Contents

Topic 2.1, 2.2 Structure and Function of Nucleic Acids & DNA Replication

CJC

3 **(a)** State how **one** structural feature of DNA contributes to its stability as a hereditary material.

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..... [1]

The "trombone" model of DNA replication postulates that two DNA polymerase enzymes work together in a protein complex during DNA replication.

Fig. 3.1 shows a DNA molecule undergoing DNA replication.

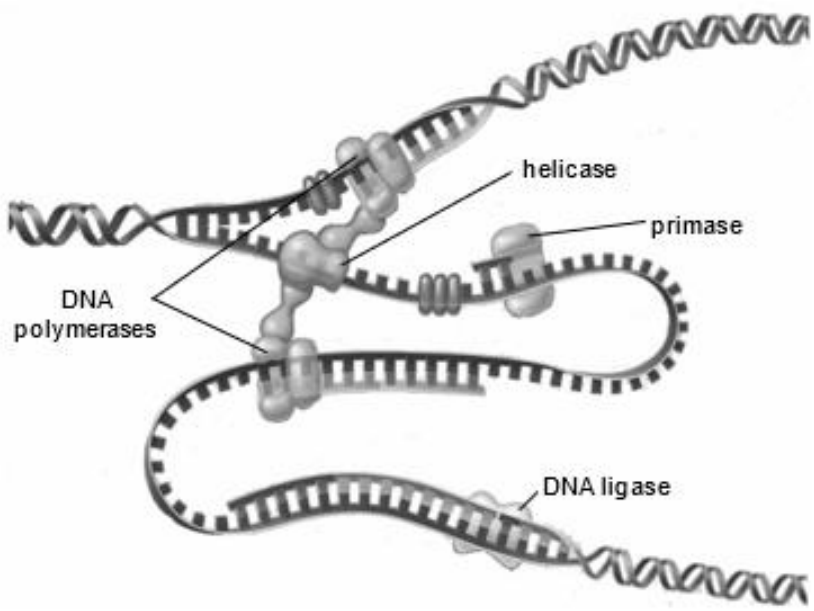

Fig. 3.1

(b) With reference to Fig. 3.1,

(i) compare how the synthesis of the lagging strand differs from that of the leading strand.

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..... [2]

(ii) suggest why it is necessary for two DNA polymerase enzymes to work together in a protein complex during DNA replication.

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..... [2]

(c) Explain how the end replication problem arises.

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..... [2]

[Total: 7]

MI

3.

(a) State the function of DNA and describe its property that allow it to perform this function.

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..... [2]

(b) In a somatic cell of a eukaryote, 20% of the nitrogenous base in the nuclear DNA is thymine.

Calculate the percentage of nitrogenous base in the nuclear DNA of this cell that is guanine and explain your answers.

Show your working clearly in the space below.

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..... [3]

(c) Fig. 3.1 shows the process of DNA replication.

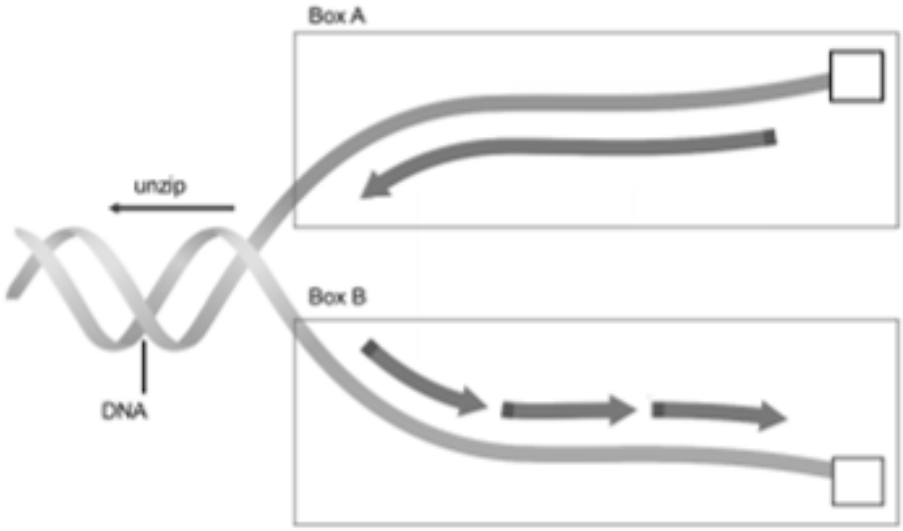

Fig. 3.1

(i) In the boxes provided in Fig. 3.1, label the end of DNA with either

- 3' or
- 5'.

[1]

(ii) Explain why DNA is replicated differently in Box **A** and Box **B**.

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..... [2]

(d) During protein synthesis, DNA is used as a template to form mRNA and the resulting mRNA is used as a template to form polypeptide chain.

Describe 2 other differences between the enzyme used to form mRNA and the enzyme used to form polypeptide chain.

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..... [2]

[Total :10]

Topic 2.3 Cell and Nuclear Division

ASRJC

2 Fig. 2.1 is a photomicrograph of root tip cells at different stages in the cell cycle.

A cell in interphase and telophase are labelled.

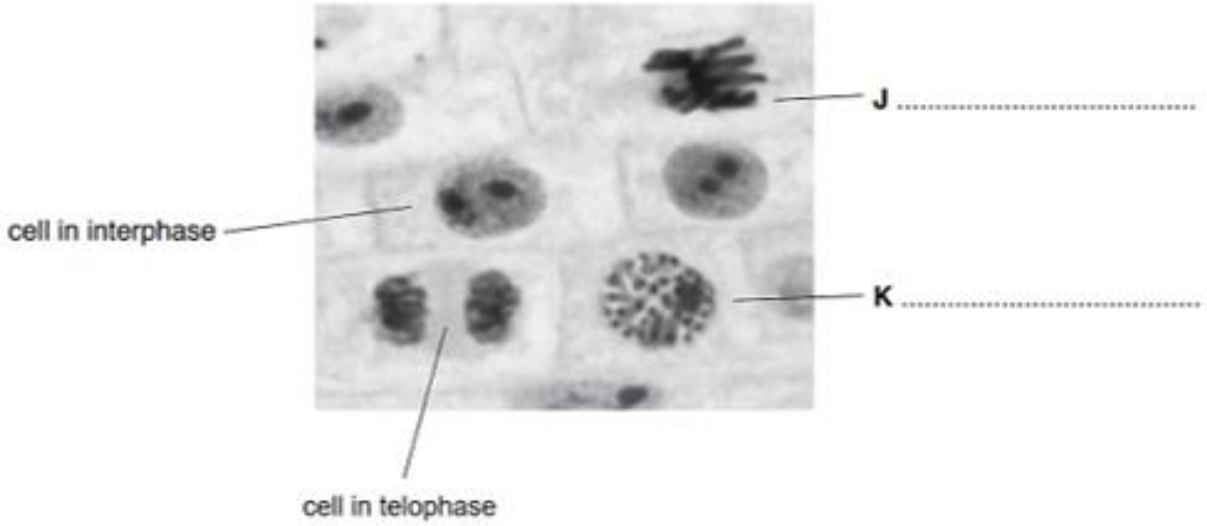

Fig. 2.1

(a) **(i)** Complete Fig. 2.1 by naming the stage of mitosis shown in each of cells **J** and **K.** [1]

(ii) State **one** feature of the cell in interphase, **visible** in Fig. 2.1, that shows this cell is **not** in early interphase.

.....

.....[1]

(b) Describe the events that occur in telophase.

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.....[2]

(c) Reduction division happens in meiosis.

(i) Describe the events that cause reduction division.

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.....[2]

(ii) Explain the need for reduction division during meiosis.

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.....[3]

[Total: 9]

JPJC

7 **(a)** Fig. 7.1 is a photomicrograph of animal cells in stages of the mitotic cell cycle.

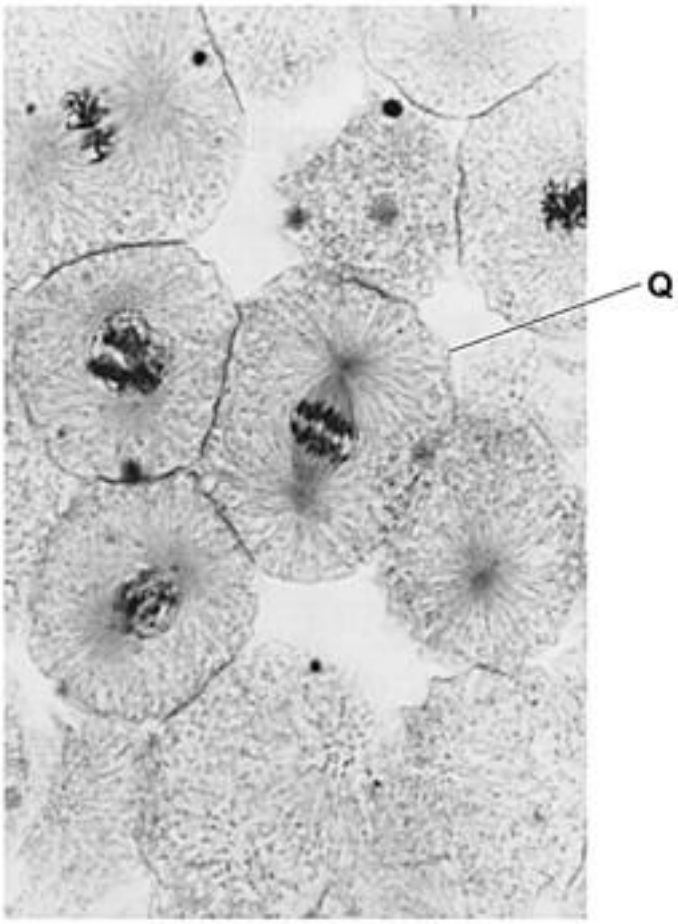

Fig. 7.1

(i) Name the stage of mitosis shown in cell Q.

..... [1]

(ii) Outline the roles of mitosis in a healthy animal.

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..... [2]

(b) Uncontrolled mitosis can cause cancer in humans.

Paclitaxel is a drug used in the treatment of some forms of cancer.

Researchers investigated the effect of Paclitaxel on the mitotic cell cycle of cancer cells.

- The cancer cells were grown for two days and then divided into groups.
- Each group was treated with a different concentration of Paclitaxel.

After 28 hours (one cell cycle):

- the percentage of cells in stages of mitosis was calculated.
- the ratio of the number of cells in anaphase to the number of cells in metaphase was determined.

The results of the investigation are shown in Fig. 7.2.

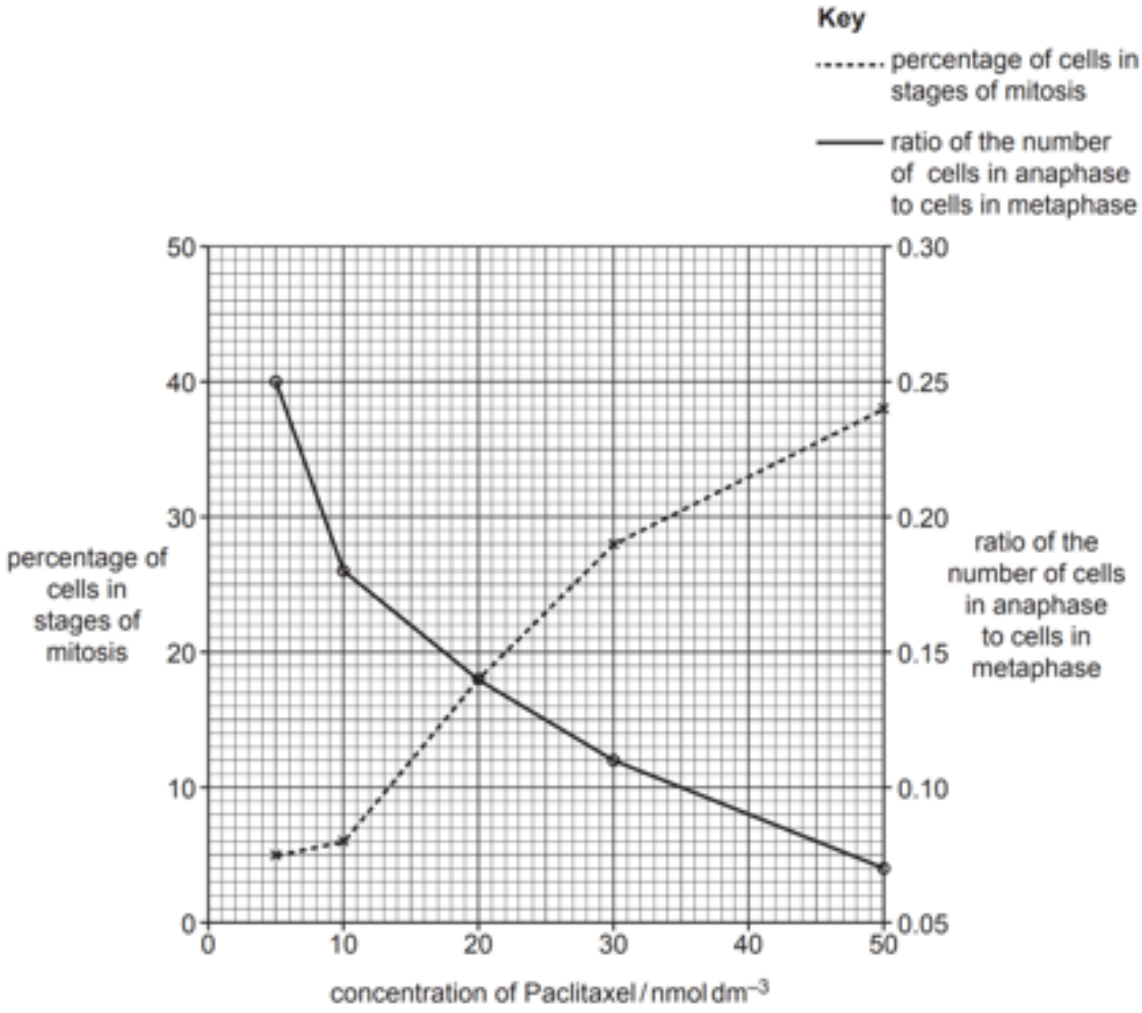

Fig. 7.2

With reference to Fig. 7.2, describe the results and suggest an explanation for the effect of Paclitaxel on the mitotic cell cycle.

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..... [4]

(c) Multiple myeloma is a type of cancer in the bone marrow where some of the stem cells start to produce abnormal blood cells.

Some treatments available are stem cell transplantation, immunotherapy and chemotherapy.

(i) In stem cell transplantation, stem cells are collected from the bone marrow of the person with multiple myeloma. Healthy stem cells are isolated and grown in the laboratory. Radiation is then used to destroy all stem cells and cancerous cells in the bone marrow. Finally, large numbers of the healthy stem cells grown in the laboratory are returned to the bone marrow.

Suggest the role of stem cells in this treatment of multiple myeloma.

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..... [3]

[Total: 10]

TMJC

3 Fig. 3.1 shows a pair of homologous chromosomes during one of the stages of meiosis.

The letters **G** to **M** represent the dominant alleles of seven genes and the letters **g** to **m** represent the recessive alleles of the same seven genes.

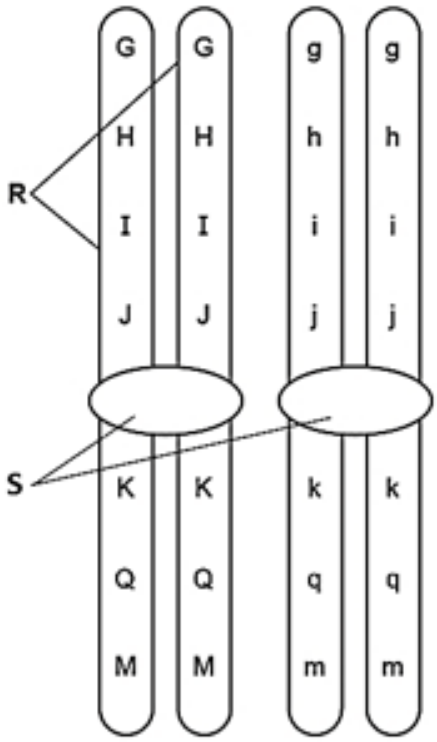

Fig. 3.1

(a) (i) Name the structures labelled **R** and **S** on Fig. 3.1. [2]

R .....

S .....

(ii) State **three** features visible on Fig. 3.1 that identify the chromosomes as a homologous pair. [3]

1 .....

.....

2 .....

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3 .....

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(iii) Fig. 3.2 shows the same two chromosomes a little later in the same stage of meiosis. Crossing over is starting to occur at point **T**.

Fig. 3.3 shows an outline of the same two chromosomes after crossing over has occurred.

Complete Fig. 3.3 by writing in the letters of the alleles along both chromosomes. Take care to clearly show the difference between letters representing dominant alleles and letters representing recessive alleles. [2]

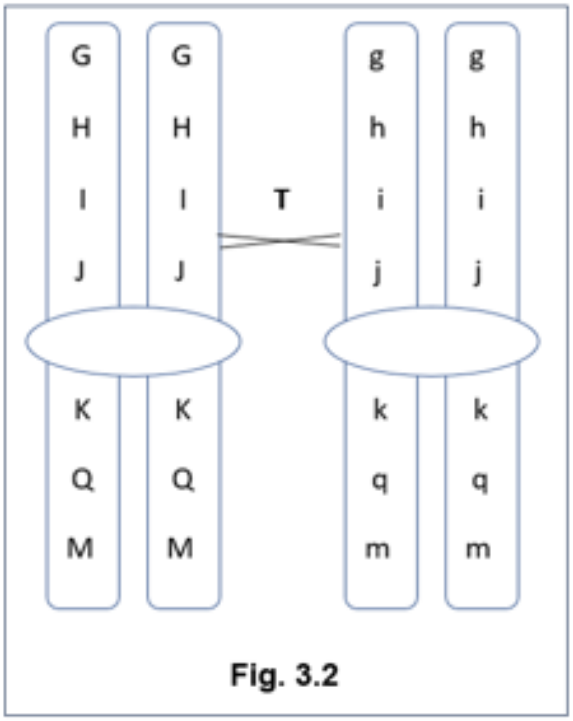

Fig. 3.2

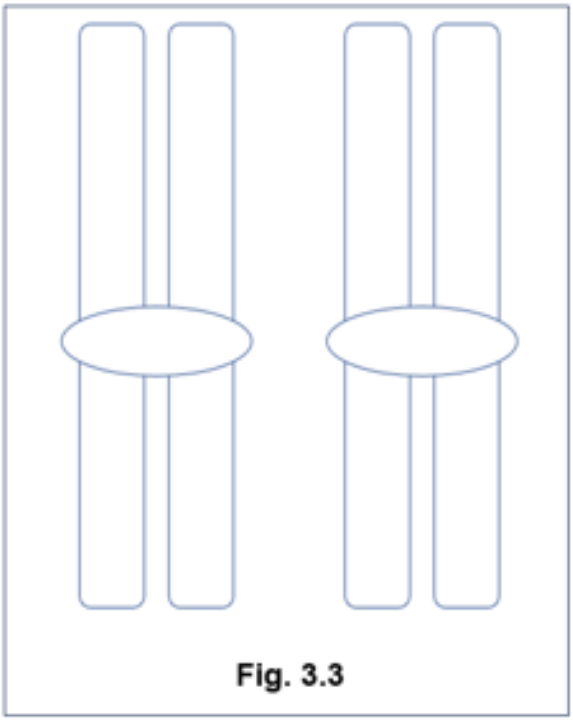

Fig. 3.3

(b) State the stage in meiosis in which crossing over occurs. [1]

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(c) Crossing over results in genetic variation.

Explain how random assortment of homologous chromosomes also results in genetic variation. [2]

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(d) The diploid number of chromosomes of this organism is ten.

In the absence of crossing over or mutation, state the number of genetically unique kinds of gametes that might be formed by one individual. [1]

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[Total: 11]

Topic 2.4, 2.5 Gene Expression and Regulation

ACJC

4 Fig. 4.1 outlines the production of a protein in a eukaryotic cell.

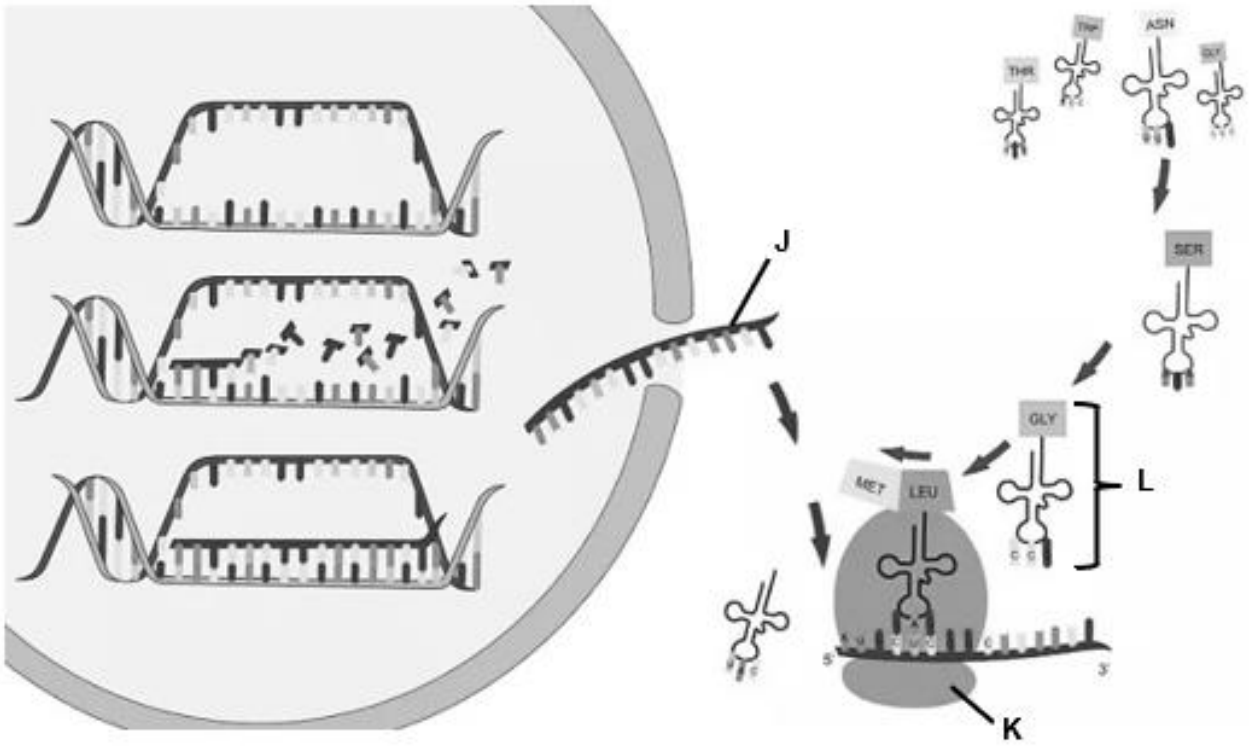

Fig. 4.1

(a) Identify structures **J**, **K** and **L**.

J .....

K .....

L ..... [3]

(b) Outline the events in protein synthesis that continue from what is shown in Fig. 4.1 which leads to the synthesis of a complete polypeptide.

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..... [3]

(c) Protein T, not shown in Fig. 4.1, is normally involved in the initiation of translation.

The unfolded protein response (UPR) is a cellular stress response that is triggered by an accumulation of unfolded or misfolded proteins after a high rate of translation. During the UPR, a kinase known as PERK is activated and acts on protein T.

Suggest how the activation of PERK prevents the accumulation of unfolded or misfolded proteins.

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..... [2]

ACJC

6 Gene expression can be controlled at the transcriptional level. An investigation was carried out to find out the effect of an enhancer sequence on the transcription of a gene.

Fig. 6.1 summarises the results of the investigation, with five different experiments, **(a) to (e)**:

(a) shows the gene without an enhancer, while **(b)** to **(e)** shows the gene under the influence of the enhancer at different arrangements.

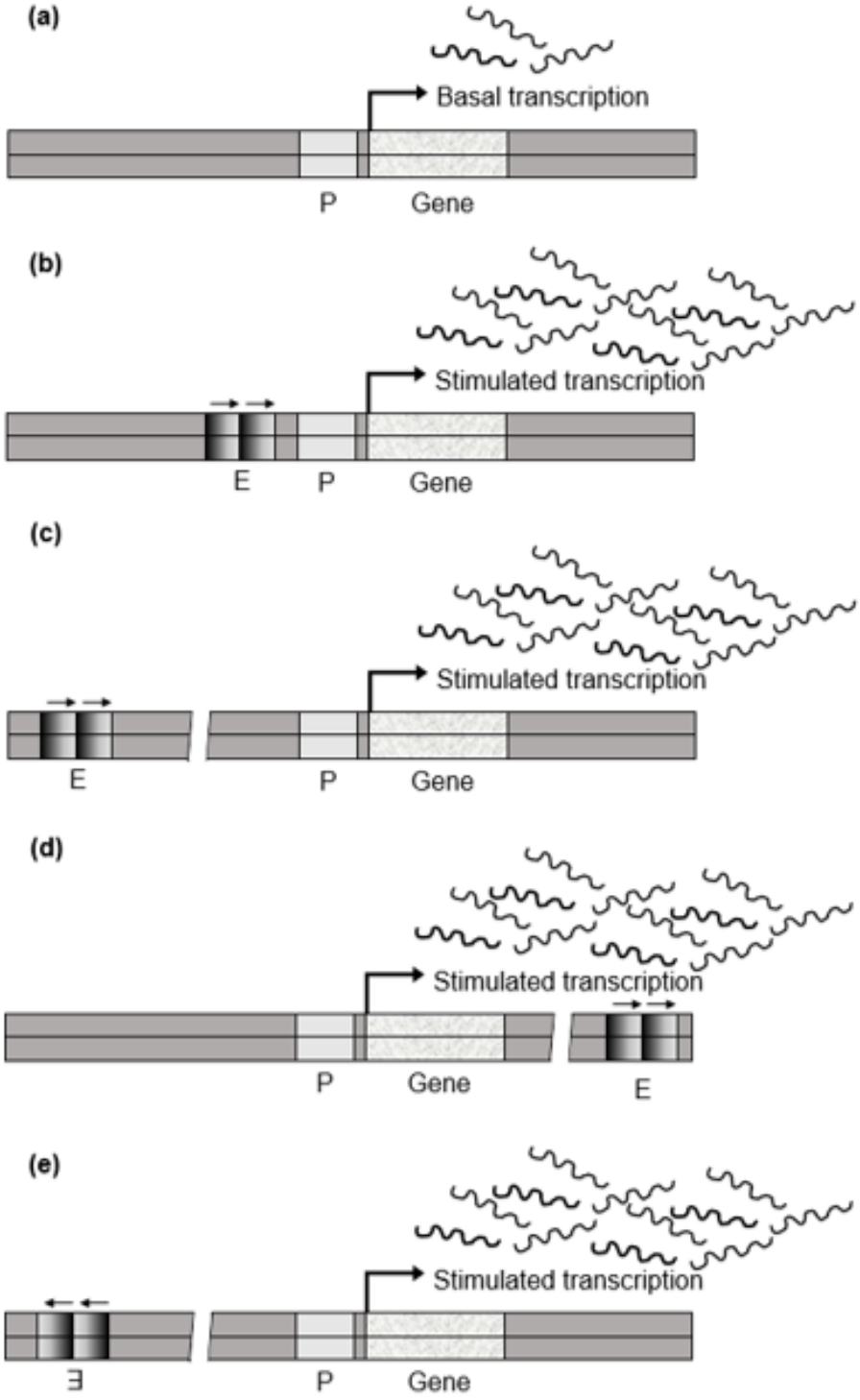

Fig. 6.1

(a) Describe the function of the enhancer.

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..... [3]

(b) Explain how the enhancer is able to perform its function in the new location shown in Fig. 6.1**(c)**.

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..... [2]

(c) With reference to Fig. 6.1**(d)** and 6.1**(e)**, describe the relationship between the activity of the enhancer and its arrangements.

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..... [2]

Specificity protein 1, Sp1, was found to stimulate transcription of a gene. It was discovered that Sp1 binds specifically to GC box sequences found within the gene. In order to study Sp1 further, the method shown in Fig. 6.2 was used to obtain and purify Sp1.

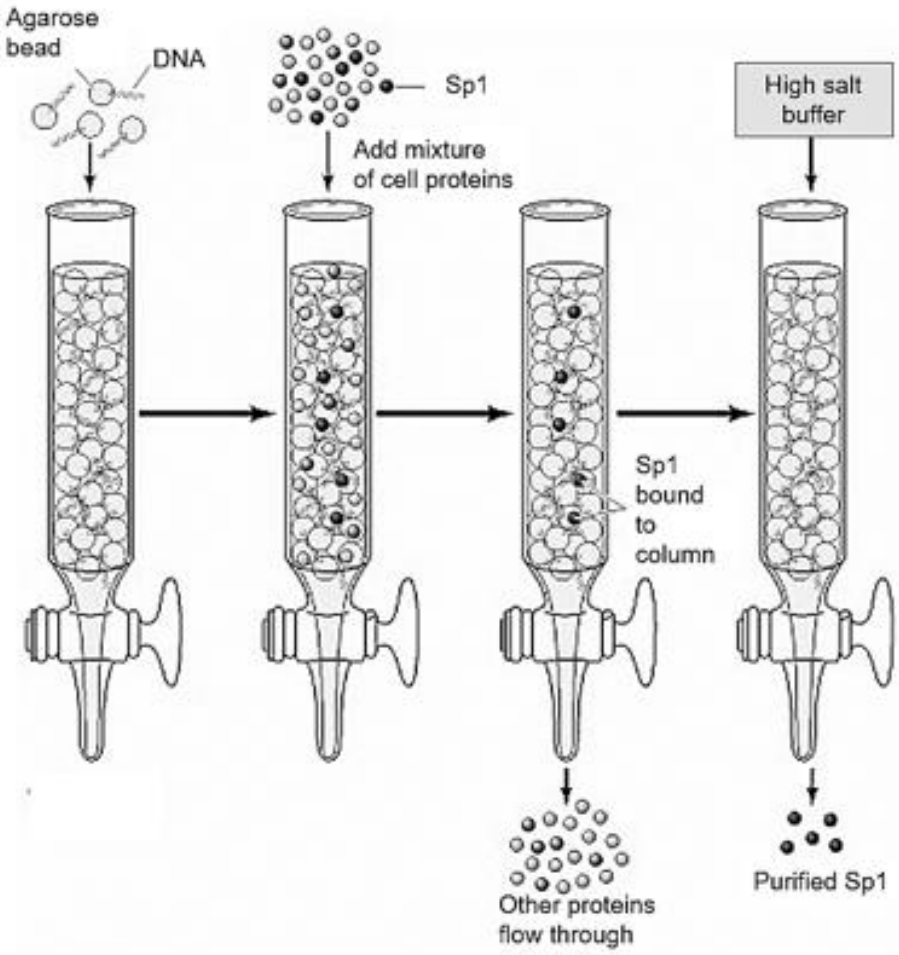

Fig. 6.2

(d) With reference to Fig. 6.2, suggest how the method purifies Sp1.

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..... [3]

[Total: 10]

CJC

4 Nucleolus is a large and dense region inside the nucleus. It consists of a fibrous part and a granular part. The large and small ribosomal subunits can be found within the granular part.

(a) Based on the information above as well as your knowledge on transcription and translation, outline how ribosomes are formed.

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..... [4]

Gene expression in eukaryotic cells is regulated at multiple levels.

Transferrin receptors (TfRs) are cell surface receptors and are involved in the uptake of extracellular iron.

The regulation of expression of transferrin receptors (TfRs) is illustrated in Fig. 4.1.
It involves the interaction between Iron Response Element-Binding Protein (IRE-BP) and Iron Response Element (IRE) which is a loop structure found on the mRNA.

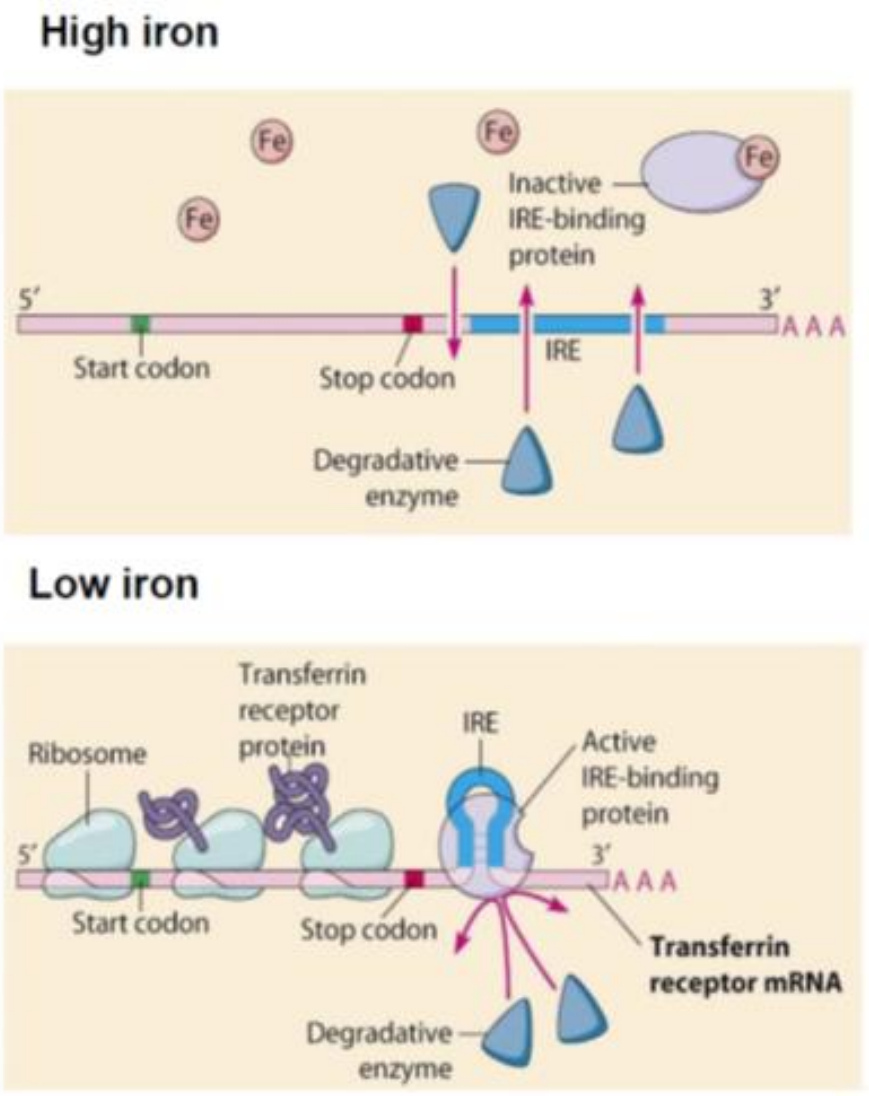

Fig 4.1

With reference to Fig 4.1.,

(b) **(i)** state the level of regulation shown.

..... [1]

(ii) explain how low cytosolic iron concentration results in an increased uptake of extracellular iron.

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..... [4]

Another example of a protein, Ferritin proteins; are involved in the storage of iron taken into the cell by TfRs.

The regulation of ferritin expression is illustrated in Fig. 4.2.

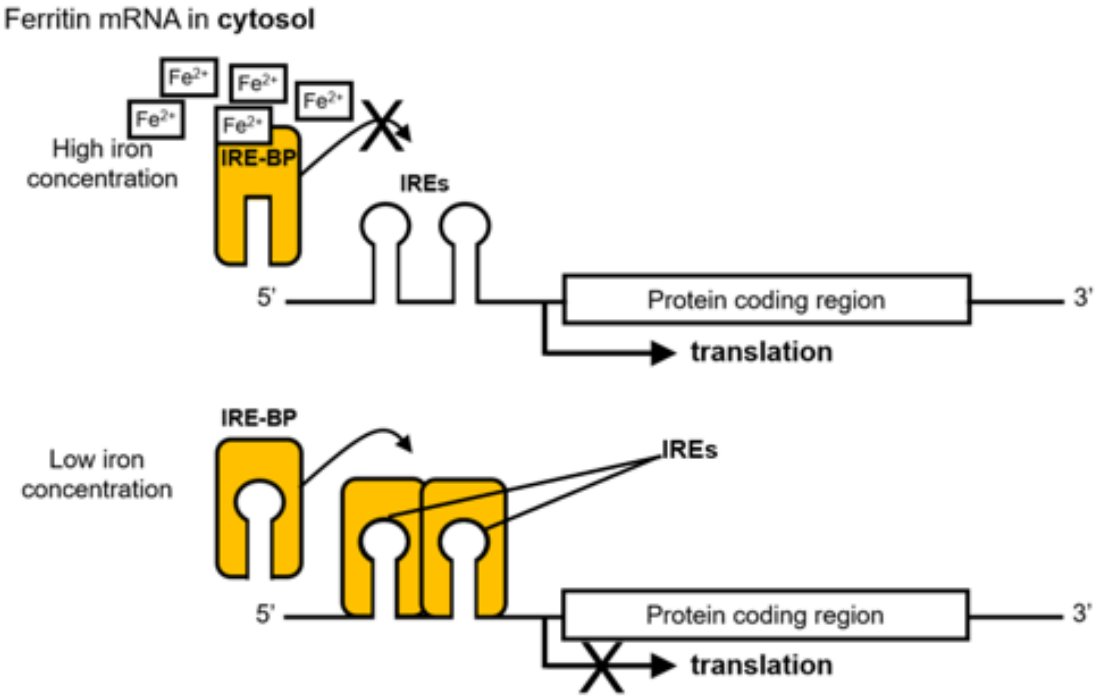

Fig 4.2

(c) Outline two differences in the regulation of ferritin and TfR expression.

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..... [2]

[Total: 11]

JPJC

4 **(a)** Explain the meaning of the terms:

(i) gene

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.....

..... [1]

(ii) codon.

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..... [1]

(b) Fig. 4.1 shows part of the sequence of events in the assembly of the enzyme lysozyme which consists of 129 amino acids.

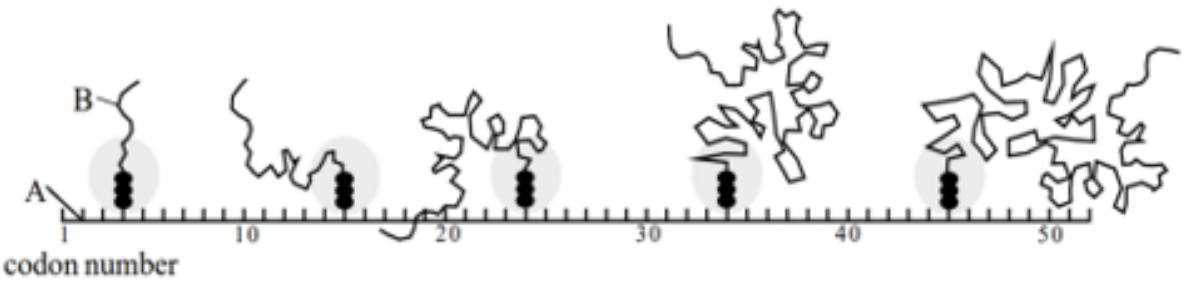

Fig. 4.1

(i) Identify structures **A**, and **B**.

A .....

B .....

[2]

(ii) Describe how the α-helix of the secondary structure of lysozyme is held together.

.....

.....

..... [1]

(iii) Describe one reason why ATP is required for assembly of lysozyme.

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..... [1]

(c) Lysozyme is capable of splitting a polysaccharide found in the bacterial cell wall.

Fig. 4.2 shows the structure of this polysaccharide. Lysozyme catalyses the hydrolysis of the β (1-4) bond between N-acetylglucosamine and N-acetylmuramic acid residues of bacterial cell walls.

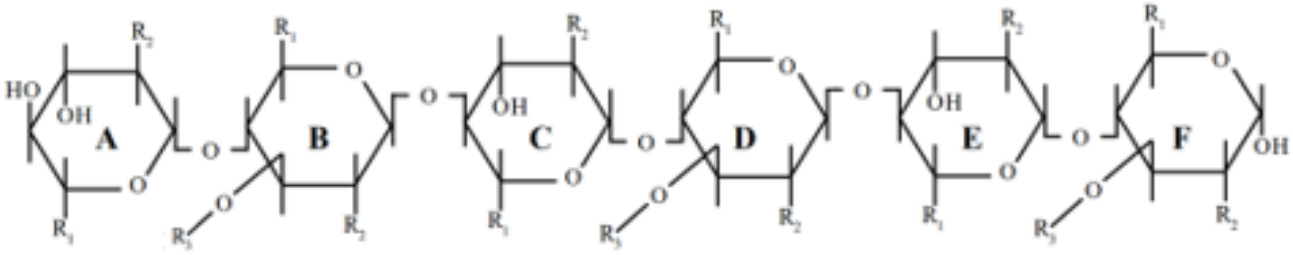

Fig. 4.2

Explain the significance of the assembly process shown in Fig. 4.1 to the activity o lysozyme shown in Fig. 4.2.

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..... [4]

[Total: 10]

SAJC

QUESTION 6

(a) DNA codes for proteins within the cell. Some regions of DNA are described as non-coding.

(i) Explain why some regions of DNA can be described as 'non-coding'.

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.....[2]

(ii) Non-coding regions of DNA show more variation than coding regions. This makes non-coding regions useful in DNA profiling. DNA profiling can be used in cases of paternity and forensics.

Suggest why non-coding regions of DNA show more variation.

.....

.....[1]

(b) In eukaryotic cells, all RNA molecules are synthesised as pre-RNA (primary RNA transcript) and undergo some form of post-transcriptional modification to form mature RNA. For example, DNA template strand is transcribed into pre-mRNA which then undergoes post-transcriptional modification to form mature mRNA. tRNA and rRNA similarly are first synthesised as pre-tRNA and pre-rRNA, and undergo post-transcriptional modification to form mature tRNA and rRNA, respectively.

Fig. 6.1 shows part of a **pre-tRNA** molecule. Geneticists identified two mutations that can affect **this** pre-tRNA, as shown in **Fig. 6.1**.

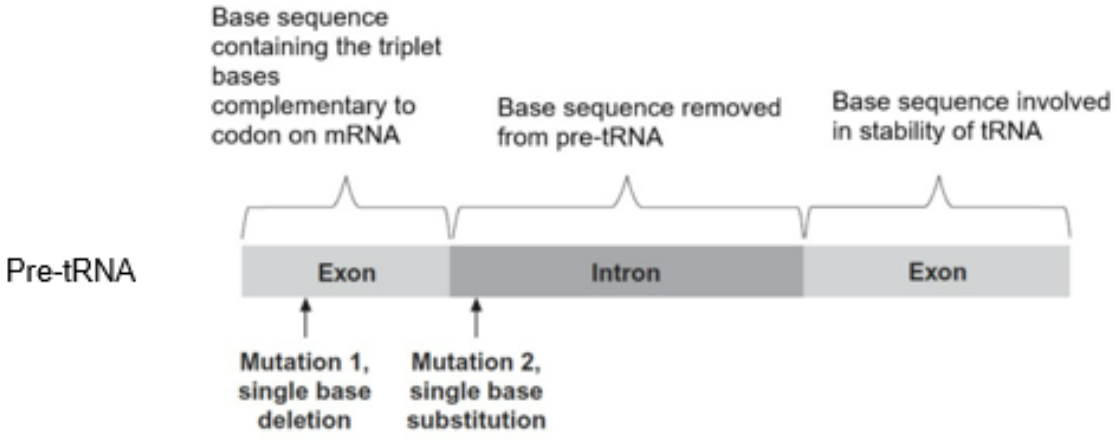

Fig. 6.1

(i) Assuming the mature tRNA formed from this mutated pre-tRNA, binds to the same amino acid as the non-mutated tRNA, explain why **mutation 1** to **this** pre-tRNA leads to the production of a less functional protein.

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.....[3]

(ii) Suggest possible effect(s) **mutation 2** might have on the tRNA produced using your content knowledge and understanding of post-transcriptional modification of mRNA.

Explain your answer.

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.....[2]

(c) Contrast between translation in eukaryote and prokaryote.

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.....[1]

[Total: 9]

Topic 2.6, 2.7 DNA Mutation and Chromosomal Aberrations

MI

4. Sickle cell anaemia is most commonly caused by the haemoglobin variant HbS, a result of a point mutation.

(a) Describe the effect of the point mutation to haemoglobin.

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..... [4]

(b) Sickle cell anaemia can be treated with a drug called hydroxyurea which induces the formation of fetal haemoglobin (HbF). HbF is normally found in fetus and newborn. When present in individuals with sickle cell anaemia, HbF prevents sickling of red blood cells.

(i) Suggest how formation of HbF would be induced.

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..... [2]

(ii) Suggest how elevated levels of HbF may reduce the symptoms of sickle cell anaemia.

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..... [2]

(c) Sickle cell anaemia is caused by a somatic mutation as it affects the somatic cell. On the other hand, germline mutation affects the gametes.

Explain why somatic mutation may have milder consequence than germline mutation.

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..... [2]

(d) Mitochondrial complex I deficiency is the most common mitochondrial disorder present in childhood. It can be caused by mutation in mitochondrial DNA (mtDNA) or mutation in nuclear DNA.

The characteristics of the deficiency caused by mutations in mtDNA are:

- a cell in an ovary produces gametes with different proportion of normal mitochondria and mitochondria that contains the mtDNA mutation
- a person has disease symptoms when the proportion of mutant mitochondria in their cells exceed a certain threshold
- the severity of disease symptoms, and the age at which they appear, can vary greatly in the children of one woman.

In a family with history of mitochondrial complex I deficiency that is caused by mutation in a nuclear DNA, the probability of a child inheriting the mutation can be predicted.

Suggest why, in families where mitochondrial complex I deficiency is caused by mtDNA mutation, it is not possible to predict the probability of a child inheriting the mutation.

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..... [2]

[Total :12]

SAJC

QUESTION 4

The red blood cells of patients with sickle cell anaemia have reduced oxygen-carrying capacity.

(a) Explain how gene mutation causes a lowering of solubility of haemoglobin S in sickle red blood cells.

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.....[5]

The early detection of sickle cell anaemia can help to reduce mortality rates and early intervention can be initiated to manage the disease effectively. Different techniques have been developed to detect the sickle cell disease and the carrier states with high sensitivity and specificity.

One of the ways is through electrophoresis of the isolated haemoglobin molecules. In this technique, red blood cell lysates containing the haemoglobin molecules are separated at alkaline pH. Under this condition, all haemoglobin molecules show a negative charge.

Fig. 4.1 shows the results obtained from the separation of haemoglobin molecules isolated from red blood cell lysates of individuals who are normal (genotype Hb^AHb^A) and who have sickle cell anaemia (genotype Hb^SHb^S). Equal concentrations of the proteins were loaded in each well before separation.

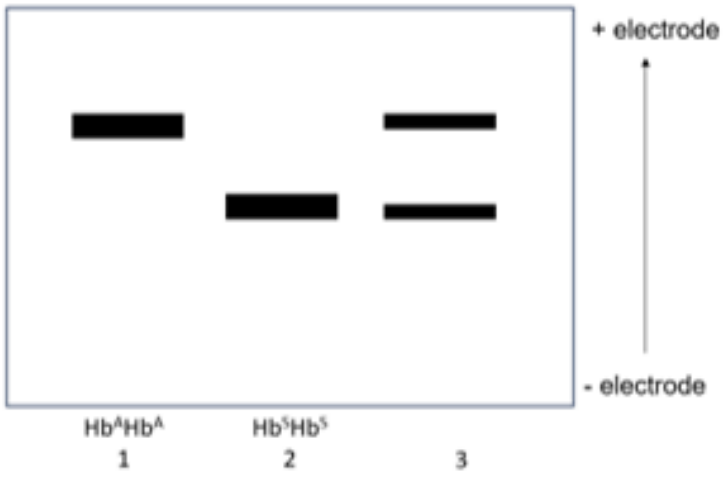

Fig. 4.1

(b) Explain how gel electrophoresis may be used to distinguish between the HbA and HbS proteins in lanes 1 and 2 respectively.

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.....[1]

(c) State the genotype of the individual who provided the red blood cell lysates in lane 3. Explain your answer.

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.....[2]

The term "relative fitness" describes the total number of offspring an organism has, compared to the average number of offspring for the population.

Fig. 4.2 shows the relative fitness of individuals who are normal, diagnosed with sickle cell anaemia (SC) and sickle cell trait (SCT). Individuals with sickle cell trait do not usually show symptoms of sickle cell anaemia although they only have a copy of the normal allele.

The fitness levels of all 3 groups of individuals have been studied in both areas with and without malaria. Those with sickle cell anaemia generally show low fitness levels due to the development of various health complications which affect their overall well-being and ability to conceive or carry a pregnancy to term.

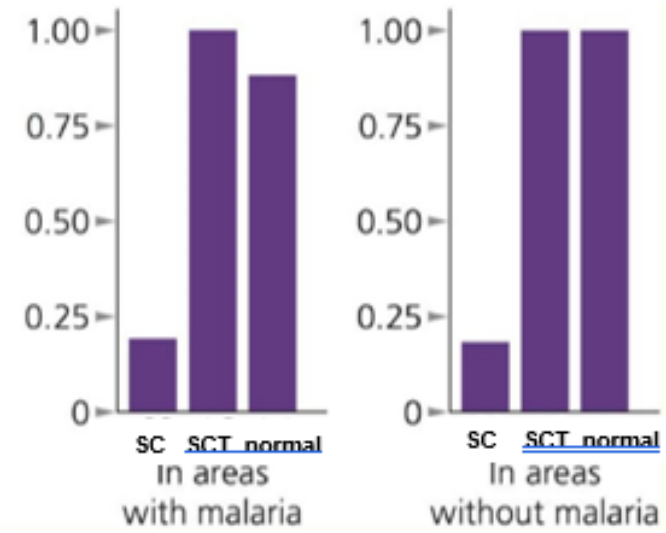

Fig. 4.2

(d) With reference to Fig. 4.2, account for the difference in relative fitness of individuals who are normal and those who show the sickle cell trait.

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.....[4]

[Total : 12]

Topic 2.8 Molecular Biology of Cancer

JPJC

Refer to JPJCQ7 in Topic 2.3 Cell and Nuclear Division.

Topic 2.9 Genetics of Viruses

ACJC

5 SARS-CoV-2 is the virus responsible for the COVID-19 pandemic. It is a positive-sense single-stranded RNA enveloped virus which uses its viral spike glycoproteins to infect human cells bearing ACE2 receptors.

Fig. 5.1 shows the viral reproductive cycle within an infected cell.

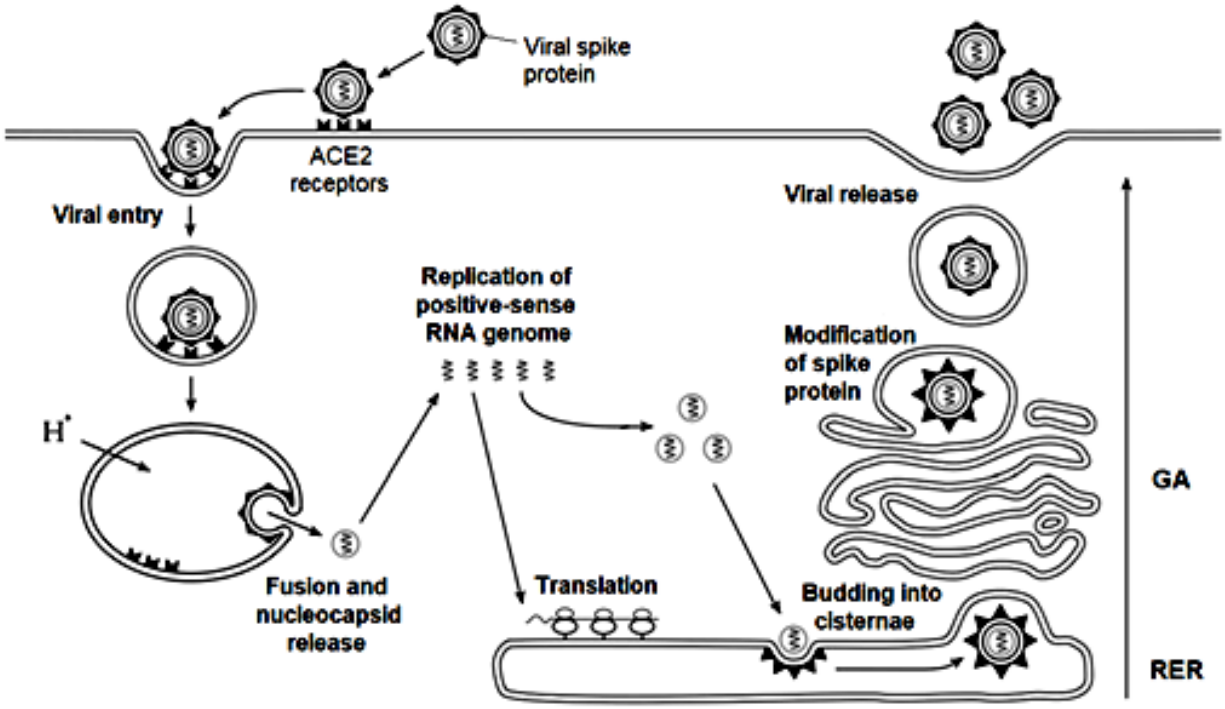

Fig. 5.1

(a) Compare the reproductive cycle of SARS-CoV-2 shown in Fig. 5.1 with that of the influenza virus.

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..... [4]

(b) Many different strains of SARS-CoV-2 have been detected since the start of the pandemic, a feature that is shared with the influenza virus.

Explain how new strains of the influenza virus may arise.

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..... [3]

The polymerase chain reaction (PCR) test has been considered the most accurate test for diagnosing an infection by SARS-CoV-2.

To conduct the PCR test, viral RNA is first extracted from the patient's biological sample, then reverse transcribed to form complementary DNA (cDNA). The cDNA is then amplified through PCR, and the presence of significant quantities of amplified DNA indicates a positive test result.

(c) Explain the basis for the high accuracy of the PCR test.

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..... [2]

Another method used for diagnosis is the antigen rapid test (ART), which can be done without the need for analysis in a laboratory and can yield results within minutes.

Scientists evaluated the effectiveness of ART in diagnosing COVID-19 in 500 patients, where the presence of SARS-CoV-2 in the patients were then confirmed by PCR testing. The results are shown in Table 5.1.

Table 5.1

result by ART	result by PCR	
	positive	negative
positive	111	3
negative	9	377

The sensitivity of a diagnostic test is defined as its ability to detect a disease when a patient is confirmed to be infected.

The specificity of a diagnostic test is its ability to correctly indicate the absence of disease when a patient is confirmed as not infected.

Both measures of sensitivity and specificity are calculated as percentages.

(d) Calculate the sensitivity and specificity of the ART method, showing your workings in the space provided.

[2]

[Total: 11]

JPJC

5 Scientists have produced structures known as virosomes, which are used as transport vehicles for cellular delivery of biologically active macromolecules into the cytoplasm of target cells. Biologically active macromolecules are carried in the central area. Virosomes do not cause disease.

Fig. 5.1 is a diagram of a section through a virosome.

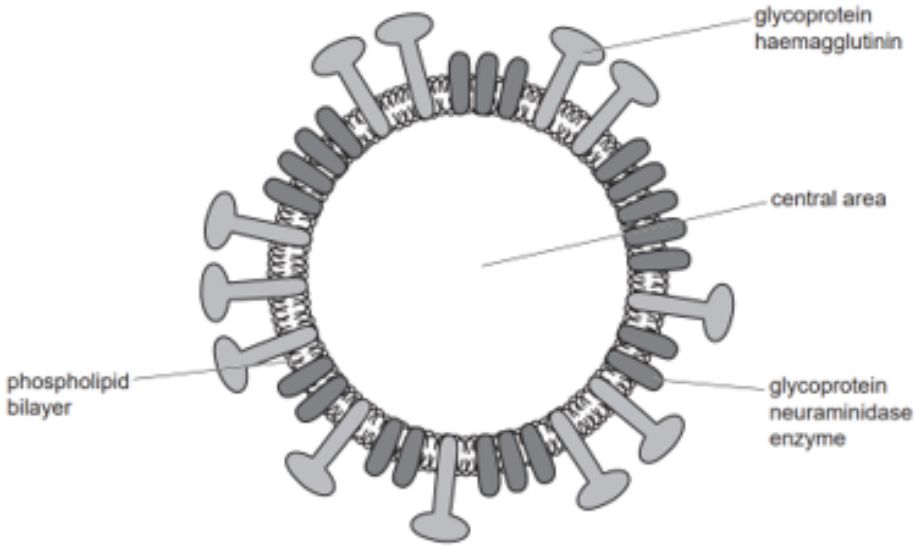

Fig. 5.1

(a) State one difference between the structure of a virosome and an influenza virus.

.....

.....

..... [1]

(b) The glycoproteins haemagglutinin and neuraminidase are found in the influenza virus and in the virosomes. Haemagglutinin binds to a receptor in the cell surface membrane of target cells.

Explain how the virosomes deliver biologically active macromolecules into target cells.

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..... [3]

(c) In influenza virus, neuraminidase removes parts of the host cell receptors that bind to haemagglutinin. This helps newly-formed viruses to leave host cells.

Drugs have been developed to act on neuraminidase. These drugs prevent viruses from leaving host cells.

Suggest and explain how these drugs act to prevent viruses leaving cells.

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..... [3]

(d) The number of cases of influenza is reported to the World Health Organization (WHO) by countries throughout the world so that global data are collected. Fig. 5.2 shows the global data collected between January 2008 and December 2012.

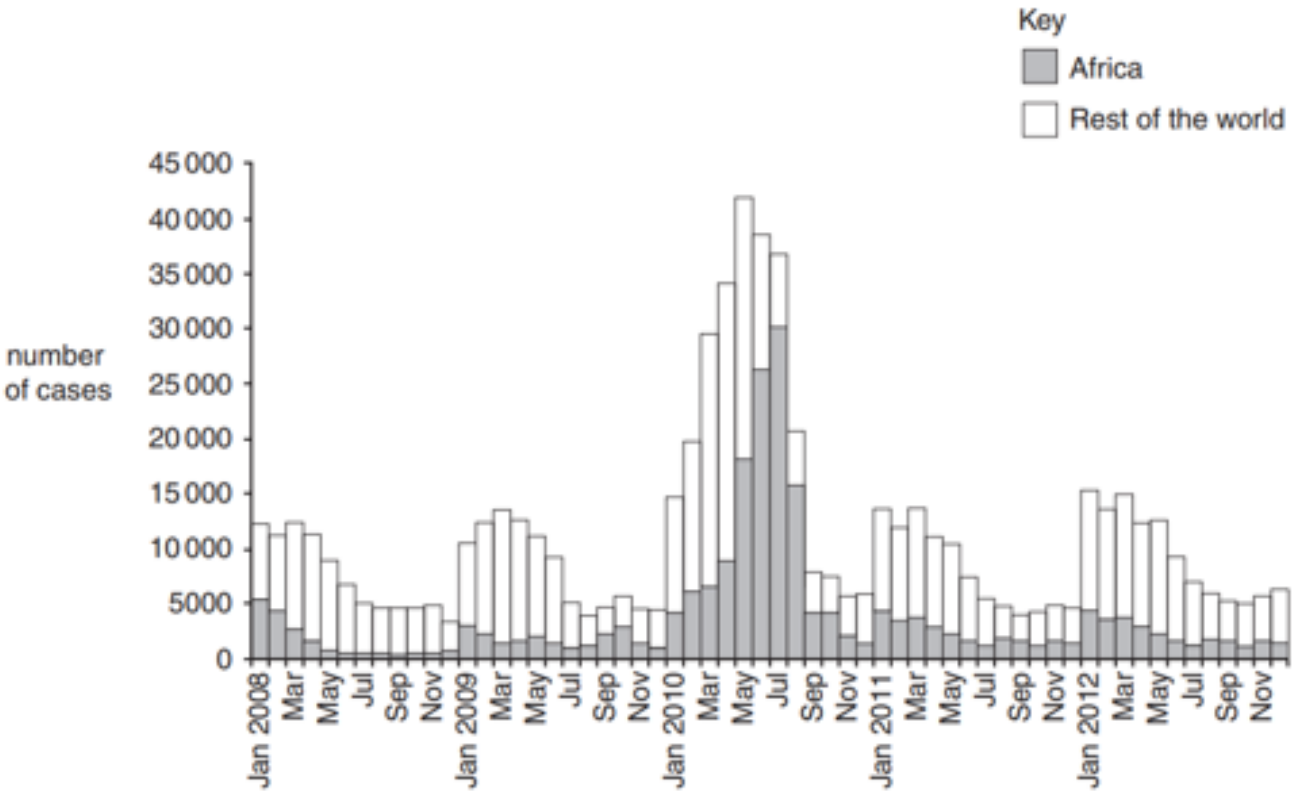

Fig. 5.2

Use the data in Fig. 5.2 to describe the pattern shown in the number of cases of influenza reported to the WHO between January 2008 and December 2012.

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..... [3]

[Total: 10]

Topic 2.10 Molecular Techniques

ASRJC

7 Cystic fibrosis (CF) is a genetic disease caused by mutations of the gene that encodes the cystic fibrosis transmembrane conductance regulator (CFTR).

The most common cystic fibrosis mutation, ΔF508, is a three-base-pair (bp) deletion of codon 508 at exon 10 of the *CFTR* gene.

Another cystic fibrosis mutation is the G551D mutation. This mutation creates a recognition site for the restriction enzyme *MboI* at codon 551. This recognition site is not present in the normal allele.

The *CFTR* gene is amplified via the polymerase chain reaction (PCR) and digested with a restriction enzyme, *MboI*.

(a) Explain the role of primers in the PCR used to amplify the *CFTR* gene.

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.....[2]

(b) Fig. 7.1 shows the results of agarose gel electrophoresis of *MboI* digest products.

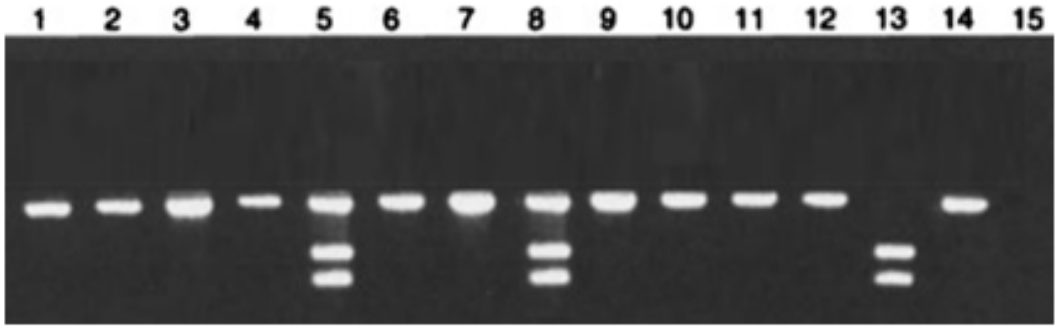

Fig. 7.1

Explain which individual(s) is/are carrier(s) of the disease.

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.....[2]

(c) To detect cystic fibrosis in individuals, a modified Southern hybridisation technique is used.

Target DNA is amplified using PCR and then denatured and labelled with biotin and streptavidin-alkaline phosphatase.

The amplified target DNA is then allowed to hybridise with a specific probe that is attached to a nitrocellulose membrane strip. When a chromogen is added, it is converted to a purple precipitate by alkaline phosphatase.

If the allele is present, it will hybridise with the respective probe. This leads to the conversion of the chromogen to a purple precipitate by alkaline phosphatase. This will then show up as a band on the strip.

Fig. 7.2 shows the PCR products of seven individuals (in lanes A – G) which are tested with four different probes. The following probes are used for the respective alleles:

probes	alleles to detect
W.ΔF508	ΔF508 wild-type allele
M.ΔF508	ΔF508 mutant allele
W.G551D	G551D wild-type allele
M.G551D	G551D mutant allele

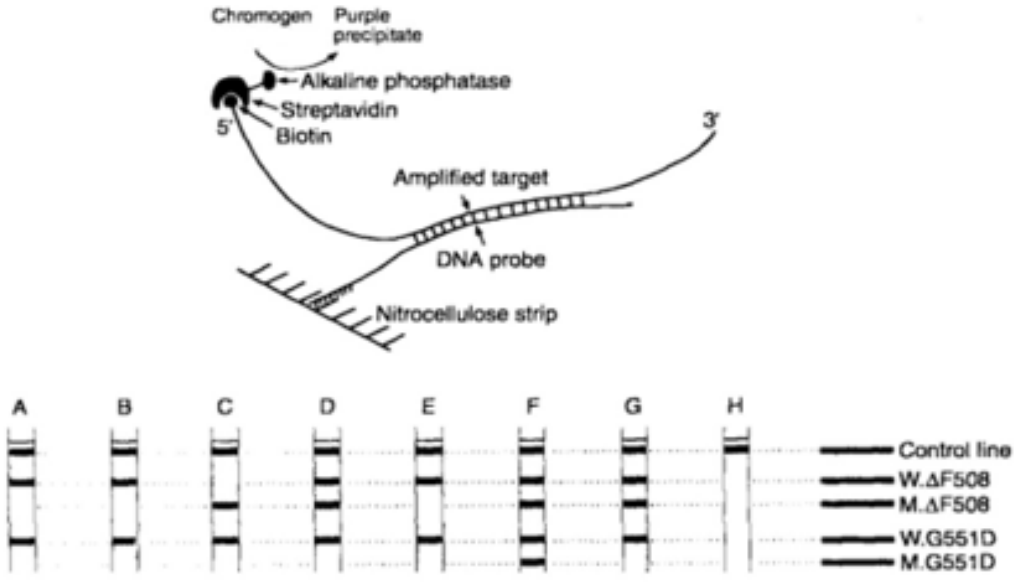

Fig. 7.2

With reference to the information provided in Fig. 7.2,

(i) describe the characteristics of the probes used.

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..... [2]

(ii) explain which of the individual(s) is/are affected by the disease.

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.....[2]

(iii) explain the purpose of H.

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.....[1]

[Total: 9]

CJC

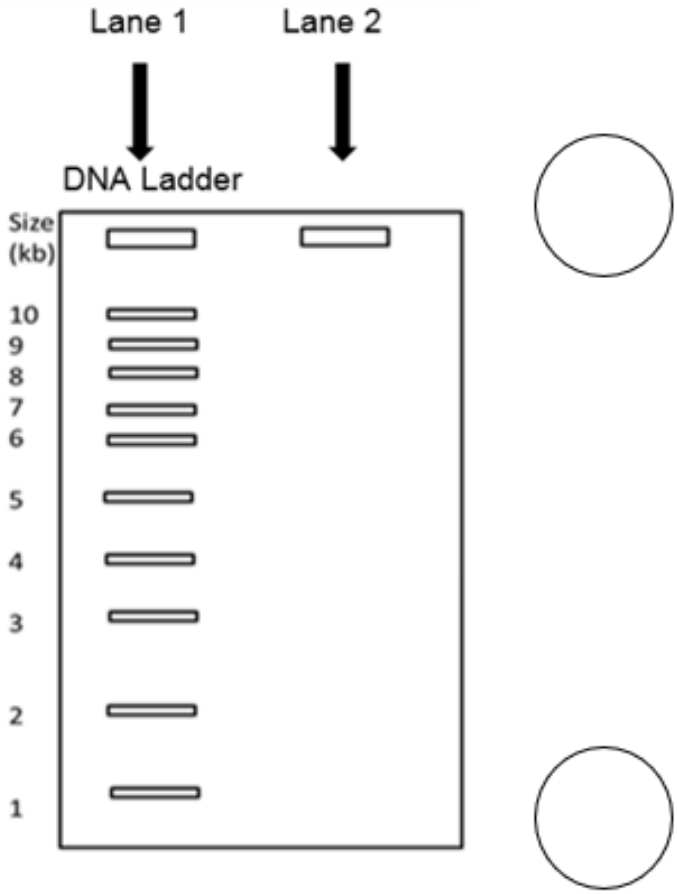
5 **(a)** Fig. 5.1 below shows a 18kb piece of DNA with positions of the restriction sites for restriction enzyme ***HaeIII***.

Fig 5.1

A restriction digestion using ***HaeIII*** enzyme was performed, followed by gel electrophoresis.

A DNA ladder, which is a set of DNA fragments of known lengths, is used to estimate the size of unknown DNA molecules, in lane 1.

On the diagram below,



(i) label the electrode / terminus with "**+**" or "– " in the circles provided. [1]

(ii) show the band pattern of the sample after the restriction digestion and gel electrophoresis in **lane 2**. [2]

(b) Explain how the separated DNA bands in the agarose gel can be visualised.

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..... [2]

(c) Sometimes, DNA bands do not appear as distinct bands but as a smear instead.

Fig. 5.2. shows an example of the smear.

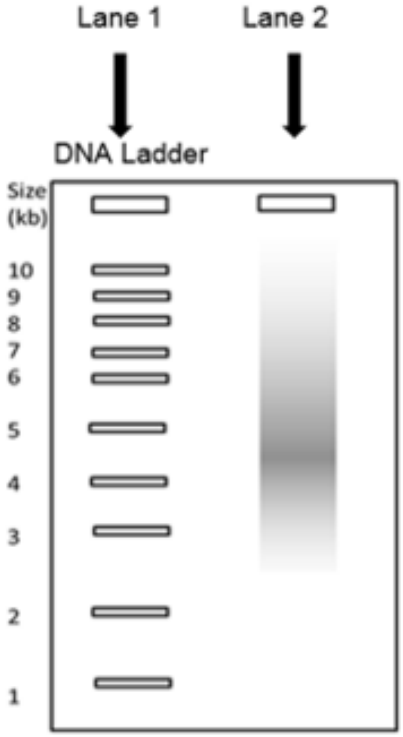

Fig 5.2

Suggest how the smear in Fig. 5.2 is formed.

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..... [2]

(d) Polymerase Chain Reaction (PCR) is a molecular technique commonly performed before agarose gel electrophoresis.

Discuss the advantage and limitation of PCR.

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..... [2]

[Total: 9]

JPJC

6 The polymerase chain reaction (PCR) is used to produce large amounts of DNA from a very small original sample. The main stages of a PCR are shown in Fig. 6.1.

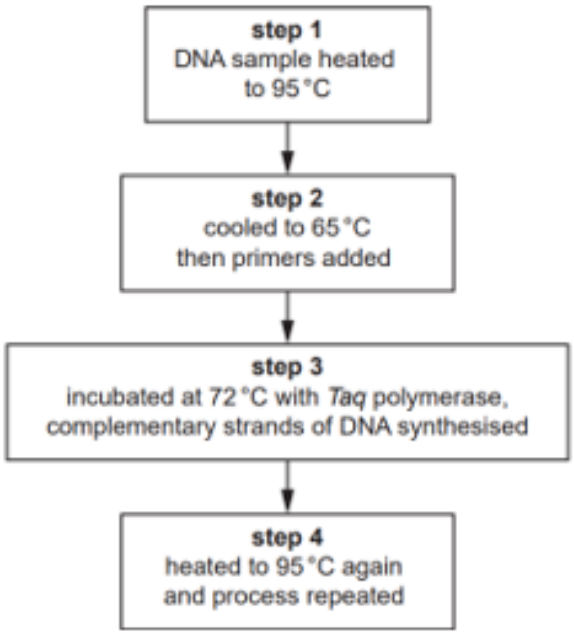

Fig. 6.1

(a) **(i)** Explain why the DNA sample is heated to 95 °C in **step 1**.

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..... [2]

(ii) Explain why primers are added in **step 2**.

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..... [2]

(iii) Explain why the enzyme *Taq* polymerase is used in **step 3**.

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..... [2]

(b) After an organism dies, its DNA gradually breaks down. However, cells in bones that were buried hundreds of years ago may still yield small amounts of DNA that can be extracted, amplified using PCR and then analysed. Mitochondrial DNA (mtDNA) is often used because there are usually more than 100 copies of it in one cell, compared with only two copies of nuclear DNA.

For example, in 1994, mtDNA from bones that had been found in a grave in Russia was analysed to confirm that these were the remains of the royal family, who were known to have been killed in 1918. The mtDNA extracted from the bones was compared with the mtDNA from a living relative of the family.

The family tree of the Russian royal family and some of their relatives is shown in Fig. 6.2.

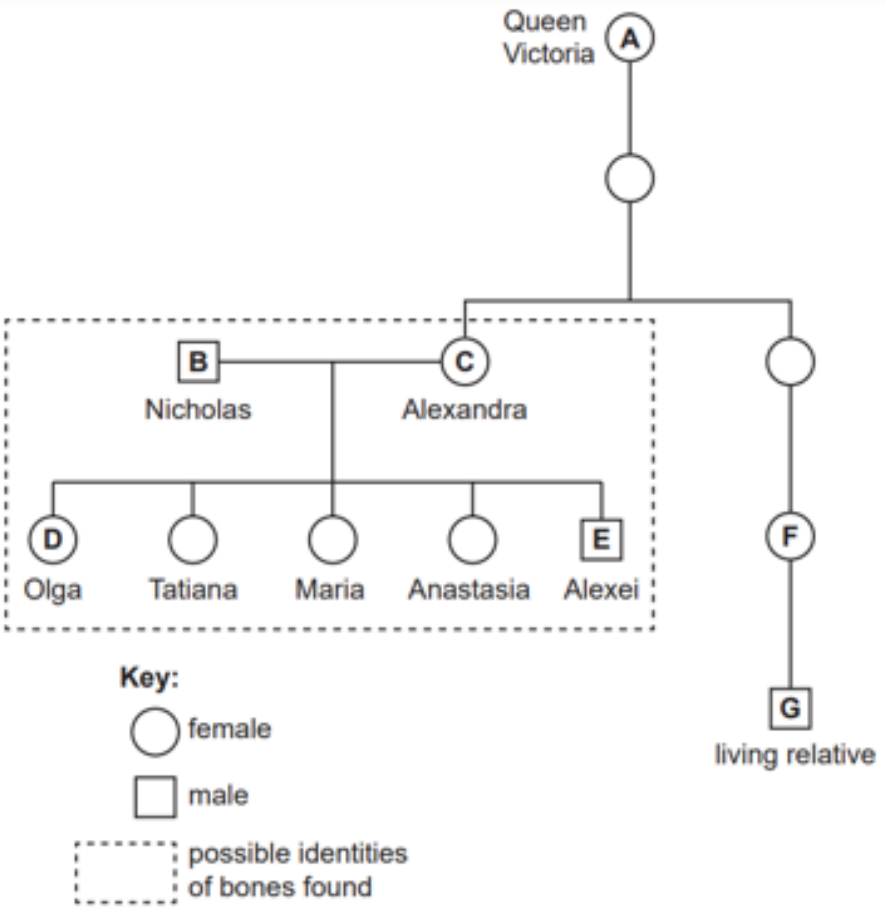

Fig. 6.2

(i) Explain why there are usually more than 100 copies of mtDNA in a cell, but only two copies of nuclear DNA.

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..... [2]

(ii) All of the mitochondria in a zygote come from the egg, not the sperm.

List the **letters** of the people in the family tree in Fig. 6.2 who would be expected to have mtDNA identical to the mtDNA of the living relative, G.

..... [1]

(c) Despite the widespread application of PCR, the technology still has some limitations. Outline one limitation of PCR.

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..... [1]

[Total: 10]

MI

5. Polymerase chain reaction (PCR) is a molecular technique commonly used in molecular biology.

(a) Describe the aim of PCR.

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..... [1]

(b) Describe what occurs in the

(i) first stage,

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..... [2]

(ii) second stage,

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..... [2]

(iii) third stage of PCR.

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..... [2]

(c) State the number of DNA molecules formed after 100 cycles of PCR.

..... [1]

In the set-up of PCR, all the required components are placed in a machine called thermocycler. The enzyme responsible for PCR has a half-life of around 40 minutes at 95°C.

(d) With reference to your knowledge of PCR, explain why a half-life of 40 minutes at 95°C allow many cycles of PCR before the enzyme needs to be replaced.

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..... [2]

[Total :10]

Topic 2.11 Genetics of Bacteria

ASRJC

5 The *lac* operon is a segment of DNA on the chromosome of *Escherichia coli*. The structural genes of the *lac* operon are only fully expressed when the bacteria are exposed to high lactose concentrations.

Fig. 5.1 is a diagram showing the *lac* operon and a nearby region of the *E. coli* genome.

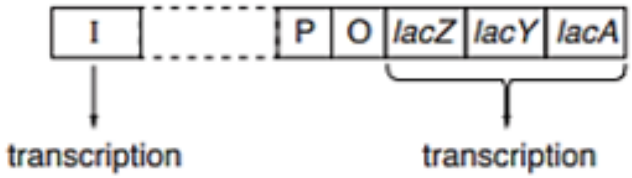

Fig. 5.1

(a) Describe a difference in control between the *lac* operon and the *trp* operon.

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.....[1]

(b) In an experiment, β-galactosidase concentration was measured in the presence of different concentrations of lactose and glucose. The results are shown in Fig. 5.2.

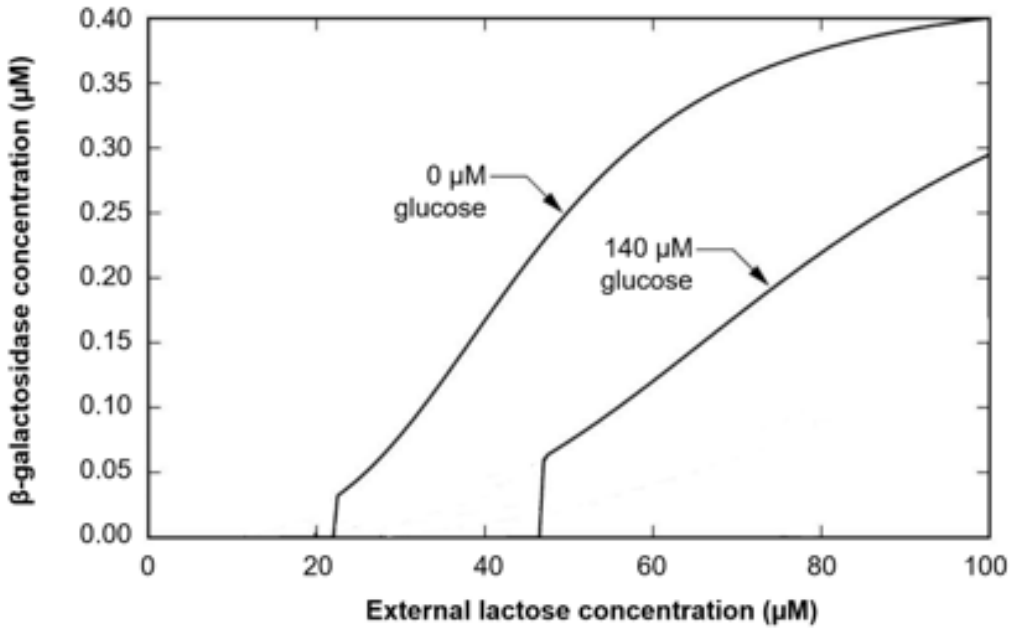

Fig. 5.2

With reference to Fig. 5.2, explain the effect of increasing glucose concentration on the *lac* operon.

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.....[4]

(c) A student claimed that if a mutant *E. coli* strain had a mutated *lacZ* gene where one nucleotide was added, no functional β-galactosidase and permease would be produced.

Discuss the validity of his claim.

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.....[4]

[Total: 9]

CJC

7 Arabinose operon (*ara* operon) is an example of operon in *Escherichia coli*.

This operon has three structural genes, *araB*, *araA* and *araD*, which encode for enzymes needed for the catabolism of arabinose, a five-carbon sugar that can be used by *E. coli* as an alternative carbon source. These genes are not normally expressed in *E. coli*. However, when arabinose is present in the environment, the three Ara enzymes are produced.

(a) Based on the information above, state if the *ara* operon is an inducible or repressible system.

..... [1]

Fig. 7.1 shows the structure of the *ara* operon as well as its regulatory genes.

The promoter site is situated within *araI* and the CAP site (CAP-binding site) is the site where cAMP-CAP binds.

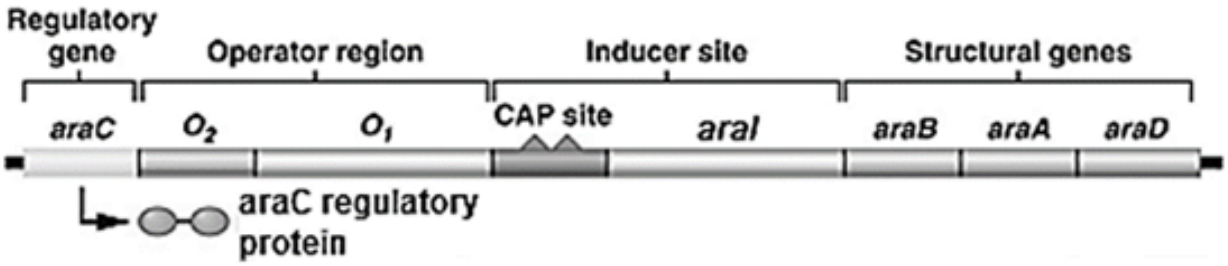

Fig. 7.1

(b) Compare the structure of the *ara* operon and *lac* operon.

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..... [4]

The *ara* operon has both positive and negative regulation, being activated in the presence of arabinose. This is illustrated in Fig. 7.2 below.

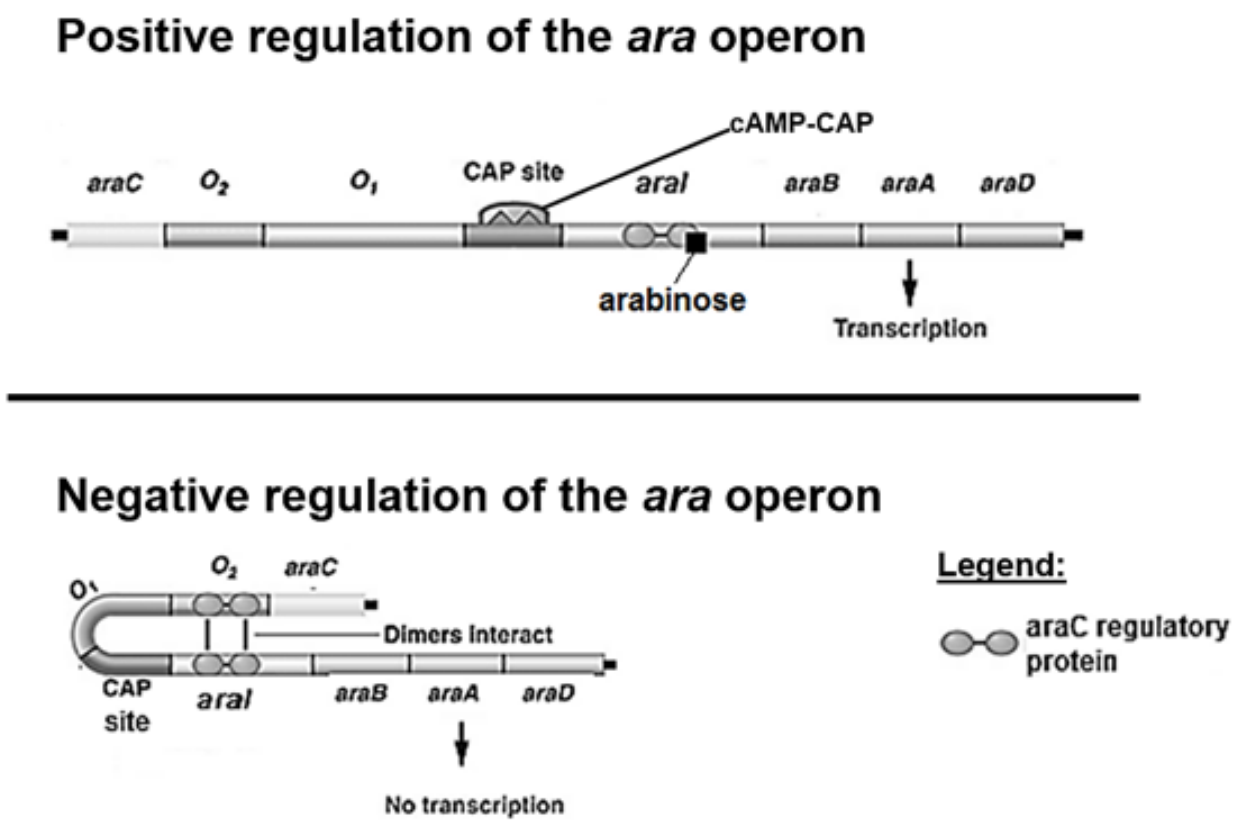

Fig. 7.2

With reference to Fig. 7.2,

(c) Describe and explain the **positive** regulation of the *ara* operon.

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..... [3]

(d) Contrast the **negative** regulation of the *ara* operon with that of the *lac* operon.

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..... [2]

(e) A deletion occurred at the beginning of the *araA* gene.
Predict the effect of this deletion on the gene product of the downstream structural gene, *araD*.
Explain your answer.

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..... [2]

[Total: 12]

CJC

8 Fig. 8.1 shows a classic experiment used to show that physical contact between bacterial cell is necessary for conjugation to happen.

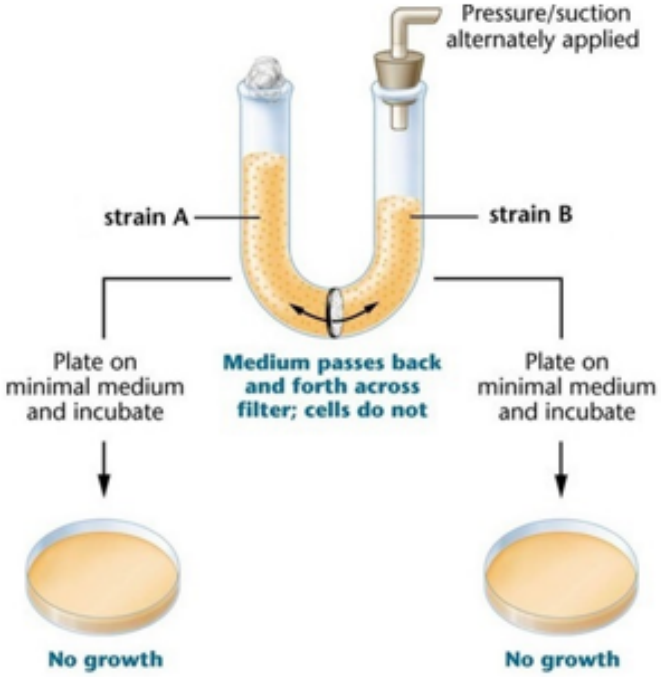

Fig. 8.1

A student tried to replicate the experiment but did not get the result shown in Fig. 8.1.

Instead, he observed a few bacterial colonies which are hybrids of strains A and B. Afterwards, he realised that he forgot to add in DNase when carrying out the experiment.

(a) Briefly describe the role of DNase in this experiment.

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..... [1]

(b) How does the lack of DNase in the experiment result in the growth of the hybrid bacterial colonies?

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..... [2]

The student above repeated the experiment, this time with DNase added. However, he continued to observe a few bacterial colonies which are hybrids of strains A and B.

The student confirmed that the filter was undamaged and there was no cross contamination between bacterial strain A and B.

(c) Suggest how bacterial colonies which are hybrids of strains A and B could have been obtained in the repeated experiment. Explain your answer.

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..... [2]

Bacteria reproduce asexually via binary fission to produce genetically identical daughter bacterial cells. Nevertheless, binary fission is considered one of the processes contributing to the extensive genetic variation in bacteria.

(d) Explain how binary fission contributes to the extensive genetic variation in bacteria.

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..... [2]

[Total: 7]

MI

6. Prokaryote reproduces via asexual process, producing clones of daughter cells.

Despite this, prokaryotes exhibit a wide range of adaptations, suggesting the presence of genetic variation in prokaryotes. Genetic recombination and random mutation can lead to the variation in prokaryotic genome. However, the probability of random mutation occurring in the population is relatively low.

(a) State why random mutation may still lead to genetic variation in prokaryote despite its low rate.

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..... [1]

Genetic recombination is the combination of DNA from two sources into the genome of an individual. In prokaryotes, transformation is one such processes.

(b) Describe how transformation leads to genetic variation in prokaryotic genome.

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..... [2]

Transformation is exploited in the laboratory to make copies of eukaryotic genes in large amount. Fig. 6.1 shows how insulin gene is inserted into bacterial plasmid.

In Fig. 6.2, the resulting plasmid is added to bacteria cell that is treated with calcium chloride ($CaCl_2$). Usually, the bacterial cell is also subjected to heat shock to facilitate the uptake of plasmid DNA. Bacteria cells that have successfully taken up the plasmid DNA is known as transformed bacterial cells. The transformed bacterial cells are plated on agar plate with suitable growth medium, allowing scientists to identify cells that express the insulin gene.

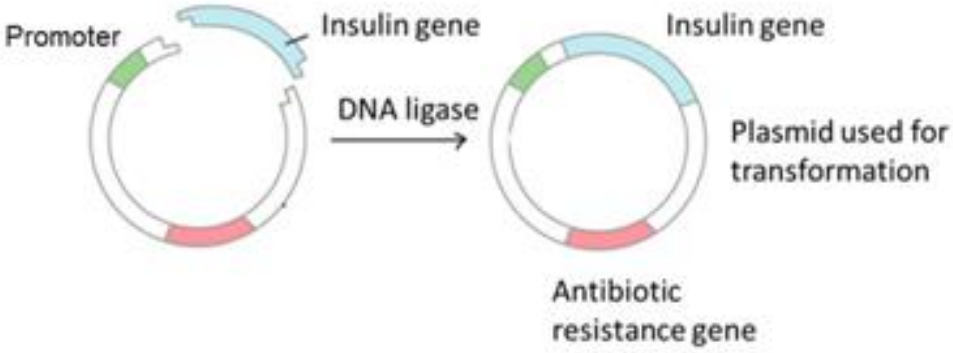

Fig. 6.1

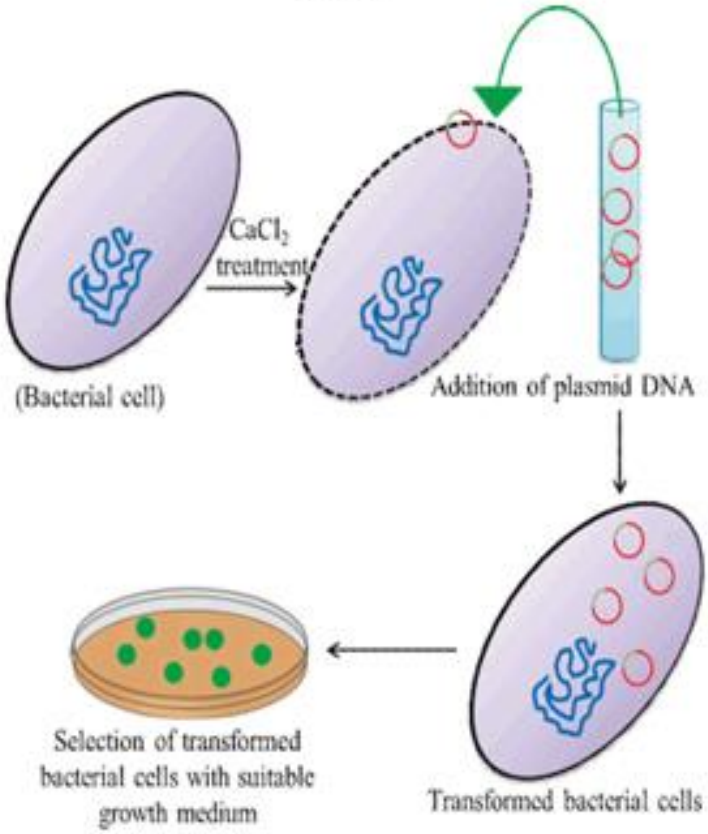

Fig. 6.2

(c) With reference to all the information provided and your knowledge of molecular biology,

(i) explain why insulin gene obtained from eukaryote can be added into plasmid DNA.

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..... [2]

(ii) explain the importance of growing bacterial cells on agar plate with suitable growth medium.

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..... [2]

(iii) suggest how calcium chloride and heat shock can facilitate the uptake of plasmid DNA into the bacterial cell.

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..... [2]

After selection of transformed bacterial cell, DNA analysis is conducted to check if plasmid in bacteria contains the insulin gene.

(d) Outline how Southern Blot is used to detect the presence of insulin gene.

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..... [4]

[Total :13]

TMJC

6 The *lac* operon is a section of DNA present in the genome of *Escherichia coli*. The structural genes of the *lac* operon are only fully expressed when the bacteria is exposed to high lactose concentrations.

Fig. 6.1 is a diagram showing the *lac* operon and a nearby region of the *E. coli* genome.

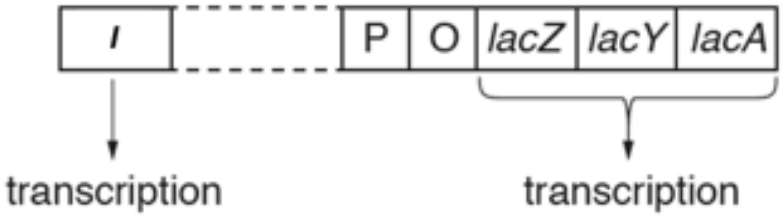

Fig. 6.1

(a) With reference to Fig. 6.1, complete Table 6.1 to identify two structural genes and its products. [2]

Table 6.1

structural gene	name of the gene product

(b) Gene *I* is transcribed all the time to produce its protein. This is known as constitutive expression.

Explain why Gene *I* is constitutively expressed. [3]

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In an investigation into the growth of *E. coli*, a sample of the bacterium was grown in a medium that contained limited concentrations of glucose and lactose. The population size of *E. coli* was measured at regular intervals.

Fig. 6.2 shows the population growth curve obtained for this investigation.

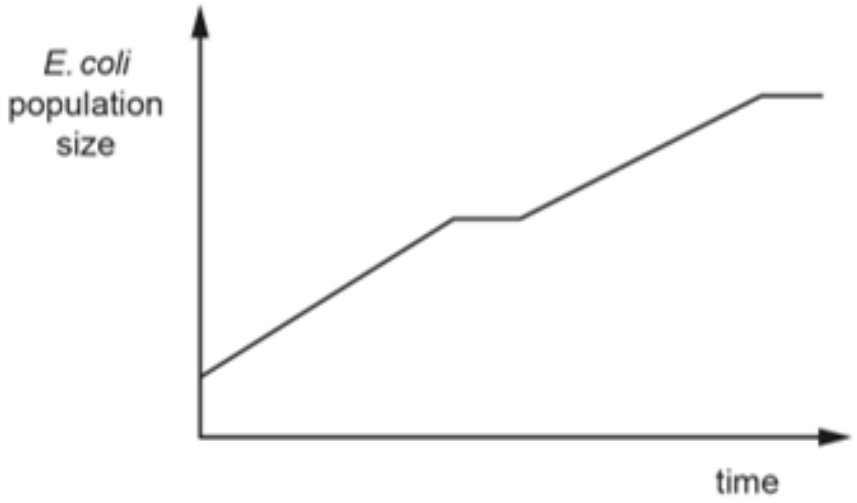

Fig. 6.2

(c) Describe **and** suggest explanations for the population growth curve shown in Fig. 6. [4]

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[Total: 9]

Topic 2.12 Inheritance

ACJC

7 A recombinant frequency of 1% indicates a distance of 1 centimorgan (cM) between two genes on a chromosome. Recombinant frequency can be calculated using the equation:

$$\text{Recombinant frequency} = \frac{\text{Number of recombinant offspring}}{\text{Number of total offspring}} \times 100\%$$

In tomato plants, the two genes controlling the height of plant and the type of leaf are on the same homologous pair of chromosomes. They are located 12 cM apart on chromosome 1. The allele **D**, for a tall plant, is dominant to the allele **d**, for a dwarf plant. The allele **M**, for normal leaves, is dominant to the allele **m**, for mottled leaves.

Pure-breeding tall tomato plants with mottled leaves are crossed with pure-breeding dwarf plants with normal leaves. All the F1 tomato plants are tall with normal leaves. The F1 tomato plants then undergo a test cross, which gives 250 offspring. There are equal numbers between the two parental phenotypes and equal numbers between the recombinant phenotypes.

(a) State the genotype of both plants from the parental generation.

.....

..... [2]

(b) Draw a genetic diagram of the test cross to show the observed results, clearly indicating the numbers of individuals in each different phenotypic class in the offspring.

[5]

Table 7.1 shows the height of 50 tomato plants measured in metres, to the nearest cm.

Table 7.1

1.52	2.05	2.39	2.14	1.84	1.65	1.91	2.34	1.04	2.95
1.72	2.28	2.32	2.00	2.11	1.66	1.74	1.97	2.21	1.43
2.08	1.76	2.68	1.91	2.07	1.85	2.19	2.14	1.99	1.57
2.06	2.45	1.82	1.11	2.68	1.86	2.19	1.56	2.78	1.23
2.83	2.01	2.44	2.04	2.63	1.90	2.21	1.37	2.57	2.54

(c) State and explain the type of genetic variation shown in the height of tomato plants.

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..... [3]

[Total: 10]

ASRJC

6 A plant breeder crossed a plant from a pure-bred line of tomato plants with red fruit with uniform pigmentation with a plant from another pure-bred line of plants with orange-coloured fruit, but with unattractive dark patches. The resulting generation all produced red fruit with dark patches.

Plants from this generation were interbred. The resulting progeny showed the following numbers of plants in each of the three phenotypes:

red fruit with dark patches	98
red fruit with uniform pigmentation	46
orange fruit with dark patches	44

The height of the plants was also measured and the data collected is shown in Table 6.1.

Table 6.1

height / cm	number of tomato plants
131 – 135	3
136 – 140	9
141 – 145	21
146 – 150	12
151 – 155	2

(a) Distinguish between the two types of variation shown in fruit colour and height in the tomato plants.

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..... [2]

(b) The genes involved in the cross above are hypothesised to be completely linked. State the meaning of the term 'complete linkage'.

.....

..... [1]

(c) Using the symbols **R/r** for the gene for colour and **D/d** for the gene for pigmentation, draw a genetic diagram to show how the second cross could lead to the three phenotypes.

[4]

(d) The χ^2 distribution table (Table 6.2) and equation to calculate χ^2 are shown below. Using the formula, the calculated χ^2 value for the cross was 0.38.

$$\chi^2 = \Sigma \frac{(O-E)^2}{E}$$

Table 6.2

degree of freedom	probability, p				
	0.10	0.05	0.02	0.01	0.001
1	2.71	3.84	5.41	6.64	10.83
2	4.61	5.99	7.82	9.21	13.82
3	6.25	7.82	9.84	11.35	16.27
4	7.78	9.49	11.67	13.28	18.47

(i) State the null hypothesis.

.....

.....[1]

(ii) Using the calculated value of χ^2 and Table 6.2, explain what conclusion can be drawn from the data.

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.....[2]

[Total: 10]

CJC

6 Pineapples have three types of leaves: spiny, spiny-tipped and non-spiny.

There are two gene loci involved in the formation of leaves in pineapple, the loci for **A/a** and **B/b** respectively.

The presence of allele **B** results in the formation of spiny-tipped leaves, while the presence of allele **b** results in the formation of spiny leaves.

The presence of allele **A** will prevent the formation of spiny-tipped leaves as well as spiny leaves.

In a particular study, homozygous plants were crossed to produce the F1 generation, which were then allowed to interbreed with the following results:

(a) Complete the table below. [2]

Cross	Phenotypes		
	Parents	F_1 Ratio	F_2 Ratio
1	non-spiny X spiny	All non-spiny	12 non-spiny: 3 spiny-tipped: 1 spiny
2	spiny X spiny		
3	spiny tipped X spiny		

(b) Draw a genetic diagram to explain cross 1. [5]

Fig. 6.1 below shows the inheritance of colour blindness.

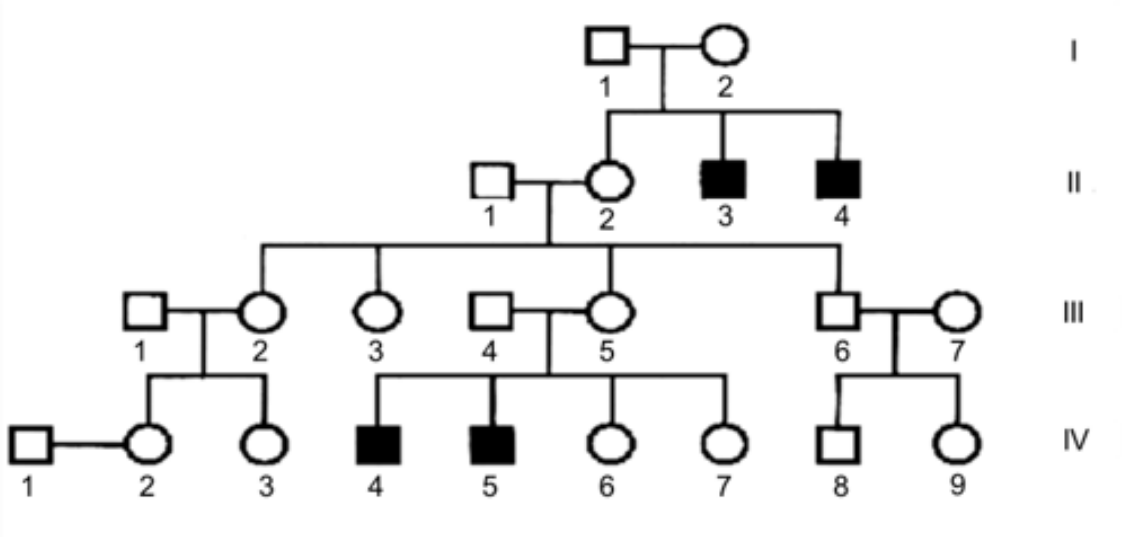

Fig 6.1

With reference to the Fig.6.1,

(c) explain the mode of inheritance of colour blindness.

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..... [2]

(d) Calculate the probability that the offspring of individual **Gen IV-1** and individual **Gen IV-2**, is a carrier. Show your working. [2]

[Total: 11]

JPJC

8 Two unlinked genes control the production of yellow flavone pigment in petals of Dahlia flowers. The petal colour also depends on the degree of hydroxylation of colourless precursor of the flavone pigment.

The dominant allele, **A**, of one gene produces dark yellow pigment due to higher degree of hydroxylation by the gene product. No pigment is produced by the recessive allele, **a**. The dominant allele, **B**, of the second gene produces a light yellow pigment due to lower degree of hydroxylation by the gene product. The recessive allele, **b**, has no hydroxylation effect.

When no yellow pigment is produced the petals are white.

This is an example of dominant epistasis.

(a) Explain the term dominant epistasis in this context.

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..... [3]

(b) Plants with the genotypes **AABB** and **aabb** were crossed and the resulting F1 generation was test-crossed.

Draw a genetic diagram of the test-cross to show the genotypes and phenotypes of the parents and offspring.

State the ratio of phenotypes of the offspring.

ratio of phenotypes .....

[5]

(c) Explain why it would be useful to carry out a chi-squared test on these results. No calculations are required to answer this question.

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..... [2]

[Total: 10]

MI

8. In the sweet pea plant, one gene codes for flower colour and one gene codes for pollen grain shape.

Flower colour is either purple or red, and the allele **F** coding for purple flowers is dominant over the allele **f** coding for red flowers.

Pollen grain shape is either long or round, and the allele **G** coding for long pollen grains is dominant over allele **g** coding for round pollen grains.

(a) A dihybrid cross was carried out between homozygous dominant and homozygous recessive sweet pea plant parents to produce the F1 generation.

The offspring from the F1 generation were crossed to produce the F2 generations. Table 8.1 shows the actual results of the cross.

Table 8.1

Phenotypes of F2 offspring	Number of individuals
purple flowers, long pollen grains	294
purple flowers, round pollen grains	24
red flowers, long pollen grains	25
red flowers, round pollen grains	57

(i) Explain how the results support the fact that this is a non-mendelian inheritance.

.....

.....

.....

..... [2]

(ii) Draw a genetic diagram to show the actual cross between the two offspring from the F1 generation.

[4]

(b) A test cross was carried out with sweet pea plants known to be heterozygous for both flower colour and pollen grain shape.

The results of the test cross are shown in Table 8.2.

Table 8.2

Phenotypes of F2 offspring	Observed number of individuals (O)	Expected number of individuals (E)	(O-E)	$(O-E)^2$	$\frac{(O-E)^2}{E}$
purple flowers, long pollen grains	105				
purple flowers, round pollen grains	15				
red flowers, long pollen grains	16				
red flowers, round pollen grains	64				

(i) Chi-squared test was carried out to investigate if the results in Table 8.2 were significantly different from those expected.

The formula for chi-squared test is shown in Fig. 8.1.

$$X^2 = \Sigma \left\{ \frac{(O-E)^2}{E} \right\}$$

Fig. 8.1

Complete Table 8.2 and calculate the value of χ^2. Show your working clearly in the space below.

Value of χ^2 : ..... [4]

(ii) The results of a test cross can be used to determine a crossover value (COV). A crossover value is the percentage of the total number of offspring showing recombination.

The COV can be calculated using the formula below.

$$\text{COV} = \frac{\text{number of recombinants}}{\text{total number of individuals}} \; x \; 100$$

Calculate the COV from the **observed results** shown in Table 8.2. Show your working clearly in the space below.

COV : .....% [2]
[Total :12]

SAJC

QUESTION 5

Chronic Granulomatous Disease (CGD) is characterized by defects in the enzyme NADPH oxidase, causing phagocytes (for examples, neutrophils) to improperly clear invading pathogens.

X-CGD is the most common type of CGD and primarily affects males, with less females getting the disease. It is caused by a mutation in a gene on the sex chromosome.

(a) With reference to the information provided above, state the mode of inheritance of CGD.

Mode of inheritance: ..... [1]

(b) Explain your answer for (a).

.....

.....

.....

.....

.....[2]

(c) A woman not affected by CGD marries a man not affected with CGD. Their daughters are all not affected, while they have some sons who are affected, and some sons who are not.

Using the symbols A/a, construct a genetic cross diagram to show the outcomes.

[4]

(d) Use the genetic cross diagram in (c) to find the probability of the couple getting a son with CGD.

.....[1]

Further research showed that the gene mutation causing CGD is a substitution mutation.

Fig. 5.1 shows a pedigree tree to show the inheritance of CGD, and Fig. 5.2 shows the outcomes of gel electrophoresis after isolation of the NADPH oxidase gene from each individual. The bands are made visible with the staining of ethidium bromide.

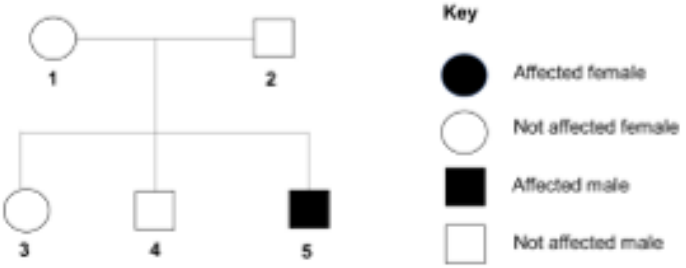

Fig. 5.1

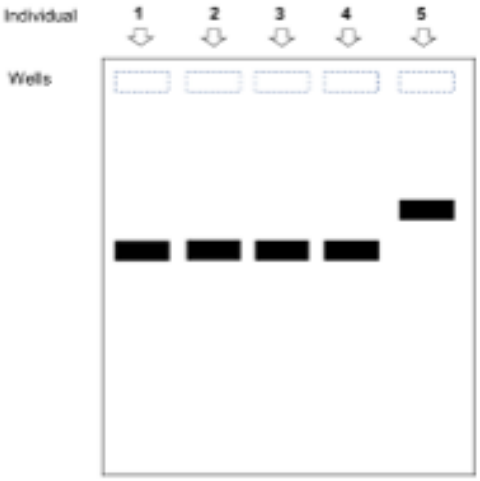

Fig. 5.2

e) With reference to Fig. 5.1 and Fig. 5.2, identify one error in the gel electrophoresis outcomes shown in Fig. 5.2. Explain your answer.

Error:

.....

Explanation:

.....

.....

[2]

Researchers are trying a new treatment for CGD, where they removed stem cells which are capable of differentiating into blood cells from the patients themselves, and genetically modified them so that they no longer carried the unwanted mutation. Then, the edited stem cells were returned to their bodies, ready to produce healthy new lymphocytes such as B lymphocytes and T lymphocytes.

(f) State the name of the stem cells which are capable of differentiating into blood cells and list two properties of these stem cells.

Name of stem cell: .....[1]

Properties: .....

.....

.....

.....[2]

(g) The new treatment mentioned above is able to overcome ethical concerns on obtaining stem cells from blastocysts derived from oocytes (eggs). Elaborate on **one** such ethical concerns.

.....

.....[1]

[Total:14]

TMJC

5 A piebald fur dog is one that has white spotted pattern and is often associated with deafness.

Pure bred brown fur dogs with normal hearing were crossed with pure bred piebald, deaf dogs. All F1 generation were brown fur dogs with normal hearing. The F1 generation were then inbred and gave offspring with the following observed numbers and phenotypes as shown in Table 5.1 below.

(a) In Table 5.1,

- indicate the expected number of progeny of the F2 phenotypes if the genes assorted independently.
- complete the rest of Table 5.1 to calculate the chi-squared value. [4]

Table 5.1

Phenotypes	Number of F2 progeny		$(O-E)^2$	$\frac{(O-E)^2}{E}$ (2 d.p.)
	Observed (O)	Expected (E)		
Brown fur, normal hearing	30			
Piebald fur, deaf	24			
Brown fur, deaf	7			
Piebald fur, normal hearing	3			

$$\chi^2 = \sum \frac{(O-E)^2}{E}$$

=

(b) The critical chi-squared value for these results at a probability of $p = 0.05$ is 7.81.

State what can be concluded about the inheritance from the chi-squared value you have calculated in **(a)**. [3]

.....

.....

.....

.....

.....

.....

.....

(c) Explain the difference between the expected and observed number in the F2 progeny. [4]

.....

.....

.....

.....

.....

.....

.....

.....

.....

(d) Draw a genetic diagram to show the results of a cross between the F1 generation.

Use the symbols **B** and **b** to represent alleles for fur colour and **D** and **d** to represent the alleles for hearing. [4]

[Total: 15]