Assessment Schedule – 2020

Biology: Demonstrate understanding of gene expression (91159)

Evidence Statement

<table>
<tr><th>Q</th><th>Expected coverage</th><th>Achievement</th><th>Merit</th><th>Excellence</th></tr>
<tr>
<td>ONE
(a)</td>
<td>
<table>
<tr><th></th><th>Normal CFTR gene sequence</th><th>Mutated sequence causing cystic fibrosis</th></tr>
<tr><td>DNA template strand</td><td>TAA TAG AAA CCA CAA</td><td>TAA TAA CCA CAA</td></tr>
<tr><td>mRNA strand</td><td>AUU AUC UUU GGU GUU</td><td>AUU AUU GGU GUU</td></tr>
<tr><td>amino acid sequence</td><td>Ile Ile Phe Gly Val</td><td>Ile Ile Gly Val</td></tr>
</table>
</td>
<td>• Correct mRNA sequences OR Correct Amino acid sequences for incorrect mRNA sequence.</td>
<td>• All of the table is filled out correctly.</td>
<td></td>
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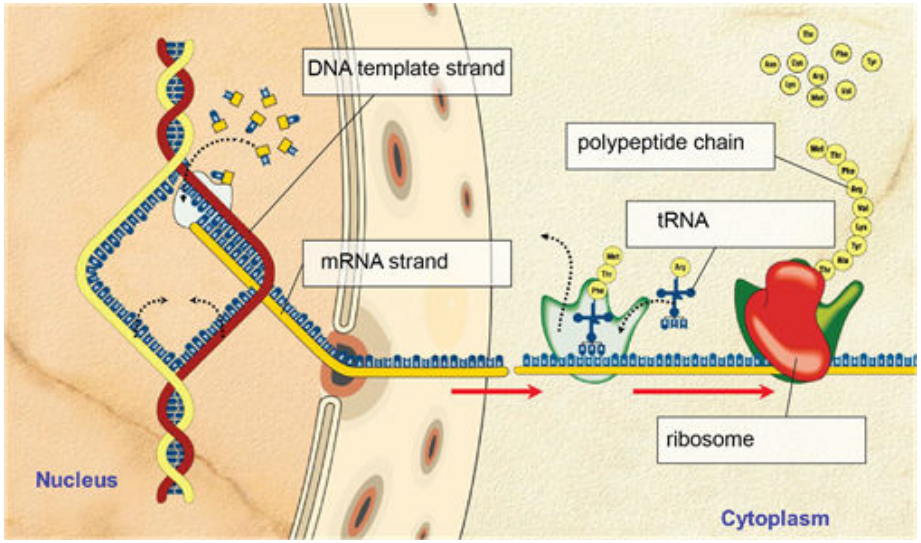
(b)	The mutation that has occurred is a deletion mutation where the three bases G, A and A have been removed from the DNA sequence. As three bases have been deleted this means that a frame shift has not occurred. A frame shift is where after a mutation, all the triplets are incorrect (as they are not "read" in their correct threes). Frame shifts can be deadly and are usually more serious than mutations that don't result in frameshifts, like substitution mutations (where a base(es) is changed with another). This is because a substitution (or a three base deletion / addition) will only (usually) 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. Comparatively, a frame shift will result in all the amino acids after the mutation to be incorrect. This will result in the final protein taking on a vastly different final shape. As a protein's shape relates to its function. This drastic change in shape will render the protein completely non-functional. As well as this, a frame shift will likely change the position of the stop codon. Either the stop codon will come early, terminating the polypeptide chain early or it will come too late, extending the length of the polypeptide chain. The change in the stop codon position further contributes to the drastic change in the final proteins structure. In our specific mutation the triplet TAG has changed to TAA, due to redundancy due to degeneracy of the code (ie different triplets, and therefore codons can code for the same amino acid) this doesn't change the amino acid produced (Ile). Due to three bases being removed however, the new mutated DNA strand is now missing the triplet (and therefore codon) which codes for the amino acid Phe. As no frame shift has occurred the stop codon will still be in the correct place and thus the shape of the protein will not be drastically different to its correct shape. However the missing amino acid has changed the shape of the final membrane protein enough to cause it not to function correctly resulting in cystic fibrosis.	• States the mutation is a deletion mutation / three bases have been removed. • Defines triplet. OR Defines codon. • Briefly describes a frameshift *(eg every triplet / codon after the mutation will be incorrect).* • Briefly describes how the location of the stop codon will change in a frameshift. • States that a frame shift has not occurred. • Identifies an Amino acid in the chain has been lost / removed. • Identifies that the mutation changes the final shape OR function of the protein. • Describes that there are multiple codons for the same amino acid.	• Explains what degeneracy of the code is and how this relates back to TAG and TAA coding for the same amino acid. • Explains how frameshifts affect the codon / triplet and therefore amino acid sequence. • Explains the consequence of the location of a stop codon changing after a frameshift. • Explains that a change in amino acid sequence will result in a change to the proteins shape AND affect the proteins function. • Explains that as three bases have been deleted, a frameshift has not occurred.	• Discusses how the deletion in the DNA, missing codon in the mRNA, and thus missing amino acid, will result in the final protein not folding correctly and therefore its shape will be incorrect and the protein will be unable to carry out its function. • Compares and contrasts the severity of frameshift mutations to this specific deletion mutation by discussing what frame shifts are and their resulting consequences.

N0	N1	N2	A3	A4	M5	M6	E7	E8
No response, No relevant evidence	Provides one statement from Achievement column	Provides two statements from Achievement column	Provides three statements from Achievement column	Provides four statements from Achievement column	Provides two statements from Merit column	Provides three statements from Merit column	Provides one statement from Excellence column	Provides both statements from Excellence column

Q	Expected coverage	Achievement	Merit	Excellence
TWO (a)		• THREE labels correct.		

(b)	The DNA template strand is the strand that the mRNA is made from. It is complementary to the mRNA strand. mRNA is "messenger" RNA and is the short single strand of RNA that is made off the template strand. tRNA is "transfer" RNA that is complementary to the mRNA. Its anticodon ensures that the correct amino acid is added to the growing polypeptide chain (as the Ribosome reads the mRNA). The Ribosome is the structure that "reads" the mRNA and, through the use of tRNA, adds amino acids that correspond to each codon on the mRNA until a stop codon is reached. The polypeptide chain is the name for the chain of Amino acids which will fold together to make a protein. Transcription is the process where mRNA is made from the template strand of DNA in the nucleus. This process involves the DNA unwinding into two single strands. An enzyme will then bind to the template strand of DNA at the promoter region. This enzyme will move along the DNA adding complementary RNA bases (A-U, C-G) until a terminating region is reached. The completed strand of mRNA will now leave the nucleus. The two strands of DNA will bind back together. This is the end of transcription. Translation is the process where the mRNA is read by the ribosome in the cytoplasm and an amino acid chain is created based on the codons on the mRNA. This process involves the Ribosome binding to the start of the mRNA strand. The ribosome will move along the mRNA reading it in threes (codons). tRNA, with three complementary bases (called anticodons) to the mRNA codons, will enter the ribosome and drop off their attached amino acid. The tRNA will then leave for another tRNA to enter the ribosome and drop off its amino acid. These amino acids are joined together by the ribosome forming a polypeptide chain. This process continues until the ribosome encounters a stop codon. At this point the mRNA will leave the ribosome, the polypeptide chain will also detach. This is the end of translation. Both processes are similar in that they use the complementary base pairing rule to ensure that no mistakes are made. As well as this both processes need a template to "read off", the DNA template strand for transcription, and the mRNA strand for translation. Contrastingly however, transcription takes place in the nucleus whereas translation happens in the cytoplasm. This is important as DNA is a valuable molecule and cannot leave the nucleus (due to its size but also to reduce the risk of it becoming damaged). Translation needs to occur in the cytoplasm as this is where the ribosomes / amino acids are located. Both processes are required to build a polypeptide chain as they each have a separate role. Without transcription the template strand could not be made into transportable mRNA, and thus the instructions for making the protein / polypeptide chain could never reach a ribosome to be translated. Without translation, even if transcription occurred there would be no process by which the information in the mRNA strand could be translated into protein. For this reason both processes are required to build a polypeptide chain.	• Describes template strand. • Describes mRNA. • Describe tRNA. • Describes ribosome. • Describes polypeptide chain. • Briefly describes transcription. • Briefly describes translation. • States a similarity between both processes . OR States a difference between both processes. • Briefly describes why both processes are needed.	• Explains how Transcription occurs. • Explains why transcription is necessary *(ie DNA can't leave the nucleus).* • Explains how translation occurs. • Explains why translation is necessary *(ie converts the instructions in DNA / mRNA into protein which carry of essential functions).* • Gives a reason for a similarity between both processes *(eg both processes use the base pairing rule to ensure their final product is correct).* • Gives a reason for a difference between both processes *(e.g. transcription needs to occur in the Nucleus as this is where DNA is located. Translation happens in the cytoplasm as this is where ribosomes are located).*	• Thorough discussion of both the process of transcription and translation including **at least one similarity and difference**. • Thorough discussion of both the process of transcription and translation including a justification as to why both processes are necessary.

N0	N1	N2	A3	A4	M5	M6	E7	E8
No response, No relevant evidence	Provides one statement from Achievement column	Provides two statements from Achievement column	Provides three statements from Achievement column	Provides four statements from Achievement column	Provides two statements from Merit column	Provides three statements from Merit column	Provides 1 bullet point from Excellence column	Provides both statements from Excellence column

Q	Expected coverage	Achievement	Merit	Excellence
THREE	An enzyme is a biological catalyst made of protein. 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. Each enzyme can only catalyse one specific reaction due to its unique shape. In a normal / functioning metabolic pathway, gene 1 codes for enzyme 1 which converts the first protein into phenylalanine. Phenylalanine is then the substrate in the next reaction where gene 2 codes for enzyme 2 which converts phenylalanine into tyrosine. If gene 2 is mutated this means that enzyme 2 will not function as its shape will be incorrect (due to incorrect amino acids being coded for). This will mean that enzyme 2 is unable to catalyse the reaction of converting phenylalanine into tyrosine. This ultimately will cause phenylalanine to build up in the person causing the PKU disorder. For Phenylalanine to be produced only gene 1 and therefore Enzyme 1 needs to be functioning (gene 2 could be mutated / enzyme 2 could be non-functional). However for Tyrosine to be produced both gene 1 and gene 2 (and their corresponding enzymes) need to be functioning. If there is a mutation in one gene (1 or 2) then Tyrosine will not be able to be produced. This is because without gene 1 / enzyme 1 phenylalanine will not be produced so enzyme 2 will have no substrate to convert into Tyrosine. If gene 1 is functioning but gene 2 is not, although Phenylalanine will be made there is no functioning enzyme to convert this into tyrosine.	• Describes an enzyme. • Describes the relationship between genes and enzymes / how one gene codes for a specific / one enzyme. • Defines metabolic pathway. • Identifies gene 2 as causing PKU disorder. • Briefly describes that for tyrosine to be produced both genes / enzymes need to be correct / functional. • Briefly describes that for phenylalanine to be produced gene 1 / enzyme 1 needs to be correct / functional OR a mutation to gene 1 will result in no phenylalanine being produced. • Briefly describes how a functional metabolic pathway works.	• Explains the relationship between genes, enzymes and products in the metabolic pathway *(ie Explains how a / the "normal" metabolic pathway functions).* • Explains that only gene 1 and enzyme 1 need to be functional to produce phenylalanine OR Explains that a mutation to gene 2 / a non-functioning enzyme 2 will not influence the production of phenylalanine. • Briefly explains that if any gene / enzyme in the pathway is mutated / non-functional, tyrosine will not be produced. • Explains why a mutated gene will result in the enzyme being non-functional (ie the incorrect amino acids are included / enzyme will be the wrong shape). • Explains why a mutation to gene 2 results in a block to the metabolic pathway and how this relates to the build-up of phenylalanine.	• Clear discussion of how phenylalanine and tyrosine are produced in a functioning metabolic pathway, as well as how and why a mutation to gene 2 will result in a build-up of phenylalanine. • Clear discussion of how phenylalanine and tyrosine are produced in a functioning metabolic pathway highlighting that there are two places on the metabolic pathway where a mutation will result in Tyrosine not being produced.

N0	N1	N2	A3	A4	M5	M6	E7	E8
No response, No relevant evidence	Provides one statement from Achievement column	Provides two statements from Achievement column	Provides three statements from Achievement column	Provides four statements from Achievement column	Provides two statements from Merit column	Provides three statements from Merit column	Provides a bullet point from Excellence column	Provides both statements from Excellence column

Cut Scores

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