Introduction

- Sudden heritable change in genetic material or character of an organism is known as **mutation**
- Individuals showing these changes are known as **mutants**
- An individual showing an altered phenotype due to mutation are known as **variant**
- Factor or agents causing mutation are known as **mutagens**
- Mutation which causes changes in base sequence of a gene are known as **gene mutation** or **point mutation**

History

- English farmer **Seth Wright** recorded case of mutation first time in 1791 in male lamb with unusual short legs
- The term mutation is coined by **Hugo de Vries** in 1900 by his observation in Oenothera
- Systematic study of mutation was started in 1910 when **Morgan** genetically analyzed white eye mutant of Drosophila
- **H. J. Muller** induced mutation in Drosophila by using X- rays in 1927 ; he was awarded with Nobel prize in 1946

Characteristics of Mutation

- Generally mutant alleles are recessive to their wild type or normal alleles
- Most mutations have harmful effect, but some mutations are beneficial
- Spontaneous mutations occurs at very low rate
- Some genes shows high rate of mutation such genes are called as mutable gene
- Highly mutable sites within a gene are known as hot spots.
- 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

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

- **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

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

- **Based on direction of mutation**

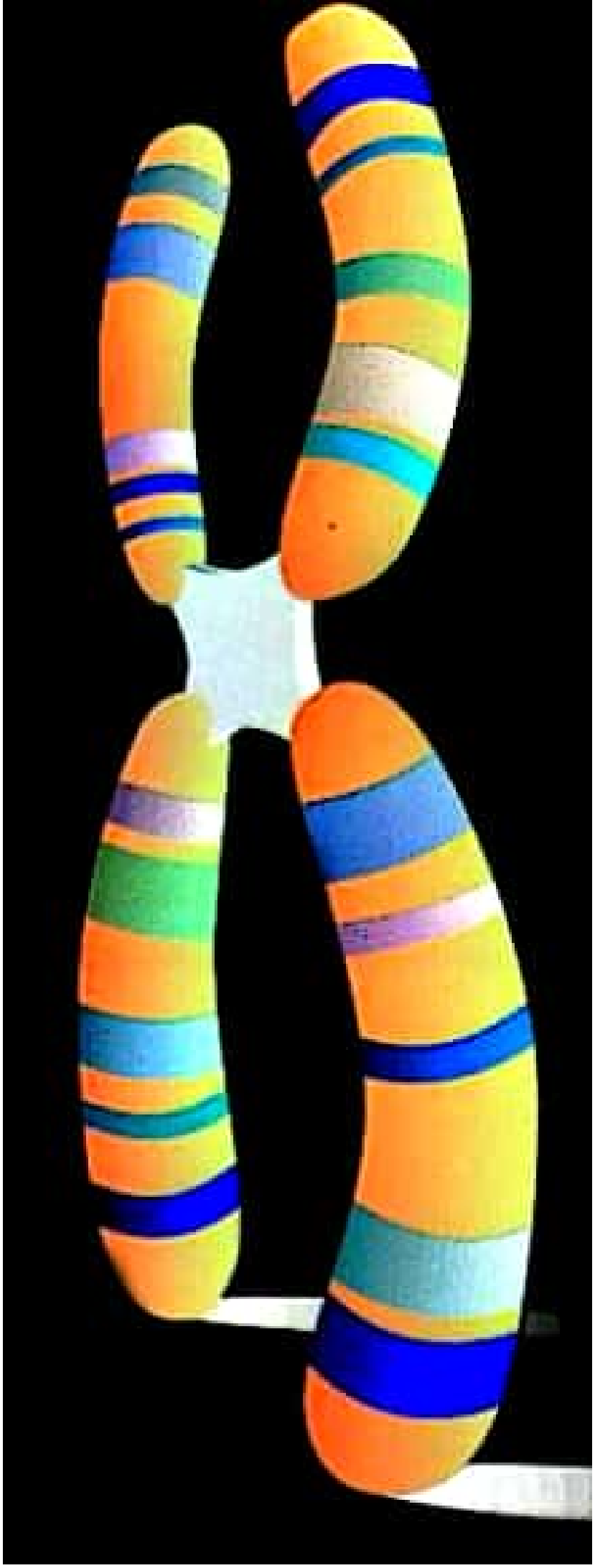
1. Forward mutation- When mutation occurs from the normal/wild type allele to mutant allele are known as forward mutation

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

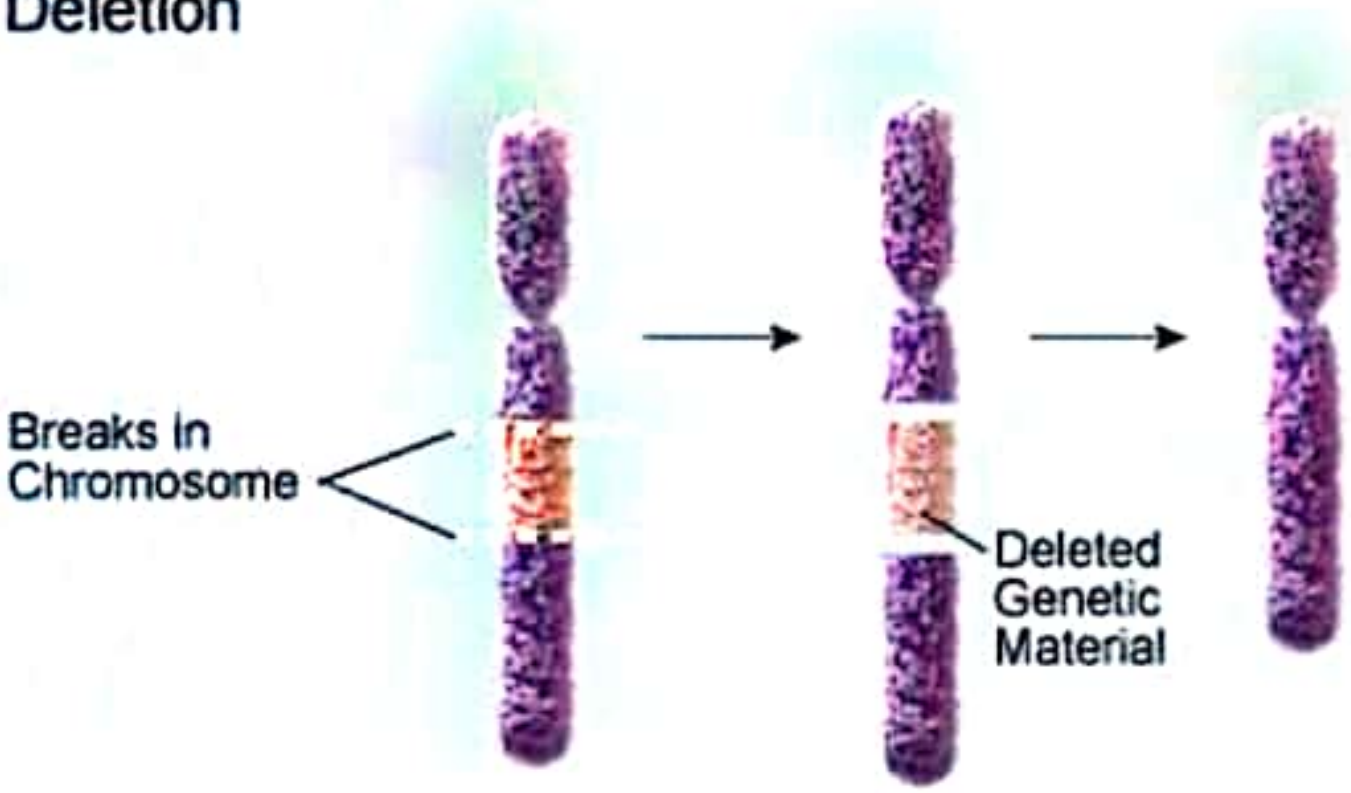
2. **Biochemical mutation-** mutation which affect the production of biochemicals and which does not not show any phenotypic character are known as biochemical mutation



Chromosome Mutations

- Five types exist:
  - Deletion
  - Inversion
  - Translocation
  - Nondisjunction
  - Duplication

Deletion

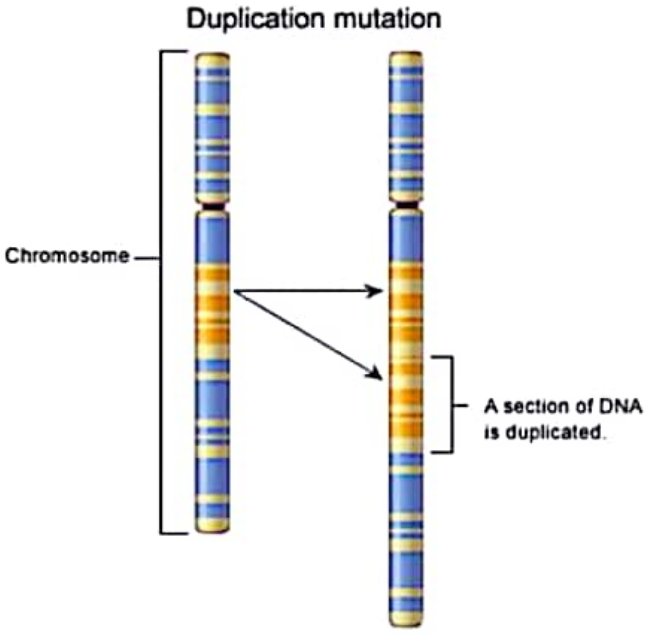


Inversion

- Chromosome segment **breaks off**
- Segment flips around **backwards**
- Segment **reattaches**

Duplication

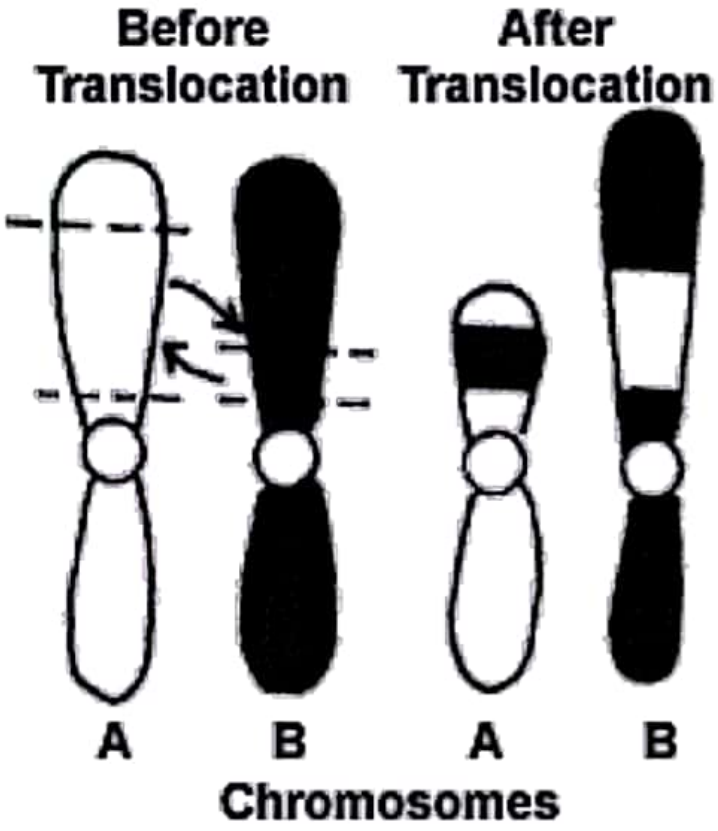
- Occurs when a gene **sequence is repeated**



Translocation

- Involves **two chromosomes** that aren't homologous
- **Part** of one chromosome is **transferred to another** chromosomes

Translocation



Nondisjunction

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

1. Original

Types of Gene Mutations

- Include:
  - Point Mutations
  - Substitutions
  - Insertions
  - Deletions
  - Frameshift

Point Mutation

- Change of a **single** nucleotide
- Includes the deletion, insertion, or substitution of **ONE** nucleotide in a gene

Point Mutation

- **Sickle Cell disease** is the result of one nucleotide substitution
- Occurs in the **hemoglobin gene**

Frameshift Mutation

- **Inserting or deleting** one or more nucleotides
- Changes the "**reading frame**" like changing a sentence
- **Proteins** built **incorrectly**