

Preface

A Note from the Authors

The science of genetics is less than 150 years old, but its accomplishments within that short time have been astonishing. Gregor Mendel first described genes as abstract units of inheritance in 1865; his work was ignored and then “rediscovered” in 1900. Thomas Hunt Morgan and his students provided experimental verification of the idea that genes reside within chromosomes during the years 1910–1920. By 1944, Oswald Avery and his coworkers had established that genes are made of DNA. James Watson and Francis Crick published their pathbreaking structure of DNA in 1953. Remarkably, less than 50 years later (in 2001), an international consortium of investigators deciphered the sequence of the 3 billion nucleotides in the human genome. Twentieth century genetics made it possible to identify individual genes and to understand a great deal about their functions.

Today, scientists are able to access the enormous amounts of genetic data generated by the sequencing of many organisms’ genomes. Analysis of these data will result in a deeper understanding of the complex molecular interactions within and among vast networks of genes, proteins, and other molecules that help bring organisms to life. Finding new methods and tools for analyzing these data will be a significant part of genetics in the twenty-first century.

Our fourth edition of *Genetics: From Genes to Genomes* emphasizes both the core concepts of genetics and the cutting-edge discoveries, modern tools, and analytic methods that will keep the science of genetics moving forward.

Our Focus—An Integrated Approach

Genetics: From Genes to Genomes represents a new approach to an undergraduate course in genetics. It reflects the way we, the authors, currently view the molecular basis of life.

We integrate:

- **Formal genetics:** the rules by which genes are transmitted.
- **Molecular genetics:** the structure of DNA and how it directs the structure of proteins.

- **Digital analysis, genomics, and proteomics:** recent technologies that allow a comprehensive analysis of the entire gene set and its expression in an organism.
- **Human genetics:** how genes contribute to health and diseases, including cancer.
- **The unity of life-forms:** the synthesis of information from many different organisms into coherent models.
- **Molecular evolution:** the molecular mechanisms by which biological systems and whole organisms have evolved and diverged.
- **Systems biology:** the multidisciplinary, integrated study of life processes that may lead to new ways to analyze, detect, and treat disease.

The strength of this integrated approach is that students who complete the book will have a strong command of genetics as it is practiced today by both academic and corporate researchers. These scientists are rapidly changing our understanding of living organisms, including ourselves. Ultimately, this vital research may create the ability to replace or correct detrimental genes—those “inborn errors of metabolism,” as researcher Archibald Garrod called them in 1923, as well as the later genetic alterations that lead to the many forms of cancer.

The Genetic Way of Thinking

Modern genetics is a molecular-level science, but an understanding of its origins and the discovery of its principles is a necessary context. To encourage a genetic way of thinking, we begin the book by reviewing Mendel’s principles and the chromosomal basis of inheritance. From the outset, however, we aim to integrate organism-level genetics with fundamental molecular mechanisms.

Chapter 1 presents the foundation of this integration by summarizing the main biological themes we explore. In Chapter 2, we tie Mendel’s studies of pea-shape inheritance to the action of an enzyme that determines whether a pea is round or wrinkled. In the same chapter, we point to the relatedness of the patterns of heredity in all organisms. Chapters 3–5 cover extensions to Mendel, the chromosome theory of inheritance, and the fundamentals of gene linkage and mapping. Starting in Chapter 6, we focus on the physical characteristics of DNA, on mutations, and on how DNA encodes, copies, and transmits biological information.

Beginning in Chapter 9, we move into the digital revolution in DNA analysis with a look at modern genetics techniques, including gene cloning, hybridization, PCR, and microarrays. We explore how bioinformatics, an emergent analytical tool, can aid in string matching and in discovery of genome and proteome features.

The understanding of molecular and computer-based techniques carries into our discussion of chromosome specifics in Chapters 12–14, and also informs our analysis of gene regulation in Chapters 15, 16, and 17, the last of which provides an in-depth discussion of the cell cycle and its disruption in cancers. Chapter 18 describes the use of genetic tools at the molecular level to uncover the complex interactions of eukaryotic development.

Chapters 19 and 20 cover population genetics, with a view of how molecular tools have provided information on species relatedness and on genomes changes at the molecular level over time. Finally, in Chapter 21 we explore systems biology, an integrated field utilizing input from several disciplines. We consider the impact a systems approach could have on the identification and treatment of disease.

Throughout our book, we present the scientific reasoning of some of the ingenious researchers of the field—from Mendel, to Watson and Crick, to the collaborators on the Human Genome Project. We hope student readers will see that genetics is not simply a set of data and facts, but also a human endeavor that relies on contributions from exceptional individuals.

Student-Friendly Features

We have taken great pains to help the student make the leap to a deeper understanding of genetics. Numerous features of this book were developed with that goal in mind.

- **One Voice Genetics:** *Genes to Genomes* has a friendly, engaging reading style that helps students master the concepts throughout this book. The writing style provides the student with the focus and continuity required to make the book successful in the classroom.
- **Visualizing Genetics** The highly specialized art program developed for this book integrates

photos and line art in a manner that provides the most engaging visual presentation of genetics available. Our Feature Figure illustrations break down complex processes into step-by-step illustrations that lead to greater student understanding. All illustrations are rendered with a consistent color theme—for example, all presentations of phosphate groups are the same color, as are all presentations of mRNA.

- **Accessibility** Our intention is to bring cutting-edge content to the student level. A number of more complex illustrations are revised and segmented to help the student follow the process. Legends have been streamlined to highlight only the most important ideas, and throughout the book, topics and examples have been chosen to focus on the most critical information.
- **Problem Solving** Developing strong problem-solving skills is vital for every genetics student. The authors have carefully created problem sets at the end of each chapter that allow students to improve upon their problem-solving ability.
- **Solved Problems** which cover topical material with complete answers provide insight into the step-by-step process of problem solving.
- **Review Problems** offer more than 600 questions involving a variety of levels of difficulty that develop excellent problem-solving skills. The problems are organized by chapter section and in order of increasing difficulty within each section for ease of use by instructors and students. Answers to selected problems are in the back of the book. The companion Study Guide and Solutions Manual by Debra Nero (available separately) provides detailed analysis of strategies to solve all of the end-of-chapter problems.

Problems

Vocabulary

1. For each of the terms in the left column, choose the best matching phrase in the right column.

a. reciprocal translocation	1. lacking one or more chromosomes or having one or more extra chromosomes
b. gynandromorph	2. movement of short DNA elements
c. pericentric	3. having more than two complete sets of chromosomes
d. paracentric	4. exact exchange of parts of two nonhomologous chromosomes
e. euploids	5. excluding the centromere
f. polyploidy	6. including the centromere
g. transposition	7. having complete sets of chromosomes
h. aneuploids	8. mosaic combination of male and female tissue

Section 13.1

2. For each of the following types of chromosomal aberrations, tell: (i) whether an organism heterozygous for the aberration will form any type of loop in the chro-

mosomes during prophase I of meiosis; (ii) whether a chromosomal bridge can be formed during anaphase I in a heterozygote, and if so, under what condition; (iii) whether an acentric fragment can be formed during anaphase I in a heterozygote, and if so, under what condition; (iv) whether the aberration can suppress meiotic recombination; and (v) whether the two chromosomal breaks responsible for the aberration occur on the same side or on opposite sides of a single centromere, or if the two breaks occur on different chromosomes.

- reciprocal translocation
- paracentric inversion
- small tandem duplication
- Robertsonian translocation
- paracentric inversion
- large deletion

3. In flies that are heterozygous for either a deletion or a duplication, there will be a looped-out region in a preparation of polytene chromosomes. How could you distinguish between a deletion or a duplication using polytene chromosome analysis?