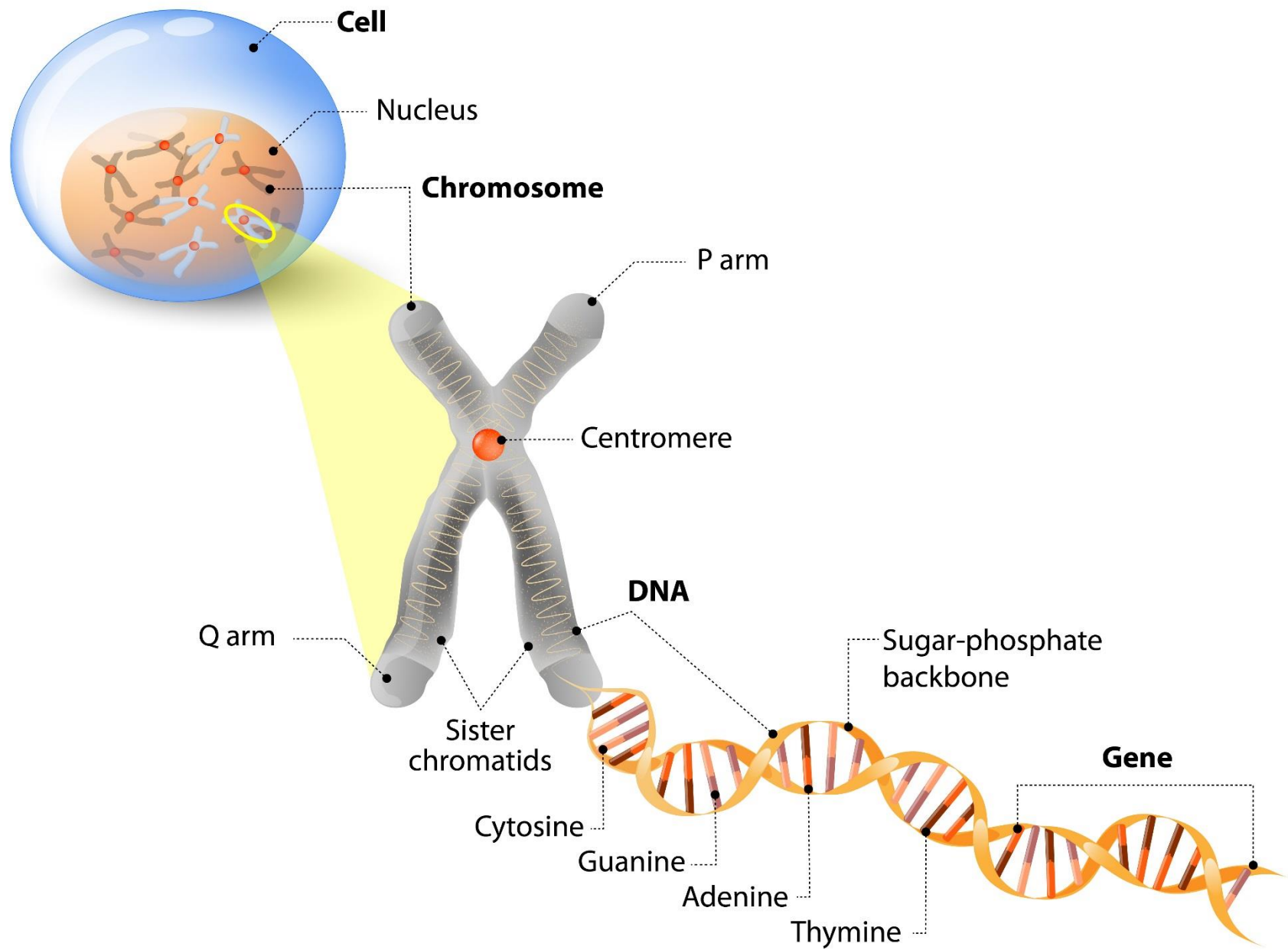
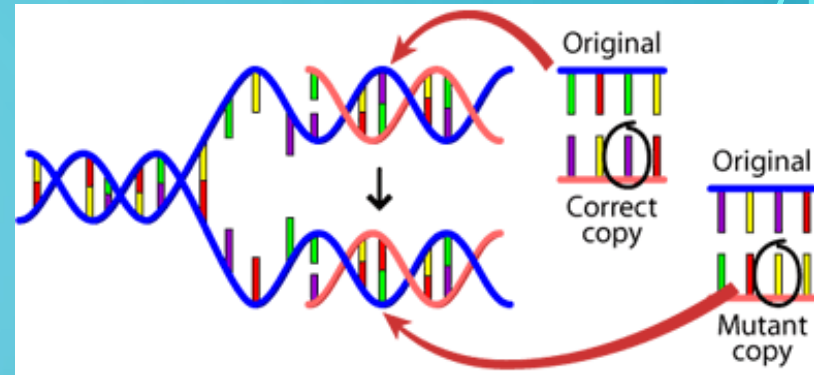




# MUTATIONS



# WHEN DO MUTATIONS OCCUR?



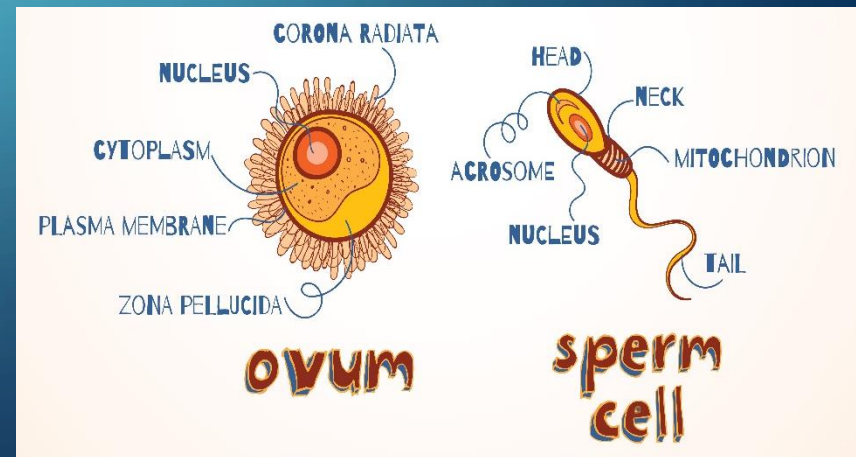
- While most DNA replicates with fairly high accuracy, mistakes do happen.
- DNA polymerase sometimes inserts the wrong nucleotide or too many or too few nucleotides into a sequence. This is called a mutation.
  - A **mutation** is a change in the DNA sequence that arises during **DNA replication**.
  - Most of these mistakes are fixed through various DNA repair processes.

# MUTATIONS

- Some replication errors make it past these mechanisms and become permanent mutations.
  - These altered nucleotide sequences can then be passed down from one cellular generation to the next
- When the genes for the DNA repair enzymes themselves become mutated, mistakes begin accumulating at a much higher rate.
  - In eukaryotes, such mutations can lead to cancer.

# WHICH MUTATIONS GET PASSED DOWN?

- Mutations may occur in somatic cells (body cells) but aren't passed to offspring
- May occur in gametes (eggs & sperm) and can be passed to offspring



# TYPES OF MUTATIONS

- **Mutations** are changes in genetic information
  - Gene mutations result from changes in a single gene.
- 2 main types of gene mutation:
  - Point Mutation – Affect one nucleotide; occurring at a single point on the gene. Usually one nucleotide is **substituted** for another nucleotide.
  - Frameshift Mutation – **Inserting** an extra nucleotide or **deleting** a nucleotide causes the entire code to “*shift*”. Insertions and deletions result in the “shifting” of the genetic code.

# GENE MUTATION- POINT MUTATIONS

- single base change
  - silent mutation
    - no amino acid change
      - Due to redundancy in genetic code
  - Missense mutation
    - change amino acid
  - Nonsense mutation
    - change to stop codon

# GENE MUTATION- FRAMESHIFT MUTATIONS

- A mutation is one in which one or more nucleotides are **inserted** or **deleted**
- Changes the “reading **frame**” like changing a sentence
- **Proteins** built **incorrectly** as a result because the whole DNA **sequence** is changed



# FRAMESHIFT MUTATION

- Original:

- The fat cat ate the wee rat.

- Frame Shift :

- **The fat caa tet hew eer at.**

- What caused the original sentence to change?

# Frameshift Mutation

<b>ATG</b>	<b>GAA</b>	<b>GCA</b>	<b>CGT</b>
<b>Met</b>	<b>Glu</b>	<b>Ala</b>	<b>Gly</b>



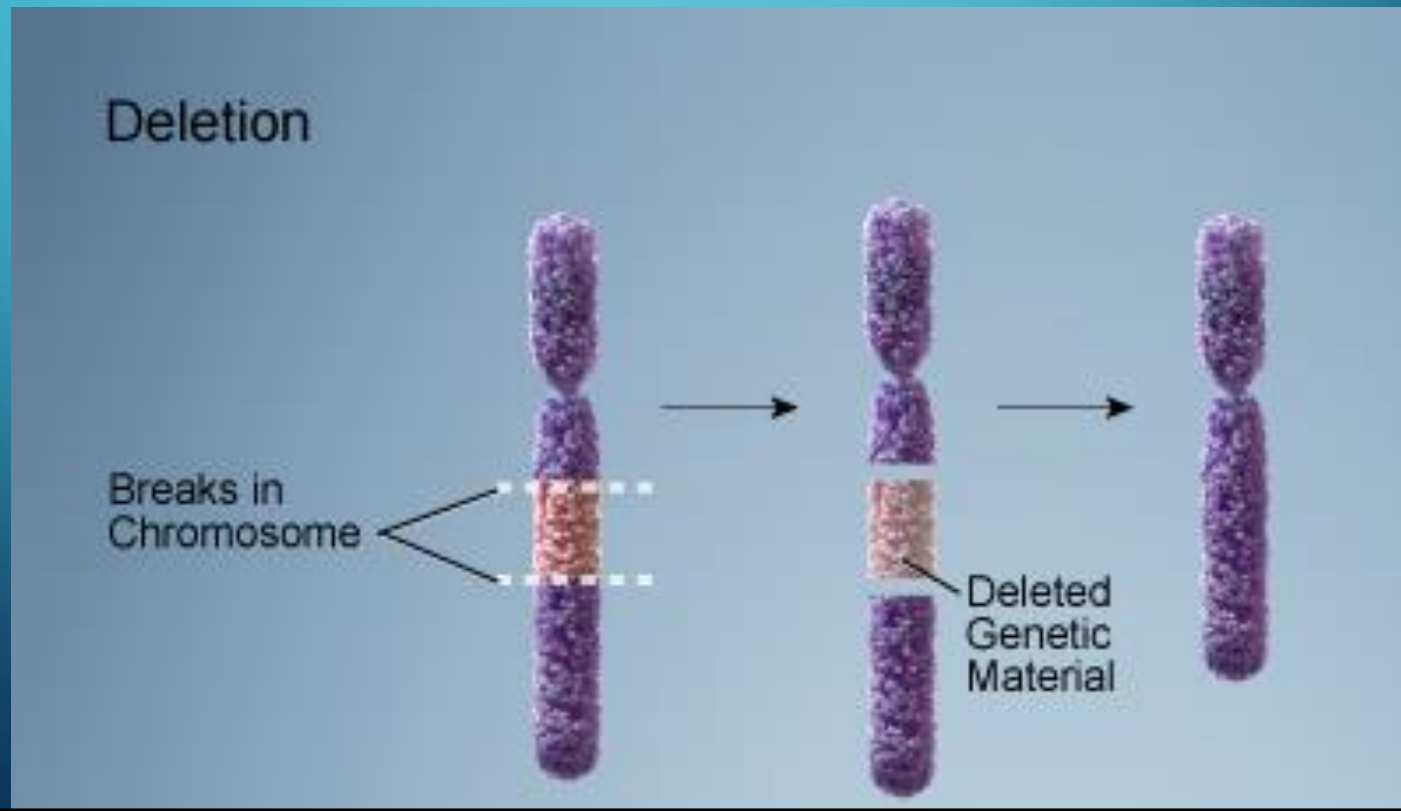
<b>ATG</b>	<b>AAG</b>	<b>CAC</b>	<b>GT</b>
<b>Met</b>	<b>Lys</b>	<b>His</b>	

# TYPES OF MUTATIONS

- **Mutations** are changes in genetic information
  - Chromosomal mutations involve changes whole chromosomes in terms of its number or structure.
- 5 main types of chromosomal mutations:
  1. Deletion
  2. Inversion
  3. Translocation
  4. Nondisjunction
  5. Duplication

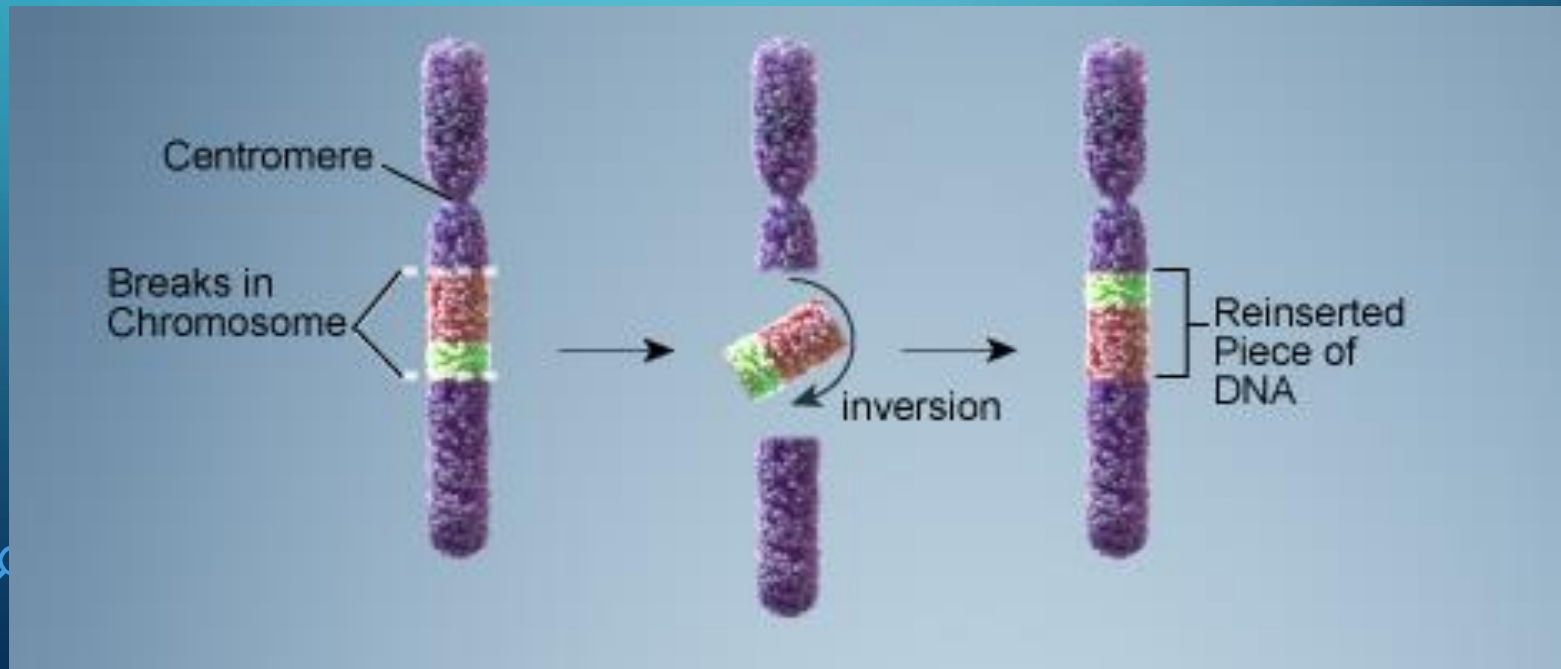
# CHROMOSOMAL MUTATION- DELETION

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



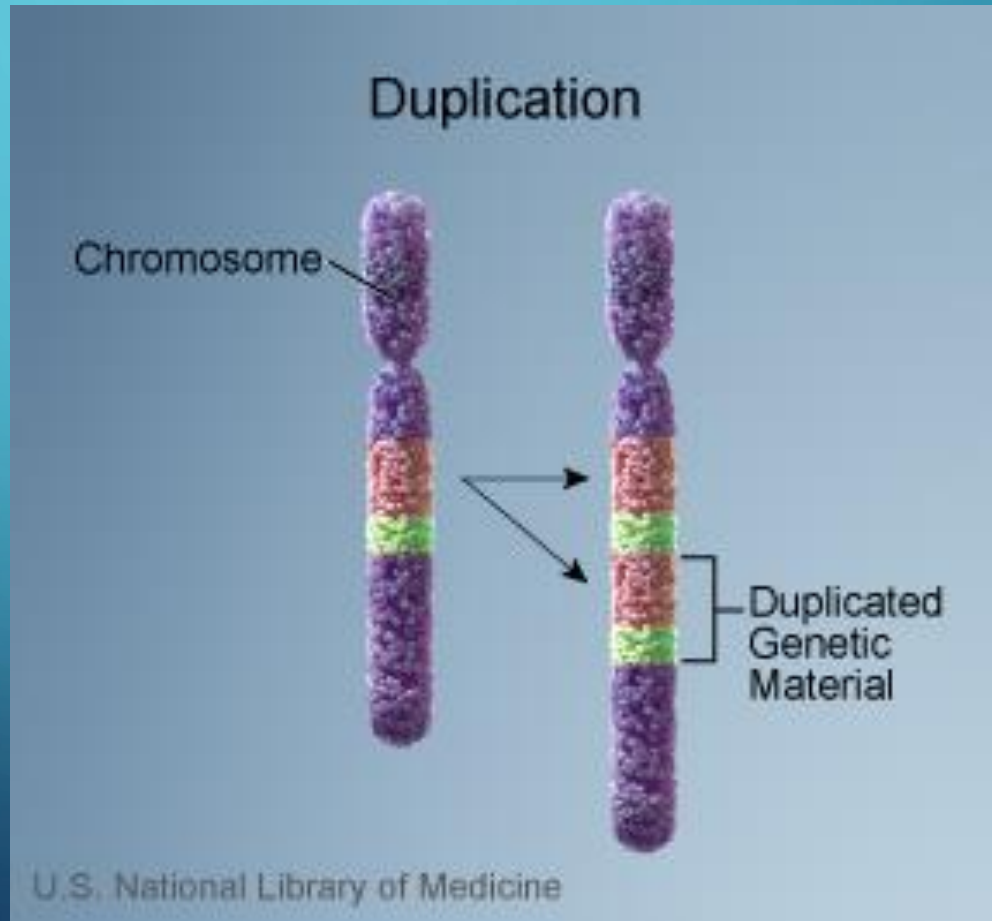
# CHROMOSOMAL MUTATION- INVERSION

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



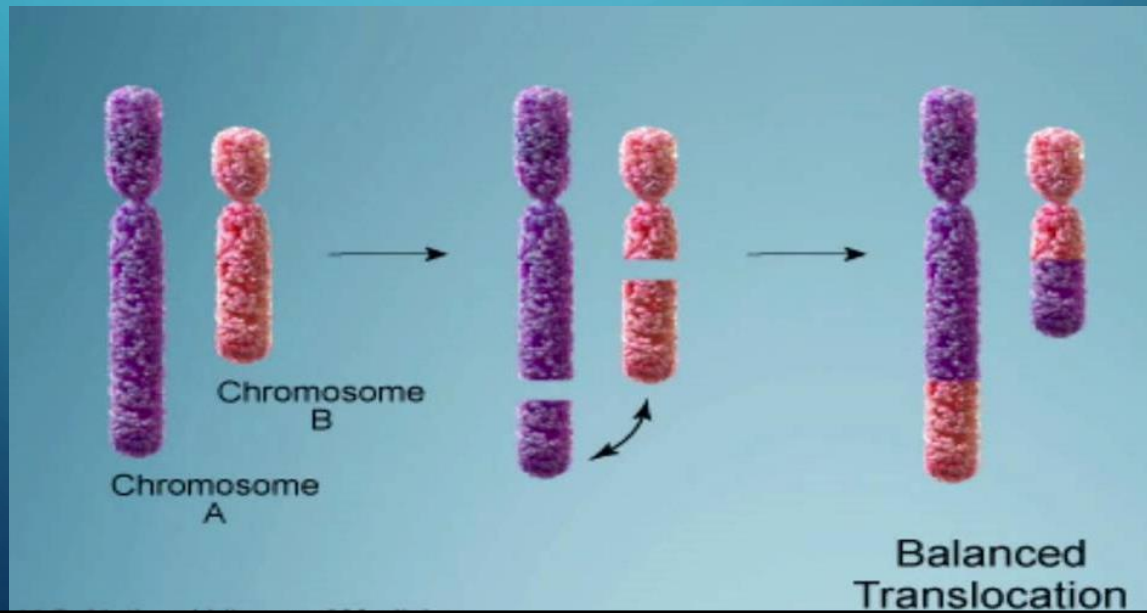
# CHROMOSOMAL MUTATION- DUPLICATION

- Occurs when an entire gene **sequence** is **repeated**



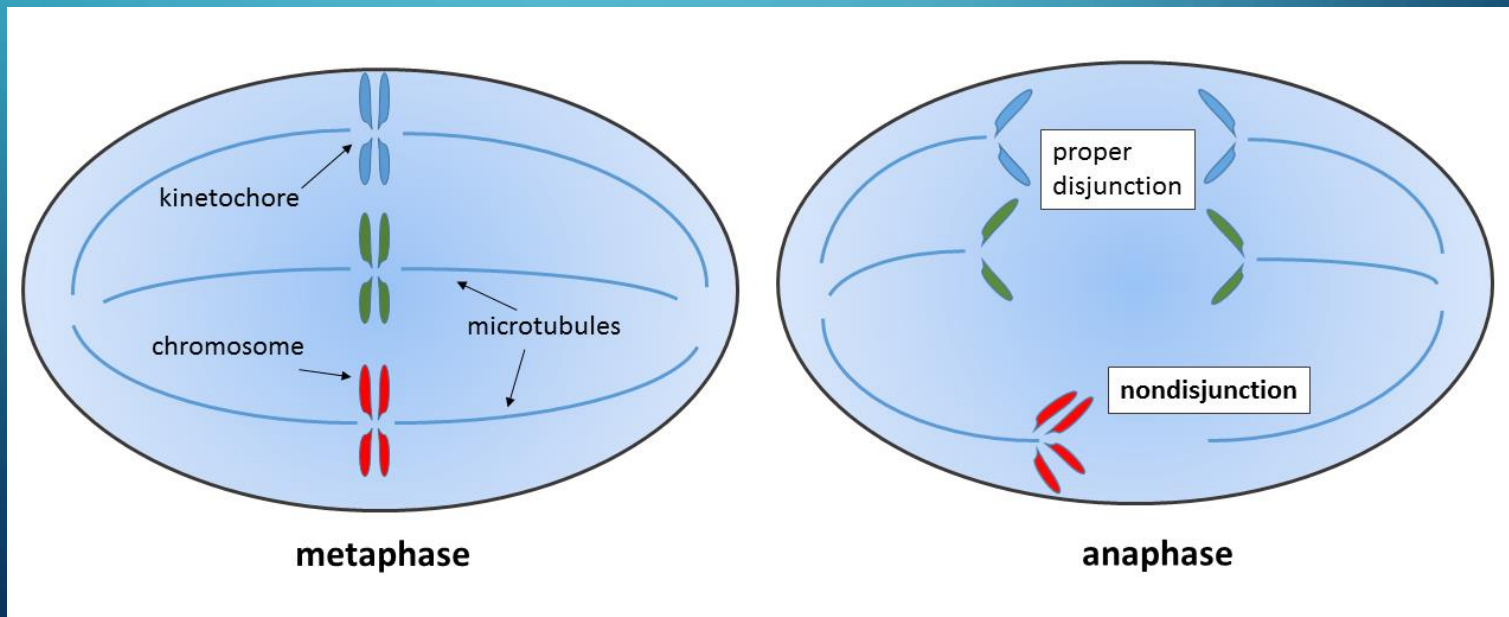
# CHROMOSOMAL MUTATION- TRANSLOCATION

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



# CHROMOSOMAL MUTATION- NONDISJUNCTION

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





# EFFECTS OF MUTATIONS

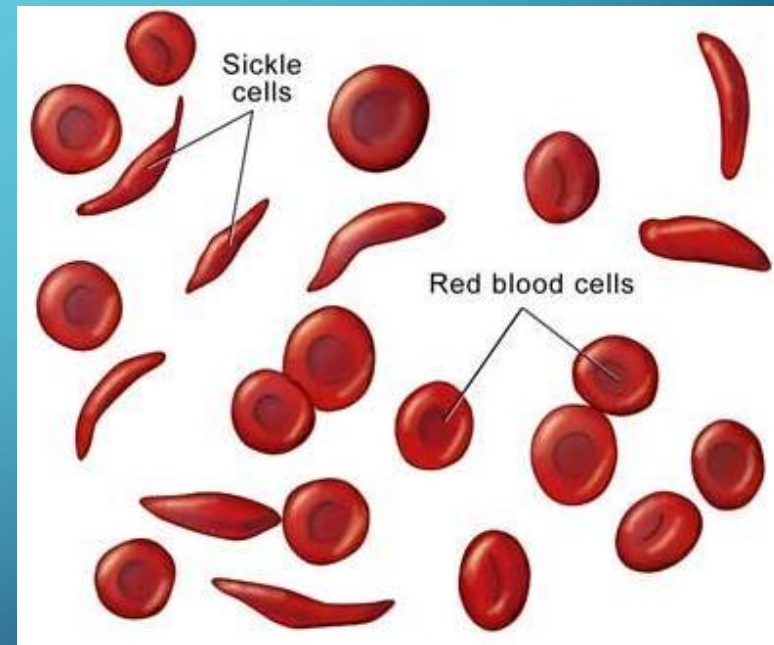
- Mutations may or may not affect an organism
  - Some mutations arise from mutagens- chemical or physical agents in the environment.
    - For example: some pesticides, tobacco smoke, X-rays, and environmental pollutants.
  - **Thus, the effects of mutations on genes vary widely. Some have little or no effect; and some produce beneficial variations. Some negatively disrupt gene function.**
  - Most mutations are neutral; they have little or no effect on the expression of genes or the function of proteins for which they code.

# EFFECTS OF MUTATIONS

- Whether a mutation is negative or beneficial depends on how its DNA changes relative to the organism's situation.
- Without mutations, organisms could not evolve, because mutations are the source of genetic variability in a species.
- Some of the most harmful mutations are those that dramatically change protein structure or gene activity.

# SICKLE CELL DISEASE

Sickle cell disease is a disorder associated with the changes in the shape of red blood cells. It is caused by a **point mutation** in one of the polypeptides found in hemoglobin, the blood's principal oxygen-carrying protein.



# EPIGENETICS

- Epigenetics is the study of heritable changes in gene expression that **doesn't** involve changes to the **DNA sequence**
- Certain circumstances in life can cause genes to be silenced or expressed over time (**gene expression**)
- **Environmental Factors** can regulate gene expression (*stress, diet, behavior, toxins & others*)
- <https://www.youtube.com/watch?v=AvB0q3mg4sQ>