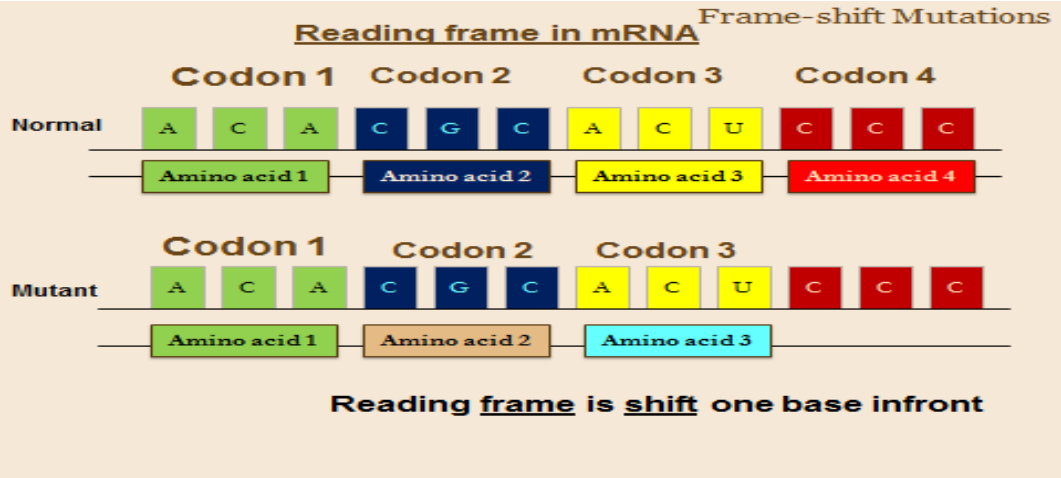
Genetics

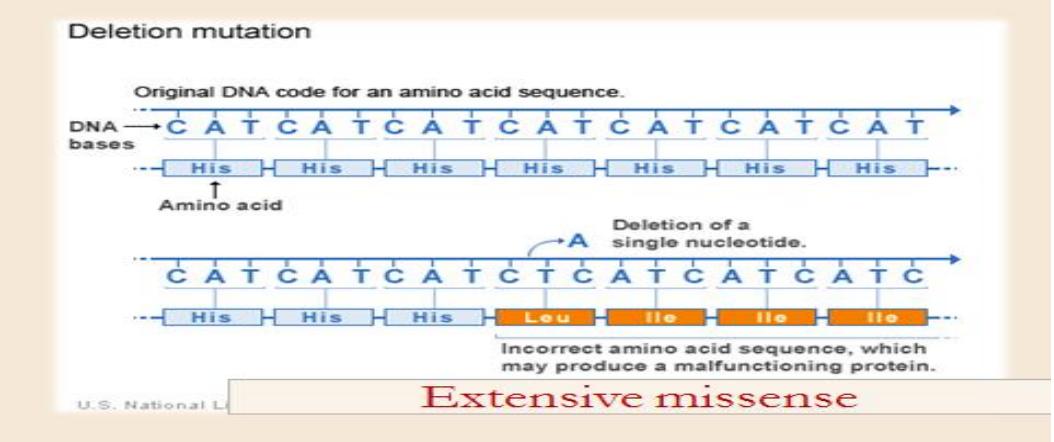
Biology Dept.3rd stage

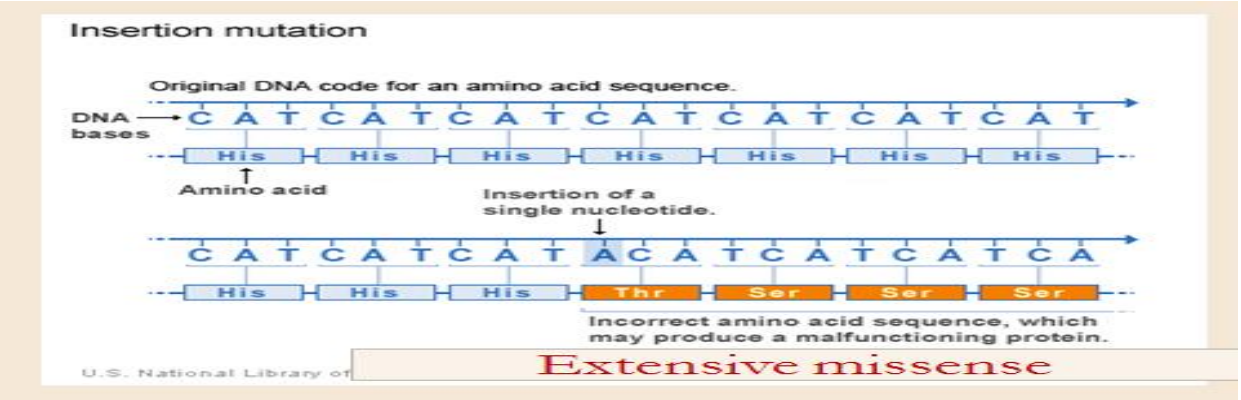
Asst. Professor. Dr. Kazhal M. Sulaiman

Lec-8- Insertions and deletions

Extra base pairs may be added or deleted from the DNA of a gene. The number of bases can range from a few to thousands. Insertions and deletions of one or two bases or multiples of one or two cause frameshifts (shift the reading frame). These can have devastating effects because the mRNA is translated in new groups of three nucleotides and the protein being produced may be useless.

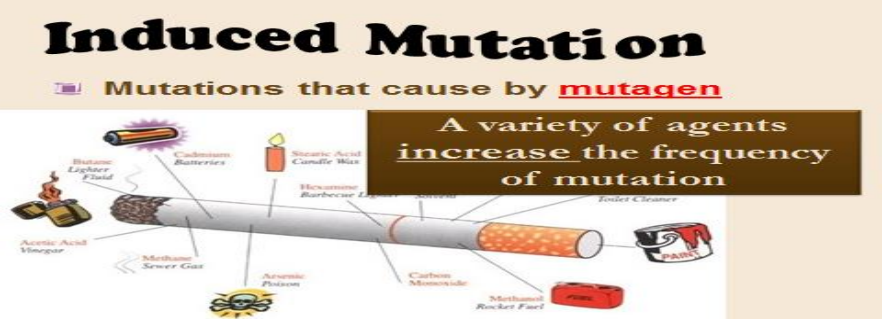


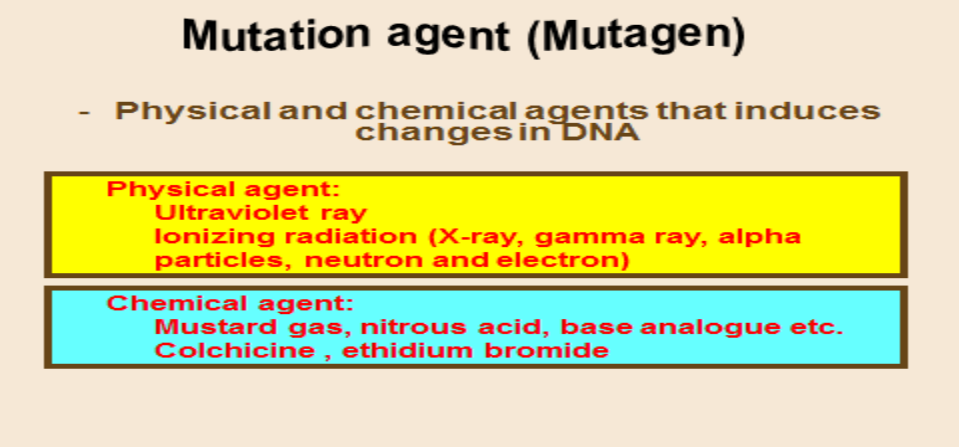




2-Induced mutation

Induced mutations on the molecular level can be caused by:





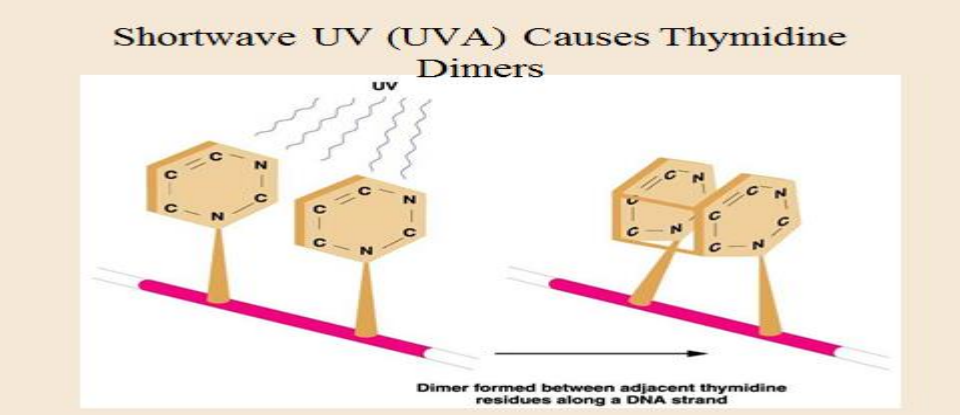
Physical agent

Physical agent	Effect
Ionizing radiation	• -breaks in one or both strands (can lead to rearrangements, deletions, chromosome loss, death if unrepaired; this is from stimulation of recombination) • -damage to/loss of bases (mutations) • -crosslinking of DNA to itself or proteins
UV radiation	pyrimidine dimers in DNA

Radiation •

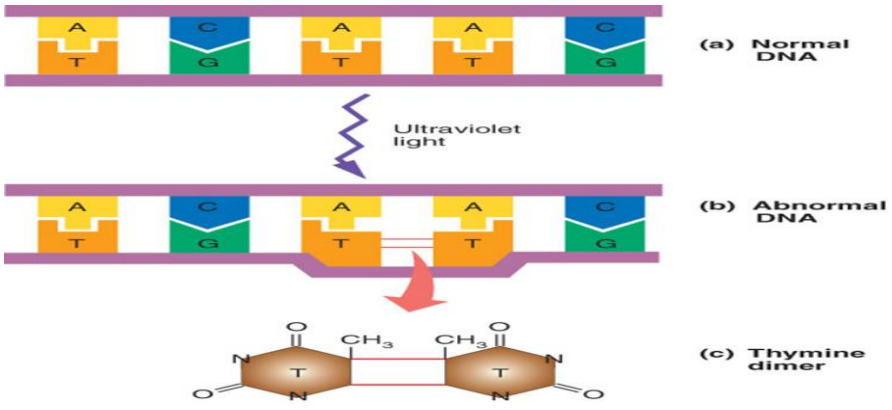
Ultraviolet radiation (nonionizing radiation). Two nucleotide bases in DNA – cytosine and thymine – are most vulnerable to radiation that can change their properties. UV light can induce adjacent pyrimidine bases in a DNA strand to become covalently joined as a pyrimidine dimer

•



- Chemical agent

Chemical agent	Effect
nitrous acid-	It causes C to U, meC to T, and A to hypoxanthine deaminations
ethyl methanesulfonate	mutagenic and recombinogenic, or mispair to result in mutations upon DNA replication.
Colchicine	Prevent the formation of spindle fibre



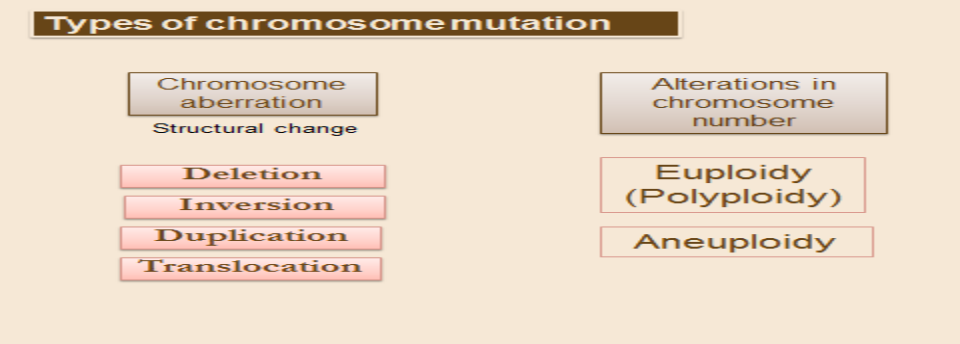
chromosomal aberrations or chromosomal mutations

Definition:

- **Abnormalities ~ in chromosomal structure (chromosome aberration) & changes in chromosome number (aneuploidy / euploidy)**

The changes in the genome involving a chromosome or part of a chromosome are called as **chromosomal aberrations** or **chromosomal mutations**. Mutation are observed in all organisms from bacteria to man and arise all of a sudden. They may be dominant or recessive, sex-linked or

autosomal, germinal or somatic, lethal or non-lethal possibly because they disturb the genic balance



They are heritable changes and are of following types-

A. Changes in structure of chromosomes

1-Change in number of genes (Addition or Deletion)

2- Rearrangement of genes (Inversion or Translocation)

B-Changes in number of chromosomes

1-Change in number of part of chromosome set (Aneuploidy)

2-Change in number of entire chromosome set (Haploidy or Polyploidy)

STRUCTURAL CHANGES IN CHROMOSOMES

The structural changes in chromosomes which appear phenotypically are known as chromosomal mutations or aberrations.

1. Deficiency or Deletion.

It involves the loss or absence of a part of chromosome involving one or more genes. Genic balance is usually disturbed and this affects the phenotype. If deletion occurs in the chromosome of gametes, it will be transmitted to the next generation.

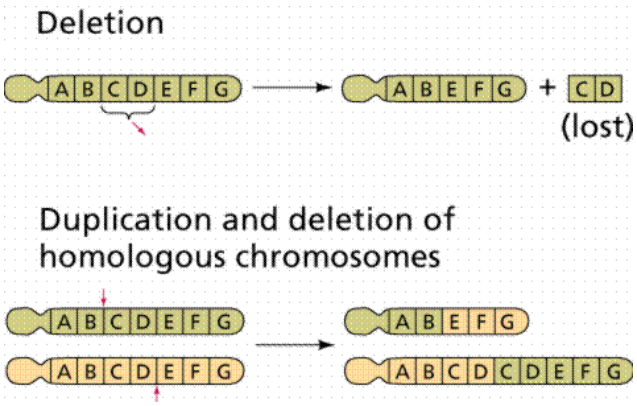


Fig.7.1. Effect of deletion and duplication on a chromosome. In the second diagram, CD genes are deleted from one chromosome and are inserted into another, causing its duplication.

Deletion can be terminal or intercalary.

Terminal deletion involves loss of chromosome segment from its one end. Due to this, one of the paired chromosome appears to be longer than the other.

Intercalary deletion involves loss of intermediate chromosome segment. Due to this, the normal chromosome forms a loop near the deleted region of its homologue as only identical regions pair with each other. An interesting example of deletion in humans is ***Cri du chat syndrome***, where the short arm of chromosome 5, i.e. 5p (p means short arm; q is long arm) is deleted. Individuals having this syndrome have a distinctive cat-like cry, are mentally retarded, moon face, low set ears and small head (*microcephally*).

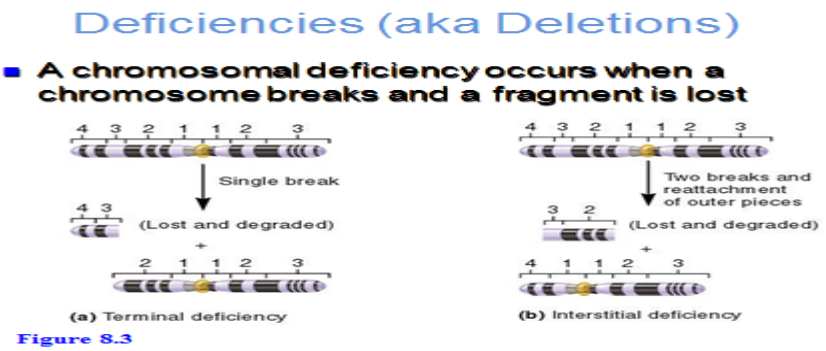


Figure 8.3

- Deletion generally produce striking genetic and physiological effects.
- When homozygous, most deletions are lethal, because most genes are necessary for life and a homozygous deletion would have zero copies of some genes.
- When heterozygous, the genes on the normal homologue are hemizygous: there is only 1 copy of those genes.