

Preface

Introduction

Rarely does very much change between the ever-shrinking window of time that separates consecutive editions of a textbook. That certainly isn't true for the fourth edition of *Human Genetics: Concepts and Applications*. The sequencing of the human genome, new with the millennium, is something that Gregor Mendel could not have fathomed, and that Francis Crick once only imagined in the context of unravelling the genetic instructions of a simple bacterium. Yet it has been done.

Whether deciphering the sequence of our genetic blueprints comes to represent a true paradigm shift in the science of genetics remains to be seen. Some have argued that it does not change what has come before but merely continues it, albeit on an enormous, systematic scale. Some have argued that the genome project was not creative or clever, comparing it to climbing Mt. Everest simply because it is there.

Although its impact on science is unclear, human genome information will almost certainly impact health care and, therefore, the average citizen. Once not even included in medical school training, human genetics and genomics are now explaining the underpinnings of many diseases. Medical consumers are asking for information on genetic testing, and the media has so hyped genetic research that some patients are even demanding treatments and procedures that are still many years in the future. Ironically, at the same time that people are looking to gene-based tests and treatments with great hope, rejection of genetically-modified (GM) crops is growing, fueled more by politics and eco-

nomics than science. The fact that both types of technologies—gene therapy and agricultural biotechnology—use much the same methods of gene transfer and expression, yet evoke such opposite responses, indicates that not everyone is familiar with basic genetic principles. A survey in the United Kingdom, for example, found that people are avoiding “GM foods” because of a fear of consuming DNA!

This book is written for the citizens of the future who will evaluate new medical options and brave new foods, and decide for themselves whether a new technology is valuable, potentially dangerous, unethical, or useless. Being informed in the coming age of genomics requires understanding what genes are, and how they function and interact with each other and environmental stimuli. While Mendel's laws, the DNA double helix, protein synthesis and population dynamics will always form the foundation of the field, the study of human genetics must now embrace much more. Completion of the human genome project has catapulted human genetics to a new level, one that has evolved from the single-gene-at-a-time approach of the last decades of the last century to a more multifactorial view.

Human Genetics: Concepts and Applications, Fourth Edition weaves the thread of genomics throughout the clear and exciting discussion of gene structure and function and biotechnology. Changes to this edition include increased emphasis on clarity and evenness of level, with several new pedagogical features added to ease learning. Updating is everywhere. The book's unique reliance on recounting the experiences of real people remains, bolstered by inclusion of more of my experiences as a genetic counselor.

What's New and Exciting About this Edition

Easier Learning

Particular care has been taken in this revision to provide a clear framework of basic principles. After reading a chapter, students should be able to identify the main concepts and place them into the larger context of genetics. New transitions have been added, chapters are more closely linked, figures are more consistent, and a new host of pedagogical tools have been added to ease learning. These aids include:

- Introductory outlines with summaries of major topics
- Numbered main headings in text, chapter introductions, and chapter summaries
- Brief, straightforward narrative introductions that get to the point fast
- Many new summary tables that encapsulate concepts
- New figures with step-by-step descriptions
- New questions and problems
- Summary of key concepts at the end of each major section
- Websites and OMIM references with each chapter

A Sense of Reality

Human Genetics: Concepts and Applications, Fourth Edition “puts a name on” and personalizes the material. It is real, relevant, and connected to everyday life.

“**In Their Own Words**” essays are written by individuals who have, or are close to people who have, inherited disease, providing a different view from the researchers who contribute the essays in most textbooks. The essays introduce:

- Don Miller, the first recipient of gene therapy for hemophilia (chapter 1)
- Stefan Schwartz, who has Klinefelter Syndrome (Chapter 11)
- Kathy Naylor, whose little girl died of cri-du-chat syndrome (Chapter 11)

- Blaine Detheridge-Newsom, a teen who has spina bifida (Chapter 14)
- Sandra Thomas, founder of the American Hemochromatosis Society (Chapter 18)

Bioethics: Choices for the Future essays, new to this edition, delve deeper into scientific puzzles and societal responses that may influence our own future.

- Considering Cloning (Chapter 3)
- Beryllium Sensitivity Screening (Chapter 14)
- Pig Parts (Chapter 15)
- The Ethics of a Recombinant Drug: EPO (Chapter 17)
- Gene Therapy Fatalities (Chapter 18)
- The Butterfly that Roared (Chapter 19)
- Technology Too Soon? The Case of ICSI (Chapter 20)

Coverage of **Genetic Counseling**, a special combination of scientific, medical, and psychological skills to educate and comfort people facing the possibility of inherited illness, appears throughout this edition.

- *BRCA1—A Genetic Counseling Nightmare* and Table 16.5 *Reasons Why Genetic Counseling for Familial Breast Cancer is Complex* (other books get it wrong!)
- Down Syndrome recurrence risks based on age and family history (Chapter 11)
- *Scenes from a Sickle Cell Disease Clinic* (Chapter 18)
- Discussion of how genetic counseling relates to other health care professions
- *Genetic Counseling Quandaries and Challenges* based on actual cases (Chapter 18)
- New Chapters 1 and 21 cover genetic counseling as part of 21st century genetic medicine

Not Just Up-to-Date—Ahead

Previous editions of *Human Genetics: Concepts and Applications* covered genetic markers, antisense technology, gene target-

ing, and human embryonic stem cells before they became headlines. This new edition continues that up-to-the-minute coverage with updates of current technologies and introduction of new ones, such as vegetable vaccines (Chapter 15), semen pharming (Chapter 17), chimera-plasty (Chapter 18), rhizosecretion and bioremediation (Chapter 19), and pharmacogenomics and DNA microarrays (Chapter 21). Yet at the same time, the book traces discoveries and developments that led to today's and tomorrow's technologies. **Technology Timelines** chronicle the gestation and birth of transplantation (Chapter 15), patenting life (Chapter 17), assisted reproductive technologies (Chapter 20) and the human genome project (Chapter 21).

Significant Changes in Content

Major goals of this revision are to engage the student with relevant coverage and to update the instructor with the latest developments in the field, but the main thrust of this revision is to ensure that the **fundamental concepts** of genetics are clearly presented to students. Significant content changes that address this goal include:

- A new section on calculating risk (Chapter 1)
- Added coverage of the cell membrane (Chapter 2)
- More material on the cell cycle, apoptosis and stem cells (Chapter 2)
- A new section on multiple births (Chapter 3)
- A clear explanation of the meaning of dominance and recessiveness (Chapter 4)
- Real examples of “linkage mapping” (Chapter 5)
- Clearer coverage of genomic imprinting (Chapter 6)
- More structured discussions of polygenic and multifactorial traits (Chapter 7)
- Expanded coverage of DNA repair disorders (Chapter 8)

- Simplified discussion of gene expression (Chapter 9)
- New sections on globin disorders and prion disorders (Chapter 10)
- Story of the development of prenatal testing (Chapter 11)
- Clear step-by-step discussion of Hardy-Weinberg mathematics (Chapter 12)
- Augmented discussion of balanced polymorphisms (Chapter 13)
- Balanced discussion of mitochondrial Eve and the multiregional hypothesis (Chapter 14)
- Coverage of innate immunity and new vaccines (Chapter 15)
- New section on the epidemiology of cancer (Chapter 16)
- Expanded discussion of how to make recombinant DNA (Chapter 17)
- New coverage of genetic counseling (Chapter 18)
- Discussion of controversy over genetically modified foods (Chapter 19)
- New discussion of the ethics of reproductive technology (Chapter 20)
- New chapter on functional genomics—beyond the Human Genome Project (Chapter 21)

Supplements

As a full service publisher of quality educational products, McGraw-Hill does much more than just sell textbooks to your students. We create and publish an extensive array of print, video, and digital supplements to support instruction on your campus. Orders of new (versus used) textbooks help us to defray the cost of developing such supplements, which is substantial. Please consult your local McGraw-Hill representative to learn about the availability of the supplements that accompany *Human Genetics: Concepts and Applications*.

For the Student

Case Study Workbook in Human Genetics, Second Edition by Ricki Lewis. 0-07-232530-5

This workbook is specifically designed to support the concepts presented in *Human Genetics* through new real cases adapted from recent scientific and medical journals, with citations included. It 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. An **answer manual** is available for the instructor.

Genetics: From Genes to Genomes CD ROM

This CD covers the most challenging concepts in the course and makes them more understandable through presentation of full-color narrated animations and interactive exercises. Icons in the text indicate related topics on the CD.

For the Instructor

Instructor's Manual and Test Item File

prepared by Jack Fabian, Keene State College
In addition to chapter outlines, answers to in-text questions, and additional questions with answers that have supported previous editions, Jack Fabian has added a number of new features to this edition. These include:

- An overview section that summarizes the material in each chapter
- A list of transparencies, Web resources, and CD presentations that support each chapter
- Ideas for classroom instruction
- A list of Internet resources and activities

Moreover, multiple choice questions and answers that instructors may use for testing are provided for each chapter. The test

item file is also available in **computerized form** compatible with either Windows or Macintosh.

Transparencies

A set of transparencies showing key illustrations from the text is available for adopters. Additional images are available for download on the book's website.

Website

Get Online! Visit us at www.mhhe.com/lewisgenetics

Explore this dynamic website that provides additional resources for both student and instructor including:

- Images and tables from the text available for downloading
- Case histories and opinion articles for discussion
- Online quizzes to support study
- Resource articles and popular press coverage
- Support groups and information sites for genetic diseases
- Internet links to related Websites

Instructors will also find a link to **PageOut: The Course website Development Center** to create a course website. Its powerful features help create a customized, professionally designed Website for your human genetics course, yet it is incredibly easy to use. There is no need to know any coding. Save time and valuable resources by typing your course information into the provided templates.