

Microbyte:

GENETIC MUTATIONS



Changes in the genetic code

- **Mutation:** Any change to the DNA sequence in the genome
 - Can be detrimental or beneficial to the organism
- Causes of mutations:
 - **Spontaneous mutation:** A random change arising from errors in DNA replication
 - **Induced mutations:** A change induced by exposure to a mutagens (e.g. radiation)

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- Causes of mutations:
 - **Spontaneous mutation:** A random change arising from errors in DNA replication
 - **Induced mutations:** A change induced by exposure to a mutagens (e.g. radiation)
- Types of mutations:
 - **Point mutation:** affect only a single nucleotide of a gene; involve addition, deletion, or substitution of a single nucleotide.
 - Substitution point mutations can result in no change to the amino acid (*silent*), a change to the amino acid (*missense*), or a premature stop codon (*nonsense*).
 - Insertion or deletion point mutations can result in frameshift mutations; however, not all frameshift mutations are point mutations
 - **Frameshift mutation:** one or more bases are inserted or deleted. Insertions and deletions will *shift* the *frame* of codons read during translation and may lead to a completely different chain of amino acids being produced.

DNA coding strand: ATGGGAACTTAG

original

DNA coding strand: ATGGGTACTTAG

point mutation (*silent*)

Separate into triplets, add the template sequence, then transcribe and translate:

DNA coding strand: ATG GGAA ACT TAG

DNA template strand: TAC CCTT TGA ATC

mRNA transcript: AUG GGAA ACU UAG

amino acid sequence: Met Gly Thr Stop

original

DNA coding strand: ATG GGT ACT TAG

DNA template strand: TAC CCA TGA ATC

mRNA transcript: AUG GGU ACU UAG

amino acid sequence: Met Gly Thr Stop

point mutation (*silent*)

DNA coding strand: ATGGGAACTTAG

original

DNA coding strand: ATGGGACCTTAG

point mutation (*missense*)

Separate into triplets, add the template sequence, then transcribe and translate:

DNA coding strand: ATG GGA ACT TAG

DNA template strand: TAC CCT TGA ATC

mRNA transcript: AUG GGA ACU UAG

amino acid sequence: Met Gly Thr Stop

original

DNA coding strand: ATG GGA CCT TAG

DNA template strand: TAC CCT GGA ATC

mRNA transcript: AUG GGA CCU UAG

amino acid sequence: Met Gly **Pro** Stop

point mutation (*missense*)

DNA coding strand: ATGGGA ACTTAG

original

DNA coding strand: ATGTGA ACTTAG

point mutation (*nonsense*)

Separate into triplets, add the template sequence, then transcribe and translate:

DNA coding strand: ATG GGA ACT TAG

DNA template strand: TAC CCT TGA ATC

mRNA transcript: AUG GGA ACU UAG

amino acid sequence: Met Gly Thr Stop

original

DNA coding strand: ATG TGA ACT TAG

DNA template strand: TAC ACT TGA ATC

mRNA transcript: AUG UGA ACU UAG

amino acid sequence: Met **Stop**

point mutation (*nonsense*)

Mutations

To view the relevant gif:
[click here](#)

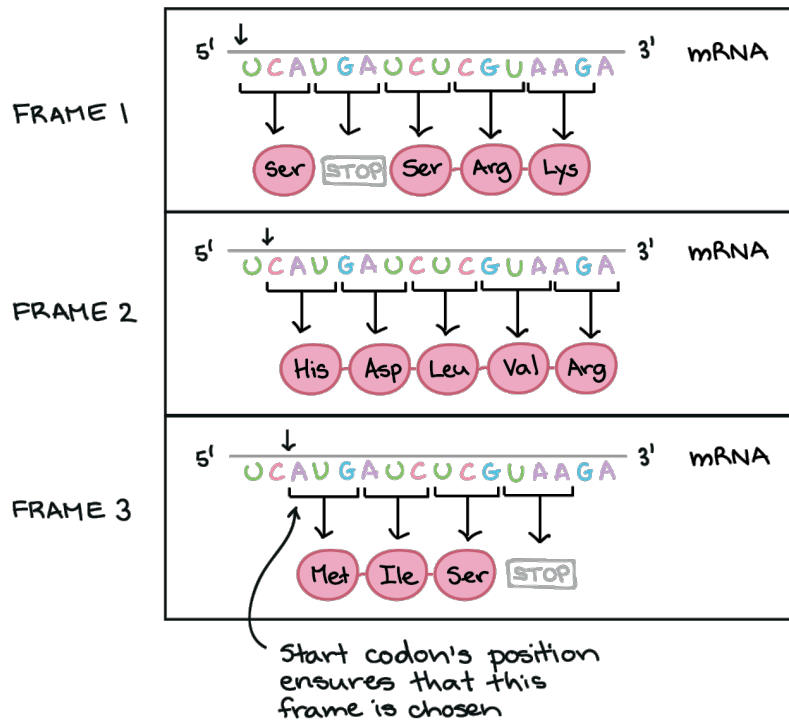
Point mutations involving the *substitution* of a single nucleotide in the DNA sequence result in a different mRNA codon read during translation and lead to one of three outcomes:

silent mutation: the amino acid stays the same

missense mutation: a change in amino acid

nonsense mutation: a premature stop codon

Reading frames and frameshift mutations



- The **reading frame** references how the sequence of an mRNA transcript is divided into codons during translation.
- Recall that:
one triplet → one codon → one amino acid
- So, ultimately the reading frame of a gene is specified in the triplet nucleotides of the DNA sequence.

image credit:
Khan Academy

- When a nucleotide(s) are inserted or deleted from the DNA sequence of a gene, the reading frame for that gene *shifts* - hence: **frameshift mutation**.
- A frameshift mutation results in the mRNA transcript specifying a different set of codons and when the ribosome reads that mutated transcript it will translate an entirely different amino acid sequence.

If a single nucleotide is inserted or deleted the frameshift mutation is also a *point mutation*

DNA coding strand: ATGGGA ACTTAG

original

DNA coding strand: ATGGTGAACTTAG

frameshift insertion (*point*)

Separate into triplets, add the template sequence, then transcribe and translate:

DNA coding strand: ATG GGA ACT TAG

DNA template strand: TAC CCT TGA ATC

mRNA transcript: AUG GGA ACU UAG

amino acid sequence: Met Gly Thr Stop

original

DNA coding strand: ATG GTG AAC TTA G

DNA template strand: TAC CAC TTG AAT C

mRNA transcript: AUG GUG AAC UUA G

amino acid sequence: Met Val Asn Leu

frameshift insertion (*point*)

DNA coding strand: ATGGGA ACTTAG

original

DNA coding strand: ATGG~~G~~AACTTAG

frameshift deletion (*point*)

Separate into triplets, add the template sequence, then transcribe and translate:

DNA coding strand: ATG GGA ACT TAG

DNA template strand: TAC CCT TGA ATC

mRNA transcript: AUG GGA ACU UAG

amino acid sequence: Met Gly Thr Stop

original

DNA coding strand: ATG ~~GAA~~ ~~CTT~~ ~~AG~~

DNA template strand: TAC ~~CTT~~ ~~GAA~~ ~~TC~~

mRNA transcript: AUG ~~GAA~~ ~~CUU~~ ~~AG~~

amino acid sequence: Met ~~Glu~~ ~~Leu~~

frameshift deletion (*point*)

DNA coding strand: ATGGGA ACTTAG

original

DNA coding strand: ATGGGGTA ACTTAG

frameshift insertion

Separate into triplets, add the template sequence, then transcribe and translate:

DNA coding strand: ATG GGA ACT TAG

DNA template strand: TAC CCT TGA ATC

mRNA transcript: AUG GGA ACU UAG

amino acid sequence: Met Gly Thr Stop

original

DNA coding strand: ATG GGG TAA CTT AG

DNA template strand: TAC CCC ATT GAA TC

mRNA transcript: AUG GGG UAA CUU AG

amino acid sequence: Met Gly Stop

frameshift insertion

Mutations

When one or more bases are *inserted* or *deleted* in DNA it shifts the “reading frame” of codons read during translation; hence: **frameshift mutation**.

One or two base frameshifts lead to an entirely different sequence of amino acids and therefore a different protein. This can be extremely detrimental to a cell.

Three base frameshifts are not usually as harmful given that the DNA code is read in triplets.

To view the relevant gif:
[click here](#)

Point mutations involving the *substitution* of a single nucleotide in the DNA sequence result in a different mRNA codon read during translation and lead to one of three outcomes:

silent mutation: the amino acid stays the same

missense mutation: a change in amino acid

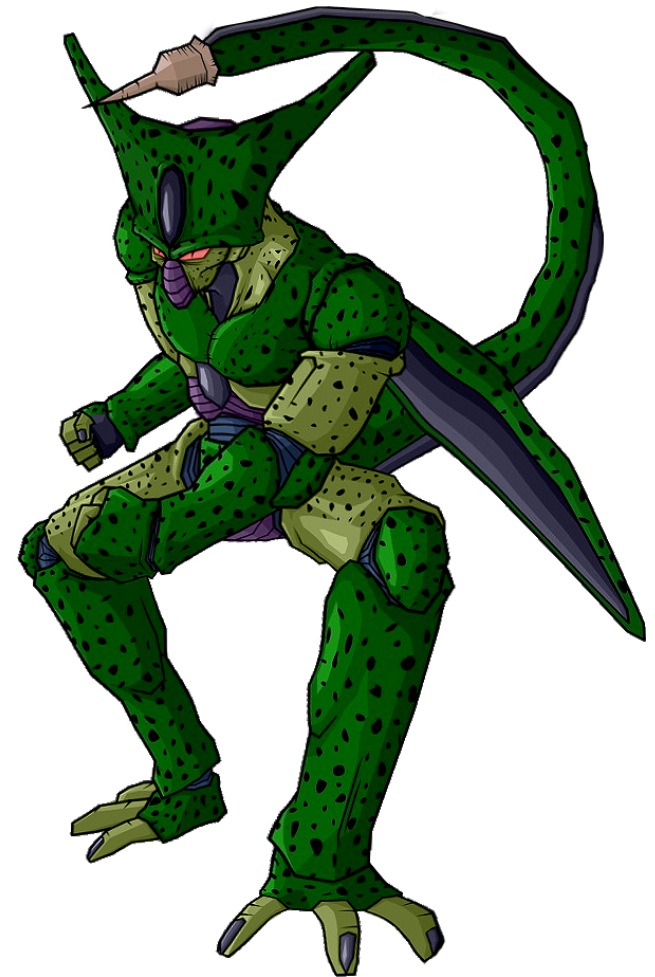
nonsense mutation: a premature stop codon

Effects of Mutations: Positive and Negative

- Many mutations are not repaired
- Effects of mutations depend on the nature of the mutation and the strategies available to the organism
- Mutations are permanent* and heritable
- Passed on to the offspring of organisms and new viruses and become a long-term part of the gene pool
- A small number of mutations contribute the success of the individual and the population
- Variant strains with alternate ways of expressing a trait can more readily adapt, survive, and reproduce

*sometimes a mutated gene will mutate back into its original sequence - this is called a **back mutation**

Imperfect Cell
(from Dragon Ball Z)



Micro is Magikal!

