

## Big Picture

Mutations occur when mistakes are made during transcription or replication of DNA. There are several kinds of mutations that occur in organisms. Mutations usually happen when chromosomes break or when nucleotides on a chromosome are changed, inserted, or deleted. Everyone has mutations, and most mutations are harmless. However, there are some mutations that can lead to serious illness or death.

## Key Terms

**Mutation:** A change in the bases of DNA/RNA.

**Mutagen:** Something that causes mutation.

**Germline Mutation:** A mutation in a gamete (can be passed onto offspring).

**Somatic Mutation:** Mutation that occurs in a somatic (non-gamete) cell.

**Chromosomal Alteration:** Mutation that changes chromosome structure.

**Point Mutation:** One nucleotide is changed.

**Frameshift Mutation:** When a nucleotide is deleted or inserted.

**Genetic Disorder:** Disease caused by gene mutations.

## Causes of Mutations

Most **mutations** are accidental and happen randomly as the result of mistakes during DNA replication or transcription

- Example: ATGA → ATCA

Mutations can also be caused by **mutagens** in the environment. Examples:

### Chemicals

- Cigarette smoke: contains dozens of mutagenic chemicals
- Nitrate and nitrate Preservatives: in hotdogs and other processed meats
- Barbecuing: create mutagenic chemicals in foods
- Benzoyl peroxide: common ingredient in acne products

### Infectious Agents

- *Helicobacter pylori*: bacteria spread through contaminated food
- Human papillomavirus (HIV): sexually transmitted virus

### Radiation

- UV radiation: both natural light and tanning beds
- X-rays: medical, dental, airport security screening

## Types of Mutations

- **Germline mutations** are serious because it can be passed onto the offspring of the organism. Only germline mutations can change offspring.
- **Somatic mutations** are not as serious because they can only occur in one cell and its daughter cells. The mutation cannot be passed onto offspring.

Mutations affect the genetic material. The effect can be a **chromosomal alteration**, **point mutation**, or **frameshift mutation**.

**Chromosomal alterations** happen when part of a chromosome breaks off and doesn't rejoin correctly.

- Serious mutation: often result in death or serious **genetic disorder**
- Types of chromosomal alterations: deletion, duplication, inversion, insertion, translocation.

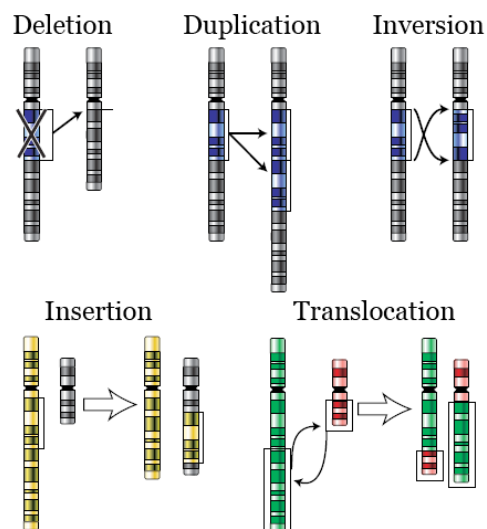


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Figure: Types of chromosomal alterations.

# MUTATION CONT.

## Types of Mutations (cont.)

**Point mutations** tend to be less serious, but the effect depends on how the mutation changes the genetic code.

Types of point mutations:

- Silent mutation: mutated codon codes for the same amino acid (no effect on organism)
  - Several codons code for the same amino acid, so if a point mutation changes CUU (leucine) to CUG, CUA, or CUC, there is no effect because all three also code for leucine
- Missense: mutated codon codes for a different amino acid
  - May have some effect on organism, but it depends on the situation
- Nonsense: mutated codon is a stop codon that stops protein synthesis early
  - Usually this has a serious effect

**Frameshift mutations** can greatly affect protein synthesis. Since codons are groups of 3 nucleotides, adding/deleting a nucleotide shifts everything over and changes every codon afterwards, which completely changes the amino acids produced.

- Example: AUG-ACU-UCG-GCC = start-threonine-serine-alanine

Now, assume an insertion occurs in this sequence. Let's say a C nucleotide is inserted after the start codon AUG:

- AUG-CAC-UUC-GGC-C = start-histidine-phenylalanine-glycine

The amino acid sequence changed dramatically even though only one nucleotide was inserted.

## Effects of Mutations

### Neutral mutations:

- Most mutations do not have a negative or a positive effect
- Many mutations are repaired before protein synthesis occurs
  - Cells containing DNA that cannot be repaired are usually prevented from dividing
- Silent point mutations are also neutral mutations because the amino acids in the protein do not change

### Beneficial mutations:

- Mutations that have a positive effect
- The new version of the protein helps the organism to adapt to changes in the environment
  - Beneficial mutations are necessary for evolution
- Tend to be passed onto more offspring and become more common over time

### Harmful mutations:

- Mutations that have a negative effect
- Result in genetic disorder or cancer
  - Cancer usually result from mutations in genes that regulate the cell cycle, which allow cells with damaged DNA to continuously divide

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