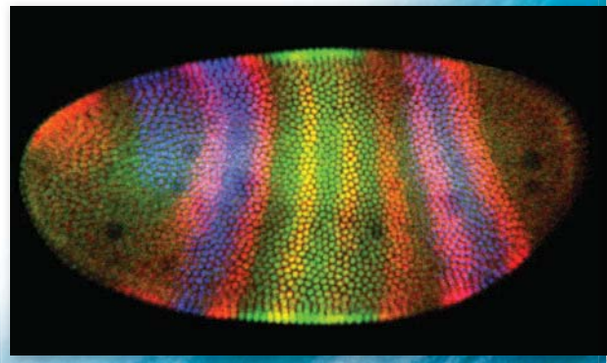
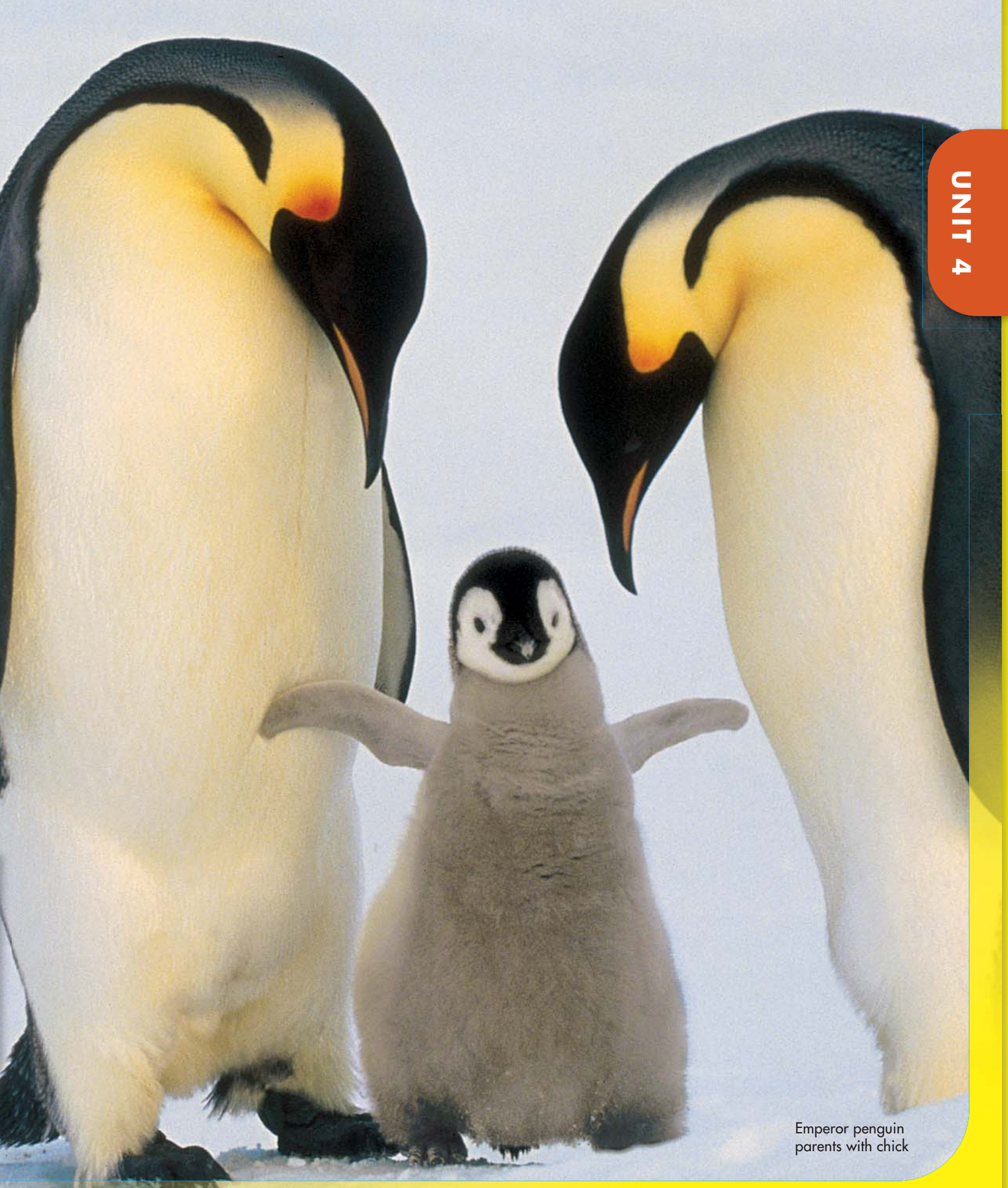
UNIT 4 Heredity

Eggs of the red-eyed tree frog stuck to the underside of a leaf



Fruit fly embryo, marked to show pattern of genes being expressed



Emperor penguin parents with chick

DISCOVERIES IN SCIENCE

Heredity and Genetics

1865

Gregor Mendel publishes the results of his studies of genetic inheritance in pea plants. Although his work is not widely known until much later, Mendel is remembered as the founder of the science of genetics.

Gregor Mendel

1879

After staining cells with Perkins dye and viewing them under a microscope, Walter Fleming identifies chromatin in cells. Soon after, he observes and describes all stages of mitosis, using terms such as *metaphase, anaphase and telophase.*

1905

Nettie Maria Stephens describes how human gender is determined by the X and Y chromosomes.

Nettie Stevens

1909

The Elements of Heredity, by Wilhelm Johannsen, a Danish biologist, is revised and translated into German. In the book, Johannsen develops many of the concepts of modern genetics, particularly phenotype and genotype. This book becomes a founding text of genetics.

1913

Alfred Henry Sturtevant, an undergraduate student at Columbia University, determines the relative location of genes on a fruit fly chromosome. He publishes a genetic map showing the order of genes and their relative distance from each other.

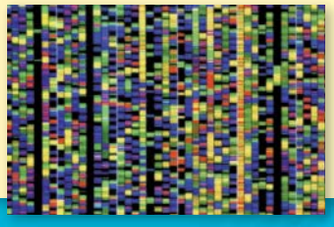
1915

Thomas Hunt publishes the book *Mechanism of Mendelian Heredity,* which explains the phenomenon of sex-linked traits observed in fruit flies.

***Drosophila melanogaster* (fruit fly)**

1989

Francis Collins and Lap-Chee Tsui identify a mutant version of a gene on chromosome 7 that causes cystic fibrosis. Discovery of the gene leads to the development of tests that can determine whether potential parents are carriers of the gene.

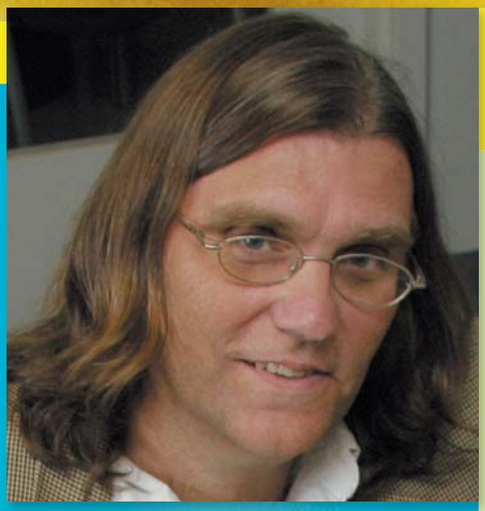


Genetic sequences on a computer screen

2003

The Human Genome Project is completed. Research teams around the world collaborated to identify all genes and decode the sequence of all DNA in human cells.

Albino peacock



BIOLOGY CAREER

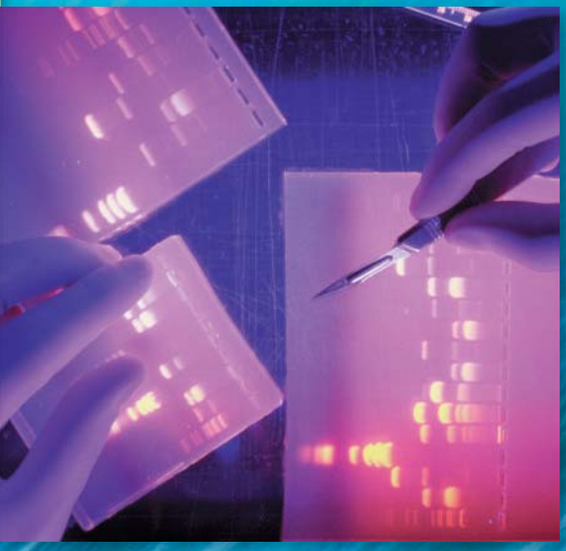
Genetics Researcher

Rob Desalle

Rob DeSalle is a curator in the Division of Invertebrate Zoology at the American Museum of Natural History in New York City. His current research focuses on molecular evolution in a variety of organisms, including pathogenic bacteria and insects.

DeSalle studies molecular evolution through comparative genomics, which is the study of similarities and differences between the genomes of various species or strains within species. Comparing the genomes of species can help determine how the species are related.

DeSalle also helped found the Conservation Genetics Program at the American Museum of Natural History. This program uses the tools of molecular genetics to help protect wildlife around the world. For example, DeSalle helped develop a genetic test to determine if caviar sold in the United States was illegally harvested from endangered species of sturgeon in the Caspian Sea.



Genetic analysis by gel electrophoresis

UNIT 4

Chapter Planner

14 Genes in Action

		Standards	Teach Key Ideas
CHAPTER OPENER, pp. 316–317	15 min.	*National Science Education Standards*	
SECTION 1 Mutation and Genetic Change, pp. 319–324 › Mutation: The Basis of Genetic Change › Several Kinds of Mutations › Effects of Genetic Change › Large-Scale Genetic Change	45 min.	LSCell 3, LSGene 3, LSEvol 1, UCP5, SI1	**Bellringer Transparency** **Transparencies** C26 The Major Types of Mutations **Visual Concepts** Mutation • Types of Gene Mutations • Genetic Disorder • Comparing X-Linked and Sex-Influenced Traits • Sickle-Cell Anemia • Hemophilia • Huntington's Disease • Disorders Due to Nondisjunction
SECTION 2 Regulating Gene Expression, pp. 325–329 › Complexities of Gene Expression › Gene Regulation in Prokaryotes › Gene Regulation in Eukaryotes › The Many Roles of Proteins	90 min.	LSCell 2, LSCell 4, LSCell 6, LSMat 4, UCP3, SPSP1	**Bellringer Transparency** **Visual Concepts** Operon • Repression of Transcription in the *lac* Operon • Regulator Gene • Activation of Transcription in the *lac* Operon • Enhancers for Control of Gene Expression • Comparing Introns and Exons
SECTION 3 Genome Interactions, pp. 330–337 › Genomes and the Diversity of Life › Moving Beyond Chromosomes › Multicellular Development and Aging	45 min.	LSCell 6, LSEvol 1, LSEvol 3, UCP4, SPSP1, SPSP5, HNS3	**Bellringer Transparency**
			See also PowerPoint® Resources

Chapter Review and Assessment Resources

- SE Super Summary, p. 338
- SE Chapter Review, p. 339
- SE Standardized Test Prep, p. 341
- Review Resources
- Chapter Tests A and B
- Holt Online Assessment

CHAPTER FastTrack

To shorten instruction due to time limitations, eliminate the Skills Practice Lab.

Basic Learners

- TE Chromosomal Mutation Models, p. 321
- TE Ultraviolet Radiation and Sunscreen, p. 322
- TE Vocabulary Refresher, p. 323
- TE Exon Splicing, p. 328
- TE Distinguishing Data, p. 331
- Directed Reading Worksheets*
- Active Reading Worksheets*
- Lab Manuals, Level A*
- Study Guide* ■
- Note-taking Workbook*
- Special Needs Activities and Modified Tests*

Advanced Learners

- TE Advantage of Sickle Cell, p. 320
- TE Down Syndrome, p. 323
- TE Nobel Winners in Genetics, p. 327
- TE Endosymbiont Theory, p. 331
- TE Retroviruses, p. 332
- TE The Mitochondrial DNA Question, p. 335
- Critical Thinking Worksheets*
- Concept Mapping Worksheets*
- Science Skills Worksheets*
- Lab Datasheets, Level C*

Key

SE Student Edition
TE Teacher's Edition
Chapter Resource File
Workbook
Transparency
CD or CD-ROM
* Datasheet or blackline master available
■ Also available in Spanish

All resources listed below are also available on the Teacher's One-Stop Planner.

Why It Matters	Hands-On	Skills Development	Assessment
Build student motivation with resources about high-interest applications.	SE **Inquiry Lab** Where Is the Protein, p. 317*■	TE **Reading Toolbox** Assessing Prior Knowledge, p. 316 SE **Reading Toolbox,** p. 318	
TE **Sickle Cell Disease,** p. 320 TE **Radiation and Cancer,** p. 322	SE **Quick Lab** Make a Model of Mutations, p. 321*■	SE **Reading Toolbox** Finding Examples, p. 320 TE **Reading Toolbox** Finding Examples, p. 320	SE **Section Review** TE **Formative Assessment** **Spanish Assessment***■ **Section Quiz**■
TE **Demonstration** Using an Analogy, p. 325 TE **Lactose Intolerance,** p. 326 TE **Demonstration** Modeling the Regulation Process, p. 327 TE **Demonstration** Splicing, p. 328	SE **Quick Lab** A Model of Introns and Exons, p. 328*■ SE **Skills Practice Lab** Protein Detection, p. 336*■	SE **Reading Toolbox** Sample Comparison Table, p. 326 TE **Reading Toolbox** Sample Comparison Table, p. 326	SE **Section Review** TE **Formative Assessment** **Spanish Assessment***■ **Section Quiz**■
TE **Demonstration** Relative Size of Genomes, p. 331 TE **A Passion for Research,** p. 332 SE **Forensic Genealogy,** p. 335	**Skills Practice Lab** Introduction to Agarose Gel Electrophoresis*■ **Exploration Lab** DNA Whodunnit*■	SE **Reading Toolbox** Prefixes, p. 333 TE **Reading Toolbox** Prefixes, p. 333 TE **Reading Toolbox** Visual Literacy, p. 335	SE **Section Review** TE **Formative Assessment** **Spanish Assessment***■ **Section Quiz**■
	See also Lab Generator		**See also Holt Online Assessment Resources**

Resources for Differentiated Instruction

English Learners

TE Vocabulary, p. 326
TE Exon Splicing, p. 328
Directed Reading Worksheets*
Active Reading Worksheets*
Lab Manuals, Level A*
Study Guide*■
Note-taking Workbook*
Multilingual Glossary

Struggling Readers

TE Mutation, Mutant, or Mutagen?, p. 319
TE Vocabulary Refresher, p. 323
TE Vocabulary, p. 326
TE Distinguishing Data, p. 331
TE Key-Term Fold, p. 333
Directed Reading Worksheets*
Active Reading Worksheets*
Lab Manuals, Level A*
Study Guide*
Note-taking Workbook*
Special Needs Activities and Modified Tests*

Special Education Students

TE Chromosomal Mutation Models, p. 321
Directed Reading Worksheets*
Active Reading Worksheets*
Lab Manuals, Level A*
Study Guide*■
Note-taking Workbook*
Special Needs Activities and Modified Tests*

Alternative Assessment

Science Skills Worksheets*
Section Quizzes*■
Chapter Tests A, B, and C*■

Chapter 14

Overview

The purpose of this chapter is to expand on the roles of genes—how they can change, how their expression is regulated, and how they control development and lifespan of the cell. The universality of the genetic code is reiterated by comparing genomes and seeing interactions at the genetic level.

READING TOOLBOX

Assessing Prior Knowledge Students should be familiar with the following concepts:
- heredity
- structure of genes
- gene expression
- triplet code for amino acids

Visual Literacy Ask students whether extra legs provide an adaptive advantage for the frogs in the photo. (Students may think extra legs would be an advantage. However, the extra legs hamper the agility of these frogs. Thus, the frogs pictured are more likely targets for predators.)

Chapter 14

Genes in Action

Preview

Why It Matters

Knowing the genetic code is not enough to understand how genes work. To understand our own bodies, we must study thousands of genes, proteins, and other molecules that interact as our bodies grow and develop.

These frogs have extra legs! When many mutated or deformed organisms are found in one area, scientists want to find out why.

Chapter Correlations

National Science Education Standards

LSCell 2 Most cell functions involve chemical reaction.

LSCell 3 Cells store and use information to guide their functions.

LSCell 4 Cell functions are regulated.

LSCell 6 Cells can differentiate and form complete multicellular organisms.

LSGene 3 Changes in DNA (mutations) occur spontaneously at low rates.

LSEvol 1 Species evolve over time.

LSEvol 3 Natural selection and its evolutionary consequences provide a scientific explanation for the fossil record of ancient life forms as well as for the striking molecular similarities observed among the diverse species of living organisms.

LSMat 4 The complexity and organization of organisms accommodates the need for obtaining, transforming, transporting, releasing, and eliminating the matter and energy used to sustain the organism.

UCP3 Change, constancy, and measurement

UCP4 Evolution and equilibrium

UCP5 Form and function

SI1 Abilities necessary to do scientific inquiry

SPSP1 Personal and community health

SPSP5 Natural and human-induced hazards

HNS3 Historical perspectives

InquiryLab

15 min

Where Is the Protein?

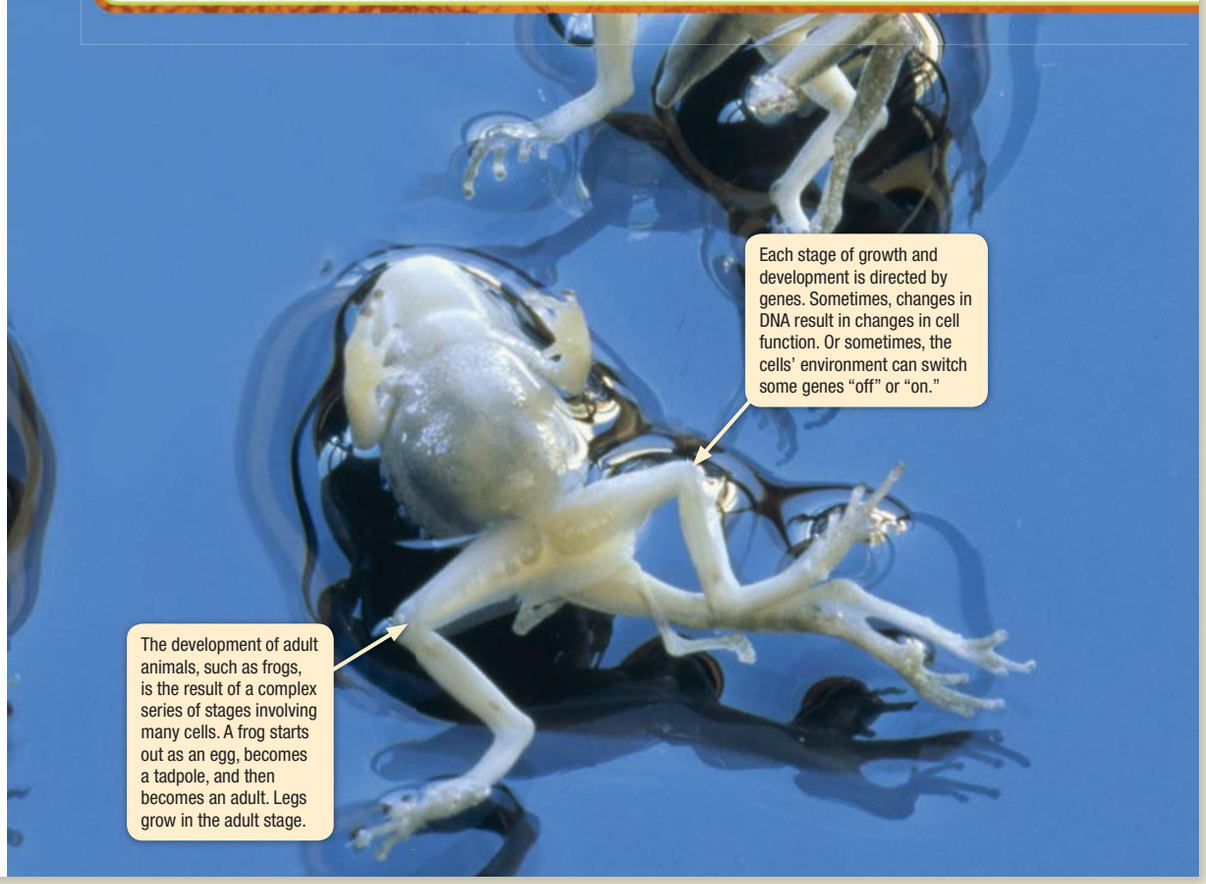
Protein test strips are inexpensive and easy-to-use measuring tools. Sold in local pharmacies, these strips are purchased by individuals who must monitor the concentration of protein in their urine. The strips can also be used to confirm the presence of protein in various foods.

Procedure

1. Work with a partner. Label **five small cups** "A," "B," "C," "D," and "E."
2. Use a **mortar and pestle** to crush **20 lentil beans.** Place the crushed beans into cup A.
3. Place **1 g of instant oatmeal** into cup B.
4. Pour **10 mL of water** into cups A and B. Swirl each cup to mix its contents.
5. Pour **10 mL of water** into cup C.
6. Pour **10 mL of milk** into cup D.
7. Pour **10 mL of fruit juice** into cup E.
8. Obtain **five protein test strips.** Follow the label instructions to detect and measure the presence of protein in each cup.

Analysis

1. **Identify** the cups in which protein was present.
2. **Identify** the cups that had the most and least amounts of protein.
3. **Identify** the control group in this experiment. Explain its purpose.



Each stage of growth and development is directed by genes. Sometimes, changes in DNA result in changes in cell function. Or sometimes, the cells' environment can switch some genes "off" or "on."

The development of adult animals, such as frogs, is the result of a complex series of stages involving many cells. A frog starts out as an egg, becomes a tadpole, and then becomes an adult. Legs grow in the adult stage.

InquiryLab

Teacher's Notes Using a blender to pulverize the lentils will save time. Protein test strips are also known as urine reagent test strips. They are available locally at most pharmacies. Such strips can be used in other activities as a convenient way to measure pH or detect ketones and nitrates. Make sure students rinse the graduated cylinder between steps 6 and 7.

Materials

- small paper or plastic cups (5)
- mortar and pestle
- lentil beans (20)
- instant oatmeal, 1 g
- graduated cylinders, 10 or 25 mL
- water, 30 mL
- milk, 10 mL
- fruit juice, apple, 10 mL
- multiple test sticks or protein test sticks for urine analysis (5)

Answers to Analysis

1. Protein should be detected in cups A, B, and D.
2. Cup A should have the most protein. Cup B should have the least amount of protein.
3. Cup C, water, was the control. It was used to show the effect of a liquid that is unreactive on the strip.

READING TOOLBOX

Using Words

1. Accept answers that are synonymous with "cannot be changed."
2. Accept answers that suggest a change that allows something to "cross" over into another state.

Using Language

1. Accept reasonable answers. Students will likely offer physical features of themselves, animals, and possibly plants. Traits may include hair color, eye color, height, or flower color.
2. inherit, inheritable, inheritance, inherent, inherence, heritage, or heredity

Using Science Graphics

Sample answer:

	Lactose absent	Lactose present
Similarities	RNA polymerase at promoter	RNA polymerase at promoter
Differences	repressor attached	lactose binds to repressor, releasing it

READING TOOLBOX

These reading tools can help you learn the material in this chapter. For more information on how to use these and other tools, see **Appendix: Reading and Study Skills.**

Using Words

Prefixes A prefix is a word part that is attached to the beginning of a word. Prefixes add to the meaning of words. For example, the prefix *im-* means "not." So, *immovable* means "not able to be moved." This table shows some additional prefixes that you may see in this chapter.

Prefixes

Prefix	Meaning
in- or *im-*	not
mut-	change
trans-	cross
homeo-	the same

Your Turn Use the table to answer the questions that follow.

1. In your own words, define *immutable.*
2. What do you think the word *transmutation* means?

Using Language

Finding Examples When you are reading scientific explanations, finding examples can help you put a concept into practical terms. Thinking of your own examples will help you remember what you read.

Your Turn For each category of items below, brainstorm as many examples as you can think of that could fit into the category.

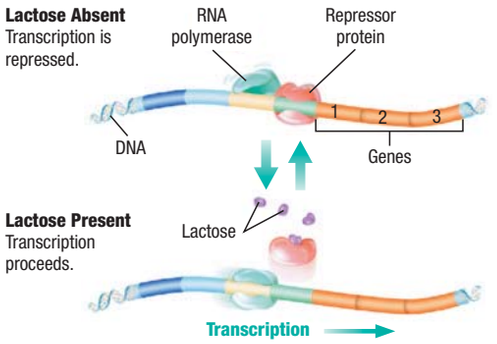
1. hereditary traits
2. words that include the word part *-her-.*

Using Science Graphics

Comparison Table A comparison table can help you understand what is happening in two similar situations. For example, this graphic shows the effect of lactose on specific genes in some prokaryotic cells. If you compare the two situations shown, you will find similarities and differences.

Your Turn Make a comparison table like the one shown here to compare what happens when lactose is present and when lactose is absent.

1. Draw a table with two columns.
2. Label the first column "Lactose absent."
3. Label the second column "Lactose present."
4. Use the graphic as a reference as you list the structures and events that are similar or different.



	Lactose absent	*Lactose present*
Similarities		
Differences		

Section 1

Mutation and Genetic Change

Key Ideas	Key Terms	Why It Matters
What is the origin of genetic differences among organisms? What kinds of mutations are possible? What are the possible effects of mutations? How can genetic change occur on a larger scale?	mutation nondisjunction polyploidy	Understanding mutation is key to understanding the differences among organisms over time.

In general, *mutation* simply means "change," and any organism that has changed from some previous or normal state can be called a *mutant*. So, a frog that has extra legs may be called a *mutant,* although the extra legs may or may not have a genetic cause.

Mutation: The Basis of Genetic Change

In genetics, a **mutation** is a change in the structure or amount of the genetic material of an organism. A genetic *mutant* is an individual whose DNA or chromosomes differ from some previous or normal state. **For the most part, genetic differences among organisms originate as some kind of genetic mutation.** Every unique allele of every gene began as a mutation of an existing gene.

Causes of Mutations Mutations occur naturally as accidental changes to DNA or to chromosomes during the cell cycle. Recall that enzymes repair most DNA that is mismatched during replication, but rarely, some DNA is not repaired. Other kinds of mistakes are possible, as you will learn. Also, the rate of mutation can be increased by some environmental factors. Such factors, called *mutagens,* include many forms of radiation and some kinds of chemicals.

Effects of Mutations Because of the way DNA is translated, a mutation can have many possible effects. A small change in DNA may affect just one amino acid in the protein that results from a gene. However, as you will see, other results are possible. A mutation may have no effect, or may harm or help in some way. The effect depends on where and when the mutation occurs. We notice mutations when they cause an unusual trait or disease, such as *sickle cell anemia,* shown in **Figure 1.** However, many mutations may go unnoticed.

Reading Check ***Where do new alleles come from?*** ***(See the Appendix for answers to Reading Checks.)***

mutation a change in the structure or amount of the genetic material of an organism

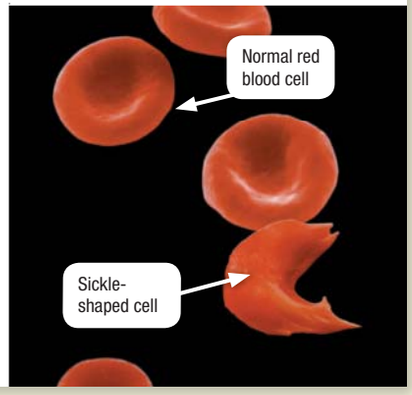


Figure 1 One out of 500 African Americans has sickle cell anemia, which is caused by a mutation in the gene that produces hemoglobin. Blood cells with the defective hemoglobin tend to bend, rupture, and get stuck.

Differentiated Instruction

Struggling Readers

Mutation, Mutant, or Mutagen? Students with reading difficulties often do not recognize the subtle differences between words. Review the meanings of *mutation, mutant* and *mutagen.* Ask students to come up with ways that will help them remember the differences. (Accept meaningful answers.) LS Verbal

Key Resources

Transparencies
C26 The Major Types of Mutations

Visual Concepts
Mutation
Types of Gene Mutations
Genetic Disorder
Comparing X-Linked and Sex-Influenced Traits
Sickle-Cell Anemia
Hemophilia
Huntington's Disease
Disorders Due to Nondisjunction

Section 1

Focus

This section describes the causes and effects of mutations. Students learn that mutations are classified as changes that occur during DNA replication, during gene expression, and within chromosomes during meiosis.

Bellringer
Use the Bellringer transparency to prepare students for this section.

Teach

Teaching Key Ideas

Movie Mutants Ask students for examples of mutant organisms they've seen in movies or cartoons. Ask them what characteristics were altered by mutation. (The most common response will be an increase in size or strength. Other responses will include humans with unusual animal qualities, such as gills.) Tell students that in this chapter, they will see that mutations are more common, and less obvious, than they might think.

Teaching Key Ideas

Identifying Terms Refer students to **Figure 1.** Explain that the *mutation* is a change in the gene for making hemoglobin. The *mutant* is the sickle-shaped cell. LS Visual

Teach, *continued*

READING TOOLBOX

Finding Examples Possible responses are the following:
- insertion: the cat sat e
- deletion: tec ata te
- nonsense: the cat (stop)

LS **Visual**

Teaching Key Ideas

Kinds of Mutations Have students look at **Figure 2** and identify the three original, correct amino acids. Next, look at the *silent mutation,* and identify the point mutation. (A for T) Why is this mutation silent? (It still codes for proline.) Next, look at the *missense mutation.* Why is it so named? (T for G results in a different amino acid, Glu.) Conclude with the *frameshift mutation.* Ask what has been inserted. (G) What is the result? (From this point, all the codons change.) LS **Visual**

go.hrw.com
interact online

Students can interact with "Kinds of Mutations" by going to go.hrw.com and typing in the keyword HX8GNXF2.

READING TOOLBOX

Finding Examples Use the phrase "the cat ate" to create examples of mutations. For example, a point mutation could change the letter *c* to *b* and would result in "the bat ate." The new phrase is also a missense mutation. Use the original phrase to make examples of an insertion, a deletion, and a nonsense mutation.

Several Kinds of Mutations

DNA and chromosomes are involved in many processes, so there are many kinds of mutations. Most mutations involve a misplacement of a nucleotide in a DNA segment. A mutation may change the results of a gene (when the gene is translated and transcribed), but not all mutations do so. **Different kinds of mutations are recognized as either changes in DNA or changes in the results of genes,** as shown in **Figure 2.**

Mutations as Changes in DNA During DNA replication, the wrong nucleotide may be paired or placed in a sequence.

Point Mutation A *point* mutation is a change of a single nucleotide in a sequence from one kind of base to another.

Insertion or Deletion Rarely, errors in replication can cause the *insertion* or *deletion* of one or more nucleotides in a sequence.

Mutations as Changes in Results of Genes Changes in a DNA sequence may affect the results of genes in many ways.

Silent Mutation A mutation is *silent* when it has no effect on a gene's function. Point mutations are often silent because the genetic code is redundant (each amino acid has multiple codons).

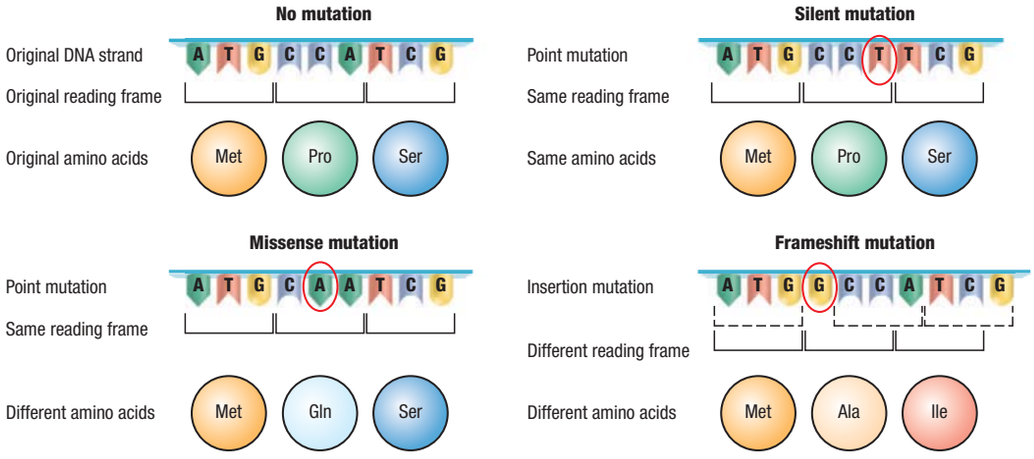
Missense Mutation A *missense* mutation results when a codon is changed such that the new codon codes for a different amino acid. This kind of mutation is also called a *replacement* mutation.

Frameshift Mutation Recall that the genetic code is "read" in "words" of three letters each (codons). The *reading frame* of a sequence depends on the starting point for reading. An insertion or deletion can shift the reading frame, or cause a *frameshift*. In this case, the remaining sequence may be "read" as different codons.

Figure 2 A mutation is a change, insertion, or deletion of one or more nucleotides in a gene. The change may or may not result in a different amino acid sequence within a protein.

go.hrw.com
interact online
Keyword: HX8GNXF2

Kinds of Mutations



Why It Matters

Sickle Cell Disease A point mutation in DNA that codes for normal hemoglobin and that results in CAT rather than CTT replaces valine for glutamine. The resulting hemoglobin causes the red blood cell (RBC) to deform to a sickle shape, which causes the effects of this disease. This example illustrates the large-scale effects of a mutation in just one nucleotide base.

Differentiated Instruction

Advanced Learners/GATE

Advantage of Sickle Cell Have students investigate the heterozygous advantage of the sickle cell trait from an evolutionary perspective.

LS **Logical**

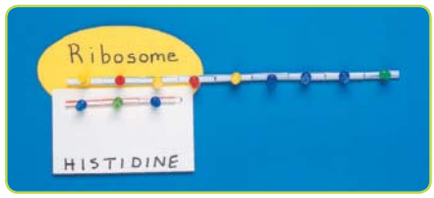
Hands-On

QuickLab

15 min

Make a Model of Mutations

You have learned about (and may have built models of) DNA replication and gene expression. Now, challenge yourself to build (or add to) a model that demonstrates each type of mutation described in this section.



Analysis

1. **List** each mutation type on **12 separate sheets of paper.** Work with a partner.
2. **Demonstrate** each mutation type by using **assorted materials** (or models that you have built previously).
3. **Draw** the "before" and "after" state for each mutation.
4. CRITICAL THINKING **Critiquing Models** Trade your drawings with another group. What is accurate and useful about their model? What could be improved? Write down your comments for the other group.

Nonsense Mutation A *nonsense* mutation results when a codon is changed to a "stop" signal. In this case, the resulting string of amino acids may be cut short, and the protein may fail to function.

More or Fewer Amino Acids If an insertion or deletion is a multiple of 3, the reading frame will be preserved. However, the protein that results may have a few more or less amino acids in it. An insertion or deletion of many codons is likely to disrupt the resulting protein's structure and function.

Chromosomal Mutations ❯ **In eukaryotic cells, the process of meiosis creates the chance of mutations at the chromosomal level.** Recall that during this process, chromosomes pair up and may undergo *crossover*. Usually, the result is an equal exchange of alleles between homologous chromosomes. But errors in the exchange can cause *chromosomal mutations*, as shown in **Figure 3.**

Deletion A *deletion* occurs when a piece of a chromosome is lost. At the end of meiosis, one of the cells will lack the genes from that missing piece. Such deletions are usually harmful.

Duplication A *duplication* occurs when a piece remains attached to its homologous chromosome after meiosis. One chromosome will then carry both alleles for each of the genes in that piece.

Inversion An *inversion* occurs when a piece reattaches to its original chromosome, but in a reverse direction.

Translocation A *translocation* occurs when a chromosome piece ends up in a completely different, nonhomologous chromosome.

Gene Rearrangement A chromosomal mutation can move an entire gene to a new location. Such a change, called a *gene rearrangement,* is likely to disrupt the gene's function in other ways, as you will learn.

❯ **Reading Check** ***Why are point mutations often silent?***

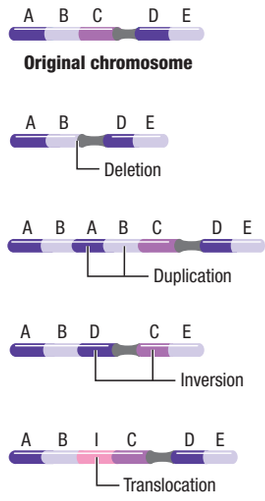


Figure 3 Four kinds of chromosomal mutations can result from errors in crossover during meiosis. ❯ **How are the types of chromosomal mutations similar to the types of smaller-scale mutations?**

Differentiated Instruction

Basic Learners/Special Education Students

Chromosomal Mutation Models Give students modeling clay or dough to demonstrate the chromosomal mutations illustrated in **Figure 3.** Elongate pieces in different colors and textures to represent the genes. Blind and visually impaired students can use models with shape or texture to understand the five types of chromosomal mutations. LS Visual

QuickLab

Teacher's Notes Students should construct examples of all DNA mutations. Make sure they record the original and changed sequences. Encourage peer evaluation. Visually impaired students will need 12 objects with four different geometric shapes to create their models.

Materials
- amino acid codon chart
- paper, (12 sheets)
- pushpins, 4 different colors or other multi-colored objects (12)
- assorted other materials, such as paperclips, straws, tape, and stickers

Answers to Analysis

To answer these questions, students will need accurate models of the following 12 mutation types: point, insertion or deletion, silent, missense, frameshift, nonsense, more or fewer amino acids, deletion, duplication, inversion, translocation, and gene rearrangement. The models also should identify the different categories.

Teaching Key Ideas

Chromosomal Mutations Unlike DNA-level mutations, which affect amino acid sequences, chromosomal mutations affect entire genes or groups of genes.

Answers to Caption Questions

Figure 3: Insertions, deletions, and substitutions are possible.

Teach, *continued*

Teaching Key Ideas

Melanoma Melanoma is named for melanin, a brownish-black pigment found under the skin. Use the diagrams in **Figure 4** to discuss melanoma in terms of mutation. Ask what the *mutagen* is that sometimes causes melanoma. (exposure to ultraviolet rays) Ask if melanoma is inherited. (No, it is found in somatic cells.) What does a melanoma look like? (a very dark, irregularly shaped growth) Stress that melanoma is a malignant form of cancer. It is easily detected and can be treated successfully if identified in early stages. Explain that, although most cancers occur in somatic cells and are thus not heritable, the genetic risk of specific cancers can be inherited. For example, a specific allele has been linked to an increased risk of one type of melanoma. LS Visual

Answers to Caption Questions

Figure 4: No, only mutations in germ cells can be passed on.

Effects of Genetic Change

Many genetic changes will cause no change in the appearance or function of organisms. Moreover, many changes in the DNA of cells may not be passed on to other cells by mitosis or meiosis. **The results of genetic change may be harmful, beneficial, or neutral; most changes are neutral and may not be passed on to offspring.** Mutations that occur in gametes can be passed on to offspring, but mutations in body cells affect only the individual in which they occur.

Heritable or Not Multicellular eukaryotes have two primary cell types: germ cells and somatic cells. *Germ cells* make up gametes, and *somatic cells* make up the rest of the body. Mutations can occur in either type of cell. However, if a mutation occurs in a somatic cell, that genetic change will be lost when the owner of the cell dies. For example, a mutation in a person's lung cell could cause the cell to grow into lung cancer. The mutated genes in the cancer cells will not be transferred to the person's children.

Only a mutation in a germ cell may be passed on to the next generation. However, any such mutation may be silent or have little effect. Only rarely do mutations cause dramatic changes in future generations.

If a mutation occurs in a somatic cell, the change may be silent or it may change the function of the cell. Recall that most tissues are derived from a few parent cells. So, if a mutation occurs in a parent cell, all cells that arise by mitosis from that cell will have copies of the mutation. If the new cells can function at all, each will have the altered structure or function caused by the mutation. If the other parent cells were normal, the resulting tissue may include both normal tissue and mutant tissue.

ACADEMIC VOCABULARY
dramatic vivid or striking

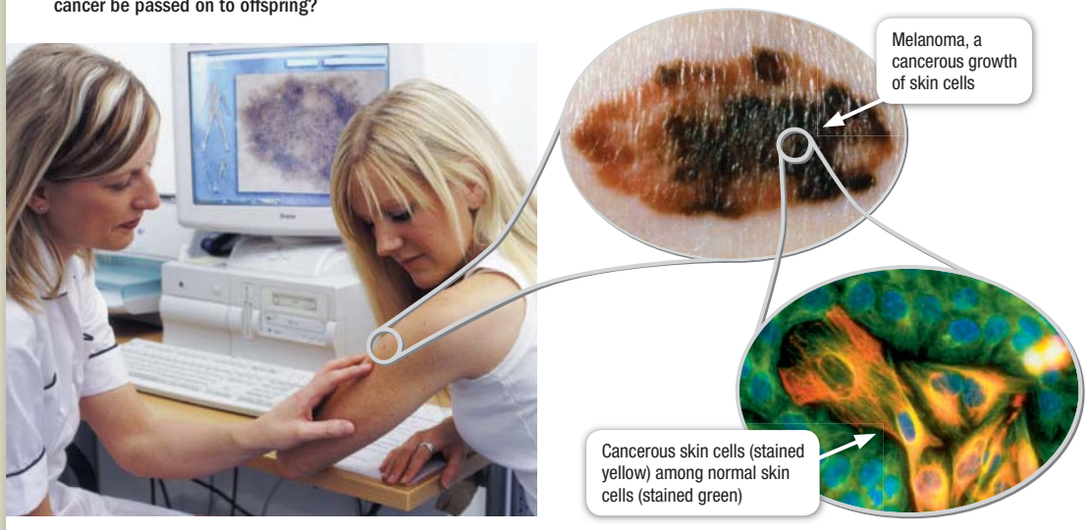


Figure 4 Melanoma is a type of skin cancer caused by mutations in melanocytes, the cells that make skin pigment. Melanoma is an example of a somatic cell cancer. **Can this kind of cancer be passed on to offspring?**

Why It Matters

Radiation and Cancer In 1986, a nuclear explosion occurred in Chernobyl, Ukraine. The release of radiation has been linked to over 5,000 cases of thyroid cancer in individuals who were children at the time of the incident, and who drank milk from cows that ingested radioactive iodine while grazing. Radiation has been linked to the incidence of cancer as a primary mutagen.

Differentiated Instruction

Basic Learners

Ultraviolet Radiation and Sunscreen Have students investigate the incidence of melanoma in sunbelt states—Florida, Texas, Arizona, and New Mexico. Have them find out the science behind sunscreens. Have students use the information and work in pairs to create consumer awareness posters about the dangers of melanoma and prevention strategies. This information is especially important for teens, who often don't adequately protect themselves from the dangers of prolonged exposure to the sun. LS Visual, Verbal

Some Human Genetic Disorders

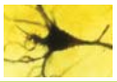
Disorder	Inheritance pattern	Major physical symptoms	Genetic effect of mutant allele	Number of cases (United States)
Sickle cell anemia 	recessive	poor blood circulation; pain; damage to organs such as liver, kidney, lungs, and heart	abnormal hemoglobin in red blood cells	72,000
Tay-Sachs disease 	recessive in most cases	deterioration of central nervous system; death in early childhood	defective form of an enzyme in nerve cells	< 100
Cystic fibrosis 	recessive	mucus buildup in organs such as lungs, liver, and pancreas; difficulty breathing and digesting; shortened life span	defective form of an enzyme in secretory cells	30,000
Hemophilia A (classical) 	recessive, sex-linked	failure of blood to clot; excessive bleeding and bruising when injured	defective form of a protein for blood clotting	18,000
Huntington disease 	dominant	gradual deterioration of brain tissue in middle age; shortened life expectancy	abnormal protein in brain cells	30,000

Figure 5 Genetic disorders are caused by inherited mutations that disrupt the normal function of a gene. › **Why are genetic disorders relatively rare?**

Tumors and Cancers Certain genes control the normal growth, division, and specialization of cells in bodies. Mutations in these genes can cause a normal somatic cell to "lose control" and begin growing and dividing abnormally. The group of cells that grows will become a *tumor.* If the tumor cells begin to invade other parts of the body, they become a form of *cancer*. An example of a somatic cell tumor is shown in **Figure 4.** Note that although cancers result from somatic cell mutations, not all somatic cell mutations cause cancer.

New Alleles You previously learned that for any given gene, many alleles, or variations, may exist. Now, you should see that any new allele must begin as a mutation of an existing allele. Most new alleles are simply the result of silent mutations, so these changes make little difference to the organisms in which they occur. However, sometimes a new allele can cause a change in a gene's function. Depending on the gene, the result may be harmful or beneficial to the organism.

Genetic Disorders Harmful effects produced by inherited mutations (defective alleles) are called *genetic disorders*. Several human genetic disorders are summarized in **Figure 5.** Often, such a disorder results because a mutation has altered the normal function of a gene. However, a person may still have one allele of the original, functioning gene. For this reason, many disorders are recessive—that is, the disorder develops only in a person who is homozygous for the mutated allele. So, two heterozygous people may be healthy, yet have children who develop a genetic disorder. A person who is heterozygous for such an allele is said to be a *carrier* of the disorder.

› **Reading Check** ***How are mutations related to cancer?***

SCILINKS
www.scilinks.org
Topic: Genetic Disorders
Code: HX80652

Differentiated Instruction

Advanced Learners/GATE

Down Syndrome Have students investigate Down syndrome, with an emphasis on the correlation between nondisjunction during oogenesis and the age of the mother. Have them represent this connection in a graphic form. LS Verbal

Basic Learners/Struggling Readers

Vocabulary Refresher To understand genetic disorders, students should review the following terms from the chapter on Mendel's work: *allele, heterozygous, homozygous,* and *carrier.* Then, have them describe the inheritance pattern for each disorder listed in **Figure 5.** (recessive disorders—one recessive allele comes from each parent; dominant disorders—offspring needs only one dominant allele for the disorder; recessive sex-linked—recessive alleles are passed on via X or Y chromosomes) LS Verbal

Answers to Caption Questions

Figure 5: Mutations are relatively rare and any mutation can be carried on recessive alleles.

Teaching Key Ideas

Genetic Disorders Genetic disorders result only from mutations that are inherited and produce defective alleles that disrupt the function of the gene. For all except one of the genetic disorders presented on this page, an individual would need to inherit a recessive allele from *both* parents in order to have the disorder. However, this is not the case with Huntington disease, which is caused by a dominant allele, so either parent can pass on the defective allele. Ask why the Huntington allele remains in the population. (Huntington's symptoms are not detected until later in life—30s and 40s, by which time the allele may have been unknowingly been passed on.)

Teach, *continued*

Answers to Caption Questions

Figure 6: missing chromosome (usually fatal), polyploidy

Teaching Key Ideas

Large-Scale Genetic Change In multicellular organisms, offspring (zygtoes) that receive incomplete chromosomes cannot usually develop because essential parts of their "blueprint" are missing. However, offspring that receive additional genes or chromosomes may have various results. Many plant species have enlarged parts due to chromosomal duplications.

Teaching Key Ideas

Nondisjunction Ask students how *nondisjunction,* which causes *trisomy 21,* differs from *polyploidy.* (In nondisjunction, only one pair of chromosomes fails to separate; in polyploidy, there is an extra set of all chromosomes because all chromosomes fail to separate.)

Close

Formative Assessment

Which type of point mutation has no effect on a gene's function?

A. missense (Incorrect. Missense mutations can change the amino acid sequence.)

B. nonsense (Incorrect. The stop codon prevents further translation.)

C. silent (Correct! A changed codon still calls for the same amino acid.)

D. insertion (Incorrect. Insertions could change all codons in the sequence.)

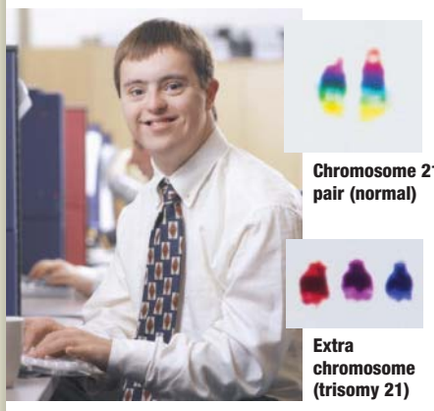


Figure 6 Most people with Down syndrome have an extra copy of chromosome 21. The extra chromosome can be seen in a karyotype. **What other conditions can result from accidents in chromosome sorting?**

nondisjunction (NAHN dis JUHNK shuhn) a failure of homologous chromosomes to separate during meiosis I or the failure of sister chromatids to separate during mitosis or meiosis II

polyploidy (PAH lee PLOY dee) an abnormal condition of having more than two sets of chromosomes

Large-Scale Genetic Change

At another scale, accidents can happen to entire sets of chromosomes. **Very large-scale genetic change can occur by misplacement, recombination, or multiplication of entire chromosomes.**

Recombination During Crossover Genetic recombination through sexual reproduction has many important consequences. Recall that during the *crossover* step of meiosis, the alleles from one parent are recombined with the alleles from the other parent. So, meiosis creates new combinations of alleles in offspring. Over time, sexual reproduction and meiotic recombination maintain genetic variety within a population.

Errors in Sorting Chromosomes Each of your chromosomes has thousands of genes. Together, these genes control cell structure and function. So, all 46 chromosomes (23 pairs) are needed for your body to develop and function normally. Human embryos with missing chromosomes rarely survive. Humans with an extra chromosome may survive but do not develop normally.

Nondisjunction Recall that when gametes form by meiosis, each pair of chromosomes separates in the step called *disjunction*. When the pairs fail to separate properly, the error is called **nondisjunction.** For example, nondisjunction of chromosome 21 can lead to a disabling condition called *Down syndrome,* or *trisomy 21,* as shown in **Figure 6.** In this case, one of the parent's gametes received both copies of chromosome 21 instead of one. When that gamete joined with a normal gamete, the child received three copies instead of two.

Polyploidy The largest scale of genetic change can happen if the entire genome is duplicated. Such duplication can occur—rarely—during meiosis, by nondisjunction of *all* chromosomes. The result is a cell with multiple sets of chromosomes, a condition known as **polyploidy.** A polyploid cell has genetic material "to spare." In future offspring, mutations can happen in some genes without losing the functions of the original genes. Thus, polyploidy is another way that organisms can change over time. Polyploidy is common in plants.

Reading Check ***How can a child be born with extra chromosomes?***

Section 1 Review

KEY IDEAS

1. **Identify** the primary mechanism for genetic change and differences among organisms.
2. **List** the kinds of mutations.
3. **Relate** the possible kinds of mutations to their effects.
4. **Relate** changes in chromosome number to possible results.

CRITICAL THINKING

5. **Evaluating Significance** Compare DNA mutations with chromosomal mutations in terms of the severity of the results of each.
6. **Justifying Conclusions** You read in a magazine that all mutations are bad. Do you agree? Explain.

USING SCIENCE GRAPHICS

7. **Visualizing** Look at **Figure 2** in this section. Notice that it shows only a single strand of the original DNA sequence and a final amino acid sequence. Sketch the matching DNA and RNA strands for the steps in between. Review the steps of gene expression if needed.

Answers to Section Review

1. mutation
2. DNA mutations: point mutations, and insertions/deletions; chromosomal mutations: deletions, duplications, inversions, translocations, and gene rearrangement
3. Point mutations, insertions, and deletions cause changes in amino acid sequences. Chromosomal mutations involve the addition, deletion, or rearrangement of entire genes.
4. Nondisjunction in animals produces individuals that are not as fit as their counterparts. However, polyploidy can be beneficial by producing offspring with more desirable qualities.
5. DNA mutations are less severe because they may or may not cause a change in one gene. Chromosomal mutations typically affect many genes.
6. No, mutations cause changes that are sometimes beneficial to a species. Mutations have contributed to the variety of life on Earth.
7. Sample answer (with no mutation):

DNA:	TAC	GGT	ACG
mRNA:	AUG	CCA	UCG
amino acid:	met	pro	ser

Section 2

Regulating Gene Expression

Key Ideas	Key Terms	Why It Matters
❯ Can the process of gene expression be controlled? ❯ What is a common form of gene regulation in prokaryotes? ❯ How does gene regulation in eukaryotes differ from gene regulation in prokaryotes? ❯ Why are proteins so important and versatile?	operon transcription factor intron exon domain	Understanding gene regulation may enable us to treat or prevent diseases that were previously unbeatable.

How do butterflies develop from caterpillars? We now know that genes determine traits such as patterns on butterfly wings, as shown in **Figure 7.** And we know that every cell in an individual starts with the same genes. So, in a butterfly's lifetime, every trait of every gene is not always "at work."

Complexities of Gene Expression

Scientists have learned that gene expression (transcription and translation) can be regulated. It is now clear that not all genes are expressed in every cell, nor are many genes expressed all of the time.

❯ **Cells have complex systems that regulate whether or not specific genes are expressed. Expression depends on the cell's needs and environment.**

Through *gene regulation,* a given genetic sequence can be expressed in different ways—in different bodies or tissues, under different conditions, or at different times. Thus, gene regulation accounts for changes during development as well as differences among organisms that have similar genes. One benefit of gene regulation is that cells can use energy and materials efficiently.

Recall that many steps take place in the expression of a gene. Also, other molecules play a role in the processes. Because complex interactions happen at each step, there are many opportunities to regulate gene expression. So, nearly every step in the process of gene expression can be regulated or controlled.

A molecular system that controls the expression of a specific gene is called a *genetic switch*. Like a light switch, a genetic switch can be turned "on" or "off." Often, the switch is triggered by factors or conditions outside the cell. Also, the product of one gene may serve to regulate another gene in the same organism.

❯ **Reading Check** *Are all genes expressed all of the time?*

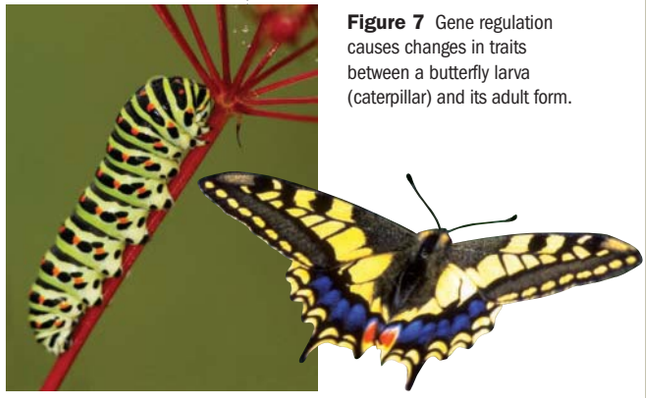


Figure 7 Gene regulation causes changes in traits between a butterfly larva (caterpillar) and its adult form.

Key Resources

Visual Concepts

Operon

Repression of Transcription in the *lac* Operon

Regulator Gene

Activation of Transcription in the *lac* Operon

Enhancers for Control of Gene Expression

Comparing Introns and Exons

Section 2

❯ Focus

This section explores the methods by which prokaryotes and eukaryotes regulate gene expression. Students will also learn that cells express only a small percentage of their genes at any given time.

Bellringer

Use the Bellringer transparency to prepare students for this section.

❯ Teach

Demonstration

Using an Analogy Hold up a copy of the student textbook. Ask students to imagine that the 1,000 pages represent all the genetic information in one of their cells. A typical cell uses only 3 to 5 percent of its total information. This equates to 30 to 50 pages of the book. Flip through this number of pages, about one chapter, to illustrate. LS Visual

Teaching Key Ideas

Genetic Switch Ask how the development of a butterfly illustrates the factors regulating gene expression. (Only those genes to make a larva functional are turned on; those genes that direct butterfly development are turned off until that stage is present.) LS Visual

Teach, *continued*

Answers to Caption Questions

Figure 8: common in prokaryotes, uncommon in eukaryotes

Teaching Key Ideas

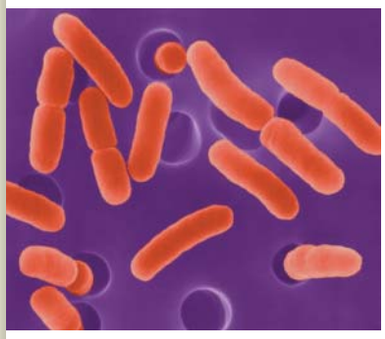
The *lac* Operon Ask students to speculate on how the operon got its name. (It controls the "operation" of enzymes that break down lactose.) The *lac* operon is a system found in *E. coli* bacteria that illustrates how the environment controls gene regulation. This system includes a promoter and operator site, a repressor protein, and the section of DNA to be transcribed, three genes that enable the bacterium to use the lactose in its environment. This system limits the expression of these genes only to times when lactose is available, thereby conserving energy and materials.

READING TOOLBOX

Sample Comparison Table

	Prokaryotes	Eukaryotes
Similarities	Promoter turns on system	More complex. Operons are rarely found
Differences	Operons control gene expression	Transcription factors, activators, repressors, enhancers

LS **Visual**



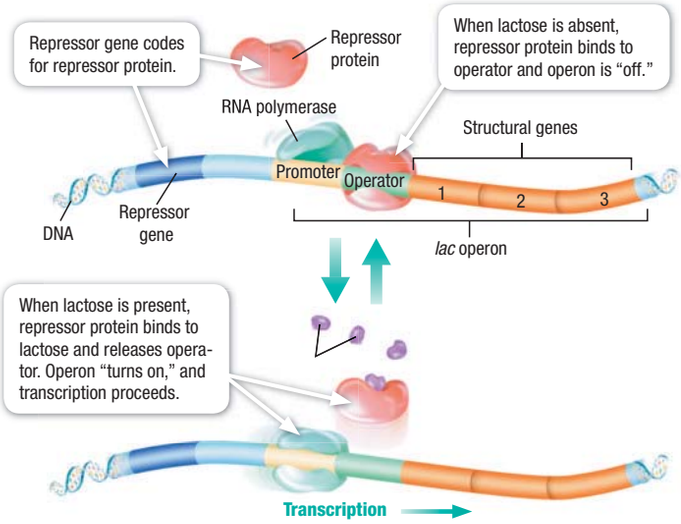


Figure 8 The *lac* operon controls the genes that code for the proteins that help a bacterium use lactose. The operon "turns on" and expresses the genes only in the presence of lactose. ❯ **How common are operons?**

READING TOOLBOX

Comparison Table Make a comparison table to compare **Figure 8** and **Figure 9.** Which roles do proteins have in the eukaryotic system but not in the prokaryotic system?

Gene Regulation in Prokaryotes

Scientists have studied and compared gene expression in prokaryotes and eukaryotes. Each has very different ways of regulating how genes are expressed. One reason for the differences can be found in the structure of genes in the two kinds of organisms.

❯ **The major form of gene regulation in prokaryotes depends upon operons that respond to environmental factors.** An **operon** is a gene-regulation system in which adjacent DNA segments control the expression of a group of genes with closely related functions. Operons are common in bacteria but uncommon in eukaryotes.

Interactions with the Environment Recall that bacteria are single cells that must get food directly from the environment. Given a stable environment, a bacterium will need a steady supply of proteins and will tend to keep expressing the same genes in the same way. But if the environment changes, a cascade of changes in gene expression may result. In a way, the environment "flips a switch."

The *lac* Operon Example An example of gene regulation is found in the bacterium *Escherichia coli*. Usually, when you eat or drink a dairy product, the chemical lactose ("milk sugar") is digested by *E. coli* cells living in your gut. These cells can use the lactose for energy or for other needs. But first, the cells must attach to, absorb, and then break down the lactose. These tasks require three different enzymes, each of which is coded for by a different gene.

The system that involves the *lac* genes is called the *lac operon* and is shown in **Figure 8.** This system includes the three genes plus a *promoter* site and an *operator* site. When lactose is available, the system "turns on" and the three genes are transcribed. When lactose is absent, the system "turns off" and transcription is blocked.

Why It Matters

Lactose Intolerance Humans who are lactose intolerant lack enzymes that will break down lactose into monosaccharide sugars for use. People who consume lactose products and are lactose intolerant experience gas, bloating, and diarrhea. Lactase enzymes are available as dietary supplements to remedy this problem.

Differentiated Instruction

Struggling Readers/English Learners

Vocabulary Students may need help with unfamiliar words on these two pages: *activator, enhancer, Escherichia coli (E. coli), lac, lactose, operator, operon, promoter, repressor gene,* and *repressor protein.* Have students create a key-term fold for these words to use while they are reading this section. Give them other reference materials, including the dictionary, to help them write reasonable definitions for each term. LS **Verbal**

Gene Regulation in Eukaryotes

Eukaryotic cells, too, must turn genes on and off in response to signals from their environment. However, **gene regulation in eukaryotes is more complex and variable than gene regulation in prokaryotes.** To begin with, gene expression in eukaryotes involves more steps and interactions than gene expression in prokaryotes.

As you shall see, regulation can occur before transcription, after transcription, or after translation. Furthermore, in eukaryotes, a nuclear membrane separates these processes. So, each process can be regulated separately.

Eukaryotic gene regulation is unique in other ways. Operons are very rare in eukaryotic cells. Also, groups of genes with related functions may be scattered on different chromosomes and controlled by multiple factors. Finally, much of the DNA in eukaryotes may never be transcribed, and even less is ultimately translated into proteins.

operon (AHP uhr AHN) a unit of adjacent genes that consists of functionally related structural genes and their associated regulatory genes

transcription factor an enzyme that is needed to begin and/or continue genetic transcription

Controlling Transcription Like prokaryotic cells, eukaryotic cells have proteins that regulate transcription. But many more proteins are involved, and the interactions are more complex. Most often, the genetic switch involves the first step of transcription, when RNA polymerase binds to the promoter region. The proteins involved in this kind of genetic switch are called **transcription factors.**

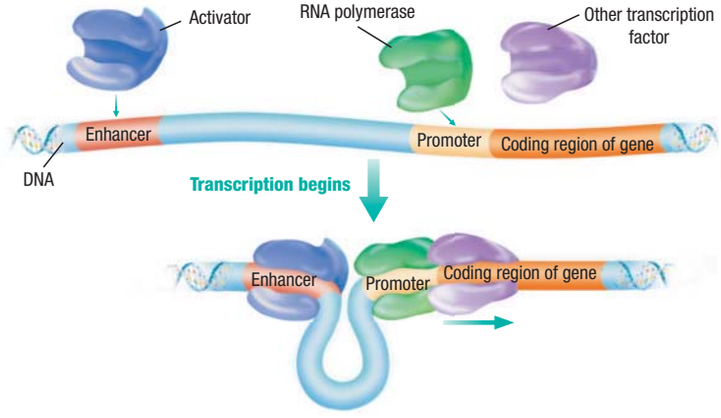
ACADEMIC VOCABULARY

regulate to control, direct, or govern; to adjust

As shown in **Figure 9,** transcription factors interact with RNA polymerases around promoter regions of DNA. A given gene can be influenced by many transcription factors. Some transcription factors act as *activators,* and some act as *repressors.*

One kind of DNA sequence that can be bound by an activator is called an *enhancer*. Enhancers are often located thousands of bases away from the promoter. A loop in the DNA forms as the factors interact at the promoter site. Each factor may also affect other factors.

Reading Check ***Which parts of gene expression can be regulated?***



go.hrw.com
interact online
Keyword: HX8GNXF9

Figure 9 Control of transcription is complex in eukaryotes. For example, an activator may bind to an enhancer site and also to RNA polymerase. This action will activate another transcription factor, and finally transcription will begin.
Why is gene regulation more complex in eukaryotes than in prokaryotes?

Teach

Demonstration

Modeling the Regulation Process Cut out shapes from colored paper to represent the components of the *lac* operon. Show the repressor without lactose bound to the operator, and then show it removed from the operator site, because lactose is bound to it. Ask students why transcription is allowed to proceed. (The repressor, when bound to lactose, changes shape, disengaging the operator site to allow transcription.) LS Visual

Teaching Key Ideas

Eukaryotic Gene Regulation A eukaryote gene has more control elements than a prokaryote gene. Typically there are distal enhancer elements and proximal control elements next to the promoter, which is adjacent to the gene. Other differences are the presence of introns and exons.

Answers to Caption Questions

Figure 9: More steps are involved in gene expression in eukaryotes.

go.hrw.com
interact online

Students can interact with "Gene Regulation" by going to go.hrw.com and typing in the keyword HX8GNXF9.

Differentiated Instruction

Advanced Learners/GATE

Nobel Winners in Genetics Have students investigate how double-stranded RNA interferes with eukaryotic transcription, which was studied by 2006 Nobel Prize winners Andrew Fire and Craig Mello. LS Verbal

MISCONCEPTION ALERT

Gene Expression in Individual Cells Students may think that all genes are expressed in a eukaryotic organism. Clarify that not all genes are expressed in a particular cell. Each cell contains the full complement of DNA, but only a small portion is actually needed for that cell. For example, a gene to make an enzyme needed or produced in a liver cell will never be expressed in a skin cell. The enzyme will be used to aid in digestion, which is not a function of a skin cell. Different conditions signal that genes will be turned on relative to the function of the cell.

Teach, *continued*

QuickLab

Teacher's Notes Make sure that students keep the letters in the order of the original as they cut and separate them. Use the text picture as a guide.

Materials
- masking tape, 20 cm
- pens, 2 different colors

Answers to Analysis

1. exons: the strip that spells "protein;" introns: the remaining strip
2. There would be extra and possibly different amino acids, so a different protein would be synthesized.

Demonstration

Splicing To distinguish between the terms *slice* and *splice*, show how electrical wires are joined together for *splicing*.

Hands-On

QuickLab

10 min

A Model of Introns and Exons

You can model introns and exons with masking tape.

Procedure

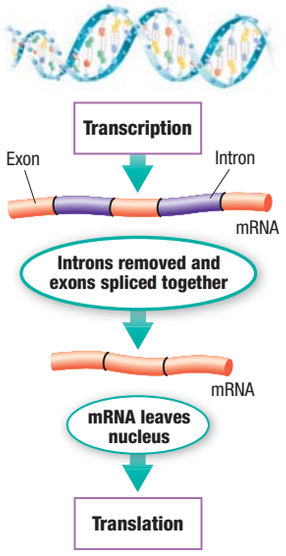
1. Place a **15 to 20 cm strip of masking tape** on your desk. The tape represents a gene.
2. Use **two colored pens** to write letters on the tape, exactly as shown in the example here. Space the letters to take up the entire length of the tape. The segments in one color represent introns; those in the other color represent exons.
3. Lift the tape. Working from left to right, use **scissors** to cut apart each group of letters of the same color.
4. Stick the pieces of tape to your desk as you cut them. Make two strips of matching colors, and join the pieces in their original order.

appropriately joined

Analysis

1. **Determine** from the resulting two strips which strip represents "introns" and which represents "exons."
2. CRITICAL THINKING **Predicting Results** What might happen to the protein if an intron were not removed?

Figure 10 After transcription in eukaryotes, the entire new mRNA segment may not be translated into proteins. Instead, introns are removed, and only exons are translated.



Processing RNA After Transcription It is simplest to think of a gene as a string of nucleotides that code for a protein. However, this simple arrangement is usually found only in prokaryotes. In eukaryotes, many genes contain *noncoding* sequences, or segments of code that will not be translated into amino acids. The noncoding segments are called **introns,** while those portions of the gene that *do* code for amino acids and will be translated are called **exons.**

RNA Splicing Exons and introns are handled in a process called *RNA splicing,* as shown in **Figure 10.** After a eukaryotic gene is transcribed, the introns are removed with the help of certain proteins. The exons that remain are *spliced,* or rejoined together, to form a smaller mRNA molecule. Finally, the spliced mRNA leaves the nucleus and is then translated.

Alternative Splicing The splicing of eukaryotic genes creates additional opportunities for variation over time. Because each exon encodes a different part of a protein, cells can occasionally shuffle exons between genes and thus make new proteins. The thousands of proteins in human cells appear to result from shuffling and recombining a few thousand exons. Some human genes, such as those for hemoglobin, are made up of multiple copies of similar exons.

Processing Proteins After Translation After translation, a chain of amino acids is formed, but the protein may not go directly into action. Further chemical changes may alter the structure and function of the protein. Such changes may affect the protein's shape, stability, or interactions with other molecules.

Final Destination A newly made protein may be needed in a specific location within the cell. The process of getting proteins to their correct destination is called *protein sorting.* Protein sorting occurs in many parts of the cell, such as the Golgi apparatus.

MISCONCEPTION ALERT

Protein Complexity Most proteins range from 150 to 400 amino acids in length. All of the protein synthesis modeling students have done so far does not represent anything close to a real protein. Their models are short segments of a polypeptide.

Differentiated Instruction

Basic Learners/English Learners

Exon Splicing Have pairs of students join at least five colored plastic-coated paper clips and five aluminum paper clips together in any order to represent DNA. Then, have students copy the sequence with a second set of paperclips to represent *transcription.* Have students remove the metal clips as a model for introns being removed. They then join the colored clips together to represent exon *splicing.*

LS Visual / Kinesthetic

Sorting Signals Protein sorting is often directed by *sorting signals,* small parts of a protein that bind to other molecules within the cell. Some signals bind the protein to its final location in the cell. Some signals bind proteins to ribosomes while translation is in progress, and sends them together to the ER for further processing. This variation is another example of the complexity of genes.

The Many Roles of Proteins

Recall that proteins are complex strings of amino acids that do much of the work in cells. The diversity of protein structures relates to the many functions that proteins serve in cells. These functions range from forming the cell's shape to regulating gene expression. Proteins range in size from about 50 amino acids to more than 25,000 amino acids. The average protein is about 250 amino acids.

Protein Structure Because they can form many shapes, proteins can serve many roles. **› The sequence of amino acids in a protein determines its three-dimensional structure and chemical behavior.** In turn, this folding determines the function of the protein, as shown in **Figure 11.** Some parts of a protein that have a specific chemical structure and function are protein **domains.** A protein may have several domains, each with a specific function. In eukaryotes, each domain is usually the result of a specific exon. Finally, large proteins may be made up of several smaller proteins, or *subunits.*

Proteins in Gene Expression Proteins serve important roles in gene expression. For example, several forms of RNA polymerase function to make mRNA, tRNA, and rRNA. Other proteins serve as *regulatory proteins* by binding to genetic switches in specific genes.

Because transcription is more complex in eukaryotes than in prokaryotes, more proteins are involved in the process. Likewise, more enzymes and structural proteins are required for translation in eukaryotes. Even after translation, additional steps may be needed to make a protein fully active in its proper place in a cell.

› **Reading Check** ***What determines a protein's shape?***

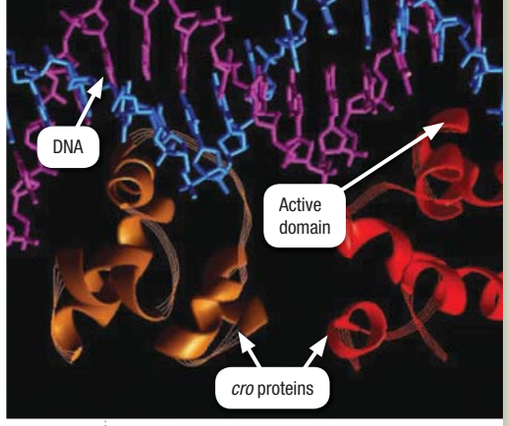


Figure 11 Many of the proteins involved in gene regulation have a shape that fits closely with DNA or RNA molecules. The example shown here is a model of two molecules of bacterial *cro* protein (orange and red) binding to a molecule of DNA (blue and purple). The parts of the protein molecules that are chemically active are called *active domains.*

intron a nucleotide sequence that is part of a gene and that is transcribed from DNA into mRNA but not translated into amino acids

exon one of several nonadjacent nucleotide sequences that are part of one gene and that are transcribed, joined together, and then translated

domain in proteins, a functional unit that a has a distinctive pattern of structural folding

Section 2 Review

› KEY IDEAS

1. **Generalize** the ways that gene expression can be regulated.
2. **Describe** an example of gene regulation in prokaryotes.
3. **Identify** how gene regulation in eukaryotes is unique.
4. **Relate** protein structure to function in gene expression and regulation.

CRITICAL THINKING

5. **Proposing Mechanisms** Propose one other mechanism, not yet mentioned, for gene regulation in either prokaryotes or eukaryotes.
6. **Using Models** Use letters and words to show how a sequence could be spliced in several ways.

ALTERNATIVE ASSESSMENT

7. **Electronic Research** Use electronic resources to find three-dimensional computer models of proteins. Be sure that the models are based on scientific research. Make a display of several models.

Teaching Key Ideas

Protein Structure The primary structure, or the amino acid sequence, of a protein determines how atoms and functional groups will interact. By pleating and folding a three-dimensional molecule is produced that has a specific region of activity. "Form dictating function" makes proteins the most diverse group of biomolecules.

› Close

Formative Assessment

What is the function of the *lac* operon?

A. It makes genes. (Incorrect. It is a system made up of genes and other components.)

B. It makes lactase enzymes. (Incorrect. There are three genes, none of which specifically code for lactase.)

C. It makes a lactose sugar. (Incorrect. Lactose sugar engages the repressor, which is part of the *lac* operon system.)

D. It switches "on" the *lac* genes. (Correct. When the repressor is engaged by lactose, the operator is turned "on.")

Answers to Section Review

1. interactions of the genetic material and the cellular environment
2. In the presence of an environmental trigger, the *lac* operon switches "on" to start transcription.
3. Regulation can occur before or after transcription, or after translation. Each process can be regulated separately. Operons are rare in eukaryotic cells. Genes with related functions may be separated and controlled by multiple factors. Much of the DNA in eukaryotes is not fully transcribed and translated.
4. The sequence of amino acids determines the three-dimensional configuration that produces an active domain on the protein.
5. Sample answer: The presence of an antibiotic in the environment may inhibit gene expression in prokaryotes.
6. Have students create a display of the color-enhanced models they find.
7. Check that student displays are based on scientific sources and show 3-D models of proteins. If possible, display these in the classroom.

Section 3

Genome Interactions

Key Ideas	Key Terms	Why It Matters
What can we learn by comparing genomes? Can genetic material be stored and transferred by mechanisms other than chromosomes? What are the roles of genes in multicellular development?	genome plasmid transposon cell differentiation apoptosis	We can understand how our own bodies work by comparing our genetic systems to those of other organisms.

Focus

The purpose of this section is to expand the concept of the gene to the level of a genome. Mobile genes, and research on how certain genes control development and cell growth are also discussed.

Bellringer

Use the Bellringer transparency to prepare students for this section.

Teach

Teaching Key Ideas

Comparing Gene Ratios Refer students to **Figure 12.** Ask what the percentages mean. (comparison of genes that are the same as in humans) Ask what they can infer from these percentages in regard to evolution. (Those organisms with similar percentages could be closely related in evolutionary history.) Stress that a genome is a summary of DNA found in *all* members of the species, not just one individual. LS Visual

Answer to Caption Question

Figure 12: We can study organisms that have genes similar to ours to learn more about our own genetics.

Do you share genes with bacteria? In a way, you do. About 10% of human genes are nearly identical to bacterial genes. **Figure 12** shows the similarity between human genes and genes of other organisms.

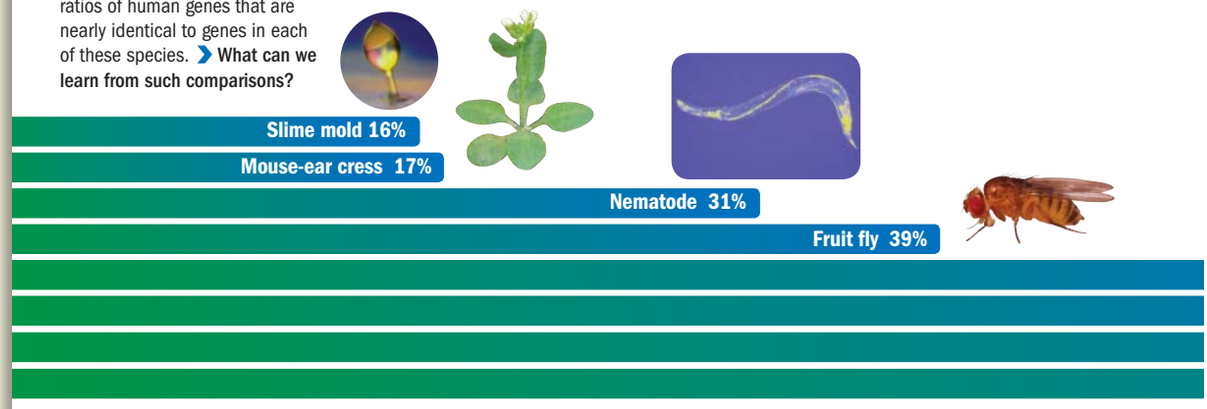
Genomes and the Diversity of Life

Studying genomes has revolutionized how we look at gene regulation and gene expression. Recall that a **genome** is all of the DNA that an organism or species has within its chromosomes. A genome contains all the genes needed to make more of that organism. Today, the genomes of hundreds of organisms have been extensively studied.

Comparisons among the genetic systems of many organisms reveal basic biological similarities and relationships.

Universal Code With few exceptions, the genetic code is the same in all organisms. For example, the codon GUC codes for the amino acid valine in bacteria, in eagles, in plants, and in your own cells. For this reason, the genetic code is often described as being universal. However, some exceptions exist to the universal aspects of the genetic code. For example, some bacteria use a slightly different set of amino acids in making proteins.

Figure 12 This graph shows the ratios of human genes that are nearly identical to genes in each of these species. **What can we learn from such comparisons?**



Genome Sizes Genome size can be measured as an amount of DNA or a number of genes. Either way, genome size is only roughly related to complexity. Genomes in microbes range from 400,000 to millions of base pairs and include from 400 to 9,300 genes. Eukaryote genomes range from 100 million to more than 3 billion base pairs with 6,000 to 100,000 genes. The human genome has about 25,000 genes. Some plants have more than 100,000 genes.

DNA Versus Genes Not all DNA in a cell is part of a gene or even part of a chromosome. Special kinds of DNA include the following:

- **Plasmids in Prokaryotes** Recall that bacterial DNA is usually stored in one long, circular chromosome. However, most bacteria have extra pieces of DNA called **plasmids.** These small, circular DNA segments are replicated independently and can be transferred between cells. So, plasmids are an important source of genetic variation in bacteria.
- **Noncoding DNA in Eukaryotes** Eukaryotes have a great deal of *noncoding* DNA. For example, introns are transcribed but never translated. Also, long stretches of repeating sequences exist that are never transcribed. The function of most noncoding DNA is unclear.
- **DNA in Cell Organelles** Recall that mitochondria and chloroplasts, shown in **Figure 13,** are organelles that have special roles in eukaryotic cells. Chloroplasts enable plants to harvest energy from sunlight. Mitochondria act as the source of energy for cell function. Each of these organelles has its own small genome that is separate from that in the nucleus. These genomes code for proteins and RNAs (rRNA and tRNA) that assist in the function of each organelle.

Endosymbiotic Theory Why do mitochondria and chloroplasts have their own DNA? Scientists suspect that each organelle had its origin in ancient bacterial cells. This idea is known as the *endosymbiotic theory*. For example, chloroplast-like bacteria could have been engulfed, but not killed, by larger cells. Each kind of cell may have benefited from this relationship. Over time, the cells would live together in a close relationship called *symbiosis*.

› **Reading Check** ***What kinds of organisms have large genomes?***

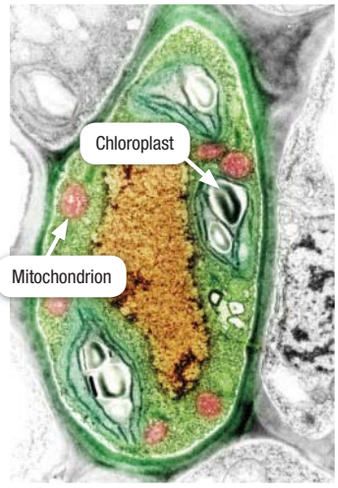
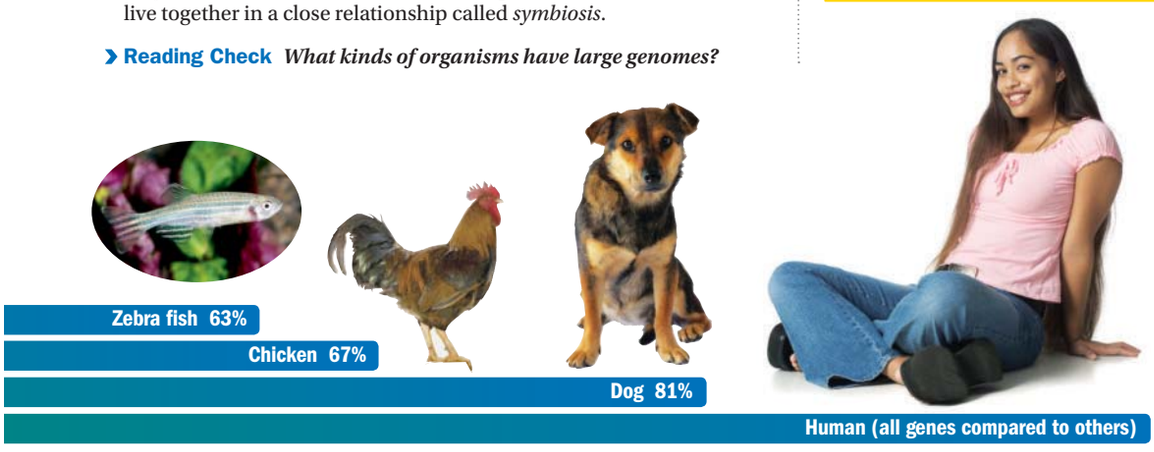


Figure 13 Chloroplasts and mitochondria have their own DNA. Each organelle's genome is stored and replicated separately from the chromosomes of the cell.

genome (JEE NOHM) the complete genetic material contained in an individual or species

plasmid (PLAZ mid) a genetic structure that can replicate independently of the main chromosome(s) of a cell



Differentiated Instruction

Advanced Learners/GATE

Endosymbiont Theory Students should investigate endosymbiont theory, particularly the work of Lynn Margulis. Have them create a table comparing prokaryotes to chloroplasts and mitochondria. In addition to physical comparison, the table should include studies on antibiotic response. LS Visual

Basic Learners/Struggling Readers

Distinguishing Data Help students with the first paragraph on this page by asking questions about the data. How many genes are in the human genome? (25,000) How many genes can be found in eukaryotes? (6,000 to 100,000 genes) Is the human genome the largest of all eukaryotes? How do you know? (No, 100,000 > 25,000) About how many base pairs are found in the largest genomes? (3 billion) What is this number in expanded notation? (3,000,000,000)
LS Verbal

Demonstration

Relative Size of Genomes Show students the following lengths of thread to represent the relative lengths of DNA in a bacterium such as *E. coli.* Use a 10 m loop to represent the main bacterial chromosome, billions of base pairs long. Use a mere 1 cm loop to represent a plasmid, thousands of base pairs long. Use a 2 mm segment to represent one gene, just tens or hundreds of base pairs. Finally, show a gelatin capsule (#0, 2 cm) to represent a bacterium magnified 10,000 times, into which this much DNA would fit (relative to length, not width).
LS Visual

Teaching Key Ideas

Endosymbiont Theory Recall that symbiosis describes two different organisms living in a close relationship for the benefit of at least one. Mitochondria and chloroplasts are membrane-bound organelles that are similar in size to bacteria and have their own DNA. It has been hypothesized that these organelles were once free-living prokaryotes that developed a symbiotic relationship with a larger cell. The DNA within these organelles is not part of the cell's chromosomes.

Teach, *continued*

Teaching Key Ideas

Mobile Genes The spotted and streaked colors on Indian corn result from genes that have been *transposed* from one chromosome to the middle of a coding sequence on another chromosome, which changes gene expression at the new location. These "jumping genes" illustrate how the movement of genetic elements in a cell can cause change.

Teaching Key Ideas

Retroviruses These viruses are named because of their ability to commandeer a cell's DNA using their own RNA as a template, thus reversing the process of transcription and changing the cell's genome. Mention that HIV is a retrovirus.

Answers to Caption Questions

Figure 14: MGEs can transfer antibiotic resistance genes between cells.

***Staphylococcus* bacteria**

Figure 14 We use antibiotic chemicals in drugs and cleaning products in an attempt to kill bacteria. However, some bacteria have become resistant to most antibiotics. **What is the role of mobile genetic elements in antibiotic resistance?**

SCILINKS
www.scilinks.org
Topic: Antibiotic Resistance
Code: HX80081

Moving Beyond Chromosomes

We now know that cells can interact at the genetic level. And we know that genetic material exists outside of chromosomes. The closer we study genetics, the more complexities we find.

Small bits of genetic material can be stored, moved, and changed by a variety of interactions.

Mobile Genetic Elements Plasmids are just one kind of *mobile genetic element (MGE).* MGEs are units of DNA or RNA that are sometimes *transposed,* or moved as a functional unit, from one place to another in a genome. Other MGEs are transposons and viruses.

Transposons Sets of genes that are transposed randomly are *jumping genes,* or **transposons.** When a transposon moves to a new place, it may inactivate a nearby gene, much like an operon does. All organisms seem to have transposons in their genomes. Some bacteria have transposons that jump between plasmids and chromosomes.

Viruses In terms of structure and function, transposons are similar to viruses. *Viruses* are very small, nonliving particles that consist of DNA or RNA inside a protein coating. Viruses infect cells by using the cells' own replication processes to make new virus copies. Sometimes, viruses take away copies of the cells' DNA or leave some DNA behind. Thus, viruses can move genetic material between cells. Certain kinds of RNA viruses, called *retroviruses,* produce DNA that becomes part of the host cell's genome.

Genetic Change The discovery of MGEs has helped us further understand genetic change. It has also enabled us to manipulate genetic change for our own purposes, as you will learn. MGEs cause genetic change by bringing together new combinations of genes. Furthermore, MGEs can transfer genetic material between individuals and even between species. For example, the genome of *Escherichia coli* (common gut bacteria) is about 15% similar to that of *Salmonella* (food-borne, illness-causing bacteria). Scientists suspect that the similar genetic sequences are the result of MGEs being passed between the species.

Antibiotic Resistance Like mutations, transpositions may have helpful or harmful effects. And what helps one organism may harm another. An effect that is helpful to bacteria but harmful to humans is the evolution of antibiotic resistance. Antibiotic chemicals are often used to prevent or combat bacterial infections, as shown in **Figure 14.** But if just one bacterial cell has a gene that makes the cell resist the effect of a particular antibiotic, that cell may survive and reproduce. Furthermore, the gene could be passed to other bacteria as part of an MGE. Scientists fear that this process is indeed happening, because increasing numbers and kinds of bacteria are becoming resistant to each of the antibiotics that have been produced.

Reading Check ***How are transposons and viruses similar?***

Why It Matters

A Passion for Research Barbara McClintock introduced the idea of jumping genes in the 1950s, and her ideas were not accepted for over 30 years. In 1983, at the age of 81, she was awarded the Nobel Prize for her research on transposons. She continued her experiments at Cold Spring Harbor until her death at age 90. Her tenacity in pursuing her beliefs in the face of adversity is an example of a dedicated scientist.

Differentiated Instruction

Advanced Learners/GATE

Retroviruses Have students investigate RNA retroviruses such as HIV and how they use reverse transcriptase to change the genetic makeup of a host cell. LS Verbal

Multicellular Development and Aging

You have learned that external or environmental cues can regulate gene expression in cells. In multicellular eukaryotes, gene regulation can also happen because of internal cues. In particular, the development of an embryo involves complex gene regulation. Many cells will develop from one beginning cell. And different kinds of cells will develop to have different functions in different parts of the body.

› Each cell within a developing body will express specific genes. Gene expression depends on the cell's age and location within the body.

transposon (trans POH ZAHN) a genetic sequence that is randomly moved, in a functional unit, to new places in a genome

cell differentiation the process by which a cell becomes specialized for a specific structure or function during multicellular development

Cell Differentiation In the process of **cell differentiation,** each new cell is modified and specialized as the cells multiply to form a body. Gene regulation plays an important role in this process. *Homeotic* genes are examples of genes that regulate differentiation. Scientists first discovered these genes in fruit flies. Mutations in these genes can cause one body part, such as a leg, to develop in place of another body part, such as an antenna.

As scientists studied many genomes, they found that many kinds of organisms have homeotic genes. And these genes always seem to control similar developmental processes by similar mechanisms. All homeotic genes code for proteins that regulate the expression of other genes. Many homeotic genes contain a similar sequence of 180 bases. This sequence, called a *homeobox,* codes for a DNA-binding domain in the resulting protein.

In general, the genetic regulation of development seems to be similar in all animals. A specific set of homeotic genes, called *hox,* is found in all animals that have a head end and a tail end. Hox genes direct development relative to body position, as shown in **Figure 15.**

› Reading Check ***What is a homeobox?***

READING TOOLBOX

Prefixes The prefix *homeo-* is used several times on this page. Use the Reading Toolbox page to find the meaning of the prefix *homeo-*. Write a definition for *homeotic* and *homeobox* in your own words.

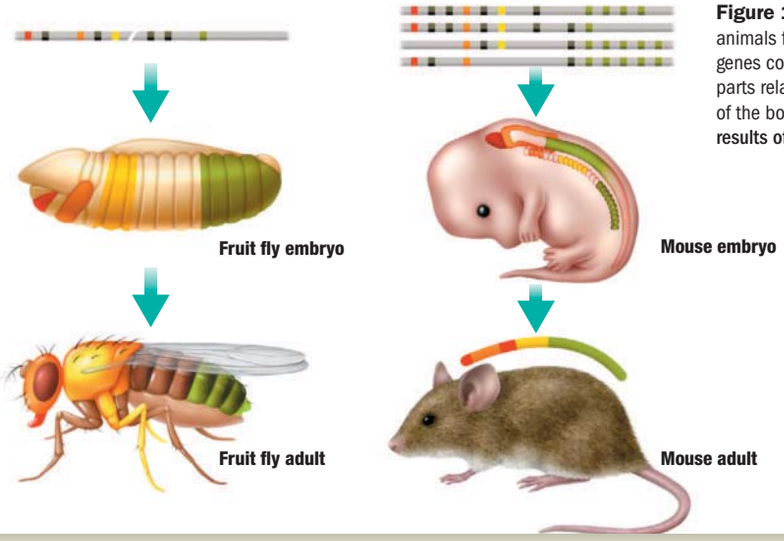


Figure 15 *Hox* genes are found in animals from insects to mammals. These genes control the development of body parts relative to the head and tail ends of the body. **› What are some possible results of mutations in these genes?**

READING TOOLBOX

Prefixes *homeo:* the same, similar; *homeotic:* being the same or similar; *homeobox:* the same "boxes" of genes
LS Verbal

Teaching Key Ideas

Using Visuals Ask students to look at **Figure 15** and compare the hox genes in the fruit fly and mouse embryos. Certain segments of the gene are color-coded to show matching sections of development in both the fly and mouse. LS Visual

Answers to Caption Questions

Figure 15: body parts developing in the wrong place

Differentiated Instruction

Struggling Readers

Key-Term Fold Have students add the following terms to the key-term fold they created in Section 2: *homeotic gene, homeobox, hox, CDK, cyclin,* and *telomeres.* LS Verbal

Teach, *continued*

Answers to Caption Questions

Figure 16: Some cells need to be removed or replaced for normal development or to repair damage.

Close

Formative Assessment

Which of the following can add new genes to a eukaryotic genome?

A. transposons (Incorrect. These genes are part of the genome; they just move.)

B. homeotic genes (Incorrect. These genes are developmental and already part of the genome.)

C. plasmids (Incorrect. These circular rings of DNA occur in prokaryotes.)

D. retroviruses (Correct! When these viruses invade a cell, they use their RNA to create new DNA in the host cell.)

Figure 16 Sunburn is the result of apoptosis—or cell suicide. When skin cells are heavily damaged by over-exposure to the sun, a genetic switch in the cells may signal the cells to stop functioning. **Why is apoptosis important in multicellular organisms?**

apoptosis (AP uhp TOH sis) in multicellular organisms, a genetically controlled process that leads to the death of a cell; programmed cell death

Cell Growth and Maintenance Although scientists have long been aware of the *cell cycle*, only recently have they begun to understand how genes regulate the cell cycle and cell growth. In 2001, three scientists received the Nobel Prize for discovering the genetic systems that regulate the cell cycle. The scientists identified two kinds of proteins that regulate the cell cycle: *CDK* and *cyclin*. These proteins are present in all eukaryotes and drive the cell cycle forward. The CDK molecules function like an engine, and the cyclins function like gears. Together, they control the speed of the cell cycle. Cancer results when control of cells has been lost because either the "engine" or the "gears" malfunction.

Cell Death and Aging In multicellular organisms, all cells have arisen from the division of other cells. But most of these cells stop dividing once the organism is mature. In fact, almost all body cells are "programmed" to age and die. At some point, the cell will simply shut down all functioning, gradually shrink, and eventually fall apart. This process of cellular "suicide" is known as **apoptosis.** Apoptosis seems to occur in consistent steps, much like other cellular processes, such as mitosis. Scientists are still studying the genetic systems that may control apoptosis.

Function of Apoptosis Why do some cells need to die? In some cases, the full development of a body part requires the removal of some cells. For example, apoptosis is responsible for the loss of a tadpole's tail as the tadpole becomes an adult. Likewise, human fingers and toes are formed through the loss of in-between tissue in the embryonic limbs. Also, apoptosis is at work when sunburned skin begins to peel off, as shown in **Figure 16.**

Telomeres Aging has many effects on cells. An example is the effect of aging on the ends of chromosomes (called *telomeres*). As cells divide repeatedly, the telomeres lose nucleotides and become shortened. In older cells, this shortening may cause mishandling of the chromosomes during mitosis and thus result in nonfunctioning cells. However, telomere shortening is not the only cause of aging.

Reading Check ***What are the roles of proteins in the cell cycle?***

Section 3 Review

KEY IDEAS

1. **Justify** comparing the genetic systems of various life-forms.
2. **List** mechanisms other than chromosomes by which genetic material may be stored and moved.
3. **Relate** gene expression to multicellular development.

CRITICAL THINKING

4. **Forming Hypotheses** Could any other cell organelles have arisen through endosymbiosis? If so, what findings may support such a hypothesis?
5. **Predicting** What could be the result of a mutation in a hox gene?
6. **Logical Reasoning** Is apoptosis a useful mechanism for prokaryotes? Explain your answer.

ALTERNATIVE ASSESSMENT

7. **Gallery of Genetic Curiosities** Create a poster, slide show, or other display that exhibits mutants and other interesting examples of genetic complexity. Be sure to provide a caption and a reference source for each of your images.

Answers to Section Review

1. Comparing genetic systems reveals basic biological similarities and relationships among organisms.
2. plasmids, transposons, and viruses
3. Genes cause differentiation of parts during development.
4. Possibly, if other organelles were found to have their own DNA or RNA and protective membranes.
5. A mutation in a hox gene would result in abnormal body development.
6. Sample answer: No, because apoptosis is related to development in multicellular organisms.
7. Accept all well-researched and well-presented displays. The investigation could be limited to exploring one mutant in detail.

Why It Matters

Forensic Genealogy

BIOTECHNOLOGY

Genealogy (JEE nee AHL uh jee) is the study of family histories. Forensic genealogy can involve finding lost relatives, identifying bodies, or confirming a claim of parenthood.

Clues in DNA

Because DNA is passed from parents to offspring, scientists can use DNA to find hereditary links between people. Samples of DNA can be analyzed to find similarities and differences. People who are related by birth will share at least some of the DNA of their common ancestors. DNA can be extracted from living cells and from dead cells in hair or bone.

Mother's or Father's DNA

Every cell in a person's body contains DNA from both parents. However, chromosome recombination makes it hard to tell which DNA came from which parent. But two kinds of DNA are unique. One kind is the DNA in a Y chromosome in males. The Y chromosome always comes from the father.

Mitochondrial DNA is also unique. The DNA in the mitochondria of cells is unrelated to the DNA in the nucleus. In humans, the mitochondria in all cells have been copied from the mother's egg cell. So, your mitochondrial DNA is the same as your mother's.

Analyses of these two kinds of DNA have solved crimes and mysteries. In some cases, people have been able to learn their true family history. In other cases, the identity of a dead body has been confirmed (or not) based on DNA comparisons with living or dead relatives.

Who is buried here? This tomb in the Cathedral of Seville, Spain, is supposed to contain the remains of Christopher Columbus. But the history of the remains is disputed, and some people claim that the true remains lie elsewhere. DNA analysis may solve the case.

Whose coffin is this? In 2005, Hurricane Katrina caused disastrous flooding in areas along the coast of the Gulf of Mexico. Many coffins floated out of burial sites. To help find the living or dead relatives of unidentified bodies, scientists could compare DNA samples. For example, mitochondrial DNA will be identical among siblings.

Quick Project Find out the latest findings from DNA analyses of the supposed remains of Christopher Columbus.

Differentiated Instruction

Advanced Learners/GATE

The Mitochondrial DNA Question Have students investigate how mtDNA sampling has been used to compare Neanderthals to modern *Homo sapiens.* LS Verbal

Why It Matters

Teacher's Notes Each cell's approximately 1,700 mitochondria contain an identical loop of DNA containing 37 genes. Mitochondrial DNA, mtDNA, contained in the cytoplasm of the egg, is retained during fertilization, while the mtDNA of the sperm is not, because only the nuclear DNA joins the egg nucleus. As a result, all of the mtDNA in all cells of an individual are that of the mother. Because this DNA is passed only through the mother, it can be used to trace human ancestry.

READING TOOLBOX

Visual Literacy Refer students to the picture of the coffin, and ask them to infer what might remain of a body that was buried for a long time. (bones, teeth, hair) Explain that hair shafts, bone, and teeth contain much more mitochondrial DNA (mtDNA) than nuclear DNA; this is another reason why mitochondrial DNA is used in forensics. LS Visual

Answer to Research

Both Spain and the Dominican Republic claim to have the authentic remains of Columbus. Students should report whether DNA testing has been used and what results, if any, were reported.

Lab

Time Required

135 min (45 min/day over 3 days)

Ratings

Teacher Prep	
Student Setup	
Concept Level	
Cleanup	

Safety Cautions

Review with students all safety procedures related to the use of chemicals and electricity. Consult labels and packaging for guidelines regarding the handling and disposal of chemical solutions and samples. Ensure that students use electrical equipment in a safe, dry area, that electrical outlets have ground-fault protection devices (GFCIs), and that wires are properly connected before power is applied.

Tips and Tricks

Timing To finish this lab over two or three class periods, pause at the end of step 5, step 11, and step 14.

- Initial setup and loading of the chamber: 30–45 min
- Running the gel: 0.5–24 h (exact time depends on the applied voltage; higher voltage, faster rate)
- Staining process: 2–24 h
- Rinsing the gels: 5–15 min

Materials If you have not performed electrophoresis before, consider buying a "starter" kit or a PAGE Protein Analysis kit to gain familiarity with the process and materials. To make your own chambers, consult the Chapter Resource File for this chapter.

- For fish samples, obtain small amounts of fresh shark, sturgeon, walleye, catfish, perch, trout, or salmon; variety is best.
- For protein separation, it is best to use polyacrylamide (PAGE) gels (15% Tris-HCl) and vertical electrophoresis chambers. An alternative is to use agarose gels (4% low-melt) and horizontal chambers, but these will take longer and will not separate the proteins clearly. Note time and voltage differences for running each type of gel.
- It is safest and easiest to buy gels that are precast in trays that fit your electrophoresis chamber. You can precast your own agarose gels, but raw polyacrylamide is hazardous.
- Always read and follow any specific instructions that come with gel materials and your electrophoresis chamber.
- Use caution when working with high voltages. Each chamber should come with an appropriate high voltage DC source.
- To save time, prepare the protein extracts (steps 2–5) ahead of time and store them on ice.
- The protein buffer solution (also called solubilization buffer) should be appropriate to the kind of gel being used. If using PAGE gels, use SDS buffer. This contains SDS (sodium dodecyl sulfate, a detergent to dissolve and polarize proteins), tris buffer (to adjust pH), glycerol (to weight down the samples in the wells), and bromophenol blue (a dye that can be seen during electrophoresis, to show the "front" of the moving molecules). If using agarose gels, use a TBE (Tris-Borate-EDTA) buffer and add bromophenol blue. Follow any directions that come with the buffer solution.

Skills Practice

Chapter 14 Lab

Objectives

- Perform a protein assay to detect the results of gene expression.
- Use gel electrophoresis and staining to detect size differences.
- Infer the presence of similar genes in different species.

Materials

- lab apron, safety goggles, and disposable gloves
- fish muscle samples (3 to 6 unknowns)
- microtubes, flip-top or screw-top (6 to 12)
- protein buffer solution with dye
- water bath
- precast gel for electrophoresis chamber
- micropipettes or tips, sterile, disposable (3 to 6)
- electrophoresis chamber with power supply and wires
- running buffer solution
- gel staining tray
- protein stain solution
- water, distilled

Safety

Protein Detection

Because genes code for proteins, the presence of specific proteins in cells indicates the presence of specific genes. In this lab, you will detect the presence of specific proteins in several species of fishes. You will separate the different proteins by using gel electrophoresis.

Electrophoresis relies on a simple fact of biochemistry: opposite charges attract one another, and like charges repel. So, molecules that have a charge can be pulled around by an electric field. To separate molecules by electrophoresis, the molecules can be pulled through a microscopic "obstacle course" that will slow down larger molecules. If the "course" is "run" for a time, the molecules will be sorted by size.

In *gel electrophoresis*, the "obstacle course" is a slab of jellylike material, simply referred to as a *gel*. Several types of gels can be used to separate samples of DNA, RNA, or proteins. The samples can be stained to see where the parts ended up.

Procedure

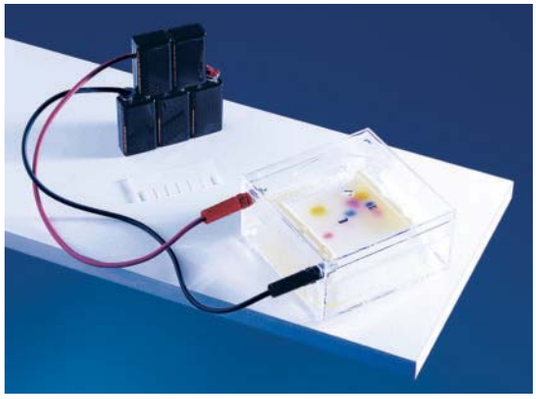
Prepare Protein Samples

1. Put on a lab apron, safety goggles, and gloves. Read all procedures, and prepare to collect your data. For each sample of fish muscle, record the type of fish, and assign it a code letter. Then, mark the letter onto two microtubes.
2. **CAUTION: Never eat or taste food in the lab.** Obtain a small piece of each fish muscle sample. Place each piece in a microtube that has the correct code label.
3. **CAUTION: Never taste chemicals or allow them to contact your skin.** For each sample, add enough protein buffer solution to cover the sample piece. Cap the tube, then gently flick it to mix the contents. The buffer will cause some of the proteins from each fish muscle sample to become suspended in the solution.
4. Let the tubes sit at room temperature for 5 min. Then, pour just the liquid from each into the second tube with the matching label. Keep the samples on ice until used.
5. **CAUTION: Use extreme caution when working with heating devices.** Heat the samples in the water bath at 95 °C for 5 min. The heat will cause the proteins from each fish muscle sample to denature.

Separate Proteins by Gel Electrophoresis

6. Examine the gel that is precast within its chamber. Note the row of small wells along one edge. These wells are where you will place the samples to be separated. Keep the gel level as you work.
7. Slowly add running buffer solution to the chamber. Add just enough to flood the wells and to cover the gel surface with buffer about 2 mm deep. Be careful not to damage the gel while pouring.

8. Using a clean micropipette, transfer 10 µL of one sample solution into one well. Be careful not to overflow the well or puncture the gel with the pipette tip. Record the sample's "lane" position.
9. Repeat step 8 for each sample. Make sure to use a clean pipette for each transfer.
10. **CAUTION: Use caution when working with electrical equipment.** Assemble the electrophoresis chamber and power source as directed by your teacher. With your teacher's approval, connect the power supply to the chamber electrodes. The negative terminal should be connected to the electrode closest to the wells, and the positive terminal should be connected to the opposite electrode.
11. Leave the chamber running but undisturbed for the amount of time specified by your teacher. During this time, the samples and dye should move toward the positive side of the gel.



View the Separated Proteins

12. When the moving front of the dye has migrated across the entire gel, disconnect the electrodes from the power source. Gently transfer the gel to the staining tray.
13. **CAUTION: Dispose of materials as directed by your teacher.** Gently pour off the buffer solution into an appropriate container as directed by your teacher.
14. Slowly pour the protein stain solution over the staining tray, and then wait for the amount of time specified by your teacher.
15. Destain the gel by soaking and rinsing it several times in distilled water. Dispose of the rinse water as directed by your teacher. Some of the stain will remain on the proteins in the gel. Draw, photograph, or photocopy the gel for analysis.
16. Clean up and dispose of your lab materials and waste according to your teacher's instructions. Wash your hands before leaving the lab.

SCILINKS
www.scilinks.org
Topic: Gene Expression
Code: HX80642

Analyze and Conclude

1. SCIENTIFIC METHODS **Organizing Data** On your picture of the gel, mark the position of each visible band in each lane of the unknown samples.
2. SCIENTIFIC METHODS **Analyzing Data** Compare the numbers and positions of visible bands among all lanes. Identify which bands of the unknown samples appear to match each other. Identify which of the samples share the most similarities.

Extension

3. **Evolutionary Relationships** Make a table to compare the protein bands from each fish. Use the table to infer which fish are most closely related by heredity and which are least related. Try to draw a "family tree" showing the evolutionary relationships among these fish.

Answers to Analysis and Conclusions

1. Check that students have accurately depicted and annotated their gels.
2. All of the fish samples should have at least one band that matches (the myosin heavy chain), while some should have a few other bands in common (actin, tropomyosin, or myosin light chains). If size standards were used, some of the samples should have a few bands of smaller size (farther migration in the gel) than others.

Answer to Extension

3. Students should organize a table and sketch a "tree" that shows increasing degrees of relatedness, from more-ancient types of fish (for example, shark) near the base, to more-recent types of fish (for example, trout and salmon) on branches nearest each other at the tips of the tree.

- The water bath should be temperature-controlled; an automatic incubator is best.
- For Step 14, use a Coomassie stain.

Procedure Tips

- Unless directed otherwise, try to keep all samples, gels, and solution cool by standing in crushed ice or refrigerating.
- To reduce student time in the lab, perform steps 1 through 5 ahead of time and store samples in tubes on ice.
- In step 11, if using a vertical PAGE system, run at 200 V for 30 min; if using horizontal agarose, run at 100 V for 45 min; if using batteries, you may need several hours. Watch the advancing front of dye to gauge how fast electrophoresis is proceeding.
- In step 13, be sure to remove the gel to a separate tray before adding stain. Never add stain to the electrophoresis chamber.
- In step 14, leave the stain for several hours or overnight.

Troubleshooting

- If the samples run the wrong way when running the gel, the electrodes are reversed.
- To keep one hand steady while using a pipette, hold the wrist with the other hand, and lean the elbow of the other hand on the table. You can sometimes see scientists doing this in photos.

Enhancement To help identify bands in the gels, use standard reagents that contain known substances, to compare to the samples. Standards should be loaded in another lane of the gels and will show the expected location of those substances in your test samples. Look for actin and myosin standards to help locate these proteins in the fish muscle samples. You might also use a molecular-mass or "kaleidoscope" marker.

Key Resources

- Holt Lab Generator
- Lab Datasheet (Levels A, B, C)
- Holt Science Biology Video Labs
- Virtual Investigations

Chapter 14

SUPER SUMMARY

Have students connect the major concepts in this chapter through an interactive Super Summary. Visit **go.hrw.com** and type in the keyword **HX8GNXS** to access the Super Summary for this chapter.

Reteaching Key Ideas

Genetic Change Have students give examples of the effects of genetic change. Emphasize that those genetic changes that do not interfere with the viability of the organism produce variations that may promote future success. LS Verbal

Gene Expression Have students review transcription to identify only exons as mRNA. Then use the example of the *lac* operon to illustrate how the environment controls gene expression. Reinforce that most genes in eukaryotes are not expressed. LS Verbal

Interactions With the Genome Ask students to explain why transposons, plasmids and homeoboxes are considered part of the genome. LS Logical

Chapter 14 Summary

go.hrw.com
SUPER SUMMARY
Keyword: HX8GNXS

Key Ideas	Key Terms

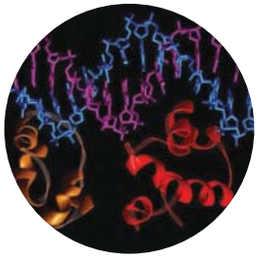
1 Mutation and Genetic Change

- For the most part, genetic differences among organisms originate as some kind of mutation.
- Different kinds of mutations are recognized as either changes in DNA or changes in the results of genes. In eukaryotic cells, the process of meiosis creates the chance of mutations at the chromosome level.
- The results of genetic change may be harmful, beneficial, or neutral; most changes are neutral and may not be passed on to offspring.
- Very large-scale genetic change can occur by misplacement, recombination, or multiplication of entire chromosomes.

mutation (319)
nondisjunction (324)
polyploidy (324)

2 Regulating Gene Expression

- Cells have complex systems that regulate whether or not specific genes are expressed, depending on the cell's needs and environment.
- The major form of gene regulation in prokaryotes depends upon operons that respond to environmental factors.
- Gene regulation in eukaryotes is more complex and variable than gene regulation in prokaryotes.
- The sequence of amino acids in a protein determines its three-dimensional structure and chemical behavior.



operon (326)
transcription factor (327)
intron (328)
exon (328)
domain (329)

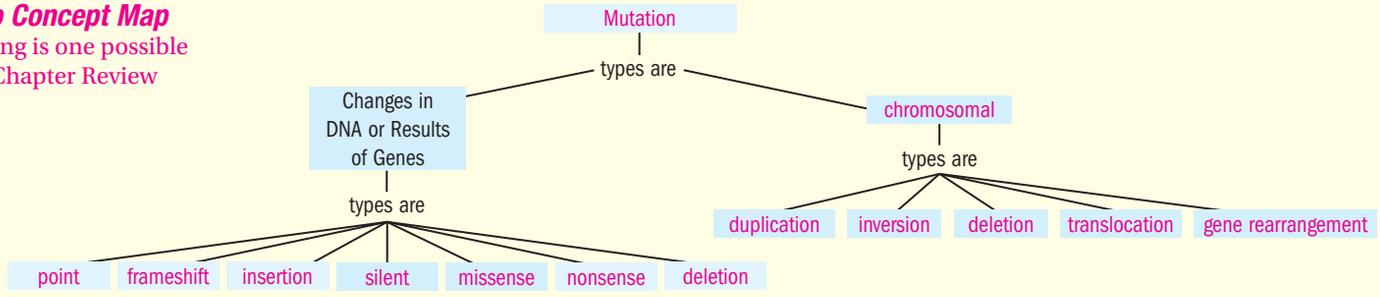
3 Genome Interactions

- Comparisons among the genetic systems of many organisms reveal basic biological similarities and relationships.
- Small bits of genetic material can be stored, moved, and changed by a variety of interactions.
- Each cell within a developing body will express specific genes, depending on the cell's age and location within the body.

genome (370)
plasmid (331)
transposon (332)
cell differentiation (333)
apoptosis (334)

Answer to Concept Map

The following is one possible answer to Chapter Review question 2.



Chapter 14 Review

READING TOOLBOX

1. **Prefixes** Use a dictionary to look up the meanings of the prefixes *non-* and *dis-* and the word *junction*. Use this information to explain the meaning of the term *nondisjunction*.
2. **Concept Map** Construct a concept map that differentiates the many kinds of mutation. Try to use the following terms: *mutation, point, insertion, deletion, silent, missense, frameshift, nonsense, amino acids, chromosomal, deletion, duplication, inversion, translocation,* and *gene rearrangement*.

Using Key Terms

3. Use the following terms in the same sentence: *gene regulation, operon, transcription factor, prokaryote,* and *eukaryote.*

In each of the following sentences, replace the incorrect term(s) with the correct key term(s).

4. New alleles arise from *gene regulation*.
5. *Mutation* is the condition of having multiple sets of chromosomes.

Understanding Key Ideas

6. All genetic differences among organisms originate as some kind of
   a. DNA. **c.** mutation.
   b. gene. **d.** amino acid.
7. A point mutation occurs when
   a. a gene's location changes.
   b. long segments of a gene are lost.
   c. gametes are forming during meiosis.
   d. one nucleotide is replaced with a different nucleotide.
8. How does the karotype of a person with Down syndrome differ from a normal karotype?
   a. It lacks a chromosome.
   b. It has two sex chromosomes.
   c. It has twice the number of chromosomes.
   d. It has an extra copy of a single chromosome.
9. The *lac* operon shuts off the production of lactose enzymes when
   a. lactose is present. **c.** glucose is present.
   b. lactose is absent. **d.** glucose is absent.
10. Which of the following is found primarily in eukaryotic genomes?
    a. introns **c.** operators
    b. operons **d.** promoters
11. Plasmids are
    a. long pieces of eukaryotic DNA.
    b. circular pieces of bacterial DNA.
    c. broken pieces of large chromosomes.
    d. pieces of viral RNA that move between species.
12. An example of a mobile genetic element is
    a. a transposon. **c.** a cancer cell.
    b. a bacterium. **d.** a hox gene.
13. Programmed cell death is called
    a. aging. **c.** apoptosis.
    b. cyclin. **d.** transposition.

Explaining Key Ideas

14. **Explain** how a genetic disorder can result from a mutation.
15. **Summarize** the role of genetic switches in gene expression.
16. **Outline** the roles of proteins in gene expression.
17. **List** ways that genetic material can be transferred by mechanisms other than chromosomes.

This image shows that specific genes regulate the development of specific parts of animal bodies. Use the image to answer the following question(s).

18. **Describe** the role of *hox* genes in development.
19. **Identify** which kind(s) of mutation could result in a group of homeotic genes being duplicated.

Assignment Guide

Section	Questions
1	1, 2, 4, 5, 6, 7, 8, 14, 19, 20, 22, 23
2	3, 9, 10, 15, 16, 24, 27, 32
3	11, 12, 13, 17, 18, 21, 25, 26, 28, 29, 30, 31

Review

Reading Toolbox

1. *Non* means "not," *dis* means "opposite," and *junction* means "joining together." Therefore, *nondisjunction* most likely means "not the opposite of joining together" which is simply "staying together."
2. See previous page for answer to concept map.

Using Key Terms

3. *Gene regulation* in *prokaryotes* involves an *operon*; *eukaryotes* rely more on *transcription factors.*
4. New alleles arise from *mutations.*
5. *Polyploidy* is the condition of having multiple sets of chromosomes.

Understanding Key Ideas

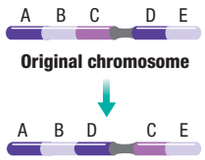
6. c 7. d 8. d 9. b
10. a 11. b 12. a 13. c

Explaining Key Ideas

14. A mutation can change one or more of the amino acids for which a gene codes. If the protein doesn't function normally, the cell(s) may not function normally.
15. Genetic switches start or stop the transcription and translation of specific genes under specific conditions.
16. Proteins serve as enzymes during replication, transcription, and translation. Proteins interact with DNA and other proteins to form genetic switching systems.
17. Exchange of plasmids in prokaryotes, reverse transcription by retroviruses, and transposons transfering genetic material.
18. Hox genes control the differentiation of cell types in animals that have a head to tail orientation.
19. a chromosomal duplication

Using Science Graphics

This image shows a specific type of mutation. Use the image to answer the following question.



20. What type of mutation is shown in the image?
- **a.** chromosomal deletion
- **b.** chromosomal inversion
- **c.** chromosomal duplication
- **d.** chromosomal translocation

21. Use this table to make a graph. Be sure to choose the appropriate graph type and to add labels.

Genetic Similarity Between Humans and Other Species*

Common name	Genus name	Similarity
Dog	*Canis*	81%
Mouse	*Mus*	79%
Rat	*Rattus*	70%
Chicken	*Gallus*	67%
Pufferfish	*Takifugu*	65%
Zebrafish	*Danio*	63%
Fruit fly	*Drosophila*	39%
Bee	*Apis*	33%
Nematode	*Caenorhabditis*	31%
Mouse-ear cress	*Arabidopsis*	17%
Rice	*Oryza*	15%
Slime mold	*Dictyostelium*	16%
Yeast	*Saccharomyces*	11%
Bacterium	*Escherichia*	10%

* percentage of human genes that are similar to genes in each of these species

Critical Thinking

22. Applying Logic What are the possible effects of a point mutation on the results of a gene? Starting with the same codon, give specific examples.

23. Applying Related Information Can genetic changes due to nondisjunction occur in bacteria?

24. Evaluating an Argument A classmate states that damage to introns is very likely to affect the synthesis of a protein but damage to exons is not. Argue against this statement.

25. Analyzing Language Scientists often state that the genetic code is universal. Explain what this statement means.

26. Inferring Relationships What does the presence of similar homeotic gene sequences among many eukaryotes suggest about evolutionary relationships among these organisms?

Connecting Key Ideas

27. Genomes and Reproduction In what ways do mobile genetic elements and sexual reproduction have similar functions?

28. Genomes and Ecology What is the main difference between the symbiosis of lichen and the endosymbiosis that is thought to have led to mitochondria and chloroplasts?

29. Genomes and Evolution Antibiotic resistance in bacteria is often used as an example of coevolution. Explain the connection.

Methods of Science

30. Experimental Setup The scientist who first suspected, and later discovered, the existence of transposons was Barbara McClintock. She conducted her main experiments by using corn kernels that were the product of many generations of self-fertilization of the same corn plants. Why did she use this kind of corn?

Writing for Science

31. Writing for an Audience Draft a lecture for younger students entitled "Are We Controlled by DNA?"

Technology Skills

32. Using Computer Graphics Use library or Internet resources to find information about technologies for three-dimensional modeling and visualization of complex biological molecules. Bring or show examples to your class of three-dimensional models of DNA, RNA, and proteins.

Using Science Graphics

20. b

21. Most students will construct a bar graph. Suggest that they place percentages on the *x*-axis and common names on the *y*-axis. This arrangement makes the graph easier to interpret.

Critical Thinking

22. Sample answer:

Silent: the codon UAU (which codes for Tyr) could be changed to UAC (which STILL codes for Tyr).

Missense: the codon UAU (which codes for Tyr) could be changed to UGU (which codes for Cys).

Nonsense: the codon UAU (which codes for Tyr) could be changed to UAA (which codes for "stop").

23. No, bacteria have only one long circular chromosome and do not replicate like a eukaryote.

24. The opposite is more likely to be true. Damage to introns, the non-coding segments, will not affect protein synthesis, but damage to exons, expressed segments, will alter protein synthesis.

25. The same four nucleotides are found in all organisms, and the same triplet combinations dictate the same amino acids to produce proteins.

26. The similarity of homeotic sequences in eukaryotes suggests that hox genes are inherited from a common ancestor.

27. Mobile genetic elements create new genetic combinations; alleles are exchanged and recombined in sexual reproduction. Both processes increase genetic variation.

28. Alga and fungus live somewhat independently in a lichen. Mitochondria and chloroplasts have lost their independence.

29. Sample answer: Humans use antibiotics to combat bacteria; bacterial genes change over time; some bacteria become resistant to antibiotics; these bacteria may pass on the resistance genes. So, the frequency of antibiotic resistance increases as humans use more antibiotics.

Methods of Science

30. If corn plants cross-fertilize, rather than self-fertilize, new genes can be introduced and effect changes. She needed to ensure that the genetic changes she observed had come from the same plant.

Writing for Science

31. Student products should emphasize the fact that DNA is the "blueprint" for protein synthesis. They should also include examples of the many uses of proteins in the body to illustrate that we are some what controlled by DNA.

Technology Skills

32. Check students' work for accurate images from reputable sources of information.

Chapter 14 Standardized Test Prep

TEST TIP If you are allowed to write on a test, mark out answers and options that you know are not possible.

Science Concepts

1. Which mutation can happen only to a chromosome?

A point **C** deletion
B silent **D** frameshift

2. Somatic cell cancer is the result of damage to

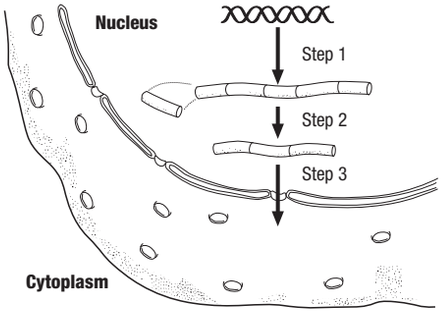
F genes that are found only in skin cells.
G genes that are found only in germ cells.
H genes that control head-to-tail orientation.
J genes that control the ability of cells to divide.

3. In the bacterium *Escherichia coli*, the *lac* operon causes the bacterium to produce certain enzymes only in the presence of lactose. The operon system is an example of

A gene mutation.
B gene regulation.
C gene duplication.
D gene differentiation.

Using Science Graphics

This diagram is a model of processes in a cell. Use the diagram to answer the following question(s).



4. If step 1 is transcription and step 3 is translation, what is happening at step 2?

F mutation of DNA **H** splicing of RNA
G copying of DNA **J** copying of RNA

5. The cell shown must belong to a(n)

A bacterium. **C** gamete.
B eukaryote. **D** prokaryote.

This table shows the genetic code, or codons of RNA that are translated into specific amino acids. Use the table to answer the following question(s).

First base	Second base				Third base
	U	C	A	G	
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	stop	stop	A
	Leu	Ser	stop	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	C
	Ile	Thr	Lys	Arg	A
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	C
	Val	Ala	Glu	Gly	A
	Val	Ala	Glu	Gly	G

6. Which of the following pairs of codons demonstrates the redundancy of the genetic code?

F AAC and AAA **H** ACA and ACG
G AUA and AUG **J** UGA and UGG

7. Suppose that the third base in the codon CUC is accidentally changed from C to G so that the codon is now CUG. What kind of mutation has occurred?

A a silent mutation
B a missense mutation
C a nonsense mutation
D a frameshift mutation

Writing Skills

8. Extended Response Write a brief essay entitled "The Pros and Cons of Genetic Mutation." Be sure to organize your essay logically, and provide examples to support your points.

State Resources

For specific resources for your state, visit go.hrw.com and type in the keyword **HSHSTR**.

Test Practice with Guided Reading Development

Answers

1. C **2.** J **3.** B
4. H **5.** B **6.** H
7. A
8. Student essays should describe the potential effects of mutations, including both positive and negative effects. Evaluate essays based on length, organization, grammar usage and mechanics. Essays should be one to three paragraphs long. Consider having students trade essays for peer review.

TEST DOCTOR

Question 4 Mutation of DNA happens before Step 1, transcription, so **F** is incorrect. Copying of DNA is replication, which is not connected to any of these processes, so **G** is incorrect. Step 2 shows how exons are spliced, so **H** is correct. Copying of RNA in transcription is identified as Step 1, not 2, so **J** is incorrect.

Question 5 Only eukaryotes have nuclei, so **A** and **D** are both incorrect, and **B** is correct. There is no way to know whether this cell is a gamete, so **C** is incorrect.

Question 7 Because the change produces the same amino acid, leucine, this is a silent mutation, and **A** is correct. A missense mutation would need to produce a different amino acid, so **B** is incorrect. Because the mutation did not produce a stop codon, it cannot be a nonsense mutation, so **C** is incorrect. **D** is incorrect because there was no insertion to produce a frameshift.