

Preface

Human Genetics for Everyone

Truth is indeed stranger than fiction. When I began writing this textbook 15 years ago with a glimpse of a future where two college roommates take tailored genetic tests, I could never have imagined that today we would be ordering such tests from websites. We send in our DNA on cheek swabs or in saliva samples to learn about our genetic selves. We may receive risk estimates of future health concerns, or take ancestry tests that reveal our pasts, noting which parts of the world our forebears likely came from and maybe even who our distant cousins are. I'm amazed.

Ricki Lewis

Today, human genetics is for everyone. It is about our variation more than about our illnesses, and increasingly about the common rather than the rare. Once an obscure science or an occasional explanation for an odd collection of symptoms, human genetics is now part of everyday conversation. At the same time, it is finally being recognized as the basis of medical science. Despite the popular tendency to talk of “a gene for” this or that, we now know that for most traits and illnesses, several to many genes interact with each other and environmental influences. By coming to know our genetic backgrounds, we can control our environments in more healthful ways. Genetic knowledge is, therefore, both informative and empowering. This book shows you how and why this is true.

What Sets this Book Apart

Current Content

As a member of the Information and Education Committee of the American Society of Human Genetics, an instructor of “Genethics,” genetic counselor, and long-time science writer, Dr. Lewis is aware of research news and government policy changes before they are published. The most exciting new developments find their way into each edition of *Human Genetics: Concepts and Applications*, sometimes in the words of the people they directly affect. A few of the most compelling updates to this edition include

- Direct-to-consumer genetic testing
- Genome-wide association (GWA) studies: promises and perils
- Gene expression profiling and personalized medicine
- Human microbiome project
- Human variation and ancestry
- GINA (Genetic Information Nondiscrimination Act)
- Induced pluripotent stem cells (reprogramming)

This new edition also reflects the shift in focus in the field of human genetics from rare single-gene inheritance to more common multifactorial traits and disorders.

The Human Touch

Human genetics is about people, and their voices echo throughout these pages. Most are real, some are composites, and many are based on the author’s experience as a science writer, genetic counselor, and hospice volunteer.

Compelling Stories and Case Studies Lewis enlivens her clear presentation of genetic concepts with compelling stories and cases like the following:

- A young fashion magazine editor keeping her leukemia at bay thanks to a drug developed through genetic research (Ch. 18, p. 366)
- A man freed from a 25-year prison term following reconsideration of DNA evidence (Ch. 14, p. 265)
- A father whose little girl has a condition so rare that it doesn’t even have a name (Ch. 4, p. 69)

Practical Application of Human Genetics Recognizing that the goal of most introductory science courses is to better inform future voters and consumers, the author provides practical application of the content to students’ lives. Topics of particular interest to students include:

- The role that genes play in disease susceptibility, physical characteristics, body weight, and behaviors, with an eye toward the dangers of genetic determinism
- Biotechnologies, including genetic testing, gene therapy, stem cell therapy, gene expression profiling, genome-wide association studies, and personalized medicine
- Ethical concerns that arise from the interface of genetic information and privacy, such as infidelity testing, ancestry testing, and direct-to-consumer genetic testing

The Lewis Guided Learning System

Each chapter is framed with a set of pedagogical features designed to reinforce the key ideas in the chapter and prompt students to think more deeply about the application of the content they have just read.

Dynamic Art

Outstanding photographs and dimensional illustrations, vibrantly colored, are featured throughout *Human Genetics*. Students will learn from a variety of figure types, including process figures with numbered steps, micro to macro representations, and the combination of art and photos to relate stylized drawings to real-life structures.

New to this Edition!

New and updated information is integrated throughout the chapters, and a few features from past editions have been moved. Highlights from the revision are included here.

Chapter 1 *Overview of Genetics*

- Updates on the Genetic Information Nondiscrimination Act and the Human Microbiome Project
- New Figure 1.8 Diseaseome—diseases are connected in unexpected ways
- New *Bioethics: Choices for the Future*, “Genetic Testing and Privacy”

Chapter 2 *Cells*

- Stem cell coverage now stresses reprogrammed cells, with two new figures and a new *Bioethics: Choices for the Future*, “Should You Bank Your Stem Cells?”
- New *In Their Own Words*, “A Little Girl with Giant Axons”

Chapter 4 *Single-gene Inheritance*

- New chapter opener “His Daughter’s DNA,” about a father’s quest to solve a genetic mystery
- New section 4.1, A Tale of Two Families

Chapter 5 *Beyond Mendel’s Laws*

- New Reading 5.1, “The Genetic Roots of Alzheimer Disease”
- New Table 5.3, Types of Genetic Markers

Chapter 6 *Matters of Sex*

- New chapter opener, “A Controversial Hypothesis: Mental Illness, Mom, and Dad”
- New Reading 6.2, “Rett Syndrome—A Curious Inheritance Pattern”

Chapter 7 *Multifactorial Traits*

- New Figure 7.1, Anatomy of a trait—rare single-gene disorders versus common SNP patterns
- New section 7.4, Genome-wide association studies (including new figures 7.9 and 7.11)

Chapter 8 *Genetics of Behavior*

- New section 8.5, How nicotine is addictive and raises cancer risk
- New section 8.8, Autism (includes new Figure 8.9, Understanding autism)

Chapter 9 *DNA Structure and Replication*

- New *Bioethics: Choices for the Future*, “Infidelity Testing”

Chapter 11 *Gene Expression and Epigenetics*

- New Figure 11.7, Control of gene expression (transcription factors and microRNAs)
- New text on the evolving definition of a gene

Chapter 12 *Gene Mutation*

- New chapter opening case study, “The Amerithrax Story”
- New Figure 12.1, Animal models of human diseases
- New Figure 12.11, Using copy number variants in healthcare

Chapter 13 *Chromosomes*

- New *Bioethics: Choices for the Future*, “The Denmark Study: Screening for Down Syndrome”

Chapter 16 *Human Ancestry*

- New *Bioethics: Choices for the Future*, “Indigenous Peoples”
- Expanded coverage of markers, haplogroups, and migration
- New Reading 16.2 “Should You Take a Genetic Ancestry Test?”

Chapter 17 *Genetics of Immunity*

- Shortened and reorganized to stress genetics

Chapter 18 *Genetics of Cancer*

- New Table 18.2, Processes and Pathways Affected in Cancer
- The cancer genome

Chapter 19 *Genetic Technologies: Amplifying, Modifying, and Monitoring DNA*

- Expanded and updated information on DNA patents
- New section 19.5, Silencing DNA (RNAi, antisense, and knockouts)

Chapter 20 *Genetic Testing and Treatment*

- New section 20.1, “Geneticists find zebras, and some horses” (including new figure 20.1)
- New information on direct-to-consumer tests and CLIA regulations
- Gene therapy to treat hereditary blindness in an 8-year-old

Chapter 22 *Genomics*

- New chapter opener, “20,000 Genomes and Counting”
- New Reading 22.1, “The First Three Humans to Have Their Genomes Sequenced”

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This book continually evolves thanks to input from instructors and students. Please let me know your thoughts and suggestions for improvement. (ralewis@nycap.rr.com)