CSIR NET Life Science Unit 8

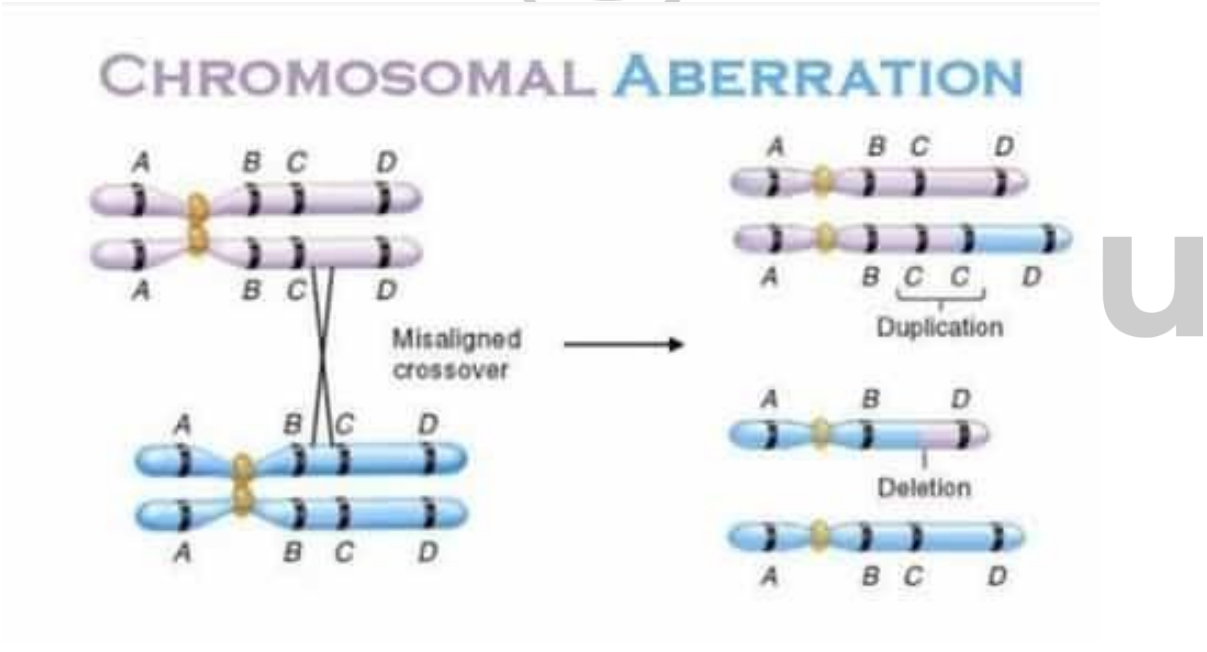
Structural and numerical alterations of chromosomes

Introduction

Chromosomes are thread-like structures found within the nucleus of both animal and plant cells. Protein and a single molecule of deoxyribonucleic acid make up each chromosome (DNA). DNA is passed along from parents to children and carries the precise instructions that distinguish each living being.

There are 46 chromosomes in the human body, with 23 pairs of chromosomes in almost every cell. Our mother gives us half of our chromosomes, and our father gives us the other half. Autosomes are the initial 22 pairs of chromosomes. The sex chromosomes, X and Y, make up the 23rd pair. Males have one X and one Y chromosome in each cell, while females have two X chromosomes in each cell.

Chromosome aberration: Abnormal structure or number of chromosomes; includes deficiency, duplication, inversion, translocation, aneuploidy, polyploidy, or any other change from the normal pattern.



Numerical Alterations:

Numerical chromosomal aberration refers to changes in the number of chromosomes in the diploid set. It is also known as ploidy. Euploidy and aneuploidy are the two kinds of ploidy.

Depending on the specific anomaly, chromosomal abnormalities can have a variety of impacts. Down syndrome, for example, is caused by an extra copy of chromosome 21. (trisomy 21). Miscarriage, illness, and growth and development issues can all be caused by chromosomal abnormalities.

Aneuploidy, or an aberrant chromosome number caused by an extra or missing chromosome, is the most frequent type of chromosomal abnormality. The majority of people with aneuploidy have trisomy (three copies of a chromosome) rather than monosomy (one copy of a chromosome). The most well-known example of chromosomal aneuploidy is Down syndrome.

The other examples are trisomy 18 (Edward's syndrome); trisomy 13; 45, X (Turner syndrome); 47, XXY (Klinefelter syndrome); 47, XYY; and 47, XXX.

- **Euploidy**

Euploidy is a change in the number of chromosomes caused by the increase or decrease of a full set of chromosomes increasing or decreasing. Euploidy is in three different forms: monoploidy, diploidy, and polyploidy.

- **Diploidy**

Somatic cells in most plants and animals have two sets of chromosomes. The joining of two gametes during fertilisation results in diploidy.

- **Polyploidy**

Polyploidy occurs when one or more sets of chromosomes are added to the diploid set. It's prevalent in plants but uncommon in animals. There are two types of polyploidy: autopolyploidy and allopolyploidy.

- **Autopolyploidy**

Autopolyploidy occurs when an organism's genome is added by one or more haploid copies. Apple is an autotetraploid, while watermelon, grapes, and banana are autotriploids.

- **Allopolyploidy**

Allopolyploidy is defined as an increase in one or more haploid sets of chromosomes from two separate species.

- **Aneuploidy**

Aneuploidy is a change in the diploid set of organisms that involves one or two chromosomes. Hypoploidy and hyperdiploidy are the two forms.

Structural Alterations

The structure of a chromosome can be altered in a variety of ways.

- **Deletions:** A mutation (a genetic aberration) in which a section of a chromosome or a sequence of DNA is deleted during DNA replication is known as deletion (also known as gene deletion, deficiency, or deletion mutation). From a single base to a whole chromosome, any number of nucleotides can be removed.

- **Duplications:** A type of mutation that involves the production of one or more copies of any component of DNA, such as a gene or even an entire chromosome.

- **Translocations:** A portion of a chromosomal segment gets transferred to another chromosome. Translocation can be divided into two categories. Segments from two separate chromosomes have been swapped in a reciprocal translocation. A complete chromosome has connected to another at the centromere in a Robertsonian translocation.

- **Inversions:** An inversion is a type of chromosome rearrangement in which a chromosome segment is reversed end to end. When a single chromosome breaks and rearranges inside itself, it is called an inversion. Paracentric and pericentric inversions are the two forms of inversions.

- **Paracentric inversions:** The centromere is not included, and both breaks occur in one chromosomal arm. The centromere is included in pericentric inversions, and each arm has a breakpoint.