

# Mutations



Occasionally, cells make mistakes when copying their DNA.

- If the base pairs don't stay in the correct order, a mutation can occur
- **Mutation**: when DNA is damaged or changed in such a way that alters the genetic message carried by that gene.



There are two different types of mutations:



- **Gene Mutations:** mutations that change a single gene
- **Chromosomal Mutations:** mutations that change whole chromosomes.

# Gene Mutations

- Gene mutations that involve a change in one nucleotide are called *point mutations*.
  - Occur at a single “point”
  - Usually happen during replication.
  - 3 different kinds of point mutations:
    1. Substitution
    2. Insertion
    3. Deletion

# 1. Substitution

- One nitrogen base is changed to a different nitrogen base.
  - Usually only affects one amino acid
  - Sometimes have no effect at all

Ex:

CCC

CCA

Proline



Proline

Even though a cytosine was swapped for an adenine, the code still calls for the same amino acid, so this mutation would cause no effect at all

CCC

ACC

Proline



threonine

The cytosine being swapped out for adenine caused a wrong amino acid to be coded for

## 2. Insertions

- When one nitrogen base is inserted into the DNA sequence.
  - The effect can be dramatic, changing the amino acid sequence from that point on.

CGACCCATT

CGA CCC ATT

CGACICCATT

CGA CTC CAT T



# 3. Deletions

- When one nitrogen base is deleted out of the sequence
  - Effect can be dramatic, affecting all the amino acids after the deletion.

CCGTACAGG

CCG TAC AGG

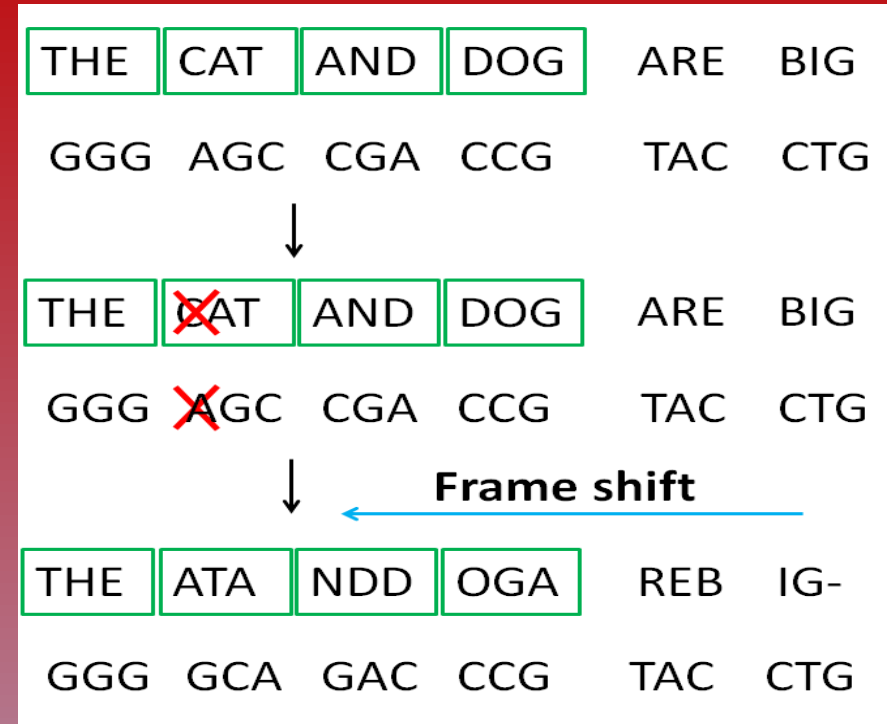
CCGTACAGG

CCG ACA GG\_\_



# Frameshift Mutations

- Insertions and deletions can make dramatic changes in the DNA sequence.
  - Another name for them is: **frameshift mutations**.
- Shifting changes the codons, therefore changing how the codons are read
  - Leads to the incorrect proteins being created



**TAT TGG CTA GTA CAT**  
**Tyr Trp Leu Val His**

**TAC TCG GCT AGT ACA T**  
**Tyr Ser Ala Ser Thr**



# Check Yourself

Name the type of mutation:

1. **TAC GGC AGC TGA**      **Insertion**  
**TAC GCG CAG CTG**

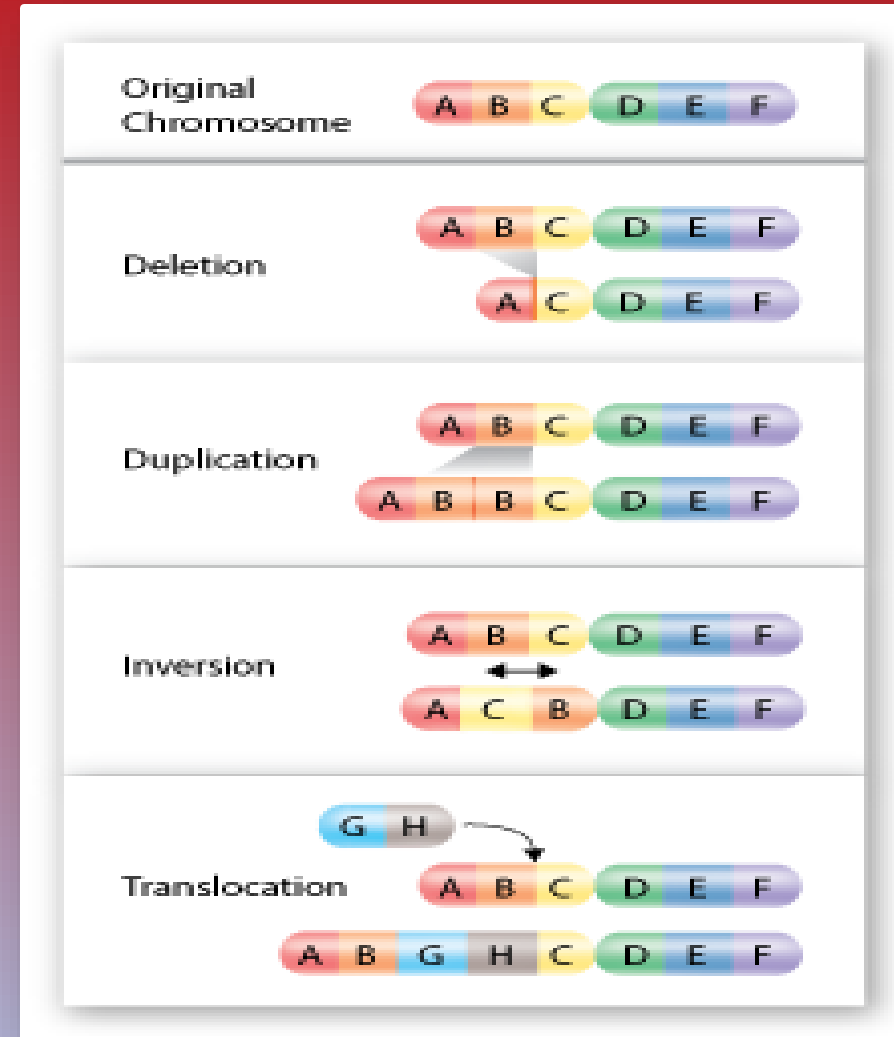
2. **AAC TGA CGC GAC**  
**AAC TGA CGC TAC**      **Substitution**

# Chromosomal Mutations

- Involve changes in whole chromosome.

## Four types:

- Deletion
  - Missing a gene
- Duplication
  - Two genes where one should be
- Inversion
  - Reversing gene positions
- Translocation
  - Set of genes where they don't belong



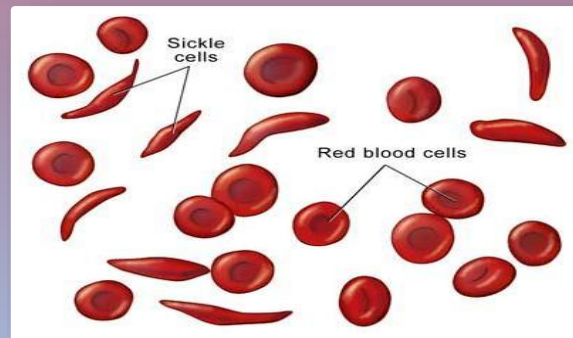
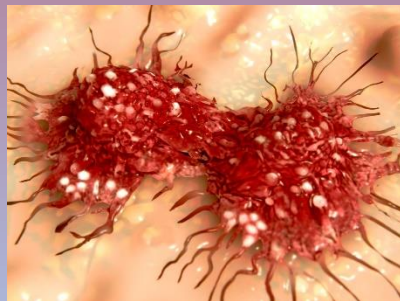
# Why do mutations happen?

Mutations happen for multiple reasons:

- Error in genetic replication
- Stressful environments
- **Mutagens:** chemical or physical agents in the environment
  - X-Rays/UV Light/Harmful Chemicals
  - ex: Cigarette smoking leads to over 80% of lung cancers

# Harmful effects of mutations

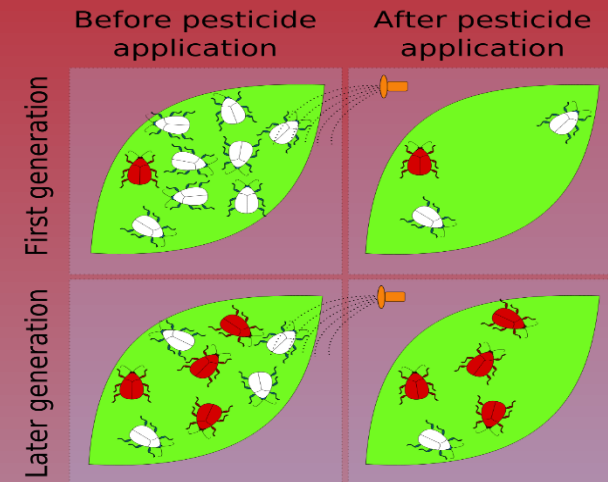
- When incorrect proteins are made can lead to:
  - Diseases
  - Cancers (uncontrolled cell growth)
  - Deformities



# Benefits of mutations

In some cases, random mutations benefit certain organisms

- Changes can potentially help an organism survive, this is what drives evolution via natural selection!
  - *Example:* Insects that (over time) are able to resist chemical pesticides
- Farmers also use genetically modified plants that are genetically mutated to be bigger and fuller crop.



# Bacteria Evolving Antibiotic Resistance

