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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.**

Excellence

TOTAL 24

### 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
		<u>UUC</u> 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>G</u> AT CCG	TTA TGC <u>AA</u> G CCG
mRNA	AAU ACG UUA GGC	AAU ACG CUA GGC	AAU ACG UUC GGC
Amino acid	Asn Thr Leu Gly	Asn Thr Leu Gly	Asn Thr Phe 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.

Mutation is a random, permanent change to DNA base sequence. The mutation 1 & 2 are substitution mutations, where a base is replaced by another in DNA base sequence. The 1st mutation is a silent mutation because the mutation in DNA triplet, changing AAT to GAT, means Leu to Leu to CUA, then amino acid from Leu to Leu. re same amino acid Leu. This is due to degeneracy of genetic code due to redundancy. There are 64 codons but only 20 amino acids & ~~each~~ multiple codon code for same amino acid, so that the mutated codon still codes for same acid Leu. Amino acid sequence is unchanged, and polypeptide chain (PPC) is correct so that protein folding, shape & function are functional at CFTR protein. This means NO symptom of cystic fibrosis (CF) (less severe).

Whereas in mutation 2, triplet change from AAT to AAG, a silent codon change from Leu to Leu, changing amino acid from Leu to Phe. As only one amino acid has changed, the effect may be minimal, but will still result in an incorrect polypeptide chain, so that

CFTR protein folding & shape is incorrect & CFTR protein is non-functional / cannot regulate salt & water balance, leading to symptoms of excess mucus in lungs & organs. However, substitution mutations do not increase or decrease the DNA base sequence, so that amino acid sequence stays the same length & position of start & stop codons in mutant stays same. (No reading frame shift) (Less severe)

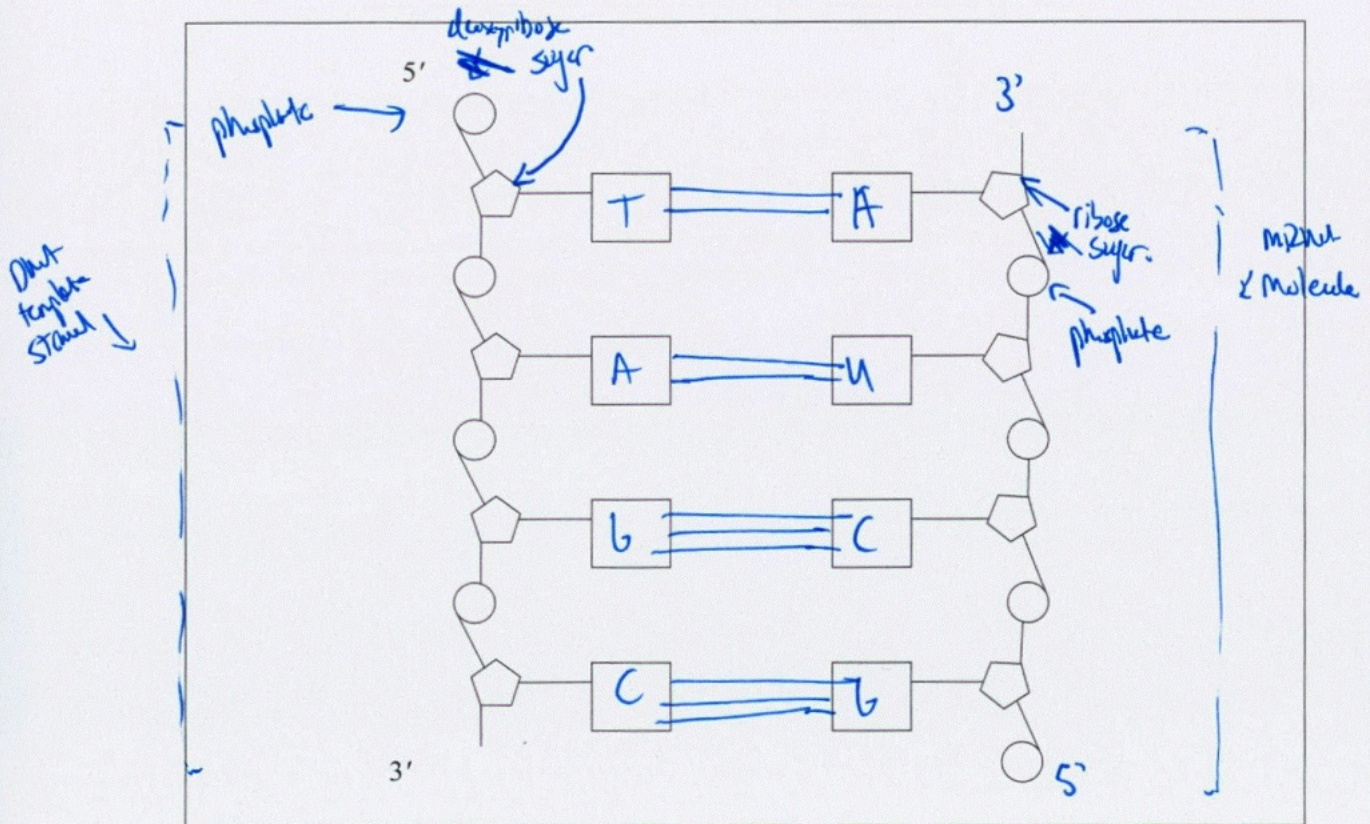
Whereas frameshift mutations, like insertion (adding base to DNA sequence) or deletion (removing base from DNA sequence) increases or decreases the DNA base sequence respectively, changing the length of the amino acid (AA) sequence drastically from the point of mutation onwards. The frameshift can also code for a early stop codon, terminating the amino acid sequence & polypeptide chain (PPC) earlier / shortening it. Hence, frameshift mutations drastically change the amino acid sequence resulting polypeptide chain (nonsense mutations) so that protein folding & shape are incorrect & CFTR protein is non-functional, so CFTR protein cannot regulate the salt & water balance in cells, leading to build-up of thick mucus in lungs & other organs which is much more severe vs substitution mutations.

## 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.

Transcription is the process that copies the DNA template strand to make complementary mRNA strand. Its purpose is to transport the mRNA <sup>to make polypeptide chain (protein)</sup> from DNA in nucleus out to ribosomes in cytoplasm, instead of DNA to prevent DNA getting damaged. ✓ T E E T m o n

- ① Transcription starts when the enzyme binds to promoter sequence.
- ② Enzyme unwinds, unravels, unzips & separates the DNA strands, exposing the bases.
- ③ Enzyme copies the DNA template strand to make complementary mRNA strand using free RNA nucleotides & complementary base pairing rule (A/U / C/G).
- ④ Transcription stops when enzyme reaches terminator sequence.
- ⑤ mRNA & enzyme detach from DNA template strand.
- ⑥ mRNA moves from the nucleus into cytoplasm to dock to ribosome.

codons are 3 consecutive bases on mRNA <sup>strand (messenger RNA)</sup>, which are complementary to anticodons, which are 3 consecutive bases on tRNA molecule. mRNA, messenger RNA carries the genetic code to make polypeptide chain from DNA in nucleus to ribosomes in cytoplasm instead of DNA to prevent DNA getting damaged. whereas tRNA (transfer RNA) carries the amino acid to the ribosomes in the cytoplasm to be added to the growing polypeptide chain (PPC).

Translation is the process where information on mRNA is read to make polypeptide chain/protein. Its purpose is to create the functional protein that can carry out life processes using the amino acid sequence. T R E A T T.

- ① Translation starts when the ribosome binds to the start codon on mRNA (AUG).
- ② Ribosome reads each codon on mRNA & matches/pairs it with the complementary anticodon on tRNA using complementary base pairing rule (A/U / C/G), bringing in specific amino acid (AA).
- ③ Each time the ribosome reads the codon, tRNA attaches to mRNA, leaving behind the amino acid.
- ④ Amino acids are added to growing polypeptide chain.
- ⑤ Translation stops when ribosome reaches stop codon on mRNA.
- ⑥ Polypeptide chain (PPC) is released.

DNA is not directly translated into the polypeptide chain, since =

① DNA is a double-stranded molecule, & ribosomes can only read single-stranded mRNA like mRNA, so DNA needs to firstly be transcribed into mRNA to be getting read by ribosomes. Also, ribosomes are only found in the cytoplasm, NOT the nucleus!

② DNA is too large to leave the nucleus pores or also too valuable to get damaged leaving the nucleus. Hence, DNA gets transcribed to transportable mRNA in order to leave nucleus pores (mRNA is small enough) to go to the ribosomes in the cytoplasm.   
 to be translated to the polypeptide chain

Further emphasising the importance of having transportable mRNA to reach the ribosomes in cytoplasm.

Hence, mRNA instead of DNA is directly translated into polypeptide chain.

### QUESTION THREE: The environment and gene expression in plants

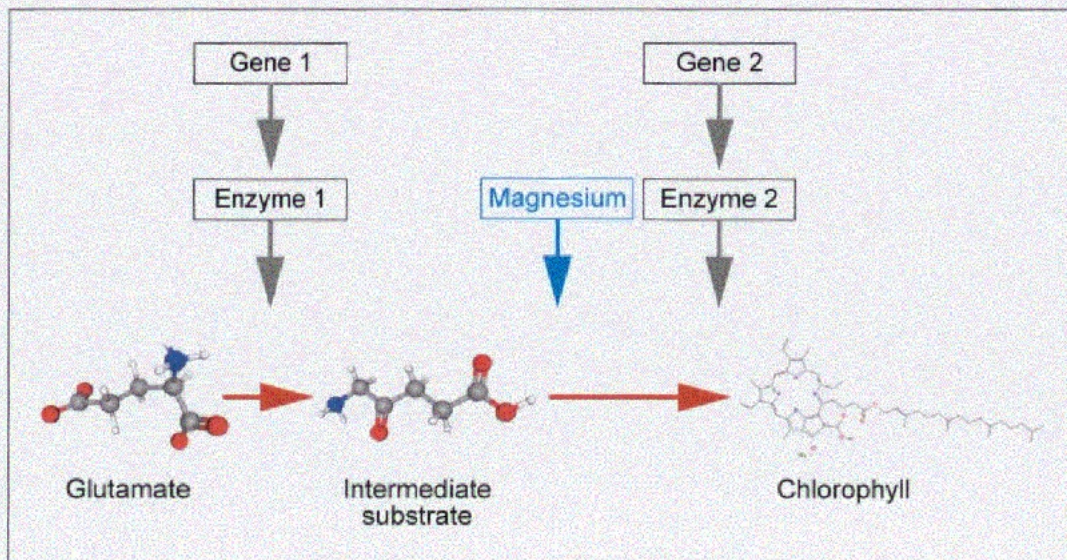
Chlorophyll is the pigment responsible for the green colour in leaves. The production of chlorophyll is influenced by both genetic factors and environmental conditions such as magnesium availability. In plants with chlorosis, genetic mutations or the lack of magnesium availability can lead to reduced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



*Gene is a section of DNA that codes for particular protein*

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.

Metabolic pathway is a series of enzyme catalysed biochemical rxn connected by their intermediates, where the product of one rxn becomes the substrate for the other. One gene codes for one enzyme. Enzyme is a biological catalyst that speed up rxn. Enzyme is specific to rxn only catalyse one rxn due to its unique shape. Many genes have many enzymes are involved in the metabolic pathway. Gene 1 codes for enzyme 1, which catalyses & converts the substrate <sup>substrate</sup> into product, intermediate substrate. Gene 2 codes for enzyme 2. With the <sup>presence of</sup> reactant of Mg, enzyme 2 catalyses & converts the intermediate substrate into final product (last product of metabolic pathway).

Mutation is a permanent & random change to DNA base sequence <sup>caused by</sup> (chemical, physical, biological) or error in DNA replication before cell division. A mutation in gene 1 <sup>(end of gene 2)</sup> can terminate chlorophyll production. Mutation in gene 1 codes for a non-functional enzyme 1 so that enzyme 1 loses its active site & cannot bind to catalyse / convert substrate into product, intermediate substrate. This leads to an accumulation of substrate & an absence of intermediate substrate & final product of chlorophyll despite gene 2 & enzyme 2 being functional, as the metabolic pathway has been terminated. Whereas in gene 2 mutation, enzyme 2 loses its active site & cannot bind to catalyse / convert intermediate product to final product (Chlorophyll). This leads to an accumulation of intermediate substrate & an absence of final product, Chlorophyll, despite gene 1 & enzyme 1 can still be functional. The absence of chlorophyll mean that chlorosis ensues, resulting in yellow/pale leaves.

Environmental factor is an internal or external factor that changes the phenotype without changing the genotype of organism / plant e.g. Mg availability. Phenotype of leaf color is produced by interactions between the genotype (which is genetically identical in isosexually reproducing plants) and the environmental factor. Mg availability. Mg availability can influence

Genotype + Environment → Phenotype

The gene expresses a limit potential genotype expression having full growth potential of green strategy least from being realised due to low Mg availability. This is since Mg is a reactant in the 2nd rxn in metabolic pathway & is needed for the enzyme 2 to catalyse & convert intermediate substrate into final product chlorophyll. Without Mg, this rxn cannot occur, leading to a buildup of intermediate substrate & an absence of final product chlorophyll, leading to chlorosis & yellow/pale leaves.

eg UV light<sup>12</sup>, alcohol, viruses.  
Mg is not a mutagen (environmental factor that can cause permanent random change to DNA base sequence) since Mg does not change the genotype of organism.

In Genotype + Environment  $\rightarrow$  Phenotype

As genotype is the same in plants that asexually reproduce (genetically identical), then when the phenotype of leaf color changes from green to yellow, the environmental factor of Mg availability must be wrong it.

When Mg is abundant/available in the soil, the Mg is present as a reactant so that enzyme 2 can catalyze & convert the intermediate substrate into final product (chlorophyll). Hence, the growth potential of the plant is fully realized due to the abundance of environmental factor Mg, allowing phenotype of leaf color to be fully expressed in green leaves instead of yellow leaves.  
This is since chlorophyll is now being produced, preventing chlorosis.

## Excellence

**Subject:** Biology

**Standard:** 91159

**Total score:** 24

Q	Grade score	Marker commentary
One	E8	<p>The response identified mutation 1 and 2 as substitution mutations and explained the effects on amino acids sequence. It linked amino acid sequence to final protein shape, functioning, and symptoms or no symptoms of cystic fibrosis.</p> <p>It compared substitution mutation to a frameshift mutation. The effects of frameshift mutations is linked to amino acid sequence / length and final protein shape / functioning and severity of cystic fibrosis symptoms.</p>
Two	E8	<p>The response discussed:</p> <ul style="list-style-type: none"><li>• the relationship between codons, anticodons, tRNA, mRNA, and amino acids</li><li>• the reasons DNA is not directly translated into a polypeptide chain and</li></ul> <p>The response also explained transcription.</p>
Three	E8	<p>The response discussed the specific metabolic pathway:</p> <ul style="list-style-type: none"><li>• including how chlorosis can be cause by mutations to genes 1, 2, or both</li><li>• including how chlorosis can be caused by magnesium availability linked to enzyme 2's functionality.</li></ul>