Chromosomal Mutation-II
(Cytogenetics : Changes in Chromosomes Number)

Each species has a characteristic number of chromosomes in the nuclei of its gametes and somatic cells. The gametic chromosome number constitutes a basic set of chromosomes called **genome**. A gamete cell contains single genome and is called **haploid**. When haploid gametes of both sexes (male and female) unite in the process of fertilization a**diploid** zygote with two genomes is formed.

However, sometimes irregularities occur in nuclear division and causes Changes in number of whole chromosomes is called **heteroploidy** Heteroploidy may involve entire sets of chromosomes **(euploidy**), or loss or addition of single whole chromosomes (**aneuploidy**). ss or addition of single whole chromosomes (**aneuploidy**). Each may produce phenotypic changes.

A. EUPLOIDY Changes in complete sets of chromosomes **:**

1- Monoploid. One set of chromosomes (1N) is characteristically found in the nuclei of some lower organisms such as fungi. Monoploids in higher organisms are usually smaller and less vigorous than the normal diploids. Few monoploid animals survive. A notable exception exists in male bees and wasps. Monoploid plants are known but are usually sterile. monoploidy is common in plants and rare in animals.

2- Triploid : Three sets of chromosomes (3N) can originate by the union of a monoploid gamete (1N) with a diploid gamete (2N). The extra set of chromosomes of the triploid is distributed in various combinations to the germ cells, resulting in genetically unbalanced gametes. Because of the sterility that characterizes triploids, they are not commonly found in natural populations.

3- Tetraploid : Four sets of chromosomes (4N) can arise in body cells by the somatic doubling of the chromosome number. Doubling is accomplished either spontaneously or it can be induced in high frequency by exposure to chemicals such as the alkaloid colchicine. Tetraploids are also produced by the union of unreduced diploid (2rt) gametes.

Colchicine: is a drug and its aqueous solution is found to prevent the formation and organization of spindle fibres, so the metaphase chromosomes of the affected cells (called **C-metaphase** or **colchicine metaphase**) do not move to a metaphase plate and remain scattered in the cytoplasm. Even the process of cytokinesis is prevented by colchicine and with duplications of chromosomes the number goes on increasing. As colchicine interferes with spindle formation, its effects are limited to dividing and meristematic cells.

Phenotypic Effects of Polyploidy

The increase in the genome's size beyond the diploid level is often caused following detectable phenotypic characteristics in a polyploid organism :

(i) Morphological effect of polyploidy. The polyploidy is invariably related with **gigantism**.

(ii) Physiological effect of polyploidy. The ascorbic acid content has been reported to be higher in tetraploid cabbages and tomatoes than in corresponding diploids.

(iii) Effect on fertility of polyploidy. The most important effect of polyploidy is that it reduces the fertility of polyploid plants in variable degrees.

(iv) Evolution through polyploidy. Interspecific hybridization combined with polyploidy offers a mechanism whereby new species may arise suddenly in natural populations.

Polyploidy in humans have been found in liver cells and cancer cells. In them polyploidy is whether complete or as a mosaic, it leads to gross abnormalities and death.

2. Aneuploidy:

Variations in chromosome number may occur that do not involve whole sets of chromosomes, but only parts of a set.

1- Monosomic. Diploid organisms that are missing one chromosome of a single pair are monosomics with the genomic formula 2n - I. The single chromosome without a pairing partner may go to either pole during meiosis, but more frequently will lag at anaphase and fails to be included in either nucleus. In animals, loss of one whole chromosome often results in genetic unbalance. which is manifested by high mortality or reduced fertility.

2- Trisomic. Diploids which have one extra chromosome are represented by the chromosomal formula In N + 1 . One of the pairs of chromosomes has an extra member, so that a trivalent structure may be formed during meiotic prophase. If 2 chromosomes of the trivalent go to one pole and the third goes to the opposite pole, then gametes will be in + 1.

Trisomy can produce different phenotypes, depending upon which chromosome of the complement is present in triplicate. In humans, the presence of one small extra chromosome (autosome 21) has a very deleterious effect resulting in Down syndrome, formerly called "mongolism".

3- Tetrasotnic. When one chromosome of an otherwise diploid organism is present in quadruplicate, this is expressed as 2« + 2. A quadrivalent may form for this particular chromosome during meiosis which then has the same problem as that discussed for autotetraploids.

4- Nullisomy. An organism which has lost a chromosome pair is a nullosomic. The nullosomic organism has the genomic formula (2n–2). A nullosomic diploid often does not survive, however, a nullosomic polyploid (e.g., hexaploid wheat, 6x–2) may survive but exhibit reduced vigour and fertility.