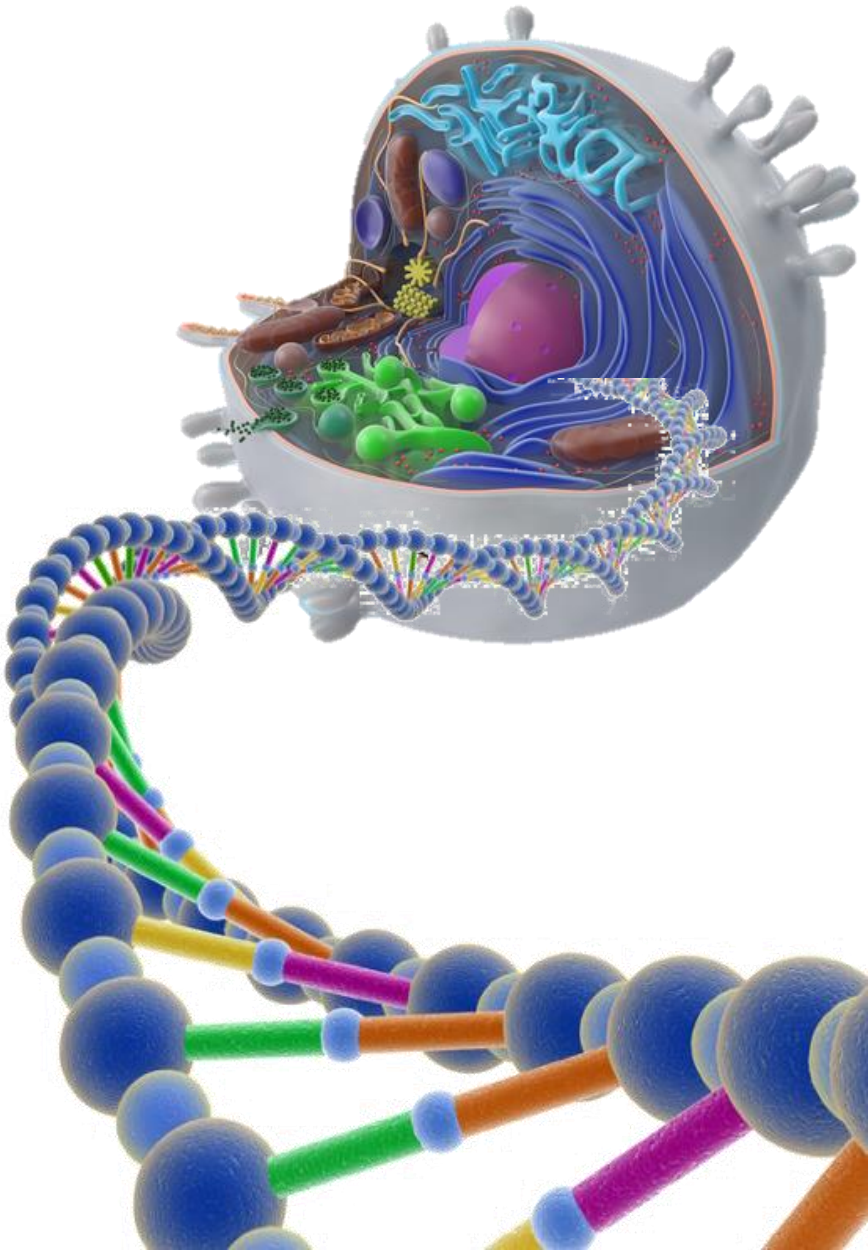

GENETIC
MUTATIONS

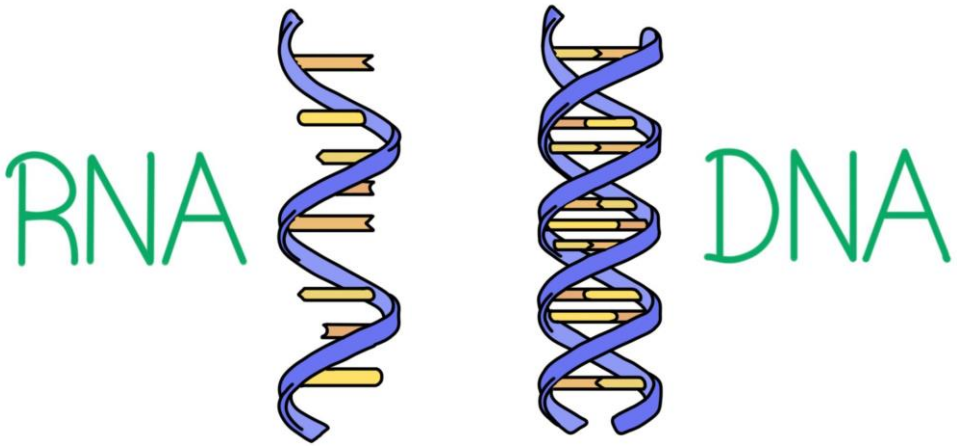
Mistakes Happen

- Sometimes, cells make mistakes.
- These mistakes can have a range of effects.
- When they occur in DNA or RNA, they are called <u>mutations</u>.



Mutations

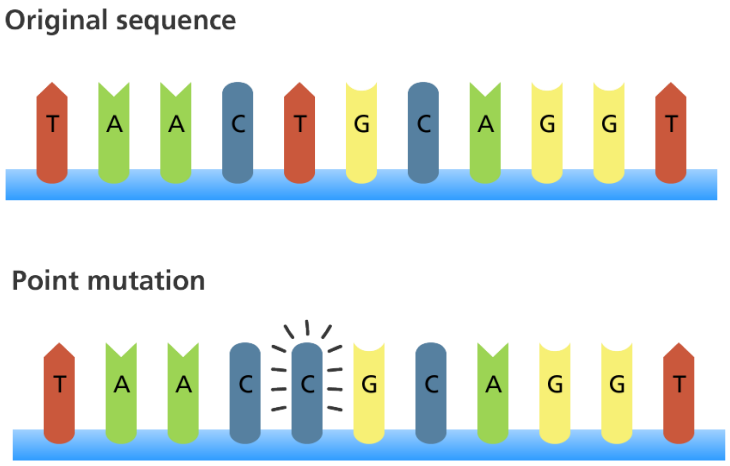
- A mutation is a change in an organism's nucleic acids.



- There are two types of mutations:
  - **Gene mutations** (genetics)
  - **Chromosomal mutations** (heredity)

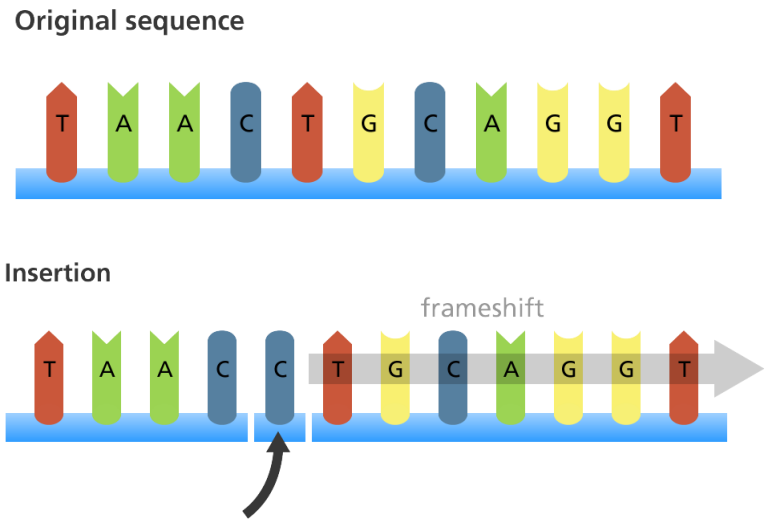
Gene Mutations

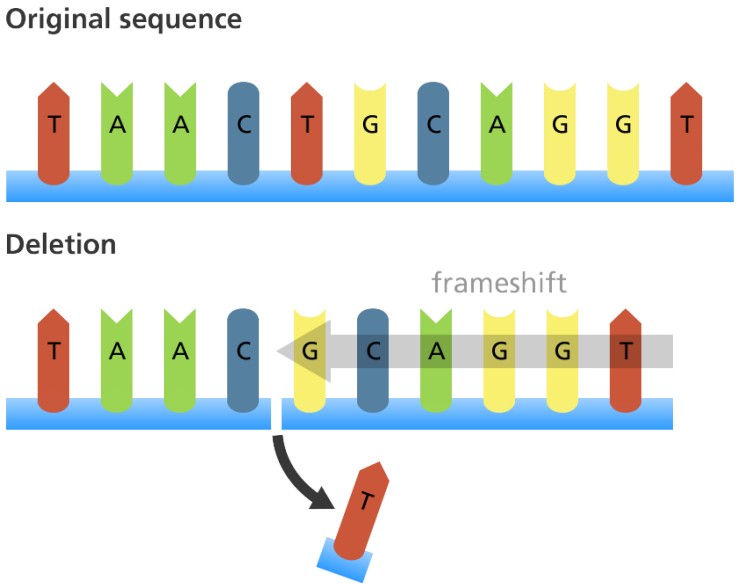
- **Point mutation** (aka: substitution)
  - a mutation in which one nucleotide is put in place of the correct nucleotide
  - Usually, a mistake like this is caught and fixed by DNA polymerase.
  - If not, the substitution may permanently change an organism's DNA.
  - Ex. sickle cell anemia



Gene Mutations

- **Frame shift mutation**
  - involves the insertion or deletion of a nucleotide in a DNA sequence
  - This shifts the entire sequence by one or more nucleotides (tends to have a bigger impact).
  - Ex. cystic fibrosis





Gene Mutations

THE DOG SAT.

TED OGS AT.

The deletion changes the reading frame, which results in codons that might code for different amino acids.

Gene Mutations

THE DOG SAT.

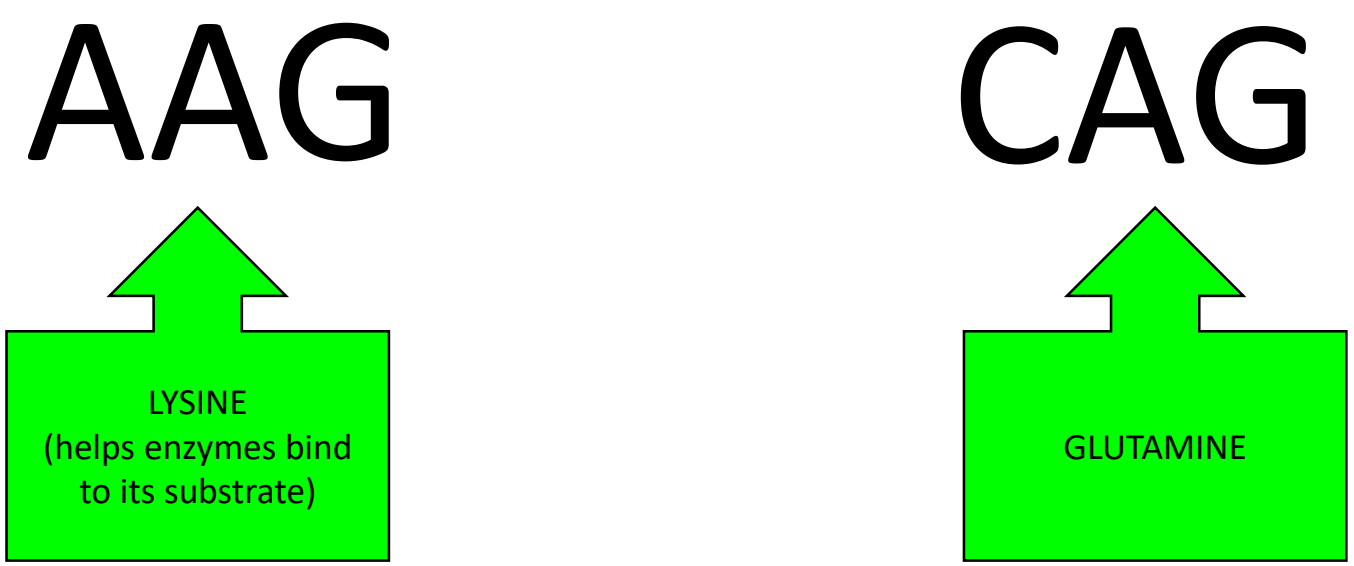
T

THT EDO GSA T.

The insertion changes the reading frame, which results in codons that might code for different amino acids.

Gene Mutations

- **Mutations may or may not affect an organism.**
  - Ex. Suppose a substitution occurs in a coding region of DNA that changes an AAG codon to CAG.



This substitution could affect protein folding and destroy the protein's function. This is known as a missense mutation.

Gene Mutations

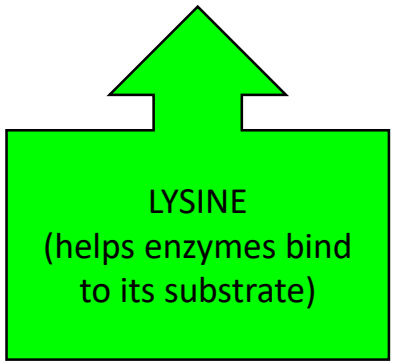
- **Mutations may or may not affect an organism.**
  - Ex. Suppose a substitution occurs in a coding region of DNA that changes an UAC codon to UAA.

This substitution would result in a premature stop codon. This is known as a nonsense mutation.

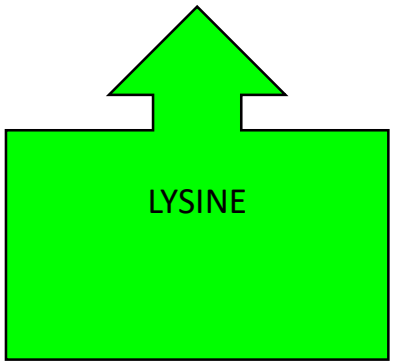
Gene Mutations

- **Mutations may or may not affect an organism.**
  - Remember: Some codons code for the same amino acid.
  - Let's suppose an "A" is substituted for the "G"

AAG



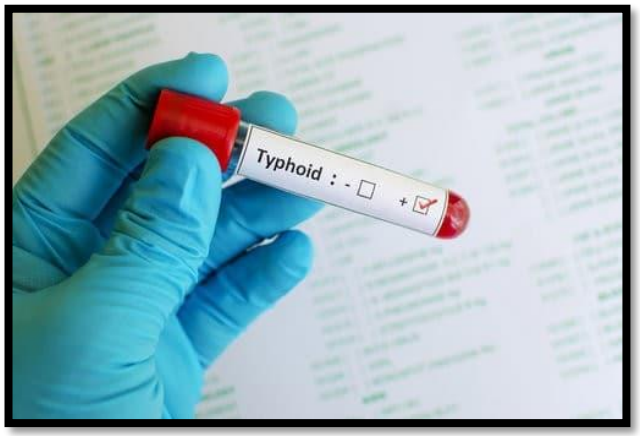
AAA



This substitution could have no affect. This is called a "silent" mutation.

Gene Mutations

- Mutations are caused by several factors.
  - replication errors
  - mitosis
  - meiosis
  - protein synthesis
  - mutagens



- Mutations are not uncommon.
  - Some are harmful. (ex. cystic fibrosis)
  - Some are helpful. (ex. immunity to typhoid fever)
  - Some have no affect. (ex. red hair)

- Organisms have many tools to repair them.
  - DNA has a built-in proofreader (DNA polymerase).
  - Sometimes DNA polymerase misses an error.

Mutagens

- ❑ultraviolet (UV) rays from the sun
- ❑industrial chemicals
- ❑carcinogens
- ❑infectious agents (viruses & bacteria)

Mutations Explained

Video Link: https://youtu.be/eDbK0cxKKsk