Helicate

HELICATE · A-LEVEL BIOLOGY · TOPIC BANK

3.8.1 Alteration of the sequence of bases in DNA can alter the structure of proteins

Topic question bank

Version 3 · 17 October 2026

Name

Date

QUESTIONS	TOTAL MARKS	TIME	YOUR SCORE
8	50	No time limit	/ 50

Contents

Subtopic	Questions	Marks
Types of gene mutation and the mutation rate Specification 3.8.1	01–03	16
Effects of gene mutations on the encoded polypeptide Specification 3.8.1	04–06	19
Gene mutations and protein function Specification 3.8.1	07–08	15
Total		**50**

Instructions

- Answer the questions in the spaces provided. Show all your working.
- The marks for each question are shown in brackets. Use them to judge how much detail to give.
- Work through a subtopic at a time, then mark your answers with the matching mark scheme.

Original questions by Helicate for AQA A-level Biology (7402), specification section 3.8.1. Not affiliated with or endorsed by AQA.

Do not write outside the box

Types of gene mutation and the mutation rate 3.8.1

0 1 . 1 The table below describes three gene mutations.
Complete the table by naming each type of gene mutation.

[3 marks]

	Description of the gene mutation	Type of gene mutation
1	The order of a group of bases in the gene is reversed.	
2	A group of bases is copied, and the copy is inserted next to the original group.	
3	One base in the gene is replaced by a different base.	

0 1 . 2 The DNA base sequence of part of a gene is TACGGATTCAAG
Which sequence was produced from it by an inversion?
Tick (✓) one box.

[1 mark]

TACGGATTCTTCAAG ☐

TACGGACTTAAG ☐

TACGATTCAAG ☐

TACGGATTGAAG ☐

4

Turn over for the next question

Do not write outside the box

0 2 Which statement about mutagenic agents is correct?
Tick (✓) one box.

[1 mark]

Gene mutations cannot happen without a mutagenic agent. ☐

Mutagenic agents increase the rate of gene mutation. ☐

Mutagenic agents only cause frame shifts. ☐

Mutagenic agents only cause mutations in genes that code for enzymes. ☐

1

Turn over for the next question

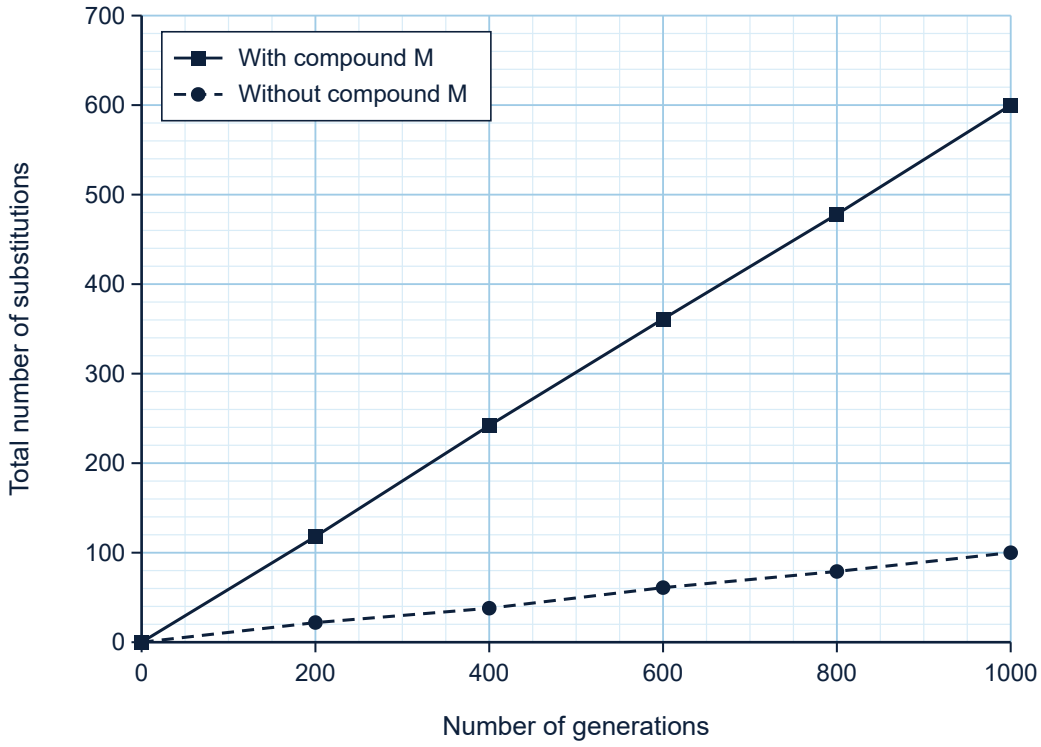
Do not write outside the box

Scientists investigated whether compound M is a mutagenic agent in yeast, a single-celled fungus. They:

- grew a population of yeast from a single cell
- used cells from this population to start 80 separate populations, called lines
- grew 40 lines in a medium containing compound M and 40 lines in the same medium without compound M
- every 200 generations, found the base sequence of all the DNA in one cell from each line
- compared each base sequence with the DNA of the original cell, then identified and counted the gene mutations of each type.

Figure 1 shows the total number of substitutions found in each group of 40 lines.

Figure 1



Question 3 continues on the next page

Do not write outside the box

0 3 . 1 The lines grown without compound M also gained gene mutations.
Explain why.

[2 marks]

0 3 . 2 The DNA of each yeast cell contains 1.2×10^7 base pairs. The mutation rate can be given as the number of substitutions per base pair per generation.
Use Figure 1 to calculate the mutation rate for the lines grown without compound M.
Give your answer in standard form.

[2 marks]

Answer _____ **per base pair per generation**

0 3 . 3 Use Figure 1 to calculate how many times greater the number of substitutions after 1000 generations was in the lines grown with compound M than in the lines grown without it.

[1 mark]

Answer _____ **times**

Question 3 continues on the next page

Do not write outside the box

Table 1 shows the number of each type of gene mutation the scientists found in each group of lines after 1000 generations. Each mutation was counted in one type only.

Table 1

Type of gene mutation	Number found in the 40 lines	
	Without compound M	With compound M
Substitution	100	600
Addition of one or more bases (not a duplication)	4	5
Deletion of one or more bases	7	6
Duplication	2	1
Inversion	1	2

0 3 . 4 The scientists concluded that compound M increases the rate of all types of gene mutation.
Use Table 1 to evaluate this conclusion.

[3 marks]

Question 3 continues on the next page

Do not write outside the box

0 3 . 5 All 80 lines were started from cells that came from one single yeast cell.
Suggest why.

[2 marks]

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0 3 . 6 Suggest why the scientists used 40 lines in each group rather than only one line.

[1 mark]

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11

Turn over for the next question

Do not write outside the box

Effects of gene mutations on the encoded polypeptide 3.8.1

0 4 . 1 Which describes a frame shift?
Tick (✓) one box.

[1 mark]

- Every base triplet downstream of the mutation is changed. ☐
- Only the base triplet that contains the mutation is changed. ☐
- The order of the bases in part of the gene is reversed. ☐
- A base triplet is copied and the copy is inserted next to the original triplet. ☐

0 4 . 2 A gene codes for a polypeptide of 450 amino acids.
Which gene mutation is most likely to change the largest number of amino acids in this polypeptide?
Tick (✓) one box.

[1 mark]

- Deletion of three bases near the start of the gene ☐
- Deletion of one base near the end of the gene ☐
- Addition of one base near the start of the gene ☐
- Substitution of one base near the start of the gene ☐

2

Turn over for the next question

Do not write outside the box

Gene H codes for a polypeptide. Table 2 shows the DNA base sequence of part of gene H and of three mutated forms of this part, **P**, **Q** and **R**. Each sequence is read in triplets, starting from the first base shown.

Table 2

Sequence	DNA base sequence
Normal	GCTTCAGACCTGAAAGGT
P	GCTTCATGACCTGAAAGGT
Q	GCTTCACTGAAAGGT
R	GCTTCACCAGTGAAAGGT

Table 3 shows the amino acid coded for by each DNA base triplet in these sequences.

Table 3

DNA triplet	Amino acid	DNA triplet	Amino acid
AAA	Lysine	GAC	Aspartic acid
AGG	Arginine	GCT	Alanine
CCA	Proline	GGT	Glycine
CCT	Proline	GTG	Valine
CTG	Leucine	TCA	Serine
GAA	Glutamic acid	TGA	Stop (the polypeptide ends)

0 5 . 1 Name the type of gene mutation that produced sequence **P**.

[1 mark]

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Question 5 continues on the next page

Do not write outside the box

0 5 . 2 Mutation **P** and mutation **Q** both change the number of bases in gene H.
Use Table 2 and Table 3 to explain why mutation **P** has a much greater effect on the encoded polypeptide than mutation **Q**.

[4 marks]

0 5 . 3 Sequence **R** was produced by an inversion.
Use Table 2 and Table 3 to describe the effect of this mutation on the encoded polypeptide, and explain why it does not cause a frame shift.

[2 marks]

7

Turn over for the next question

Do not write outside the box

Dystrophin is a protein in muscle cells that helps to hold the muscle fibres together. A deletion in the gene coding for dystrophin can cause muscular dystrophy, in which the muscles become weaker over time.

- In the severe form of muscular dystrophy, the muscle cells contain little or no functional dystrophin.
- In the mild form, the muscle cells contain dystrophin that still works, but less well than normal.

Doctors studied 200 people with muscular dystrophy caused by a deletion in this gene. For each person, they found the number of bases deleted from the exons of the gene. Table 4 shows their results.

Table 4

Number of bases deleted from the exons	Number of people	
	With the severe form	With the mild form
A multiple of three	9	61
Not a multiple of three	118	12

0 6 . 1 Calculate the percentage of the people with a deletion that was **not** a multiple of three who had the severe form.
Give your answer to 1 decimal place.

[2 marks]

Answer _____ **%**

Question 6 continues on the next page

Do not write outside the box

0 6 . 2 Use Table 4 and your knowledge of gene mutations to explain the relationship between the number of bases deleted and the form of muscular dystrophy.

[5 marks]

0 6 . 3 Nine people had the severe form, even though the number of bases deleted was a multiple of three.
Suggest **two** explanations for this.

[2 marks]

1

2

Question 6 continues on the next page

Do not write outside the box

0 6 . 4 Twelve people had the mild form, even though the number of bases deleted was **not** a multiple of three.
Suggest **one** explanation for this.

[1 mark]

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10

Turn over for the next question

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Gene mutations and protein function 3.8.1

Enzyme G in a species of bacterium is a single polypeptide of 340 amino acids. The amino acid at position 172 is part of the active site. No other amino acid listed in Table 5 is part of the active site.

Scientists made four mutated versions of the gene for enzyme G. They purified the enzyme made from each version and measured its initial rate of reaction at 30 °C and pH 7.0. Table 5 shows their results.

Table 5

Version	Mutation in the gene	Change in the polypeptide	Initial rate of reaction / μmol of product min^{-1} mg^{-1} of enzyme
Normal	None	None	8.4
1	Substitution in triplet 172	Aspartic acid replaced by asparagine at position 172	0.3
2	Substitution in triplet 12	Serine replaced by threonine at position 12	8.2
3	Duplication of triplet 250	Extra glycine after position 250	8.3
4	Deletion of triplet 301	Lysine at position 301 missing	5.0

0 7 . 1 Calculate the percentage decrease in the initial rate of reaction of the enzyme from version 4 compared with the normal enzyme.
Give your answer to 1 decimal place.

[2 marks]

Answer ..... **%**

Question 7 continues on the next page

Do not write outside the box

0 7 . 2 Use Table 5 to explain why the enzyme from version 1 has a much lower initial rate of reaction than the enzyme from version 2.

[3 marks]

0 7 . 3 The scientists concluded that changes to the polypeptide that are not in the active site do not affect the activity of enzyme G.
Use Table 5 to evaluate this conclusion.

[3 marks]

0 7 . 4 Give **one** variable, other than temperature and pH, that the scientists should have kept the same when they measured the initial rates of reaction.

[1 mark]

9

Turn over for the next question

Do not write outside the box

0 8 Glucose enters some cells by facilitated diffusion through a carrier protein called GT. GT has a binding site for glucose.
Some gene mutations in the gene coding for GT stop GT from transporting glucose. Other gene mutations in this gene have no effect on glucose transport.
Explain how gene mutations in this gene can have these different effects.

[6 marks]

6

END OF QUESTIONS

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