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91159



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Mana Tohu Mātauranga o Aotearoa
New Zealand Qualifications Authority

Level 2 Biology 2025

91159 Demonstrate understanding of gene expression

Credits: Four

Achievement	Achievement with Merit	Achievement with Excellence
Demonstrate understanding of gene expression.	Demonstrate in-depth understanding of gene expression.	Demonstrate comprehensive understanding of gene expression.

Check that the National Student Number (NSN) on your admission slip is the same as the number at the top of this page.

You should attempt ALL the questions in this booklet.

If you need more room for any answer, use the extra space provided at the back of this booklet.

Check that this booklet has pages 2–16 in the correct order and that none of these pages is blank.

Do not write in the margins (//////). This area will be cut off when the booklet is marked.

YOU MUST HAND THIS BOOKLET TO THE SUPERVISOR AT THE END OF THE EXAMINATION.

Merit

TOTAL 17

QUESTION ONE: Mutation effects on protein – cystic fibrosis

Cystic fibrosis is a genetic disorder caused by mutations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which encodes a protein that helps regulate salt and water balance in cells. Mutations in this gene can disrupt the function of the CFTR protein, leading to the build-up of thick mucus in the lungs and other organs.

Table 1. mRNA (codon): Amino Acid

		Second Position				
		U	C	A	G	
First Position	U	UUU Phe	UCU Ser	UAU Tyr	UGU Cys	U
		UUC Phe	UCC Ser	UAC Tyr	UGC Cys	C
		UUA Leu	UCA Ser	UAA STOP	UGA STOP	A
		UUG Leu	UCG Ser	UAG STOP	UGG Trp	G
	C	CUU Leu	CCU Pro	CAU His	CGU Arg	U
		CUC Leu	CCC Pro	CAC His	CGC Arg	C
		CUA Leu	CCA Pro	CAA Gln	CGA Arg	A
		CUG Leu	CCG Pro	CAG Gln	CGG Arg	G
	A	AUU Ile	ACU Thr	AAU Asn	AGU Ser	U
		AUC Ile	ACC Thr	AAC Asn	AGC Ser	C
		AUA Ile	ACA Thr	AAA Lys	AGA Arg	A
		AUG Met	ACG Thr	AAG Lys	AGG Arg	G
G	GUU Val	GCU Ala	GAU Asp	GGU Gly	U	
	GUC Val	GCC Ala	GAC Asp	GGC Gly	C	
	GUA Val	GCA Ala	GAA Glu	GGA Gly	A	
	GUG Val	GCG Ala	GAG Glu	GGG Gly	G	

- (a) Part of a DNA sequence and its associated mRNA and amino acid sequence are shown in Table 2 below. Complete the mRNA and amino acid sequences for both mutated sequences in the table. The DNA mutations are underlined.

Table 2

	Normal sequence	Mutated sequence 1	Mutated sequence 2
DNA template strand	TTA TGC AAT CCG	TTA TGC <u>GAT</u> CCG	TTA TGC <u>AAG</u> CCG
mRNA	AAU ACG UUA GGC	AAU ACG <u>CUA</u> GGC	AAU ACG <u>UUC</u> GGC
Amino acid	Asn Thr Leu Gly	Asn Thr <u>Leu</u> Gly	Asn Thr <u>Phe</u> Gly

(b) Discuss the effects of these mutations on the amino acid sequence and final protein.

In your answer, include discussion of:

- the type of point mutations shown, and the severity of these mutations compared to other types, such as frame shift or stop codons (nonsense mutations) ✓
- the effects of these mutations on the amino acid sequence and final protein produced ✓
- the effect of these mutations on the final functioning of the CFTR protein and how this relates to the symptoms of cystic fibrosis.

A mutation is a permanent change in the DNA sequence that is the only source of new alleles. "alleles is an alternative form of a gene". The Normal DNA strand shown are TTA TGC AAT CCG these are in ~~parts~~ triplets where ~~the~~ triplets "TTA" are transcribed onto mRNA in the nucleus through the complementary base pairing rule "T-A and C-G" with the exception that in RNA the T is substituted with U. So it becomes A-U instead. These triplets then form codons on the mRNA where translation by the tRNA matches with the codons with anticodons and matches amino acids in the cytoplasm joining with ^{peptide bonds making} polypeptide chains. These polypeptide chains then fold forming the protein. In mutated sequence a substitution point mutation occurs where the A is substituted for the G in the third triplet. These do not cause a frameshift as there are of equal length polypeptide chain as there was before. This mutation produces an amino acid ~~len~~ that is of the same as the amino acid on the non mutated sequence. This therefore is a same sense mutation which means that tho a base in the DNA sequence was changed it still produced the same amino acid "len" as the normal sequence. Therefore the protein formed had the same functional capacity as it did before. This is due to degeneracy and redundancy in the mRNA as several base sequences code for an amino acid. This shows the non severe effect a ~~non sense~~ ^{same sense} mutation has on a ~~amino~~ protein function. This also shows us that the

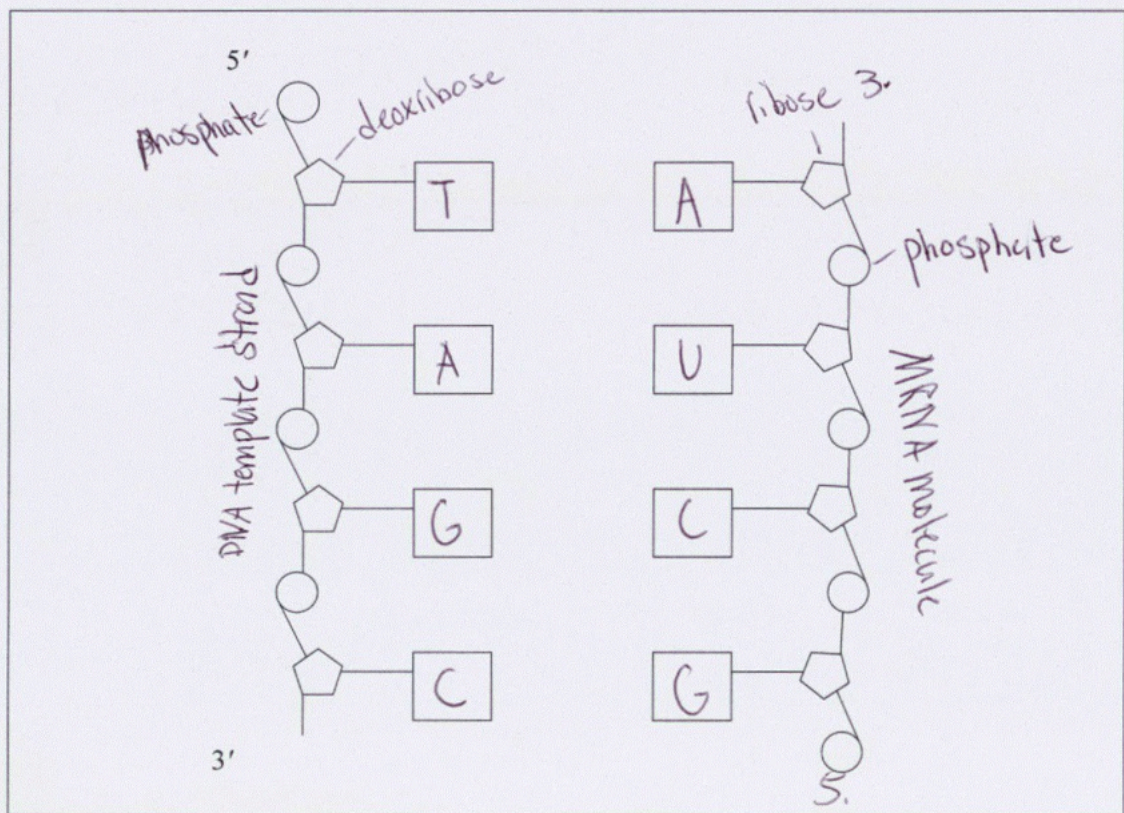
Cystic fibrosis was "not" caused by the mutation in mutated Sequence 1 ~~as~~ as the protein should still function to clear mucus, regulate salt and water in the lungs. The second mutated sequence shows a "substitution point mutation" where a base is changed and swapped. Where the third triplet last letter T is substituted for a G. This leads to a no frame shift or length change in the polypeptide chain but causes a different amino acid to be formed through the different mRNA codon (UUC) instead ~~of~~ giving the incorrect amino acid (Phe). This leads to a missense mutation where one incorrect amino acid has formed. This means that the protein may no longer function in regulating salt and water balance causing cystic fibrosis. This is far more severe than the same sense mutation as the protein will have a reduced function without this Leu amino acid. However it is not as serious as a nonsense reading frame shift as this would cause whole polypeptide chains to form incorrectly and/or causing a shortening or lengthening of the amino acid due to missing stop codon. This would form a totally non-functioning protein and would be the most severe form of cystic fibrosis. These final mutation on Sequence 1 will not cause cystic fibrosis and will still lead to the normal sequence formed. Furthermore the 2nd mutated sequence will cause cystic fibrosis due to the missense mutation causing an incorrect amino acid forming. This will cause the CFTR protein to not be able to regulate salt and water as well, leading to a build up of mucus in lungs causing the cystic fibrosis.

QUESTION TWO: Protein synthesis

(a) Complete the diagram of transcription below.

In your answer:

- fill in the DNA template strand containing the bases thymine (T), adenine (A), guanine (G), and cytosine (C) ✓
- complete the corresponding mRNA strand, showing complementary base pairing ✓
- label the ribose and deoxyribose on the DNA and mRNA strands ✓
- label the phosphates on both the DNA and mRNA strands ✓
- label the 5' and 3' ends of the mRNA molecule ✓
- label the DNA template strand and the mRNA molecule. ✓



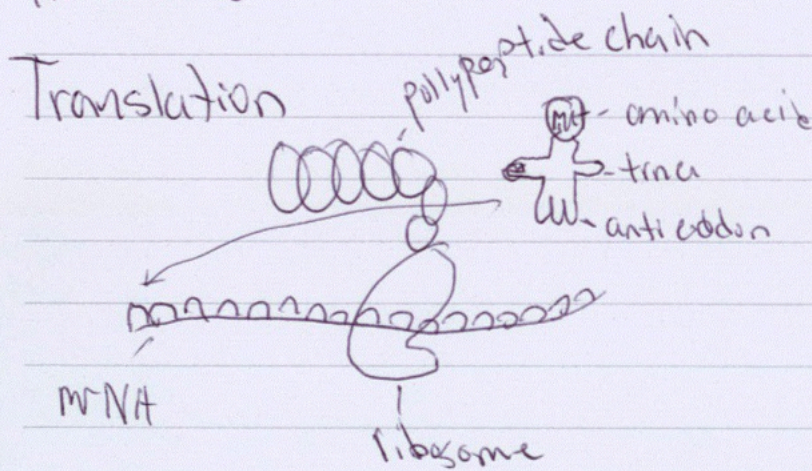
(b) Discuss the importance of transcription and translation, and why the DNA strand is not directly translated into a polypeptide chain.

In your answer, include discussion of:

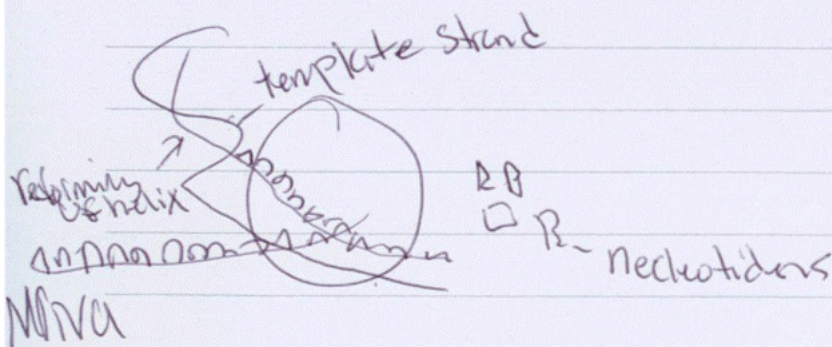
- the purpose of transcription and translation, and the steps involved with each ✓
- the relationship between codons, anticodons, tRNA, mRNA, and amino acids during translation ✓
- TWO reasons why the DNA strand cannot be directly translated into a polypeptide chain, focusing on the roles of transcription and mRNA in this process. ✓

The Transcription begins in the nucleus where RNA polymerase enzyme unzips the 2 helical strands of DNA into ~~into~~ where one strand is the template DNA strand and one is the other. At this point mRNA "messenger RNA" begins matching the bases with the complementary base pairing rules A-T and C-G however RNA uses A-U instead of T and therefore substitutes the T for U, where adenine, uracil, guanine and cytosine for DNA. At this point the floating base pairs nucleotides in the nucleus form with the complementary base pairing and then forms mRNA using the DNA template strand. The DNA is double stranded and a large molecule and therefore ~~takes time~~ therefore cannot fit out of the nucleus pores to move to the cytoplasm. Therefore the mRNA a single strand not as stable and small molecule is able to fit out of the pore and travel to the cytoplasm for translation. Furthermore the DNA is used for every single cell in our body and needs to be protected. Therefore never leaves the nucleus which is done by the mRNA. As the mRNA leaves the nucleus through the pores the DNA is rejoined. As we move to the cytoplasm the mRNA is read by the cytoplasmic ribosome inside the cytoplasm. It reads the promoter region "start codon" AUG on the mRNA to know the translation process must begin. As each codon "3 base triplet on the mRNA" is read the tRNA "transport RNA" matches the anti-codon to the codon on the mRNA through the ^{complementary} base pairing rule. These tRNA carry a amino acid which when the anticodon matches attaches to the adjacent amino acid through peptide bonds. This process repeats until several amino acids bond together until the ribosome reads a stop codon on the mRNA at this point the amino acid chain "polypeptide chain" detaches off of the tRNA, where

The tRNA further detaches off and the old mRNA gets dissolved by the enzymes. This process repeats, ~~until~~ once several poly peptide chains are made then they fold and form structures we call proteins. The key point on why DNA cannot be directly translated is due to the size "cannot leave pores" and need to protect the DNA therefore the DNA does not leave the nucleus and cannot be directly translated



Transcription



QUESTION THREE: The environment and gene expression in plants

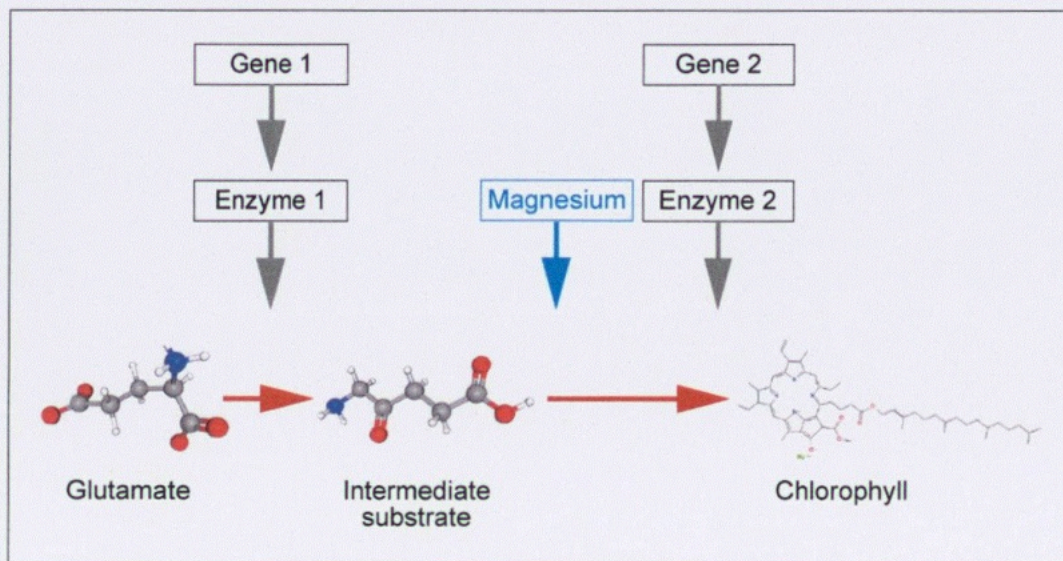
Chlorophyll is the pigment responsible for the green colour in leaves. The production of chlorophyll is influenced by both genetic factors and environmental conditions such as magnesium availability. In plants with chlorosis, genetic mutations or the lack of magnesium availability can lead to reduced chlorophyll production, resulting in yellow or pale leaves.



Strawberry leaf showing chlorosis symptoms.

Healthy strawberry leaf.

A simplified metabolic pathway that makes chlorophyll is shown below:



Discuss how genes, enzymes, and the environment regulate the production of chlorophyll in plants and cause the yellowing of leaves due to chlorosis.

In your answer include discussion of:

- a metabolic pathway ✓
- the metabolic pathway shown above, using the terms substrate, enzyme, gene, and final product ✓
- how DNA mutations and magnesium availability can affect chlorophyll production and leaf colour. ✓

A metabolic pathway is a set of essential biochemical reactions formed through catalysts that produce a product. The metabolic pathway for the essential life process for plants of production of chlorophyll is listed. It begins with the glutamate, where the Gene 1 produces a ~~catalyst~~/enzyme 1 that converts glutamate "substrate" into an intermediate substrate. At this point magnesium is a catalyst that helps gene 2 \rightarrow Enzyme 2 to produce the final "product" of the chlorophyll. Furthermore the intermediate substrate becomes the substrate for the ~~second~~ enzyme 2 which used in combination with the magnesium forms the chlorophyll. The condition chlorosis is a mutation "a percent change in the dna sequence" or lack of magnesium availability could affect to the metabolic pathway of chlorophyll production. Specifically genotype + environment = phenotype. The genotype for the metabolic pathway is already there for producing chlorophyll in both healthy and not healthy leaves however due to the environmental factor "enough magnesium or not" has caused different phenotypes (a green or yellow leaf). In the case of chlorosis this catalyst of magnesium needed for enzyme 2 to convert intermediate substrate into chlorophyll was lacking. This meant that glutamate for both plants were converted properly through enzyme 1. however the lack of magnesium caused enzyme 2 to not be able to convert enough intermediate substrate into chlorophyll. This caused a lack of green pigment for the plant and mostly likely resulted in the death as chlorophyll is needed for photosynthesis. However as the plant with enough magnesium converted enough ~~the~~ intermediate substrate with the enzyme 2 this means it

had the healthy green hue we love. DNA mutations could further affect the metabolic pathway by changing a gene, enzyme. This is what happened to the strawberry plant, let's say gene 1 was unable to produce the enzyme 1 then there would be a build up of glutamate which would not be converted. This would cause no intermediate substrate to be produced to be converted by gene 2 enzyme 2 to chlorophyll therefore the plant would be very yellow and die.

Merit

Subject: Biology

Standard: 91159

Total score: 17

Q	Grade score	Marker commentary
One	E7	The response compared substitution mutation to frameshift mutation and discussed the frameshift causing change in amino acid length (shortening), linking to functionality and severity of cystic fibrosis symptoms.
Two	M6	The response identified the structure of the DNA and mRNA. The response explained: <ul style="list-style-type: none">• the complementary nature of the DNA and mRNA code• the reasons why DNA is not directly translated into a polypeptide chain• translation.
Three	A4	The response described: <ul style="list-style-type: none">• a metabolic pathway• a biological catalyst• a final product• gene + environment produces the phenotype• a substrate.