

## 12-4 Mutations

Mutations are changes in the genetic material.

### Kinds of Mutations

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

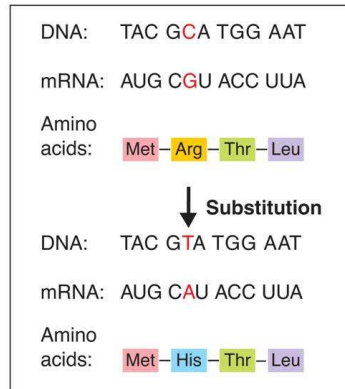
Mutations that produce changes in whole chromosomes are known as **chromosomal mutations**.

### Gene Mutations

Gene mutations involving a change in one or a few nucleotides are known as **point mutations** because they occur at a single point in the DNA sequence.

Point mutations include substitutions, insertions, and deletions.

Substitutions usually affect no more than a single amino acid.



The effects of insertions or deletions are more dramatic.

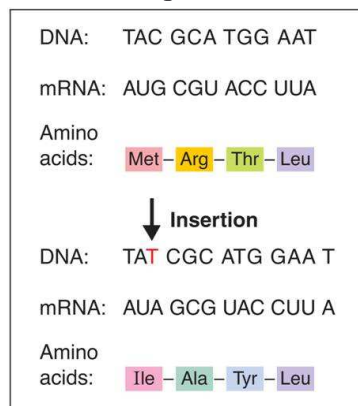
The addition or deletion of a nucleotide causes a shift in the grouping of codons.

Changes like these are called **frameshift mutations**.

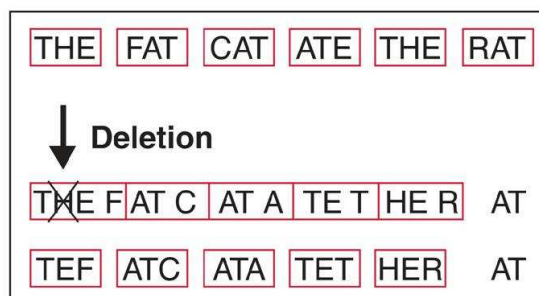
Frameshift mutations may change every amino acid that follows the point of the mutation.

Frameshift mutations can alter a protein so much that it is unable to perform its normal functions.

In an insertion, an extra base is inserted into a base sequence.



In a deletion, the loss of a single base is deleted and the reading frame is shifted.



## Chromosomal Mutations

Chromosomal mutations involve changes in the number or structure of chromosomes. Chromosomal mutations include deletions, duplications, inversions, and translocations. Deletions involve the loss of all or part of a chromosome.



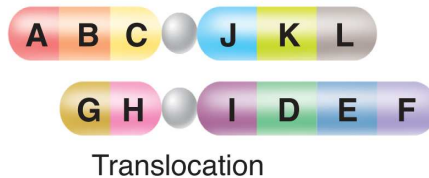
Duplications produce extra copies of parts of a chromosome.



Inversions reverse the direction of parts of chromosomes.



Translocations occur when part of one chromosome breaks off and attaches to another.



## Significance of Mutations

Many mutations have little or no effect on gene expression.

Some mutations are the cause of genetic disorders.

Beneficial mutations may produce proteins with new or altered activities that can be useful.

**Ploidy** is the condition in which an organism has extra sets of chromosomes.