Lesson 08 : Chromosomal Mutations

Chromosomal mutations are major alterations affecting the structure or number of chromosomes in a cell. Unlike point genetic mutations that impact one or more nucleotides within a gene, chromosomal mutations involve large DNA segments or even entire chromosomes. They can have dramatic effects on the phenotype and are often the cause of severe genetic diseases.

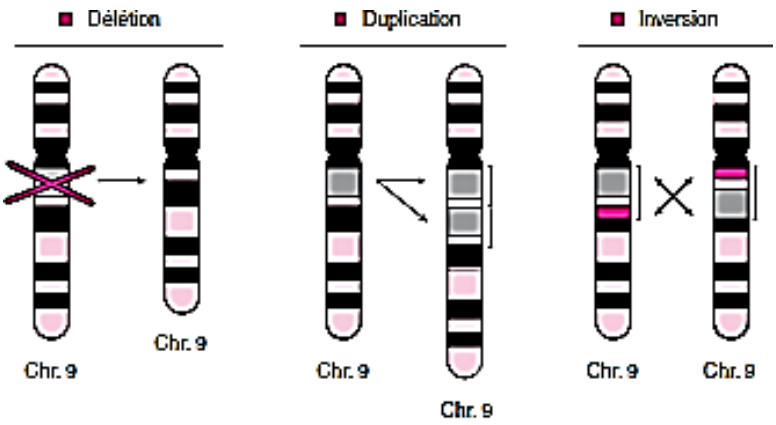
I. Types of Chromosomal Mutations

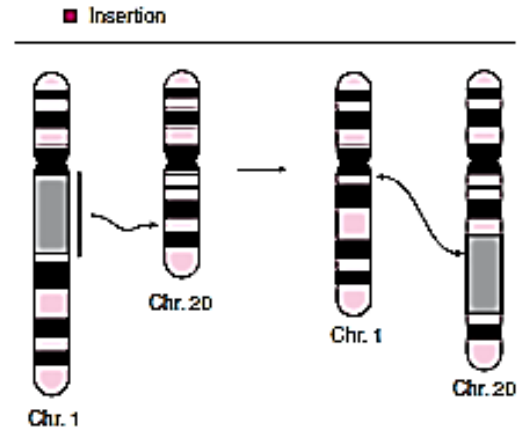
Chromosomal mutations are divided into two main categories: structural chromosomal mutations and numerical chromosomal mutations.

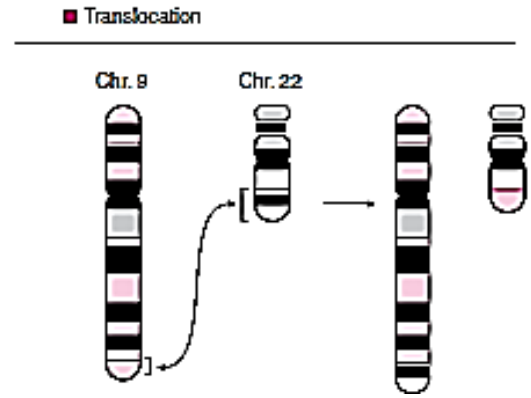
1. Structural Chromosomal Mutations

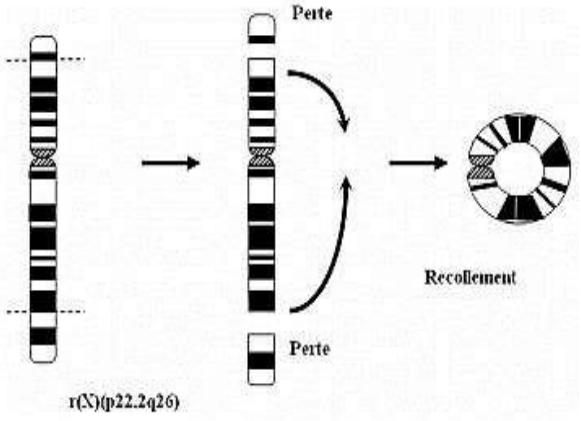
These mutations alter the internal structure of one or more chromosomes. They include:

- **Deletions**
  Loss of a chromosome segment. This can lead to the loss of several genes, potentially causing lethality or severe diseases such as Cri-du-chat syndrome.
- **Duplications**
  A chromosome segment is duplicated, resulting in an extra copy of genes in that region. This may lead to gene overexpression and disorders.
- **Inversions**
  A chromosome segment is flipped and reintegrated into the chromosome. This can disrupt genes at the breakpoints or within the inversion, affecting their function.
- **Translocations**
  A chromosome segment is transferred to another non-homologous chromosome. There are two main types:
  - **Reciprocal translocations:** Exchange of segments between two non-homologous chromosomes.
  - **Robertsonian translocations:** Fusion of two acrocentric chromosomes (chromosomes with very short p arms) at the centromere.









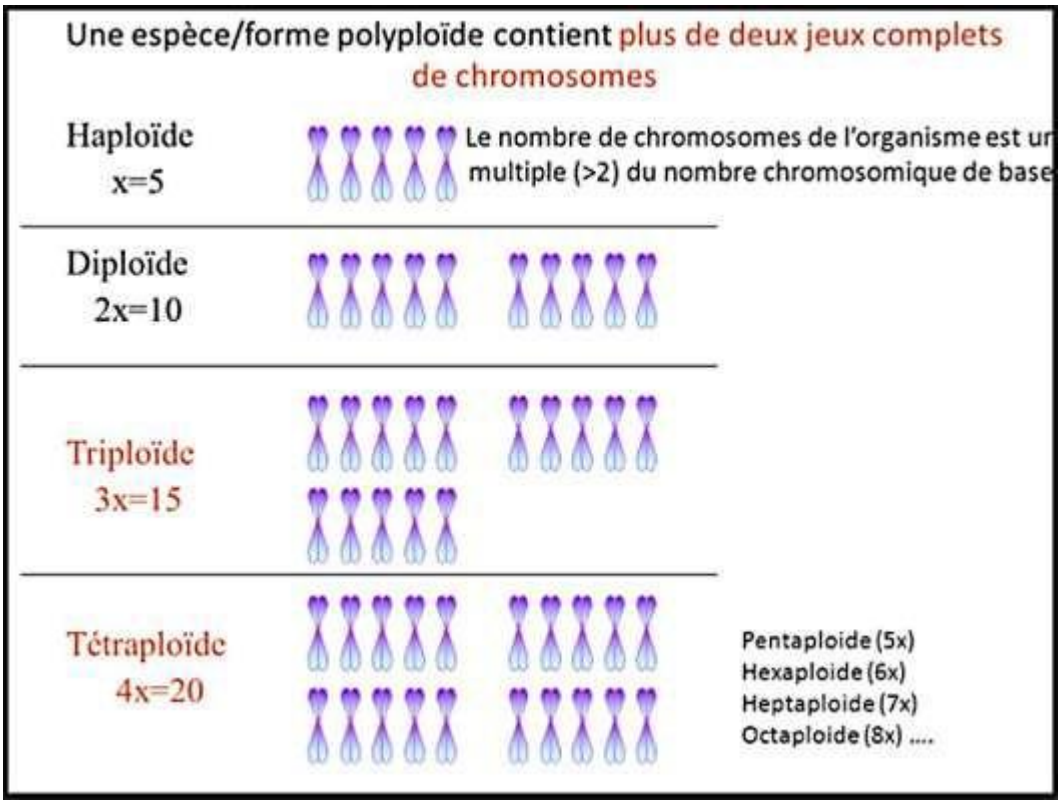
2. Numerical Chromosomal Mutations

These mutations affect the total number of chromosomes in an organism. They include:

- **Euploidy**

  Numerical variations that affect the entire chromosome set equally (each chromosome undergoes the same numerical change). Two types of euploidy are:

- **Monoploidy (Haploidy):** The basic chromosomal number is represented once (n).
- **Polyploidy:** The basic chromosomal number is represented multiple times. One-third of angiosperms (flowering plants) have more than two sets of chromosomes. Types include:
  - **Diploidy:** Two sets of chromosomes (2n), common in most animals and complex multicellular organisms.
  - **Triploidy:** Three sets of chromosomes (3n).
  - **Tetraploidy:** Four sets of chromosomes (4n).

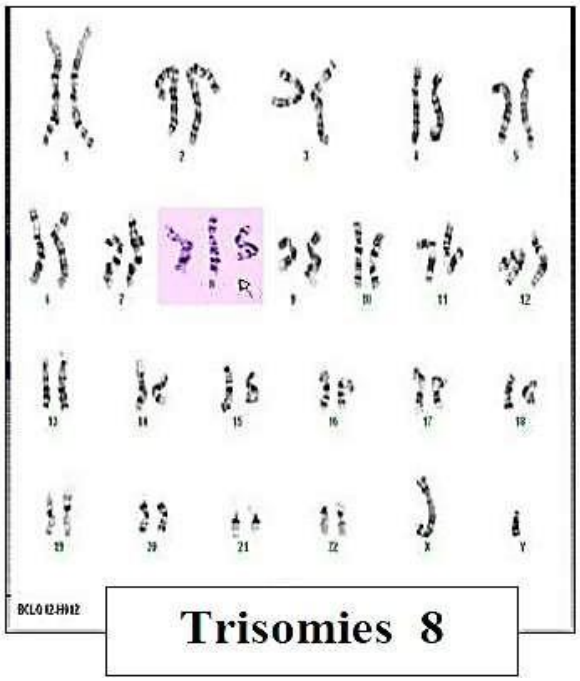


- **Aneuploidy**

  Numerical chromosomal variations that do not equally affect all chromosomes. Types include:
  - Monosomic: 2n - 1
  - Trisomic: 2n + 1
  - Nullisomic: 2n - 2, usually lethal in diploids.
  - Tetrasomic: 2n + 2

- **Polyploidies**

  The presence of additional chromosome sets. This phenomenon is more common in plants than in animals.



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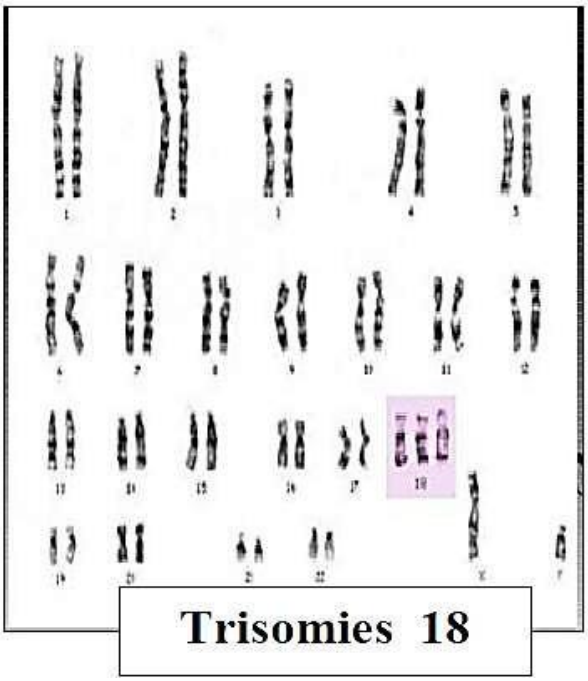

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Mehanisms of Chromosomal Mutations

Chromosomal mutations may arise through various mechanisms, including:

- **Errors during meiosis**
  - Chromosomal nondisjunction: During meiosis, homologous chromosomes or sister chromatids fail to separate properly, leading to abnormal chromosome distribution in daughter cells.
- **Errors during DNA replication**

  Mistakes during DNA replication can lead to deletions, duplications, or inversions.
- **Abnormal reciprocal exchanges**

  During meiosis, segment exchanges between non-homologous chromosomes may occur, resulting in translocations.
- **Exposure to mutagenic agents**

  Physical (radiation) or chemical agents can cause chromosome breaks, leading to structural mutations

Consequences of Chromosomal Mutations

The effects of chromosomal mutations depend on the type of mutation and the genes affected. Potential consequences include:

- **Major genetic disruptions**
  Loss or duplication of essential genes may result in developmental disorders, congenital malformations, or severe diseases such as certain cancers (e.g., chronic myeloid leukemia caused by a translocation between chromosomes 9 and 22, creating the Philadelphia chromosome).
- **Fertility issues**
  Translocations may cause fertility problems due to the inability of altered chromosomes to properly pair during meiosis.
- **Chromosomal syndromes**
  Chromosomal mutations underlie many genetic syndromes, including:
  - **Down syndrome (Trisomy 21):** Presence of an extra chromosome 21.
  - **Klinefelter syndrome (XXY):** Presence of an extra X chromosome in males.
  - **Turner syndrome (XO):** Absence of one X chromosome in females.
- **Cancers**
  Some chromosomal mutations are directly associated with cancer development. For instance, the translocation between chromosomes 8 and 14 is linked to Burkitt lymphoma.