

Preface

Genetics on the Personal Level

Genetics, like other sciences, is built around units: a DNA nucleotide, a gene, a chromosome, a cell, a family, and a population. But the truly important unit of inheritance is the person.

The person has been the focus of this book through its seven editions, even as research has shifted from a single gene approach to considering entire genomes. With this revision, however, came an experience that has opened my eyes in a new way to the connections among the various levels of genetics. I have come to know a young man who has Huntington disease. My new friend is unable to speak or move, but he communicates with what remains of his facial expression, especially his eyes. I am astonished at how an extra bit of DNA out on a tip of the fourth largest chromosome can slowly drown a small part of the brain in protein, diminishing the body while leaving the person intact. The mutation reverberates through my friend's family, striking many through the generations.

Knowing someone with an inherited disease renews my wonder at how, most of the time, our 24,000 or so genes work just fine, and their variants create the distinctions that make us individuals. New to this edition, glimpses of personal impacts of genetics open each chapter. They span the familiar (infections and lactose intolerance), the exceedingly rare (inborn errors of metabolism), the practical (forensics) to the just weird (improving pig manure). All capture attention and catapult the reader into the chapter material. Within the chapters, a feature called Relating the Concepts refocuses the discussion on the initial vignette. Other personal touches from editions past remain, such as "In Their Own Words" boxed essays, in-text examples, and questions, problems, and cases at the end of each chapter.

When we step back from individual views of genetics, a bigger picture emerges. Reading 16.1 encapsulates much recent research in *What Makes Us Human?* It continues ideas from the opening essay, which evokes a time when more than one type of hominid roamed the earth. The final chapter considers the human genome in the context of other species with whom we share the planet and includes Figure 22.9, a periodic table-like chart comparing selected organisms whose genomes have been sequenced. I look forward to updating it as we learn more.

What Sets This Book Apart

Unique Writing Style

From the beginning, the clarity, flavor, and immediacy of *Human Genetics: Concepts and Applications* has been uniquely interesting and accessible to non-scientist readers. Into the book flows my 25 years of experience as a journalist, 20 years as a genetic counselor, and my PhD background in genetics. As a frequent contributor to the magazine *The Scientist* for 15 years, I have had access to scientific meetings and researchers that are not available to everyone. From my network of sources, I learn of new research results before they are published. This allows me to keep *Human Genetics: Concepts and Applications* current—and that's vital for a scientific field that is racing ahead as fast as genetics/genomics.

My approach is straightforward: present the essential concepts in clear language, then demonstrate them with the very best, sometimes quirky, examples. Students appreciate this approach to writing and learn from it.

Pedagogical Tools

Pedagogical aids ensure that students can identify and understand basic concepts and

relate the many examples and cases. Chapters open with an outline and an essay. At the end of each major section, **Key Concepts** reinforce important core material. **Relating the Concepts** questions are embedded in the narrative, where they connect chapter content to the opening essay. **Chapter Summaries** review content, calling attention to important vocabulary.

Each chapter ends with a great variety of **Review Questions** to measure content knowledge and **Applied Questions** to practice using that knowledge. Both sets of questions are written to engage students in understanding the mechanisms of genetics and enable them to master content from a basic to a more advanced level—often with humor. Following the Applied Questions are **Cases and Research Results**, many of them new to this edition. Answers to all questions are at the end of the book.

Web Activities encourage students to dig deeper. They provide an opportunity to find the newest genetic information and to use some of the latest tools and databases in genetic analysis.

Dynamic Art

Art Presented in Steps Illustrating an entire biological process in one figure can overwhelm students and hinder understanding. Therefore, in this edition, appropriate figures have been reorganized, and summary tables and step-by-step figures have been added where appropriate to help students focus on the important concepts.

For example, building a polypeptide from genetic instructions lies at the crux of understanding genetics, and is a difficult topic more easily learned in pieces. The figures in Chapter 10, *Gene Action: From DNA to Protein*, clarify concepts by gradually explaining the various components of the concept.

- Orientations of DNA and mRNA are uniform in all figures in chapter 10.

- New figure 10.6 is small but important: it depicts the physical relationships among tRNA, mRNA, and amino acid.
- In previous editions, figure 10.7 showed 3 stages of transcription in part a and simultaneous transcription in part b. It is now 2 figures. Similarly, translation elongation and termination, once a single figure, are now two.
- New figure 10.19 summarizes the entire process: From DNA to RNA to protein.
- The final section, on protein folding, now includes a table of disorders—kuru might be unfamiliar, but Alzheimer's disease and Parkinson's disease are not.

Chapter 14, *When Allele Frequencies Stay Constant*, has two new figures (14.4 and 14.5) that calculate carrier frequencies for two disorders. Figure 15.9 in chapter 15, *Changing Allele Frequencies*, uses colored shapes to trace how nonrandom mating, migration, genetic drift, mutations, and natural selection change allele frequencies, driving evolution. This new edition retains the entire figure, but also breaks it down, introducing each component separately before the culmination.

Comparisons Used to Aid Understanding

In contrast to art presented in steps, some concepts in genetics reflect dichotomies, and so presenting them together can provide clarity. Figure 5.13, for example, contrasts independent assortment and linkage. Figure 7.10 and the accompanying discussion distinguish a linkage study from an association study. Table 14.5 compares VNTRs and STRs, two classes of DNA repeats used in DNA profiling.

New to This Edition

- Each chapter now opens with a **case study** that provides students with a glimpse of the personal impact of genetics.
- **Relating the Concepts** sections pose questions related to the opening case study, so students apply concepts to real-life situations.
- **New and updated information has been integrated throughout the text**, giving students a modern perspective of human genetics.

Chapter 1, *Overview of Genetics*

- Boxed reading introduces DNA structure and function, so instructors can easily cover DNA or Mendel first.
- New section on metagenomics discusses sampling DNA from ecosystems.

Chapter 4, *Mendelian Inheritance*

- Illustrations reworked so the short and tall pea plants are easier to distinguish.
- Several new end-of-chapter questions.

Chapter 5, *Exceptions and Extensions to Mendel's Laws*

- New figure compares independent assortment to linkage.

Chapter 8, *The Genetics of Behavior*

- Fuller explanation of neuron function begins the chapter.

Chapter 14, *When Allele Frequencies Stay Constant*

- Section on forensics is rewritten for clarity and currency.
- Chapter 15, *Changing Allele Frequencies*
 - *Reading, Antibiotic Resistance: Genomics to the Rescue?* revised to include information on methicillin-resistant staph aureus (MRSA) and microbial genome sequences that determine how resistance arises.
 - Figure 15.9 from the sixth edition, *Forces that change allele frequencies*, is broken down into its parts at the appropriate section of text, and the parts reunited into a complete figure at the end of the chapter.

Chapter 18, *Genetics of Cancer*

- New section, table, and figures on Origins of Cancer Cells.

Chapter 19, *Genetic Technologies*

- **Reorganized** to cover patenting, amplifying, modifying, and monitoring DNA.

Chapter 20, *Genetic Testing, Genetic Counseling, and Gene Therapy*

- Emphasizes genetic testing, counseling, and privacy.

Chapter 22, *The Age of Genomics*

- Offers more information on comparative genomics including a new figure: "A periodic table of genomes" that highlights 9 sequenced (non-human) genomes.
- **Unparalleled coverage of stem cells** Dr. Lewis' specialty is stem cells, and coverage in this text reflects her frequent conversations with leading researchers in this field. The information and illustrations on stem cells are unparalleled.
 - In Chapter 2, *Cells*, four illustrations accompany the clear narrative, progressing from basic to applied views of stem cells. Figures 2.23 and 2.24 stress the defining characteristic of self-renewal, as well as depicting the route to differentiation of daughter cells. Figure 2.25 walks through somatic cell nuclear transfer, and figure 2.26 considers how stem cells can heal a heart.
 - The stem cell theme continues in figure 11.4, which depicts development of the pancreas into a uniquely dual structure—from a single type of progenitor cell.
 - A new section in Chapter 18, *Genetics of Cancer*, explores the role of stem cells in causing cancer, with several new figures (18.8, 18.9, 18.10).
- **New problems, cases, and research questions** are scattered throughout the text. The popular **Solving a Problem** feature introduced in the last edition includes new problems.
- **Focus Placed on the Important Concepts** Fewer bold terms and a more concise glossary emphasize the important terms—if a term isn't directly related to genetics, or if it is a disorder name, it isn't emphasized for memorization. At the same time, all disorders are identified by their Online Mendelian Inheritance in Man number, to ease acquiring further information. Suggested Readings have been moved to the Online Learning Center, providing space for more end-of-chapter exercises.

Teaching and Learning Supplements

McGraw-Hill offers various tools and teaching products to support the seventh edition of *Human Genetics: Concepts and Applications*. Students can order supplemental study materials by contacting their local bookstore. Instructors can obtain teaching aids by calling the Customer Service Department at 800-338-3987, visiting the text website at www.mhhe.com/lewisgenetics7, or contacting your local McGraw-Hill sales representative.

Digital Content Manager

This multimedia collection of visual resources allows instructors to utilize artwork from the text in multiple formats to create customized classroom presentations, visually based tests and quizzes, dynamic course website content, or attractive printed support materials. The digital assets on this cross-platform CD-ROM are grouped by chapter within the following easy-to-use folders:

- **Art Library**—All textbook art in a format compatible with presentation or word processing software.
- **Photo Library**—All photos from the textbook are available in color.
- **PowerPoint Presentations**—Ready-made presentations cover each chapter of the textbook.
- **Active Art Library**—Key figures from the textbook are saved in manipulable layers that can be isolated and customized to meet the needs of the lecture environment. Build images from simple to complex to suit your lecture style.
- **Animations Library**—Numerous full-color animations of key processes are provided. Harness the visual impact of processes in motion by importing these files into classroom presentations or course websites.

Instructor Testing and Resource CD-ROM (ITRCD)

The ITRCD is a cross-platform CD-ROM providing a wealth of resources for the instructor. Supplements featured on this

CD-ROM include a computerized test bank utilizing McGraw-Hill's EZ Test. EZ Test is a flexible and easy-to-use electronic testing program. The program allows instructors to create tests from book-specific items. It accommodates a wide range of question types, and instructors may add their own questions. Multiple versions of the test can be created and any test can be exported for use with course management systems such as WebCT, BlackBoard or PageOut. The program is available for Windows and Macintosh environments.

Other assets on the ITRCD are grouped within easy-to-use folders. The Instructor's Manual is available, and Word files of the test bank are included for those instructors who prefer to work outside of the test generator software.

Instructor's Manual

The Instructor's Manual, prepared by William Perry Baker of Midwestern University, is available on the ITRCD or through the Instructor Resources of the Online Learning Center, (www.mhhe.com/lewisgenetics7). The manual includes chapter outlines and overviews, a chapter-by-chapter resource guide to use of visual supplements, answers to questions in the textbook, additional questions and answers for each chapter, and Internet resources and activities.

Overhead Transparencies

A set of 100 full-color transparencies showing key illustrations from the textbook is available for adopters.

For the Student

Genetics: From Genes to Genomes CD-ROM

This easy-to-use CD covers the most challenging concepts in the course and makes them more understandable through presentation of full-color animations and interactive exercises.

Online Learning Center (OLC)

Get online at www.mhhe.com/lewisgenetics7. The OLC offers an extensive array of learning and teaching tools. Explore this dynamic site

designed to help you get ahead and stay ahead in your study of human genetics. Some of the activities you will find on the website include:

- Self-quizzes to help you master material in each chapter
- Flash cards to ease learning of new vocabulary
- Case studies to practice application of your knowledge of human genetics
- Links to resource articles, popular press coverage, and support groups

Case Workbook to accompany Human Genetics, seventh edition, by Ricki Lewis

This workbook is specifically designed to support the concepts presented in *Human Genetics* through real cases adapted from recent scientific and medical journals, interviews, and meetings. The workbook has been extensively updated and provides practice for constructing and interpreting pedigrees; applying Mendel's laws; reviewing the relationships of DNA, RNA, and proteins; analyzing the effects of mutations; evaluating phenomena that distort Mendelian ratios; designing gene therapies; and applying new genomic approaches to understanding inherited disease. A special set of exercises at the end of the workbook link concepts across chapters. An answer key is available for the instructor.

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