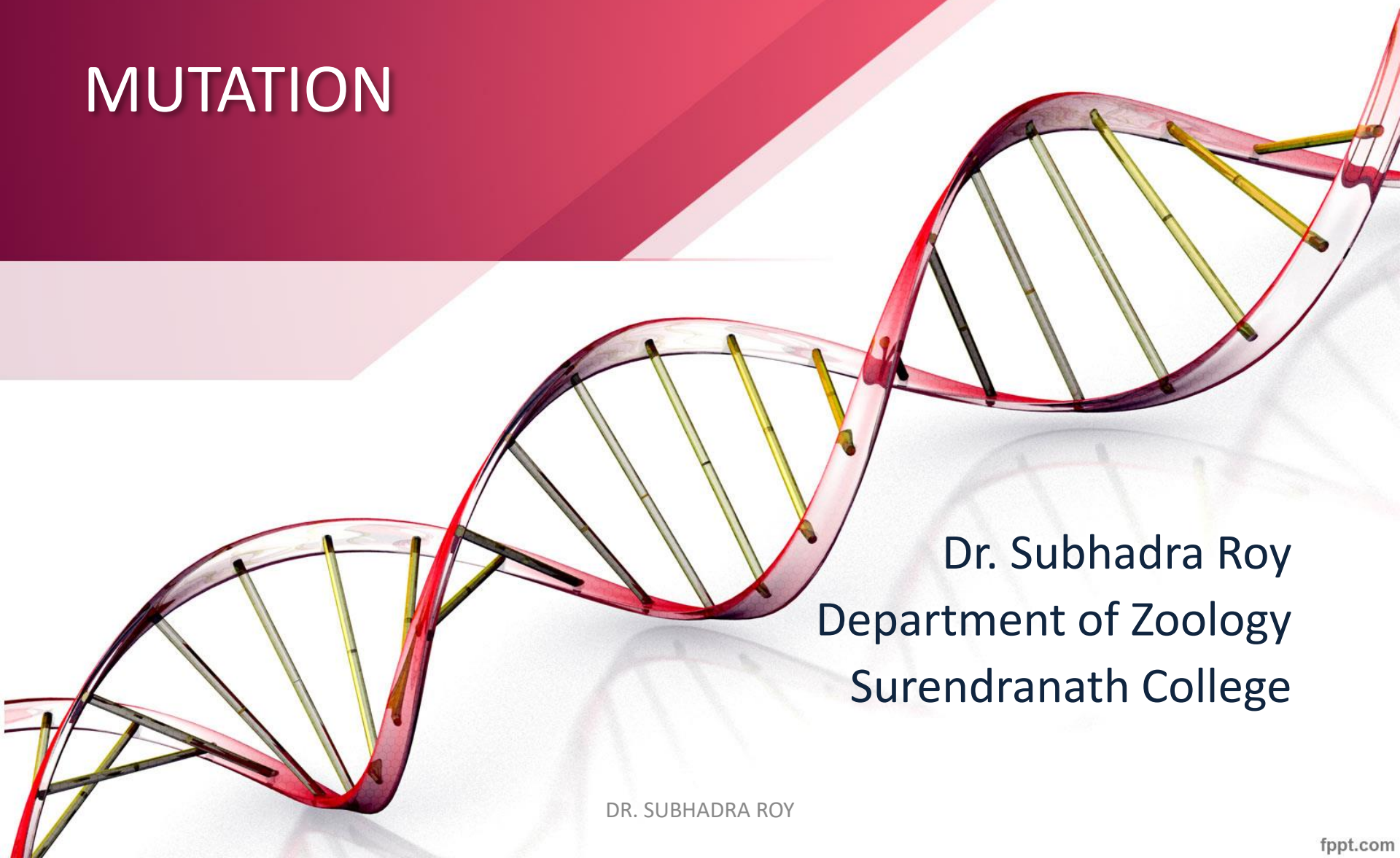
MUTATION



Dr. Subhadra Roy
Department of Zoology
Surendranath College

Hugo De Vries

Mutation theory-

The theory states that evolution is a jerky process where new varieties and species are formed by mutations (discontinuous variations) that function as raw material of evolution.

Important definitions

- Mutation- An alteration in the genetic material (the genome) of a cell of a living.
- Chromosomal mutation- Changes or alterations in the structures of chromosomes involving changes either in the total number of genes or gene loci in a chromosome or their rearrangement.
- Gene mutation- A gene **mutation** is a permanent alteration in the DNA sequence that makes up a gene
- Engage your Audience

Important definitions

- Mutagen- Any agent that causes an increase in the spontaneous rate of mutation.
- Mutant- A cell or organism carrying an altered or mutant allele.
- Mutation rate- The frequency with which mutations take place at a given locus or in a population.
- Muton - In the fine structure analysis of the gene, the smallest unit of mutation, corresponding to a single base change.

TYPES OF CHROMOSOMAL MUTATIONS

Structural changes in chromosomes

Changes involving the number of the gene loci

- Deficiency or Deletion
- Duplication

Changes involving the arrangement of gene loci

- Translocation
- Inversion

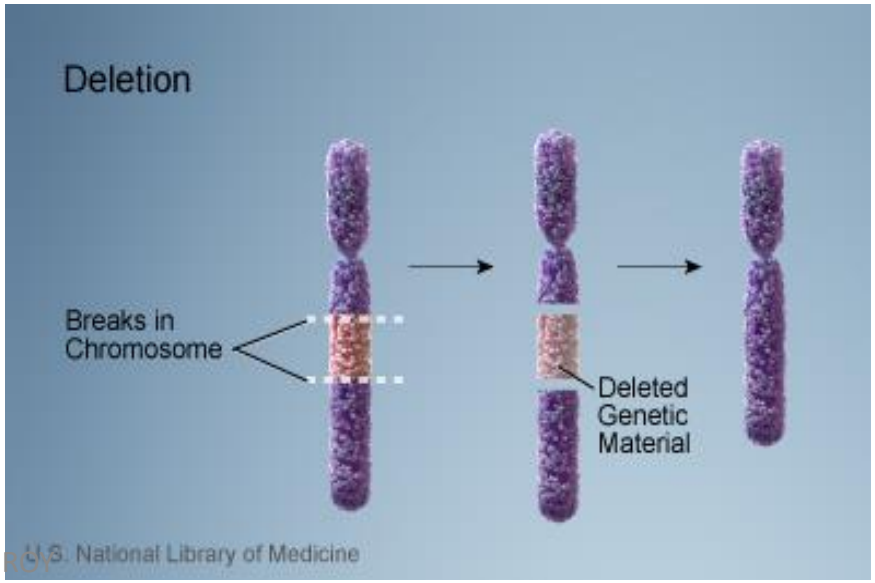
Deficiency or Deletion

Results due to loss of a part of a chromosome. Deletion occuring on only one of the two homologous chromosomes (i.e. present in heterozygous condition) can be tolerated by an organism. At meiosis, such individuals will form a loop in a bivalent that can be observed at pachytene stage.

In presence of a deficiency, a Recessive allele will behave like dominant allele , a phenomenon called **pseudodominance.**

Examples include

- waltzing mice
- Cri-du chat syndrome in human



Duplication

Obtained due to addition of a part of a chromosome.

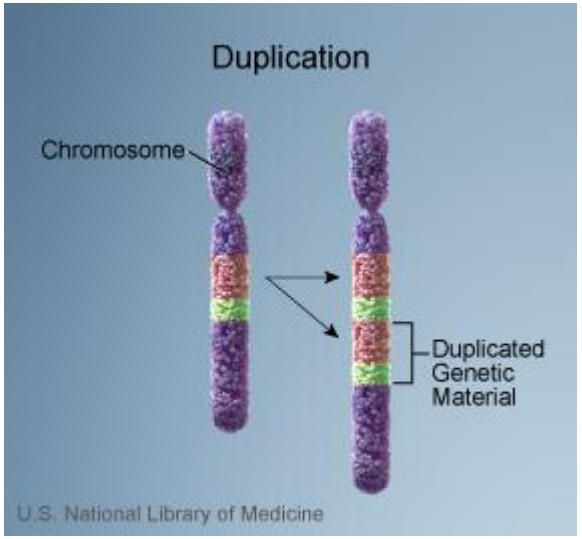
Addition of chromosome segment may occur at any of the following positions:

- In adjacent region
- At a displaced position of the same arm
- On the different arm of the same

Chromosome

- On a different chromosome.

Example includes Bar eye character in Drosophila

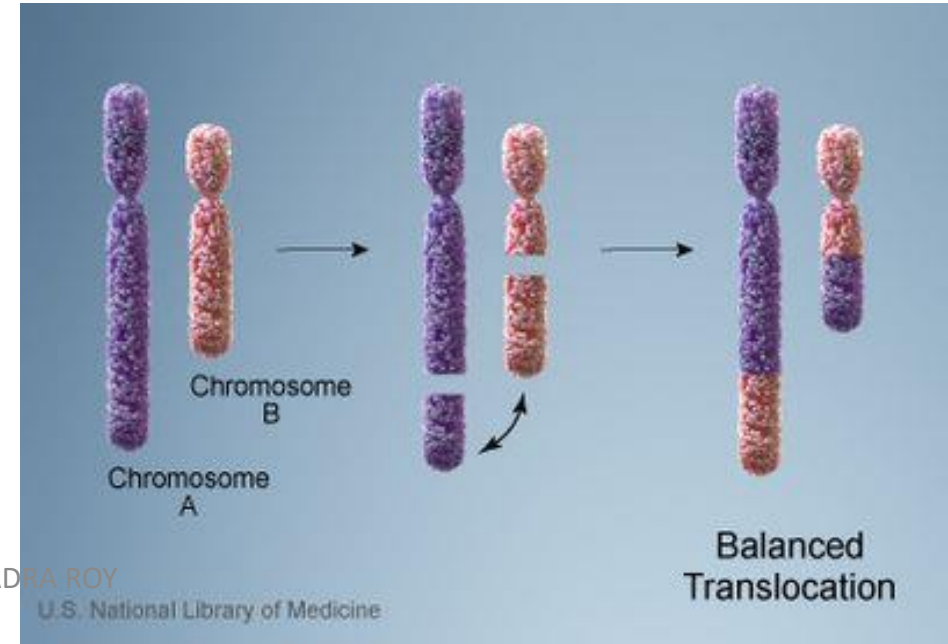


Translocation

Includes all types of unilateral or bilateral transfer of chromosome segments from one chromosome to another. Mutual exchange of chromosome segments between two pairs of non-homologous chromosomes is called reciprocal translocations or segmental interchanges.

Example-

In chronic myeloid leukemia, Philadelphia chromosome is formed by translocation between chromosomes 9 and 22.



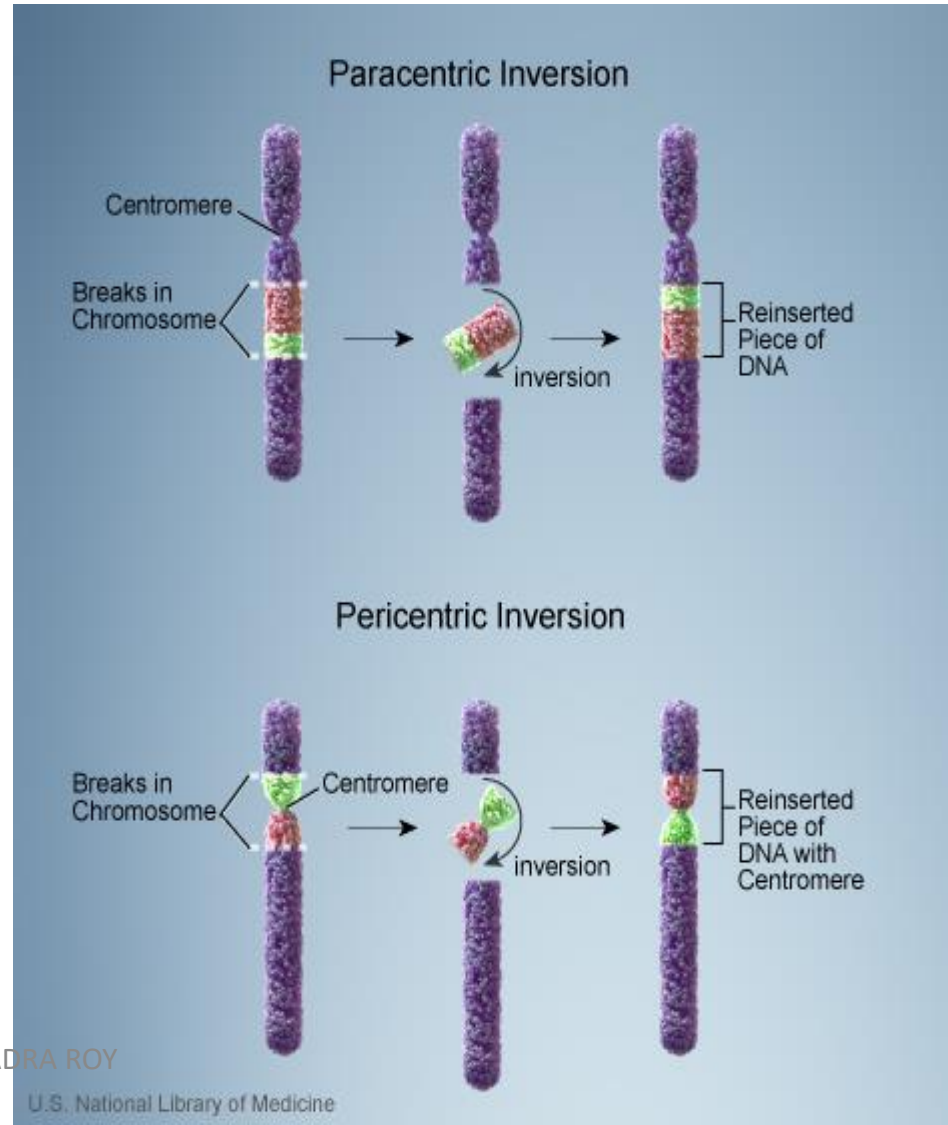
Inversion

It is formed when there are two breaks in a chromosome and the intercalary segment reunites in reverse order. Inversions are often called crossover suppressors. It can be of two types:

i. Paracentric inversion

ii. Pericentric inversion

Example- The most common inversion seen in humans is on chromosome 9. This inversion is generally considered to increase the risk for miscarriage or infertility for some affected individuals



TYPES OF CHROMOSOMAL MUTATIONS

Numerical changes in chromosomes (Heteroploidy)

Aneuploidy

- Monosomy
- Nullisomy
- Trisomy
- Tetrasomy

Euploidy

- Monoploidy
- Haploidy
- Polyploidy

Aneuploidy

Aneuploidy means presence of chromosome number which is different than a multiple of basic chromosome number. It can be

- Hypoploidy- loss of one or more chromosomes
  - Monosomy (2n-1)
  - Nullisomy (2n-2)
- Hyperploidy-addition of one or more chromosomes
  - Trisomy (2n+1)
  - Tetrasomy (2n+2)

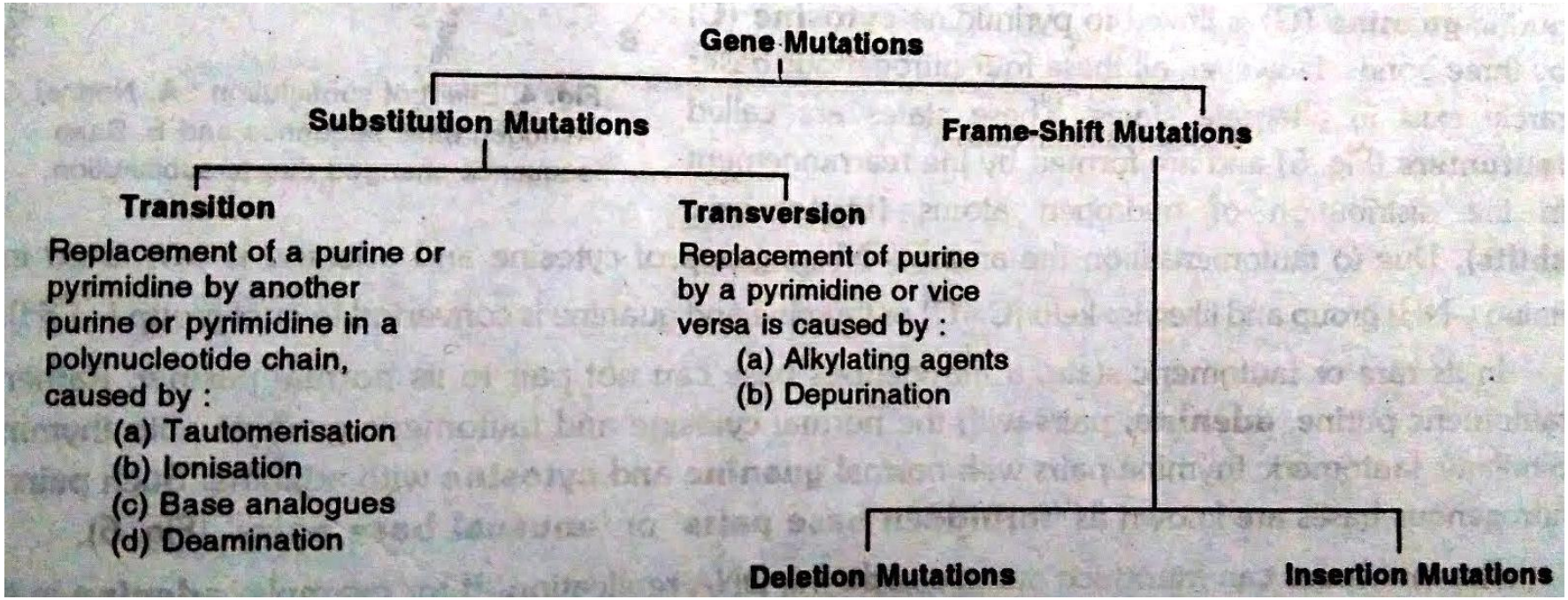
Gene mutation or Point mutation

A mutation that can be mapped to a single locus is called gene or point mutation. This include changes in gene structure or composition.

Detection of mutations

- ClB method in Drosophila
- Muller-5 method in Drosoplila
- Balanced lethals for detecting induced mutations in autosomes
- Detection of mutations in microorganisms

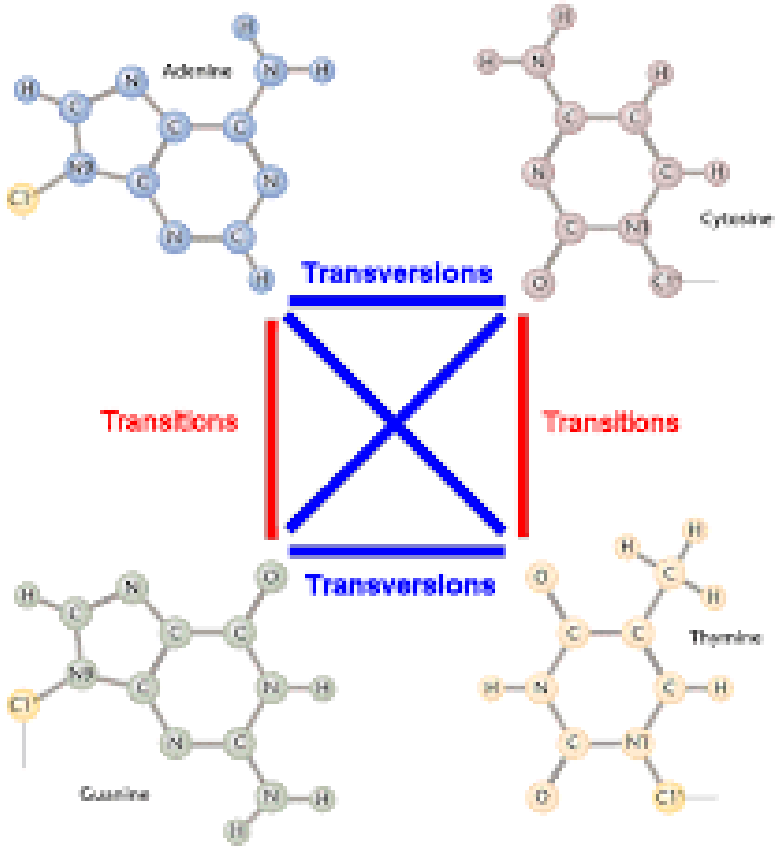
Types of gene mutations



Substitution mutations

Substitution mutations

Are a type of mutation in which a single nucleotide is substituted with a different nucleotide. Examples of (base-pair) substitutions: a purine is substituted with a different purine (A → G) or a pyrimidine, for a different pyrimidine (C → T). This type of substitution mutation is referred to as transition. Another type of base-pair substitution is the so-called transversion. In transversion, a purine is substituted with a pyrimidine or a pyrimidine with a purine.

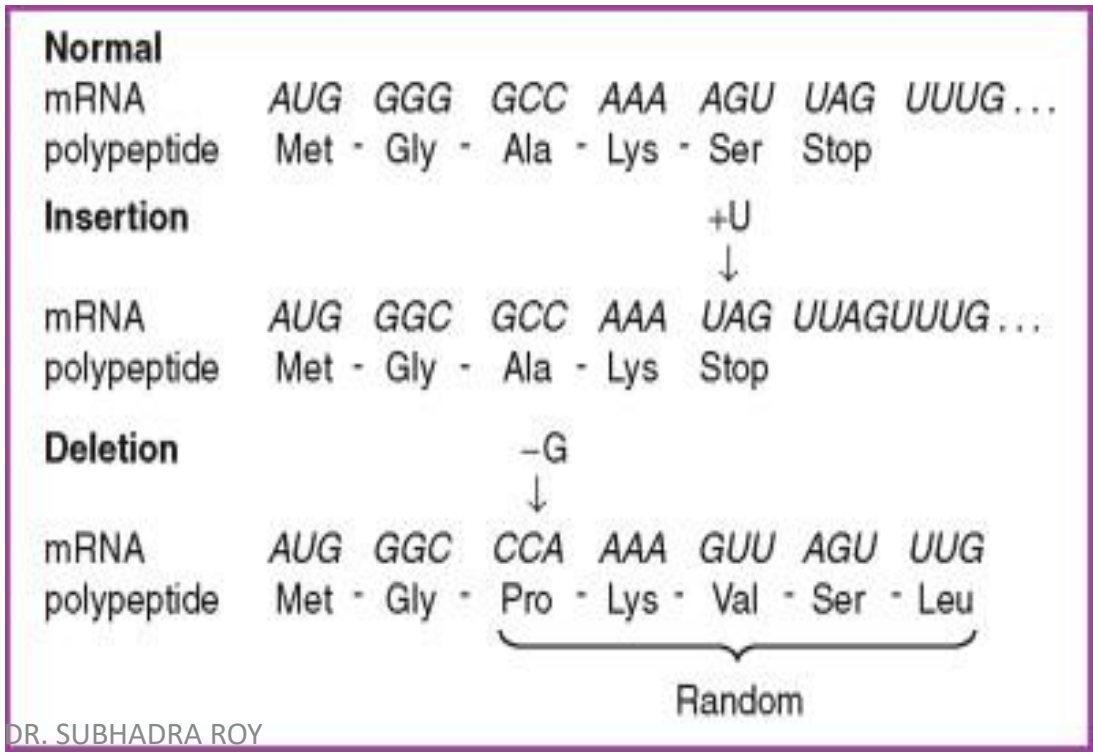


Frameshift Mutation

Frameshift mutation

Frameshift mutations are deletions or additions of 1, 2, or 4 nucleotides that change the ribosome reading frame.

A **frameshift mutation** occurs when nucleotides are inserted into or deleted from the DNA and cause a "shift" in the reading of mRNA codons.



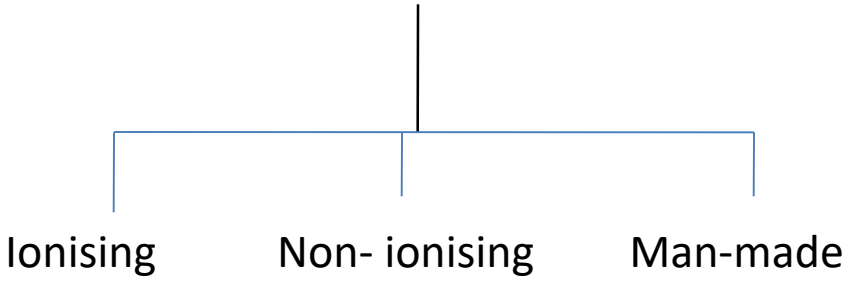
Induced mutation

Induced mutations are alterations in the gene after it has come in contact with mutagens and environmental causes.

In 1927, **H.J. Muller** showed for the first time that mutations could be induced in Drosophila by use of external agents or mutagens. He was awarded Nobel Prize in 1946.

Causes of induced mutations (Mutagens)

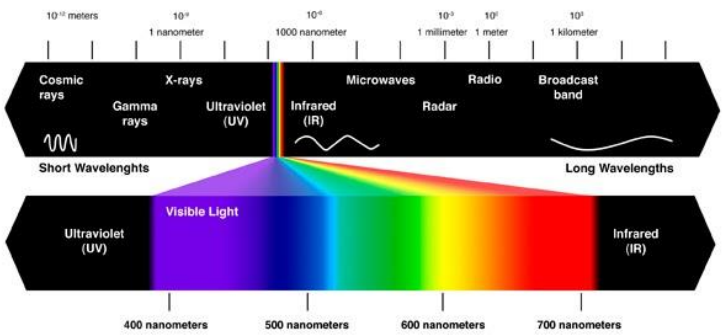
Radiations



Chemicals

- Alkylating agents (ENU)
- Methylating agents (EMS)
- Polycyclic hydrocarbons
- DNA intercalating agents
- DNA crosslinker
- Oxidative damage

Radiations



Ionising Radiations

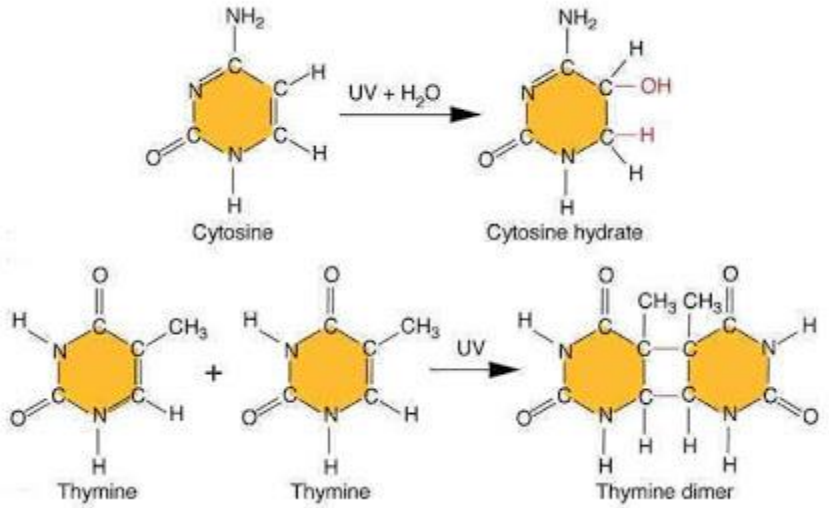
X-rays, gamma rays, alpha and beta rays, neutrons, protons & other fast moving particles.

Non-ionising radiations

Have longer wavelengths and and carry much lower energy. Examples- Ultraviolet light rays.

Man-made radiations

These include the ionising and non-ionising radiations that the experimental animals are exposed to.



Chemical agents

TABLE 25.3

Some commonly used chemical mutagens and their mode of action

Group of mutagen	*Name of chemical*	*Mode of action*
1. Alkylating Agents	Ethyl Methane Sulphonate	*AT* ↔ *GC* Transitions
	Methyl Methane Sulphonate	Transitions
	Ethyl Ethane Sulphonate	*GC* ↔ *AT* Transitions
	Ethylene Imines	Transitions.
2. Base Analogues	5 Bromo Uracil	*AT* ↔ *GC* Transitions
	2. Amino Purine	*AT* ↔ *GC* Transitions
3. Acridine Dyes	Acriflavin, Proflavin	Deletion addition and Frameshifts.
4. Others	Nitrous Acid	*AT* ↔ *GC* Transitions
	Hydroxylamine	*GC* ↔ *AT* Transitions
	Sodium Azide	Transitions

THANK YOU