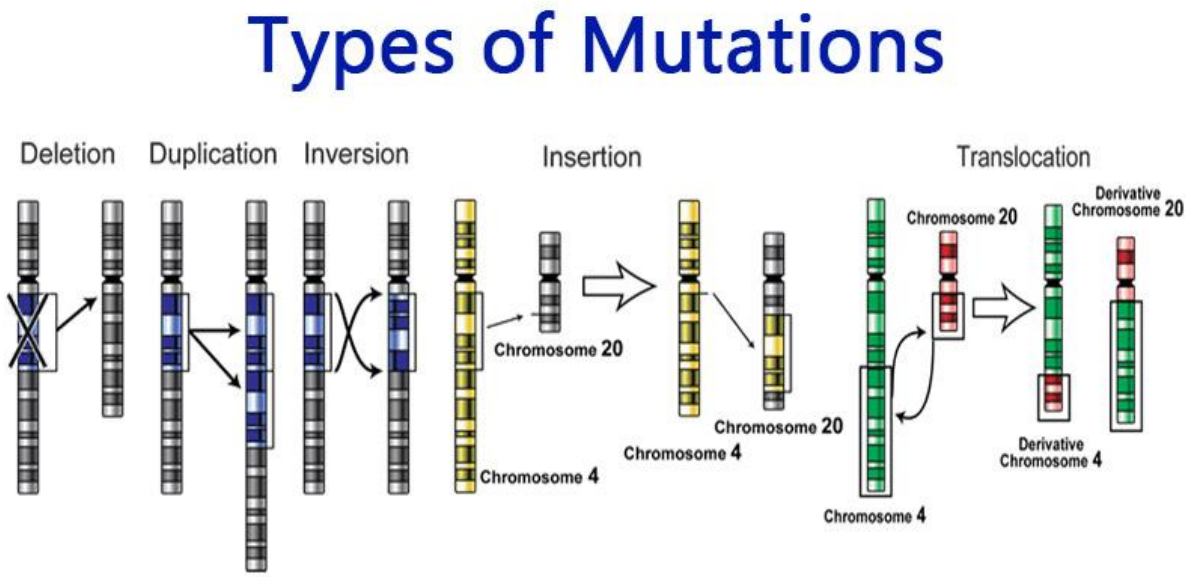
Mutation and its types

Mutation is a process that produces a gene or chromosome that differs from the wild type. The mutation may result due to changes either on the gene or the chromosome itself.

Thus, broadly mutation maybe:

1. **Gene mutation** where the allele of a gene changes.
2. **Chromosome mutation** where segments of chromosomes, whole chromosomes, or entire sets of chromosomes change.



Types of Mutations

There are various schemes for classification of different kind of mutations. Depending on:

A. The Type of Cell Involved

1. **Somatic mutations**

- 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 mutations**

- 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

B. Mode of Origin

(1) Spontaneous mutations

The spontaneous mutations occur suddenly in the nature and their origin is unknown. They are also called "background mutation" and have been reported in many organisms such as, Oenothera, maize, bread molds, microorganisms (bacteria and viruses), Drosophila, mice, man, etc.

(2) Induced mutations

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.

C. Direction of Mutation

According to their mode of direction following types of mutations have been recognised:

1. Forward mutations

In an organism when mutations create a change from wild type to abnormal phenotype, then that type of mutations are known as forward mutations. Most mutations are forward type.

2. Reverse or back mutations

The forward mutations are often corrected by error correcting mechanism, so that an abnormal phenotype changes into wild type phenotype.

D. Size and Quality

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

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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.

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.**

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

2. Multiple mutations or gross 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.

E. Phenotypic Effects

1. **Morphological mutations** are mutations that affect the outwardly visible properties of an organism (i.e. curly ears in cats)

2. **Lethal mutations** are mutations that affect the viability of the organism (i.e. Manx cat).
3. **Conditional mutations** are mutations in which the mutant allele causes the mutant phenotype only in certain environments (called the restrictive condition).

In the permissive condition, the phenotype is no longer mutant.

Example. Siamese cat – mutant allele causes albino phenotype at the restrictive temperature of most of the cat body but not at the permissive temperature in the extremities where the body temperatures is lower.

4. **Biochemical mutations** are mutations that may not be visible or affect a specific morphological characteristic but may have a general affect on the ability to grow or proliferate.

For example, the bacterium Escherichia coli does not require the amino acid tryptophan for growth because they can synthesize tryptophan. However, there are E. coli mutants that have mutations in the trp genes. These mutants are auxotrophic for tryptophan, and tryptophan must be added to the nutrient medium for growth.

F. Magnitude of Phenotypic Effect

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.

G. Loss of Function or Gain of Function

1. **Loss of function mutation**

Loss of function mutation is also called inactivating mutations, result in the gene product having less or no function (being partially or wholly inactivated).

2. **Gain of function mutations**

The gain of function mutations also called activating mutations, change the gene product such that its effect gets stronger (enhanced activation) or even is superseded by a different and abnormal function.

H. Type of Chromosome Involved

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.

I. Chromosomal Mutation and Types

- The changes in the genome involving chromosome parts, whole chromosomes, or whole chromosome sets are called chromosome aberrations or chromosome mutations.
- Chromosome mutations have proved to be of great significance in applied biology—agriculture (including horticulture), animal husbandry and medicine.

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

a. Structural changes in chromosomes:

1. **Changes in number of genes**

(a) Loss: **Deletion** which involves loss of a broken part of a chromosome.

(b) Addition: **Duplication** which involves addition of a part of chromosome.

2. **Changes in gene arrangement:**

(a) Rotation of a group of genes 180^0 within one chromosome: **Inversion** in which broken segment reattached to original chromosome in reverse order.

(b) Exchange of parts between chromosomes of different pairs: **Translocation** in which the broken segment becomes attached to a non- homologous chromosome resulting in new linkage relations.

b. Changes in number of chromosomes:

1. **Euploidy**

- It involves the loss, or gain, of whole chromosome set.
- The term euploidy (Gr., eu = even or true; ploid = unit) designates genomes containing chromosomes that are multiples of some basic number (x).
- The euploids are those organisms which contain balanced set or sets of chromosomes in any number.
- The number of chromosomes in a basic set is called the monoploid number, x.
- Those euploid types whose number of sets is greater than two are called polyploid.
- Thus, 1x is monoploid, 2x is diploid; and the polyploid types are 3x (triploid), 4x (tetraploid), 5x (pentaploid), 6x (hexaploid) and so on.
- Mutation due to Euploidy refers to the state of having a chromosome number that is an exact multiple of a basic chromosome set. This means the number of chromosome sets is increased in euploidy.

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.

1. **Nullisomy (2n-2)** is the loss of both chromosomes of the homologous pair. This conditions may be lethal in most organisms.
2. **Monosomy (2n-1)** is the loss of a single chromosome of the homologous pair.
3. **Trisomy** is the gain of an extra chromosome (2n+1). Klinefelter syndrome (44+XXY/XYY) and Down syndrome are examples of trisomy.

Missense mutation A base change that converts one codon into another. Many missense mutations are silent because the encoded amino acid remains the same or the amino acid substitution is sufficiently subtle so as not to compromise activity of the enzyme. Missense mutations that have a marked effect often lie in the active site or grossly disrupt protein folding.

Nonsense mutation A base change that converts a codon within the coding sequence into a stop codon. Note that there is only a limited set of sense codons that can be converted to a stop codon by a single base change. Nonsense mutations lead to a truncated protein product. Nonsense mutations that lie early in the gene sequence will completely inactivate the gene. Sometimes nonsense mutations that lie late in the gene sequence will not disrupt gene function.

Frameshift mutation The addition or deletion of a base or bases such that the coding sequence is shifted out of register. Note that addition or deletion of a multiple of three bases does not cause a frameshift. After the frameshift mutation is encountered, missense codons will be read up to the first stop codon. Like nonsense mutations, frameshift mutations usually lead to complete inactivation of the gene.

SUGGESTED READING

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