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.

Scientists have found several factors that increase the numbers of deformities in frogs. These factors include UV radiation, pesticides, and parasites. Parasites invade the frogs' bodies and may disrupt development.

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.

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

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.)***

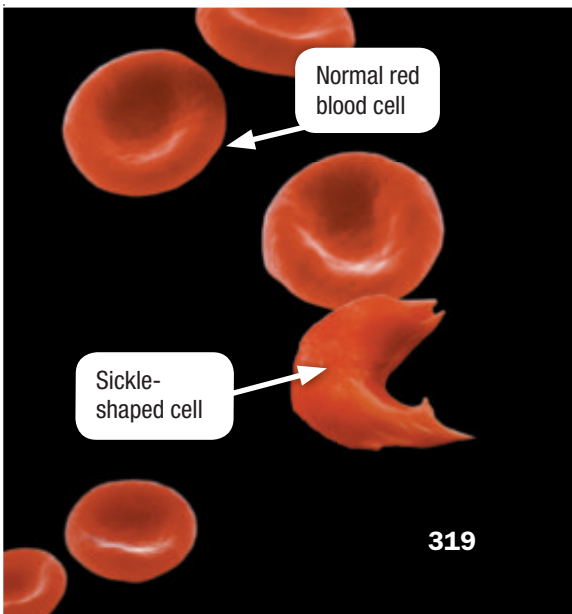

Figure 1 One out of 500 African Americans has sickle cell anemia, which is caused by a mutation in the gene that produces hemoglobin. Blood cells with the defective hemoglobin tend to bend, rupture, and get stuck.

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.

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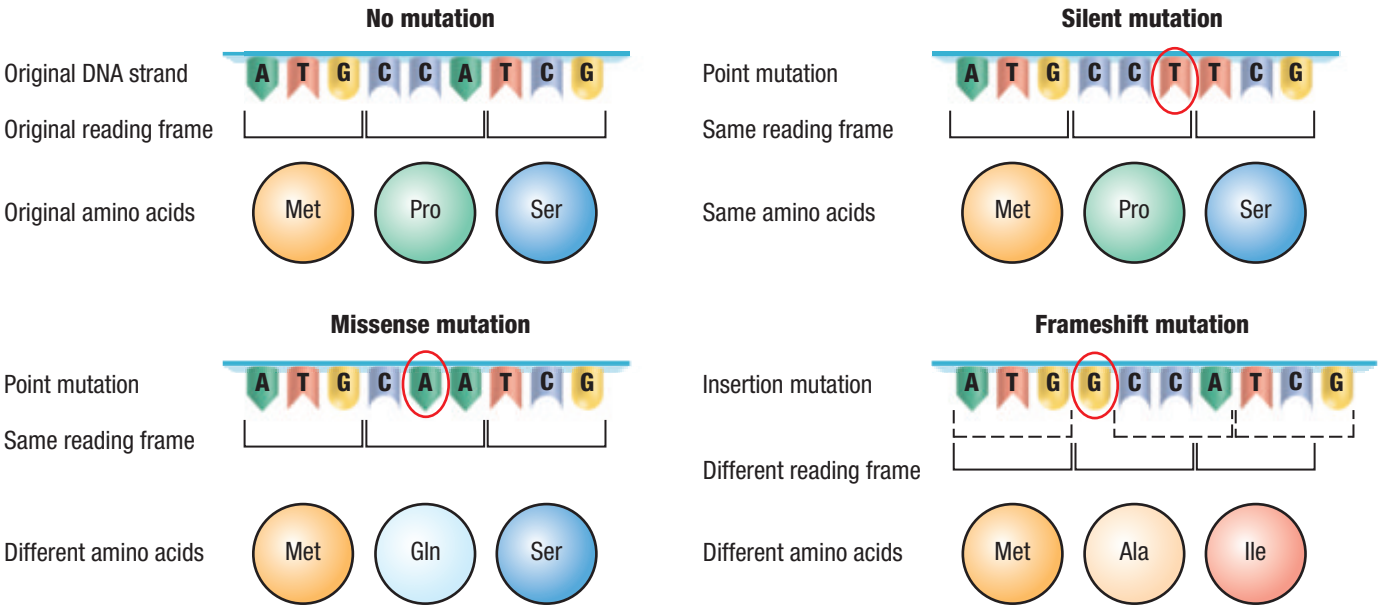
Figure 2 A mutation is a change, insertion, or deletion of one or more nucleotides in a gene. The change may or may not result in a different amino acid sequence within a protein.

Kinds of Mutations



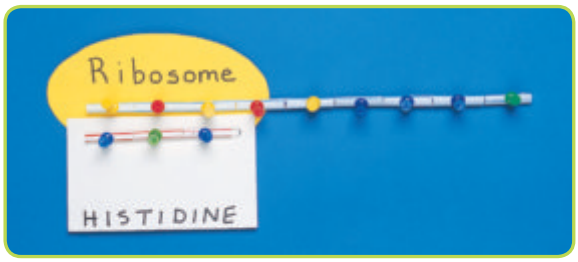
Hands-On

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.

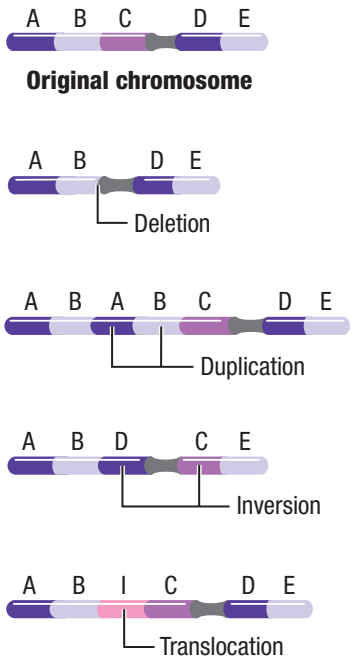
Inversion An *inversion* occurs when a piece reattaches to its original chromosome, but in a reverse direction.

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?**



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

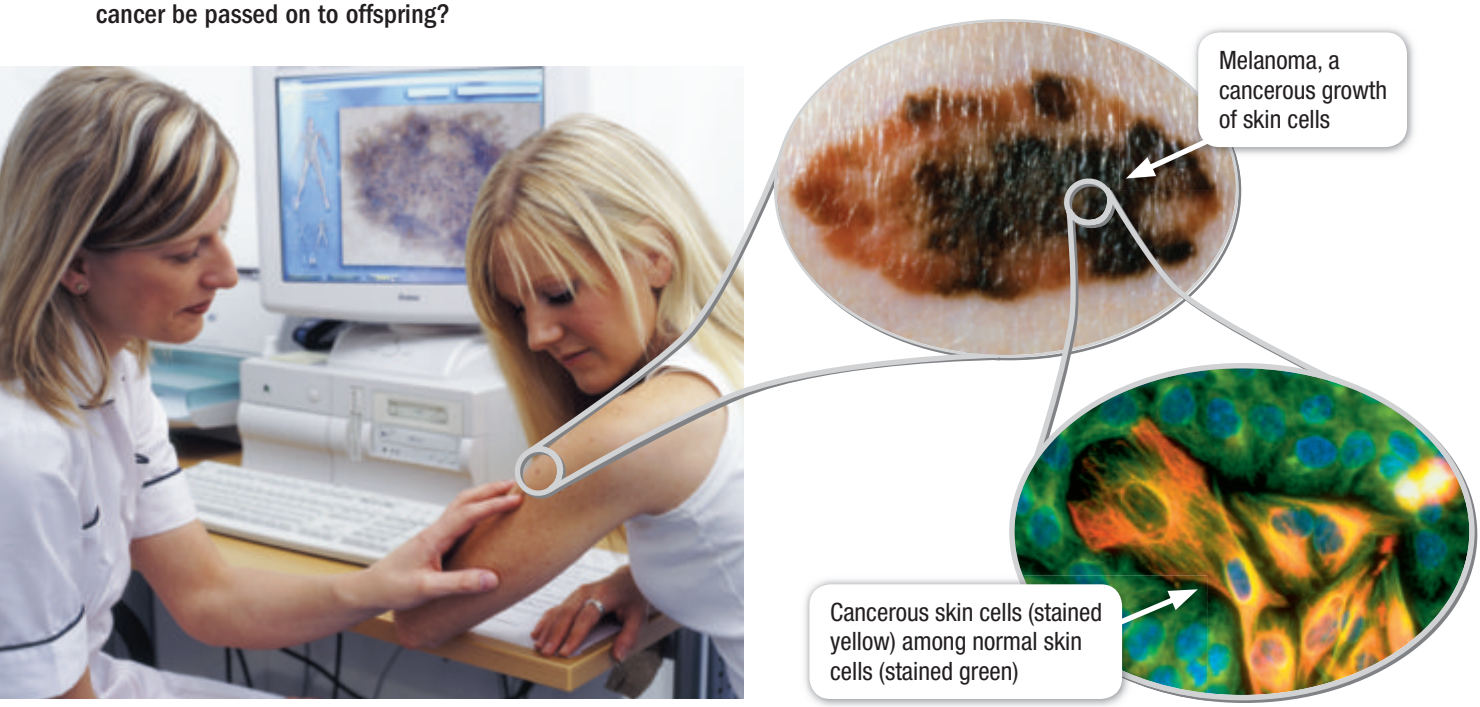


Figure 4 Melanoma is a type of skin cancer caused by mutations in melanocytes, the cells that make skin pigment. Melanoma is an example of a somatic cell cancer. **› Can this kind of cancer be passed on to offspring?**

Some Human Genetic Disorders

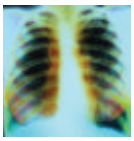
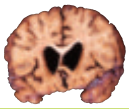
Disorder	Inheritance pattern	Major physical symptoms	Genetic effect of mutant allele	Number of cases (United States)
Sickle cell anemia 	recessive	poor blood circulation; pain; damage to organs such as liver, kidney, lungs, and heart	abnormal hemoglobin in red blood cells	72,000
Tay-Sachs disease 	recessive 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?***

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Topic: Genetic Disorders
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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.

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 protein 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?***

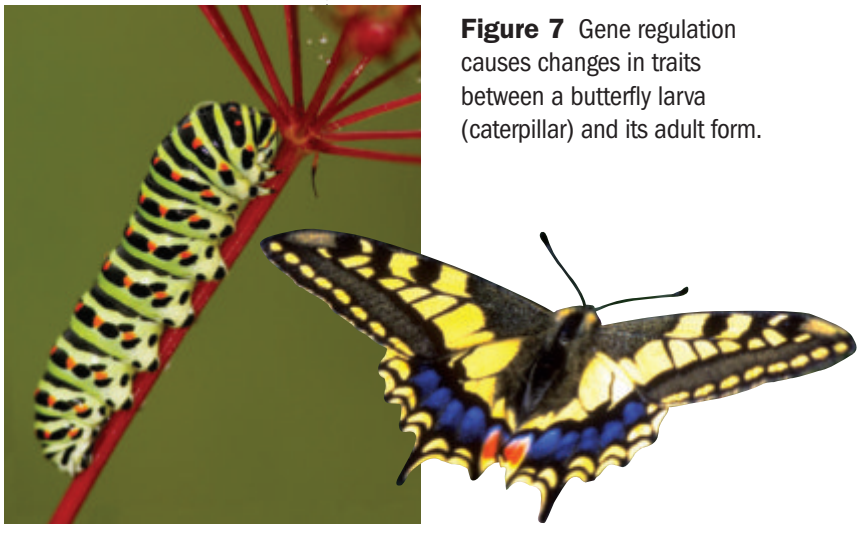
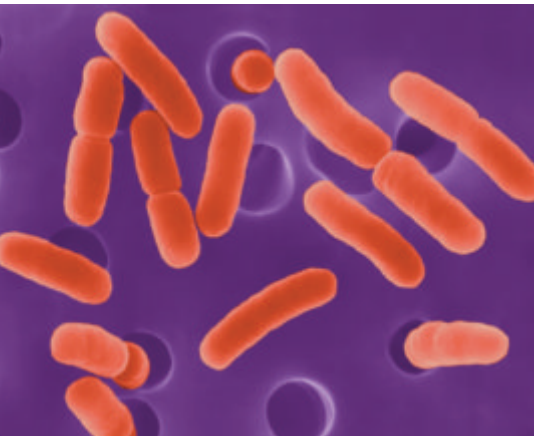


Figure 7 Gene regulation causes changes in traits between a butterfly larva (caterpillar) and its adult form.



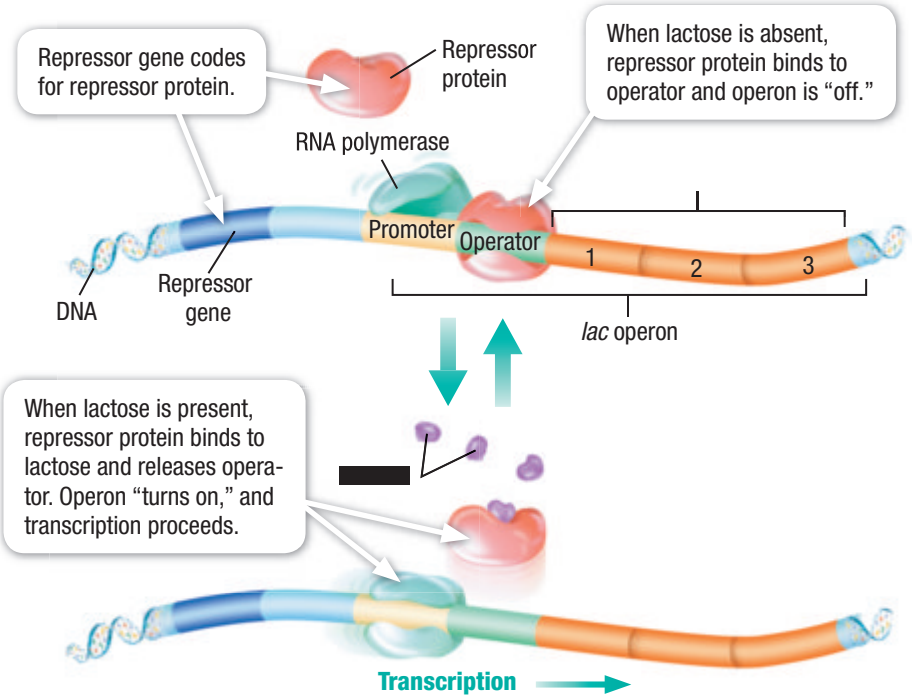


Figure 8 The *lac* operon controls the genes that code for the proteins that help a bacterium use lactose. The operon "turns on" and expresses the genes only in the presence of lactose. ❯ **How common are operons?**

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.

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 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?***

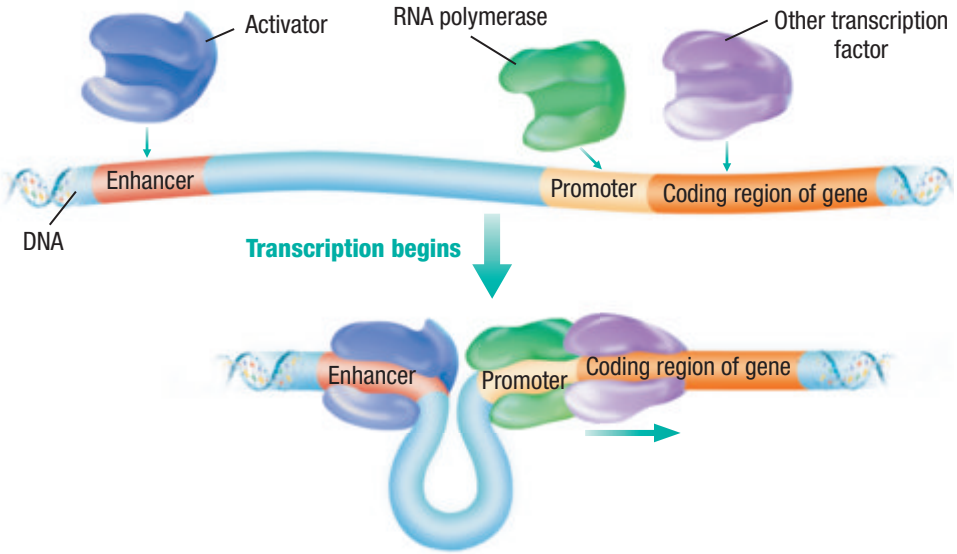


Figure 9 Control of transcription is complex in eukaryotes. For example, an activator may bind to an enhancer site and also to RNA polymerase. This action will activate another transcription factor, and finally transcription will begin.
› Why is gene regulation more complex in eukaryotes than in prokaryotes?

Hands-On

QuickLab

10 min

A Model of Introns and Exons

You can model introns and exons with masking tape.

Procedure

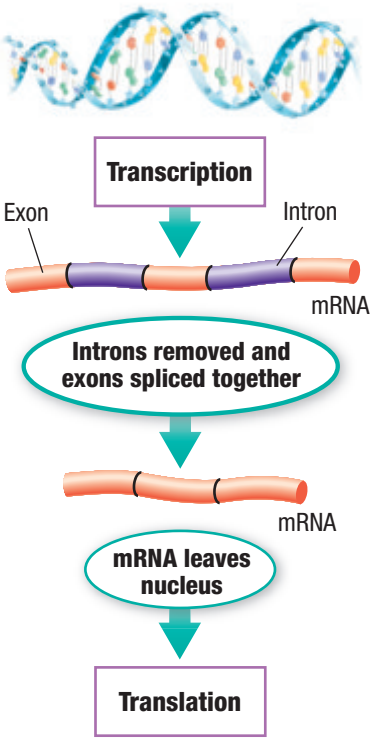
1. Place a **15 to 20 cm strip of masking tape** on your desk. The tape represents a gene.
2. Use **two colored pens** to write letters on the tape, exactly as shown in the example here. Space the letters to take up the entire length of the tape. The segments in one color represent introns; those in 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.

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?***

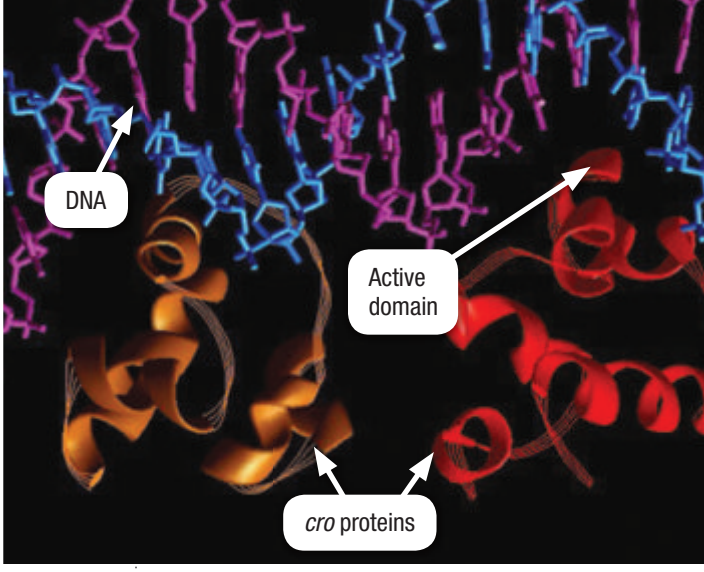


Figure 11 Many of the proteins involved in gene regulation have a shape that fits closely with DNA or RNA molecules. The example shown here is a model of two molecules of bacterial *cro* protein (orange and red) binding to a molecule of DNA (blue and purple). The parts of the protein molecules that are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

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.

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.

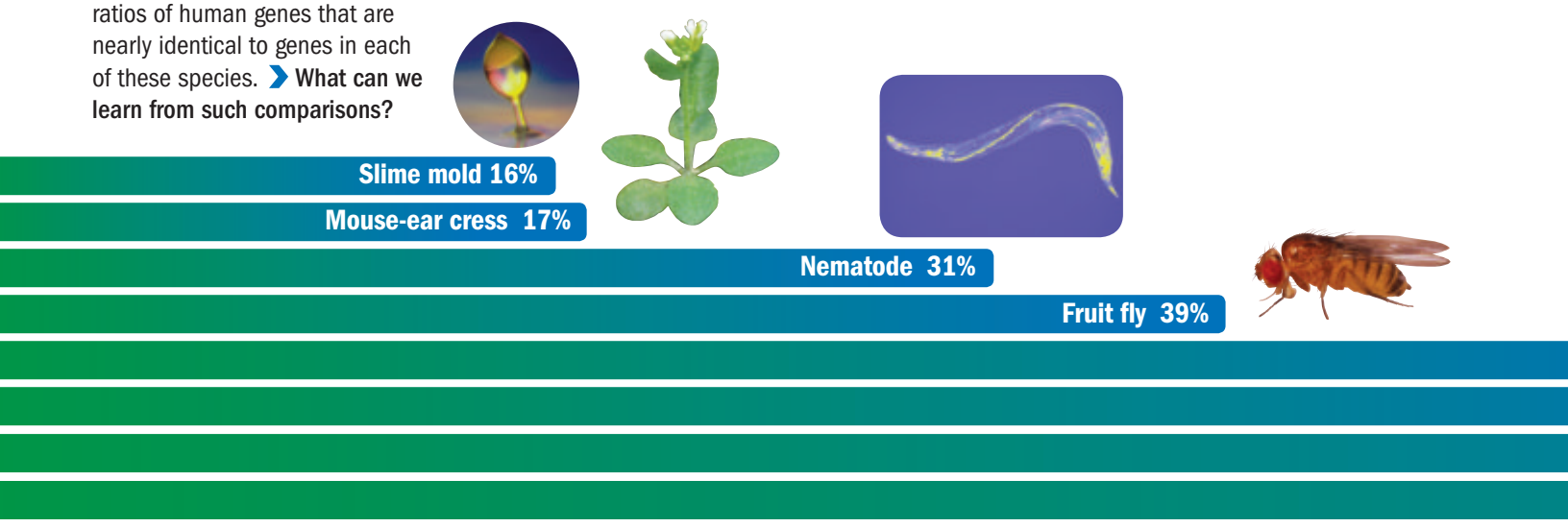


Figure 12 This graph shows the ratios of human genes that are nearly identical to genes in each of these species. ❯ **What can we learn from such comparisons?**

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 30,000 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?***

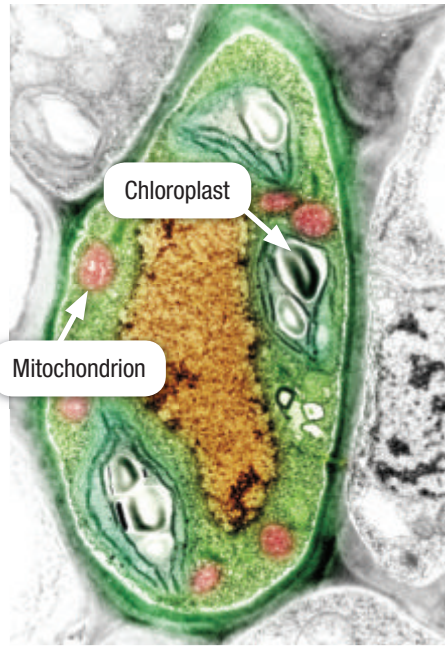


Figure 13 Chloroplasts and mitochondria have their own DNA. Each organelle's genome is stored and replicated separately from the chromosomes of the cell.

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

Zebra fish 63%

Chicken 67%

Dog 81%

Human (all genes compared to others)

***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?***

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.

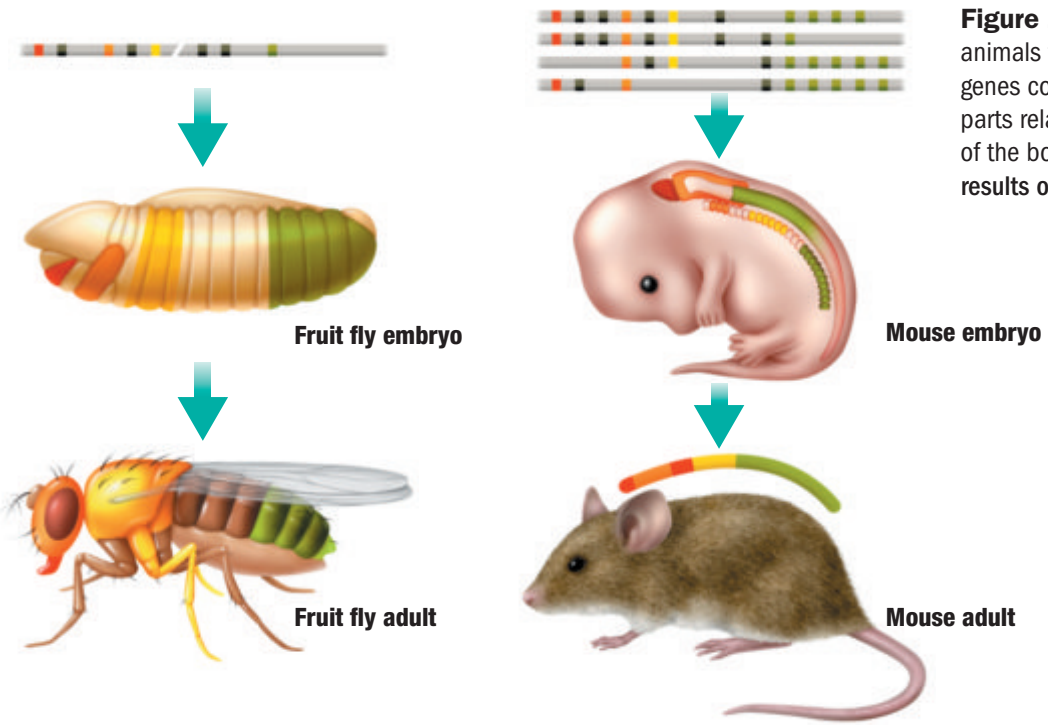


Figure 15 *Hox* genes are found in animals from insects to mammals. These genes control the development of body parts relative to the head and tail ends of the body. › **What are some possible results of mutations in these genes?**

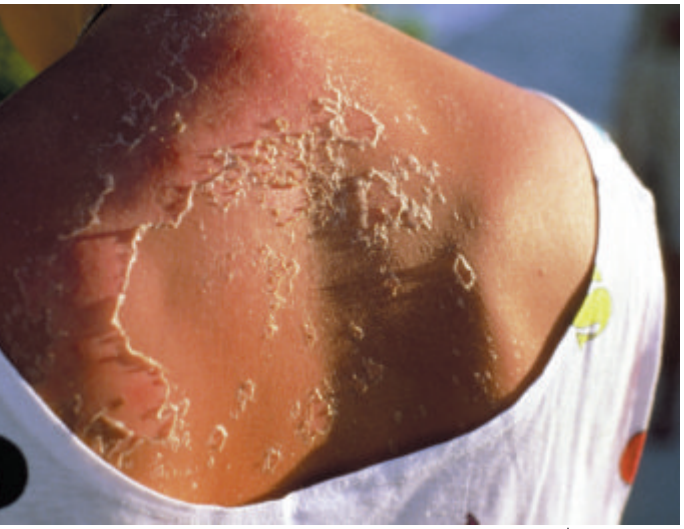


Figure 16 Sunburn is the result of apoptosis—or cell suicide. When skin cells are heavily damaged by overexposure to the sun, a genetic switch in the cells may signal the cells to stop functioning. **› Why is apoptosis important in multicellular organisms?**

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.

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

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 gene expression in 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** Could apoptosis occur in 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.

Chapter 14 Summary

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SUPER SUMMARY
Keyword: HX8GNXS

Key Ideas	Key Terms

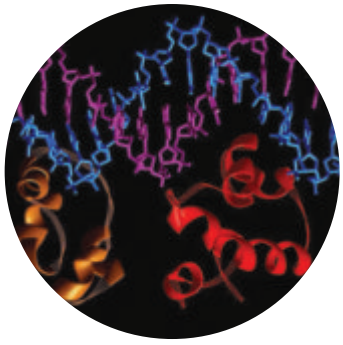
1 Mutation and Genetic Change

- 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 expression in eukaryotes is more complex and variable than gene expression 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)