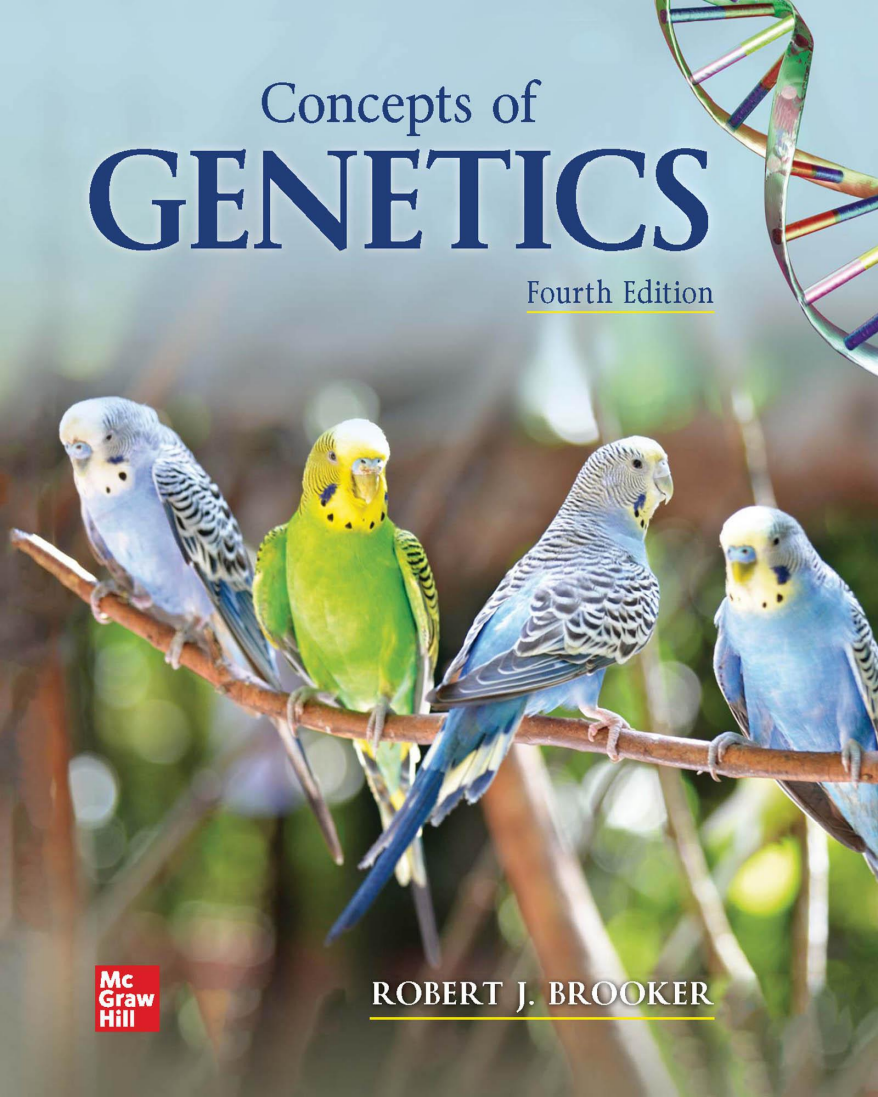
Concepts of GENETICS

Fourth Edition



ROBERT J. BROOKER

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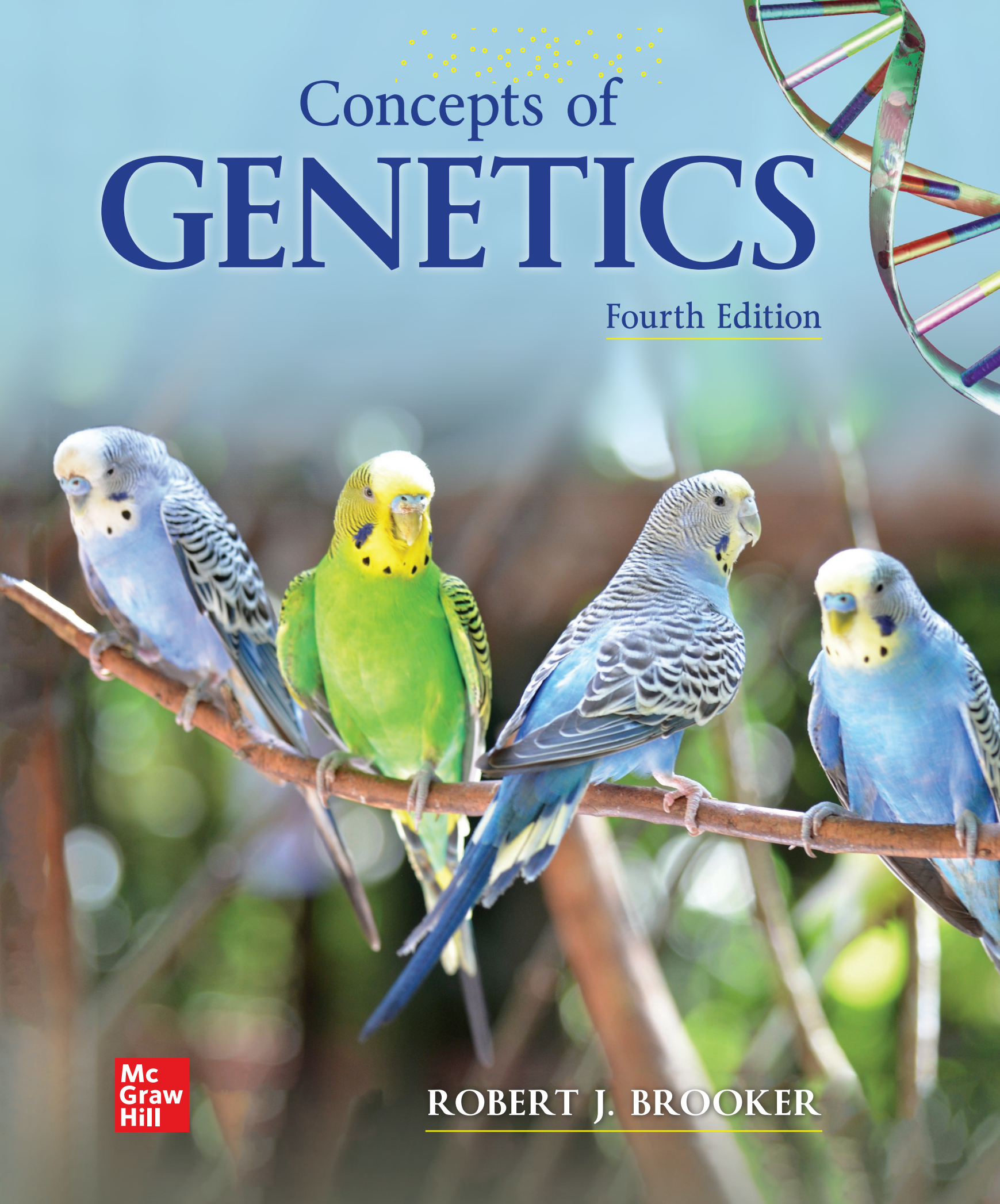

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CONCEPTS OF GENETICS, FOURTH EDITION

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ABOUT THE AUTHOR

Robert J. Brooker is a professor in the Department of Genetics, Cell Biology, and Development and the Department of Biology Teaching and Learning at the University of Minnesota–Minneapolis. He received his B.A. in biology from Wittenberg University in 1978 and his Ph.D. in genetics from Yale University in 1983. At Harvard, he conducted postdoctoral studies on lactose permease, which is the product of the *lacY* gene of the *lac* operon. He continued his work on transporters at the University of Minnesota. Dr. Brooker's laboratory has also investigated the structure, function, and regulation of manganese and iron transporters found in bacteria and *C. elegans*. At the University of Minnesota, he teaches undergraduate courses in biology and genetics, and he has taught online classes for over a decade.

Robert Brooker

DEDICATION

To my wife, Deborah, and our children,
Daniel, Nathan, and Sarah

BRIEF CONTENTS

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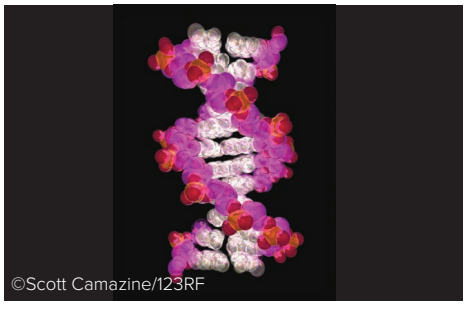
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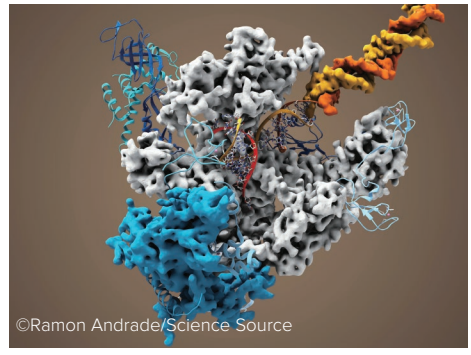
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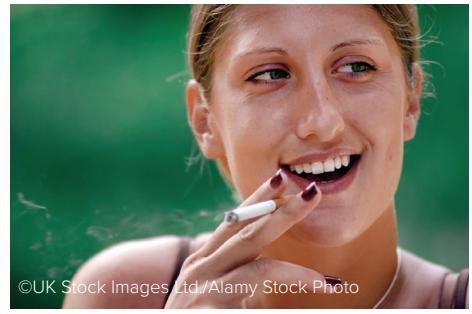
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PREFACE

Concepts of Genetics, 4e, is intended for students who want to gain a conceptual grasp of the various fields of genetics. The content reflects current trends in genetics, and the pedagogy is based on educational research. As an author, researcher, and teacher, I want to provide a textbook that gets students actively involved in learning genetics. I have worked with a talented team of editors, illustrators, and media specialists who have helped me make the fourth edition of *Concepts of Genetics* a fun learning tool.

An effective textbook should strive to accomplish four goals:

1. It should help students improve their critical-thinking skills. As described shortly, this goal is a key emphasis of this textbook.
2. The content needs to be comprehensive, accurate, and up-to-date.
3. Students should be exposed to the techniques and methodologies they will need to become successful in their fields of work.
4. The pedagogy should inspire students.

The hard work that has gone into the fourth edition of *Concepts of Genetics* has been aimed at achieving these goals. Instead of being a collection of "facts and figures," *Concepts of Genetics*, Fourth Edition, by Robert Brooker, is intended to be an engaging and motivating textbook in which critical-thinking skills are strongly emphasized. We welcome your feedback so we can make future editions even better!

CRITICAL-THINKING SKILLS

Critical thinking is the mental process of actively and skillfully conceptualizing, applying, analyzing, synthesizing, and/or evaluating information gathered from, or generated by, various experiences including observation, experimentation, reflection, and/or communication. It provides a guide to belief and action. A person who is skillful at critical thinking can draw reasonable conclusions from a collection of information, and discriminate between useful and less useful details to solve problems and to make decisions. If a primary goal for your students is to improve their critical-thinking skills, *Concepts of Genetics,* 4e, is an excellent choice. Here are eleven good reasons why:

1. **Each section of every chapter begins with a set of learning outcomes.** These outcomes help students understand what they should be learning and what critical-thinking skills they should be mastering. Similarly, each section ends with a summary of the key concepts.

LEARNING OUTCOMES

Each section begins with one or more Learning Outcomes. These allow a student to appreciate the skills and knowledge they will gain if they master the material.

23.4 GENETIC DRIFT

Learning Outcomes:

1. Define *genetic drift*.
2. Explain how population size affects genetic drift, and calculate the probabilities of the outcomes of this process.
3. Compare and contrast the bottleneck effect and the founder effect.

REVIEWING THE KEY CONCEPTS

These bulleted lists at the end of each section help students identify important concepts. Students should understand these concepts before moving on to the next section.

23.4 REVIEWING THE KEY CONCEPTS

- Genetic drift involves changes in allele frequencies due to random fluctuations. Over the long run, it often results in allele fixation or loss. The effect of genetic drift occurs more rapidly in small populations (see Figure 23.14).
- Two mechanisms that can influence genetic drift are the bottleneck effect and the founder effect (see Figure 23.15).

2. **A feature called *Genetic TIPS* provides a consistent approach to help students solve problems in genetics.** Starting with Chapter 2, typically 3 to 6 of these TIPS are are found in each chapter. They are directly aimed at helping students improve their critical-thinking skills.

GENETIC TIPS

Problem solving is a skill that genetics students need to master. ***Genetic TIPS*** *help students solve problems in genetics. This approach has three components: First, the student is made 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 a* ***P****roblem-Solving* ***S****trategy to tackle the question.* ***More Genetic TIPS*** *are presented at the end of the chapter, allowing for additional practice in strengthening problem-solving skills.*

Genetic TIPS

The Question: A cat is born with two X chromosomes and one Y chromosome. One of the X chromosomes carries the black fur allele and the other carries the orange fur allele. Would you expect this cat to be a male or female? Would it be calico?

Topic: What topic(s) in genetics does this question address?

The topics are sex determination and X-chromosome inactivation.

Information: What information do you know based on the question and your understanding of the topic?

From the question, you know the composition of sex chromosomes in a cat and the fur color alleles carried on the cat's X chromosomes. From your understanding of the topics, you may remember that the Y chromosome determines maleness in mammals and that X-chromosome inactivation occurs and only one X chromosome remains active in somatic cells.

Problem-Solving Strategy: Predict the outcome.

With regard to sex determination, you would predict that the cat is a male because the Y chromosome causes maleness. You would also predict that random X-chromosome inactivation would occur in this cat's somatic cells, because the cells contain two X chromosomes. The cat is heterozygous

3. **In genetics and other fields of biology, a key component of critical thinking is the ability to follow the scientific method.** Many chapters in this textbook have one or two "feature investigations" that are presented according to the scientific method. They begin with a hypothesis (or goal) and then describe the experiment in a series of steps. The data are then presented and discussed, so that students can understand how data analysis allows scientists to reach conclusions based on evidence.

The Genome of Tobacco Mosaic Virus Is Composed of RNA

We now know that bacteria, archaea, protists, fungi, plants, and animals all use DNA as their genetic material. In 1956, Alfred Gierer and Gerhard Schramm isolated RNA from tobacco mosaic virus (TMV), which infects plant cells. When this purified RNA

BACKGROUND OBSERVATIONS

Each experiment begins with a description of the information that led researchers to study a hypothesis-driven or discovery-based problem. Detailed information about the researchers and the experimental challenges they faced help students to understand actual research.

THE HYPOTHESIS OR THE GOAL

The student is given a possible explanation for the observed phenomenon that will be tested or the question researchers were hoping to answer. This section reinforces the scientific method and allows students to experience the process for themselves.

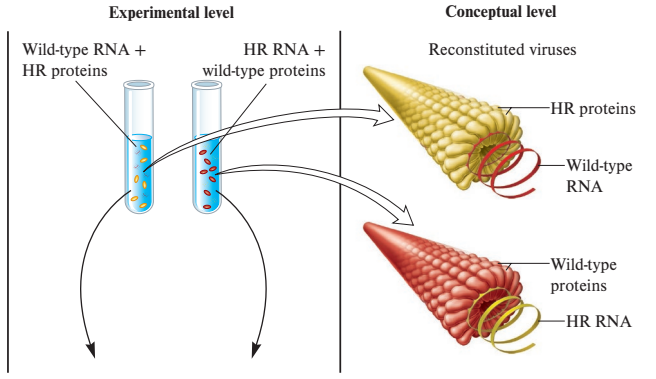
▶ THE HYPOTHESIS

RNA is the genetic material of TMV.

▶ TESTING THE HYPOTHESIS — FIGURE 10.2 **Evidence that RNA is the genetic material of TMV.**

Starting material: Purified preparations of RNA and proteins from wild-type TMV and from the Holmes ribgrass (HR) strain of TMV.

1. Mix together wild-type RNA and HR proteins or HR RNA and wild-type proteins. Allow time for the RNA and proteins to assemble into intact viruses. These are called reconstituted viruses.



TESTING THE HYPOTHESIS OR ACHIEVING THE GOAL

This section illustrates the experimental process, including the actual steps followed by scientists to test their hypothesis or study a question. Science comes alive for students with this detailed look at experimentation.

THE DATA

Actual data from the original research paper help students understand how real-life research results are reported. Each experiment's results are discussed in the context of the larger genetic principle to help students understand the implications and importance of the research.

▶ THE DATA

Composition of Reconstituted Virus Placed on Tobacco Leaves	*Symptoms on Tobacco Leaves*	*Amino Acids Found in Newly Made Viral Proteins Following Infection: Methionine*	*Histidine*
Wild-type RNA and HR protein	Like wild-type TMV	No	No
HR RNA and wild-type protein	Like HR TMV	Yes	Yes

Source: Data adapted from Fraenkel-Conrat, H., & Singer, B. (1957) Virus Reconstitution II. Combination of Protein and Nucleic Acid from Different Strains, *Biochimica et Biophysica Acta*, vol. 24, no.3, 540–548.

▶ INTERPRETING THE DATA

As seen in the data, the outcome of infection depended on the RNA that was found in the reconstituted virus but not the protein. If wild-type RNA was used, the leaves developed symptoms that were typical of wild-type TMV, and the capsid proteins of newly made viruses lacked methionine or histidine. In contrast, if the

INTERPRETING THE DATA

This discussion, which examines whether the experimental data supported or disproved the hypothesis or provided new information to propose a hypothesis, gives students an appreciation for scientific interpretation.

4. **Each section of every chapter ends with multiple-choice questions**. Formative assessment at the end of each section allows students to critically evaluate their mastery of the material before moving on to the next section.

COMPREHENSION QUESTIONS

Multiple choice questions found at the end of each section allow students an opportunity to test their knowledge of key information and concepts. This helps students better identify what they know and don't know before tackling more concepts. Answers are provided at the end of the chapter.

7.3 COMPREHENSION QUESTIONS

Answer the multiple-choice questions based on the following experiment:

P generation: True-breeding flies with red eyes and long wings were crossed to flies with white eyes and miniature wings. All F_1 offspring had red eyes and long wings.

The F_1 female flies were then crossed to males with white eyes and miniature wings. The following results were obtained for the F_2 generation:

129 red eyes, long wings
133 white eyes, miniature wings
71 red eyes, miniature wings
67 white eyes, long wings

1. What is/are the phenotypes of the recombinant offspring of the F_2 generation?
   a. red eyes, long wings
   b. white eyes, miniature wings
   c. red eyes, long wings; and white eyes, miniature wings
   d. red eyes, miniature wings; and white eyes, long wings
2. The recombinant offspring of the F_2 generation were due to crossing over that occurred
   a. during spermatogenesis in the P generation males.
   b. during oogenesis in the P generation females.
   c. during spermatogenesis in the F_1 males.
   d. during oogenesis in the F_1 females.
3. What is the map distance between the two genes for eye color and wing length?
   a. 32.3 mu
   b. 34.5 mu
   c. 16.2 mu
   d. 17.3 mu

5. **Most figures have Concept Check questions so that students can determine if they understand the key points in the figure.** Many of these questions probe students' understanding of genetic mechanisms.

CONCEPT CHECK QUESTIONS

Students can test their knowledge and understanding with Concept Check questions that are associated with the figure legends. These questions often go beyond simple recall of information and ask students to apply or interpret information presented in the illustrations.

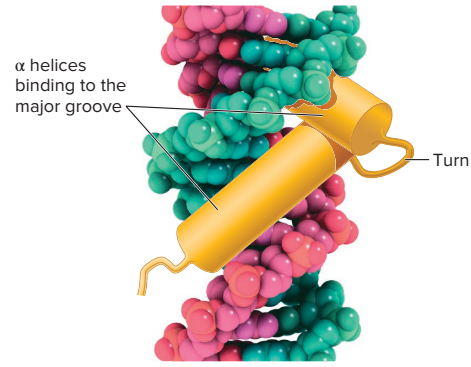
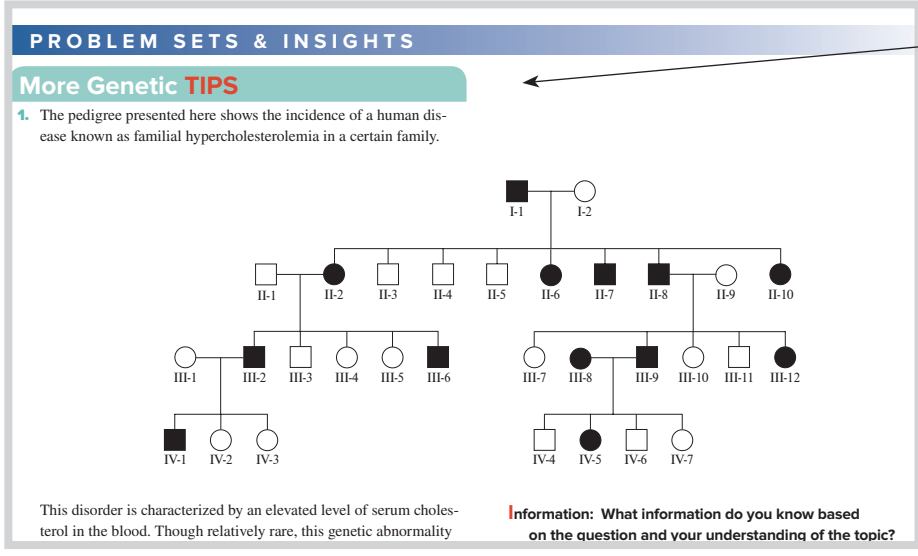


FIGURE 14.6 The binding of σ factor protein to the DNA of a promoter. Part of the protein contains two α helices connected by a turn, termed a helix-turn-helix motif. Two α helices of the protein can fit within the major groove of the DNA. Amino acid side chains within the α helices form hydrogen bonds with the bases in the DNA.

Concept Check: Why is it necessary for portions of the sigma (σ) factor protein to fit into the major groove?

6. ***More Genetic TIPS* and 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 at the end of the textbook so that students can assess their learning.



PROBLEM SETS & INSIGHTS

More Genetic TIPS

1. The pedigree presented here shows the incidence of a human disease known as familial hypercholesterolemia in a certain family.

This disorder is characterized by an elevated level of serum cholesterol in the blood. Though relatively rare, this genetic abnormality

Information: What information do you know based on the question and your understanding of the topic?

MORE GENETIC TIPS

*Like the Genetic TIPS within the chapter, these problems provide more practice in developing problem-solving skills before the students work through more problems unaided. The Genetic TIPS help the student identify the primary question (the **T**opic), evaluate the question based on the student's knowledge of the topic (**I**nformation), and then the student is guided through the solution revealing a **P**roblem-Solving **S**trategy. These provide a reference for when students encounter similar problems later.*

CONCEPTUAL QUESTIONS

These questions test the understanding of basic genetic principles. The student is given many questions with a wide range of difficulty. Some require critical-thinking skills, and some require the student to write coherent answers in an essay form.

Conceptual Questions

C1. The process of binary fission begins with a single mother cell and ends with two daughter cells. Would you expect the mother and daughter cells to be genetically identical? Explain why or why not.

C2. What is a homolog? With regard to genes and alleles, how are homologs similar to and different from each other?

C3. What is a sister chromatid? Are sister chromatids genetically similar or identical? Explain.

C4. With regard to sister chromatids, which phase of mitosis is the organization phase, and which is the separation phase?

C5. A species is diploid with three chromosomes per set. Make a drawing that shows what the chromosomes look like in the G_1 and G_2 phases of the cell cycle.

C6. How does the attachment of kinetochore microtubules to the kinetochore differ in metaphase of meiosis I from metaphase of mitosis? Discuss what you think would happen if a sister chromatid was not attached to a kinetochore microtubule.

C7. For the following events, specify whether each occurs during mitosis, meiosis I, or meiosis II:

a. Separation of conjoined chromatids within a pair of sister chromatids

Application and Experimental Questions

E1. List three advantages of using pea plants as an experimental organism.

E2. Explain the technical difference between a cross-fertilization experiment and a self-fertilization experiment.

E3. How long did it take Mendel to complete the experiment in Figure 3.5?

E4. For all seven characters described in the data of Figure 3.5, Mendel allowed the F_2 plants to self-fertilize. He found that when F_2 plants with recessive traits were allowed to self-fertilize, they always bred true. However, when F_2 plants with dominant traits were crossed, some bred true but others did not. A summary of Mendel's results is shown in the following table.

The Ratio of True-Breeding and Non-True-Breeding Parents of the F_2 Generation

F_2 Parents	True-Breeding	Non-True-Breeding	Ratio
Round	193	372	1:1.93
Yellow	166	353	1:2.13
Gray	36	64	1:1.78
Smooth	29	71	1:2.45
Green	40	60	1:1.5
Axial	33	67	1:2.08
Tall	28	72	1:2.57
TOTAL:	525	1059	1:2.02

When considering the data in this table, keep in mind that they describe whether or not the F_2 generation parents with a domi-

APPLICATION AND EXPERIMENTAL QUESTIONS

These questions test the ability to analyze data, design experiments, or appreciate the relevance of experimental techniques.

QUESTIONS FOR STUDENT DISCUSSION/COLLABORATION

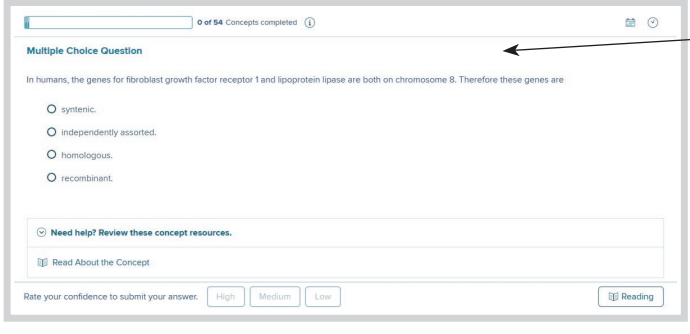
These questions encourage students to consider broad concepts and practical problems. Some questions require a substantial amount of computational activities, which can be worked on as a group.

Questions for Student Discussion/Collaboration

1. Consider this cross of pea plants: *Tt Rr yy Aa* × *Tt rr Yy Aa*, where *T* = tall, *t* = dwarf, *R* = round, *r* = wrinkled, *Y* = yellow, *y* = green, and *A* = axial, *a* = terminal. What is the expected phenotypic outcome of this cross? One group of students should solve this problem by making one big Punnett square, and another group should solve it by making four single-gene Punnett squares and using the multiplication method. Time each other to see who gets done first.
2. A cross was made between two pea plants, *TtAa* and *Ttaa*, where *T* = tall, *t* = dwarf, and *A* = axial, *a* = terminal. What is the probability that the first three offspring that you observe will be tall with axial flowers or dwarf with terminal flowers and the fourth offspring will be tall with axial flowers? Discuss what operation(s) (e.g., product rule or binomial expansion equation) you used and in what order you used them.
3. Consider this four-factor cross: *Tt Rr yy Aa* × *Tt RR Yy aa*, where *T* = tall, *t* = dwarf, *R* = round, *r* = wrinkled, *Y* = yellow, *y* = green, and *A* = axial, *a* = terminal. What is the probability that the first three plants you observe will have round seeds? What is the easiest way to solve this problem?

7. **The textbook material is supported by digital learning tools found in Connect.** Instructors can assign questions and activities in Connect. Assignments due before class time or following an in-class activity help students prepare or review. See the section on Connect on page xviii.

8. **McGraw-Hill SmartBook 2.0 is an adaptive learning tool for textbook navigation.** It is available in Connect and has been shown to strengthen recall and increase retention so that students can move beyond memorizing and apply their learning in meaningful ways.

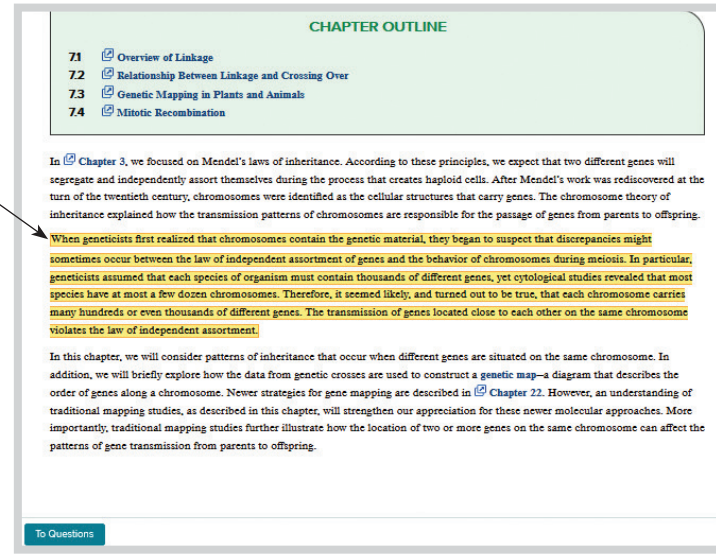


SMARTBOOK 2.0 ADAPTIVE ASSESSMENTS

Assessment based on Learning Outcomes adapts to each student's individual needs, pinpointing knowledge gaps and focusing on concepts that require additional study.

SMARTBOOK 2.0 ADAPTIVE READING

Yellow highlights help students easily identify their assigned content, and the highlighting changes based on student performance.


CHAPTER OUTLINE

7.1 Overview of Linkage
7.2 Relationship Between Linkage and Crossing Over
7.3 Genetic Mapping in Plants and Animals
7.4 Mitotic Recombination

In Chapter 3, we focused on Mendel's laws of inheritance. According to these principles, we expect that two different genes will segregate and independently assort themselves during the process that creates haploid cells. After Mendel's work was rediscovered at the turn of the twentieth century, chromosomes were identified as the cellular structures that carry genes. The chromosome theory of inheritance explained how the transmission patterns of chromosomes are responsible for the passage of genes from parents to offspring.

When geneticists first realized that chromosomes contain the genetic material, they began to suspect that discrepancies might sometimes occur between the law of independent assortment of genes and the behavior of chromosomes during meiosis. In particular, geneticists assumed that each species of organism must contain thousands of different genes, yet cytological studies revealed that most species have at most a few dozen chromosomes. Therefore, it seemed likely, and turned out to be true, that each chromosome carries many hundreds or even thousands of different genes. The transmission of genes located close to each other on the same chromosome violates the law of independent assortment.

In this chapter, we will consider patterns of inheritance that occur when different genes are situated on the same chromosome. In addition, we will briefly explore how the data from genetic crosses are used to construct a genetic map—a diagram that describes the order of genes along a chromosome. Newer strategies for gene mapping are described in Chapter 22. However, an understanding of traditional mapping studies, as described in this chapter, will strengthen our appreciation for these newer molecular approaches. More importantly, traditional mapping studies further illustrate how the location of two or more genes on the same chromosome can affect the patterns of gene transmission from parents to offspring.

To Questions

9. **"Genes → Traits" are descriptions added to figure legends that help students relate the concepts they have learned in molecular genetics with the traits that occur at the level of a whole organism.** The **Genes → Traits** descriptions remind students that molecular and cellular phenomena ultimately lead to the traits that are observed in each species. This insight allows them to apply their knowledge in new ways.

GENES → TRAITS

Because genetics is such a broad discipline ranging from the molecular level to populations, many students have trouble connecting the concepts they learn in molecular genetics with the traits that occur at the level of an organism. To make this connection more meaningful, certain figures have a "Genes→Traits" feature that reminds students that molecular and cellular phenomena ultimately lead to traits observed in organisms.

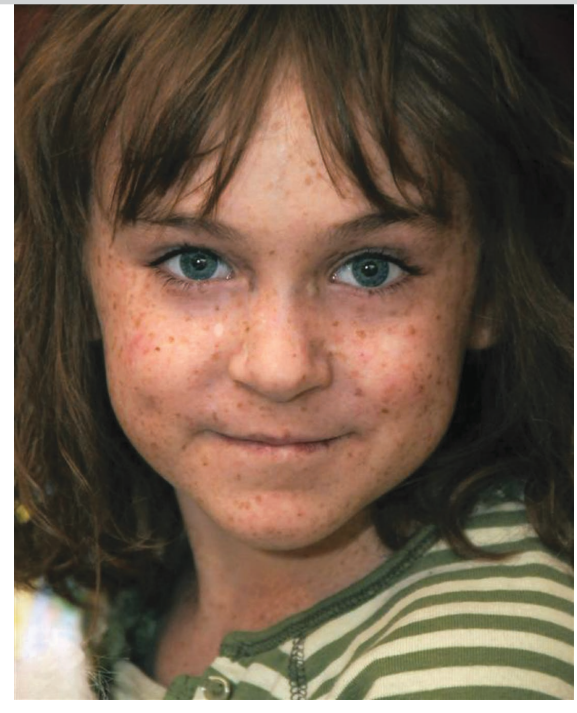


FIGURE 19.18 An individual affected with xeroderma pigmentosum.

Genes → Traits Xeroderma pigmentosum is caused by a defect in one of seven different genes that encode proteins of the NER system. Affected individuals have an increased sensitivity to sunlight because of an inability to repair UV-induced DNA lesions. In addition, they may also have pigmentation abnormalities, many premalignant lesions, and a high predisposition to skin cancer.

Barcroft Media/Getty Images

10. **Working with education specialists, the author has crafted interactive exercises in which the students can make their own choices in problem-solving activities and predict what the outcomes will be.** Many of these exercises are focused on inheritance patterns and human genetic diseases (e.g., see Chapters 5 and 22). In addition, many interactive exercises are found in the molecular chapters. These types of exercises engage students in the learning process. The interactive exercises are found in the eBook, and the corresponding material in the chapter is indicated with an Interactive Exercise icon.

11. **Our media specialists have created over 50 animations for a variety of genetic processes.** These animations were made specifically for this textbook and use the art from the textbook. The animations literally make many of the figures in the textbook "come to life." The animations are found in the eBook and the corresponding material in the chapter is indicated with an Online Animation icon.

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 out 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 flipping the classroom or active learning. For many instructors, the classroom has become more learner-centered rather 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 students understand if their 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 the memorization of facts that are easily forgotten. Students become trained to "think like scientists" and to develop problem-solving skills that enable them to apply scientific reasoning. In other words, active learning fosters critical thinking.

A common concern among instructors who are beginning to try out active learning is that they think they will have to teach their students 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, *Concepts of Genetics*, Fourth Edition, is intended to provide students with a resource that can be effectively used out of the classroom. In particular, when students are expected to learn textbook material on their own, it is imperative that they are given formative assessment on a regular basis so they can gauge whether or not they are mastering the material. Formative assessment is a major feature of this textbook and is bolstered by McGraw-Hill Connect®—a state-of-the art digital assignment and assessment platform. In *Concepts of Genetics,* Fourth Edition, formative assessment is provided in multiple ways:

1. Answers are provided for: (1) the multiple-choice questions throughout the chapter; (2) Concept Check questions following the figure legends; and (3) end-of-chapter questions.
2. As mentioned, McGraw-Hill SmartBook is an adaptive learning tool available in Connect that guides students through the textbook material. As they move through each chapter, they are asked questions to determine if they have mastered the material. If they get a question wrong, they can highlight the material that they need to review.
3. The set of learning outcomes at the beginning of each section provides a road map for students to appreciate what they should be learning. Likewise, the summaries at the end of each section reinforce the key concepts.

HOW WE EVALUATED YOUR NEEDS

ORGANIZATION: MENDEL FIRST VERSUS MOLECULAR FIRST

In surveying many genetics instructors, it became apparent that most people fall into two camps: **Mendel first** versus **Molecular first.** I have taught genetics both ways. As a teaching tool, this textbook has been written with these different teaching strategies in mind. The organization and content lend themselves to various teaching formats.

Chapters 2 through 10 are largely inheritance chapters, whereas Chapters 23 and 24 examine population and quantitative genetics. The bulk of the molecular genetics is found in Chapters 11 through 22, although I have tried to weave a fair amount of molecular genetics into Chapters 2 through 10 as well. The information in Chapters 11 through 22 does not assume that a student has already covered Chapters 2 through 10. Actually, each chapter is written with the perspective that instructors may want to vary the order of their chapters to fit their students' needs.

For those who like to discuss inheritance patterns first, a common strategy would be to cover Chapters 1 through 10 first, and then possibly 23 and 24. (However, many instructors like to cover quantitative and population genetics at the end. Either way works fine.) The more molecular and technical aspects of genetics would then be covered in Chapters 11 through 22. Alternatively, if you like the "Molecular first" approach, you would probably cover Chapter 1, then skip to Chapters 11 through 22, then return to Chapters 2 through 10, and then cover Chapters 23 and 24 at the end of the course. This textbook was written in such a way that either strategy works well.

ACCURACY

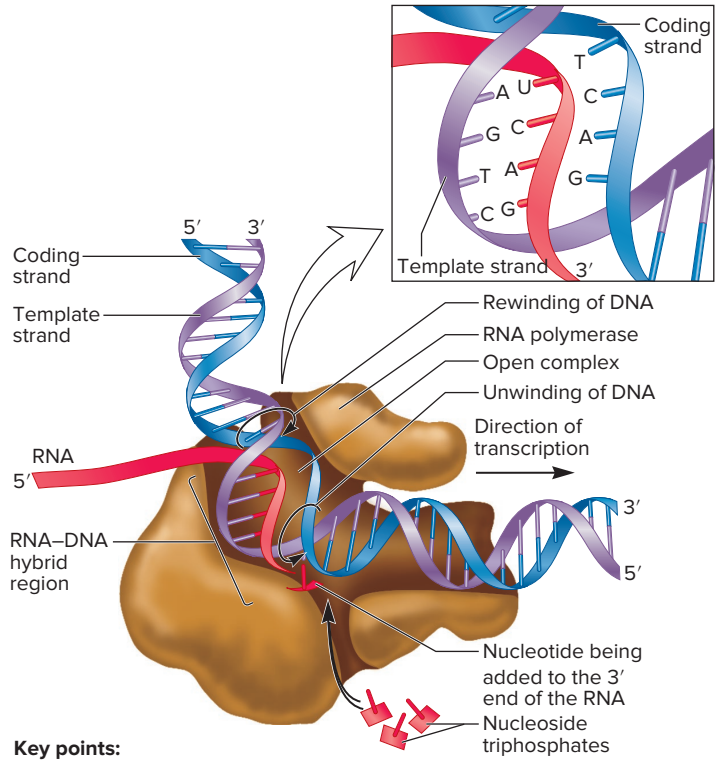
Both the publisher and I acknowledge that inaccuracies can be a source of frustration for both the instructor and students. Therefore, throughout the writing and production of this textbook we have worked very hard to catch and correct errors during each phase of development and production.

Each chapter has been reviewed by faculty members who teach the course or conduct research in genetics or both. In addition, a developmental editor has gone through the material to check for accuracy in art and consistency between the text and art. When the problem sets were first developed, we had a team of students work through all of the problems and one developmental editor also checked them. The author personally checked every question and answer when the chapters were completed for this edition.

ILLUSTRATIONS

In surveying students whom I teach, I often hear it said that most of their learning comes from studying the figures. Likewise, instructors frequently use the illustrations from a textbook as a central teaching tool. For these reasons, a great amount of effort has gone into the illustrations. The illustrations are created with four goals in mind:

1. **Completeness.** For most figures, it should be possible to understand an experiment or genetic concept by looking at the illustration alone. Students have complained that it is difficult to understand the content of an illustration if they have to keep switching back and forth between the figure and text. In cases where an illustration shows the steps in a scientific process, the steps are described in brief statements that allow the students to understand the whole process (e.g., see Figure 17.10). Likewise, such illustrations should make it easier for instructors to explain these processes in the classroom.
2. **Clarity.** The figures have been extensively reviewed by students and instructors. This has helped us to avoid drawing things that may be confusing or unclear. Aside from being unmistakably represented, all new elements within each figure are clearly labeled.
3. **Consistency.** Before we began to draw the figures, we generated a style sheet that contained recurring elements that are found in many places in the textbook. Examples include the DNA double helix, DNA polymerase, and fruit flies. We agreed on the best way(s) to draw these elements and also what colors they should be. Therefore, as students and instructors progress through this textbook, they become accustomed to the way things should look.
4. **Realism.** An important emphasis of this textbook is to make each figure as realistic as possible. When drawing macroscopic elements (e.g., fruit flies, pea plants), the illustrations are based on real images, not on cartoonlike simplifications. Our most challenging goal, and one that we feel has been achieved most successfully, is the realism of our molecular drawings. Whenever possible, we have tried to depict molecular elements according to their actual structures, if such structures are known. For example, the ways we have drawn RNA polymerase, DNA polymerase, DNA helicase, and ribosomes are based on their crystal structures. When a student sees a figure in this textbook that illustrates an event in transcription, RNA polymerase is depicted in a way that is as realistic as possible (e.g., Figure 14.8 below).



Key points:

- RNA polymerase slides along the DNA, creating an open complex as it moves.
- The DNA strand known as the template strand is used to make a complementary copy of RNA, resulting in an RNA–DNA hybrid.
- RNA polymerase moves along the template strand in a 3′ to 5′ direction, and RNA is synthesized in a 5′ to 3′ direction using nucleoside triphosphates as precursors. Pyrophosphate is released (not shown).
- The complementarity rule is the same as the AT/GC rule except that U is substituted for T in the RNA.

Laguna Design/Science Source

WRITING STYLE

Motivation in learning often stems from enjoyment. If you enjoy what you're reading, you are more likely to spend longer amounts of time with it and focus your attention more crisply. The writing style of this book is meant to be interesting, down to earth, and easy to follow. Each section of every chapter begins with an overview of the contents of that section, usually with a table or

figure that summarizes the broad points. The section then examines how those broad points were discovered experimentally, as well as explaining many of the finer scientific details. Important terms appear in the text in a boldface font. These terms are also found at the end of the chapter and in the glossary.

There are various ways to make a genetics book interesting and inspiring. The subject matter itself is pretty amazing, so it's not difficult to build on that. In addition to describing the concepts and experiments in ways that motivate students, it is important to draw on examples that bring the concepts to life. In a genetics book, many of these examples come from the medical realm. This textbook contains lots of examples of human diseases that convey some of the underlying principles of genetics. Students often say they remember certain genetic concepts because they remember how defects in certain genes can cause disease. For example, defects in DNA repair genes cause a higher predisposition to develop cancer. In addition, I have tried to be evenhanded in providing examples from the microbial and plant world. Finally, students are often interested in applications of genetics that affect their everyday lives. Because we frequently hear about genetics in the news, it's inspiring for students to learn the underlying basis for such technologies. Chapters 20 and 21 are devoted to genetic technologies, and applications of these and other technologies are found throughout this textbook. By the end of their genetics course, students should come away with a greater appreciation for the influence of genetics in their lives.

SIGNIFICANT CONTENT CHANGES TO THE FOURTH EDITION

In addition to the usual updates to material based on new research information, other additions and changes to the fourth edition have been made, as described next.

- Chapter 2. Reproduction and Chromosome Transmission: The topic of bacterial cell division has been updated with regard to the role of FtsZ in forming a septum between daughter cells (see new inset to Figure 2.4).
- Chapter 4. Sex Determination and Sex Chromosomes: In Figure 4.7, which concerns X-chromosome inactivation, the nucleation step has been revised.
- Chapter 5. Extensions of Mendelian Inheritance: A new section has been added that describes how gene modification plays a role in the color of parakeet feathers (see new Table 5.4).
- Chapter 8. Variation in Chromosome Structure and Number: The topic of copy number variation in humans and other species has been updated.
- Chapter 9. Genetics of Bacteria: The topic of transduction has been simplified.
- Chapter 10. Genetics of Viruses: New information on coronavirus has been added, including a new micrograph (see Figure 10.5).
- Chapter 11. Molecular Structure of DNA and RNA: Figure 11.15a has been revised based on known structures of bulge loops in RNA molecules.
- Chapter 12. Molecular Structure of Chromosomes and Transposition: The discussion of eukaryotic chromosome structure has been updated based on new knowledge regarding their compaction. Figures 12.16b, 12.17, 12.19, and 12.20 are new figures that are replacing figures in previous editions.
- Chapter 14. Gene Transcription and RNA Modification: The topic of splicing has been revised to mention that the spliceosome is a metalloribozyme.
- Chapter 16. Gene Regulation in Bacteria: The subsection on riboswitches has been updated to show that a riboswitch has an aptamer domain and an expression platform. Figures 16.16 and 16.17 have been revised.
- Chapter 17. Gene Regulation in Eukaryotes: The chapter has a new section (Section 17.5) called Heterochromatin: Function, Structure, Formation, and Maintenance, which includes four new figures (see Figures 17.15 through 17.18).
- Chapter 18. Non-Coding RNAs: The description of PIWI-interacting RNAs has been updated to distinguish between transcriptional silencing via piRITS versus RNA degradation via piRISC (see Figure 18.9).
- Chapter 20. Molecular Technologies: New information has been added on the use of dead Cas9 to activate gene expression, including a new figure (see Figure 20.19).
- Chapter 21. Genomics: Updated genomic information has been added in several places throughout the chapter.
- Chapter 22. Medical Genetics and Cancer: In the Fourth Edition, a greater effort has been made to distinguish between disorders that follow a simple Mendelian inheritance pattern versus those that involve mutations in multiple genes and are genetically more complex.
- Chapter 24. Quantitative Genetics: Figure 24.10 has been replaced with a new illustration that conveys the concept of selective breeding more clearly.

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 greatly indebted to a variety of people. This textbook has gone 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.

Let me begin by acknowledging the many people at McGraw-Hill Education whose efforts are amazing. My highest praise goes to Ian Townsend (Portfolio Manager), who managed many aspects of this project. I also would like to thank Elizabeth Sievers (Senior Product Developer) for her patience in overseeing this project and her contributions to the digital components. Liz is the glue that holds textbook development and textbook production together. Other people at McGraw-Hill have played key roles in producing an actual book and the supplements that go along with it. In particular, Jessica Portz (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 Beth Cray (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 as well as creating an awesome cover. Finally, I would like to thank Kelly Brown (Marketing Manager), whose major efforts begin once the fourth edition is published!

With regard to the content of the book, Deborah Brooker (Freelance Developmental Editor) has worked closely with me in developing the art for this textbook for all of the editions. She has scrutinized each figure for clarity and logic. I would also like to thank Jane Hoover (Freelance Copy Editor) for her superb copy-editing. Her crisp understanding of the material allows her to edit it in a meaningful way, which has significantly improved the text's clarity. She is at the top of the list for copyeditors.

I would also like to extend my thanks to everyone at MPS Limited, including the many artists who have played important roles in developing the illustrations for this textbook. Also, the folks at MPS Limited worked with great care in the paging of the book, making sure that the figures and relevant text are as close to each other as possible. Likewise, the people at MPS Limited have done a great job of locating many of the photographs that have been used in this textbook.

Finally, I want to thank Alexey Nikitin, *Grand Valley State University* and the many scientists who provided feedback and recommendations over the editions. Their broad insights and constructive suggestions were an important factor that shaped its final content and organization. I am truly grateful for their time and effort.

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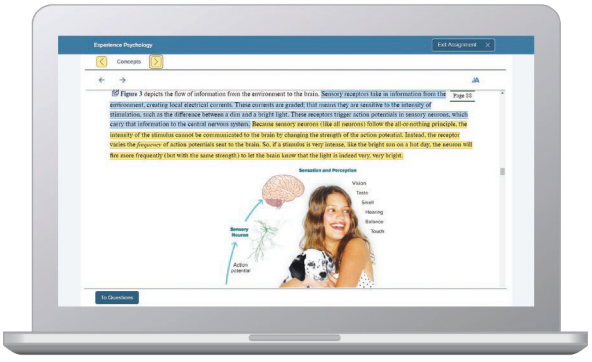

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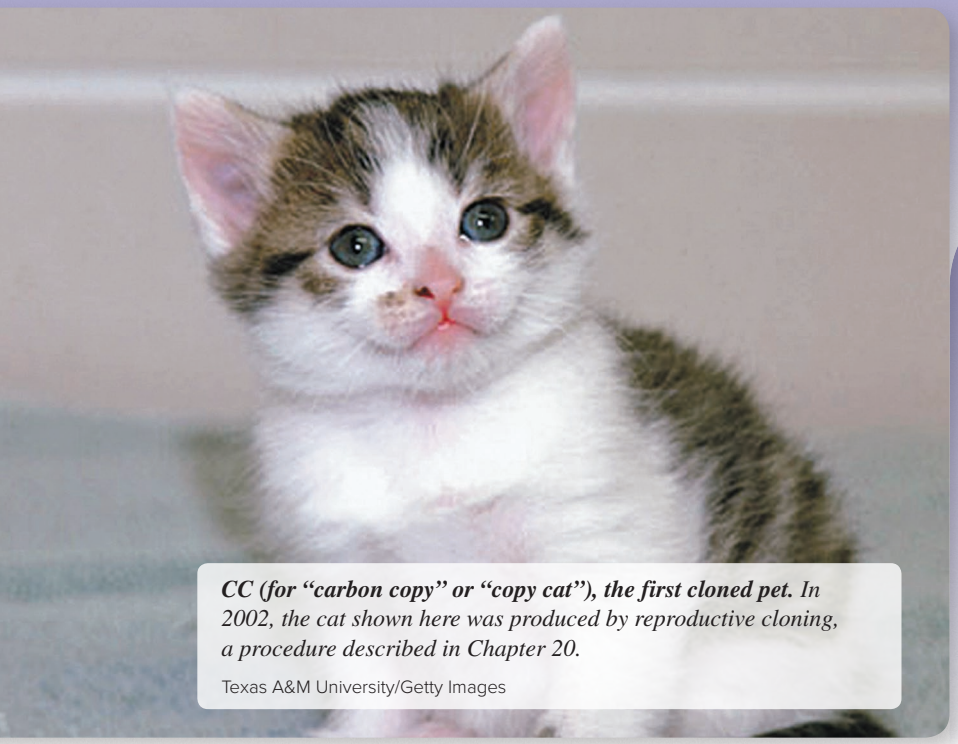


CC (for "carbon copy" or "copy cat"), the first cloned pet. *In 2002, the cat shown here was produced by reproductive cloning, a procedure described in Chapter 20.*

Texas A&M University/Getty Images

CHAPTER OUTLINE

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 (NIH) and the Department of Energy joined forces with international partners to decipher the massive amount of information contained in our **genome**—the **deoxyribonucleic acid (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 of 90% of the human genome. The completed sequence, published in 2003, has an accuracy greater than 99.99%; fewer than 1 mistake was made in every 10,000 base pairs (bp)!

In 2008, a more massive undertaking, called the 1000 Genomes Project, was launched, with the goal of attaining 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, such as 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 lead to improvements in modern medicine by providing better diagnoses of diseases and allowing the development of new treatments for them.

A controversial example of a genetic technology is mammalian cloning. In 1997, Ian Wilmut and his colleagues produced clones of sheep, using mammary cells from an adult animal (**Figure 1.2**). More recently, such cloning has been achieved for 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. Cloning of livestock 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. As discussed in Chapter 20, this prospect has raised serious ethical questions. Within the past few years, legislation that involves bans on human cloning has been introduced.

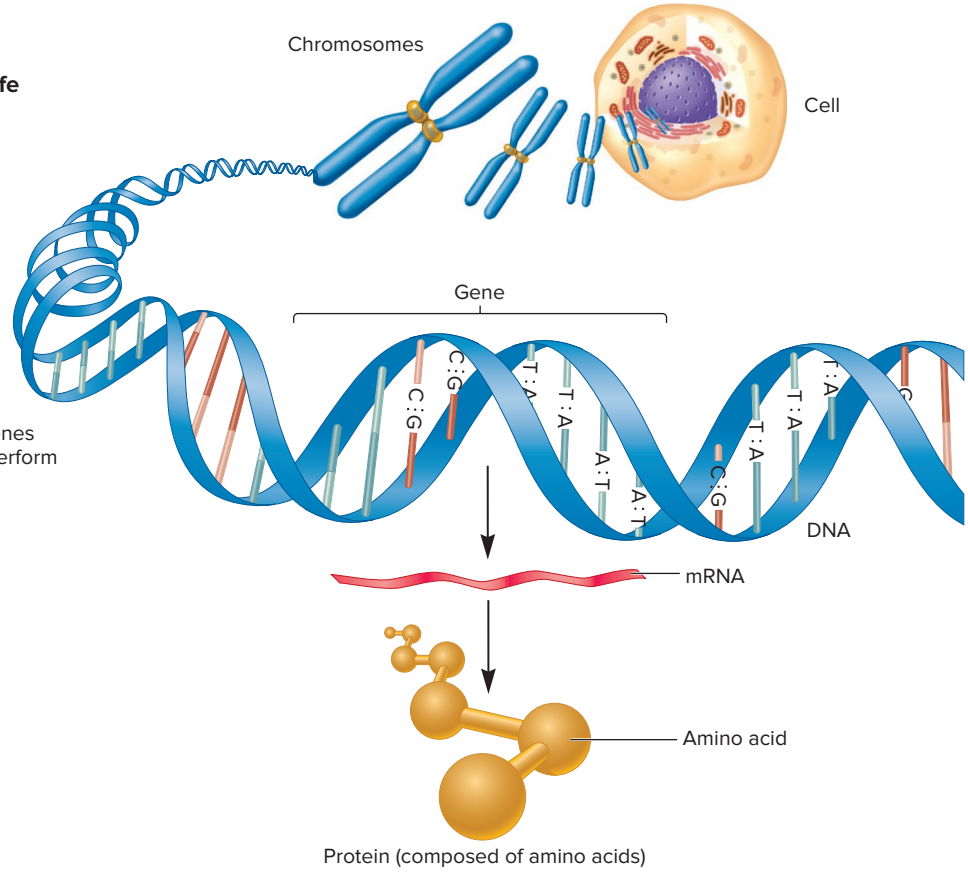


FIGURE 1.1 **The Human Genome Project.** The human genome is a complete set of human chromosomes. People have two sets of chromosomes, one set from each parent. Collectively, each set of chromosomes is composed of a DNA sequence that is approximately 3 billion base pairs long. As discussed later, most 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 protein-coding 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?*

FIGURE 1.2 **The cloning of a mammal.** The lamb on the left is Dolly, the first mammal to be cloned. She was cloned from a cell of a Finn Dorset (a white-faced sheep). The sheep on the right is Dolly's surrogate mother, a Blackface ewe. A description of how Dolly was produced is presented in Chapter 20.

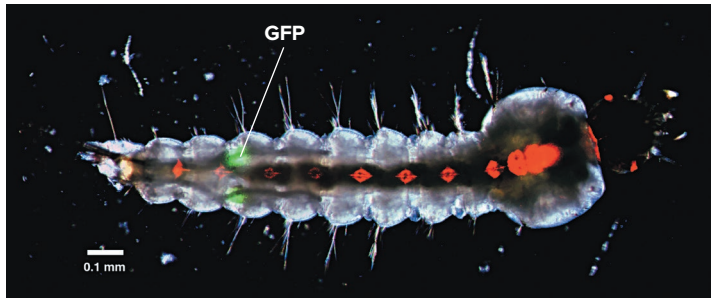
R. Scott Horner KRT/Newscom

Concept Check: *What ethical issues may be associated with human cloning?*

Finally, genetic technologies provide the means of modifying 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" 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.

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

(a) GFP expressed in mice



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

FIGURE 1.3 Introduction of a jellyfish gene into laboratory mice and mosquitoes. (a) A gene that naturally occurs in certain 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)** *GFP* 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 female mosquitoes?

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 breed only once. 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. Outline how DNA stores the information to make proteins.
3. Explain how proteins are largely responsible for cell structure and function.

Genetics is the branch of biology that focuses on 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, but such a vague definition does not do justice to the exciting characteristics of genes as intricate molecular units that manifest themselves as critical contributors to cell structure and function.

- 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—a linear sequence of amino acids that folds into a unit that constitutes a protein or part of a protein.
- 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. An 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 characteristics.

In this section, we will examine the general features of life with an emphasis on the molecular level. 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 a cohesive review of topics covered 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 24.

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 cellular molecules are **nucleic acids** (i.e., DNA and RNA), **proteins**, **carbohydrates**, and **lipids**. Three of these—nucleic acids, proteins, and carbohydrates—exist as **macromolecules** that are composed of many repeating units of smaller building blocks. Proteins, RNA, and carbohydrates can be 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. **Figure 1.4** illustrates this concept.

- Nucleotides are small organic molecules.
- Nucleotides are linked to each other and form the building blocks of DNA, which is a macromolecule.
- DNA is a component of chromosomes, which also contain proteins that contribute to chromosome structure.
- Within a eukaryotic cell, the chromosomes are contained in a compartment called the cell nucleus. The nucleus is bounded by a double membrane that is composed of lipids and proteins and shields the chromosomes from the rest of the cell. The nucleus is an example of an **organelle**—a membrane-bound compartment with a specialized function. The cell nucleus protects the chromosomes from mechanical damage and provides a single compartment for genetic activities such as gene transcription.
- Finally, cellular molecules, macromolecules, and organelles 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**. As we will learn throughout this textbook, proteins are the "workhorses" of all living cells. The range of functions among different types of proteins is truly remarkable. Some examples include the following:

- Proteins help determine the shape and structure of a given cell. For example, the protein known as tubulin can assemble into large structures known as microtubules, which provide a cell with internal structure and organization.
- Proteins are inserted into cell membranes and aid in the transport of ions and small molecules across the membrane.

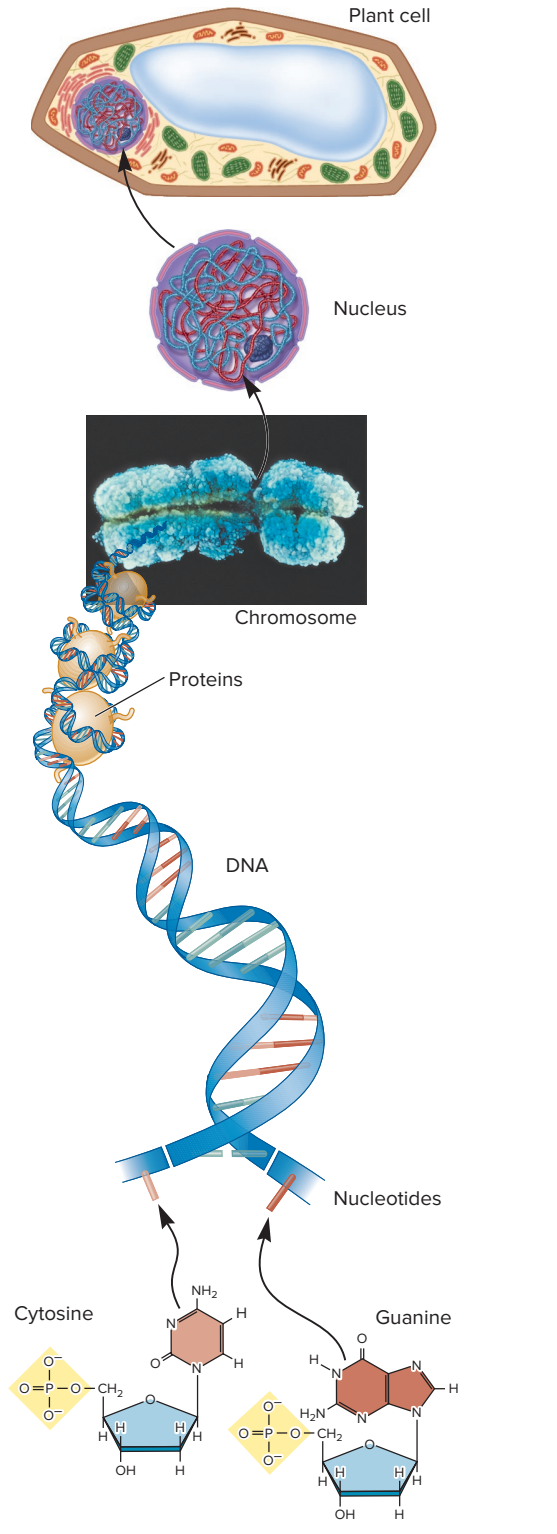


FIGURE 1.4 Molecular organization of a living cell. Cellular structures are constructed from smaller building blocks. In this example, DNA is formed from the linkage of nucleotides, producing a very long macromolecule. The DNA associates with proteins to form a chromosome. The chromosomes are located 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?

- Proteins may also function as biological motors. An interesting case is the protein known as myosin, which is involved in the contractile properties of muscle cells.
- Within multicellular organisms, certain proteins function in cell-to-cell recognition and signaling. For example, the hormone insulin is secreted by endocrine cells and binds to the insulin receptor proteins found within the plasma membrane of target cells.
- **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. These enzymes are important in the utilization of energy.

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

As mentioned, 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 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 task is extremely complicated 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 of the numbers of proteins produced by complex eukaryotes 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 of which 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.

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

- The sequence of amino acids in a polypeptide causes it to fold into a particular structure; 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.

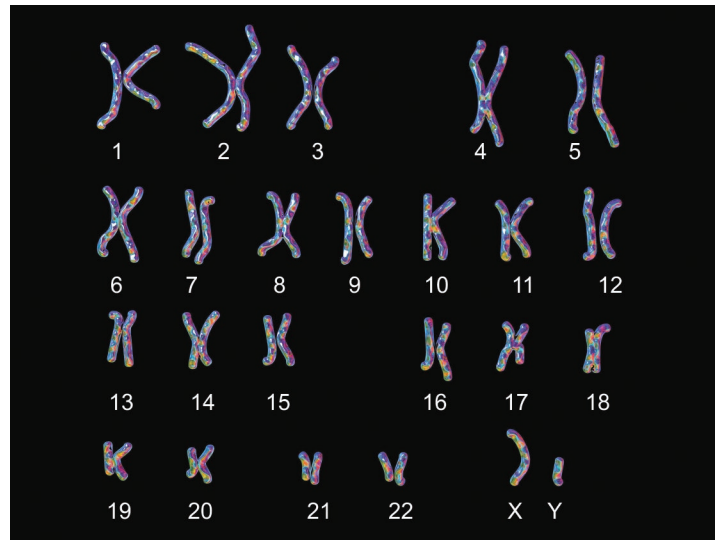


FIGURE 1.5 A micrograph of the 46 chromosomes found in a cell from a human male.

Kateryna Kon/Shutterstock

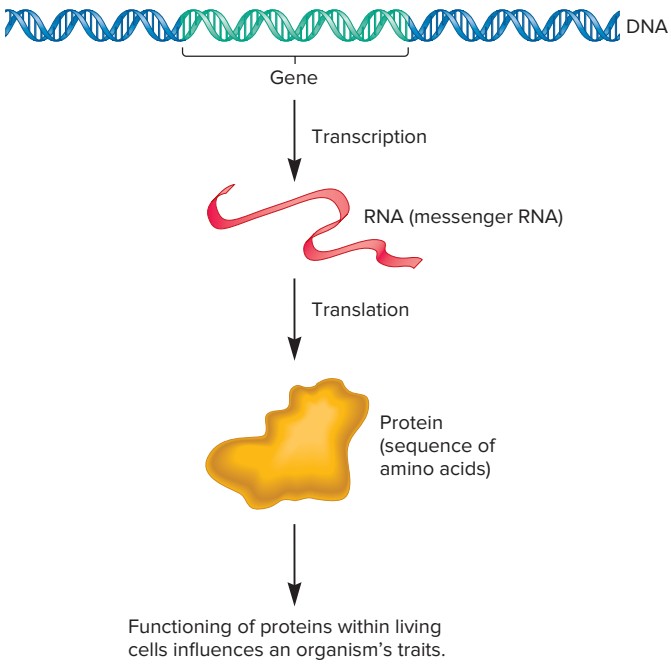
Concept Check: Which types of macromolecules are found in chromosomes?

In living cells, DNA is found within large structures known as **chromosomes**. Figure 1.5 is a micrograph of the 46 chromosomes in a cell from a human male, which are found in pairs. 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 carry about 1000 different genes.

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 is accessed in a stepwise process (Figure 1.6).

1. In the first step, known as **transcription**, the DNA sequence within a gene is copied into a nucleotide sequence of **ribonucleic acid (RNA)**. Most genes encode RNAs that contain the information for the synthesis of a particular polypeptide. This type of RNA is called **messenger RNA (mRNA)**.
2. During the process of **translation**, the sequence of nucleotides in an mRNA provides the information (using the genetic code) to produce the amino acid sequence of a polypeptide.
3. A polypeptide folds into a three-dimensional structure. As mentioned, a protein is a functional unit. Some proteins are composed of a single polypeptide, and other proteins consist of two or more polypeptides.
4. The functioning of proteins largely determines cell structure and function.



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 functional protein, thereby influencing an organism's traits.

Concept Check: Where is the information to make a polypeptide stored?

1.1 REVIEWING THE KEY CONCEPTS

- Living cells are composed of nucleic acids (DNA and RNA), proteins, carbohydrates, and lipids.
- The entire collection of proteins a cell makes at a given time is its proteome. The proteome largely determines the structure and function of a cell (see Figure 1.4).
- DNA, which is found within chromosomes, stores the information needed to make 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).

1.1 COMPREHENSION QUESTIONS

1. Which of the following is *not* a constituent of a cell's proteome?
   a. An enzyme
   b. A motor protein
   c. A receptor 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, traits, and the environment.
4. Describe how genes are transmitted in sexually reproducing species.
5. Describe the process of evolution.

A trait is any characteristic that an organism displays. In genetics, we place traits into different categories.

- **Morphological traits** 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.
- **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 Within Cells Leads to an Organism's Traits

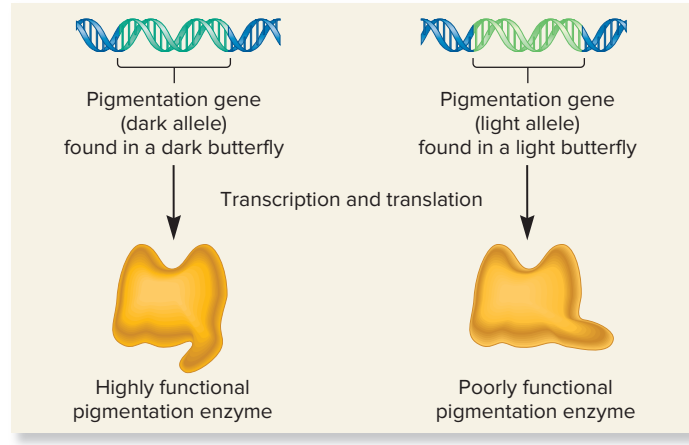
A complicated, yet very exciting, aspect of genetics is that the field's observations and theories span four levels of biological organization: molecules, cells, organisms, and populations. This broad scope can make it difficult to appreciate the relationship

between genes and traits. To understand this connection, we need to relate the following four phenomena:

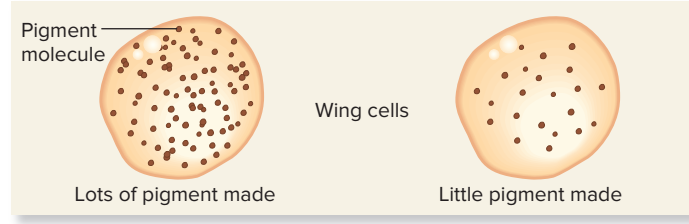
1. As we learned in Section 1.1, genes are expressed at the **molecular level**. In other words, gene transcription and translation lead to the production of a particular protein, which is a molecular process.
2. Proteins often function at the **cellular level**. The function of a protein within a cell 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.

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 the others are very light. Let's consider how we can explain this trait at the molecular, cellular, organism, and population levels.

1. 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 can exist in two different forms called **alleles**. In this example, one allele results in 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.
2. At the cellular level (Figure 1.7b), the functional differences between the 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.

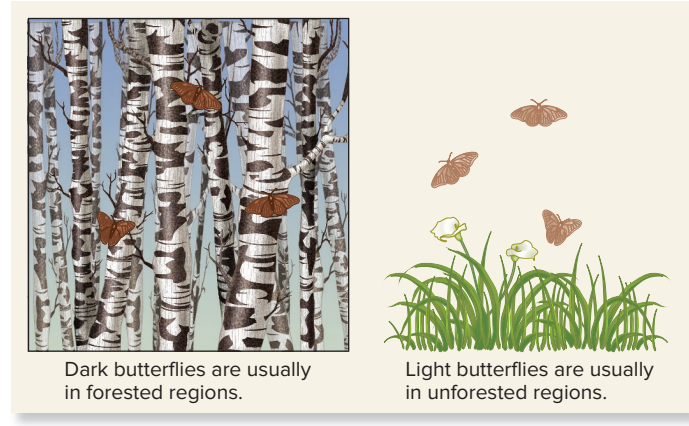


(a) Molecular level



(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?

3. 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.
4. Finally, at the population level (Figure 1.7d), geneticists want to know why a species of butterfly has some members with dark wings and others 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 was perched on a dark surface such as a tree trunk. In contrast, the 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 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.

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

Concept Check: Why do these two frogs look so different?

Inherited Differences in Traits Are Due to Genetic Variation

In Figure 1.7, we considered how gene expression can lead to variation in a trait of an organism, specifically, 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 black hair, and others have brown hair; some petunias have white flowers, but 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 the outcome of traits. At the molecular level, genetic variation can be attributed to different types of modifications.

- Small or large differences can occur within gene sequences. When such changes initially occur, they are called **gene mutations**, which are heritable changes in the genetic material. Gene 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 specifies.
- 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.
- 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 within the sequences of 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. Some human genetic disorders are the result of chromosomal alterations. An 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 six sets of chromosomes, whereas wild species typically have two sets (**Figure 1.9b**).

Traits Are Governed by Genes and by the Environment

In our discussion thus far, we have considered the role that genes play in the outcome of traits. Another critical factor is the **environment**—the surroundings in which an organism exists. A variety of factors in an organism's environment profoundly affect 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. The term **norm of reaction** refers to the effects of environmental variation on an individual's traits.

(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)** Wheat plants. Cultivated wheat has six sets of chromosomes.

(a) Stockbyte / Alamy; (b) Pixtal/age fotostock

Concept Check: Are these examples of gene mutations, variation in chromosome structure, or variation in chromosome number?

FIGURE 1.10 **Environmental influence on the outcome of PKU.** This girl with PKU has developed properly because she followed a diet that is very low in phenylalanine.

©Noah Goodrich/Zumapress/Newscom

Concept Check: What would have been the consequences if this girl had followed a standard diet, which contains a higher amount of phenylalanine?

External influences may dictate the way that genetic variation is manifested in an individual. An interesting example is the human genetic disorder **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.

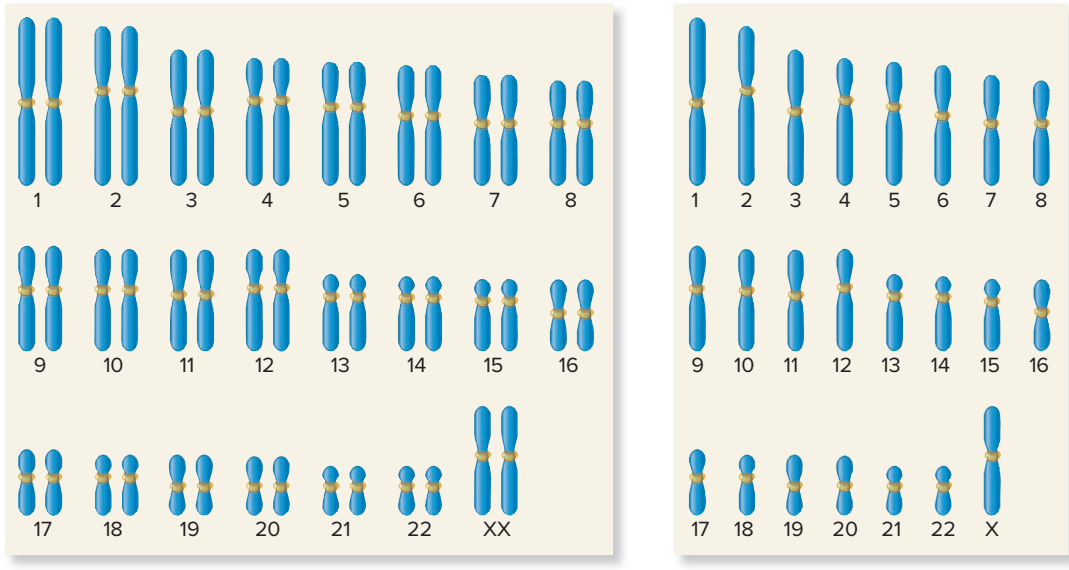
A rare variation in the sequence of the phenylalanine hydroxylase gene results in a nonfunctional version of this protein. Individuals with two copies of this rare, inactive allele cannot metabolize phenylalanine properly. Such individuals represent about 1 in 8000 births in the United States. When given 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. On such a diet, PKU individuals manifest a variety of detrimental traits, including mental impairment, underdeveloped teeth, and foul-smelling urine. In contrast, when PKU individuals are identified at birth and raised on a restricted diet that is low in phenylalanine, they develop normally (**Figure 1.10**). Fortunately, through routine screening of newborns, most affected babies in the United States are now diagnosed and treated early. 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 topic of inheritance. How are traits passed from parents to offspring? The foundation for our understanding of inheritance came from the studies of pea plants by Gregor Mendel in the nineteenth century. His work revealed that genetic determinants, 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 1.11a**). With the exception of the sex chromosomes (X and Y), each chromosome in a homologous pair contains the same kinds of genes. For example, both copies of human chromosome 12 carry the gene that encodes phenylalanine hydroxylase, which was discussed previously. 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 1.11b**).
- 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



(a) Chromosomal composition found in most female human cells (46 chromosomes)

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

FIGURE 1.11 **The complement of human chromosomes in somatic cells and gametes.** **(a)** A schematic drawing of the 46 chromosomes of a human female. With the exception of the sex chromosomes, chromosomes 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?

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

As proposed 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 of the anteater, enabling it to feed on ants located in the ground. 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 survive and 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 survive and reproduce in its native 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 1.12** depicts the evolution of the modern-day horse. 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 producing adaptations to changing global climates. Over North America, where much of horse evolution occurred, 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. The changes seen in horses' teeth are consistent with a shift from eating tender leaves to eating grasses and other types of vegetation that are tougher and require more chewing.

1.2 REVIEWING THE KEY CONCEPTS

- 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 1.8–1.10).
- 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 1.11).
- Evolution refers to changes in the genetic composition of a population from one generation to the next (see Figure 1.12).

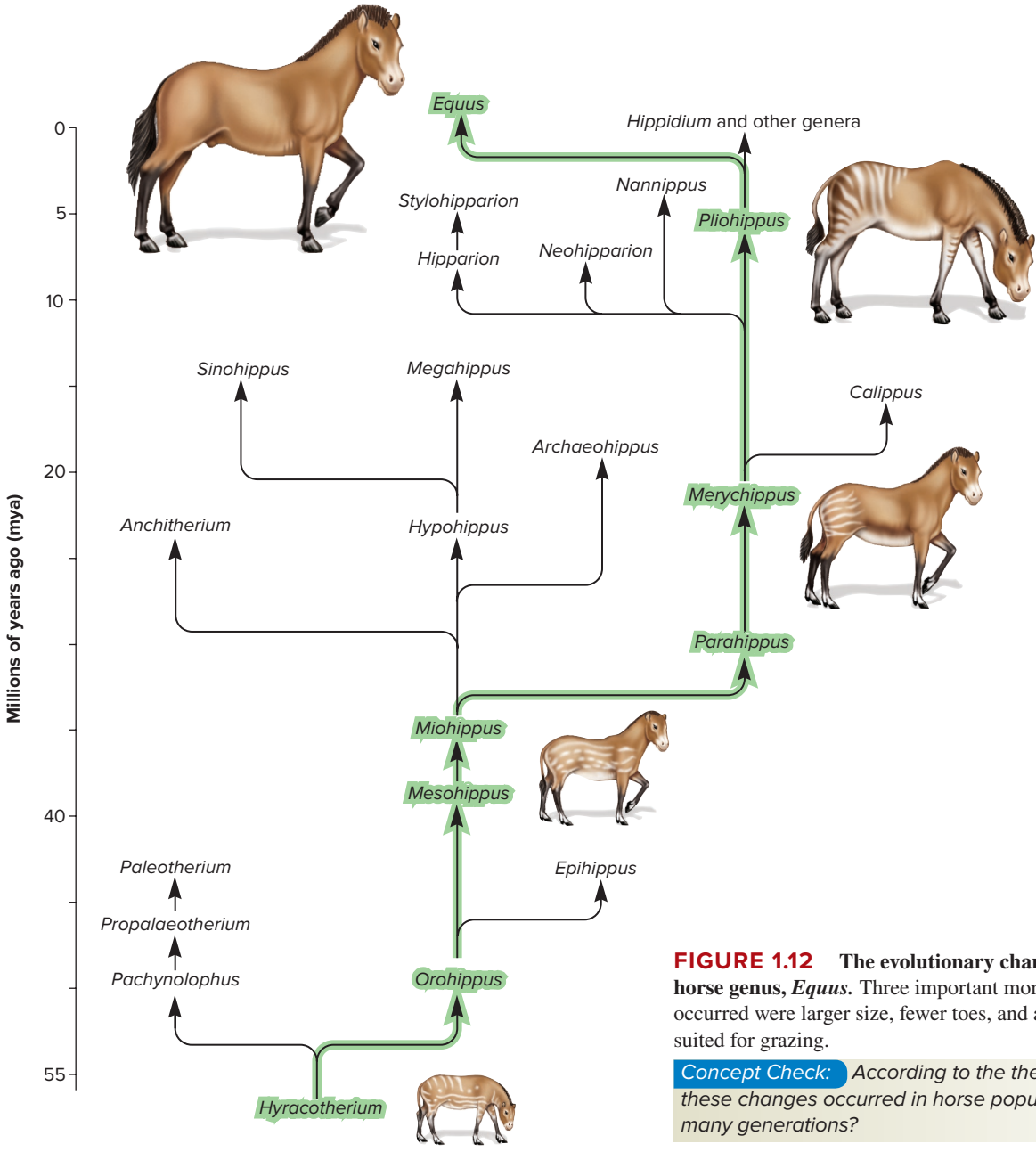


FIGURE 1.12 **The evolutionary changes that led to the modern horse genus, *Equus*.** Three important morphological changes that occurred were larger size, fewer toes, and a shift toward a jaw structure suited for grazing.

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

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.

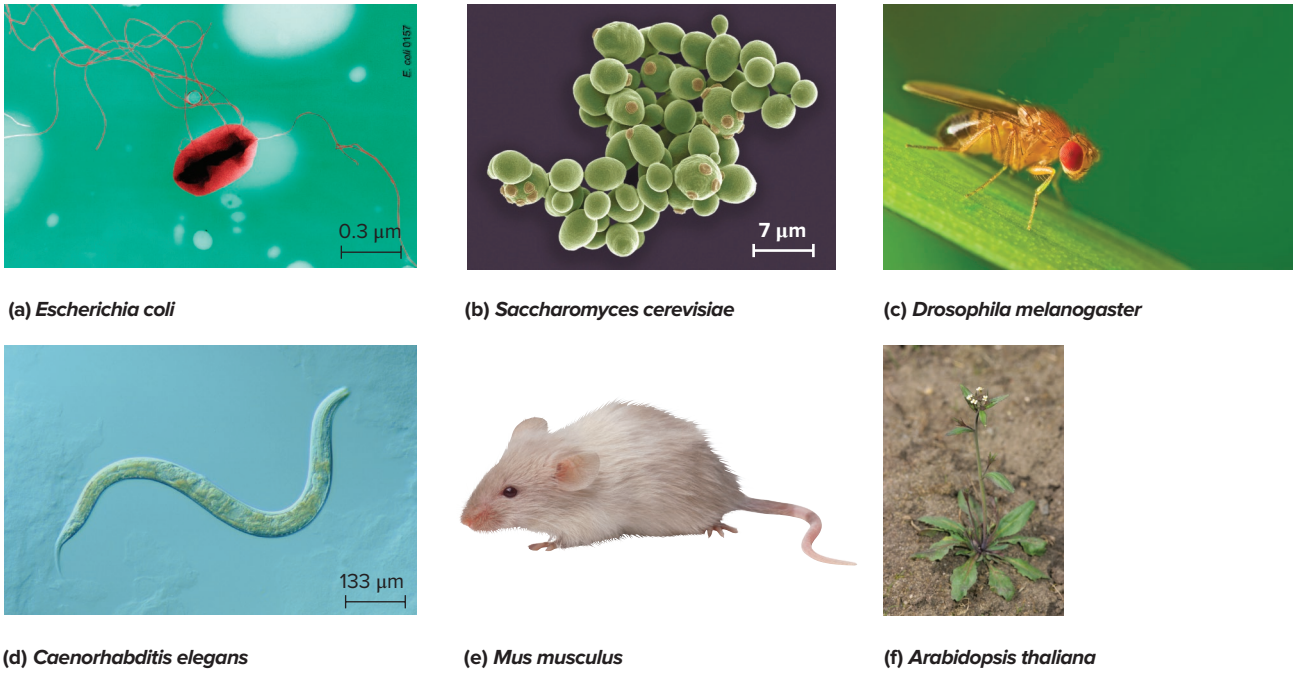
Genetics is a broad discipline encompassing molecular, cellular, organism, and population biology. Many scientists who are interested in genetics have been trained in supporting disciplines such as biochemistry, biophysics, cell biology, mathematics, microbiology, population biology, ecology, agriculture, 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 1.13** shows some examples of model organisms, including *Escherichia coli* (a bacterium), *Saccharomyces cerevisiae* (a yeast), *Drosophila melanogaster* (fruit fly), *Caenorhabditis elegans* (a nematode worm), *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 will examine the general questions that scientists in these areas are attempting to answer.

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 genetic determinants, which we now call genes, are passed as discrete units from parents to offspring via sperm and egg cells. Since Mendel's pioneering studies of the 1860s, our knowledge of genetic transmission has greatly increased. Many patterns of genetic transmission are more complex than the simple Mendelian patterns that are described in Chapter 3. The additional complexities of transmission genetics are examined in Chapters 4 through 10.

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



(a) *Escherichia coli* (b) *Saccharomyces cerevisiae* (c) *Drosophila melanogaster*

(d) *Caenorhabditis elegans* (e) *Mus musculus* (f) *Arabidopsis thaliana*

FIGURE 1.13 **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) CDC/ Peggy S. Hayes & Elizabeth H. White, M.S.; (b) Science Photo Library/Alamy Stock Photo; (c) janeff/iStockphoto/Getty Images; (d) Sinclair Stammers/Science Source; (e) G.K. & Vikki Hart/Getty Images; (f) WILDLIFE GmbH/Alamy Stock Photo

Concept Check: *Can you think of another example of a model organism?*

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 3, the ratio of tall to dwarf offspring (3:1) provides important information concerning the inheritance pattern of the height trait.

Throughout Chapters 2 to 10, we will learn how researchers try to answer many fundamental questions concerning the passage of genetic material from cell to cell and the passage of traits from parents to offspring.

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 effect of a gene mutation that eliminates the function of a gene. This is called a **loss-of-function mutation**, and the resulting 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 theory of evolution by natural selection proposed by Darwin provided a biological 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. The work of population geneticists 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 REVIEWING THE KEY CONCEPTS

- Model organisms are studied by many different researchers so that they can compare their results and determine scientific principles that apply more broadly to other species (see Figure 1.13).
- Genetics is traditionally divided into transmission genetics, molecular genetics, and population genetics, though overlap occurs among these fields.

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

Regardless of what field of genetics they work in, 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 genes 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 these observations. Along the way, the student is provided with many bits and pieces of experimental techniques and data. Although this textbook provides you with many observations and hypotheses, it attempts to go one step further. Many of the following 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. The five steps are as follows:

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(s) 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. Hopefully, you will find this a more interesting and rewarding way to learn about genetics. 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 alternative approaches and 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 improve your skills of formulating hypotheses, designing experiments, and interpreting data. Also, some of the questions in the problem sets are aimed at refining these skills.

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

As you 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 24 contain solved problems named **Genetic TIPS**, which stands for **T**opic, **I**nformation, and **P**roblem-solving **S**trategy. 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 CUU 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 (see arrow). Because codons specify amino acids, this alteration may change the third amino acid to something else. Note: If you look ahead to Chapter 15 (see Table 15.1), 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 24, 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.* Because genetics is an experimental science, many problems involve the analysis of data, which are the product of experiments. 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 better understand genetic concepts and to make predictions.

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

1.4 REVIEWING THE KEY CONCEPTS

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

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

Page 1. genome, deoxyribonucleic acid (DNA)
Page 3. genetics, gene, traits
Page 4. nucleic acids, proteins, carbohydrates, lipids, macromolecules, organelle, proteome
Page 5. enzymes, nucleotides, polypeptides, genetic code, codon, amino acid, chromosomes, gene expression, transcription, ribonucleic acid (RNA), messenger RNA (mRNA), translation
Page 6. morphological traits, physiological traits, behavioral traits
Page 7. molecular level, cellular level, organism level, species, population level, alleles
Page 8. genetic variation, morphs, gene mutations, environment, norm of reaction
Page 9. phenylketonuria (PKU), diploid, homologs, somatic cells, gametes, haploid
Page 10. biological evolution, evolution, natural selection
Page 12. model organisms, genetic cross
Page 13. genetic approach, loss-of-function mutation, loss-of-function allele, hypothesis testing, scientific method
Page 14. discovery-based science

CHAPTER SUMMARY

- The complete genetic composition of a cell is called 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 1.1–1.3).

1.1 The Molecular Expression of Genes

- Living cells are composed of nucleic acids (DNA and RNA), proteins, carbohydrates, and lipids.
- The entire collection of proteins a cell makes at a given time is its proteome. The proteome largely determines the structure and function of a cell (see Figure 1.4).
- DNA, which is found within chromosomes, stores the information needed to make proteins (see Figure 1.5).
- Most genes encode polypeptides, which are the units that make up 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 1.8–1.10).
- 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 1.11).
- Evolution refers to changes in the genetic composition of a population from one generation to the next (see Figure 1.12).

1.3 Fields of Genetics

- Model organisms are studied by many different researchers so that they can compare their results and determine scientific principles that apply more broadly to other species (see Figure 1.13).
- Genetics is traditionally divided into transmission genetics, molecular genetics, and population genetics, though overlap occurs among these fields.

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. 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 are critical to cell structure and function.

Problem-Solving Strategy: Relate structure and function.

One strategy for solving 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 structure and function 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 receive and transmit signals 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.

2. A human gene called *CFTR* (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. Is each of the following examples a description of 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. Usually, mutant genes that cause severe diseases are relatively rare. 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 are very sick, 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 levels. This concept is described in Figure 1.7.

Problem-Solving Strategies: Make a drawing. Compare and contrast.

One strategy to solve this problem is to make a drawing of the descriptions of parts A through D and decide if you are drawing something at the molecular, cellular, organism, or population level. For example, if you drew the description in part B, you would draw a protein, which is a molecule. If you drew the description in part C, you would draw a cell in which a salt imbalance is present. Another strategy to solve this problem is 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.

Conceptual Questions

C1. At the molecular level, what is a gene? Where are genes located?

C2. Briefly explain how gene expression occurs at the molecular level.

C3. A human gene called the β-globin gene encodes a polypeptide that functions as a subunit of the protein known as hemoglobin. Hemoglobin carries oxygen within red blood cells. In human populations, the β-globin gene can be found as the more common allele, called the Hb^A allele, but 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.

C4. 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?

C5. What is the cause of Down syndrome?

C6. The text describes how the trait of phenylketonuria (PKU) is greatly influenced by the environment. 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.

C7. What is meant by the term *diploid*? Which cells of the human body are diploid, and which cells are not?

C8. What is a DNA sequence?

C9. What is the genetic code?

C10. 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

C11. 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 for that animal to reproduce.

C12. What are the primary interests of researchers working in the following fields of genetics?

A. Transmission genetics

B. Molecular genetics

C. Population genetics

Application and Experimental Questions

E1. Pick any example of a genetic technology, and describe how it has directly affected your life.

E2. What is a genetic cross?

E3. 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?

E4. Figure 1.5 shows a micrograph of the common number of chromosomes from a human cell. If you created this type of display using cells from a person with Down syndrome, what would you expect to see?

E5. Many organisms are studied by geneticists. Of the following species, do you think it is more likely that each of them would 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

E6. Pick any trait you like in any species of wild plant or animal. The trait must somehow vary among different members of the species. For example, some butterflies have dark wings and others have light wings (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 explains 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.

Answers to Comprehension Questions

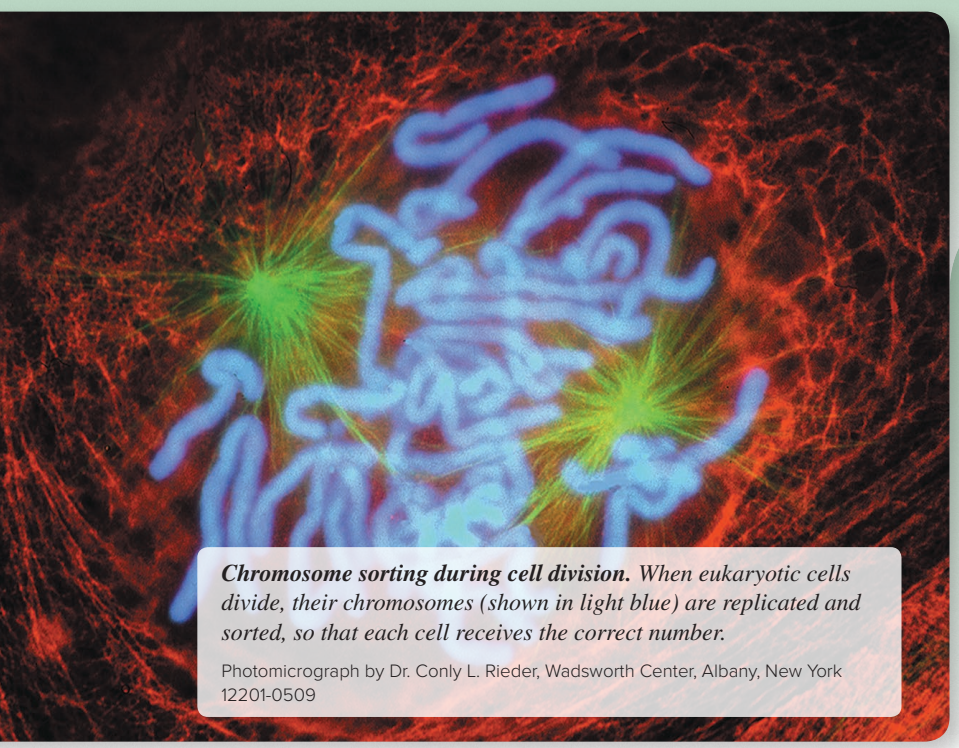
1.1: d, c, b, b

1.2: d, d, a, c

1.3: d, a

1.4: d

Note: All answers are available for the instructor in Connect; the answers to even-numbered questions and all Concept Check questions are in Appendix B.



Chromosome sorting during cell division. *When eukaryotic cells divide, their chromosomes (shown in light blue) are replicated and sorted, so that each cell receives the correct number.*

Photomicrograph by Dr. Conly L. Rieder, Wadsworth Center, Albany, New York 12201-0509

2

CHAPTER OUTLINE

REPRODUCTION AND CHROMOSOME TRANSMISSION

Reproduction is the biological process by which new cells or new organisms are produced. In this chapter, we will first survey reproduction at the cellular level, paying close attention to the inheritance of chromosomes. An examination of chromosomes at the microscopic level provides us with insights regarding the inheritance patterns of traits, which we will consider in Chapter 3. To appreciate this relationship, we will examine how cells distribute their chromosomes during the process of cell division. We will see that in bacteria and most unicellular eukaryotes, simple cell division provides a way to reproduce asexually. Next we will explore a form of cell division called meiosis that produces cells with half the number of chromosomes. This form of cell division is needed for sexual reproduction, which is the formation of a new individual following the union of two gametes. This chapter will end with a discussion of how sexual reproduction occurs in animals and plants.

2.1 GENERAL FEATURES OF CHROMOSOMES

Learning Outcomes:

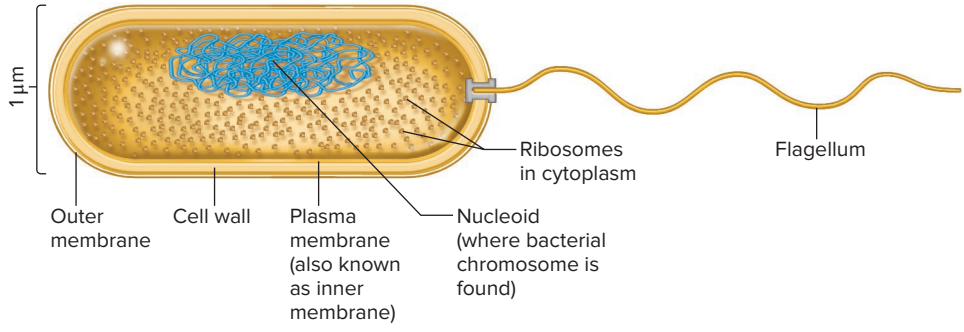
1. Define the term *chromosome*.
2. Outline key differences between prokaryotic and eukaryotic cells.
3. Describe the procedure for making a karyotype.
4. Summarize the similarities and differences between homologous chromosomes.

Chromosomes are structures within living cells that contain the genetic material. Genes are physically located within chromosomes. Biochemically, each chromosome contains a very long segment of DNA, which is the genetic material, and proteins, which are bound to the DNA and provide it with an organized structure. In eukaryotic cells, this complex between DNA and proteins is

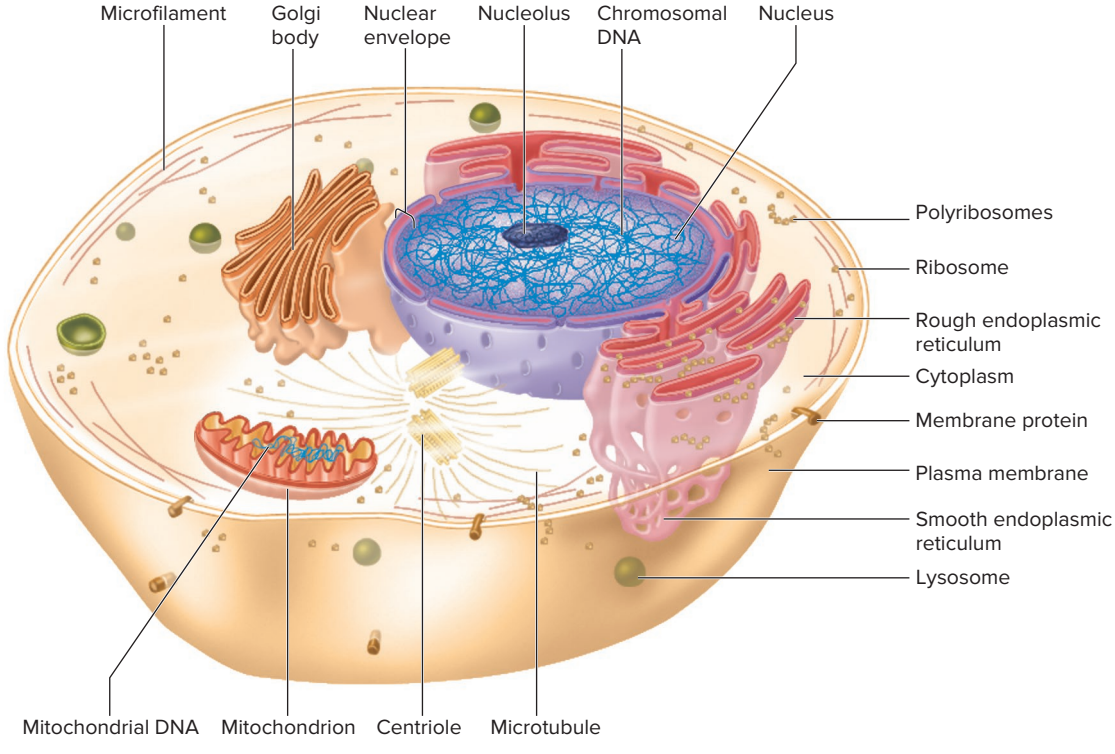
called **chromatin**. In this chapter, we will focus on the cellular mechanics of chromosome transmission to better understand the patterns of gene transmission that we will consider in Chapters 3 through 7. In particular, we will examine how chromosomes are copied and sorted into newly made cells. In later chapters, particularly Chapters 11 and 12, we will examine the molecular features of chromosomes in greater detail.

Before we begin a description of chromosome transmission, we need to consider the distinctive cellular differences between prokaryotic and eukaryotic species. Bacteria and archaea are referred to as **prokaryotes**, from the Greek meaning "prenucleus" because their chromosomes are not contained within a membrane-bound nucleus in the cell. Prokaryotes usually have a single type of circular chromosome in a region of the cytoplasm called the **nucleoid** (**Figure 2.1a**). The cytoplasm is enclosed by a plasma membrane that regulates the uptake of nutrients and the excretion of waste products. Outside the plasma membrane is a rigid cell wall that protects the cell from breakage. Certain species of bacteria also have an outer membrane on the exterior side of the cell wall.

Eukaryotes, from the Greek meaning "true nucleus," include some simple species, such as single-celled protists and some fungi (such as yeast), and more complex multicellular species, such as plants, animals, and other fungi. The cells of eukaryotic species have internal membranes that enclose highly specialized compartments (**Figure 2.1b**). These compartments form membrane-bound



(a) Prokaryotic cell



(b) Eukaryotic cell

FIGURE 2.1 **The basic organization of cells. (a)** A bacterial cell. The example shown here represents a typical bacterium such as *Escherichia coli*, which has an outer membrane. **(b)** A eukaryotic cell. The example shown here is a typical animal cell.

Concept Check: Eukaryotic cells exhibit compartmentalization. What does this mean?

organelles with specific functions. For example, a particularly conspicuous organelle is the **nucleus**, which is bounded by two membranes that constitute the nuclear envelope. Most of the genetic material is found within chromosomes that are located in the nucleus. In addition to the nucleus, certain organelles in eukaryotic cells contain a small amount of their own DNA. These include the mitochondrion, which plays a role in ATP synthesis, and, in plant cells, the chloroplast, which functions in photosynthesis. The DNA found in these organelles is referred to as extranuclear, or extrachromosomal, DNA to distinguish it from the DNA that is found in the cell nucleus. We will examine the role of mitochondrial and chloroplast DNA in Chapter 6.

In this section, we will focus on the composition of chromosomes found in the nucleus of eukaryotic cells. As you will learn, eukaryotic species contain genetic material that comes in sets of linear chromosomes.

Eukaryotic Chromosomes Are Examined Cytologically to Prepare a Karyotype

Insights into inheritance patterns have been gained by observing chromosomes under the microscope. **Cytogenetics** is the field of genetics that involves the microscopic examination of chromosomes. The most basic observation that a **cytogeneticist** can make is the examination of the chromosomal composition of a particular cell. For eukaryotic species, this is usually accomplished by observing the chromosomes as they are found in actively dividing cells. When a cell is preparing to divide, the chromosomes become more tightly coiled, which shortens them, thereby increasing their diameter. The consequence of this shortening is that distinctive shapes and numbers of chromosomes become visible with a light microscope. Each species has a particular chromosome composition. For example, most human cells contain 23 pairs of chromosomes, for a total of 46. On rare occasions, some individuals may inherit an abnormal number of chromosomes or a chromosome with an abnormal structure. Such abnormalities can often be detected by a microscopic examination of the chromosomes within actively dividing cells. In addition, a cytogeneticist may examine chromosomes as a way to distinguish two closely related species.

Figure 2.2a shows the general procedure for preparing human chromosomes to be viewed by microscopy. In this example, the cells were obtained from a sample of human blood; more specifically, the chromosomes within leukocytes (a type of white blood cell) were examined. Blood cells are a type of **somatic cell**, which is any cell of the body that is not a **gamete** or a precursor to a gamete. As discussed later, gametes are involved with sexual reproduction.

1. After the blood cells have been removed from the body, they are treated with one chemical that stimulates them to begin cell division and another chemical that halts cell division during mitosis, which is described later in this chapter.
2. As shown in Figure 2.2a, these actively dividing cells are subjected to centrifugation to concentrate them. The concentrated preparation is then mixed with a hypotonic solution that makes the cells swell. This swelling causes the chromosomes to spread out within the cell, thereby making it easier to see each individual chromosome.
3. After a second centrifugation step, the cells, which are at the bottom of the tube, are treated with a fixative that chemically freezes them so the chromosomes can no longer move around. The cells are then treated with a chemical dye that binds to the chromosomes and stains them. As discussed in greater detail in Chapter 8, this gives chromosomes a distinctive banding pattern that greatly enhances geneticists' ability to visualize and uniquely identify them (look ahead to Figure 8.1c, d). The cells are then placed on a slide and viewed with a light microscope.

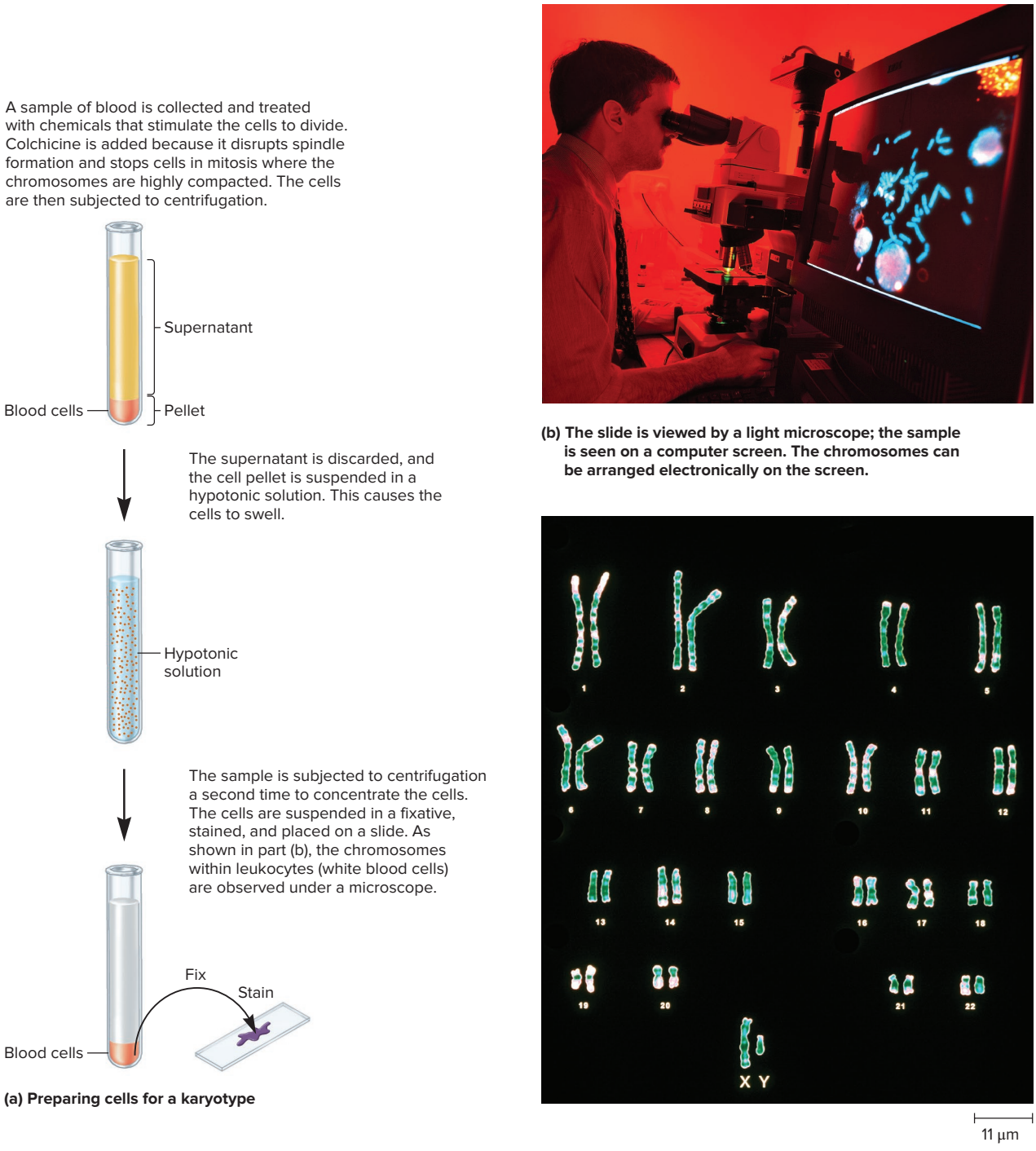
In a cytogenetics laboratory, the microscopes are equipped with a camera that can photograph the chromosomes. In recent years, advances in technology have allowed cytogeneticists to view microscopic images on a computer screen (**Figure 2.2b**). On the screen, the chromosomes can be arranged in a standard way, usually from largest to smallest. As seen in **Figure 2.2c**, the human chromosomes are lined up, and a number is assigned to designate each type of chromosome. An exception is the sex chromosomes, which are designated with the letters X and Y. An organized representation of the chromosomes within a cell is called a **karyotype**. A karyotype reveals how many chromosomes are found within an actively dividing somatic cell.

Eukaryotic Chromosomes Are Inherited in Sets

Most eukaryotic species are **diploid** or have a diploid phase in their life cycles, which means that each type of chromosome is a member of a pair. A diploid cell has two sets of chromosomes. In humans, for example, most somatic cells have 46 chromosomes—two sets of 23 each. Other diploid species, however, have different numbers of chromosomes in their somatic cells. For example, the dog has 39 chromosomes per set (78 total), the fruit fly has 4 chromosomes per set (8 total), and the tomato plant has 12 per set (24 total).

When a species is diploid, the members of a pair of chromosomes are called **homologs**; each type of chromosome is found in a homologous pair. As shown in Figure 2.2c, a human somatic cell has two copies of chromosome 1, two copies of chromosome 2, and so forth. Within each pair, the chromosome on the left is a homolog to the one on the right, and vice versa. In each pair, one chromosome was inherited from the mother and its homolog was inherited from the father. The two chromosomes in a homologous pair are nearly identical in size, have the same banding pattern, and contain a similar composition of genetic material. If a particular gene is found on one copy of a chromosome, it is also found on the other homolog.

However, the two homologs may carry different versions of a given gene, which are called **alleles**. In Chapter 3, we will see that some alleles are dominant, meaning that they mask the expression of alleles that are recessive. As an example, let's consider a gene in humans, called *Herc2*, which is one of a few different genes that affect eye color. The *Herc2* gene is located on chromosome 15 and comes in alleles that result in brown or blue eyes. In a person with brown eyes, one copy of chromosome 15 may carry a dominant brown allele, whereas its homolog may carry a



(a) Preparing cells for a karyotype

(b) The slide is viewed by a light microscope; the sample is seen on a computer screen. The chromosomes can be arranged electronically on the screen.

(c) For a diploid human cell, two complete sets of chromosomes from a single cell constitute a karyotype of that cell.

FIGURE 2.2 The procedure for making a human karyotype.

(b) David Parker/Science Source; (c) ©Leonard Lessin/Science Source

Concept Check: How do you think the end result of karyotype preparation would be affected if the blood cells were not treated with a hypotonic solution?

recessive blue allele. One copy was inherited from the mother and the other from the father.

At the molecular level, how similar are homologous chromosomes? The answer is that the sequence of bases of one homolog usually differs from the sequence of the other homolog by less than 1%. For example, the DNA sequence of chromosome 1 that you inherited from your mother is more than 99% identical to the sequence of chromosome 1 that you inherited from your father.

Nevertheless, it should be emphasized that the sequences are not identical. The slight differences in DNA sequences provide the allelic differences in genes. Again, if we use the eye color gene as an example, a slight difference in DNA sequence distinguishes the brown and blue alleles. However, the striking similarities between homologous chromosomes do not apply to the pair of sex chromosomes—X and Y. These chromosomes differ in size and genetic composition. Certain genes that are found on the X chromosome are not found on the Y chromosome, and vice versa. The X and Y chromosomes are not considered homologous chromosomes, though they do have short regions of homology.

Figure 2.3 shows two homologous chromosomes with three different genes. An individual carrying these two chromosomes is **homozygous** for the dominant allele of gene *A*, which means that both homologs carry the same allele. The individual is **heterozygous**, *Bb,* for the second gene, meaning that the homologs carry different alleles. For the third gene, the individual is homozygous for a recessive allele, *c*. The physical location of a gene is called its **locus** (plural: **loci**). As seen in Figure 2.3, for example, the locus of gene *C* is toward one end of this chromosome, whereas the locus of gene *B* is more in the middle.

2.1 REVIEWING THE KEY CONCEPTS

- Chromosomes are structures that contain the genetic material, which is DNA.
- Prokaryotic cells are simple and lack cell compartmentalization, whereas eukaryotic cells contain a cell nucleus and other compartments (see Figure 2.1).
- Chromosomes can be examined under the microscope. An organized representation of the chromosomes from a single cell is called a karyotype (see Figure 2.2).
- In eukaryotic species, the chromosomes are found in sets. Eukaryotic cells are often diploid, which means that each type of chromosome occurs in a homologous pair (see Figure 2.3).

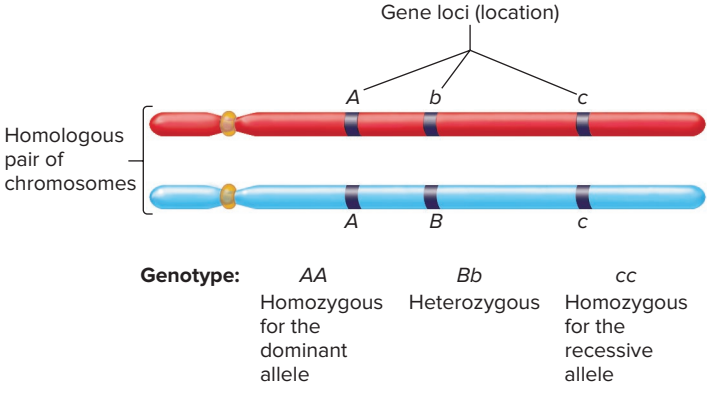


FIGURE 2.3 **A comparison of homologous chromosomes.** Each of the homologous chromosomes in a pair carries the same types of genes, but, as shown here, the alleles may or may not be different.

Concept Check: How are homologs similar to each other, and how are they different?

2.1 COMPREHENSION QUESTIONS

1. Which of the following is *not* found in a prokaryotic cell?
   a. Plasma membrane
   b. Ribosome
   c. Cell nucleus
   d. Cytoplasm
2. When a karyotype is prepared, which of the following steps is carried out?
   a. Treat the cells with a chemical that causes them to begin cell division.
   b. Treat the cells with a hypotonic solution that causes them to swell.
   c. Expose the cells to chemical dyes that bind to the chromosomes and stain them.
   d. All of the above steps are carried out.
3. How many sets of chromosomes are found in a human somatic cell, and how many chromosomes are within one set?
   a. 2 sets, with 23 in each set
   b. 23 sets, with 2 in each set
   c. 1 set, with 23 in each set
   d. 23 sets, with 1 in each set

2.2 CELL DIVISION

Learning Outcomes:

1. Describe the process of binary fission in bacteria.
2. Outline the phases of the eukaryotic cell cycle.

Now that we have an appreciation for the chromosomal composition of living cells, we can consider how chromosomes are copied and transmitted when cells divide. One purpose of cell division is **asexual reproduction**. In this process, a preexisting cell divides to produce two new cells. By convention, the original cell is usually called the mother cell, and the two new cells are the daughter cells. When species are unicellular, the mother cell is judged to be one individual, and the two daughter cells are two new separate organisms. Asexual reproduction is how bacterial cells proliferate. In addition, certain unicellular eukaryotes, such as the amoeba and baker's yeast (*Saccharomyces cerevisiae*), can reproduce asexually.

Another purpose of cell division is to achieve **multicellularity**. Species such as plants, animals, most fungi, and some protists are derived from a single cell that has undergone repeated cell divisions. Humans, for example, begin as a single fertilized egg; repeated cell divisions produce an adult with many trillions of cells. The precise transmission of chromosomes during every cell division is critical so that all cells of the body receive the correct amount of genetic material.

In this section, we will consider how the process of cell division requires the duplication, organization, and distribution of the chromosomes. In bacteria, which have a single circular chromosome, the division process is relatively simple. Prior to cell division, bacteria duplicate their circular chromosome; they then distribute a copy into each of the two daughter cells. This process, known as binary fission, is described first. Eukaryotes have multiple numbers of

chromosomes that occur as sets. This added complexity requires a more complicated sorting process, called mitosis, so that each newly made cell receives the correct number and types of chromosomes. In this section, we will examine how eukaryotic cell division follows a series of stages called the cell cycle.

Bacteria Reproduce Asexually by Binary Fission

As discussed earlier (see Figure 2.1a), bacterial species are typically unicellular, although individual bacteria may associate with each other to form pairs, chains, or clumps. Unlike eukaryotes, which have their chromosomes in a separate nucleus, the circular chromosome of a bacterium is in direct contact with the cytoplasm. In Chapter 12, we will consider the molecular structure of bacterial chromosomes in greater detail.

The capacity of bacteria to divide is really quite astounding. Some species, such as *Escherichia coli* (*E. coli*), a common species of the intestine, can divide every 20 to 30 minutes. As shown in **Figure 2.4**, bacteria reproduce by a process called **binary fission**.

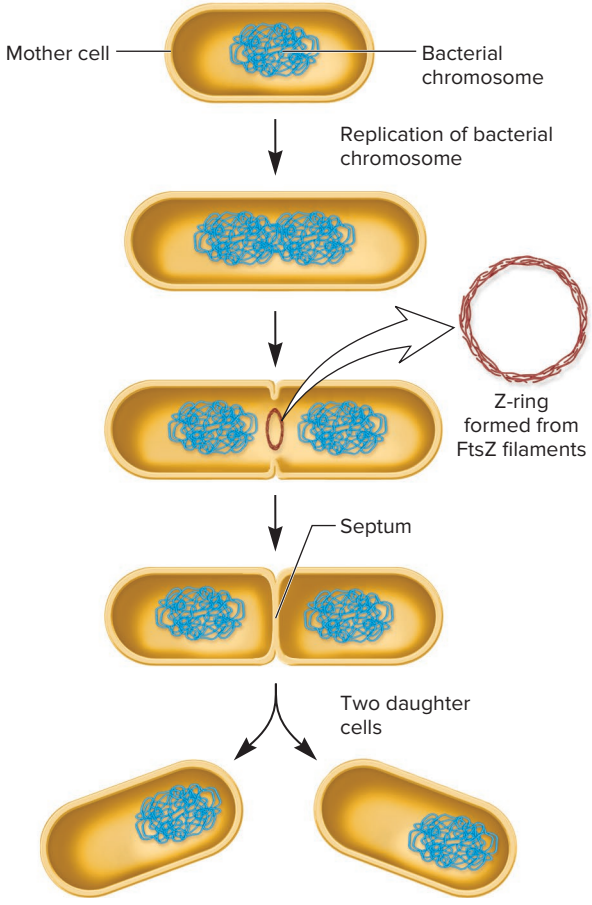


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FIGURE 2.4 Binary fission: the process by which bacterial cells divide. Prior to division, the chromosome replicates to produce two identical copies. These two copies segregate from each other, with one copy going to each daughter cell.

Concept Check: What is the function of the FtsZ protein during binary fission?

1. Prior to cell division, bacterial cells copy, or replicate, their chromosomal DNA. This produces two identical copies of the genetic material, as shown at the top of Figure 2.4.
2. A protein called FtsZ is the first protein to move to the division site that will separate the two daughter cells. Filaments composed of FtsZ protein assemble into a structure called a Z-ring (see inset to Figure 2.4).
3. FtsZ recruits other proteins to produce a septum, which is a new cell wall between the daughter cells. The filaments within the Z-ring are thought to pull on each other and tighten to promote the formation of a septum. FtsZ is evolutionarily related to a eukaryotic protein called tubulin. As discussed later in this chapter, tubulin is the main component of microtubules, which play a key role in chromosome sorting in eukaryotes. Both FtsZ and tubulin form structures that provide cells with organization and play key roles in cell division.
4. As a result of binary fission, a bacterial cell called the mother cell has divided into two daughter cells. Each daughter cell receives a copy of the chromosomal genetic material. Except when rare mutations occur, the daughter cells are usually genetically identical because they contain exact copies of the genetic material from the mother cell.

Binary fission is an asexual form of reproduction because it does not involve genetic contributions from two different gametes. On occasion, bacteria can exchange small pieces of genetic material with each other. We will consider some interesting mechanisms of such genetic exchange in Chapter 9.

Eukaryotic Cells Advance Through a Cell Cycle to Produce Genetically Identical Daughter Cells

The common outcome of eukaryotic cell division is to produce two daughter cells that have the same number and types of chromosomes as the original mother cell. This requires a replication and division process that is more complicated than simple binary fission. Eukaryotic cells that are destined to divide advance through a series of phases known as the **cell cycle** (**Figure 2.5**). These phases are named G for gap, S for synthesis (of the genetic material), and M for mitosis. There are two G phases: G_1 and G_2. The term *gap* originally described the gaps between S phase and mitosis in which it was not microscopically apparent that significant changes were occurring in the cell. However, we now know that both gap phases are critical periods in the cell cycle that involve many molecular changes. In actively dividing cells, the G_1, S, and G_2 phases are collectively known as **interphase**.

In addition, cells may remain permanently, or for long periods of time, in a phase of the cell cycle called G_0. A cell in the G_0 phase is either temporarily not advancing through the cell cycle or, in the case of terminally differentiated cells, such as most nerve cells in an adult mammal, will never divide again. In other words, the G_0 phase is a nondividing stage.

Let's consider the key steps in these four phases.

1. During the **G_1 phase**, a cell may prepare to divide. Depending on the cell type and the conditions it encounters, a cell in the G_1 phase may accumulate molecular changes and reach a

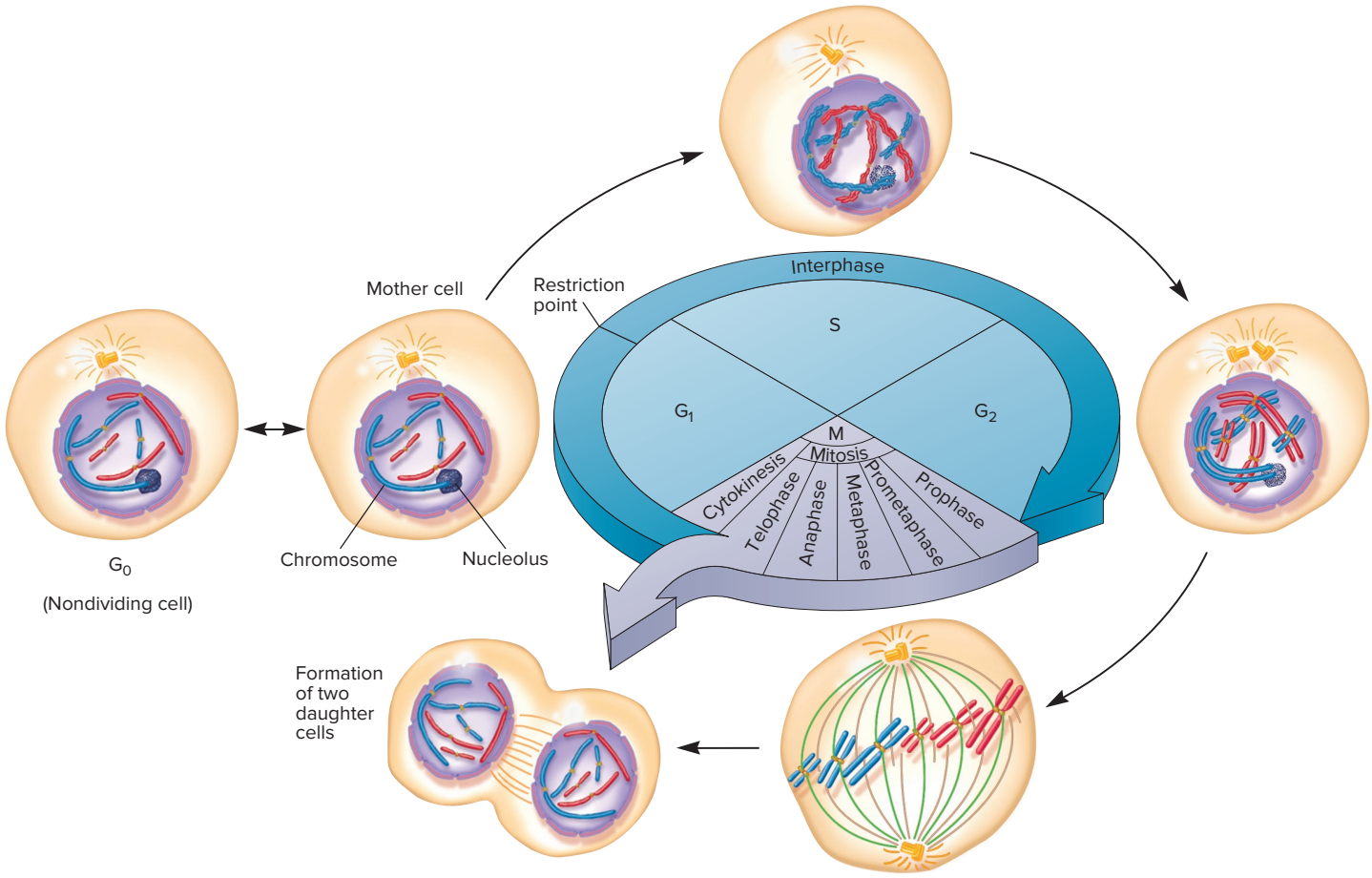


FIGURE 2.5 **The eukaryotic cell cycle.** Dividing cells advance through a series of phases, denoted G_1, S, G_2, and M phases. This diagram shows the advancement of a cell through mitosis to produce two daughter cells. The original mother cell had three pairs of chromosomes, for a total of six individual chromosomes. By the G_2 phase, these have replicated to yield 12 chromatids found in six pairs of sister chromatids. After mitosis and cytokinesis are completed, each of the two daughter cells contains six individual chromosomes, just like the mother cell. Note: The chromosomes in G_0, G_1, S, and G_2 phases are not condensed (look ahead to Figure 2.8a). In this drawing, they are shown partially condensed so they can be easily counted.

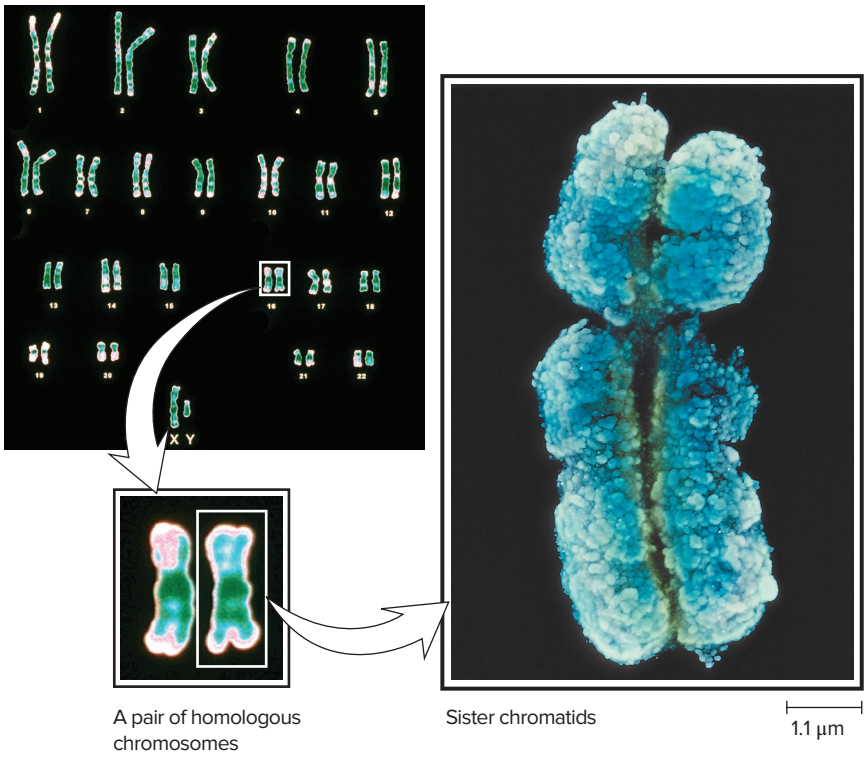
Concept Check: *What is the difference between the G_0 and G_1 phases?*

restriction point and thereby be committed to a pathway that leads to cell division. After a cell has reached a restriction point, it will advance through the remainder of G_1, and then proceed through the S, G_2, and M phases to complete the cell cycle.

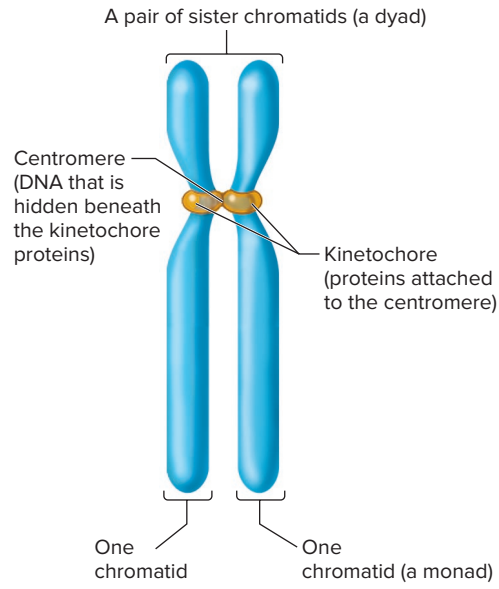
2. Once past the restriction point, the cell then advances to the **S phase**, during which the chromosomes are replicated. After replication, the two copies are called **chromatids**. They are joined to each other at a region of DNA called the **centromere** to form a unit known as a pair of **sister chromatids**, or a **dyad** (**Figure 2.6**). A single chromatid within a dyad is called a **monad**. An unreplicated chromosome can also be called a monad. When S phase is completed, a cell has twice as many chromatids as it had chromosomes in the G_1 phase. The **kinetochore** is a group of proteins that are bound to the centromere. These proteins help to hold the sister chromatids together and also play a role in chromosome sorting, as discussed later.
3. During the **G_2 phase**, the cell accumulates the materials that are necessary for nuclear and cell division.
4. Finally, the cell advances into the **M phase** of the cell cycle, when **mitosis** occurs. The primary purpose of mitosis is to distribute the replicated chromosomes, dividing one cell nucleus into two nuclei, so that each daughter cell receives the same complement of chromosomes. For example, a human cell in the G_2 phase has 92 chromatids, which are found in 46 pairs. During mitosis, these pairs of chromatids are separated and sorted in such a way that each daughter cell receives 46 chromosomes.
5. Two daughter cells are formed by a process called cytokinesis.

2.2 REVIEWING THE KEY CONCEPTS

- Bacteria divide by binary fission (see Figure 2.4).
- To divide, eukaryotic cells advance through a cell cycle (see Figure 2.5).
- Prior to cell division, eukaryotic chromosomes are replicated to form sister chromatids (see Figure 2.6).



(a) Homologous chromosomes and sister chromatids



(b) Schematic drawing of sister chromatids

FIGURE 2.6 **Chromosomes following DNA replication. (a)** The photomicrograph at the upper left shows a human karyotype. The photomicrograph on the right shows a chromosome in the form called a dyad, or a pair of sister chromatids. This chromosome is in the metaphase stage of mitosis, which is described later in the chapter. Note: Each of the 46 chromosomes that are viewed in a human karyotype (upper left) is actually a pair of sister chromatids. Look closely at the two insets. **(b)** A schematic drawing of sister chromatids. This structure has two chromatids that lie side by side. As seen here, each chromatid is a distinct unit. The two chromatids are held together by kinetochore proteins that bind to each other and to the centromere of each chromatid.

Concept Check: *What is the difference between homologs and the chromatids within a pair of sister chromatids?*

2.2 COMPREHENSION QUESTIONS

1. Binary fission
   a. is a form of asexual reproduction.
   b. is a way for bacteria to reproduce.
   c. begins with a single mother cell and produces two genetically identical daughter cells.
   d. All of the above are true of binary fission.
2. Which of the following is the correct order of phases of the eukaryotic cell cycle?
   a. G_1, G_2, S, M
   b. G_1, S, G_2, M
   c. G_1, G_2, M, S
   d. G_1, S, M, G_2
3. What critical event occurs during S phase of the eukaryotic cell cycle?
   a. The cell either prepares to divide or commits to not dividing.
   b. DNA replication produces pairs of sister chromatids.
   c. The chromosomes condense.
   d. The single nucleus is divided into two nuclei.

2.3 MITOSIS AND CYTOKINESIS

Learning Outcomes:

1. Describe the structure and function of the mitotic spindle apparatus.
2. List and describe the phases of mitosis.
3. Outline the key differences between animal and plant cells with regard to cytokinesis.

As we have seen, eukaryotic cell division involves a cell cycle in which the chromosomes are replicated and then sorted so that each daughter cell receives the same amount of genetic material. This process ensures genetic consistency from one cell generation to the next. In this section, we will examine the stages of mitosis and cytokinesis in greater detail.

The Mitotic Spindle Apparatus Organizes and Sorts Eukaryotic Chromosomes

Before we discuss the events of mitosis, let's first consider the structure of the **mitotic spindle apparatus** (also known simply as

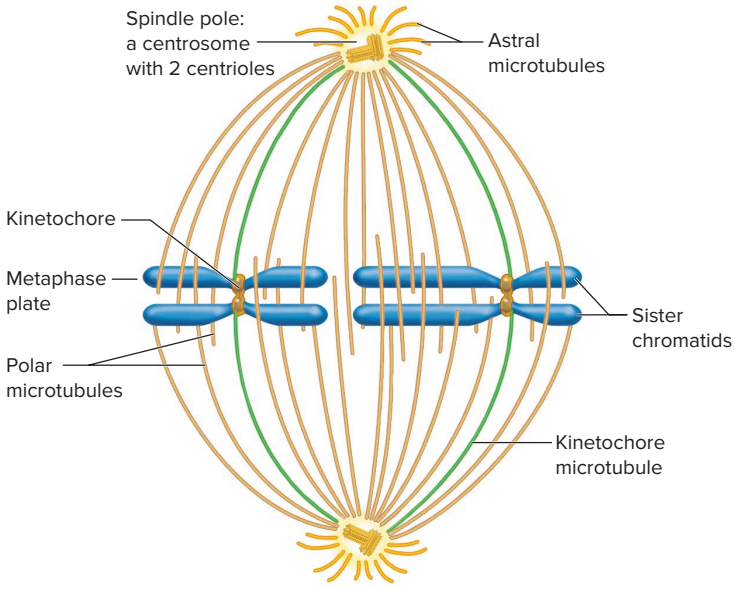


FIGURE 2.7 The structure of the spindle apparatus in a typical animal cell. A single centrosome duplicates during S phase, and the two centrosomes separate at the beginning of M phase. The spindle apparatus is formed from microtubules that are rooted in the centrosomes. Each centrosome is located at a spindle pole. The astral microtubules emanate away from the region between the poles. They help position the spindle apparatus within the cell. However, astral microtubules are not found in many species, such as plants. The polar microtubules project into the region between the two poles; they play a role in pole separation. The kinetochore microtubules are attached to the kinetochore of sister chromatids.

Concept Check: Where are the two ends of a kinetochore microtubule?

the **spindle apparatus** or the **mitotic spindle**), which is involved in the organization and sorting of chromosomes (**Figure 2.7**). The spindle apparatus is formed from **microtubule-organizing centers (MTOCs)**, which are structures found in eukaryotic cells from which microtubules grow. Microtubules are produced from the rapid polymerization of tubulin proteins. In animal cells, the spindle apparatus is formed from two MTOCs called **centrosomes**. Each centrosome is located at a **spindle pole**. A pair of **centrioles** at right angles to each other is found within each centrosome. Centrosomes and centrioles are found in animal cells but not in all eukaryotic species. For example, plant cells do not have centrosomes. Instead, the nuclear envelope functions as an MTOC for the formation of the spindle apparatus in plant cells.

The spindle apparatus of a typical animal cell has three types of microtubules (see Figure 2.7).

- The astral microtubules emanate outward from the centrosome toward the plasma membrane. They are important for the positioning of the spindle apparatus within the cell.
- The polar microtubules project toward the region where the chromosomes are found during mitosis—the region between the two spindle poles. Polar microtubules that overlap with each other play a role in the separation of the two poles. They help to "push" the poles away from each other.
- The kinetochore microtubules have attachments to kinetochores, which are protein complexes bound to the centromeres of individual chromosomes.

The spindle apparatus allows cells to organize and separate chromosomes so that each daughter cell receives the same complement of chromosomes. This sorting process, known as mitosis, is described next.

The Transmission of Chromosomes During the Division of Eukaryotic Cells Involves a Process Known as Mitosis

In **Figure 2.8**, the process of mitosis is shown for a diploid animal cell. In the simplified diagrams below the micrographs in the figure, the original mother cell contains six chromosomes; it is diploid ($2n$) and has three chromosomes per set ($n = 3$). One set is shown in blue, and the homologous set is red. As discussed next, mitosis is subdivided into phases known as prophase, prometaphase, metaphase, anaphase, and telophase.

Prophase Prior to mitosis, the cells are in interphase, during which the chromosomes are **decondensed**—less tightly compacted—and found in the nucleus (Figure 2.8a). At the start of mitosis, in **prophase**, the chromosomes have already replicated to produce 12 chromatids that are joined as six pairs of sister chromatids (Figure 2.8b). As prophase proceeds, the nuclear membrane begins to dissociate into small vesicles. At the same time, the chromatids **condense** into more compact structures that are readily visible by light microscopy. The spindle apparatus also begins to form, and the nucleolus, which is the site of ribosome assembly, disappears.

Prometaphase As mitosis advances from prophase to prometaphase, the centrosomes move to opposite ends of the cell and establish two spindle poles, one within each of the future daughter cells. Once the nuclear membrane has completely fragmented into vesicles, the spindle fibers can interact with the sister chromatids. This interaction occurs in a phase of mitosis called **prometaphase** (Figure 2.8c). How do sister chromatids become attached to the spindle apparatus? Initially, microtubules form rapidly and can be seen growing out from the two poles. As a microtubule grows, if its end happens to make contact with a kinetochore, the end is said to be captured and remains firmly attached to the kinetochore. This random process is how sister chromatids become attached to kinetochore microtubules. Alternatively, if the end of a microtubule does not collide with a kinetochore, the microtubule eventually depolymerizes and retracts to the centrosome. As the end of prometaphase nears, the kinetochore on a pair of sister chromatids is attached to kinetochore microtubules from opposite poles. As these events are occurring, the sister chromatids undergo jerky movements as they are tugged, back and forth, between the two poles. By the end of prometaphase, the spindle apparatus is completely formed.

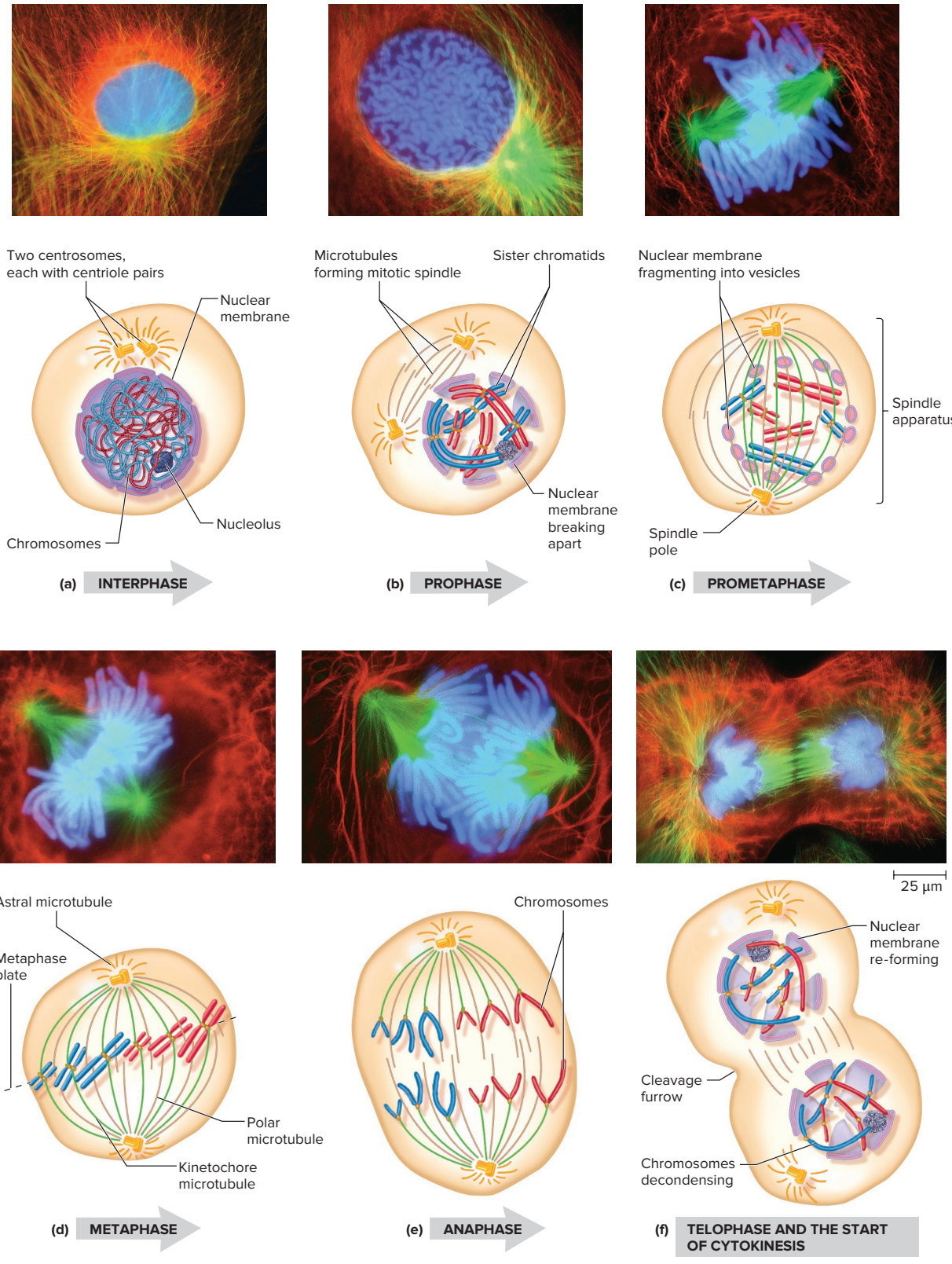


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FIGURE 2.8 **The process of mitosis in an animal cell.** The two rows of micrographs illustrate cells of a fish embryo advancing through mitosis. The chromosomes are stained in blue and the microtubules are green. Below the micrographs are schematic diagrams that emphasize the sorting and separation of the chromosomes. In the diagrams, the original diploid cell is shown with six chromosomes (three in each set). At the start of mitosis, these have already replicated into 12 chromatids. After cytokinesis is completed, the final result is two daughter cells, each containing six chromosomes.

(a-f) Photomicrographs by Dr. Conly L. Rieder, Wadsworth Center, Albany, New York 12201-0509

Concept Check: *During which phase of mitosis are sister chromatids separated and sent to opposite poles?*

Metaphase Eventually, the pairs of sister chromatids align themselves along a plane called the **metaphase plate**. As shown in Figure 2.8d, when this alignment is complete, the cell is in **metaphase** of mitosis. At this point, each pair of chromatids (each dyad) is attached to both poles by kinetochore microtubules. The pairs of sister chromatids have become organized into a single row along the metaphase plate. When this organizational process is finished, the chromatids can be equally distributed into two daughter cells.

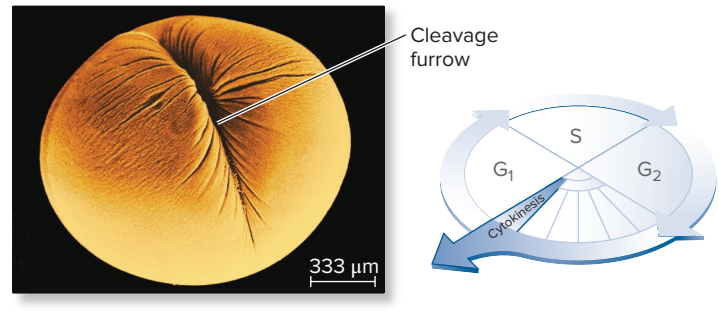
Anaphase At **anaphase**, the connection that is responsible for holding each pair of chromatids together is broken (Figure 2.8e). Each chromatid, or monad, now an individual chromosome, is linked to only one of the two poles. As anaphase proceeds, the chromosomes move toward the pole to which they are attached. This movement is due to the shortening of the kinetochore microtubules. In addition, the two poles themselves move farther apart due to the elongation of the polar microtubules, which slide in opposite directions as a result of the actions of motor proteins.

Telophase During **telophase**, the chromosomes reach their respective poles and decondense. The nuclear membrane now reforms to produce two separate nuclei. In Figure 2.8f, this process has produced two nuclei that contain six chromosomes each. The nucleoli have also reappeared.

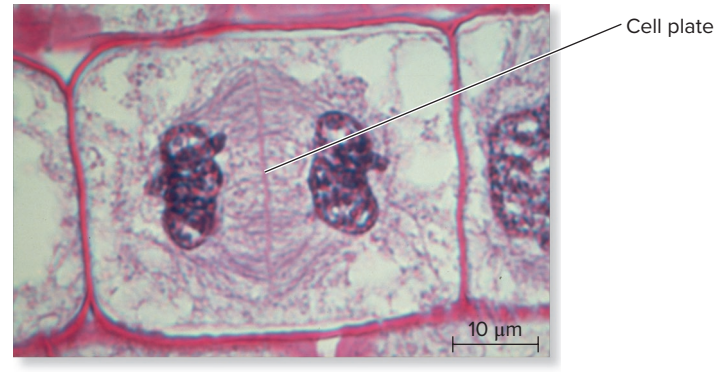
Cytokinesis In most cases, mitosis is quickly followed by **cytokinesis**, in which the two nuclei are segregated into separate daughter cells. Likewise, cytokinesis also segregates cell organelles such as mitochondria and chloroplasts into daughter cells. In animal cells, cytokinesis begins shortly after anaphase. A contractile ring, composed of myosin motor proteins and actin filaments, assembles adjacent to the plasma membrane. Myosin hydrolyzes ATP, which shortens the ring and thereby constricts the plasma membrane to form a **cleavage furrow** that ingresses, or moves inward (**Figure 2.9a**). Ingression continues until a midbody structure is formed that physically pinches the single cell into two.

In plants, the two daughter cells are separated by the formation of a **cell plate** (**Figure 2.9b**). At the end of anaphase, Golgi-derived vesicles carrying cell wall materials are transported to the equator of a dividing cell. These vesicles are directed to their locations via microtubules and actin filaments that serve as tracks for vesicle movement. The fusion of these vesicles gives rise to the cell plate, which is a membrane-bound compartment. The cell plate begins in the middle of the cell and expands until it attaches to the mother cell's wall. Once this attachment has taken place, the cell plate undergoes a process of maturation and eventually separates the mother cell into two daughter cells.

Outcome of Mitotic Cell Division Mitosis and cytokinesis ultimately produce two daughter cells having the same number of chromosomes as the mother cell. Barring rare mutations, the two daughter cells are genetically identical to each other and to the mother cell from which they were derived. The critical consequence of this sorting process is to ensure genetic consistency from one generation of somatic cells to the next. The development of multicellularity relies on the repeated process of mitosis and cytokinesis. In diploid organisms that are multicellular, most of the somatic cells are diploid and genetically identical to each other.



(a) Cleavage of an animal cell



(b) Formation of a cell plate in a plant cell

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FIGURE 2.9 Cytokinesis in animal and plant cells. **(a)** In an animal cell, cytokinesis involves the formation of a cleavage furrow. **(b)** In a plant cell, cytokinesis occurs via the formation of a cell plate between the two daughter cells.

(a) © Don W. Fawcett/Science Source; (b) ©Ed Reschke

Concept Check: What causes the cleavage furrow in a dividing animal cell to ingress?

Genetic TIPS

The Question: What are the functional roles of the spindle apparatus in an animal cell? Explain how these functions are related to the three types of microtubules: astral, polar, and kinetochore microtubules.

Topic: What topic in genetics does this question address?

The topic is mitosis. More specifically, the question is about the role of the spindle apparatus.

Information: What information do you know based on the question and your understanding of the topic?

From the question, you know there are three types of microtubules. From your understanding of the topic, you may remember the structure of the spindle apparatus, which is

shown in Figure 2.7. Also, Figure 2.8 shows the roles that the spindle apparatus plays during mitosis.

Problem-Solving Strategy: Define key terms. Describe the steps.

One strategy to begin solving this problem is to make sure you understand the key terms. In particular, you may want to look up the meaning of *spindle apparatus* and *microtubules*, if you don't already know what those terms mean. After you understand the key terms, another useful problem-solving strategy is to describe the steps of mitosis, and think about the roles of the types of microtubules in the various steps. These steps are shown in Figure 2.8. You may also want to refer back to Figure 2.7 to appreciate the structure of the spindle apparatus.

Answer:

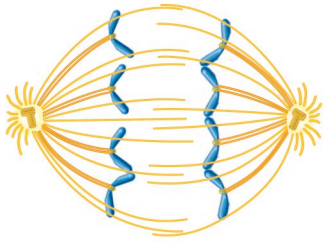
- The spindle apparatus is involved in sorting the chromosomes and promoting the division of one cell into two daughter cells.
- The polar microtubules overlap with each other and push the poles apart during anaphase.
- The astral microtubules help to orient the spindle apparatus in the cell.
- The kinetochore microtubules attach to chromosomes and aid in their sorting. Their roles are to align the chromosomes at the metaphase plate and to pull the chromosomes to the poles during anaphase.

2.3 REVIEWING THE KEY CONCEPTS

- Chromosome sorting in eukaryotes is achieved via the mitotic spindle apparatus (see Figure 2.7).
- A common way for eukaryotic cells to divide is by mitosis and cytokinesis. Mitosis is divided into prophase, prometaphase, metaphase, anaphase, and telophase. During cytokinesis, animal cells divide by forming a cleavage furrow and plant cells form a cell plate (see Figures 2.8 and 2.9).

2.3 COMPREHENSION QUESTIONS

1. During which phase of mitosis does the nuclear envelop re-form?
   a. Prometaphase
   b. Metaphase
   c. Anaphase
   d. Telophase
2. Which phase of mitosis is depicted in the drawing below?



   a. Prophase
   b. Prometaphase
   c. Metaphase
   d. Anaphase
   e. Telophase

2.4 MEIOSIS

Learning Outcomes:

1. List and describe the phases of meiosis.
2. Outline the key differences between mitosis and meiosis.

In the previous section, we considered the process in which a eukaryotic cell can divide by mitosis and cytokinesis so that a mother cell produces two genetically identical daughter cells. Diploid eukaryotic cells may also divide by an alternative process called **meiosis** (from the Greek meaning "less"). During meiosis, **haploid** cells, which contain a single set of chromosomes, are produced from a cell that was originally diploid. For this to occur, the chromosomes must be correctly sorted and distributed in a way that reduces the chromosome number to half its original value. In the case of humans, for example, each gamete must receive half of the total number of 46 chromosomes. But not just any 23 chromosomes will do—a gamete must receive one chromosome from each of the 23 pairs. In this section, we examine how the phases of meiosis lead to the formation of cells with a haploid complement of chromosomes.

Meiosis Produces Cells That Are Haploid

The process of meiosis bears striking similarities to mitosis. Like mitosis, meiosis begins after a cell has advanced through the G_1, S, and G_2 phases of the cell cycle. However, meiosis involves two successive divisions rather than one (as in mitosis). Prior to meiosis, the chromosomes are replicated in S phase to produce pairs of sister chromatids. This single replication event is then followed by two sequential cell divisions called meiosis I and II. As in mitosis, each of these divisions is subdivided into prophase, prometaphase, metaphase, anaphase, and telophase.

Prophase of Meiosis I **Figure 2.10** emphasizes some of the important events that occur during prophase of meiosis I, which is further subdivided into stages known as leptotene, zygotene, pachytene, diplotene, and diakinesis.

1. During the **leptotene** stage, the replicated chromosomes begin to condense and become visible with a light microscope.
2. Unlike prophase of mitosis, the **zygotene** stage of prophase of meiosis I involves a recognition process known as **synapsis**, in which the homologous chromosomes recognize each other and begin to align themselves along their entire lengths. As this occurs, a **synaptonemal complex** forms between the homologs. This complex is thought to promote the binding of homologs to each other. However, the synaptonemal complex may not be required for the pairing of homologous chromosomes, because some species, such as *Aspergillus nidulans* and *Schizosaccharomyces pombe*, completely lack such a complex, yet their chromosomes synapse correctly.
3. At **pachytene**, the homologs have become completely aligned. The associated chromatids are known as **bivalents**. Each bivalent contains two pairs of sister chromatids, or a total of four chromatids. A bivalent is also called a **tetrad** (from the prefix *tetra-*, meaning "four") because it is composed of four chromatids—that is, four monads.

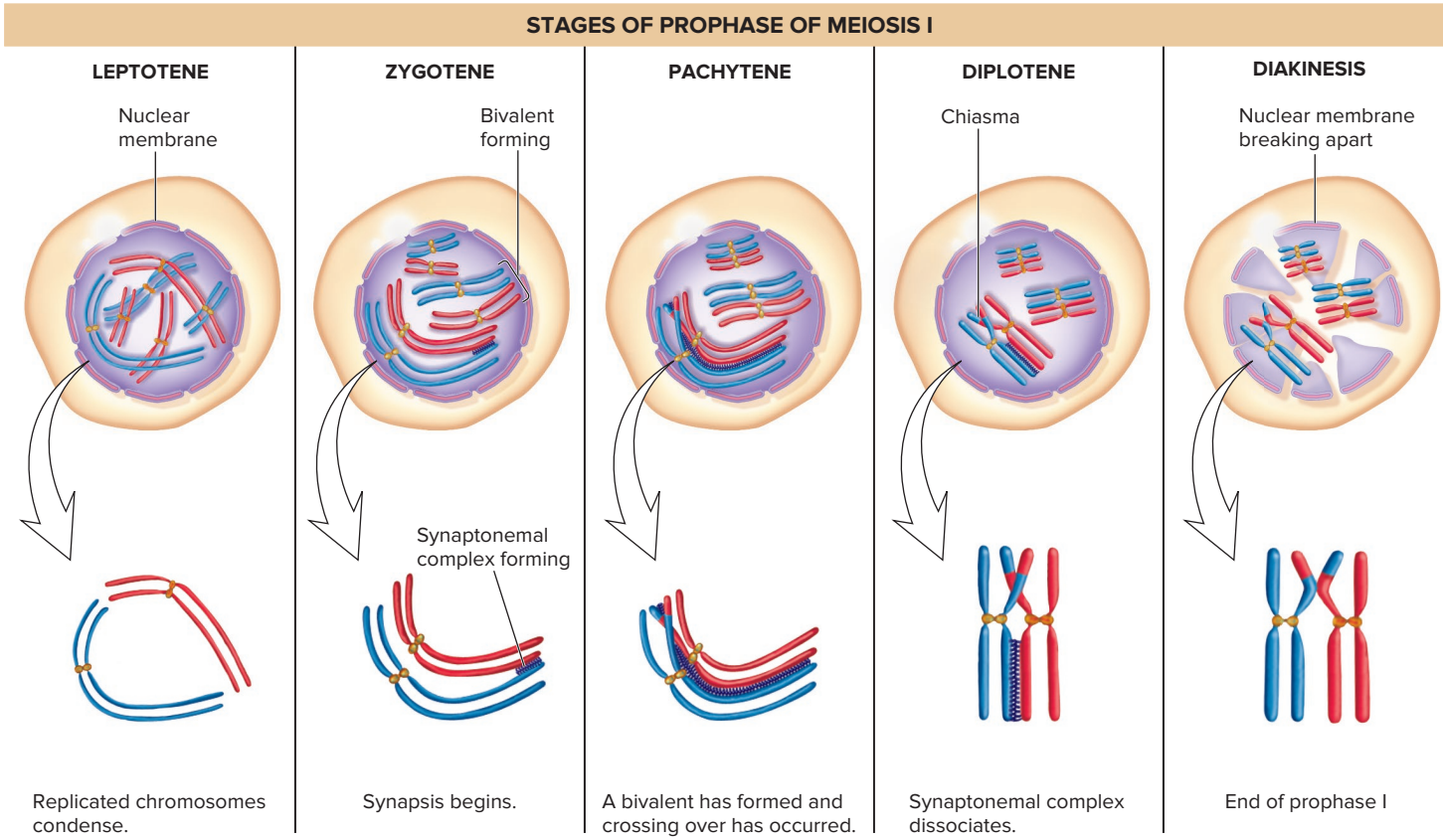


FIGURE 2.10 **The events that occur during prophase of meiosis I.** Note: For simplicity, the nucleolus is not shown in this figure.

Concept Check: *What is the end result of crossing over?*

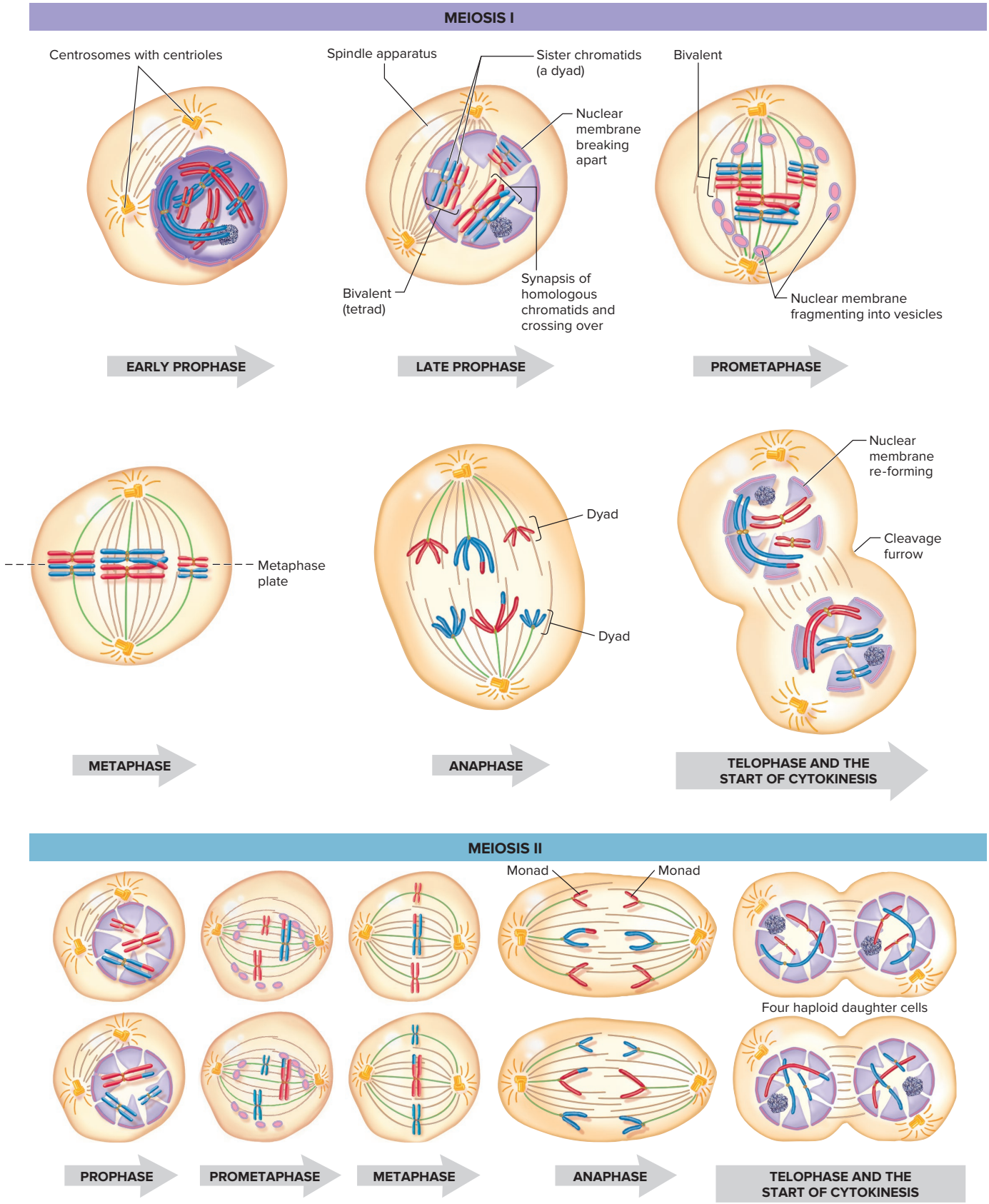
Prior to the pachytene stage, when synapsis is complete, an event known as crossing over usually occurs. **Crossing over** involves a physical exchange of chromosome pieces that results in an exchange of genetic information. Depending on the size of the chromosome and the species, an average eukaryotic chromosome incurs from a couple to a couple of dozen crossovers. During spermatogenesis in humans, for example, an average chromosome undergoes slightly more than 2 crossovers, whereas chromosomes in certain plant species may undergo 20 or more crossovers. In Figure 2.10, crossing over has occurred at a single site between two of the larger chromatids. The connection that results from crossing over is called a **chiasma** (plural: **chiasmata**), because it physically resembles the Greek letter chi, χ. We will consider the genetic consequences of crossing over in Chapter 7 and the molecular process of crossing over in Chapter 13.

4. By the end of the **diplotene** stage, the synaptonemal complex has largely disappeared. The chromatids within a bivalent pull apart slightly, and it becomes easier to see under a microscope that a bivalent is actually composed of four chromatids.
5. In the last stage of prophase of meiosis I, **diakinesis**, the synaptonemal complex completely disappears.

Prometaphase of Meiosis I Figure 2.10 emphasizes the pairing and crossing over that occur during prophase of meiosis I. In **Figure 2.11**, we turn our attention to the general events in meiosis. Prophase of meiosis I is followed by prometaphase, in which the spindle apparatus is completely formed and the chromatids are attached via kinetochore microtubules.

Metaphase of Meiosis I At metaphase of meiosis I, the bivalents are organized along the metaphase plate. However, their pattern of alignment is strikingly different from that observed during mitosis (refer back to Figure 2.8d). Before we consider the rest of meiosis I, a particularly critical feature for you to appreciate is how the bivalents are aligned along the metaphase plate. In particular, the pairs of sister chromatids (the dyads) are aligned in a double row rather than a single row, as occurs in mitosis. Furthermore, the arrangement of sister chromatids within this double row is random with regard to the blue and red homologs. In Figure 2.11, one of the blue homologs is above the metaphase plate and the other two are below, whereas one of the red homologs is below the metaphase plate and other two are above.

In an organism that produces many gametes, meiosis can produce a different arrangement of homologs in various cells—for example, three blues above and none below, or none above and three below, and so on. Because most eukaryotic species have several chromosomes per set, the sister chromatids can be randomly aligned along the metaphase plate in many possible ways. Let's consider humans, who have 23 chromosomes per set. The possible number of different random alignments equals 2^n, where n is the number of chromosomes per set. Thus, in humans, there are 2^{23},



ONLINE ANIMATION

FIGURE 2.11 **The stages of meiosis in an animal cell.** See text for details.

Concept Check: *How do the four cells at the end of meiosis differ from the original mother cell?*

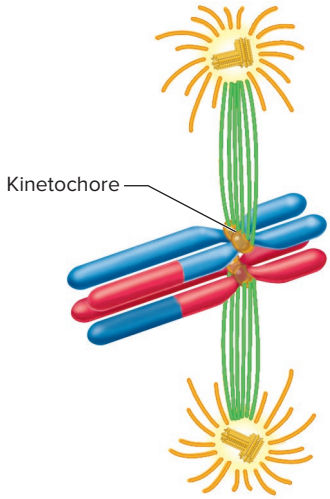


FIGURE 2.12 Attachment of the kinetochore microtubules to replicated chromosomes during meiosis. The kinetochore microtubules from a given pole are attached to one pair of chromatids in a bivalent, but not both. Therefore, each pair of sister chromatids is attached to only one pole.

Concept Check: How is the attachment of chromosomes to kinetochore microtubules during metaphase of meiosis I different from their attachment during metaphase of mitosis?

or over 8 million, possibilities! Because the homologs are genetically similar but not identical, we see from this calculation that the random alignment of homologous chromosomes provides a mechanism to promote a vast amount of genetic diversity.

In addition to the random arrangement of homologs within a double row, a second distinctive feature of metaphase of meiosis I is the attachment of kinetochore microtubules to the sister chromatids (**Figure 2.12**). One pair of sister chromatids is linked to one of the poles, and the homologous pair is linked to the opposite pole. This arrangement is quite different from the kinetochore attachment sites during mitosis, in which a pair of sister chromatids is linked to both poles (see Figure 2.8d).

Anaphase of Meiosis I During anaphase of meiosis I, the two pairs of sister chromatids within a bivalent separate from each other (see Figure 2.11). However, the connection that holds sister chromatids together does not break. Instead, each joined pair of chromatids migrates to one pole, and the homologous pair of chromatids moves to the opposite pole. Another way of saying this is that the two dyads within a tetrad separate from each other and migrate to opposite poles.

Telophase of Meiosis I and Cytokinesis Finally, at telophase of meiosis I, the sister chromatids have reached their respective poles, and decondensation occurs in many, but not all, species. The nuclear membrane may re-form to produce two separate nuclei. In the example shown in Figure 2.11, the end result of meiosis I is two cells, each with three pairs of sister chromatids. A reduction division has occurred. The original diploid cell had its chromosomes in homologous pairs, but the two cells produced at the end of meiosis I are considered to be haploid; they do not have pairs of homologous chromosomes.

Meiosis II The sorting events that occur during meiosis II are similar to those that occur during mitosis, but the starting point is different. For a diploid organism with six chromosomes, mitosis begins with 12 chromatids that are joined as six pairs of sister chromatids (refer back to Figure 2.8). In other words, mitosis begins with six dyads in this case. By comparison, the two cells that begin meiosis II each have six chromatids that are joined as three pairs of sister chromatids; meiosis II begins with three dyads. Otherwise, the steps that occur during prophase, prometaphase, metaphase, anaphase, and telophase of meiosis II are analogous to a mitotic division.

Meiosis versus Mitosis If we compare the outcome of meiosis (see Figure 2.11) to that of mitosis (see Figure 2.8), the results are quite different. In the examples we examined, mitosis produced two diploid daughter cells with six chromosomes each, whereas meiosis produced four haploid daughter cells with three chromosomes each. In other words, meiosis halved the number of chromosomes per cell. **Table 2.1** compares the key differences among mitosis, meiosis I, and meiosis II.

With regard to alleles, the results of mitosis and meiosis are also different. The daughter cells produced by mitosis are genetically identical. However, the haploid cells produced by meiosis are not genetically identical to each other because they contain only one homologous chromosome from each pair. In Chapter 3, we will consider how gametes may differ in the alleles that they carry on their homologous chromosomes.

TABLE 2.1

A Comparison of Mitosis, Meiosis I, and Meiosis II

Phase	Event	Mitosis	Meiosis I	Meiosis II
Prophase	Synapsis	No	Yes	No
Prophase	Crossing over	Rarely	Commonly	Rarely
Prometaphase	Attachment to the poles	A pair of sister chromatids to both poles	A pair of sister chromatids to one pole	A pair of sister chromatids to both poles
Metaphase	Alignment along the metaphase plate	Sister chromatids	Bivalents	Sister chromatids
Anaphase	Separation of:	Sister chromatids	Bivalents	Sister chromatids
End result		Two diploid cells		Four haploid cells

Genetic TIPS

The Question: If a diploid cell contains four chromosomes (i.e., two per set), how many possible random arrangements of homologs can occur during metaphase of meiosis I?

Topic: What topic in genetics does this question address?

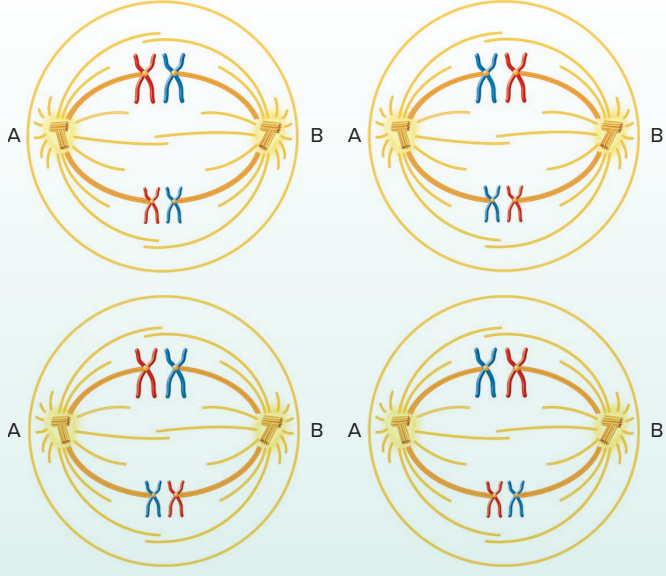
The topic is meiosis. More specifically, the question is about metaphase of meiosis I.

Information: What information do you know based on the question and your understanding of the topic?

From the question, you know that a cell that started with two pairs of homologous chromosomes has entered meiosis and is now in metaphase of meiosis I. From your understanding of the topic, you may remember that bivalents align along the metaphase plate (see Figure 2.11). The orientations of the homologs within the bivalents are random.

Problem-Solving Strategies: Make a drawing. Make a calculation.

One strategy to solve this problem is to make a drawing in which the homologs are different colors, such as red and blue. Note: The spindle poles are labeled A and B in the drawing below. The alignment occurs relative to the spindle poles.



Another strategy is to make a calculation in which the number of different random alignments equals 2^n, where n is the number of chromosomes per set.

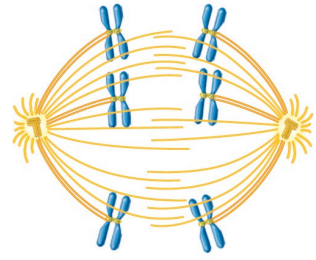
Answer: As seen in the drawing, the number of random alignments is 4. From a calculation, the number of different random alignments equals 2^n. So the possible number of arrangements in this case is 2^2, which is 4.

2.4 REVIEWING THE KEY CONCEPTS

- Another way for eukaryotic cells to divide is via meiosis, which produces four haploid cells. During prophase of meiosis I, homologs synapse, and crossing over may occur (see Figures 2.10–2.12, Table 2.1).

2.4 COMPREHENSION QUESTIONS

1. When does crossing over usually occur, and what is the end result?
   a. It occurs during prophase of meiosis I, and the end result is the exchange of pieces between homologous chromosomes.
   b. It occurs during prometaphase of meiosis I, and the end result is the exchange of pieces between homologous chromosomes.
   c. It occurs during prophase of meiosis I, and the end result is the separation of sister chromatids.
   d. It occurs during prometaphase of meiosis I, and the end result is the separation of sister chromatids.
2. Which phase of meiosis is depicted in the following drawing?



   a. Metaphase of meiosis I
   b. Metaphase of meiosis II
   c. Anaphase of meiosis I
   d. Anaphase of meiosis II

2.5 SEXUAL REPRODUCTION

Learning Outcomes:

1. Define *sexual reproduction.*
2. Describe how animals make sperm and egg cells.
3. Explain how plants alternate between haploid and diploid generations.

In the previous section, we considered how a diploid cell divides by meiosis to produce cells with half the genetic material of the original mother cell. This process is critical for sexual reproduction, which is a common way for eukaryotic organisms to produce offspring. During **sexual reproduction**, gametes are made that contain half the amount of an organism's genetic material. These gametes fuse with each other in the process of fertilization to begin the life of a new organism.

Gametes are highly specialized cells that are produced by a process called **gametogenesis**. As discussed previously, gametes are typically haploid, which means they contain half the number of chromosomes found in diploid cells. Haploid cells are represented by 1*n* and diploid cells by 2*n*, where *n* refers to a set of chromosomes. A haploid gamete contains a single set of chromosomes, whereas a diploid cell has two sets. For example, a diploid human cell contains two sets of chromosomes, for a total of 46, but a human gamete (sperm or egg cell) contains only a single set of 23 chromosomes.

Some simple eukaryotic species are **isogamous**, which means that the gametes are morphologically similar. Examples of isogamous organisms include many species of fungi and algae. Most eukaryotic species, however, are **heterogamous**—they produce two morphologically different types of gametes. Male gametes, or