

## 11.MUTATIONS

Mutation in a broad sense include all those heritable changes which alter the phenotype of an individual. Thus, mutation can be defined as a sudden heritable change in the character of an organism which is not due to either segregation or recombination.

- ❖ The term mutation was first used by Hugo de Vries to describe the sudden phenotypic changes which were heritable, while working with *Oenothera lamarckiana*.
- ❖ But the earliest record of mutations dates to 1791 when Seth Wright noticed a male lamb with unusually short legs in his flock of sheep. This lamb served as a source of short leg trait for the development of Ancon breed of sheep.
- ❖ However, the systematic studies on mutations were started in 1910 by T.H. Morgan who used *Drosophila melanogaster* for his studies.
- ❖ In 1927, H.J. Muller demonstrated for the first time the artificial induction of mutations by using x-rays in *Drosophila*.
- ❖ Similarly, in 1928, L.J. Stadler demonstrated an increase in the rate of mutations due to x-rays in barley and maize.
- ❖ Induction of mutations by chemicals in fungus *Aspergillus* was demonstrated by R.A. Steinberg in 1939.
- ❖ C. Auerbach and J.N. Robson in 1946 used chemicals to induce mutations in *Drosophila*.
- ❖ The first plant breeding programme using mutations (mutation breeding) was initiated in 1929 in Sweden, Germany and Russia.
- ❖ In India, it was initiated in early 1930s.

### Terminology

**Muton:** The smallest unit of gene capable of undergoing mutation and it is represented by a nucleotide.

**Mutator gene:** A gene which causes another gene or genes to undergo spontaneous mutation.

**Mutable genes:** Genes which show very high rates of mutation as compared to other genes.

**Mutant:** An organism or cell showing a mutant phenotype due to mutant allele of a gene. **Mutagen:** A physical or chemical agent which induces mutation.

**Hot spots:** Highly mutable sites within a gene.

**Gene mutations or point mutations:** The changes which alter the chemical structure of a gene at molecular level.

**Characteristic features of mutations:**

- 1) Mutations are mostly recessive and very rarely dominant e.g. epiloia in human, *Notch* wing in *Drosophila* etc.
- 2) Most mutations have harmful effects and very few (less than 0.1 %) are beneficial.
- 3) Mutations are random event in term of time of their occurrence and the gene in which they occur.
- 4) Mutations may be due to a change in a gene, a group of genes or in entire chromosome.
- 5) If gene mutations are not lethal, the mutant individuals may survive. However, chromosomal mutations are generally lethal and such mutants do not survive.
- 6) If mutation occur at both loci simultaneously, the mutants can be identified in  $M_1$  generation. However, if it is restricted to one locus only, (dominant to recessive) the effect can be seen only in  $M_2$  generation.
- 7) Macro-mutations are visible and can be easily identified, while micro-mutations cannot be seen with naked eye and need special statistical tests (or statistical analysis).
- 8) Many of the mutants show sterility.
- 9) Most mutants are of negative selection value.
- 10) Mutation for altogether new character generally does not occur.
- 11) Mutations are random i.e. they can occur in any tissue or cell of an organism. However, some genes show higher mutation rate than others.
- 12) Mutations can be sectorial. The branches arising from mutated sector show mutant characters.
- 13) Mutations are recurrent i.e. the same mutation may occur again and again.
- 14) Induced mutations commonly show pleiotropy often due mutation in closely linked genes.

- 15) Fine structure analysis of genes has shown that mutations producing the same phenotypic effect may be located at different sites within the same gene.
- 16) Mutations occur in both forward (from wild type allele to mutant allele) and reverse (from mutant allele to wild type allele) direction. Generally, the rate of forward mutations are much higher than those for reverse mutations.

**Classification of mutations:** Mutations can be classified in several ways.

**1. Based on direction of mutations:**

- a) *Forward mutation:* Any change from wild type allele to mutant allele
- b) *Backward mutation or reverse mutation:* A change from mutant allele to wild type

**2. Based on source / cause of mutations:**

- a) *Spontaneous mutation:* Mutation that occur naturally
- b) *Induced mutation:* Mutation that originates in response to mutagenic treatment

**3. Based on tissue of origin:**

- a) *Somatic mutation:* A mutation in somatic tissue
- b) *Germinal mutation:* A mutation in germline cells or in reproductive tissues

**4. Based on effect on survival:**

- a) *Lethal mutation:* Mutation which kills the individual that carries it. (survival 0%)
- b) *Sub-lethal mutation:* When mortality is more than 50% of individuals that carry mutation
- c) *Sub-vital mutation:* When mortality is less than 50% of individual that carry mutation.
- d) *Vital mutation:* When all the mutant individuals survive (survival-100%)

**5. Based on trait or character effected:**

- a) *Morphological mutation:* A mutation that alters the morphological features of an individual.

- b) **Biochemical mutation:** A mutation that alters the biochemical function of an individual.

**6. Based on visibility or quantum of morphological effect produced:**

- a) **Macro-mutations:** Produce a distinct morphological change in phenotype (which can be detected easily without any confusion due to environmental effects). Generally found in qualitative characters. Eg : colour of flowers, height of plant etc.
- b) **Micro-mutations:** Mutations with invisible phenotypic changes, (which can be easily confused with effects produced due to environment). Generally observed in quantitative characters.

**7. Based on the site of mutation or on cytological basis:**

- a) **Chromosomal mutations:** Mutations associated with detectable changes in either chromosome number or structure.
- b) **Gene or point mutations:** Mutations produced by alterations in base sequences of concerned genes.
- c) **Cytoplasmic mutations:** Mutations associated with the changes in chloroplast DNA (cpDNA) and mitochondrial DNA (mtDNA).

**8. Molecular basis:** point or gene mutations can be subdivided as

- a) **Base substitution**
- b) **Deletion**
- c) **Addition**
- d) **Transposition**
- e) **Trinucleotide expansion**

**9. Effect on the expression of neighbouring genes:**

- a) **Polar mutations:** some gene mutations suppress the expression of other genes located next to them on one side (downstream side).
- b) **Nonpolar mutations:** affect the expression of only those genes within they are located.

## 10. Intensity of characters expression:

- a) **Amorphic mutations:** almost total loss of the expression of the affected phenotype as they produce totally non-functional gene products.
- b) **Hypomorphic mutations:** when a mutation leads to partial loss in the expression of a trait.
- c) **Hypermorphic mutation:** the mutant alleles lead to an increased expression than that of wild type allele, r<sup>II</sup> mutants of phage T<sub>4</sub> of *E. coli* (in terms of plaque size).

**I. Spontaneous mutations:** Spontaneous mutations occur naturally without any apparent cause. There are two possible sources of origin of these mutations.

1. Due to error during DNA replication.
2. Due to mutagenic effect of natural environment Eg. UV rays from sunlight

The rate of spontaneous mutations is very low. 1 in 10 lakhs i.e.  $10^{-6}$ . But different genes may show considerably different mutation rates. In crop plants, some varieties were developed through spontaneous mutations are given below in tabular form.

Sr. no.	Crop	Variety
1	Rice	GEB-24, Dee-Geo-Woo-Gen
2	Wheat	Norin
3	Groundnut	TMV-10
4	Sorghum	Co-4 (coimbatore 4)

**II. Induced mutations:** Mutations can be induced artificially through treatment with either physical or chemical mutagens. The exploitation of induced mutations for crop improvement is called **mutation breeding**. The rate of induced mutations is very high. The induced mutations did not differ from spontaneous mutations in expression. Examples of popular induced mutants in crop plants are listed below.

<b>Sr. no.</b>	<b>Crop</b>	<b>Mutant variety</b>	<b>Parent</b>	<b>Mutagen</b>
1	Rice	Jagannath, Mahsuri mutant	T-141 Mahsuri	X-rays $\gamma$ -rays
2	Wheat	Sharbati sonara NP-836	Sonara 64 NP-799	UV rays x rays
3	French Beans	Pusa Parvati Pusa Lal Meeruti	Wax podded Meeruti	x-rays x-rays
4	Tomato	S-12	Sioux	$\gamma$ -rays
5	Castor	Aruna	HC-6	Thermal neutrons
6	. Cotton	MCU 7 MCU 10	1143 EE MCU 4	x-rays $\gamma$ -rays