

Preface

Human Genetics Touches Us All

When I wrote the first edition of this book, in 1992, I could never have imagined that today, thousands of people would have had their genomes sequenced. Nor could I have imagined, when the first genomes were sequenced a decade later, that the process could take under a day, for less than \$1,000. Of course, understanding all the information in a human genome will take much longer.

Each subsequent edition opened with a scenario of two students taking genetic tests, which grew less hypothetical and more real over time, even reaching the direct-to-consumer level. This new edition reflects the translation of gene and genome testing and manipulation from the research lab to the clinic.

The eleventh edition opens with "Eve's Genome" and ends with "Do You Want Your Genome Sequenced?" In between, the text touches on what exome and genome sequencing have revealed about single-gene diseases so rare that they affect only a single family, to clues to such common and complex conditions as intellectual disability and autism. Exome and genome sequencing are also important in such varied areas as understanding our origins, solving crimes, and tracking epidemics. In short, DNA sequencing will affect most of us.

As the cost of genome 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 about the common as well as 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, and health care professionals must be fluent in the field's language and concepts. 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 to mold who we are.

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 medical news writer, blogger, professor, and genetic counselor, along with her expertise in genetics. Updates to this edition include

- Genetic tests, from preconception to old age
- Disease-in-a-dish stem cell technology
- From Mendel to molecules: family exome analysis
- Allelic diseases: one gene, more than one disease
- Admixture of archaic and modern humans
- Gene silencing and genome editing
- Cancer genomes guide treatment
- The reemergence of gene therapy
- Personal genome sequencing: promises and limitations

The transition of genetics to genomics catalyzed slight reorganization of the book. The order of topics remains, but material that had been boxed or discussed in later chapters because it was once new technology has been moved up as the "applications" become more integrated with the "concepts." The book has evolved with the science.

The Human Touch

Human genetics is about people, and their voices echo throughout these pages. They speak in the narrative as well as in many new chapter introductions, boxes, stories, and end-of-chapter questions and cases.

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 knows the little girl in the "In Their Own Words" essay in chapter 2 and on the cover with her dog, who is 1 of about 70 people in the world with giant axonal neuropathy. Perhaps there is no more heart-wrenching image of Mendelian inheritance than the chapter 4 opening photo of a daughter and father, who died from Huntington disease within weeks of each other.

Clinical 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, archaic human genome sequencing, gene therapy, familial DNA searches, exome sequencing, cell-free fetal DNA testing, and personal genome sequencing
- Ethical concerns that arise from the interface of genetic and genomic 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 material. “*The Big Picture*” encapsulates the overall theme of the chapter. The chapter opening essay and figure grab attention. Content flows logically through three to

five major sections per chapter that are peppered with high-interest boxed readings (“*In Their Own Words*,” “*Clinical Connections*,” “*Bioethics: Choices for the Future*,” “*A Glimpse of History*,” and “*Technology Timelines*”). End-of-chapter pedagogy progresses from straight recall to applied and creative questions and challenges.

Dynamic Art

Outstanding photographs and dimensional illustrations, vibrantly colored, are featured throughout *Human Genetics: Concepts and Applications*. 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.