Assessment Schedule – 2021

Biology: Demonstrate understanding of gene expression (91159)

Evidence Statement

Q	Expected Coverage	Achievement	Merit	Excellence
ONE (a)	DNA is a double-stranded molecule made up of a phosphate, deoxyribose sugar, and nitrogen base. It contains the bases A, T, C, and G. RNA is a single-stranded molecule, made up of phosphate, ribose sugar, and nitrogen bases A, U, G, and C. RNA is a single-stranded nucleic acid, while DNA is double-stranded. In RNA the base U replaces the base T of DNA. Both DNA and RNA contain phosphate, and both are made up of repeating nucleotides (with varying bases). DNA is located only in the nucleus and is long-lived, whereas RNA is made in the nucleus and travels to the cytoplasm, and is short lived.	• Describes two (or more) differences between DNA and RNA, e.g. ribose v deoxyribose; uracil v thymine; single v double. • Describes two (or more) similarities between DNA and RNA (e.g. same three bases, both can be found in the nucleus, phosphate etc). OR Describes ONE similarity and ONE difference.	• Explains a similarity and a difference between RNA and DNA. OR Explains TWO similarities. OR Explains TWO differences. (*E.g. DNA is longer than RNA because DNA contains many genes. Both DNA and (m)RNA can be found in the nucleus as this is where transcription takes place*).	

(b)	DNA is made up of two strands. The template strand's function is to provide the correct sequence of amino acids in the protein by being "read" by enzymes during transcription. The coding is exactly what the mRNA copy of the template strand will be (except it contains thymine rather than Uracil). During transcription, the DNA is unwound, and the coding and template strands are separated. After transcription, these two strands wind back up. The coding strands' function is to increase the durability of the DNA strand. It reduces the risk that the DNA can be damaged and, as well as this, in the event that the template strand is damaged, acts as a template for repairing enzymes. For example, if a base was removed from the template strand and in the opposite location on the coding strand there was an adenine base, enzymes will repair the template strand by inserting a thymine base (following the complementary base paring rule). The coding strand ensures that the code on the template strand is current, and thus the resulting amino acid chains and final protein(s) are correct. Transcription is the process that makes mRNA. This process occurs in the nucleus and involves the DNA unwinding into two single strands at the promoter region. An enzyme will come along and will add bases complementary to the DNA template strand until a terminating region is reached. The completed single strand of mRNA will now leave the nucleus via a nuclear pore. As mRNA is single stranded, it is small enough to fit through nuclear pores, allowing it to reach a ribosome. The function of mRNA is to make sure that the (correct) code / instructions for how to make a polypeptide chain, and therefore protein, reaches the ribosome so that translation can occur. Accurate transcription is ensured by the base pairing rule. The size of the bases determines which bases can bind together. A large (double ring / purines) base can complement only a small (single ring / pyrimidines) base. For example, A can bind only with T, because A is large and T is small. Adenine and guanine are both large, so can't fit together in either DNA or RNA. In addition, the placement of hydrogen bonds prevents other bonding combinations. A and T form the same number of hydrogen bonds together, and C and G form the same number of hydrogen bonds together. Adenine can't bind with cytosine, because they have different numbers of hydrogen bonds and can't chemically fit together.	• Defines the coding strand OR describes its function. • Defines / describes the template strand OR describes its function. • Briefly describes the function of mRNA. • States that mRNA is made in the nucleus. • Briefly describes / defines transcription. • Identifies / states that the base pairing rule enables accurate / correct transcription.	• Explains the function of coding strand (*E.g. increases the durability to the DNA strand. Or similar)*. • Explains the function of the template strand (*E.g. provides the "template" for the enzymes to read off. Or similar)*. • Explains the function of mRNA (*E.g. carries the 'message' from the DNA to the ribosome for translation as the DNA cannot leave the nucleus*. *Or similar.*) • Explains the process of transcription (needs to include detail around how the enzyme "knows" where to start or finish). • Explains the complementary nature of DNA and mRNA, by mentioning the size of the bases (double vs single ring) OR the number of hydrogen bonds between the bases.	• Comprehensive discussion of the function of the mRNA strand AND the coding AND template strands in DNA by clearly links order of DNA bases to order of amino acids in the polypeptide, which creates the folding / joining / needed to make the protein functional. • Comprehensive discussion that includes how the DNA is transcribed (needs to include detail around how the enzyme "knows" where to start and finish),. AND Links the size of the nucleotide bases with accurate transcription, e.g. since single ring / small bases can only fit with double ring / larger bases, the base pairing rule ensures accurate transcription OR Links the chemical properties / number of hydrogen bonds that can form between nucleotide bases with the reason only certain combinations are possible. Therefore, the base pairing rule ensures accurate transcription.

NØ	N1	N2	A3	A4	M5	M6	E7	E8
No response; no relevant evidence.	Provides any ONE statement from Achievement column.	Provides any TWO statements 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 TWO bullet points.

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<table>
<tr><th>Q</th><th>Expected Coverage</th><th>Achievement</th><th>Merit</th><th>Excellence</th></tr>
<tr>
<td>TWO
(a)</td>
<td>
<table>
<tr><th></th><th>Normal PAH gene sequence</th><th>Point mutation 1</th><th>Point mutation 2</th></tr>
<tr><td>DNA template strand</td><td>TAT GGA GCC GGG</td><td>TAT GGA ACC GGG</td><td>TAT GGA TCC GGG</td></tr>
<tr><td>mRNA strand</td><td>AUA CCU CGG CCC</td><td>AUA CCU UGG CCC</td><td>AUA CCU AGG CCC</td></tr>
<tr><td>Amino acid sequence</td><td>Ile Pro Arg Pro</td><td>Ile Pro Trp Pro</td><td>Ile Pro Arg Pro</td></tr>
</table>
</td>
<td>• Three boxes correct (must include at least one mRNA and Amino acid sequence / box).</td>
<td>• All mRNA and amino acid sequences are correct.</td>
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(b)	Mutations can be caused by mutagens (substances such as UV radiation, which damage the DNA) or by errors during DNA replication. Either way, the final base sequence has changed. Point mutation one is a base substitution (as only one base / nucleotide has been affected), and has changed the third amino acid from ARG to TRP. The second point mutation is also a substitution; however this is a silent mutation where the change in the base sequence has coded for the same amino acid ARG. This is due to the degeneracy of the code leading to redundancy. There are 20 amino acids, however 64 triplets / codons. This means that an amino acid will usually have more than one codon / triplet that codes for it. No reading frame shift has occurred in either of the mutations (where the bases are moved to the left or right by a base deletion or insertion that is not a multiple of three), and therefore there will be no effect on the start or stop codons for either of these mutations. However ,mutation one will have an effect on the final protein (the enzyme phenylalanine) that is produced. The change in amino acid will result in the amino acid chain folding incorrectly. This will mean that its shape is slightly altered, and thus its function will be compromised (it will not be able to catalyse the reaction that is intended). However, for the silent mutation, this is not the case. As the same amino acid is coded, this means that the amino acid chain will fold in the exact way that it was intended, and thus the enzyme will function exactly the same as if the mutation had not happened.	• Describes a cause of mutations. OR Lists multiple examples of mutagens (*E.g. mutations are caused by things such as smoking, UV radiation etc*). • Describes a point mutation. • Describes a substitution mutation. OR Identifies a substitution mutation has occurred. • Describes a silent mutation. • Identifies a frame shift has not occurred / start / stop codons not affected. • Describes the function of an enzyme. • States enzyme will still function / have same shape / folding for Point mutation 2. OR Enzyme wont function / will have a different shape /folding for Point mutation 1.	• Point mutation 1's change in amino acid will result in the protein changing shape / folding differently which will affect its function. OR Point mutation 2 does not cause a change in amino acid so its shape / folding AND function will be unaffected. • Briefly explains the cause of silent mutations (ie more codons than amino acids / redundancy in the code) . • Explains how / why neither mutation will affect the start or stop codons / cause a frameshift, as the length of the gene / number / order of amino acids will still be the same. • Explains the change in shape of the protein seen in mutation 1 might be slight / small so the enzyme may still function to some degree.	• Discusses why point mutation 1 will not affect the position of the start / stop codons (i.e. does not cause a frameshift / mutation occurs in the middle of the gene) and how the change in the amino acid will impact protein folding, shape AND function. • Discusses how point mutation 2 is a silent mutation, by comprehensively explaining the cause of redundancy in the code, linking this to how the sequence / length of amino acids will still be the same and thus the folding / shape of the protein will be the same and therefore the enzyme will function normally.

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THREE	A metabolic pathway is a series of enzyme-controlled reactions where the product of one reaction becomes the substrate for the next (intermediates). A person with PKU must stick to a low protein diet their entire life, due to the fact that they have a mutation in their DNA (Gene 2) that codes for the PAH enzyme. As a result, they cannot produce this enzyme (or the enzyme that they do produce is the incorrect shape to carry out its function). As a result, if a person ingests protein, this will be converted into phenylalanine by protease (which is coded for by Gene 1) however the phenylalanine cannot be converted into tyrosine and then into melanin, so will build up in the body. This is true, even if all the other enzymes in the metabolic pathway are functioning. However, even if a person has the gene mutation for Gene 2, they can avoid developing PKU by not eating protein. This is possible because they can skip out this part of the metabolic pathway by consuming foods containing tyrosine / a tyrosine supplement. This way no phenylalanine builds up in the body (as the protease enzyme has no substrate / protein to catalyse into phenylalanine, but tyrosine is still present for the enzyme tyrosinase (coded for by Gene 3) to convert into melanin. If a person with a mutation in Gene 2 did not stick to a low protein diet their entire life, they would develop the symptoms of PKU, due to the fact that the protease enzyme, will be catalysing protein into phenylalanine, but their non functioning or absent PAH enzyme would not convert this into tyrosine. The resulting build up / high levels of phenylalanine in the blood would become toxic and start damaging the person's brain. Comparatively unlike PKU, albinism cannot be "cured" by changing your diet. This is because the mutation which causes albinism is in Gene 3. This results in the enzyme Gene 3 codes for, tyrosinase, to not be present or to be non functional (as it is the wrong shape). Consequently, tyrosine is not able to be converted into melanin. As no foods contain melanin (it must be made by the tyrosinase enzyme in the body), there is no diet / environment which can provide melanin, and thus the person will have albinism. A person could develop both PKU and albinism in two ways. The first way is where there is a mutation to Gene 2, the person does not eat enough foods containing tyrosine, and continues to eat protein. The protein consumed would result in a build-up of phenylalanine (causing PKU), and the lack of tyrosine could mean that, even though Gene 3 still codes for tyrosinase, this enzyme does not have enough tyrosine / substrate to convert into melanin (resulting in albinism). The second way someone could have both PKU and albinism is when they	• Briefly describes / defines a metabolic pathway. • Describes how an environmental factor can prevent PKU. (*E.g. no / low protein intake means no phenylalanine is made.)* • States that a change in diet does not affect DNA / correct a mutation. (*E.g. environment doesn't change genotype therefore phenotype still the same.*) • Describes how consuming tyrosine can complete the pathway for someone with a mutation to gene 2 / PKU. • Describes the cause of PKU (*E.g. Phenylalanine is not being converted to tyrosine OR there is a mutation in Gene 2.*) • Describes the relationship between genes, enzymes. (*E.g. one gene codes for one specific enzyme OR Gene 1 codes for the production of Protease etc.*) • Describes one way that someone could have PKU AND albinism. (*E.g. mutation to both Gene 2 and 3 OR mutation to Gene 2 (allow Gene 1) and no intake of Tyrosine.*)	• Explains the relationship between genes enzymes and products for this specific metabolic pathway. • Clearly explains how a person develops PKU by explaining the role of Gene 1 and Protease and Gene 2 and PAH Enzyme in the metabolic pathway. • Explains how a change in diet / the environment cannot affect / correct the mutation, and thus someone with PKU will always produce no / a non functioning PAH enzyme. • Explains how / why the symptoms of PKU / PKU can be avoided by someone sticking to a low-protein diet for their entire life OR Explains why someone with a mutation to gene 3 / with albinism cannot be "cured" by changing their environment / diet (i.e. you cannot consume melanin). • Explains how a person develops albinism by explaining the relationship between Gene 3 tyrosinase, and tyrosine and melanin. OR Explains that even if Gene 3 is working / non mutated. no melanin will be produced / a person will have albinism if	• Comprehensive discussion that includes an explanation of this specific metabolic pathway, as well as how someone could have both PKU AND albinism by having a mutation to Gene 2, as well as not consuming tyrosine OR Having a mutation to Gene 2 and Gene 3. • Comprehensive discussion that includes an explanation of this specific metabolic pathway, including how / why the symptoms of PKU / PKU can be avoided by someone sticking to a low-protein diet for their entire life AND Explains why someone with a mutation to gene 3 / with albinism cannot be "cured" by changing their environment / diet (i.e. you cannot consume melanin).

	have mutations in both Gene 2 and Gene 3, and they consume protein in their diet. If this was the case, the protein consumed would be converted into phenylalanine by the protease enzyme; however the mutated Gene 2 would mean that there was no functioning PAH enzyme present to convert this into tyrosine. This would result in PKU. However, unlike the previous example, no amount of tyrosine consumed would result in melanin being produced. This is because Gene 3 is mutated, so the enzyme tyrosinase is either not present or is not the correct shape for tyrosine to fit into its active site, and thus be catalysed into Melanin, resulting in Albinism.		Gene 2 is mutated and a person is not ingesting tyrosine. • Explains one way that someone could have both PKU AND Albinism.	

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Cut Scores

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