

Q1.(a) Explain how the structure of DNA is related to its functions.

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Scientists investigated three genes, **C**, **D** and **E**, involved in controlling cell division.

They studied the effect of mutations in these genes on the risk of developing lung cancer.

The scientists analysed genes **C**, **D** and **E** from healthy people and people with lung cancer.

- If a person had a normal allele for a gene, they used the symbol N.
- If a person had two mutant alleles for a gene, they used the symbol M.

They used their data to calculate the risk of developing lung cancer for people with different combinations of N and M alleles of the genes. A risk value of 1.00 indicates no increased risk. The following table shows the scientists' results.

Gene C	Gene D	Gene E	Risk of developing lung cancer
N	N	N	1.00
M	N	N	1.30
N	N	M	1.78
N	M	N	1.45

N = at least one copy of the normal allele is present

M = two copies of the mutant allele are present

- (b) What do these data suggest about the relative importance of the mutant alleles of genes **C**, **D** and **E** on **increasing** the risk of developing lung cancer? Explain your answer.

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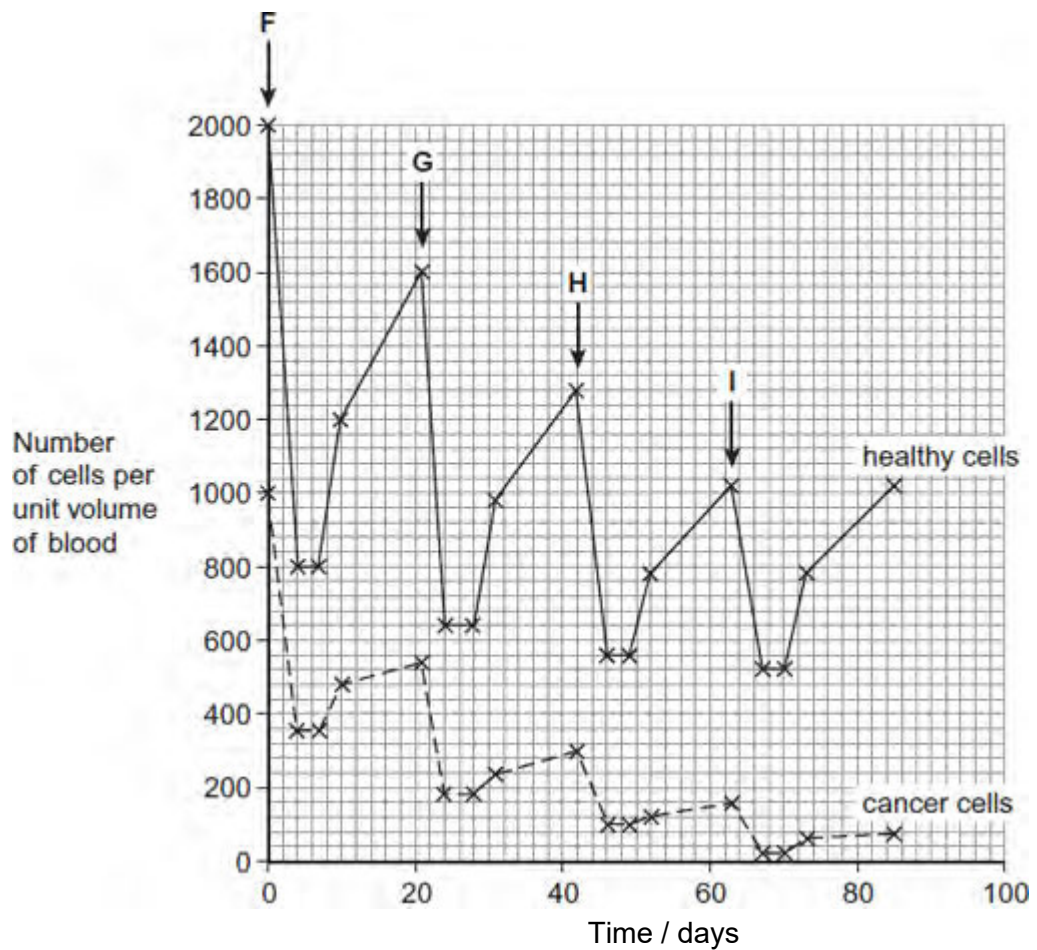
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Chemotherapy is the use of a drug to treat cancer. The drug kills dividing cells. The figure below shows the number of healthy cells and cancer cells in the blood of a patient receiving chemotherapy. The arrows labelled **F** to **I** show when the drug was given to the patient.



- (c) Calculate the rate at which healthy cells were killed between days 42 and 46.

..... cells killed per unit volume of blood per day

(1)

- (d) Describe similarities and differences in the response of healthy cells and cancer cells to the drug between times **F** and **G**.

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- (e) More cancer cells could be destroyed if the drug was given more frequently.
Suggest why the drug was **not** given more frequently.

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(Total 15 marks)

Q2. The Amish are a group of people who live in America. This group was founded by 30 Swiss people, who moved to America many years ago. The Amish do not usually marry people from outside their own group.

One of the 30 Swiss founders had a genetic disorder called Ellis-van Creveld syndrome. People with this disorder have heart defects, are short and have extra fingers and toes. Ellis-van Creveld syndrome is caused by a faulty allele.

In America today, about 1 in 200 Amish people are born with Ellis-van Creveld syndrome. This disorder is very rare in people in America who are not Amish.

- (a) In America today, there are approximately 1250 Amish people who have Ellis-van Creveld syndrome. Use the information provided to calculate the current Amish population of America.

Amish population

(1)

- (b) The faulty allele that causes Ellis-van Creveld syndrome is the result of a mutation of a gene called *EVC*. This mutation leads to the production of a protein that has one amino acid missing.

- (i) Suggest how a mutation can lead to the production of a protein that has one amino acid missing.

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- (ii) Suggest how the production of a protein with one amino acid missing may lead to a genetic disorder such as Ellis-van Creveld syndrome.

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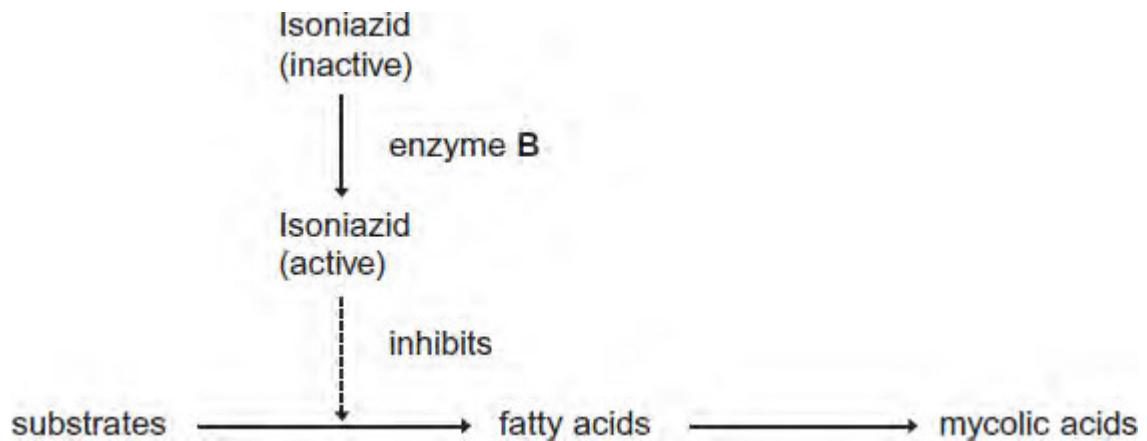
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(Total 5 marks)

Q3. Mycolic acids are substances that form part of the cell wall of the bacterium that causes tuberculosis. Mycolic acids are made from fatty acids. Isoniazid is an antibiotic that is used to treat tuberculosis. The diagram shows how this antibiotic inhibits the production of mycolic acids in this bacterium.



- (a) Treatment with isoniazid leads to the osmotic lysis of this bacterium. Use information in the diagram to suggest how.

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- (b) Human cells also produce fatty acids. Isoniazid does not affect the production of these fatty acids.

Use information in the diagram to suggest **one** reason why isoniazid does **not** affect the production of fatty acids in human cells.

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- (c) A mutation in the gene coding for enzyme **B** could lead to the production of a non-functional enzyme. Explain how.

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(3)
(Total 6 marks)

Q4.Phenylketonuria is a disease caused by mutations of the gene coding for the enzyme PAH. The table shows part of the DNA base sequence coding for PAH. It also shows a mutation of this sequence which leads to the production of non-functioning PAH.

DNA base sequence coding for PAH	C	A	G	T	T	C	G	C	T	A	C	G
DNA base sequence coding for non-functioning PAH	C	A	G	T	T	C	C	C	T	A	C	G

- (a) (i) What is the maximum number of amino acids for which this base sequence could code?

(1)

- (ii) Explain how this mutation leads to the formation of non-functioning PAH.

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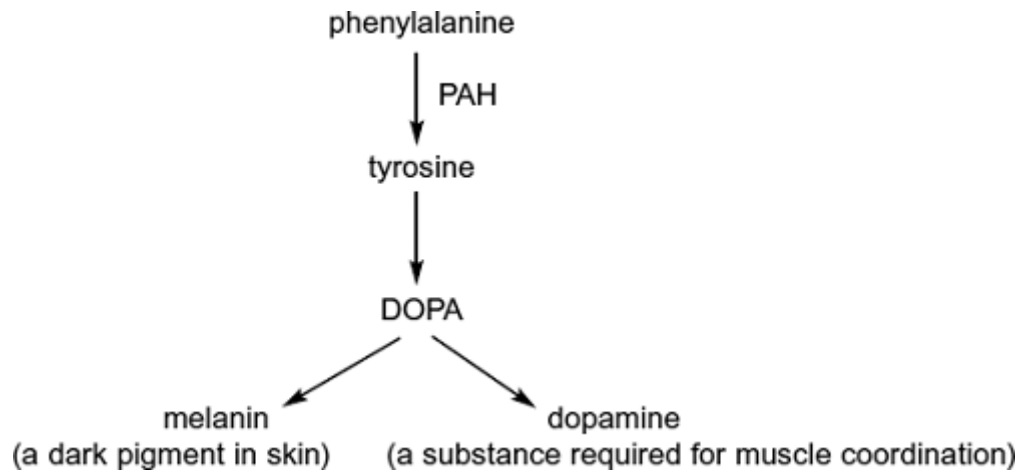
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PAH catalyses a reaction at the start of two enzyme-controlled pathways. The diagram shows these pathways.



- (b) Use the information in the diagram to give **two** symptoms you might expect to be visible in a person who produces non-functioning PAH.

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- (c) One mutation causing phenylketonuria was originally only found in one population in central Asia. It is now found in many different populations across Asia. Suggest how the spread of this mutation may have occurred.

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(1)

Q5. (a) What name is used for the non-coding sections of a gene?

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(1)

Figure 1 shows a DNA base sequence. It also shows the effect of two mutations on this base sequence. **Figure 2** shows DNA triplets that code for different amino acids.

Figure 1

Original DNA base sequence	A	T	T	G	G	C	G	T	G	T	C	T
Amino acid sequence												
Mutation 1 DNA base sequence	A	T	T	G	G	A	G	T	G	T	C	T
Mutation 2 DNA base sequence	A	T	T	G	G	C	C	T	G	T	C	T

Figure 2

DNA triplets	Amino acid
GGT, GGC, GGA, GGG	Gly
GTT, GTA, GTG, GTC	Val
ATC, ATT, ATA	Ile
TCC, TCT, TCA, TCG	Ser
CTC, CTT, CTA, CTG	Leu

(b) Complete **Figure 1** to show the sequence of amino acids coded for by the original DNA base sequence.

(1)

(c) Some gene mutations affect the amino acid sequence. Some mutations do not.

Use the information from **Figure 1** and **Figure 2** to explain

- (i) whether mutation **1** affects the amino acid sequence

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- (ii) how mutation **2** could lead to the formation of a non-functional enzyme.

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- (d) Gene mutations occur spontaneously.

- (i) During which part of the cell cycle are gene mutations most likely to occur?

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- (ii) Suggest an explanation for your answer.

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(1)

(Total 9 marks)

