types of diabetes:
  - a. Diabetes mellitus type I (insulin-dependant) there is a deficiency of insulin, due to damage to the  $\beta$ -cells.
  - b. Diabetes mellitus type II (non insulin-dependant) the cells in the body become unable to respond to insulin.
  - c. Diabetes insipidus the pituitary gland is unable to secrete ADH, hence copious dilute urine is produced.
- 17. There are three possible fates of excess amino acids:
  - a. Deamination the amino group of the amino acid is removed, and converted into ammonia (NH<sub>3</sub>). This reacts with CO<sub>2</sub>, in the ornithine cycle, to form urea, which reenters the blood to be removed by the kidneys  $2NH_3 + CO_2 \ge CO(NH_2)_2 + H_2O$ .
  - b. Transamination the amino acid is converted into a different amino acid (e.g. an essential amino acid being converted into a non-essential one).
  - c. Used in the production of plasma proteins.
- 18. Nitrogen is excreted as different compounds, depending upon the animal:
  - a. Ammonia aquatic animals, as the excess water will dilute the toxic NH<sub>3</sub>.
  - b. Uric acid birds, as the semi-solid waste helps to conserve water.
  - c. Urea terrestrial animals, to give hypertonic urine.
- 19. A renal dialysis machine works on the principle of separation by molecular size hence the urea and other waste products are filtered out of the blood, whilst useful substances remain.
- 20. Ultrafiltration is the filtration blood under pressure, to produce a filtrate identical to tissue fluid:
  - a. The afferent arteriole, entering the glomerulus, is wider than the efferent arteriole leaving it. This, along with the arterioles splitting into many smaller arterioles, creates a high hydrostatic pressure.
  - b. Blood is forced into the Bowman's capsule through a three-layer filter, consisting of the capillary endothelium, the basement membrane (a fine feature), and the pedicels (projections on the podocytes).
- 21. Selective reabsorption is the process of reabsorbing the useful substances, such as glucose, amino acids, water soluble vitamins and minerals, and most of the water and salts, from the glomerular filtrate back into the blood-stream:
  - a. The microvilli in the walls of the proximal and distal convoluted tubules provide a large surface area for the reabsorption of substances.
  - b. Water passes from the filtrate into the blood (lower water potential due to the plasma proteins) by osmosis, and many of the dissolved substances diffuse back into the blood.
  - Active transport is used to keep urea in the filtrate, and to aid the reabsorption of other substances.
- 22. The loop of Henle uses a countercurrent multiplier system to reabsorb water:
  - a. Sodium and chloride ions are actively pumped out of the ascending limb into the interstitial fluid. The ascending limb is impermeable to water.
  - b. Water is drawn out of the descending limb by osmosis, into the more concentrated interstitial fluid.
  - c. At the bottom of the loop, the filtrate is very concentrated, due to the water loss, so the active transport of sodium and chloride ions in the ascending limb makes it dilute again.
  - d. As the fluid passes down the collecting duct, more water is reabsorbed into the increasingly concentrated medulla.
  - e. Urine passes down the ureters to the bladder, and empties through the urethra.
- 23. Osmoreceptors in the hypothalamus monitor the osmotic blood concentration, and baroreceptors all over the circulatory system monitor blood pressure:
  - a. When blood pressure is too low, and blood concentration is too high, the hypothalamus sends impulses to the pituitary gland to release ADH (anti-diuretic hormone).
  - b. ADH increases the water permeability of the distal convoluted tubule and collecting duct.
  - c. More water is reabsorbed from the tubules back into the blood, so more concentrated urine is produced, blood pressure is raised, and blood concentration decreases.
  - d. Alcohol inhibits the production of ADH, so large quantities of dilute urine are produced.
- 24. The light entering the eye is focused upon the fovea centralis on the retina. The process of adapting the eye to distance is accommodation:
  - a. Most of the refraction is done by the cornea, which has the same refractive index as the aqueous humour.

- b. The lens achieves fine focusing with the ciliary muscles (circular):
  - i. For a distant object, the ciliary muscles relax and the suspensory ligaments tighten, to make the lens flat and thin.
  - ii. For a nearby object, the ciliary muscles contract and the suspensory ligaments slacken, to make the lens more spherical.
- 25. The iris controls the pupil size, hence the amount of light entering the eye:
  - a. In dim light, the radial muscles contract and the circular relax, to dilate the pupil.
  - b. In bright light, the circular muscles contract and the radial relax, to constrict the pupil.
- 26. The retina contains two types of photoreceptors, called rods and cones:
  - a. Rods respond to dim light, and are responsible for peripheral vision. There are a number of rods connected to a single bipolar cell, hence a low visual acuity. The visual pigment contained in rods is rhodopsin.
  - b. Cones respond to bright light, and are responsible for central and colour vision. There is only one bipolar cell per cone, hence a high visual acuity. Cones are mainly located around the centre of the retina (fovea), rather than the peripheral. The visual pigment contained in cones is iodopsin.
- 27. Rod cells function as follows:
  - a. In the dark, gated Na<sup>+</sup> channels are kept open by cyclic GMP. This allows the constant production of glutamate neurotransmitters, which inhibit the bipolar cell.
  - b. In the light, rhodopsin splits into opsin and retinal, due to the photosensitive retinal changing from a cis to a trans isomer.
  - c. Opsin is an enzyme that starts a series of reactions to reduce cGMP levels. This closes the ion channels, so no neurotransmitter is released and the bipolar cell is activated.
  - d. When it becomes dark again, it takes slightly longer for the rhodopsin to reform, hence the time needed for dark adaptation.
- 28. Visual acuity is the ability to see detail i.e. to distinguish between distinct points that are close together. This depends upon the degree of retinal convergence i.e. how many photoreceptors share the same bipolar cell.
- 29. The trichromatic theory of colour vision states that there are three types of cone (red, green and blue), and that each detects a different wavelength of light. Thus it is the wavelength of the light reflected from an object that determines its colour, and correspondingly the relative stimulation of the three cone types on the retina.
- 30. Problems with the trichromatic theory:
  - a. Colour constancy colours appear the same, regardless of the surrounding light conditions the visual cortex tends to keep a colour.
  - b. A colour changes, depending upon the colours that are surrounding it.
  - c. After staring at a strongly coloured image, staring away results in the formation of a 'phantom' image in the complementary colours to the original.
- 31. The left visual cortex receives visual information from the left side of both eyes, and the right visual cortex that from the right side of both eyes. Roughly 20% of all the neurones from both eyes go to the midbrain. The optic nerves cross at the optic chiasma.
- 32. There are three types of neurone:
  - a. Sensory neurones carry information from sense organs (transducers) to the CNS.
  - b. Motor neurones carry information from the CNS to muscles and glands.
  - c. Intermediate neurones connect sensory and motor neurones.
- 33. The spinal nerves branch off to the arms at the brachial plexus, and to the legs at the sciatic plexus.
- 34. There are two types of motor neurone:
  - a. Myelinated neurones a sheath of myelin is formed from Schwann cells around the axon. The junctions of these sheaths are the nodes of Ranvier. This speeds up the conduction (action potentials jump from node to node by salutatory conduction), and insulates to prevent cross conducting.
  - b. Non-myelinated neurones there is no myelin sheath; hence the nerve transmission is much slower.
- 35. In nervous tissue, white matter consists mostly of myelinated nerve fibres (axons), whereas grey matter is mainly cell bodies.
- 36. Glial cells are packed between neurones to form neuroglia tissue:
  - a. Provides mechanical support and electrical insulation.
  - b. Schwann cells are specialised glial cells, forming myelin sheaths.
  - c. Control the nutrient and ionic balance, and break down neurotransmitters.
- 37. Neurones contain nissl granules (mitochondria, free ribosomes and rough ER):

- a. These generate enzymes involved in impulse transmission and synthesis of trophic factors.
- b. They regulate growth and differentiation of nervous tissue.
- 38. A resting potential is the charge across the axon membrane (about -70mV) when it is not transmitting an impulse:
  - a. A sodium-potassium pump actively transports three  $Na^+$  ions out of the axon, for every two  $K^+$  ions that it transports in.
  - b. K<sup>+</sup> ions diffuse out more rapidly than the Na<sup>+</sup> ions diffuse in, so there are more positive ions outside the membrane than inside, hence the resting potential.
- 39. An action potential is a brief reversal of the resting potential, when an impulse is transmitted:
  - a. At a synapse, the neurotransmitter opens ligand-gated Na<sup>+</sup> and then K<sup>+</sup> channels in the postsynaptic membrane.
  - b. As the Na<sup>+</sup> channels open first, the Na<sup>+</sup> ions rapidly diffuse out of the axon, resulting in depolarisation of the membrane.
  - c. The  $K^+$  channels then open, allowing the  $K^+$  ions to rapidly diffuse back into the axon, hence repolarisation of the membrane takes place.
  - d. The Na<sup>+</sup> channels close, and cannot be opened, for a period of about 1ms called the absolute refractory period hence an impulse cannot be transmitted by the neurone during this time. The relative refractory period takes place for about 2ms after this, whereby a stimulus must be stronger than usual in order to be transmitted.
  - e. The current resulting from the flow of ions causes adjacent voltage-gated Na<sup>+</sup> and K<sup>+</sup> channels to open, thus the action potential moves along the axon.
- 40. The all-or-none rule states that a stimulus must have a minimum intensity to initiate an action potential if the intensity is above the threshold level, then the impulse is fully transmitted; if not then there is no impulse at all.
- 41. In order for a large enough impulse to be created, summation of impulses can take place:
  - a. Temporal summation many small impulses summate over time at the same synapse.
  - b. Spatial summation many impulses at different synapses on the same cell allow an impulse to be transmitted, e.g. many rods are needed for one bipolar cell to fire.
- 42. Accommodation is the temporary response to ignore a stimulus, if it is a permanent feature of the current environment. Adaptation is a more permanent response, whereby an increasingly strong stimulus is required for the same response (e.g. the effects of nicotine).
- 43. Synapses are gaps between neurones to allow chemical control of impulses:
  - a. An action potential arrives at the end of a pre-synaptic neurone.
  - b. Ca<sup>2+</sup> ion channels open, and the calcium ions diffuse into the cell.
  - c. This causes the synaptic vesicles (containing neurotransmitter) to move towards the presynaptic membrane, where exocytosis takes place, to release the neurotransmitter.
  - d. The neurotransmitter diffuses across the synaptic cleft, to protein receptor molecules on the post-synaptic membrane, which triggers the action potential in the membrane. This creates an excitatory post-synaptic potential (EPSP).
  - e. An inhibitory post-synaptic potential (IPSP) can be set up by some neurotransmitters, whereby the impulse is inhibited. Negative (Cl<sup>-</sup>) ion channels open to make the membrane increasingly negative, hence reducing the chance of an action potential being set up this allows for the creation of neural pathways.
- 44. A number of different neurotransmitters are used throughout the body:
  - a. Acetylcholine (cholinergic synapses) most motor neurones (somatic/voluntary nervous system), neuromuscular junctions, parasympathetic synapses.
  - b. Noradrenaline (adrenergic synapses) sympathetic synapses.
  - c. Serotonin and dopamine used in the brain.
- 45. When the neurotransmitter reaches the post-synaptic neurone, it is broken down by enzymes, and diffuses back across the synapse to the pre-synaptic neurone. Acetylcholine is broken down into acetic acid and choline by the enzyme acetylcholinesterase.
- 46. Drugs can have various effects on synapses:
  - a. Hallucinogens (e.g. LSD) mimic actions of other neurotransmitters.
  - b. Nicotine similar to acetylcholine, so the body becomes addicted to it.
  - c. Curare and atropine block acetylcholine.
  - d. Muscarine mimics acetylcholine.
- 47. The neuromuscular junction (motor endplate) initiates muscle contraction:
  - a. The action potential arrives at the motor endplate, and acetylcholine is released. This crosses the synapse to the sarcolemma, where it initiates an action potential.

- b. The action potential moves along the sarcolemma and into a T-tubule.
- As a result of the action potential, the sarcoplasmic reticulum becomes permeable to Ca<sup>2+</sup> ions, which rapidly diffuse out to the myofibrils.
- d. This initiates the ratchet mechanism of muscle contraction.
- e. Temporal summation of impulses is used to provide a stronger contraction of the muscle.
- 48. There are a number of levels of organisation in the central nervous system:
  - Spinal cord involved in reflex arcs, whereby the higher levels are informed but not involved.
  - b. Hindbrain:
    - i. Medulla oblongata basis of the autonomic nervous system, involved in homeostasis, control of non-skeletal muscles (cardiac and smooth (peristalsis)).
    - ii. Cerebellum controls body movement and maintains balance.
    - iii. Reticular activating system (RAS) filters and coordinates incoming stimuli, controls consciousness, arousal from sleep, motivation and sustained concentration.
  - c. Midbrain:
    - i. Pons Pneumotaxis centre, involved in homeostasis.
    - ii. Optic chiasma crossing of the optic nerves, important in vision.
  - d. Forebrain:
    - i. Thalamus directs sensory information to the correct part of the cerebral cortex.
    - ii. Hypothalamus site of thirst, hunger, sex drive. Important in homeostasis (thermoregulation, osmoregulation), links with the pituitary gland by the infundibular stalk.
    - iii. Cerebrum cerebral hemispheres are the site of all higher mental activity, linked by the corpus callosum. The cerebral cortex (surface of the cerebrum) is the site of activity. The right cerebral hemisphere controls and responds to the left side of the body, and vice-versa.
- 49. The cerebral cortex is divided into different functional areas:
  - a. Sensory areas receive information from the sense organs.
  - b. Motor areas control voluntary muscles in the body.
  - c. Association areas interpret sensory information in the light of previous experience (memory). The speech association area allows language to be recognised from sound.
- 50. The visual cortex contains different levels of cells:
  - a. Simple cells respond to small groups of rods and cones, to give colour recognition.
  - b. Complex cells receive information from several simple cells to allow object recognition.
  - c. Hypercomplex cells receive information from several complex cells, to recognise changes in lines and perceive movement.
- 51. The brain and the spinal cord are protected by:
  - a. Bone the skull and vertebral column respectively.
  - b. The spinal and cranial meninges three layers of tough membrane.
  - c. Cerebrospinal fluid cushions and bathes the brain, filling the ventricles, and cushions the spinal cord, acting as a shock absorber in both cases.
- 52. The sympathetic and parasympathetic divisions of the ANS have opposing functions:
  - a. Iris parasympathetic constricts pupil, sympathetic dilates.
  - b. Ciliary muscle parasympathetic accommodates near vision.
  - c. Lacrimal gland parasympathetic secretes tears.
  - d. Urinary bladder wall parasympathetic contracts, sympathetic relaxes.
- 53. Skeletal muscles occur in antagonistic pairs, held together by connective tissue, with a tendon at each end, attached to the bones. Bones are connected by ligaments (containing elastin). Striated muscle fibres have many nuclei in the sarcoplasm, which lie near to the surface of the fibre.
- 54. Collagen is a fibrous protein contained in tendons and bones, preventing them from stretching or breaking by making then less brittle and slightly bendy.
- 55. Arthropods have exoskeletons as their cuticle, so the muscles are attached to the inner surface of this. Exoskeletons have a disadvantage in that they are very heavy in larger organisms, and they must be shed in order for the organism to grow.
- 56. Skeletal muscle consists of muscle fibres, each containing many myofibrils:
  - a. These consist of thick filaments of myosin, each surrounded by six thin actin filaments in a spiral fashion.

- b. The myofibril is striped containing light zones (actin only) and dark zones (containing myosin). The H-zone contains myosin only, hence is slightly lighter than the rest of the dark zone that contains both actin and myosin.
- c. When the myofibril contracts, the actin slides over the myosin, thus the light bands get shorter, the H-zone gets smaller, and the dark zone overall stays the same length.
- 57. The sliding filament hypothesis of muscle contraction (ratchet mechanism):
  - a. When the muscle is at rest, the actin cannot bond to the myosin, as tropomyosin molecules are between the binding sites.
  - b. When the neuromuscular junction activates an impulse, Ca<sup>2+</sup> ions are released by the sarcoplasmic reticulum.
  - c. The calcium ions bind to, and change the shape of troponin molecules.
  - d. Troponin displaces tropomyosin, so the myosin heads can bind to the actin.
  - e. The myosin head pulls backwards, pulling the actin over the myosin.
  - f. An ATP molecule fixes to the myosin head, causing it to detach.
  - g. The ATP provides the energy to return the myosin head to its original position.
  - n. The myosin head re-attaches to another actin binding site, further along the chain.

## Genetics

- 1. Basic definitions in genetics:
  - a. Genotype the genetic constitution of an organism, i.e. the combination of alleles.
  - b. Phenotype the observable features of an organism, which are a combination of its genes and environmental factors.
  - c. Gene a length of DNA coding for a specific polypeptide (characteristic).
  - d. Chromosome one long DNA molecule, with genes along its length. Each chromosome is a linkage group.
  - e. Locus the position of a gene on a chromosome. The distance between two genes determines the frequency of cross-overs (recombinations) between them.
  - f. Allele an alternative form of a gene, occupying the same locus on the homologous chromosomes. Alleles may be dominant, recessive, or codominant.
  - g. Homozygous both alleles are the same (i.e. both dominant or both recessive).
  - h. Heterozygous the pair of alleles are different (i.e. one dominant and one recessive).
- 2. Meiosis takes place in two meiotic divisions:
  - a. First division homologous chromosomes each form pairs of chromatids, and crossingover of alleles takes place between them:
    - i. Interphase before meiosis, DNA replicates to 4 copies of each chromosome.
    - ii. Prophase I chromosomes become visible, centromeres move to opposite poles of cell. Each homologous pair comes together to form a bivalent. Crossing over occurs between the bivalent forms at chiasmata.
    - iii. Metaphase I bivalents arrange themselves on the equator of the spindle.
    - iv. Anaphase I chromatid pairs split apart to opposite poles of the cell.
    - v. Telophase I cytokinesis begins, to form two new cells.
  - b. Second division the chromatids are pulled apart to produce four haploid nuclei, each with a copy of each homologous chromosome:
    - i. Interphase a resting time between the two divisions.
    - ii. Prophase II a new spindle forms, perpendicular to the first.
    - iii. Metaphase II chromosomes line up on the equator of the spindle.
    - iv. Anaphase II chromatids are pulled apart to opposite poles of the cell.
    - v. Telophase II cytokinesis begins, to produce four haploid cells.
- 3. Meiosis can be advantageous, due to the sources of variation in offspring:
  - a. Reassortment (independent assortment) in each homologous pair, there is a paternal chromosome and a maternal one. During anaphase I, these are separated, and hence can be reshuffled into any combination.
  - b. Random fertilisation any female gamete can fertilise any male gamete, hence there is a large variation in the possible zygotes, due to the variation across the gametes.
  - c. Recombination (crossing over) during prophase I, the bivalents cross over at chiasmata, thus parts of the chromatids are crossed from one to the other. The combination of paternal and maternal alleles on the chromatid is therefore altered.
- 4. In humans there are 23 pairs of chromosomes 22 of the pairs are autosomes, and there is one pair of sex chromosomes, which can be XX or XY this is determined by the sperm, as all the ova will have the X chromosome.

- 5. Mendel's laws of genetics:
  - a. Law of segregation during meiosis, the two members of any pair of alleles possessed by an individual, segregate into separate gametes and thus into different offspring. In other words, alleles will not alter one another, but retain their integrities during replication.
  - b. Law of independent assortment during meiosis, all combinations of alleles are distributed to daughter nuclei with equal probability – so for the genotype AaBb, then AB, Ab, aB and ab are all formed in equal meiotic frequency.
- 6. Monohybrid inheritance refers to the inheritance of a single gene with two alleles:
  - a. For two alleles, A and a, there are three possible genotypes (in a 1:2:1 ratio):
    - i. Homozygous dominant (AA) dominant allele is expressed.
    - ii. Homozygous recessive (aa) recessive allele is expressed.
    - iii. Heterozygous (Aa) dominant allele is expressed.
  - b. There will be two phenotypes, in a 3:1 ratio.
- 7. Codominant alleles are ones that are both expressed in the phenotype, for example blood groups:
  - a. There are three alleles  $-I^A$  and  $I^B$  are dominant, and  $I^O$  is recessive.
  - b.  $I^A$  and  $I^B$  code for A and B proteins on red blood cells, whereas  $I^O$  codes for no relevant
    - i.  $I^{O}I^{O}$  blood group O.

    - ii. I<sup>A</sup>I<sup>A</sup> or I<sup>A</sup>I<sup>O</sup> blood group A.
      iii. I<sup>B</sup>I<sup>B</sup> or I<sup>B</sup>I<sup>O</sup> blood group B.
    - iv.  $I^A I^B blood group \ \widetilde{AB}$  (codominance).
- 8. Dihybrid crosses involve two separate genes, each with two alleles, at the same time. The results are organised in a Punnett square, as there will be 16 genotypes.
  - a. With parents of AABB and aabb, F1 will all be AaBb.
  - b. F2 will then give a 9:3:3:1 ratio of phenotypes, assuming the functions of the genes (biochemically) are independent.
- 9. Epistasis is where the presence of one allele affects the expression of another, due to a biochemical interaction - if two enzymes are produced by genes A and B respectively, then if A is not expressed, then the pathway cannot proceed regardless of whether B can be produced or not:
  - a. In wild mice, two genes are needed for the agouti (grey/brown) colour:
    - i. If A and B are present, the mice will be agouti.
    - ii. For AAbb or Aabb, the mice will be black (due to lack of enzyme B).
    - iii. For aa, regardless of whether or not there is enzyme B, the mice will be albino, as enzyme A is not present, and B depends on A.
  - b. This will result, in this case, in a 9:3:4 ratio of phenotypes.
- 10. Sex-linked inheritance occurs when the gene occurs on the sex chromosomes. Males cannot be carriers, if the faulty allele is on the X chromosome, as they will only have one copy of the gene. Men tend to be much more affected, as women must have both faulty alleles, whereas men only need to have one (women are often carriers though):
  - a. Red/green colour blindness recessive on X chromosome.
  - b. Pattern baldness dominant on Y chromosome.
  - c. Haemophilia recessive on X chromosome.
- 11. There are two types of genetic variation:
  - a. Discontinuous variation tends to be coded for by a single gene, e.g. the human ABO blood group system. There are only certain, specific, categories, with nothing in-between.
  - Continuous variation usually polygenic (controlled by many genes), for example height, mass, intelligence. The population forms a normal distribution.
- 12. Genetic variation can be caused by:
  - Variation during meiosis (see above).
  - b. Mutations an error occurs during DNA replication or transcription, altering the DNA sequence. A chromosome mutation is when this happens to large sections of DNA or genes, during cell division.
  - Environmental factors the expression of genes can be affected by diet, disease, or temperature for example. Exposure to mutagens will increase the chances of a mutation.
- 13. The Hardy-Weinberg allows gene frequencies to be predicted:
  - a. The total frequency of phenotype expression is given by p and q, for the dominant and recessive alleles respectively -p+q=1. b. Squaring gives  $p^2+2pq+q^2=1$ , whereby  $p^2$  is the frequency of AA, 2pq is the frequency
  - of Aa, and  $q^2$  is the frequency of aa.
  - This is based on the assumptions that:

- i. There is a large population.
- ii. Breeding is completely random.
- iii. There is no natural selection.
- iv. There are no mutations of the alleles.
- 14. It is very difficult to remove an allele from a large population e.g. for a 10% frequency, if the population is 10, then only 1 must die, but if the population is 5000, then 500 must die.
- 15. Natural selection is the process in which those organisms whose genes give them a selective advantage (positive selection pressure) are more likely to survive, reproduce and pass their genes on to the next generation. A new feature is introduced into a species by a random mutation, then the selection pressure determines whether or not it propagates through the population evolution does not take place due to environmental factors, but environmental factors determine the relevance of a particular mutation.
- 16. Artificial selection is when breeding is controlled for specific characteristics. This results in the formation of new breeds, but not new species (e.g. *Canis familiaris*).
- 17. A stabilising selection pressure occurs when there is no pressure to change i.e. staying the same will be the most advantageous for the species.
- 18. A balancing selection pressure occurs when a particular genotype can be both advantageous and disadvantageous e.g. sickle cell anaemia, whereby the aa genotype results in death, but the Aa phenotype does not, and results in malarial resistance. Hence the disease is widespread in Africa.
- 19. A species is defined as a population or group of similar individuals that can reproduce to produce fertile offspring.
- 20. The evolution of a new species (speciation) takes place by:
  - a. Isolation part of the population becomes geographically isolated, so cannot breed with the rest of the population.
  - b. Natural selection the two environments will be different, with different selection pressures, so different phenotypes and alleles are favoured.
  - c. Speciation over generations, genetic differences accumulate, so the two populations cannot interbreed if brought back together.
- 21. The large-scale movement of populations (immigration and emigration) is important in evolution.
- 22. Evidence for evolution comes from:
  - a. Fossil records.
  - b. Common blood pigments (e.g. haemoglobin in most chordates, haemocyanin in arthropods and molluscs).
  - c. Similar larval forms:
    - i. Annelids (segmented worms) and molluscs (shelled animals).
    - ii. Echinoderms (5-sided animals) and chordates (vertebrates).
  - d. Similar embryological development in reptiles, fish and mammals.
- 23. The pentadactyl limb is a good example of evolution:
  - a. 1 upper limb, 2 lower limbs and 5 digits.
  - b. Adaptive radiation of this form gave such structures as wings, hooves etc.
- 24. Similarities between organisms can be:
  - a. Analogous functional similarity (different in origin and structure), e.g. bird wing and arthropod wing.
  - b. Homologous similarity in position, origin and structure e.g. the pentadactyl limb.
- 25. Taxonomy is the classification of organisms using the Linnaeus system (based on Latin). The classification is phylogenetic (based on evolutionary history), and organisms are classified according to similarities in morphology, anatomy, biochemistry, behaviour, genetics and DNA.
- 26. There are seven layers of classification:
  - a. Kingdom, phylum, class, order, family, genus and species.
  - b. Every organism has two names (written in italics or underlined) a generic name (its genus, with a capital letter) and a specific name (its species, in lowercase).
- 27. There are five kingdoms of living organism (viruses are unclassified):
  - Kingdom Animalia multicellular eukaryotes, heterotrophic nutrition, radial (simple) or bilateral (complex) symmetry.
  - b. Kingdom Plantae multicellular eukaryotes, cellulose cell walls, photoautotrophs.
  - c. Kingdom Fungi eukaryotes, chitin cell walls, heterotrophic nutrition (parasitic or saprophytic), reproduce by spore production.
  - d. Kingdom Protoctista eukaryotes with characteristics excluding them from any of the other kingdoms (e.g. amoeba, algae and slime moulds).
  - e. Kingdom Prokaryotae no nucleus, no membrane-bound organelles, circular DNA.