Mutation

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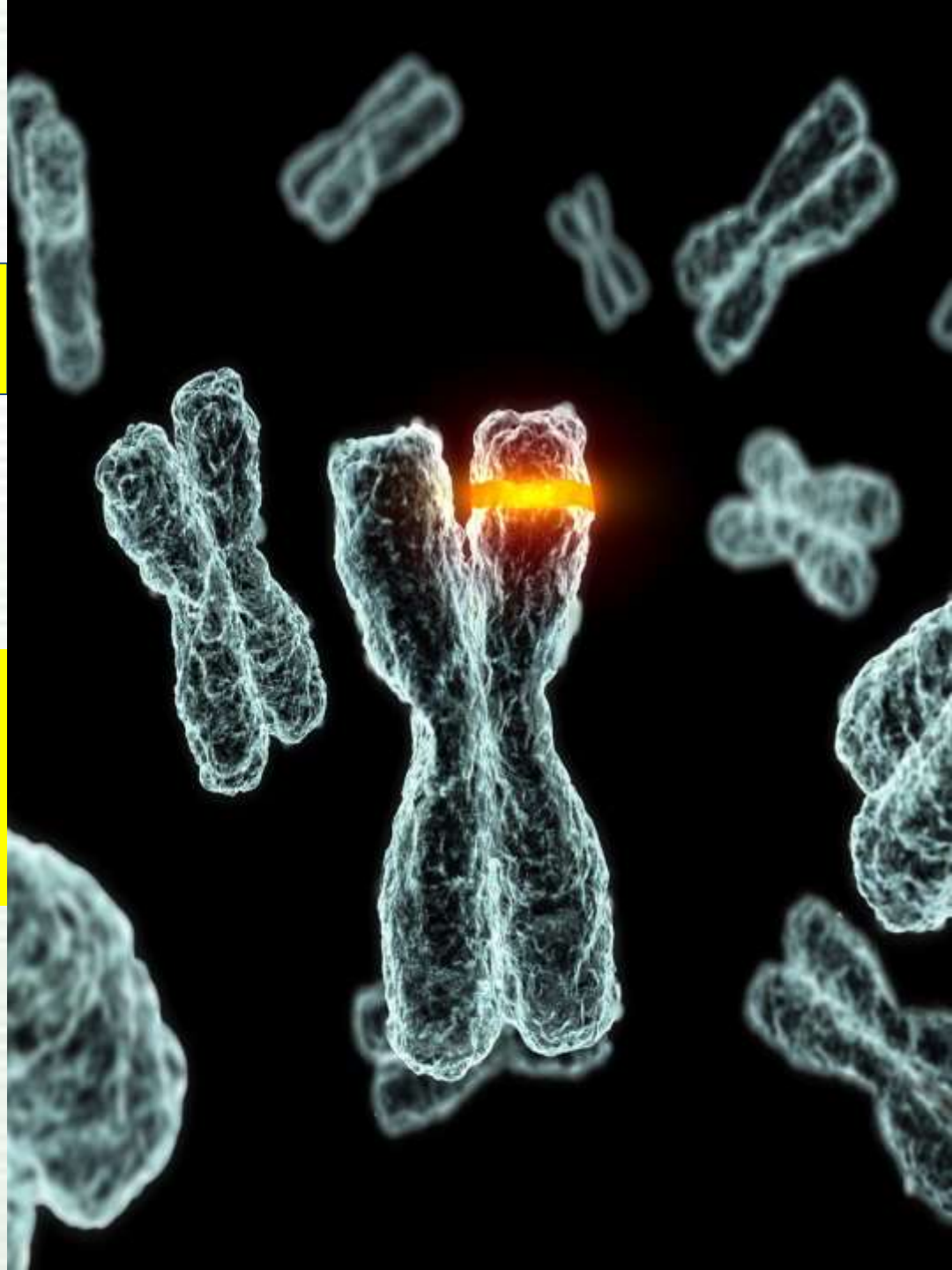


Introduction

A mutation is a change or alteration happens in a DNA, gene or chromosome due to intrinsic or extrinsic factors such as an error in replication or exposure to UV light, respectively.

✓*Change in the nucleotide sequence of the DNA",* is a mutation.

✓The mutation is an important biological process in nature. It can be helpful or harmful. For instance, the mutation creates variations in nature by providing new alleles in nature and hence helps in evolution.

✓The word mutation was similar to the French word "mutacioun" which literally means "process of changing."

✓Although the "mutation" world was originally derived from the Latin word "mutare". The meaning of this is "to change."

History

✓The term “mutation” was coined by ***Hugo De Vries*** in 1890. However, before him, Seth Wright, an English farmer noticed mutation first time in his unusual short-lege male lambs during 1791. He fails to define the process.

✓After the findings of Hugo de Vries, the mechanism of mutation was studied by Morgan in 1910.

✓In 1927, ***H. J. Muller*** performed experiments of artificial mutagenesis. Using the X-rays he had introduced mutation in Drosophila. For that, he was awarded Nobel prize in 1946.

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 with in 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.
2. Most mutations have harmful effects and very few (less than 0.1 %) are beneficial.
3. They may be due to a change in a gene, a group of genes or in entire chromosome.
4. If gene mutations are not lethal, the mutant individuals may survive.
5. If mutation occur at both loci simultaneously, the mutants can be identified in M1 generation. However, if it is restricted to one locus only, (dominant to recessive) the effect can be seen only in M2 generation.
6. Macro-mutations are visible and can be easily identified, while micro - mutations can not be seen with naked eye and need special statistical tests.
7. Many of the mutants show sterility.
8. Most mutants are of negative selection value.
9. Mutation for altogether new character generally does not occur.
10. Mutations are random i.e. they can occur in any tissue or cell of an organism.
11. Mutations can be sectorial. mutated sector show mutant characters.
12. Mutations are recurrent i.e. the same mutation may occur again and again.
13. Induced mutations commonly show pleiotropy often due mutation in closely linked genes.

Classification of mutations:

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%)

Classification of mutations cont.

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 with out any confusion due to environmental effects) 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).

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

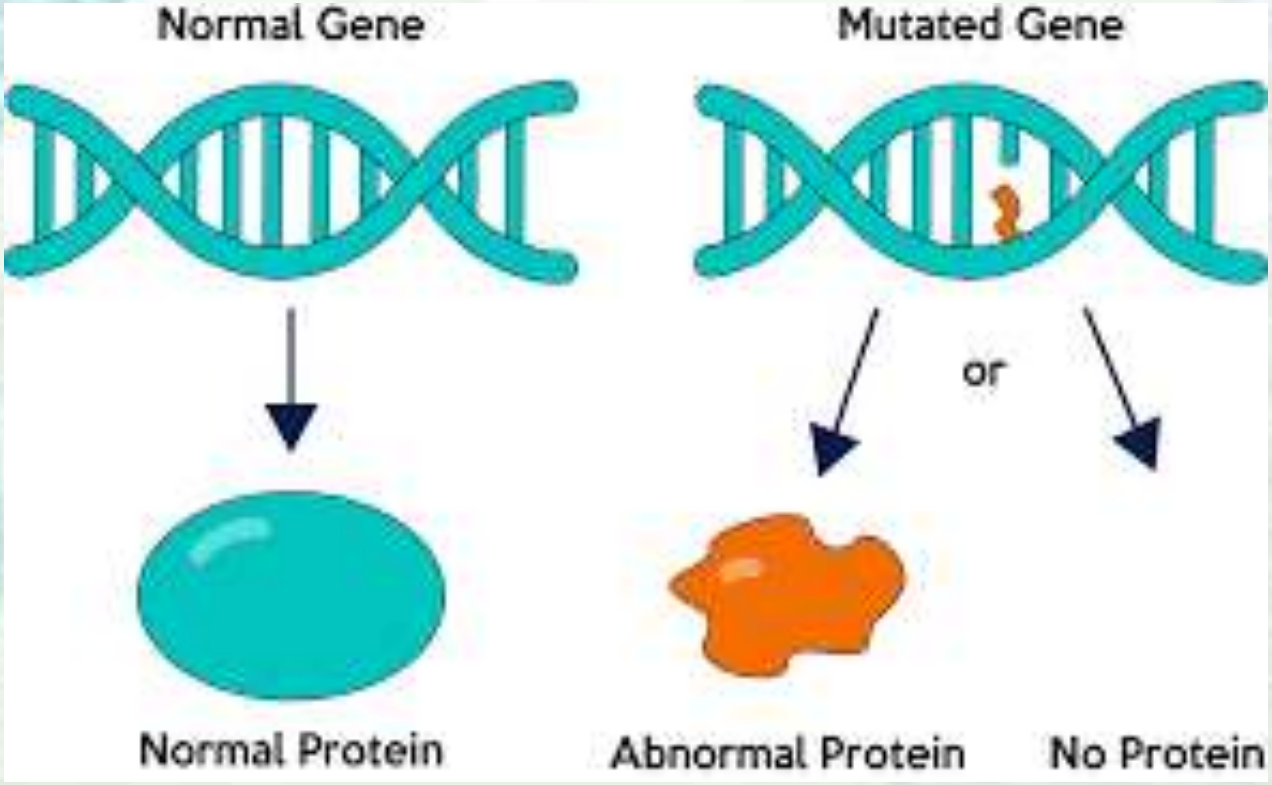
a) **Chromosomal mutations:** Mutations associated with detectable changes in either chromosome number or structure.

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).

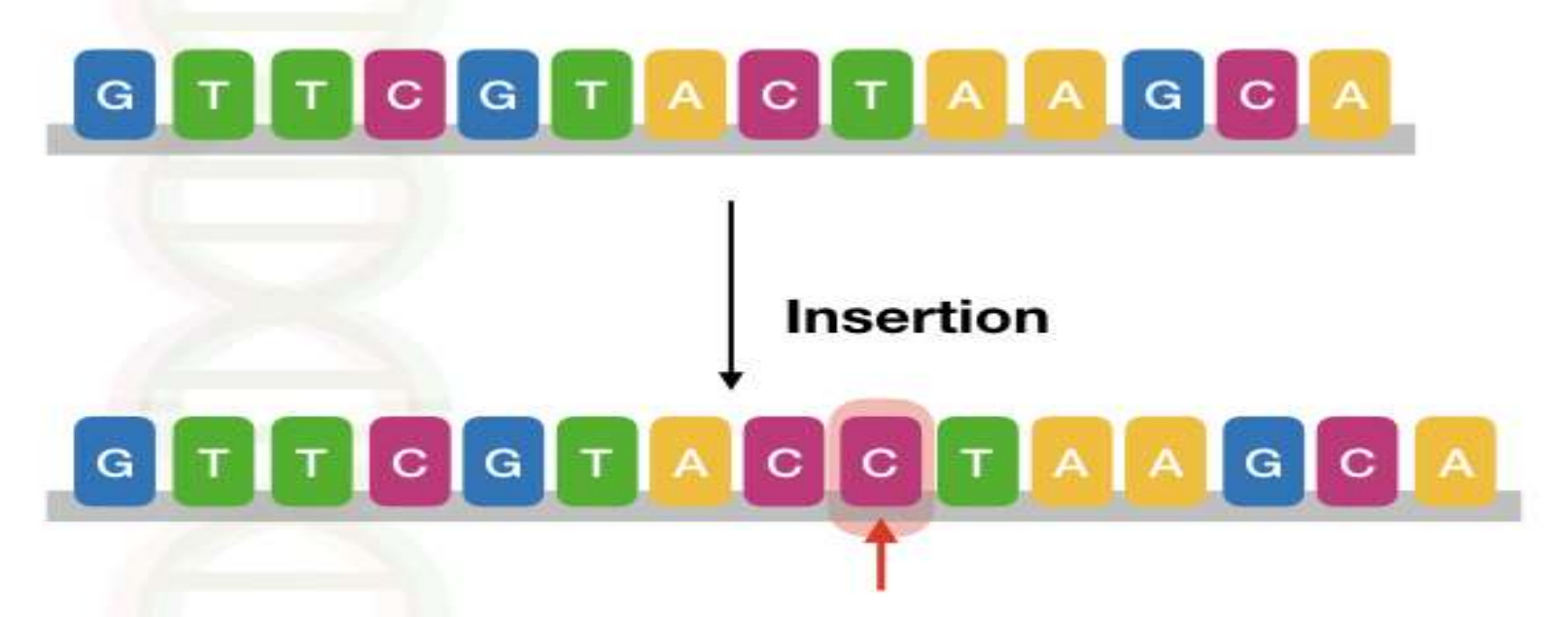
Gene Mutation

Mutation or series of mutations occur in the sequence of a gene that changes the function of a gene is referred to as gene mutations.

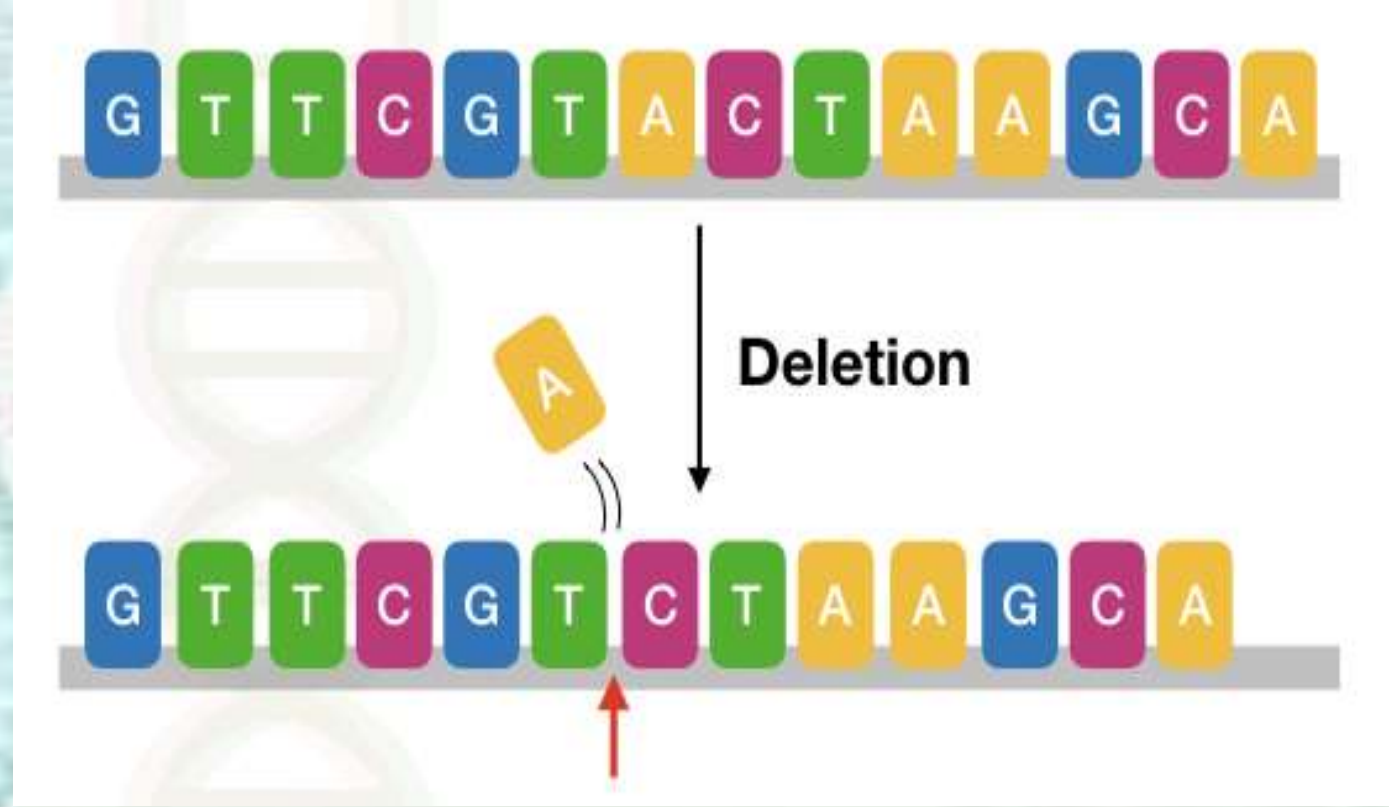


***Point mutation*–** Change in the single base of the DNA. It's often known as single nucleotide polymorphism. Read our amazing article on Point mutation.

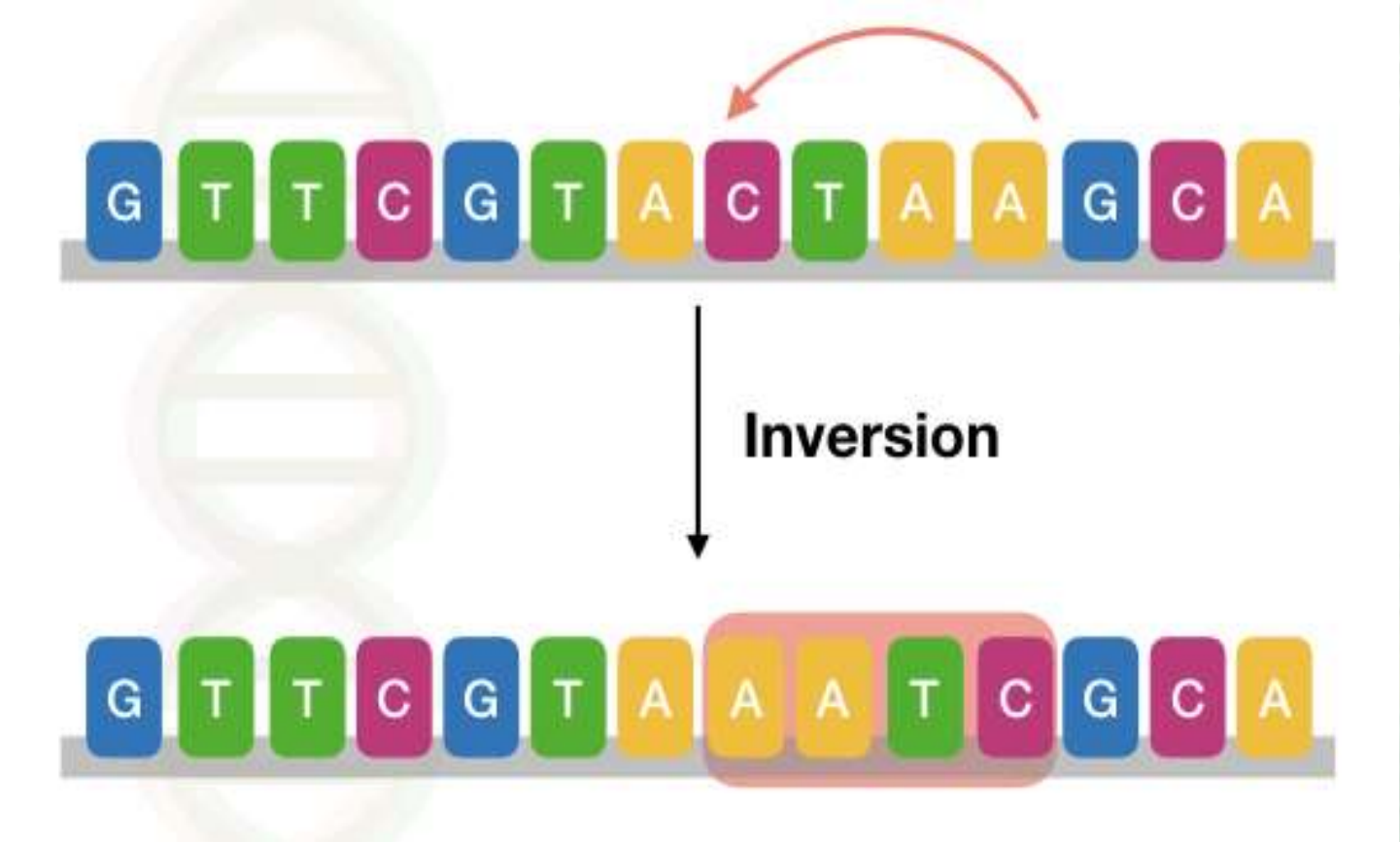
***Insertion*–** insertion or addition of a base into the gene sequence. Often known as addition mutation.



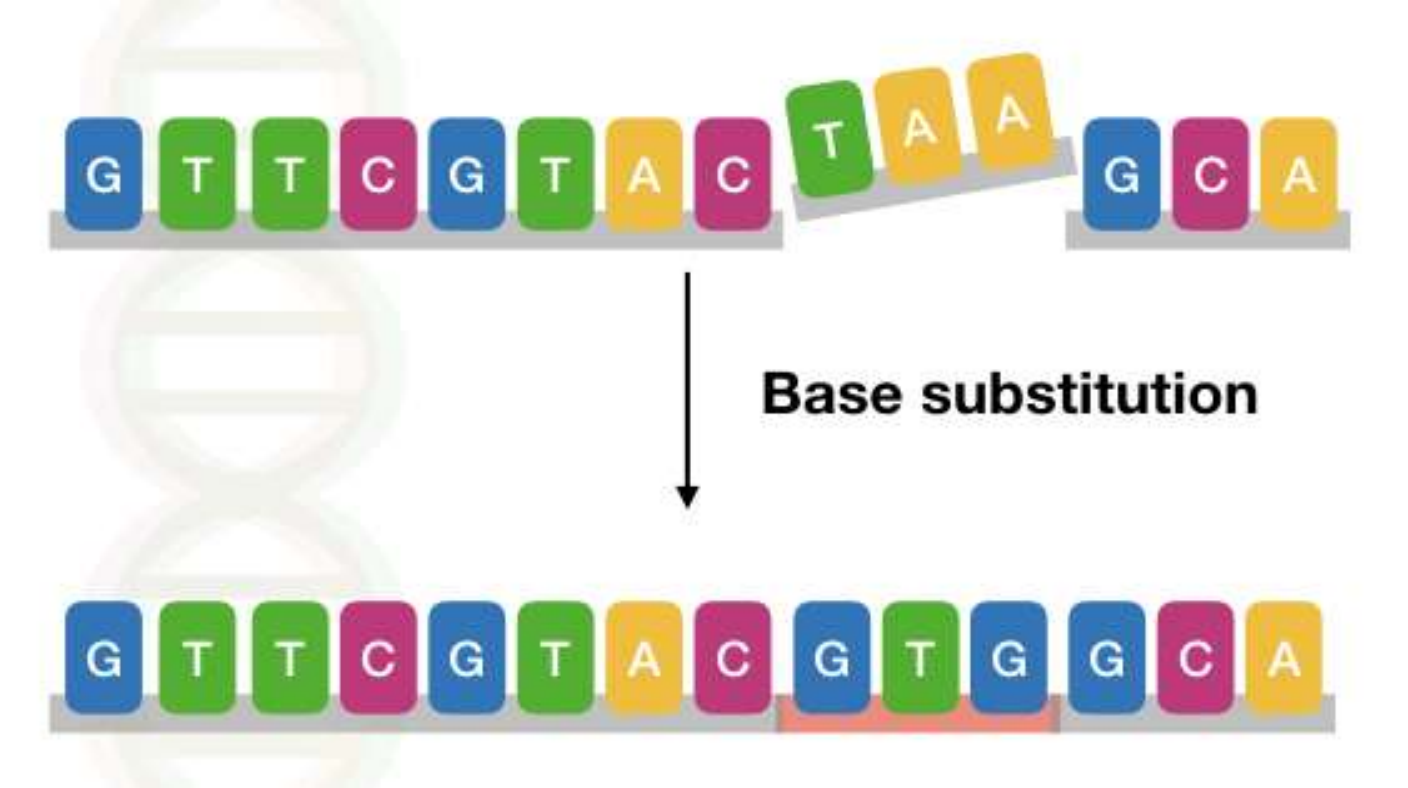
Deletion – When a base or some bases deleted from the gene sequence.



***Inversion*– When some gene sequences inverted and inserted back into the original sequence.**



Substitution– When some bases of a gene sequence are replaced by other bases.



Forward mutation:

Genetic Mutation from wild-type to mutant or evolution of new mutation from wild-type allele is called a forward mutation. The forward mutation leads to the evolution of new traits in the population.

Backward mutation:

✓A mutation is a unidirectional process, but sometimes some mutation gives original (wild-type) allele back to population, such mutation is called the backward or back mutation.

✓Back mutation is a very rare and unusual phenomenon in nature.

✓It gives original phenotype back into the population by true back mutation or by the occurrence of a secondary mutation.

✓In true back mutation, a mutation occurred at the same location as it occurred during forward-mutation. In simple words, it gives the wild-type codon back to the population.

Silent mutation:

It is non-expressive. In silent mutation, a new codon codes for the same amino acid as the wild-type one.

Mis-sense mutation:

A codon originated from a nucleotide change that will code for different amino acids. It can lead to alteration or loss of function in protein.

Nonsense mutation:

A stop codon is added to the premature protein. It stops protein synthesis because a stop codon ends synthesis of protein and results in a premature protein or truncated protein.

Frameshift mutation:

Base pair alteration causes abnormal reading frame which ultimately results in an abnormal protein formation.

A specific reading frame has a start codon and a stop codon. In between both codons, a definite coding sequence is present.

In a frameshift mutation, alteration in DNA leads to shifting of this reading frame from one place to another in a genome. So the position of start or stop codon changes.

ORIGINAL DNA:

corresponds to the amino acid leucine

FRAMESHIFT MUTATION

every amino acid that follows will be altered

SILENT MUTATION

leucine

MISSENSE MUTATION

valine

NONSENSE MUTATION

stop codon

Spontaneous Mutation

✓Spontaneous mutations happen without any known reasons.

✓It arises due to metabolic errors, replication errors or due development errors.

Spontaneous mutations are rare and occur without any reason. It's originated by birth.

✓Larger genes are more prone to spontaneous mutation because the chance of replication error is higher in larger genes.

✓The rate of spontaneous mutation is 10^{-5} per gene per generation during replication.

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.

CROP	VARIETY
1.Rice	GEB-24, Dee-Geo-Woo-Gen
2.Wheat	Norin
3.Groundnut	TMV-10
4.Sorghum	Co-4 (coimbatore 4)

Induced mutation is resulting from exposure of an organism to mutagenic agents. The general mutagenic agents are radiation, UV light and chemicals.

✓The UV light is responsible for xeroderma pigmentation and skin cancer, it causes uncontrolled cell division by penetrating into the skin.

✓Due to the lower energy of the UV light, it can not penetrates other tissues of us but it can penetrate the skin cells and activates oncogenes.

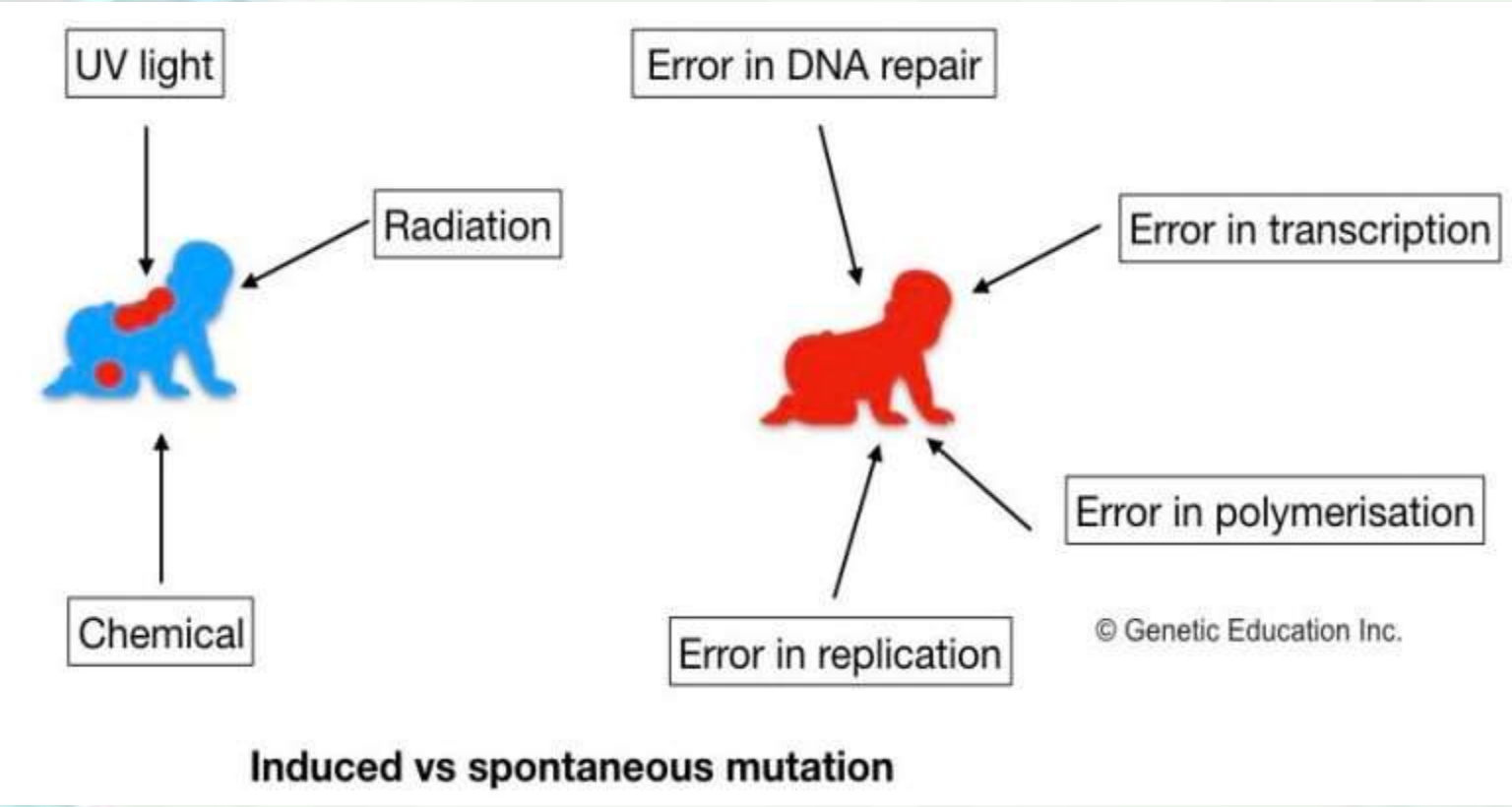
✓Chemicals such as alkylating agents and base analogues are common chemical mutagens. However, every chemical is somehow mutagenic to our DNA.

✓Furthermore, climate change and lifestyle play a major role in acquiring mutations.

Induced mutations:

- **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.

Crop	Mutant Variety	Original variety	Mutagene
1. Rice	Jagannath,	T-141	X-Ray
	Mahsuri mutant	Mahsuri	Y-Ray
2. Wheat	Sharbati sonara	Sonara-64	UV Ray
	NP-836	NP-799	X-Ray
3. Tomato	S-12	Sioux	Y-Ray
4. Castor	Aruna	HC-6	Thermal neutrons
5. Cotton	MCU-7	1143 EE	X-Ray
	MCU-10	MCU-4	Y-Ray

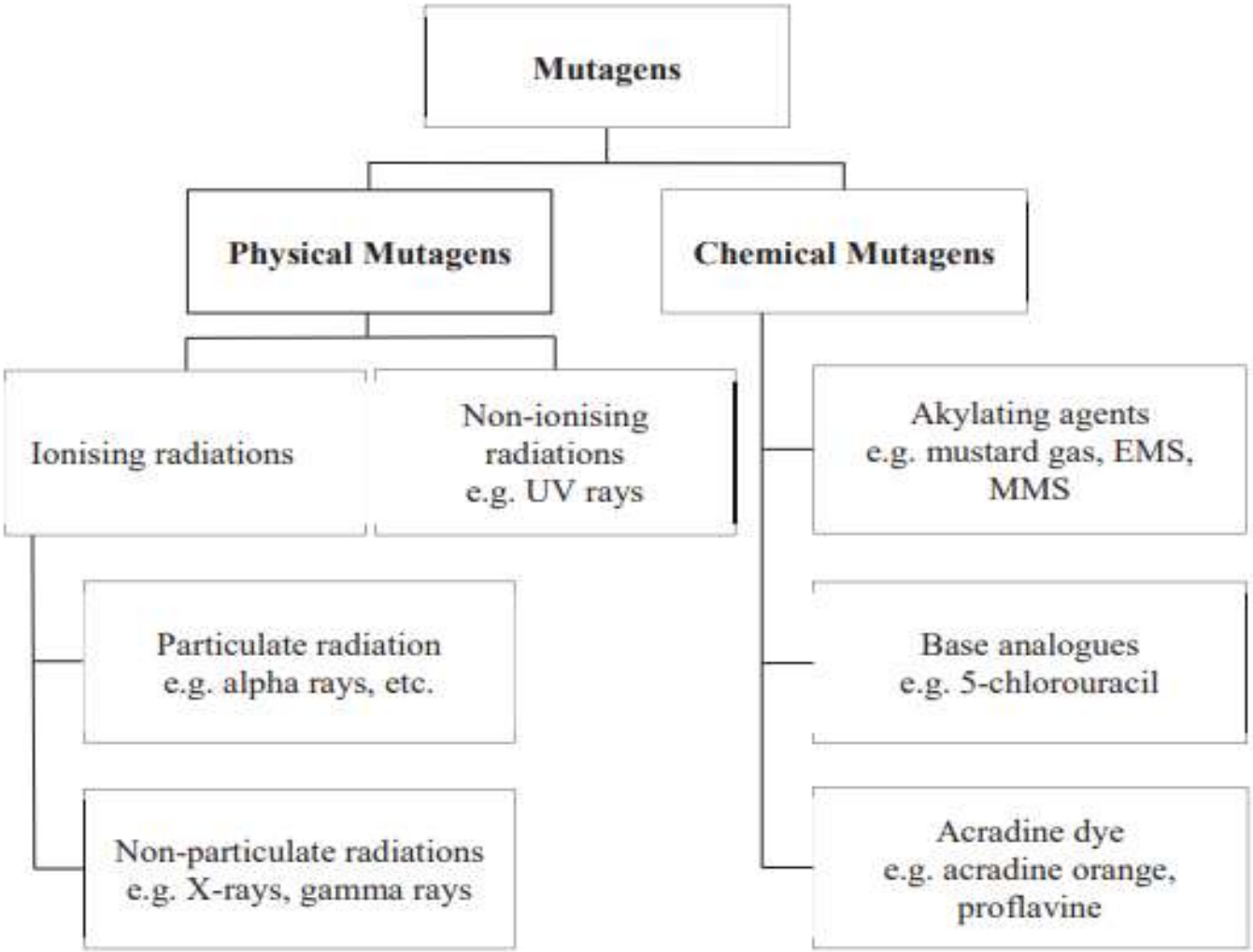


Induced vs spontaneous mutation

✓Spontaneous mutations are stable, inherited and occur infrequently in populations. The rate of inheritance depends on the types of inheritance pattern.

✓Spontaneous mutations are miraculous. It remains recessive in the parental organism and not show off symptoms. Its effect shows in consecutive generations.

✓In contrast, induced mutations immediately show its effect on target organism.



Common mutagens use in plant mutation induction

Detection of sex linked lethal mutations in *Drosophila* by Muller's ClB technique

- Muller developed a system, ClB technique for detecting recessive sexlinked lethal mutations induced by X-ray treatment in *Drosophila*.
- He used a heterozygous (ClB) stock of Drosophila, which has a special X-chromosome, a large part of which is inverted (paracentric inversion).
- This acts as a crossover suppressor in the inverted region and is designated by C.
- A recessive lethal (l) gene and the dominant gene for bar (B) eye are located within the inverted segment.
- As a result, the l and B are always inherited together in the same chromosome.
- The other X-chromosome was normal.
- Consequently all ClB females identified by the bar eye shape, are heterozygous for this chromosome, while the males having the ClB chromosome do not survive [because of the l (lethal) gene].
- The male flies were irradiated with X-rays for the induction of sex-linked recessive lethal mutations. Such males are crossed with ClB females.

- In the F1, half of the females will have the ClB chromosome, which are easily identified by the bar-shaped eyes.
- The remaining half of the females will not have the ClB chromosome and are rejected.
- All the surviving F1 males will have the normal chromosome from the ClB females, while those receiving the ClB chromosome will die due to the lethal gene 'l'.
- Each F1 ClB females is mated to a normal male.
- Each F1 ClB female will have one ClB X-chromosome and one X-chromosome from the mutagen treated male parent.
- So, half of the male progeny receiving ClB X-chromosome will die.
- The remaining half male progeny will receive their X-chromosome from their mutagen treated grand father which may or may not carry the induced mutation.
- In case lethal mutation was induced, no males will be observed.
- On the other hand, if no lethal mutation was induced, half of the males will survive.
- Thus the ClB method was the simple, rapid and most efficient method for detecting sex-linked lethal mutations.

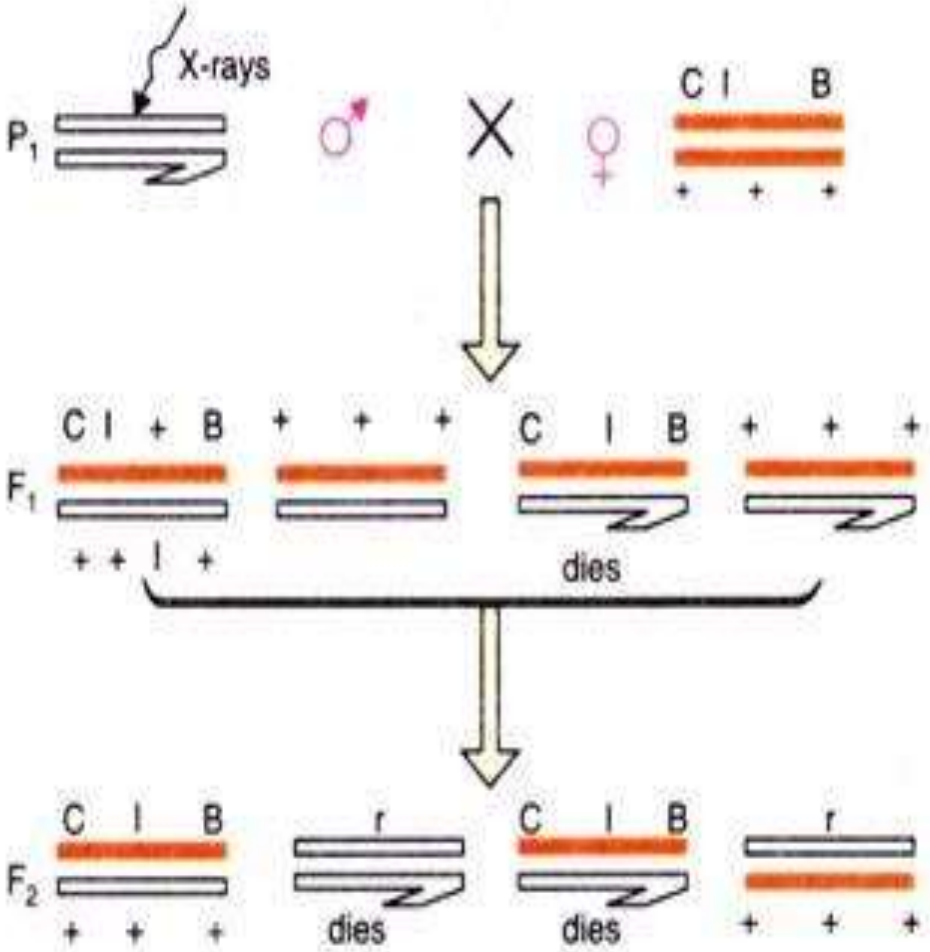


Fig 46.10. Diagram of Muller's CIB technique for detecting sex-linked (X-linked), recessive lethal mutations.

Detection of mutations in plants

- Generally the seeds of a variety or strain are treated with the mutagens and grown to obtain M1 plants.
- The M1 plants are selfed to avoid out crossing due to partial male sterility in M1 plants. The seeds thus obtained represent the M2 generation.
- The M2 plants are grown and the plants having mutant features are counted.
- Then the frequency of a given mutation is estimated as per cent ratio between the number of plants exhibiting a mutant phenotype in M2 and the total number of plants in M2.

$$\text{Mutation frequency (\%)} = \frac{\text{Number of plants exhibiting mutant phenotype in M2}}{\text{Total number of plants in M2}} \times 100$$

Significance of mutations in Plant Breeding:

1. When a variety is exceptionally good except for one or few characters.
2. When a recessive character is desirable and transfer of that character from wild species is difficult.
3. When a desirable character is linked with an undesirable character.
4. If there is no known source of resistance gene in the available germplasm.
5. To create variability.
6. To develop male sterile lines.
7. To create variations in vegetatively propagated plants.

THANK YOU