M.Sc Mazin Eidan Hadi
mazin.eidan@uomus.edu.iq

Mutations

What is a mutation?

A mutation is a permanent change in the sequence of DNA. In order for an observable effect, mutations must occur in gene exons or regulatory elements. Changes in the non-coding regions of DNA (introns and junk DNA) generally do not affect function.

What causes mutations?

Mutations can be caused by external (exogenous) or endogenous (native) factors, or they may be caused by errors in the cellular machinery. Physical or chemical agents that induce mutations in DNA are called mutagens and are said to be mutagenic.

Exogenous factors: environmental factors such as sunlight, radiation, and smoking can cause mutations.

Endogenous factors: errors during DNA replication can lead to genetic changes as can toxic by-products of cellular metabolism.

What are the consequences of mutations?

Mutations can be advantageous and lead to an evolutionary advantage of a certain genotype. Mutations can also be deleterious, causing disease, developmental delays, structural abnormalities, or other effects.

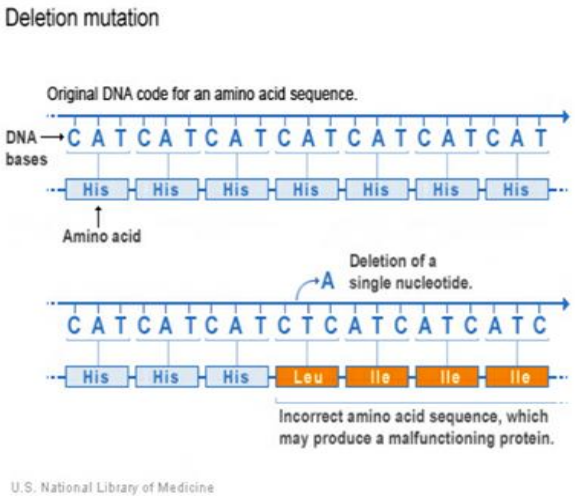
Types of Mutations:

There are many different ways that DNA can be changed, resulting in different types of mutation. Here is a quick summary of a few of these:

1- Deletion:

Genetic material is removed or deleted. A few bases can be deleted or it can be complete or partial loss of a chromosome. See figure below

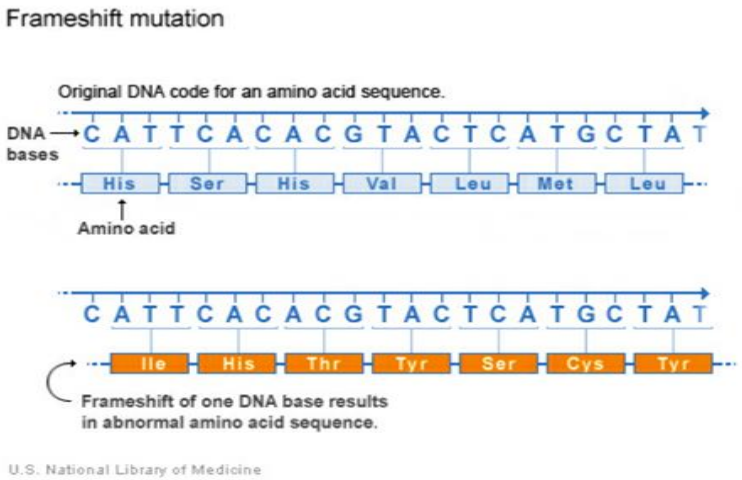
M.Sc Mazin Eidan Hadi
mazin.eidan@uomus.edu.iq



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

2- Frameshift

The insertion or deletion of a number of bases that is not a multiple of 3. This alters the reading frame of the gene and frequently results in a premature stop codon and protein truncation.

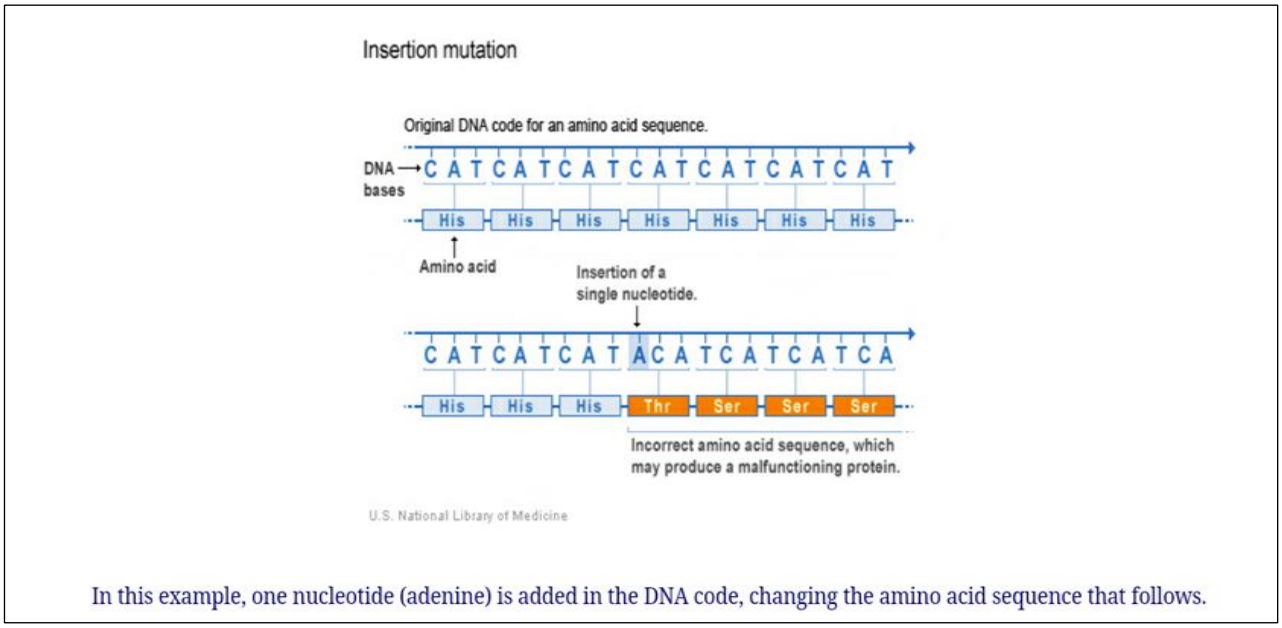


A frameshift mutation changes the amino acid sequence from the site of the mutation.

M.Sc Mazin Eidan Hadi
mazin.eidan@uomus.edu.iq

3- Insertion

When genetic material is put into another region of DNA. This may be the insertion of 1 or more bases, or it can be part of one chromosome being inserted into another, non-homologous chromosome.



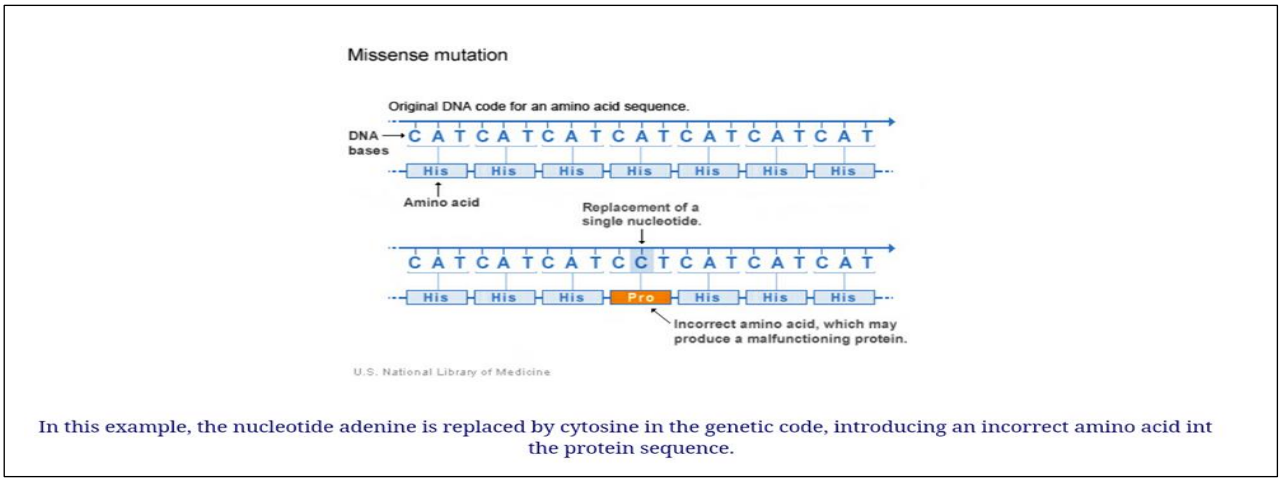
In this example, one nucleotide (adenine) is added in the DNA code, changing the amino acid sequence that follows.

4- Point

A single base change in DNA sequence. A point mutation may be silent, missense, or nonsense.

a- Missense

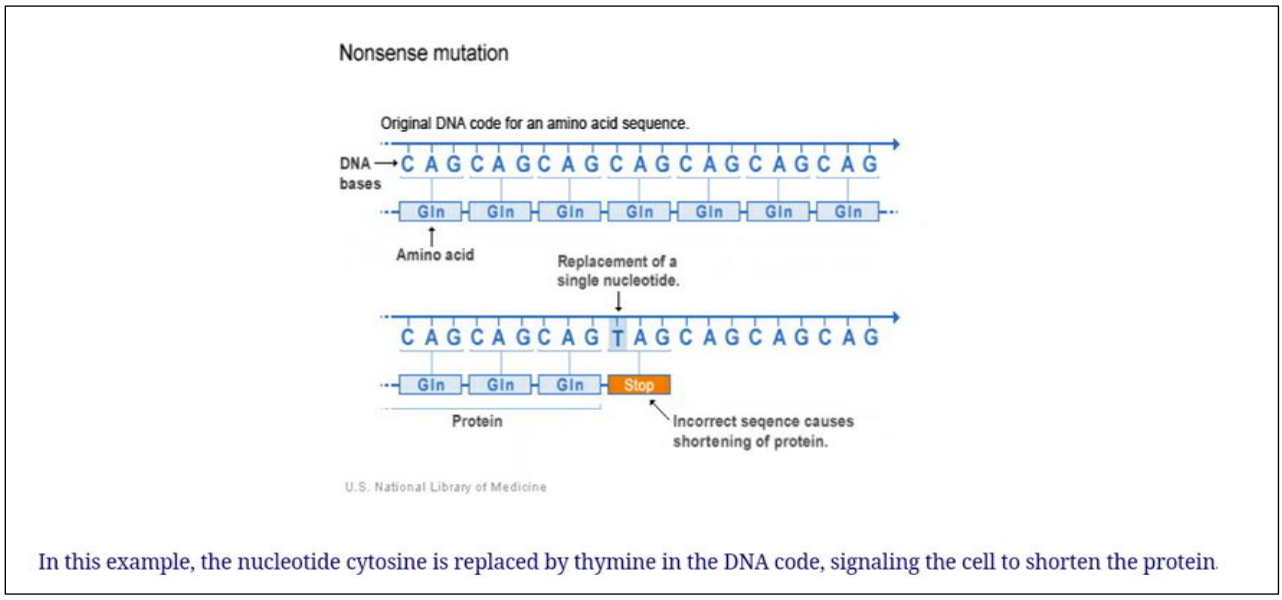
A change in DNA sequence that changes the codon to a different amino acid. Not all missense mutations are deleterious, some changes can have no effect. Because of the ambiguity of missense mutations, it is often difficult to interpret the consequences of these mutations in causing disease.



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

b- Nonsense

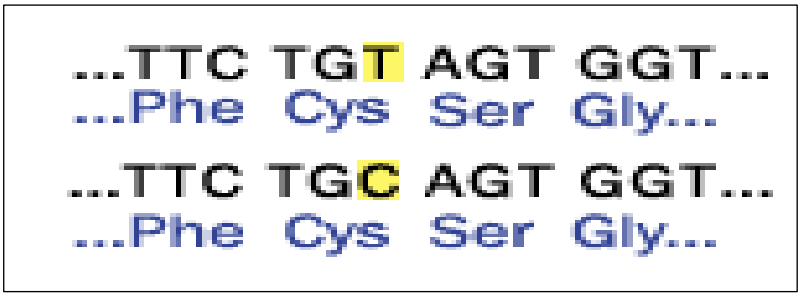
A change in the genetic code that results in the coding for a stop codon rather than an amino acid. The shortened protein is generally non-function or its function is impeded.



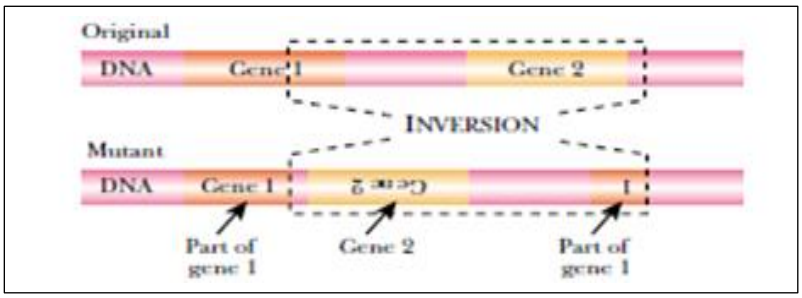
In this example, the nucleotide cytosine is replaced by thymine in the DNA code, signaling the cell to shorten the protein.

c- Silent

A change in the genetic sequence that does not change the protein sequence. This can occur because of redundancy in the genetic code where an amino acid may be encoded for by multiple codons.



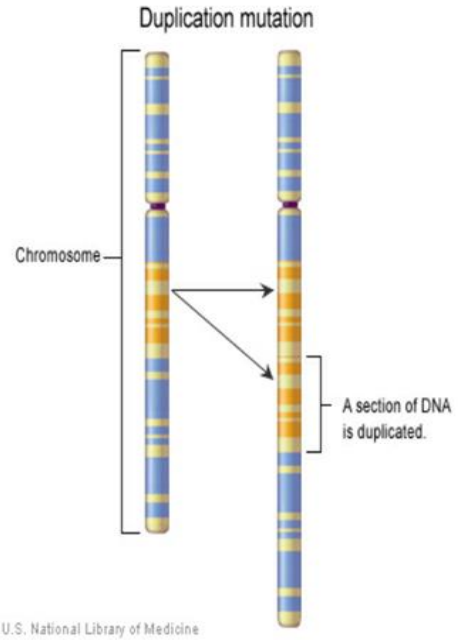
5- **Inversion**: a segment of DNA is inverted, but remains at the same overall location.



M.Sc Mazin Eidan Hadi
mazin.eidan@uomus.edu.iq

6- Duplication

A segment of DNA is duplicated; the second copy usually remains at the same location as the original.



A section of DNA is accidentally duplicated when a chromosome is copied.