

Genetics Section 8.7

I. Mutations

*Mutation = *any change of DNA in a gene*.

-Mutations can be caused by *errors* in replication, transcription, cell division, or by external agents.

A. Mutation in gametes.

1. Mutations can affect **reproductive cells** by *changing the sequence of nucleotides within* a sperm or ovum.
2. If the **mutated gamete** takes part in fertilization, the **altered gene** would become part of the *genetic makeup of the offspring*.

3. The mutation can produce a *new trait* or it may result in *problems in the cell*.
4. In some cases, gene mutations may have *positive effects* for a species.

B. Mutations in somatic cells.

1. Damage to a body cell may impair the *function of the cell*.

Ex: A cell in the stomach may lose its ability to make HCL acid needed for digestion, or skin cells may lose elasticity.

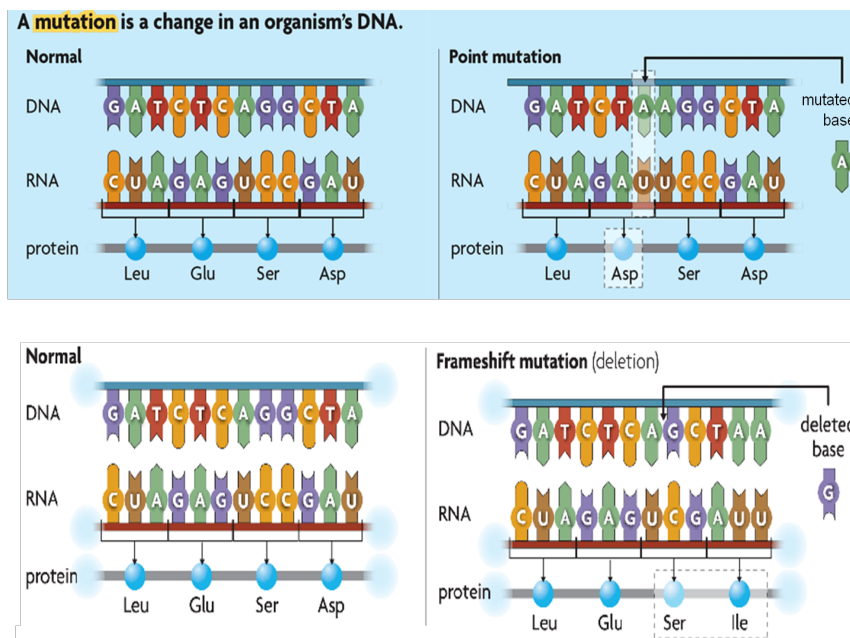
2. When the damaged cell *replicates and divides*, the new cell may affect the genes that control cell division, thus this can cause *cancer*.

II. There are two types of mutations: *chromosomal or gene*

A. Gene function can seriously be affected by mutations in individual genes. There are two types:

- 1) Point mutation - a change in *single* nucleotide
- 2) Frameshift mutations - involve the *addition or deletion* that alters every codon (set of nucleotides) from the point of the mutation on; it can completely change the *protein product* produced by a gene.

In general, point mutations are *less harmful* than frameshift, because only one codon is affected

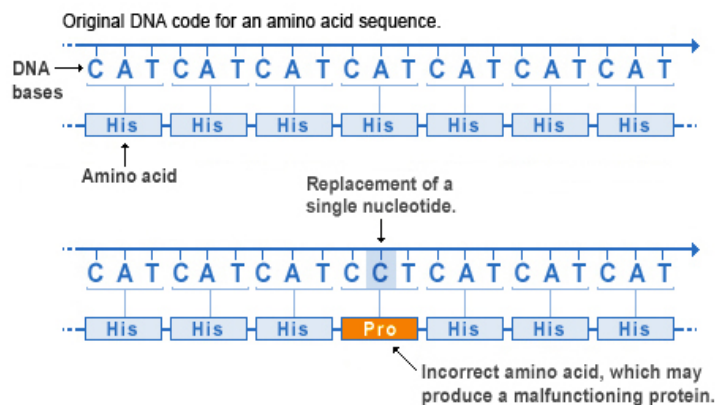


3. **Missense mutation** is a point mutation in which a single nucleotide change results in a codon that codes for a different amino acid. Sometimes it is considered a **silent mutation** because the **change has no effect on the protein produced**.

Another type of substitution is a **nonsense mutation** in which a **codon is changed to a premature stop codon** that results in truncation (shorten) of the resulting protein.

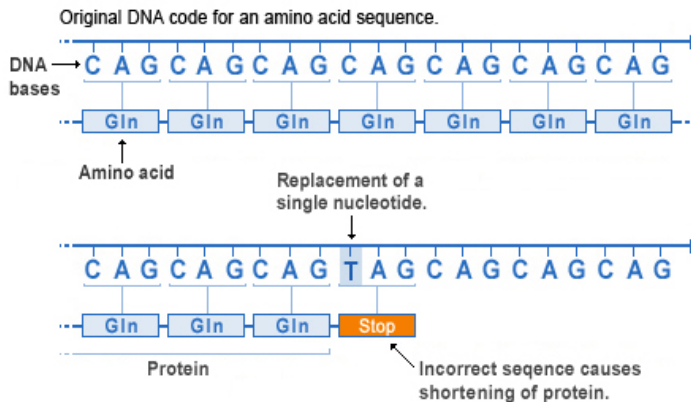
Missense or nonsense mutations can render the resulting protein partially or totally **nonfunctional**

Missense mutation



In this example, **the nucleotide adenine is replaced by cytosine** in the genetic code, introducing an **incorrect amino acid** into the protein sequence.

Nonsense mutation



In this example, the nucleotide cytosine is replaced by thymine in the DNA code, signaling the cell to shorten the protein.

B. Chromosomal mutations can involve:

- 1) Segments of chromosomes causing a shift in the order of nucleotides sequences include:

Deletions - involving the loss of part of a chromosome.

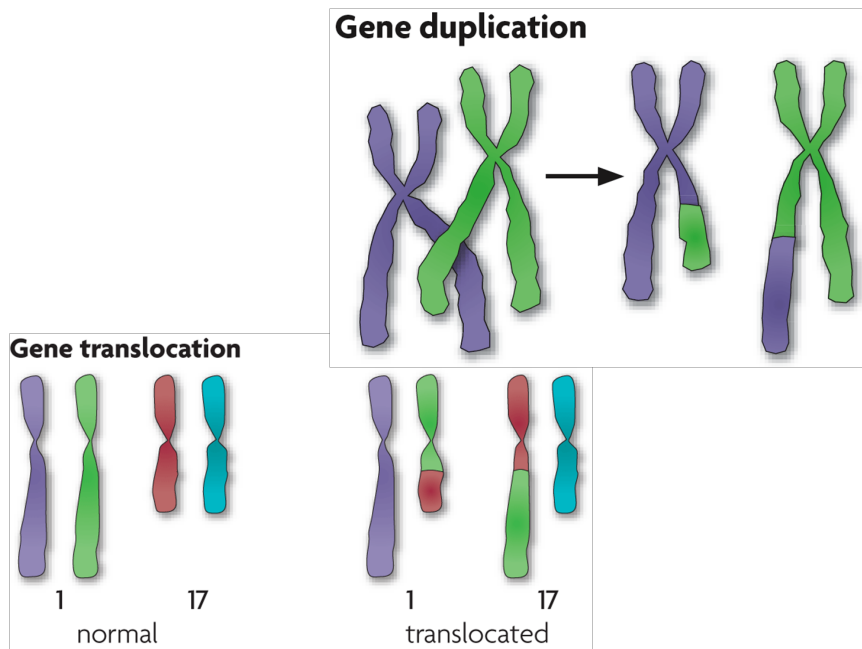
***Duplications** - a segment of a chromosome is repeated.

Inversions - when a part of a chromosome becomes reversed of its usual direction.

***Translocation** - a part of one chromosome breaks off and attaches to another.

-These types of mutations occur more often in **plants; and are deadly to animals**.

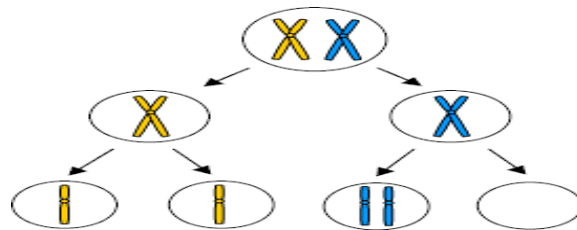
-Such mutations affect the distribution of genes to the gametes during **meiosis**.



2) Whole chromosomes, individually or in sets.

- ***Nondisjunction*** - a type of mutation that involves whole chromosomes. When ***homologs fail to separate during meiosis***.

-Depending on the gamete fertilized there are ***more or less*** the normal number of chromosomes present in an organisms cell.



3. Mutations may or may not affect phenotype.

-**Chromosomal mutations** tend to have a **big effect**.

-Some **gene mutations** change **phenotype**.

-A mutation may cause a **premature stop** codon.

-A mutation may change **protein shape** or **the active site**.

-A mutation may change **gene regulation**.

III. Causes of Mutations

A. 1. **Replication** errors can cause **mutations**.

2. **Mutagens**, such as UV ray and chemicals, can cause mutations.

3. Some **cancer drugs** use mutagenic properties to kill cancer cells.

B. Repair of DNA

1) The enzyme, **DNA Polymerase**, proofreads the DNA and replace incorrect nucleotides with correct nucleotides.

2) These repair mechanisms work well, but are **not perfect**. So, some nucleotides remain **uncorrected**.

