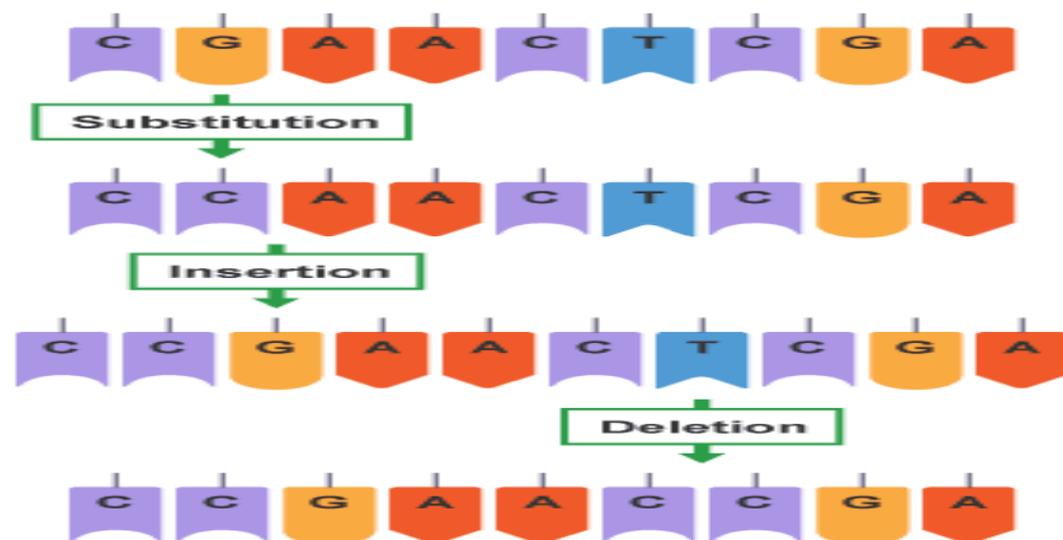


## mutations

A mutation is a change in the DNA sequence of an organism. These changes can arise from errors during DNA replication, exposure to mutagens (like radiation or chemicals), or viral infections. Mutations can be beneficial, harmful, or have no noticeable effect on the organism. They can occur in both somatic cells (body cells) or germline cells (sperm and egg cells). Germline mutations can be passed on to offspring, while somatic mutations are not inherited



What causes mutations?

**DNA Replication Errors:** During cell division, DNA replication is not perfect, and occasional mistakes can occur, leading to mutations.

**Mutagens:** Environmental factors like radiation (UV light, X-rays), certain chemicals (found in cigarette smoke, for example), and even some viruses can damage DNA and induce mutations.

**Spontaneous Mutations:** Some mutations arise randomly without any apparent cause.

## **Types of Mutations:**

### Point Mutations:

These involve changes to a single nucleotide base in the DNA sequence, such as substitutions, insertions, or deletions.

### Chromosomal Aberrations:

These are larger-scale mutations that affect the structure or number of chromosomes. Examples include deletions (loss of a DNA segment), duplications (extra copies of a DNA segment), inversions (flipping a segment of DNA), and translocations (moving a segment of DNA to a different chromosome).

### Impact of Mutations:

#### Harmless:

Many mutations have no noticeable effect on the organism. These can be silent mutations that don't change the protein produced or mutations in non-coding regions of the DNA.

#### Harmful:

Some mutations can disrupt gene function, leading to diseases like cancer or genetic disorders. For example, mutations in tumor suppressor genes can lead to uncontrolled cell growth.

#### Beneficial:

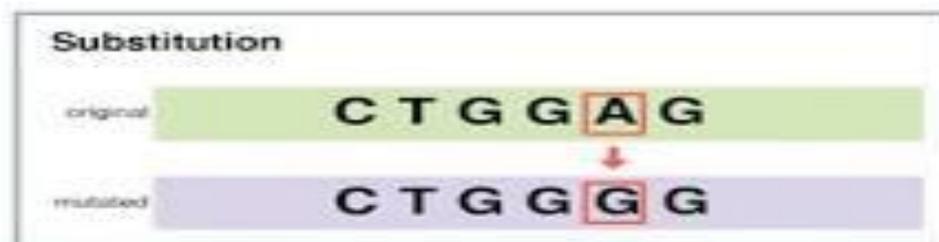
In rare cases, mutations can be advantageous, providing an organism with a new trait that helps it survive and reproduce more effectively. These beneficial mutations are the basis of evolution.

what are the 4 types of mutations?

There are four types of chromosomal mutations: deletion, duplication, inversion, and translocation.

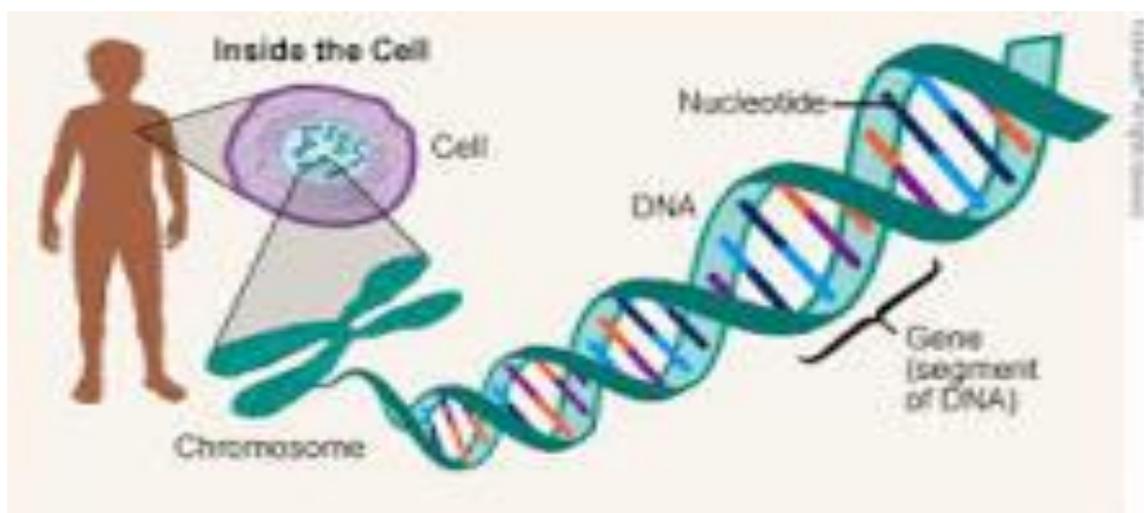
What is mutation, for example?

For example, sickle cell anemia is caused by a substitution in the beta-hemoglobin gene, which alters a single amino acid in the protein produced. change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called silent mutations



What are some examples of mutations?

Hereditary mutations include cystic fibrosis, hemophilia, and sickle cell disease. Other mutations can happen on their own during a person's life. These are called sporadic, spontaneous, or new mutations. They affect only some cells.



## Type of mutatiuons

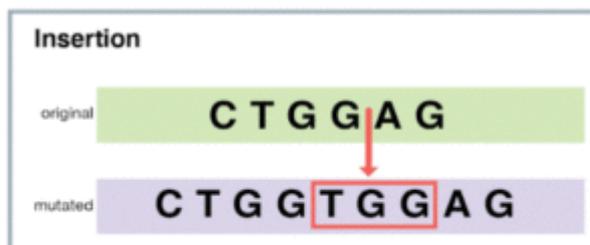
### Substitution

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). Such a substitution could:

1. change a codon to one that encodes a different amino acid and cause a small change in the protein produced. For example, [sickle cell anemia](#) is caused by a substitution in the beta-hemoglobin gene, which alters a single amino acid in the protein produced.
2. change a codon to one that encodes the same amino acid and causes no change in the protein produced. These are called silent mutations.
3. change an amino-acid-coding codon to a single “stop” codon and cause an incomplete protein. This can have serious effects since the incomplete protein probably won't function.

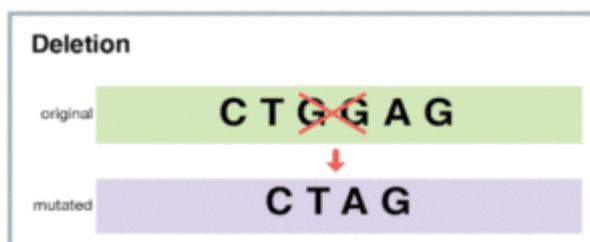
### Insertion

Insertions are mutations in which extra base pairs are inserted into a new place in the DNA.



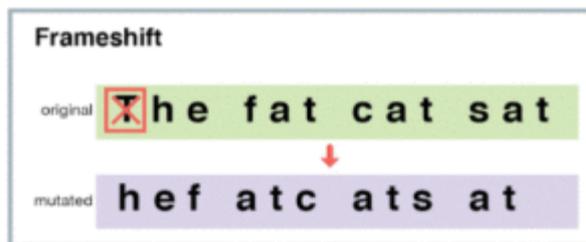
### Deletion

Deletions are mutations in which a section of DNA is lost, or deleted.



## Frameshift

Since protein-coding DNA is divided into codons three bases long, insertions and deletions can alter a gene so that its message is no longer correctly parsed. These changes are called frameshifts



For example, consider the sentence, “The fat cat sat.” Each word represents a codon. If we delete the first letter and parse the sentence in the same way, it doesn’t make sense.

In frameshifts, a similar error occurs at the DNA level, causing the codons to be parsed incorrectly. This usually generates truncated proteins that are as useless as “hef atc ats at” is uninformative.