# MUTATION

**Mutation** is defined as **a change in genotype** that influence or character of an organism or altered releated phenotype. Mutation is the gene or chromosome that results from a mutational process **Mutagens are chemical or physical agents that interact with DNA to cause mutation**s. Factor or agents causing mutation are known as mutagens .An individual showing an altered phenotype due to mutation are known as variant .mutant is the organism or cell whose changed phenotype is attributed to a mutation

## CHARACTERISTICS OF MUTATION

1-Generally mutant alleles are recessive to their wild type or normal alleles

2- Most mutations have harmful effect, but some mutations are beneficial

3-Spontaneous mutations occurs at very low rate

4-Some genes shows high rate of mutation such genes are called as mutable gene

5-Highly mutable sites within a gene are known as hot spots

6-Mutation can occur in any tissue/cell (somatic or germinal) of an organism

## Classification of mutation

**Based on the survival of an individual**

1.**Lethal mutation** – when mutation causes death of all individuals undergoing mutation are known as lethal

2. **Sub lethal mutation** - causes death of 90% individuals

3. **Sub vital mutation**– such mutation kills less than 90% individuals

4.**Vital mutation** -when mutation don't affect the survival of an individual are known as vital

5.**Supervital mutation –** This kind of mutation enhances the survival of individual

**BASED ON CAUSES OF MUTATION**

**1. Spontaneous mutation**- Spontaneous mutation occurs naturally without any cause. The rate of spontaneous mutation is very slow eg- Methylation followed by deamination of cytosine. Rate of spontaneous mutation is higher in eukaryotes than prokaryotes. Eg. UV light of sunlight causing mutation in bacteria They are also called "background mutation

**2. Induced Mutation-** Mutations produced due to treatment with either a chemical or physical agent are called induced mutation . The agents capable of inducing such mutations are known as mutagen. use of induced mutation for crop improvement program is known as mutation breeding. Eg. X- rays causing mutation in cereals Besides naturally occurring spontaneous mutations, the mutations can be induced artificially in the living organisms by exposing them to abnormal environment such as radiation, certain physical conditions (i.e., temperature) and chemicals

**BASED ON TISSUE OF ORIGIN**

**1. Somatic mutation-** A mutation occurring in somatic cell is called somatic mutation. In asexually reproducing species somatic mutations transmits from one progeny to the next progeny Mutations that are in the somatic tissues of the body.
Mutations are not transmitted to progeny.The extent of the phenotypic effect depends upon whether the mutation is dominant or recessive (dominant mutations generally have a greater effect. The extent of the phenotypic effect depends upon whether it occurs early or late in development (early arising mutations have a greater effect

**2. Germinal Mutation-** When mutation occur in gametic cells or reproductive cells are known as germinal mutation. In sexually reproductive species only germinal mutation are transmitted to the next generation Mutations that are in the germ tissues of the body. Mutations may be transmitted to progeny .Dominant mutations are seen in first generation after the mutation occurs.If a female gamete containing an X-linked mutation is fertilized, the males will show the mutant phenotype Recessive mutations will only be seen upon the chance mating with an individual carrying the recessive allele too; thus, the recessive mutation may remain hidden for many generations

**BASED ON DIRECTION OF MUTATION**

**1. Forward mutation-** When mutation occurs from the normal/wild type allele to mutant allele are known as forward mutation

**2. Reverse mutation-** When mutation occurs in reverse direction that is from mutant allele to the normal/wild type allele are known as reverse mutation

**Type of trait affected**

**1. Visible mutation-** Those mutation which affects on phenotypic character and can be detected by normal observation are known as visible mutation

**2. Biochemical mutation-** mutation which affect the production of biochemicals and which does not not show any phenotypic character are known as biochemical mutation

Silent - changes a codon but codes for the same amino acid

Missense - substitutions that change a codon for one amino acid into a codon for a different amino acid

Nonsense -substitutions that change a codon for one amino acid into a stop codon.

According to size following two types of mutations have been recognized:

**1. Point mutation**

When heritable alterations occur in a very small segment of DNA molecule, i.e., a single nucleotide or nucleotide pair, then this type of mutations are called “point mutations”. The point mutations may occur due to following types of subnucleotide change in the DNA and RNA.

**Deletion mutations.** The point mutation which is caused due to loss or deletion of some portion (single nucleotide pair) in a triplet codon of a cistron or gene is called deletion mutation.

**Insertion or addition mutation.** The point mutations which occur due to addition of one or more extra nucleotides to a gene or cistron are called insertion mutations.

**2.Frameshift mutations.**

The mutations which arise from the insertion or deletion of individual nucleotides and cause the rest of the message downstream of the mutation to be read out of phase, are called **frameshift mutations.**

3. **Substitution mutation.** A point mutation in which a nucleotide of a triplet is replaced by another nucleotide, is called substitution mutation.

**4. Multiple mutations**

When changes involving more than one nucleotide pair, or entire gene, then such mutations are called gross mutations. The gross mutations occur due to rearrangements of genes within the genome. It may be:

1.The rearrangement of genes may occur within a gene. Two mutations within the same functional gene can produce different effects depending on gene whether they occur in the cis or trans position.

2.The rearrangement of gene may occur in number of genes per chromosome. If the numbers of gene replicas are non-equivalent on the homologous chromosomes, they may cause different types of phenotypic effects over the organisms.

3.Due to movement of a gene locus new type of phenotypes may be created, especially when the gene is relocated near heterochromatin. The movement of gene loci may take place due to following method:

**(i) Translocation.** Movement of a gene may take place to a non-homologous chromosome and this is known as translocation.

**(ii) Inversion.** The movement of a gene within the same chromosome is called inversion.

**According to their phenotypic effects following kinds of mutations may occur:**

**1.Dominant mutations**

The mutations which have dominant phenotypic expression are called dominant mutations. For example, in man the mutation disease aniridia (absence of iris of eyes) occurs due to a dominant mutant gene.

**2.Recessive mutations**

Most types of mutations are recessive in nature and so they are not expressed phenotypically immediately. The phenotypic effects of mutations of a recessive gene is seen only after one or more generations, when the mutant gene is able to recombine with another similar recessive gene.

**3.Isoalleles**

Some mutations alter the phenotype of an organism so slightly that they can be detected only by special techniques. Mutant genes that give slightly modified phenotypes are called isoalleles. They produce identical phenotypes in homozygous or heterozygous combinations.

**General Types**

**Gene mutation** :This involves the change in nucleotide sequence of gene.

**Mechanisms for gene mutation**

Errors in DNA replication .Errors in DNA repair Environmental mutagen causes DNA damage that is not repaired correctly. Transposons and insertion sequences (a mobile DNA

elements that can move from one location in the chromosome to another; the element may "jump" into a gene thereby mutating it

**2. Chromosome mutation** is the segments of chromosomes, whole chromosomes, or entire sets of chromosomes change

According to the types of chromosomes, the mutations may be of following two kinds:

**1.Autosomal mutations.** This type of mutation occurs in autosomal chromosomes.

**2.Sex chromosomal mutations.** This type of mutation occurs in sex chromosomes.

**Chromosome mutation are 5 types**

Chromosome mutations are inherited once they occur and are of the following types

a) **Deletion** which involves loss of a broken part of a chromosome.
b) **Inversion** in which broken segment reattached to original chromosome in reverse order
c) **Duplication** which involves when gene sequence of chromosome. repeated
d) **Translocation** in which the broken segment becomes attached to a non- homologous chromosome resulting in new linkage relations.
e) **Nondisjunction:** Failure of chromosome to separate during meiosis which cause gamete to have many to few chromosome.

**Changes in number of chromosomes:**

**1.Euploidy**It involves the loss, or gain, of whole chromosome set.

**Polyploidy**:Addition of one or more sets of chromosomes.

They may be further:

**(a) Autopolyploidy**. The autopolyploidy involves polyploidy, in which the same basic set of chromosomes are multiplied.

**(b) Allopolyploidy.** The polyploidy results due the doubling of chromosome number in a F1 hybrid which is derived from two distinctly different species. The resultant species is called an allopolyploid.

**2.Aneuploidy**

It involves the loss, or gain, of a part of the chromosome set.It refers to a condition in which one or a few chromosomes are added or deleted from the normal chromosome number. Hence, the number of chromosomes in aneuploidy can be greater or smaller than the number of chromosomes in the wild type.

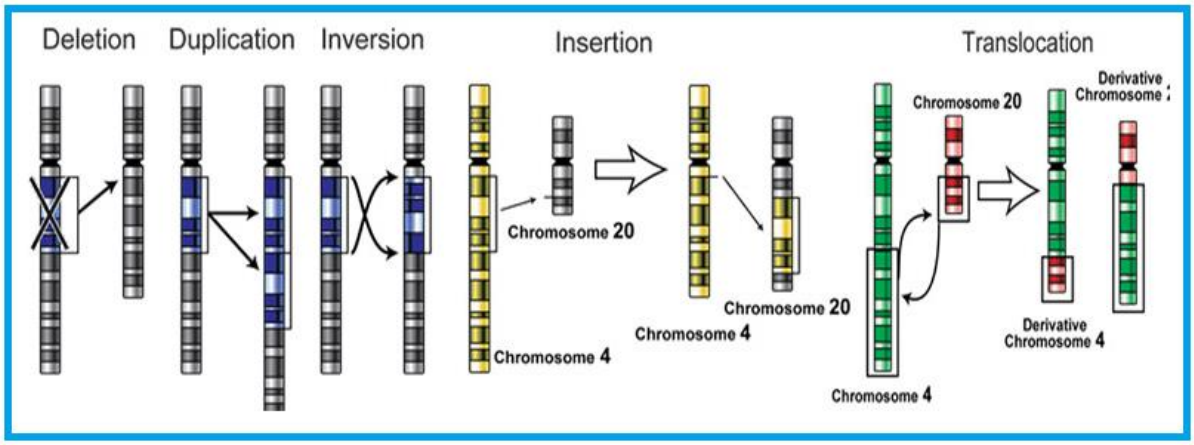
Various types of aneuploidy can be identified as: nullisomy, monosomy, and trisomy.

**a)Nullisomy (2n-2)** is the loss of both chromosomes of the homologous pair. This conditions may be lethal in most organisms.

**b)Monosomy (2n-1)** is the loss of a single chromosome of the homologous pair.

**c)Trisomy** is the gain of an extra chromosome (2n+1).

**Fig 1. Chromosomal Mutation**

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