Preface

Genetics is a relatively young science that has made major strides since the beginning of the twentieth century. A closer examination reveals that progress in genetics, like all science, moves forward in fits and starts. At the moment, unexpected discoveries are opening new subdisciplines, re-energizing old ones, and expanding the role of genetics as one of the foundations of biology. For example, epigenetics is providing answers to old questions and is rapidly emerging as an important field linking the environment with the genome and providing insights into the evolution of our species. New approaches in gene therapy offer hope that this field can finally live up to the hopes and expectations that it can be used to treat genetic disorders. Other findings are also rapidly moving from the laboratory to medical practice. These include discoveries about the role of stem cells in the development of cancer and the mobilization of the immune system to fight cancer. The contents of this edition, like the others that have preceded it, have been extensively rewritten and updated to reflect these discoveries, but as with past editions, the underlying rationale and aims have remained constant.

This book is written for a one-term human genetics course for students in humanities, social sciences, business, engineering, and other fields. It assumes that the students who come to this course will have little or no background in biology, chemistry, or mathematics and will have personal, professional, or intellectual reasons for wanting to learn something about human genetics. The book is also intended to serve those who will become *consumers* of genetic-based health care services and those who may become *providers* of health care services.

Because genetic knowledge and technology is rapidly being transferred to many areas of our society, it is imperative that the general public, elected officials, and policy makers outside the scientific community have a working knowledge of genetics to help shape how genetics and its associated technologies will be used in our society. To communicate this knowledge, *Human Heredity* is written to transmit the principles of genetics in a straightforward and accessible way, without unnecessary jargon, detail, or the use of anecdotal stories in place of research-based content. Some descriptive chemistry is used after an appropriate introduction and definition of terms. In the same vein, no advanced math skills are required to calculate elementary probabilities or to calculate genotype and allele frequencies.

Goals of the Text

From its beginnings, this book has held to a few simple goals for teaching students about human genetics. This edition continues that tradition with the following goals:

1. Present the concepts underlying human genetics in clear, concise, jargon-free language to give students a working knowledge of genetics. Each chapter presents a limited number of clearly stated concepts and examples to assist learning a complex topic.
2. Begin each chapter with a relevant example in the form of a case study that nonmajors can understand and which provides examples that students can apply to themselves, their families, and their work environments.

3. Examine the social, cultural, and ethical implications associated with the use of genetic technology.
4. Explain the origin, nature, and amount of genetic diversity present in the human population and how that diversity has been shaped by natural selection.

To achieve these goals, emphasis has been placed on clear writing and the use of accompanying photographs and artwork that teach rather than merely illustrate the ideas under discussion.

Organization

Although it is without formal divisions, the text is organized into four sections: Chapters 1 through 7 cover cell division, transmission of traits from generation to generation, and development. Chapters 8 through 12 emphasize molecular genetics, mutation, and cancer. Chapters 13 through 16 include recombinant DNA technology, genomics, and biotechnology. These chapters cover the basic methods of genetic technology and how they are used in agriculture, medicine, and the biotechnology industry. In addition, these chapters cover genetic screening, genetic testing, and genetic counseling. Chapters 17 through 19 cover specialized topics: the immune system, the genetics of behavior, and population genetics and human evolution.

Instructors teaching genetics to nonmajors come from many different backgrounds and use a wide range of instructional formats, including active learning, peer-to-peer instruction, and adaptive learning. To facilitate this array of approaches, the book is organized to allow both students and instructors to use the material no matter what order of topics is selected. After the first section, the chapters can be used in any order. Within each chapter, outlines and end-of-chapter activities let the instructor and students easily identify and explore central ideas.

What's New in the Eleventh Edition

Each chapter has been updated to reflect the latest advances in genetics. Listed below are some of the most significant revisions in this edition.

Chapter 1: A Perspective on Human Genetics

- New chapter opening photo
- New opening case study on translational medicine
- Revised and updated text throughout

Chapter 2: Cells and Cell Division

- Text edited throughout for clarity
- Revised Figure 2.6 The Nucleus
- Revised Figure 2.7 The Cell Cycle

Chapter 3: Transmission of Genes from Generation to Generation

- Revised Section 3-3 Mendel's Experimental Design
- Revised Section 3-4 Crossing Pea Plants

Chapter 4: Pedigree Analysis in Human Genetics

- New chapter opening photo
- Edited and revised Section 4-2 Pedigree Analysis
- Revised Section 4-3 Autosomal Recessive Traits
- Revised Section 4-4 Autosomal Dominant Traits
- Replaced Figure 4.20 OMIM Home Page
- Redrawn Figure 4.23 Common Autosomal Trait

Chapter 5: The Inheritance of Complex Traits

- Revised Section 5-2 Polygenic Traits Are Controlled by Two or More Genes
- Revised Section 5-3 Complex Traits and Variation in Phenotype
- Revised Section 5-9 Skin Color and IQ Are Complex Traits

- Replaced and updated Figure 5.12 Obesity in the United States
- New Figure 5.17 Skin Color and Latitude

Chapter 6: Cytogenetics: Karyotypes and Chromosome Aberrations
- Revised Section 6-2 The Human Chromosome Set
- Section 6-4 Analyzing Karyotypes, new subsection on noninvasive prenatal diagnosis
- Revised Section 6-7 Sex Chromosome Aneuploidy
- New Figure 6.1 Human Chromosome
- New Figure 6.2 Telomeres
- New Figure 6.12 Free Fetal DNA

Chapter 7: Development and Sex Determination
- New chapter opening photo
- Revised Section 7-2 The Human Reproductive System
- Revised Section 7-3 Human Development
- Revised Figure 7.10 Sex Determination
- Revised Figure 7.11 The Segregation of Sex Chromosomes

Chapter 8: The Structure, Replication, and Chromosomal Organization of DNA
- Revised Section 8-3 The Chemistry of DNA
- Revised Section 8-6 DNA Replication

Chapter 9: Gene Expression and Gene Regulation
- Revised Section 9-4 Tracing the Flow of Genetic Information
- Revised Section 9-6 Translation Requires the Interaction of Several Components
- Revised Section 9-9 Several Mechanisms Regulate the Expression of Genes
- New Figure 9.5 mRNA Processing
- New Figure 9.8 Transfer RNA Molecule
- New Figure 9.13 Prion Protein Folding
- Revised Table 9.2 Amino Acids Commonly Found in Proteins

Chapter 10: From Proteins to Phenotypes
- Revised Section 10-4 Phenylketonuria: A Mutation That Affects an Enzyme
- Revised Section 10-8 Pharmacogenetics and Pharmacogenomics

Chapter 11: Genome Alterations: Mutation and Epigenetics
- Reorganized and revised entire chapter
- Revised and expanded Section 11-3 Detecting Mutations and Measuring Mutation Rates
- New Section 11-4 Mutations Can Be Spontaneous or Induced
- Revised Section 11-5 Mutations at the Molecular Level
- Revised Section 11-6 Mutations Can Be Repaired
- Revised Section 11-8 Epigenetic Changes
- New Figure 11.4 Errors in DNA Replication
- New Figure 11.5 Base Pairing in Tautomeric Shifts
- New Figure 11.13 Proofreading in DNA Polymerase
- Revised Figure 11.14 Base-Pair Substitutions
- New Figure 11.15 A DNA Repair System
- New Figure 11.19 Epigenetic Changes to DNA
- New Figure 11.24 The Hypothalamus

Chapter 12: Genes and Cancer
- Revised Section 12-5 Cancer-Causing Mutations
- Revised Section 12-7 Mutant Cancer Alleles
- Revised Section 12-10 Genomics, Epigenetics, and Cancer
- Revised Figure 12.5 The Eukaryotic Cell Cycle
- New Figure 12.6 Normal and Mutant Tumor-Suppressor Genes
- Revised Figure 12.16 Gene Fusion in 9;22 Translocation
- New Figure 12.19 Cigarette Consumption and Lung Cancer
- Updated Table 12.1 Estimated New Cancer Cases
- Updated Table 12.3 Colorectal Cancer in the United States
- New Table 12.7 Cancer-Related Genes Inactivated by Hypermethylation

Chapter 13: An Introduction to Genetic Technology

- Revised Section 13-5 Finding a Specific Gene in a Library
- New Figure 13.12 Extinct Ground Sloth

Chapter 14: Biotechnology and Society

- Revised Section 14-6 DNA Profiles as Tools for Identification
- Revised Table 14.1 Some Products Made by Recombinant DNA Technology

Chapter 15: Genomes and Genomics

- Revised Section 15-6 What Have We Learned So Far About the Human Genome?
- New Section 15-8 The Human Microbiome Is Our Other Genome
- Revised Section 15-9 Proteomics Is an Extension of Genomics
- Revised Figure 15.5 History and Timeline for Genome Projects
- New Figure 15.11 Single Nucleotide Polymorphisms
- New Figure 15.14 Body Sites Sampled for Human Microbiome Project

Chapter 16: Reproductive Technology, Genetic Testing, and Gene Therapy

- Revised Section 16-3 Assisted Reproductive Technologies
- Revised Section 16-5 Genetic Testing and Screening
- Section 16-6 Therapy for Genetic Disorders, new subsection on exon skipping therapy
- New Figure 16.7 Stages in the IVF Procedure
- New Figure 16.8 Injection of Single Sperm into Egg
- Revised Figure 16.16 Gene Therapy
- New Figure 16.17 Exon Skipping
- Revised and updated Figure 16.18 Gene Therapy Clinical Trials 2014
- New Table 16.3 History of Gene Therapy

Chapter 17: Genes and the Immune System

- Revised Section 17-7 Organ Transplants Must Be Immunologically Matched
- Revised Section 17-8 Disorders of the Immune System
- New Figure 17.16 Herrick Twins
- New Figure 17.18 Jim Finn
- New Figure 17.19 Allergic Response
- Revised Table 17.5 Some Autoimmune Diseases

Chapter 18: Genetics of Behavior

- Reorganized and revised
- Revised Section 18-2 Models, Methods, and Phenotypes in Studying Behavior
- New Section 18-3 The Nervous System Is the Focus of Behavior Genetics
- Revised Section 18-4 Single-Gene Mutations Cause Behavioral Disorders
- Revised Section 18-5 Huntington Disease Is a Model for Neurogenerative Disorders
- Revised Section 18-6 Animal Models: The Search for Behavior Genes
- Revised Section 18-7 The Genetics of Complex Behavioral Disorders
- Section 18-8 Genetics and Social Behavior, new subsection on addictive behavior
- New Figure 18.2 The Human Nervous System
- New Figure 18.3a Synapses and Synaptic Transmission
- New Figure 18.11 Genetic Relationship Between Psychiatric Disorders
- New Figure 18.14 Heritability of Addictive Behaviors
- Revised Figure 18.15 Metabolism of Alcohol
- Revised Table 18.2 Selected Neurotransmitters and Some Processes They Affect
- New Table 18.3 Selected Recreational Drugs and the Neurotransmitters They Mimic
- New Table 18.4 Some Behavioral Disorders Associated with Synaptic Defects
- New Table 18.5 Important Risk Factor Genes for Alzheimer Disease
- New Table 18.6 Genes Involved in Nicotine Addiction

Chapter 19: Population Genetics and Human Evolution

- Revised Section 19-7 The Evolutionary History and Spread of Our Species
- Revised Section 19-8 Genomics and Human Evolution
- New Figure 19.11 Hominin Evolution
- New Figure 19.14 Cave in Denisova, Siberia

Features of the Book

Numbered Chapter Outlines

The beginning of each chapter contains an outline of the primary headings, providing an overview of the main concepts, secondary ideas, and examples. To help students grasp the central points, many of the headings are written as narratives or summaries of the ideas that follow. These outlines also serve as convenient starting points for students to review the material in each chapter. To make the outlines more useful, they have been numbered sequentially and used to organize the summary, the questions, and the problems at the end of each chapter. In this way, students can relate examples and questions to specific topics in the chapter more easily and clearly.

First Section Case Studies

The first section of each chapter contains a case study that is directly related to the main ideas of the chapter, often drawn from real life. Topics include the use of DNA fingerprinting in court cases, the cloning of milk cows, the use of exome sequencing to diagnose a genetic disorder, and the development of *in vitro* fertilization (IVF) and the birth of Louise Brown—the first IVF baby. These case studies are designed to promote student interest in the topics covered in the chapter and to demonstrate that laboratory research often has a direct impact on everyday life. These case studies are linked to another case presented in the ***Genetics in Practice*** section at the end of the chapter.

The Genetic Revolution

The Genetic Revolution is a feature that emphasizes the past, present, and future impact of genetic technology on our daily lives, from genetic testing at birth to the future of cancer therapy. Accompanying questions are designed to be used for classroom discussion, research topics, and student presentations.

Exploring Genetics

Exploring Genetics feature boxes present ideas and applications that are related to and extend the central concepts in a chapter. Some of these examine controversies that arise as genetic knowledge is transferred into technology and services. Accompanying questions are designed to be used for classroom discussion, research topics, and student presentations.

Marginal Glossary

A glossary in the page margins gives students immediate access to definitions of terms as they are introduced in the text. This format also allows definitions to be identified when students are studying or preparing for examinations. The definitions have been gathered into an alphabetical glossary at the back of the book. Because an understanding of the concepts of genetics depends on understanding the relevant terms, more than 350 terms are included in the glossary.

End-of-Chapter Features

Genetics in Practice: Relevant Case Studies

A case study is included at the end of each chapter, illustrating the impact of genetics in our society. These contain scenarios and examples of genetic issues related to health, reproduction, personal decision making, public health, and ethics. Many of the case studies and the accompanying questions can be used for classroom and other activities.

Summary

Each chapter ends with a summary that restates the major ideas covered in the chapter. The beginning outline and ending summary for each chapter use the same content and

order to emphasize major concepts and their applications. Each point of the summary outline is followed by a brief restatement of the chapter material covered under the same heading. This helps students recall the concepts, topics, and examples presented in the chapter. It is hoped that this organization will minimize the chance that they will attempt to learn by rote memorization.

Questions and Problems

The summary's focus on the chapter's main points is continued in the ***Questions and Problems*** at the end of each chapter. The questions and problems are presented under the headings from the chapter outline. This allows students to relate the problems and questions to specific topics presented in the chapter, focus on concepts they find difficult, and work the problems that illustrate those topics. The questions and problems are designed to test students' knowledge of the facts and their ability to reason from the facts to conclusions. To this end, they use an objective question format and a problem-solving format. Because some quantitative skills are necessary in human genetics, almost all chapters include some problems that require students to organize the concepts in the chapter and use those concepts in reasoning to a conclusion. Answers to selected problems are provided in an appendix. Answers to all questions and problems are available in the Instructor's Manual on the password-protected Instructor Companion Site.

Pedagogical Features

Genomic Databases as Resources

To make students aware of the array of genomic resources available to them, genetic disorders mentioned in the book are referenced by their indexing numbers from the comprehensive catalog available online as *Online Mendelian Inheritance in Man (OMIM)*. OMIM (updated daily) contains text, pictures, and videos, along with literature references. Through Entrez, OMIM is cross-linked to databases containing DNA sequences, protein sequences, chromosome maps, and other resources. Students and an informed public need to be aware of the existence and relevance of such databases, and to be up to date, textbooks must incorporate these resources.

Students can use OMIM to obtain detailed information about a genetic disorder, its mode of inheritance, its phenotype and clinical symptoms, mapping information, biochemical properties, the molecular nature of the disorder, and a bibliography of relevant papers. In the classroom, OMIM and its links are valuable resources for student projects and presentations.

Online Learning and Teaching Solutions

The online learning and teaching solutions that accompany this edition are designed to aid student learning as well as to assist the instructor in preparing lectures and examinations and in keeping abreast of the latest developments in the field. Instructor materials are available to qualified adopters. Please consult your local Cengage learning consultant for details. You may also visit the Brooks/Cole biology site at ***www.cengage.com/biology*** to see samples of these materials, request a desk copy, locate your learning consultant, or purchase a copy online.

MindTap for Biology

MindTap is a fully online, highly customizable learning experience built upon Cengage Learning content. MindTap combines student learning tools—readings, multimedia, activities, and assessments—into a singular Learning Path that guides students through their course. Instructors personalize the experience by customizing authoritative Cengage Learning content and learning tools, including the ability to add their own content in the Learning Path via apps that integrate into the MindTap framework seamlessly with Learning Management Systems. New to this edition! Chapter opening videos, assignable homework, and a digital Study Guide.

Cengage Learning Testing Powered by Cognero

Cengage Learning Testing Powered by Cognero is a flexible, online system that allows you to:

- author, edit, and manage test bank content from multiple Cengage Learning solutions
- create multiple test versions in an instant
- deliver tests from your LMS, your classroom, or wherever you want

Start right away!

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- No special installs or downloads needed
- Create tests from school, home, the coffee shop—anywhere with Internet access

What will you find?

- Simplicity at every step. A desktop-inspired interface features drop-down menus and familiar, intuitive tools that take you through content creation and management with ease.
- Full-featured test generator. Create ideal assessments with your choice of 15 question types (including true/false, multiple choice, opinion, and essay). Multi-language support, an equation editor, and unlimited metadata help ensure your tests are complete and compliant.
- Cross-compatible capability. Import and export content into other systems.

Instructor Companion Site

Everything you need for your course in one place! This collection of book-specific lecture and class tools is available online via ***www.cengage.com/login***. Access and download PowerPoint presentations, images, instructor's manual, videos, and more.

Cooperative Learning: Making Connections in General Biology, Second Edition

A collection of separate, ready-to-use, short cooperative activities that have broad application for first-year biology courses. They fit perfectly with any style of instruction, whether in large lecture halls or flipped classrooms. The activities are designed to address a range of learning objectives, such as reinforcing basic concepts, making connections between various chapters and topics, data analysis and graphing, developing problem solving skills, and mastering terminology. Since each activity is designed to stand alone, this collection can be used in a variety of courses and with any text. Authored by Mimi Bres and Arnold Weisshaar.

A Problem-Based Guide to Basic Genetics

Provides students with a thorough and systematic approach to solving transmission genetics problems, along with numerous solved problems and practice problems. Written and illustrated by Donald Cronkite of Hope College.

Virtual Biology Laboratories: Genetics and Genetics 2 (Pedigree Analysis) Modules

These "virtual" online experiments expose students to the tools used in modern biology, support and illustrate lecture material, and allow students to "do" science by performing experiments, acquiring data, and using the data to explain biological phenomena.

Gene Discovery Lab

This is a CD-ROM lab manual that provides a virtual laboratory experience for the student in doing experiments in molecular biology. It includes experiments that use nine of the most common molecular techniques in biology, an overview of scientific method and experimental techniques, and Web links to provide access to data and other resources.

Acknowledgments

Over the course of eleven editions, many reviewers have given their time to improve the pedagogy, presentation of concepts, and ways of inspiring students. From edition to edition, a number of reviewers went to extraordinary lengths to keep my ideas and writing on the straight and narrow path and to make suggestions that have greatly improved the book. George Hudock of Indiana University, H. Eldon Sutton of the University of Texas, and Werner Heim of Colorado College generously gave me access to their collective wisdom, and helped me learn and relearn many of the nuances involved in writing about genetics. I am most grateful for their efforts.

In the last edition, Daniel Friderici of Michigan State University examined the text, figures, and problems from a student's point of view, and helped me present each chapter's important concepts in a straightforward and engaging way. In addition, I greatly appreciate his many suggestions on how to improve the end-of-chapter questions, problems, and how to frame the answers so that the questions become effective teaching tools. I am also very grateful to Patricia Matthews of Grand Valley State University who spent many hours scrutinizing the text, helping me clarify and streamline my writing, pointing out inconsistencies in word use, and improving the flow of ideas throughout the text.

To all the reviewers who helped in the preparation of this and previous editions, I offer my thanks and gratitude for their efforts.

Ted W. Fleming, *Bradley University*
Daniel Friderici, *Michigan State University*
Pamela L. Hanratty, *Indiana University*
Bradley Isler, *Ferris State University*
Mary King Kananen, *Pennsylvania State University, Altoona*
Brenda Knotts, *Eastern Illinois University*
Clint Magill, *Texas A&M University*
Robert L. Snyder, *State University of New York, Potsdam*
Jan Trybula, *State University of New York, Potsdam*
Jo Ann Wilson, *Florida Gulf Coast University*
Elizabeth T. Wood, *University of Arizona*
Denise Woodward, *Pennsylvania State University*

At Cengage Learning, it was once again a pleasure to work with Peggy Williams, Senior Product Manager. Her vision about how to increase the pedagogical value of texts and her extensive knowledge of the market have strengthened and enhanced the book. Hal Humphrey was the content project manager who pulled together all the resources and people needed to put this edition together. The content developer, Suzannah Alexander, oversaw the preparation of this edition. Her attention to detail and gentle nudging kept the project on schedule.

Lauren Oliveira , Casey Lozier, and Kellie Petruzzelli coordinated the digital package for the book. Photo research was handled by Priya Subbrayal at PreMedia Global, whose hard work provided many excellent choices for photos.

Lynn Lustberg at MPS Limited eased the book through all the twists and turns involved in production.

Contacting the Author

I welcome questions and comments from faculty and students about the book or about questions and issues related to human genetics. Please contact me at: cummings.chicago@gmail.com.

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