HELICATE · A-LEVEL BIOLOGY · TOPIC BANK

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

Mark scheme

Version 3 · 17 October 2026

TOTAL MARKS	QUESTIONS
50	8

How to use this mark scheme

;	Separates marking points. Each semicolon marks the end of one creditable point, and each point is worth one mark.
;;	Two marks for this answer (used for calculations).
/	Alternative words or phrases that are credited for the same point.
OR	An alternative answer for the same mark.
( )	Words in brackets are not needed to gain the mark.
max	More marking points are listed than marks available. Award up to the number of marks shown.
Accept	An alternative answer that gains credit.
Reject	A wrong answer. It does not gain credit and cancels a correct point it contradicts.
Ignore	Neither credited nor penalised. Mark the rest of the answer.
ECF	Error carried forward: credit correct working that uses an earlier wrong answer.
AO	Assessment objective: AO1 knowledge and understanding; AO2 applying knowledge; AO3 analysing, interpreting and evaluating.

Where an answer is correct but not listed, credit it if it is biologically accurate and answers the question at the depth the marks require. Right + wrong = wrong: a contradiction cancels a correct point.

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Types of gene mutation and the mutation rate 3.8.1

Question	Marking guidance	Mark	Comments
01.1	1. Row 1: inversion; 2. Row 2: duplication; 3. Row 3: substitution;	**3** (3 × AO1)	2. **Reject** addition (a copy inserted next to the original group is a duplication) 3. **Accept** (single-)base substitution **Ignore** 'gene' or 'mutation' added to the name, eg 'inversion mutation'

Question	Marking guidance	Mark	Comments
01.2	TACGGACTTAAG;	**1** (1 × AO2)	The bases TTC (bases 7–9) are reversed to CTT; the other options show a duplication (TTC copied), a deletion (one G removed) and a substitution (C replaced by G)

Question	Marking guidance	Mark	Comments
02	Mutagenic agents increase the rate of gene mutation;	**1** (1 × AO1)	

Question	Marking guidance	Mark	Comments
03.1	1. Gene mutations occur spontaneously/randomly; 2. (Mutations) arise during DNA replication (before each cell division/in every generation);	**2** (2 × AO1)	1. **Accept** other mutagenic agents in the environment or produced by the cell, eg UV light or chemicals produced in metabolism 1. **Ignore** 'mutations happen without compound M' (repeats the stem) 2. **Accept** errors in copying/base pairing during DNA replication **Reject** the yeast mutate to adapt to the medium OR mutations are caused by selection

Question	Marking guidance	Mark	Comments
03.2	Correct answer of 2.1×10^{-10} for 2 marks;; Accept for 1 mark: $100 \div (40 \times 1000 \times 1.2 \times 10^{7})$ **OR** 2.5×10^{-3} (substitutions per line per generation) **OR** a correct answer not in standard form, eg 0.000 000 000 21 **OR** 8.3×10^{-9} (number of lines not taken into account)	**2** (2 × AO2)	**Accept** 2.08×10^{-10} for 2 marks **Accept** answers that use the gradient of the line (0.1 substitutions per generation in 40 lines)

Question	Marking guidance	Mark	Comments
03.3	6 (times);	**1** (1 × AO2)	600 ÷ 100. Accept 6.0 **Accept** 6 for ratios of gradients of the two lines

Question	Marking guidance	Mark	Comments
03.4	1. (Supports, substitutions only) 600 substitutions with compound M compared with 100 without it OR 6 times as many substitutions; 2. (Against) the numbers of additions, deletions, duplications and inversions are similar with and without compound M, eg 6 and 7 deletions, so M does not appear to increase these types; 3. The numbers of the other types are very small (eg 1 and 2 inversions), so an increase in these types might not be detected; 4. No statistical test/no measure of variation between lines, so not known whether the differences are significant OR differences could be due to chance; 5. Only one concentration of compound M was used, so a higher concentration might increase the other types (the claim about all types is not tested fully);	**3 max** (3 × AO3)	**Max 2** if no data from Table 1 are used 2. **Accept** any correct pair of values from the four rows other than substitution

Question	Marking guidance	Mark	Comments
03.5	1. All the lines start with the same DNA base sequence OR all the cells are genetically identical (at the start); 2. So any differences from the original cell are (new) mutations that arose during the investigation; 3. So differences between the two groups are due to compound M, not to genetic differences between the starting cells;	**2 max** (2 × AO3)	**Ignore** 'to make it a fair test' unqualified

Question	Marking guidance	Mark	Comments
03.6	Gene mutations are rare/there are few mutations in each line, so more lines give more mutations to count OR to give a more reliable/representative (mean) rate OR to reduce the effect of chance differences between lines;	**1** (1 × AO3)	**Ignore** 'more accurate' or 'fair test' unqualified

Effects of gene mutations on the encoded polypeptide 3.8.1

Question	Marking guidance	Mark	Comments
04.1	Every base triplet downstream of the mutation is changed;	**1** (1 × AO1)	

Question	Marking guidance	Mark	Comments
04.2	Addition of one base near the start of the gene;	**1** (1 × AO2)	Adding one base near the start causes a frame shift, so all the base triplets downstream are changed

Question	Marking guidance	Mark	Comments
05.1	Addition (of a base/of T);	**1** (1 × AO2)	**Accept** insertion **Reject** duplication/substitution

Question	Marking guidance	Mark	Comments
05.2	1. (In P) adding one base causes a frame shift, so all the base triplets after/downstream of the mutation are changed; 2. (In P) the third triplet becomes TGA/a stop triplet, so the polypeptide ends after the serine shown OR the polypeptide is terminated early/shorter; 3. (In Q) three bases/one whole triplet (GAC) is deleted, so (only) one amino acid/aspartic acid is missing (from the polypeptide); 4. (In Q) there is no frame shift OR the triplets after the deletion are unchanged, so the rest of the amino acid sequence (leucine, lysine, glycine) is unchanged;	**4** (1 × AO1) (3 × AO2)	**Max 3** if no triplet or amino acid from Table 2 or Table 3 is quoted 1. 'Frame shift' alone does not gain point 1: the idea that all later/downstream triplets are changed is needed 1. and 2. Accept codon(s) for triplet(s) 2. **Accept** premature stop codon 3. **Reject** 'aspartic acid is not produced/formed' **Reject** amino acids produced/formed by the DNA, eg 'different amino acids are formed'

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Question	Marking guidance	Mark	Comments
05.3	1. At least one named change, eg aspartic acid replaced by proline OR leucine replaced by valine (and the rest of the amino acids are the same); 2. The number of bases is unchanged/no bases are added or removed, so the triplets after the inversion (AAA, GGT) are read in the same way OR all later triplets are unchanged;	**2** (2 × AO2)	1. 'Two amino acids change' without naming any change from Table 3 does not gain point 1 1. **Reject** amino acids produced/formed 2. **Accept** codons for triplets 2. **Accept** 'the reading frame is unchanged' only with the idea that no bases are added or removed

Question	Marking guidance	Mark	Comments
06.1	Correct answer of 90.8 (%) for 2 marks;; Accept for 1 mark: 118 ÷ 130 **OR** a correct answer not given to 1 decimal place, eg 90.77 or 91	**2** (2 × AO2)	**Reject** 59.0 (÷ 200) for 2 marks

Question	Marking guidance	Mark	Comments
06.2	1. Deleting a number of bases that is not a multiple of three causes a frame shift, so all the base triplets downstream of the deletion are changed; 2. (So) the amino acids after the deletion are different OR a premature stop triplet/codon is formed, so the polypeptide is shorter/terminated; 3. (So) dystrophin has a different tertiary structure/cannot function, giving the severe form (in 118 of 130 people); 4. Deleting a multiple of three bases removes whole triplets/does not cause a frame shift, so the triplets after the deletion are unchanged; 5. (So) only a few amino acids are missing and the rest of the amino acid sequence/primary structure is unchanged; 6. (So) dystrophin is shorter but still (partly) functional, giving the mild form (in 61 of 70 people);	**5 max** (2 × AO1) (3 × AO2)	**Max 3** if only one type of deletion is explained **Max 4** if no data from Table 4 are used, eg 118 of 130 or 91% severe; 61 of 70 or 87% mild 1. 'Frame shift' alone does not gain point 1: the idea that all later/downstream triplets are changed is needed 1. and 4. Accept codons for triplets 3. **Accept** 'little or no functional dystrophin' 3. **Reject** 'no protein is produced'. Accept a shorter/non-functional polypeptide **Reject** amino acids produced/formed by the gene **Reject** '3°' for tertiary

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Question	Marking guidance	Mark	Comments
06.3	1. The missing amino acids are in a part of dystrophin that is essential for its function/for binding OR their loss changes the (bonds and) tertiary structure, so dystrophin cannot function; 2. A large number of bases/amino acids is deleted (so much of the protein is missing); 3. The deletion removes parts of two triplets, forming a new triplet that is a stop triplet (so the polypeptide is shorter); 4. The person has another mutation (in the dystrophin gene or in another gene) that affects the muscles;	**2 max** (2 × AO3)	1. Bonds need not be named for this point **Ignore** environmental factors unqualified

Question	Marking guidance	Mark	Comments
06.4	A second mutation that adds bases (an addition) makes the total change a multiple of three/restores the reading frame, so there is no frame shift beyond it;	**1** (1 × AO3)	**Accept** duplication (it also adds bases) **Reject** 'the deletion is in an intron' (the deletion is in the exons) **Reject** a deletion near the end of the gene

Gene mutations and protein function 3.8.1

Question	Marking guidance	Mark	Comments
07.1	Correct answer of 40.5 (%) for 2 marks;; Accept for 1 mark: (8.4 − 5.0) ÷ 8.4 **OR** 59.5 (%) (version 4 as a percentage of normal) **OR** a correct answer not given to 1 decimal place, eg 40.48 or 40	**2** (2 × AO2)	**Reject** 68.0 (÷ 5.0) for 2 marks

Question	Marking guidance	Mark	Comments
07.2	1. In version 1 the amino acid changed is in the active site/at position 172; 2. (In version 1) the R group of aspartic acid is replaced by that of asparagine, so an ionic/hydrogen bond in the active site is lost/changed; 3. (So) the active site changes shape/the tertiary structure changes, so the substrate is no longer complementary OR fewer enzyme-substrate complexes form; 4. In version 2 the amino acid changed is not in the active site/is at position 12, so the shape of the active site/tertiary structure is (largely) unchanged;	**3 max** (1 × AO1) (2 × AO2)	**Max 2** if version 2 is not addressed (point 4) 2. Bonds must be named. Ignore 'bonds in different places' unqualified 2. **Reject** a bonding change described for both versions 2. **Accept** H bonds for hydrogen bonds 3. **Reject** '3°' for tertiary. Ignore quaternary/3D structure 3. **Accept** E-S complex / ES complex. E-S alone only if 'enzyme-substrate complex' is written elsewhere. Ignore ESC **Reject** amino acids produced/formed

Question	Marking guidance	Mark	Comments
07.3	1. (Supports) versions 2 and 3 change the polypeptide outside the active site, and their rates (8.2 and 8.3) are similar to normal (8.4); 2. (Against) version 4 changes the polypeptide outside the active site but its rate is reduced (to 5.0/by about 40%); 3. A missing amino acid may change bonds/folding elsewhere in the polypeptide, so the tertiary structure/active site shape changes; 4. Amino acids far apart in the polypeptide/primary structure may be close to the active site in the tertiary structure; 5. Only one mutation at each position/only four versions were tested OR no repeats/no measure of variation, so not known whether small differences (eg 8.2 and 8.4) are significant;	**3 max** (3 × AO3)	**Max 2** if no data from Table 5 are used **Max 2** if only points that support or only points that are against the conclusion are given 3. Bonds need not be named for this point

Question	Marking guidance	Mark	Comments
07.4	Concentration of substrate OR volume of the reaction mixture OR concentration/mass of enzyme;	**1** (1 × AO3)	**Ignore** 'amount of substrate' unqualified **Ignore** the version of the gene (the independent variable)

Question	Marking guidance	Mark	Comments
08	1. (Gene mutation is a) change in the DNA base sequence OR a named mutation, eg addition/deletion/inversion of bases; 2. Addition/deletion (of bases, not a multiple of three) causes a frame shift, so all base triplets downstream are changed; 3. A (premature) stop triplet/codon may form, so the polypeptide is shorter/terminated; 4. Change in the amino acid sequence/primary structure (of GT); 5. Change in (the positions of) hydrogen/ionic/disulfide bonds; 6. Change in the tertiary structure (of GT); 7. (So) the binding site changes shape/is no longer complementary to glucose, so glucose cannot bind/be transported; 8. No effect: the genetic code is degenerate, so the same amino acid is coded for; 9. No effect: a changed amino acid does not change the tertiary structure/binding site, eg it has a similar R group or is far from the binding site; 10. No effect: the mutation is in an intron/non-coding DNA;	**6 max** (5 × AO1) (1 × AO2)	**Max 4** if none of points 8–10 is given **Max 4** if none of points 2–7 is given 1. 'DNA' must be stated. Reject a change in the amino acid sequence for point 1 2. 'Frame shift' alone does not gain point 2: the idea that all later/downstream triplets are changed is needed 2. and 3. Accept codons for triplets 5. Bonds must be named. Accept H bonds for hydrogen bonds 6. **Reject** '3°' for tertiary. Ignore quaternary/3D structure 7. **Reject** active site (GT is not an enzyme) 8. **Accept** a description of degenerate, eg some amino acids are coded for by more than one triplet. Reject 'DNA is degenerate' OR 'degenerate code of amino acids' **Reject** amino acids produced/formed by the DNA

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