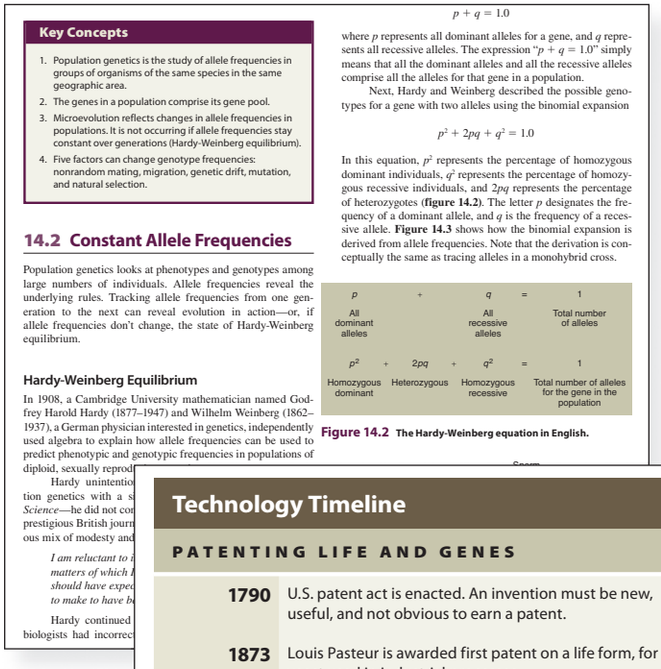
The Lewis Guided Learning System

Chapter Outline focuses attention on the major concepts.

Numbered Sections help organize the content.

Chapter Openers show how the content relates to real life.

Key Concepts boxes summarize what a student should know before leaving each numbered section.



Key Concepts

1. Population genetics is the study of allele frequencies in groups of organisms of the same species in the same geographic area.
2. The genes in a population comprise its gene pool.
3. Microevolution reflects changes in allele frequencies in populations. It is not occurring if allele frequencies stay constant over generations (Hardy-Weinberg equilibrium).
4. Five factors can change genotype frequencies: nonrandom mating, migration, genetic drift, mutation, and natural selection.

14.2 Constant Allele Frequencies

Population genetics looks at phenotypes and genotypes among large numbers of individuals. Allele frequencies reveal the underlying rules. Tracking allele frequencies from one generation to the next can reveal evolution in action—or, if allele frequencies don't change, the state of Hardy-Weinberg equilibrium.

Hardy-Weinberg Equilibrium

In 1908, a Cambridge University mathematician named Godfrey Harold Hardy (1877–1947) and Wilhelm Weinberg (1862–1937), a German physician interested in genetics, independently used algebra to explain how allele frequencies can be used to predict phenotypic and genotypic frequencies in populations of diploid, sexually reprod

Hardy unintentio
tion genetics with a s
Science—he did not co
prestigious British journ
ous mix of modesty and

I am reluctant to i
matters of which I
should have expec
to make to have b

Hardy continued
biologists had incorrec

$$p + q = 1.0$$

where p represents all dominant alleles for a gene, and q represents all recessive alleles. The expression "$p + q = 1.0$" simply means that all the dominant alleles and all the recessive alleles comprise all the alleles for that gene in a population.

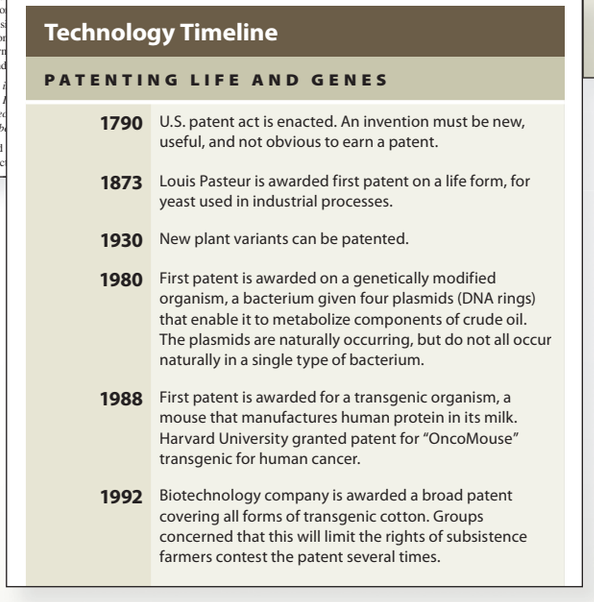
Next, Hardy and Weinberg described the possible genotypes for a gene with two alleles using the binomial expansion

$$p^2 + 2pq + q^2 = 1.0$$

In this equation, p^2 represents the percentage of homozygous dominant individuals, q^2 represents the percentage of homozygous recessive individuals, and $2pq$ represents the percentage of heterozygotes (**figure 14.2**). The letter p designates the frequency of a dominant allele, and q is the frequency of a recessive allele. **Figure 14.3** shows how the binomial expansion is derived from allele frequencies. Note that the derivation is conceptually the same as tracing alleles in a monohybrid cross.

p	+	q	=	1
All dominant alleles		All recessive alleles		Total number of alleles
p^2	+ $2pq$	+ q^2	=	1
Homozygous dominant	Heterozygous	Homozygous recessive		Total number of alleles for the gene in the population

Figure 14.2 **The Hardy-Weinberg equation in English.**



Technology Timeline

PATENTING LIFE AND GENES

1790 U.S. patent act is enacted. An invention must be new, useful, and not obvious to earn a patent.

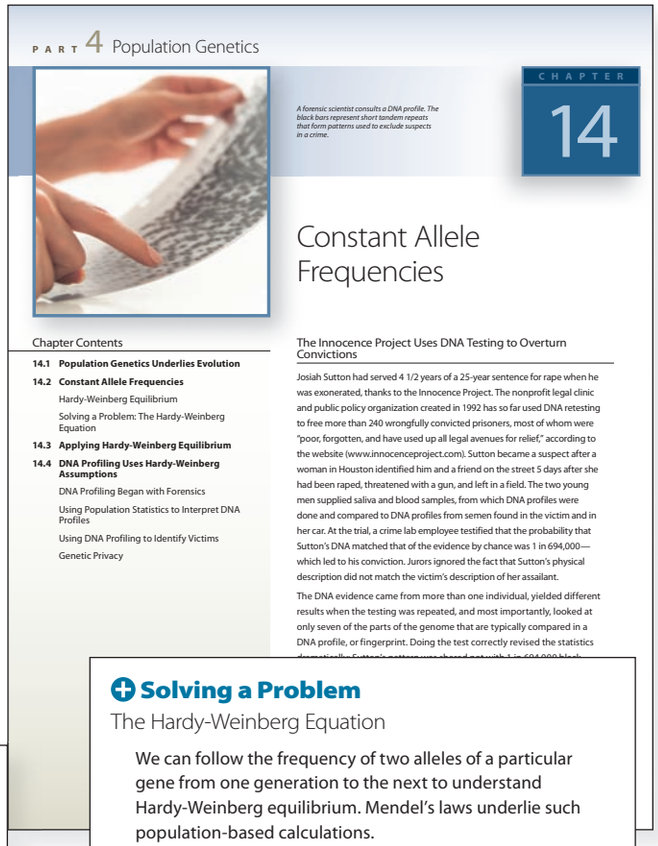
1873 Louis Pasteur is awarded first patent on a life form, for yeast used in industrial processes.

1930 New plant variants can be patented.

1980 First patent is awarded on a genetically modified organism, a bacterium given four plasmids (DNA rings) that enable it to metabolize components of crude oil. The plasmids are naturally occurring, but do not all occur naturally in a single type of bacterium.

1988 First patent is awarded for a transgenic organism, a mouse that manufactures human protein in its milk. Harvard University granted patent for "OncoMouse" transgenic for human cancer.

1992 Biotechnology company is awarded a broad patent covering all forms of transgenic cotton. Groups concerned that this will limit the rights of subsistence farmers contest the patent several times.



PART 4 Population Genetics

CHAPTER 14

A forensic scientist consults a DNA profile. The black bars represent short tandem repeats that form patterns used to exclude suspects in a crime.

Constant Allele Frequencies

Chapter Contents

The Innocence Project Uses DNA Testing to Overturn Convictions

Josiah Sutton had served 4 1/2 years of a 25-year sentence for rape when he was exonerated, thanks to the Innocence Project. The nonprofit legal clinic and public policy organization created in 1992 has so far used DNA retesting to free more than 240 wrongfully convicted prisoners, most of whom were "poor, forgotten, and have used up all legal avenues for relief," according to the website (www.innocenceproject.com). Sutton became a suspect after a woman in Houston identified him and a friend on the street 5 days after she had been raped, threatened with a gun, and left in a field. The two young men supplied saliva and blood samples, from which DNA profiles were done and compared to DNA profiles from semen found in the victim and in her car. At the trial, a crime lab employee testified that the probability that Sutton's DNA matched that of the evidence by chance was 1 in 694,000—which led to his conviction. Jurors ignored the fact that Sutton's physical description did not match the victim's description of her assailant.

The DNA evidence came from more than one individual, yielded different results when the testing was repeated, and most importantly, looked at only seven of the parts of the genome that are typically compared in a DNA profile, or fingerprint. Doing the test correctly revised the statistics

Solving a Problem

The Hardy-Weinberg Equation

We can follow the frequency of two alleles of a particular gene from one generation to the next to understand Hardy-Weinberg equilibrium. Mendel's laws underlie such population-based calculations.

Consider an autosomal recessive trait: a middle finger shorter than the second and fourth fingers. If we know the frequencies of the dominant and recessive alleles, then we can calculate the frequencies of the genotypes and phenotypes and trace the trait through the next generation. The dominant allele *D* confers normal-length fingers; the recessive allele *d* confers a short middle finger (**figure 14.4**). We can deduce the frequencies of the dominant and recessive alleles by observing the frequency of homozygous recessives, because this phenotype—short finger—reflects only one genotype. If 9 out of 100 individuals in a population have short fingers—genotype *dd*—the frequency is 9/100 or 0.09. Since *dd* equals q^2, then q equals 0.3. Since $p + q = 1.0$, knowing that q is 0.3 tells us that p is 0.7.

Next, we can calculate the proportions of the three genotypes that arise when gametes combine at random:

Homozygous dominant = *DD*
= 0.7 × 0.7 = 0.49
= 49 percent of individuals in generation 1

Homozygous recessive = *dd*
= 0.3 × 0.3 = 0.09
= 9 percent of individuals in generation 1

Heterozygous = *Dd* + *dD*
= $2pq$ = (0.7)(0.3) + (0.3)(0.7) = 0.42
= 42 percent of individuals in generation 1

Solving a Problem sections walk students step-by-step through the process of performing a genetic analysis.

In-Chapter Review Tools, such as chapter glossaries and timelines of major discoveries, are handy tools for reference and study.

Each chapter ends with a point-by-point **Chapter Summary.**

Review Questions assess content knowledge.

Summary

11.1 Gene Expression Through Time and Tissue

1. Changes in gene expression occur over time at the molecular level (globin switching), at the tissue level (blood plasma), and at the organ/gland level (pancreas development).
2. **Proteomics** catalogs the types of proteins in particular cells, tissues, organs, or entire organisms under specified conditions.

11.2 Control of Gene Expression

3. Acetylation of certain histones enables the transcription of associated genes. Phosphorylation and methylation are also important in **chromatin remodeling.**
4. MicroRNAs bind to certain mRNAs, blocking translation.

11.3 Maximizing Genetic Information

5. A small part of the genome encodes protein, but these genes specify a much greater number of proteins.
6. Alternate splicing, use of introns, and cutting proteins translated from a single gene contribute to protein diversity.

11.4 Most of the Human Genome Does *Not* Encode Protein

7. The nonprotein-encoding part of the genome includes viral sequences, noncoding RNAs, introns, promoters and other controls, and repeats.

www.mhhe.com/lewisgenetics9

Answers to all end-of-chapter questions can be found at **www.mhhe.com/lewisgenetics9**. You will also find additional practice quizzes, animations, videos, and vocabulary flashcards to help you master the material in this chapter.

Review Questions

1. Why is control of gene expression necessary?
2. Define *epigenetics.*
3. Distinguish between the type of information that epigenetics provides and the information in the DNA sequence of a protein-encoding gene.
4. Describe three types of cells and how they differ in gene expression from each other.
5. Explain how a mutation in a promoter can affect gene expression.
6. What is the environmental signal that stimulates globin switching?
7. How does development of the pancreas illustrate differential gene expression?
8. How do histones control gene expression, yet genes also control histones?
9. Name a mechanism that silences transcription of a gene and a mechanism that blocks translation of an mRNA.
10. What controls whether histones enable DNA wrapped around them to be transcribed?
11. What are two ways that microRNA functioning is complex?
12. Describe three ways that the number of proteins exceeds the number of protein-encoding genes in the human genome.
13. How can alternate splicing generate more than one type of protein from the information in a gene?
14. In the 1960s, a gene was defined as a continuous sequence of DNA, located permanently at one place on a chromosome, that specifies a sequence of amino acids from one strand. List three ways this definition has changed.
15. Give an example of a discovery mentioned in the chapter that changed the way we think about the genome.

Applied Questions

1. Several new drugs inhibit the enzymes that either put acetyl groups on histones or take them off. Would a drug that combats a cancer caused by too little expression of a gene that normally suppresses cell division add or remove acetyl groups?
2. Chromosome 7 has 863 protein-encoding genes, but many more proteins. The average gene is 69,877 bases, but the average mRNA is 2,639 bases. Explain both of these observations.
3. CHARGE syndrome (MIM 214800) causes heart defects, visual problems, facial palsy, blocked nostrils, and difficulty swallowing. A mutation in a gene called *Chd1* causes the condition. The protein product of this gene recognizes and binds methyl groups on certain histones. Explain how this mutation leads to pleiotropy (multiple symptoms).
4. How many different proteins encompassing two exons can be produced from a gene that has three exons?
5. Many people with trisomy 21 Down syndrome (an extra chromosome 21; see section 13.3) who survive into adulthood develop early-onset Alzheimer disease. The APP gene which when mutant causes this form of Alzheimer disease is on chromosome 21. Explain how this form of Alzheimer disease in trisomy 21 individuals differs from the same disorder caused by a mutation in the APP promoter in a person who has the normal two copies of chromosome 21.

Web Activities

6. Gene expression profiling tests began to be marketed just a few years ago. Google "Oncotype DX," "MammaPrint," or simply "gene expression profiling in cancer" and describe how classifying a particular cancer based on gene expression profiling can improve diagnosis and/or treatment. (Or apply this question to a different type of disease.)

Case Studies and Research Results

7. Jerrold is 38 years old. His body produces too much of the hormone estrogen, which has enlarged his breasts. He had a growth spurt and developed pubic hair by age 5, and then his growth dramatically slowed so that his adult height is well below normal. He has a very high-pitched voice and no facial hair, which reflect the excess estrogen. Jerrold's son, Timmy, is 8 years old and has the same symptoms.

   Jerrold and Timmy have an overactive gene for aromatase, an enzyme required to synthesize estrogen. Five promoters control expression of the gene in different tissues, and each promoter is activated by a different combination of hormonal signals. The five promoters lead to estrogen production in skin, fat, brain, gonads (ovaries and testes) and placenta. In premenopausal women, the ovary-specific promoter is highly active, and estrogen is abundant. In men and postmenopausal women, however, only small amounts of estrogen are normally produced, in skin and fat. The father and son have a wild type aromatase gene, but high levels of estrogen in several tissues, particularly fat, skin, and blood. They do, however, have a mutation that turns around an adjacent gene so that the aromatase gene falls under the control of a different promoter. Suggest how this phenotype arises.
8. Margaret is 102 years old, and she still walks at least half a mile a day, albeit slowly. She is a trim vegetarian who has rarely been ill her entire life. Morris is an obese, balding 62-year-old man who has high blood pressure and colon cancer. How might their proteome portraits, such as the one in figure 11.4, differ? (Hint: Reread Reading 3.1, Genes and Longevity.)

Forensics Focus

9. Establishing time of death is critical information in a murder investigation. Forensic entomologists can estimate the "postmortem interval" (PMI), or the time at which insects began to deposit eggs on the corpse, by sampling larvae of specific insect species and consulting developmental charts to determine the stage. The investigators then count the hours backwards to estimate the PMI. Blow flies are often used for this purpose, but their three larval stages look remarkably alike in shape and color, and development rate varies with environmental conditions. With luck, researchers can count back 6 hours from the developmental time for the largest larvae to estimate the time of death.

   In many cases, a window of 6 hours is not precise enough to narrow down suspects when the victim visited several places and interacted with many people in the hours before death. Suggest a way that gene expression profiling might be used to more precisely define the PMI and extrapolate a probable time of death.

Applied Questions help students develop problem solving skills.

Web Activities encourage students to use the latest tools and databases in genetic analysis.

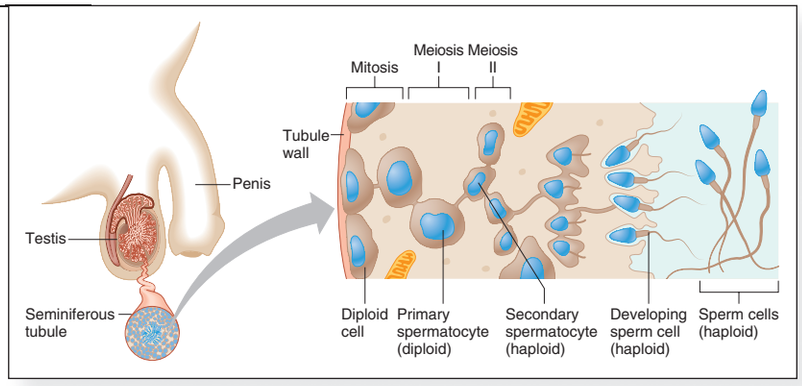
Cases and Research Results use stories taken from headlines, journals, and even fiction as the basis for questions that ask students to analyze data or predict results.

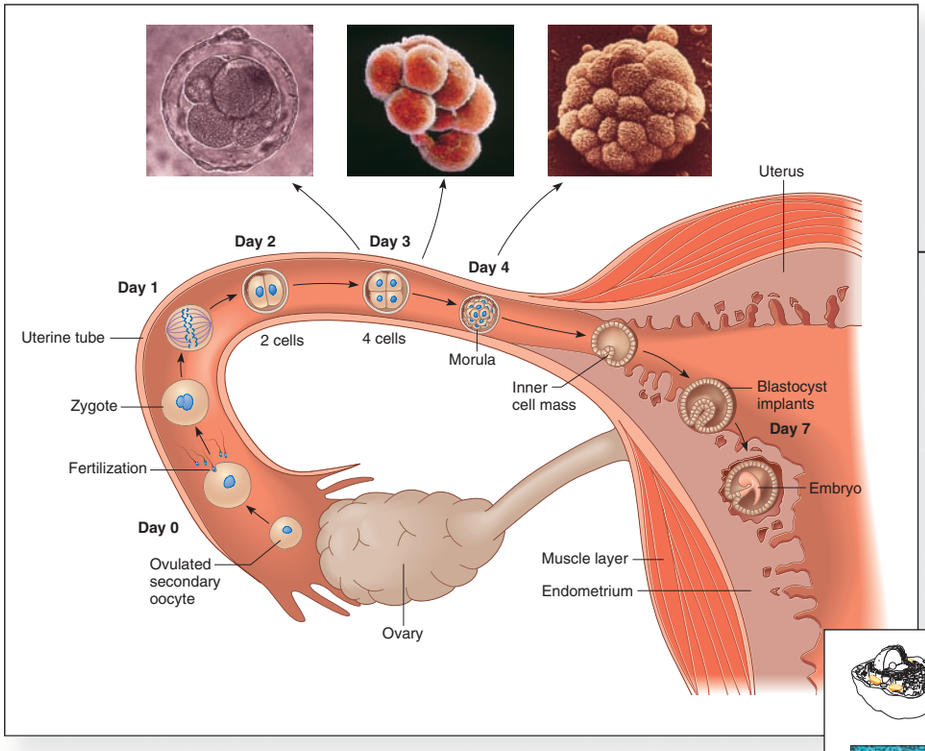
Capitalizing on students' interest in forensic science, new **Forensics Focus** questions make students think about the genetic principles involved in the collection and use of genetic information in criminal investigations.

Dynamic Art Program

Multilevel Perspective

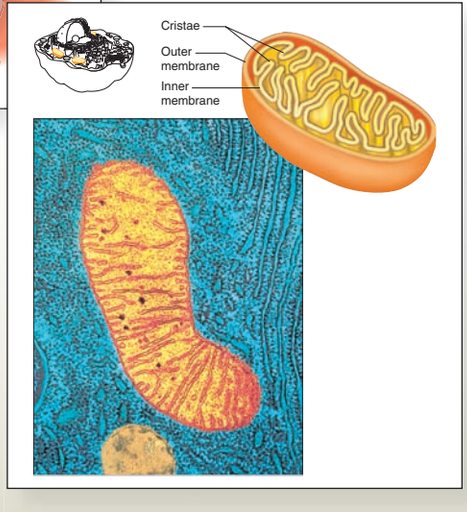
Illustrations depicting complex structures show macroscopic and microscopic views to help students see the relationship between increasingly detailed drawings.





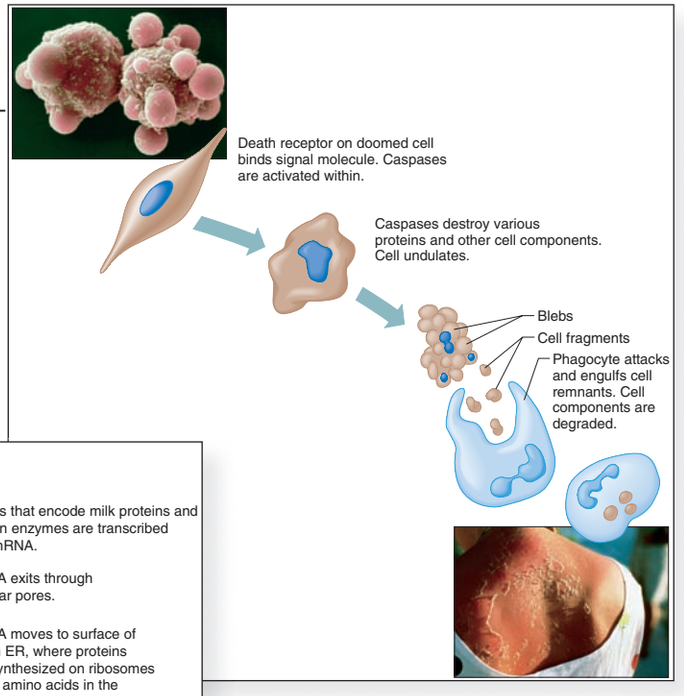
Combination Art

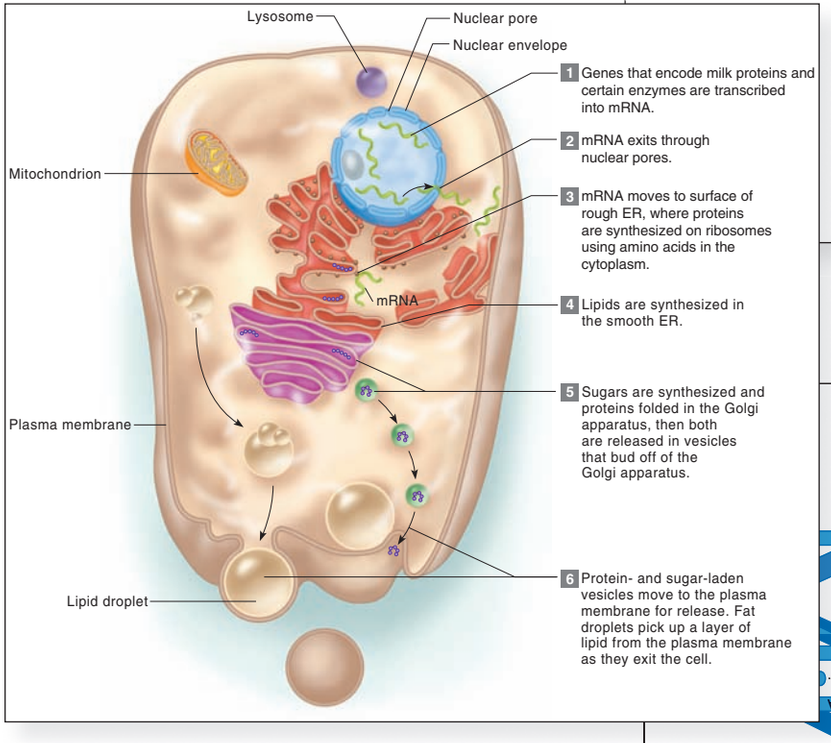
Drawings of structures are paired with micrographs to give the student the best of both perspectives: the realism of photos and the explanatory clarity of line drawings.



Complex Content in Context

Molecular and cellular information is put into a familiar context to help students make connections.





Process Figures

Complex processes are broken down into a series of smaller steps that are easy to follow. Here, organelles interact to produce and secrete a familiar substance—milk.