

# Human Heredity

*principles and issues*



*5th edition*

MICHAEL CUMMINGS

Fifth Edition

# HUMAN HEREDITY

## *Principles and Issues*

Michael R. Cummings  
*University of Illinois at Chicago*



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# Preface

When the first edition of *Human Heredity* was published in 1988, molecular biology and recombinant DNA technologies were just beginning to transform the field of human genetics. At that time, biotechnology was a fledgling industry, and the scientific disciplines of genomics and bioinformatics did not yet exist. The Human Genome Project was in the early stages of planning, and its inception was two years away. In the twelve years since then, the biotechnology industry has grown to become a twenty billion dollar component of the economy, thousands of human genes have been identified, and the sequence of most of the human genome will be completed while this edition is being used in the classroom.

Keeping in mind how genetic research, technology, and knowledge are transforming research, industry, agriculture, and our everyday lives, I have written this book for a one-term introductory human genetics course for the non-major. It assumes that the student has little or no background in biology, chemistry, or mathematics, but has an interest in learning something about human genetics. Some descriptive chemistry is used after an appropriate introduction and definition of terms. In the same vein, math is used in several places, but no advanced math skills are required to calculate elementary probabilities or to calculate genotype and allele frequencies.

To emphasize the need for non-science students to acquire an understanding of basic genetic concepts rather than a collection of facts, each chapter is organized around one or two central ideas. For example, Chapter 3 deals with Mendelian principles, and uses crosses in pea plants to explain basic concepts. The use of an experimental organism (in this case, peas) to illustrate the principles that govern the transmission of traits from generation to generation makes it easy to define each concept and provide examples that are clear cut and unambiguous. With a firm grounding in these principles, students will be able to apply them to the transmission of traits in humans, where the methods are indirect and observational rather than experimental.

The speed at which genetics is advancing and transferring knowledge into many other areas including medicine, law, agriculture, and the pharmaceutical industry makes it clear that difficult, informed decisions need to be made at many levels, from the personal to the societal. The public, elected officials, and policy makers outside the biological sciences community need to have a working knowledge of genetic principles in order to support and help shape the course of research and its applications in our society. This text, based on a hierarchical approach that links a basic set of genetic concepts, has been written to transmit these principles without unnecessary jargon, detail, or the use of anecdotal stories.

This book is written by an author who works with undergraduates on a daily basis, teaching courses in a biology curriculum to both majors and non-majors. Over the last decade, students have offered suggestions about how to introduce a topic, have identified the most effective examples and analogies in explaining concepts, and more importantly, been forthright in clarifying what does not work in the classroom. As in the past, the organization and content for this edition have been developed by incorporating classroom-tested ideas refined by student feedback. With this background, this text has been written to achieve several well-defined goals, and this edition reinforces and extends these goals:

1. Present the principles of human genetics in a clear, concise manner that gives students a working knowledge of genetics. The premise behind this

approach is that the use of a limited number of clearly presented, interlinked concepts is the best way to learn a complex subject such as genetics.

2. 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.
3. Examine the social, cultural, and ethical implications associated with the use of genetic technology.
4. 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.

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 addition, the book features up-to-date coverage and flexible organization. A conscious effort has been made to pare down unnecessary terms and jargon and to present the material in a straightforward and engaging manner.

In general, the text consists of three sections: Chapters 1–7 cover cell division, transmission of traits from generation to generation and development. Chapters 8–13 emphasize molecular genetics, recombinant DNA 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. Chapters 14–19 consider specialized topics, including cancer, the immune system, population genetics, as well as the social aspects of genetics including behavior, genetic screening, and genetic counseling.

Because courses in human genetics have a wide range of formats, 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.

## FEATURES OF THE FIFTH EDITION

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### New Chapter

Chapter 1 has been rewritten to serve as an introduction to the science of genetics, and provides an overview of the structure and scope of the field, and a timeline of major developments in human genetics. The social impact of human genetics on law and social policy is emphasized, and the need for informed decisions in the future is outlined. This chapter provides a perspective for what follows in subsequent chapters.

### New Organization

The order of chapters is the same as in the last edition. The reorganization and reordering of chapters that took place in the last edition has proven successful, and has been continued here. Mendelian inheritance and quantitative inheritance are in one section, and the chapter on mutation precedes the chapters on recombinant DNA technology and its applications. In this edition, emphasis has been placed on ensuring that each chapter focuses on one or two central ideas. As a result, material has been moved, rewritten to shift emphasis, and new sections have been added as needed.

A few examples of these changes will serve to illustrate how this focus was developed. In Chapter 2, the sections on the human chromosome set and the box on making a human karyotype have been moved to Chapter 6, leaving the central ideas of cell structure, mitosis, and meiosis intact. In addition, the essay on cell membranes has been moved from Chapter 4 to Chapter 2, and the essay from Chapter 2 on aging has been moved to Chapter 4. In Chapter 7, the section on human reproduction

has been eliminated, and the chapter now begins with human development from fertilization to birth. This places the sections on sex determination and differentiation into a broader context. In Chapter 8, the section on viruses has been eliminated, and the chapter begins with a discussion of the evidence that DNA is a carrier of genetic information. Once this has been established, a discussion of DNA as a part of human chromosomes follows, along with sections on the structure and replication of DNA. The result is a chapter that deals with a few central ideas, using clear-cut examples for each idea.

## Expanded Questions and Problems

Recognizing that many students have difficulty solving genetics problems, the end-of-chapter questions and problems have been revised and expanded. The revisions and additions have been contributed by adopters of the text who have used their experience and student input to redesign, rewrite, and add to the problem sets. The questions and problems use both an objective question and problem format, and are arranged by level of difficulty. Because some quantitative skills are necessary in human genetics, almost all chapters include some problems that require the students to organize the concepts in the chapter and use these concepts in reasoning to a conclusion. Answers to selected problems are provided in Appendix B.

## New Case Histories

To make issues in human genetics relevant to situations that students may encounter outside the classroom, a new section on case histories has been added at the end of each chapter. This section contains scenarios and examples of genetic issues related to health, reproduction, personal decision making, public health, and ethics. Many of these can be used as the basis for classroom discussions, student presentations, and role playing.

## New Topics

Many new topics have been introduced, or have received greater emphasis. It is impossible to list them all, but included among them are pharmacogenetics; background radiation exposure, radiation as a source of mutation, and DNA repair mechanisms; cloning of mammals from somatic cells; mapping genes by positional cloning, preimplantation genetic testing, the use of DNA chips in genetics, biopharming, and the ethical issues surrounding the Human Genome Project; DNA repair mutations in cancer, the role of gatekeeper and caretaker genes in cancer, genomic instability and cancer, and behavior and cancer; the search for genes controlling manic depression and schizophrenia; and genetic evidence for the spread of humans across the world.

## Guest Essays

This edition features a series of essays written by distinguished scientists, describing how they became interested in science, what they have chosen to study, and how their research relates to the larger context of human genetics. These essays are not just *about* scientists, they are written *by* scientists, giving non-majors some insight into the lives, thoughts, and motives of biologists and geneticists.

## Genetic Databases

To foster awareness of the vast array of databases dealing with genetics, the genetic disorders mentioned in the book are referenced with the indexing number assigned to them in the comprehensive catalog assembled by Victor McKusick and his colleagues.

This catalog is available in book form as *Mendelian Inheritance in Man: Catalog of Human Genes and Genetic Disorders*. It is also available at several World Wide Web sites as *Online Mendelian Inheritance in Man (OMIM)*. The online version (with daily updates) contains text, pictures, and videos along with references to the literature and links to other databases, including those related to the Human Genome Project. Students and an informed public need to be aware of the existence and relevance of such databases, and OMIM is used here in part to promote such awareness.

Students wishing to learn more about a particular genetic disorder can use OMIM to obtain detailed information about the disorder, its mode of inheritance, phenotype and clinical symptoms, mapping information, biochemical properties, the molecular nature of the defect, and availability of cloned sequences for the gene in question. In the classroom, OMIM and its links to other databases are a valuable resource for student projects and presentations.

## **New Internet Activities**

The World Wide Web (WWW) is an important and valuable resource in teaching human genetics, and the exercises included in this edition can be used to expand on concepts covered in the text, and provide detailed information about specific genetic disorders. They 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.

This edition contains updated and expanded end-of-chapter Internet activities for students. These activities use WWW resources to enhance the topics covered in the chapter, and are designed to generate interaction and thought rather than passive observation. Sites for these activities can be reached through the book's home page.

## **PEDAGOGICAL FEATURES**

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The basic organization within chapters, which has been successful as a teaching resource, has been continued in this edition. Many of these features have been updated and revised to reflect current topics, engage student interest, and improve the pedagogy.

### **Opening Vignettes**

Each chapter begins with a short prologue directly related to the main ideas of the chapter, often drawn from real life. Topics include genetic discoveries, such as the chromosomal basis of Down syndrome, the development of genetic technologies such as *in vitro* fertilization, and the birth of Louise Brown, the first IVF baby. These vignettes are designed to engage student interest in the topics covered in the chapter and to demonstrate that laboratory research often has a direct impact on everyday life.

### **Chapter Outlines**

At the beginning of each chapter, an outline provides an overview of the main concepts, secondary ideas, and examples. To help students grasp the central points, many of the headings have been rewritten 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.

### **Concepts and Controversies**

Within most chapters, students will find boxes that present ideas and applications related to the central concepts in the chapter. Some of these present interesting but tangential examples that should be of interest to the student, while others examine controversies that arise as genetic knowledge is transferred into technology and services.

## **Guest Essays**

Scattered throughout the book are essays written by prominent scientists. The essays introduce the human side of scientists, and summarize how they became interested in science, and how their work relates to larger issues in society.

## **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. These 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.

## **Sidebars**

Throughout the book, sidebars are used to highlight applications of concepts, present the latest findings, and point out controversial ideas without interrupting the flow of the text.

## **END-OF-CHAPTER-FEATURES**

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The end-of-chapter features have been revised and updated for this edition, and a new section on case histories has been added.

## **Case Studies**

As described earlier, this new section contains case histories 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.

## **Summary**

Each chapter ends with a numbered summary that restates the major ideas covered in the chapter. Beginning each chapter with an outline and ending with a summary of the major concepts and their applications helps focus the students' attention on the conceptual framework and minimizes the chance that they will attempt to learn by rote memorization of facts.

## **Questions and Problems**

The questions and problems at the end of each chapter are designed to test students' knowledge of the facts and their ability to reason from the facts to conclusions. This section has been revised and expanded by contributions from adopters of previous editions, by professionals in genetic research and health care services, and by students, who suggested many of the revisions and new problems.

## **Internet Activities**

Activities at the end of each chapter use Web sites to engage the student in activities related to the concepts discussed in the text. This section has been revised and expanded in this edition.



## For Further Reading

A list of readings is presented at the end of each chapter. These include reviews and general articles that are accessible to the non-scientist, as well as the key papers that describe discoveries covered in the chapter. The references have been updated just before publication to provide the most current coverage of the literature.

## ANCILLARY MATERIALS

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The expanded array of 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's Manual

An expanded and updated instructor's manual is available to help instructors in preparing class materials. It contains chapter outlines, chapter summaries, teaching/learning objectives, key terms, additional test questions, and discussion questions. It also contains answers to end-of-chapter questions and problems that are not given in the text.

### Test Bank

A computerized test bank, available on CD-ROM, can be customized and printed according to the instructor's preferences and needs. It contains approximately 800 test items.

### Transparencies

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

### Study Guide

A student study guide has been prepared by Nancy Shontz of Grand Valley State University. It is intended to enhance understanding of the text and course material. It includes chapter objectives and summaries, lists of terms, case worksheets (based on case studies in the text), discussion problems and questions, and other practice test items in multiple-choice, fill-in-the-blank, and modified true/false formats.

### Gene Link 2.0

A lecture/presentation tool for instructors, which allows them to select from a large database of images from this and other related Brooks/Cole texts as well as animations that can be arranged and edited to create dynamic, motivating lectures. The lectures created in this format (including animations, images, notes and URLs), can be posted on the Web.

### Online Genetics Newsletter

An electronic newsletter (on the Brooks/Cole Biology Resource Center) that features articles and essays on current topics and discoveries in the field of human and general genetics with links to related sites.

## Current Perspectives in Genetics

A reader prepared by Shelly Cummings of the University of Chicago features approximately 40 articles in molecular, classical, and human genetics. Each article begins with a brief introduction and ends with critical thinking or discussion questions. Answers are provided in the back of the book.

## Genetics on the Web: A Brief Guide to the Internet

A handy reference for students who need an introduction to research on the Internet. It includes a list of relevant URLs listed by genetics topics.

## Instructor's Edition

A special edition of the text for instructors that features extra frontmatter, including a visual preface.

## CONTACTING THE AUTHOR

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

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## ACKNOWLEDGMENTS

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A text is originally shaped by the vision and teaching philosophy of its author. By the time it reaches the fifth edition, it has been reshaped by changes in the field, and more importantly, by reviewers, editors, instructors, and students. Hopefully, with each of these changes, it becomes a more useful teaching tool that represents the distilled wisdom of those who have contributed to it. Over its lifetime, this text has had many reviewers, but three stand out for the time and effort they have expended in improving the presentation of topics, sharpening the language, and often, teaching or re-teaching me things about genetics and pedagogy. Through their patience and efforts, they have helped me become a better author and a better teacher. I owe them a great debt of gratitude, and I thank them for their contributions to this book and to the teaching of human genetics. They are: H. Eldon Sutton of the University of Texas, George Hudock of Indiana University, and Werner Heim of Colorado College.

To all the reviewers who helped in the preparation of this edition, I extend my thanks and gratitude for their efforts and many suggestions. Their efforts have enhanced the focus and presentation of the material.

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Once again, Michelle Murphy of Notre Dame University undertook the task of revising and adding to the end-of-chapter questions and problems. Writing good questions is one of the most difficult tasks in teaching, and Michelle's efforts show through in this expanded and pedagogically improved feature of the text. Shelly Cummings of the University of Chicago prepared the case histories at the end of each chapter. Her hard work and cheerful optimism have produced a new and valuable feature for this edition. The Internet activities were revised and expanded by Kim Finer of Kent State University, who is recognized as one of the pioneers in using electronic media in the classroom. She was assisted by Peter Follette.

The book has had a warm and welcoming reception in its new home at Brooks/Cole Publishing. The transition from West Publishing, via Wadsworth Publishing was guided by Gary Carlson, Executive Editor, who ensured that the transfer took place with a minimum of problems. He also worked to develop the approach and philosophy of this edition. My editor, Nina Horne, with her experience in publishing and knowledge of the marketplace, has helped me to emphasize the human aspects of human genetics, and guided the preparation of this edition. The heart and soul of this edition has been Marie Carigma-Sambilay, my project development editor. She worked tirelessly to coordinate all aspects of this project, and in spite of the inevitable delays, setbacks, and dilemmas, was always cheerful, upbeat, and optimistic. Editorial assistants Larisa Lieberman and John-Paul Ramin efficiently handled the review focus. Mary Anne Shahidi, the production coordinator, oversaw the book's production process. Her positive attitude was always a steadying influence. Chad Thomas and Terry Routley at Carlisle Communications smoothly guided the production of this edition, including the development of the design, art, and photo program. Roy Neuhaus was the creative force behind the cover design.

The concern and care expressed by the staff at Wadsworth and Brooks/Cole during my illness was heartwarming. I deeply appreciate their cards, flowers, and gifts, and I will always remember their kindness. I thank them and appreciate their efforts in overcoming the ensuing delays.

Once again, I wish to express special thanks to my colleague, Suzanne McCutcheon, who gave her time to help supervise the research of graduate students in my lab while this edition was in preparation. I also thank my students, Holly Dimitropoulos and David Wolff for their patience.

*Michael R. Cummings*

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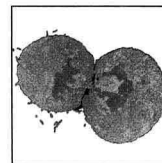
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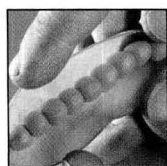




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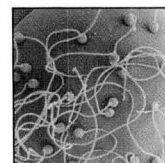
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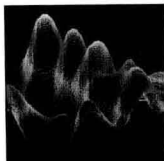
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