Genetic Mutations

Human Genetics

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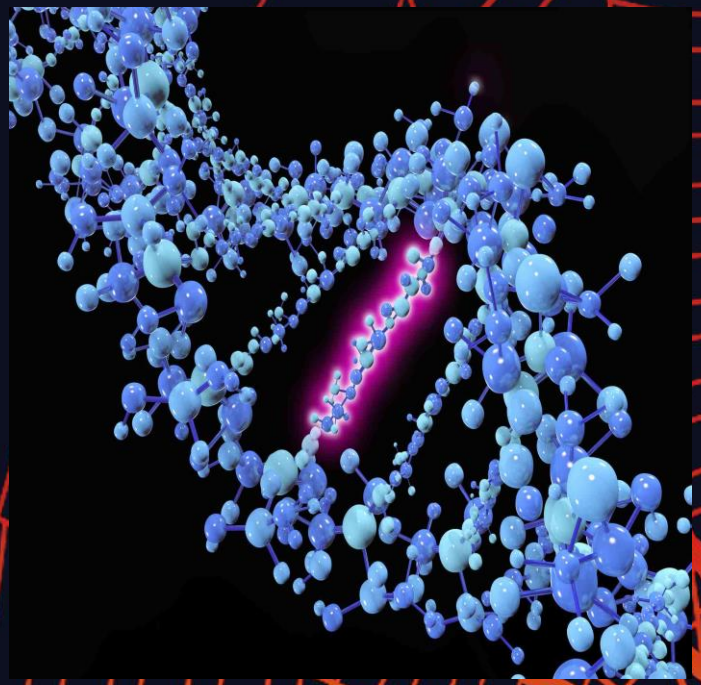
What Are Mutations?

- 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

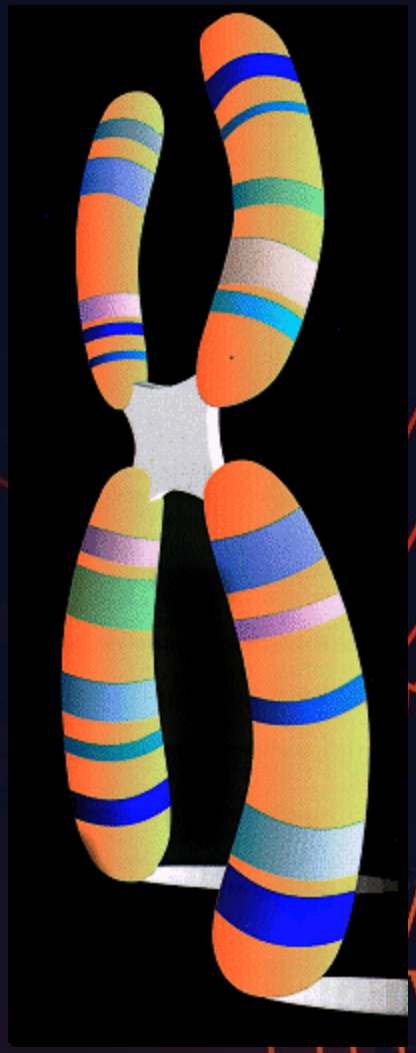
Are Mutations Helpful or Harmful?

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

Types of Mutations

Chromosome Mutations

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

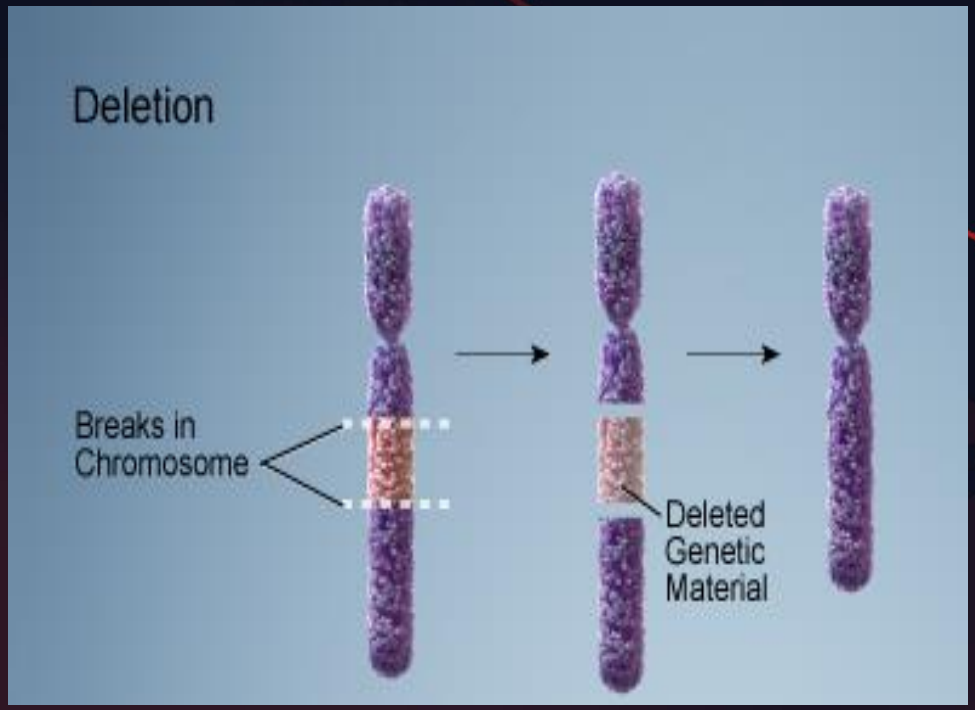


Chromosome Mutations

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

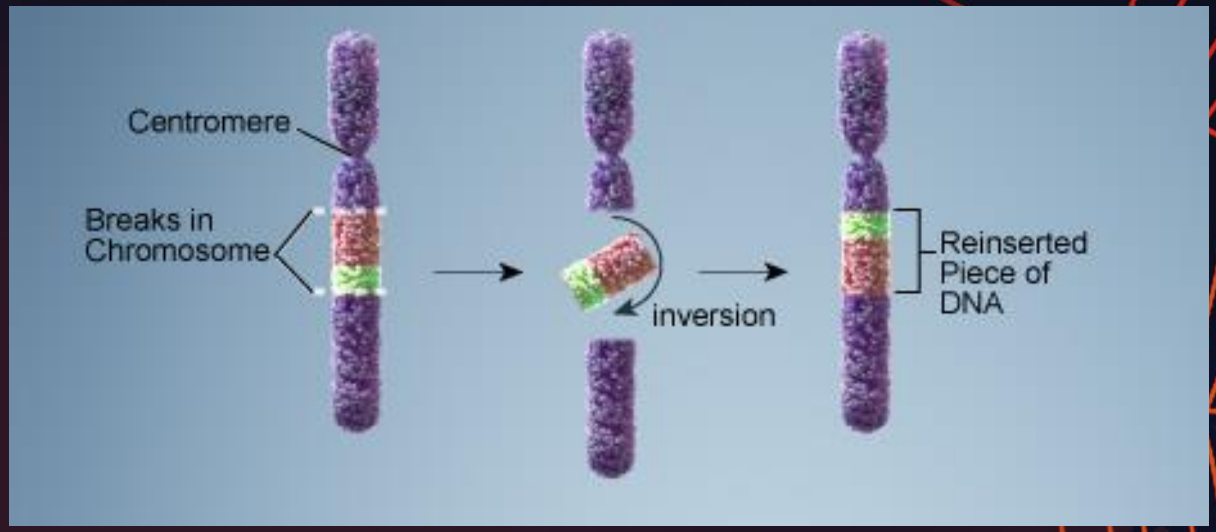
Deletion

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



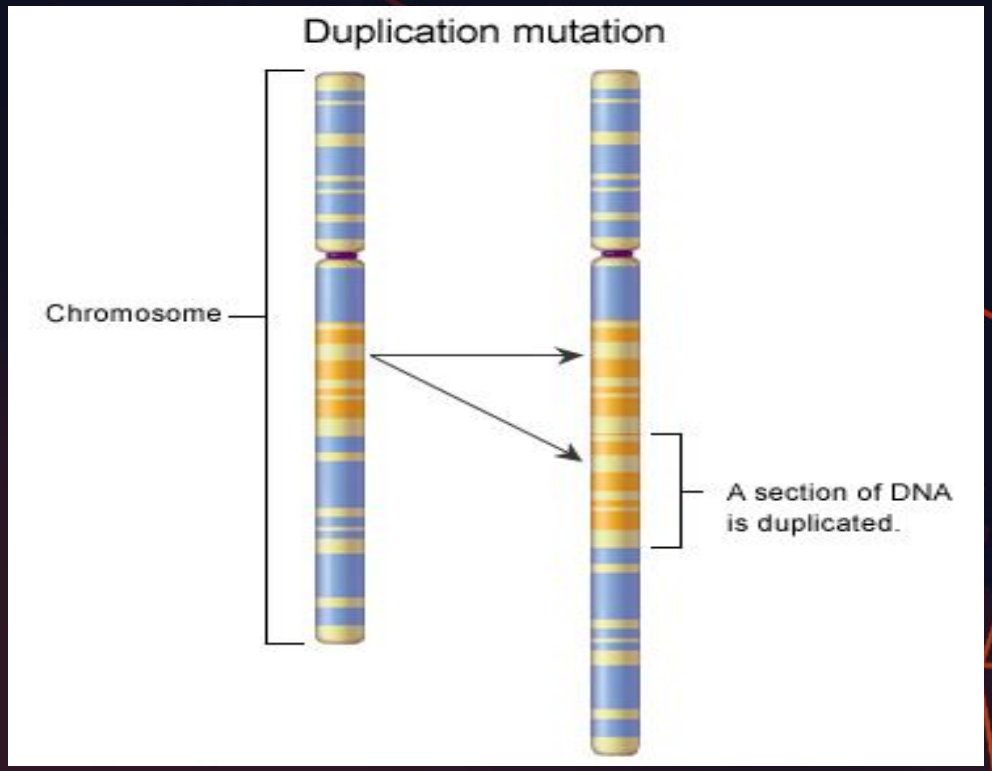
Inversion

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



Duplication

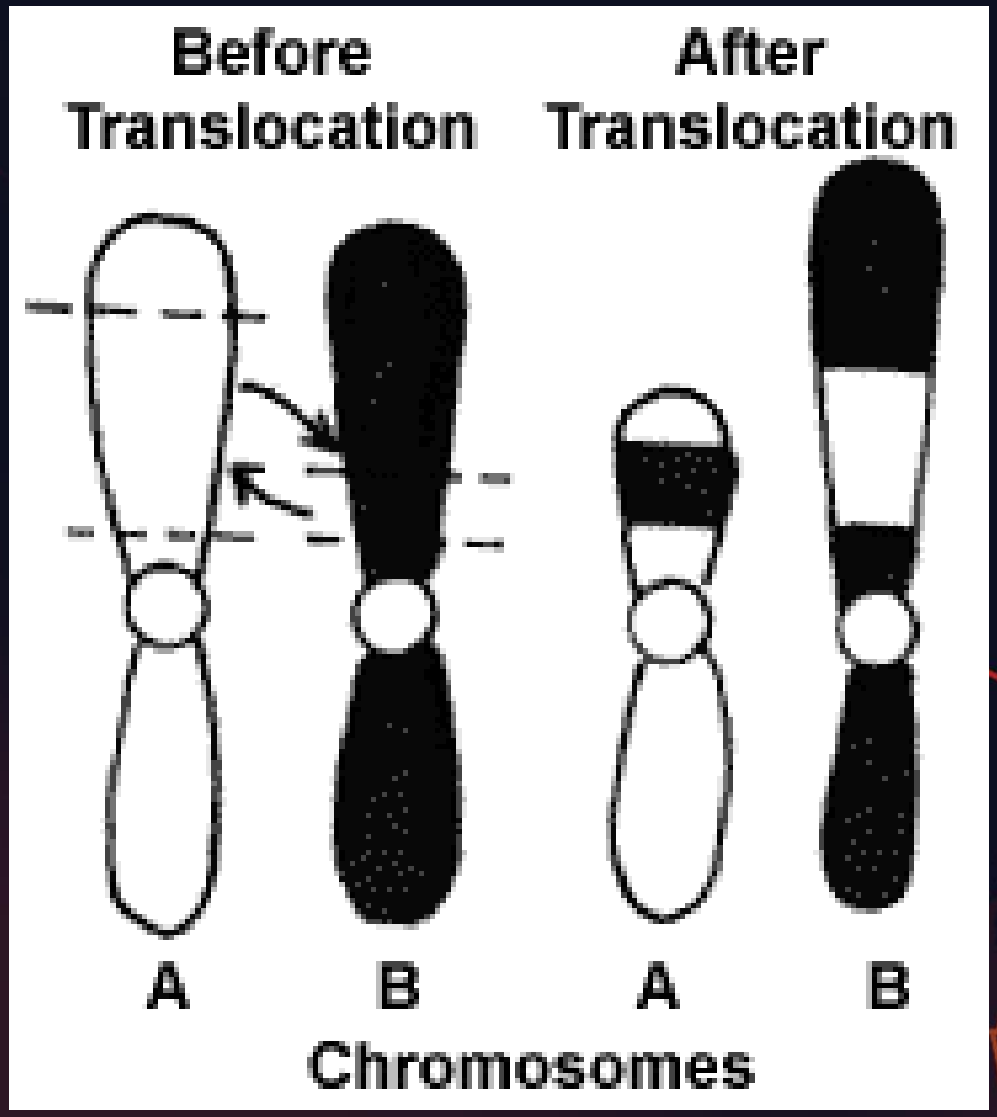
- Occurs when a gene **sequence is repeated**



Translocation

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

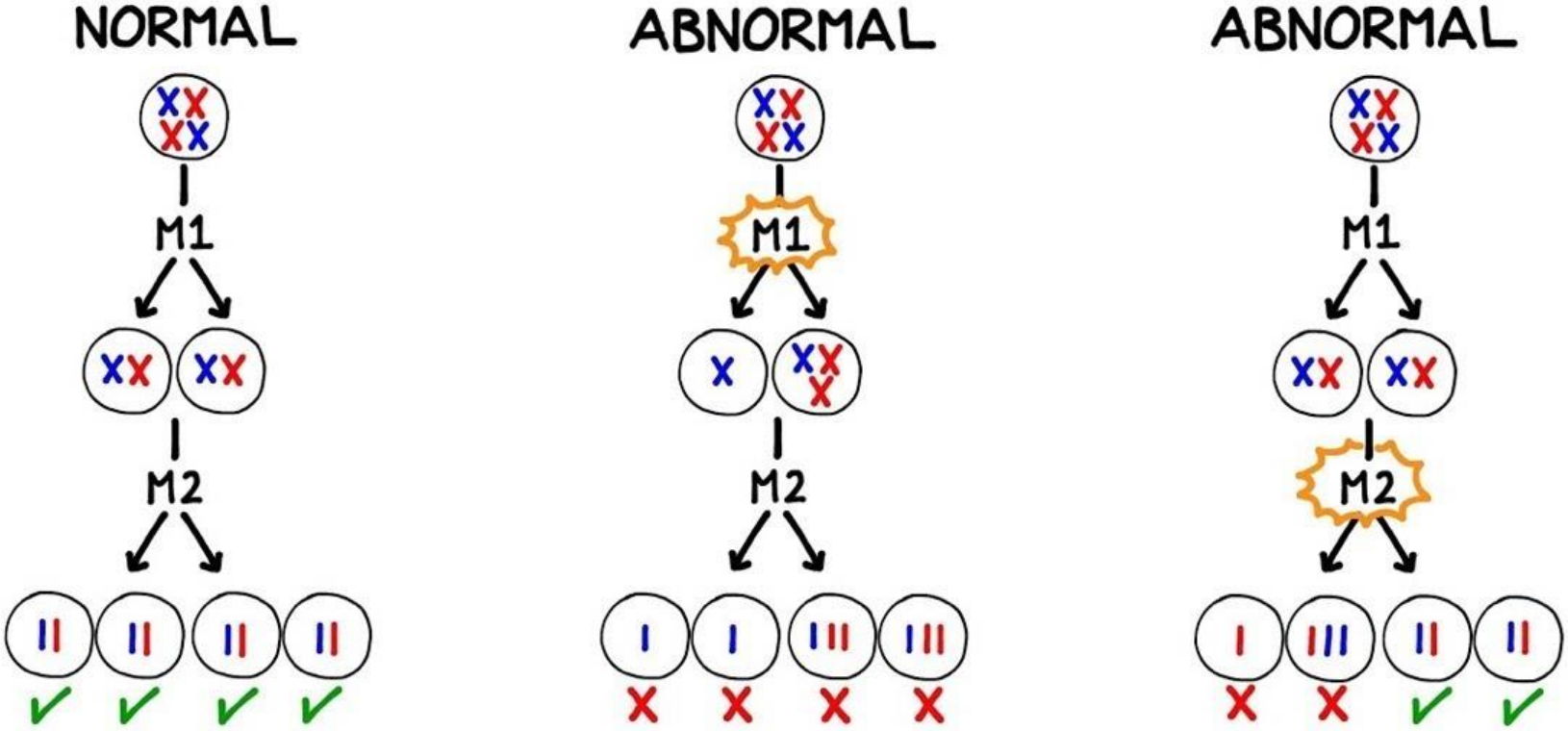
Translocation

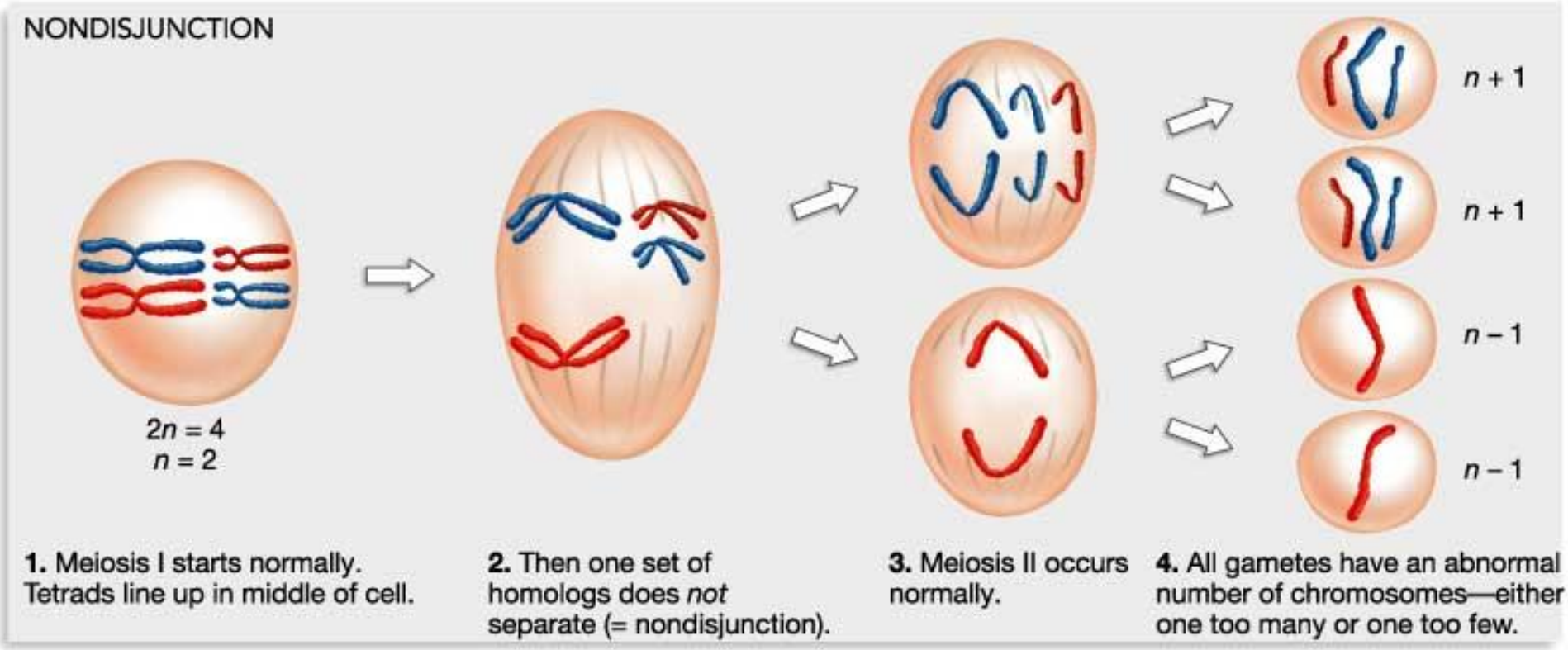


Nondisjunction

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

NON-DISJUNCTION

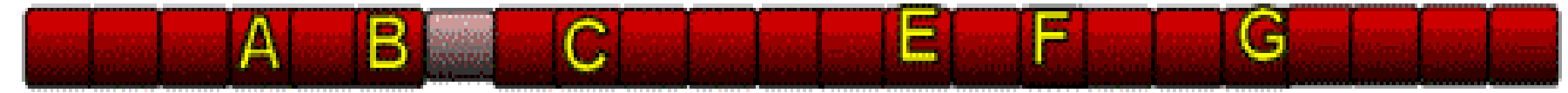



NONDISJUNCTION
2n = 4
n = 2
n + 1
n + 1
n – 1
n – 1
1. Meiosis I starts normally. Tetrads line up in middle of cell.
2. Then one set of homologs does not separate (= nondisjunction).
3. Meiosis II occurs normally.
4. All gametes have an abnormal number of chromosomes—either one too many or one too few.

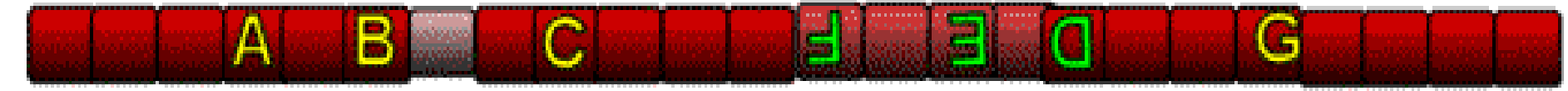
Original Chromosome

Duplication

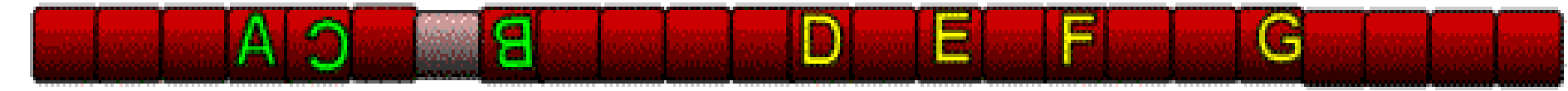
Deletion



Inversion

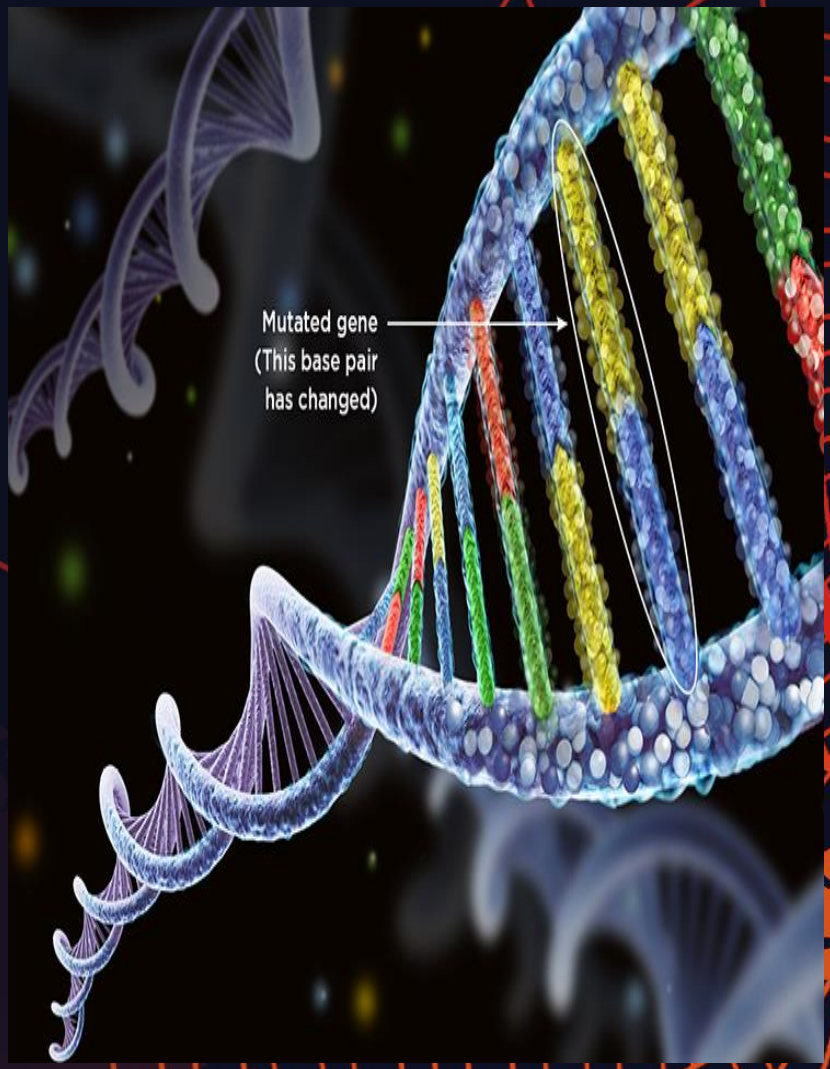


Inversion



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.

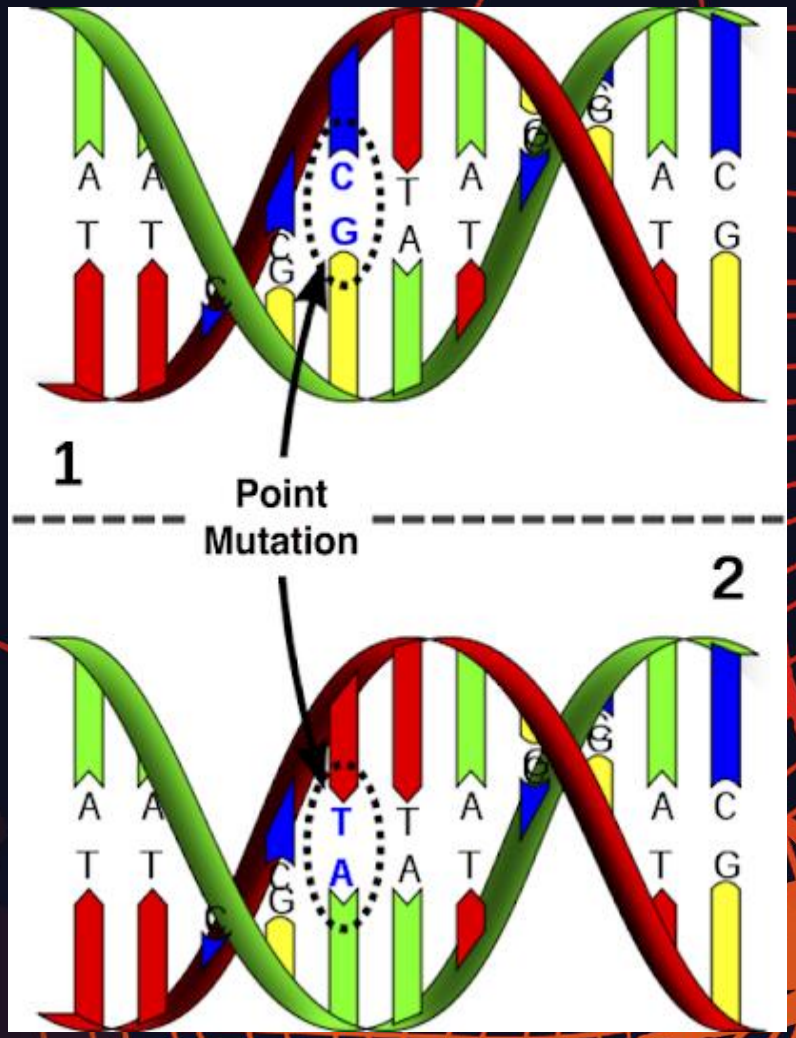


Types of Gene Mutations

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

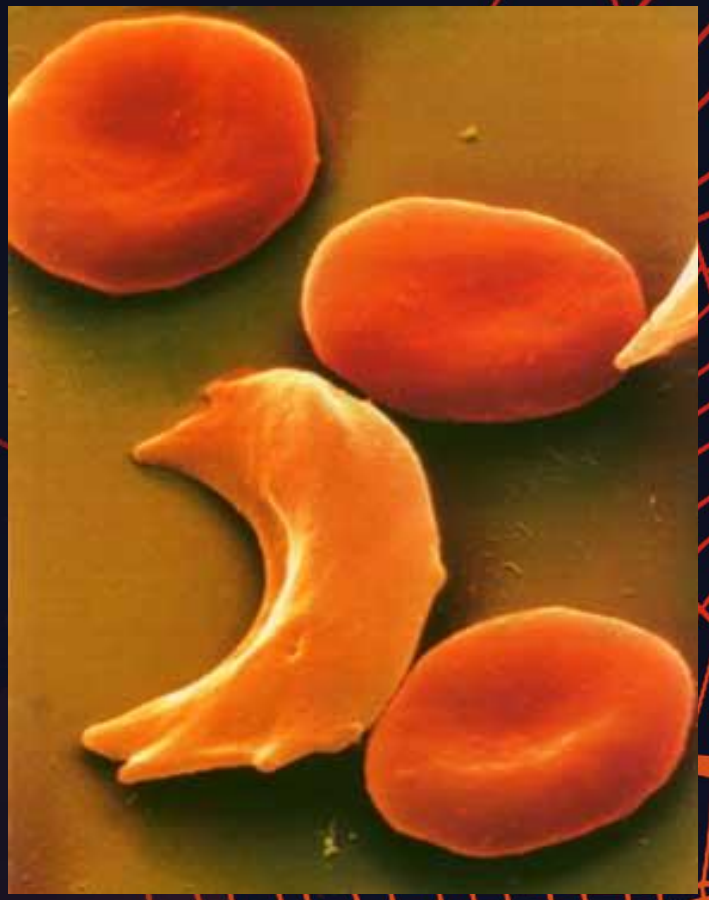
Point Mutation

- 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**



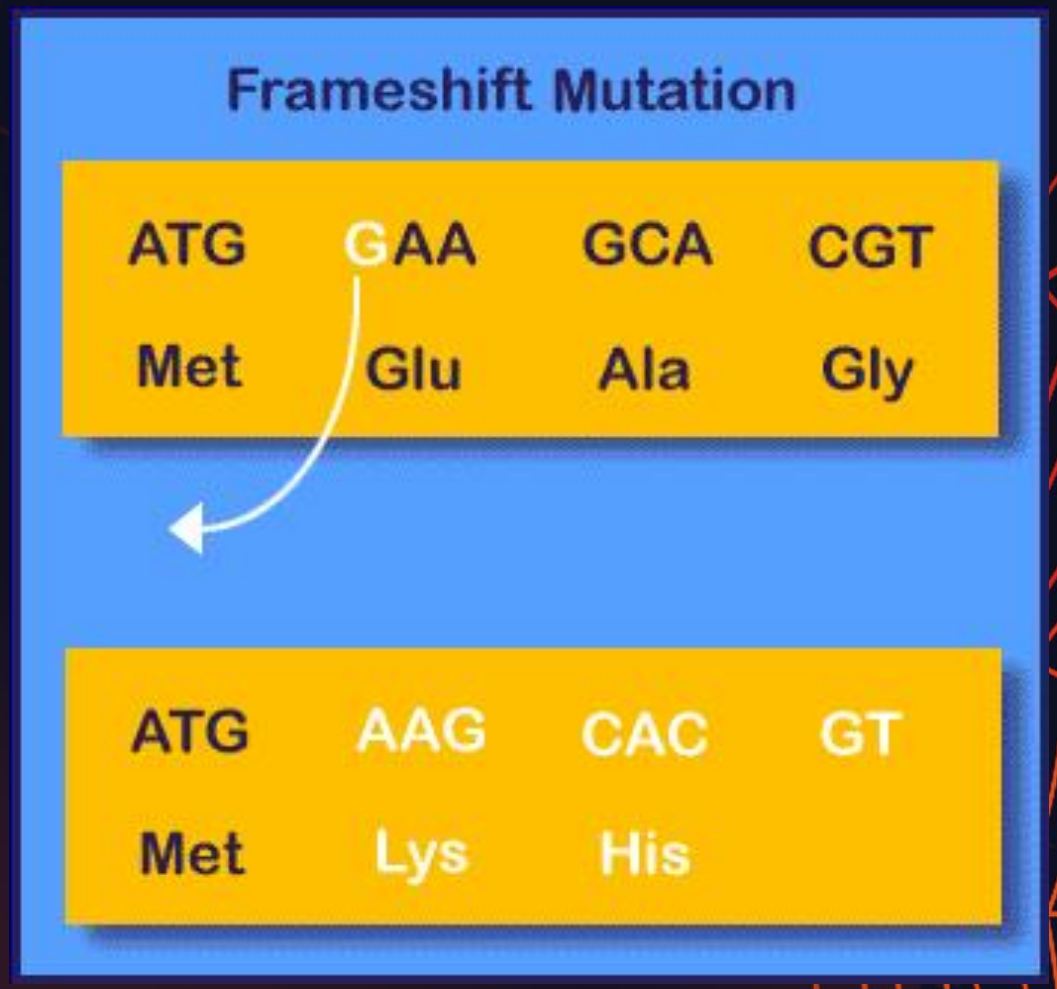
Frameshift Mutation

- **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 c**a**a tet hew eer at.**

Amino Acid Sequence Changed



THANK YOU!

- Questions
  or
  Comments