AQA A-LEVEL · 7402

BIOLOGY



EXAM QUESTION PACK

3.8.1 GENE MUTATION

SCORE	PERCENTAGE
/130	%

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Q1.

(a) There are different types of gene mutation.

Put a tick (✓) in the box next to the statement which describes **incorrectly** the effect of the mutation in an exon of a gene.

A substitution may not result in a change to the encoded amino acid.	☐
An inversion will result in a change in the number of DNA bases.	☐
A deletion will result in a frame shift.	☐
An addition will result in a frame shift.	☐

(1)

(b) Describe how alterations to tumour suppressor genes can lead to the development of tumours.

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(3)

(c) A type of malignant tumour cell divides every 8 hours.

Starting with one of these cells, how many tumour cells will be present after 4 weeks?
Assume none of these cells will die.

Give your answer in standard form.

Answer = _____

(2)

(Total 6 marks)

Q2.

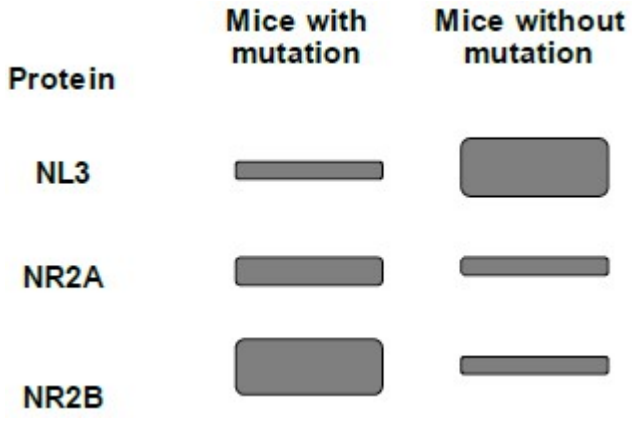
Some autism spectrum disorders (ASDs) are associated with a mutation affecting the neuroligin-3 gene. This gene codes for a protein called NL3, that is found in synapses.

Scientists investigated the effects of a mutation affecting NL3 in mice. They obtained brains from mice with the mutation and from mice without the mutation. For each type of mouse they:

- obtained a solution containing all of the proteins from synapses in one part of the brain
- separated these proteins using gel electrophoresis
- identified and measured the amount of three proteins from the solution using three different labelled antibodies.

The three proteins are parts of a postsynaptic membrane receptor.

The diagram below shows the scientists' results. Each band shows the presence of a protein. The size of a band shows the amount of the protein present.



(a) The mutation affecting NL3 in these mice was a substitution in the neuroligin-3 gene.

What is a substitution mutation?

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(1)

(b) Suggest how gel electrophoresis separated the proteins obtained from the synapses.

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(2)

(c) Each type of labelled antibody binds specifically to one of the proteins.

Explain why.

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(3)

(d) What do these data show about the effects of the mutation on the proteins?

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(2)

(e) These proteins are part of a receptor found in synapses in the part of the brain called the hippocampus. A high ratio of NR2B to NR2A protein in this receptor has been associated with good memory.

Using all of the information, suggest how the mutation affecting the NL3 protein may affect a mouse.

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(2)

(Total 10 marks)

Q3.

Mycobacterium tuberculosis causes tuberculosis. The DNA of *M. tuberculosis* contains a direct repeat (DR) region. The DR region consists of 43 different, non-coding base sequences called spacers. Each spacer is found in a specific place in the DR region. In different strains of *M. tuberculosis*, some of these spacers have been lost.

(a) (i) The DR region consists of non-coding base sequences.

What is meant by a non-coding base sequence?

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(1)

(ii) Name the process by which the base sequence of a spacer is lost from a DR region.

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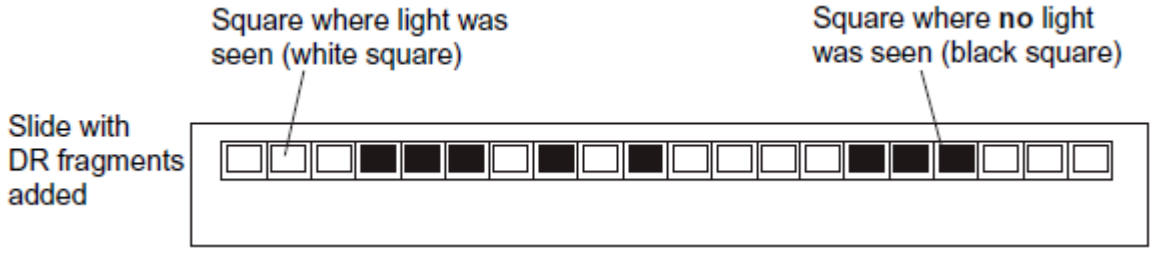
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(1)

Scientists investigated the DR regions of different strains of *M. tuberculosis*. They produced a DNA probe for each of the 43 spacer sequences. Each probe was:

- labelled with a fluorescent marker that gave off light if the probe attached to its complementary spacer
- attached to a particular square on a slide.

They obtained samples of the DR region from each strain. These were cut into small single-stranded DNA fragments. The fragments from each strain were added to a slide with the DNA probes attached. The diagram below shows their results for one strain of *M. tuberculosis* with 20 of the probes.



(b) The scientists cloned the DR region DNA *in vitro* before testing for the presence of spacers.

Give the name of the method they used to clone the DNA *in vitro*.

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(1)

(c) Explain how the use of DNA probes produced the results in the diagram.

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(3)

(d) Doctors can use the method with DNA probes to identify the specific strain of *M. tuberculosis* infecting a patient. This is very important when there is an outbreak of a number of cases of tuberculosis in a city.

Suggest and explain why it is important to be able to identify the specific strain of *M. tuberculosis* infecting a patient.

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(2)

(Total 8 marks)

Q4.

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

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(6)

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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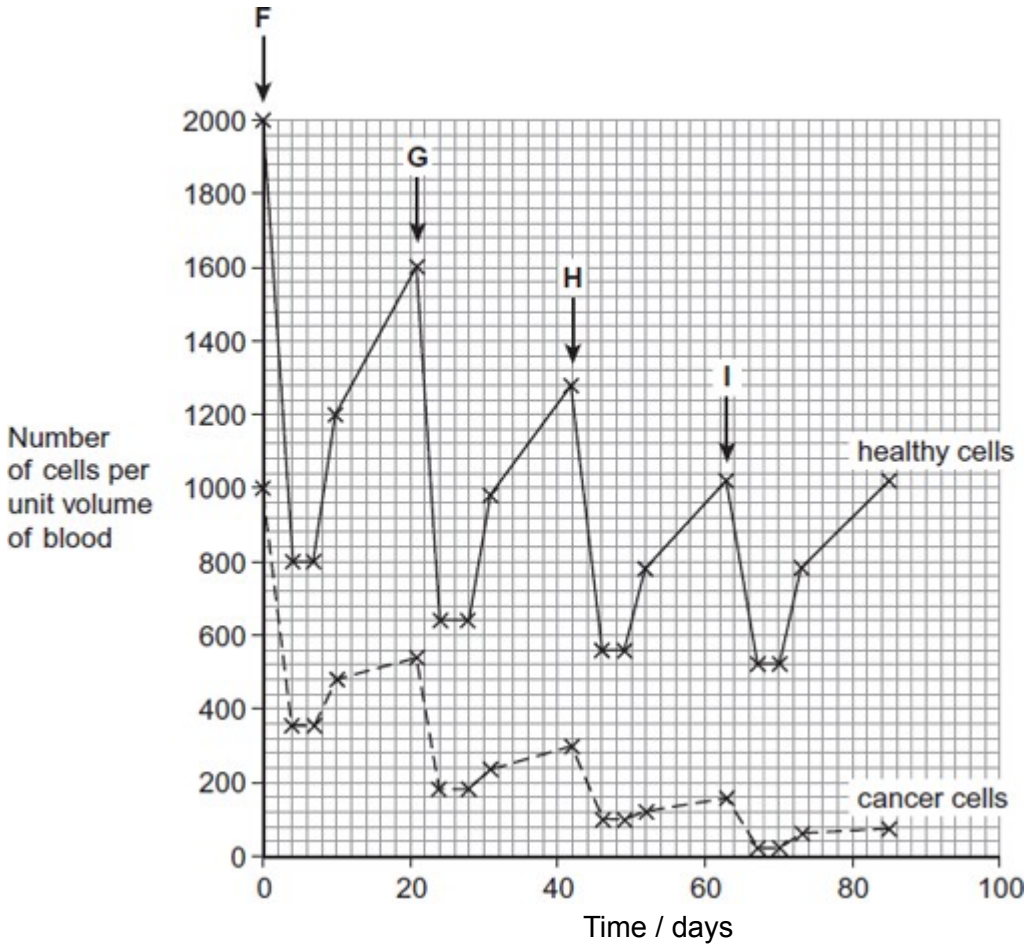
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(3)

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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(3)

(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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(2)

(Total 15 marks)

Q5.

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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(2)

(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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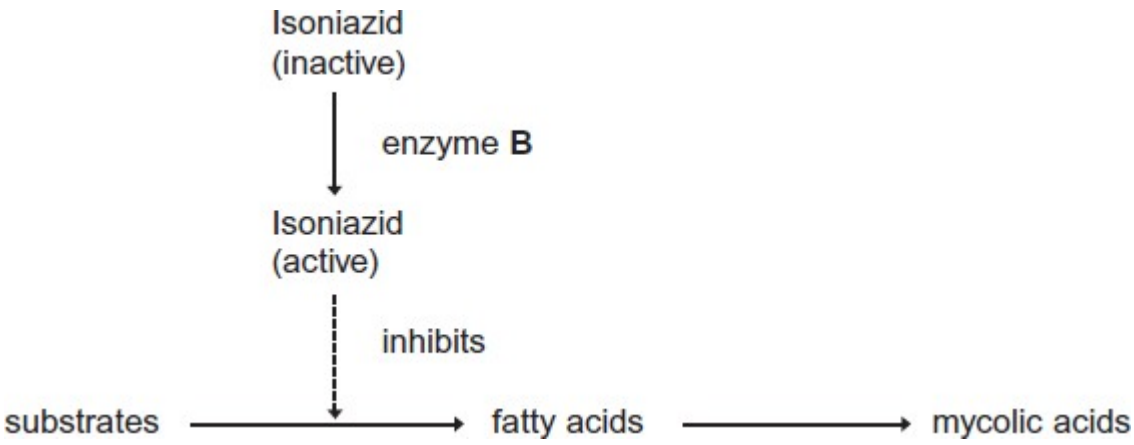
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(2)

(Total 5 marks)

Q6.

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 antibioticthat 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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(2)

(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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(1)

(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)

Q7.

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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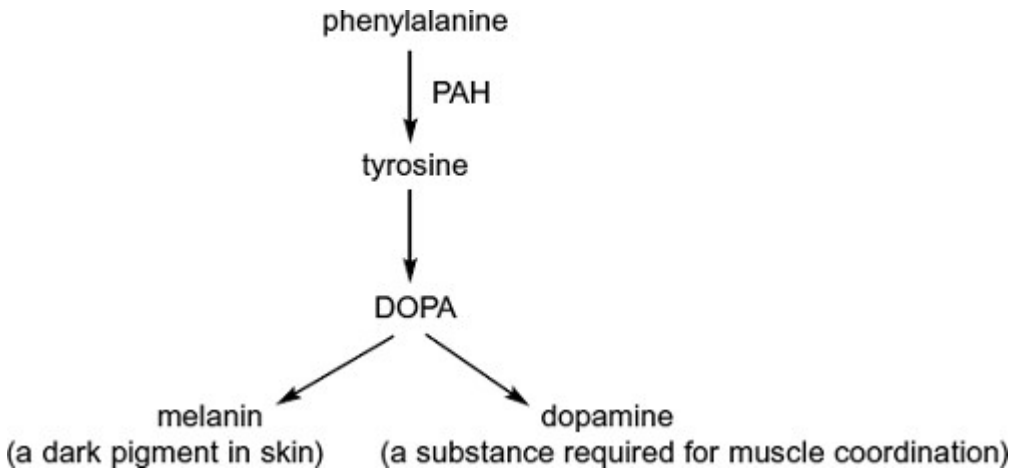
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(3)

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.

1. _____

2. _____

(2)

(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)

(Total 7 marks)

Q8.

Write an essay on using DNA in science and technology.

(Total 25 marks)

Q9.

(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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(2)

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

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(3)

(d) Gene mutations occur spontaneously.

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

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(1)

(ii) Suggest an explanation for your answer.

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(1)

(Total 9 marks)

Q10.

Read the following passage.

Soon a single drop of blood might be enough to reveal, at a very early stage, if a patient has cancer. It could also tell us what type of cancer it is and whether it is treatable. Fragments of DNA from body cells are present in blood plasma. Some of these fragments may be from cancer cells. The fragments can be detected by a new test in which a test strip containing nucleic acid binds to sections of altered DNA.

Other cancer-detecting techniques involve removing a tissue sample from a patient. The tissue sample is used to obtain mRNA. By examining the mRNA, scientists can discover whether cancer is present.

Use information from the passage and your own knowledge to answer the questions.

(a) Describe how altered DNA may lead to cancer.

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(6)

(b) Explain why fragments of DNA from cancer cells may be present in blood plasma (lines 3-4).

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(2)

(c) Explain why the nucleic acid on the test strip will only bind to altered DNA (lines 4-5).

_____

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_____

(2)

(d) This test strip will allow cancers to be detected at a very early stage. Explain why cancer is more likely to be treated successfully if the disease is detected at a very early stage.

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(2)

(e) Explain how examining mRNA (line 7) enables scientists to discover whether cancer is present.

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(3)

(Total 15 marks)

Q11.

Figure 1 shows part of a sarcomere.

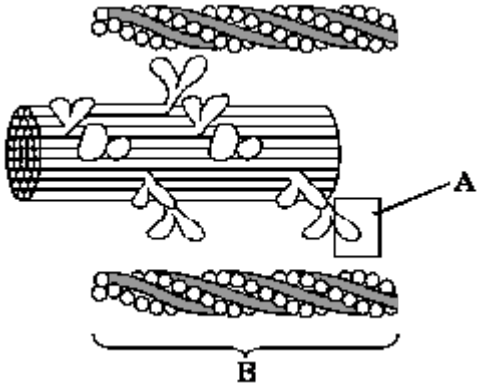


Figure 1

(a) (i) Name the main protein in structure **B**.

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(1)

(ii) Name the structure in box **A**.

_____

(1)

(b) (i) Describe how calcium ions cause the myofibril to start contracting.

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(2)

(ii) Describe the events that occur within a myofibril which enable it to contract.

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(3)

Slow and fast skeletal muscle fibres differ in a number of ways. Slow fibres get their ATP from aerobic respiration while anaerobic respiration provides fast fibres with their ATP. **Figure 2** shows a bundle of fast and slow fibres seen through an optical microscope. The fibres have been stained with a stain that binds to the enzymes which operate in the electron transport chain.

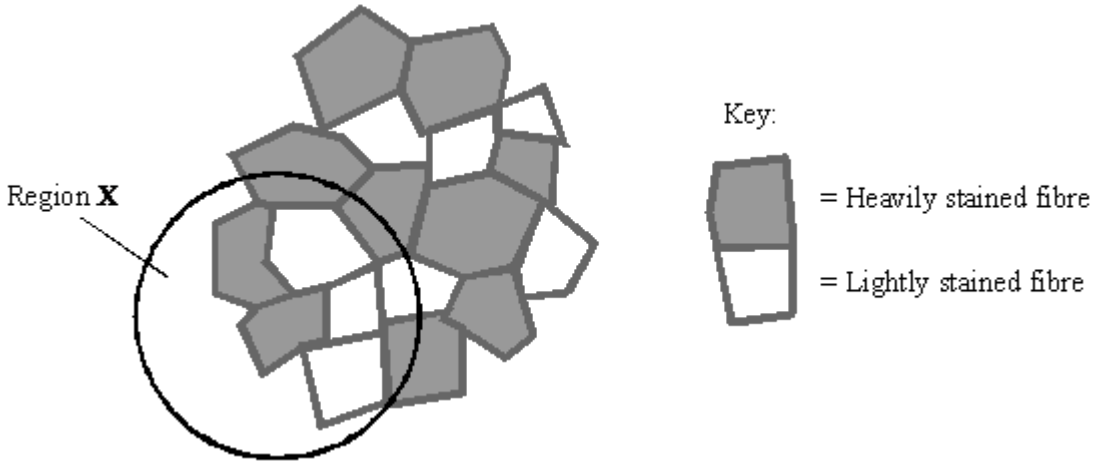


Figure 2

(c) (i) Describe how you could calculate the percentage of fast fibres in this bundle.

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_____

(1)

(ii) The figure calculated by the method in part (c)(i) may not be true for the muscle as a whole. Explain why.

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(1)

(d) The fibres in **Figure 3** correspond to those in region **X** of **Figure 2**. They were stained with a substance that binds to enzymes involved in glycolysis. Shade **Figure 3** to show the appearance of the fibres. Use the shading shown in the key.

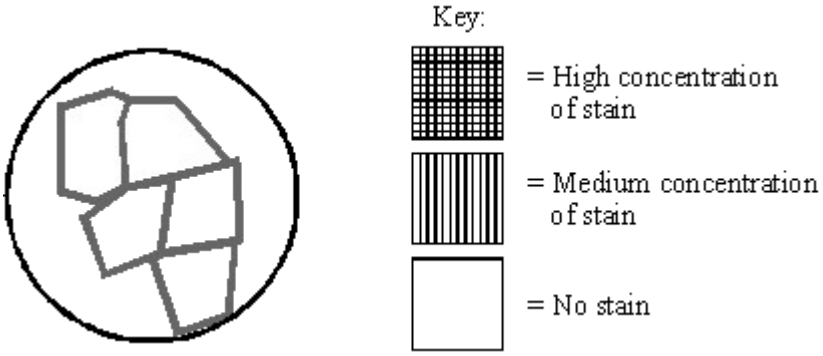


Figure 3

(2)

(e) Recent research has shown that the difference in fibre types is due in part to the presence of different forms of the protein myosin with different molecular shapes.

Explain how a new form of myosin with different properties could have been produced as a result of mutation.

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(4)

(Total 15 marks)

Q12.

One hypothesis for the cause of cancer of the colon (large intestine) is that *Clostridium* bacteria present in the gut can convert bile steroids into cancer-causing substances.

(a) Explain the presence of bile in the colon.

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(2)

(b) The concentrations of bile steroids and numbers of *Clostridium* bacteria were measured in people with colon cancer and in controls without colon cancer. The table shows the results.

Concentration of bile steroids	Number of *Clostridium* bacteria	Percentage of cancer patients	Percentage of controls	P
high	high	76	9	<0.01
high	low	13	8	<0.01
low	high	7	34	<0.01
low	low	4	49	<0.01

A statistical test showed there was a significant difference between the cancer patients and the controls in each of the four categories.

(i) Explain how the results could be used to support the hypothesis that *Clostridium* bacteria convert bile steroids into substances which cause colon cancer.

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(2)

(ii) Explain how the results indicate that other factors may be involved in causing colon cancer.

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(1)

(c) Human cells contain genes that control their growth and division. One of these genes codes for a protein that prevents cell division. The substances formed from bile steroids by *Clostridium* bacteria may cause gene mutation. Describe and explain how these substances could cause colon cancer.

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(4)

(Total 9 marks)