

## 13.3 Mutations

### Lesson Objectives

-  Define mutations and describe the different types of mutations.
-  Describe the effects mutations can have on genes.

### Lesson Summary

**Types of Mutations** **Mutations** are heritable changes in genetic information. There are two categories of mutations: gene mutations and chromosomal mutations.

- ▶ Gene mutations produce changes in a single gene. **Point mutations** involve only one or a few nucleotides. Substitutions, insertions, and deletions are all types of point mutations.
  - In a substitution, one base is changed to a different base, which may affect only a single amino acid and have no effect at all.
  - In insertions and deletions, one base is inserted or removed from the DNA sequence. Insertions and deletions are 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 mutation and can have dramatic effects on the organism.
- ▶ Chromosomal mutations produce changes in the number or structure of chromosomes. They include deletions, duplications, inversions, and translocations.
  - Deletion involves the loss of all or part of a chromosome.
  - Duplication produces an extra copy of all or part of a chromosome.
  - Inversion reverses the direction of parts of a chromosome.
  - Translocation occurs when part of one chromosome breaks off and attaches to another.

**Effects of Mutations** Genetic material can be altered by natural events or by artificial means. Errors can be made during replication. Environmental conditions may increase the rate of mutation. **Mutagens** are chemical or physical agents in the environment that cause mutations.

The effects of mutations on genes vary widely:

- ▶ Some mutations have little or no effect.
- ▶ Some mutations produce beneficial variations. One example is **polyploidy** in plants, in which an organism has extra sets of chromosomes. Polyploid plants are often larger and stronger than diploid plants. Mutations can also produce proteins with new or altered functions that can be useful to organisms in different or changing environments.
- ▶ Some mutations negatively disrupt gene function or dramatically change protein structure. Genetic disorders such as sickle cell disease can result.

## Types of Mutations

For Questions 1–8, match the term with its definition.

**Definition**

- \_\_\_\_\_ 1. The change of one base to another in a DNA sequence
- \_\_\_\_\_ 2. A change in one or a few nucleotides that occur at a single point in the DNA sequence
- \_\_\_\_\_ 3. Part of one chromosome breaks off and attaches to another
- \_\_\_\_\_ 4. A heritable change in genetic information
- \_\_\_\_\_ 5. A mutation that produces an extra copy of all or part of a chromosome
- \_\_\_\_\_ 6. A chromosomal mutation that reverses the direction of parts of a chromosome
- \_\_\_\_\_ 7. A kind of mutation that can change every amino acid that follows the point of mutation
- \_\_\_\_\_ 8. The addition of a base to the DNA sequence

**Term**

- A. mutation
- B. substitution
- C. point mutation
- D. frameshift mutation
- E. insertion
- F. translocation
- G. inversion
- H. duplication

9. Complete the table to describe the processes and outcomes of the different types of gene (point) mutations.

Type	Description	Outcome
Substitution		
Insertion		
Deletion		

10. Deletion can happen as a gene mutation or as a chromosomal mutation. What is the difference?

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## Effects of Mutations

For Questions 10–17, write the letter of the correct answer on the line at the left.

- \_\_\_\_\_ 10. The cellular machinery that replicates DNA inserts an incorrect base
- A. most of the time.
  - B. about half the time.
  - C. roughly once in every million bases.
  - D. roughly once in every 10 million bases.
- \_\_\_\_\_ 11. Small changes in genes
- A. disappear quickly.
  - B. gradually accumulate over time.
  - C. prevent the next generation from developing.
  - D. do not affect future generations.
- \_\_\_\_\_ 12. A possible mutagen is
- A. an anticodon.
  - B. translocation.
  - C. hemoglobin.
  - D. ultraviolet light.
- \_\_\_\_\_ 13. What happens when cells cannot repair the damage caused by a mutagen?
- A. The DNA base sequence changes permanently.
  - B. The DNA base sequence is not affected.
  - C. The organism is not affected.
  - D. The organism is affected temporarily.
- \_\_\_\_\_ 14. Which of the following most accurately summarizes the effects of mutations on living things?
- A. Most mutations are harmful, but some have little effect.
  - B. Many mutations have little or no effect, but some can be harmful or beneficial.
  - C. Most mutations are beneficial and a few are harmful.
  - D. About half of mutations are beneficial and half are harmful.
- \_\_\_\_\_ 15. Mutations are important to the evolution of a species because they
- A. happen over the long period of time that evolution requires.
  - B. cut out and replace damaged or useless genes.
  - C. are a source of genetic variability.
  - D. accelerate the transcription rate of DNA.
- \_\_\_\_\_ 16. Cancer is the product of a mutation that
- A. causes the uncontrolled growth of cells.
  - B. changes the structure of hemoglobin in the blood.
  - C. brings about stunted growth and severe pain.
  - D. causes a translocation in a pair of chromosomes.

- \_\_\_\_\_ 17. Polyploidy is the condition in which
- A. a piece of a chromosome breaks off and reattaches to another chromosome.
  - B. an organism has an extra set of chromosomes.
  - C. a mutagen speeds the mutation rate.
  - D. an insect develops a resistance to a pesticide.
18. In the space below, draw an example of a normal blood cell and an example of a sickle cell.



**Apply the Big idea**

19. A gene that codes for one of the polypeptide chains of the blood protein hemoglobin lies on chromosome 11 in humans. A substitution mutation in that gene causes the amino acid valine to be incorporated into hemoglobin in a place where glutamic acid would normally lie. The result is sickle cell disease. Explain how a change in a single base in DNA can bring about such a serious disorder.

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