

AQA A-LEVEL • 7402

BIOLOGY



EXAM QUESTION PACK

3.8.1 GENE MUTATION

SCORE

/130

PERCENTAGE

%

STEMUP

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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.

2

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.

This image shows a blank sheet of white paper with horizontal ruling lines. The lines are evenly spaced and extend across the width of the page. There are no margins, text, or other markings on the paper.

(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.

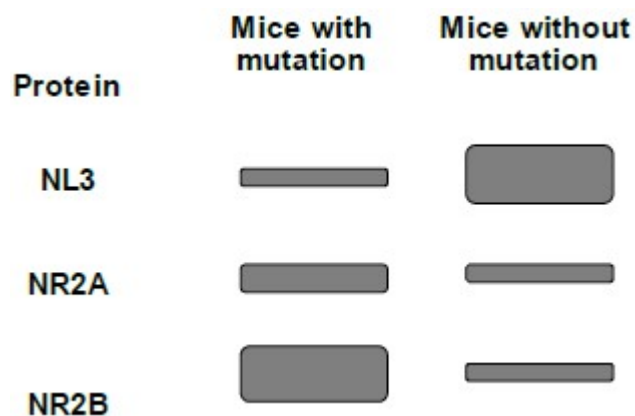
Some autism spectrum disorders (ASDs) are associated with a mutation affecting the neuroligin-3 gene. This gene codes for a protein called NL3, that is found in synapses.

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?

(1)

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

(2)

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

(3)

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

(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.

(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?

(1)

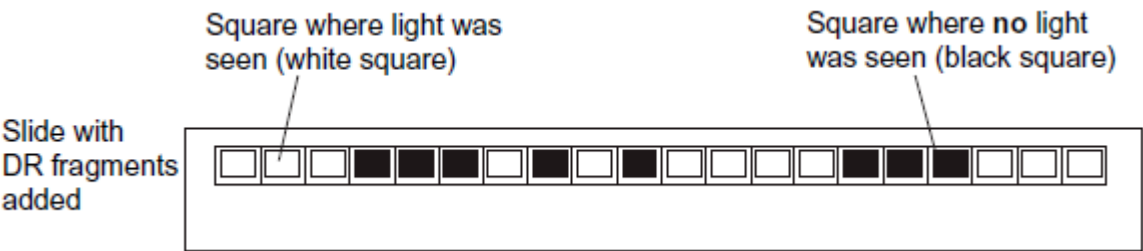
- (ii) Name the process by which the base sequence of a spacer is lost from a DR region.

(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*.

(1)

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

(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.

(2)
(Total 8 marks)

Q4.

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

(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.

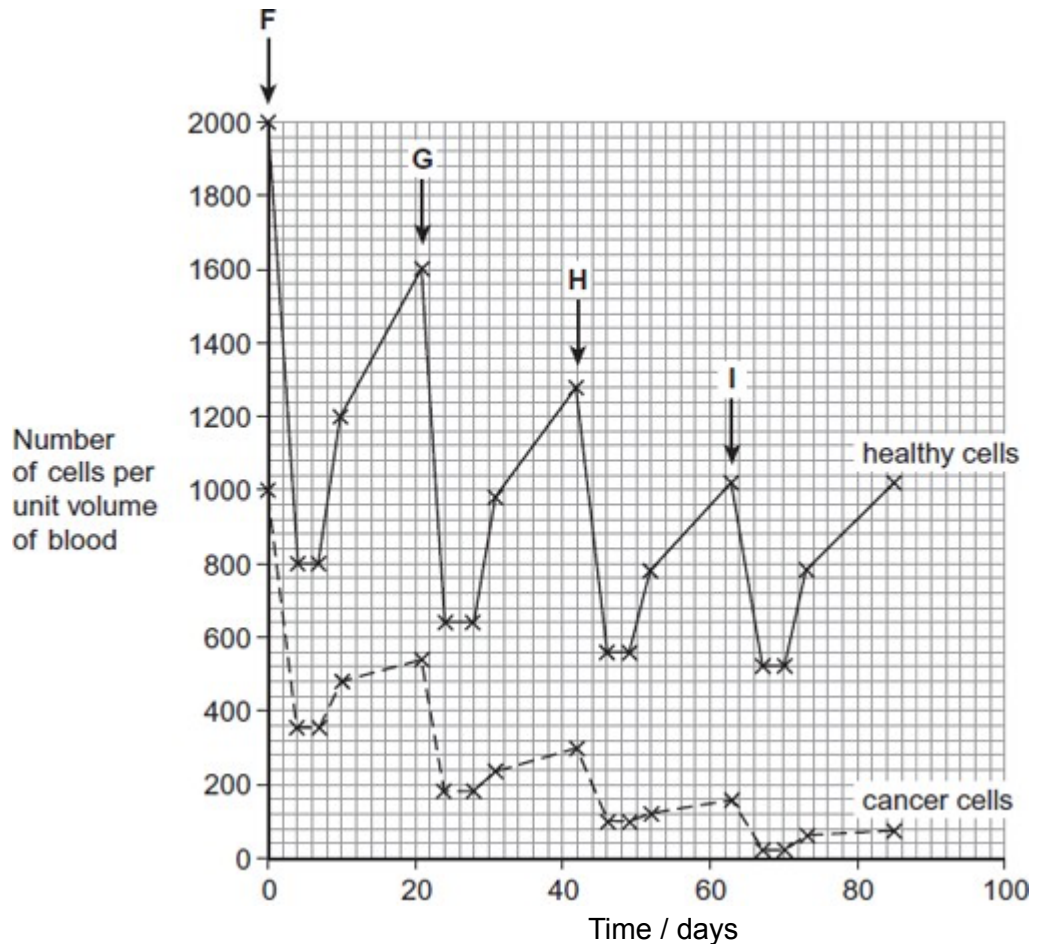
Gene C	Gene D	Gene E	Risk of developing lung cancer
N	N	N	1.00
M	N	N	1.30
N	N	M	1.78
N	M	N	1.45

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.

(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**.

(3)

- (e) More cancer cells could be destroyed if the drug was given more frequently.

Suggest why the drug was **not** given more frequently.

(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.

(2)

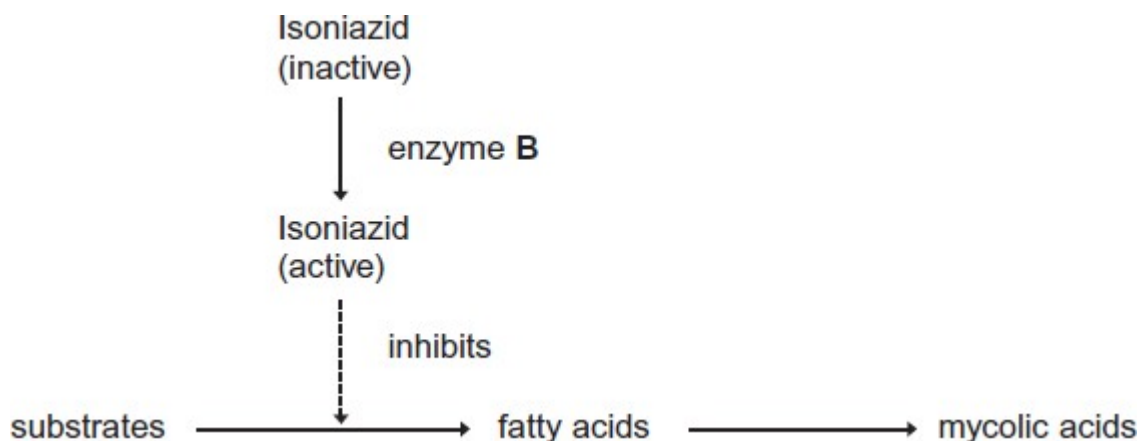
- (ii) Suggest how the production of a protein with one amino acid missing may lead to a genetic disorder such as Ellis-van Creveld syndrome.

(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 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.

(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.

(1)

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

(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

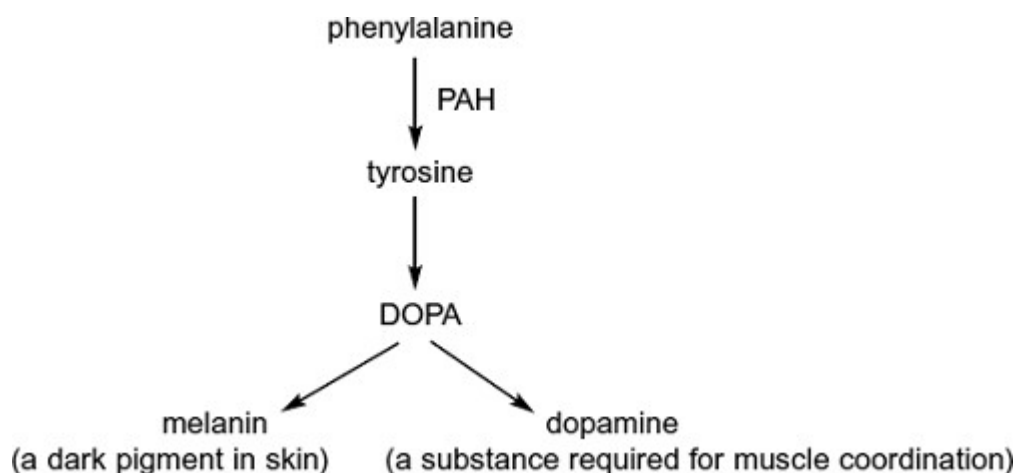
- (a) (i) What is the maximum number of amino acids for which this base sequence could code?

(1)

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

(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.

(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?

(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.
- (c) Some gene mutations affect the amino acid sequence. Some mutations do not. Use the information from **Figure 1** and **Figure 2** to explain

(1)

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

(2)

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

(3)

(d) Gene mutations occur spontaneously.

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

(1)

(ii) Suggest an explanation for your answer.

(1)

(Total 9 marks)

Q10.

Read the following passage.

5 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.

(6)

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

(2)

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

(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.

(2)

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

(3)
(Total 15 marks)

Q11.

Figure 1 shows part of a sarcomere.

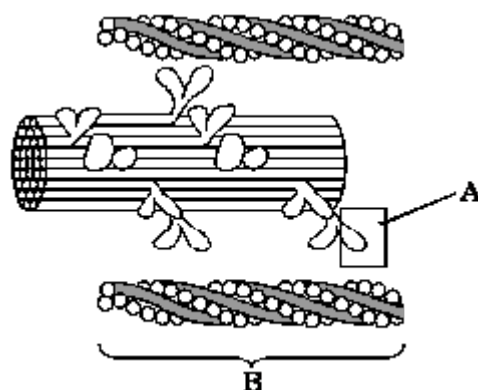


Figure 1

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

(1)

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

(1)

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

(2)

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

(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.

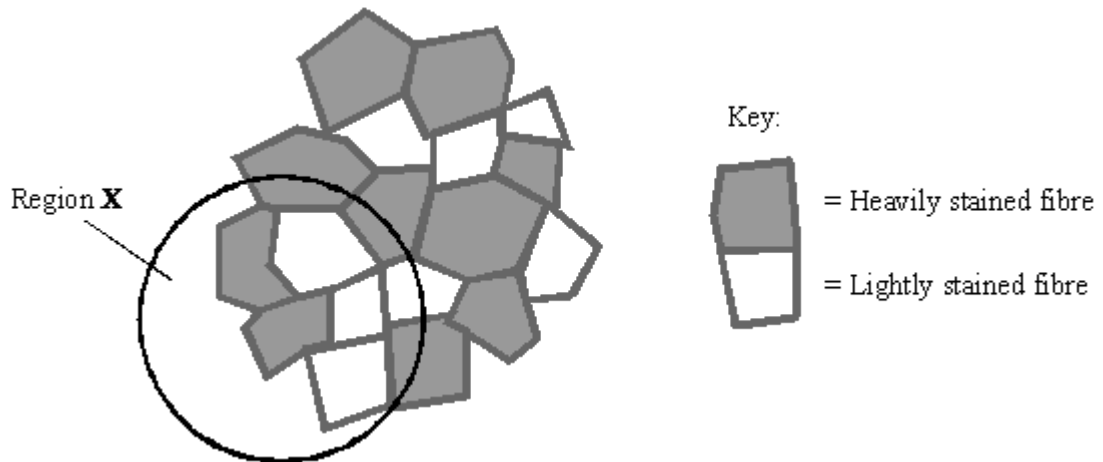


Figure 2

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

(1)

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

(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.

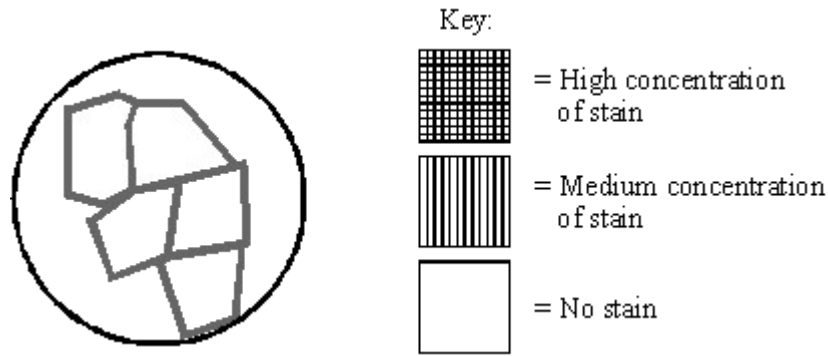


Figure 3

(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.

(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.

(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 <i>Clostridium</i> 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.

(2)

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

(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.

(4)

(Total 9 marks)

Mark schemes

Q1.

(a) Box 2.

An inversion will result in a change in the number of DNA bases.

Reject if more than one box with tick. Ignore crossed-out ticks

1

- (b) 1. (Increased) methylation (of tumour suppressor genes);
Accept abnormal methylation or hypermethylation
Ignore decreased acetylation of histones
2. Mutation (in tumour suppressor genes);
3. Tumour suppressor genes are not transcribed/expressed
OR
Amino acid sequence/primary structure altered;
Accept mRNA for transcription/transcribed
Accept tertiary structure altered
Accept different amino acid
Ignore reference to protein not being formed
4. (Results in) rapid/uncontrollable cell division;
Accept cell division cannot be regulated
Ignore growth

3 max

- (c) 1. Correct answer of $1.9/1.93 \times 10^{25} = 2$ marks;;
Accept $2 \times 10^{25} = 2$ marks
Ignore any numbers after 1.93
2. Incorrect answer but shows $84 = 1$ mark
OR
 $28 \times 3 = 1$ mark
OR
Incorrect answer but shows 672 divided by 8 = 1 mark;

2

[6]

Q2.

(a) 1. Replacement of a base by a different base (in DNA);

1

- (b) 1. (Depends on) size / mass (of protein);
2. (Depends on) charge (of protein);
Accept for 2 marks 'Smaller / more highly charged move further'

2 max

- (c) 1. Each protein has a different tertiary structure;
 2. (Each) antibody has a specific antigen / binding / variable region / site;
 3. So, (each antibody) forms different antigen-antibody complex
OR
 (each antibody) only binds to complementary (protein); 3
- (d) 1. Less NL3;
 2. More NR2A **and** NR2B; 2
- (e) 1. Higher ratio NR2B to NR2A with mutation;
Accept 'more' as equivalent to 'ratio'
 2. (Perhaps) better memory in mice with mutation; 2
- [10]**

Q3.

- (a) (i) Does not code for amino acid/tRNA/rRNA;
Accept 'does not code for production of protein/polypeptide'
Reject 'that produces/makes amino acid' 1
- (ii) Deletion mutation;
Accept 'deletion'
Ignore references to splicing 1
- (b) (The) polymerase chain reaction;
Accept PCR 1
- (c) 1. Probes are single stranded / have a specific base sequence;
 2. Complementary base sequence on (specific) spacer
OR
 3. Complementary/specific to (particular) spacer;
 4. (In white squares probe) binds (to single-stranded spacer) and glows/produces light/fluoresce;
 2. *Need idea of complementary to spacer*
 3. *Accept converse for dark squares* 3
- (d) 1. To see if strain is resistant to any antibiotics;
 2. So can prescribe effective/right antibiotic;
OR
 3. To see whether (any) vaccine works against this strain/
 see which vaccine to use/ to produce specific vaccine;

4. (So) can vaccinate potential contacts/to stop spread;

OR

5. Can test other people to see if they have the same strain/ to trace where people caught TB;
6. Allowing control of spread of disease/vaccinate/treat contacts (of people with same strain) before they get TB;

Do not allow mix and match of points from different alternative pairs

2 max

[8]

Q4.

- (a)
 1. Sugar-phosphate (backbone) / double stranded / helix **so** provides strength / stability / protects bases / protects hydrogen bonds;
Must be a direct link / obvious to get the mark
Neutral: reference to histones
 2. Long / large molecule **so** can store lots of information;
 3. Helix / coiled **so** compact;
Accept: can store in a small amount of space for 'compact'
 4. Base sequence allows information to be stored / base sequence codes for amino acids / protein;
Accept: base sequence allows transcription
 5. Double stranded **so** replication can occur semi-conservatively / strands can act as templates / complementary base pairing / A-T and G-C so accurate replication / identical copies can be made;
 6. (Weak) hydrogen bonds **for** replication / unzipping / strand separation / many hydrogen bonds **so** stable / strong;
Accept: 'H-bonds' for 'hydrogen bonds'

6

- (b)
 1. (Mutation) in **E** produces highest risk / 1.78;
 2. (Mutation) in **D** produces next highest risk / 1.45;
 3. (Mutation) in **C** produces least risk / 1.30;
Must be stated directly and not implied
E > D > C = 3 marks
Accept: values of 0.78, 0.45 and 0.30 for MP1, MP2 and MP3 respectively
If no mark is awarded, a principle mark can be given for the idea that all mutant alleles increase the risk

3

- (c) **180**;

1

- (d) **(Similarities):**

1. Same / similar pattern / both decrease, stay the same then increase;
2. Number of cells stays the same for same length of time;
Ignore: wrong days stated

(Differences):

(Per unit volume of blood)

3. Greater / faster decrease in number of healthy cells / more healthy cells killed / healthy cells killed faster;
Accept: converse for cancer cells
Accept: greater percentage decrease in number of cancer cells / greater proportion of cancer cells killed

4. Greater / faster increase in number of healthy cells / more healthy cells replaced / divide / healthy cells replaced / divide faster;
Accept: converse for cancer cells
*For **differences**, statements made must be comparative*

3 max

- (e)
1. More / too many healthy cells killed;
 2. (So) will take time to replace / increase in number;
Neutral: will take time to 'repair'
 3. Person may die / have side effects;

2 max

[15]

Q5.

- (a) 250 000;

1

- (b) (i) Loss of 3 bases / triplet = 2 marks;;
'Stop codon / code formed' = 1 mark max unless related to the last amino acid

Loss of base(s) = 1 mark;

eg triplet for last amino acid is changed to a stop codon / code = 2 marks

3 bases / triplet forms an intron = 2 marks

Accept: descriptions for 'intron' eg non-coding DNA

'Loss of codon' = 2 marks

2

- (ii)
1. Change in tertiary structure / active site;
Neutral: change in 3D shape / structure
 2. (So) faulty / non-functional protein / enzyme;
Accept: reference to examples of loss of function eg fewer E-S complexes formed

2

[5]

Q6.

- (a) 1. Cell wall not formed / production inhibited;
 1. *Q Accept: weakened cell wall, but do not accept 'cell wall is broken down'*
2. Lower water potential in bacterium;
 2. *Accept: converse*
 2. *Must be clear that the lower water potential is in the bacterium*
3. Water enters and causes lysis / expansion / pressure;
- (b) Human cells lack enzyme (**B**) / have a different enzyme / produce different fatty acids / use different substrates;
 Neutral: 'human cells do not have cell walls' as out of context
- (c) 1. Change in base sequence (of DNA / gene) leading to change in amino acid sequence / primary structure (of enzyme);
 1. *Accept: different amino acids coded for*
 1. *Reject: different amino acids produced*
2. Change in hydrogen / ionic / disulphide bonds leading to change in the tertiary structure / active site (of enzyme);
 2. *Neutral: alters 3D structure / 3D shape*
3. Substrate not complementary / cannot bind (to enzyme / active site) / no enzyme-substrate complexes form;

2 max

1

3

[6]**Q7.**

- (a) (i) 4;
- (ii) 1. Change in amino acid / (sequence of) amino acids / primary structure;
 1. *Reject = different amino acids are 'formed'*
2. Change in hydrogen / ionic / disulphide bonds alters tertiary structure / active site (of enzyme);
 2. *Alters 3D structure on its own is not enough for this marking point.*
3. Substrate not complementary / cannot bind (to enzyme / active site) / no enzyme- substrate complexes form;
- (b) 1. Lack of skin pigment / pale / light skin / albino;
2. Lack of coordination / muscles action affected;
- (c) Founder effect / colonies split off / migration / interbreeding;

1

3

2 max

Q8.

21 – 25	Extended abstract Generalised beyond specific context	Response shows holistic approach to the question with a fully integrated answer which makes clear links between several different topics and the theme of the question. Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained. No significant errors or irrelevant material. For top marks in the band, the answer shows evidence of reading beyond specification requirements.
16 – 20	Relational Integrated into a whole	Response links several topics to the main theme of the question, to form a series of interrelated points which are clearly explained. Biology is fundamentally correct A-level content and contains some points which are detailed, though there may be some which are less well developed, with appropriate use of terminology. Perhaps one significant error and, or, one irrelevant topic which detracts from the overall quality of the answer.
11 – 15	Multistructural Several aspects covered but they are unrelated	Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question. Biology is usually correct A-level content, though it lacks detail. It is usually clearly explained and generally uses appropriate terminology. Some significant errors and, or, more than one irrelevant topic.
6 – 10	Unistructural Only one or few aspects covered	Response predominantly deals with only one or two topics that relate to the question. Biology presented shows some superficial A-level content that may be poorly explained, lacking in detail, or show limited use of appropriate terminology. May contain a number of significant errors and, or, irrelevant topics.

1 – 5	Unfocused	Response only indirectly addresses the theme of the question and merely presents a series of biological facts which are usually descriptive in nature or poorly explained and at times may be factually incorrect. Content and terminology is generally below A-level. May contain a large number of errors and, or, irrelevant topics.
0		Nothing of relevance or no response.

Commentary on terms and statements in the levels mark scheme

The levels mark scheme for the essay contains a number of words and statements that are open to different interpretations. This commentary defines the meanings of these words and statements in the context of marking the essay. Many words and statements are used in the descriptions of more than one level of response. The definitions of these remain the same throughout.

Levels mark scheme word/statement	Definition
Holistic	Synoptic, drawing from different topics (usually sections of the specification)
A fully integrated answer which makes clear links between several different topics and the theme of the question	All topics relate to the title and theme of the essay; for example, explaining the biological importance of a process. When considering, for example, the importance of a process, the explanation must be at A-level standard. ‘Several’ here is defined as at least four topic areas from the specification covered. This means some sentences, not just a word or two. It does not mean using many examples from one topic area.
Biology is detailed and comprehensive A-level content, uses appropriate terminology, and is very well written and always clearly explained.	Detailed and comprehensive A-level content is the specification content. Terminology is that used in the specification. Well written and clearly explained refers mainly to biological content and use of terminology. Prose, handwriting and spelling are secondary considerations. Phonetic spelling is accepted, unless examiners are instructed not to do so for particular words; for example, glucagon, glucose and glycogen.

No significant errors or irrelevant material.	A significant error is one which significantly detracts from the biological accuracy or correctness of a described example. This will usually involve more than one word. Irrelevant material is several lines (or more) that clearly fails to address the title, or the theme of the title.
For top marks in the band, the answer shows evidence of reading beyond specification requirements.	An example that is relevant to the title and is not required in the specification content. The example must be used at A-level standard.
Response mostly deals with suitable topics but they are not interrelated and links are not made to the theme of the question.	Not addressing the biological theme of the essay (e.g. importance) at <u>A-level standard</u> .

Please note that to obtain full credit, students must use information to show **the importance of Using DNA in science and technology**.

Topics

DNA and classification	
2.2	Structure of DNA
2.3	Differences in DNA lead to genetic diversity
2.9	Comparison of DNA base sequences
Genetic engineering and making useful substances	
2.5	Plasmids
5.8	The use of recombinant DNA to produce transformed organisms that benefit humans
Other uses of DNA	
2.5	Cell cycle and treatment of cancer
5.8	Gene therapy; Medical diagnosis and the treatment of human disease; The use of DNA probes to screen patients for clinically important genes.

In order to fully address the question and reach the highest mark bands students must also include at least four topics in their answer, to demonstrate a synoptic approach to the essay.

Students may be able to show the relevance of other topics from the specification.

Note, other topics from beyond the specification can be used, providing they relate to the title and contain factually correct material of at least an A-level standard. Credit should not

be given for topics beyond the specification which are below A-level standard.

[25]

Q9.

- (a) Introns; 1
- (b) Ile Gly Val Ser; 1
- (c) (i) Has no effect / same amino acid (sequence) / same primary structure; 1
Q Reject same amino acid formed or produced.
- Glycine named as same amino acid; 1
It still codes for glycine = two marks.
- (ii) Leu replaces Val / change in amino acid (sequence) / primary structure; 3
 Change in hydrogen / ionic bonds which alters tertiary structure / active site;
Q Different amino acid formed or produced negates first marking point.
 Substrate cannot bind / no longer complementary / no enzyme-substrate complexes form;
Active site changed must be clear for third marking point but does not need reference to shape.
- (d) (i) Interphase / S / synthesis (phase); 1
- (ii) DNA / gene replication / synthesis occurs / longest stage; 1
Allow 'genetic information' = DNA.
Allow 'copied' or 'formed' = replication / synthesis

[9]

Q10.

- (a) 1 (DNA altered by) mutation;
 2 (mutation) changes base sequence;
 3 of gene controlling cell growth / oncogene / that monitors cell division;
 4 of tumour suppressor gene;
 5 change protein structure / non-functional protein / protein not formed;
 6 (tumour suppressor genes) produce proteins that inhibit cell division;
 7 mitosis;
 8 uncontrolled / rapid / abnormal (cell division);
 9 malignant tumour; max 6
- (b) cancer cells die / break open;
 releasing DNA;

- | | | |
|--|---|-------------|
| | 2 | |
| (c) normal DNA and changed DNA have different sequences;
DNA only binds to complementary sequence; | | 2 |
| (d) fewer abnormal / cancerous cells / smaller tumours;
less cell damage / less spread / fewer locations to treat; | | 2 |
| (e) mRNA base sequence has changed;
gene / DNA structure is different / has mutated;
cancer gene active / tumour suppressor gene inactive; | | 3 |
| | | [15] |

Q11.

- | | | |
|---|--|-------------|
| (a) (i) actin (<i>Accept</i> tropomyosin); | | 1 |
| (ii) myosin head; | | 1 |
| (b) (i) Ca^{2+} binds to [part of] the actin / troponin;
this causes tropomyosin to be displaced;
uncovers [myosin] binding sites [on actin] / allows actin to bind; | | max 2 |
| (ii) myosin heads bind to actin / cross bridge formation /
actomyosin formed;
myosin heads / crossbridges swivel / ratchet mechanism;
causing actin to slide relative to myosin;
energy provided by hydrolysis of ATP; | | max 3 |
| (c) (i) (number lightly stained fibres / total number of fibres) $\times$ 100;
(actual numbers are 10 / 18 $\times$ 100) | | 1 |
| (ii) sample not representative / large enough / individual muscle fibres
different sizes / contain different number of myofibrils; | | 1 |
| (d) all some stain = 1
fast dark and slow lighter = 2 | | 2 |
| (e) change in base sequence in DNA / addition / deletion / substitution of a base
in DNA of the gene which codes for myosin;
change in amino acid sequence / primary structure;
causes a different tertiary structure;
which alters the binding properties of myosin; | | 4 |
| | | [15] |

Q12.

- (a) secreted by the liver / storage / release from gall bladder into the duodenum / small

- intestine;
bile passes unchanged from small intestine to colon;
- 2
- (b) (i) chance alone has not caused the difference (between the two patients types);
high steroid high bacteria (significantly) higher percentage of cancer patients / low steroids low bacteria (significantly) higher percentage of control patients;
- 2
- (ii) some patients with low levels of one / both factor(s) have cancer;
- 1
- (c) change in code / base sequence / structure of gene;
addition / deletion / substitution;
mRNA / transcription changed;
gene product / protein structure / amino acid sequence changed / different protein;
loss of function;
uncontrolled cell division;
- 4 max

[9]

Examiner reports

Q1.

- (a) The vast majority of students (94%) realised that an inversion would not result in a change in the number of DNA bases.
- (b) This question proved to be a very effective discriminator with one in three students gaining maximum marks. Nearly all students (91%) gained at least one mark, often for referring to mutation or methylation of tumour suppressor genes. Mutation was the most frequently credited marking point, although a significant number of students mentioned both alterations for two marks. Weaker students who provided further details on mutations often limited their descriptions to a non-functional protein being produced. Better answers included changes in primary or tertiary structure. Many students describing methylation did appreciate that this could prevent transcription of tumour suppressor genes. A significant minority of students referred to uncontrollable cell growth rather than uncontrollable or rapid cell division. There were also irrelevant, and often incorrect, references to oncogenes.
- (c) Over 83% of students gained at least one mark, almost invariably for showing 84 cell divisions. Slightly more than half of these students provided further calculations leading to a correct answer for two marks. Common errors included 84^2 and 28^3 .

Q3.

- (a)
 - (i) 72% correctly wrote about not coding for amino acids. Some failed to score because they wrote about 'not producing/making amino acids'.
 - (ii) Most students ignored the fact that the DR region is in DNA and wrote about splicing/removing the non-coding sequence from mRNA. The other 22% wrote, correctly, about deletion mutations.
- (c) This question discriminated well across the range of marks. 50% obtained two marks, usually for reference to a probe binding to a complementary base sequence on spacer DNA and then linking this to production of light. Only 13% also stated that each probe has a specific base sequence. 14% failed to score. Some referred to complementary base pairing, rather than sequences.
- (d) Surprisingly, 40% scored zero. There were many very vague answers about picking the right treatment and not encouraging resistance. 20% obtained both marks, usually for the idea of identifying any antibiotics to which the strain was resistant, so an appropriate antibiotic can be selected. The other mark points relating to vaccines and tracing sources of infection were also seen.

Q4.

Parts (a), (b) and (d) proved to be good discriminators.

- (a) It was disappointing that only just below 40% of students scored at least half marks. This was mainly due to simply describing the structure of DNA, without explaining how these features relate to its functions. Some students wrote about DNA structure and function in different paragraphs. This made it unclear which feature went with which function, as no direct links had been made. In contrast, there were some truly excellent responses, which had clearly been well planned before putting pen to paper. The most common mark points awarded were for the sugar-phosphate backbone providing strength or protecting bases, the helix allowing the molecule to

be compact, weak hydrogen bonds allowing strand separation or replication and the two strands acting as templates or allowing semi-conservative replication. Relatively few students linked complementary base pairing with accurate replication or the production of identical copies of DNA. Similarly, few students referred to DNA as a large molecule that can store lots of information, or the base sequence coding for amino acids. Weaker responses often mentioned this in the context of the genetic code being degenerate. Indeed, some students thought that the base sequence causes amino acids to be *produced*. The ability to convey that *many* hydrogen bonds provide stability was rarely seen. It was also unfortunate that a number of students wasted their time by writing about irrelevant topics such as the differences between prokaryotic and eukaryotic DNA and the role of histones. There were also some lengthy accounts of DNA replication, enzyme structure and the different levels of protein structure.

- (b) Many students scored at least two marks for stating that a mutation in gene **E** produces the highest risk and a mutation in gene **C** produces the lowest risk. However, only the best responses also referred to gene **D**. Students who did not mention any of the genes usually picked up one mark for noting that all of the mutant alleles increase the risk of lung cancer. Surprisingly, some thought that a mutation in gene **D** produces the highest risk.
- (c) Just fewer than 40% of students gave the correct answer of **180**.
- (d) Two-thirds of students scored at least two marks. Many were able to identify the decrease, plateau and increase for healthy cells and cancer cells. However, relatively few made reference to the plateau occurring for the same length of time. Students who failed to gain a mark for a similarity usually ignored the plateau. Most students spotted that a greater number of healthy cells were killed or that they experienced a faster decrease in number. Similarly, it was impressive to see that some used data from the graph to calculate that a greater *proportion* of cancer cells were killed. Many students also noted the faster increase in the number of healthy cells.
- (e) Half of students scored full marks. This was usually for mentioning that too many healthy cells would be killed, which could kill the patient or cause side effects. However, relatively few appreciated that it would take time to replace the healthy cells that had been killed.

Q5.

- (a) Nearly all students gave the correct answer of **250,000**.
- (b) (i) One-third of students gained at least one mark. This question required students to apply the principle that three bases code for one amino acid to an unfamiliar context. However, other creditworthy approaches were used to explain why the faulty protein has one amino acid missing. This said, many students simply defined the term 'mutation' or repeated information given in the question stem. Consequently, there were many references to a *change* in the base sequence or amino acid sequence. Only the best responses mentioned a loss of bases. Students who took a different approach fell into one of two camps. Some suggested that a stop codon had formed for one mark. However, it was rare to see this related to the final amino acid of the protein. Similarly, others were clearly aware of introns but rarely mentioned that three bases may form an intron. Unfortunately, a minority of students provided a good response to (c) (ii) for this question part.
- (ii) One-third of students gained full marks. Many were aware that the protein

produced could be faulty or non-functional. However, the ability to explain this in terms of a change in tertiary structure or active site discriminated well. Unfortunately, some students went no further than to state that the protein would have a different primary structure. This was given in the question stem and therefore not credited.

Q6.

- (a) This proved to be an excellent discriminator. Nearly half of students scored full marks. This was usually for stating that the cell wall does not form, leading to cell lysis due to entry of water. It was usually only the best responses that referred to a lower water potential in the bacterium. Weaker responses revealed a number of misconceptions. These often referred to the cell wall being broken down or that isoniazid *caused* the cell wall to become permeable to water.
- (b) Half of students were aware that human cells may lack enzyme **B**, use different substrates, or produce different fatty acids. Weaker responses usually fell into one of two types. The first suggested the idea that isoniazid is an enzyme. This led to widespread references to enzyme inhibition and active sites on a variety of molecules. The second used the fact that human cells do not have cell walls. The question asked why isoniazid does not affect the production of *fatty acids* in human cells. Hence, reference to cell walls was out of context and was not credited.
- (c) Two-thirds of students scored full marks and all marking points were regularly seen. Weaker responses were marked by the use of scientific terms in the wrong context, e.g. 'different amino acids produced', 'base sequence of the enzyme', 'amino acid base sequence', 'amino acids coding for' and 'different hydrogen bonds form between bases'.

Q7.

- (a)
 - (i) Over 90% of students correctly determined that base sequence could code for a maximum number of four amino acids.
 - (ii) The vast majority of students gained at least one mark, often by mentioning a change in the sequence in amino acids. However, a significant number of students incorrectly referred to 'different amino acids being formed'. Most students gained a second mark for explaining that the active site/ tertiary structure would be altered. Over 50% of students gained maximum marks either by linking this to enzyme-substrate complexes not being formed or to changes in hydrogen bonds.
- (b) Most students had little difficulty in using the information to give two symptoms of phenylketonuria and gained both marks.
- (c) The majority of students obtained this mark, often by referring to migration or by describing interbreeding. However, over a third of students failed to gain credit and often accounted for the spread of phenylketonuria by horizontal or vertical gene transfer.

Q8.

Using DNA in science and technology

The very best essays from candidates who selected this option were outstanding. They reviewed, often in great detail, the relevant aspects of the specification although not always incorporating the role of DNA in the classification of organisms. Considering that

much of the content of this essay could be drawn from this unit, it was surprising how poor many answers were. Understanding of techniques was often extremely limited, particularly *in vivo* gene cloning and the use of markers. Many essays presented no more than a broad overview either emphasising ethical issues at the expense of biological detail or failing to distinguish established practice from wishful thinking.

Q9.

- (a) Less than half the candidates correctly named introns as the non-coding sections of a gene.
- (b) The vast majority of candidates correctly identified the amino acid sequence.
- (c)
 - (i) Most candidates obtained at least one mark for stating that the amino acid sequence would not change. However, less than half the candidates gained the second mark by explaining that the new base triplet would still code for glycine.
 - (ii) Most candidates gained at least one mark, often by mentioning a change in the sequence of amino acids. However, a significant number of candidates incorrectly referred to 'different amino acids being formed'. Many candidates gained a second mark for explaining that the active site/ tertiary structure would be altered. The best candidates gained maximum marks either by linking this to enzyme-substrate complexes not being formed or to changes in hydrogen or ionic bonds.
- (d)
 - (i) Almost two thirds of candidates correctly identified the part of the cell cycle as being interphase or the synthesis stage. Anaphase was a common incorrect response.
 - (ii) Most candidates obtained this mark, often by indicating that DNA replication occurs during interphase.

Q10.

- (a) Many candidates gave a good account of the changes a mutation could produce and those with clear expression achieved full marks; many scored three or four marks. Uncontrolled cell division and malignant tumors were frequently referred to and some appreciated that genes which controlled cell division could have changed. References to benign tumours or cell mutations were irrelevant in the context of this question.
- (b) Very few candidates achieved marks here, mainly because they did not read the question. Whole cells in the blood were not required, but the understanding that cancer cells could burst or die and release their DNA was.
- (c) Few seemed to understand this and restated the question without reference to the changed base sequences to which the strip would bind.
- (d) This was generally well known. The main reason for failing to gain marks was a reference to an undefined 'it' which would be growing, dividing or spreading, causing undefined damage.
- (e) Here too some candidates who understood the problem found it hard to explain that changes in the mRNA would reflect mutations in the DNA and would show that a cancer gene was active.

Q11.

- (a) (i) and (ii) A majority of candidates were able to identify A as the myosin head, although rather fewer were able to name actin as the main protein in the thin filament.
- (b) In general, the responses to this section of the question revealed a pleasing level of knowledge and understanding.
 - (i) Many candidates, including otherwise weaker candidates, were able to describe the role of calcium ions in binding to troponin and removing tropomyosin from the myosin binding sites on the actin molecule.
 - (ii) Again, a good number were able to describe the role of ATP and the two proteins in bringing about contraction of the myofibril.
- (c) (i) Only better candidates realised that to calculate the percentage of fast fibres, the number of fast fibres (lightly stained fibres) must be divided by the *total* number of fibres and this figure then multiplied by one hundred. Many weaker candidates multiplied the ratio of the two fibres by one hundred.
- (ii) Most candidates could explain that the figure obtained might not be typical as different regions of a muscle may have different proportions of the two fibres, or because the sample used is such a small one as to be not necessarily reliable.
- (d) Only really able candidates realised that *all* the fibres would undergo glycolysis, whether respiring aerobically or anaerobically. However, those respiring anaerobically would undergo glycolysis only (and not any further stages of the aerobic pathway) and so produce the enzymes used in glycolysis in greater concentrations.
- (e) Many candidates interpreted this question as another concerning natural selection, despite the clear instruction to explain how a new form of myosin could be *formed as a result of mutation*. Good candidates were able to explain how alterations to the base sequence of DNA could result in a different mRNA and, as a result, a different primary structure of the protein. They then went on to explain how this would result in different folding of the molecule, tertiary structure and properties as a result.

Q12.

This question produced a wide range of marks and discriminated well between candidates.

- (a) Most candidates described the role of bile in digestion but failed to refer to the site of production, or to recognise the distinction between the small intestine and the colon.
- (b) The concept of significance was well rehearsed by many candidates. However, few could then use the evidence effectively to support the hypothesis. Most simply related high incidence to high levels of both factors, without drawing a comparison with the control patients. Weaker candidates confused cause and effect, assuming percentage quoted indicated risk of contracting cancer, rather than patients found to have the factor present.
- (c) There were a lot of good answers with a significant minority of candidates gaining full marks. Very few mentioned changes to mRNA resulting from mutations, referring only to the influence on the process of translation. Weaker candidates interchanged base sequence of DNA with amino acid sequence of polypeptide.