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A2 Biology OCR

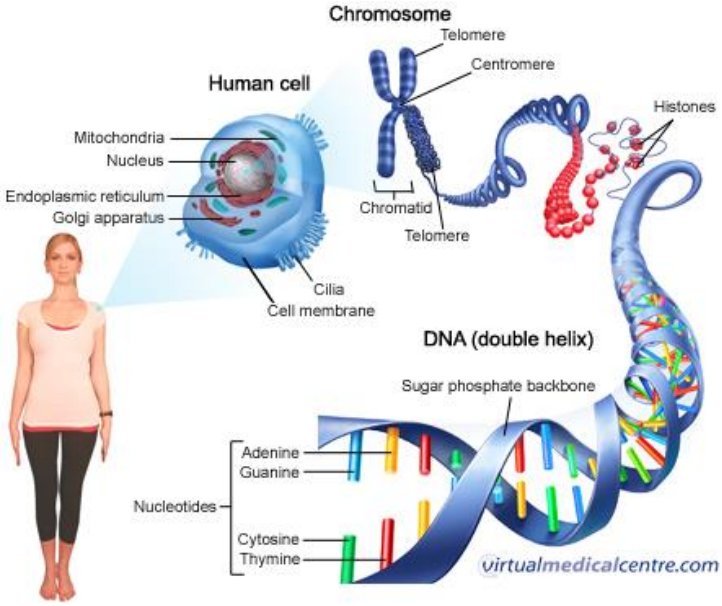
Unit F215: Control, genomes and environment

Module 1.1 Cellular control

Notes & Questions

State that genes code for polypeptides, including enzymes.

- Genes are lengths of DNA which code for 1 or more polypeptides.
- The entire library of genes in an organism is referred to as its Genome
- The Human Genome contains approximately 25,000 genes
- A Polypeptide is a polymer made from amino acids monomers.
- Polypeptides can include;
  - Antibodies
  - Enzymes
  - Hormones
  - Structural proteins such as keratin (Hair) or elastin (skin)
  - Receptor molecules involved in cell signalling



Explain the meaning of the term *genetic code*.

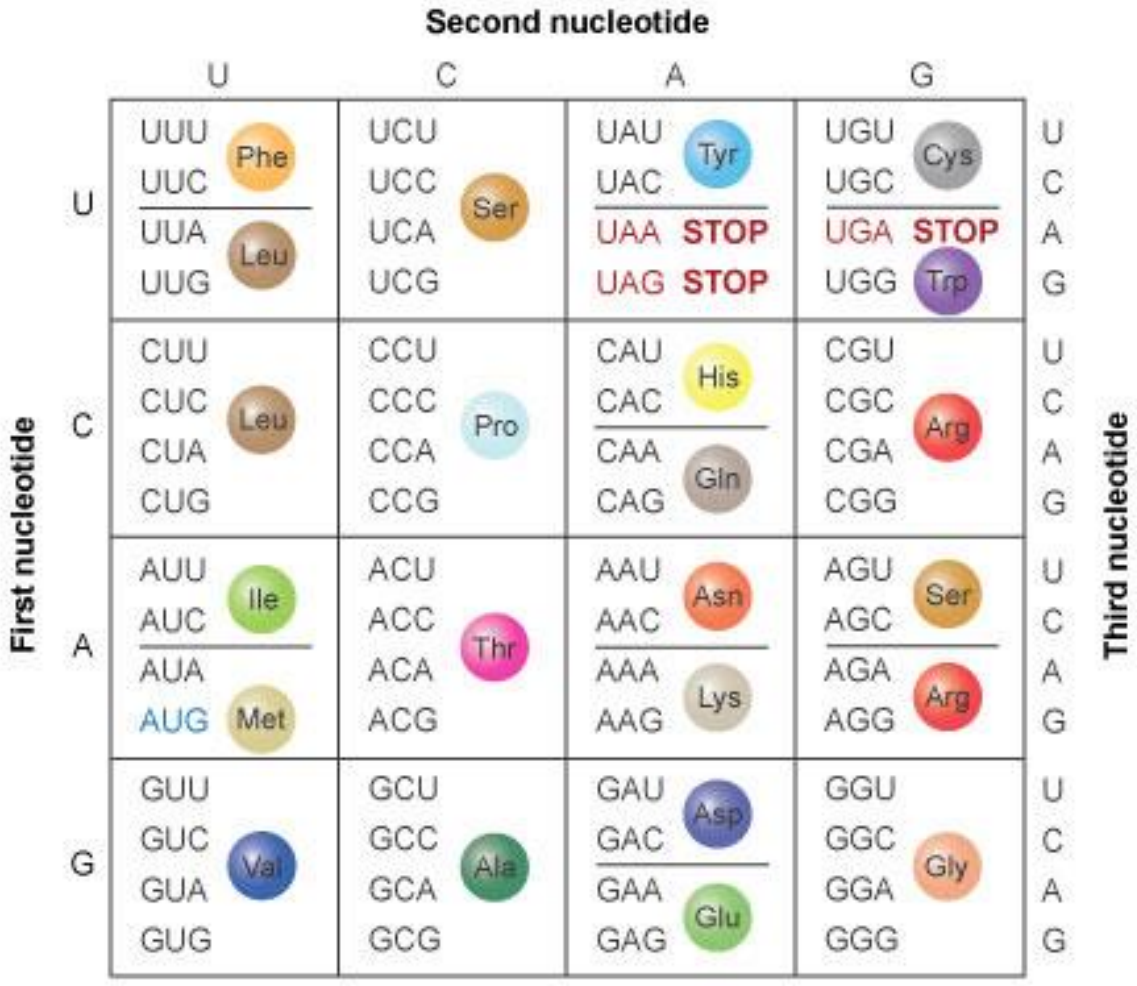
- The genetic code is the sequence of Nitrogenous bases (DNA) held in the gene.
- The code is held in the DNA double stranded Double Helix
- There are 4 different Nitrogenous bases;

Purines	**Pyrimidines**
Adenine	Thymine
Guanine	Cytosine

- There are 20 different amino acids.
- How can this genetic code containing 4 different nitrogenous bases code for all 20 different amino acids?
  - 1 Nitrogenous base code = 1 amino acid, then the 4 bases could only code for 4 amino acids ($4^1 = 4$ codes)
    i.e. A, T, C & G
  - 2 Nitrogenous base code = 1 amino acid, then the 4 bases could only code for 16 amino acids ($4^2 = 4 \times 4 = 16$ codes)
    i.e. AA, AT, AC, AG, TA, TT, TC, TG, CA, CT, CC, CG, GA, GT, GC & GG.
  - 3 Nitrogenous base code = 1 amino acid, then the 4 bases could code for 64 possibilities. That is enough codes for all 20 amino acids.
    i.e. AAA, AAT, AAC, AAG, ATA, ATT, ATC……etc

The genetic code is therefore made up of 3 base code words.

- The 3 base genetic code is said to be degenerative as there are more than one code for each amino acid
  - i.e. 64 possible codes only code for the 20 amino acids including a start code and 3 stop codes
- This is very useful because;
  - More useful amino acids have multiple codes
  - This allows some mutations at a base level to have no impact on the structure of the polypeptide formed
- The Code is transferrable in most organisms. E.g. TCT = Serine in almost all living things. Evolution?



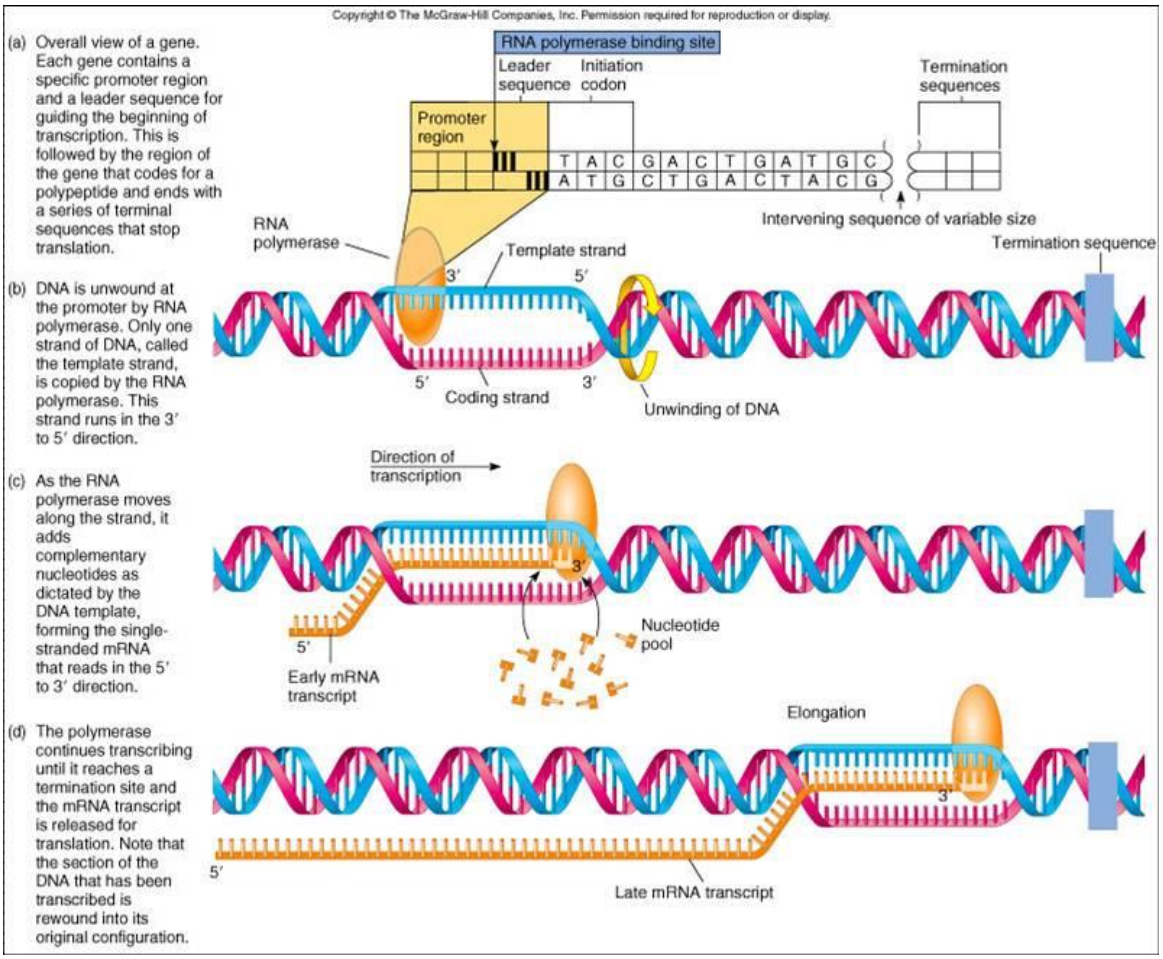
Describe, with the aid of diagrams, the way in which a nucleotide sequence codes for the amino acid sequence in a polypeptide.

- Transcription (DNA code is copied onto messenger RNA)

- Not all the DNA in the nucleus is involved – only the DNA of the gene to be transcribed is involved.

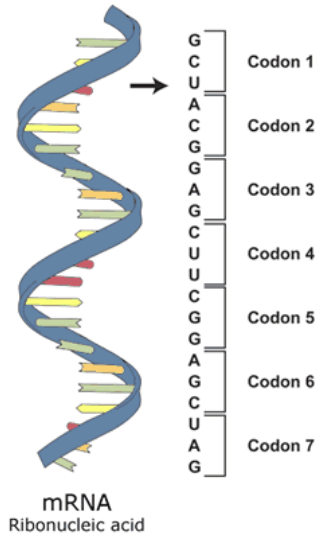
1. The gene for the desired polypeptide, dips into the nucleolus.
2. The DNA unwinds
3. The DNA unzips,
4. By Hydrogen bonds between the complementary bases of the DNA break.
5. Gene is exposed
6. RNA nucleotides align with complementary DNA nucleotides
7. U with A, A with T, G with C & C with G.
8. RNA polymerase now binds to the template strand of the DNA for the gene
9. RNA Polymerase catalyses the formation of a messenger RNA (mRNA) copy of the genetic code by building the sugar-phosphate backbone.
10. mRNA copy of the gene is formed
11. Hydrogen bonds between the mRNA and template DNA break releasing the mRNA.
12. mRNA leaves the nucleus via the nuclear pore.

- The messenger RNA is synthesised from free activated RNA nucleotides which are found in the nucleolus. They are activated through the addition of 2 additional phosphate groups.
  i.e. ATP (for AMP), GTP (for GMP), UTP (for UMP) and CTP (for CMP).

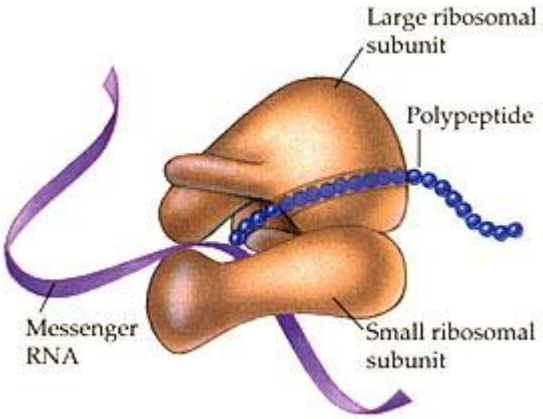


Describe, with the aid of diagrams, how the sequence of nucleotides within a gene is used to construct a polypeptide, including the roles of messenger RNA, transfer RNA and ribosomes.

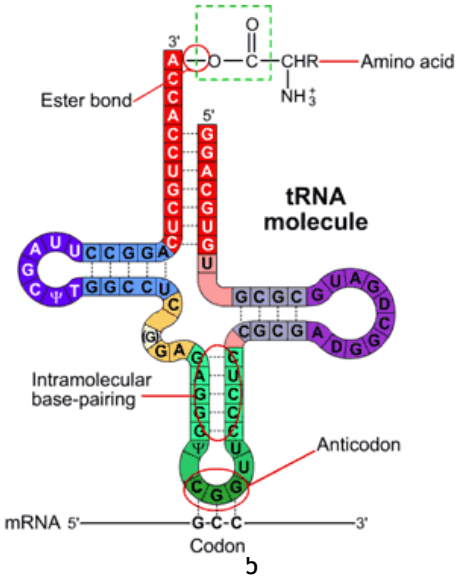
- There are 3 types of RNA are used; Messenger, Ribosomal & Transfer.
    - Messenger RNA (mRNA)
      - Single stranded
      - Carries the genetic code in 3 base codes known as codons



    - Ribosomal RNA (rRNA)
      - 2 subunits (1 large and 1 smaller subunits)
      - Binds two mRNA codons at a time and accommodates tRNA.
      - Can be found free in the cell cytoplasm, or attached to endoplasmic reticulum to form Rough ER.

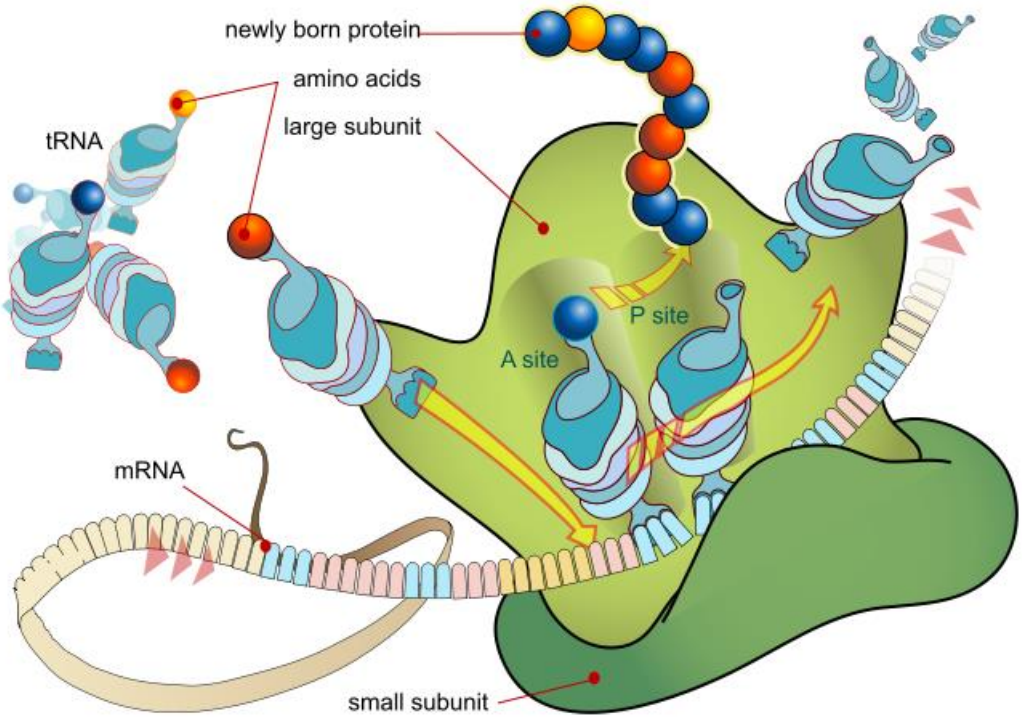


    - Transfer RNA (tRNA)
      - Made of 75 RNA nucleotides
      - 2 Binding sites, 1 called the anti-codon which is complementary to the codon on the mRNA and a second which binds the corresponding amino acid.



- Translation (mRNA code read and sequences amino acids to form a polypeptide.

1. mRNA attaches to the smaller ribosomal subunit
2. 2 mRNA codons are attached at a time
3. tRNA brings an amino acid to the ribosome/mRNA
4. Each tRNA has a specific amino acid attached
5. A tRNA with a complementary anti-codon to the mRNA codon binds.
6. This is by complementary base pairing A=U and G=C
7. The first mRNA codon is always AUG (known as the start codon) and codes for the amino acid Methionine.
8. A second tRNA with a complementary anti-codon to the second mRNA codon now attaches.
9. A peptide bond forms between the amino acids
10. The first tRNA now leaves the ribosome to collect another Methionine amino acid.
11. The mRNA nucleic acid now moves along another 3 base codon.
12. This process is known as Indexing.
13. The third codon on the mRNA is now exposed in the ribosome and so the process continues.
14. There are 3 stop codons (UAG, UAC & UAA) These have complementary anti-codons on tRNA but the tRNA molecules do not have an associated amino acid so the change ends and the polypeptide is free.



- The DNA sequence determines the mRNA sequence which determines the amino acid sequence and so the primary structure of the polypeptide.

- In Prokaryotes translation occurs immediately as transcription is occurring. This is because prokaryotes have no nuclear envelope – DNA is loose.

State that cyclic AMP activates proteins by altering their three-dimensional structure.

- Once the polypeptides have been synthesised they may not be immediately active.
- Cyclic AMP can activate polypeptides by altering their 3D structure.
- Cyclic AMP if you remember is synthesised from ATP using the enzyme adenyl cyclise.
- Cyclic AMP is referred to as the second messenger and is heavily involved with cell signalling with protein/polypeptide hormones such as insulin and adrenaline,

Explain genetic control of protein production in a prokaryote using the *lac* operon.

- The Lac Operon is an example of an inducible protein. i.e. can be turned on or off.
- E-Coli can respire both glucose and Lactose (Glucose + Galactose).
- Usually their environment is glucose rich so they do not want to make the enzymes necessary for lactose respiration.
    - Why?
        - Making proteins that are not essential at that time will tie up resources such as amino acids
        - Uses a lot of energy unnecessarily

- The lactose respiration specific enzymes are;
    - Beta-Galactosidase
        - Catalyses the hydrolysis of lactose into glucose and Galactose.
    - Lactose Permease
        - Increases the cell's permeability to lactose so it can take in more lactose from the surrounding environment.

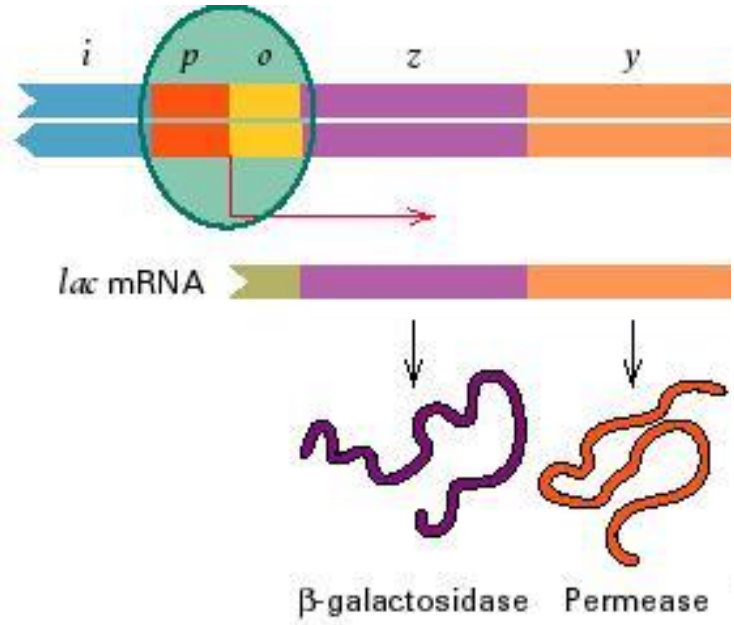
- The Lac Operon

  i = Regulatory Gene which codes for a repressor protein.

  p = Promoter region which binds RNA Polymerase

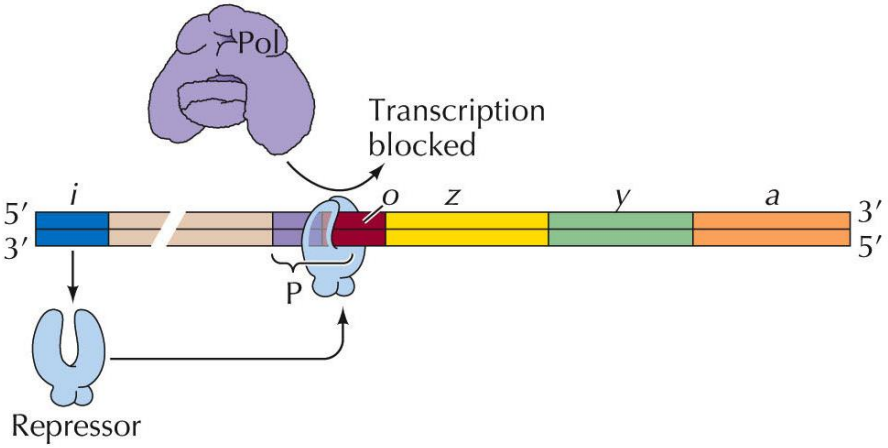
  o = Operator region which binds the repressor protein

  z = Structural gene for Beta-Galactosidase

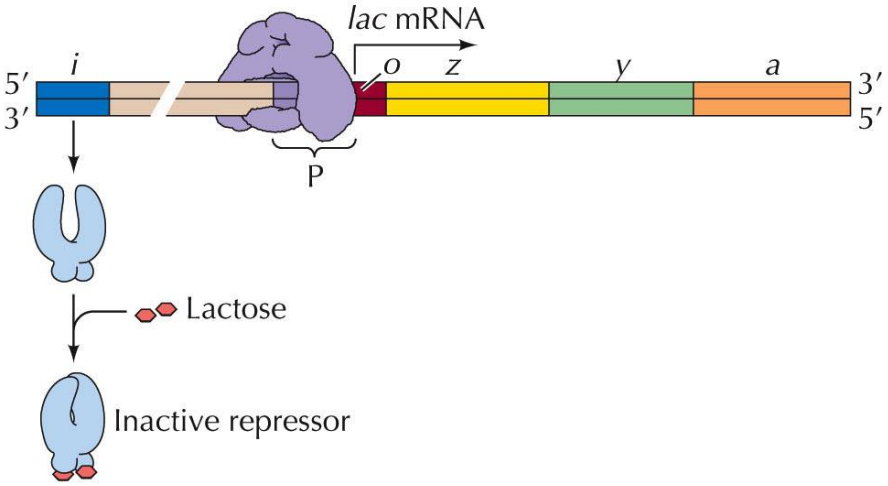


y = Structural gene for lactose permease.

- A Glucose Rich environment / Lactose Low environment
  1. The regulatory gene undergoes transcription and translation and a repressor protein is synthesised
  2. The repressor protein has 2 binding sites, one of which binds to lactose the other to the operator region.
  3. In the absence of lactose the repressor protein binds to the operator region and blocks part of the promoter region of the lac operon.
  4. This prevents RNA Polymerase binding to the promoter region
  5. This therefore prevents transcription of the structural genes Z for Beta-Galactosidase and Y for Lactose Permease.

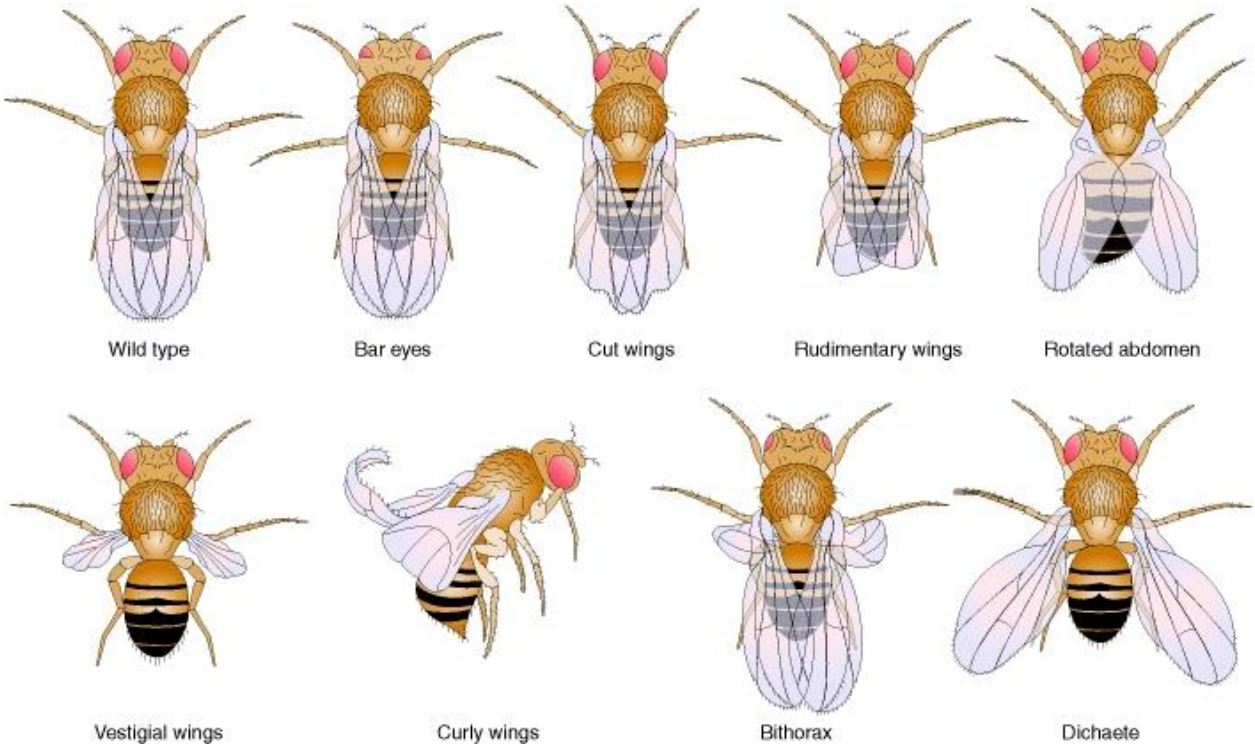


- A Glucose Low environment / Lactose Rich Environment
  1. The regulatory gene undergoes transcription and translation and a repressor protein is synthesised
  2. The repressor protein has 2 binding sites, one of which binds to lactose the other to the operator region.
  3. In the presence of lactose the repressor protein binds to the lactose, which alters the shape of the repressor protein.
  4. The repressor protein can now no longer bind to the operator region.
  5. This leaves the promoter region free to bind RNA Polymerase
  6. This therefore allows the transcription of the structural genes Z for Beta-Galactosidase and Y for Lactose Permease.
  7. Beta-Galactosidase catalyses the hydrolysis of lactose to glucose and galactose.
  8. Lactose permease increases the cell's permeability to lactose so it can take in more lactose from the surrounding environment.
  9. As the presence of lactose allows the synthesis of these proteins, lactose is referred to as a inducer molecule.

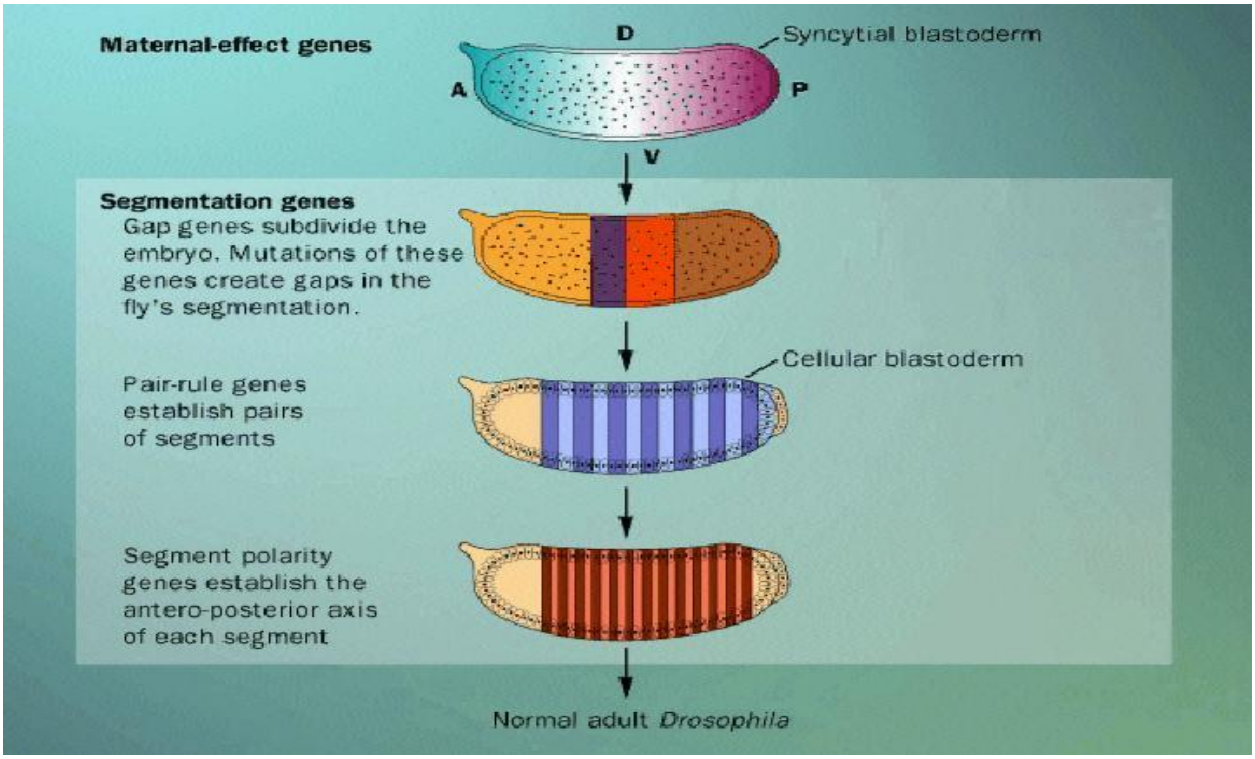


Explain that the genes that control development of body plans are similar in plants, animals and fungi, with reference to homeobox sequences.

- Homeotic genes are master genes or regulatory genes. They control the development of an organism from a zygote to offspring.
- Any mutations in these Homeotic genes are known as Homeotic mutations and much work has been carried out on Drosophila flies to investigate the effects that mutations in these genes can have on the organisms appearance



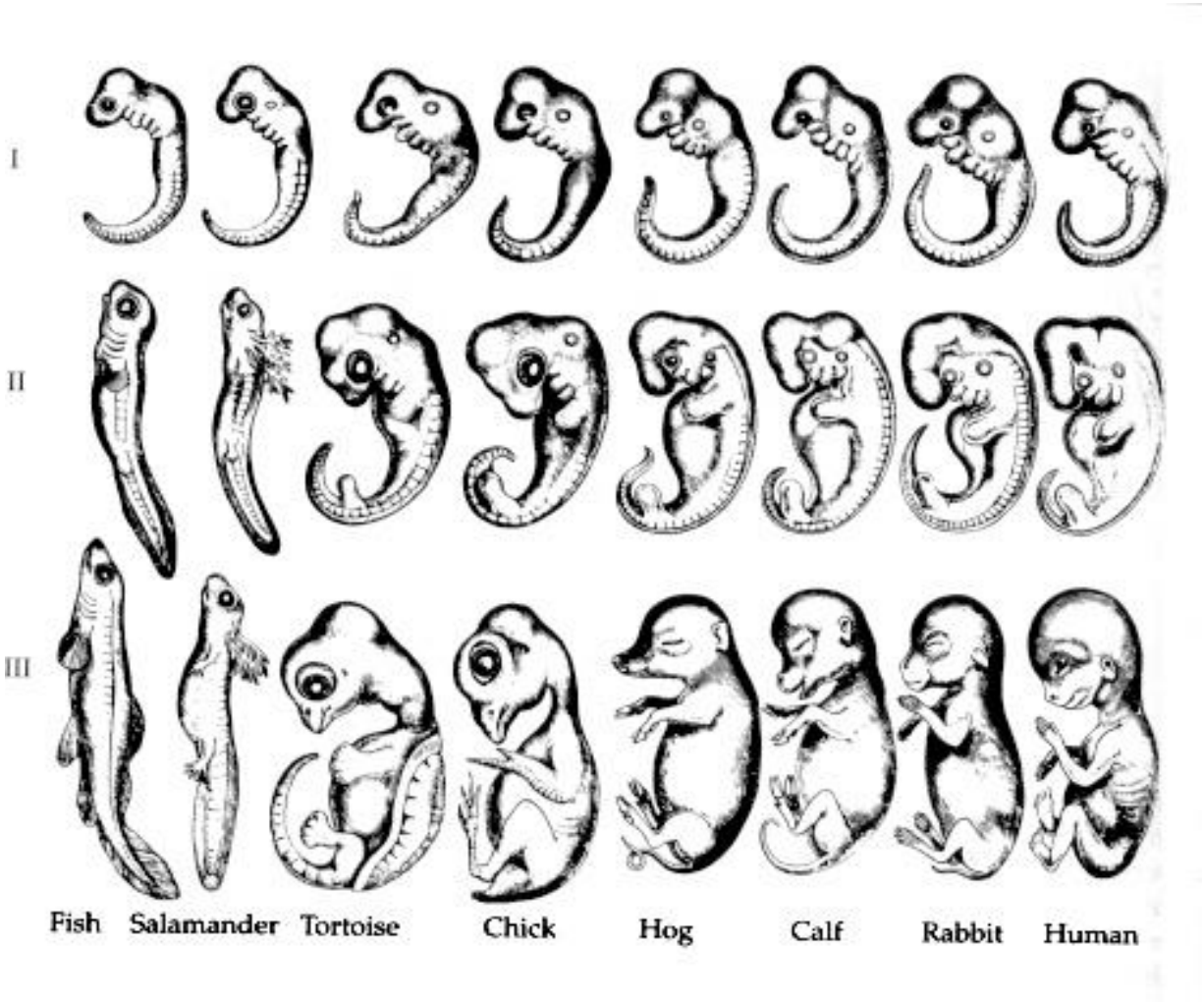
- Homeotic genes regulate the expression of other genes in an organisms genome.
- There are 3 categories of Homeotic genes



- Homeotic genes are Homeobox genes
- Homeobox genes are a type of gene which contains within its genetic code a sequence of 180 bases, which is referred to as a Homeobox.
- The Homeobox codes for a 60 amino acid long section of the polypeptide referred to as the Homeodomain.
- These Homeodomains are often transcription factors which can control the up or down regulation of transcription of other structural genes elsewhere within the organism's genome.
- Homeobox Genes are arranged in clusters known as Hox-Clusters. Each one usually containing between 9 – 11 Homeobox genes.
- Different organisms have different numbers of Hox-Clusters depending on their complexity.
    - Nematode worms – 1 Hox-Cluster
    - Drosophila worms – 2 Hox-Clusters
    - Vertebrates worms – 4 Hox-Clusters

- Homeobox genes are always expressed in a specific pattern and any deviation from this will result in abnormalities or defects to the organism's appearance.
- Retinoic acid (Vitamin A derivative) is a morphogen, activating Homeotic genes in vertebrates.
- Pregnant ladies are advised to avoid too much vitamin A (sourced from red meat and especially Liver) as it can interfere with normal gene expression.

- Homeotic genes control an organisms development and work in much the same way in all animals, plants and fungi

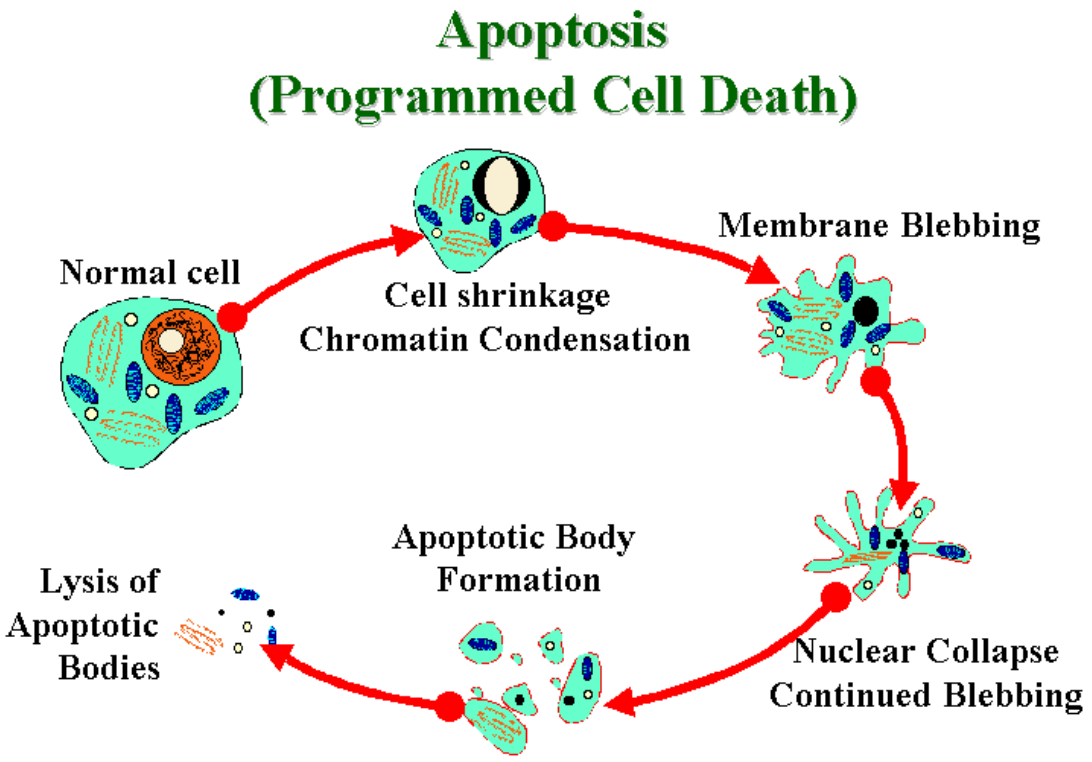


Outline how apoptosis (programmed cell death) can act as a mechanism to change body plans.

- Apoptosis is programmed cell death
- What?
  - It is ordered, tidy and does not affect neighbouring cells or tissues
- Why?
  - It removes excess cells allowing their components to be recycled into new cells.
  - It allows organisms to develop, i.e. digit formation.
  - Removes harmful cells (T-Lymphocytes that are complementary to our own antigens)
  - Removes ineffective cells (cells at the end of their lifecycle).
- How is it controlled?
  - Hormones
  - Cytokines – from infected cells

- Growth factors – protein signals which control growth
- Nitric Oxide - mitochondrial inner membranes become permeable
  - H^+ concentration dissipates
  - ATP production decreases or stops

- Apoptosis
  1. Enzymes digest the cytoskeleton
  2. Cytoplasm becomes dense as organelles become more tightly packed together
  3. Blebs form (bulging)
  4. Chromatin (DNA & proteins) condense and the nuclear envelope breaks
  5. DNA fragments
  6. The cell breaks apart forming apoptotic bodies (vesicle like structures)
  7. These bodies package the harmful hydrolytic enzymes preventing damage to other cells
  8. Phagocytosis of the cellular debris, some of which can be recycled.



- Necrosis is premature cell death
- Necrosis is caused by
  - Infection
  - Trauma
  - Toxins
- Necrosis is harmful to the body and can in some cases be fatal

- Hayflick's Limit
- This refers to the believed number of mitotic divisions a cell can undergo before it is highlighted be destroyed and its components recycled.

- If a mutation occurs in the section of DNA that is responsible for the highlighting process then the cell simply does not obey the hayflick's limit and continues to undergo mitosis
- This is described as uncontrolled cell division or in more common terms, cancer.

State that mutations cause changes to the sequence of nucleotides in DNA molecules.

- Mutations are changes to the sequence of DNA nucleotides in a gene
- They are a change to the genetic code during interphase of the cells lifecycle
- If a mutation occurs in a somatic (body) cell the effects are only present in that individual.
- If a mutation occurs in a gamete (sex) cell the effects will be present in future generations.
- There are 3 levels at which a mutation can occur
    - DNA level mutations – Changes in the arrangement of bases in an individual gene
    - Gene mutations – Changes in the chromosome, changing the arrangement of genes
    - Chromosome mutations – Addition or deletion of a chromosome

- What causes mutations?
    - Cigarette tar
    - UV Light
    - X-Rays
    - Gamma-rays

- DNA mutations can be referred to as Point mutations or Insertion/deletion/inversion mutations.

- Point Mutations (Substitutions)
    - 1 base or 1 base triplet is swapped for another
    - Original DNA sequence - the fat cat ate the wee rat
    - Mutant DNA sequence - the fat hat ate the wee rat
    - Examples
        - Sickle cell anaemia
            - Deoxygenated haemoglobin becomes fibrous in nature and cannot carry oxygen.
            - Caused by 1 amino acid VALINE replacing Glutamic acid
        - Proto-oncogenes (growth promoting genes)
            - Oncogenes →Prot-oncogenes (permenantly switched on)
            - This leads to tumours
- Deletion Mutations
    - Removal of 1 or more bases – results in a frameshift mutation
    - Original DNA sequence - the fat cat ate the wee rat
    - Mutant DNA sequence - the fat caa tet hew eer at

- Mutant DNA sequence - the fat ate the wee rat
- Example
  - Cystic Fibrosis
    - Over production of mucus in the respiratory, digestive & reproductive tracts
    - Deletion of 1 base triplet in a polypeptide 1,480 amino acids long

- Insertion Mutations
  - Addition of 1 or more bases – results in a frameshift mutation
  - Original DNA sequence - the fat cat ate the wee rat
  - Mutant DNA sequence - the fat cat hat ate the wee rat
  - Example
    - Huntington's Disorder
      - Onset in over 40s with symptoms such as forgetfulness, confusion, unbalance and slurred speech.
      - Insertion of a stutter where CAG is repeated

- Inversion Mutations
  - Inverting 1 or more base triplet
  - Original DNA sequence - the fat cat ate the wee rat
  - Mutant DNA sequence - the fat tar eew eht eta tac

Explain how mutations can have beneficial, neutral or harmful effects on the way a protein functions.

- Harmful Mutations
- Have a negative effect on a organisms phenotype and can affect its ability to survive and reproduce.
- Examples
  - Huntington's Disorder
  - Cystic Fibrosis
  - Sickle Celled Anaemia

- Neutral Mutations
- Have no affect on the phenotype or its ability to survive and reproduce.
  - Occur in non-coding regions of DNA

    - Silent mutants – changes the triplet code but it still results in the same amino acid
    - Codes for a new protein, however the protein has no effect on the organisms survivability
- Examples
    - Tongue rollers
    - Honey suckle smellers
    - Ear lobe shape
    - PTC tasters

- Beneficial Mutations
- All the living things you see around you are the result of positive mutations.

Questions

1. The bacterium *Escherichia coli* (*E. coli*) uses glucose as a respiratory substrate. In the absence of glucose, *E. coli* can use lactose. The use of a different substrate is determined by the interaction between genes and the environment.

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[Total 5 marks]

2. Cystic fibrosis (CF) in humans is caused by mutations of a gene coding for transmembrane protein (CFTR) which acts as an ion pump. A large number of different mutations of the gene have been found. Explain what is meant by a gene mutation.

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[Total 2 marks]

3. A product manufactured using microorganisms is single cell protein (SCP).

   Describe how a protein would be synthesised in the cell of a single celled fungus.

In your answer, you should make clear the sequence of the steps in the process.

[Total 10 marks]

4. DNA is found in the nucleus of a cell.

- During interphase DNA replicates.
- DNA is involved in the transcription stage of protein synthesis.

The following statements, **A** to **H**, refer to events that may take place during:

- ♦ DNA replication **only**
- ♦ transcription **only**
- ♦ **both** DNA replication **and** transcription
- ♦ **neither** DNA replication **nor** transcription.

Complete the table by marking the appropriate boxes with a tick (✓) if the event takes place or a cross (✗) if it does not take place.

		DNA replication	transcription
A	Nucleotides line up along an exposed DNA strand.		
B	The whole of the double helix 'unzips'.		
C	Uracil pairs with adenine.		
D	A tRNA triplet pairs with an exposed codon.		
E	Both DNA polynucleotide chains act as templates.		
F	Adjacent nucleotides bond, forming a sugar-phosphate backbone.		
G	The original DNA molecule is unchanged after the process.		
H	Adenine pairs with thymine.		

[Total 8 marks]

5.

(a) Part of the **DNA** base sequence coding for a protein is shown below.

A T G G C C T A A G T G

(i) State the corresponding base sequence of **mRNA**.

.....

[2]

(ii) Name the process by which the DNA code is transferred to mRNA.

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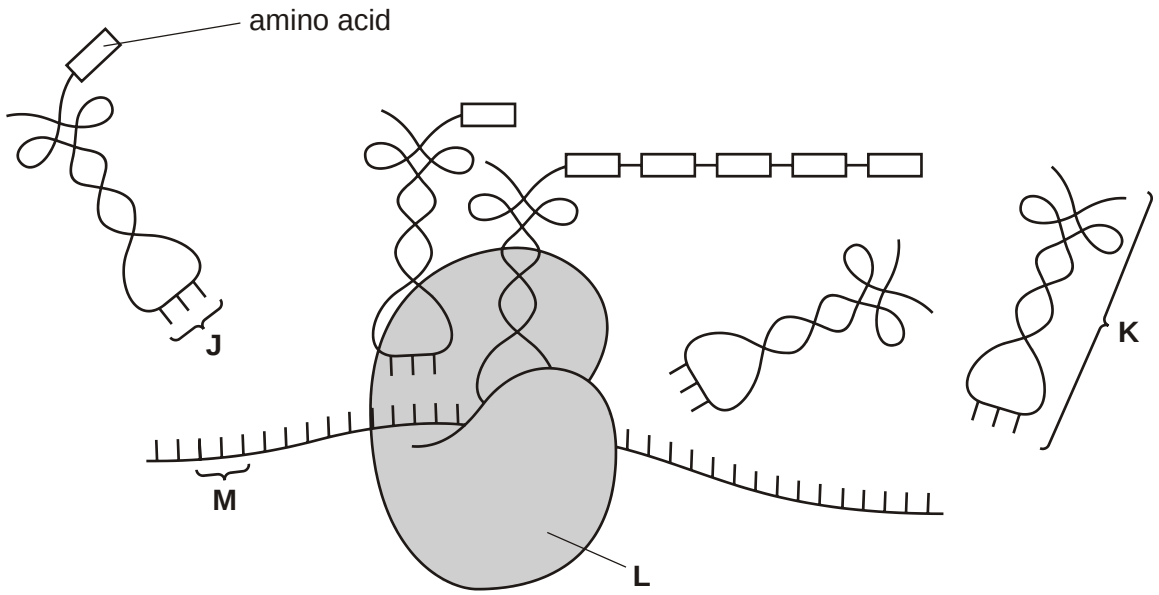
[1]

(b) The figure below is a diagram that shows the stage in protein synthesis when amino acids are joined in the correct sequence to make the primary structure of the protein.



(i) Name **J** to **M**.

The group of bases at **J** .....

K .....

.....

L .....

The group of bases at **M** .....

[4]

(ii) Using the information in the diagram to help you, explain how amino acids become arranged into the correct sequence in the primary structure of the protein.

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[4]

(c) Mistletoe is a parasitic plant that produces lectin 1, a ribosome-inactivating protein. Lectin 1 inhibits protein synthesis in the cells of the host plant.

Suggest how lectin 1 could inhibit protein synthesis.

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[2]

[Total 13 marks]

6. Liver cells damaged by hepatitis infection switch on a gene called Fas, which causes them to self-destruct. Pioneering research has produced a strikingly successful treatment for hepatitis in mice. The Fas gene was silenced by the technique of RNA interference.

RNA molecules, 21 to 23 nucleotides long, were injected into mice with hepatitis. The sequence of this 'small interfering RNA' (siRNA) matched part of the Fas gene. Once in the liver cell the two strands of the siRNA were separated so that one strand could bind to the mRNA transcript of the Fas gene. This caused the mRNA to be destroyed by enzymes, therefore preventing the gene product from being made.

This therapy prevented liver cell death and considerably increased the survival of mice with hepatitis.

(a) (i) Describe a way in which the **function** of mRNA differs from that of DNA.

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[2]

(ii) Describe **two** ways in which the **structure** of siRNA differs from mRNA.

1 .....

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2 .....

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[2]

(b) Describe how one strand of the siRNA can bind to the mRNA of the Fas gene.

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[3]

[Total: 7 marks]

7.

(a) Repeating nucleotide sequences are common in the genomes of eukaryotes, for example in the centromeres and in the regions, called introns, which appear to interrupt the genes. Repeating sequences have been referred to as 'junk DNA'.

Suggest why the term 'junk DNA' is misleading.

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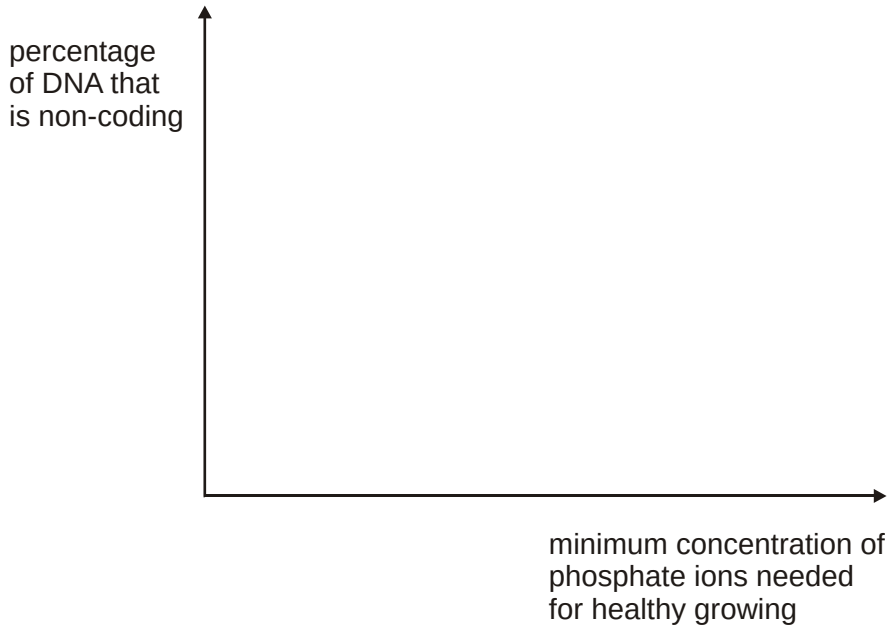
[2]

(b) Some species of plant are able to grow on soils that contain very little phosphate, while other species, for example stinging nettles, can only grow well in soils that are rich in phosphate. Each nucleotide in a DNA molecule includes a phosphate group.

If much of the non-coding DNA can be correctly regarded as functionless 'junk', there may be a correlation between the percentage of DNA that is non-coding and the minimum concentration of phosphate ions needed for healthy growth.

Draw a straight line graph, using the axes in the figure below, to show the correlation that you would predict.



[1]

(c) Name a substance **other than DNA** that is found in cells and has one or more phosphate groups as part of its chemical structure.

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[1]

[Total 4 marks]

8. An enzyme, such as amylase, has a specific 3-dimensional shape.

Explain how DNA structure determines the specific shape of enzymes.

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[Total 4 marks]