



教育部高等教育司推荐
国外优秀生命科学教学用书

遗传学基础

(第8版)(影印版)

ESSENTIALS OF GENETICS

(Eighth Edition)

William S. Klug
Michael R. Cummings
Charlotte A. Spencer
Michael A. Palladino




Pearson

高等教育出版社

Q3
110



教育部高等教育司推荐
国外优秀生命科学教学用书

遗传学基础

(第8版)(影印版)

ESSENTIALS OF GENETICS

(Eighth Edition)



William S. Klug
Michael R. Cummings
Charlotte A. Spencer
Michael A. Palladino

高等教育出版社·北京

图字：01-2016-4809 号

Authorized Adaptation from the English language edition, entitled ESSENTIALS OF GENETICS, 8E, 9780321803115 by KLUG, WILLIAM S.; CUMMINGS, MICHAEL R.; SPENCER, CHARLOTTE A.; PALLADINO, MICHAEL A., published by arrangement with Pearson Education Inc, Copyright © 2013 Pearson Education, Inc.

All rights reserved. No part of this book may be reproduced or transmitted in any form or by any means, electronic or mechanical, including photocopying, recording or by any information storage retrieval system, without permission from Pearson Education, Inc.

China Adapted edition published by PEARSON EDUCATION ASIA LTD. and HIGHER EDUCATION PRESS LIMITED COMPANY Copyright © 2016.

This Adapted edition is manufactured in the People's Republic of China, and is authorized for sale only in the People's Republic of China excluding Hong Kong, Taiwan and Macau.

本书英文原版 ESSENTIALS OF GENETICS, 8E, 9780321803115, 作者为 KLUG, WILLIAM S.; CUMMINGS, MICHAEL R.; SPENCER, CHARLOTTE A.; PALLADINO, MICHAEL A., 该书为培生教育出版集团出版。

版权所有。未经培生教育出版集团许可, 任何部分不得以任何形式、任何途径(电子版或纸质版)复制或传播, 包括影印、录制或信息存储及检索系统。

此改编版由培生教育出版集团和高等教育出版社有限公司合作出版。著作权 © 2016。

此改编版在中华人民共和国境内印制出版, 仅限于在中华人民共和国境内(但不允许在中国香港、澳门特别行政区及中国台湾地区)销售。

本书封面贴有 Pearson Education (培生教育出版集团)激光防伪标签。无标签者不得销售。

图书在版编目(CIP)数据

遗传学基础: 第8版=Essentials of Genetics, Eighth Edition:
英文/(美)克卢格(Klug, W. S.)等编著. —影印本. —北京:
高等教育出版社, 2016. 8
ISBN 978-7-04-045521-2

I. ①遗… II. ①克… III. ①遗传学-高等学校-教材-英文 IV. ①Q3

中国版本图书馆CIP数据核字(2016)第126612号

策划编辑 高新景 责任编辑 高新景 封面设计 张楠 封面摄影 俞瑜 责任印制 田甜

出版发行 高等教育出版社
社址 北京市西城区德外大街4号
邮政编码 100120
印刷 固安县铭成印刷有限公司
开本 889mm×1194mm 1/16
印张 34.5
字数 1000千字
购书热线 010-58581118
咨询电话 400-810-0598

网 址 <http://www.hep.edu.cn>
<http://www.hep.com.cn>
网上订购 <http://www.hepmall.com.cn>
<http://www.hepmall.com>
<http://www.hepmall.cn>

版 次 2016年8月第1版
印 次 2016年8月第1次印刷
定 价 85.00元

本书如有缺页、倒页、脱页等质量问题, 请到所购图书销售部门联系调换

版权所有 侵权必究

物料号 45521-00

数字课程 (基础版)

遗传学基础

(影印版)

登录方法:

1. 访问<http://abook.hep.com.cn/45521>, 进行注册。已注册的用户输入用户名和密码登录, 进入“我的课程”。
2. 点击页面右上方“绑定课程”, 正确输入教材封底数字课程账号(20位密码, 刮开涂层可见), 进行课程绑定。
3. 在“我的课程”中选择本课程并点击“进入课程”即可进行学习。课程在首次使用时, 会出现在“申请学习”列表中。

课程绑定后一年为数字课程使用有效期。如有使用问题, 请发邮件至: lifescience@pub.hep.cn。



遗传学基础 (影印版)

用户名

密码

验证码

8869

进入课程

内容介绍

纸质教材

版权信息

联系方式

“遗传学基础(影印版)”数字课程与纸质教材紧密配合, 配套资源包括书中所有插图的彩色版、习题答案。数字课程充分运用多种形式的资源, 为师生提供教学参考。

高等教育出版社

<http://abook.hep.com.cn/45521>

About the Authors

William S. Klug recently retired as Professor of Biology at The College of New Jersey (formerly Trenton State College) in Ewing, New Jersey. He served as Chair of the Biology Department for 17 years, a position to which he was first elected in 1974. He received his B.A. degree in Biology from Wabash College in Crawfordsville, Indiana, and his Ph.D. from Northwestern University in Evanston, Illinois. Prior to coming to The College of New Jersey, he was on the faculty of Wabash College as an Assistant Professor. His research interests have involved ultrastructural and molecular genetic studies of oogenesis in *Drosophila*. He taught the genetics course as well as the senior capstone seminar course in human and molecular genetics to undergraduate biology majors for 38 years. In 2002, he was the recipient of the initial teaching award given at the College of New Jersey granted to the faculty member who most challenges students to achieve high standards. He also received the 2004 Outstanding Professor Award from the Sigma Pi International, and in the same year, he was nominated as the Educator of the Year, an award given by the Research and Development Council of New Jersey.

Michael R. Cummings is Research Professor in the Department of Biological, Chemical, and Physical Sciences at Illinois Institute of Technology, Chicago, Illinois. For more than 25 years, he was a faculty member in the Department of Biological Sciences and in the Department of Molecular Genetics at the University of Illinois at Chicago. He has also served on the faculties of Northwestern University and Florida State University. He received his B.A. from St. Mary's College in Winona, Minnesota, and his M.S. and Ph.D. from Northwestern University in Evanston, Illinois. In addition to this text and its companion volumes, he has also written textbooks in human genetics and general biology for nonmajors. His research interests center on the molecular organization and physical mapping of the heterochromatic regions of human acrocentric chromosomes. At the undergraduate level, he teaches courses in Mendelian and molecular genetics, human genetics, and general biology, and has received numerous awards for teaching excellence given by university faculty, student organizations, and graduating seniors.

Charlotte A. Spencer is a retired Associate Professor from the Department of Oncology at the University of Alberta in Edmonton, Alberta, Canada. She has also served as a faculty member in the Department of Biochemistry at the University of Alberta. She received her B.Sc. in Microbiology from the University of British Columbia and her Ph.D. in Genetics from the University of Alberta, followed by postdoctoral training at the Fred Hutchinson Cancer Research Center in Seattle, Washington. Her research interests involve the regulation of RNA polymerase II transcription in cancer cells, cells infected with DNA viruses, and cells traversing the mitotic phase of the cell cycle. She has taught courses in biochemistry, genetics, molecular biology, and oncology, at both undergraduate and graduate levels. In addition, she has written booklets in the Prentice Hall Exploring Biology series, which are aimed at the undergraduate nonmajor level.

Michael A. Palladino is Dean of the School of Science and Associate Professor in the Department of Biology at Monmouth University in West Long Branch, New Jersey. He received his B.S. degree in Biology from Trenton State College (now known as The College of New Jersey) and his Ph.D. in Anatomy and Cell Biology from the University of Virginia. He directs an active laboratory of undergraduate student researchers studying molecular mechanisms involved in innate immunity of mammalian male reproductive organs and genes involved in oxygen homeostasis and ischemic injury of the testis. He has taught a wide range of courses for both majors and nonmajors and currently teaches genetics, biotechnology, endocrinology, and laboratory in cell and molecular biology. He has received several awards for research and teaching, including the New Investigator Award of the American Society of Andrology, the 2005 Distinguished Teacher Award from Monmouth University, and the 2005 Caring Heart Award from the New Jersey Association for Biomedical Research. He is co-author of the undergraduate textbook *Introduction to Biotechnology*, Series Editor for the Benjamin Cummings *Special Topics in Biology* booklet series, and author of the first booklet in the series, *Understanding the Human Genome Project*.

Dedication

There are only a few chances to publicly recognize contributions that people have made to one's professional achievements, and to express appreciation for the positive role they have played in long-standing personal interactions, which is an added bonus.

I wish to dedicate this edition, the 19th in this text series, to my editors, of all kinds, past and present, in recognition of their contributions to the success and longevity of these books: Kerry Baruch, Bob Lakemacher, Bob Rogers, Sheri Snively, Gary Carlson, Michael Gillespie, and Dusty Friedman. Over the past three decades, your insights and support have been indispensable; you are all hall-of-famers in your professions.

And with deep appreciation of their direct input to these books, I also dedicate this edition to my coauthors, Charlotte Spencer, Mike Cummings, Mike Palladino, and Harry Nickla—my four steadfast compatriots, who for decades have always been confident that we collectively could arrive at the most accurate formula to impart to students the ideal blend of the critical history, analytical thinking, and modern findings in Genetics.

To all — you have been voices of reason as well as sources of great inspiration and heartwarming friendship. It has been my good fortune that you have been a part of my life.

Preface

Essentials of Genetics is written for courses requiring a text that is briefer and less detailed than its more comprehensive companion, *Concepts of Genetics*. While coverage is thorough and modern, *Essentials* is written to be more accessible to biology majors early in their undergraduate careers, as well as to students majoring in a number of other disciplines, including agriculture, animal husbandry, chemistry, engineering, forestry, psychology, and wildlife management. Because *Essentials of Genetics* is shorter than many other texts, it is also more manageable in one-quarter and trimester courses.

Goals

In this and the previous edition of *Essentials of Genetics*, the two most important goals have been to devise modern pedagogic innovations that enhance learning, and to combine these with updated, highly accessible coverage of genetic topics of both historical and modern significance. As described below, this edition is no exception to achieving these goals, as we introduce: 1) a new set of chapters called **Special Topics in Modern Genetics**; and 2) **MasteringGenetics**, a powerful online learning and assessment system that helps students develop and practice problem-solving skills.

Beyond these new features, our overarching goals have remained the same as for previous editions. Specifically, we seek to

- Emphasize concepts rather than excessive detail.
- Write clearly and directly to students in order to provide understandable explanations of complex analytical topics.
- Emphasize problem solving, thereby guiding students to think analytically and to apply and extend their knowledge of genetics.
- Provide the most modern and up-to-date coverage of this exciting field.
- Propagate the rich history of genetics that so beautifully elucidates how information is acquired as the discipline develops and grows.
- Create inviting, engaging, and pedagogically useful figures enhanced by meaningful photographs to support student understanding.
- Provide outstanding interactive media support to guide students in understanding important concepts through animations, tutorial exercises, and assessment tools.

The above goals serve as the cornerstone of *Essentials of Genetics*. This pedagogic foundation allows the book to accommodate courses with many different approaches and lecture formats. While the book presents a coherent table of contents that represents one approach to offering a course in genetics, chapters are nevertheless written to be independent of one

another, allowing instructors to utilize them in various sequences.

New to This Edition

We are excited to make available to users of this edition two pedagogic innovations that will expand our coverage of emerging topics and enhance students' learning. At the same time, the book has undergone a thorough streamlining to help instructors and students focus on the truly essential information.

- **Special Topics in Modern Genetics**—As new research topics in genetics gain stature and evolve, either they gradually find their way into textbooks as a short section in one chapter (when they are very specific), or they are mentioned briefly in many chapters (when either they are of more general impact). In either case, the topics are difficult for students and adopters of the text to locate among all of the other coverage, and sometimes they are barely covered at all. To provide greater prominence to such topics, new to this edition is a feature that we call **Special Topics in Modern Genetics**—a series of shorter, more specialized chapters about half the length of traditional chapters that provide concise, cohesive coverage of emerging genetics topics of great interest to students and instructors. Our goal is to facilitate the delivery of a lecture on each topic as well as to provide support for students who have heard such a lecture. And should the topics not be a formal part of the class, we are confident that they are of sufficient general interest that students may wish to read them on their own. For this edition, we have selected four important topics that are valuable, unique additions to the text, providing modern in-depth coverage that would otherwise not be present:

1. Epigenetics
2. DNA Forensics
3. Genomics and Personalized Medicine

Special Topics chapters, identified by colored margin tabs, fall at the end of the book, but they can be utilized at any time during the course. The strong supporting figures that accompany each **Special Topics** chapter are available in PowerPoint to facilitate their use in classroom presentations.

- **MasteringGenetics™**—This powerful online homework and assessment program helps students to understand key topics and concepts and to build effective problem-solving skills. In-depth tutorials coach students to correct answers with hints and feedback specific to their misconceptions. Assignments are auto-graded to save time for instructors,

and the gradebook makes it easy to identify specific students who are struggling and topics that the whole class is finding difficult.

■ Updated Topics

In addition to introducing the new Special Topics mini chapters, we have revised each chapter in the text to present the most current findings in genetics. Below is a list of some of the most significant additions and updates that were incorporated.

Ch. 1: Introduction to Genetics • Revised section on the Dawn of Modern Biology • Section on DNA and RNA rewritten • Expanded discussion of gene expression • Updated coverage of biotechnology • New coverage of genomics and proteomics

Ch. 2: Mitosis and Meiosis • New coverage and a new figure involving the role of cohesin and shugoshin during mitosis and meiosis

Ch. 3: Mendelian Genetics • New section—Correlation between Independent Assortment and Meiosis

Ch. 4: Modification of Mendelian Ratios • Several new problems added

Ch. 5: Sex Determination and Sex Chromosomes • New information regarding sex determination in chickens • Updated coverage of mammalian sex determination • Updated coverage of the human Y chromosome • Updated coverage of the mechanism of X chromosome inactivation

Ch. 6: Chromosome Mutations: Variation in Number and Arrangement • Noninvasive prenatal genetic diagnosis (NIPGD) introduced

Ch. 7: Linkage and Chromosome Mapping in Eukaryotes • New information involving chromosome mapping using DNA markers and annotated databases

Ch. 8: Genetic Analysis and Mapping in Bacteria and Bacteriophages • Revised figures involving conjugation • Introduction to horizontal versus vertical gene transfer • New information on the history of multiple resistant bacteria

Ch. 9: DNA Structure and Analysis • Continued classical coverage of DNA structure and analysis

Ch. 10: DNA Replication and Recombination • New figures depicting DNA replication

Ch. 11: Chromosome Structure and DNA Sequence Organization • Introduction to the role of chromatin remodeling in epigenetic modifications • New section on telomeric DNA sequences and TERRA (telomere

repeat-containing RNA) • New coverage on chromosome banding

Ch. 12: The Genetic Code and Transcription • New figure and updated coverage depicting the action of RNA polymerase during prokaryotic transcription • Updated coverage on RNA splicing

Ch. 13: Translation and Proteins • New coverage on the dynamic role of the ribosome during translation

Ch. 14: Gene Mutation, DNA Repair, and Transposition • New Section—Alkylating, Intercalation, and Adduct-Forming Agents • New Section—Single-Gene Mutations Cause a Wide Range of Human Diseases—describes the types of human disorders caused by the various types of single-gene mutations

Ch. 15: Regulation of Gene Expression • New coverage, including a new figure, to encompass attenuation and riboswitches as metabolite-sensing RNAs • New and updated material on core promoters, focused and dispersed promoters, and promoter elements, along with two new figures

Ch. 16: The Genetics of Cancer • New section on the cancer stem cell hypothesis

Ch. 17: Recombinant DNA Technology • Major revision and reorganization of recombinant DNA techniques • Added emphasis on radioactive labeling techniques to indicate more widespread current usage of nonradioactive detection and labeling methods (e.g., probe-labeling, sequencing) • Addition of RT-PCR and quantitative real-time PCR (qPCR) techniques • New material on FISH and spectral karyotyping • Major revision of DNA sequencing technologies to include capillary electrophoresis-based computer automated sequencing and next generation sequencing technologies • Major revision of Problems and Discussion Questions section

Ch. 18: Genomics and Proteomics • New section on “10 years after the HGP,” including a new section on the Human Microbiome Project • Updated content on the human genome, including new information about copy number variations (CNVs) • Expanded content on “stone age” genomics and new data on the Neanderthal genome • Expanded content on comparative genomics to include comparisons of model organism genomes and the human genome • New section on personal genomes and sequencing of individual diploid genomes • New section on Genome 10K • Updated content on systems biology, including new figure comparing human disease gene interaction network

Ch. 19: Applications and Ethics of Genetic Engineering and Biotechnology • New section on synthetic genomes • New and updated content on direct to consumer genetic testing (DTC) and patenting genetic

information • New section and figure on genome wide association studies for studying genetic diseases • Updated discussions on synthetic genomes, direct to consumer genetic testing (DTC), and patenting genetic information

Ch. 20: Developmental Genetics • Refined discussion on evolutionary conservation of developmental mechanisms and model organisms • Clarified section on genetic analysis of embryogenesis

Ch. 21: Quantitative Genetics and Multifactorial Traits • Revised coverage of twin studies • Updated coverage of quantitative trait loci (QTLs)

Ch. 22: Population and Evolutionary Genetics • Section on macroevolution and speciation rewritten with new examples and two new figures • Expanded coverage of phylogenetic analysis including a new figure • Rewritten section on molecular clocks including a new figure • Updated coverage on comparative genomics of Neanderthals and modern humans

This list reflects the rapid growth of information in genetics.

■ Emphasis on Concepts

Essentials of Genetics focuses on conceptual issues in genetics and uses problem solving to develop a deep understanding of them. We consider a concept to be a cognitive unit of meaning that encompasses a related set of scientifically derived findings and ideas. As such, a concept provides broad mental imagery, which we believe is a very effective way to teach science, in this case, genetics. Details that might be memorized, but soon forgotten, are instead subsumed within a conceptual framework that is easily retained. Such a framework may be expanded in content as new information is acquired and may interface with other concepts, providing a useful mechanism to integrate and better understand related processes and ideas. An extensive set of concepts may be devised and conveyed to eventually encompass and represent an entire discipline—and this is our goal in this genetics textbook.

To aid students in identifying the conceptual aspects of a major topic, each chapter begins with a section called **Chapter Concepts**, which identifies the most important ideas about to be presented. Then, throughout each chapter, *Essential Points* are provided that establish the key issues that have been discussed. And in the *How Do We Know?* question that starts each chapter's problem set, students are asked to identify the experimental basis of important genetic findings presented in the chapter. As an extension of the learning approach in biology called "Science as a Way of Knowing," this feature enhances students' understanding of many key concepts covered in each chapter.

Collectively, these features help to ensure that students easily become aware of and understand the major conceptual issues as they confront the extensive vocabulary and the many

important details of genetics. Carefully designed figures also support this approach throughout the book.

■ Emphasis on Problem Solving

Helping students develop effective problem-solving skills is one of the greatest challenges of a genetics course. The feature called **Now Solve This**, integrated throughout each chapter, asks students to link conceptual understanding in a more immediate way to problem solving. Each entry provides a problem for the student to solve that is closely related to the current text discussion. A pedagogic hint is then provided to aid in arriving at the correct solution. All chapters conclude with **Insights and Solutions**, a popular and highly useful section that provides sample problems and solutions that demonstrate approaches useful in genetic analysis. These help students develop analytical thinking and experimental reasoning skills. Digesting the information in *Insights and Solutions* primes students as they move on to the lengthier *Problems and Discussion Questions* section that concludes each chapter. Here, we present questions that review topics in the chapter and problems that ask students to think in an analytical and applied way about genetic concepts. Problems are of graduated difficulty, with the most demanding near the end of each section. The addition of MasteringGenetics extends our focus on problem solving online, and it allows students to get help and guidance while practicing how to solve problems.

■ Continuing Features

The Eighth Edition has maintained a number of popular features that are pedagogically useful for students as they study genetics. Collectively, these create a platform that seeks to challenge students to think more deeply about, and thus understand more comprehensively, the information he or she has just finished studying.

- **Exploring Genomics** Appearing in 10 chapters, this feature illustrates the pervasiveness of genomics in the current study of genetics. Each entry asks students to access one or more genomics-related Web sites that collectively are among the best publicly available resources and databases. Students work through interactive exercises that ensure their familiarity with the type of genomic or proteomic information available. Exercises instruct students on how to explore specific topics and how to access significant data. Questions guide student exploration and challenge them to further explore the sites on their own. Importantly, *Exploring Genomics* integrates genomics information throughout the text, as this emerging field is linked to chapter content. This feature provides the basis for individual or group assignments in or out of the classroom.
- **Genetics, Technology, and Society Essays** Appearing in 12 chapters, this feature provides a synopsis of a topic related to a current finding in genetics that impacts directly on our current society. After each essay, a section entitled

"Your Turn" appears in which questions are posed to students along with various resources to help answer them. This innovation provides yet another format to enhance classroom interactions.

- **Case Study** This feature appears at the end of each chapter and provides the basis for enhanced classroom interactions. In each entry, a short scenario related to one of the chapter topics is presented, followed by several questions. These ask students to apply their newly acquired knowledge to real-life issues that may be explored in small-group discussions or serve as individual assignments.

■ Acknowledgments

Reviewers

All comprehensive texts are dependent on the valuable input provided by many colleagues. The following individuals provided valuable advice, constructive criticism, and/or suggestions regarding the content of the Eighth Edition:

Althea K. Alton, *Western Illinois University*; Thomas H. Alton, *Western Illinois University*; Brian Ashburner, *University of Toledo*; George Bates, *Florida State University*; Mark Brick, *Colorado State University*; Jill Buettner, *Richland College*; Susan Capasso, *St. Vincent's College*; Aaron Cassill, *University of Texas - San Antonio*; Steve Denison, *Eckerd College*; John P. Doucet, *Nicholls State University*; Kurt Elliott, *Northwest Vista College*; Lehman L. Ellis, *Our Lady of Holy Cross College*; Victor Fet, *Marshall University*; Clarence E. Fouche, *Virginia Intermont College*; Gail Fraizer, *Kent State University*; Alexandros Georgakilas, *East Carolina University*; Edward F. Golenberg, *Wayne State University*; John Gray, *University of Toledo*; Danielle Hamill, *Ohio Wesleyan University*; David Kass, *Eastern Michigan University*; Mary Kimble, *Northeastern Illinois University*; Joan Kuh, *University of Hawaii*; Joseph Kulkosky, *Chestnut Hill College*; Alan C. Leonard, *Florida Institute of Technology*; Janet Lewis, *Michigan State University*; Jeannette M. Loutsch, *University of Science and Arts of Oklahoma*; Roy B. Mason, *Mt. San Jacinto College*; Philip McClean, *North Dakota State University*; Shawn Meagher, *Western Illinois University*; Sudhir Nayak, *The College of New Jersey*; Harry Nickla, *Creighton University*; Phillip A. Ortiz, *Empire State College*; Terrence Puryear, *Northeastern Illinois University*; Thomas F. Savage, *Oregon State University*; Brian W. Schwartz, *Columbus State University*; Allan Showalter, *Ohio University*; Thomas Smith, *Southern Arkansas University*; Tatiana Tatum, *Saint Xavier University*; Pattie Thompson, *University of Texas - San Antonio*; Paul Wilson, *Trent University*; Michael Zarowitz, *California Polytechnic University - San Luis Obispo*

While we take full responsibility for any errors in this book, we gratefully acknowledge the help and input provided by the above individuals.

Contributors

We offer a special acknowledgment to those who have provided direct input during many revisions of this text. We thank Sarah Ward at Colorado State University for her contribution

of the conservation genetics chapter; Sudhir Nayak from The College of New Jersey for his input into all things genomic; David Kass at Eastern Michigan University, Katherine Uyhazi at Yale Medical School, and Tamara Mans at North Hennepin Community College for their numerous contributions to the Genetics, Technology, and Society essays; Elliott Goldstein from Arizona State University for his perpetual input into molecular genetics coverage, and Mike Guidry of LightCone Interactive and Karen Hughes of the University of Tennessee for their original contributions to the media program. We thank Jutta Heller at University of Washington—Tacoma, John Osterman at the University of Nebraska—Lincoln, Virginia McDonough at Hope College, and Kiran Misra at Edinboro University of Pennsylvania for their work on the media program. We also offer special thanks to Harry Nickla, recently retired from Creighton University. In his role as author of the *Student Handbook*, he has written many new problems and authored the Selected Answers section that appear in Appendix A. We are grateful to all of the above contributors not only for sharing their genetic expertise, but for their dedication to this project as well as the pleasant interactions they provided.

Editorial and Production Input

At Pearson, we express appreciation and high praise for the editorial guidance of Michael Gillespie, whose ideas and efforts have helped to shape and refine the features of this edition of the text. Dusty Friedman, our Project Editor, has worked tirelessly to keep the project on schedule and to maintain our standards of high quality. In addition, our editorial staff—Deborah Gale, Executive Director of Development, Laura Tommasi, Senior Media Producer, Daniel Ross, Associate Media Producer, Caroline Ross, Assistant Media Producer, Juliana Tringali, Project Editor, Zane Coleman, Content Specialist, and Tania Mlawer, Director of Editorial Content for Mastering Genetics—has provided valuable input into the current edition. They have worked creatively to ensure that the pedagogy and design of the book and media package are at the cutting edge of a rapidly changing discipline. Sudhir Nayak of The College of New Jersey provided outstanding work for the new MasteringGenetics program, and his input regarding genomics was appreciated. Camille Herrera supervised all of the production intricacies with great attention to detail and perseverance. Outstanding copyediting was performed by Betty Pessagno, for which we are most grateful. Lauren Harp has professionally and enthusiastically managed the marketing of the text. Finally, the beauty and consistent presentation of the art work is the product of Imagineering of Toronto. Without the work ethic and dedication of the above individuals, the text would never have come to fruition.

Proofreading a manuscript of a 600-page text deserves more thanks than words can offer. Our utmost appreciation is extended to the individuals who confronted this task with patience and diligence, including Virginia McDonough, Ford Lux, Matthew Gilg, and Brian Flynn, a recent graduate of The College of New Jersey.

As these many acknowledgments make clear, a text such as this is a collective enterprise. All of the above individuals deserve to share in any success this text enjoys. We want them to know that our gratitude is equaled only by the extreme dedication evident in their efforts.

Brief Contents

- 1 Introduction to Genetics 1
- 2 Mitosis and Meiosis 15
- 3 Mendelian Genetics 35
- 4 Modification of Mendelian Ratios 60
- 5 Sex Determination and Sex Chromosomes 92
- 6 Chromosome Mutations: Variation in Number and Arrangement 109
- 7 Linkage and Chromosome Mapping in Eukaryotes 131
- 8 Genetic Analysis and Mapping in Bacteria and Bacteriophages 156
- 9 DNA Structure and Analysis 177
- 10 DNA Replication and Recombination 200
- 11 Chromosome Structure and DNA Sequence Organization 222
- 12 The Genetic Code and Transcription 238
- 13 Translation and Proteins 260
- 14 Gene Mutation, DNA Repair, and Transposition 280
- 15 Regulation of Gene Expression 303
- 16 The Genetics of Cancer 330
- 17 Recombinant DNA Technology 346
- 18 Genomics, Bioinformatics, and Proteomics 367
- 19 Applications and Ethics of Genetic Engineering and Biotechnology 399
- 20 Developmental Genetics 427
- 21 Quantitative Genetics and Multifactorial Traits 446
- 22 Population and Evolutionary Genetics 465

SPECIAL TOPICS IN MODERN GENETICS 1 Epigenetics 489

SPECIAL TOPICS IN MODERN GENETICS 2 DNA Forensics 499

SPECIAL TOPICS IN MODERN GENETICS 3 Genomics and Personalized Medicine 510

GLOSSARY 520

CHAPTER 1

Introduction to Genetics 1

- 1 Genetics Has a Rich and Interesting History 2
- 2 Genetics Progressed from Mendel to DNA in Less Than a Century 3
- 3 Discovery of the Double Helix Launched the Era of Molecular Genetics 6
- 4 Development of Recombinant DNA Technology Began the Era of DNA Cloning 8
- 5 The Impact of Biotechnology Is Continually Expanding 8
- 6 Genomics, Proteomics, and Bioinformatics Are New and Expanding Fields 10
- 7 Genetic Studies Rely on the Use of Model Organisms 10
- 8 We Live in the Age of Genetics 12

GENETICS, TECHNOLOGY, AND SOCIETY

The Scientific and Ethical Implications of Modern Genetics 13

EXPLORING GENOMICS

Internet Resources for Learning about Genomics, Bioinformatics, and Proteomics 13

CASE STUDY: Extending essential ideas of genetics beyond the classroom 14

Problems and Discussion Questions 14

CHAPTER 2

Mitosis and Meiosis 15

- 1 Cell Structure Is Closely Tied to Genetic Function 16
- 2 Chromosomes Exist in Homologous Pairs in Diploid Organisms 18
- 3 Mitosis Partitions Chromosomes into Dividing Cells 20
- 4 Meiosis Creates Haploid Gametes and Spores and Enhances Genetic Variation in Species 24
- 5 The Development of Gametes Varies during Spermatogenesis and Oogenesis 28
- 6 Meiosis Is Critical to the Sexual Reproduction Cycle of All Diploid Organisms 29
- 7 Electron Microscopy Has Revealed the Cytological Nature of Mitotic and Meiotic Chromosomes 30

EXPLORING GENOMICS

PubMed: Exploring and Retrieving Biomedical Literature 31

CASE STUDY: Timing is everything 32

Insights and Solutions 32

Problems and Discussion Questions 33

CHAPTER 3

Mendelian Genetics 35

- 1 Mendel Used a Model Experimental Approach to Study Patterns of Inheritance 36
- 2 The Monohybrid Cross Reveals How One Trait Is Transmitted from Generation to Generation 37
- 3 Mendel's Dihybrid Cross Generated a Unique F_2 Ratio 40
- 4 The Trihybrid Cross Demonstrates That Mendel's Principles Apply to Inheritance of Multiple Traits 44
- 5 Mendel's Work Was Rediscovered in the Early Twentieth Century 45
- 6 Independent Assortment Leads to Extensive Genetic Variation 47
- 7 Laws of Probability Help to Explain Genetic Events 48
- 8 Chi-Square Analysis Evaluates the Influence of Chance on Genetic Data 49
- 9 Pedigrees Reveal Patterns of Inheritance of Human Traits 52

EXPLORING GENOMICS

Online Mendelian Inheritance in Man 54

CASE STUDY: To test or not to test 55

Insights and Solutions 55

Problems and Discussion Questions 57

CHAPTER 4

Modification of Mendelian Ratios 60

- 1 Alleles Alter Phenotypes in Different Ways 61
- 2 Geneticists Use a Variety of Symbols for Alleles 61
- 3 Neither Allele Is Dominant in Incomplete, or Partial, Dominance 62
- 4 In Codominance, the Influence of Both Alleles in a Heterozygote Is Clearly Evident 63
- 5 Multiple Alleles of a Gene May Exist in a Population 63
- 6 Lethal Alleles Represent Essential Genes 65
- 7 Combinations of Two Gene Pairs with Two Modes of Inheritance Modify the 9:3:3:1 Ratio 66
- 8 Phenotypes Are Often Affected by More Than One Gene 67
- 9 Complementation Analysis Can Determine If Two Mutations Causing a Similar Phenotype Are Alleles of the Same Gene 72
- 10 Expression of a Single Gene May Have Multiple Effects 73

- 11 X-Linkage Describes Genes on the X Chromosome 74
- 12 In Sex-Limited and Sex-Influenced Inheritance, an Individual's Sex Influences the Phenotype 76
- 13 Genetic Background and the Environment Affect Phenotypic Expression 77
- 14 Genomic (Parental) Imprinting and Gene Silencing 80
- 15 Extranuclear Inheritance Modifies Mendelian Patterns 81

GENETICS, TECHNOLOGY, AND SOCIETY

Improving the Genetic Fate of Purebred Dogs 84

CASE STUDY: But he isn't deaf 85

Insights and Solutions 86

Problems and Discussion Questions 87

CHAPTER 5

Sex Determination and Sex Chromosomes 92

- 1 Life Cycles Depend on Sexual Differentiation 93
- 2 X and Y Chromosomes Were First Linked to Sex Determination Early in the Twentieth Century 95
- 3 The Y Chromosome Determines Maleness in Humans 96
- 4 The Ratio of Males to Females in Humans Is Not 1.0 99
- 5 Dosage Compensation Prevents Excessive Expression of X-Linked Genes in Humans and Other Mammals 100
- 6 The Ratio of X Chromosomes to Sets of Autosomes Determines Sex in *Drosophila* 103
- 7 Temperature Variation Controls Sex Determination in Reptiles 104

GENETICS, TECHNOLOGY, AND SOCIETY

A Question of Gender: Sex Selection in Humans 106

CASE STUDY: Doggone it! 107

Insights and Solutions 107

Problems and Discussion Questions 107

CHAPTER 6

Chromosome Mutations: Variation in Number and Arrangement 109

- 1 Variation in Chromosome Number: Terminology and Origin 110
- 2 Monosomy and Trisomy Result in a Variety of Phenotypic Effects 111
- 3 Polyploidy, in Which More Than Two Haploid Sets of Chromosomes Are Present, Is Prevalent in Plants 114
- 4 Variation Occurs in the Composition and Arrangement of Chromosomes 117
- 5 A Deletion Is a Missing Region of a Chromosome 118
- 6 A Duplication Is a Repeated Segment of a Chromosome 119
- 7 Inversions Rearrange the Linear Gene Sequence 121
- 8 Translocations Alter the Location of Chromosomal Segments in the Genome 123

- 9 Fragile Sites in Human Chromosomes Are Susceptible to Breakage 125

GENETICS, TECHNOLOGY, AND SOCIETY

Down Syndrome and Prenatal Testing—The New Eugenics? 127

CASE STUDY: Fish tales 128

Insights and Solutions 128

Problems and Discussion Questions 129

CHAPTER 7

Linkage and Chromosome Mapping in Eukaryotes 131

- 1 Genes Linked on the Same Chromosome Segregate Together 132
- 2 Crossing Over Serves as the Basis of Determining the Distance between Genes during Mapping 133
- 3 Determining the Gene Sequence during Mapping Requires the Analysis of Multiple Crossovers 138
- 4 As the Distance between Two Genes Increases, Mapping Estimates Become More Inaccurate 145
- 5 Chromosome Mapping Is Now Possible Using DNA Markers and Annotated Computer Databases 146
- 6 Other Aspects of Genetic Exchange 147
- 7 Did Mendel Encounter Linkage? 149

EXPLORING GENOMICS

Human Chromosome Maps on the Internet 150

CASE STUDY: Links to autism 150

Insights and Solutions 151

Problems and Discussion Questions 152

CHAPTER 8

Genetic Analysis and Mapping in Bacteria and Bacteriophages 156

- 1 Bacteria Mutate Spontaneously and Are Easily Cultured 157
- 2 Genetic Recombination Occurs in Bacteria 157
- 3 Rec Proteins Are Essential to Bacterial Recombination 165
- 4 The F Factor Is an Example of a Plasmid 165
- 5 Transformation Is Another Process Leading to Genetic Recombination in Bacteria 166
- 6 Bacteriophages Are Bacterial Viruses 167
- 7 Transduction Is Virus-Mediated Bacterial DNA Transfer 170
- 8 Bacteriophages Undergo Intergenic Recombination 172

GENETICS, TECHNOLOGY, AND SOCIETY

From Cholera Genes to Edible Vaccines 173

CASE STUDY: To treat or not to treat 174

Insights and Solutions 174

Problems and Discussion Questions 176

CHAPTER 9

DNA Structure and Analysis 177

- 1 The Genetic Material Must Exhibit Four Characteristics 178
- 2 Until 1944, Observations Favored Protein as the Genetic Material 178
- 3 Evidence Favoring DNA as the Genetic Material Was First Obtained during the Study of Bacteria and Bacteriophages 179
- 4 Indirect and Direct Evidence Supports the Concept that DNA Is the Genetic Material in Eukaryotes 184
- 5 RNA Serves as the Genetic Material in Some Viruses 185
- 6 The Structure of DNA Holds the Key to Understanding Its Function 186
- 7 Alternative Forms of DNA Exist 192
- 8 The Structure of RNA Is Chemically Similar to DNA, but Single-Stranded 193
- 9 Many Analytical Techniques Have Been Useful during the Investigation of DNA and RNA 193

EXPLORING GENOMICS

Introduction to Bioinformatics: BLAST 196

CASE STUDY: Zigs and zags of the smallpox virus 197

Insights and Solutions 197

Problems and Discussion Questions 197

CHAPTER 10

DNA Replication and Recombination 200

- 1 DNA Is Reproduced by Semiconservative Replication 201
- 2 DNA Synthesis in Bacteria Involves Five Polymerases, as Well as Other Enzymes 205
- 3 Many Complex Issues Must Be Resolved during DNA Replication 208
- 4 A Coherent Model Summarizes DNA Replication 211
- 5 Replication Is Controlled by a Variety of Genes 211
- 6 Eukaryotic DNA Replication Is Similar to Replication in Prokaryotes, but Is More Complex 212
- 7 The Ends of Linear Chromosomes Are Problematic during Replication 213
- 8 DNA Recombination, Like DNA Replication, Is Directed by Specific Enzymes 216

GENETICS, TECHNOLOGY, AND SOCIETY

Telomeres: The Key to Immortality? 218

CASE STUDY: At loose ends 219

Insights and Solutions 219

Problems and Discussion Questions 219

CHAPTER 11

Chromosome Structure and DNA Sequence Organization 222

- 1 Viral and Bacterial Chromosomes Are Relatively Simple DNA Molecules 223
- 2 Mitochondria and Chloroplasts Contain DNA Similar to Bacteria and Viruses 224
- 3 Specialized Chromosomes Reveal Variations in the Organization of DNA 226
- 4 DNA Is Organized into Chromatin in Eukaryotes 228
- 5 Eukaryotic Genomes Demonstrate Complex Sequence Organization Characterized by Repetitive DNA 231
- 6 The Vast Majority of a Eukaryotic Genome Does Not Encode Functional Genes 235

EXPLORING GENOMICS

Database of Genomic Variants: Structural Variations in the Human Genome 235

CASE STUDY: Art inspires learning 236

Insights and Solutions 236

Problems and Discussion Questions 237

CHAPTER 12

The Genetic Code and Transcription 238

- 1 The Genetic Code Exhibits a Number of Characteristics 239
- 2 Early Studies Established the Basic Operational Patterns of the Code 239
- 3 Studies by Nirenberg, Matthaei, and Others Deciphered the Code 240
- 4 The Coding Dictionary Reveals the Function of the 64 Triplets 244
- 5 The Genetic Code Has Been Confirmed in Studies of Bacteriophage MS2 246
- 6 The Genetic Code Is Nearly Universal 246
- 7 Different Initiation Points Create Overlapping Genes 247
- 8 Transcription Synthesizes RNA on a DNA Template 247
- 9 RNA Polymerase Directs RNA Synthesis 248
- 10 Transcription in Eukaryotes Differs from Prokaryotic Transcription in Several Ways 250
- 11 The Coding Regions of Eukaryotic Genes Are Interrupted by Intervening Sequences Called Introns 252
- 12 Transcription Has Been Visualized by Electron Microscopy 255

GENETICS, TECHNOLOGY, AND SOCIETY

Nucleic Acid-Based Gene Silencing: Attacking the Messenger 256

CASE STUDY: A drug that sometimes works 257

Insights and Solutions 257

Problems and Discussion Questions 257

CHAPTER 13

Translation and Proteins 260

- 1 Translation of mRNA Depends on Ribosomes and Transfer RNAs 260
- 2 Translation of mRNA Can Be Divided into Three Steps 264
- 3 High-Resolution Studies Have Revealed Many Details about the Functional Prokaryotic Ribosome 267
- 4 Translation Is More Complex in Eukaryotes 268
- 5 The Initial Insight that Proteins Are Important in Heredity Was Provided by the Study of Inborn Errors of Metabolism 269
- 6 Studies of *Neurospora* Led to the One-Gene:One-Enzyme Hypothesis 270
- 7 Studies of Human Hemoglobin Established that One Gene Encodes One Polypeptide 270
- 8 Variation in Protein Structure Is the Basis of Biological Diversity 273
- 9 Proteins Function in Many Diverse Roles 275

EXPLORING GENOMICS

Translation Tools and Swiss-Protein for Studying Protein Sequences 276

CASE STUDY: Lost in translation 277

Insights and Solutions 277

Problems and Discussion Questions 277

CHAPTER 14

Gene Mutation, DNA Repair, and Transposition 280

- 1 Gene Mutations Are Classified in Various Ways 281
- 2 Spontaneous Mutations Arise from Replication Errors and Base Modifications 283
- 3 Induced Mutations Arise from DNA Damage Caused by Chemicals and Radiation 285
- 4 Single-Gene Mutations Cause a Wide Range of Human Diseases 288
- 5 Organisms Use DNA Repair Systems to Counteract Mutations 289
- 6 The Ames Test Is Used to Assess the Mutagenicity of Compounds 294
- 7 Transposable Elements Move within the Genome and May Create Mutations 295

EXPLORING GENOMICS

Sequence Alignment to Identify a Mutation 299

CASE STUDY: Genetic dwarfism 299

Insights and Solutions 300

Problems and Discussion Questions 300

CHAPTER 15

Regulation of Gene Expression 303

- 1 Prokaryotes Regulate Gene Expression in Response to Both External and Internal Conditions 304
- 2 Lactose Metabolism in *E. coli* Is Regulated by an Inducible System 304
- 3 The Catabolite-Activating Protein (CAP) Exerts Positive Control over the *lac* Operon 309
- 4 The Tryptophan (*trp*) Operon in *E. coli* Is a Repressible Gene System 310
- 5 Alterations to RNA Secondary Structure Also Contribute to Prokaryotic Gene Regulation 312
- 6 Eukaryotic Gene Regulation Differs from That in Prokaryotes 313
- 7 Eukaryotic Gene Expression Requires Chromatin Modifications 314
- 8 Eukaryotic Transcription Requires Specific *Cis*-Acting Sites 316
- 9 Activators and Repressors Influence Transcription When Bound to *Cis*-Acting Sites 318
- 10 Activators and Repressors Interact with General Transcription Factors and Affect Chromatin Structure 319
- 11 Posttranscriptional Gene Regulation Occurs at All the Steps from RNA Processing to Protein Modification 321
- 12 RNA-induced Gene Silencing Controls Gene Expression in Several Ways 323

EXPLORING GENOMICS

Tissue-Specific Gene Expression 325

CASE STUDY: A mysterious muscular dystrophy 326

Insights and Solutions 326

Problems and Discussion Questions 327

CHAPTER 16

The Genetics of Cancer 330

- 1 Cancer Is a Genetic Disease at the Level of Somatic Cells 331
- 2 Cancer Cells Contain Genetic Defects Affecting Genomic Stability, DNA Repair, and Chromatin Modifications 333
- 3 Cancer Cells Contain Genetic Defects Affecting Cell-Cycle Regulation and Apoptosis 334
- 4 Proto-Oncogenes and Tumor-suppressor Genes Are Altered in Cancer Cells 336
- 5 Cancer Cells Metastasize and Invade Other Tissues 339
- 6 Predisposition to Some Cancers Can Be Inherited 339
- 7 Viruses Contribute to Cancer in Both Humans and Animals 340
- 8 Environmental Agents Contribute to Human Cancers 341

GENETICS, TECHNOLOGY, AND SOCIETY

Breast Cancer: The Double-Edged Sword of Genetic Testing 342

CASE STUDY: I thought it was safe 343

Insights and Solutions 343

Problems and Discussion Questions 343

CHAPTER 17

Recombinant DNA Technology 346

- 1 Recombinant DNA Technology Began with Two Key Tools: Restriction Enzymes and DNA Cloning Vectors 347
- 2 DNA Libraries Are Collections of Cloned Sequences 352
- 3 The Polymerase Chain Reaction Is a Powerful Technique for Copying DNA 354
- 4 Molecular Techniques for Analyzing DNA 357
- 5 DNA Sequencing Is the Ultimate Way to Characterize DNA Structure at the Molecular Level 360

EXPLORING GENOMICS

Manipulating Recombinant DNA: Restriction Mapping and Designing PCR Primers 363

CASE STUDY: Should we worry about recombinant DNA technology? 364

Insights and Solutions 364

Problems and Discussion Questions 364

CHAPTER 18

Genomics, Bioinformatics, and Proteomics 367

- 1 Whole-Genome Shotgun Sequencing Is a Widely Used Method for Sequencing and Assembling Entire Genomes 368
- 2 DNA Sequence Analysis Relies on Bioinformatics Applications and Genome Databases 370
- 3 Functional Genomics Attempts to Identify Potential Functions of Genes and Other Elements in a Genome 372
- 4 The Human Genome Project Revealed Many Important Aspects of Genome Organization in Humans 373
- 5 The "Omics" Revolution Has Created a New Era of Biological Research 376
- 6 Comparative Genomics Analyzes and Compares Genomes from Different Organisms 380
- 7 Metagenomics Applies Genomics Techniques to Environmental Samples 386
- 8 Transcriptome Analysis Reveals Profiles of Expressed Genes in Cells and Tissues 386
- 9 Proteomics Identifies and Analyzes the Protein Composition of Cells 389
- 10 Systems Biology Is an Integrated Approach to Studying Interactions of All Components of an Organism's Cells 394

EXPLORING GENOMICS

Contigs, Shotgun Sequencing, and Comparative Genomics 395

CASE STUDY: Bioprospecting in Darwin's wake 396

Insights and Solutions 396

Problems and Discussion Questions 397

CHAPTER 19

Applications and Ethics of Genetic Engineering and Biotechnology 399

- 1 Genetically Engineered Organisms Synthesize a Wide Range of Biological and Pharmaceutical Products 400
- 2 Genetic Engineering of Plants Has Revolutionized Agriculture 403
- 3 Transgenic Animals with Genetically Enhanced Characteristics Have the Potential to Serve Important Roles in Biotechnology 405
- 4 Synthetic Genomes and the Emergence of Synthetic Biology 408
- 5 Genetic Engineering and Genomics Are Transforming Medical Diagnosis 409
- 6 Genome-Wide Association Studies Identify Genome Variations that Contribute to Disease 415
- 7 Gene Therapy Approaches for Treating Genetic Diseases 417
- 8 Genetic Engineering, Genomics, and Biotechnology Create Ethical, Social, and Legal Questions 420

GENETICS, TECHNOLOGY, AND SOCIETY

Personal Genome Projects and the Race for the \$1000 Genome 423

CASE STUDY: A first for gene therapy 423

Insights and Solutions 424

Problems and Discussion Questions 424

CHAPTER 20

Developmental Genetics 427

- 1 Evolutionary Conservation of Developmental Mechanisms Can Be Studied Using Model Organisms 427
- 2 Genetic Analysis of Embryonic Development in *Drosophila* Reveals How the Body Axis of Animals Is Specified 428
- 3 Zygotic Genes Program Segment Formation in *Drosophila* 431
- 4 Homeotic Selector Genes Specify Body Parts of the Adult 433
- 5 Plants Have Evolved Developmental Regulatory Systems That Parallel Those of Animals 436
- 6 *C. elegans* Serves as a Model for Cell-Cell Interactions in Development 438

GENETICS, TECHNOLOGY, AND SOCIETY

Stem Cell Wars 441

CASE STUDY: One foot or another 442

Insights and Solutions 442

Problems and Discussion Questions 442

CHAPTER 21**Quantitative Genetics and Multifactorial Traits 446**

- 1 Quantitative Traits Can Be Explained in Mendelian Terms 447
- 2 The Study of Polygenic Traits Relies on Statistical Analysis 449
- 3 Heritability Values Estimate the Genetic Contribution to Phenotypic Variability 452
- 4 Twin Studies Allow an Estimation of Heritability in Humans 455
- 5 Quantitative Trait Loci Can Be Mapped 457

GENETICS, TECHNOLOGY, AND SOCIETY

The Green Revolution Revisited: Genetic Research with Rice 460

CASE STUDY: A genetic flip of the coin 461

Insights and Solutions 461

Problems and Discussion Questions 462

CHAPTER 22**Population and Evolutionary Genetics 465**

- 1 Genetic Variation Is Present in Most Populations and Species 466
- 2 The Hardy–Weinberg Law Describes Allele Frequencies and Genotype Frequencies in Populations 467
- 3 The Hardy–Weinberg Law Can Be Applied to Human Populations 469
- 4 Natural Selection Is a Major Force Driving Allele Frequency Change 472
- 5 Mutation Creates New Alleles in a Gene Pool 475
- 6 Migration and Gene Flow Can Alter Allele Frequencies 476
- 7 Genetic Drift Causes Random Changes in Allele Frequency in Small Populations 477
- 8 Nonrandom Mating Changes Genotype Frequency but Not Allele Frequency 478
- 9 Reduced Gene Flow, Selection, and Genetic Drift Can Lead to Speciation 479
- 10 Phylogeny Can Be Used to Analyze Evolutionary History 481

GENETICS, TECHNOLOGY, AND SOCIETY

Tracking Our Genetic Footprints out of Africa 484

CASE STUDY: An unexpected outcome 485

Insights and Solutions 485

Problems and Discussion Questions 486

SPECIAL TOPICS IN MODERN GENETICS 1**Epigenetics 489**

Epigenetic Alterations to the Genome 489

BOX 1 The Beginning of Epigenetics 490

Epigenetics and Imprinting 492

Epigenetics and Cancer 494

BOX 2 What More We Need to Know about Epigenetics and Cancer 496

Epigenetics and the Environment 496

Epigenome Projects 497

Selected Readings and Resources 497

SPECIAL TOPICS IN MODERN GENETICS 2**DNA Forensics 499**

DNA Profiling Methods 499

BOX 1 The Pitchfork Case: The First Criminal Conviction Using DNA Profiling 500

BOX 2 Thomas Jefferson's DNA: Paternity and Beyond 504

BOX 3 The World Trade Center Attacks: Identifying Victims by DNA Profiling 505

BOX 4 The Pascal Della Zuana Case: DNA Barcodes and Wildlife Forensics 506

BOX 5 The Kennedy Brewer Case: Two Bite-Mark Errors and One Hit 507

Technical and Ethical Issues Surrounding DNA Profiling 508

Selected Readings and Resources 508

SPECIAL TOPICS IN MODERN GENETICS 3**Genomics and Personalized Medicine 510**

Personalized Medicine and Pharmacogenomics 510

BOX 1 The Story of Pfizer's Crizotinib 511

Personalized Medicine and Disease Diagnosis 514

BOX 2 The Pharmacogenomics Knowledge Base (PharmGKB): Genes, Drugs, and Diseases on the Web 515

Analyzing One Personal Genome 515

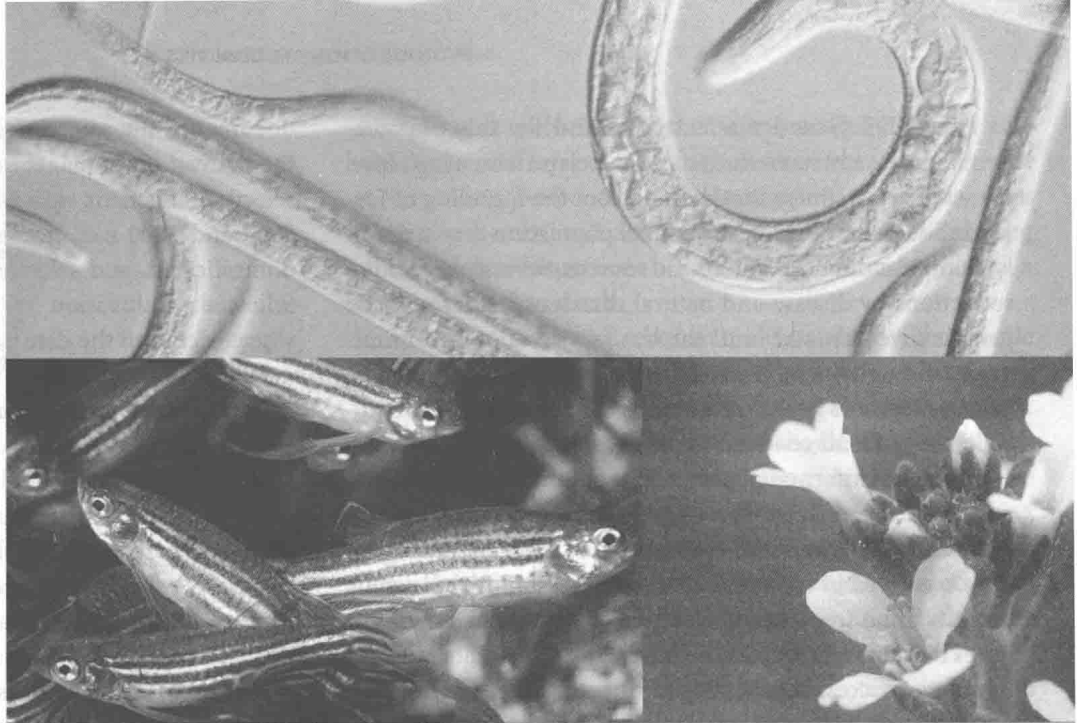
BOX 3 The Future of Personalized Genome Sequencing 517

Technical, Social, and Ethical Challenges 517

Selected Readings and Resources 518

GLOSSARY 520

Newer model organisms in genetics include the roundworm *C. elegans*, the zebrafish, *D. rerio*, and the mustard plant *A. thaliana*.



1

Introduction to Genetics

CHAPTER CONCEPTS

- Genetics in the twenty-first century is built on a rich tradition of discovery and experimentation stretching from the ancient world through the nineteenth century to the present day.
- Transmission genetics is the general process by which traits controlled by genes are transmitted through gametes from generation to generation.
- Mutant strains can be used in genetic crosses to map the location and distance between genes on chromosomes.
- The Watson-Crick model of DNA structure explains how genetic information is stored and expressed. This discovery is the foundation of molecular genetics.
- Recombinant DNA technology revolutionized genetics, was the foundation for the Human Genome Project, and has generated new fields that combine genetics with information technology.
- Biotechnology provides genetically modified organisms and their products that are used across a wide range of fields including agriculture, medicine, and industry.
- Model organisms used in genetics research are now utilized in combination with recombinant DNA technology and genomics to study human diseases.
- Genetic technology is developing faster than the policies, laws, and conventions that govern its use.

In December 1998, the Icelandic Parliament passed a law granting a biotechnology company, deCODE Genetics, a license to create and operate a database drawn from medical records of all of Iceland's 270,000 residents. The records in this Icelandic Health Sector Database (or HSD) were encoded to ensure anonymity. The new law also allowed deCODE Genetics to cross-reference the medical information from the HSD with a comprehensive genealogical database from the National Archives and to correlate information in these two databases with results from the analysis of deoxyribonucleic acid (DNA) samples collected from Icelandic donors. This combination of medical, genealogical, and genetic information forms a powerful resource available exclusively to deCODE Genetics for marketing to researchers and companies for a period of 12 years, ending in 2012.

This is not a science fiction scenario from a movie such as *Gattaca* but a real example of the increasingly complex interaction of genetics and society in the first decades of the twenty-first century. The development and use of these databases in Iceland have generated similar large-scale projects in Great Britain, Estonia, Latvia, the Kingdom of Tonga, and other countries. In the United States, smaller-scale programs, involving tens of thousands of individuals, are underway. All these databases will be used to search for susceptibility genes that control complex human diseases.