MUTATION AND IT'S TYPES

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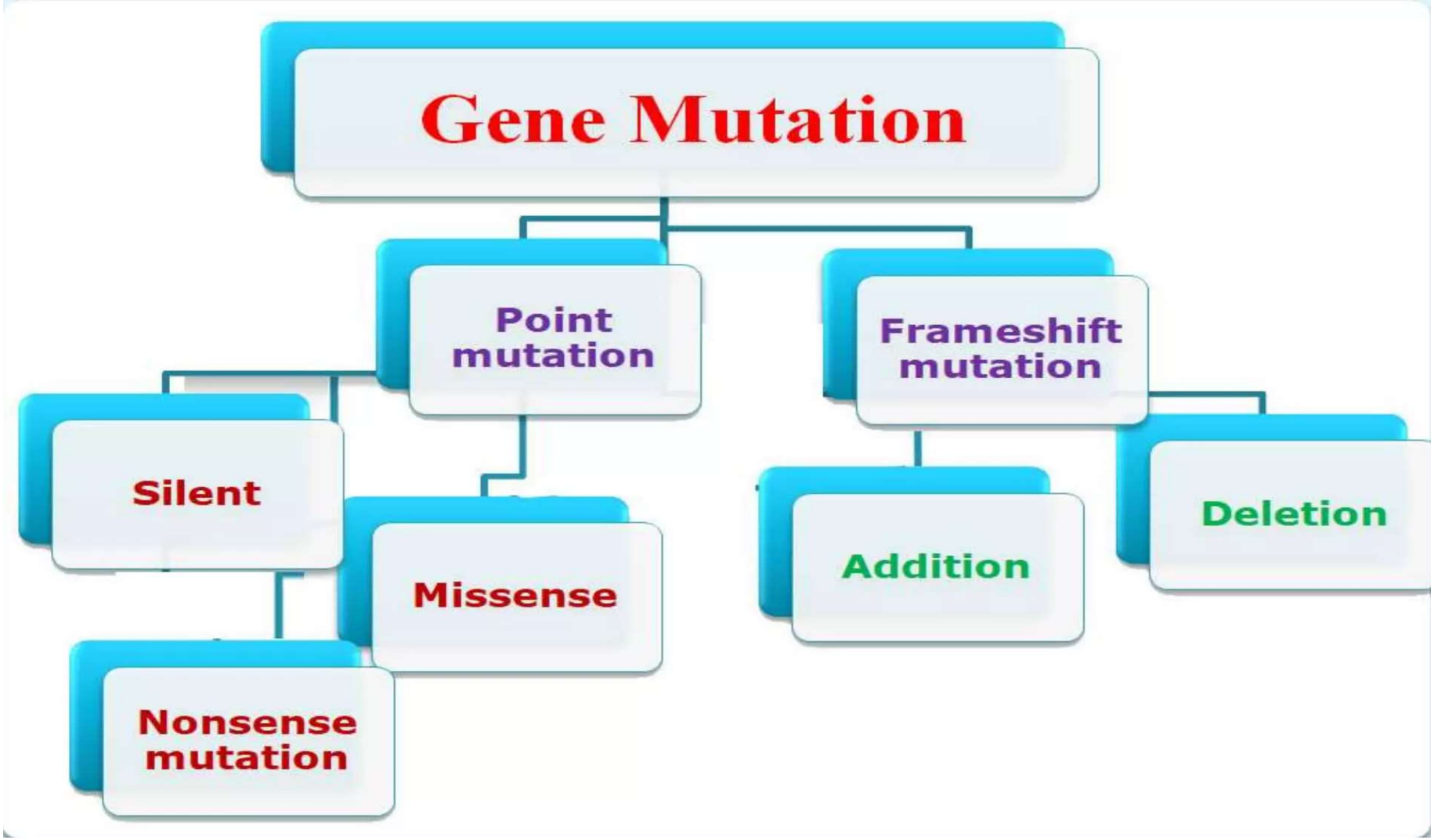
- A mutation is a change that occurs in our DNA sequence, either due to mistakes when DNA is copied or as the result of environmental factors such as UV light.

- The DNA sequence of a gene can be altered in a number of ways.

- Gene mutations have varying effects on health, depending on where they occur and whether they alter the function of essential proteins.

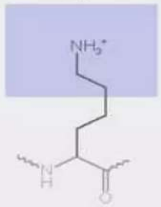
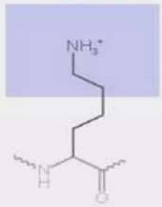
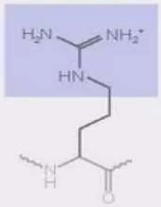
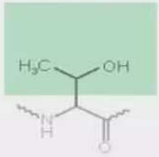
Types of Mutation

- Gene Mutation
- Chromosome Mutation


Gene Mutation
Point mutation
Frameshift mutation
Silent
Missense
Nonsense mutation
Addition
Deletion

Point mutation

A point mutation or substitution is a genetic mutation where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA.

	No mutation	Point mutations			
		Silent	Nonsense	Missense	
				conservative	non-conservative
DNA level	TTC	TTT	ATC	TCC	TGC
mRNA level	AAG	AAA	UAG	AGG	ACG
protein level	Lys	Lys	STOP	Arg	Thr
					

Silent mutations

Silent mutations are mutations in DNA that do not have an observable effect on the organism's phenotype. They are a specific type of neutral mutation.

Nonsense mutation

- A nonsense mutation is also a change in one 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.

Missense mutation

- This type of mutation is a change in one DNA base pair that results in the substitution of one amino acid for another in the protein made by a gene.

Frameshift mutation

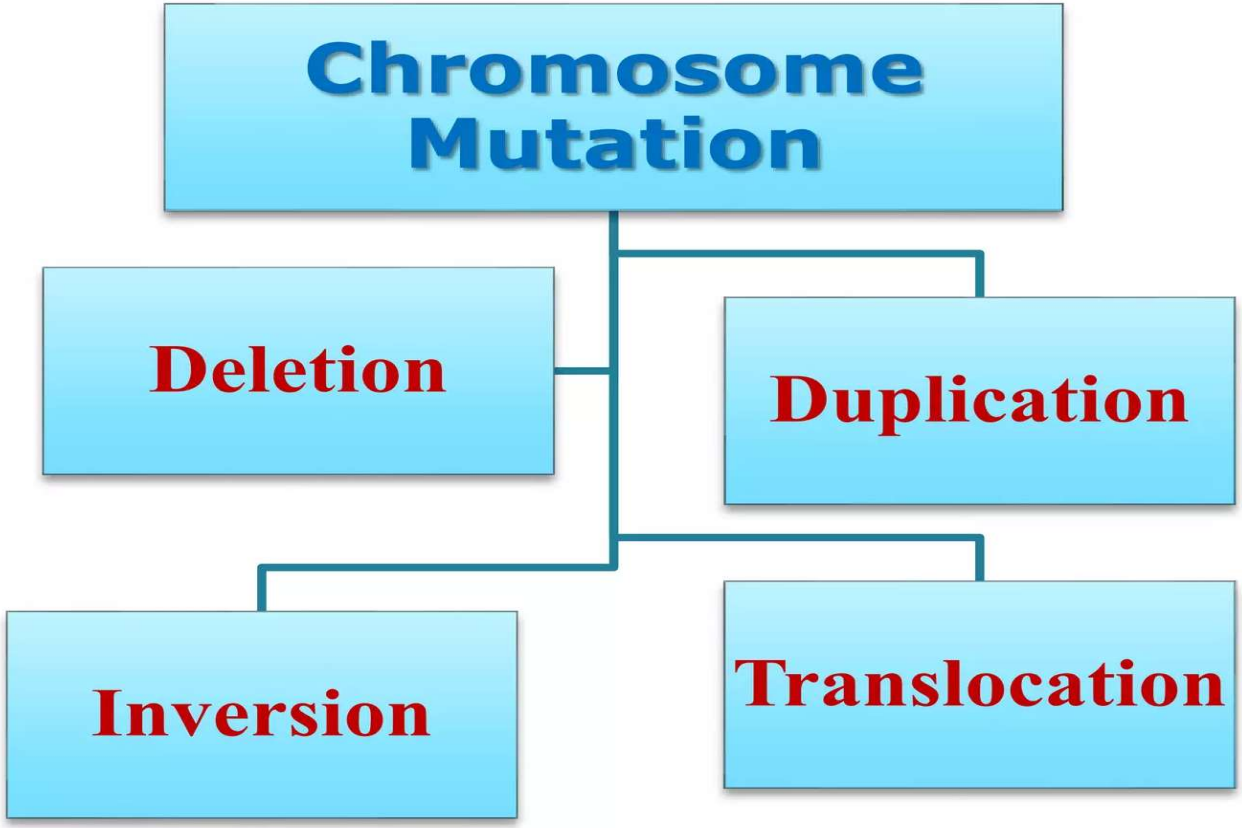
- This type of mutation occurs when the addition or loss of DNA bases changes a gene's reading frame.
- A reading frame consists of groups of 3 bases that each code for one amino acid.
- A frameshift mutation shifts the grouping of these bases and changes the code for amino acids.
- The resulting protein is usually nonfunctional.
- Insertions, deletions, and duplications can all be frameshift mutations.

Deletion

- A deletion changes the number of DNA bases by removing a piece of DNA.
- Small deletions may remove one or a few base pairs within a gene, while larger deletions can remove an entire gene or several neighboring genes.
- The deleted DNA may alter the function of the resulting protein(s).

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.


Chromosome Mutation
Deletion
Duplication
Inversion
Translocation

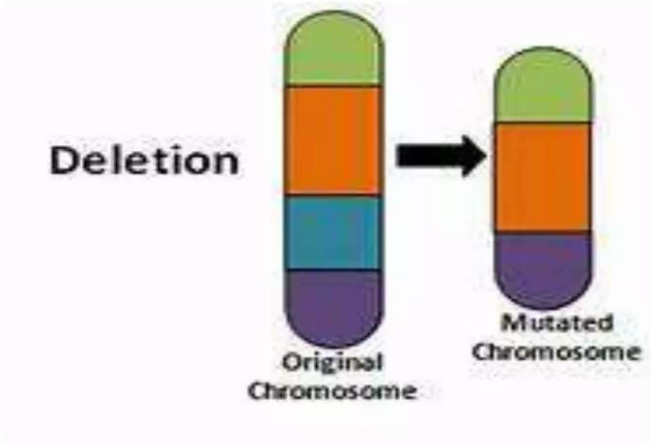
Chromosome mutation

- A chromosome mutation is an unpredictable change that occurs in a chromosome.

- These changes are most often brought on by problems that occur during meiosis (division process of gametes) or by mutagens (chemicals, radiation, etc.).

Deletion

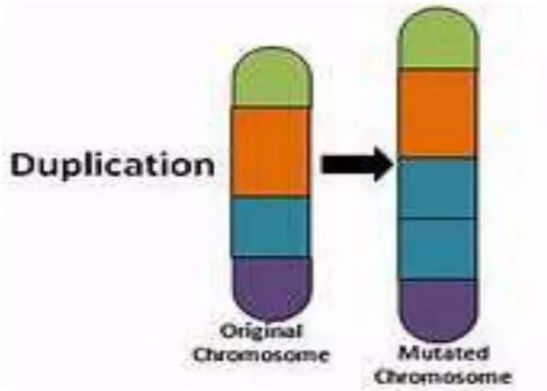
- A small-scale type of deletion mutation is one in which one or more nucleotides are lost or deleted from the chromosome



Duplication

- A type of mutation in which a portion of a genetic material or a chromosome is duplicated or replicated, resulting in multiple copies of that region.

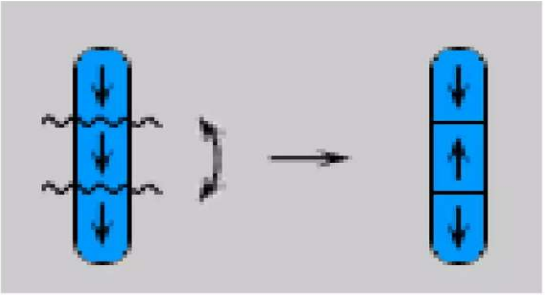
- Duplication results from an unequal crossing-over between misaligned homologous chromosomes during meiosis



Inversion Mutation

- If two breaks occur in one chromosome, sometimes the region between the breaks rotates 180 degrees before rejoining with the two end fragments.

- Such an event creates a chromosomal mutation called an inversion.



Translocation mutation

- Translocation mutations take place when a portion of a chromosome is relocated.

- The genes from one chromosome can move to another position on the same chromosome; alternatively, they can become incorporated into a different chromosome.