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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.

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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 11–18, write the letter of the correct answer on the line at the left.

_____ **11.** 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.

_____ **12.** 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.

_____ **13.** A possible mutagen is

A. an anticodon.

B. translocation.

C. hemoglobin.

D. ultraviolet light.

_____ **14.** 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.

_____ **15.** 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.

_____ **16.** 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.

_____ **17.** 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.

_____ **18.** 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.

19. In the space below, draw an example of a normal blood cell and an example of a sickle cell.

Apply the Big idea

20. 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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13.4 Gene Regulation and Expression

Lesson Objectives

- Describe gene regulation in prokaryotes.
- Explain how most eukaryotic genes are regulated.
- Relate gene regulation to development in multicellular organisms.

Lesson Summary

Prokaryotic Gene Regulation Prokaryotes do not need to transcribe all of their genes at the same time. They can conserve energy and resources by regulating their activities, producing only those genes necessary for the cell to function. In prokaryotes, DNA-binding proteins regulate genes by controlling transcription. An **operon** is a group of genes that are regulated together. An example is the *lac* operon in the bacterium *E. coli*:

- This group of three genes must be turned on together before the bacterium can use lactose as food.
- When lactose is not present, the DNA-binding protein called *lac* repressor binds to a region called the **operator**, which switches the *lac* operon off.
- When lactose binds to the repressor, it causes the repressor to fall off the operator, turning the operon on.

Eukaryotic Gene Regulation Transcription factors are DNA-binding proteins. They control the expression of genes in eukaryotes by binding DNA sequences in the regulatory regions. Gene promoters have multiple binding sites for transcription factors, each of which can influence transcription.

- Complex gene regulation in eukaryotes makes cell specialization possible.
- The process by which microRNA (miRNA) molecules stop mRNA molecules from passing on their protein-making instructions is **RNA interference (RNAi)**.
- RNAi technology holds the promise of allowing scientists to turn off the expression of genes from viruses and cancer cells, and it may provide new ways to treat and perhaps even cure diseases.

Genetic Control of Development Regulating gene expression is especially important in shaping the way a multicellular organism develops. Gene regulation helps cells undergo **differentiation**, becoming specialized in structure and function. Master control genes are like switches that trigger particular patterns of development and differentiation in cells and tissues.

- **Homeotic genes** are master control genes that regulate organs that develop in specific parts of the body.
- **Homeobox genes** share a similar 130-base DNA sequence called homeobox. They code for transcription factors that activate other genes that are important in cell development and differentiation in certain regions of the body.
- **Hox genes** are a group of homeobox genes that tell the cells of the body how to differentiate as the body grows.

Environmental factors can also affect gene expression.

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Prokaryotic Gene Regulation

1. How do prokaryotes conserve energy?

2. How do DNA-binding proteins in prokaryotes regulate genes?

3. What is an operon?

4. What is in the *lac* operon in *E. coli*?

5. What is the function of the genes in the *lac* operon of *E. coli*?

6. What turns the *lac* operon off?

7. How does a repressor protein turn off the *lac* operon?

8. How does lactose turn on the *lac* operon?

9. Complete the table to describe the role of each regulatory region or molecule in the operation of the *lac* operon.

Regulatory Region or Molecule	What It Does
Repressor protein	
Operator	
RNA polymerase	
Lactose	

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Eukaryotic Gene Regulation

10. In what two ways is gene regulation in eukaryotes different from gene regulation in prokaryotes?

a. _____

b. _____

_____

11. What is a TATA box? What does a TATA box do?

_____

_____

_____

12. What are transcription factors and what do they do?

_____

_____

13. Explain how gene regulation makes cell specialization possible.

_____

_____

14. What is microRNA and how is it related to mRNA?

_____

_____

15. Explain how the process of RNA interference works.

_____

_____

_____

_____

_____

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Genetic Control of Development

For Questions 16–23, write the letter of the correct answer on the line at the left.

_____ **16.** As an embryo develops, different sets of genes are regulated by

A. mRNA and *lac* repressors.
B. operons and operators.
C. transcription factors and repressors.
D. promoters and operators.

_____ **17.** The process through which cells become specialized in structure and function is

A. transcription.
B. gene expression.
C. differentiation.
D. RNA interference.

_____ **18.** Homeotic genes are

A. regulator genes that bind to operons in prokaryotes.
B. master control genes that regulate organs that develop in specific parts of the body.
C. parts of the silencing complex that regulates gene action through RNA interference.
D. base sequences complementary to sequences in microRNA.

_____ **19.** What role do homeobox genes play in cell differentiation?

A. They code for transcription factors that activate other genes important in cell development and differentiation.
B. They block certain gene expression.
C. They cut double-stranded loops into microRNA.
D. They attach to a cluster of proteins to form a silencing complex, which binds to and destroys certain RNA.

_____ **20.** In flies, the group of homeobox genes that determines the identities of each segment of a fly's body is the group known as

A. silencing complexes.
B. promoters.
C. operators.
D. Hox genes.

_____ **21.** Clusters of Hox genes are found in

A. flies only.
B. flies and frogs only.
C. plants only.
D. nearly all animals.

_____ **22.** The "switches" that trigger particular patterns of development and differentiation in cells and tissues are

A. mRNA molecules.
B. master control genes.
C. silencing complexes.
D. Dicer enzymes.

_____ **23.** Metamorphosis is

A. a series of transformations from one life stage to another.
B. the master switch that triggers development and differentiation.
C. the product of interactions among homeotic genes.
D. the process by which genetic information is passed from one generation to the next.

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24. Environmental factors can influence gene expression. Fill in the table below to show how organisms respond to conditions in their environment.

	Environmental Factor Influencing Gene Expression	How the Organism Responds
***E. coli* with limited food supply**	nutrient availability	
A tadpole in a drying pond		

Apply the Big Idea

25. Many research studies have shown that different species may possess some of the exact same genes but show vastly different traits. How can that happen?

_____

_____

_____

_____

_____

