# Assessment Schedule – 2018

## Biology: Demonstrate understanding of gene expression (91159)

### Evidence Statement

| Q | Expected Coverage | Achievement | Merit | Excellence |
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| ONE (a) | Triplet: 3 consecutive bases on DNA strand.<br>Codon: 3 consecutive bases on mRNA strand. | • Describes triplet **OR** codon. | | |

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| (b) | The point mutations that cause a frameshift are insertion and deletion. An insertion is when a base is inserted into the DNA sequence, and a deletion is when a base is taken out / removed from the DNA sequence. Both cause a frameshift mutation because they both change the reading frame / number of bases in a gene, either increasing or decreasing the DNA sequence.<br>mRNA: 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 on the mRNA. Hence, the reading of codons on the mRNA after the mutation will code for different amino acids. In addition, the frameshift could alter start / stop codons, which affects the length of the polypeptide and thus the structure of the protein.<br>A substitution mutation causes a silent mutation. Substitution mutation is where a single nucleotide is changed / swapped, which may / may not result in a codon that codes for a different amino acid. The swapped base may still code for the same amino acid due to redundancy in the code. The length / number of bases in a gene and the mRNA remains the same, therefore the start and stop codons will still function. The final protein may still be the same and carry out the same function, therefore the mutation is silent.<br>Translation: if an insertion or deletion mutation has occurred on the DNA sequence, this will change the codons on the mRNA and the reading frame, resulting in a completely different translation. tRNA anticodon will match with an incorrect codon and 'deliver' the incorrect amino acids from the mutation onwards. The first stop codon in the sequence will also be changed causing the polypeptide chain to be longer or shorter than normal. The final protein will be non-functional, because the entire amino acid sequence from the insertion / deletion mutation onwards is incorrect, therefore causing the folding of the final protein to be abnormal and non-functioning.<br>A substitution mutation / silent mutation may still 'deliver' the correct amino acid during translation, so the amino sequence remains the same length and same order, resulting in the final protein being able to fold correctly and carry out its specific function. This is due to redundancy / degeneracy in the code in which more than one codon codes for an amino acid. | • Briefly describes a frameshift.<br>E.g. a frameshift causes the bases to move.<br>• Identifies insertion **OR** deletion mutations as causing frameshifts **or** defines insertion or deletion.<br>• Identifies substitution mutations as causing a silent mutation **or** defines a substitution mutation<br>• Describes a silent mutation.<br>• Describes a point mutation.<br>• Describes an effect of a mutation on transcription.<br>• Describes an effect of a mutation on translation.<br>• Describes a frameshift as changing the position of stop codon. | • Explains how a frameshift / insertion / deletion affects the mRNA produced.<br>• Explains how a silent mutation affects the mRNA produced.<br>• Explains how a frameshift affects translation (ie amino acid / polypeptide chain length).<br>• Explains how a silent mutation affects translation (ie amino acid / polypeptide chain).<br>• Explains how a silent mutation links to degeneracy / redundancy of the code. | • Comprehensively shows understanding of the consequences of frameshift mutations during protein synthesis **and** its effects on the final protein shape / folding **and** function (must include change in stop codon position).<br>• Comprehensively shows understanding of the consequences of silent mutation during protein synthesis **and** its effects on the final protein shape / folding and function. |

| NØ | N1 | N2 | A3 | A4 | M5 | M6 | E7 | E8 |
|---|---|---|---|---|---|---|---|---|
| No response; no relevant evidence. | Provides any partial point statement from Achievement column. | Provides any ONE statement from Achievement column. | Provides any THREE statements from Achievement column. | Provides FOUR statements from Achievement column. | Provides any TWO statements from Merit column. | Provides any THREE statements from Merit column. | Provides the criteria for Excellence for ONE bullet point. | Provides the criteria for Excellence for BOTH bullet points. |

| Q | Expected Coverage | Achievement | Merit | Excellence |
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| TWO | Cystic fibrosis is inherited / chronic obstructive pulmonary disease is not inherited.<br>Mutation is a permanent change in the base sequence of a gene, altering an organism's genotype. This could result in a different amino acid(s) being coded for and thus the resulting protein to be altered in shape.<br>Environmental factor can be an internal or external factor that affects the organism's phenotype. It does not change the DNA sequence / does not affect genotype but alters the expression (transcription / translation) of genes.<br>The genotype of an organism and environmental conditions interact to determine the phenotype. The CF phenotype exists because a person possesses a mutation on the CFTR gene, and therefore this gene does not produce a fully functional protein and expresses the CF phenotype. This occurs regardless of the environmental factor, because the genotype is mutated, and therefore protein synthesis will always produce a non-functional protein. This is also the reason why CF cannot be cured, and an individual will always express the CF phenotype. CF can be inherited because the mutation has occurred in the genotype. When the germ line cell divides to create gametes, the mutated gene can be passed on via a chromosome into the gametes.<br><br>The COPD phenotype exists due to the environment. A COPD person does not have a mutation (has a normal genotype), therefore produces a functional CFTR protein. However, the environmental factor of coal dust and pollution interacts with the normal genotype to produce excess mucus in response to the coal dust. Hence, when the environmental factor is taken away the phenotype reverts to producing normal amounts of mucus and the individual is cured. Changes in phenotype due to environmental factors are not inherited because the DNA sequence / genotype in the gametes (or in any somatic cell) is not changed. | • Identifies CF is inherited, or COPD is not inherited.<br>• Describes a mutation and an environmental factor.<br>• Describes: genotype + environment = phenotype.<br>• Describes CF caused by genotype / mutation.<br>• Describes COPD caused by environmental factors.<br>• Describes why CF cannot be cured<br>• Describes why COPD can be cured. | • Explains how a mutation affects the genotype by linking it to DNA base sequence, amino acid or protein function.<br>• Explains how an environmental factor affects the genotype and phenotype.<br>• Uses CF or COPD example to explain how mutations and environmental factors cause phenotype, or another named example.<br>• Explains with reason why CF can be inherited (gametes).<br>• Explains with reason why COPD cannot be inherited (gametes). | • Comprehensive discussion linking the environmental factor of coal dust / pollution producing COPD phenotype, but genotype not changed, therefore could be cured and not inherited. (i.e. COPD phenotype and can be cured, because once the environmental factor is removed there is nothing stopping the genotype from being expressed).<br>• Comprehensive discussion of why CF cannot be cured and is inherited (i.e. mutation on CF gene changes genotype and is inherited AND produces CF phenotype and cannot be cured, because protein synthesis continues to make faulty proteins, which causes similar symptoms of COPD). |

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|---|---|---|---|---|---|---|---|---|
| No response; no relevant evidence. | Provides any partial point statement from Achievement column. | Provides any ONE statement from Achievement column. | Provides any THREE statements from Achievement column. | Provides any FOUR statements from Achievement column. | Provides any TWO statements from Merit column. | Provides any THREE statements from Merit column. | Provides the criteria for Excellence for ONE bullet point. | Provides the criteria for Excellence for BOTH bullet points. |

| Q | Expected Coverage | Achievement | Merit | Excellence |
|---|---|---|---|---|
| THREE | Gene B codes for the enzyme B. Enzyme B catalyses (speeds up) the reaction of the precursor tyrosine into pigment (melanin). Therefore, Gene B produces enzyme B; enzyme B and tyrosine produce either black or brown (melanin) pigment.<br>Gene C codes for enzyme C. This enzyme catalyses (speeds up) the reaction of the intermediate pigment (melanin) substrate into pigment (melanin) expression. Therefore, gene C produces enzyme C: enzyme C and intermediate produce pigment (melanin) expression.<br>A metabolic pathway is a series of biochemical reactions that are connected by their intermediates. The reactants (or substrates) of one reaction are the products of the previous one, and so on / a series of (enzyme controlled) reactions. Because there is a series of biochemical reactions, each one usually controlled by an enzyme, there are multiple places where the end result can be affected.<br>Yellow: gene B correctly functioning and produces either black or brown pigment. However, if gene C is non-functioning, any pigment produced at the start of the metabolic pathway cannot be expressed and the dog will be yellow / Gene B is non-functional and no pigment is produced. Gene C is functional but because no pigment is produced at the start of the metabolic pathway, no pigment can be expressed, and the dog is yellow. Gene D is functional and produces enzyme D, which interacts with the substrate from the previous reaction to produce normal intensity pigment, and the dog is yellow. Either gene B or gene C is non-functional to produce yellow.<br>Brown: gene B is functional, and the enzyme B interacts with tyrosine to produce brown pigment. Gene C is also functional, and the enzyme C expresses the brown pigment in the dog coat colour. Gene D is functional and produces enzyme D which interacts with the substrate from the previous reaction to produce normal intensity pigment and the dog is brown. All genes and enzymes are functional.<br>Black: gene B is functional, and the enzyme B interacts with tyrosine to produce black pigment. Gene C is also functional and enzyme C expresses the black pigment in the dog coat colour. Gene D is functional and produces enzyme D, which interacts with the substrate from the previous reaction to produce normal intensity pigment and the dog is black. All genes and enzymes are functional.<br>Dilute brown / black / yellow: as above plus Gene D is non-functioning (does not express pigment intensity normally) therefore produces enzyme D that interacts with the substrate of previous reaction to decrease the intensity of any pigment and produce a dilute coat colour. | • Describes function of gene B AND enzyme B.<br>• Describes function of gene C AND enzyme C.<br>• Describes a metabolic pathway.<br>• Describes an enzyme.<br>• Describes enzymes are coded for by a gene.<br>• Briefly describes one way a yellow coat colour is produced OR a black / brown coat colour is produced OR dilute coat colour is produced. | • Explains one way how yellow coat colour is produced.<br>• Explains how black / brown coat colour is produced.<br>• Explains how a dilute coat colour is produced.<br>• Explains the relationship between genes, enzymes, and products in relation to a metabolic pathway. | • Comprehensive discussion of how brown / black coat colour / phenotype is produced (all genes, enzymes, substrates and products named and integrated into answer).<br>• Discusses why there are two ways to produce yellow coat colour (all genes, enzymes, substrates and products named and integrated into answer).<br>• Comprehensive discussion of which enzymes need to be functioning / nonfunctioning for dilute coat colour / phenotype to be produced. |

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|---|---|---|---|---|---|---|---|---|
| No response; no relevant evidence. | Provides any partial point statement from Achievement column. | Provides any ONE statement from Achievement column. | Provides any THREE statements from Achievement column. | Provides FOUR statements from Achievement column. | Provides any TWO statements from Merit column. | Provides any THREE statements from Merit column. | Provides the criteria for Excellence for ANY bullet point. | Provides the criteria for Excellence for TWO bullet points. |

## Cut Scores

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