Preface

WHEN THE FIRST EDITION of this book was published, recombinant DNA technology was just starting to have an impact on human genetics, and the beginnings of the Human Genome Project were several years in the future. As this edition goes to press, websites advertise the availability of genetic identification cards, television crime shows routinely solve cases using DNA forensic evidence, and genomic scans use microarrays carrying the human genome to screen for disorders that will not develop for decades. The supermarket and the pharmacy are filled with the products of biotechnology, and terms such as genetically modified organisms, DNA profiles, and prenatal diagnosis are parts of everyday speech.

Human Heredity has developed in parallel with advances in human genetics. In the second edition, recombinant DNA technology moved from being a section to a freestanding chapter. In the third edition, the impact of recombinant DNA technology gradually spread into other chapters. By the fourth edition, a chapter on biotechnology became necessary, and in the fifth edition, half a chapter was devoted to the Human Genome Project. The sixth edition was published shortly after the draft sequence of the human genome was reported, giving us a glimpse into the size, organization, and evolutionary history of our genome. The complete sequence of the coding portion of the genome was reported in 2003. What is now apparent, even in the short time following completion of the human genome sequence, is that this information is transforming the diagnosis and treatment of disease and accelerating the pace of research by an order of magnitude. Genome sequence information, coupled with technology such as DNA microarrays, is causing a fundamental shift in the way human genetics research is done. These developments allow researchers to use automated processes to gather data on a scale far beyond what was imagined only a few years ago, rapidly advancing our knowledge of genetics, creating new applications and products. Reflecting these changes, a chapter devoted to genomics has been added to this edition.

I wrote this book for one-term non-major human genetics courses to help undergraduates in the humanities, social sciences, business, engineering, and other fields understand these fast-moving developments. It is written for students with little or no background in biology, chemistry, or mathematics, but who want to learn something about human genetics. 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.

Knowledge from genetics is rapidly being transferred to many other fields. The spread of technology and the issues it raises make it clear that difficult but informed decisions are required at many levels, from the personal to the political. The public, elected officials, and policy makers outside the scientific community need a working knowledge of genetics to help shape the course of research and its applications in our society. *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 content.

GOALS OF THE TEXT

Being in the lab doing research in human genetics has offered me opportunities to train and nurture the development of graduate students and to present papers at meetings, symposia, and seminars where I meet and interact with other researchers and students to exchange ideas and help shape the course of genetics research and its applications. In addition, working with clini-

cian colleagues and training medical students in my lab has given me valuable insight into the changes genetics, genomics, and technology have brought to medicine. I also work with undergraduates on a daily basis in my laboratory and through teaching courses in a biology curriculum to both majors and non-majors.

Using my research background in human genetics, my experience in training clinicians, and my classroom skills developed in teaching genetics to undergraduate and graduate students, I have written and continuously refined *Human Heredity,* combining the latest advances in research with the newest methods of teaching. In all editions, however, I have not worked in isolation. The book has been shaped by the contributions of others. Students in the course have suggested how to best introduce topics, identified the most effective examples and analogies in explaining concepts, and more important, been forthright in clarifying what does not work in the classroom. I have incorporated ideas refined by feedback from students and from faculty who have used the book at other institutions, and by comments and suggestions from reviewers. From the first edition, this book has been written to achieve several well-defined goals. This edition continues that tradition, incorporating the following goals:

1. Present the principles of human genetics in clear, concise language giving students a working knowledge of genetics. The premise in this approach is that a limited number of clearly presented, linked concepts is the best way to learn a complex subject such as genetics.
2. As in the classroom, begin the discussion of concepts at a level that students can understand and provide relevant examples that students can apply to themselves, their families, and their work environment.
3. Examine the social, cultural, and ethical implications associated with the use of genetic technology.
4. Communicate an understanding of the origin and amount of genetic diversity present in the human population, and how this diversity has been shaped by natural selection.

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

In general, the text consists of 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, genomics, and biotechnology. These chapters cover gene action, mutation, cloning, the applications of genetic technology, the Human Genome Project, and the social, legal, and ethical issues related to genetics, as well as genetic screening, genetic testing, and genetic counseling. Chapters 17 through 19 consider specialized topics, including the immune system, population genetics, as well as the social aspects of genetics, including behavior.

Courses in human genetics use a wide range of formats. To accommodate these, the book is organized so it will be easy to use, no matter what order of topics an instructor chooses. After the section on transmission genetics, the chapters can be used in any order. Within each chapter, the outline lets the instructor and students easily identify central ideas.

NEW FEATURES OF THE SEVENTH EDITION

New Chapter

Chapter 15, titled "The Human Genome Project and Genomics," summarizes what we have learned about the human genome and introduces students to the new and dynamic field of genomics. The chapter begins by reviewing genetic mapping using linkage analysis to construct genetic maps, and the slow progress made with this method. The next section introduces the use of positional cloning, a method using recombinant DNA techniques to clone, map, and sequence disease-causing genes with no previous knowledge of the gene product or its function.

To identify, catalog, and elucidate the function of all human genes, the chapter examines the history, scope, methods, and results of the Human Genome Project, and the scientific intrigue surrounding its down-to-the wire race for the sequence. The steps in genome analysis are summarized, and the major features of the human genome are outlined. The challenges of the future derived from the impact of genomics on research, disease diagnosis, and treatment are reviewed

in the last section of the chapter, using a keystone article written by genome scientists as a basis for the discussion.

Organizing the Book to Meet the Challenge of Genomics

The order of chapters has been revised, creating a middle section devoted to recombinant DNA techniques, biotechnology, and genomics as a central theme in human genetics. The reorganization and reordering of chapters has created a first section that outlines cell structure, mitosis, and meiosis (Chapter 2). Chapter 3 uses peas as a model system to cover Mendelian inheritance. Subsequent chapters in this section cover the basic methods of human genetics (Chapter 4), quantitative inheritance (Chapter 5), cytogenetics and karyotypes (Chapter 6), and human development (Chapter 7). While this first section precedes a discussion of genomics, the impact of recombinant DNA and genomics has been scattered throughout these chapters. In all cases, emphasis has been placed on ensuring that each chapter focuses on a small number of basic concepts.

The second section builds on the concepts of the first and moves the discussion to the molecular level, outlining the steps in the replication, storage, and expression of genetic information in the nucleotides of DNA molecules (Chapters 8 and 9), and the relationship between proteins and phenotype (Chapter 10). Chapter 11 explores how mutation alters phenotypes at the molecular and phenotypic level, leading into the final chapter of this section (Chapter 12), which discusses cancer, one of the consequences of mutation.

The core of the book contains chapters dealing with cloning, the impact of biotechnology, and the development of genomics and the Human Genome Project. Chapter 13 begins with a discussion of cloning, using organisms as the first set of examples. The chapter then considers the tools and methods used in cloning DNA and the methods available for analyzing clones, including DNA sequencing. Chapter 14 outlines the use of recombinant DNA techniques in biotechnology, using examples from agriculture, the development of model organisms for research, genetic testing, including the use of microarrays in genomic scanning, DNA profiles, and the ethical and social issues raised by the use of biotechnology. Chapter 15 begins by reviewing gene mapping in the pre–recombinant DNA era and the impact of positional cloning on mapping. The chapter moves on to explain the methods used in genomics and then discusses the results of the Human Genome Project and examines how genomics is used in investigating a genetic disorder. A section on proteomics as the next step in understanding our genome is followed by consideration of the ethical issues related to genomics and the future of genomics in research and medicine. Chapter 16 continues this theme by showing how biotechnology and genomics are used in reproductive technology, gene therapy, and genetic counseling. The ethical concerns about the use of biotechnology in assisted reproductive technology (ART) and gene therapy are outlined.

The final section of the book contains chapters on the immune system (Chapter 17) and behavior genetics (Chapter 18). Chapter 19 covers the essentials of population genetics and the use of molecular methods to study human evolution.

A Revised Art Program and Animations

The art program for this edition has been greatly revised and updated. Many figures have been replaced and new photos can be found throughout the book. The most significant change in the art program is the addition of dozens of **Active Figures,** art linked via the Human GeneticsNow™ website to animations that lead students step by step through the concepts. These animations are valuable assets for teaching and learning processes, including mitosis, meiosis, DNA replication, and gene expression.

Personalized Learning Resources and New Ways to Assess Learning

Recognizing that many students have difficulty solving genetics problems, the end-of-chapter questions and problems are now supplemented by Human GeneticsNow™, a password-protected website integrated with each chapter. All of the Active Figures from the text are located on this site, along with dozens more animations, interactive media, and tutorials. On Human GeneticsNow™, students can take diagnostic pre-tests that guide them to text, art, and animations that help them learn what they haven't yet mastered. After going over this personalized course of study, students finish with post-learning quizzes to assess their grasp of this new knowl-

edge. Both the pre- and post-tests results can be mailed to instructors, who can also keep track of students' progress through their own access of the site. Access to Human GeneticsNow™ comes free with every new copy of *Human Heredity*, seventh edition.

Genetics in Practice: Relevant Case Studies

To make issues in human genetics relevant to situations that students may encounter outside the classroom, case studies are included at the end of each chapter, demonstrating the effects of "Genetics in Practice" in our society. This section contains 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 discussions, student papers and presentations, and role playing. The cases and their questions are also located on the book's companion website along with links to resources for further research and exploration.

Genetic Databases as Resources

To foster awareness of the vast array of databases dealing with genetics and to integrate electronic resources into the text, genetic disorders mentioned in the book are referenced using their assigned indexing number from the comprehensive catalog assembled by Victor McKusick and his colleagues. This catalog is available online as *Online Mendelian Inheritance in Man™ (OMIM)*. OMIM™ (updated daily) contains text, pictures, and videos along with literature references. OMIM™ is cross-linked to other 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, 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.

For further reading about genetics, students can log on to InfoTrac® College Edition, an online library of articles from nearly 5,000 periodicals. This resource can be used in conjunction with electronic databases as resources for papers, class discussions, and presentations. Access to InfoTrac® is provided free with each new copy of *Human Heredity*, seventh edition.

New Internet Activities

The World Wide Web (WWW) is an important and valuable resource in teaching human genetics, and the *Human Heredity* companion website hosts quizzes, glossary, learning objectives, and a number of activities and links that can be used to expand on concepts and topics covered in the text. The website content can also be used to introduce the social, legal, and ethical aspects of human genetics into the classroom and serve as a point of contact with support groups and testing services. All of the website features, exercises, and activities described below can be easily completed online and e-mailed to instructors, making them ideal for assignments.

A new online feature is tied to the "How would you vote?" questions that follow every chapter's opening vignette. These are targeted questions about an issue related to the story and the chapter content. On the website, each "How would you vote?" question is accompanied by background information, links to helpful sites and materials, thought questions, and a chance to cast an online vote on the topic and view the resulting tallies.

Another new online feature is the inclusion of the "Genetics in Practice" case studies on the website. The cases, found at the end of each chapter, are repeated in their entirety on the website and accompanied by helpful links to resources for further exploration. This extra information makes them ideal as starting points for research projects and presentations.

In addition to the new features, this edition of *Human Heredity* contains updated end-of-chapter Internet Activities for students. These activities use WWW resources to enhance the topics covered in the chapter and are designed to develop critical thinking skills and to generate interaction and thought rather than passive observation. As with the other features, these activities are repeated on the book's companion website, along with links to websites and resources.

PEDAGOGICAL FEATURES

In this edition, the book has been substantially rewritten, and the order of chapters has been reorganized to reflect current findings and research directions, to engage student interest, and to improve the pedagogy. The basic organization within chapters, which has been successful as a teaching resource, has been continued in this edition.

(New!) Numbered Chapter Outlines

At the beginning of each chapter, an outline of the primary chapter headings provides 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 the chapter. To make the outlines more useful, they have been numbered and used to organize both the summary and the questions and problems at the end of each chapter. In this way, students can more easily and clearly relate examples and questions to specific topics in the chapter.

Opening Case Study

Each chapter begins with a short prologue 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 creation of a DNA vaccine for SARS, and the development of *in vitro* fertilization (IVF) and the birth of Louise Brown, the first IVF baby. These vignettes 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. In this edition, many of the opening stories are brand new or rewritten, and all are tied to a new feature, "How would you vote?"

(New!) How Would You Vote?

To stimulate thought and discussion, each chapter has a section, **How Would You Vote?** which presents an issue directly related to the opening story. It asks students to think about the topic and then visit the book's website where they can explore related links and cast a vote pro or con on the question posed. At the end of the chapter, the question is posed again, against the information presented in the chapter and after students have had a chance to learn more about the concepts related to the issue. These questions are intended to encourage students to think seriously about the genetic issues and concerns, provoking individual reflection and group discussion, which can be applied in a variety of ways both in and out of the classroom.

(New!) Keep in Mind Points

To keep students focused on the basic concepts in the chapter, a **Keep in Mind** box in the margin of the chapter opener contains a bulleted list of the main topics and key concepts presented in the chapter. Each item on the list is repeated in a highlighted text box at the conclusion of the section related to the concept, reinforcing the importance of the concept and providing students an aid in focusing their studies on fundamental points.

(New!) Active Figures

The most significant pedagogical addition to this edition is the use of **Active Figures,** linking art in the text to animations of important concepts, processes, and technologies discussed throughout the book. These animations convey an immediate appreciation of how a process works in a way that cannot be effectively shown in a static series of illustrations. These Active Figure animations can be found on the password-protected Human GeneticsNow™ site.

Genetic Journeys

Genetic Journeys feature boxes present ideas and applications that are related to and extend the central concepts in a chapter. The interesting but tangential examples presented provide context and connection of real examples to the ideas in the chapter.

Genetics in Society

Genetics in Society feature boxes provide a wider context to the ideas presented in the text. These essays elaborate and examine controversies that arise as genetic knowledge is transferred into technology and services.

Spotlight On . . .

Located in margins throughout the book, **Spotlight On** sidebars highlight applications of concepts, present the latest findings, and point out controversial ideas without interrupting the flow of the text.

Margin 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

The end-of-chapter features have been revised and updated for this edition. The questions are organized to reflect the order of topics in the chapter. Questions have been added to the case studies to enhance their use in the classroom, and new Internet activities have been added.

Genetics in Practice: Case Studies

Genetics in Practice case studies present specific examples of individuals and families using various genetic services, large-scale issues such as radioactive pollution, and the impact of the Human Genome Project. Many of these can be used as the basis for classroom discussions, student presentations, and role playing. The cases and their accompanying questions are also available on the book's companion website, where they are supplemented by links to other relevant sites. Students can e-mail instructors their responses to the case study questions through the website.

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 and, it is hoped, minimizes the chance 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, to focus on concepts they find difficult, and to work the problems that illustrate these 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 these 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 with Test Items.

Internet Activities

Internet Activities at the end of each chapter use websites to engage the student in activities related to the concepts discussed in the text. Internet resources are now an essential part of teach-

ing genetics, and this section introduces students to the many databases, instructional sites, and support groups available to them. The activities are repeated and expanded on the book's companion website.

ANCILLARY MATERIALS

Instructor's Resources

The ancillary materials that accompany this edition are designed to assist the instructor in preparing lectures and examinations and to help keep instructors abreast of the latest developments in the field. Instructor materials are available to qualified adopters. Please consult your local Thomson Brooks/Cole sales representative for details. You may also visit the Brooks/Cole biology site at **http://biology.brookscole.com** to see samples of these materials, request a desk copy, locate your sales representative, or purchase a copy online.

Multimedia Manager
This easy-to-use, dual-platform digital library and presentation tool provides *all* of the art, photos, and tables from the text in PowerPoint® and JPG formats, along with a pre-created PowerPoint® lecture outline for each chapter, which you can modify and adapt to your own needs. A unique feature allows you to manipulate and resize figures and remove labels to customize your presentations.

Instructor's Manual with Test Items
An expanded and updated instructor's manual is available to help instructors in preparing class materials. This manual, prepared by Carl Frankel of Pennsylvania State University, Hazleton campus, contains chapter outlines, chapter summaries, teaching/learning objectives, key terms, additional test questions, and discussion questions. It also contains answers to all the end-of-chapter questions and problems found in the book.

ExamView®
This computerized test bank, available on CD-ROM, helps you create and deliver customized tests both online and in print. ExamView® guides you through the process while its "what you see is what you get" capability allows you to see the test you are creating on screen exactly as it will print or display online.

Human Heredity Companion Website for Instructors
The password-protected site for instructors at **http://biology.brookscole.com/cummings7** contains all the features of the student site listed below, plus chapter summaries and outlines, answers to end-of-chapter questions, and other helpful instructor resources.

Human GeneticsNow™
Instructor access to Human GeneticsNow™ at **http://now.ilrn.com/cummings7** includes the ability to monitor students' progress through the various chapter tests and media assets.

CNN® Today Videos
These short clips compiled from high-interest news stories related to genetics are a great way to launch your lectures. There are now two volumes of CNN® Today Videos for Genetics, published in 2003 and 2005. Both are available on videocassette and also on CD-ROM (in Quicktime® format).

Transparency Acetates
A set of 100 color transparencies featuring key figures—including drawings, charts, and diagrams from the text—is available to adopters.

Student Resources

Human GeneticsNow™
Located online at **http://now.ilrn.com/cummings7,** Human GeneticsNow™ is an exciting new assessment-centered learning tool developed in concert with the text. The site offers diagnostic pre-tests, personalized learning plans using media and animations located on the site, and

confirming post-tests. PIN code access to Human GeneticsNow™ is packaged *free* with every new copy of the text.

Human Heredity Companion Website for Students
A valuable partner to this text, the companion website at **http://biology.brookscole.com/cummings7** features focused quizzing for each text chapter, glossary flashcards, learning objectives, "Internet Activities" with questions, "Genetics in Practice" cases with questions and links, "How would you vote?" exercises with voting tallies, annotated web links, and InfoTrac® keywords.

Study Guide
A student study guide has been prepared by Nancy Shontz of Grand Valley State University. The study guide includes chapter objectives and summaries, key terms, case worksheets (based on the "Genetics in Practice" case studies found in the text), discussion problems and questions, and other practice test items in multiple-choice, fill-in-the-blank, and modified true/false formats.

A Problem-Based Guide to Basic Genetics
Written and illustrated by Donald Cronkite of Hope College, this useful little manual provides students with a thorough and systematic approach to solving transmission genetics problems, along with numerous solved problems and practice problems.

Virtual Biology Laboratories: Genetics and Genetics 2 (Pedigree Analysis) Modules Available Fall Term 2005. These "virtual" online experiments expose students to the tools used in modern biology, support and illustrate lecture material, and allow students to actually "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 the scientific method and experimental techniques, and Web links to provide access to data and other resources.

Contacting the Author

I welcome questions and comments from faculty and students about the book or about human genetics. Please contact me at: cummings@iit.edu

Acknowledgments

When Jerry Westby of West Publishing approached me almost exactly 10 years ago to ask if I would be interested in writing a text for my undergraduate nonmajors human genetics course, I was somewhat reluctant to consider a project of that dimension. In the end, Jerry's arguments were persuasive, and over the last decade, this book has grown to be a labor of love. As human genetics becomes more entwined with social and legal issues, it is essential that nonspecialists become familiar with the concepts of genetics. I am indebted to him for his insight, his creative contributions, and his commitment to bridging the gap between scientist and nonscientist.

Over the years, many reviewers, including those who helped with this edition, have given their time to improve the pedagogy, presentation of concepts, and nuances of language. Three past reviewers have gone to extraordinary lengths to help me learn and in some cases, re-learn details of genetics and have generously given me access to their collective wisdom: George Hudock of Indiana University, H. Eldon Sutton of the University of Texas, and Werner Heim of Colorado College. I am grateful for their efforts to help make this book an effective teaching tool.

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In past editions, Michelle Murphy Whaley of the University of Notre Dame and Peter Follette took on the daunting task of revising and adding to the end-of-chapter questions and problems as well as writing questions for the Genetics in Practice case studies. For this edition, Peter once again took on the task of writing insightful questions that challenge students to think and apply their knowledge.

At Brooks/Cole, the book has had creative input from many talented individuals. I am especially indebted to my editor Nedah Rose and to Mary Arbogast, senior developmental editor, who developed a revision plan that integrates text with web-based resources including animated figures, web-linked learning assessments, and a multimedia manager for instructors. Ann Caven, the marketing manager, provided many insights during the planning stages, and her suggestions have helped make this a better book.

As the project got under way, Sarah Lowe assembled and analyzed the reviews and helped establish priorities for the revision. I was especially fortunate to have Kari Hopperstead oversee this revision. Her organizational skills kept me moving and provided me with vast amounts of analyses, information, and resources, all of which allowed me to focus on revising, updating, and writing. She was also a sounding board for my ideas, and contributed many important ideas of her own that improved the order of chapters, pedagogy, and student-oriented learning features. I am thankful for her creative input, ideas, and suggestions, which are reflected in the many new features added to this edition. Travis Metz coordinated all the web-based features of the book and helped integrate them with the text. The new artwork in this edition was done by Raychel Ciemma, Lisa Starr, and Gary Head, along with contributions from other sources. Gopa designed the layout, Hiroko Chastain designed the cover, and Sarah Harkrader oversaw the process of obtaining permissions to use figures and photos from other publications. It was a pleasure to once again work with Linda Sykes who did the photo research for this edition. Even as the deadlines tightened, she was willing to start anew looking for the best photos.

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Michael R. Cummings