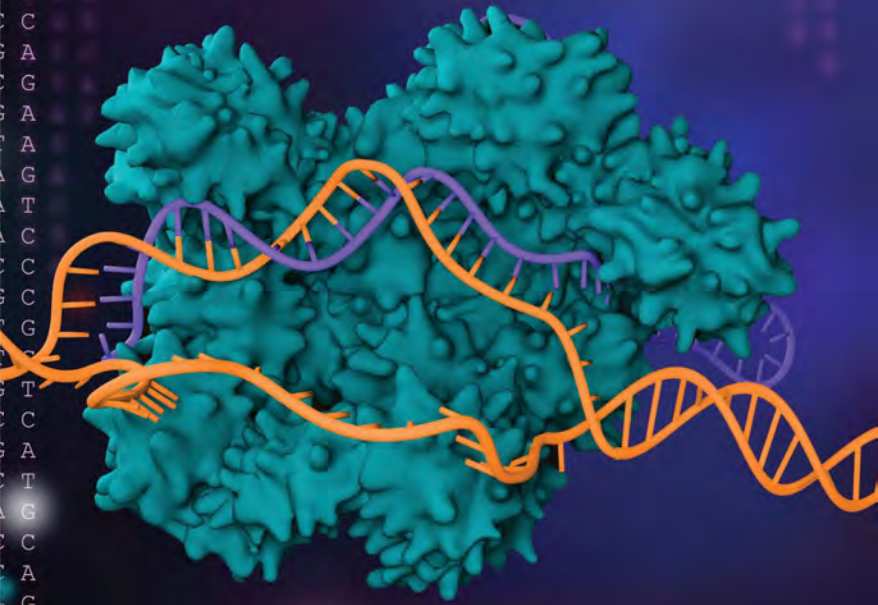
Seventh Edition

GENETICS

Analysis & Principles



Robert J. Brooker

Mc Graw Hill

GENETICS

ANALYSIS & PRINCIPLES

Seventh Edition

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GENETICS: ANALYSIS & PRINCIPLES, SEVENTH EDITION

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PREFACE

In the seventh edition of *Genetics: Analysis & Principles,* the content has been updated to reflect current trends in the field. In addition, the presentation of the content has been improved in ways that foster active learning. As an author, researcher, and teacher, I want a textbook that gets students actively involved in learning genetics. To achieve this goal, I have worked with a talented team of editors, illustrators, and media specialists who have helped me to make the seventh edition of *Genetics: Analysis & Principles* a fun learning tool.

Overall, an effective textbook needs to accomplish four goals. First, it needs to provide comprehensive, accurate, and up-to-date content in its field. Second, it needs to expose students to the techniques and skills they will need to become successful in that field. Third, an effective textbook should have pedagogical features, such as formative assessment, that foster student learning. And finally, it should inspire students so that they want to continue their studies of genetics and maybe even pursue the field as a career. The hard work that has gone into the seventh edition of *Genetics: Analysis & Principles* has been aimed at achieving all of these goals!

FLIPPING THE CLASSROOM

A trend in science education is the phenomenon that is sometimes called "flipping the classroom." This phrase refers to the idea that some of the activities that used to be done in class are now done outside of class, and vice versa. For example, instead of spending the entire class time lecturing on textbook and other materials, some of the class time is spent engaging students in various activities, such as problem solving, working through case studies, and designing experiments. This approach is called active learning. For many instructors, the classroom has become more learner centered than teacher centered. A learner-centered classroom provides a rich environment in which students can interact with each other and with their instructors. Instructors and fellow students often provide formative assessment—immediate feedback that helps each student understand if his or her learning is on the right track.

What are some advantages of active learning? Educational studies reveal that active learning usually promotes greater learning gains. In addition, active learning often focuses on skill development rather than on the memorization of facts that are easily forgotten. Students become trained to "think like scientists" and to develop a critical-thinking skills that enable them to apply scientific reasoning. A common concern among instructors who are beginning to try out active learning is that they think they will have less time to teach and therefore will cover less material. However, this may not be the case. Although students may be provided with online lectures, "flipping the classroom" typically gives students more responsibility for understanding the textbook material on their own. Along these lines, *Genetics: Analysis & Principles,* seventh edition, is intended to provide students with a resource that can be effectively used outside of the classroom. Here are several of the key pedagogical features:

- A new feature for the sixth edition called **Genetic *TIPS*** is retained in the seventh edition and provides a consistent approach to help students solve problems in genetics. In other words, it is aimed at improving their critical-thinking skills. This approach has three components. First, the student is made

GENETIC TIPS **THE QUESTION:** All of the Genetic TIPS begin with a question. As an example, let's consider the following question:

The coding strand of DNA in a segment of a gene is as follows: ATG GGC CTT AGC. This strand carries the information to make a region of a polypeptide with the amino acid sequence, methionine-glycine-leucine-serine. What would be the consequences if a mutation changed the second cytosine (C) in this sequence to an adenine (A)?

TOPIC: ***What topic in genetics does this question address?*** The topic is gene expression. More specifically, the question is about the relationship between a gene sequence and the genetic code.

INFORMATION: ***What information do you know based on the question and your understanding of the topic?*** In the question, you are given the base sequence of a short segment of a gene and told that one of the bases has been changed. From your understanding of the topic, you may remember that a polypeptide sequence is determined by reading the mRNA (transcribed from a gene) in groups of three bases called codons.

PROBLEM-SOLVING STRATEGY: ***Compare and contrast.*** One strategy to solve this problem is to compare the mRNA sequence (transcribed from this gene) before and after the mutation:

Original: AUG GGC **C**UU AGC
Mutant: AUG GGC **A**UU AGC

ANSWER: The mutation has changed the sequence of bases in the mRNA so that the third codon has changed from CUU to AUU. Because codons specify amino acids, this may change the third amino acid to something else. Note: If you look ahead to Chapter 13 (see Table 13.1), you will see that CUU specifies leucine, whereas AUU specifies isoleucine. Therefore, you would predict that the mutation would change the third amino acid from leucine to isoleucine.

aware of the ***T***opic at hand. Second, the question is evaluated with regard to the ***I***nformation that is available to the student. Finally, the student is guided through one or more ***P***roblem-Solving ***S***trategies to tackle the question.

- **Genes → Traits:** Because genetics is such a broad discipline, ranging from the molecular level to populations, many instructors have told us that it is a challenge for students to see both "the forest and the trees." It is commonly mentioned that students often have trouble connecting the concepts they have learned in molecular genetics with the traits that occur at the level of a whole organism (e.g., What does transcription have to do with blue eyes?). To try to make this connection more meaningful, certain figure legends in each chapter, designated **Genes → Traits**, remind students that molecular and cellular phenomena ultimately lead to the traits that are observed in each species (see Figure 14.8).
- **Learning Outcomes:** Each section of every chapter begins with a set of learning outcomes. These outcomes help students understand what they should be able to do once they have mastered the material in that section.
- **Formative Assessment:** When students are expected to learn textbook material on their own, it is imperative that they are regularly given formative assessment so they can gauge whether they are mastering the material. Formative assessment is a major feature of this textbook and is bolstered by Connect—a state-of-the art digital assignment and assessment platform. In *Genetics: Analysis & Principles,* seventh edition, formative assessment is provided in multiple ways.

1. As mentioned, a feature called Genetic TIPS is aimed at helping students refine their problem-solving skills.
2. Each section of every chapter ends with multiple-choice Comprehension Questions. Also, compared with previous editions, many chapters in the seventh edition are divided into more sections that are shorter in length. Formative assessment at the end of each section allows students to evaluate their mastery of the material before moving on to the next section. The answers are provided in Appendix B at the end of the book.
3. Most figures have Concept Check questions so that students can determine if they understand the key points in the figure. The answers are provided in Appendix B at the end of the book.
4. Extensive end-of chapter questions continue to provide students with feedback regarding their mastery of the material. The answers to even-numbered questions are provided in Appendix B.
5. The textbook material is supported by digital learning tools found in Connect. Instructors can assign questions and activities in Connect, and students also have access to our valuable adaptive study tool, SmartBook 2.0. With this tool, students are repeatedly given questions regarding the textbook material, and depending on their answers, they may advance in their reading, or they are given specific advice on what textbook material to go back and review.

Overall, the pedagogy of *Genetics: Analysis & Principles,* seventh edition, has been designed to foster student learning. Instead of being a collection of facts and figures, *Genetics: Analysis & Principles,* seventh edition, by Rob Brooker, is intended to be an engaging and motivating textbook in which formative assessment allows students to move ahead and learn the material in a productive way. We welcome your feedback so that we can make future editions even better!

MAJOR CONTENT CHANGES IN RECENT EDITIONS

- Coverage of the topic of epigenetics was expanded to a whole chapter in the sixth edition, which is Chapter 16. In the seventh edition, this chapter includes a new section that focuses on the structure and function of heterochromatin.
- A chapter on non-coding RNAs, Chapter 17, was added in the sixth edition. This long-overdue chapter was included in response to a remarkable explosion in our appreciation for the roles of non-coding RNAs in many aspects of molecular biology. Although two new chapters were added in recent editions, the overall page length of the seventh edition is not longer than the fifth edition.
- Discussion of the role of the CRISPR-Cas system in providing prokaryotes with a genome defense mechanism has been added in Chapter 17, and its use by researchers to edit genes is described in Chapter 21.
- Due to the expanding material in the area of medical genetics and cancer biology, the chapter on medical genetics and cancer has been split into two chapters in the seventh edition.
- The material that was covered in Chapter 20 of the sixth edition has been placed in other chapters. Homologous recombination is now in Chapter 19 and transposition is in Chapter 10.

Examples of Specific Content Changes to Individual Chapters in the Seventh Edition

- Chapter 2, Mendelian Inheritance: The discussion of the chi square test has been revised to better explain the connection to the null hypothesis.
- Chapter 4, Extensions of Mendelian Inheritance: Ever wonder why some breeds of dogs, such as collies, have white undersides with darker fur on their backs? A new subsection explains how this phenomenon is related to events during development that involve the migration and proliferation of melanocytes (see Figure 4.16).
- Chapter 6, Genetic and Linkage Mapping in Eukaryotes: The section on genetic mapping has been revised to emphasize that the pattern of allele linkage is deduced from the true-breeding P generation and that F_2 recombinants are produced by a crossover.

- Chapter 7, Genetic Transfer and Mapping in Bacteria: The section on bacterial transduction has been simplified by eliminating the discussion of cotransduction, which is not commonly used.
- Chapter 9, Molecular Structure of DNA and RNA: Based on reviewer feedback, the topic of triplex DNA was removed from this chapter.
- Chapter 10, Molecular Structure of Chromosomes and Transposable Elements: Due to their effects on chromosome structure, transposable elements (discussed in Chapter 20 in the sixth edition) are now covered in this chapter (see Section 10.5). New and revised figures regarding eukaryotic chromatin structure were added (see Figures 10.19, 10.22, and 10.23).
- Chapter 11, DNA Replication: A new model depicting the molecular structure of DNA polymerase has been added (see Figure 11.8b).
- Chapter 12, Gene Transcription and RNA Modification: New information describing how a component of the spliceosome acts as a ribozyme has been added.
- Chapter 14, Gene Regulation in Bacteria: The discussion of the *trp* operon has been simplified.
- Chapter 15, Gene Regulation in Eukaryotes I: Transcriptional and Translational Regulation: The topic of insulators has been moved to Chapter 16. They are now called "barriers" to heterochromatin formation. Discussion of the potential role of nucleosome-free regions in preventing heterochromatin spreading has been added.
- Chapter 16, Gene Regulation in Eukaryotes II: Epigenetics: A new section entitled "Heterochromatin: Formation, Structure, Formation, and Maintenance" has been added. This section has five new figures (see Figures 16.4 through 16.8). The section entitled "Role of Epigenetics in Cancer" was moved to Chapter 25.
- Chapter 17, Non-coding RNAs: New figures illustrate non-coding RNAs that act as a guide and a decoy (see Figures 17.2 and 17.3). Figure 17.10 was revised to distinguish between pRITS and pRISC, which silence the transcription and translation of transposable elements, respectively.
- Chapter 18, Genetics of Viruses: The information on the origin of HIV and the occurrence of HIV infection worldwide and in the United States has been updated.
- Chapter 19, Gene Mutation, DNA Repair, and Recombination: Figure 19.18, which concerns nucleotide excision repair, has been revised to include the function of UvrD. The section on homologous recombination (in Chapter 20 in the sixth edition) was moved to this chapter.
- Chapter 20, Molecular Technologies: This chapter was Chapter 21 in the sixth edition. The topic of real-time PCR has been divided into smaller subsections for clarity and ease of reading. Section 20.4, formerly called "Gene Mutagenesis" is now called "Gene Editing."
- Chapter 21, Biotechnology: This chapter was Chapter 22 in the sixth edition. The section entitled "Human Gene Therapy" was moved to the chapter on medical genetics (now Chapter 24).
- Chapter 22, Genomics I: Analysis of DNA: This chapter was Chapter 23 in the sixth edition. The section on physical mapping has been simplified.
- Chapter 23, Genomics II: Functional Genomics, Proteomics, and Bioinformatics: This chapter was Chapter 24 in the sixth edition.
- Chapter 24, Medical Genetics: This chapter was Chapter 25 in the sixth edition. The topic of genome-wide association (GWA) studies was added (see Figure 24.8). The section "Human Gene Therapy" was moved to this chapter. The material on cancer was moved to Chapter 25, which solely focuses on cancer.
- Chapter 25, Genetic Basis of Cancer: This topic is now featured in its own chapter, which includes four separate sections. The last section focuses on the role of epigenetics in cancer.
- Chapter 26, Developmental Genetics: The information on *Hox* genes in development and on the role of the *SRY* gene in human sex determination has been updated.
- Chapter 27, Population Genetics: Discussion of the topic of inbreeding has been expanded.
- Chapter 28, Complex and Quantitative Traits: The identification of QTLs is now covered in a separate section.
- Chapter 29, Evolutionary Genetics: The cladistics method for constructing a phylogenetic tree is compared with the UPGMA method.

Suggestions Welcome!

It seems very appropriate to use the word *evolution* to describe the continued development of this textbook. I welcome any and all comments. The refinement of any science textbook requires input from instructors and their students. These include comments regarding writing, illustrations, supplements, factual content, and topics that may need greater or less emphasis. You are invited to contact me at:

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ACKNOWLEDGMENTS

The production of a textbook is truly a collaborative effort, and I am deeply indebted to many people. All seven editions of this textbook went through multiple rounds of rigorous revision that involved the input of faculty, students, editors, and educational and media specialists. Their collective contributions are reflected in the final outcome.

Deborah Brooker (Freelance Developmental Editor) meticulously read the new material, analyzed every figure, and offered extensive feedback. Her attention to detail in this edition and previous editions has profoundly contributed to the accuracy and clarity of this textbook. I would also like to thank Jane Hoover (Freelance Copy Editor) for understanding the material and working extremely hard to improve the text's clarity. Her efforts are truly appreciated. She is at the top of the list for copy editors.

I would also like to acknowledge the many people at McGraw-Hill Education whose skills and insights are amazing. My highest praise goes to Elizabeth Sievers (Senior Product Developer), who carefully checks all aspects of textbook development and makes sure that all of the pieces of the puzzle are in place. I am also grateful to Andrew Urban (Portfolio Manager) for overseeing this project. I would like to thank other people at McGraw-Hill who have played key roles in producing the actual book and the supplements that go along with it. In particular, Jessica Portz (Senior Content Project Manager) has done a superb job of managing the components that need to be assembled to produce a book. I would also like to thank Lori Hancock (Content Licensing Specialist), who acted as an interface between me and the photo company. In addition, my gratitude goes to David Hash (Designer), who provided much input into the internal design of the book and created an awesome cover. Finally, I would like to thank Kelly Brown (Executive Marketing Manager), whose major efforts begin when the seventh edition comes out!

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Finally, I want to thank the many scientists who reviewed the chapters of this textbook. Their broad insights and constructive suggestions were an overriding factor that shaped its final content and organization. I am truly grateful for their time and compassion.

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CHAPTER OUTLINE

1.1 The Molecular Expression of Genes

1.2 The Relationship Between Genes and Traits

1.3 Fields of Genetics

1.4 The Science of Genetics

CC (for "carbon copy" or "copy cat"), the first cloned pet. *In 2002, the cat shown here was produced by reproductive cloning, a procedure described in Chapter 21.*

1 OVERVIEW OF GENETICS

Hardly a week goes by without a major news story announcing a genetic breakthrough. The increasing pace of genetic discoveries has become staggering. The Human Genome Project is a case in point. This project began in the United States in 1990, when the National Institutes of Health and the Department of Energy joined forces with international partners to decipher the massive amount of information contained in our **genome**—the DNA found within all of our chromosomes (**Figure 1.1**). Remarkably, in only a decade, the researchers working on this project determined the DNA sequence (the order of the bases A, T, G, and C) of over 90% of the human genome. The completed sequence, published in 2003, has an accuracy greater than 99.99%; less than 1 mistake was made in every 10,000 base pairs!

In 2008, a more massive undertaking, called the 1000 Genomes Project, was launched to establish a detailed understanding of human genetic variation. In this international project, researchers set out to determine the DNA sequence of at least 1000 anonymous participants from around the globe. In 2015, the sequencing of over 2500 genomes was described in the journal *Nature*.

Studying the human genome allows us to explore fundamental details about ourselves at the molecular level. The results of human genome projects have shed considerable light on basic questions, like how many genes we have, how genes direct the activities of living cells, how species evolve, how single cells develop into complex tissues, and how defective genes cause disease. Furthermore, understanding our genome may lend itself to improvements in modern medicine by leading to better diagnoses of diseases and allowing the development of new treatments for them.

The quest to unravel the mysteries within our genes has involved the invention of many new technologies. For example, researchers have developed genetic techniques to produce medicines, such as human insulin, that would otherwise be difficult or impossible to make. Human insulin is synthesized in strains of *Escherichia coli* bacteria that have been genetically altered by the addition of genes that encode the polypeptides that form this hormone. Grown in laboratories, these bacteria make large amounts of human insulin. As discussed in Chapter 21, the insulin is purified and administered to many people with insulin-dependent diabetes.

DNA, the molecule of life

The adult human body is composed of trillions of cells.

Most human cells contain the following:

- 46 human chromosomes, found in 23 pairs
- 2 meters of DNA
- Approximately 22,000 genes coding for proteins that perform most life functions
- Approximately 3 billion DNA base pairs per set of chromosomes, containing the bases A, T, G, and C

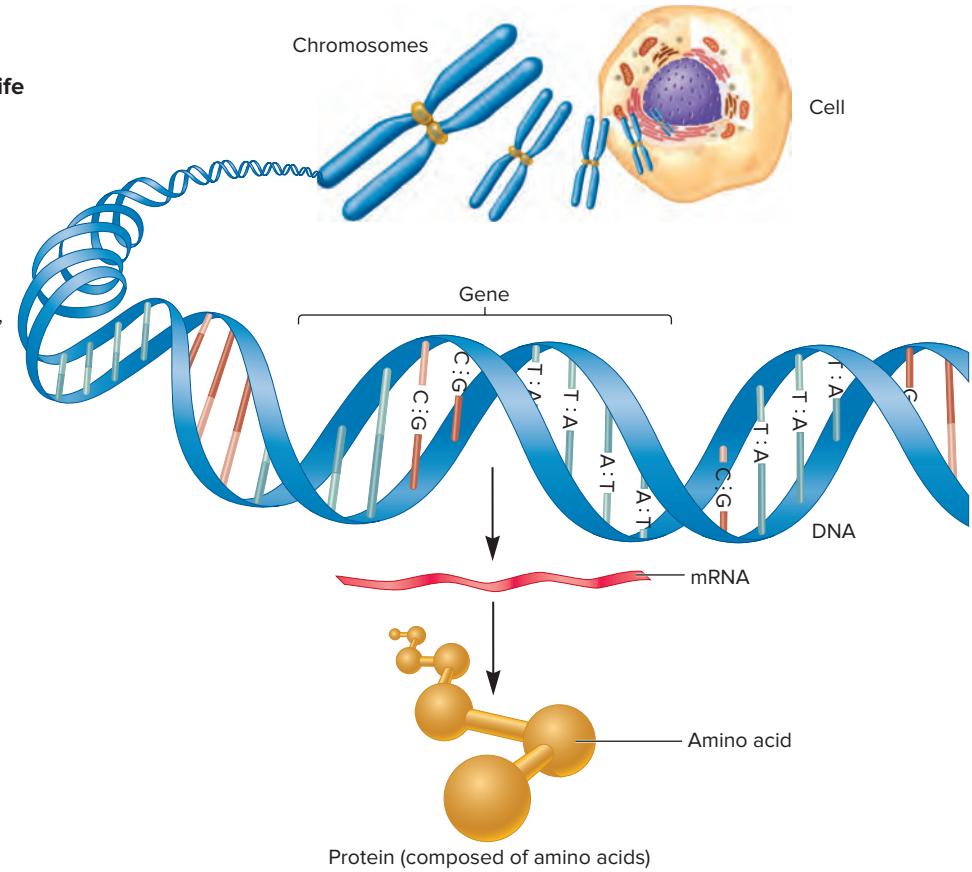


FIGURE 1.1 The human genome. The human genome is a complete set of human chromosomes. People have two sets of chromosomes—one set from each parent—which are found in the cell nucleus. The Human Genome Project revealed that each set of chromosomes is composed of a DNA sequence that is approximately 3 billion base pairs long. As discussed later, most genes are first transcribed into mRNA and then the mRNA is used to make proteins. Estimates suggest that each set of chromosomes contains about 22,000 different protein-encoding genes. This figure emphasizes the DNA found in the cell nucleus. Humans also have a small amount of DNA in their mitochondria, which has also been sequenced.

CONCEPT CHECK: How might a better understanding of our genes be used in the field of medicine?

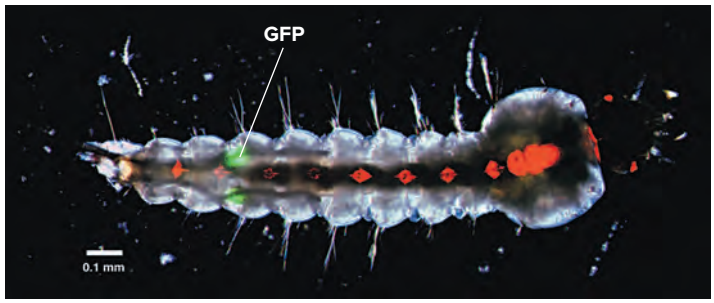
New genetic technologies are often met with skepticism and sometimes even with disdain. An example is mammalian cloning. In 1997, Ian Wilmut and his colleagues created clones of sheep, using mammary cells from an adult animal (**Figure 1.2**). More recently, such cloning has been achieved in several mammalian species, including cows, mice, goats, pigs, and cats. In 2002, the first pet was cloned, a cat named CC (for "carbon copy" or "copy cat"; see the chapter-opening photo). The cloning of mammals provides the potential for many practical applications. With regard to livestock, cloning would enable farmers to use cells from their best individuals to create genetically homogeneous herds. This could be advantageous in terms of agricultural yield, although such a genetically homogeneous herd may be more susceptible to certain diseases. However, people have become greatly concerned about the possibility of human cloning. This prospect has raised serious ethical questions. Within the past few years, legislation that involves bans on human cloning has been introduced.

Finally, genetic technologies provide the means to modify the traits of animals and plants in ways that would have been unimaginable just a few decades ago. **Figure 1.3a** shows a striking example in which scientists introduced a gene from jellyfish into mice. Certain species of jellyfish emit a "green glow"

FIGURE 1.2 The cloning of a mammal. The lamb in the front is Dolly, the first mammal to be cloned. She was cloned from the cells of a Finn Dorset (a white-faced sheep). The sheep in the back is Dolly's surrogate mother, a Blackface ewe. A description of how Dolly was produced is presented in Chapter 21.

CONCEPT CHECK: What ethical issues may be associated with human cloning?

(a) GFP expressed in mice



(b) GFP expressed in the gonads of a male mosquito

FIGURE 1.3 The introduction of a jellyfish gene into laboratory mice and mosquitoes. **(a)** A gene that naturally occurs in jellyfish encodes a protein called green fluorescent protein (GFP). The *GFP* gene was cloned and introduced into mice. When these mice are exposed to UV light, GFP emits a bright green color. These mice glow green, just like the jellyfish! **(b)** The *GFP* gene was introduced next to a gene sequence that causes the expression of GFP only in the gonads of male mosquitoes. The resulting green glow allows researchers to identify and sort males from females.

(a): ©Eye of Science/Science Source; (b): Photo taken by Flaminia Catteruccia, Jason Benton and Andrea Crisanti, and assembled by www.luciariccidesign.com

CONCEPT CHECK: Why is it useful to sort male mosquitoes from females?

produced by a bioluminescent protein called green fluorescent protein (GFP) encoded by a gene in the jellyfish genome. When exposed to blue or ultraviolet (UV) light, the protein emits a striking green-colored light. Scientists were able to clone the *GFP* gene from a sample of jellyfish cells and then introduce this gene into laboratory mice. The green fluorescent protein is made throughout the cells of their bodies. As a result, their skin, eyes, and organs give off an eerie green glow when exposed to UV light. Only their fur does not glow.

The expression of green fluorescent protein allows researchers to identify particular proteins in cells or specific body parts. For example, Andrea Crisanti and colleagues have altered mosquitoes to express GFP only in the gonads of males (**Figure 1.3b**). This enables the researchers to distinguish males from females and sort mosquitoes by sex. Why is this useful? Researchers can produce a population of mosquitoes and then sterilize the males. The ability to distinguish males from females makes it possible to release the sterile males without the risk of releasing additional females. The release of sterile males may be an effective means of controlling mosquito populations because females mate only once before they die. Mating with a sterile male prevents a female from producing offspring. In 2008, Osamu Shimomura, Martin Chalfie, and Roger Tsien received the Nobel Prize in chemistry for the discovery and development of GFP, which has become a widely used tool in biology.

Overall, as we move forward in the twenty-first century, the excitement level in the field of genetics is high, perhaps higher than it has ever been. Nevertheless, new genetic knowledge and technologies will create many ethical and societal challenges. In this chapter, we begin with an overview of genetics and then explore the various fields of genetics and their experimental approaches.

1.1 THE MOLECULAR EXPRESSION OF GENES

Learning Outcomes:

1. Describe the biochemical composition of cells.
2. Explain how proteins are largely responsible for cell structure and function.
3. Outline how DNA stores the information to make proteins.

Genetics is the branch of biology that deals with heredity and variation. It stands as the unifying discipline in biology by allowing us to understand how life can exist at all levels of complexity, ranging from the molecular to the population level. Genetic variation is the root of the natural diversity that we observe among members of the same species and among different species.

Genetics is centered on the study of genes. A gene is classically defined as a unit of heredity. At the molecular level, a **gene** is a segment of DNA that contains the information to produce a functional product. The functional product of most genes is a polypeptide, which is a linear sequence of amino acids that folds into one of the units that constitute proteins. In addition, genes are commonly described according to the way they affect **traits,** which are the characteristics of an organism. In humans, for example, we observe traits such as eye color, hair texture, and height. The ongoing theme of this textbook is the relationship between genes and traits. As an organism grows and develops, its collection of genes provides a blueprint that determines its traits.

In this section, we examine the general features of life, beginning with the molecular level and ending with populations of organisms. As will become apparent, genetics is the

common thread that explains the existence of life and its continuity from generation to generation. For most students, this chapter should serve as an overview of topics they have learned in other introductory courses such as general biology. Even so, it is usually helpful to see the "big picture" of genetics before delving into the finer details that are covered in Chapters 2 through 29.

Living Cells Are Composed of Biochemicals

To fully understand the relationship between genes and traits, we need to begin with an examination of the composition of living organisms. Every cell is constructed from intricately organized chemical substances. Small organic molecules such as glucose and amino acids are produced by the linkage of atoms via chemical bonds. The chemical properties of organic molecules are essential for cell vitality in two key ways. First, the breaking of chemical bonds during the degradation of small molecules provides energy to drive cellular processes. A second important function of these small organic molecules is their role as the building blocks for the synthesis of larger molecules. Four important categories of larger molecules are **nucleic acids** (i.e., DNA and RNA), **proteins, carbohydrates,** and **lipids.** Three of these—nucleic acids, proteins, and carbohydrates—form **macromolecules** that are composed of many repeating units of smaller building blocks. RNA, proteins, and some carbohydrates are made from hundreds or even thousands of repeating building blocks. DNA is the largest macromolecule found in living cells. A single DNA molecule can be composed of a linear sequence of hundreds of millions of building blocks called nucleotides!

The formation of cellular structures relies on the interactions of molecules and macromolecules. For example, nucleotides are connected together to make DNA, which is a constituent of chromosomes (**Figure 1.4**). In addition, DNA is associated with many proteins that provide organization to the structure of chromosomes. Within a eukaryotic cell, the chromosomes are contained in a compartment called the cell nucleus. The nucleus is bounded by a double membrane composed of lipids and proteins that shields the chromosomes from the rest of the cell. The organization of chromosomes within a cell nucleus protects the chromosomes from mechanical damage and provides a single compartment for genetic activities such as gene transcription. As a general theme, the formation of large cellular structures arises from interactions among different molecules and macromolecules. These cellular structures, in turn, are organized to make a complete living cell.

Each Cell Contains Many Different Proteins That Determine Cell Structure and Function

To a great extent, the characteristics of a cell depend on the types of proteins that it makes. The entire collection of proteins that a cell makes at a given time is called its **proteome.** The range of functions among different types of proteins is truly remarkable. Some proteins help determine the shape and structure of a given cell.

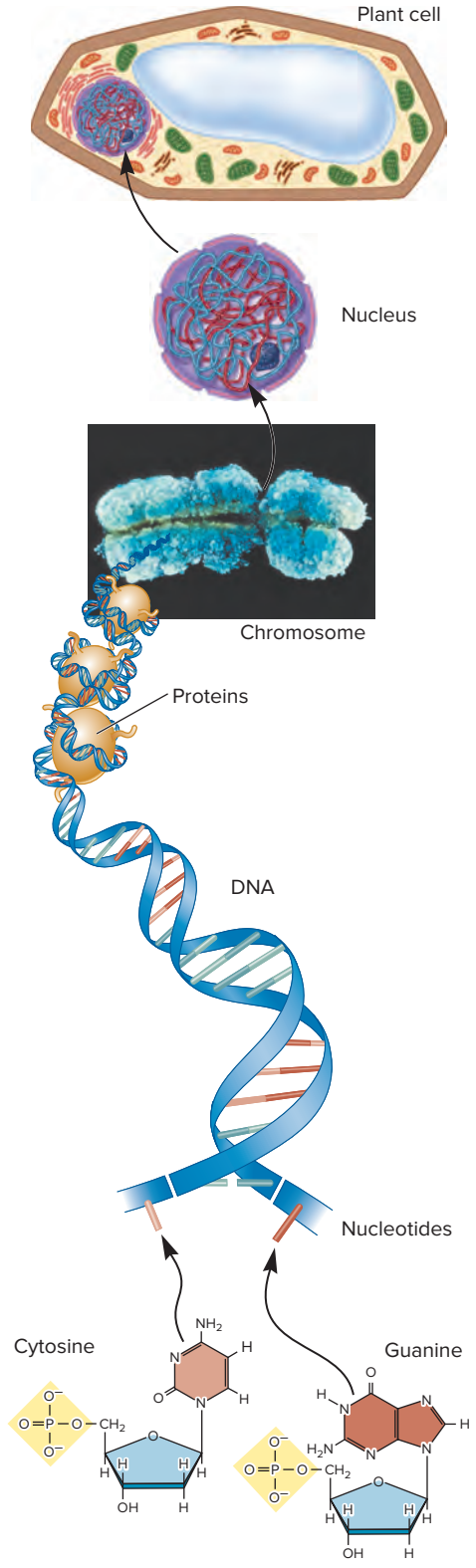


FIGURE 1.4 Molecular organization of a living cell. Cellular structures are constructed from smaller building blocks. In this example, DNA is formed from the linkage of nucleotides, producing a very long macromolecule. The DNA associates with proteins to form a chromosome. The chromosomes are located within a membrane-bound organelle called the nucleus, which, along with many other different types of organelles, is found within a complete cell.

(inset) ©Biophoto Associates/Science Source

CONCEPT CHECK: Is DNA a small molecule, a macromolecule, or an organelle?

For example, the protein known as tubulin assembles into large structures known as microtubules, which provide the cell with internal structure and organization. Other proteins are inserted into cell membranes and aid in the transport of ions and small molecules across the membrane. **Enzymes,** which accelerate chemical reactions, are a particularly important category of proteins. Some enzymes play a role in the breakdown of molecules or macromolecules into smaller units. Known as catabolic enzymes, these are important in the utilization of energy. Alternatively, anabolic enzymes and accessory proteins function in the synthesis of molecules and macromolecules throughout the cell. The construction of a cell greatly depends on its proteins that are involved in anabolism because these are required to synthesize all cellular macromolecules.

Molecular biologists have come to realize that the functions of proteins underlie the cellular characteristics of every organism. At the molecular level, proteins can be viewed as the active participants in the enterprise of life.

DNA Stores the Information for Protein Synthesis

The genetic material of living organisms is composed of a substance called **deoxyribonucleic acid,** abbreviated **DNA.** The DNA stores the information needed for the synthesis of all cellular proteins. In other words, the main function of the genetic blueprint is to code for the production of proteins in the correct cell, at the proper time, and in suitable amounts. This is an extremely complicated task because living cells make thousands of different proteins. Genetic analyses have shown that a typical bacterium can make a few thousand different proteins, and estimates for the numbers produced by complex eukaryotic species range in the tens of thousands.

DNA's ability to store information is based on its structure. DNA is composed of a linear sequence of **nucleotides.** Each nucleotide contains one of four nitrogen-containing bases: adenine (A), thymine (T), guanine (G), or cytosine (C). The linear order of these bases along a DNA molecule contains information similar to the way that groups of letters of the alphabet represent words. For example, the "meaning" of the sequence of bases ATGGGCCTTAGC differs from that of the sequence TTTAAGCTTGCC. DNA sequences within most genes contain the information to direct the order of amino acids within **polypeptides** according to the **genetic code.** In the code, a three-base sequence, called a **codon,** specifies one particular **amino acid** among the 20 possible choices. One or more polypeptides form a functional protein. In this way, the DNA can store the information to specify the proteins made by an organism.

DNA Sequence	Amino Acid Sequence
ATG GGC CTT AGC	Methionine Glycine Leucine Serine
TTT AAG CTT GCC	Phenylalanine Lysine Leucine Alanine

In living cells, the DNA is found within large structures known as **chromosomes.** **Figure 1.5** is a micrograph of the 46 chromosomes contained in a cell from a human male; this type of image is known as a **karyotype.** The DNA of an average human chromosome is an extraordinarily long, linear, double-stranded structure that contains well over a hundred million nucleotides. Along the immense length of a chromosome, the genetic information is parceled into functional units known as genes. An average-sized human chromosome is expected to contain about 1000 different protein-encoding genes.

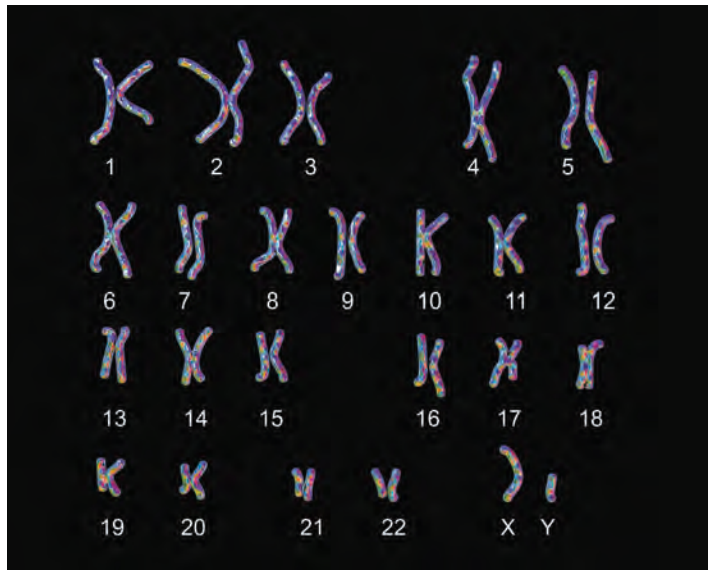
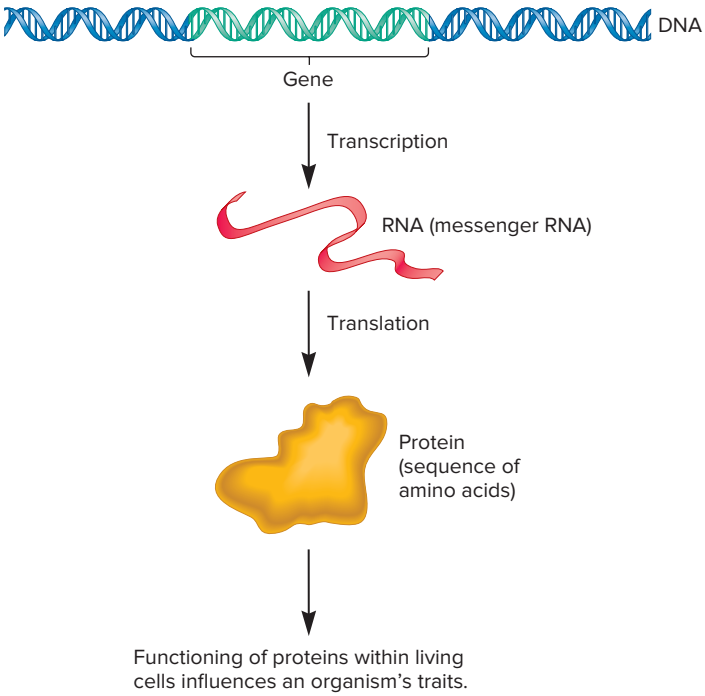


FIGURE 1.5 A micrograph of the 46 chromosomes found in a cell from a human male.

CONCEPT CHECK: Which types of macromolecules are found in chromosomes?

The Information in DNA Is Accessed During the Process of Gene Expression

To synthesize its proteins, a cell must be able to access the information that is stored within its DNA. The process of using a gene sequence to affect the characteristics of cells and organisms is referred to as **gene expression.** At the molecular level, the information within genes is accessed in a stepwise process (**Figure 1.6**). In the first step, known as **transcription,** the DNA sequence within a gene is copied into a nucleotide sequence of **ribonucleic acid (RNA). Protein-encoding genes** (also called **structural genes**) carry the information for the amino acid sequence of a polypeptide. When a protein-encoding gene is transcribed, the first product is an RNA molecule known as **messenger RNA (mRNA).** During polypeptide synthesis—a process called **translation**—the sequence of nucleotides within the mRNA determines the sequence of amino acids in a polypeptide. One or more polypeptides then fold and assemble into a functional protein. The synthesis of functional proteins largely determines an organism's traits. As discussed further in Chapter 12 (look ahead to Figure 12.1), the pathway of gene expression from DNA to RNA to protein is called the **central dogma of genetics** (also called the central dogma of molecular biology). It forms a cornerstone of our understanding of genetics at the molecular level.



ONLINE ANIMATION

FIGURE 1.6 Gene expression at the molecular level. The expression of a gene is a multistep process. During transcription, one of the DNA strands is used as a template to make an RNA strand. During translation, the RNA strand is used to specify the sequence of amino acids within a polypeptide. One or more polypeptides form a protein that functions within the cell, thereby influencing an organism's traits.

CONCEPT CHECK: Where is the information to make a polypeptide stored?

1.1 COMPREHENSION QUESTIONS

1. Which of the following is *not* a constituent of a cell's proteome?
   a. An enzyme
   b. A cytoskeletal protein
   c. A transport protein in the plasma membrane
   d. An mRNA
2. A gene is a segment of DNA that contains the information to produce a functional product. The functional product of most genes is
   a. DNA.
   b. mRNA.
   c. a polypeptide.
   d. none of the above.
3. The function of the genetic code is to
   a. promote transcription.
   b. specify the amino acids within a polypeptide.
   c. alter the sequence of DNA.
   d. do none of the above.
4. The direct result of the process of transcription is the synthesis of
   a. DNA.
   b. RNA.
   c. a polypeptide.
   d. all of the above.

1.2 THE RELATIONSHIP BETWEEN GENES AND TRAITS

Learning Outcomes:

1. Outline how the expression of genes leads to an organism's traits.
2. Define *genetic variation.*
3. Discuss the relationship between genes and traits.
4. Describe how genes are transmitted in sexually reproducing species.
5. Explain the process of evolution.

A trait is any characteristic that an organism displays. In genetics, we often focus our attention on **morphological traits,** those that affect the appearance, form, and structure of an organism. The color of a flower and the height of a pea plant are morphological traits. Geneticists frequently study these types of traits because they are easy to evaluate. For example, an experimenter can simply look at a plant and tell if it has red or white flowers. However, not all traits are morphological. **Physiological traits** affect the ability of an organism to function. For example, the rate at which a bacterium metabolizes a sugar such as lactose is a physiological trait. Like morphological traits, physiological traits are controlled, in part, by the expression of genes. **Behavioral traits** affect the ways an organism responds to its environment. An example is the mating calls of bird species. In animals, the nervous system plays a key role in governing such traits. In this section, we will examine the relationship between the expression of genes and an organism's traits.

The Molecular Expression of Genes Leads to an Organism's Traits

A complicated, yet very exciting, aspect of genetics is that the field's observations and theories span four levels of biological organization: molecules, cells, organisms, and populations. This broad scope can make it difficult to appreciate the relationship between genes and traits. To understand this connection, we need to relate the following phenomena:

1. Genes are expressed at the **molecular level.** In other words, gene transcription and translation lead to the production of a particular protein, which is a molecular process.
2. Proteins often function at the **cellular level.** The function of a protein within a cell affects the structure and workings of that cell.
3. An organism's traits are determined by the characteristics of its cells. We do not have microscopic vision, yet when we view morphological traits, we are really observing the

properties of an individual's cells. For example, a red flower has its color because its cells make a red pigment. The trait of red flower color is an observation at the **organism level.** Yet the trait is rooted in the molecular characteristics of the organism's cells.

4. A **species** is a group of organisms that maintains a distinctive set of attributes in nature. The occurrence of a trait within a species is an observation at the **population level.** Along with learning how a trait occurs, we also want to understand why a trait becomes prevalent in a particular species. In many cases, researchers discover that a trait predominates within a population because it promotes the reproductive success of the members of the population. This leads to the evolution of beneficial traits.

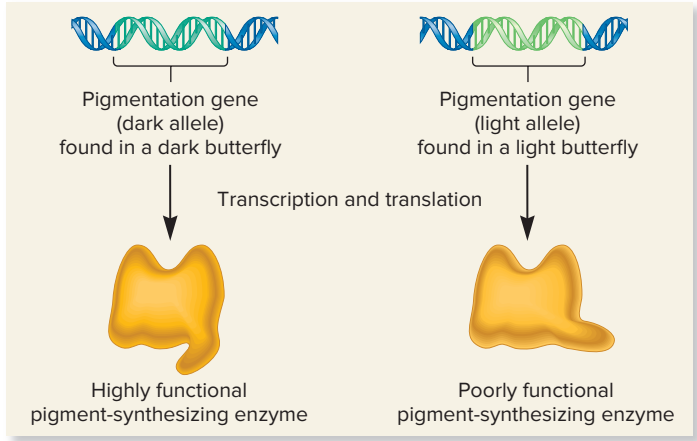
To illustrate the four levels of genetics with an example, **Figure 1.7** considers the trait of pigmentation in a species of butterflies. Some members of this species are dark colored and others are very light. Let's consider how we can explain this trait at the molecular, cellular, organism, and population levels.

At the molecular level, we need to understand the nature of the gene or genes that govern this trait. As shown in Figure 1.7a, a gene, which we will call the pigmentation gene, is responsible for the amount of pigment produced. The pigmentation gene exists in two different versions. Alternative versions of a specific gene are called **alleles.** In this example, one allele confers a dark pigmentation and the other causes a light pigmentation. Each of these alleles encodes a protein that functions as a pigment-synthesizing enzyme. However, the DNA sequences of the two alleles differ slightly from each other. This difference in the DNA sequences leads to a variation in the structure and function of the respective pigment-synthesizing enzymes.

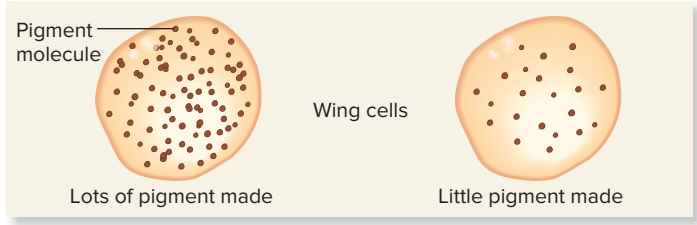
At the cellular level (Figure 1.7b), the functional differences between the two pigment-synthesizing enzymes affect the amount of pigment produced. The allele causing dark pigmentation, which is shown on the left, encodes an enzyme that functions very well. Therefore, when this gene is expressed in the cells of the wings, a large amount of pigment is made. By comparison, the allele causing light pigmentation encodes an enzyme that functions poorly. Therefore, when this allele is the only pigmentation gene expressed, little pigment is made.

At the organism level (Figure 1.7c), the amount of pigment in the wing cells governs the color of the wings. If the pigment-synthesizing enzymes produce high amounts of pigment, the wings are dark colored. If the enzymes produce little pigment, the wings are light.

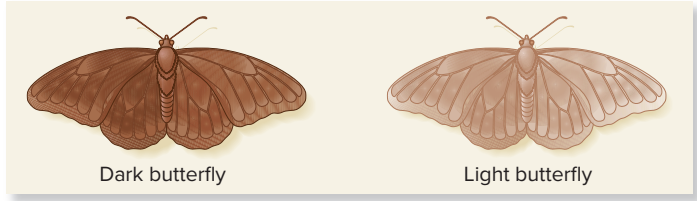
Finally, at the population level (Figure 1.7d), geneticists would like to know why a species of butterfly would contain some members with dark wings and other members with light wings. One possible explanation is differential predation. The butterflies with dark wings might better avoid being eaten by birds if they happened to live within a dimly lit forest. The dark wings would help to camouflage the butterfly if it were perched on a dark surface such as a tree trunk. In contrast, light-colored wings would be an advantage if the butterfly inhabited a brightly lit meadow. Under these conditions, a bird might be less likely to notice a light-colored butterfly that was perched on a sunlit surface. A population



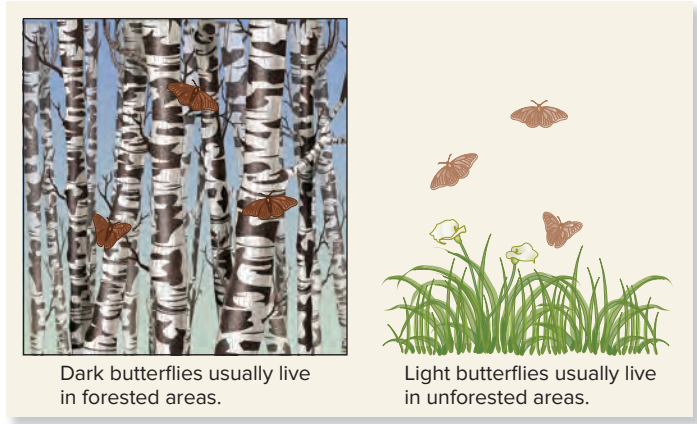
(a) Molecular level



(b) Cellular level



(c) Organism level



(d) Population level

FIGURE 1.7 The relationship between genes and traits at the (a) molecular, (b) cellular, (c) organism, and (d) population levels.

CONCEPT CHECK: Which butterfly has a more active pigment-synthesizing enzyme, the dark- or light-colored one?

geneticist might study this species of butterfly and find that the dark-colored members usually live in forested areas and the light-colored members reside in unforested areas.

Inherited Differences in Traits Are Due to Genetic Variation

In Figure 1.7, we considered how gene expression leads to variation in a trait of organisms, using the example of dark- versus light-colored wings in a species of butterflies. Variation in traits among members of the same species is very common. For example, some people have brown hair and others have blond hair; some petunias have white flowers and others have purple flowers. These are examples of **genetic variation.** This term refers to the differences in inherited traits among individuals within a population.

In large populations that occupy a wide geographic range, genetic variation can be quite striking. Morphological differences have often led geneticists to misidentify two members of the same species as belonging to separate species. As an example, **Figure 1.8** shows two dyeing poison frogs that are members of the same species, *Dendrobates tinctorius.* They display dramatic differences in their markings. Such contrasting forms within a single species are termed **morphs.** You can easily imagine how someone might mistakenly conclude that these frogs are not members of the same species.

Changes in the nucleotide sequence of DNA underlie the genetic variation that we see among individuals. Throughout this textbook, we will routinely examine how variation in the genetic material results in changes in an organism's traits. At the molecular level, genetic variation can be attributed to different types of modifications.

1. Small or large differences can occur within gene sequences. When such changes initially occur, they are called **gene mutations.** Mutations result in genetic variation in which a gene is found in two or more alleles, as previously described in Figure 1.7. In many cases, gene mutations alter the expression or function of a protein that a gene encodes.
2. Major alterations can also occur in the structure of a chromosome. A large segment of a chromosome can be lost, rearranged, or reattached to another chromosome.
3. Variation may also occur in the total number of chromosomes. In some cases, an organism may inherit one too many or one too few chromosomes. In other cases, it may inherit an extra set of chromosomes.

Variations of sequences within genes are a common source of genetic variation among members of the same species. In humans, familiar examples of sequence variation involve genes for eye color, hair texture, and skin pigmentation. Chromosome variation—a change in chromosome structure or number (or both)—is also found, but this type of change is often detrimental. Many human genetic disorders are the result of chromosomal alterations. The most common example is Down syndrome, which is due to the presence of an extra chromosome (**Figure 1.9a**). By comparison, chromosome variation in plants is common and often results in plants with superior characteristics, such as increased resistance to disease. Plant breeders have frequently exploited this observation. Cultivated varieties of wheat, for example, have many more chromosomes than the wild species does (**Figure 1.9b**).

FIGURE 1.8 Two dyeing poison frogs (*Dendrobates tinctorius*) are examples of different morphs within a single species.
(Left): ©Natalia Kuzmina/Shutterstock; (Right): ©Valt Ahyppo/Shutterstock

CONCEPT CHECK: Why do these two frogs look so different?

Traits Are Governed by Genes and by the Environment

In our discussion thus far, we have considered the role that genes play in determining an organism's traits. Another critical factor is the **environment**—the surroundings in which an organism exists. A variety of factors in an organism's environment profoundly affects its morphological and physiological features. For example, a person's diet greatly influences many traits, such as height, weight, and even intelligence. Likewise, the amount of sunlight a plant receives affects its growth rate and the color of its flowers.

An interesting example of the interplay between genes and the environment involves the human genetic disease **phenylketonuria (PKU).** Humans have a gene that encodes an enzyme known as phenylalanine hydroxylase. Most people have two functional copies of this gene. People with one or two functional copies of the gene can eat foods containing the amino acid phenylalanine and metabolize it properly.

A rare variation in the gene that encodes phenylalanine hydroxylase results in a nonfunctional version of this enzyme. Individuals

(a) (b)

FIGURE 1.9 Examples of chromosome variation. (a) A person with Down syndrome. She has 47 chromosomes rather than the common number of 46, because she has an extra copy of chromosome 21. **(b)** A wheat plant. Cultivated wheat has six sets of chromosomes.
(a): ©Stockbyte/Alamy; (b): ©Pixtal/age fotostock

CONCEPT CHECK: Do these examples constitute variation in chromosome structure or variation in chromosome number?

with two copies of this rare, inactive allele cannot metabolize phenylalanine properly. When given a standard diet containing phenylalanine, individuals with this disorder are unable to break down this amino acid. Phenylalanine accumulates and is converted into phenylketones, which are detected in the urine. Individuals with PKU can manifest a variety of detrimental traits, including mental impairment, underdeveloped teeth, and foul-smelling urine. Fortunately, through routine newborn screening in the United States, PKU is now diagnosed early. Part of the treatment is a diet that restricts phenylalanine, which is present in high-protein foods such as eggs, meat, and dairy products. Restricting phenylalanine allows the affected child to develop normally. PKU provides a dramatic example of how the environment and an individual's genes can interact to influence the traits of the organism.

During Reproduction, Genes Are Passed from Parent to Offspring

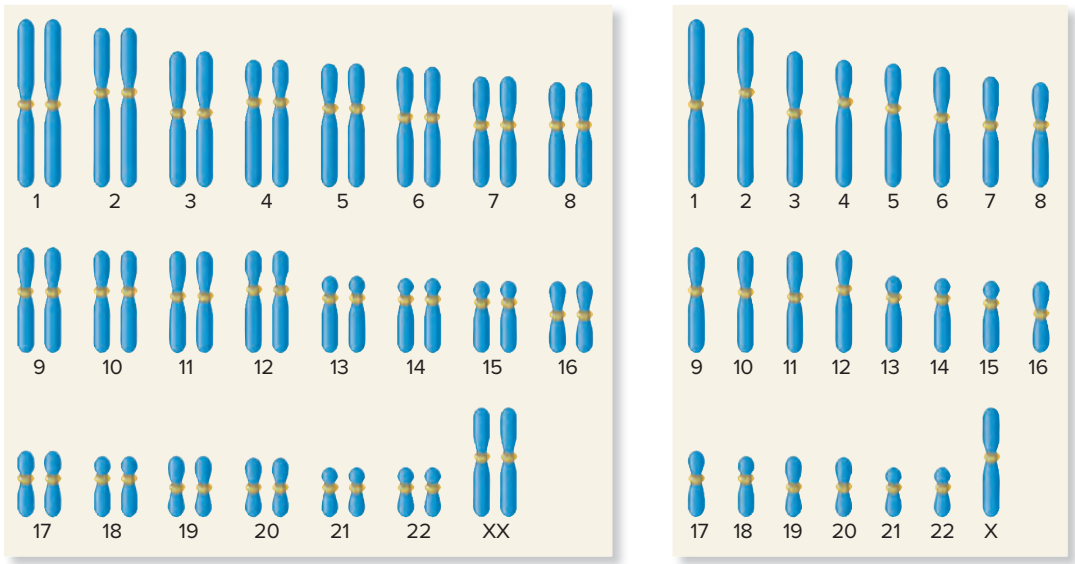
Now that we have considered how genes and the environment govern the outcome of traits, we can turn to the issue of inheritance. How are traits passed from parents to offspring? The foundation for our understanding of inheritance came from Gregor Mendel's study of pea plants in the nineteenth century. His work revealed that the genetic determinants that govern traits, which we now call genes, are passed from parent to offspring as discrete units. We can predict the outcome of many genetic crosses based on Mendel's laws of inheritance.

The inheritance patterns identified by Mendel can be explained by the existence of chromosomes and their behavior during cell division. Like Mendel's pea plants, sexually reproducing species are commonly **diploid.** This means that their cells contain two copies of each chromosome, one from each parent. The two copies are called **homologs** of each other. Because genes are located within chromosomes, diploid organisms have two copies of most genes. Humans, for example, have 46 chromosomes, which are found in homologous pairs (**Figure 1.10a**). With the exception of the sex chromosomes (X and Y), each homologous pair contains the same kinds of genes. For example, both copies of human chromosome 12 carry the gene that encodes phenylalanine hydroxylase, the enzyme that is nonfunctional in people with PKU. Therefore, each individual has two copies of this gene, which may or may not be identical alleles.

Most cells of the human body that are not directly involved in sexual reproduction contain 46 chromosomes. These cells are called **somatic cells.** In contrast, the **gametes**—sperm and egg cells—contain half that number (23) and are termed **haploid** (**Figure 1.10b**). The union of gametes during fertilization restores the diploid number of chromosomes. The primary advantage of sexual reproduction is that it enhances genetic variation. For example, a tall person with blue eyes and a short person with brown eyes may have short offspring with blue eyes or tall offspring with brown eyes. Therefore, sexual reproduction can result in new combinations of two or more traits that differ from those of either parent.

The Genetic Composition of a Species Evolves from Generation to Generation

As we have just seen, sexual reproduction has the potential to enhance genetic variation. This can be an advantage for a population of individuals as they struggle to survive and compete within their natural environment. The term **biological evolution,** or simply, **evolution,**



(a) Chromosomal composition found in human somatic cells of females (46 chromosomes)

(b) Chromosomal composition found in a human gamete (23 chromosomes)

FIGURE 1.10 The complement of human chromosomes in somatic cells and gametes. **(a)** A schematic drawing of the 46 chromosomes of a human. With the exception of the sex chromosomes, these are always found in homologous pairs in somatic cells, such as skin or nerve cells. **(b)** The chromosomal composition of a gamete, which contains only 23 chromosomes, one from each pair. This gamete contains an X chromosome. Half of the gametes from human males contain a Y chromosome instead of an X chromosome.

CONCEPT CHECK: The leaf cells of a corn plant contain 20 chromosomes each. How many chromosomes are found in a gamete made by a corn plant?

refers to the process of changes in the genetic makeup of a population from one generation to the next.

As suggested by Charles Darwin in the nineteenth century, the members of a species are in competition with one another for essential resources. Random genetic changes (i.e., mutations) occasionally occur within an individual's genes, and sometimes these changes lead to a modification of traits that promote reproductive success. For example, over the course of many generations, random gene mutations have lengthened the snout and extended the tongue of the anteater, enabling it to probe into the ground and feed on ants. When a mutation creates a new allele that is beneficial, the allele may become prevalent in future generations because the individuals carrying the allele are more likely to reproduce and pass the beneficial allele to their offspring. This process is known as **natural selection.** In this way, a species becomes better adapted to its environment.

Over a long period of time, the accumulation of many genetic changes may lead to rather striking modifications in a species' characteristics. As an example, **Figure 1.11** depicts the evolution of the modern-day horse. Over time, a variety of morphological changes occurred, including an increase in size, fewer toes, and modified jaw structure. The changes can be attributed to natural selection. Over North America, where much of horse evolution occurred,

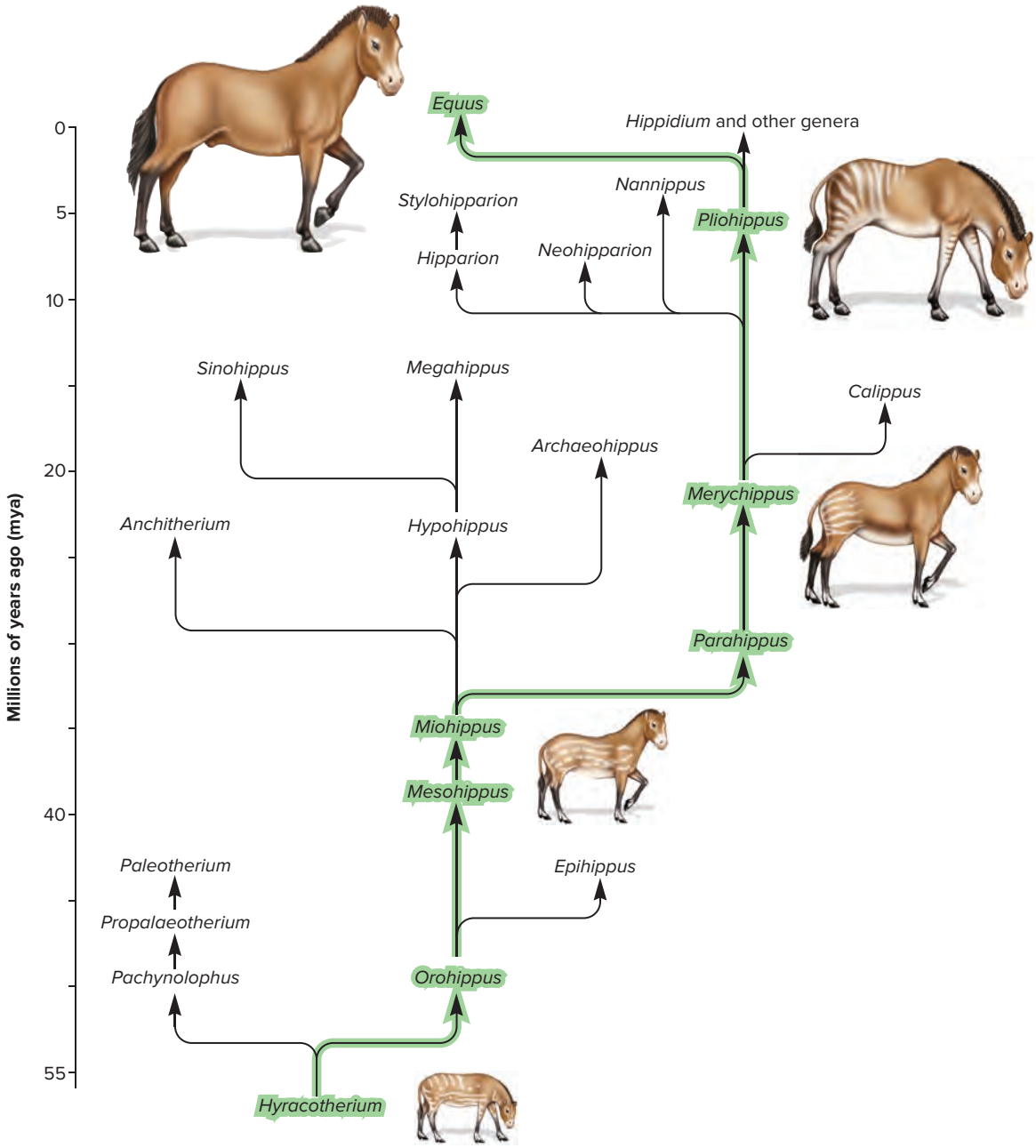


FIGURE 1.11 The evolutionary changes that led to the modern horse genus, *Equus*. Three important morphological changes that occurred were larger size, fewer toes, and a shift toward a jaw structure suited for grazing.

CONCEPT CHECK: According to the theory of evolution, why have these changes occurred in horse populations over the course of many generations?

large areas of dense forests were replaced with grasslands. The increase in size and changes in foot structure enabled horses to escape predators more easily and travel greater distances in search of food. Natural selection favored the changes seen in horses' teeth, because such changes allowed them to eat grasses and other types of vegetation that are tougher and require more chewing.

1.2 COMPREHENSION QUESTIONS

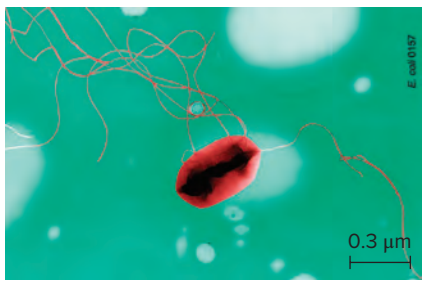
1. At which of the following levels can gene expression be observed?
   a. Molecular and cellular levels
   b. Organism level
   c. Population level
   d. All of the above
2. Variation in the traits of organisms may be attributable to
   a. gene mutations.
   b. alterations in chromosome structure.
   c. variation in chromosome number.
   d. all of the above.
3. A human skin cell has 46 chromosomes. A human sperm cell has
   a. 23.
   b. 46.
   c. 92.
   d. None of the above is the number of chromosomes in a sperm cell.
4. Evolutionary change caused by natural selection results in species with
   a. greater complexity.
   b. less complexity.
   c. greater reproductive success in their environment.
   d. the ability to survive longer.

1.3 FIELDS OF GENETICS

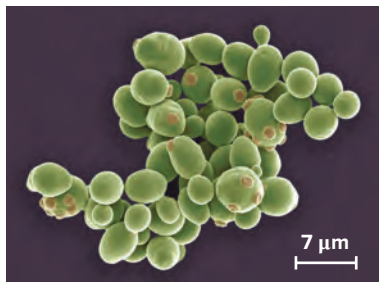
Learning Outcome:

1. Compare and contrast the three major fields of genetics: transmission, molecular, and population genetics.

Genetics is a broad discipline encompassing molecular, cellular, organism, and population biology. Many scientists who are interested in genetics have been trained in supporting disciplines such as biochemistry, biophysics, cell biology, mathematics, microbiology, population biology, ecology, agriculture, and medicine. Experimentally, geneticists often focus their efforts on **model organisms**—organisms studied by many different researchers so that they can compare their results and determine scientific principles that apply more broadly to other species. **Figure 1.12** shows some examples of model organisms: *Escherichia coli* (a bacterium), *Saccharomyces cerevisiae* (a yeast), *Drosophila melanogaster* (fruit fly), *Caenorhabditis elegans* (a nematode worm),



(a) *Escherichia coli*



(b) *Saccharomyces cerevisiae*

(c) *Drosophila melanogaster*

(d) *Caenorhabditis elegans*

(e) *Mus musculus*

(f) *Arabidopsis thaliana*

FIGURE 1.12 Examples of model organisms studied by geneticists. **(a)** *Escherichia coli* (a bacterium), **(b)** *Saccharomyces cerevisiae* (a yeast), **(c)** *Drosophila melanogaster* (fruit fly), **(d)** *Caenorhabditis elegans* (a nematode worm), **(e)** *Mus musculus* (mouse), and **(f)** *Arabidopsis thaliana* (a flowering plant).

(a): Source: Peggy S. Hayes & Elizabeth H. White, M.S./CDC; (b): ©Science Photo Library/Alamy; (c): ©Janeff/Getty Images; (d): ©Sinclair Stammers/Science Source; (e): ©G.K. & Vikki Hart/Getty Images; (f): ©WILDLIFE GmbH/Alamy

CONCEPT CHECK: Can you think of another example of a model organism?

Mus musculus (mouse), and *Arabidopsis thaliana* (a flowering plant). Model organisms offer experimental advantages over other species. For example, *E. coli* is a very simple organism that can be easily grown in the laboratory. By limiting their work to a few model organisms, researchers can more easily unravel the genetic mechanisms that govern the traits of a given species. Furthermore, the genes found in model organisms often function in a similar way to those found in humans.

The study of genetics has been traditionally divided into three areas—transmission, molecular, and population genetics—although there is some overlap of these three fields. In this section, we compare these three fields of genetics.

Transmission Genetics Explores the Inheritance Patterns of Traits as They Are Passed from Parents to Offspring

A scientist working in the field of transmission genetics examines the relationship between the transmission of genes from parent to offspring and the outcome of the offspring's traits. For example, how can two brown-eyed parents produce a blue-eyed child? Or why do tall parents tend to produce tall children, but not always? Our modern understanding of transmission genetics began with the studies of Gregor Mendel. His work provided the conceptual framework for transmission genetics. In particular, he originated the idea that factors, which we now call genes, are passed as discrete units from parents to offspring via sperm and egg cells. Since Mendel's pioneering studies in the 1860s, our knowledge of genetic transmission has skyrocketed. Many patterns of genetic transmission are more complex than the simple Mendelian patterns that are described in Chapter 2. The additional complexities of transmission genetics are examined in Chapters 3 through 8.

Experimentally, the fundamental technique used by a transmission geneticist is the **genetic cross**—the breeding of two selected individuals and then analyzing their offspring in an attempt to understand how traits are passed from parents to offspring. In the case of experimental organisms, the researcher chooses two parents with particular traits and then categorizes the offspring according to the traits they possess. In many cases, this analysis is quantitative in nature. For example, an experimenter may cross two tall pea plants and obtain 100 offspring that fall into two categories: 75 tall and 25 dwarf. As we will see in Chapter 2, the ratio of tall to dwarf offspring provides important information concerning the inheritance pattern of the height trait.

Molecular Genetics Focuses on a Biochemical Understanding of the Hereditary Material

The goal of molecular genetics, as the name of the field implies, is to understand how the genetic material works at the molecular level. In other words, molecular geneticists want to understand the molecular features of DNA and how these features underlie the expression of genes. The experiments of molecular geneticists are usually conducted within the confines of a laboratory. Their efforts frequently progress to a detailed analysis of DNA, RNA, and proteins, using a variety of techniques that are described throughout Parts III, IV, and V of this textbook.

Molecular geneticists often study mutant genes that have abnormal function. This is called a **genetic approach** to the study of a research question. In many cases, researchers analyze the effects of a gene mutation that eliminates the function of a gene. This is called a **loss-of-function mutation,** and the resulting version of the gene is called a **loss-of-function allele.** Studying the effect of such a mutation often reveals the role of the functional, nonmutant gene. For example, let's suppose that a particular plant species produces purple flowers. If a loss-of-function mutation within a given gene causes a plant of that species to produce white flowers, you might suspect that the role of the functional gene involves the production of purple pigmentation.

Studies within molecular genetics interface with other disciplines such as biochemistry, biophysics, and cell biology. In addition, advances within molecular genetics have shed considerable light on the areas of transmission and population genetics. Our quest to understand molecular genetics has spawned a variety of modern molecular technologies and computer-based approaches. Furthermore, discoveries within molecular genetics have had widespread applications in agriculture, medicine, and biotechnology.

Population Genetics Is Concerned with Genetic Variation and Its Role in Evolution

The foundations of population genetics arose during the first few decades of the twentieth century. Although many scientists of this era did not accept the findings of Mendel or Darwin, the theories of population genetics provided a compelling way to connect the two viewpoints. Mendel's work and that of many succeeding geneticists gave insight into the nature of genes and how they are transmitted from parents to offspring. The work of Darwin provided a natural explanation for the variation in characteristics observed among the members of a species. To relate these two phenomena, population geneticists have developed mathematical theories to explain the prevalence of certain alleles within populations of individuals. This work helps us understand how processes such as natural selection have resulted in the prevalence of individuals that carry particular alleles.

Population geneticists are particularly interested in genetic variation and how that variation is related to an organism's environment. In this field, the frequencies of alleles within a population are of central importance.

1.3 COMPREHENSION QUESTIONS

1. Which of the following is *not* a model organism?
   a. *Mus musculus* (laboratory mouse)
   b. *Escherichia coli* (a bacterium)
   c. *Saccharomyces cerevisiae* (a yeast)
   d. *Sciurus carolinensis* (gray squirrel)
2. A person studying the rate of transcription of a particular gene is working in the field of
   a. molecular genetics.
   b. transmission genetics.
   c. population genetics.
   d. None of the above is correct.

1.4 THE SCIENCE OF GENETICS

Learning Outcomes:

1. Describe what makes genetics an experimental science.
2. Outline different strategies for solving problems in genetics.

Science is a way of knowing about our natural world. The science of genetics allows us to understand how the expression of our genes produces the traits that we possess. In this section, we will consider how scientists attempt to answer questions via experimentation. We will also consider general approaches for solving problems.

Genetics Is an Experimental Science

Researchers typically follow two general types of scientific approaches: hypothesis testing and discovery-based science. In **hypothesis testing,** also called the **scientific method,** scientists follow a series of steps to reach verifiable conclusions about the world. Although scientists arrive at their theories in different ways, the scientific method provides a way to validate (or invalidate) a particular hypothesis. Alternatively, research may also involve the collection of data without a preconceived hypothesis. For example, researchers might analyze the genes found in cancer cells to identify those that have become mutant. In this case, the scientists may not have a hypothesis about which particular genes may be involved. The collection and analysis of data without the need for a preconceived hypothesis is called **discovery-based science** or, simply, discovery science.

In traditional science textbooks, the emphasis often lies on the product of science. That is, many textbooks are aimed primarily at teaching the student about the observations scientists have made and the hypotheses they have proposed to explain those observations. Along the way, the student is provided with many bits and pieces of experimental techniques and data. Likewise, this textbook also provides you with many observations and hypotheses. However, it attempts to go one step further. Most chapters contain one or two figures presenting experiments that have been "dissected" into five individual components to help you to understand the entire scientific process:

1. Background information is provided so that you can appreciate observations that were known prior to conducting the experiment.
2. Most experiments involve the testing of a hypothesis via the scientific method. In those cases, the figure presenting the experiment states the hypothesis the scientists were trying to test. In other words, what scientific question were the researchers trying to answer?
3. Next, the figure follows the experimental steps the scientists took to test the hypothesis. The figure presents two parallel illustrations labeled "Experimental Level" and "Conceptual Level." The experimental level helps you to understand the techniques that were used. The conceptual level helps you to understand what is actually happening at each step in the procedure.
4. The raw data from the experiment are then presented.
5. Last, an interpretation of the data is offered within the text.

The rationale behind this approach is that it enables you to see the experimental process from beginning to end. As you read through the chapters, the experiments will help you to see the relationship between science and scientific theories.

As a student of genetics, you will be given the opportunity to involve your mind in the experimental process. As you are reading an experiment, you may find yourself thinking about different approaches and alternative hypotheses. Different people can view the same data and arrive at very different conclusions. As you progress through the experiments in this book, you will enjoy genetics far more if you try to develop your skills at formulating hypotheses, designing experiments, and interpreting data. Also, some of the questions in the problem sets are aimed at refining these skills.

Finally, it is worthwhile to point out that science is a social discipline. As you develop your skills at scrutinizing experiments, it is fun to discuss your ideas with other people, including fellow students and faculty members. Keep in mind that you do not need to "know all the answers" before you enter into a scientific discussion. Instead, it is more rewarding to view science as an ongoing and never-ending dialogue.

Genetic TIPS Will Help You to Improve Your Problem-Solving Skills

As your progress through this textbook, your learning will involve two general goals:

- You will gather foundational knowledge. In other words, you will be able to describe core concepts in genetics. For example, you will be able to explain how DNA replication occurs and describe the proteins that are involved in this process.
- You will develop problem-solving skills that allow you to apply that foundational knowledge in different ways. For example, you will learn how to use statistics to determine if a genetic hypothesis is consistent with experimental data.

The combination of foundational knowledge and problem-solving skills will enable you not only to understand genetics, but also to apply your knowledge in different situations. To help you develop these skills, Chapters 2 through 29 contain solved problems named Genetic TIPS, which stands for Topic, Information, and Problem-solving Strategy. These solved problems follow a consistent pattern.

GENETIC TIPS **THE QUESTION:** All of the Genetic TIPS begin with a question. As an example, let's consider the following question:

The coding strand of DNA in a segment of a gene is as follows: ATG GGC CTT AGC. This strand carries the information to make a region of a polypeptide with the amino acid sequence, methionine-glycine-leucine-serine. What would be the consequences if a mutation changed the second cytosine (C) in this sequence to an adenine (A)?

TOPIC: ***What topic in genetics does this question address?*** The topic is gene expression. More specifically, the question is about the relationship between a gene sequence and the genetic code.

INFORMATION: ***What information do you know based on the question and your understanding of the topic?*** In the question, you are given the base sequence of a short segment of a gene and told that one of the bases has been changed. From your understanding of the topic, you may remember that a polypeptide sequence is determined by reading the mRNA (transcribed from a gene) in groups of three bases called codons.

PROBLEM-SOLVING STRATEGY: ***Compare and contrast.*** One strategy to solve this problem is to compare the mRNA sequence (transcribed from this gene) before and after the mutation:

Original: AUG GGC **C**UU AGC
Mutant: AUG GGC **A**UU AGC

ANSWER: The mutation has changed the sequence of bases in the mRNA so the third codon has changed from CUU to AUU. Because codons specify amino acids, this alteration may change the third amino acid to something else. Note: If you look ahead to Chapter 13 (see Table 13.1), you will see that CUU specifies leucine, whereas AUU specifies isoleucine. Therefore, you would predict that the mutation would change the third amino acid from leucine to isoleucine.

Throughout Chapters 2 through 29, each chapter will contain several Genetic TIPS. Some of these will be within the chapter itself and some will precede the problem set that is at the end of each chapter. Though there are many different problem-solving strategies, Genetic TIPS will focus on ten strategies that will help you to solve problems. You will see these ten strategies over and over again as you progress through the textbook:

1. *Define key terms.* In some cases, a question may be difficult to understand because you don't know the meaning of one or more key terms in the question. If so, you will need to begin your problem-solving by defining such terms, either by looking them up in the glossary or by using the index to find the location in the text where the key terms are explained.
2. *Make a drawing.* Genetic problems are often difficult to solve in your head. Making a drawing may make a big difference in your ability to see the solution.
3. *Predict the outcome.* Geneticists may want to predict the outcome of an experiment. For example, in Chapters 3 through 6, you will learn about different ways to predict the outcome of genetic crosses. Becoming familiar with these methods will help you to predict the outcomes of particular experiments.
4. *Compare and contrast.* Making a direct comparison between two things, such as two RNA sequences, may help you to understand how they are similar and how they are different.
5. *Relate structure and function.* A recurring theme in biology and genetics is that structure determines function. This relationship holds true at many levels of biology, including the molecular, microscopic, and macroscopic levels. For some questions, you will need to understand how certain structural features are related to their biological functions.
6. *Describe the steps.* At first, some questions may be difficult to understand because they may involve mechanisms that occur in a series of several steps. Sometimes, if you sort out the steps, you may identify the key step that you need to understand to solve the problem.
7. *Propose a hypothesis.* A hypothesis is an attempt to explain an observation or data. Hypotheses may be made in many forms, including statements, models, equations, and diagrams.
8. *Design an experiment.* Experimental design lies at the heart of science. In many cases, an experiment begins with some type of starting material(s), such as strains of organisms or purified molecules, and then the starting materials are subjected to a series of steps. The experiments featured throughout the textbook will also help you refine the skill of designing experiments.
9. *Analyze data.* The results from an experiment produce data that can be analyzed in order to accept or reject a hypothesis. Many of the Genetic TIPS give you practice at analyzing data. For example, a variety of different statistical methods are used to analyze data and make conclusions about what the data mean.
10. *Make a calculation.* Genetics is a quantitative science. Researchers have devised mathematical relationships to understand and predict genetic phenomena. Becoming familiar with these mathematical relationships will help you to better understand genetic concepts and to make predictions.

For most problems throughout this textbook, one or more of these strategies may help you to arrive at the correct solution. Genetic TIPS will provide you with practice at applying these ten problem-solving strategies.

1.4 COMPREHENSION QUESTION

1. The scientific method involves which of the following?
   a. The collection of observations and the formulation of a hypothesis
   b. Experimentation
   c. Data analysis and interpretation
   d. All of the above

KEY TERMS

Introduction: genome
1.1: genetics, gene, traits, nucleic acids, proteins, carbohydrates, lipids, macromolecules, proteome, enzymes, deoxyribonucleic acid (DNA), nucleotides, polypeptides, genetic code, codon, amino acid, chromosomes, karyotype, gene expression, transcription, ribonucleic acid (RNA), protein-encoding genes (structural genes), messenger RNA (mRNA), translation, central dogma of genetics
1.2: morphological traits, physiological traits, behavioral traits, molecular level, cellular level, organism level, species, population level, alleles, genetic variation, morphs, gene mutations, environment, phenylketonuria (PKU), diploid, homologs, somatic cells, gametes, haploid, biological evolution (evolution), natural selection
1.3: model organisms, genetic cross, genetic approach, loss-of-function mutation, loss-of-function allele
1.4: hypothesis testing, scientific method, discovery-based science

CHAPTER SUMMARY

- The complete genetic composition of a cell is its genome. The genome encodes all of the proteins a cell can make. Many key discoveries in genetics are related to the study of genes and genomes (see Figures 1.1, 1.2, 1.3).

1.1 The Molecular Expression of Genes

- Living cells are composed of nucleic acids (DNA and RNA), proteins, carbohydrates, and lipids (see Figure 1.4).
- The entire collection of proteins a cell makes at a given time is its proteome. The proteome determines the structure and function of a cell.
- DNA, which is found within chromosomes, stores the information needed to make cellular proteins (see Figure 1.5).
- Most genes encode polypeptides that are units within functional proteins. Gene expression at the molecular level involves transcription to produce mRNA and translation to produce a polypeptide (see Figure 1.6).

1.2 The Relationship Between Genes and Traits

- Genetics, which governs an organism's traits, is studied at the molecular, cellular, organism, and population levels (see Figure 1.7).
- Genetic variation underlies variation in traits. In addition, the environment plays a key role (see Figures 1.8, 1.9).
- During reproduction, genetic material is passed from parents to offspring. In many species, somatic cells are diploid and have two sets of chromosomes, whereas gametes are haploid and have a single set (see Figure 1.10).
- Evolution refers to changes in the genetic composition of a population from one generation to the next (see Figure 1.11).

1.3 Fields of Genetics

- Genetics is traditionally divided into transmission genetics, molecular genetics, and population genetics, though overlap occurs among these fields. Many geneticists study model organisms (see Figure 1.12).

1.4 The Science of Genetics

- Researchers in genetics carry out hypothesis testing or discovery-based science.
- Genetic TIPS are aimed at improving your ability to solve problems.

PROBLEM SETS & INSIGHTS

MORE GENETIC TIPS **1.** A human gene called the *CFTR* gene (for cystic fibrosis transmembrane regulator) encodes a protein that functions in the transport of chloride ions across the cell membrane. Most people have two copies of a functional *CFTR* gene and do not have cystic fibrosis. However, a mutant version of the *CFTR* gene is found in some people. If a person has two mutant copies of the gene, he or she develops the disease known as cystic fibrosis. Are the following descriptions of this disease related to genetics at the molecular, cellular, organism, or population level?

A. People with cystic fibrosis have lung problems due to a buildup of thick mucus in their lungs.

B. The mutant *CFTR* gene encodes a defective chloride transporter.

C. A defect in the chloride transporter causes a salt imbalance in lung cells.

D. Scientists have wondered why the mutant *CFTR* gene is relatively common. In fact, it is the most common mutant gene that causes a severe disease in people of Northern European descent.

One possible explanation why cystic fibrosis is so common is that people who have one copy of the functional *CFTR* gene and one copy of the mutant gene may be more resistant to diarrheal diseases such as cholera. Therefore, even though individuals with two mutant copies have the disorder, people with one mutant copy and one functional copy might have a survival advantage over people with two functional copies of the gene.

TOPIC: ***What topic in genetics does this question address?*** The topic is the different levels at which genetics is studied, ranging from the molecular to the population level.

INFORMATION: ***What information do you know based on the question and your understanding of the topic?*** The question describes the disease called cystic fibrosis. Parts A through D give descriptions of various aspects of the disease. From your understanding of the topic, you may remember that genetics can be studied at the molecular, cellular, organism, and population level. This concept is described in Figure 1.7.

PROBLEM-SOLVING STRATEGY: ***Make a drawing. Compare and contrast.*** One strategy to solve this problem is to make a drawing of what is described in each of parts A through D and decide if you are drawing something at the molecular, cellular, organism, or population levels. For example, if you drew the description in part B, you would be drawing a protein, which is a molecule. If you drew the description in part C, you would be drawing a cell in which a salt imbalance is present. Another strategy to solve this problem would be to compare and contrast parts A, B, C, and D with each other. For example, if you compared part A and part D, you might realize that part A is describing something in one person, whereas part D is describing the occurrence of the mutant gene in multiple people.

ANSWER:

A. Organism level. This is a description of a trait at the level of an entire individual.

B. Molecular level. This is a description of a gene and the protein it encodes.

C. Cellular level. This is a description of how protein function affects the cell.

D. Population level. This is a possible explanation of why two alleles of the gene occur within a population.

2. Most genes encode proteins. Explain how proteins produce an organism's traits. Provide examples.

TOPIC: ***What topic in genetics does this question address?*** The topic is the relationship between genes and traits. More specifically, the question is about how proteins, which are encoded by genes, produce an organism's traits.

INFORMATION: ***What information do you know based on the question and your understanding of the topic?*** In the question, you are reminded that most genes encode proteins and that proteins play a role in producing an organism's traits. From your understanding of the topic, you may remember that proteins carry out a variety of functions that determine cell structure and function.

PROBLEM-SOLVING STRATEGY: ***Relate structure and function.*** One strategy you can use to solve this problem is to consider the relationship between protein structure and function. Think about examples in which the structure and function of proteins govern the structure and function of living cells. Also, consider how the structures and functions of cells determine an organism's traits.

ANSWER: The structure and function of proteins govern the structure and function of living cells. For example, specific proteins help determine the shape and structure of a given cell. The protein known as tubulin can assemble into large structures known as microtubules, which provide the cell with internal structure and organization. The proteins that a cell makes are largely responsible for the cell's structure and function. For example, the proteins made by a nerve cell cause the cell to be very elongated and to be able to transmit signals to and from other cells. The structure of a nerve cell provides animals with many traits, such as the ability to sense the temperature of their environment and the ability to send signals to their muscles to promote movement.

Conceptual Questions

C1. Pick any example of a genetic technology, and describe how it has directly affected your life.

C2. At the molecular level, what is a gene? Where are genes located?

C3. Most genes encode polypeptides, which are functional units of proteins. Explain how the structure and function of proteins produce an organism's traits.

C4. Briefly explain how gene expression occurs at the molecular level.

C5. A human gene called the β-globin gene encodes a polypeptide that functions as a subunit of the protein known as hemoglobin. Hemoglobin is found within red blood cells; it carries oxygen. In human populations, the β-globin gene can be found as the common allele called the Hb^A allele, and it can also be found as the Hb^S allele. Individuals who have two copies of the Hb^S allele have the disease called sickle cell disease. Are the following descriptions examples of genetics at the molecular, cellular, organism, or population level?

A. The Hb^S allele encodes a polypeptide that functions slightly differently from the polypeptide encoded by the Hb^A allele.

B. If an individual has two copies of the Hb^S allele, that person's red blood cells take on a sickle shape.

C. Individuals who have two copies of the Hb^A allele do not have sickle cell disease, but they are not resistant to malaria. People

who have one Hb^A allele and one Hb^S allele do not have sickle cell disease, and they are resistant to malaria. People who have two copies of the Hb^S allele have sickle cell disease, and this disease may significantly shorten their lives.

D. Individuals with sickle cell disease have anemia because their red blood cells are easily destroyed by the body.

C6. What is meant by the term *genetic variation*? Give two examples of genetic variation not discussed in this chapter. What causes genetic variation at the molecular level?

C7. What is the cause of Down syndrome?

C8. The text describes how the detrimental symptoms associated with the disease phenylketonuria (PKU) are caused by a faulty gene. However, a change in diet can prevent these symptoms. Pick a trait of your favorite plant species, and explain how genetics and the environment may play important roles in the outcome of that trait.

C9. What is meant by the term *diploid*? Which cells of the human body are diploid, and which cells are not?

C10. What is a DNA sequence?

C11. What is the genetic code?

C12. Explain the relationship between each of these pairs of genetic terms:

A. Gene and trait

B. Gene and chromosome

C. Allele and gene

D. DNA sequence and amino acid sequence

C13. With regard to biological evolution, which of the following statements is incorrect? Explain why.

A. During its lifetime, an animal evolves to become better adapted to its environment.

B. The process of biological evolution has produced species that are better adapted to their environments.

C. When an animal is better adapted to its environment, the process of natural selection makes it more likely that the animal will reproduce.

C14. What are the primary interests of researchers working in the following fields of genetics?

A. Transmission genetics

B. Molecular genetics

C. Population genetics

Experimental Questions

E1. What is a genetic cross?

E2. The technique known as DNA sequencing (described in Chapter 20) enables researchers to determine the DNA sequence of genes. Would this technique be used primarily by transmission geneticists, molecular geneticists, or population geneticists?

E3. Figure 1.5 shows a micrograph of chromosomes from a normal human cell. If you created this kind of image using a cell from a person with Down syndrome, what would you expect to see?

E4. Many organisms are studied by geneticists. Do you think each of the following species would be more likely to be studied by a transmission geneticist, a molecular geneticist, or a population geneticist? Explain your answer. Note: More than one answer may be possible for a given species.

A. Dogs

B. *E. coli*

C. Fruit flies

D. Leopards

E. Corn

E5. Pick any trait you like in any species of wild plant or animal. The trait must somehow vary among different members of the species (see Figure 1.7). Note: When picking a trait to answer this question, do not pick the trait of wing color in butterflies.

A. Summarize all of the background information that you already have (from personal observations) regarding this trait.

B. Propose a hypothesis that would explain the genetic variation within the species. For example, in the case of the butterflies, your hypothesis might be that the dark butterflies survive better in dark forests and the light butterflies survive better in sunlit fields.

C. Describe the experimental steps you would follow to test your hypothesis.

D. Describe the possible data you might collect.

E. Interpret your data.

Note: All answers appear in Connect; the answers to the even-numbered questions and all of the Concept Check and Comprehension Questions are in Appendix B.

CHAPTER OUTLINE

2.1 Mendel's Study of Pea Plants
2.2 Law of Segregation
2.3 Law of Independent Assortment
2.4 Studying Inheritance Patterns in Humans
2.5 Probability and Statistics

The garden pea, studied by Mendel.

2 MENDELIAN INHERITANCE

An appreciation for the concept of heredity can be traced far back in human history. Hippocrates, a Greek physician, was the first person to provide an explanation for hereditary traits (around 400 B.C.E.). He suggested that "seeds" are produced by all parts of the body and then collected and transmitted to the offspring at the time of conception. Furthermore, he hypothesized that these seeds cause certain traits of the offspring to resemble those of the parents. This idea, known as pangenesis, was the first attempt to explain the transmission of hereditary traits from generation to generation.

The first systematic studies of genetic crosses were carried out by German botanist Joseph Kölreuter from 1761 to 1766. In crosses between different strains of tobacco plants, he found that the offspring were usually intermediate in appearance between the two parents. This observation led Kölreuter to conclude that both parents make equal genetic contributions to their offspring. Furthermore, his observation was consistent with the blending hypothesis of inheritance. According to this idea, the factors that dictate hereditary traits could blend together from generation to generation. The blended traits would then be passed to the next generation. Before the 1860s, the popular view, which combined the notions of pangenesis and blending inheritance, was that hereditary traits were rather malleable and could change and blend over the course of one or two generations. However, the pioneering work of Gregor Mendel would prove instrumental in refuting this viewpoint.

In this chapter, we will first examine the outcome of Mendel's crosses of pea plants. We begin our inquiry into genetics here because the inheritance patterns observed in peas are fundamentally related to inheritance patterns found in other eukaryotic species, such as corn, fruit flies, mice, and humans. We will discover how Mendel's insights into the patterns of inheritance in pea plants revealed some simple rules that govern the process of inheritance. In Chapters 3 through 8, we will explore more complex patterns of inheritance and also consider the role that chromosomes play as the carriers of the genetic material.

In the last section of this chapter, we will examine some general concepts in probability and statistics. How are statistical methods useful? First, probability calculations allow us to predict the outcomes of simple genetic crosses, as well as the outcomes of more complicated crosses described in later chapters. In addition, we will learn how to use statistics to test the validity of genetic hypotheses that attempt to explain the inheritance patterns of traits.

2.1 MENDEL'S STUDY OF PEA PLANTS

Learning Outcomes:

1. Describe the characteristics of pea plants that make them a suitable organism to study genetically.
2. Outline the steps that Mendel followed to make crosses between different strains of pea plants.
3. List the seven characteristics of pea plants that Mendel chose to study.

Gregor Johann Mendel, born in 1822, is now remembered as a pioneer of genetics (**Figure 2.1**). He grew up on a small farm in Hyncice (formerly Heinzendorf) in northern Moravia, which was then a part of Austria and is now a part of the Czech Republic. Instead of becoming a farmer, however, Mendel entered the Augustinian monastery of St. Thomas, completed his studies for the priesthood, and was ordained in 1847. Soon after becoming a priest, Mendel worked for a short time as a substitute teacher. To continue that role, he needed to obtain a teaching license from the government. Surprisingly, he failed the licensing exam due to poor answers in the areas of physics and natural history. Therefore, Mendel then enrolled at the University of Vienna to expand his knowledge in these two areas. Mendel's training in physics and mathematics taught him to perceive the world as an orderly place, governed by natural laws. In his studies, Mendel learned that these natural laws could be stated as simple mathematical relationships.

In 1856, Mendel began his historic studies on pea plants. For 8 years, he grew and crossed thousands of pea plants in a small 23- by 115-foot garden. He kept meticulously accurate records that included quantitative data concerning the outcomes of his crosses. He published his work, entitled *Experiments on Plant Hybrids*, in 1866. This paper was largely ignored by scientists at that time, possibly because of its title, which did not reveal the key observations he made. Another reason his work went unrecognized could be a lack of understanding of chromosomes and their transmission, a topic we will discuss in Chapter 3. Nevertheless, Mendel's ground-breaking work allowed him to propose the natural laws that now provide a framework for our understanding of genetics.

Prior to his death in 1884, Mendel reflected, "My scientific work has brought me a great deal of satisfaction and I am convinced that it will be appreciated before long by the whole world." Sixteen years later, in 1900, the work of Mendel was independently rediscovered by three biologists with an interest in plant genetics: Hugo de Vries of Holland, Carl Correns of Germany, and Erich von Tschermak of Austria. Within a few years, the influence of Mendel's studies was felt around the world. In this section, we will examine Mendel's experimental approach.

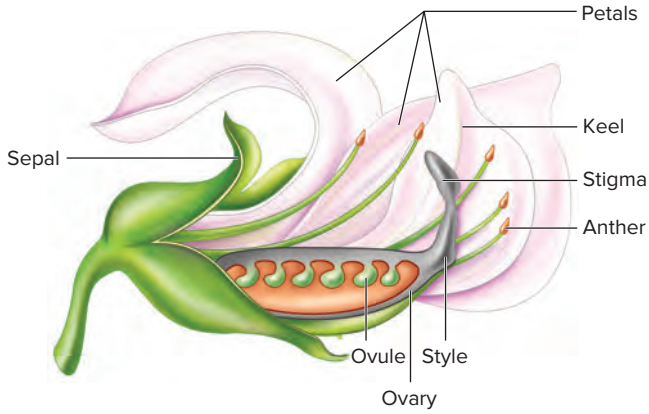
Mendel Chose Pea Plants as His Experimental Organism

Mendel's study of genetics grew out of his interest in ornamental flowers. Prior to his work with pea plants, many plant breeders had conducted experiments aimed at obtaining flowers with new colors. When two distinct individuals with different characteristics are bred to each other, the experiment is called a **cross,** or a **hybridization,** and the offspring are referred to as **hybrids.** For example, a hybridization experiment could involve a cross between a purple-flowered plant and a white-flowered plant. Mendel was particularly intrigued by the consistency with which offspring of subsequent generations showed characteristics of one or the other parent. His educational training in physics and the natural sciences led him to consider that this regularity might be rooted in natural laws that could be expressed mathematically. To uncover these laws, he realized that he needed to carry out quantitative experiments in which the numbers of offspring carrying certain traits were carefully recorded and analyzed.

FIGURE 2.1 **Gregor Johann Mendel.**
Source: National Library of Medicine

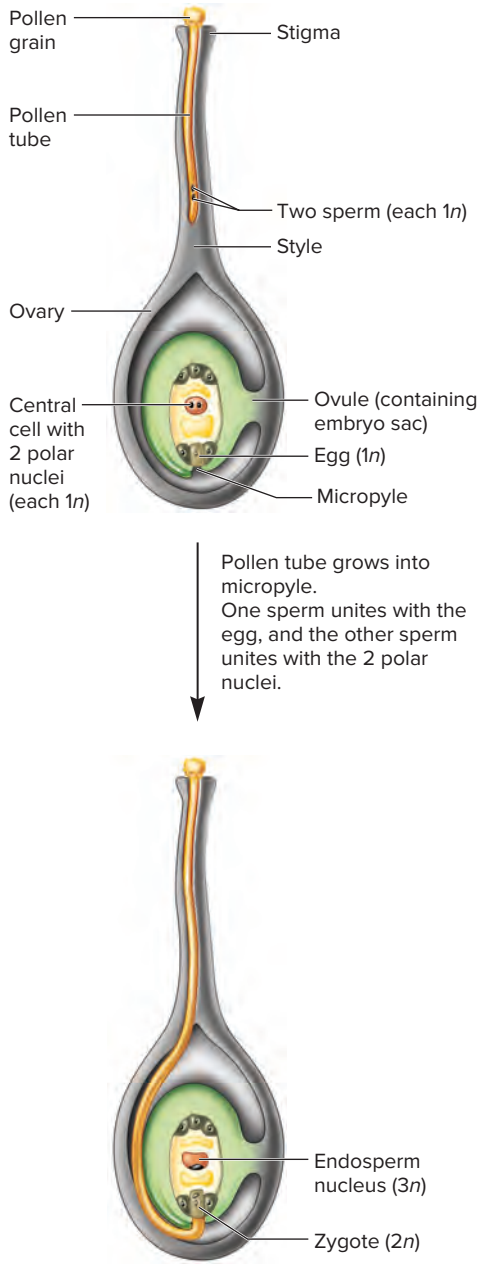
Mendel chose the garden pea, *Pisum sativum*, to investigate the natural laws that govern plant hybrids. The reproductive features of this plant are shown in **Figure 2.2**. Several properties of this species were particularly advantageous for studying plant hybridization. First, the species was available in several varieties, which varied in height and in the appearance of their flowers, seeds, and pods.

A second important feature of pea plants for Mendel's studies was the ease of making crosses. The term **gamete** is used to



(a) Structure of a pea flower

(b) A flowering pea plant



(c) Pollination and fertilization

FIGURE 2.2 Flower structure and pollination in pea plants. **(a)** The pea flower produces both pollen and egg cells. The pollen grains are produced within the anthers, and the egg cells are produced within the ovules that are contained within the ovary. The keel is two modified petals that are fused and enclose the anthers and ovaries. In this drawing, some of the keel is not shown so that the internal reproductive structures of the flower can be seen. **(b)** Photograph of a flowering pea plant. **(c)** A pollen grain must first land on the stigma. After this occurs, the pollen grain sends out a long tube through which two sperm cells travel toward an ovule to reach an egg cell. The fusion between a sperm and an egg cell results in fertilization and creates a zygote. The second sperm fuses with a central cell containing two polar nuclei to create the endosperm. The endosperm provides nutritive material for the developing embryo.

CONCEPT CHECK: Prior to fertilization, where is a pea plant's male gamete located?

describe haploid reproductive cells that fuse to form a zygote. In flowering plants, reproduction occurs by a pollination event (Figure 2.2c). Male gametes **(sperm cells)** are produced within **pollen grains** that form in the **anthers,** and the female gametes **(egg cells)** are produced within **ovules** that form in the **ovaries.** For fertilization to occur, a pollen grain lands on the **stigma,** which stimulates the growth of a pollen tube. This tube enables sperm cells to enter the stigma and migrate toward an ovule. Fertilization takes place when a sperm enters the micropyle, an opening in the ovule wall, and fuses with an egg cell.

In some experiments, Mendel allowed plants to reproduce by **self-fertilization,** which means that the pollen and eggs are derived from the same plant. In peas, two modified petals are fused to form a keel that encloses the reproductive structures of a flower. Because of this covering, pea plants naturally reproduce by self-fertilization. Usually, pollination occurs even before the flower opens.

In other experiments, however, Mendel wanted to make crosses between different plants. How did he accomplish this goal? Fortunately, pea plants contain relatively large flowers that are easy to manipulate, making it possible to cross two particular plants and study the outcomes. This process, known as **cross-fertilization,** requires that the pollen from one plant be placed on the stigma of another plant. The procedure is shown in **Figure 2.3**. Mendel was able to pry open immature flowers and remove the anthers before they produced pollen. Therefore, these flowers could not self-fertilize. He then obtained pollen from another plant by gently touching its mature anthers with a paintbrush. Mendel applied this pollen to the stigma of the flower that already had its anthers removed. In this way, he was able to cross-fertilize his pea plants, thereby obtaining any type of hybrid he wanted.

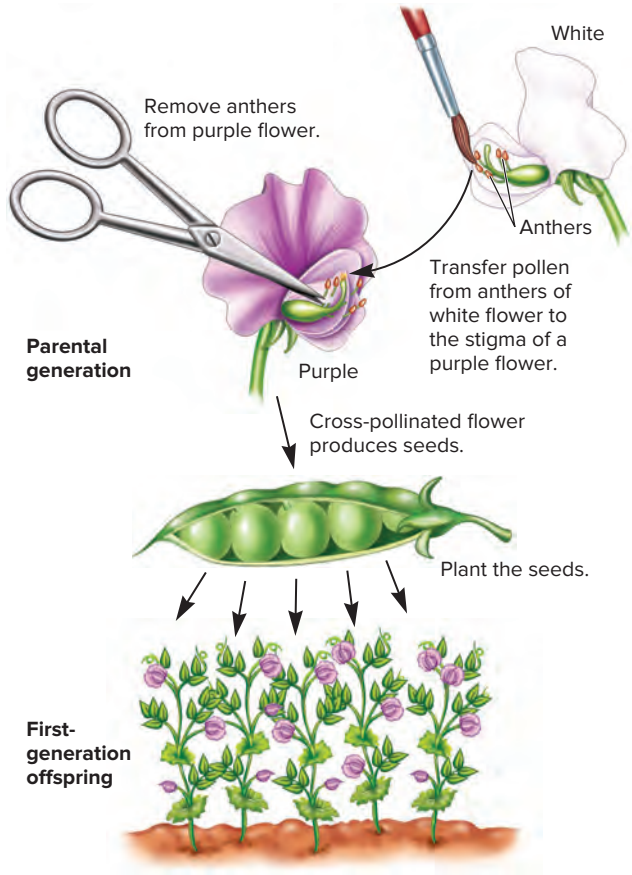


FIGURE 2.3 How Mendel cross-fertilized pea plants. This illustration depicts a cross between one plant with purple flowers and another with white flowers. The offspring from this cross are the result of pollination of the purple flower using pollen from a white flower.

CONCEPT CHECK: In this experiment, which plant, the white- or purple-flowered one, is providing the egg cells, and which is providing the sperm cells?

Mendel Studied Seven Characteristics That Bred True

When he initiated his studies, Mendel obtained several varieties of peas that were considered to be distinct. These plants had many different morphological characteristics. The general characteristics of an organism are called **characters.** The term **trait,** or **variant,** is typically used to describe the specific properties of a character. For example, eye color is a character of humans, and blue eye color is the trait (or variant) found in some people.

Over the course of 2 years, Mendel tested his pea strains to determine if their characteristics bred true. Breeding true means that a trait does not vary in appearance from generation to generation. For example, if the seeds from a pea plant were yellow, the next generation would also produce yellow seeds. Likewise, if these offspring were allowed to self-fertilize, all of their offspring would also produce yellow seeds, and so on. A variety that continues to produce the same trait after several generations of self-fertilization is called a **true-breeding line,** or **true-breeding strain.**

Mendel next concentrated his efforts on the analysis of characters that were clearly distinguishable between different true-breeding lines. **Figure 2.4** illustrates the seven characters that Mendel eventually chose to follow in his breeding experiments. All seven were found in two variants. For example, one character he followed was height, which was found in two variants: tall and dwarf plants.

2.1 COMPREHENSION QUESTIONS

1. Experimental advantages of using pea plants include which of the following?
   a. They came in several different varieties.
   b. They were capable of self-fertilization.
   c. They were easy to cross.
   d. All of the above were advantages.
2. With regard to Mendel's experiments, the term *cross* refers to an experiment in which
   a. the gametes come from different individuals.
   b. the gametes come from a single flower of the same individual.
   c. the gametes come from different flowers of the same individual.
   d. Both a and c are true.
3. To avoid self-fertilization in his pea plants, Mendel had to
   a. spray the plants with a chemical that damaged the pollen.
   b. remove the anthers from immature flowers.
   c. grow the plants in a greenhouse that did not contain pollinators (e.g., bees).
   d. do all of the above.

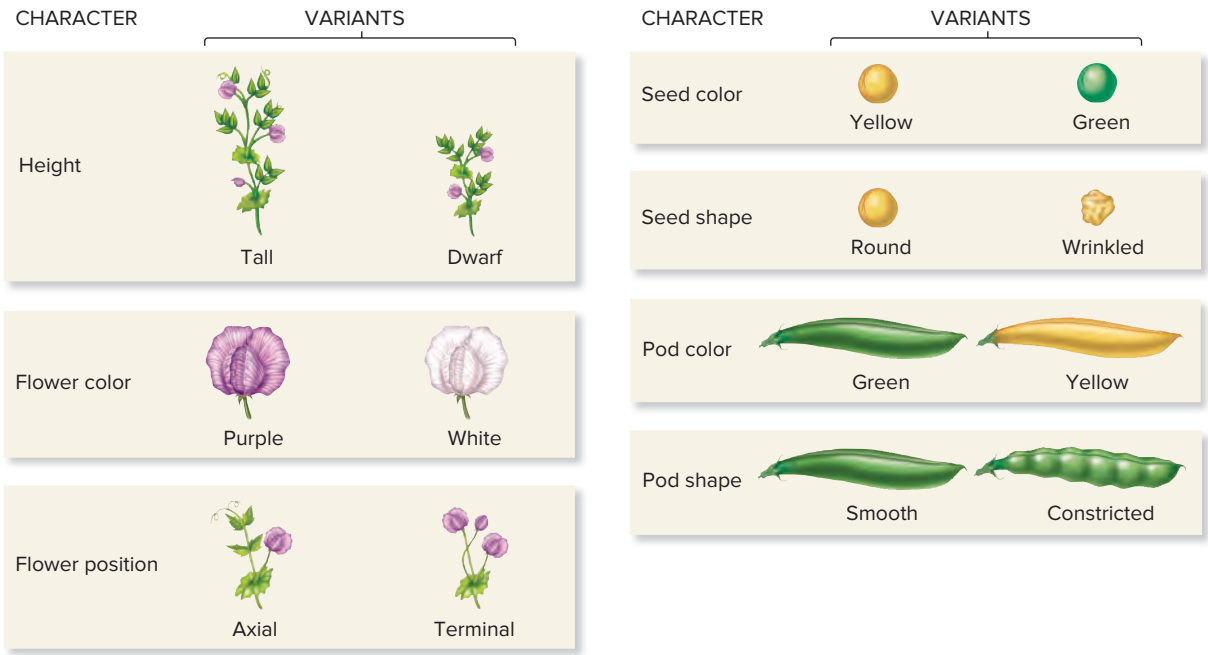


FIGURE 2.4 An illustration of the seven characters that Mendel studied. Each character was found as two variants that were decisively different from each other.

CONCEPT CHECK: What do we mean when we say a line is true-breeding?

2.2 LAW OF SEGREGATION

Learning Outcomes:

1. Analyze Mendel's experiments involving single-factor crosses.
2. State Mendel's law of segregation, and explain how it is related to gamete formation and fertilization.
3. Predict the outcome of a single-factor cross or a self-fertilization experiment using a Punnett square.

As discussed in the previous section, Mendel carried out self-fertilization or cross-fertilization experiments with his pea plants. In this section, we will examine how he studied the inheritance of characters by crossing variants to each other. A cross in which an experimenter observes one character is called a **single-factor cross.** A cross between two parents with different variants for a given character produces single-character hybrids, also known as **monohybrids.** As you will learn, this type of experimental approach led Mendel to propose the law of segregation.

EXPERIMENT 2A

Mendel Followed the Outcome of a Single Character for Two Generations

Prior to conducting his studies, Mendel did not already have a hypothesis to explain the formation of hybrids. However, his educational background caused him to realize that a quantitative analysis of crosses might uncover mathematical relationships that would otherwise be mysterious. His experiments were designed to determine the relationships that govern hereditary traits. This rationale is called an **empirical approach.** Laws deduced from an empirical approach are known as empirical laws.

Mendel's experimental procedure is shown in **Figure 2.5**. He began with true-breeding plants that differed in a single character. These are termed the **parental generation,** or **P generation.** Crossing true-breeding parents to each other, called a P cross, produces the offspring that constitute the **F_1 generation,** or first filial generation (from the Latin *filius*, meaning "son"). As seen in the data, all plants of the F_1 generation showed the trait of one parent but not the other. This prompted Mendel to follow the transmission of this character for one additional generation. To do so, the plants of the F_1 generation were allowed to self-fertilize to produce a second generation called the **F_2 generation,** or second filial generation.

THE GOAL (DISCOVERY-BASED SCIENCE)

Mendel speculated that the inheritance pattern for a single character may follow quantitative natural laws. The goal of this experiment was to uncover such laws.

ACHIEVING THE GOAL **FIGURE 2.5** **Mendel's analysis of single-factor crosses.**

Starting material: Mendel began his experiments with true-breeding strains of pea plants that varied in only one of seven different characters (see Figure 2.4).

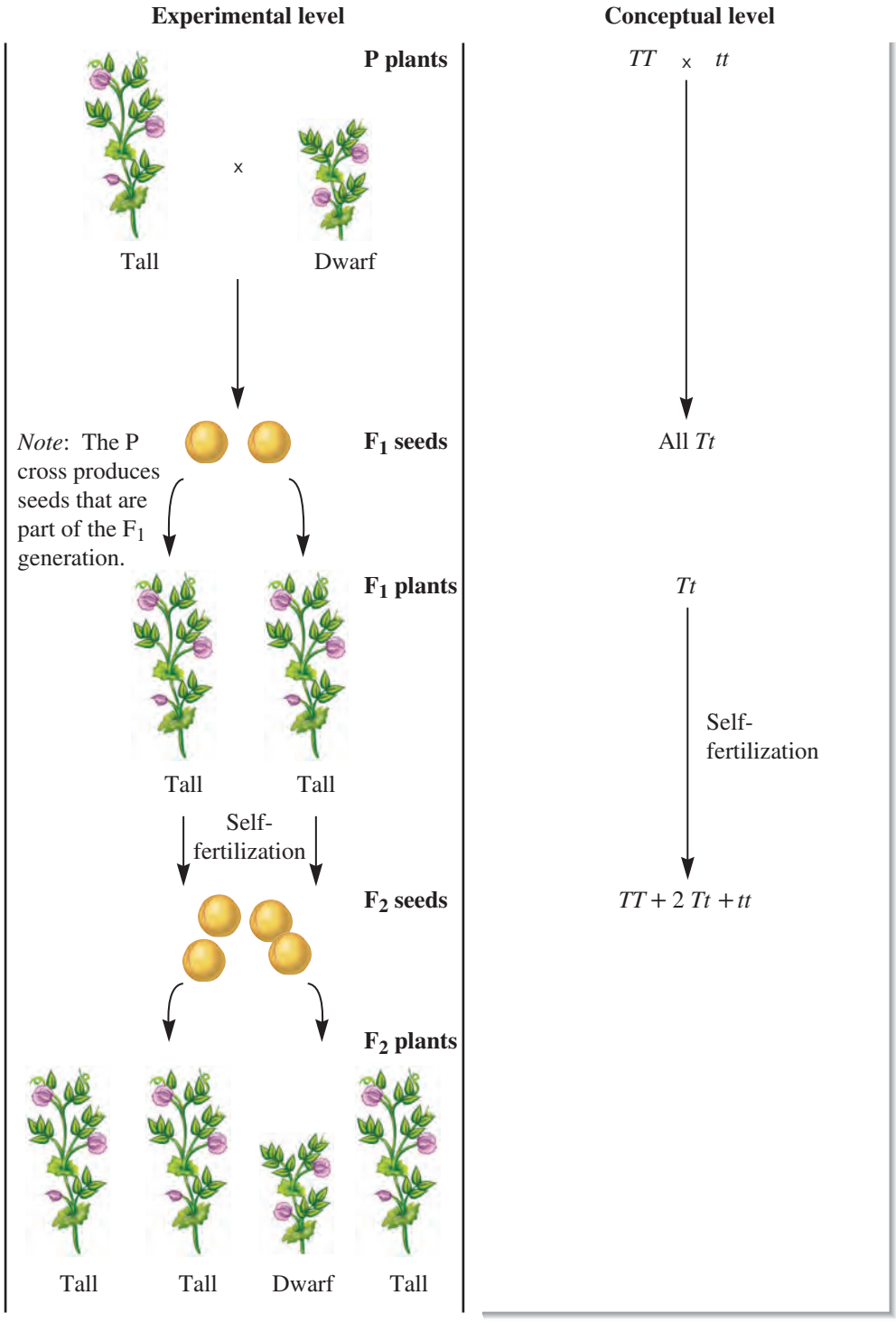
1. For each of seven characters, Mendel cross-fertilized two different true-breeding strains. Keep in mind that each cross involved two plants that differed in regard to only one of the seven characters studied. The illustration at the right shows one cross between a tall and dwarf plant. This is called a P (parental) cross.

2. Collect the F_1 generation seeds. The following spring, plant the seeds and allow the plants to grow. These are the plants of the F_1 generation.

3. Allow the F_1 generation plants to self-fertilize. This produces seeds that are part of the F_2 generation.

4. Collect the F_2 generation seeds and plant them the following spring to obtain the F_2 generation plants.

5. Analyze the traits found in each generation.



THE DATA

P cross	*F_1 generation*	*F_2 generation*	*Ratio of traits in F_2 generation*
Tall × dwarf height	All tall	787 tall, 277 dwarf	2.84:1
Purple × white flowers	All purple	705 purple, 224 white	3.15:1
Axial × terminal flowers	All axial	651 axial, 207 terminal	3.14:1
Yellow × green seeds	All yellow	6,022 yellow, 2,001 green	3.01:1
Round × wrinkled seeds	All round	5,474 round, 1,850 wrinkled	2.96:1
Green × yellow pods	All green	428 green, 152 yellow	2.82:1
Smooth × constricted pods	All smooth	882 smooth, 299 constricted	2.95:1
Total	All dominant	14,949 dominant, 5,010 recessive	2.98:1

Source: Mendel, Gregor, "Versuche über Plflanzenhybriden, Verhandlungen des naturforschenden Vereines in Brünn, Bd. IV für das Jahr 1865," *Abhandlungen,* 1866, 3–47.

INTERPRETING THE DATA

The data in the table are the results of producing an F_1 generation via cross-fertilization and an F_2 generation via self-fertilization of the F_1 plants. A quantitative analysis of these data allowed Mendel to propose three important ideas:

1. Mendel's data argued strongly against a blending mechanism of heredity. In all seven cases, the F_1 generation displayed traits that were distinctly like one of the two parents rather than traits intermediate in character. His first proposal was that one variant for a particular character is **dominant** to another variant. For example, the variant of green pods is dominant to that of yellow pods. The term **recessive** is used to describe a variant that is masked by the presence of a dominant trait but reappears in subsequent generations. Yellow pods and dwarf height are examples of recessive variants. They can also be referred to as recessive traits.
2. When a true-breeding plant with a dominant trait was crossed to a true-breeding plant with a recessive trait, the dominant trait was always observed in the F_1 generation. In the F_2 generation, most offspring displayed the dominant trait, but some showed the recessive trait. How did Mendel explain this observation? Because the recessive trait appeared in the F_2 generation, he made a second proposal—the genetic determinants of traits are passed along as "unit factors" from generation to generation. His data were consistent with a **particulate theory of inheritance,** in which the genetic determinants that govern traits are inherited as discrete units that remain unchanged as they are passed from parent to offspring. Mendel referred to the genetic determinants as unit factors, but we now call them genes (from the Greek, *genesis*, meaning "birth," or, *genos*, meaning "origin").
3. When Mendel compared the numbers of dominant and recessive traits in the F_2 generation, he noticed a recurring pattern. Within experimental variation, he always observed approximately a 3:1 ratio between the dominant and the recessive trait. As described next, this quantitative approach allowed him to make a third proposal—genes **segregate** from each other during the process that gives rise to gametes.

Mendel's 3:1 Ratio Is Consistent with the Law of Segregation

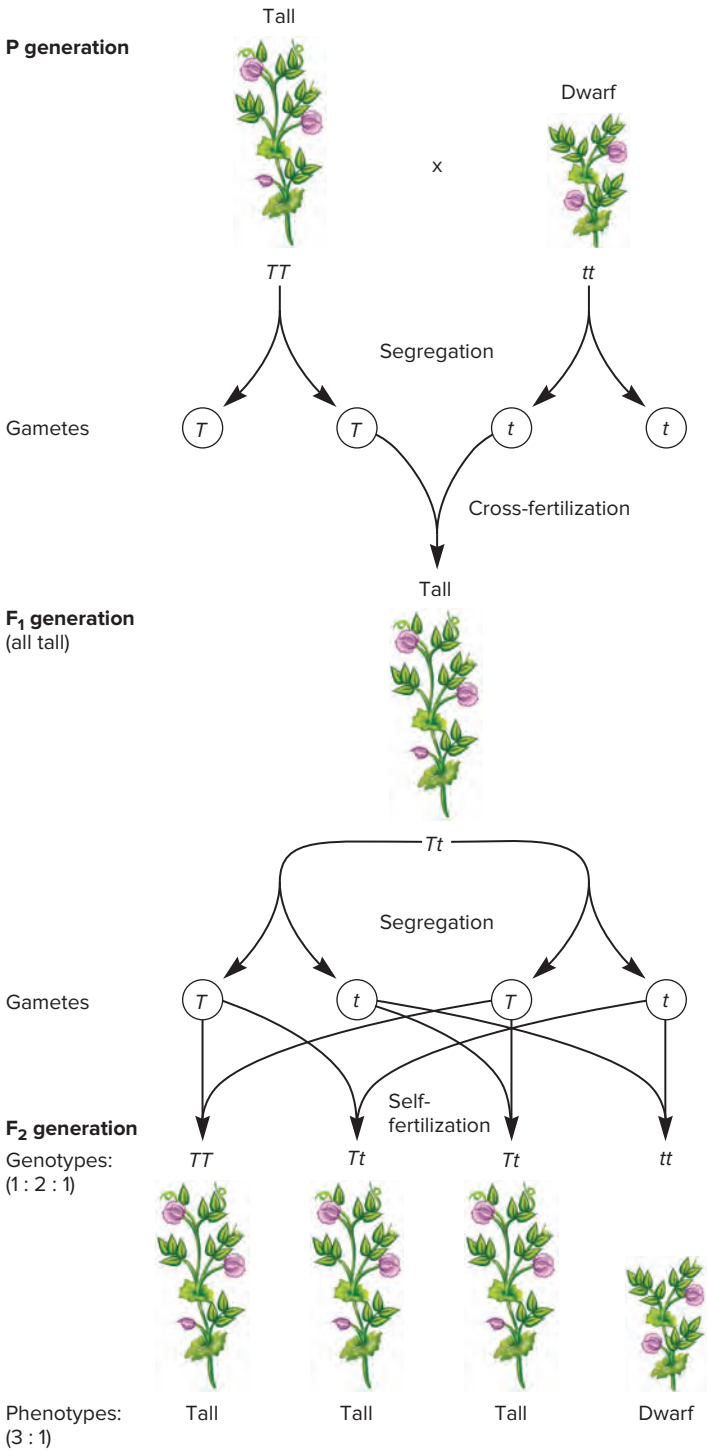
Mendel's research was aimed at understanding the laws that govern the inheritance of traits. At that time, scientists did not understand the molecular composition of the genetic material or its mode of transmission during gamete formation and fertilization. We now know that the genetic material is composed of deoxyribonucleic acid (DNA), a component of chromosomes. Each chromosome contains hundreds to thousands of shorter segments that function as genes—a term that was originally coined by Danish botanist Wilhelm Johannsen in 1909. A **gene** is defined as a unit of heredity that may influence the outcome of a trait in an organism. Each of the seven pea plant characters that Mendel studied is influenced by a different gene.

Most eukaryotic species, such as pea plants and humans, have their genetic material organized into pairs of chromosomes, as discussed in Chapter 3. For this reason, eukaryotes have two copies of most genes. These copies may be the same or they may differ. The term **allele** (from the Latin *alius* meaning "other") refers to an alternative form of a particular gene. For example, the height gene in pea plants is found as a tall allele and a dwarf allele. With this modern knowledge, the results shown in Figure 2.5 are consistent with the idea that each parent transmits only one copy of each gene (i.e., one allele) to each offspring. Using modern terminology, **Mendel's law of segregation** states the following:

> *The two copies of a gene segregate (or separate) from each other during the process that gives rise to gametes.*

Therefore, only one copy of each gene is found in a gamete. At fertilization, two gametes combine randomly, potentially producing different allelic combinations.

Let's use Mendel's cross of tall and dwarf pea plants to illustrate how alleles are passed from parents to offspring (**Figure 2.6**). The letters *T* and *t* are used to represent the alleles of the gene that determines plant height. By convention, the uppercase letter represents the dominant allele (*T* for tall height, in this case), and the recessive allele is represented by the same letter in lowercase



INTERACTIVE EXERCISE

FIGURE 2.6 Mendel's law of segregation. This illustration shows a cross between a true-breeding tall plant and a true-breeding dwarf plant and the subsequent segregation of the tall (*T*) and dwarf (*t*) alleles in the F_1 and F_2 generations.

CONCEPT CHECK: With regard to the *T* and *t* alleles of pea plants, explain what segregation means.

(*t*, for dwarf height). For the P cross, both parents are true-breeding plants. Therefore, each one has identical copies of the height gene. When an individual has two identical copies of a gene, the individual is said to be **homozygous** with respect to that gene. (The prefix *homo-* means "like," and the suffix *-zygo* means "pair.") In the P cross, the tall plant is homozygous for the tall allele *T*, and the dwarf plant is homozygous for the dwarf allele *t*. In contrast, the F_1 generation is **heterozygous,** with the genotype *Tt*, because every individual carries one copy of the tall allele and one copy of the dwarf allele. A heterozygous individual carries different alleles of a gene. (The prefix *hetero-* means "different.")

The term **genotype** refers to the genetic composition of an individual. *TT* and *tt* are the genotypes of the P generation in this experiment. The term **phenotype** refers to observable traits of an organism. In the P generation, the plants exhibit a phenotype that is either tall or dwarf. In some cases, plants have different genotypes yet the same phenotype. For example, both *TT* and *Tt* plants are tall.

The law of segregation predicts that the phenotypes of the F_2 generation will be tall and dwarf in a ratio of 3:1 (see Figure 2.6). The parents of the F_2 generation are heterozygous. Due to segregation, their gametes can carry either a *T* allele or a *t* allele, but not both. Following self-fertilization, *TT*, *Tt*, and *tt* are the possible genotypes of the F_2 generation. By randomly combining these alleles, the genotypes are produced in a 1:2:1 ratio. Because *TT* and *Tt* both produce tall phenotypes, a 3:1 phenotypic ratio is observed in the F_2 generation.

A Punnett Square Can Be Used to Predict the Outcome of Crosses and Self-Fertilization Experiments

An easy way to predict the outcome of simple genetic crosses and self-fertilization experiments is to use a **Punnett square,** a method originally proposed by British geneticist Reginald Punnett. To construct a Punnett square, you must know the genotypes of the parents. With this information, the Punnett square enables you to predict the types of offspring the parents are expected to produce and in what proportions.

Step 1. *Write down the genotypes of both parents. (In a self-fertilization experiment, a single parent provides the sperm and egg cells.)* Let's consider an example in which a heterozygous tall plant is crossed to another heterozygous tall plant. The plant providing the sperm (via pollen) is the male parent and the plant providing the eggs is the female parent.

Male parent: *Tt*

Female parent: *Tt*

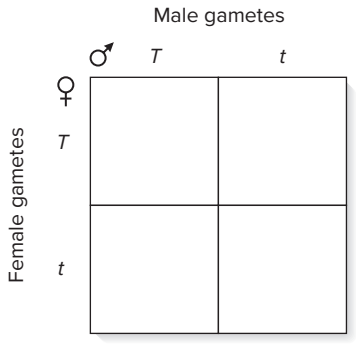
Step 2. *Write down the possible gametes that each parent can make.* Remember that the law of segregation tells us that a gamete carries only one copy of each gene.

Male gametes: *T* or *t*

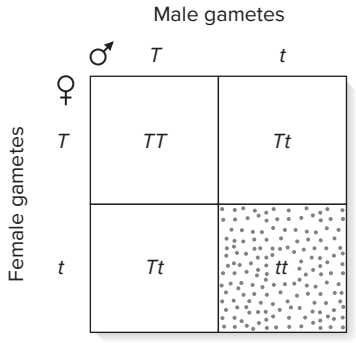
Female gametes: *T* or *t*

Step 3. *Create an empty Punnett square.* In the examples shown in this textbook, the number of columns equals the number of male gametes, and the number of rows equals the number of female gametes. Our example has two rows and

two columns. Place the male gametes across the top of the Punnett square and the female gametes along the side.



Step 4. *Fill in the possible genotypes of the offspring by combining the alleles of the gametes in the empty boxes.*



Step 5. *Determine the relative proportions of genotypes and phenotypes of the offspring.* The genotypes are obtained directly from the Punnett square. They are contained within the boxes that have been filled in. In this example, the genotypes are *TT*, *Tt*, and *tt* in a 1:2:1 ratio. To determine the phenotypes, you must know the dominant/recessive relationship between the alleles. For plant height, *T* (tall) is dominant to *t* (dwarf). The genotypes *TT* and *Tt* are tall, whereas the genotype *tt* is dwarf. Therefore, our Punnett square shows us that the ratio of phenotypes is 3:1, or 3 tall plants : 1 dwarf plant.

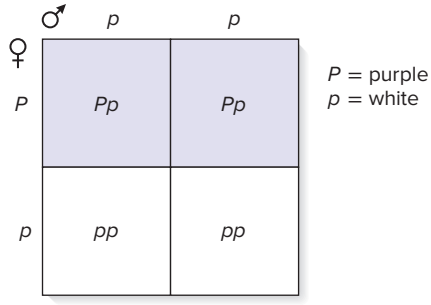
GENETIC TIPS

THE QUESTION: One pea plant that is heterozygous with regard to flower color (purple is dominant to white) is crossed to a plant with white flowers. What are the predicted outcomes of genotypes and phenotypes for the offspring?

TOPIC: ***What topic in genetics does this question address?*** The topic is Mendelian inheritance. More specifically, the question is about a single-factor cross.

INFORMATION: ***What information do you know based on the question and your understanding of the topic?*** From the question, you know that one plant is heterozygous for flower color. If *P* is the purple allele and *p* the white allele, the genotype of this plant is *Pp*. The other plant exhibits the recessive phenotype, so its genotype must be *pp*. From your understanding of the topic, you may remember that alleles segregate during gamete formation and parents each pass one allele to their offspring; the two alleles combine at fertilization.

PROBLEM-SOLVING STRATEGY: ***Predict the outcome.*** One strategy to solve this type of problem is to use a Punnett square to predict the outcome of the cross. The Punnett square is shown next.



ANSWER: The ratio of offspring genotypes is 1 *Pp* : 1 *pp*. The ratio of the phenotypes is 1 purple : 1 white.

2.2 COMPREHENSION QUESTIONS

1. A pea plant is *Tt*. Which of the following statements is correct?
   a. Its genotype is *Tt,* and its phenotype is dwarf.
   b. Its phenotype is *Tt,* and its genotype is dwarf.
   c. Its genotype is *Tt,* and its phenotype is tall.
   d. Its phenotype is *Tt,* and its genotype is tall.
2. A *Tt* pea plant is crossed to a *tt* plant. What is the expected ratio of phenotypes for offspring from this cross?
   a. 3 tall : 1 dwarf
   b. 1 tall : 1 dwarf
   c. 1 tall : 3 dwarf
   d. 2 tall : 1 dwarf

2.3 LAW OF INDEPENDENT ASSORTMENT

Learning Outcomes:

1. Analyze Mendel's experiments involving two-factor crosses.
2. State Mendel's law of independent assortment.
3. Predict the outcome of a two-factor cross using a Punnett square.
4. Define *loss-of-function allele,* and explain why such alleles are useful to study.

Though his experiments as described in Figure 2.5 revealed important ideas regarding a hereditary law, Mendel realized that additional insights might be uncovered if he conducted

more complicated experiments. In this section, we will examine how he conducted crosses in which he simultaneously investigated the pattern of inheritance for two different characters. In other words, he carried out **two-factor crosses** in which he followed the inheritance of two different characters within the same groups of individuals. These experiments led to the formulation of a second law—the law of independent assortment.

EXPERIMENT 2B

Mendel Also Analyzed Crosses Involving Two Different Characters

To illustrate Mendel's work, we will consider an experiment in which one of the characters was seed shape, found in round or wrinkled variants, and the second character was seed color, which existed as yellow and green variants. In this two-factor cross, Mendel followed the inheritance pattern for both characters simultaneously.

What results are possible from a two-factor cross? One possibility is that the genetic determinants for these two different characters are always linked to each other and inherited as a single unit (**Figure 2.7a**). If this were the case, the F_1 offspring could produce only two types of gametes, *RY* and *ry*. A second possibility is they are not linked and can assort themselves independently into gametes (**Figure 2.7b**). If independent assortment occurred, an F_1 offspring could produce four types of gametes, *RY*, *Ry*, *rY*, and *ry*. Keep in mind that the results of Figure 2.5 have already shown us that a gamete carries only one allele for each gene.

The experimental protocol for this two-factor cross is shown in **Figure 2.8**. Mendel began with two different strains of true-breeding pea plants that were different in seed shape and seed color. One plant was produced from seeds that were round and yellow; the other plant from seeds that were wrinkled and green. When these plants were crossed, the seeds, which contain the plant embryo, are considered part of the F_1 generation. As expected, the data revealed that the F_1 seeds displayed a phenotype of round and yellow. This phenotype was observed because round and yellow are dominant traits. It is the F_2 generation that supports the independent assortment model and refutes the linkage model.

THE HYPOTHESES

The inheritance pattern for two different characters follows one or more quantitative natural laws. Two possible hypotheses are described in Figure 2.7.

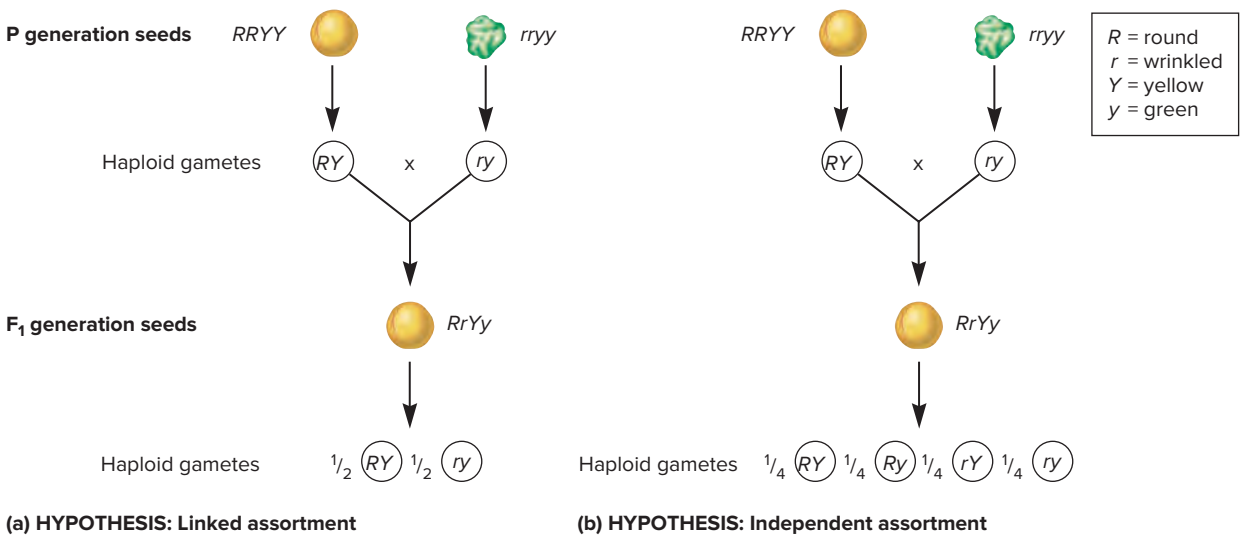


FIGURE 2.7 Two hypotheses to explain how two different genes assort during gamete formation. **(a)** According to the hypothesis of linked assortment, the two genes always stay associated with each other. **(b)** In contrast, the independent assortment hypothesis proposes that the two different genes randomly segregate into haploid cells.

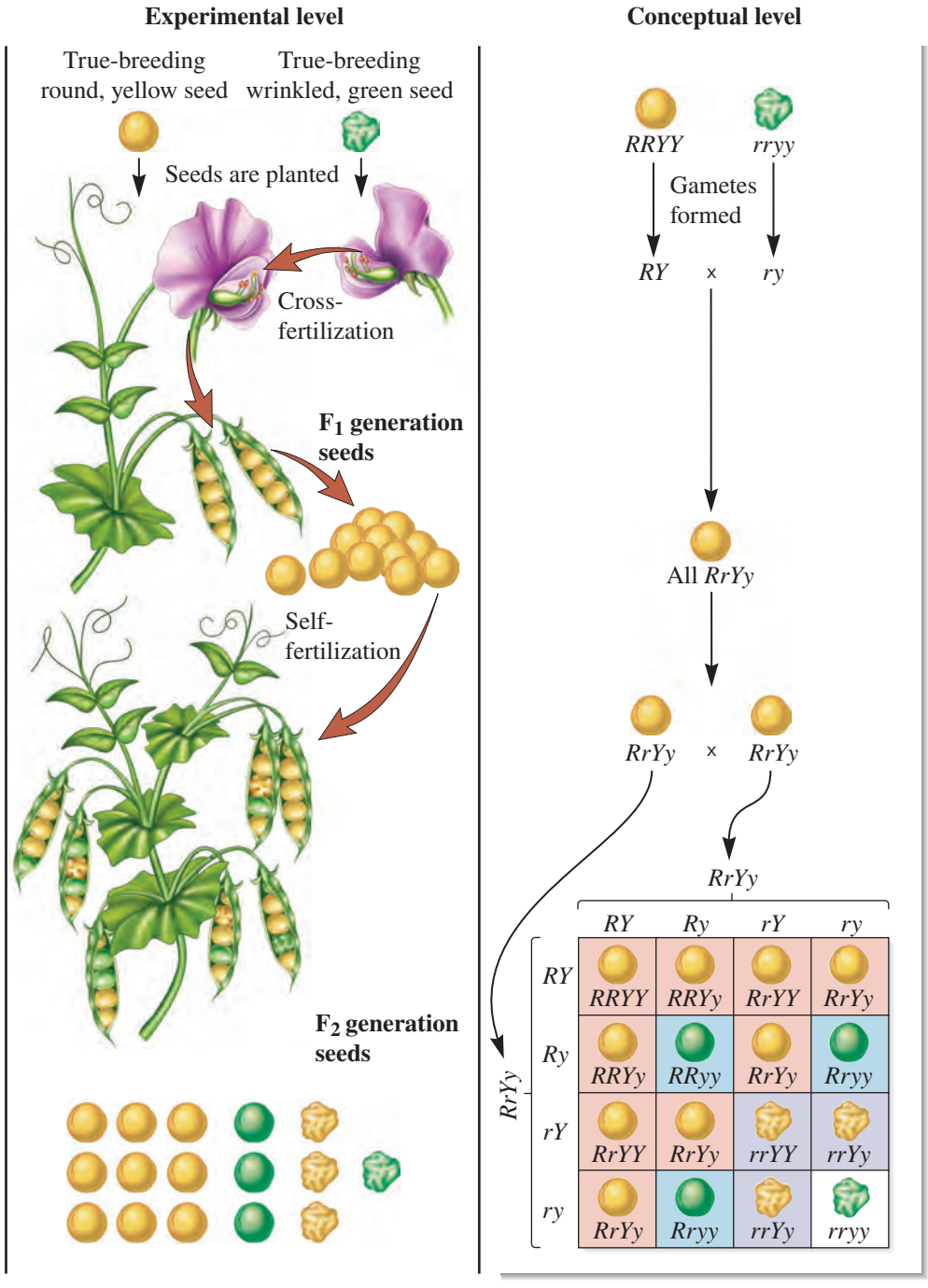
CONCEPT CHECK: According to the hypothesis of linked assortment shown here, what is linked? Are two different genes linked, or are two different alleles of the same gene linked, or both?

TESTING THE HYPOTHESES

FIGURE 2.8 Mendel's analysis of two-factor crosses.

Starting material: In this experiment, Mendel began with two types of true-breeding strains of pea plants that were different with regard to two characters. One strain produced round, yellow seeds (*RRYY*); the other strain produced wrinkled, green seeds (*rryy*).

1. Cross the two true-breeding strains to each other. This produces F_1 generation seeds.
2. Collect many seeds and record their phenotype.
3. F_1 seeds are planted and grown, and the F_1 plants are allowed to self-fertilize. This produces seeds that are part of the F_2 generation.
4. Analyze the characteristics found in the F_2 generation seeds.



THE DATA

P cross	F_1 generation	F_2 generation
Round, yellow seeds × wrinkled, green seeds	All round, yellow seeds	315 round, yellow seeds 108 round, green seeds 101 wrinkled, yellow seeds 32 wrinkled, green seeds

INTERPRETING THE DATA

In addition to seeds that were like those of the parental generation, the F_2 generation also had seeds that were round and green and seeds that were wrinkled and yellow. These two categories of F_2 seeds are called **nonparental** because these combinations of traits were not found in the true-breeding plants of the parental generation.

The occurrence of nonparental variants contradicts the linked-assortment hypothesis (see Figure 2.7a). According to that model, the *R* and *Y* alleles should be linked together and so should the *r* and *y* alleles. If this were the case, the F_1 plants could only produce gametes that were *RY* or *ry*. These would combine to produce *RRYY* (round, yellow), *RrYy* (round, yellow), or *rryy* (wrinkled, green) seeds in a 1:2:1 ratio. Nonparental seeds could, therefore, not be produced. However, Mendel did not obtain this result. Instead, he observed a phenotypic ratio of 9:3:3:1 in the F_2 generation.

Mendel's Two-Factor Crosses Led to the Law of Independent Assortment

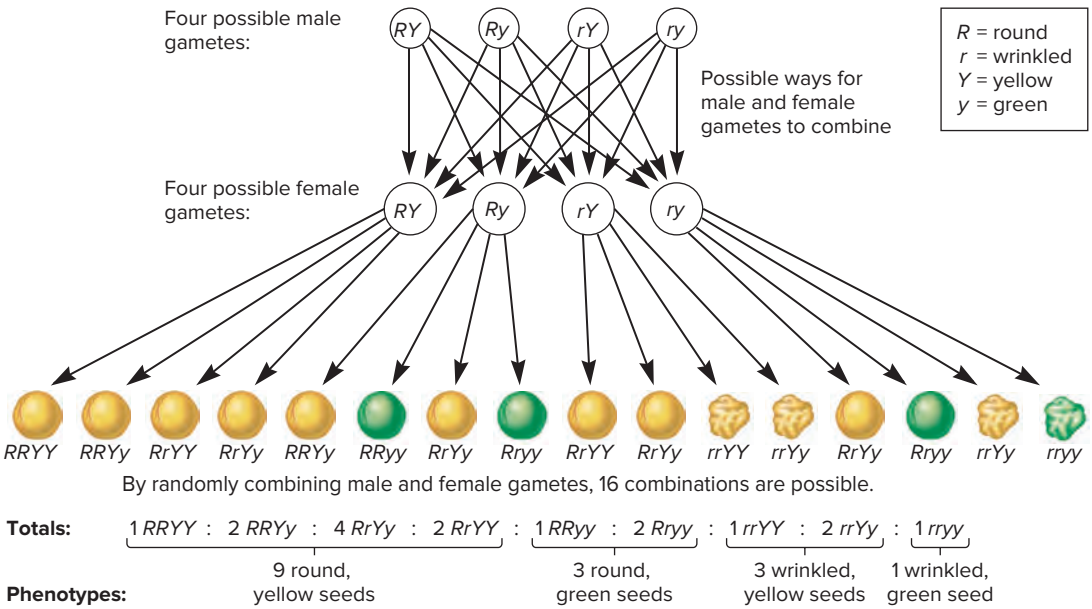
Mendel's results from many two-factor crosses rejected the linked-assortment hypothesis and, instead, supported the hypothesis that different characters assort themselves independently. Using modern terminology, **Mendel's law of independent assortment** states the following:

> *Two different genes will randomly assort their alleles during the process that gives rise to gametes.*

In other words, the allele for one gene will be found within a resulting gamete independently of whether the allele for a different gene is found in the same gamete. In the example given in Figure 2.8, the round and wrinkled alleles are assorted into haploid gametes independently of the yellow and green alleles. Therefore, a heterozygous *RrYy* parent can produce four different gametes—*RY*, *Ry*, *rY*, and *ry*—in equal proportions.

In an F_1 self-fertilization experiment, any two gametes can combine randomly during fertilization. This allows for 4^2, or 16, possible offspring, although some offspring will be genetically identical to each other. As shown in **Figure 2.9**, these 16 possible combinations result in seeds with the following phenotypes: 9 round, yellow; 3 round, green; 3 wrinkled, yellow; and 1 wrinkled, green. This 9:3:3:1 ratio is the expected outcome when a plant that is heterozygous for both genes is allowed to self-fertilize. Mendel was clever enough to realize that the data for his two-factor experiments were close to a 9:3:3:1 ratio. In Figure 2.8, for example, his F_1 generation produced F_2 seeds with the following characteristics: 315 round, yellow seeds; 108 round, green seeds; 101 wrinkled, yellow seeds; and 32 wrinkled, green seeds. If we divide each of these numbers by 32 (the number of plants with wrinkled, green seeds), the phenotypic ratio of the F_2 generation is 9.8 : 3.4 : 3.2 : 1.0. Within experimental error, Mendel's data approximated the predicted 9:3:3:1 ratio for the F_2 generation.

The law of independent assortment held true for two-factor crosses involving all of the characters that Mendel studied in pea plants. However, in other cases, the inheritance pattern of two different genes is consistent with the linked-assortment hypothesis described in Figure 2.7a. In Chapter 6, we will examine the inheritance of genes that are linked because they are close to each other



INTERACTIVE EXERCISE

FIGURE 2.9 Mendel's law of independent assortment.

Genes→Traits This self-fertilization experiment involves a parent that is heterozygous for seed shape and seed color (*RrYy*). Four types of male gametes are possible: *RY, Ry, rY,* and *ry*. Likewise, four types of female gametes are possible: *RY, Ry, rY,* and *ry*. These four types of gametes are the result of the independent assortment of the seed shape and seed color alleles relative to each other. During fertilization, any one of the four types of male gametes can unite with any one of the four types of female gametes, resulting in 16 combinations.

CONCEPT CHECK: Why does independent assortment promote genetic variation?

within the same chromosome. As we will see, linked genes do not assort independently.

An important consequence of the law of independent assortment is that a single individual can produce a vast array of genetically different gametes. As mentioned in Chapter 1, diploid species have pairs of homologous chromosomes, which may differ with respect to the alleles they carry. When an offspring receives a combination of alleles that differs from those in the parental generation, this phenomenon is termed **genetic recombination.** One mechanism that accounts for genetic recombination is independent assortment. A second mechanism, discussed in Chapter 6, is crossing over, which can reassort alleles that happen to be linked along the same chromosome.

The phenomenon of independent assortment is rooted in the random pattern in which the pairs of chromosomes assort themselves during the process of meiosis, a topic addressed in Chapter 3. When two different genes are found on different chromosomes, they randomly assort into haploid cells (look ahead to Figure 3.16). If a species contains a large number of chromosomes, this creates the potential for an enormous amount of genetic diversity. For example, human cells contain 23 pairs of chromosomes. These pairs separate and randomly assort into gametes during meiosis. The number of different gametes an individual can make equals 2^n, where n is the number of pairs of chromosomes. Therefore, a human can make 2^{23}, or over 8 million, possible gametes, due to independent assortment. The capacity to make so many genetically different gametes enables a species to produce a great diversity of individuals with different combinations of traits. This variety of phenotypes allows environmental factors to select for those combinations of traits that favor reproductive success.

A Punnett Square Can Be Used to Solve Independent Assortment Problems

As already depicted in Figure 2.8, we can make a Punnett square to predict the outcome of experiments involving two or more genes that assort independently. Let's see how such a Punnett square is made by considering a cross between two plants that are heterozygous for height and seed color (**Figure 2.10**). This cross is *TtYy* × *TtYy*. When we construct a Punnett square for this cross, we must keep in mind that each gamete has a single allele for each of two genes. In this example, the four possible gametes from each parent are

TY, *Ty*, *tY*, and *ty*

In this two-factor cross, we need to make a Punnett square containing 16 boxes. The phenotypes of the resulting offspring are predicted to occur in a ratio of 9:3:3:1.

The Multiplication and Forked-Line Methods Can Also Be Used to Solve Independent Assortment Problems

In crosses involving three or more genes, the construction of a single large Punnett square becomes very unwieldy. For example, in a three-factor cross between two pea plants that are *Tt Rr Yy*, each parent can make 2^3, or 8, possible gametes. Therefore, the Punnett

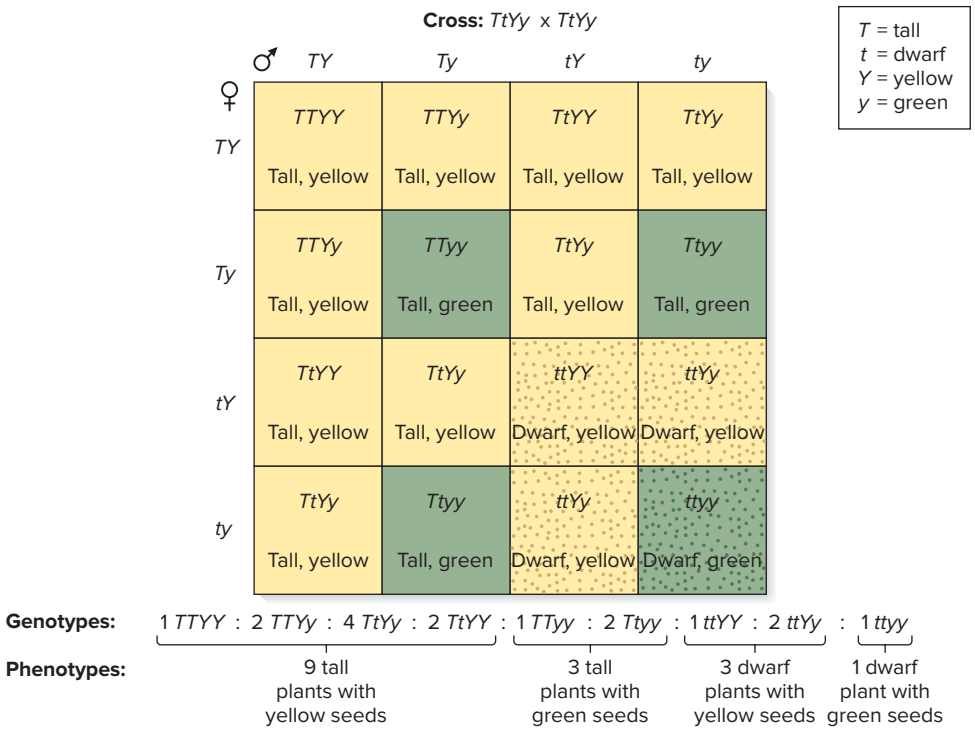


FIGURE 2.10 A Punnett square for a two-factor cross. The Punnett square shown here involves a cross between two pea plants that are heterozygous for height and seed color. The cross is *TtYy* × *TtYy*.

CONCEPT CHECK: If a parent plant is *Ttyy,* how many different types of gametes can it make?