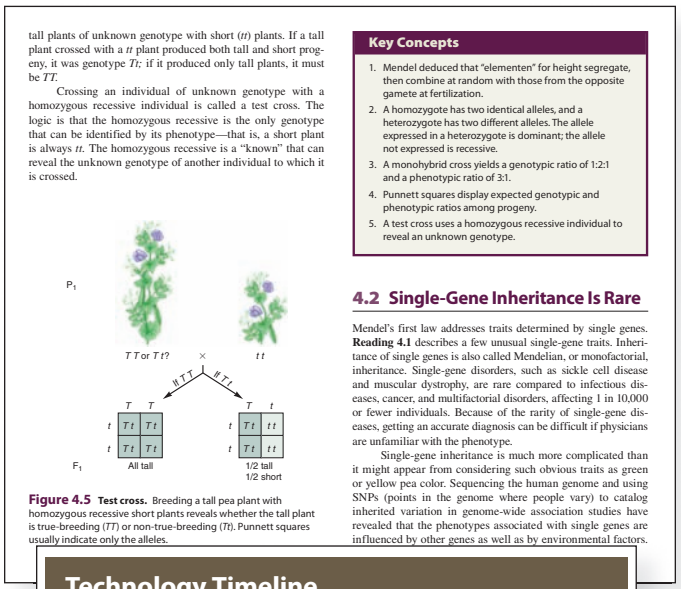
The Lewis Guided Learning System

Learning Outcomes enable students to practice and apply learning in the real world.

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.



tall plants of unknown genotype with short (*tt*) plants. If a tall plant crossed with a *tt* plant produced both tall and short progeny, it was genotype *Tt;* if it produced only tall plants, it must be *TT*.

Crossing an individual of unknown genotype with a homozygous recessive individual is called a test cross. The logic is that the homozygous recessive is the only genotype that can be identified by its phenotype—that is, a short plant is always *tt*. The homozygous recessive is a "known" that can reveal the unknown genotype of another individual to which it is crossed.

Figure 4.5 **Test cross.** Breeding a tall pea plant with homozygous recessive short plants reveals whether the tall plant is true-breeding (*TT*) or non-true-breeding (*Tt*). Punnett squares usually indicate only the alleles.

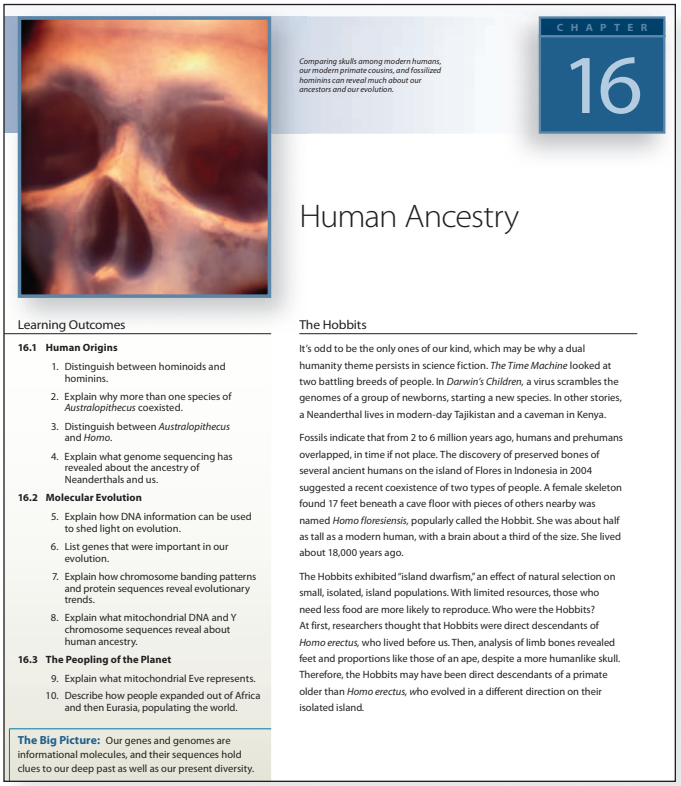
Key Concepts

1. Mendel deduced that "elementen" for height segregate, then combine at random with those from the opposite gamete at fertilization.
2. A homozygote has two identical alleles, and a heterozygote has two different alleles. The allele expressed in a heterozygote is dominant; the allele not expressed is recessive.
3. A monohybrid cross yields a genotypic ratio of 1:2:1 and a phenotypic ratio of 3:1.
4. Punnett squares display expected genotypic and phenotypic ratios among progeny.
5. A test cross uses a homozygous recessive individual to reveal an unknown genotype.

4.2 Single-Gene Inheritance Is Rare

Mendel's first law addresses traits determined by single genes. **Reading 4.1** describes a few unusual single-gene traits. Inheritance of single genes is also called Mendelian, or monofactorial, inheritance. Single-gene disorders, such as sickle cell disease and muscular dystrophy, are rare compared to infectious diseases, cancer, and multifactorial disorders, affecting 1 in 10,000 or fewer individuals. Because of the rarity of single-gene diseases, getting an accurate diagnosis can be difficult if physicians are unfamiliar with the phenotype.

Single-gene inheritance is much more complicated than it might appear from considering such obvious traits as green or yellow pea color. Sequencing the human genome and using SNPs (points in the genome where people vary) to catalog inherited variation in genome-wide association studies have revealed that the phenotypes associated with single genes are influenced by other genes as well as by environmental factors.



CHAPTER 16

Comparing skulls among modern humans, our modern primate cousins, and fossilized hominins can reveal much about our ancestors and our evolution.

Human Ancestry

Learning Outcomes

16.1 Human Origins

1. Distinguish between hominoids and hominins.
2. Explain why more than one species of *Australopithecus* coexisted.
3. Distinguish between *Australopithecus* and *Homo*.
4. Explain what genome sequencing has revealed about the ancestry of Neanderthals and us.

16.2 Molecular Evolution

5. Explain how DNA information can be used to shed light on evolution.
6. List genes that were important in our evolution.
7. Explain how chromosome banding patterns and protein sequences reveal evolutionary trends.
8. Explain what mitochondrial DNA and Y chromosome sequences reveal about human ancestry.

16.3 The Peopling of the Planet

9. Explain what mitochondrial Eve represents.
10. Describe how people expanded out of Africa and then Eurasia, populating the world.

The Big Picture: Our genes and genomes are informational molecules, and their sequences hold clues to our deep past as well as our present diversity.

The Hobbits

It's odd to be the only ones of our kind, which may be why a dual humanity theme persists in science fiction. *The Time Machine* looked at two battling breeds of people. In *Darwin's Children,* a virus scrambles the genomes of a group of newborns, starting a new species. In other stories, a Neanderthal lives in modern-day Tajikistan and a caveman in Kenya.

Fossils indicate that from 2 to 6 million years ago, humans and prehumans overlapped, in time if not place. The discovery of preserved bones of several ancient humans on the island of Flores in Indonesia in 2004 suggested a recent coexistence of two types of people. A female skeleton found 17 feet beneath a cave floor with pieces of others nearby was named *Homo floresiensis,* popularly called the Hobbit. She was about half as tall as a modern human, with a brain about a third of the size. She lived about 18,000 years ago.

The Hobbits exhibited "island dwarfism," an effect of natural selection on small, isolated, island populations. With limited resources, those who need less food are more likely to reproduce. Who were the Hobbits? At first, researchers thought that Hobbits were direct descendants of *Homo erectus,* who lived before us. Then, analysis of limb bones revealed feet and proportions like those of an ape, despite a more humanlike skull. Therefore, the Hobbits may have been direct descendants of a primate older than *Homo erectus,* who evolved in a different direction on their isolated island.

Technology Timeline

PATENTING LIFE AND GENES

1790	U.S. patent act enacted. A patented invention must be new, useful, and not obvious.
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 awarded on a genetically modified organism, a bacterium given four DNA rings that enable it to metabolize components of crude oil.
1988	First patent 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 awarded patent for all forms of transgenic cotton. Groups concerned that this will limit the rights of subsistence farmers contest the patent several times.
1996–1999	Companies patent partial gene sequences and certain

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

Bioethics: Choices for the Future boxes include Questions for Discussion.

Bioethics: Choices for the Future

Banking Stem Cells

The parents-to-be were very excited by the DVD that came in the mail shortly after they began seeing an obstetrician:

> Bank your baby's cord blood stem cells and benefit from breakthroughs. Be prepared for the unknowns in life.

The short film profiled children who were saved from certain deadly diseases because their parents had stored their umbilical cord blood. The statistics quoted were persuasive: More than 70 diseases are currently treatable with cord blood transplants, and 10,000 procedures have already been done.

With testimonials like that, it is little wonder that parents collectively spend more than $100 million per year to store cord blood. The ads and statistics are accurate but misleading, because of what they *don't* say. Most people never actually use the umbilical cord blood stem cells that they store. The scientific reasons go beyond the fact that treatable diseases are very rare. In addition, cord blood stem cells are not nearly as pluripotent as some other stem cells, limiting their applicability. Perhaps the most compelling reason that stem cell banks are rarely used is based on logic: For a person with an inherited disease, *healthy* stem cells are required—not his or her own, which could cause the disease all over again. The patient needs a donor.

Commercial cord blood banks may charge more than $1,000 for the initial collection plus an annual fee. However, the U.S. National Institutes of Health and organizations in many other nations have supported not-for-profit banks for years, and do not charge fees. Donations of cord blood to these facilities are not to help the donors directly, but to help whoever can use the cells.

As stem cell science has leaped forward, both commercial cell banks and anecdotal reports of successes have captured much media attention. This was the case for an 18-month-old boy whose cerebral palsy greatly improved after he was treated with his own cord blood cells. Whether he would have improved without the treatment isn't known.

Commercial stem cell banks are not just for newborns. One company, for example, offers to bank "very small embryonic-like stem cells" for an initial charge of $7,500 and a $750 annual fee, "enabling people to donate and store their own stem cells when they are young and healthy for their personal use in times of future medical need." The cells come from a person's blood and, in fact, one day may be very useful, but the research has yet to be done supporting use of the cells in treatments.

Questions for Discussion

1. Storing stem cells is not regulated by the U.S. government the way that a drug or a surgical procedure is because it is a service that will be helpful for treatments not yet invented. Do you think such banks should be regulated, and if so, by whom and how?
2. What information do you think that companies offering to store stem cells should present on their websites?
3. Do you think that advertisements for cord blood storage services that have quotes and anecdotal reports, but do not mention that most people who receive stem cell transplants do not in fact receive their own cells, are deceptive? Or do you think it is the responsibility of the consumer to research and discover this information?
4. How can medical consumers become aware that the government funds facilities to store stem cells?
5. It is likely that in the future, stem cell–based treatments will be possible, following large-scale clinical trials. What is the fairest way to prepare for this type of future medical treatment?

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

Review Questions assess content knowledge.

Applied Questions help students develop problem-solving skills.

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, **pseudogenes,** introns, promoters and other controls, and repeats.

www.mhhe.com/lewisgenetics10

Answers to all end-of-chapter questions can be found at **www.mhhe.com/lewisgenetics10.** 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.

Web Activities

1. 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.)

Forensics Focus

1. 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. Blowflies 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.

Case Studies and Research Results

1. 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.

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

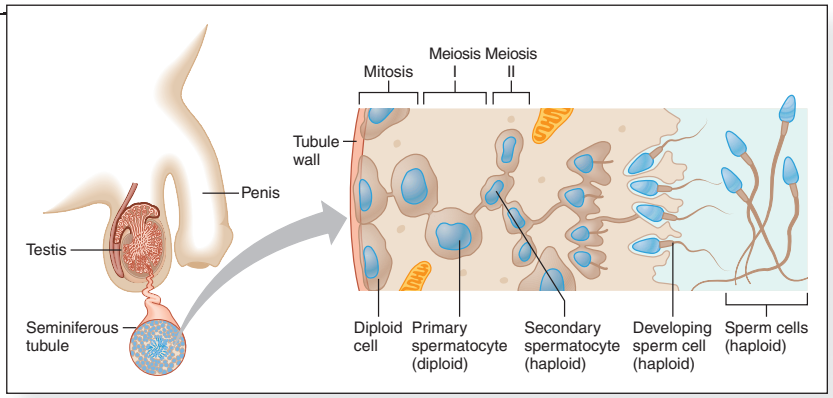
Capitalizing on students' interest in forensic science, new **Forensics Focus** questions make students think about the genetic principles involved in the collection and use of genetic information in criminal investigations.

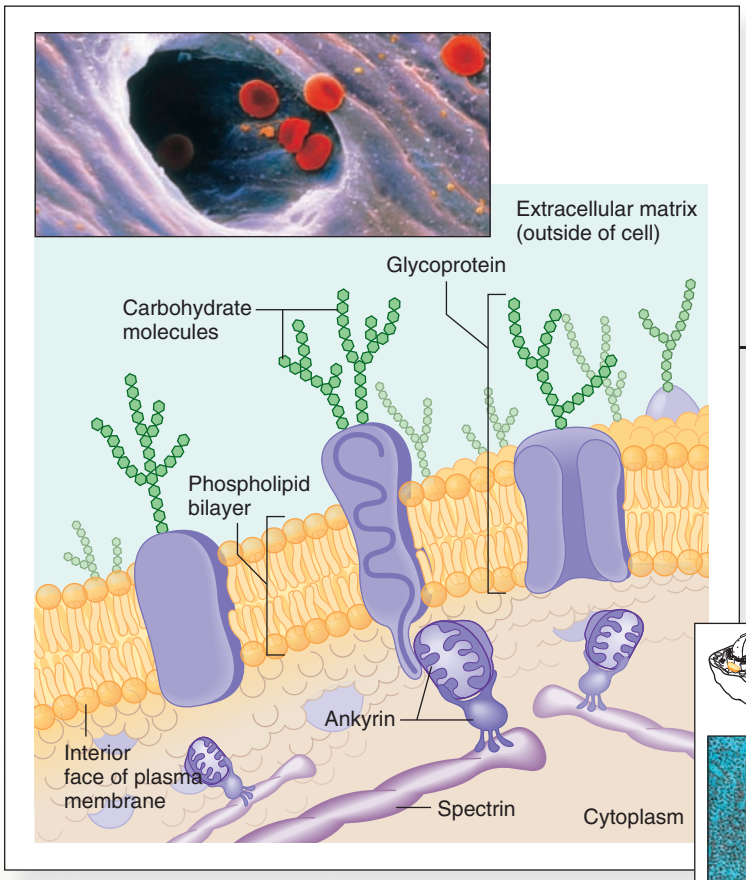
Cases and Research Results use stories based on accounts in medical and scientific journals; real clinical cases; posters and reports from professional meetings; and fiction to ask students to analyze data and predict results.

Dynamic Art Program

Multilevel Perspective

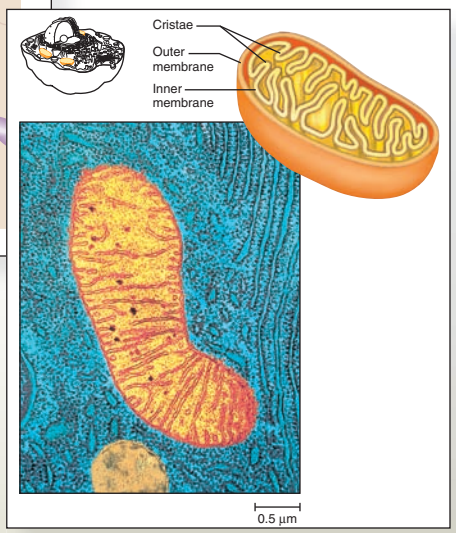
Illustrations depicting complex structures show macroscopic and microscopic views to help students see the relationship between increasingly detailed drawings.





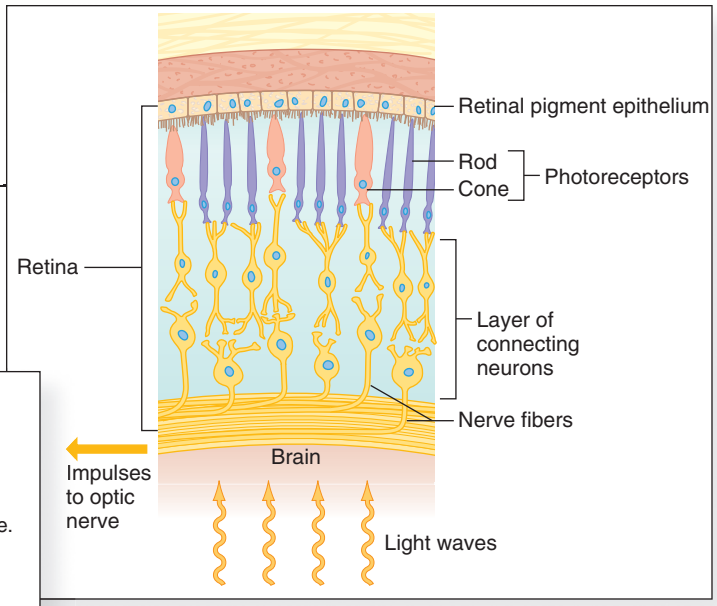
Combination Art

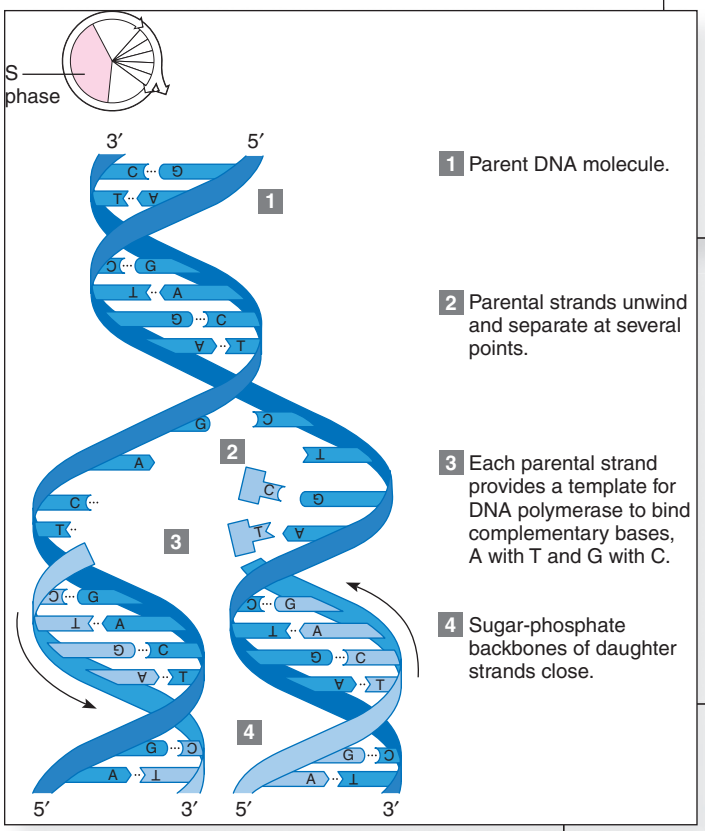
Drawings of structures are paired with micrographs to give the student the best of both perspectives: the realism of photos and the explanatory clarity of line drawings.



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.