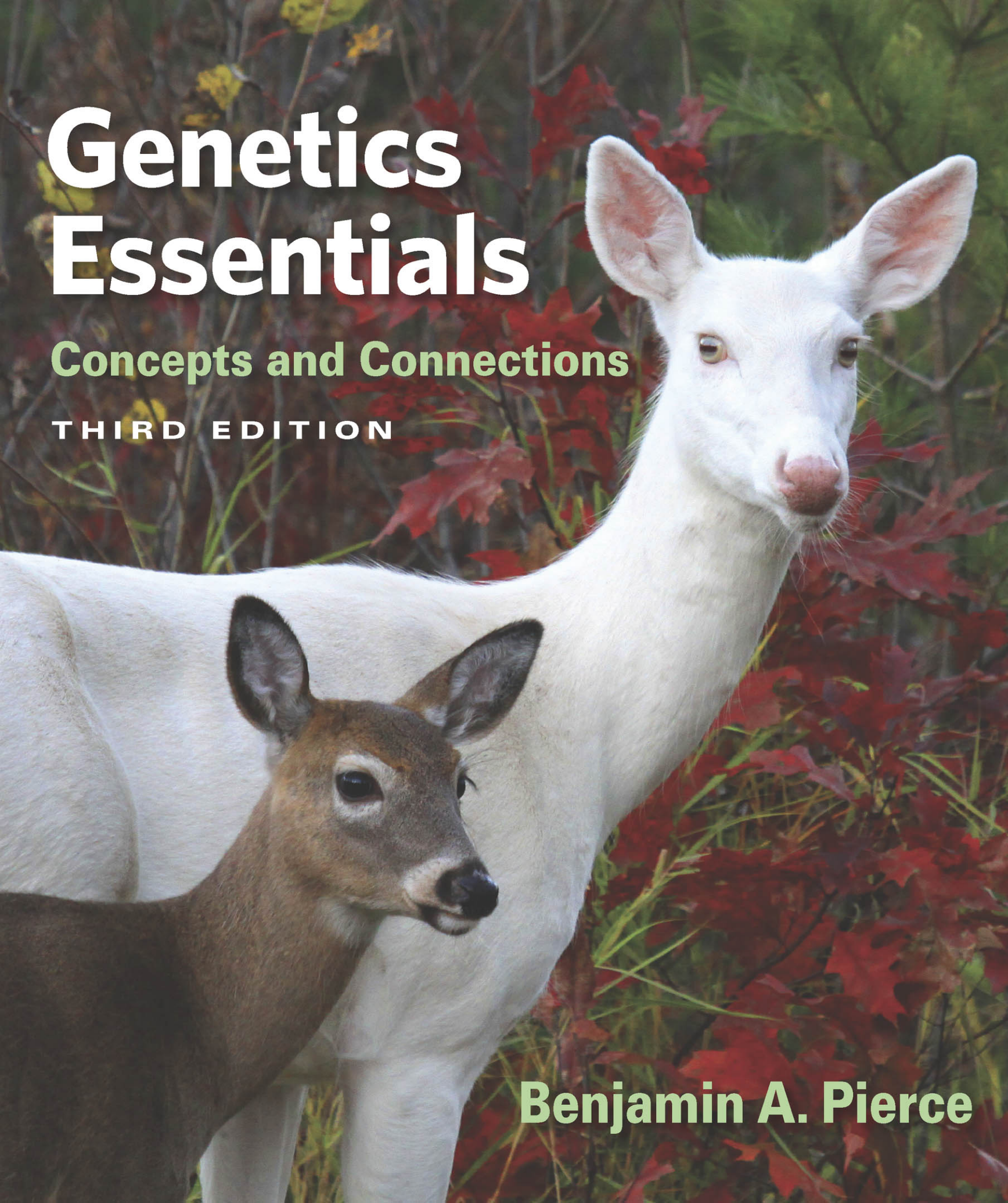
Genetics
Essentials
Concepts and Connections
THIRD EDITION
Benjamin A. Pierce

Genetics Essentials

Concepts and Connections

Benjamin A. Pierce

Southwestern University

W. H. Freeman & Company

A Macmillan Education Imprint

New York

Publisher: Kate Ahr Parker
Senior Acquisitions Editor: Lauren Schultz
Developmental Editor: Anna Bristow
Media and Supplements Editor: Anna Bristow
Editorial Assistant: Shannon Moloney
Marketing Assistant: Bailey James
Project Editor: J. Carey Publishing Service
Manuscript Editor: Norma Sims Roche
Director of Design, Content Management: Diana Blume
Cover and Text Design: Jerry Wilke Design
Illustrations: Dragonfly Media Group
Illustration Coordinator: Janice Donnola
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Photo Researcher: Christine Buese and Eileen Liang
Production Coordinator: Lisa McDowell
Composition: codeMantra
Printing and Binding: RR Donnelley/Willard
Front Cover/Title Page Photo: Michael Crowley/Getty Images

Library of Congress Control Number: 2014950827

ISBN-13: 978-1-4641-9075-9
ISBN-10: 1-4641-9075-5

Printed in the United States of America

First printing

W. H. Freeman and Company
41 Madison Avenue
New York, NY 10010

www.whfreeman.com

To the students who enroll in my genetics class each year and continually inspire me with their intelligence, curiosity, and enthusiasm

Brief Contents

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Letter from the Author

Marlene Tyrrell

Genetics is among the most exciting and important biology courses that you will take. Almost daily, we are bombarded with examples of the relevance of genetics: the discovery of genes that influence human diseases, traits, and behaviors; the use of DNA testing to trace disease transmission and to solve crimes; the use of genetic technology to develop new products. And today, genetics is particularly important to the student of biology, serving as the foundation for many biological concepts and processes. It is truly a great time to be learning genetics!

Although genetics is important and relevant, mastering the subject is a significant challenge for many students. The field encompasses complex processes and is filled with detailed information. Genetics is often the first biology course in which students must develop problem-solving skills and apply what they have learned to novel situations.

My goal as author of your textbook is to help you overcome these challenges and excel at genetics. As we make our journey together through introductory genetics, I'll share what I've learned in my 35 years of teaching genetics, give you advice and encouragement, motivate you with stories of the people, places, and experiments of genetics, and keep our focus on the major concepts.

Genetics Essentials: Concepts and Connections was written in response to requests from instructors and students for a more streamlined and focused genetics textbook that covers less content than a full-length genetics textbook. It has as its foundation my more comprehensive *Genetics: A Conceptual Approach*, which is now in its fifth edition.

At Southwestern University, my office door is always open, and my own students frequently drop by to share their own approaches to learning as well as their experiences, concerns, and triumphs. I would love to hear from you—by email (pierceb@southwestern.edu), by telephone (512-863-1974), or in person (Southwestern University, Georgetown, Texas).

Ben Pierce
Professor of Biology
and holder of the Lillian Nelson Pratt Chair,
Southwestern University

Preface

Welcome to the third edition of *Genetics Essentials: Concepts and Connections*, a brief genetics textbook designed specifically for your one-semester course. The title *Genetics Essentials: Concepts and Connections* precisely conveys the major goals of the book: to help students uncover major concepts of genetics and make connections among those concepts so as to have a fuller understanding of genetics. The text maintains the features that made the first two editions so effective: simple and instructive illustrations, accessible writing style, a strong emphasis on problem solving, and useful pedagogical features throughout the book.

Hallmark Features

- **Key Concepts and Connections** Throughout the book, I've included pedagogical devices to help students focus on the major concepts of each topic.
  - **Concepts boxes** throughout each chapter summarize the key points of the preceding section. ***Concept Check*** questions allow students to quickly assess their understanding of the material they've just read. Concept Checks are in multiple-choice or short-answer format, and answers are listed at the end of each chapter.
  - **Connecting Concepts** sections compare and contrast processes or integrate ideas across sections and chapters to help students see how different genetics topics relate to one another. All major concepts are listed in the ***Concepts Summary*** at the end of each chapter.

CONCEPTS

The Punnett square is a shorthand method of predicting the genotypic and phenotypic ratios of progeny from a genetic cross.

✓ CONCEPT CHECK 4

If the F_1 plant depicted in Figure 3.4 is backcrossed to the parent with round seeds, what proportion of the progeny will have wrinkled seeds? (Use a Punnett square.)

a. $^3/_4$ b. $^1/_2$
c. $^1/_4$ d. 0

- **Accessibility** I have intentionally used a friendly and conversational writing style so that students will find the book inviting and informative. The **introductory stories** at the beginning of every chapter draw students into the material. These stories highlight the relevance of genetics to daily life and feature new research in genetics, the genetic basis of human disease, hereditary oddities, and other interesting topics. Four of the eighteen chapters have new introductory stories in this edition.
- **Clear, Simple Illustrations** The attractive and instructive illustrations continue to play a pivotal role in reinforcing the key concepts presented in each chapter. Because many students are visual learners, I have worked closely with the illustrators to make sure that the main point of each illustration is easily identified and understood. Most include narratives that take students through a process step by step or that point out important features of a structure or experiment. Throughout the book there are illustrations that facilitate a student's understanding of the experimental process by posing a question, describing experimental methodology, presenting results, and drawing a conclusion that reinforces the major concept being addressed.
- **Emphasis on Problem Solving** I believe that problem solving is essential to the mastery of genetics. It is also among the most difficult skills for a student to learn. **Newly reformatted Worked Problems** (see **New and Reorganized Content** on p. xvi) walk students through a key problem and review important strategies to consider when tackling a problem of a similar type. **Try Problem** links point to end-of-chapter problems that students can work to test their understanding of the material just read, all with answers in the back of the book so students can check their results. At the end of each

CONDITIONAL PROBABILITY Thus far, we have used probability to predict the chances of producing certain types of progeny given only the genotypes of the parents. Sometimes we have additional information that modifies, or "conditions," the probability, a situation termed **conditional probability**. For example, assume that we cross two heterozygous pea plants (*Tt* × *Tt*) and obtain a tall offspring plant. What is the probability that this tall plant is heterozygous (*Tt*)? You might assume that the probability would be $^1/_2$, the probability of obtaining a heterozygous offspring in a cross between two heterozygotes. In this case, however, we have some additional information—the phenotype of the offspring plant—which modifies that probability. When two heterozygous individuals are crossed, we expect $^1/_4$ *TT*, $^1/_2$ *Tt*, and $^1/_4$ *tt* progeny. We know that the offspring in question is tall, so we can eliminate the possibility that it has genotype *tt*. Tall progeny must be either genotype *TT* or genotype *Tt*, and in a cross between two heterozygotes, these genotypes occur in a 1:2 ratio. Therefore, the probability that a tall offspring plant is heterozygous (*Tt*) is two out of three, or $^2/_3$.
▶TRY PROBLEMS 24 and 25

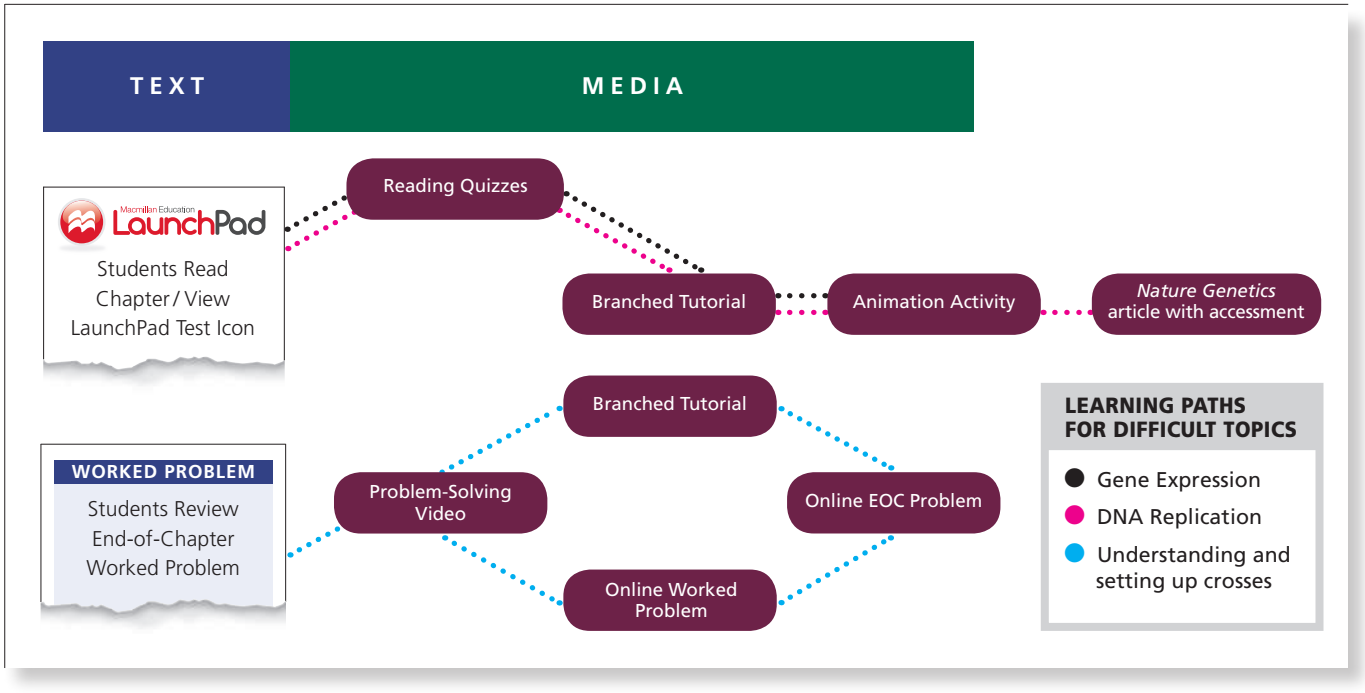
chapter are a wide range of questions and problems, organized by chapter section and subdivided into Comprehension Questions, Application Questions and Problems, and Challenge Questions. Some of these questions draw on examples from published papers and are marked by a data analysis icon.

- **Streamlined Content** To provide students taking a brief genetics course with the most important concepts, I've shortened the book considerably. The third edition of *Genetics Essentials: Concepts and Connections* is about 250 pages shorter than *Genetics: A Conceptual Approach*, a reduction of more than 35%.

New to the Third Edition

The third edition builds on successful features of the first two editions of *Genetics Essentials: Concepts and Connections* and provides an up-to-date look at the field.

- **Connected Learning Paths for Core Topics** At the heart of this revision is the connection between the book and its media. The media resources were developed alongside the text in order to create a connected learning path from the book throughout the media package. Through extensive reviewing we identified the genetics topics and skills that students find most difficult. We then designed our media to include the types of resources that instructors felt would best help students understand these topics, study, and practice problem solving—a key skill that is crucial to understanding genetics. To give you an idea of how our media resources connect with the book, here are a few examples of the learning paths we visualized for two of the topics that are consistently noted by instructors as difficult for their students.



- **New Problem-Solving Videos** offer students valuable help by reviewing basic problem-solving strategies. The problem-solving videos demonstrate an instructor working through problems that students find difficult in a step-by-step manner.
- **New Online Branched Tutorials** identify where students have difficulty and route them through the steps of the problem according to the answers they enter. Hints allow students to receive feedback along the way, as if they were working the problem with an instructor.

- **New Online Worked Problems** allow students to step through a worked problem online in the same format that they've seen in the book. Hints provide feedback as they work through the problem.
- **Updated and New Animations/Simulations** help students understand key processes in genetics by outlining them in a step-by-step manner. The animations and simulations now include assessment questions at the end to help students evaluate whether they understood the concept or technique they viewed.
- **Comprehensively Revised Assessment** All media resources have undergone extensive rewriting, reviewing, and accuracy checking.
  - **Online End-of-Chapter Problems** include nearly all of the end-of-chapter problems from the text, converted to a self-graded multiple-choice and drop-down menu format for quizzing, homework, or student practice.
  - **LearningCurve** allows students to test their comprehension of the chapter concepts with questions similar to the Comprehension Questions at the end of each chapter. The system adapts to each student's individual level of preparedness by giving them questions at varying levels of difficulty depending on whether they answer a question without any help, if they need help but eventually get the question right, or if they are unable to answer the question. Links to the appropriate e-Book section, hints, and feedback help students realize where they need more practice on a topic.
  - **New Online Reading Quizzes** test whether students have comprehended the basic concepts in the chapter they've just read.
  - **The Test Bank** contains at least 50 multiple-choice and short-answer questions per chapter.
- ***Nature Genetics* Articles with Assessment** engage students with primary research and encourage critical thinking. Specifically selected for both alignment with text coverage and exploration of identified difficult topics, the *Nature Genetics* articles include assessment questions that can be automatically graded. Some of the open-ended (non-multiple-choice) questions are also suitable for use in flipped classrooms and active learning discussions either in class or online.

All of the new media resources for *Genetics: A Conceptual Approach* will be available in our new LaunchPad system. **LaunchPad** is a dynamic, fully integrated learning environment that brings together all of our teaching and learning resources in one place. It also contains the fully interactive **e-Book** and other newly updated resources for students and instructors, including the following:

- **Newly Updated Clicker Questions** allow instructors to integrate active learning into the classroom and to assess students' understanding of key concepts during lectures. Available in Microsoft Word and PowerPoint, numerous questions are based on the Concept Check questions featured in the textbook.
- **New Lecture PPTs** have been developed to minimize preparation time for new users of the book. These files offer suggested lectures, including key illustrations and summaries that instructors can adapt to their teaching styles.
- **Textbook Images and Tables** are offered as high-resolution JPEG files. Each image has been fully optimized to increase type sizes and adjust color saturation. These images have been tested in a large lecture hall to ensure maximum clarity and visibility.
- The ***Solutions and Problem-Solving Manual*** (written by Jung H. Choi, Georgia Institute of Technology, and Mark E. McCallum, Pfeiffer University) contains complete answers and worked-out solutions to all questions and problems in the textbook. The manual has been reviewed for accuracy and has been found to be an indispensable tool for success by students. The *Solutions Manual* is also available in print (ISBN: 1-4641-5092-3).

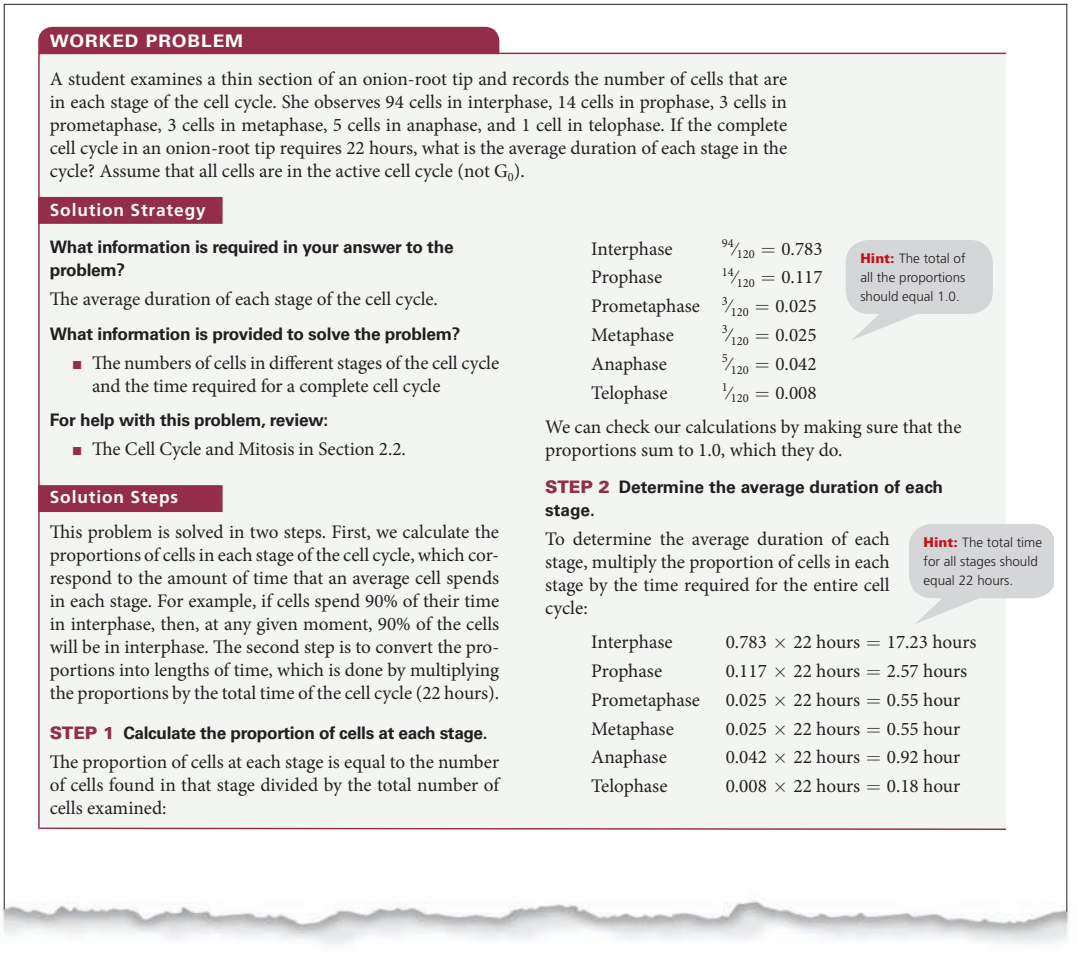
New and Reorganized Content

The third edition addresses recent discoveries in genetics corresponding to our ever-changing understanding of inheritance, the molecular nature of genetic information, epigenetics, and genetic evolution.

NEW Epigenetics Content The coverage of **epigenetics** has been significantly updated and expanded in **Chapter 12**. Briefer discussions of epigenetics are now in Chapters 4, 8, and 16.

Other new and updated content includes the following:

- New molecular genetics section: "The Molecular Nature of Alleles" (Chapter 3)
- New section on conditional probability (Chapter 3)
- Extensively revised section on dosage compensation (Chapter 4)
- Significantly expanded section on chromosome rearrangements (Chapter 6)
- Expanded discussion of the discovery of DNA structure, including Franklin's contribution (Chapter 8)
- New section on CRISPR RNAs (Chapter 10)
- New section: "Long Noncoding RNAs Regulate Gene Expression" (Chapter 10)
- Updated and expanded discussion of changes in chromatin structure (Chapter 12)
- New section on Illumina sequencing (Chapter 14)
- Revised section on DNA fingerprinting (Chapter 14)
- Updated and expanded discussion of genome-wide association studies (Chapter 15)
- Updated discussion of metagenomics (Chapter 15)
- New section on evolution through changes in gene regulation (Chapter 18)



WORKED PROBLEM

A student examines a thin section of an onion-root tip and records the number of cells that are in each stage of the cell cycle. She observes 94 cells in interphase, 14 cells in prophase, 3 cells in prometaphase, 3 cells in metaphase, 5 cells in anaphase, and 1 cell in telophase. If the complete cell cycle in an onion-root tip requires 22 hours, what is the average duration of each stage in the cycle? Assume that all cells are in the active cell cycle (not G_0).

Solution Strategy

What information is required in your answer to the problem?

The average duration of each stage of the cell cycle.

What information is provided to solve the problem?

- The numbers of cells in different stages of the cell cycle and the time required for a complete cell cycle

For help with this problem, review:

- The Cell Cycle and Mitosis in Section 2.2.

Solution Steps

This problem is solved in two steps. First, we calculate the proportions of cells in each stage of the cell cycle, which correspond to the amount of time that an average cell spends in each stage. For example, if cells spend 90% of their time in interphase, then, at any given moment, 90% of the cells will be in interphase. The second step is to convert the proportions into lengths of time, which is done by multiplying the proportions by the total time of the cell cycle (22 hours).

STEP 1 Calculate the proportion of cells at each stage.

The proportion of cells at each stage is equal to the number of cells found in that stage divided by the total number of cells examined:

Interphase	$^{94}/_{120} = 0.783$
Prophase	$^{14}/_{120} = 0.117$
Prometaphase	$^{3}/_{120} = 0.025$
Metaphase	$^{3}/_{120} = 0.025$
Anaphase	$^{5}/_{120} = 0.042$
Telophase	$^{1}/_{120} = 0.008$

Hint: The total of all the proportions should equal 1.0.

We can check our calculations by making sure that the proportions sum to 1.0, which they do.

STEP 2 Determine the average duration of each stage.

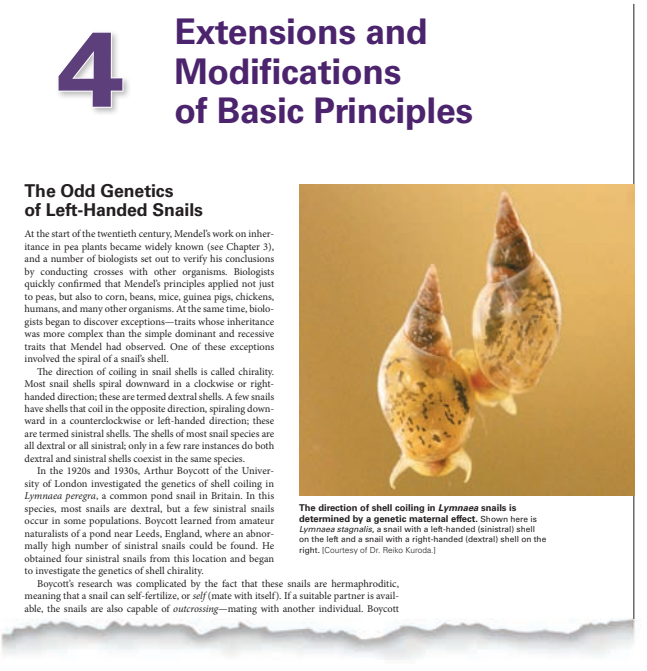
To determine the average duration of each stage, multiply the proportion of cells in each stage by the time required for the entire cell cycle:

Hint: The total time for all stages should equal 22 hours.

Interphase	$0.783 \times 22 \text{ hours} = 17.23 \text{ hours}$
Prophase	$0.117 \times 22 \text{ hours} = 2.57 \text{ hours}$
Prometaphase	$0.025 \times 22 \text{ hours} = 0.55 \text{ hour}$
Metaphase	$0.025 \times 22 \text{ hours} = 0.55 \text{ hour}$
Anaphase	$0.042 \times 22 \text{ hours} = 0.92 \text{ hour}$
Telophase	$0.008 \times 22 \text{ hours} = 0.18 \text{ hour}$

- **Reformatted Worked Problems** walk students through difficult quantitative concepts in an easy to follow manner, presenting a Solution Strategy and Solution Steps for each worked problem. Hint balloons remind students of key points to keep in mind or refer them back to a specific part of the text to review.

- **NEW Figure-Based End-of-Chapter Problems** bring a visual aspect to problem solving, engaging students closely with figures in each chapter and helping them assess their understanding of key concepts and processes.
- **NEW Introductory Stories** Each chapter begins with a brief **introductory story** that illustrates the relevance of a genetic concept that students will learn in the chapter. These stories—a favorite feature of past editions—give students a glimpse of what's going on in the field of genetics today and help to draw them into the chapter. Among the new introductory story topics are "The Odd Genetics of Left-Handed Snails" and "Building a Better Banana." End-of-chapter problems specifically address concepts discussed in most introductory stories, both old and new.



4 Extensions and Modifications of Basic Principles

The Odd Genetics of Left-Handed Snails

At the start of the twentieth century, Mendel's work on inheritance in pea plants became widely known (see Chapter 3), and a number of biologists set out to verify his conclusions by conducting crosses with other organisms. Biologists quickly confirmed that Mendel's principles applied not just to peas, but also to corn, beans, mice, guinea pigs, chickens, humans, and many other organisms. At the same time, biologists began to discover exceptions—traits whose inheritance was more complex than the simple dominant and recessive traits that Mendel had observed. One of these exceptions involved the spiral of a snail's shell.

The direction of coiling in snail shells is called chirality. Most snail shells spiral downward in a clockwise or right-handed direction; these are termed dextral shells. A few snails have shells that coil in the opposite direction, spiraling downward in a counterclockwise or left-handed direction; these are termed sinistral shells. The shells of most snail species are all dextral or all sinistral; only in a few rare instances do both dextral and sinistral shells coexist in the same species.

In the 1920s and 1930s, Arthur Boycott of the University of London investigated the genetics of shell coiling in *Lymnaea peregra*, a common pond snail in Britain. In this species, most snails are dextral, but a few sinistral snails occur in some populations. Boycott learned from amateur naturalists of a pond near Leeds, England, where an abnormally high number of sinistral snails could be found. He obtained four sinistral snails from this location and began to investigate the genetics of shell chirality.

Boycott's research was complicated by the fact that these snails are hermaphroditic, meaning that a snail can self-fertilize, or *self* (mate with itself). If a suitable partner is available, the snails are also capable of *outcrossing*—mating with another individual. Boycott

The direction of shell coiling in *Lymnaea* snails is determined by a genetic maternal effect. Shown here is *Lymnaea stagnalis*, a snail with a left-handed (sinistral) shell on the left and a snail with a right-handed (dextral) shell on the right. [Courtesy of Dr. Reiko Kuroda.]

Acknowledgments

Every day as I enter the classroom, I feel a rush of excitement in anticipation of exploring with my students the concepts, knowledge, and questions that constitute the field of genetics. For me, teaching genetics is never dull or routine. The field is constantly changing, with important new advances occurring weekly, and the information is interesting and relevant. But mostly, my pleasure in teaching genetics derives from the students who have filled my classes, whose energy, intelligence, curiosity, and humor have been a source of inspiration and motivation for the past 35 years. I have also learned from students worldwide who have used earlier editions of this book and kindly shared with me—through emails and phone calls—their thoughts about the book and how it could be improved. I thank my own teachers, Dr. Raymond Canham and Dr. Jeffrey Mitton, for introducing me to genetics and showing me how to teach and do scholarship.

I am indebted to Southwestern University for providing a stimulating environment in which quality teaching and research flourish. My daily life is greatly enriched by my colleagues in the Biology Department, who sustain me with their friendship, collegiality, and advice. I am grateful to President Edward Burger and Provost James Hunt for helping to create this supportive academic environment and for their friendship and encouragement.

The creation of modern science textbooks is a team effort, and I have been blessed to work with an outstanding team at W. H. Freeman and Company. Publisher Susan Winslow has been the guiding force for the development of this third edition of *Genetics Essentials*. Lauren Schultz, Senior Acquisitions Editor for the Life Sciences team, was a wonderful shepherd of the project, providing ideas, encouragement, and support throughout the project. I had the pleasure of working with Developmental Editors Anna Bristow and Lisa Samols, whose positive attitudes, superb organizational skills, and attention to detail made this edition a success.

Norma Sims Roche served as manuscript editor; she provided many valuable editorial suggestions and ensured that the prose was clear and engaging. Project Editor Jennifer Carey expertly managed the production of this third edition. I thank Dragonfly Media Group for creating and revising the book's illustrations and Matthew McAdams for coordinating the illustration program. Thanks to Lisa McDowell at W. H. Freeman for coordinating the composition and manufacturing phases of production. Diana Blume developed the book's design and the cover for this edition. I thank Christine Buese and Eileen Liang

for photo research. Anna Bristow coordinated the development of the excellent media and supplements that accompany the book, assisted by Shannon Moloney and Nandini Ahuja. Joseph Ahlander, Ellen France, Robert Fowler, Idit Hazan, Brian Kreiser, Joshua Loomis, Amy McMillan, Marcie Moehnke, Brian C. Ring, Douglas Thrower, Lauren Tranter, and Daniel Williams developed and reviewed assessment questions.

I extend special thanks to the W. H. Freeman sales representatives, regional managers, and regional sales specialists. To know and work with this group of professionals has been a pleasure and privilege. Ultimately, their hard work and good service account for the success of Freeman books.

A number of colleagues served as reviewers of the textbook, kindly lending me their technical expertise and teaching experience. Their assistance is gratefully acknowledged; any remaining errors are entirely my responsibility.

Marlene Tyrrell, my spouse and best friend for 35 years, and our children and their spouses—Sarah, Matt, Michael, and Amber—provide love, support, and inspiration for everything I do.

My gratitude goes to the reviewers of this new edition of *Genetics Essentials:*

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1 Introduction to Genetics

Albinism among the Hopis

Rising a thousand feet above the desert floor, Black Mesa dominates the horizon of the Enchanted Desert and provides a familiar landmark for travelers passing through northeastern Arizona. Black Mesa is not only a prominent geological feature: more significantly, it is the ancestral home of the Hopi Native Americans. Fingers of the mesa reach out into the desert, and alongside or on top of each finger is a Hopi village. Most of the villages are quite small, with only a few dozen inhabitants, but they are incredibly old. One village, Oraibi, has existed on Black Mesa since 1150 A.D. and is the oldest continually occupied settlement in North America.

In 1900, Alěs Hrdliěka, an anthropologist and physician working for the American Museum of Natural History, visited the Hopi villages of Black Mesa and reported a startling discovery. Among the Hopis were 11 white people—not Caucasians, but white Hopi Native Americans. These people had a genetic condition known as albinism (**Figure 1.1**).

A Hopi pueblo on Black Mesa. Albinism, a genetic condition, arises with high frequency among the Hopi people and occupies a special place in the Hopi culture. [Photograph No. 79-AA-S02, "Walpi, Arizona 1941," 1941; Ansel Adams Photographs of National Parks and Monuments. Records of the National Park Service/National Archives at College Park, MD.]

Albinism is caused by a defect in one of the enzymes required to produce melanin, the pigment that darkens our skin, hair, and eyes. People with albinism either don't produce melanin or produce only small amounts of it, and consequently, have white hair, light skin, and no pigment in the irises of their eyes. Melanin normally protects the DNA of skin cells from the damaging effects of ultraviolet radiation in sunlight, and melanin's presence in the developing eye is essential for proper eyesight.

The genetic basis of albinism was first described by the English physician Archibald Garrod, who recognized in 1908 that the condition was inherited as an autosomal recessive trait, meaning that a person must receive two copies of an albino mutation—one from each parent—to have albinism. In recent years, the molecular natures of the mutations that lead to albinism have been elucidated. Albinism in humans is caused by defects in any one of several different genes that control the synthesis and storage of melanin. Many different types of mutations can occur in each gene, any one of which may lead to albinism. The form of albinism found among the Hopis is most likely oculocutaneous albinism (albinism affecting the eyes and skin) type 2, caused by a defect in the *OCA2* gene on chromosome 15.

The Hopis are not unique in having people with albinism among the members of their tribe. Albinism is found in almost all human ethnic groups and is described in ancient writings: it has probably been present since humankind's beginnings. What is

1.1 Albinism among the Hopi Native Americans. The Hopi girl in the middle of this photograph, taken about 1900, has albinism. [©The Field Museum, #CSA118. Charles Carpenter.]

unique about the Hopis is the high frequency of albinism in their population. In most human groups, albinism is rare, present in only about 1 in 20,000 people. In the villages on Black Mesa, it reaches a frequency of 1 in 200, a hundred times greater than in most other populations.

Why is albinism so frequent among the Hopis? The answer to this question is not completely known, but geneticists who have studied albinism among the Hopis speculate that the high frequency of the albino gene is at least partly related to the special place that albinism occupied in the Hopi culture. For much of their history, the Hopis considered members of their tribe with albinism to be important and special. People with albinism were considered pretty, clean, and intelligent. Having a number of people with albinism in one's village was considered a good sign, a symbol that the people of the village contained particularly pure Hopi blood. Members of the tribe with albinism performed in Hopi ceremonies and held positions of leadership, often as chiefs, healers, and religious leaders.

Hopis with albinism were also given special treatment in everyday activities. The Hopis have farmed small garden plots at the foot of Black Mesa for centuries. Every day throughout the growing season, the men of the tribe trek to the base of Black Mesa and spend much of the day in the bright southwestern sunlight tending their corn and vegetables. With little or no melanin in their skin, people with albinism are extremely susceptible to sunburn and have increased incidences of skin cancer when exposed to the sun. Furthermore, many don't see well in bright sunlight. Therefore, male Hopis with albinism were excused from farming and allowed to remain behind in the village with the women of the tribe, performing other duties.

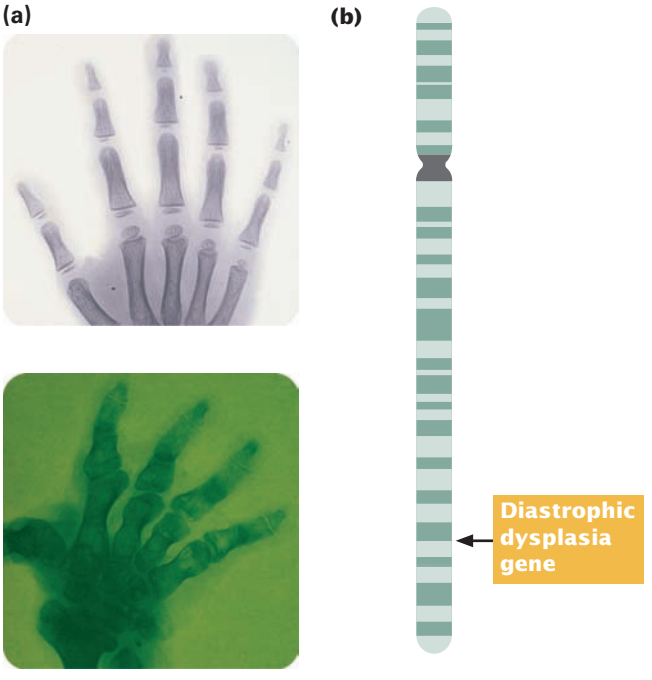
Throughout the growing season, the men with albinism were the only male members of the tribe in the village with the women during the day, and thus they enjoyed a mating advantage, which helped to spread their albino genes. In addition, the special considerations given to Hopis with albinism allowed them to avoid the detrimental effects of albinism: increased skin cancer and poor eyesight. The small size of the Hopi tribe probably also played a role by allowing chance to increase the frequency of the albino gene. Regardless of the factors that led to the high frequency of albinism, the Hopis greatly respected and valued the members of their tribe who possessed this particular trait. Unfortunately, people with genetic conditions in many societies are often subject to discrimination and prejudice. ▶TRY PROBLEMS 1 AND 22

Genetics is one of the most rapidly advancing fields of science, with important new discoveries reported every month. Look at almost any major news source and chances are that you will see articles related to genetics: the completion of another organism's genome, such as that of the monarch butterfly; the discovery of genes that affect major diseases, including multiple sclerosis, depression, and cancer; analyses of DNA from long-extinct animals such as the woolly mammoth; or the identification of genes that affect skin pigmentation, height, or learning ability in humans. Even among advertisements, you are likely to see ads for genetic testing to determine a person's ancestry, paternity, and susceptibility to diseases and disorders. These new findings and applications of genetics often have significant economic and ethical implications, making the study of genetics relevant, timely, and interesting.

This chapter introduces you to genetics and reviews some concepts that you may have encountered briefly in a preceding biology course. We begin by considering the importance of genetics to each of us, to society, and to students of biology. We then turn to the history of genetics and how the field as a whole developed. The final part of the chapter presents some fundamental terms and principles of genetics that are used throughout the book.

1.1 Genetics Is Important to Us Individually, to Society, and to the Study of Biology

Albinism among the Hopis illustrates the important role that genes play in our lives. This one genetic defect, among the 20,000 genes that humans possess, completely changes the life of a Hopi who possesses it. It alters his or her occupation, role in Hopi society, and relations with other members of the tribe. We all possess genes that influence our lives in significant ways. Genes affect our height, weight, hair color, and skin pigmentation. They influence our susceptibility to many diseases and disorders (**Figure 1.2**) and even contribute to

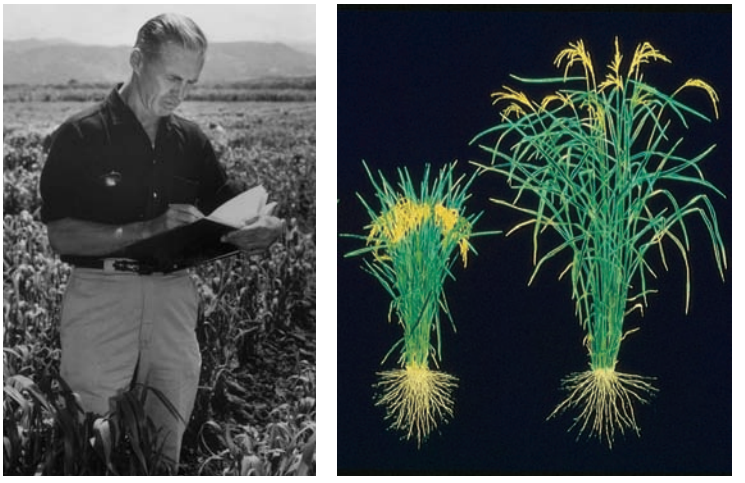


1.2 Genes influence susceptibility to many diseases and disorders. (a) An X-ray of the hand of a person suffering from diastrophic dysplasia (bottom), a hereditary growth disorder that results in curved bones, short limbs, and hand deformities, compared with an X-ray of a normal hand (top). (b) This disorder is due to a defect in the *SLC26A2* gene on chromosome 5. [Part a: (top) Biophoto Associates/Science Source; (bottom) Reprinted from Johanna Hästbacka et al., The diastrophic dysplasia gene encodes a novel sulfate transporter: Positional cloning by fine structure linkage disequilibrium mapping. *Cell* 78(6):1073–1087, 1994. © 1994 Elsevier. Courtesy of Prof. Eric Lander, Whitehead Institute, MIT.]

our intelligence and personality. Genes are fundamental to who and what we are.

Although the science of genetics is relatively new compared with sciences such as astronomy and chemistry, people have understood the hereditary nature of traits and have practiced genetics for thousands of years. The rise of agriculture began when people started to apply genetic principles to the domestication of plants and animals. Today, the major crops and animals used in agriculture are quite different from their wild progenitors, having undergone extensive genetic alterations that increase their yields and provide many desirable traits, such as disease and pest resistance, special nutritional qualities, and characteristics that facilitate harvest. The Green Revolution, which expanded food production throughout the world in the 1950s and 1960s, relied heavily on the application of genetics (**Figure 1.3**). Today, genetically engineered corn, soybeans, and other crops constitute a significant proportion of the food produced worldwide.

The pharmaceutical industry is another area in which genetics plays an important role. Numerous drugs and



1.3 In the Green Revolution, genetic techniques were used to develop new high-yielding strains of crops. (Left) Norman Borlaug, a leader in the development of new varieties of wheat that led to the Green Revolution. Borlaug was awarded the Nobel Peace Prize in 1970. (Right) A modern, high-yielding rice plant (left) and a traditional rice plant (right). [Left: © Bettmann/CORBIS. Right: IRRI.]

food additives are synthesized by fungi and bacteria that have been genetically manipulated to make them efficient producers of these substances. The biotechnology industry employs molecular genetic techniques to develop and mass-produce substances of commercial value. Growth hormone, insulin, clotting factor, enzymes, antibiotics, vaccines, and many drugs are now produced commercially by genetically engineered bacteria and other cells (**Figure 1.4**). Genetics has also been used to produce bacteria that remove minerals from ore, break down toxic chemicals, and inhibit damaging frost formation on crop plants.

1.4 The biotechnology industry uses molecular genetic methods to produce substances of economic value. [© Andrew Brookes/Corbis.]

Genetics also plays a critical role in medicine. Physicians recognize that many diseases and disorders have a hereditary component, including not only rare genetic disorders such as sickle-cell anemia and Huntington disease, but also many common diseases such as asthma, diabetes, and hypertension. Advances in genetics have resulted in important insights into the nature of diseases such as cancer and in the development of diagnostic tests, including those that identify pathogens and defective genes. Gene therapy—the direct alteration of genes to treat human diseases—has now been administered to thousands of patients, although its use is still experimental and limited.

The Role of Genetics in Biology

Although an understanding of genetics is important to all people, it is critical to the student of biology. Genetics provides one of biology's unifying principles: all organisms use genetic systems that have a number of features in common. Genetics also undergirds the study of many other biological disciplines. Evolution, for example, is genetic change that takes place over time, so the study of evolution requires an understanding of genetics. Developmental biology relies heavily on genetics: tissues and organs develop through the regulated expression of genes (**Figure 1.5**). Even such fields as taxonomy, ecology, and animal behavior are making increasing use of genetic methods. The study of almost any field of biology or medicine is incomplete without a thorough understanding of genes and genetic methods.

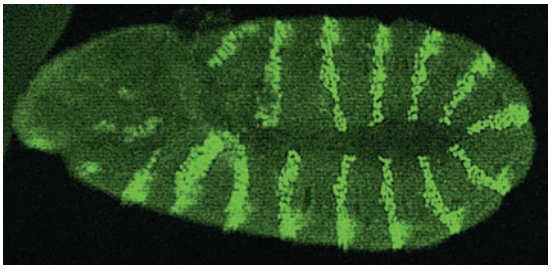
Genetic Diversity and Evolution

Life on Earth exists in a tremendous array of forms and features in almost every conceivable environment. Life is also characterized by adaptation: many organisms are exquisitely suited to the environment in which they are found. The history of life is a chronicle of new forms of life emerging, old forms disappearing, and existing forms changing.

Despite their tremendous diversity, living organisms have an important feature in common: all use similar genetic systems. The complete set of genetic instructions for any organism is its **genome**. All genomes are encoded in nucleic acids—either DNA or RNA. The coding system for genomic information is also common to all life: all genetic instructions are in the same format and, with rare exceptions, the code words are identical. Likewise, the processes by which genetic information is copied and decoded are remarkably similar for all forms of life. These common features of heredity suggest that all life on Earth evolved from the same primordial ancestor that arose between 3.5 billion and 4 billion years ago. Biologist Richard Dawkins describes life as a river of DNA that runs through time, connecting all organisms past and present.

The fact that all organisms have similar genetic systems means that the study of one organism's genes reveals principles that apply to other organisms. Investigations of how bacterial DNA is replicated (copied), for example, provide information that applies to the replication of human DNA. It also means that genes will function in foreign cells, which makes genetic engineering possible. Unfortunately, the similarity of genetic systems is also the basis for diseases such as AIDS (acquired immune deficiency syndrome), in which viral genes are able to function—sometimes with alarming efficiency—in human cells.

Life's diversity and adaptation are products of evolution, which is simply genetic change over time. Evolution is a two-step process: first, inherited differences arise randomly, and then the proportion of individuals with particular differences increases or decreases. Genetic variation is therefore the foundation of all evolutionary change and is ultimately the basis of all life as we know it. Furthermore, techniques of molecular genetics are now routinely used to decipher evolutionary relationships among organisms; for example, recent analysis of DNA isolated from Neanderthal fossils has provided insight into the relationship between Neanderthals and modern humans, demonstrating that Neanderthals and the ancestors of modern humans probably interbred some 30,000 to 40,000 years ago. Genetics, the study of genetic variation, is critical to understanding the past, present, and future of life.



1.5 The key to development lies in the regulation of gene expression. This early fruit-fly embryo illustrates the localized expression (indicated by bright green) of the *engrailed* gene, which helps determine the development of body segments in the adult fly. [Steven Paddock.]

CONCEPTS

Heredity affects many of our physical features as well as our susceptibility to many diseases and disorders. Genetics contributes to advances in agriculture, pharmaceuticals, and medicine and is fundamental to modern biology. All organisms use similar genetic systems, and genetic variation is the foundation of the diversity of all life.

✓ CONCEPT CHECK 1

What are some of the implications of all organisms having similar genetic systems?

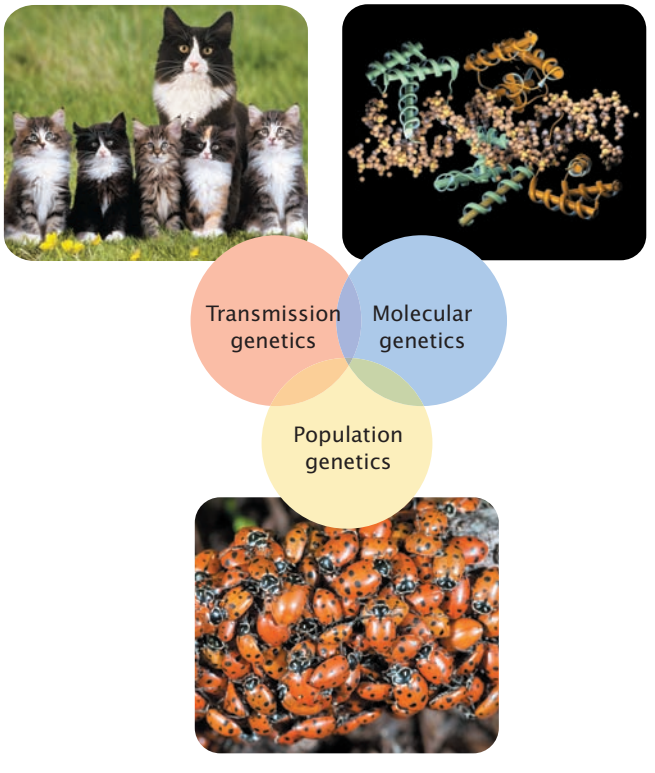
a. That all life forms are genetically related
b. That research findings on one organism's gene function can often be applied to other organisms
c. That genes from one organism can often exist and thrive in another organism
d. All of the above

Divisions of Genetics

The study of genetics consists of three major subdisciplines: transmission genetics, molecular genetics, and population genetics (**Figure 1.6**). **Transmission genetics** (also known as classical genetics) encompasses the basic principles of heredity and how traits are passed from one generation to the next. This subdiscipline addresses the relation between chromosomes and heredity, the arrangement of genes on chromosomes, and gene mapping. Here, the focus is on the individual organism—how an individual inherits its genetic makeup and how it passes its genes to the next generation.

Molecular genetics concerns the chemical nature of the gene itself: how genetic information is encoded, replicated, and expressed. It includes the cellular processes of replication, transcription, and translation (by which genetic information is transferred from one molecule to another) and of gene regulation (the processes that control the expression of genetic information). The focus in molecular genetics is the gene—its structure, organization, and function.

Population genetics explores the genetic composition of groups of individuals of the same species (populations) and how that composition changes over time and space. Because evolution is genetic change, population genetics is fundamentally the study of evolution. The focus of this subdiscipline is the group of genes found in a population.



1.6 Genetics can be subdivided into three interrelated fields. [Top left: Juniors Bildarchiv/Alamy. Top right: Martin McCarthy/Getty Images. Bottom: Stuart Wilson/Science Source.]

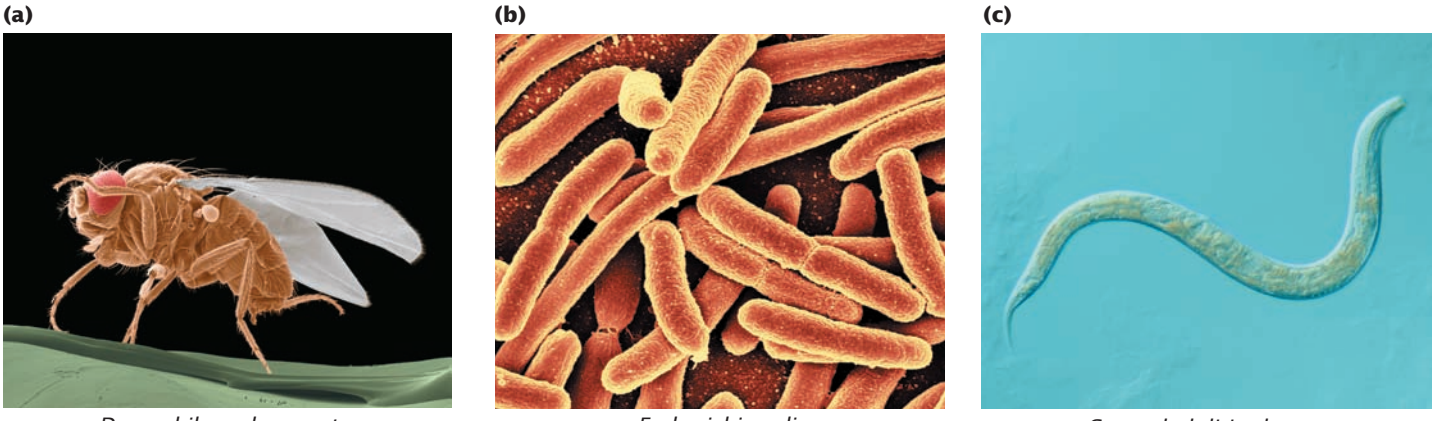
The division of the study of genetics into these three subdisciplines is convenient and traditional, but we should recognize that these subdisciplines overlap and that each one can be further divided into a number of more specialized fields, such as chromosomal genetics, biochemical genetics, quantitative genetics, and so forth. Alternatively, the study of genetics can be subdivided by organism (fruit-fly, corn, or bacterial genetics), and each of these organisms can be studied at the level of transmission, molecular, or population genetics. Modern genetics is an extremely broad field, encompassing many interrelated subdisciplines and specializations. TRY PROBLEM 17

Model Genetic Organisms

Through the years, genetic studies have been conducted on thousands of different species, including almost all major groups of bacteria, fungi, protists, plants, and animals. Nevertheless, a few species have emerged as **model genetic organisms**: organisms with characteristics that make them particularly useful for genetic analysis and about which a tremendous amount of genetic information has accumulated. Six model organisms that have been the subject of intensive genetic study are *Drosophila melanogaster*, a fruit fly; *Escherichia coli*, a bacterium present in the gut of humans and other mammals; *Caenorhabditis elegans*, a nematode (also called a roundworm); *Arabidopsis thaliana*, the thale cress plant; *Mus musculus*, the house mouse; and *Saccharomyces cerevisiae*, baker's yeast (**Figure 1.7**). These species are the organisms of choice for many genetic researchers, and their genomes were sequenced as a part of the Human Genome Project (described in Chapter 15).

At first glance, this group of lowly and sometimes unappreciated creatures might seem to be unlikely candidates for model organisms. However, all possess traits that make them particularly suitable for genetic study, including a short generation time, large but manageable numbers of progeny, adaptability to a laboratory environment, and the ability to be housed and propagated inexpensively. The life cycles, genomic characteristics, and features that make these model organisms useful for genetic studies are included in special illustrations in later chapters for five of the six species. Other species that are frequently the subject of genetic research and considered model genetic organisms include *Neurospora crassa* (bread mold), *Zea mays* (corn), *Danio rerio* (zebrafish), and *Xenopus laevis* (clawed frog). Although not generally considered a model genetic organism, humans have also been subjected to intensive genetic scrutiny.

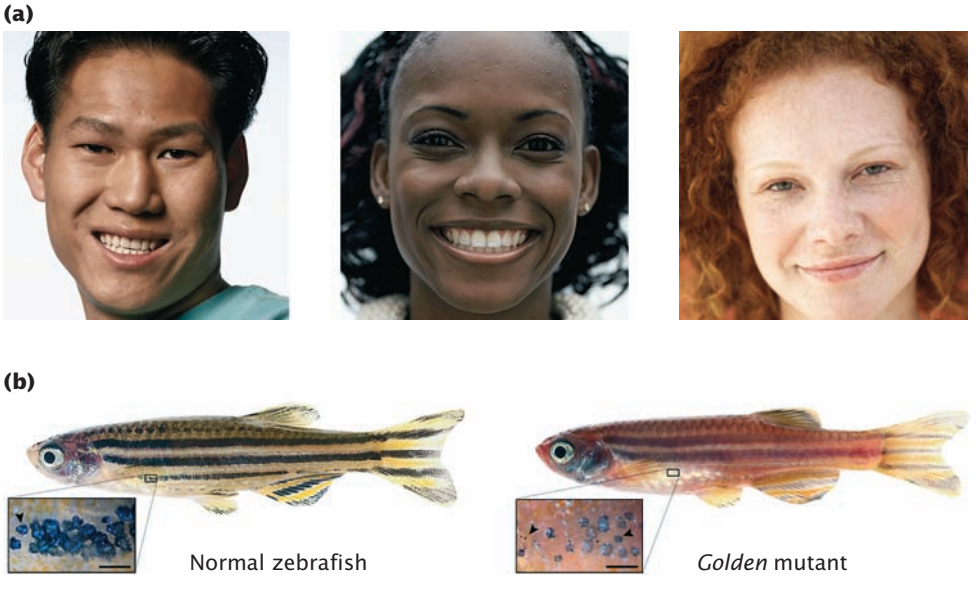
The value of model genetic organisms is illustrated by the use of zebrafish to identify genes that affect skin pigmentation in humans. For many years, geneticists have recognized that differences in pigmentation among human ethnic groups



1.7 Model genetic organisms are species with features that make them useful for genetic analysis. Organisms (a) through (e) are discussed in more detail on the pages referred to below each. [Part a: SPL/Science Source. Part b: Pasieka/Science Source. Part c: Sinclair Stammers/Science Source. Part d: Peggy Greb/ARS/USDA. Part e: AP Photo/Joel Page. Part f: Biophoto Associates/Science Source.]

(**Figure 1.8a**) are genetic, but the genes causing these differences were largely unknown. The zebrafish has become an important model in genetic studies because it is a small vertebrate that produces many offspring and is easy to rear in the laboratory. The mutant zebrafish called *golden* has light pigmentation due to the presence of fewer, smaller, and less dense pigment-containing structures called melanosomes in its cells (**Figure 1.8b**). Light skin in humans is similarly due to fewer and less dense melanosomes in pigment-containing cells.

Keith Cheng and his colleagues hypothesized that light skin in humans might result from a mutation that is similar to the *golden* mutation in zebrafish. Taking advantage of the ease with which zebrafish can be manipulated in the laboratory, they isolated and sequenced the gene responsible for the *golden* mutation and found that it encodes a protein that takes part in calcium uptake by melanosomes. They then searched a database of all known human genes and found a similar gene called *SLC24A5*, which encodes the same function in human cells. When they examined human populations, they found that light-skinned Europeans typically possess one form of this gene, whereas darker-skinned Africans, East Asians, and Native Americans usually possess a different form of the gene. Many other genes also affect pigmentation in humans, as illustrated by mutations in the *OCA2* gene that produce albinism among the Hopis (discussed in the introduction to this chapter). Nevertheless, *SLC24A5* appears to be responsible for 24% to 38% of the differences in pigmentation between Africans and Europeans. This example illustrates the power of model organisms in genetic research. However, we should not forget that all



1.8 The zebrafish, a model genetic organism, has been instrumental in helping to identify genes encoding pigmentation differences among humans. (a) Human ethnic groups differ in degree of skin pigmentation. (b) The zebrafish *golden* mutation is caused by a gene that controls the amount of melanin in melanosomes. [Part a: Barbara Penoyar/Getty Images; Amos Morgan/Getty Images; Stockbyte/Getty Images. Part b: Keith Cheng/Jake Gittlen, Cancer Research Foundation Penn State College of Medicine.]

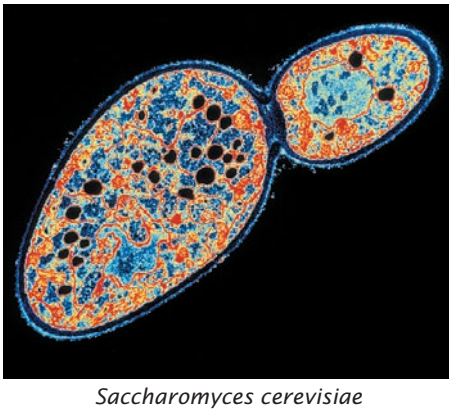
(d)

Arabidopsis thaliana
Thale-cress plant (pp. 331–333)

(e)

Mus musculus
House mouse (pp. 393–394)

(f)



Saccharomyces cerevisiae
Baker's yeast

organisms possess unique characteristics and that sometimes the genetics of models do not accurately reflect the genetic systems of other organisms.

CONCEPTS

The three major divisions of genetics are transmission genetics, molecular genetics, and population genetics. Transmission genetics examines the principles of heredity; molecular genetics deals with the gene and the cellular processes by which genetic information is transferred and expressed; population genetics concerns the genetic composition of groups of organisms and how that composition changes over time and space. Model genetic organisms are species that have received special emphasis in genetic research: they have characteristics that make them useful for genetic analysis.

✓ CONCEPT CHECK 2

Would the horse make a good model genetic organism? Why or why not?

1.2 Humans Have Been Using Genetics for Thousands of Years

Although the science of genetics is young—almost entirely a product of the past 100 years or so—people have been using genetic principles for thousands of years.

The Early Use and Understanding of Heredity

The first evidence that people understood and applied the principles of heredity in earlier times is found in the domestication of plants and animals, which began between approximately 10,000 and 12,000 years ago in the Middle East. The first domesticated organisms included wheat, peas, lentils, barley, dogs, goats, and sheep (**Figure 1.9a**). By 4000 years ago, sophisticated genetic techniques were already in use in the Middle East. The Assyrians and Babylonians developed several hundred varieties of date palms that differed in fruit size, color, taste, and time of ripening (**Figure 1.9b**). Other crops and domesticated animals

(a)

(b)

1.9 Ancient peoples practiced genetic techniques in agriculture. (a) Modern wheat, with larger and more numerous seeds that do not scatter before harvest, was produced by interbreeding at least three different wild species. (b) Assyrian bas-relief sculpture showing artificial pollination of date palms at the time of King Assurnasirpalli II, who reigned from 883 to 859 B.C. [Part a: Scott Bauer/ARS/USDA. Part b: Image copyright © The Metropolitan Museum of Art. Image source: Art Resource, NY.]

were developed by cultures in Asia, Africa, and the Americas in the same period.

The ancient Greeks gave careful consideration to human reproduction and heredity. Greek philosophers developed the concept of **pangenesis**. This concept suggested that specific pieces of information travel from various parts of the body to the reproductive organs, from which they are passed to the embryo (**Figure 1.10a**). Pangenesis led the ancient Greeks to propose the notion of the **inheritance of acquired characteristics**, in which traits acquired in a person's lifetime become incorporated into that person's hereditary information and are passed on to offspring; for example, people who developed musical ability through diligent study would produce children who are innately endowed with musical ability. Although incorrect, these ideas persisted through the twentieth century.

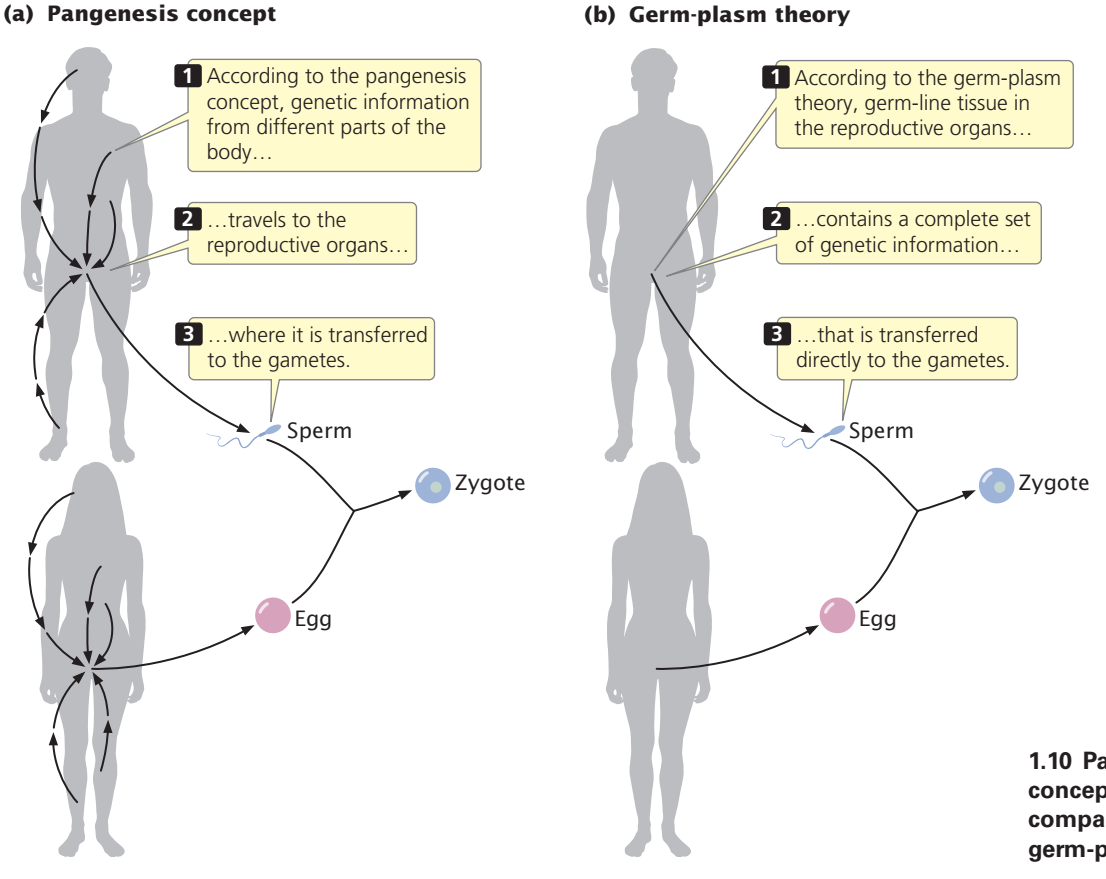
Additional developments in our understanding of heredity occurred during the seventeenth century. Dutch eyeglass makers began to put together simple microscopes in the late 1500s, enabling Robert Hooke (1635–1703) to discover cells in 1665. Microscopes provided naturalists with new and exciting vistas on life, and perhaps it was excessive enthusiasm for this new world of the very small that gave rise to the idea of **preformationism**. According to preformationism, inside the egg or sperm there exists a fully formed miniature adult, a *homunculus*, which simply enlarges during development (**Figure 1.11**). Preformationism meant that all traits were inherited from only one parent—from the father if the homunculus was in the sperm or from the mother if it was in the egg. Although many observations suggested that offspring possess a mixture of traits from both parents, preformationism remained a popular concept throughout much of the seventeenth and eighteenth centuries.

Another early notion of heredity was **blending inheritance**, which proposed that the traits of offspring are a blend, or mixture, of parental traits. This idea suggested that the genetic material itself blends, much as blue and yellow pigments blend to make green paint. It also suggested that after having been blended, genetic differences could not be separated in future generations, just as green paint cannot be separated into blue and yellow pigments. Some traits do *appear* to exhibit blending inheritance; however, thanks to Gregor Mendel's research with pea plants, we now understand that individual genes do not blend.

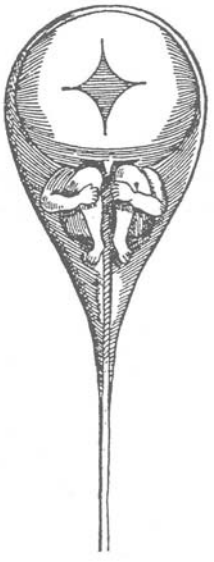
The Rise of the Science of Genetics

In 1676, Nehemiah Grew (1641–1712) reported that plants reproduce sexually by using pollen from the male sex cells. With this information, a number of botanists, including Gregor Mendel (1822–1884; **Figure 1.12**), began to experiment with crossing plants and creating hybrids. Mendel went on to discover the basic principles of heredity in the 1860s.

Developments in cytology (the study of cells) in the 1800s had a strong influence on genetics. Building on the work of



1.10 Pangenesis, an early concept of inheritance, compared with the modern germ-plasm theory.

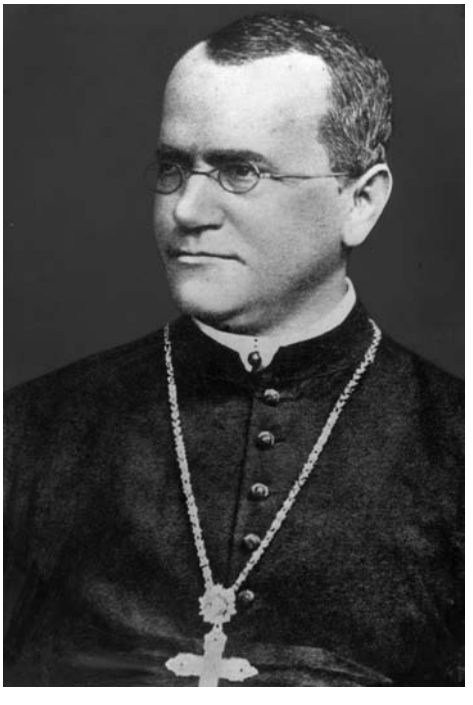


1.11 Preformationists in the seventeenth and eighteenth centuries believed that a sperm or an egg contains a fully formed human (the homunculus). Shown here is a drawing of a homunculus inside a sperm. [Science Source.]

others, Matthias Jacob Schleiden (1804–1881) and Theodor Schwann (1810–1882) proposed the **cell theory** in 1839. According to this theory, all life is composed of cells, cells arise only from preexisting cells, and the cell is the fundamental unit of structure and function in living organisms. Biologists interested in heredity began to examine cells to see what takes place in the course of cell reproduction. Walther Flemming (1843–1905) observed the division of chromosomes in 1879 and published a superb description of mitosis. By 1885, biologists generally recognized that the cell nucleus contains the hereditary information.

Charles Darwin (1809–1882), one of the most influential biologists of the nineteenth century, put forth the theory of evolution through natural selection and published his ideas in *On the Origin of Species* in 1859. Darwin recognized that heredity was fundamental to evolution, and he conducted extensive genetic crosses with pigeons and other organisms. He never understood the nature of inheritance, however, and this lack of understanding was a major omission in his theory of evolution.

In the last half of the nineteenth century, cytologists demonstrated that the nucleus had a role in fertilization. Near the close of the nineteenth century, August Weismann (1834–1914) finally laid to rest the notion of the inheritance of acquired characteristics. He cut off the tails of mice for 22 consecutive generations and showed that the tail length in descendants remained stubbornly long. Weismann proposed the **germ-plasm theory**, which holds that the cells in the reproductive organs carry a complete set of genetic information that is passed to the egg and sperm (**Figure 1.10b**). This theory, and some of the other early theories of heredity that we have discussed up to this point, are summarized in **Table 1.1.**



1.12 Gregor Mendel was the founder of modern genetics. Mendel first discovered the principles of heredity by crossing different varieties of pea plants and analyzing the pattern of the transmission of traits in subsequent generations. [Hulton Archive/Getty Images.]

TABLE 1.1 Early concepts of heredity

Concept	Proposed	Correct or incorrect
Pangenesis	Genetic information travels from different parts of the body to reproductive organs.	Incorrect
Inheritance of acquired characteristics	Acquired traits become incorporated into hereditary information.	Incorrect
Preformationism	Miniature organism resides in sex cells; thus all traits are inherited from one parent.	Incorrect
Blending inheritance	Genes blend and mix.	Incorrect
Germ-plasm theory	All cells contain a complete set of genetic information.	Correct
Cell theory	All life is composed of and cells arise only from cells.	Correct
Mendelian inheritance	Traits are inherited according to specific principles proposed by Mendel.	Correct