

12-4 Mutations



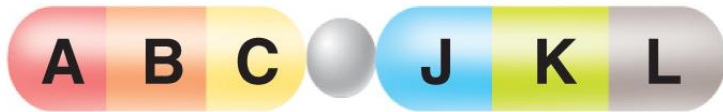
Original chromosome



Deletion



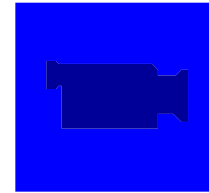
Duplication



Translocation



Inversion



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.

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 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.

DNA: TAC GCA TGG AAT

mRNA: AUG CGU ACC UUA

Amino acids:

Met – Arg – Thr – Leu



Insertion

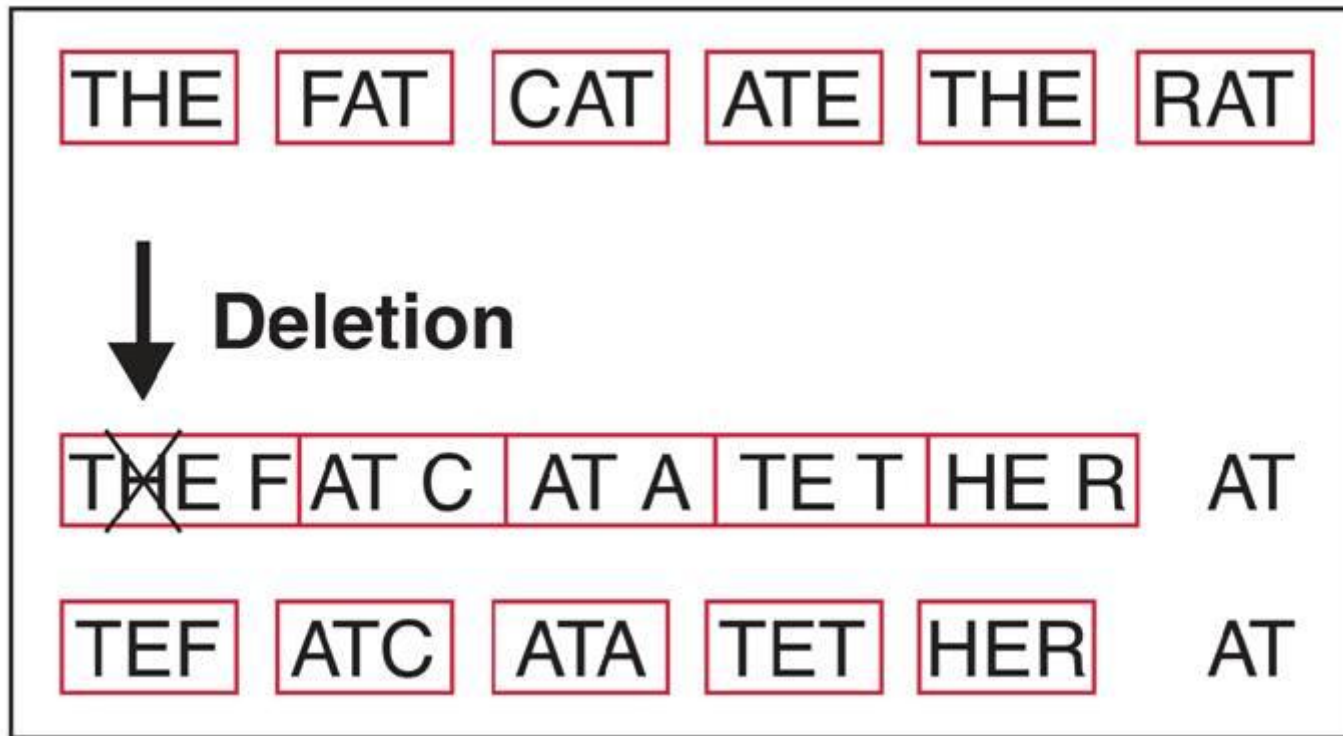
DNA: TAT CGC ATG GAA T

mRNA: AUA GCG UAC CUU A

Amino acids:

Ile – Ala – Tyr – Leu

In a deletion, 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.



Original chromosome



Deletion

Duplications produce extra copies of parts of a chromosome.



Original chromosome

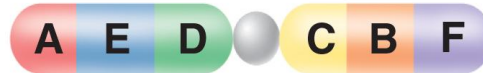


Duplication

Inversions reverse the direction of parts of chromosomes.



Original chromosome



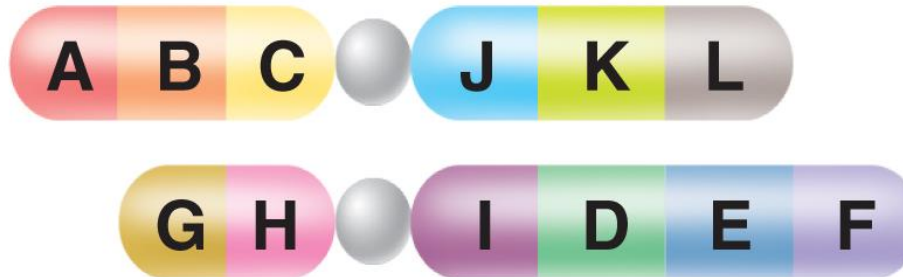
Inversion



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.

Polyploid Karyotype

Trisomy 21



12-4 Section QUIZ

Continue to:

Section QUIZ

- or -

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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.
- b. deletion.
- c. inversion.
- d. point mutation.

2

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

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.
- c. duplication.
- d. point mutation.

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.
- d. a substitution.

5 When two different chromosomes exchange some of their material, the mutation is called a(an)

- a. inversion.
- b. deletion.
- c. substitution.
- d. translocation.