# Chapter Nine



| 3.5.1 Stimuli, both internal and external, are detected and lead to a response. | |
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| **Survival and response** | Organisms increase their chance of survival by responding to changes in their environment.<br>Tropisms as responses to directional stimuli that can maintain the roots and shoots of flowering plants in a favourable environment.<br>Taxes and kineses as simple responses that can maintain a mobile organism in a favourable environment.<br>A simple reflex arc involving three neurones. The importance of simple reflexes in avoiding damage to the body. |
| **Control of heart rate** | The role of chemoreceptors and pressure receptors, the autonomic nervous system and effectors in controlling heart rate. |
| **Receptors** | The basic structure of a Pacinian corpuscle as an example of a receptor. The creation of a generator potential on stimulation.<br>The Pacinian corpuscle should be used as an example to illustrate the following.<br>• Receptors only respond to specific stimuli<br>• Stimulation of receptor membranes produces deformation of stretch-mediated sodium channels, leading to the establishment of a generator potential.<br>Differences in sensitivity and visual acuity as explained by differences in the distribution of rods and cones and the connections they make in the optic nerve. |

# Neurones

- **Neurones →** specialised nerve cells adapted to rapidly carrying electrochemical changes called nerve impulses.



- **Structure →**
- **Cell body –** contains nucleus and many RER – production of protons and neurotransmitters.
- **Dendrons –** extensions of cell body – subdivide into dendrites  carry nerve impulses towards cell body.
- **Axon –** single long fibre – carry nerve impulses away from cell body.
- **Schwann cells –** surround axon, protect it and provide electrical insulation – carry out phagocytosis and nerve regeneration.
- **Myelin sheath –** covers axon – made up of schwann cell membranes – myelinated neurones transmit nerve impulses faster than unmyelinated neurones.
- **Nodes of ranvier –** gaps between adjacent schwann cells where there is no myelin sheath.

# Regulation of Blood Glucose



- **Role of the pancreas →**
- Situated behind the stomach.
- Produces enzymes for digestion and hormones for regulating blood glucose.
- Made up of mainly enzyme-producing cells.
- There are also hormone-producing cells called the **Islets of Langerhans.**

- **There are two types of Islets of Langerhans →**
- **α-cells →** larger and produce glucagon.
- **β-cells →** smaller and produce insulin.

- **Regulation of blood glucose →**
- Glucose is the main substrate for respiration – mammals must contain a relatively constant level.
- If levels rise too high, it lowers the water potential of blood and creates osmotic problems that can cause dehydration.

- **Normal level →** 90mg per $100cm^3$
- **Blood glucose comes from three sources →**
- **Diet –** breakdown of carbohydrates.
- **Glycogenolysis –** break down of glycogen – stored in liver and muscle cells.
- **Gluconeogenesis –** production of new glucose from sources other than carbohydrates e.g. glycerol and amino acids.

# Chapter Thirteen



| 3.5.5 | Negative feedback helps maintain an original internal state in the context of a dynamic equilibrium. Positive feedback also occurs. |
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| **Principles** | Negative feedback restores systems to their original level.<br>The possession of separate mechanisms involving negative feedback controls departures in different directions from the original state, giving a greater degree of control.<br>Positive feedback results in greater departures from the original levels.<br>Positive feedback is often associated with a breakdown of control systems, e.g. in temperature control.<br>**Candidates should be able to** interpret diagrammatic representations of negative and positive feedback. |
| **Control of mammalian oestrus** | The mammalian oestrous cycle is controlled by FSH, LH, progesterone and oestrogen.<br>The secretion of FSH, LH, progesterone and oestrogen is controlled by interacting negative and positive feedback loops.<br>**Candidates should be able to** interpret graphs showing the blood concentrations of FSH, LH, progesterone and oestrogen during a given oestrous cycle. Changes in the ovary and uterus lining are **not** required. |

# Chapter Fourteen



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| **Gene mutation** | Gene mutations might arise during DNA replication. The deletion and substitution of bases.<br>Gene mutations occur spontaneously. The mutation rate is increased by mutagenic agents. Some mutations result in a different amino acid sequence in the encoded polypeptide. Due to the degenerate nature of the genetic code, not all mutations result in a change to the amino acid sequence of the encoded polypeptide.<br>The rate of cell division is controlled by proto-oncogenes that stimulate cell division and tumour suppressor genes that slow cell division. A mutated proto-oncogene, called an oncogene, stimulates cells to divide too quickly. A mutated tumour suppressor gene is inactivated, allowing the rate of cell division to increase. |

# Structure of Ribonucleic Acid



- **mRNA →**
- Long strand in single helix.
- Manufactured when DNA forms a pair or copy of part of one of its strands.
- Leaves nucleus through nuclear pores and enter cytoplasm.
- Acts as a template upon which proteins are built.
- Easily broken down – exists only while needed to manufacture a given protein.

- **Transfer RNA (tRNA) →**
- Relatively small molecule – around 80 nucleotides.
- Single stranded chain folded into a clover-leaf shape – one end of chain extends beyond the other – amino acids can easily attach.
- At opposite end, sequence of three other bases known as the anticodon – for each amino acid there is a different sequence of organic bases on the anticodon.
- During protein synthesis, the anticodon pairs with three complementary bases that make up the codon on mRNA.
- Lines up amino acids on mRNA template during protein synthesis.

# Chapter Fifteen



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| **Regulation of transcription and translation** | Transcription of target genes is stimulated only when specific transcriptional factors move from the cytoplasm into the nucleus.<br>The effect of oestrogen on gene transcription.<br><br>Small interfering RNA (siRNA) as a short, double-strand of RNA that interferes with the expression of a specific gene.<br><br>**Candidates should be able to**<br>• interpret data provided from investigations into gene expression<br>• interpret information relating to the use of oncogenes and tumour suppressor genes in the prevention, treatment and cure of cancer<br>• evaluate the effect on diagnosis and treatment of disorders caused by hereditary mutations and those caused by acquired mutations. |

# Totipotency & Cell Specialism



- Although all cells contain all genes, only certain genes are expressed in any one cell at any one time.
- Some genes are permanently expressed, others are switched on and off when needed.
- Differentiated cells differ from each other because they produce different proteins.

- **Totipotent cells** → cells that can mature into any body cell.

- **Ways in which genes are prevented from expressing themselves →**
- Preventing transcription and mRNA production.
- Breaking down mRNA before its genetic code is translated.

- Most cells, once matured, can no longer develop into other cells – they lose their totipotency.
- Few totipotent cells exist in mature animals – these are adult stem cells.
- Stem cells are undifferentiated dividing cells in adult animal tissues – need to be constantly replaced.
- Stem cells are found in the intestine lining, skin and bone marrow.
- Embryonic stem cells are found at the earliest stage of development of an embryo.

- Mature plants have many totipotent cells.
- So cells from most plant species can be used to clone new plants.

# Chapter Sixteen



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| | Many human diseases result from mutated genes or from genes that are useful in one context but not in another, e.g. sickle cell anaemia.<br><br>DNA sequencing and the PCR are used to produce DNA probes that can be used to screen patients for clinically important genes. The use of this information in genetic counselling, e.g. for parents who are both carriers of defective genes and, in the case of oncogenes, in deciding the best course of treatment for cancers.<br><br>**Candidates should** understand the principles of these methods. They should be aware that methods are continuously updated and automated. |
| **Genetic fingerprinting** | An organism's genome contains many repetitive, non-coding base sequences. The probability of two individuals having the same repetitive sequences is very low.<br><br>The technique of genetic fingerprinting in analysing DNA fragments, that have been cloned by PCR, and its use in determining genetic relationships and in determining the genetic variability within a population.<br><br>**Candidates should be able to**<br><br>• explain the biological principles that underpin genetic fingerprinting techniques<br>• interpret data showing the results of gel electrophoresis to separate DNA fragments<br>• explain why scientists might use genetic fingerprints in the fields of forensic science, medical diagnosis, animal and plant breeding. |

# In Vivo Gene Cloning
# The Use of Vectors



- **Antibiotic-resistance markers →**
- The gene that was cut out was responsible for resistance to a second antibiotic – bacteria with the required gene won't be resistant
- **Process →**
- Bacteria cells are cultured by spreading them thinly on a nutrient agar plate.
- Each will grow into a genetically identical colony.
- Tiny samples are transferred onto a replica plate with the second antibiotic.
- Colonies killed must have taken up the required gene.
- Colonies are in the exact same place as on the original plate so they can be easily identified.

- **Fluorescent markers →**
- A more rapid method is the transference of a gene from a jellyfish into the plasmid.
- This gene produces a green fluorescent protein (GFP).
- The gene to be cloned is transplanted into the centre of the GFP gene.
- Any bacterial cell that is to be cloned will not be able to produce GFP.
- Those that fluoresce do not have the gene to be cloned.

- **Enzyme markers →**
- Another gene marker is the gene that produces the enzyme lactase.
- Lactase will turn a particular colourless substrate blue.
- The required gene is transplanted into the gene that makes lactase.
- If a plasmid with the required gene is present in the bacterial cell, it will not produce lactase and change the colour of the colourless substrate.

# Locating & Sequencing Genes



- **Gel electrophoresis →**
- DNA fragments are placed on to an agar gel and a voltage is applied.
- The larger the fragment, the more slowly they move.
- Smaller fragments therefore move further.
- The fragments of different lengths are separated.
- The fragment distance is compared against a known reference called a DNA ladder.
- The fragment with one nucleotide will only contain a terminator which can be easily identified.
- This can be done for the fragment with two nucleotides, then three and so on.
- The fragment can therefore be sequenced.

- Only fragments up to around 500 bases can be sequenced.
- Larger genes must be cut into smaller fragments by restriction endonucleases.

- **Restriction mapping →**
- Cutting DNA with a series of different restriction endonucleases.
- The fragments are then separated by gel electrophoresis.
- The distance between the recognition sites can be determined by the patterns of fragments that are produced.

# Screening for Clinically Important Genes



- Screening can determine the probabilities of a couple having offspring with a genetic disorder.
- Potential parents who are at risk can obtain advice from a genetic counsellor.

- Cancers may develop as a result of mutations that prevent the tumour-suppressor genes inhibiting cell division.
- Mutations of both alleles must be present to inactivate the gene.
- If a mutated gene is detected by genetic screening, individuals who are at a greater risk of cancer can make informed decisions about their lifestyle.

- Genetic counsellors help individuals to understand the results and implications of the screening to make appropriate decisions.
- Genetic counsellors research family histories of inherited diseases to advise parents on the likelihood of it arising in their children.