Quadrant II – Transcript and Related Materials

Programme : Bachelor of Science (First Year)

Subject : Zoology

Paper Code : ZOC 102

Paper Title : Diversity of Chordates and Genetics

Unit : 10

Module Name : Types of Chromosomal Aberrations

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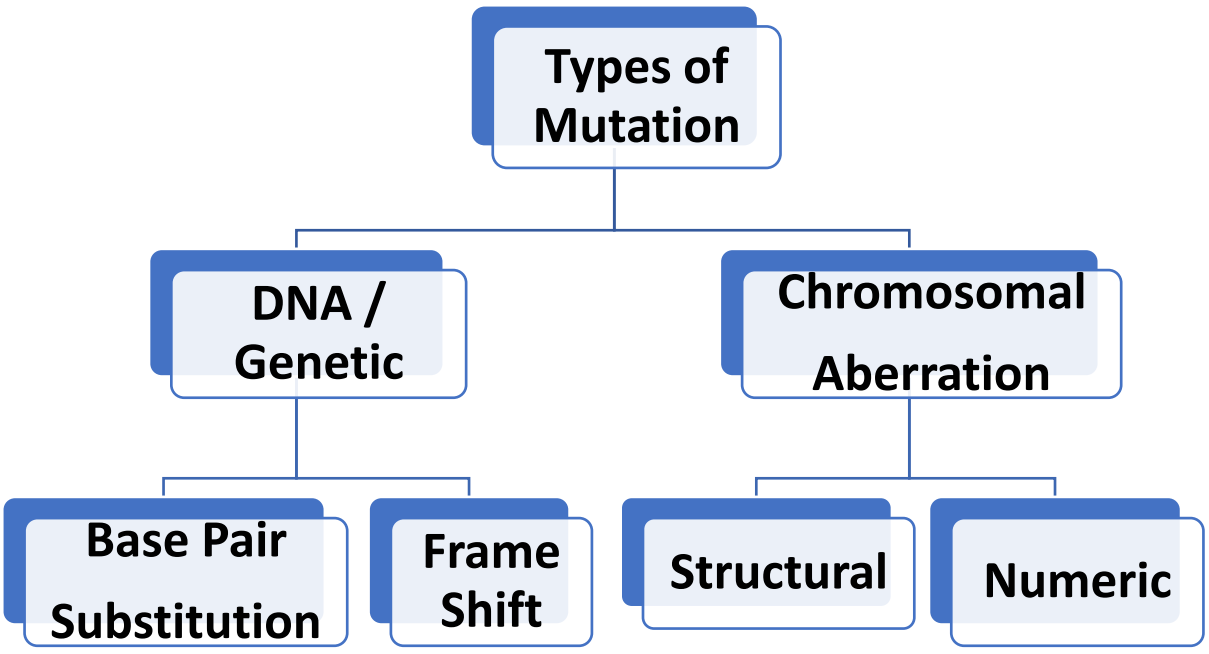
Notes

Mutation

A mutation is an alteration in the nucleotide sequence of the genome of an organism, virus, or extra chromosomal DNA. The Mutation can be classified into two types i.e. 1. Mutation in DNA or Nucleotide bases and 2. Mutation in Chromosome. The mutation at chromosomal level is referred as Chromosomal aberrations.



Chromosomal aberrations

Chromosomal aberrations, or abnormalities, are changes to the structure or number of chromosomes, which are strands of condensed genetic material. Chromosomal Aberration may cause medical /Genetic disorders in organism. Chromosomal aberrations are further classified into two types;

1. Structural Chromosomal Aberration
2. Numerical Chromosomal Aberrations

1. Structural Chromosomal Aberration

Loss or gain of chromosomal segments / Gene in a chromosome is termed as Structural Chromosomal Aberration. They are of following types

a) Deletion: Loss of a segment of the genetic material in a chromosome is called Deletion. If the loss of chromosomal segment is from the tip of the chromosome it is referred as **terminal deletion** where as if the loss of chromosomal segment is from central body it is referred as **Intercalary deletion.**

b) Duplication: When a segment of chromosome presents more than once in the chromosome then its called duplication.

c) Inversion: When an order of the gene in chromosome in particular segment is reversed by 180, it is referred as Inversion. If the inversion of chromosome segment also includes centromere then it's termed as **Pericentric Inversion** while if the inversion doesn't involve centromere its termed as **Paracentric Inversion**.

d) Translocation: Interchange of Chromosomal segment between two chromosomes occur then called as translocation.

2. Numerical Chromosomal Aberrations

Alteration in the number of chromosomes from the diploid set is called as numerical chromosomal Aberrations. It is also known as Ploidy. These are usually caused by a failure of chromosome division, which results in cells with an extra chromosome or a deficiency in chromosomes. Gametes with these anomalies can result in conditions such as Down syndrome (who have 47 chromosomes instead of 46), or Turner syndrome (45 chromosomes). Numerical chromosomal Aberrations are classified into following types.

A) Euploidy: Euploidy is a chromosomal variation that involves the entire set of chromosomes in a cell or an organism. Euploidy is more tolerated in plants than in animals. There may be a single set (monoploidy), two sets (diploidy), or multiple sets (polyploidy, i.e. triploid, tetraploid, pentaploid, hexaploid, etc.) of chromosomes.

B) Aneuploidy: It is the second major category of chromosome mutations in which chromosome number is abnormal. An aneuploid is an individual organism whose chromosome number differs from the wild type by part of a chromosome set. Generally, the aneuploid chromosome set differs from wild type by only one or a small number of

chromosomes. The deletion of one or more chromosome form a single chromosomal set in an individual or organism is referred as **Hypoploidy** (2n-1, 2n-2, 2n-3, etc) where as The addition of one or more chromosome in a single chromosomal set of an individual or organism is referred as **Hyperploidy** (2n+1, 2n+2, 2n+3, etc).