
14.4

# Types of Mutations

Now and then cells make mistakes in copying their own DNA, inserting the wrong base or even skipping a base as a strand is put together.

These variations are called **mutations**, from the Latin word *mutare*, meaning “to change.”

Mutations are heritable changes in genetic information.

# Types of Mutations

All mutations fall into two basic categories:

Those that produce changes in a single gene are known as gene mutations.

Those that produce changes in whole chromosomes are known as chromosomal mutations.

# Gene Mutations

Mutations that involve changes in one or a few nucleotides are known as **point mutations** because they occur at a single point in the DNA sequence. They generally occur during replication.

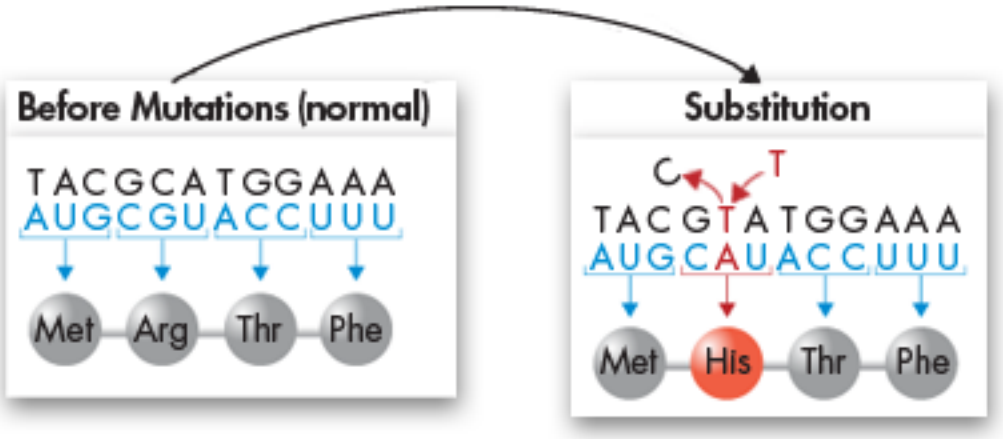
If a gene in one cell is altered, the alteration can be passed on to every cell that develops from the original one.

Point mutations include substitutions, insertions, and deletions.

# Substitutions

In a substitution, one base is changed to a different base.

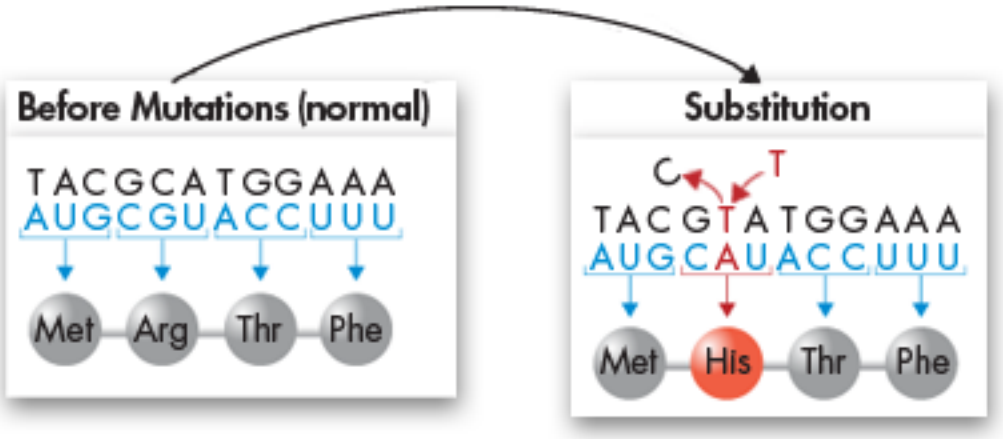
Substitutions usually affect no more than a single amino acid, and sometimes they have no effect at all.



# Substitutions

In this example, the base cytosine is replaced by the base thymine, resulting in a change in the mRNA codon from CGU (arginine) to CAU (histidine).

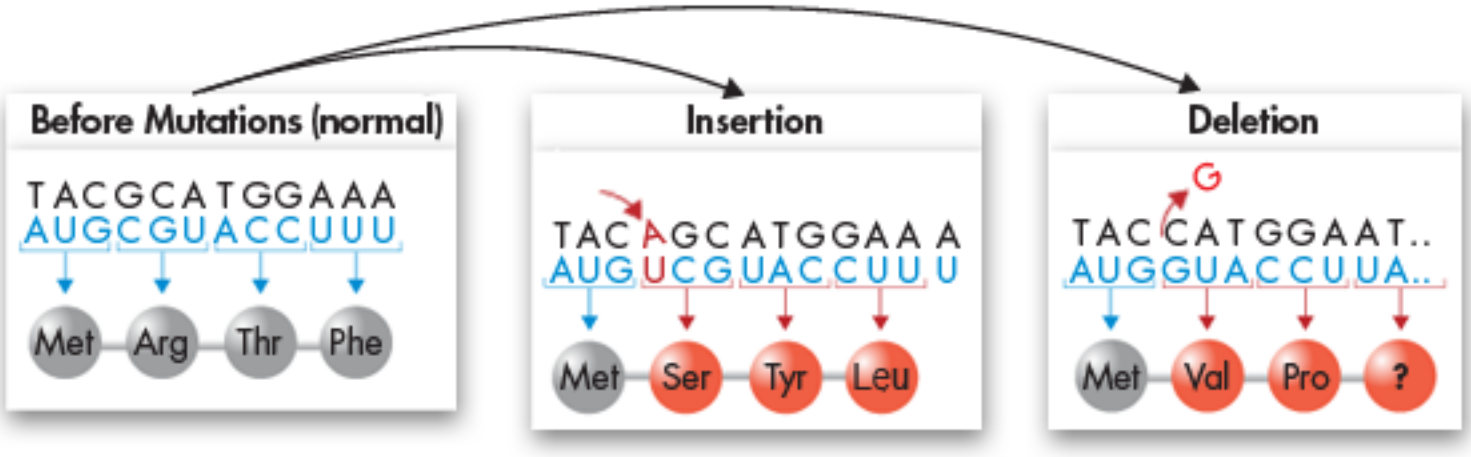
However, a change in the last base of the codon, from CGU to CGA for example, would still specify the amino acid arginine.



# Insertions and Deletions

Insertions and deletions are point mutations in which one base is inserted or removed from the DNA sequence.

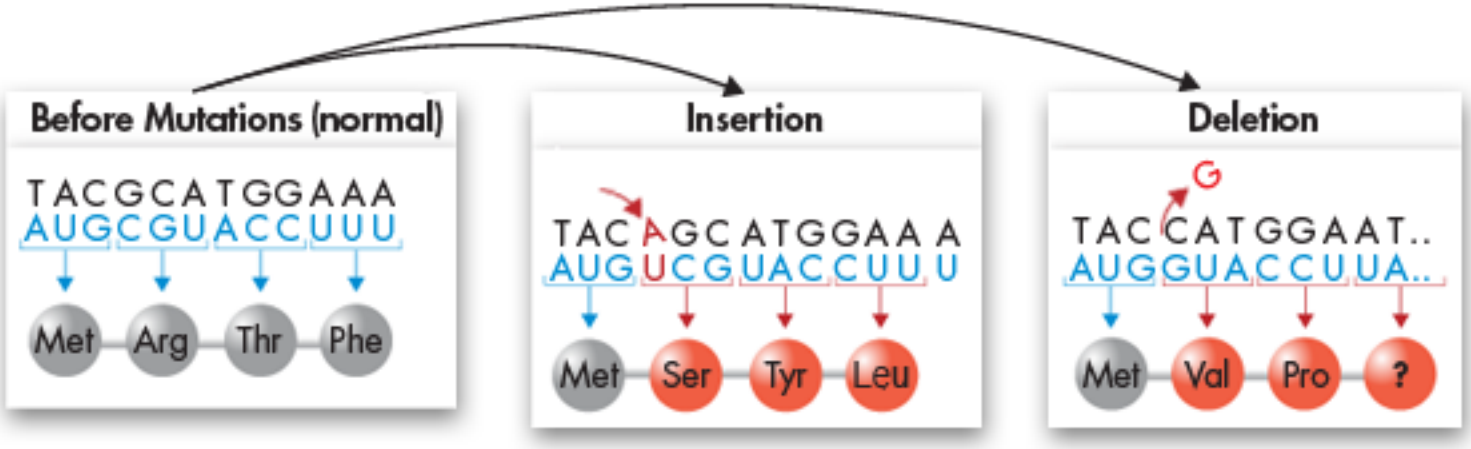
If a nucleotide is added or deleted, the bases are still read in groups of three, but now those groupings shift in every codon that follows the mutation.



# Insertions and Deletions

Insertions and deletions are also called **frameshift mutations** because they shift the "reading frame" of the genetic message.

Frameshift mutations can change every amino acid that follows the point of the mutation and can alter a protein so much that it is unable to perform its normal functions.



# Chromosomal Mutations

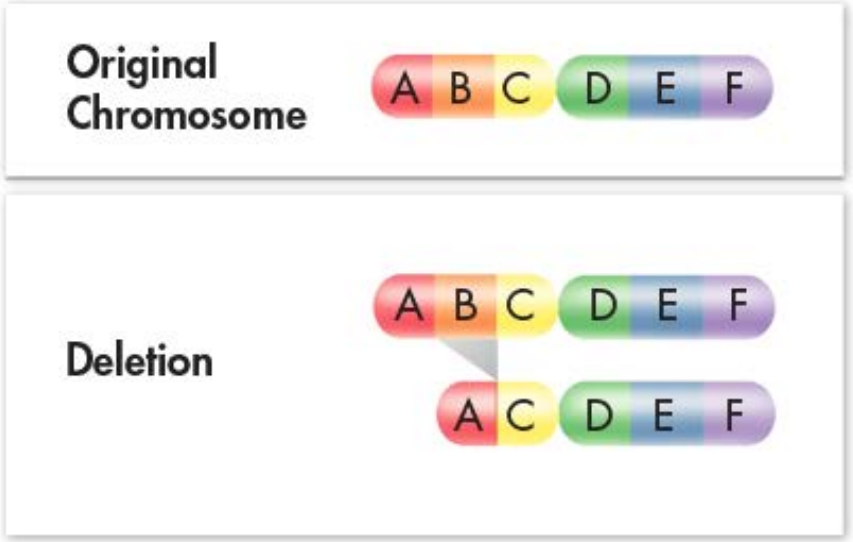
Chromosomal mutations involve changes in the number or structure of chromosomes.

These mutations can change the location of genes on chromosomes and can even change the number of copies of some genes.

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

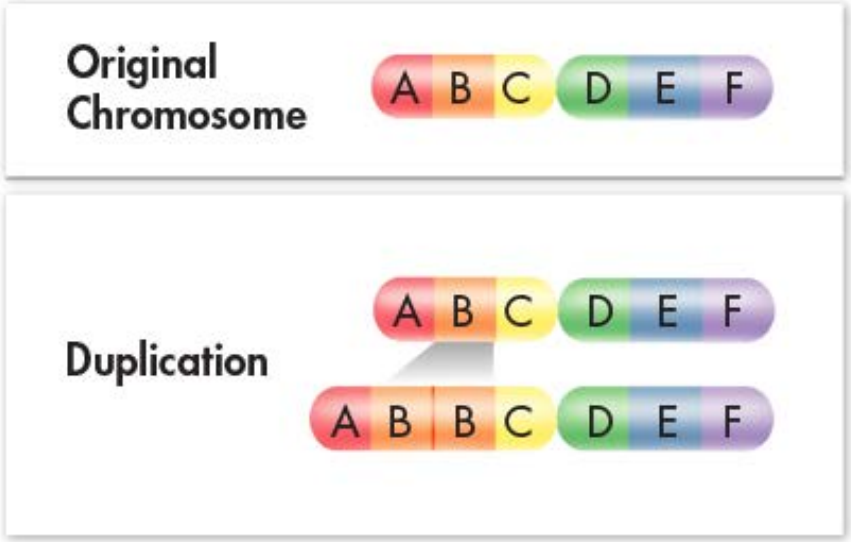
# Chromosomal Mutations

Deletion involves the loss of all or part of a chromosome.



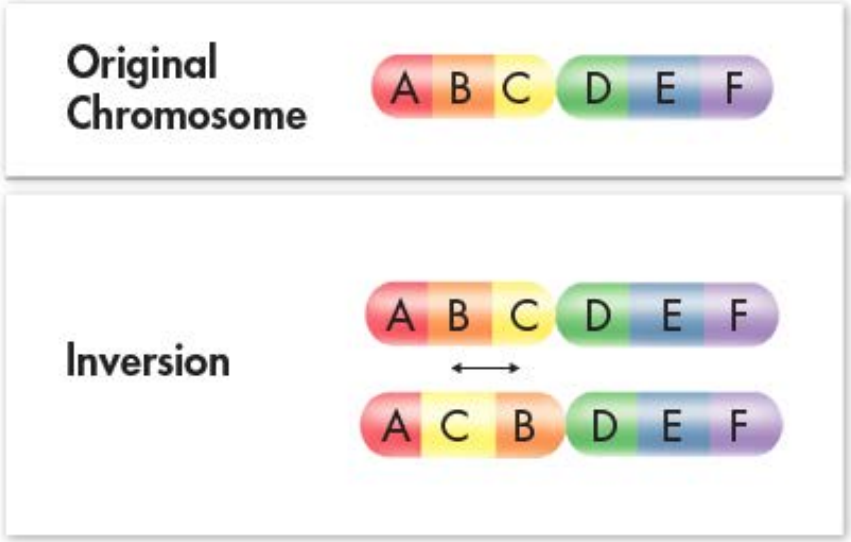
# Chromosomal Mutations

Duplication produces an extra copy of all or part of a chromosome.



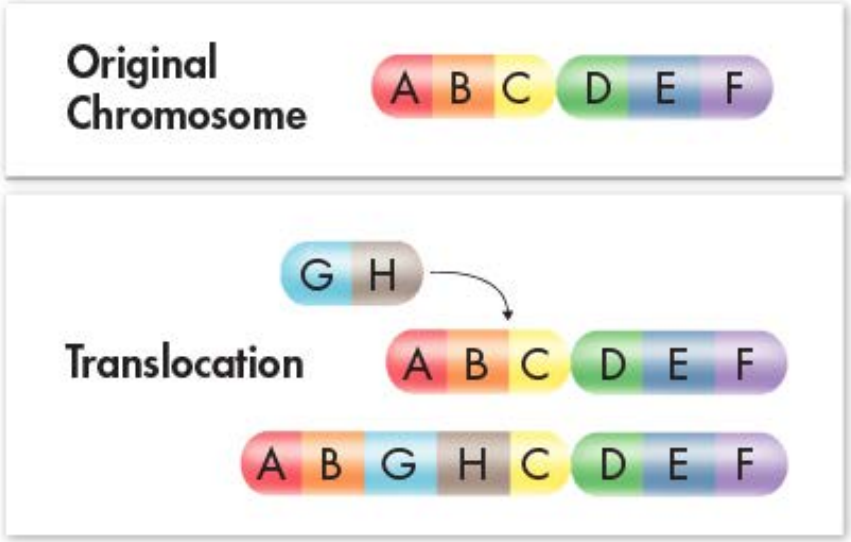
# Chromosomal Mutations

Inversion reverses the direction of parts of a chromosome.



# Chromosomal Mutations

Translocation occurs when part of one chromosome breaks off and attaches to another.



# Mutagens

Some mutations arise from **mutagens**, chemical or physical agents in the environment.

Chemical mutagens include certain pesticides, a few natural plant alkaloids, tobacco smoke, and environmental pollutants.

Physical mutagens include some forms of electromagnetic radiation, such as X-rays and ultraviolet light.

# Mutagens

If these mutagens interact with DNA, they can produce mutations at high rates.

Some compounds interfere with base-pairing, increasing the error rate of DNA replication.

Others weaken the DNA strand, causing breaks and inversions that produce chromosomal mutations.

Cells can sometimes repair the damage; but when they cannot, the DNA base sequence changes permanently.

# Harmful and Helpful Mutations

The effects of mutations on genes vary widely. Some have little or no effect; and some produce beneficial variations. Some negatively disrupt gene function.

Whether a mutation is negative or beneficial depends on how its DNA changes relative to the organism's situation.

Mutations are often thought of as negative because they disrupt the normal function of genes.

However, without mutations, organisms cannot evolve, because mutations are the source of genetic variability in a species.

# Harmful Effects

Some of the most harmful mutations are those that dramatically change protein structure or gene activity.

The defective proteins produced by these mutations can disrupt normal biological activities, and result in genetic disorders.

Some cancers, for example, are the product of mutations that cause the uncontrolled growth of cells.

# Harmful Effects

Sickle cell disease is a disorder associated with changes in the shape of red blood cells. Normal red blood cells are round. Sickle cells appear long and pointed.

Sickle cell disease is caused by a point mutation in one of the polypeptides found in hemoglobin, the blood's principal oxygen-carrying protein.

Among the symptoms of the disease are anemia, severe pain, frequent infections, and stunted growth.

# Beneficial Effects

Some of the variation produced by mutations can be highly advantageous to an organism or species.

Mutations often produce proteins with new or altered functions that can be useful to organisms in different or changing environments.

For example, mutations have helped many insects resist chemical pesticides.

Some mutations have enabled microorganisms to adapt to new chemicals in the environment.

# Beneficial Effects

Plant and animal breeders often make use of “good” mutations.

For example, when a complete set of chromosomes fails to separate during meiosis, the gametes that result may produce triploid (3N) or tetraploid (4N) organisms.

The condition in which an organism has extra sets of chromosomes is called **polyploidy.**

# Beneficial Effects

Polyploid plants are often larger and stronger than diploid plants.

Important crop plants—including bananas and limes—have been produced this way.

Polyploidy also occurs naturally in citrus plants, often through spontaneous mutations.