

CONCEPTS OF
GENETICS

TWELFTH EDITION

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Library of Congress Cataloging-in-Publication Data

Names: Klug, William S., author. | Cummings, Michael R., author. | Spencer,

Charlotte A., author. | Palladino, Michael A., author. | Killian, Darrell J., author.

Title: Concepts of genetics / William S. Klug (The College of New Jersey),

Michael R. Cummings (Illinois Institute of Technology), Charlotte A. Spencer, Michael A. Palladino (Monmouth University), Darrell J. Killian (Colorado College).

Description: Twelfth edition. | Hoboken, New Jersey : Pearson Education, Inc., 2018. | Includes index.

Identifiers: LCCN 2017047484 | ISBN 9780134604718

Subjects: LCSH: Genetics—Textbooks.

Classification: LCC QH430 .K574 2018 | DDC 572.8—dc23

LC record available at <https://lcn.loc.gov/2017047484>

Courseware Portfolio Manager: Michael Gillespie
Content Producer: Brett Coker
Managing Producer: Mike Early
Product Marketing Manager: Christa Pesek Pelaez
Courseware Director, Content Development: Ginnie Simone Jutson
Courseware Senior Analysts, Content Development: Sonia Divittorio and Barbara Price
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Rich Media Content Producer: Chloe Veylit
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Full-Service Project Managers: Michelle Gardner and Heidi Aguiar
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Illustrations: Imagineering Art
Art Coordinator: Stephanie Marquez
Design Manager: Maria Guglielmo
Cover and Interior Designer: Tamara Newnam
Rights & Permissions Project Manager: Angelica Aranas
Rights & Permissions Management: Eric Schrader
Photo Researcher: Eric Schrader
Manufacturing Buyer: Stacey J. Weinberger

Cover Illustration: H. Adam Steinberg, artforscience.com



ISBN 10: 0-134-60471-7; ISBN 13: 978-0-134-60471-8 (Student edition)
ISBN 10: 0-134-81892-X; ISBN 13: 978-0-134-81892-4 (Books à la Carte edition)
1 18
www.pearson.com

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Dedication

We dedicate this edition to our long-time colleague and friend Harry Nickla, who sadly passed away in 2017. With decades of experience teaching Genetics to students at Creighton University, Harry's contribution to our texts included authorship of the *Student Handbook and Solutions Manual* and the test bank, as well as devising most of the Extra Spicy problems at the end of each chapter. He was also a source of advice during the planning session for each new edition, and during our many revisions. We always appreciated his professional insights, friendship, and conviviality. We were lucky to have him as part of our team, and we miss him greatly.

WSK, MRC, CAS, MAP, and DJK

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Preface

It is essential that textbook authors step back and look with fresh eyes as each edition of their work is planned. In doing so, two main questions must be posed: (1) How has the body of information in their field—in this case, Genetics—grown and shifted since the last edition? (2) Which pedagogic innovations that are currently incorporated into the text should be maintained, modified, or deleted? The preparation of the 12th edition of *Concepts of Genetics*, a text well into its fourth decade of providing support for students studying in this field, has occasioned still another fresh look. And what we focused on in this new edition, in addition to the normal updating that is inevitably required, were three things:

1. The importance of continuing to provide comprehensive coverage of important, emerging topics.

In this regard, we continue to include a unique approach in genetics textbooks that offers readers a set of abbreviated, highly focused chapters that we label **Special Topics in Modern Genetics**. In this edition, these provide unique, cohesive coverage of six important topics: *CRISPR-Cas and Genomic Editing*, *DNA Forensics*, *Genomics and Precision Medicine*, *Genetically Modified Foods*, *Gene Therapy*, and *Advances in Neurogenetics: The Study of Huntington Disease*. The initial and final chapters in this series are both new to this edition.

2. The recognition of the vastly increased knowledge resulting from the study of gene regulation in eukaryotes.

To that end, the single chapter on this topic in previous editions has been expanded to three chapters: “Transcriptional Regulation in Eukaryotes” (Chapter 17), “Posttranscriptional Regulation in Eukaryotes” (Chapter 18), and “Epigenetic Regulation of Gene Expression” (Chapter 19). This extended coverage reflects many recent discoveries that reveal that RNA in many forms other than those that are essential to the process of transcription and translation (mRNA, tRNA, and rRNA) play critical roles in the regulation of eukaryotic gene activity. As well, it is now clear based on molecular studies related to epigenetics that this topic is best taught as an integral part of eukaryotic gene regulation. This new material provides the student exposure to modern coverage of a significant research topic.

3. The importance of providing an increased emphasis on ethical considerations that genetics is bringing into everyday life.

Regarding this point, we have converted the essay feature *Genetics, Technology, and Society* to one with added emphasis on ethics and renamed it *Genetics, Ethics, and Society*. Approximately half the chapters have new or revised essays. In addition, the feature called *Case Study*, which appears near the end of all chapters, has been recast with an increased focus on ethics. Both of these features increase the opportunities for active and cooperative learning.

Goals

In the 12th edition of *Concepts of Genetics*, as in all past editions, we have five major overarching goals. Specifically, we have sought to:

- Emphasize the basic concepts of genetics.
- Write clearly and directly to students, providing understandable explanations of complex, analytical topics.
- Maintain our strong emphasis on and provide multiple approaches to problem solving.
- Propagate the rich history of genetics, which so beautifully illustrates how information is acquired during scientific investigation.
- Create inviting, engaging, and pedagogically useful full-color figures enhanced by equally helpful photographs to support concept development.

These goals collectively serve as the cornerstone of *Concepts of Genetics*. This pedagogic foundation allows the book to be used in courses with many different approaches and lecture formats.

Writing a textbook that achieves these goals and having the opportunity to continually improve on each new edition has been a labor of love for all of us. The creation of each of the twelve editions is a reflection not only of our passion for teaching genetics, but also of the constructive feedback and encouragement provided by adopters, reviewers, and our students over the past four decades.

New to This Edition

New to this edition are four chapters. Two are Special Topics in Modern Genetics entries entitled “CRISPR-Cas and Genome Editing” and “Advances in Neurogenetics: The Study of Huntington Disease.” Both cover cutting-edge information and represent very recent breakthroughs in genetics. CRISPR, a genome-editing tool, is a straightforward technique that allows specific, highly accurate modification of DNA sequences within genes and is thus a powerful tool in the world of genetic research and gene therapy. In addition to this chapter, we call your attention to the introduction to Chapter 1 for an introduction to CRISPR and to also note that we have chosen this gene-editing system as the subject matter illustrated on the cover. Special Topics Chapter 6 illustrates the many of advances that have been made in the study of human neurogenetics. Huntington disease, a monogenic human disorder, has been subjected to analysis for over 40 years using every major approach and technique developed to study molecular genetics, and as such, exemplifies the growing body of information that has accrued regarding its causes, symptoms, and future treatment.

Additional new chapters arise from a major reorganization and expansion of our coverage of regulation of gene expression in eukaryotes, where we have split our previous coverage into three parts: transcriptional regulation (Chapter 17), posttranscriptional regulation (Chapter 18), and epigenetic regulation (Chapter 19). Chapter 18 includes much of the content previously contained in the Special Topics chapter *Emerging Roles of RNA* in the previous edition. Chapter 19, focused on epigenetics, is an expansion of the content previously contained in the *Epigenetics* Special Topics chapter from the previous edition.

Collectively, the addition of these four new chapters provides students and instructors with a much clearer, up-to-date presentation to these important aspects of genetics.

Continuing Pedagogic Features

We continue to include features that are distinct from, and go beyond, the text coverage, which encourage active and cooperative learning between students and the instructor.

- **Modern Approaches to Understanding Gene Function** This feature highlights how advances in genetic technology have led to our modern understanding of gene function. Appearing in many chapters, this feature prompts students to apply their analytical thinking skills, linking the experimental technology to the findings that enhance our understanding of gene function.
- **Genetics, Ethics, and Society** This feature provides a synopsis of an ethical issue related to a current finding in genetics that impacts directly on society today. It includes a section called *Your Turn*, which directs students to related resources of short readings and Web sites to support deeper investigation and discussion of the main topic of each essay.
- **Case Study** This feature, at the end of each chapter, introduces a short vignette of an everyday genetics-related situation, followed by several discussion questions. Use of the Case Study should prompt students to relate their newly acquired information in genetics to ethical issues that they may encounter away from the course.
- **Evolving Concept of the Gene** This short feature, integrated in appropriate chapters, highlights how scientists’ understanding of the gene has changed over time. Since we cannot see genes, we must infer just what this unit of heredity is, based on experimental findings. By highlighting how scientists’ conceptualization of the gene has advanced over time, we aim to help students appreciate the process of discovery that has led to an ever more sophisticated understanding of hereditary information.
- **How Do We Know Question** Found as the initial question in the *Problems and Discussion Questions* at the end of each chapter, this feature emphasizes the pedagogic value of studying how information is acquired in science. Students are asked to review numerous findings discussed in the chapter and to summarize the process of discovery that was involved.
- **Concept Question** This feature, found as the second question in the *Problems and Discussion Questions* at the end of each chapter, asks the student to review and comment on common aspects of the Chapter Concepts, listed at the beginning of each chapter. This feature places added emphasis on our pedagogic approach of conceptual learning.
- **Mastering Genetics** This robust online homework and assessment program guides students through complex topics in genetics, using in-depth tutorials that coach students to correct answers with hints and feedback specific to their misconceptions. New content for the 12th edition of *Concepts of Genetics* includes tutorials on emerging topics such as CRISPR-Cas, and Dynamic Study Modules, interactive flash cards that help students master basic content so they can be more prepared for class and for solving genetics problems.

New and Updated Topics

We have revised each chapter in the text to present the most current, relevant findings in genetics. Here is a list of some of the most significant new and updated topics covered in this edition.

Chapter 1: Introduction to Genetics

- New introductory vignette that discusses the discovery and applications of the genome-editing CRISPR-Cas system
- Updated section “We Live in the Age of Genetics”

Chapter 7: Sex Determination and Sex Chromosomes

- Updated content on the XIST gene product as a long noncoding RNA
- New insights about a novel gene involved in temperature-sensitive differentiation of snapping turtles and lizards, as well as the impact of climate change on sex, sex reversal, and sex ratios

Chapter 9: Extranuclear Inheritance

- Updated information on mtDNA disorders and nuclear DNA mismatches

Chapter 11: DNA Replication and Recombination

- New coverage of the role of telomeres in disease, aging, and cancer
- New and expanded coverage of telomeres and chromosome stability, explaining how telomeres protect chromosome ends

Chapter 13: The Genetic Code and Transcription

- New coverage on transcription termination in bacteria
- New section entitled “Why Do Introns Exist?”
- Updated coverage on RNA editing

Chapter 14: Translation and Proteins

- New coverage of eukaryotic closed-loop translation, including a new figure
- Revised coverage of Beadle and Tatum’s classic experiments
- Expanded coverage on the posttranslational modifications of proteins
- New coverage of the insights gleaned from the crystal structure of the human 80S ribosome

Chapter 15: Gene Mutation, DNA Repair, and Transposons

- New and revised coverage on transposons, focusing on the mechanisms of transposition by both retrotransposons and DNA transposons, as well as a

discussion of how transposition creates mutations.

Two new tables and five new figures are included

- Reorganization of the mutation classification section with table summaries
- New and expanded coverage of human germ-line and somatic mutation rates

Chapter 17: Transcriptional Regulation in Eukaryotes

- Revised chapter organization focuses specifically on transcriptional regulation
- Revised coverage of regulation of the *GAL* gene system in yeast with an updated figure
- New coverage on genetic boundary elements called insulators

Chapter 18: Posttranscriptional Regulation in Eukaryotes

- New chapter that greatly expands upon the previous coverage of posttranscriptional gene regulation in eukaryotes
- Revised and expanded coverage of alternative splicing and its relevance to human disease
- Expanded coverage on RNA stability and decay with a new figure
- Updated coverage of noncoding RNAs that regulate gene expression with a new figure
- Enriched coverage of ubiquitin-mediated protein degradation with a new figure

Chapter 19: Epigenetic Regulation of Gene Expression

- New chapter emphasizing the role of epigenetics in regulating gene expression, including coverage of cancer, transmission of epigenetic traits across generations, and epigenetics and behavior
- New coverage on the recently discovered phenomenon of monoallelic expression of autosomal genes
- Updated coverage of epigenome projects

Chapter 20: Recombinant DNA Technology

- Increased emphasis on the importance of whole-genome sequencing approaches
- New coverage of CRISPR-Cas as a gene editing approach, including a new figure
- Updated content on next-generation and third-generation sequencing

Chapter 21: Genomic Analysis

- Increased emphasis on the integration of genomic, bioinformatic, and proteomic approaches to analyzing genomes and understanding genome function

- A new section entitled “Genomic Analysis Before Modern Sequencing Methods,” which briefly summarizes approaches to mapping and identifying genes prior to modern sequencing
- Reorganized and revised content on the Human Genome Project. Updated content on personal genome projects and new content on diploid genomes and mosaicism and the pangenome to emphasize human genetic variations
- New coverage of the Human Microbiome Project including a new figure displaying microbiome results of patients with different human disease conditions
- New coverage of *in situ* RNA sequencing

Chapter 22: Applications of Genetic Engineering and Biotechnology

- Updated content on biopharmaceutical products including newly approved recombinant proteins, DNA vaccine trials to immunize against Zika virus, genetically modified organisms, and gene drive in mosquitos to control the spread of Zika
- New coverage of genes essential for life and how synthetic genomics is being applied to elucidate them. Clarification of prognostic and diagnostic genetics tests and the relative value of each for genetic analysis
- New content on DNA and RNA sequencing
- New section entitled “Screening the Genome for Genes or Mutations You Want,” which discusses how scientists can look at genetic variation that confers beneficial phenotypes
- New section entitled “Genetic Analysis by Personal Genomics Can Include Sequencing of DNA and RNA” that expands coverage of personal genome projects and new approaches for single-cell genetic analysis of DNA and RNA

Chapter 23: Developmental Genetics

- New section entitled “Epigenetic Regulation of Development”
- New coverage of DNA methylation and progressive restriction of developmental potential
- Expanded coverage of binary switch genes and regulatory networks

Chapter 24: Cancer Genetics

- Extended coverage of environmental agents that contribute to human cancers, including more information about both natural and human-made carcinogens
- New section entitled “Tobacco Smoke and Cancer” explaining how a well-studied carcinogen induces a wide range of genetic effects that may lead to mutations and cancer

- New section entitled “Cancer Therapies and Cancer Cell Biology,” describing the mechanisms of chemotherapies and radiotherapies as they relate to cancer cell proliferation, DNA repair, and apoptosis

Chapter 25: Quantitative Genetics and Multifactorial Traits

- Updated coverage on quantitative trait loci (QTLs)
- Revised and expanded section entitled “eQTLs and Gene Expression”

Chapter 26: Population and Evolutionary Genetics

- New coverage on vertebrate evolution
- New coverage of phylogenetic trees
- Updated coverage on the origins of the human genome
- New section entitled “Genotype and Allele Frequency Changes”
- New coverage on pre- and post-zygotic isolating mechanisms

Special Topic Chapter 1: CRISPR-Cas and Genome Editing

- New chapter on a powerful genome editing tool called CRISPR-Cas
- Up-to-date coverage on CRISPR-Cas applications, the patenting of this technology, and the ethical concerns of human genome editing

Special Topic Chapter 2: DNA Forensics

- New section on the still controversial DNA phenotyping method, including new explanations of how law-enforcement agencies currently use this technology

Special Topic Chapter 3: Genomics and Precision Medicine

- New section entitled “Precision Oncology,” including descriptions of two targeted cancer immunotherapies: adoptive cell transfer and engineered T-cell therapies
- Updated pharmacogenomics coverage, including a description of new trends in preemptive gene screening for pharmacogenomic variants as well as the pGEN4Kids program, a preemptive gene screening program that integrates DNA analysis data into patient electronic health records

Special Topic Chapter 4: Genetically Modified (GM) Foods

- New section entitled “Gene Editing and GM Foods” describing how scientists are using the new techniques of gene editing (including ZFN, TALENS, and CRISPR-Cas) to create GM food plants and animals,

and how these methods are changing the way in which GM foods are being regulated

- A new box entitled “The New CRISPR Mushroom” describing the development and regulatory approval of the first CRISPR-created GM food to be approved for human consumption

Special Topic Chapter 5: Gene Therapy

- Updated coverage of gene therapy trials currently underway
- Reordered chapter content to highlight emergence of CRISPR-Cas in a new section entitled “Gene Editing”
- Substantially expanded content on CRISPR-Cas including a brief summary of some of the most promising trials in humans and animals to date
- Incorporation of antisense RNA and RNA interference into a new section entitled “RNA-based Therapeutics,” including updated trials involving spinal muscular atrophy
- Updated content on roles for stem cells in gene therapy
- New content on combining gene editing with immunotherapy
- New ethical discussions on CRISPR-Cas and germline and embryo editing

Special Topic Chapter 6: Advances in Neurogenetics: The Study of Huntington Disease (HD)

- New chapter that surveys the study of HD commencing around 1970 up to the current time
- Coverage of the genetic basis and expression of HD, the mapping and isolation of the gene responsible for the disorder, the mutant gene product, molecular and cellular alterations caused by the mutation, transgenic animal models of HD, cellular and molecular approaches to therapy, and a comparison of HD to other inherited neurodegenerative disorders

Strengths of This Edition

- **Organization** —We have continued to attend to the organization of material by arranging chapters within major sections to reflect changing trends in genetics. Of particular note is the expansion of our coverage of the regulation of gene expression in eukaryotes, now reorganized into three chapters at the end of Part Three. Additionally, Part Four continues to provide organized coverage of genomics into three carefully integrated chapters.
- **Active Learning** —A continuing goal of this book is to provide features within each chapter that small groups of students can use either in the classroom or as assignments outside of class. Pedagogic research continues to support the value and effectiveness of such active and cooperative learning experiences. To this end, there are

four features that greatly strengthen this edition: *Case Study*; *Genetics, Ethics, and Society*; *Exploring Genomics*; and *Modern Approaches to Understanding Gene Function*. Whether instructors use these activities as active learning in the classroom or as assigned interactions outside of the classroom, the above features will stimulate the use of current pedagogic approaches during student’ learning. The activities help engage students, and the content of each feature ensures that they will become knowledgeable about cutting-edge topics in genetics.

Emphasis on Concepts

The title of our textbook—*Concepts of Genetics*—was purposefully chosen, reflecting our fundamental pedagogic approach to teaching and writing about genetics. However, the word “concept” is not as easy to define as one might think. Most simply put, we consider a concept to be *a cognitive unit of meaning—an abstract representation that encompasses a related set of scientifically derived findings and ideas*. Thus, a concept provides a broad mental image that, for example, might reflect a straightforward snapshot in your mind’s eye of what constitutes a chromosome; a dynamic vision of the detailed processes of replication, transcription, and translation of genetic information; or just an abstract perception of varying modes of inheritance.

We think that creating such mental imagery is the very best 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 and nearly impossible to forget. 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 topics about to be presented. Each chapter ends with a section called *Summary Points*, which enumerates the five to ten key points that have been discussed. And in the *How Do We Know?* question that starts each chapter’s problem set, students are asked to connect concepts to experimental findings. This question is then followed by a *Concept Question*, which asks the student to review and comment on common aspects of the Chapter Concepts. Collectively, these features help to ensure that students engage in, 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 our conceptual approach throughout the book.

Emphasis on Problem Solving

As authors and teachers, we have always recognized the importance of enhancing students' problem-solving skills. Students need guidance and practice if they are to develop into strong analytical thinkers. To that end, we present a suite of features in every chapter to optimize opportunities for student growth in the important areas of problem solving and analytical thinking.

- **Now Solve This** Found several times within the text of each chapter, each entry provides a problem similar to ones found at the end of the chapter that is closely related to the current text discussion. In each case, a pedagogic hint is provided to offer insight and to aid in solving the problem.
- **Insights and Solutions** As an aid to the student in learning to solve problems, the *Problems and Discussion Questions* section of each chapter is preceded by what has become an extremely popular and successful section. *Insights and Solutions* poses problems or questions and provides detailed solutions and analytical insights as answers are provided. The questions and their solutions are designed to stress problem solving, quantitative analysis, analytical thinking, and experimental rationale. Collectively, these constitute the cornerstone of scientific inquiry and discovery.
- **Problems and Discussion Questions** Each chapter ends with an extensive collection of *Problems and Discussion Questions*. These include several levels of difficulty, with the most challenging (*Extra-Spicy Problems*) located at the end of each section. Often, Extra-Spicy Problems are derived from the literature of genetic research, with citations. Brief answers to all even-numbered problems are presented in Appendix B. The *Student Handbook and Solutions Manual* answers every problem and is available to students whenever faculty decide that it is appropriate.
- **How Do We Know?** Appearing as the first entry in the *Problems and Discussion Questions* section, this question asks the student to identify and examine the experimental basis underlying important concepts and conclusions that have been presented in the chapter. Addressing these questions will aid the student in more fully understanding, rather than memorizing, the endpoint of each body of research. This feature is an extension of the learning approach in biology first formally described by John A. Moore in his 1999 book *Science as a Way of Knowing—The Foundation of Modern Biology*.
- **Mastering Genetics** Tutorials in Mastering Genetics help students strengthen their problem-solving skills while exploring challenging activities about key genetics

content. In addition, end-of-chapter problems are also available for instructors to assign as online homework. Students will also be able to access materials in the Study Area that help them assess their understanding and prepare for exams.

For the Instructor

Mastering Genetics— <http://www.masteringgenetics.com>

Mastering Genetics engages and motivates students to learn and allows you to easily assign automatically graded activities. Tutorials provide students with personalized coaching and feedback. Using the gradebook, you can quickly monitor and display student results. Mastering Genetics easily captures data to demonstrate assessment outcomes. Resources include:

- New Dynamic Study Modules, which are interactive flashcards, provide students with multiple sets of questions with extensive feedback so they can test, learn, and retest until they achieve mastery of the textbook material. These can be assigned for credit or used for self-study, and they are powerful preclass activities that help prepare students for more involved content coverage or problem solving in class.
- New tutorials on topics like CRISPR-Cas will help students master important, challenging concepts.
- In-depth tutorials that coach students with hints and feedback specific to their misconceptions
- An item library of thousands of assignable questions including end-of-chapter problems, reading quizzes, and test bank items. You can use publisher-created prebuilt assignments to get started quickly. Each question can be easily edited to match the precise language you use.
- Over 100 Practice Problems are like end-of-chapter questions in scope and level of difficulty and are found only in Mastering Genetics. Solutions are not available in the Student Solutions Manual, and the bank of questions extends your options for assigning challenging problems. Each problem includes specific wrong answer feedback to help students learn from their mistakes and to guide them toward the correct answer.
- eText 2.0 provides a dynamic digital version of the textbook, including embedded videos. The text adapts to the size of the screen being used, and features include student and instructor note-taking, highlighting, bookmarking, search, and hot-linked glossary.
- A gradebook that provides you with quick results and easy-to-interpret insights into student performance.

Downloadable Instructor Resources

The Instructor Resources for the 12th edition offers adopters of the text convenient access to a comprehensive and innovative set of lecture presentation and teaching tools. Developed to meet the needs of veteran and newer instructors alike, these resources include:

- The JPEG files of all text line drawings with labels individually enhanced for optimal projection results (as well as unlabeled versions) and all text tables.
- Most of the text photos, including all photos with pedagogical significance, as JPEG files.
- The JPEG files of line drawings, photos, and tables preloaded into comprehensive PowerPoint® presentations for each chapter.
- A second set of PowerPoint® presentations consisting of a thorough lecture outline for each chapter augmented by key text illustrations.
- PowerPoint® presentations containing a comprehensive set of in-class clicker questions for each chapter.
- An impressive series of concise instructor animations adding depth and visual clarity to the most important topics and dynamic processes described in the text.
- In Word and PDF files, a complete set of the assessment materials and study questions and answers from the testbank. Files are also available in TestGen format.

TestGen EQ Computerized Testing Software

(013483223X/9780134832234) Test questions are available as part of the TestGen EQ Testing Software, a text-specific testing program that is networkable for administering tests. It also allows instructors to view and edit questions, export the questions as tests, and print them out in a variety of formats.

For the Student

Student Handbook and Solutions Manual

(0134870085 / 9780134870083) Authored by Michelle Gaudette (*Tufts University*) and Harry Nickla (*Creighton University-Emeritus*). This valuable handbook provides a detailed step-by-step solution or lengthy discussion for every problem in the text. The handbook also features additional study aids, including extra study problems, chapter outlines, vocabulary exercises, and an overview of how to study genetics.

Mastering Genetics— <http://www.masteringgenetics.com>

Used by over one million science students, the Mastering platform is the most effective and widely used online

tutorial, homework, and assessment system for the sciences; it helps students perform better on homework and exams. As an instructor-assigned homework system, Mastering Genetics is designed to provide students with a variety of assessment tools to help them understand key topics and concepts and to build problem-solving skills. Mastering Genetics tutorials guide students through the toughest topics in genetics with self-paced tutorials that provide individualized coaching with hints and feedback specific to a student's individual misconceptions. Students can also explore the Mastering Genetics Study Area, which includes animations, the eText, *Exploring Genomics* exercises, and other study aids. The interactive eText 2.0 allows students to highlight text, add study notes, review instructor's notes, and search throughout the text.

Acknowledgments

Contributors

We begin with special acknowledgments to those who have made direct contributions to this text. First of all, we are pleased to acknowledge the work of Michelle Gaudette, who has assumed responsibility for writing the *Student Handbook and Solutions Manual* and the answers in Appendix B. We much appreciate this important contribution. We also thank Jutta Heller of the University of Washington—Tacoma, Christopher Halweg of North Carolina State University, Pamela Osenkowski of Loyola University—Chicago, and John Osterman of the University of Nebraska—Lincoln for their work on the media program. Steven Gorsich of Central Michigan University, Virginia McDonough of Hope College, Cindy Malone of California State University—Northridge, Pamela Marshall of Arizona State University West, and Brad Mehrrens of University of Illinois all made important contributions to the instructor resources program. We are grateful to all of these contributors not only for sharing their genetic expertise, but for their dedication to this project as well as the pleasant interactions they provided.

Proofreaders and Accuracy Checking

Reading the manuscript of an 800+ page textbook deserves more thanks than words can offer. Our utmost appreciation is extended to Ann Blakey, *Ball State University*, Jutta Heller, *University of Washington—Tacoma*, and Valerie Oke, *University of Pittsburgh*, who provided accuracy checking of many chapters, and to Kay Brimeyer, who proofread the entire manuscript. They confronted this task with patience and diligence, contributing greatly to the quality of this text.

Reviewers

All comprehensive texts are dependent on the valuable input provided by many reviewers. While we take full responsibility for any errors in this book, we gratefully acknowledge

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the help provided by those individuals who reviewed the content and pedagogy of this edition:

Kyle Belozarov, York University
Ann Blakey, Ball State University
Michelle Boissiere, Xavier University of Louisiana
Gregory Booton, The Ohio State University
Mary Bryk, Texas A&M University
Soochin Cho, Creighton University
Claire Cronmiller, University of Virginia
Johnny El-Rady, University of South Florida
Christy Fillman, University of Colorado—Boulder
Edison Fowlks, Hampton University
Shaghayegh Harbi, New York University
Christopher Harendza, Montgomery County
Community College
Benjamin Harrison, University of Alaska—Anchorage
Steven Karpowicz, University of Central Oklahoma
David Kass, Eastern Michigan University
Oliver Kerscher, The College of William and Mary
Kirkwood Land, University of the Pacific
Howard Laten, Loyola University Chicago
Haiying Liang, Clemson University
Te-Wen Lo, Ithaca College
Kari Loomis, University of Massachusetts—Amherst
TyAnna Lovato, University of New Mexico
Matthew Marcello, Pace University
Pamela Marshall, Arizona State University West
Jeff Maughan, Brigham Young University
Virginia McDonough, Hope College
Craig Miller, University of California—Berkeley
Anna Newman, University of Houston
Pamela Osenkowski, Loyola University Chicago
Stephen Page, Community College of Baltimore County
Dennis Ray, The University of Arizona
Jennifer Schisa, Central Michigan University
Randall Small, The University of Tennessee—Knoxville
Paul Small, Eureka College
Doug Thrower, University of California—Santa Barbara

Harald Vaessin, The Ohio State University
Meena Vijayaraghavan, Tulane University

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

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. Brett Coker, our Content Producer, has worked tirelessly to keep the project on schedule and to maintain our standards of high quality. Sonia DiVittorio provided detailed feedback in her role as developmental editor. In addition, our editorial team—Ginnie Simone Jutson, Director of Content Development; Barbara Price, Senior Development Editor; Margot Otway, Senior Development Editor; Sarah Jensen, Senior Content Developer for Mastering Genetics; and Chloe Veylit, Rich Media Producer—have 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. Summer Giles, Editorial Assistant, helped manage and edit the instructor resources. Shaghayegh Harbi authored engaging, insightful tutorials for Mastering Genetics. Michelle Gardner supervised all the production intricacies with great attention to detail and perseverance. Outstanding copyediting was performed by Lucy Mullins, for which we are most grateful. Kelly Galli and Christa Pelaez have professionally and enthusiastically managed the marketing of the text. Finally, the beauty and consistent presentation of the artwork are the product of Imagineering of Toronto. Without the work ethic and dedication of the above individuals, the text would never have come to fruition.