

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; and (2) What pedagogic innovations might be devised and incorporated into the text that will unquestionably enhance students' learning? The preparation of the 11th edition of *Concepts of Genetics*, a text now entering 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 two things: (1) the need to increase the opportunities for instructors and students to engage in **active and cooperative learning approaches**, either within or outside of the classroom; and (2) the need to provide more **comprehensive coverage of important, emerging topics** that do not yet warrant their own traditional chapters.

Regarding the first point, and as discussed in further detail below, we have added a new feature called **Modern Understanding of Gene Function**, which appears within many chapters. In addition, we have retained the very popular **Genetics, Technology, and Society** essays that appear at the end of many chapters. This feature includes an active learning format called *Your Turn*, which directly engages the student with provocative assignments. These features are in addition to **Exploring Genomics** entries, and together, these all may serve as the basis for interactions between small groups of students, either in or out of the classroom. Regarding emerging topics, 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 uniquely cohesive coverage of six important topics: *Epigenetics*, *Emerging Roles of RNA*, *Genomics and Personalized Medicine*, *DNA Forensics*, *Genetically Modified Foods*, and *Gene Therapy*. Three of these (*RNA*, *GM Foods*, and *Gene Therapy*) are new to this edition.

Goals

In the 11th edition of *Concepts of Genetics*, as in all past editions, we have five major goals. Specifically, we have sought to:

- Emphasize the basic concepts of genetics.

- Write clearly and directly to students in order to provide 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 us. The creation of each of the eleven 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 three decades.

New to This Edition

- **Special Topics in Modern Genetics**—We have been pleased with the popular reception that the Special Topics in Modern Genetics chapters has received. First introduced in the tenth edition, 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 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*, and *Gene Therapy*. For all Special Topic chapters, we have added a series of questions that send the student back into the chapter to review key ideas or provide the basis of personal contemplations and group discussions.

- **Modern Approaches to Understanding Gene Function**—This new feature highlights how advances in genetic technology have led to our modern understanding of gene

function. Appearing in many chapters, this feature also prompts students to apply their analytical thinking skills, linking the experimental technology to the findings that enhance our understanding of gene function.

- **Evolving Concept of the Gene** Also new to this edition is a short feature, integrated in appropriate chapters, that highlights how scientists' understanding of what a gene is 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.
- **Concept 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 Chapter Concepts, listed at the beginning of each chapter. This feature places added emphasis on our pedagogic approach of conceptual learning.
- **Neurogenetics** A major change that is evident in the Table of Contents involves Chapter 24. Previously entitled Behavior Genetics, this chapter has been reworked and redefined to reflect the wealth of information regarding the impact of genetics on the field of neurobiology, linking genetic analysis to brain function and brain disorders. Thus, the fully revised Chapter 24 is now entitled Neurogenetics.
- **MasteringGenetics** 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 *Concepts of Genetics 11e* includes a library of Practice Problems that are like end of chapter questions and only appear in MasteringGenetics. These problems offer a valuable extension of questions available for assignments. These questions include wrong answer feedback to help students learn from their mistakes.

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. 3: Mendelian Genetics

- New Understanding Gene Function section: Identifying Mendel's Gene for Regulating White Flower Color in Peas
- New end-of-chapter problems based on Migaloo, the albino hump-backed whale

Ch. 7: Sex Determination and Sex Chromosomes

- New coverage on paternal age effects (PAEs) in humans
- New Understanding Gene Function section: *Drosophila Sxl* Gene Induces Female Development

Ch. 8: Chromosome Mutations: Variation in Number and Arrangement

- Updated coverage of fragile-X syndrome
- New Understanding Gene Function section: Mouse Models of Down Syndrome

Ch. 9: Extranuclear Inheritance

- New coverage on mitochondrial swapping and the prevention of mtDNA-based disorders

Ch. 11: 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 coverage of homologous recombination and gene conversion
- New coverage of replication of telomeric DNA
- New Understanding Gene Function section: Lethal Knockouts of DNA Ligase Genes

Ch. 12: DNA Organization in Chromosomes

- New coverage of kinetochore structure and function

Ch. 14: Translation and Proteins

- Revision of all ribosome figures

Ch. 15: Gene Mutation, DNA Repair, and Transposition

- New data on spontaneous mutation rates in humans.
- Reorganization and updates for mutation classification
- Updated coverage of xeroderma pigmentosum and DNA repair mechanisms
- New Understanding Gene Function section: Transposon-Mediated Mutations Reveal Genes Involved in Colorectal Cancer

Ch. 16: Regulation of Gene Expression in Prokaryotes

- Updated coverage of gene regulation by riboswitches

Ch. 17: Regulation of Gene Expression in Eukaryotes

- Expanded coverage of chromatin modifications
- Introduction of Weintraub and Groudine's experiments involving chromatin structure and transcription regulation
- Updated coverage of promoter and enhancer structures and functions
- Updated coverage of the mechanisms of transcription activation and repression
- Updated section on RNA silencing
- New section: ENCODE Data Are Transforming Our Concepts of Eukaryotic Gene Regulation
- New Understanding Gene Function section: MicroRNAs Regulate Ovulation in Female Mice

Ch. 18: Developmental Genetics

- New section: Binary Switch Genes and Signaling Pathways, including four new figures
- New Understanding Gene Function section: Single-Gene Signaling Mechanism Reveals Secrets to Head Regeneration in Planaria

Ch. 19: Cancer and Regulation of the Cell Cycle

- New coverage of the progressive nature of colorectal cancers
- Revised and updated coverage of driver and passenger mutations
- Expanded coverage of the role of viruses in human cancers

Ch. 20: Recombinant DNA Technology

- Expanded coverage and new figure on third-generation DNA sequencing
- New section on gene targeting that includes content and figures on gene knockout animals, conditional knockouts, and transgenic animals

Ch. 21: Genomics, Bioinformatics, and Proteomics

- Updated coverage of the Human Microbiome Project
- New content introducing exome sequencing
- Updated coverage of personal genome projects
- Revised and expanded coverage of the Encyclopedia of DNA Elements (ENCODE) Project
- New content and figure covering chromatin-immunoprecipitation (ChIP) and ChIP-sequencing (ChIPSeq)
- New Case Study discussing the microbiome as a risk factor for disease

Ch. 22: Applications and Ethics of Genetic Engineering and Biotechnology

- New section and figure on Synthetic Biology for Bioengineering Applications

- New coverage and figure on deducing fetal genome sequences from maternal blood
- Updated coverage of prenatal genetic testing
- Updated coverage of the minimal genome
- New coverage involving 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 PDQ content and additional new questions
- New Genetics, Technology, and Society essay: Privacy and Anonymity of Genomic Data
- New Case Study on cancer-killing bacteria

Ch. 23: Quantitative Genetics and Multifactorial Traits

- Updated coverage of quantitative trait loci (QTL) in studying multifactorial phenotypes

Ch. 24: Neurogenetics

- Conversion of the Behavior Genetics chapter to one entitled Neurogenetics, with emphasis on molecular events in the brain and nervous system related to behavior. Includes 10 new figures and 3 new tables.
- New Understanding Gene Function section: *RbAp48* and a Potential Molecular Mechanism for Age-Related Memory Loss

Ch. 25: Population and Evolutionary Genetics

- New coverage on macroevolution and the rate of speciation
- Revision of the discussion of the molecular clock
- Two new sections: The Complex Origins of Our Genome and Our Genome Is a Mosaic

Special Topic Chapter 1: Epigenetics

- New section: MicroRNAs and Long Non-Coding RNAs

Special Topic Chapter 2: Emerging Roles of RNA

- New chapter on the newly discovered emerging roles of RNA—focuses on the diverse functions of RNAs with an emphasis on noncoding RNA

Special Topic Chapter 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 Chapter 4: Genomics and Personalized Medicine

- New coverage on personal genomics and cancer, including 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 Chapter 5: Genetically Modified Foods

- New chapter on genetically modified foods—the genetic technology behind them, the promises, debates, and controversies

Special Topic Chapter 6: Gene Therapy

- New chapter on the modern aspects of gene therapy—provides up-to-date applications of gene therapy in humans

Glossary

- Many definitions refined
- Approximately two dozen new terms

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 which, 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. 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. 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 feature new to this edition called *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 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.

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 Part IV, which combines the chapter providing foundational coverage of Recombinant DNA Technology with two chapters involving Genomics.
- **Pedagogy**—As discussed above, one of the major pedagogic goals of this edition 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 five features that greatly strengthen this edition.
 - **Case Study** This feature, at the end of each chapter, introduces a short vignette (a "Case") of an everyday encounter related to genetics, followed by a series of discussion questions. Use of the Case Study should prompt students to relate their newly acquired information in genetics to issues that they may encounter away from the course.
 - **Genetics, Technology, and Society** This feature provides a synopsis of a topic related to a current finding in genetics that impacts directly on our current society. It now includes a new 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.
 - **Exploring Genomics** This feature extends the discussion of selected topics present in the chapter by exploring new findings resulting from genomic studies. Students are directed to Web sites that provide the "tools" that research scientists around the world rely on for current genomic information.

Whether instructors use these activities as active learning in the classroom or as assigned interactions outside of class, the above features will stimulate the use of current pedagogic approaches to students' 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 Problem Solving

As authors and teachers, we have always recognized the importance of teaching students how to become effective problem solvers. Students need guidance and practice if they are to develop strong analytical thinking skills. 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 those 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. This feature closely links the text discussion to 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 or 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 current literature of genetic research, with citations. Brief answers to all even-numbered problems are presented in Appendix B.
- **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 end-point 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*.

- **MasteringGenetics** Tutorials in MasteringGenetics 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

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
- 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.
- A new category of Practice Problems are like end of chapter questions in scope and level of difficulty and are found only in MasteringGenetics. 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.
- A gradebook that provides you with quick results and easy-to-interpret insights into student performance

TestGen EQ Computerized Testing Software

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

MasteringGenetics—

<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, MasteringGenetics 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. 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 the MasteringGenetics Study Area, which includes animations, the eText, *Exploring Genomics* exercises, and other study aids. The interactive eText allows students to highlight text, add study notes, review instructor's notes, and search throughout the text.

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