Mutation

In genetics, a mutation is a permanent change of the nucleotide sequence of the genome of an organism, virus, or extra chromosomal genetic element.

Mutations result from unrepaired damage to DNA or to RNA genomes (typically caused by radiation or chemical mutagens), errors in the process of replication, or from the insertion or deletion of segments of DNA by mobile genetic elements.

Mutations may or may not produce discernible changes in the observable characteristics (phenotype) of an organism.

Mutations play a part in both normal and abnormal biological processes including: evolution, cancer, and the development of the immune system.

Mutation can result in several different types of change in sequences. Mutations in genes can either have no effect, alter the product of a gene, or prevent the gene from functioning properly or completely.

Description

Mutations can involve the duplication of large sections of DNA, usually through genetic recombination. These duplications are a major source of raw material for evolving new genes, with tens to hundreds of genes duplicated in animal genomes every million years.

Novel genes are produced by several methods, commonly through the duplication and mutation of an ancestral gene, or by recombining parts of different genes to form new combinations with new functions.

Changes in chromosome number may involve even larger mutations, where segments of the DNA within chromosomes break and then rearrange.

Nonlethal mutations accumulate within the gene pool and increase the amount of genetic variation. The abundance of some genetic changes within the gene pool can be reduced by natural selection, while other "more favorable" mutations may accumulate and result in adaptive changes.

For example, a butterfly may produce offspring with new mutations. The majority of these mutations will have no effect; but one might change the color of one of the butterfly's offspring, making it harder (or easier) for predators to see. If this color change is advantageous, the chance of this butterfly's surviving and producing its own offspring are a little better, and over time the number of butterflies with this mutation may form a larger percentage of the population.

Neutral mutations are defined as mutations whose effects do not influence the fitness of an individual. These can accumulate over time due to genetic drift. It is believed that the overwhelming majority of mutations have no significant effect on an organism's fitness. Also, DNA repair mechanisms are able to mend most changes before they become permanent mutations, and many organisms have mechanisms for eliminating otherwise-permanently mutated somatic cells.

Induced mutation

Induced mutations on the molecular level can be caused by:-

- Chemicals
    - Hydroxylamine NH2OH
    - Base analogs
    - Alkylating agents (e.g., *N*-ethyl-*N*-nitrosourea)
    - Agents that form DNA adducts (e.g., ochratoxin A metabolites)
    - DNA intercalating agents (e.g., ethidium bromide)
    - DNA crosslinkers

    - Oxidative damage
- Nitrous acid converts amine groups on A and C
- Radiation
    - Ultraviolet radiation (non ionizing 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. UV radiation, in particular longer-wave UVA, can also cause oxidative damage to DNA.

Mutations

What Are Mutations?

- Changes in the nucleotide sequence of DNA
- May occur in somatic cells (aren't passed to offspring)
- May occur in gametes (eggs & sperm) and be passed to offspring

Are Mutations Helpful or Harmful?

- Mutations happen regularly
- Almost all mutations are neutral
- Chemicals & UV radiation cause mutations
- Many mutations are repaired by enzymes
- Some type of skin cancers and leukemia result from somatic mutations
- Some mutations may improve an organism's survival (beneficial)

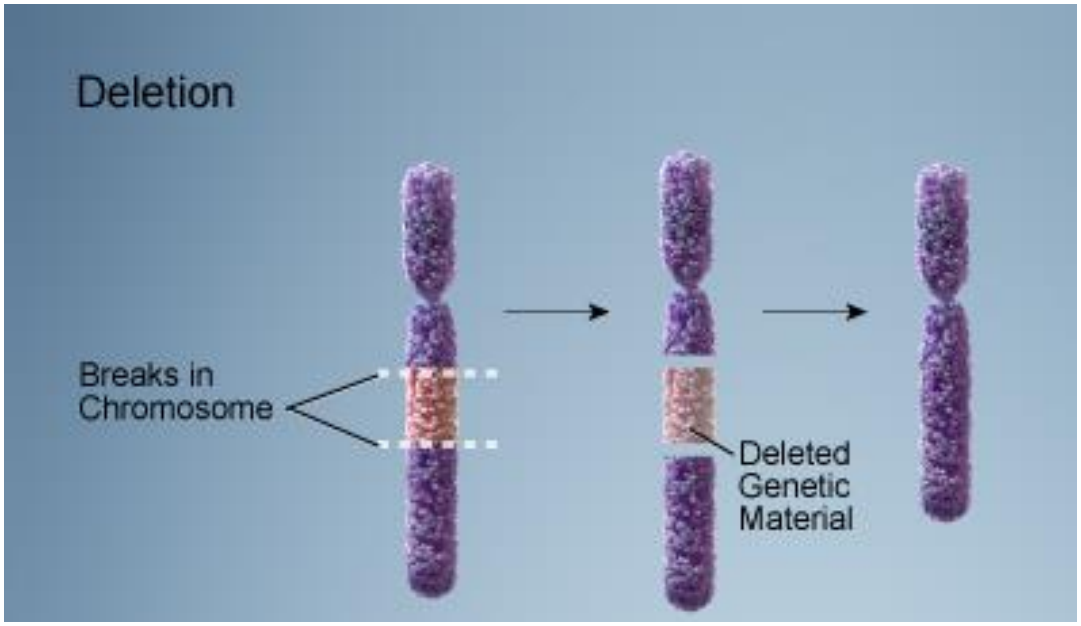
Types of Mutations

Chromosome Mutations

- May Involve:
  - **Changing the structure** of a chromosome
  - The **loss or gain** of part of a chromosome
- Five types exist:
  - **Deletion** –**Inversion** –**Translocation--Nondisjunction --Duplication**

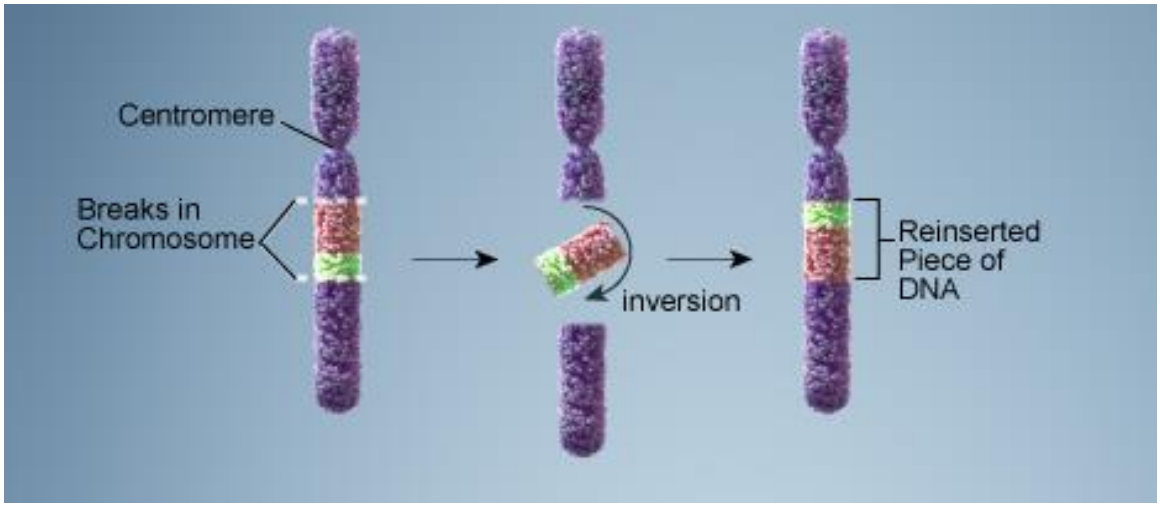
Deletion

- Due to **breakage**
- A **piece** of a chromosome is **lost**



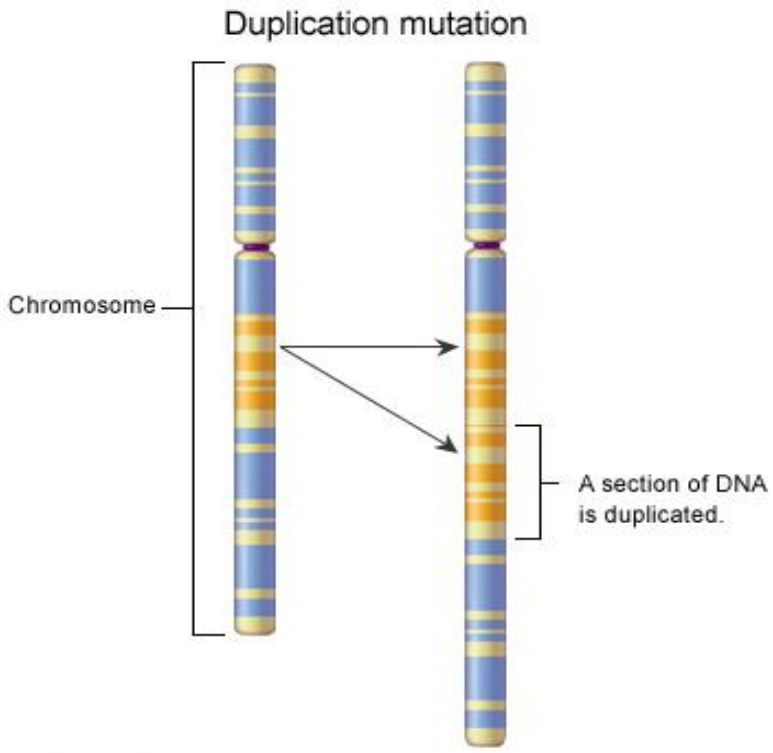
Inversion

- Chromosome segment **breaks off**
- Segment flips around **backwards**
- Segment **reattaches**



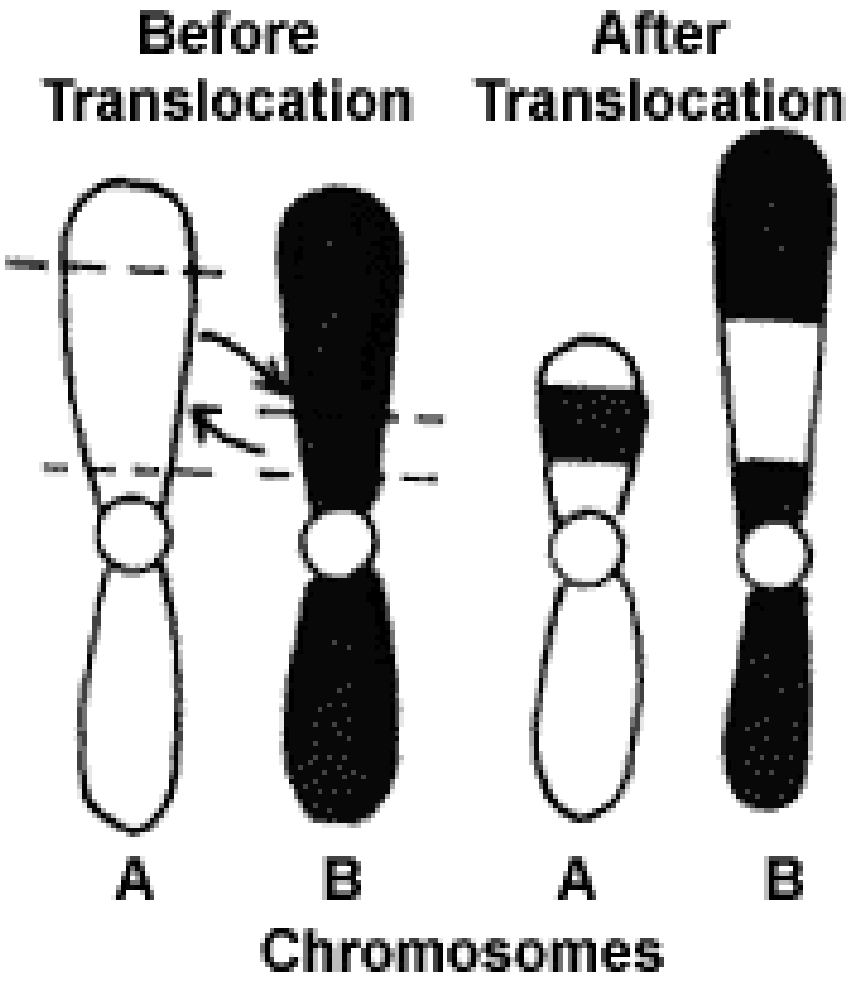
Duplication

- Occurs when a gene **sequence is repeated**



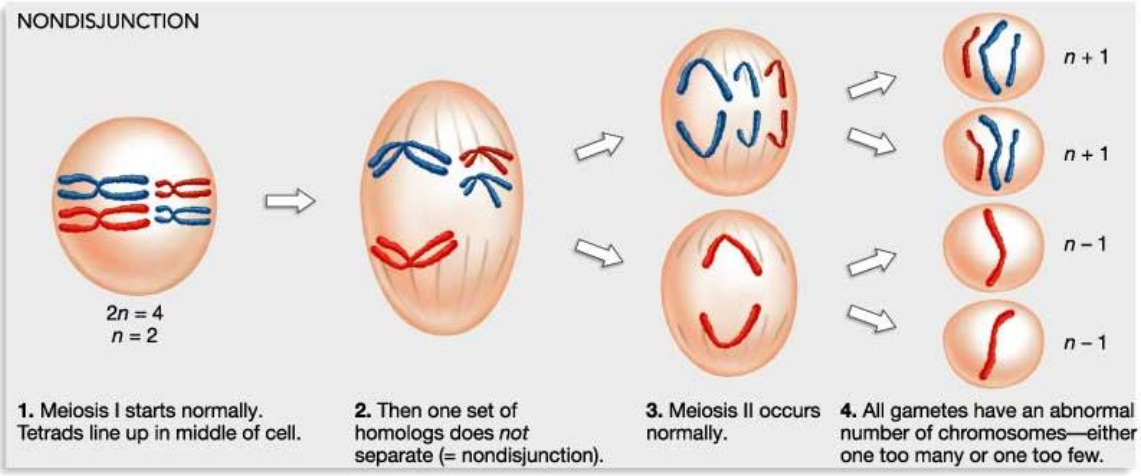
Translocation

- Involves **two chromosomes** that aren't homologous
- **Part** of one chromosome is **transferred to another** chromosomes

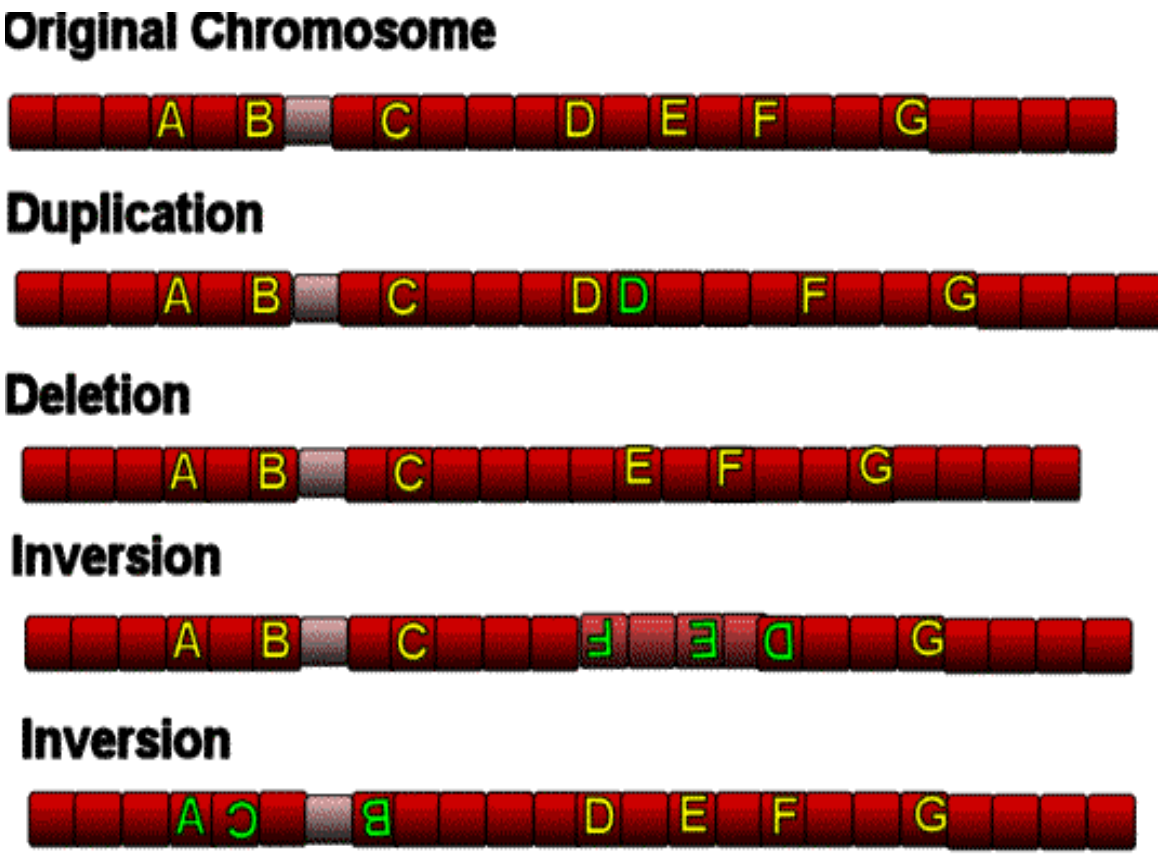


Nondisjunction

- **Failure** of chromosomes **to separate** during meiosis
- Causes gamete to have **too many or too few chromosomes**



Chromosome Mutation Animation



Gene Mutations

- Change in the **nucleotide sequence** of a gene
- May only involve a single nucleotide
- May be due to **copying errors**, **chemicals**, **viruses**, etc.

Types of Gene Mutations

- Include:
  - Point Mutations –Substitutions—Insertions—Deletions--Frameshift

Point Mutation

- Change of a **single** nucleotide
- Includes the deletion, insertion, or substitution of **ONE** nucleotide in a gene

- **Sickle Cell disease** is the result of one nucleotide substitution
- Occurs in the **hemoglobin gene**

Frameshift Mutation

- **Inserting or deleting** one or more nucleotides
- Changes the "**reading frame**" like changing a sentence
- **Proteins** built **incorrectly**
- Original:
    - The fat cat ate the wee rat.
- Frame Shift ("a" added):
    - The fat caa tet hew eer at.

Amino Acid Sequence Changed