Assessment Schedule – 2025

Biology: Demonstrate understanding of gene expression (91159)

Assessment Criteria

Achievement	Achievement with Merit	Achievement with Excellence
Demonstrate ***understanding*** involves: • defining, using annotated diagrams or models to explain, and giving characteristics of, or an account of, gene expression.	*Demonstrate* ***in-depth understanding*** involves: • providing reasons how or why biological ideas and processes affect gene expression.	*Demonstrate* ***comprehensive understanding*** involves: • linking biological ideas and processes about gene expression; explanations may involve justifying, relating, evaluating, comparing, and contrasting, or analysing.

Cut Scores

Not Achieved	Achievement	Achievement with Merit	Achievement with Excellence
0–7	8–13	14–18	19–24

Evidence

Question One

<table>
<tr><th>Expected Coverage</th><th>Achievement</th><th>Achievement with Merit</th><th>Achievement with Excellence</th></tr>
<tr><td>(a)
<table>
<tr><th></th><th>Normal sequence</th><th>Mutated sequence 1</th><th>Mutated sequence 2</th></tr>
<tr><td>DNA template strand</td><td>TTA TGC AAT CCG</td><td>TTA TGC GAT CCG</td><td>TTA TGC AAG CCG</td></tr>
<tr><td>mRNA</td><td>AAU ACG UUA GGC</td><td>AAU ACG CUA GGC</td><td>AAU ACG UUC GGC</td></tr>
<tr><td>Amino acid</td><td>Asn Thr Leu Gly</td><td>Asn Thr Leu Gly</td><td>Asn Thr Phe Gly</td></tr>
</table></td><td>One Achievement point for either correct row OR column.</td><td></td><td></td></tr>
<tr><td>(b)
Both mutations are classified as point substitution mutations, which involve the substitution of a single nucleotide in the DNA sequence.
Substitution: as only one base / nucleotide has been (permanently changed) on the DNA.
A mutation is a permanent change in the DNA (sequence) / genetic code / gene.
Same-sense / silent: one base is swapped but doesn't change the amino acid.
Missense: one base is swapped and changes one amino acid.
Nonsense: one base is changed and changes into a stop codon.
Degeneracy / redundancy of the code means that an amino acid will usually have more than one codon / triplet that codes for it.
Point mutation 1 involves the substitution of A for G in the third codon of the sequence, resulting in the same amino acid (Leu) due to the degeneracy of the genetic code. This type of mutation is termed a silent mutation as it does not alter the protein's amino acid sequence. The absence of a change in the amino acid means that the CFTR protein keeps its shape and remains functional.</td><td>Identifies:
• substitution / describes a substitution / point mutation
• identifies that no frame shift has occurred / no nonsense mutation / no change in the start / stop codon / no change in length of gene.

Describes:
• substitution mutation is less severe than a frameshift
• a mutation as a permanent change mutation in the DNA bases
• mutation 1 is least severe (compared to frameshift) / no effect on final protein compared to other mutation</td><td>Explains:
• point mutation 1 is a substitution and identifies no change in amino acid sequence
• point mutation 2 is a substitution mutation and identifies the change in one amino acid (Leu-Phe)
OR
point mutation 2 is a substitution causes only small change in the polypeptide chain, i.e. a small change in overall function)
• point mutation 1 (the substitution makes no change due to degeneracy in the genetic code)
• degeneracy and how it relates back to either the triplet / codon coding for the same amino acid</td><td>Provides detailed discussion of:
• mutation 1 the effect of the mutation on the amino acid sequence and the functioning of the final CFTR protein and no symptoms
AND
mutation 2 the effect of the mutation on the amino acid sequence and the functioning of the final CFTR protein and symptoms occur
• mutation 2 is a substitution mutation therefore less severe than a frameshift because an insertion / deletion causes a lot of aa downstream of the mutation to change, causing the length / aa number / position of stop codon to change significantly, causing the shape</td></tr>
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Expected Coverage	Achievement	Achievement with Merit	Achievement with Excellence
Point mutation 2 involves a different substitution where A is replaced by U in the third codon, resulting in a phenylalanine (Phe) instead of leucine (Leu). This change alters the final amino acid sequence, potentially affecting the folding and functionality of the CFTR protein. The change from Leu to Phe can lead to a misfolded protein, with an altered shaped that cannot function correctly. This mutation shows that a single-point substitution nucleotide change can significantly impact protein function AND that single-point substitutions can have an effect on protein shape and function. In contrast, frame shift mutations, which involve the insertion or deletion of nucleotides, can lead to more drastic changes in the protein, often resulting in a completely non-functional protein or a protein truncated by a premature stop codon (nonsense mutation). Insertion / deletion mutation cause a frameshift mutation because they both change the number of bases / length of a gene, either increasing or decreasing the DNA sequence. By adding or removing a single base, every codon / triplet from the point of the mutation is affected (reading frameshift), which would therefore create different codons and different amino acid sequence. The frameshift could also alter start / stop codons, which affects the length of the amino acid sequence and thus the structure / folding of the final protein. Insertion and deletion mutation affect the final protein the most / more than substitution mutations. This is because a substitution may only affect one triplet / codon, and therefore only one amino acid in the polypeptide chain. If only one amino acid is changed, this will change the shape of the protein; however, this change might only be slight. In contrast, insertion and deletion mutations may result in all the amino acids after the mutation to be incorrect. This will result in the final protein folding differently. This drastic change in shape will render the protein completely non-functional. These mutations may also change the position of the stop triplet / codon either by being too early and terminating the polypeptide chain early / causing a shorter polypeptide chain or too late extending the polypeptide chain. **Magnitude of impact:** The key distinction is that point substitution mutations often produce no change, or minor or specific changes in protein function; however, frameshift mutations can lead to extensive changes that disrupt the protein's overall structure and function.	• mutation 2 effect minor severity (compared to frameshift) / effects the final protein compared to another mutation • mutation 1 as a silent / same-sense mutation / same amino acid produced (Leu produced even though mutation) / protein will function • mutation 2 as missense mutation / change in one amino acid (Phe produced instead of Leu) / protein will not function • degeneracy / redundancy but not linked to context.	(e.g. GAT/CUA and ATT/UUA code for the same aa (M4)). • there is no insertion / deletion, so no reading frame shift has occurred that would affect all amino acids downstream / so a large change in the polypeptide chain OR if an insertion / deletion were to occur there would be a frameshift so that would affect all the aa downstream / so a large change in the polypeptide chain • no change to stop codon so amino acids number / length the same / protein length the same OR nonsense mutation / change to stop codon (due to frame shift so amino acid number / length would change a lot) / protein length changed (M6).	to be different, no functional protein and more severe symptoms.

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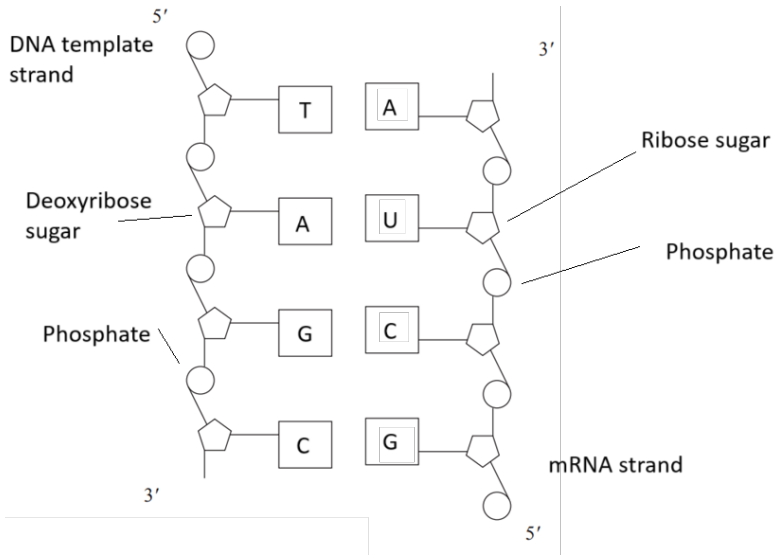
The functional ramifications of these mutations are significant for cystic fibrosis. A properly functioning CFTR protein is critical for maintaining ion and fluid balance in epithelial cells. With Point mutation 1, the amino acid sequence means that the CFTR protein can still regulate salt and water transport effectively, showing no symptoms of cystic fibrosis. However, Point mutation 2 causes aa phenylalanine instead of leucine, which hinders the protein's ability to function correctly. This causes the symptoms of cystic fibrosis, such as thick mucus in the lungs and digestive tract.			

N1	N2	A3	A4	M5	M6	E7	E8
ONE evidence point at Achievement.	TWO evidence points at Achievement.	THREE evidence points at Achievement.	FOUR evidence points at Achievement.	TWO evidence points at Merit.	THREE evidence points at Merit.	ONE evidence point at Excellence.	TWO evidence points at Excellence.

N0 = No response; no relevant evidence.

Question Two

Expected Coverage	Achievement	Achievement with Merit	Achievement with Excellence
(a)  	• All correct base pairing rule shown between DNA-mRNA / base pairing rules described in b). • Labels correctly TWO of the following on the diagram: - deoxyribose sugar and ribose sugar - DNA strand and mRNA strand - phosphate on DNA strand and phosphate on mRNA strand - 5' 3' on DNA strand and 3' 5' on mRNA strand.	Full diagram labelled correctly and base pairing rule correct.	
(b) Codon: the sequence of three (consecutive) bases/nucleotides on the mRNA strand. Anticodon: three (consecutive) bases on a tRNA molecule. mRNA structure: a single-stranded short molecule made up of phosphate, ribose sugar, and nitrogen bases A, U, G, and C. mRNA function: carries the code / gene / genetic information out of the nucleus. tRNA structure has an anticodon (3 bases) and an amino acid. tRNA function: carries the amino acid to the ribosome and 'drops' it off (for polypeptide chain). Translation is described: mRNA is used to create a (functional) protein / polypeptide chain. Transcription is the first step in this process. It occurs in the nucleus, where a specific segment of DNA, containing the code for a protein, is	Describes: • a codon • an anticodon • tRNA structure • mRNA structure • transcription (start and end point) • ONE reason why DNA strand not directly translated into a polypeptide chain (valid reason) • translation (start and end point) • tRNA function • mRNA function.	Explains: • process of transcription, including start place in the strand (e.g. codon / promotor sequence, base pairing rule forming mRNA strand, idea of elongation, and a stop place in the strand, e.g. stop codon / terminator sequence, occurs in nucleus) • translation, including that it occurs at ribosomes, start place, e.g. codon, idea of amino dropped off / polypeptide formation, stop place, e.g. codon idea) • triplets / codons determine the correct / match / corresponding /	Provides detailed discussion of: • the relationship between codons, anticodons, tRNA, mRNA, and amino acids (translation) (*must have idea of specific aa*) • the importance of transcription and translation and relationship • TWO reasons DNA is not directly translated into a polypeptide chain AND explanation of transcription.

Expected Coverage	Achievement	Achievement with Merit	Achievement with Excellence
transcribed into messenger RNA (mRNA). This is necessary because DNA, being double-stranded and located in the nucleus, cannot directly participate in protein synthesis. During transcription, the DNA strand is opened, and an enzyme reads the DNA's nucleotide triplets (called triplets) and transcribes them into complementary mRNA codons. These codons are sets of three nucleotides that correspond to specific amino acids. This process ensures that the genetic code can be safely transported from the nucleus to the cytoplasm, where the ribosomes are located. Translation is the next phase, which occurs in the cytoplasm, in the ribosomes. Here, the mRNA is read codon by codon. Each codon on the mRNA corresponds to a specific amino acid. tRNA molecules carry amino acids to the ribosome, with each tRNA containing an anticodon – a set of three nucleotides complementary to the mRNA codons. As the ribosome reads the mRNA, the tRNA molecules bind to the corresponding codons via their anticodons, ensuring that the amino acids are placed in the correct sequence. This sequence of amino acids forms the polypeptide chain, which eventually folds into a functional protein. The role of transcription is to convert the genetic information in the DNA into a transportable form / message, the mRNA. This process allows the information to exit the nucleus and be used in protein synthesis. Without transcription, the DNA code would remain locked in the nucleus and could not be utilised by the ribosomes to produce proteins. The role of translation is to interpret the code carried by the mRNA and ensure the correct sequence of amino acids into a polypeptide chain. The ribosome, tRNA, and mRNA work together to decode the genetic information and link the amino acids in the proper order, forming a protein. The importance of translation and transcription: • Translation of the mRNA template converts nucleotide-based genetic information into amino acids chains to create a polypeptide chain / actual protein. • Without translation, the correct order (sequence) of amino acids would not occur and the polypeptide / protein would not fold into a specific 3-D shape. • Transcription converts DNA code into a 'usable' form (mRNA).		specific amino acids in the polypeptide chain • the complementary nature (correctly used in the answer anywhere) of DNA and mRNA or of mRNA and tRNA • C binds with G and T / U with A due to the number (2 and the 3) of hydrogen bonds (*accept on Q2a diagram but must label H-bonds*) *OR* C binds with G and A binds with T / U because of the size of the bases; double ring purines can bind only with single ring pyrimidines • the importance of transcription / translation. • TWO reasons why DNA strand not directly translated into a polypeptide chain (*valid reason linked to something correct*).	

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• Transcription ensures only the correct / specific protein is made because only one gene is transcribed at a time. • Transcription 'sets' the sequence for translation. The order of bases on mRNA determines order of aa. Translation uses mRNA molecule to make aa sequence. Reasons why a polypeptide chain is not directly translated from the DNA strand: • Ribosomes are used to make polypeptide chains, and are not found in the nucleus / ribosome cannot bind to DNA. • Ribosomes are capable of translating only single-stranded mRNA. • DNA is only one copy of the gene but a cell can produce many mRNA via transcription; therefore many copies of the same gene / protein in response to cell demands. If translation was to occur in the nucleus directly from the DNA template strand, it would be slow, as only one molecule of protein could be produced at a time by each cell, as there is only one copy of the needed DNA. • As proteins are large molecules, these may not be able to leave the nucleus, as they would be too large to pass through the pores of the nuclear membrane. • Maintain DNA integrity (keep it safe from damage). Blueprint, working copy of mRNA created with genetic information / DNA cannot leave the nucleus because it will get broken down / damaged / destroyed by enzymes in the cytoplasm.			

N1	N2	A3	A4	M5	M6	E7	E8
ONE evidence point at Achievement.	TWO evidence points at Achievement.	THREE evidence points at Achievement.	FOUR evidence points at Achievement.	THREE evidence points at Merit.	FOUR evidence points at Merit.	ONE evidence point at Excellence.	TWO evidence points at Excellence.

N0 = No response; no relevant evidence.

Question Three

Expected Coverage	Achievement	Achievement with Merit	Achievement with Excellence
An enzyme is a biological catalyst made of protein / protein that speed up a chemical reaction. A metabolic pathway is a series of chemical reactions where the product of one reaction is the substrate for the next. In a metabolic pathway, one gene codes for one enzyme. A gene is a section of DNA that codes for a protein / enzyme / trait. A substrate is a substance that an enzyme 'acts' on / binds with (to produce a product). Each enzyme can catalyse only one specific reaction due to its unique shape. The process involves several enzymes that convert glutamate into an intermediate and into chlorophyll, the pigment responsible for the green colour of leaves. Gene 1 encodes enzyme 1, which catalyses the conversion of glutamate (substrate) into an intermediate product. Gene 2 encodes enzyme 2, which further converts the intermediate product into chlorophyll (final product) if magnesium is present. DNA mutations in the gene 1 or gene 2 coding for enzyme 1 and enzyme 2 can cause chlorosis. If gene 1 is mutated, enzyme 1 may be non-functional or incorrectly shaped, preventing the conversion of glutamate into the intermediate substrate. This stops the first step, leading to a lack of chlorophyll production and to yellowing of leaves. If gene 2 is mutated, enzyme 2 could be non-functional or incorrectly shaped so cannot catalyse the final conversion into chlorophyll. The intermediate substrates build up, also causing chlorosis. Magnesium is an essential nutrient / co-factor / co-enzyme in the conversion of intermediates into chlorophyll. Without sufficient magnesium, enzyme 2 cannot convert the intermediate product into chlorophyll. This prevents chlorophyll production, resulting in chlorosis where leaves turn yellow.	Describes: • a metabolic pathway (series / linked idea) • an enzyme (*have to linked to biology not generic*) • the relationship between genes and enzymes / how one gene codes for one enzyme or a specific enzyme OR definition of a gene • that gene 1, coding / makes for enzyme 1, / gene 2, coding / makes for enzyme 2 • for chlorophyll to be produced, both genes / enzymes need to be correct / functional/no mutations • the intermediate substrate to be produced gene 1 / enzyme 1 needs to be correct / functional *OR* a mutation to gene 1 will result in no intermediate substrate being formed (particle pathway described) *OR* that for chlorophyll to be produced, gene 2 / enzyme 2 needs to be correct / functional *OR* a mutation to gene 2 will result in no chlorophyll being formed • lots / enough Mg makes chlorophyll (*do not accept opposite, e.g. low Mg because given in source material*)	Explains: • the relationship between two genes, and two enzymes, and three molecules for this specific metabolic pathway; need to identify gene codes for enzymes • how low magnesium would prevent enzyme 2 from 'working' correctly / OR high / enough Mg enables enzyme 2 to work correctly / function (*enzyme 2 needs to be linked to Mg specifically and its functionality*) • gene 1 or gene 2 mutated, causing enzyme 1 or 2 not to 'work' / function therefore no chlorophyll made / leaves are yellow / chlorosis (need to understand gene 1/2 codes for enzyme 1/2) • normal genotype / no mutation interacts with enough/high Mg (environment) to make green leaves OR normal genotype / no mutation interactions with low Mg to make yellow leaves • mutation in either gene, it does not matter how much Mg is available, yellow leave will still be produced.	Provides detailed discussion of: • the specific metabolic pathway, including how chlorosis can be caused by mutations in gene 1 or gene 2 or both • the specific metabolic pathway, including how chlorosis can be caused by magnesium deficiency • the specific metabolic pathway discussed and interaction between genotype, environment, and phenotype identified linked to context.

Expected Coverage	Achievement	Achievement with Merit	Achievement with Excellence
The plants genotype interacts with the environmental factor of Mg to produce a specific phenotype of green leaves / genotype + environment = phenotype.	• gene + environment ⇨ phenotype *OR* genes / enzymes working correctly but low Mg causes yellow leaves / chlorosis • a substrate • final product as chlorophyll / define final product as end point of metabolic pathway.		

N1	N2	A3	A4	M5	M6	E7	E8
ONE evidence point at Achievement.	TWO evidence points at Achievement.	THREE evidence points at Achievement.	FOUR evidence points at Achievement.	TWO evidence points at Merit.	THREE evidence points at Merit.	ONE evidence point at Excellence.	TWO evidence points at Excellence.

N0 = No response; no relevant evidence.