

# 12-4 Mutations



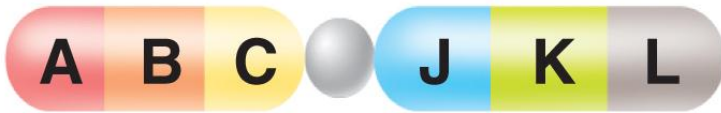
Original chromosome



Deletion



Duplication



Translocation



Inversion



**Mutations are changes in the genetic material.**



## Gene Mutations

### Point mutations

→ involving one or a few nucleotides

→ include substitutions, insertions, and deletions.

Substitutions usually affect no more than a single amino acid.

DNA: TAC GCA TGG AAT

mRNA: AUG CGU ACC UUA

Amino acids: Met — Arg — Thr — Leu

↓ Substitution

DNA: TAC GTA TGG AAT

mRNA: AUG CAU ACC UUA

Amino acids: Met — His — Thr — Leu

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

→ called **frameshift mutations**.

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

→ 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.

DNA: TAC GCA TGG AAT

mRNA: AUG CGU ACC UUA

Amino acids: Met – Arg – Thr – Leu

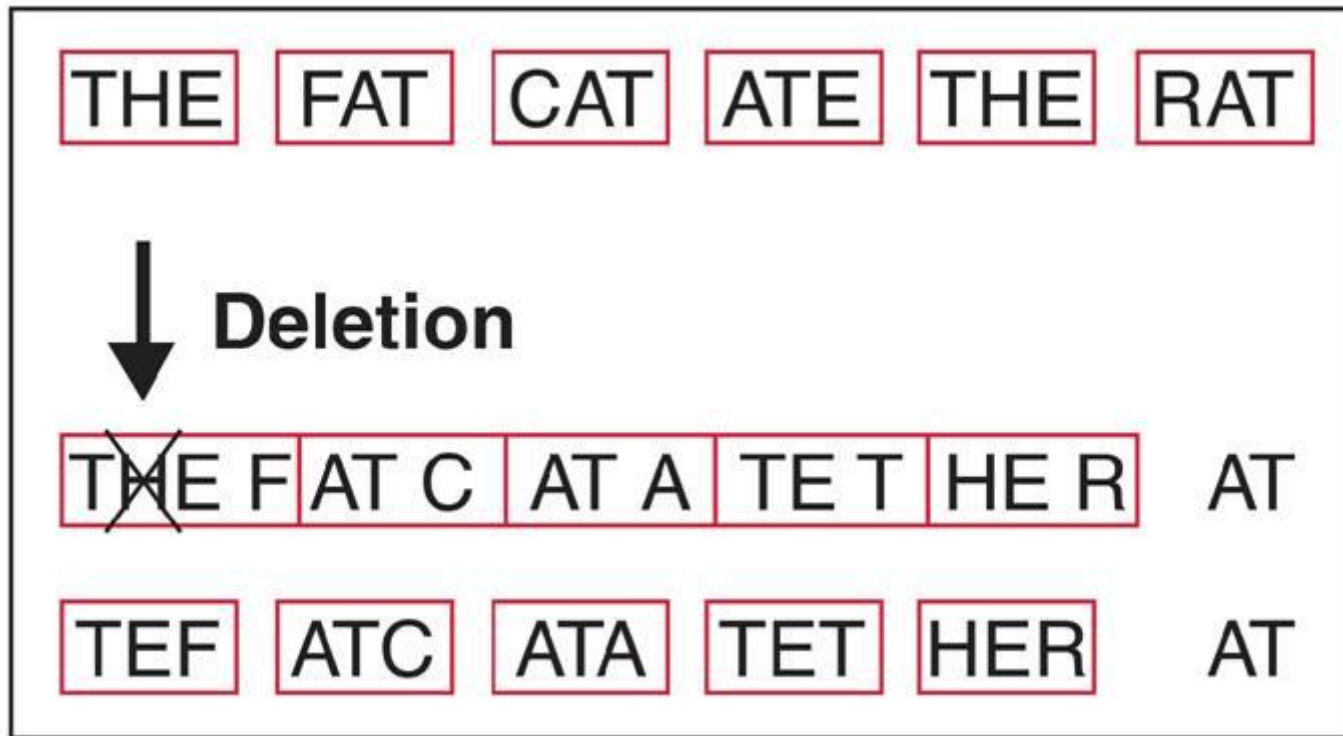
↓ Insertion

DNA: TAT CGC ATG GAA T

mRNA: AUA GCG UAC CUU A

Amino acids: Ile – Ala – Tyr – Leu

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



## Chromosomal Mutations

- involve changes in the number or structure of chromosomes.
- include deletions, duplications, inversions, and translocations.



Deletions involve the loss of all or part of a chromosome.



Original chromosome



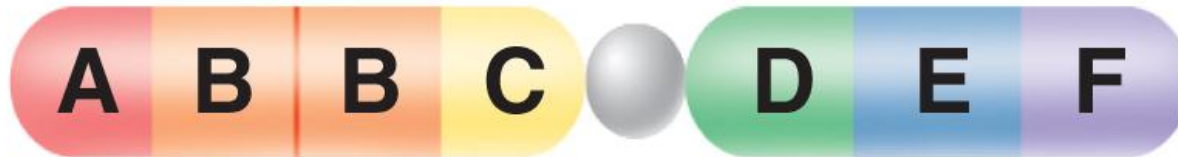
Deletion



Duplications produce extra copies of parts of a chromosome.



Original chromosome



Duplication

Inversions reverse the direction of parts of chromosomes.



Original chromosome



Inversion

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



Original chromosome



Translocation

# 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.

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

# 12-4 Section QUIZ

Continue to:

**Section QUIZ**

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## 12-4 Section QUIZ

1

A mutation in which all or part of a chromosome is lost is called a(an)

a. duplication.

A

b. deletion.

c. inversion.

d. point mutation.



## 12-4 Section QUIZ

**2** A mutation that affects every amino acid following an insertion or deletion is called a(an)

- A** a. frameshift mutation.
- b. point mutation.
- c. chromosomal mutation.
- d. inversion.

## 12-4 Section QUIZ

**3** A mutation in which a segment of a chromosome is repeated is called a(an)

a. deletion.

b. inversion.

**A** c. duplication.

d. point mutation.

## 12-4 Section QUIZ

**4** The type of point mutation that usually affects only a single amino acid is called

- a. a deletion.
- b. a frameshift mutation.
- c. an insertion.

**A** d. a substitution.

## 12-4 Section QUIZ

- 5** When two different chromosomes exchange some of their material, the mutation is called a(an)
- a. inversion.
  - b. deletion.
  - c. substitution.
  - A** d. translocation.

**END OF SECTION**