

Preface

Human Genetics Comes Full Circle

I like to peek at the ending to a novel. I just can't wait to know what will happen to the characters. Human genetics has become the same way, and so I invite you to read the very last section of this tenth edition now, at the beginning. It is about the personal genome sequencing of one man, and how he is using the information. As the cost of such sequencing plummets, we all may be able to look to our genomes for echoes of our pasts and hints of our futures—if we so choose. We may also learn what we can do to counter our inherited tendencies and susceptibilities. Genetic knowledge is informative and empowering. This book shows you how and why this is true.

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 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 genes interact with each other and environmental influences.

What Sets This Book Apart

Current Content

The exciting narrative writing style, with clear explanations of concepts and mechanisms propelled by stories, reflects Dr. Lewis's eclectic experience as a science writer, instructor, and genetic counselor, coupled with her expertise in genetics. Updates to this landmark edition include

- Genetic tests, from preconception to old age
- From Mendel to molecules: family genome analysis
- Allelic diseases: one gene, two diseases
- Denisova hominins, coexisting with us and Neanderthals
- Disease-in-a-dish stem cell technology
- The reemergence of gene therapy
- Personal genome sequencing: promises and limitations

This edition continues the shift in focus from rare single-gene inheritance to more common multifactorial traits and diseases.

The Human Touch

Human genetics is about people, and their voices echo throughout these pages.

Compelling Stories and Cases When the parents of children with visual loss stood up at a conference to meet other families with the same very rare inherited disease, Dr. Lewis was there, already composing the opening essay to chapter 5. She met the little girl in Reading 2.2, who is 1 in 51 in the world with her disease, as well as the magazine editor in Reading 18.1, who was one of the first to benefit from a wonder drug for her leukemia. In the photo that starts chapter 20, Dr. Lewis reads birthday cards to Max, who had gene therapy for Canavan disease. Readers of earlier editions of this book made the cards for Max, who has appeared here since age 3. The experiences of real people flesh out the science of genetics.

Practical Application of Human Genetics A working knowledge of the principles and applications of human genetics is critical to being an informed citizen and health care consumer. Broad topics of particular interest include

- The roles that genes play in disease risk, physical characteristics, and behavior, with an eye toward the dangers of genetic determinism
- Biotechnologies, including next-generation DNA sequencing, genetic testing, stem cell technology, gene expression profiling, genome-wide association studies, gene therapy, familial DNA searches, whole exome sequencing, cell-free fetal DNA testing, and personal genome sequencing
- Ethical concerns that arise from the interface of genetic information and privacy.

The Lewis Guided Learning System

Each chapter begins with two views of the content. “*Learning Outcomes*” embedded in the table of contents guide the student in mastering content. “*The Big Picture*” encapsulates the overall theme of the chapter. Content flows logically through three to five major sections per chapter that are peppered with high-interest boxed readings (“*In Their Own Words*,” “*Readings*,” “*Bioethics: Choices for the Future*,” and “*Technology Timelines*”). End-of-chapter pedagogy progresses from straight recall to applied and creative questions and challenges. Overall the pedagogical features of each chapter reinforce key ideas and prompt students to think more deeply about the application of the content.

Dynamic Art

Outstanding photographs and dimensional illustrations, vibrantly colored, are featured throughout *Human Genetics*. Figure types include 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. Highlights from the revision are included here.

Chapter 1 *Overview of Genetics*

- A university encourages genetic testing of incoming freshmen—until stopped.

Chapter 2 *Cells*

- A “disease-in-a-dish” for a rare condition opens the chapter.

Chapter 3 *Meiosis and Development*

- Can sirtuins slow aging?

Chapter 4 *Single-Gene Inheritance*

- Family genome analysis bridges Mendelian and molecular genetics.

Chapter 5 *Beyond Mendel's Laws*

- Genetic heterogeneity is seen through the eyes of families at a conference for hereditary blindness.

Chapter 6 *Matters of Sex*

- Wiskott-Aldrich syndrome opens the chapter.
- Hemophilia B affected the royal families, not hemophilia A.

Chapter 7 *Multifactorial Traits*

- “Genetics of Athletics” opens the chapter.
- More emphasis on epigenetics.
Faces: a compelling example of normal variation revealed with genome-wide association studies.

Chapter 8 *Genetics of Behavior*

- Behavioral disorders may not be distinct at the genetic level.
- How are autism and schizophrenia related?

Chapter 10 *Gene Action: From DNA to Protein*

- “Whole exome sequencing” opens the chapter.

Chapter 11 *Gene Expression and Epigenetics*

- How famine sowed the seeds of schizophrenia, thanks to epigenetics.

Chapter 12 *Gene Mutation*

- Restoring protein folding to treat cystic fibrosis opens the chapter.
- Allelic diseases explain old mysteries.

Chapter 14 *Constant Allele Frequencies*

- Forensics update: touch DNA, familial DNA searches, fish fraud.

Chapter 15 *Changing Allele Frequencies*

- Update on dog and cat genetics.
- “Mutiny on the Bounty” and migraine: a profound founder effect.

Chapter 16 *Human Ancestry*

- Exploiting the Havasupai in *Bioethics: Choices for the Future*.
- Neanderthal genome reveals we interbred.
- Denisova hominins shared Europe with our ancestors and Neanderthals.
- DNA of the Khoisan is the most diverse.
- A newly discovered paleo-Eskimo from Greenland reveals an ancient migration route.

Chapter 17 *Genetics of Immunity*

- The Berlin patient: curing leukemia and HIV.
- A new view of allergy, due to mutation.

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

- Experimental evolution and the Gulf oil spill.

Chapter 20 *Genetic Testing and Treatment*

- Max Randell turns 13, thanks to gene therapy for Canavan disease.
- Preconception comprehensive carrier testing for 400+ diseases.
- Cell-free fetal DNA reveals entire fetal genomes.
- Repurposed drugs treat common genetic diseases.
- Gene therapy returns with three successes.

Chapter 21 *Reproductive Technologies*

- “The Twiblings” open the chapter.
- The ethics of postmortem gamete retrieval, male and female.

Chapter 22 *Genomics*

- Updates on genome projects.
- Sequencing the *Cacao* genome (chocolate).
- The human microbiome.
- How useful is a personal genome sequence?