



MUTATION AND ITS ROLE

Presented by:
Dr. Ankit Kumar Singh
Assistant Professor
Department of Botany
Marwari College
Lalit Narayan Mithila University
Darbhanga
ankitbhu30@gmail.com

- Mutation can be defined as a sudden heritable change in the character or nucleotide sequence of an organism which is not due to either segregation or recombination
- In broad sense mutation includes all the heritable changes which alters the phenotype of an organisms.

- The term mutation was first used in *Oenothera lamarckiana* by Hugo de Vries for the sudden phenotypic changes which are heritable.
- 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*

Characteristic features of mutations

- Mutations are mostly recessive and very rarely dominant.
 - Most mutations have harmful effects and very few are beneficial.
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- Mutation may be due to a change in a gene, a group of genes or in entire chromosome.
 - If gene mutations are not lethal, the mutant individuals may survive.
 - Mutations are random i.e. they can occur in any tissue or cell of an organism
 - Mutations are recurrent i.e. the same mutation may occur again and again
 - Highly mutable sites within a gene are known as hot spots
 - Spontaneous mutations occurs at very low rate
 - Generally mutant alleles are recessive to their wild type or normal alleles

Classification of mutation

Based on causes of mutation:

Based on cause of mutation mutation are two types

A. Spontaneous mutation: Spontaneous mutation occurs naturally without any cause. The rate of spontaneous mutation is very slow.

B. 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.

Based on tissue of origin

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.

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.

Based on direction of mutation

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

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

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%)

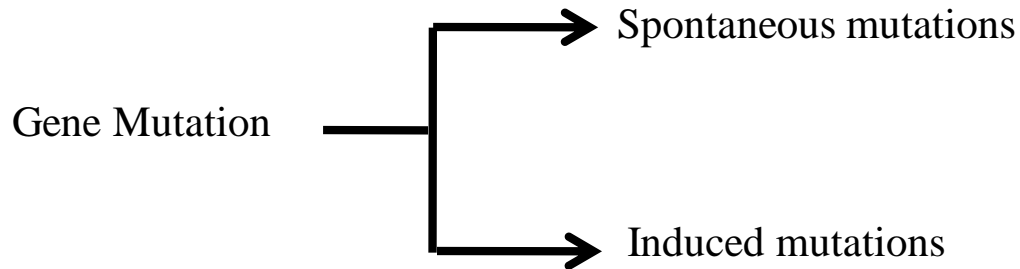
Based on the site of mutation

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

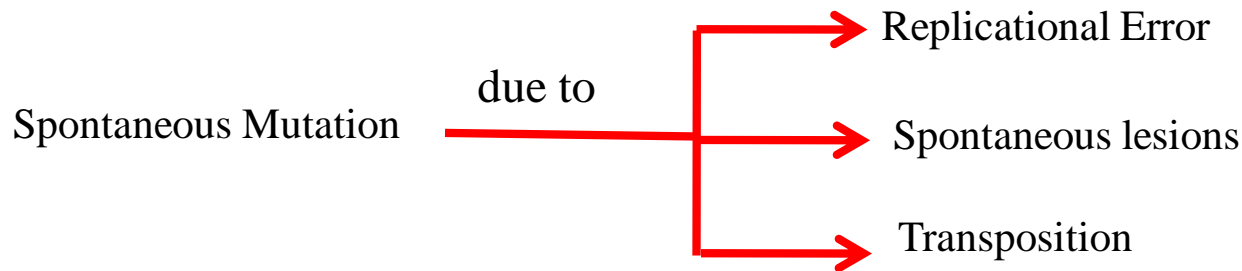
Gene or point mutations: Mutations produced by alterations in base sequences of concerned genes.

Cytoplasmic mutations: Mutations associated with the changes in chloroplast DNA (cpDNA) and mitochondrial DNA (mtDNA).

Molecular basis of gene mutation



Spontaneous mutations



Spontaneous mutations occur naturally without any apparent cause.

- There are three possible sources of origin of these mutations.
- The rate of spontaneous mutations is very low. 1 in 10 lakhs.
But different genes may show considerably different mutation rates.
- In crop plants some varieties were developed through spontaneous 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.

Mutations can be induced artificially using some agents known as mutagens. It may be

1. Physical mutagens
2. Chemical mutagens

1. Physical mutagens

Physical mutagens include ionizing and non ionizing radiations

Ionizing radiations: They work through the release of ions. They have deep penetrating capacity. Examples: x-rays, β -rays, etc.

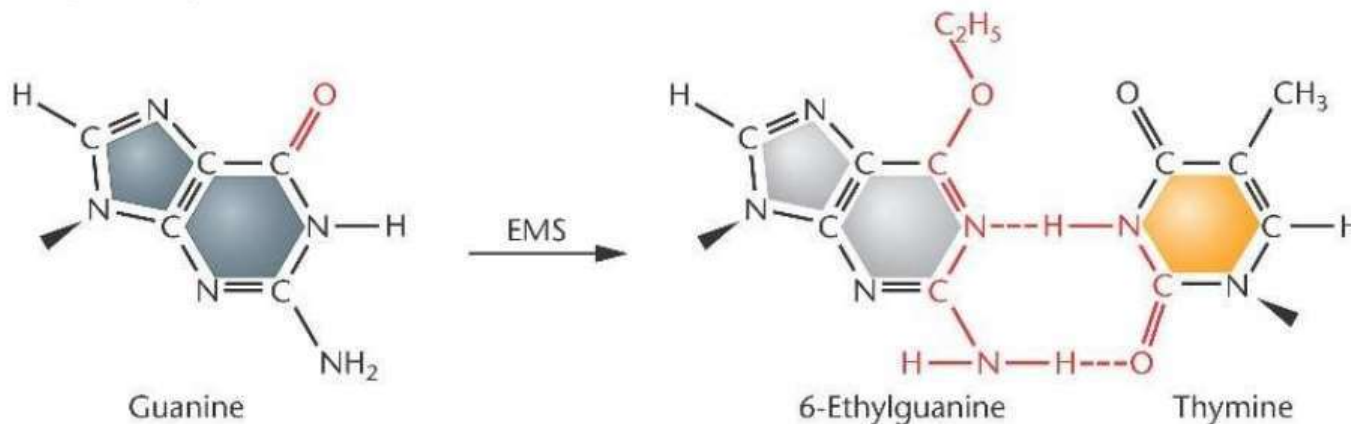
Non-ionizing radiations : They function through excitation and have a very low penetrating capacity. They are used for studies on bacteria and viruses. Example : UV rays

2. Chemical mutagens

Alkylating agents

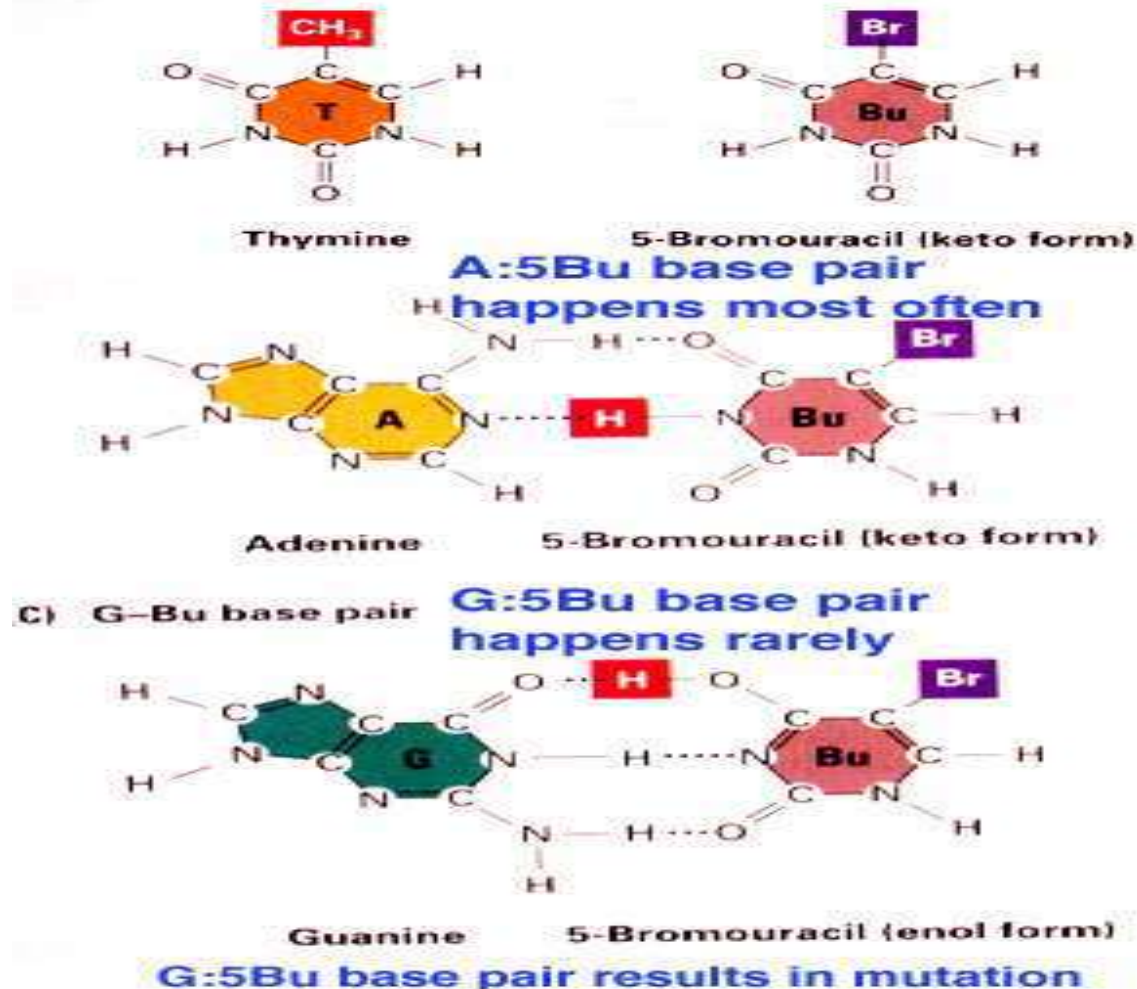
- This is the most powerful group of mutagens.
- These are the chemicals which are mainly used to induce mutations in cultivated plants.
- They induce mutations especially transitions and transversions by adding an alkyl group (either ethyl or methyl) at various positions in DNA.
- Alkylation produces mutation by changing hydrogen bonding in various ways.

Examples- Dimethyl sulphonate (DMS), Ethyl methane sulphonate (EMS), Nitrosomethyl Urea (NMU), Nitrosoethyl Urea (NEU), Methyl methane sulphonate (MMS).



Base analogues

- These are chemicals which are very similar to DNA bases, such chemicals are sometimes incorporated in DNA in place of normal bases during replication.
- Thus they can cause mutation by wrong base pairing.
- An incorrect base pairing results in transitions or transversions after DNA replication. Example: 5-bromouracil, 3-bromodeoxy uridine, 2-amino purine



Acridine dyes

- These are positively charged and they insert themselves between two base pairs of DNA.
- This is known as intercalation. Replication of intercalated DNA molecules results in addition or deletion of one or few base pairs which produces frame shift mutations.
- Acridine dyes Eg: proflavin, acriflavin, acridine orange, etc. are very effective mutagens

Miscellaneous

- Hydroxyl amine produce chromosomal aberrations.
- Nitrous acid (deaminating agent) has strong mutagenic activity in a variety of viruses and micro organisms. But not useful in higher plants

Molecular basis of mutations

- The term mutation is presently used to cover only those changes which alter the chemical structure of the gene at molecular level.
- Such changes are commonly referred to as “point mutations”. Point mutations involve a change in the base sequence of a gene which results in the production of a mutant phenotype.
- Point mutations can be subdivided into the following three classes on the basis of molecular change associated with them.
 1. Base substitution
 2. Base deletion
 3. Base addition

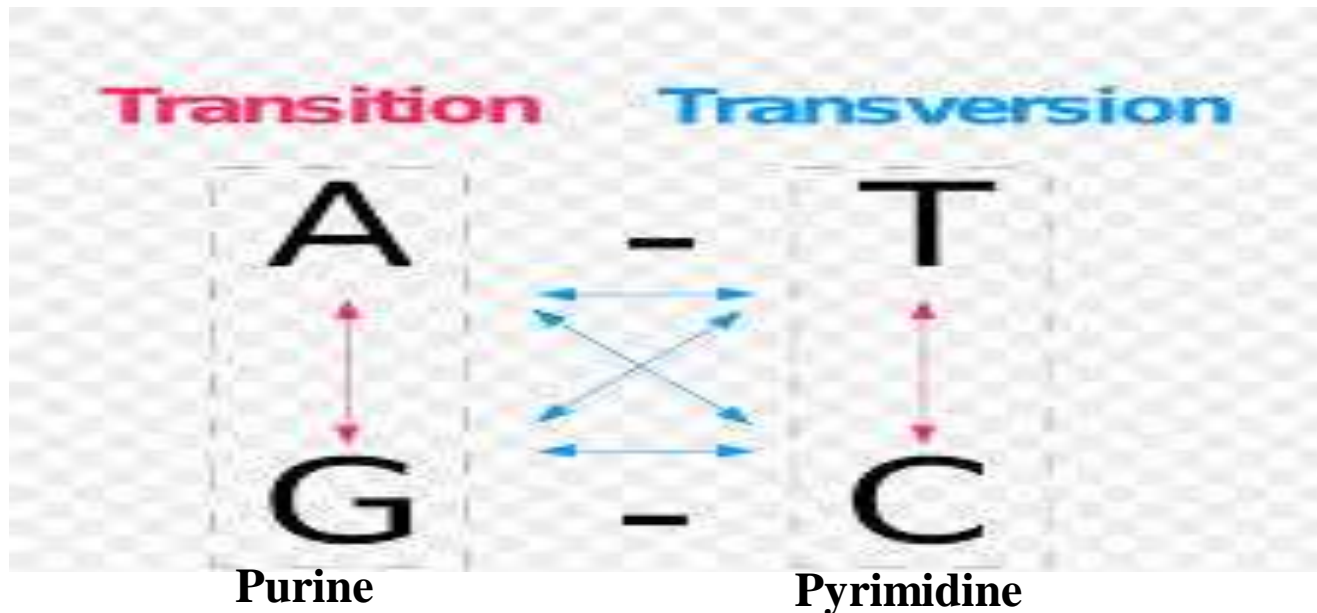
- DNA replication occurs when one double-stranded DNA molecule creates two single strands of DNA
- A single point mutation can change the whole DNA sequence. Changing one purine or pyrimidine may change the amino acid that the nucleotides code for.
- A point mutation, or single base substitution, is a type of mutation that causes the replacement of a single base nucleotide with another nucleotide of the genetic material, DNA or RNA
- Point mutations may arise from spontaneous mutations that occur during DNA replication. The rate of mutation may be increased by mutagens
- Replication errors that lead to substitution, insertion, or deletion mutations.

Base substitution

When a single base in a DNA molecule is replaced by another base it is known as base substitution. This can be of two types.

❖ **Transversion:** The substitution of a purine by a pyrimidine or of a pyrimidine by a purine in DNA or RNA is known as transversion.

❖ **Transition:** The substitution of a purine by another purine or of a pyrimidine by another pyrimidine base in DNA or RNA is known as transition.



Base deletion : In base deletion, one or more bases are altogether deleted

Base addition: There is insertion of one or more bases.

Frameshift mutations

The mutations which arise due to addition or deletion of nucleotides in mRNA are known as frameshift mutations, because the reading frame of base triplets (codons) beyond the point of addition or deletion is altered as a consequence of such mutations.

ACG AGG ACU GCA UAC CA...

Thr Arg Thr Ala Tyr

Normal Translation

A CGA GGA CUG CAU ACC A...

Arg Gly Leu His Thr

+1 Frameshifted Translation

AC GAG GAC UGC AUA CCA...

Glu Asp Cys Ile Pro

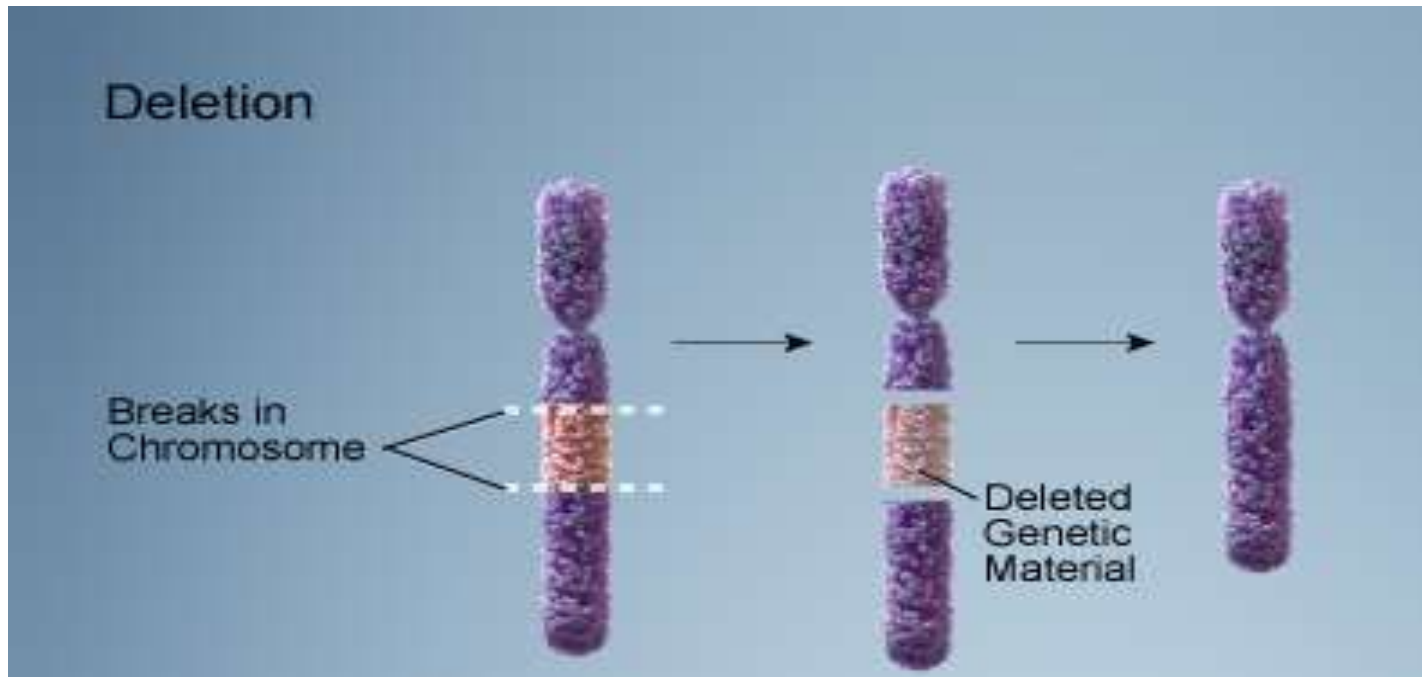
-1 Frameshifted Translation

Chromosome Mutations

Chromosomal mutations involve the change in the structure of chromosome. It is following types

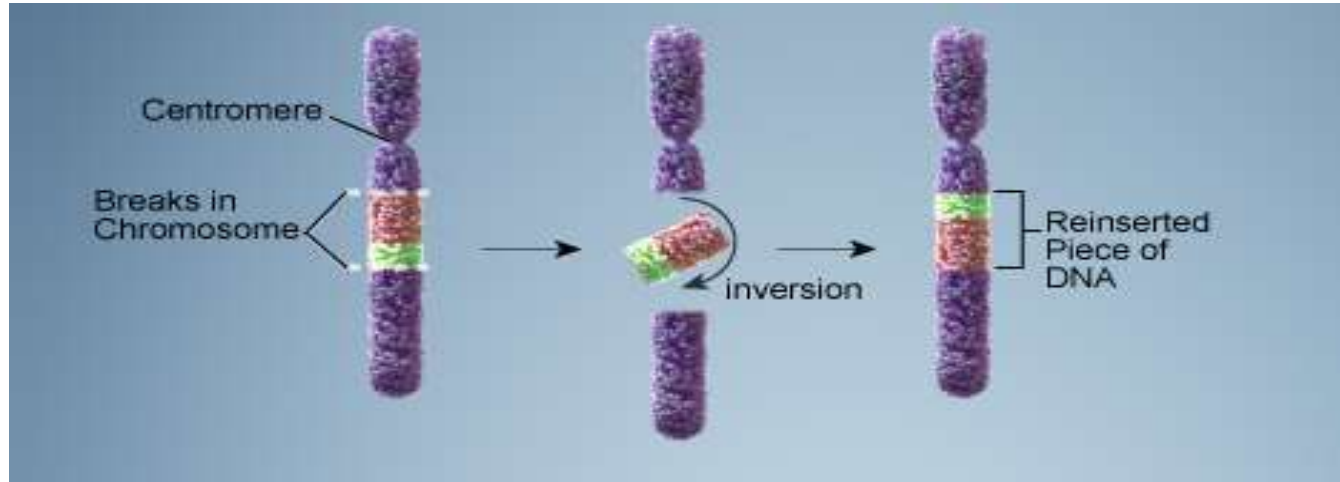
1. Deletion
2. Inversion
3. Translocation
4. Nondisjunction
5. Duplication

1. Deletion : Due to breakage in chromosome a piece of a chromosome is lost

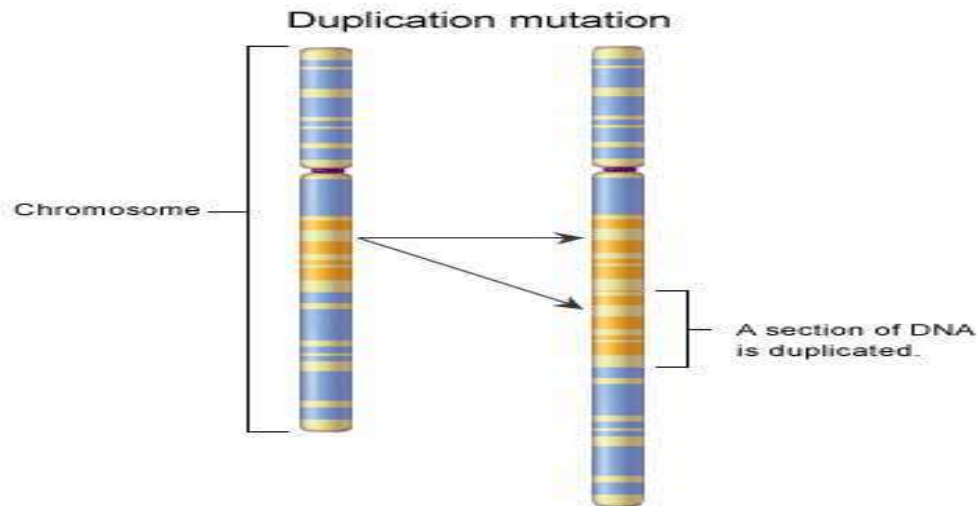


2. Inversion

Chromosome segment breaks off. After breakage segment flips around backwards and thereafter segment reattached

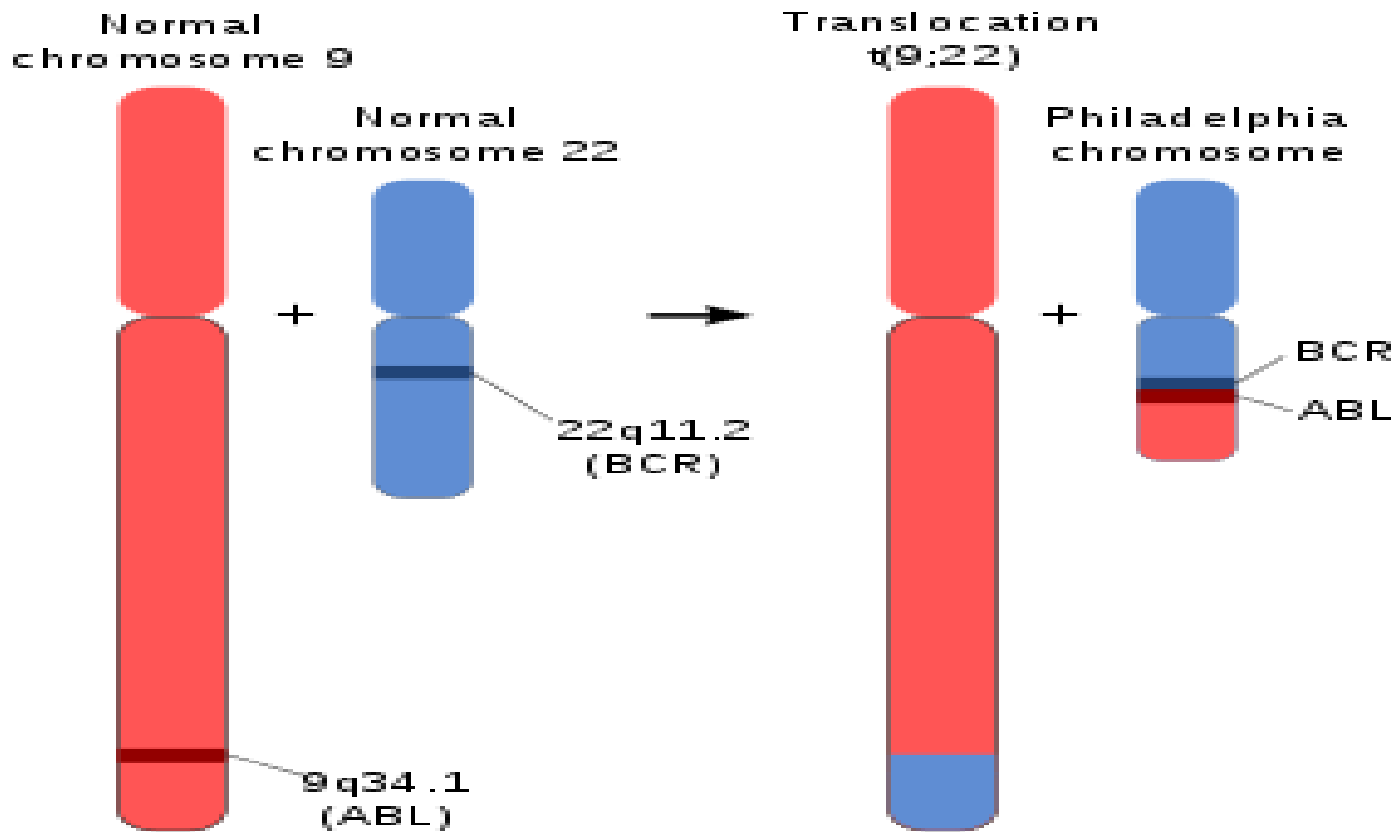


3. Duplication: Occurs when a gene sequence is repeated



4. Translocation

➤ Involves two chromosomes that are not homologous. Part of one chromosome is transferred to another chromosomes



5. Nondisjunction

- Failure of chromosomes to separate during meiosis
- Causes gamete to have too many or too few chromosomes
- Examples of Nondisjunction:
 - Down Syndrome
 - Turner Syndrome
 - Klinefelter's Syndrome

ROLE OF MUTATION

Mutation is generally deleterious and recessive but mutations also play beneficial role

- Different varieties in different crop plants have been obtained by mutation.
- Mutation create variations in vegetatively propagated plants
- Mutation leads to development of male sterile lines
- Mutation leads to speciation and play key role in evolution

- In wheat *Sharbati sonora* variety has been developed by gamma-rays mutation on Sonora 64 variety . **This variety is responsible for Green revolution in India.**
- In rice Reimei variety has been developed by mutation.
- In Castor Aruna variety has been developed by mutation.
- Penicillin production has been increased enormously by mutation.
- Besides these varieties, a number of other variety have been produced in different plants

Thank You !!