

MUTATION

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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

Are Mutations Helpful or Harmful?

- Some type of **skin cancers and leukemia** result from **somatic mutations**
- Some mutations may **improve** an organism's **survival** (beneficial)

Types of Mutations

The background is a dark blue gradient with a subtle pattern of small white dots. On the right side, there are several technical graphics: a large circular gauge with a scale from 0 to 210, a smaller circular gauge with a scale from 0 to 100, and a dashed circular arrow. On the left side, there is a partial circular arrow graphic.

Chromosome Mutations

- May Involve:
 - Changing the structure of a chromosome
 - The **loss or gain** of part of a chromosome

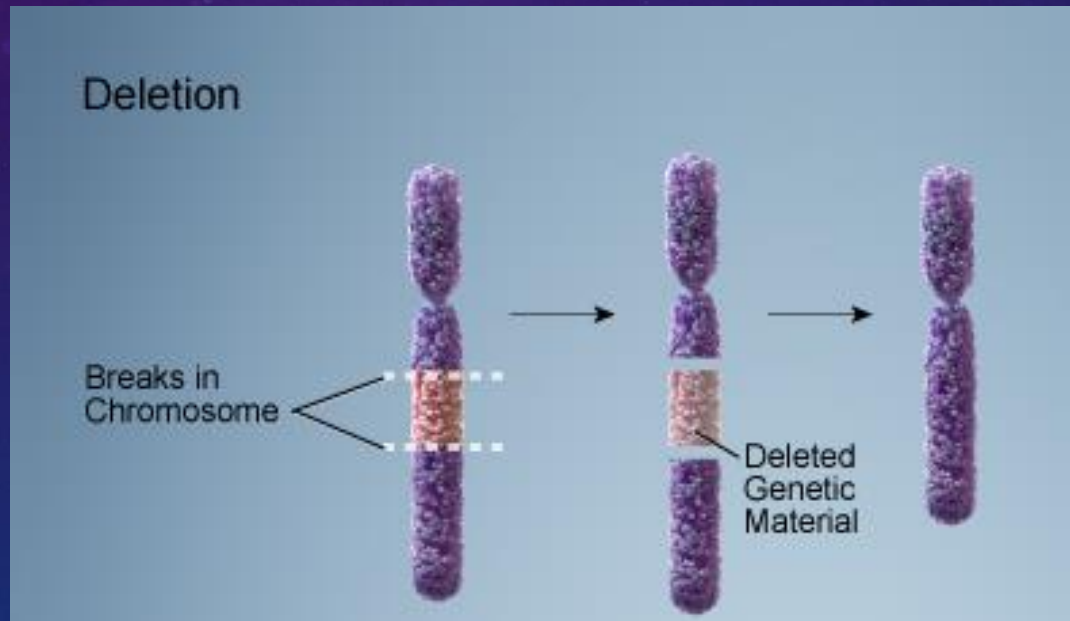


Chromosome Mutations

- Five types exist:
 - Deletion
 - Inversion
 - Translocation
 - Nondisjunction
 - Duplication

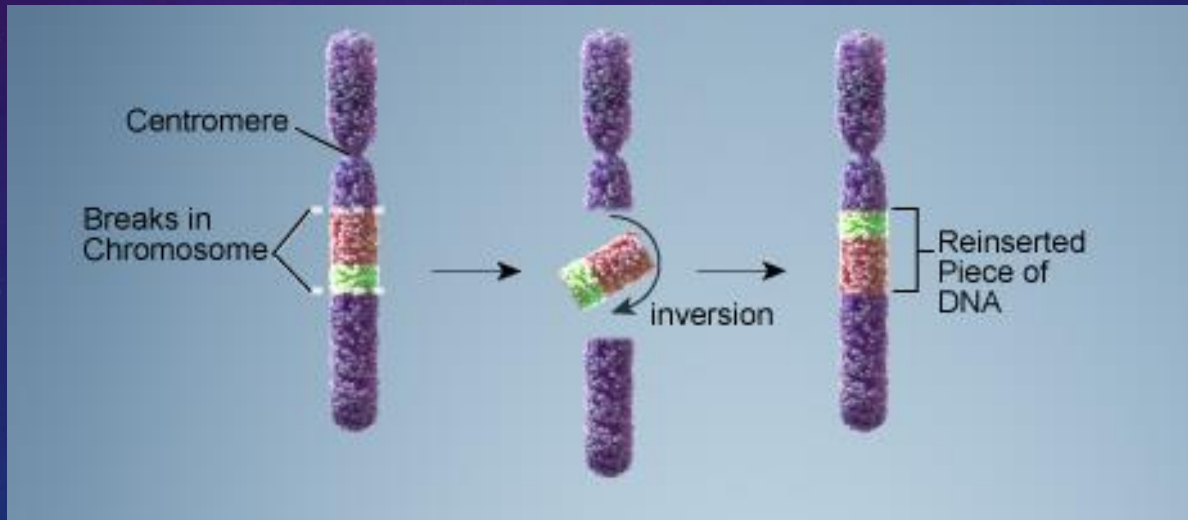
Deletion

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



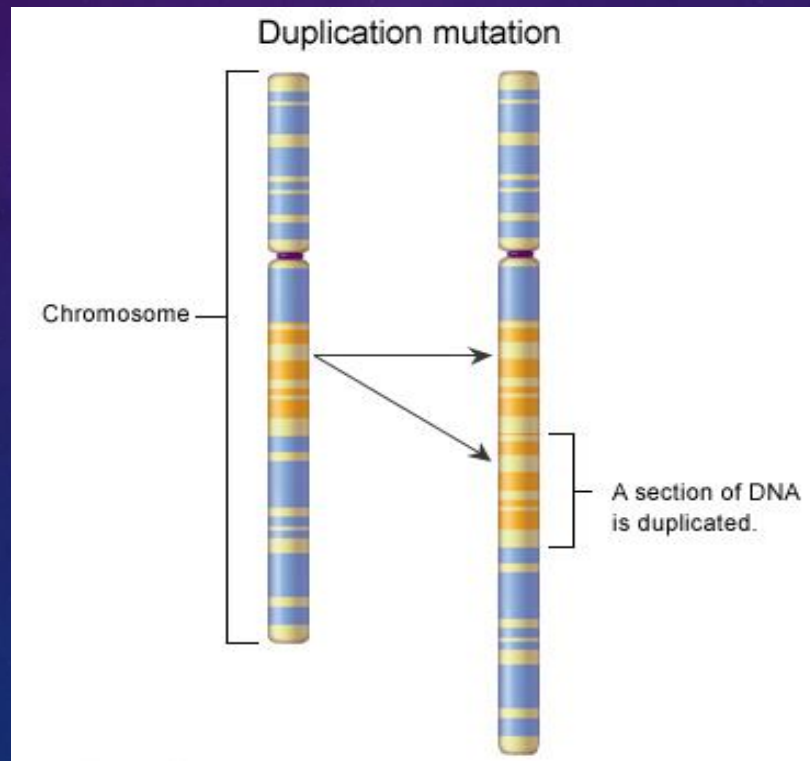
Inversion

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



Duplication

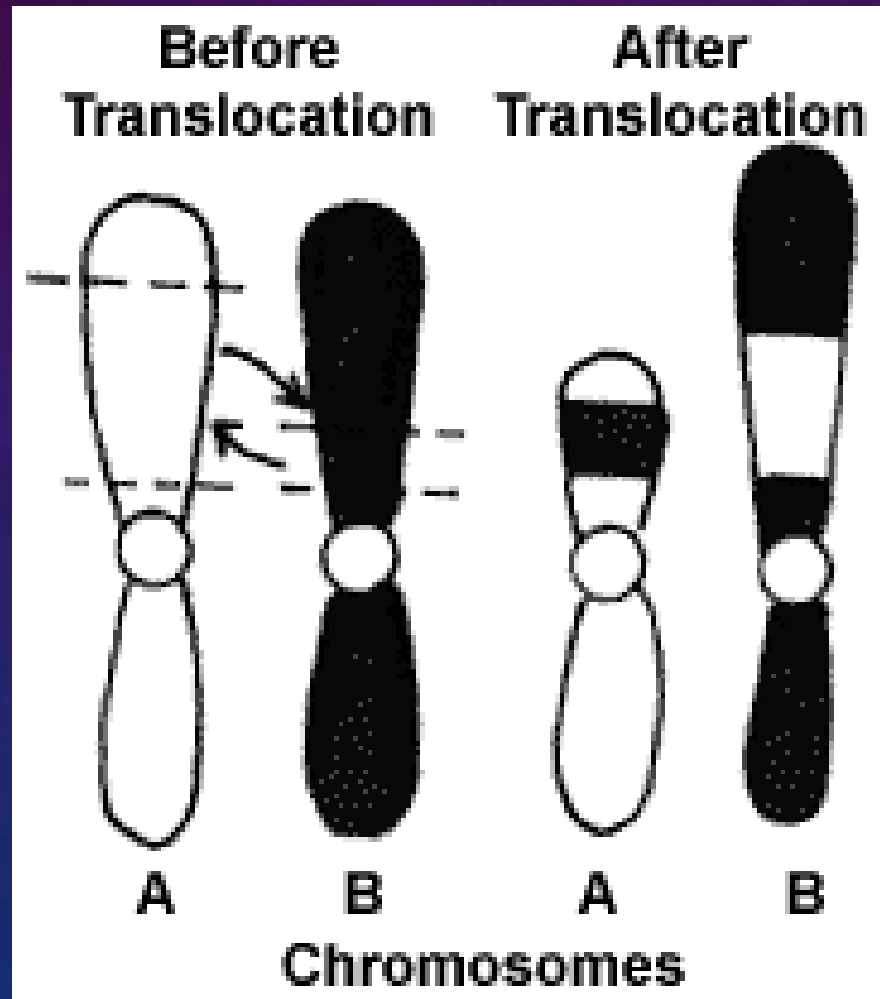
- Occurs when a gene **sequence** is **repeated**



Translocation

- Involves **two chromosomes** that are **NOT** homologous
- **Part** of one chromosome is **transferred to another** chromosome

Translocation



Nondisjunction

- **Failure** of chromosomes to **separate** during meiosis
- Causes gamete to have **too many** or **too few** chromosomes
- Disorders:
 - **Down Syndrome** - three 21st chromosomes
 - **Turner Syndrome** - single X chromosome
 - **Klinefelter's Syndrome** - XXY chromosomes

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

Point Mutation

- **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**

Frameshift Mutation

- Original:
 - The fat cat ate the wee rat.
- Frame Shift (“a” added):
 - The fat caa tet hew eer at.

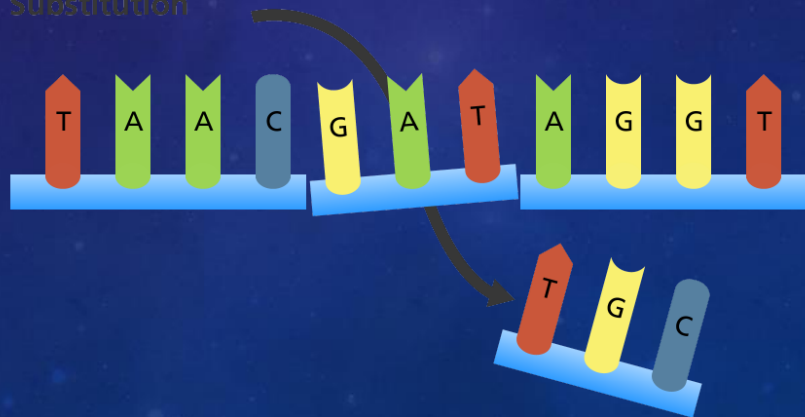
SUBSTITUTION MUTATION

A **substitution** is a **mutation** that exchanges one base for another (i.e., a change in a single "chemical letter" such as switching an A to a G)

Original sequence



Substitution



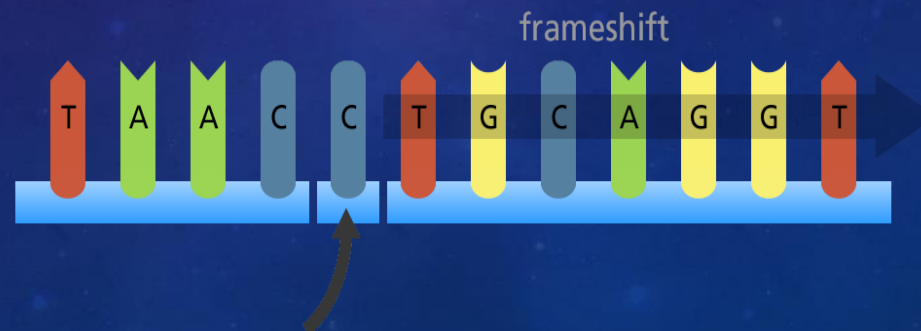
INSERTION MUTATION

- The addition of one or more nucleotide base pairs into a DNA sequence

Original sequence

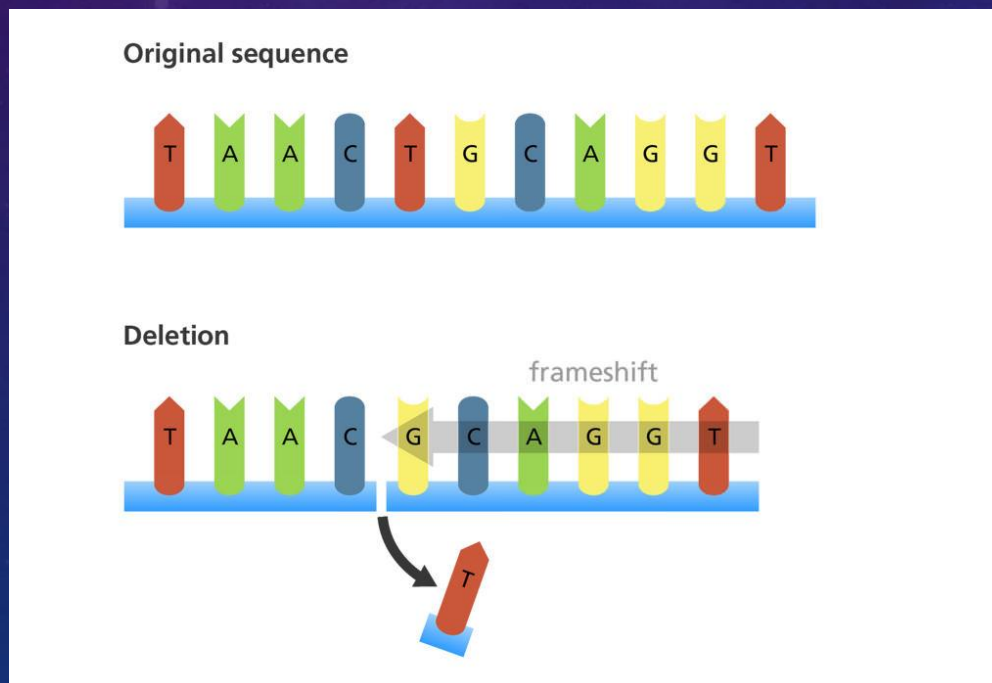


Insertion



DELETION MUTATION

- A part of a chromosome or a sequence of DNA is lost during DNA replication.
- Any number of nucleotides can be deleted, from a single base to an entire piece of chromosome



THANK YOU

