

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, as well as to students majoring in a number of other disciplines, including agriculture, animal husbandry, chemistry, nursing, 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 edition of *Essentials of Genetics*, the two most important goals have been to introduce pedagogic innovations that enhance learning and to provide carefully updated, highly accessible coverage of genetic topics of both historical and modern significance. As new tools and findings of genetics research continue to emerge rapidly and grow in importance in the study of all subdisciplines of biology, instructors face tough choices about what content is truly essential as they introduce the discipline to novice students. We have thoughtfully revised each chapter in light of this challenge, by selectively scaling back the detail or scope of coverage in the more traditional chapters in order to provide expanded coverage and broader context for the more modern, cutting-edge topics. Our aim is to continue to provide efficient coverage of the fundamental concepts in transmission and molecular genetics that lay the groundwork for more in-depth coverage of emerging topics of growing importance—in particular, the many aspects of the genomic revolution that is already relevant to our day-to-day lives as well as the relatively new findings involving epigenetics and noncoding RNAs.

While we have adjusted this edition to keep pace with changing content and teaching practices, we remain dedicated to the core principles that underlie this book. 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

In addition to streamlining core chapters and updating information throughout the text, key improvements to this edition include three additional chapters in the Special Topics in Modern Genetics unit, end of chapter questions in Special Topics chapters, and a new feature exploring scientists' evolving understanding of the concept of the gene.

- **Special Topics in Modern Genetics** We have been pleased with the popular reception to the Special Topics in Modern Genetics chapters. Our goal has been to provide abbreviated, cohesive coverage of important topics in genetics that are not always easily located in textbooks. Professors have used these focused, flexible chapters in a multitude of ways: as the backbone of lectures, as inspiration for student assignments outside of class, and as the basis of group assignments and presentations.

New to this edition are chapters on topics of great significance in genetics:

- Emerging Roles of RNA
- Genetically Modified Foods
- Gene Therapy

For all Special Topics chapters, we have added a series of questions that send the student back into the chapter to review key ideas or that provide the basis of personal contemplations and group discussions.

- **Evolving Concept of the Gene** Also new to this edition is a short feature, integrated in appropriate chapters, that highlights how scientists' understanding of a 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.

- **Concepts Question** A new 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 Key Concepts, listed at the beginning of each chapter. This feature places added emphasis on our pedagogic approach of conceptual learning.
- **MasteringGenetics** This powerful 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 *Essentials of Genetics* includes a robust library of Practice Problems—found only in MasteringGenetics—that are like end of chapter questions in scope and difficulty. These questions include wrong answer feedback specific to a student's error, helping build students' problem-solving and critical thinking skills. These problems are not included in the Student Solution Manual and offer a valuable new resource of problems for practice or for credit.

New and Updated Topics

While 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 new and updated topics present in this edition.

Ch. 1: Introduction to Genetics • New chapter introduction vignette emphasizing translational medicine

Ch. 2: Mitosis and Meiosis • Updated coverage of kinetochore assembly and the concept of disjunction • Expanded coverage of checkpoints in cell cycle regulation

Ch. 4: Modification of Mendelian Ratios • New section on mitochondria, human health, and aging

Ch. 5: Sex Determination and Sex Chromosomes • Updated coverage on paternal age effects (PAEs) in humans • New content regarding the primary sex ratio in humans

Ch. 6: Chromosome Mutations • New information on Fragile X Syndrome and the *FMRI* gene • New information regarding gene families as linked to gene duplications

Ch. 7: Linkage and Chromosome Mapping in Eukaryotes • Introduction of "sequence maps" in humans based on the use of DNA markers

Ch. 10: DNA Replication and Recombination • Updated coverage of DNA Pol III holoenzyme • Revised figures involving DNA synthesis • New coverage of the initiation of bacterial DNA synthesis • New information on DNA recombination • New coverage of replication of telomeric DNA • Revision of the GTS essay: Telomeres: The Key to Immortality

Ch. 11: Chromosome Structure and DNA Sequence Organization • Updated coverage of chromatin remodeling • New information on H3 histone substitution in centromeric DNA • New coverage regarding the transcript of Alu sequences

Ch. 12: The Genetic Code and Transcription • Extended coverage of promoter elements in eukaryotes • Introduction of the process of RNA editing • Revision of figures involving ribosomes and transcription

Ch. 13: Translation and Proteins • Revision of all ribosome figures • New information on initiation, elongation during translation in eukaryotes

Ch. 14: Gene Mutation, DNA Repair, and Transposition • Reorganization and updates for mutation classification • Updated coverage of xeroderma pigmentosum and DNA repair mechanisms

Ch. 15: Regulation of Gene Expression • Updated coverage of gene regulation by riboswitches • Expanded coverage of chromatin modifications • Updated coverage of promoter and enhancer structures and functions • Updated coverage of the mechanisms of transcription activation and repression

Ch. 16: The Genetics of Cancer • New coverage of the progressive nature of colorectal cancers • Revised and updated coverage of driver and passenger mutations

Ch. 17: Recombinant DNA Technology • Streamlined content on recombinant DNA techniques to deemphasize older techniques and focus on more modern methods • New figure on FISH • Expanded coverage on next-generation and third-generation sequencing • New section on gene-targeting approaches includes content and figures on gene knockout animals and transgenic animals • Revised PDQ content

Ch. 18: Genomics, Bioinformatics, and Proteomics • Updated content on the Human Microbiome Project • New content introducing exome sequencing • Updated content on personal genome projects • Revised and expanded coverage of the Encyclopedia of DNA Elements (ENCODE) Project • New figure on genome sequencing technologies • New Case Study on the microbiome as a risk factor for disease

Ch. 19: Applications and Ethics of Genetic Engineering and Biotechnology • New section on synthetic biology for bioengineering applications • New material and figure on deducing fetal genome sequences from maternal blood • Revised and updated content on prenatal genetic testing • Moved content on GM crops to ST 5 • Moved content on gene therapy to ST 6 • Updated discussion on synthetic genomes • Revised and streamlined content on DNA microarrays given the changing role of microarrays in gene testing (relative to whole-genome, exome, and RNA sequencing) • New content on genetic analysis by sequencing individual

genomes for clinical purposes and single-cell sequencing

- Revised ethics section to include additional discussion on the analysis of whole-genome sequences, preconception testing, DNA patents, and destiny predictions
- Major revision of end of chapter questions
- New GTS essay on the privacy and anonymity of genomic data
- New Case Study on genetically modified bacteria for cancer treatment

Ch. 20: Developmental Genetics • New introductory section on the key steps to the differentiated state • New section on the role of binary switch genes and regulatory programs in controlling organ formation, including new figures

Ch. 21: Quantitative Genetics and Multifactorial Traits • New section on limitations of heritability studies • Updated coverage of multifactorial genotypes and expanded coverage of the tomato genome and implications for future improvement in tomato strains • Revised coverage of eQTLs

Ch. 22: Population and Evolutionary Genetics

- Revised and updated section on detecting genetic variation and the application of new technology to detect variation in DNA and in genomes
- Extensively revised and updated section on the process of speciation
- The section on use of phylogenetics to investigate evolutionary history has been improved and expanded with new examples
- Information on human evolution has been completely revised and updated with new information about the genomics of extinct human species and their relationship to our species
- Five new figures have been added throughout the chapter to accompany the added text

Special Topic 1: Epigenetics • Heavily revised section on imprinting • New ideas on the role of epigenetics in cancer accompany the coverage of the role of somatic mutation in cancer • New section on epigenetic modification of behavior in model organisms and humans

Special Topic 2: Emerging Roles of RNA • New chapter that focuses on the recently discovered functions of RNAs with an emphasis on noncoding RNAs • An introduction to CRISPR/Cas technology in gene editing • Explanation of mechanisms of microRNA and long noncoding RNA gene regulation • Discussion of extracellular RNAs in cell–cell communication and disease diagnosis • Coverage of RNA-induced transcriptional silencing

Special Topic 3: DNA Forensics • New coverage describing how DNA can be inadvertently transferred to a crime scene, leading to false arrests • New coverage of DNA phenotyping

Special Topic 4: Genomics and Personalized Medicine • New coverage on personal genomics and cancer, including a new story of one person's successful experience using “omics” profiling to select a personalized cancer treatment • Updated coverage of personalized

medicine and disease diagnostics • Updated coverage of recent studies using “omics” profiles to predict and monitor disease states

Special Topic 5: Genetically Modified Foods • New chapter on genetically modified foods—the genetic technology behind them, the promises, debates, and controversies

Special Topic 6: Gene Therapy • New chapter on the modern aspects of gene therapy • Provides up-to-date applications of gene therapy in humans

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 Ninth 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 numerous 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 many 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 Studies** 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.

For the Instructor

MasteringGenetics—

<http://www.masteringgenetics.com>

MasteringGenetics 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. MasteringGenetics easily captures data to demonstrate assessment outcomes. Resources include:

- In-depth tutorials that coach students with hints and feedback specific to their misconceptions.
- A new, robust library of **Practice Problems** offers more opportunities to assign challenging problems for student homework or practice. These questions include targeted wrong answer feedback to help students learn from their mistakes. They appear only in MasteringGenetics, and solutions are not included in the Student Solutions Manual.
- An item library of assignable questions including end of chapter problems, test bank questions, and reading quizzes. You can use publisher-created prebuilt assignments to get started quickly. Each question can be easily edited to match the precise language you use.
- A gradebook that provides you with quick results and easy-to-interpret insights into student performance.

Instructor Resource DVD

(0-134-14365-5)



The Instructor Resource DVD offers adopters of the text convenient access to 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.
- An impressive series of concise instructor animations adding depth and visual clarity to the most important topics and dynamic processes described in the text.
- The instructor animations preloaded into PowerPoint® presentation files for each chapter.

- PowerPoint® presentations containing a comprehensive set of in-class Classroom Response System (CRS) questions for each chapter.
- In Word files, a complete set of the assessment materials and study questions and answers from the test bank, the text's in-chapter text questions, and the student media practice questions.

TestGen EQ Computerized Testing Software (0-134-20303-8)

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

Authored by Harry Nickla, Creighton University (Emeritus)
(0-134-18998-1)

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.

MasteringGenetics—

<http://www.masteringgenetics.com>

Used by over a million science students, the Mastering platform is the most effective and widely used online tutorial, homework, and assessment system for the sciences. Perform better on exams with MasteringGenetics. As an instructor-assigned homework system, MasteringGenetics is designed to provide students with a variety of assessments to help them understand key topics and concepts and to build problem-solving skills. MasteringGenetics 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 MasteringGenetics' Study Area, which includes animations, the eText, *Exploring Genomics* exercises, and other study aids. The interactive eText 2.0 allows students to access their text on mobile devices, highlight text, add study notes, review instructor's notes, and search throughout the text, 24/7.

Acknowledgments

Contributors

We begin with special acknowledgments to those who have made direct contributions to this text. Foremost, we are pleased to thank Dr. Darrell Killian of Colorado College for writing the Special Topic chapter on Emerging Roles of RNA. We much appreciate this important contribution.

We also thank Christy Filman of the University of Colorado—Boulder, Jutta Heller of the University of Washington—Tacoma, Christopher Halweg of North Carolina State University, Pamela Osenkowski of Loyola University—Chicago, John Osterman of the University of Nebraska—Lincoln, and Fiona Rawle of the University of Toronto—Mississauga for their work on the media program. Virginia McDonough of Hope College and Cindy Malone of California State University—Northridge contributed greatly to the instructor resources. We also express special thanks to Harry Nickla, recently retired from Creighton University. In his role as author of the *Student Handbook and Solutions Manual* and the test bank, he has reviewed and edited the problems at the end of each chapter and has written many of the new entries as well. He also provided the brief answers to selected problems that appear in the Appendix.

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 detailed manuscript of textbook deserves more thanks than words can offer. Our utmost appreciation is extended to Michelle Gaudette, Tufts University, and Kirkwood Land, University of the Pacific, who provided accuracy checking of many chapters, and to Joanna Dinsmore, 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 the help provided by those individuals who reviewed the content and pedagogy of this edition:

Soochin Cho, *Creighton University*; Mary Colavito, *Santa Monica College*; Kurt Elliott, *Northwest Vista College*; Edison Fowlks, *Hampton University*; Yvette Gardner, *Clayton State University*; Theresa Geiman, *Loyola University—Maryland*; Christopher Harendza, *Montgomery County Community College*; Lucinda Jack, *University of Maryland*; David Kass, *Eastern Michigan University*; Kirkwood Land, *University of the Pacific*; Te-Wen Lo, *Ithaca College*; Matthew Marcello, *Pace University*; Virginia McDonough, *Hope College*; Amy McMillan, *SUNY Buffalo State*; Sanghamitra Mohanty, *University of Texas—Austin*; Sudhir Nayak, *The College of New Jersey*; Pamela Osenkowski, *Loyola University—Chicago*; John Osterman, *University of Nebraska—Lincoln*; Pamela Sandstrom, *University of Nevada—Reno*; Adam Sowalsky, *Northeastern University*; Brian Stout, *Northwest Vista College*; James D. Tucker, *Wayne State University*; Jonathan Visick, *North Central College*; Fang-Sheng Wu, *Virginia Commonwealth University*; Lev Yampolsky, *East Tennessee State University*

Special thanks go to Mike Guidry of LightCone Interactive and Karen Hughes of the University of Tennessee for their original contributions to the media program.

As these 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. 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. 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 team—Ginnie Simione-Jutson, Executive Director

of Development, Chloé Veylit, Media Producer, and Tania Mlawer, Director of Editorial Content for MasteringGenetics—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. Sudhir Nayak of The College of New Jersey provided outstanding work for the MasteringGenetics program and his input regarding genomics is much appreciated. Margaret Young and Rose Kernan 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 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.

Acknowledgments

We begin with special appreciation to those who have made direct contributions to this text. For their help, we are pleased to thank Dr. Robert K. Miller of Colorado College for his special contribution to the text. We also appreciate the contributions of those who 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. Sudhir Nayak of The College of New Jersey provided outstanding work for the MasteringGenetics program and his input regarding genomics is much appreciated. Margaret Young and Rose Kernan 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 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.