



# GENE MUTATIONS AND VARIATIONS

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# MUTATIONS (FROM GREEN BOOK)

- Any alteration in an organism's DNA is called a mutation.
- Many mutations don't impact the organism, particularly in eukaryotes, as much of the DNA doesn't affect its physical traits.
- Mutations that do affect traits often lead to lethality, as most genes are vital for survival.
- A minority of mutations result in visible, non-lethal changes in the organism's appearance.

# CHARACTERISTICS OF MUTATION

**Nature of Change** - alteration in the sequence of nucleotides in DNA.

**Frequency** - randomized frequency occur at a very low frequency. Physical and chemical mutagens can enhance this rate.

**Mutation Rate** - mutable genes, mutator genes, anti-mutator genes.

**Direction of Change** - wild type to mutant allele.

**Effects Site of Mutation** - generally lethal, only 0.1% of them are useful.

**Type of Event** - any gene, any cell and any stage.

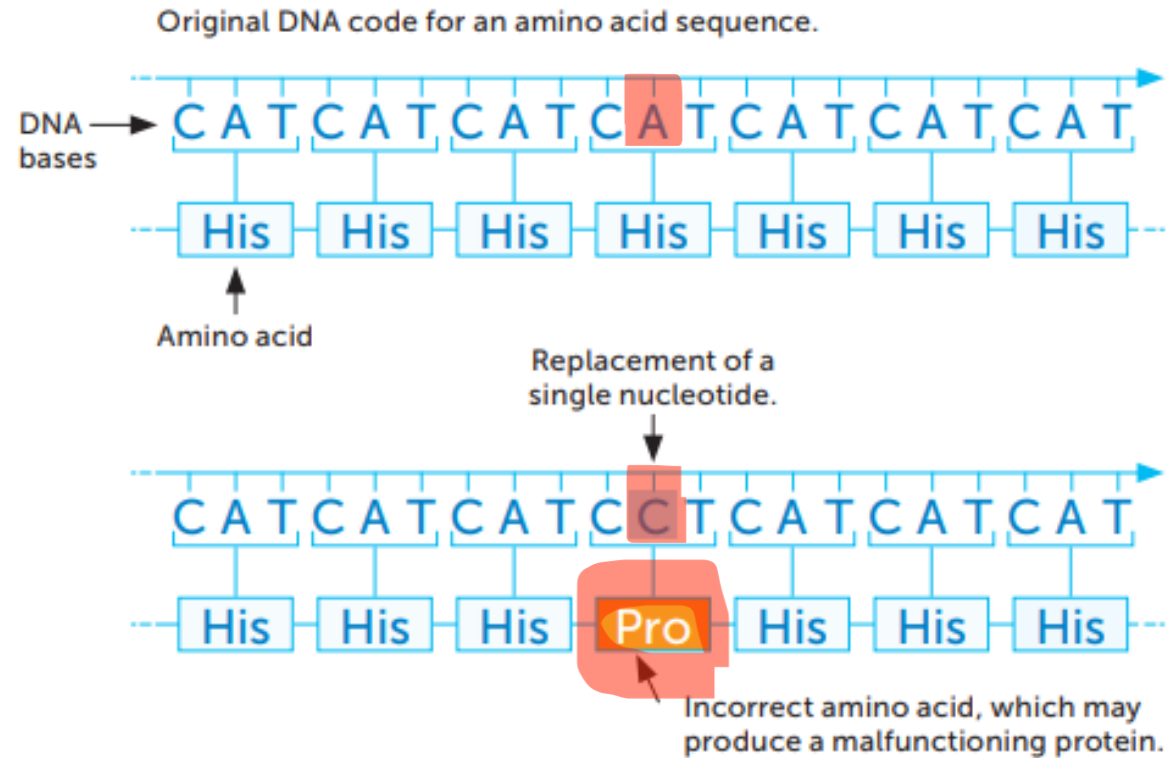
**Recurrence** - same mutation may occur repeatedly.

**Site of Mutation** - Hot spots - 500-1000 mutational sites.

## MISSENSE MUTATION

A mutation that **changes one single base pair** causes the substitution of a different **amino acid** in the resulting protein.

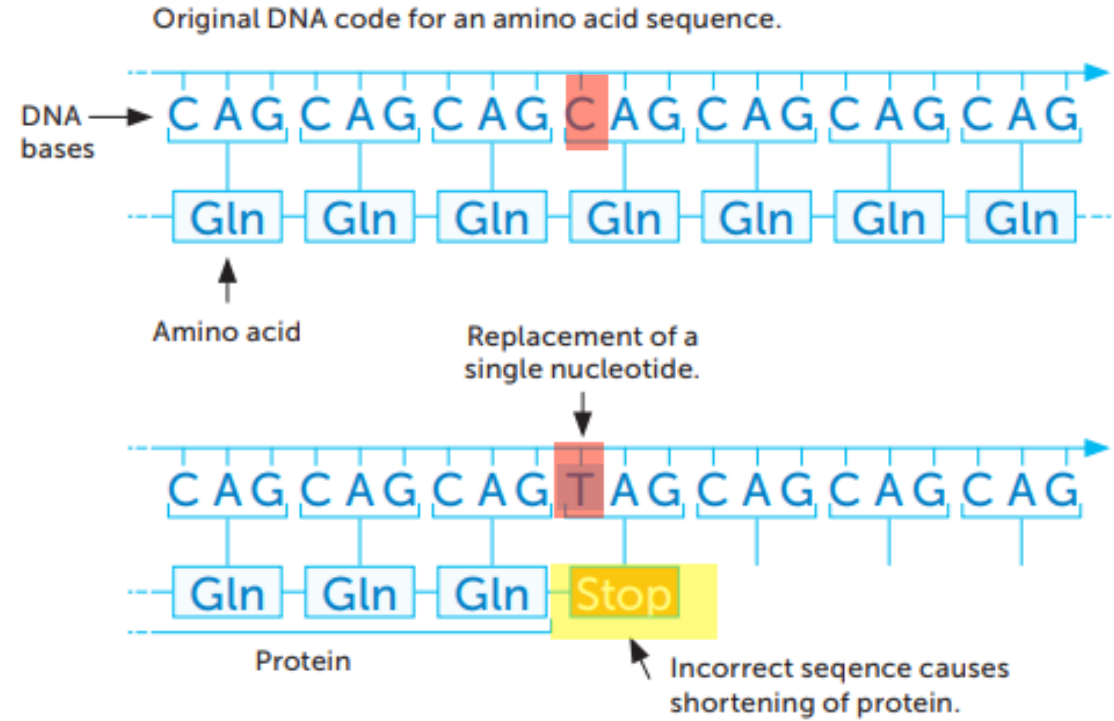
In this example, **the nucleotide adenine is replaced by cytosine in the genetic code**, introducing an incorrect amino acid into the protein sequence.



**Figure 1: Missense mutation**

## NONSENSE MUTATION

A nonsense mutation is also a change in single DNA base pair. Instead of substituting one amino acid for another, however, the altered DNA sequence prematurely signals the cell to stop building a protein. This type of mutation results in a shortened protein that may function improperly or not at all.



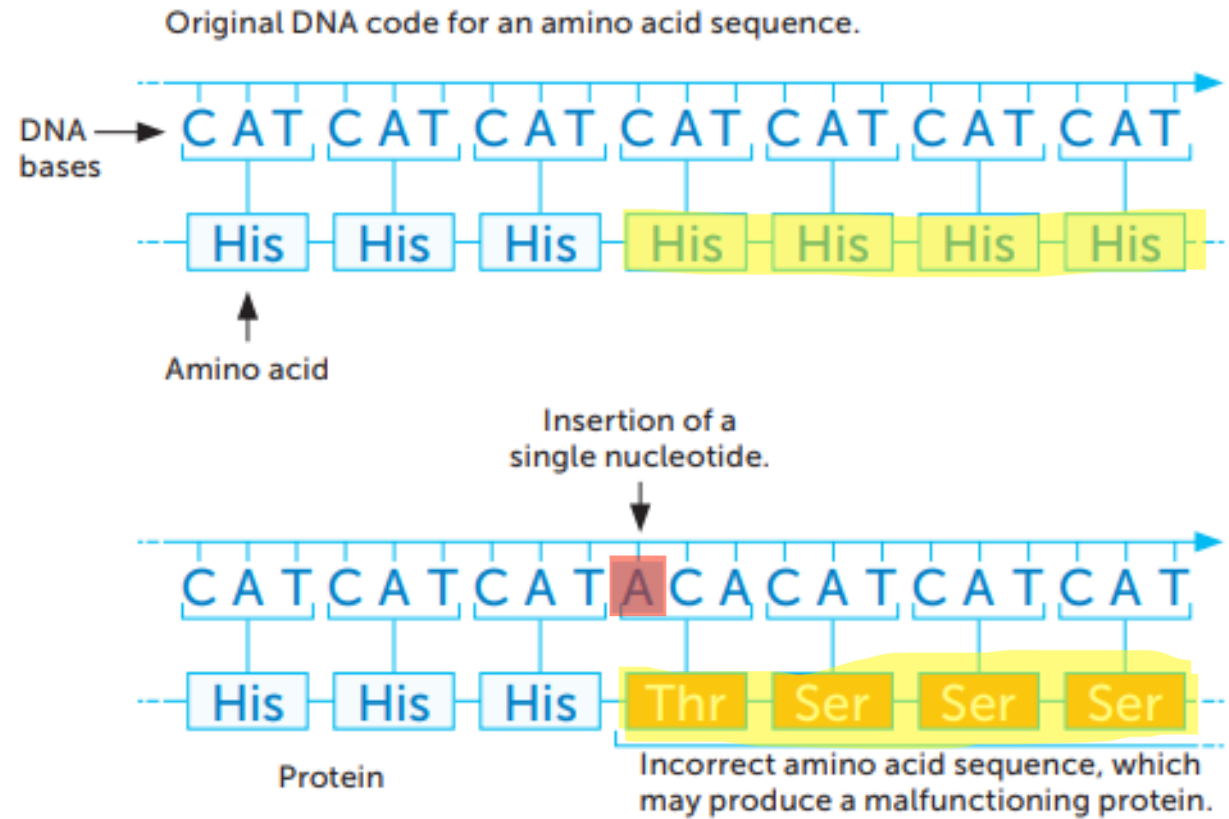
**Figure 2: Nonsense mutation**

In this example, the nucleotide cytosine is replaced by thymine in the DNA code, signaling the cell to shorten the protein.

# INSERTION

An insertion changes the number of DNA bases in a gene by adding a piece of DNA. As a result, the protein made by the gene may not function properly.

In this example, one nucleotide (adenine) is added in the DNA code changing the amino acid sequence that follows.

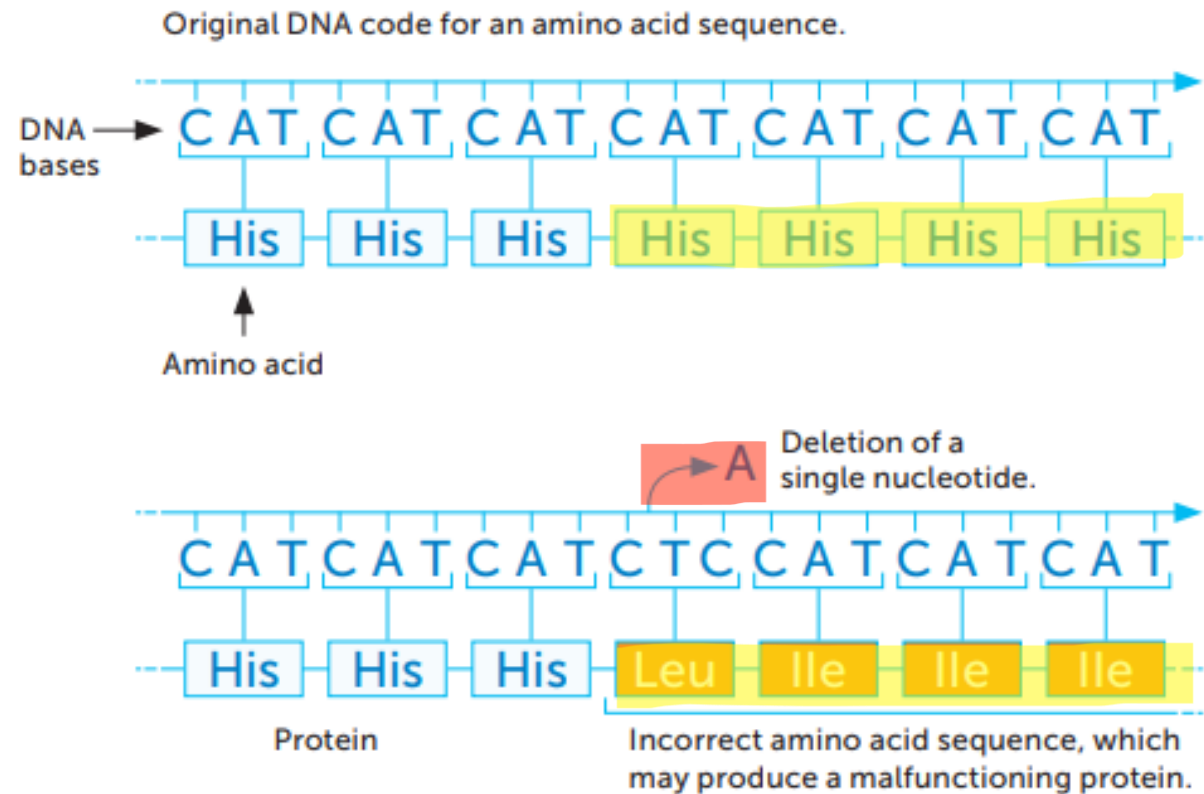


**Figure 3: Insertion**

# DELETION

A deletion changes the number of DNA bases by removing a piece of DNA.

In this example, one nucleotide (adenine) is deleted from the DNA code, changing the amino acid sequence that follows.



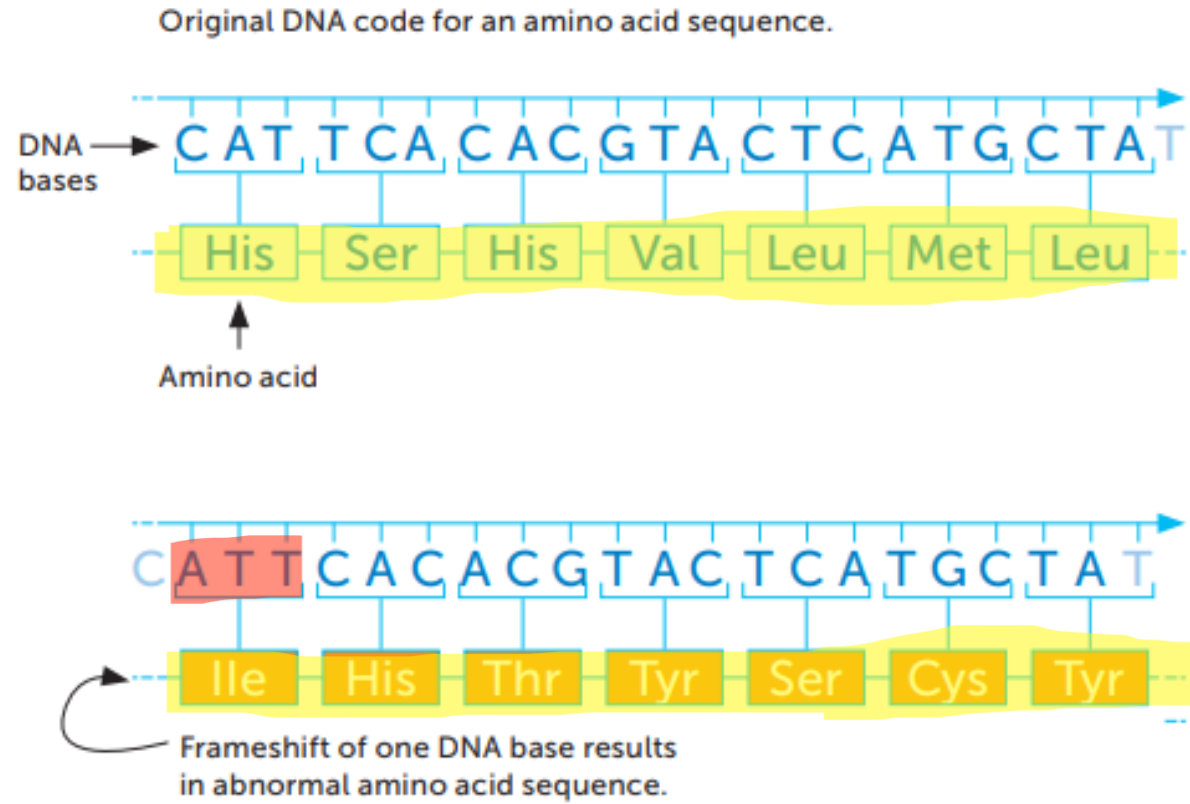
**Figure 4: Deletion**



## FRAMESHIFT MUTATION

This type of mutation occurs when the addition or loss of DNA bases changes a gene's reading frame.

A frameshift mutation shifts the grouping of these bases and changes the code for amino acids. The resulting protein is usually nonfunctional.



**Figure 5: A frameshift mutation changes the amino acid sequence from the site of the mutation.**



# ANSWER THE QUESTIONS - EXTENSION ACTIVITIES

What type of mutation is seen here?

How did the mutation affect the amino acid sequence?

Vazhdo prej faqe 56 në libër!



## Extension Activities



Original DNA sequence:	TAC	ACC	TTG	GCG	ACG	ACT
mRNA transcript:	AUG	UGG	AAC	CGC	UGC	UGA
amino acids:	Methionine	Threonine	Asparagine	Arginine	Cysteine	Stop

Original DNA sequence:	TAC	ACC	TTG	GGA	CGA	CT
mRNA transcript:	AUG	UGG	AAC	CCU	GAU	
amino acids:	Methionine	Threonine	Asparagine	Proline	Aspartic acid	