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# 2

91159



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## Level 2 Biology 2022

### 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 2–12 in the correct order and that none of these pages is blank.

Do not write in any cross-hatched area (X). This area may be cut off when the booklet is marked.

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

**Excellence**

**TOTAL**

**22**

## QUESTION ONE: PROTEIN SYNTHESIS

Translation is an important step in protein synthesis.



Source: <https://www.thinglink.com/scene/750792545688092673>

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

In your answer:

- ✓ • describe protein synthesis
- explain translation
- discuss the relationship between codons, anticodons, tRNA, mRNA, and amino acids
- justify with TWO reasons why DNA is not directly translated into a polypeptide chain.

Protein synthesis is the process that ~~trans~~ creates a <sup>functional</sup> protein from the genetic code found in DNA. It first involves the process of transcription which creates a strand of mRNA complementary to the template strand on DNA, which then travels ~~into~~ out of the nucleus via a nuclear pore and into the cytoplasm, where it attaches to a ribosome to begin the process of translation. Translation is the process which forms a polypeptide chain of <sup>specific complementary</sup> amino acids based off of the codons in the mRNA strand. This alone doesn't form a functioning protein, ~~and~~ in order for protein synthesis to be complete, the primary structure of the protein (polypeptide chain of amino acids) needs to undergo folding in order to



★ A codon is a group of three consecutive bases on mRNA

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hold the correct shape and hence function properly as a protein. ~~During~~ ★ Translation begins when the ribosome reads the start codon. From here, the ribosome (an organelle <sup>in the cytoplasm</sup> that reads codons during translation) brings in the correct tRNA molecule that ~~has~~ has a complementary anticodon to the codon on the mRNA strand according to the <sup>complementary</sup> base pairing rules for mRNA, where adenine pairs with uracil, and cytosine pairs with guanine. This tRNA also carries a specific amino acid which is coded for by the mRNA codon. This process repeats, and the <sup>specific</sup> amino acids brought in by the tRNA molecules form peptide bonds between one another to form a polypeptide chain of amino acids. This polypeptide grows until the ribosome reaches the stop codon which ends translation and signals for the ribosome to release the mRNA strand and the newly formed polypeptide chain so that it can begin the folding process to form a functional protein. DNA is a very valuable double stranded, helix-shaped molecule that contains an individual's original copy of their genetic information. One reason why DNA isn't directly translated into a polypeptide chain is that, ~~the~~ travelling outside of the nucleus where it is stored safely into the cytoplasm would risk this original copy of the genetic code getting damaged which would cause disastrous consequences if segments of DNA were damaged as they could no longer be transcribed into correct mRNA strands or translated into <sup>functional</sup> polypeptide chain that would fold to be a functional protein. ~~mRNA is~~ In addition, DNA is a very long molecule so it would be unnecessary and inefficient for it to travel through the cytoplasm and be read fully by the ribosome just to find the small section that codes for a protein. In both instances mRNA is better suited for being translated into a polypeptide chain because it is a copy <sup>of the DNA</sup> so it doesn't matter if it gets damaged in the cytoplasm, and because it's a shorter molecule that only carries the information needed to form a <sup>correct</sup> polypeptide chain.



## QUESTION TWO: THE GENETIC CODE

A mutation in the gene coding for the enzyme tyrosinase causes albinism, a condition that results in a decrease in the production of the pigment melanin. These individuals have albino phenotypes, because melanin gives pigment to their skin, hair, and eyes.



Source: <https://www.quora.com/Can-animals-have-albinism>

(a) Describe what a mutation is.

A mutation is a <sup>random</sup> permanent change in the base sequence of DNA.

(b) Different point mutations can cause albinism.

Discuss the effect of point mutations on final proteins. *major, minor/minimal*

In your answer:

- ✓ describe an insertion, deletion, and substitution mutation
- ✓ name the type of point mutation that is unlikely to change the protein, and explain why
- ✓ name the type of point mutation that would change the protein the most, and explain why *frameshift*
- ✓ discuss how these mutations affect the length and expression of the DNA base sequence *start & stop codons changing length of chain*
- ✓ discuss the degeneracy in the genetic code.

Insertion mutations are when a base is randomly added to the DNA base sequence. This causes a reading 'frameshift'



\* because the polypeptide chain of amino acids is unchanged and can correctly fold.

as every codon after this mutation changes as each base moves one space to the ~~left~~<sup>right</sup>. Whereas, deletion mutations are when a DNA base is randomly removed from the DNA base sequence. This also causes a reading 'frameshift' as each codon after this mutation changes as each base ~~after this mutation~~ moves ~~to~~ one space to the left. A substitution mutation is when a base is randomly swapped out and replaced with another base in the DNA base sequence. There are multiple different types of substitution mutations, for example a missense substitution mutation is where the codon has changed and ~~the~~ codes for a different amino acid that may or may not affect the final functioning of the protein. While, a silent substitution mutation is when although the codon has changed and has a different sequence of three bases, the amino acid coded for is the same. This is ~~due to~~ because the genetic code is redundant due to degeneracy within the code, as multiple codons can code for the same specific amino acid. ~~Also~~ Also, a nonsense substitution mutation is when the <sup>new</sup> codon ~~then~~ coded for is a stop codon which would hence stop translation prematurely and shorten the DNA base sequence that forms a (shortened) polypeptide chain. The point mutation that is unlikely to change the protein's <sup>shape and</sup> ability to function is a silent / same sense substitution, as explained above where the amino acids in the polypeptide chain are all the same, meaning the polypeptide chain can fold correctly, leading to a functional protein that is unchanged. Although frameshift mutations such as insertion and deletion mutations have a major effect on the final protein because

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all of the codons after the mutation are different, forming a different polypeptide chain of amino acids that is unlikely to be able to fold to produce a functional protein, nonsense substitution mutations have <sup>the ability to make</sup> an even larger impact on the final protein because they can stop translation early, causing the final polypeptide chain of amino acids to be incomplete, so it can't fold properly at all, resulting in a completely non-functional (shortened) protein. Insertion mutations make the DNA base sequence longer and causes the expression of this genetic code (phenotype) to be entirely incorrect and may result in <sup>if the gene coding for the enzyme tyrosinase is mutated, the</sup> albinism as <sup>the</sup> protein producing melanin ~~could~~ <sup>will</sup> be non-functional. Additionally, deletion mutations make the DNA base sequence shorter and could affect the organisms phenotype in the same way as described for insertion mutations.

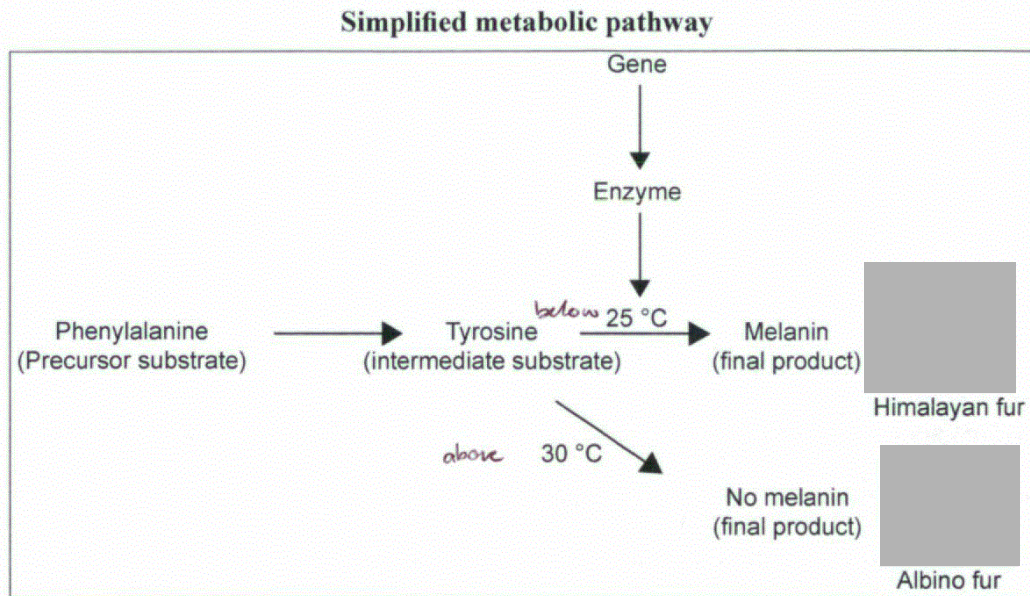
~~silent~~ <sup>silent</sup> & missense Substitution mutations don't change the length of the DNA base sequence unless it's a nonsense substitution mutation which shortens the DNA base sequence as described previously. Even though <sup>missense</sup> substitution mutations don't change the length, they could still cause albinism, while a silent substitution mutation will result in the normal phenotype as the enzyme tyrosinase that produces melanin and prevents albinism is fully functional.



### QUESTION THREE: GENE EXPRESSION AND ENVIRONMENT

Himalayan rabbits show a mutant form of albinism that is temperature sensitive.

At lower temperatures (below 25 °C), the gene produces melanin (black) in fur. The enzyme is inactive at higher temperatures (above 30 °C) and produces no melanin (white) in fur. The average body temperature of a Himalayan rabbit is 37 °C.



Adapted from: <https://arba.net/recognized-breeds/> and <https://animalcorner.org/rabbit-breeds/himalayan-rabbit-breed/>

Discuss how genes, enzymes, and the environment control the expression of melanin in Himalayan rabbits.

In your answer:

- ✓ describe what a metabolic pathway is
- ✓ explain a metabolic pathway using the terms precursor, substrate, intermediate, enzyme, gene, and final product
- ✓ discuss why Himalayan rabbits have dark melanin expression in their body extremities such as nose, ears, feet, and tail, and not in their core body
- discuss the environmental conditions necessary for Himalayan rabbits to express melanin (all black) AND no melanin (all white) fur colour for their entire life span.

A metabolic pathway is a series of enzyme controlled reactions where the product of one reaction becomes the substrate of the next. In this metabolic pathway, the precursor ~~and~~ substrate (phenylalanine) is converted to tyrosine which is an intermediate substrate by an enzyme (a biological catalyst that forms products from substrate's) which was coded for by a specific gene (not shown in the metabolic

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pathways above). Genes code for the enzymes that can catalyse the reaction from a specific substrate to a specific product. Tyrosine is then converted to the pigment, melanin if the <sup>body</sup> temperature of Himalayan rabbits is below  $25^{\circ}\text{C}$  by an enzyme coded for by a gene<sup>resulting in the mutant form of albinism</sup>. However, if the rabbit's body temperature is above  $30^{\circ}\text{C}$  then the enzyme will be nonfunctional<sup>(inactive)</sup> (caused by this environmental factor) and won't be able to catalyse the reaction ~~between~~<sup>from</sup> tyrosine to melanin, so no melanin is produced, resulting in the Himalayan rabbit's final phenotype showing the normal form of albinism. Himalayan rabbits have dark melanin expression on areas<sup>of their body</sup> such as their nose, ears, and feet because these areas are colder, while the core<sup>body</sup> temperature of the rabbit is warmer. Because these areas are colder and ~~the~~ ~~are~~ likely less than  $25^{\circ}\text{C}$ , the enzyme<sup>converting tyrosine to melanin</sup> will be active and will result in only these specifically colder areas expressing the black fur phenotype, while the rabbit's core area will show white fur as the temperature is hotter than  $30^{\circ}\text{C}$  and is too warm for the enzyme producing melanin to function. Environmental factors are internal or external factors that could affect the final expression of an organism's phenotype. In order for Himalayan rabbits to express a completely black phenotype, the enzyme ~~catalysing~~<sup>catalysing</sup> the reaction from tyrosine to melanin needs to be functioning and active. Therefore all parts of the rabbit's body need to be below  $25^{\circ}\text{C}$ . So, the temperature of the rabbit's surrounding external environment needs to decrease (the rabbit could move to a colder environment). This external temperature needs to be consistently low for the rest



of the rabbit's life span because this will ensure that the enzyme remains functioning and producing melanin. ~~The oppos~~ Whereas, in order for a rabbit to produce no melanin for the rest of their life span, the enzyme needs to be inactive which occurs at body temperatures above  $30^{\circ}\text{C}$ . This is possible, as the average body temperature of a rabbit is  $37^{\circ}\text{C}$ . The rabbit's external environment would need to increase in temperature, in order for its internal temperature to remain high and keep the enzyme inactive so no melanin is produced.







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<b>Standard</b>	91159	<b>Display ID</b>	62165519	<b>Total score</b>	22
		<b>NSN</b>			
<b>Q</b>	<b>Grade score</b>	<b>Annotation</b>			
1	E7	<p>How mRNA/tRNA have specific amino acid by mentioning complementary nature.</p> <p>Explains translation including the idea of a start and stop codon.</p> <p>Explains the function of mRNA as a message/template of the gene/genetic code.</p> <p>Discusses translation and two reasons why DNA is not directly translated into a polypeptide chain.</p>			
2	E8	<p>Discusses degeneracy of the code is able to buffer the effect of substitution mutation and therefore the amino acid sequence mostly likely remains the same therefore folds (shape) the same and the function will be the same.</p> <p>Discuss how deletion/insertion mutation most likely causes the amino acid sequence to change and therefore the shape (folding) of the protein will be different and will effect function.</p>			
3	E7	<p>Explains the relations between genes, enzymes and products of a metabolic pathway.</p> <p>Discusses how the rabbits have black fur in all parts of their body for their entire life.</p>			