Gene Mutations

These practice questions can be used by students and teachers and is

Suitable for AQA A Level 7402 Biology Topic Question

Level: AQA A LEVEL 7402

Subject: Biology

Exam Board: AQA A Level 7402

Topic: Gene Mutations

1

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.

_____

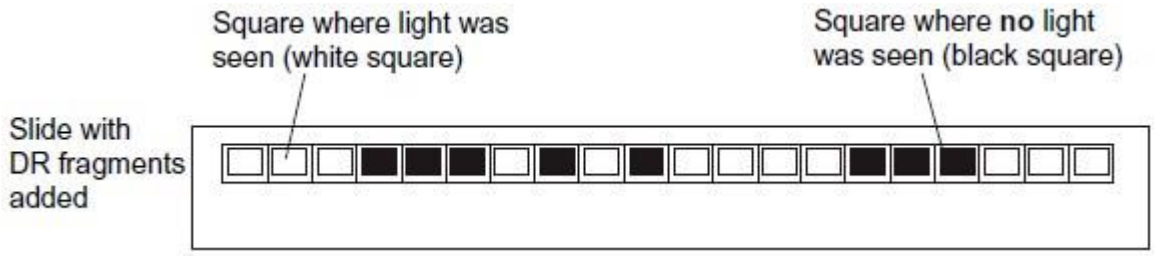
_____

(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 itscomplementary 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)

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

(Extra space)

(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.

_____

_____

_____

_____

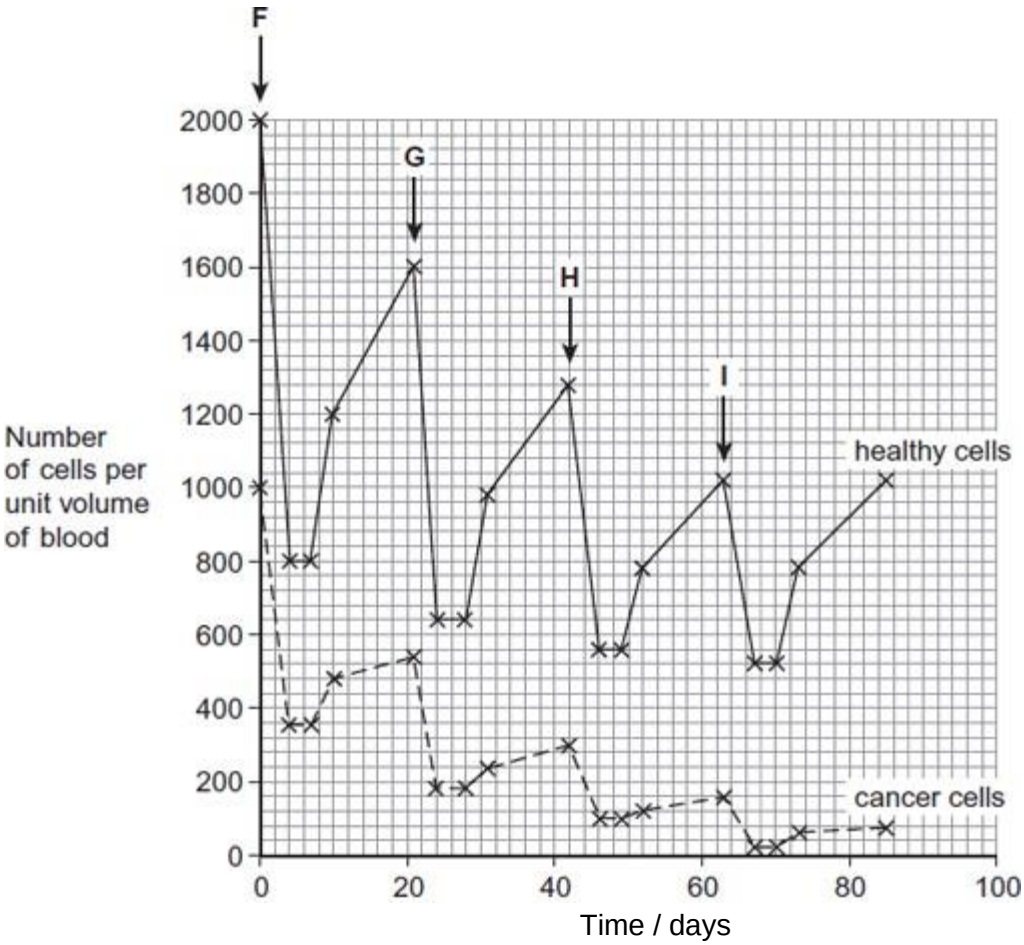
_____

_____

_____

(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 thedrug between times **F** and **G**.

_____

_____

_____

(Extra space) _____

(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)

3 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 Creveldsyndrome. 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 agene 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 aminoacid missing.

_____

_____

_____

_____

_____

(2)

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

_____

_____

_____

_____

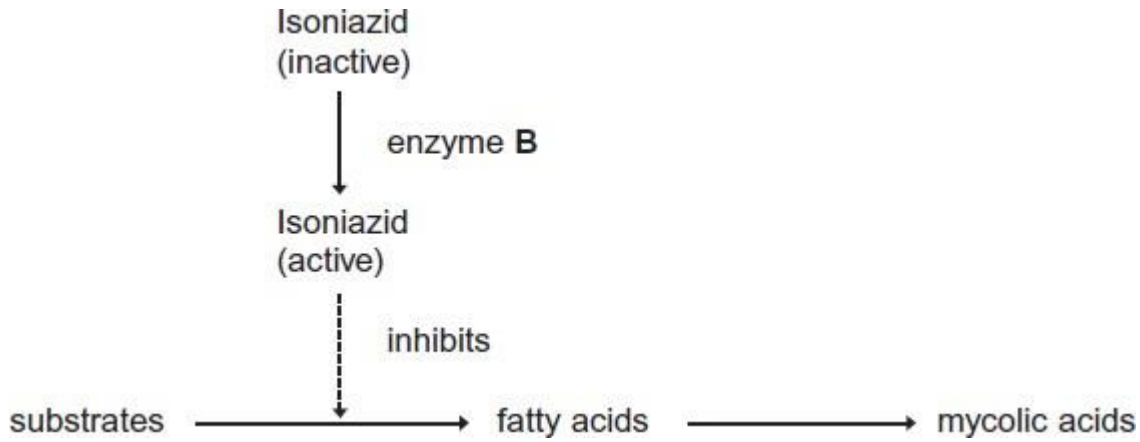
_____

(2)

(Total 5 marks)

4 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 thediagram to suggest how.

_____

_____

_____

_____

_____

(2)

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

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.

(Extra space)

(3)

(Total 6 marks)

5 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?

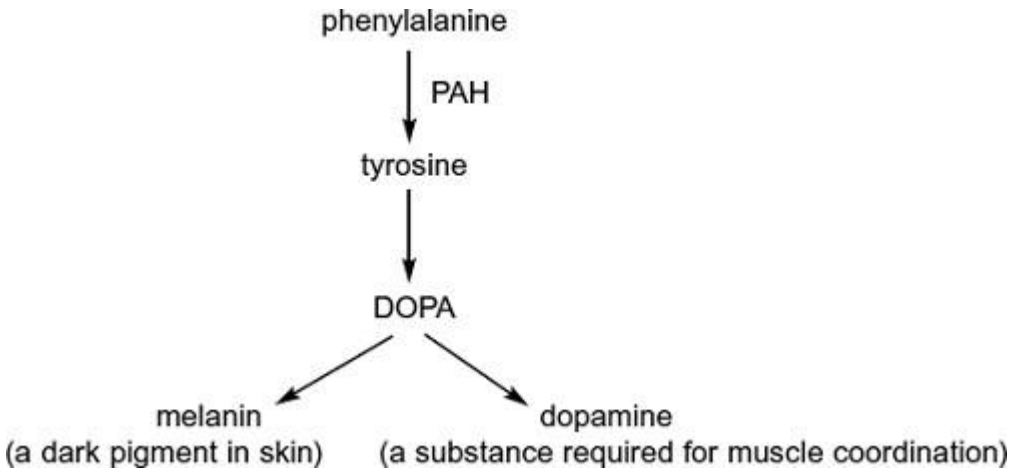
(1)

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

(*Extra space*)_____

(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 incentral Asia. It is now found in many different populations across Asia. Suggest how the spread of this mutation may have occurred.

_____

_____

_____

(1)

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

6

_____

(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

_____

_____

_____

_____

(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)

Essay

7

You should write your essay in continuous prose.

Your essay will be marked for its scientific accuracy.

It will also be marked for your selection of relevant material from different parts of the specification and for the quality of your written communication.

The maximum number of marks that can be awarded is

Scientific	16
Breadth of knowledge	3
Relevance	3
Quality of written communication	3

Write an essay on the following topic:

Using DNA in science and technology

(Total 25 marks)

Read the following passage.

8

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 5 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 ismore 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 ispresent.

(3)

(Total 15 marks)

9 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.

S (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 *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.

_____

_____

_____

_____

(2)

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

_____

_____

(1)

S (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)

10 **Figure 1** shows part of a sarcomere.

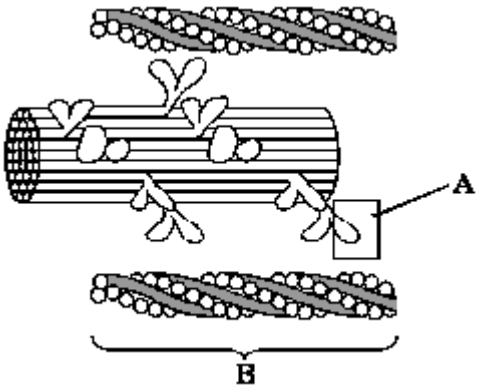

Figure 1

(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.

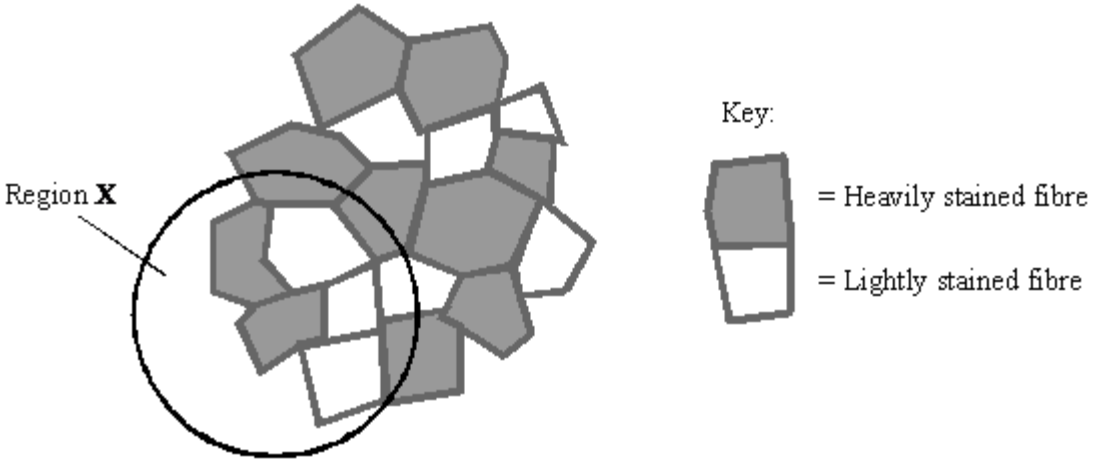

Figure 2

S (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.

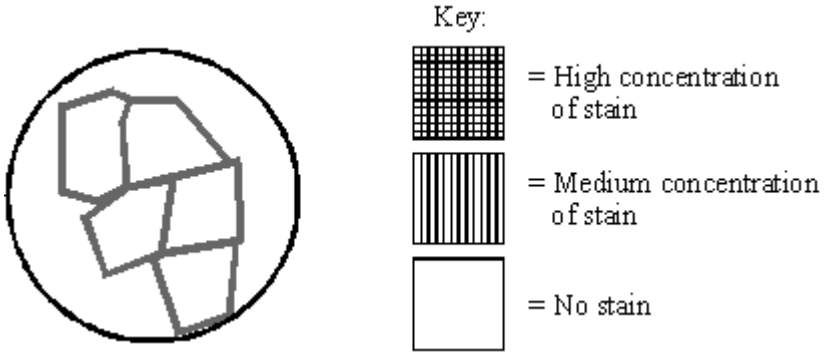

Figure 3

(2)

S (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)

Mark schemes

(a) (i) Does not code for amino acid/tRNA/rRNA;

1

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) andglows/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/ seewhich 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/ totrace 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]

(a) 1. Sugar-phosphate (backbone) / double stranded / helix **so** provides strength / stability

2

/ 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 foramino 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 <u>percentage</u> decrease in number of cancer cells / greater <u>proportion</u> of cancer cells killed

4. Greater / faster increase in number of healthy cells / more healthy cellsreplaced / 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] (a) 250 000;

3 **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] (a) 1. Cell wall not formed / production inhibited;

4

1. ***Q*** *Accept: weakened cell wall, but do not accept 'cell wall is broken down'*

2. Lower <u>water potential</u> in bacterium;

2. Accept: converse

2. Must be clear that the lower water potential is in the bacterium

3. <u>Water enters</u> and causes lysis / expansion / pressure;

2 max

(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

1

(c) 1. Change in base sequence (of DNA / gene) leading to change in amino acidsequence / 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 tertiarystructure / active site (of enzyme);

2. Neutral: alters 3D structure / 3D shape

3. Substrate not complementary / cannot bind (to enzyme / active site) / noenzyme-substrate complexes form;

3

[6] (a) (i) 4;

5 **1**

(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 markingpoint.

3. Substrate not complementary / cannot bind (to enzyme / active site) / noenzyme- substrate complexes form;

3

(b) 1. Lack of skin pigment / pale / light skin / albino;

2. Lack of coordination / muscles action affected;

2 max

(c) Founder effect / colonies split off / migration / interbreeding;

Allow description of interbreeding e.g. reproduction between individuals from different populations

1

[7] (a) Introns;

6 **1**

(b) Ile Gly Val Ser;

1

(c) (i) Has no effect / same amino acid (sequence) / sameprimary structure;

***Q** Reject same amino acid formed or produced.*

1

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;

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.

3

(d) (i) Interphase / S / synthesis (phase);

1

(ii) DNA / gene replication / synthesis occurs / longest stage;*Allow 'genetic information' = DNA.*

Allow 'copied' or 'formed' = replication / synthesis

1

[9]

Essay Using DNA in science and technology

7

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.

8

(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

9

[15] (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] (a) (i) actin (*Accept* tropomyosin);

10

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) × 100;
(actual numbers are 10 / 18 × 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 DNAof 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]