



# Exercises on DNA sequence mutations

24 February 2026

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## 1 – Targeted outcomes

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1. Define the term 'mutation' as

*'any spontaneous change in the genetic code'*

2. Explain how DNA mutation can result in beneficial, harmful or minimal effects on the functioning of an organism.
3. State some factors which can cause a mutation.
4. Describe
  - i. Substitution mutations
  - ii. Insertion mutations
  - iii. Deletion mutations
  - iv. Chromosomal number mutations
5. Describe different consequences of mutations on the amino acid sequence and resultant protein function using a codon chart.

## 2 – Exercises

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In each of the following *three* scenarios,

- i. Indicate where within the *DNA sequence* the *mutation* has occurred.
- ii. State what *type* of mutation has occurred and describe the *nature* of this mutation type.
- iii. State one potential factor which could have caused the mutation.
- iv. Transcribe the *DNA sequence* to *mRNA*, and convert the resulting *mRNA* sequence to an *amino acid sequence* using the provided codon chart, then explain the consequence of the *DNA mutation* on said *amino acid sequence* and the resultant protein's function.
- v. State whether and explain why the effect of the mutation on the organism would be either *harmful, beneficial, or minimal*.

*(Note: the DNA sequences used in these exercises are not the actual DNA sequences of the genes discussed)*

## Scenario 1

The *LCT* (*Lactase*) gene provides instructions for making the enzyme *lactase*. Lactase breaks down *lactose*, the natural sugar in milk and dairy products.

*Lactose intolerance* is the inability to fully digest *lactose* due to insufficient *lactase* enzyme production in the small intestine as a result of *LCT* gene inactivity – potentially causing diarrhea and stomach cramps within 30 minutes to 2 hours of dairy consumption.

*Lactase persistence* results from a mutation in the *LCT* gene that allows it to remain active into adulthood, allowing individuals to continue to digest *lactate*.

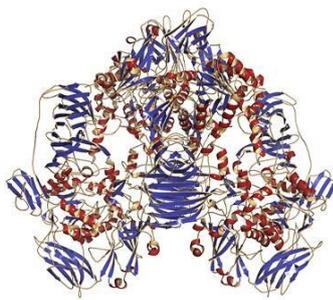


Figure 1. Three-dimensional shape of the lactase enzyme.

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

### Lactose intolerance

<i>LCT</i> gene (coding)	ATGAATCCAGCTTGA
<i>mRNA</i> transcription	
Amino acid sequence	

### Lactase persistence

<i>LCT</i> gene (coding)	ATGAAACCAGCTTGA
<i>mRNA</i> transcription	
Amino acid sequence	

## Scenario 2

The *PADI3* gene encodes the *peptidyl arginine deiminase 3 (PAD3)* enzyme. *PAD3* converts arginine residues to citrulline in proteins, which allows hair follicles to form.

*Uncombable hair syndrome (UHS)* is a rare hair shaft disorder characterized by dry, frizzy, silvery-blond or straw coloured hair (see *Figure 2*) that stands out from the scalp and cannot be combed flat. *UHS* most commonly results from a mutation in the *PADI3* gene.



*Figure 2. Uncombable hair syndrome.*

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

### *Combable hair*

<i>PADI3</i> gene (template)	ATGCACGAGCGATGA
<i>mRNA</i> transcription	
<i>Amino acid</i> sequence	

### *Uncombable hair syndrome*

<i>PADI3</i> gene (template)	ATGCACGAGACGATGA
<i>mRNA</i> transcription	
<i>Amino acid</i> sequence	

### Scenario 3

In a species of bacteria called *Paenarthobacter ureafaciens* (KI72), the *nylA* gene can encode for the *NylA* enzyme. *NylA* allows KI72 to consume nylon waste, such as old activewear and toothbrush bristles.

The *nylA* gene likely originated in KI72 from a mutation of a pre-existing gene, which we will call *ABC*.



Figure 3. Three-dimensional shape of the *NylA* enzyme.

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

*KI72* incapable of producing *NylA* and consuming nylon

<i>ABC</i> gene (template)	ATGCTGCCAGATTGA
mRNA transcription	
Amino acid sequence	

*KI72* capable of producing *NylA* and consuming nylon

<i>nylA</i> gene (template)	ATGCTGCCAATTGA
mRNA transcription	
Amino acid sequence	

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