1.8 The Control of Gene Expression: 30-minute Pack

AQA 7402 · Calculator allowed · about 30 minutes

Total Marks

Name: _____ Date: ____ / ____ / _____

Answer ALL questions. Show all your working.

1 Gene mutations occur when the base sequence of DNA is changed.

(a) A single base substitution changes one DNA triplet but, because of the degenerate nature of the genetic code, the amino acid coded for does not change. Which term correctly describes this type of mutation? **(1)**

- ☐ A) Nonsense mutation
- ☐ B) Silent mutation
- ☐ C) Frameshift mutation
- ☐ D) Missense mutation

(b) Explain why a base deletion is more likely than a single base substitution to have a significant effect on the polypeptide produced. **(3)**

(c) State one reason why a mutation occurring within an intron is unlikely to affect the amino acid sequence of the final protein. **(1)**

(Total for Question 1 is 5 marks)

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2 Epigenetics refers to heritable changes in gene expression that occur without a change in the base sequence of DNA. Two epigenetic mechanisms are DNA methylation and histone modification.

(a) Define the term "epigenetics". **(1)**

(b) Describe how increased methylation of DNA can silence a gene. **(3)**

(c) Acetylation of histone proteins generally increases the rate of transcription of associated genes. Explain how this occurs. **(3)**

(d) During development, cells with an identical genome become specialised into different cell types (differentiation). Explain how epigenetic mechanisms allow this to happen. **(2)**

(Total for Question 2 is 9 marks)

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3 This question is based on a core practical technique used in genetic profiling. Restriction enzymes were used to cut DNA samples into fragments, which were then separated using gel electrophoresis. Because the relationship between fragment size and migration distance is not linear, a calibration graph is produced by plotting log10(fragment size in base pairs, bp) against migration distance (mm) for DNA fragments of known size. Two calibration points obtained were: a 100 bp fragment migrated 52 mm, and a 1000 bp fragment migrated 18 mm.

(a) Describe how DNA fragments are separated by gel electrophoresis, referring to charge and size. **(2)**

(b) Show that the gradient of the calibration line, plotting log10(size/bp) against migration distance (mm), is -0.0294 (to 3 significant figures), using the two calibration points given. **(2)**

(c) Calculate the size, in bp, of an unknown DNA fragment that migrated 40 mm on the same gel. Give your answer to 3 significant figures. **(3)**

(d) Explain why, when comparing DNA fingerprints from two different samples, both samples must be cut using the same restriction enzyme before electrophoresis. **(2)**

(Total for Question 3 is 9 marks)

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Mark scheme · 1.8 The Control of Gene Expression: 30-minute Pack

Question 1

(a) B cao **B1**

(a) Answer: B) Silent mutation

(b) a deletion causes a frameshift: the reading frame (grouping into triplets/codons) is shifted from the point of the mutation onwards oe **B1**

(b) every subsequent codon is changed, which may alter every amino acid after the mutation and/or create a premature stop codon oe **B1**

(b) a substitution only ever affects one triplet/codon, so at most only one amino acid changes, and due to the degenerate code it may not change the amino acid (and therefore the protein) at all oe **B1**

(b) Answer: A deletion shifts the reading frame so that every codon after the mutation is altered, whereas a substitution changes at most one codon/amino acid and, because the code is degenerate, may have no effect at all.

(c) introns do not code for amino acids and are removed (spliced out) from the pre-mRNA before translation, so a mutation there does not appear in the mature mRNA/protein oe **B1**

(c) Answer: Introns are removed by splicing before translation, so a mutation within an intron is not present in the mature mRNA and does not affect the protein.

Question 2

(a) heritable changes in gene function/expression that do not involve a change to the base/nucleotide sequence of DNA oe **B1**

(a) Answer: Epigenetics is the study of heritable changes in gene expression that occur without any change to the base sequence of DNA.

(b) methyl groups are added to cytosine bases in the DNA, often in the promoter region of a gene oe **B1**

(b) increased methylation physically prevents transcription factors and/or RNA polymerase from binding to the DNA, and/or attracts proteins that cause the chromatin to condense (heterochromatin formation) oe **B1**

(b) transcription of the gene is inhibited/prevented, so no mRNA (and therefore no protein) is produced from that gene oe **B1**

(b) Answer: Methyl groups added to cytosine bases (often in the promoter) block transcription factor/RNA polymerase binding and can condense chromatin structure, preventing transcription of the gene.

(c) acetyl groups added to histone proteins reduce the overall positive charge on the histones oe **M1**

(c) this reduces the strength of the (electrostatic/ionic) association between the (negatively charged) DNA and the histones oe **M1**

(c) chromatin becomes less condensed (more open/euchromatin), making the DNA more accessible to transcription factors/RNA polymerase, increasing the rate of transcription oe **A1**

(c) Answer: Acetylation reduces the positive charge on histones, loosening the DNA-histone association so chromatin becomes less condensed; this makes the gene more accessible to transcription factors/RNA polymerase, increasing transcription.

(d) different patterns of DNA methylation and/or histone modification are established in different cell lineages during development, permanently switching off genes not needed in that cell type oe **B1**

(d) only the genes needed for a particular cell's specialised function remain able to be transcribed/expressed, so cells with the same genome develop different structures and functions oe **B1**

(d) Answer: Different epigenetic marks (methylation/histone modification patterns) are established in different cell lineages, permanently silencing genes not needed for that cell's role, so only a subset of genes remains expressed, producing specialised cells from an identical genome.

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Question 3

(a) DNA fragments are negatively charged (due to phosphate groups) and are loaded into wells in an agarose gel; a (direct) electric current is applied across the gel oe **B1**

(a) fragments move towards the positive electrode (anode); smaller fragments move further/faster through the pores of the gel than larger fragments in a given time, separating the fragments by size oe **B1**

(a) Answer: DNA fragments, being negatively charged, migrate towards the anode when a current is applied; smaller fragments move further through the gel than larger fragments in the same time, separating them by size.

(b) correct substitution: gradient = (log10(100) - log10(1000)) / (52 - 18) = (2 - 3) / 34 shown **M1**

(b) gradient = -1/34 = -0.0294 (3 sf) cso **A1**

(b) Answer: -0.0294 log10(bp) per mm

(c) correct intercept found: c = 3 - (-0.0294 x 18) = 3.53 (3 sf) oe (or equivalent method using either calibration point) **M1**

(c) log10(size) = (-0.0294 x 40) + 3.53 = 2.35 (3 sf) shown **M1**

(c) size = $10^{2.35}$ = 225 bp (awrt 220-230 bp) cao **A1**

(c) Answer: Approximately 225 bp

(d) the same restriction enzyme cuts DNA at the same specific (recognition) base sequence in both samples oe **B1**

(d) this produces comparable/corresponding fragments in each sample, so differences in the resulting banding patterns reflect real differences in the DNA sequence between the samples, allowing valid comparison oe **B1**

(d) Answer: Using the same restriction enzyme ensures both samples are cut at the same recognition sequences, producing comparable fragments so that any differences in the banding pattern are due to genuine sequence differences between the samples.

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