Principles of GENETICS

EIGHTH EDITION

Principles of GENETICS

EIGHTH EDITION

Eldon John Gardner
Utah State University

Michael J. Simmons
University of Minnesota

D. Peter Snustad
University of Minnesota

JOHN WILEY & SONS, INC.
New York Chichester Brisbane Toronto Singapore

Acquisitions Editor	Sally Cheney
Managing Editor	Joan Kalkut
Production Manager	Joe Ford
Cover and Text Designer	Dawn L. Stanley
Production Supervisor	Savoula Amanatidis
Manufacturing Manager	Lorraine Fumoso
Copy Editor	Marjorie Shustak
Photo Researcher	John Schultz
Photo Research Assistant	Hilary Newman
Photo Research Manager	Stella Kupferberg
Illustrations	John Balbalis John Wiley Illustration Dept. J/B Woolsey Associates
Cover Illustration	Steve Jenkins

Library of Congress Cataloging in Publication Data:

Gardner, Eldon John, 1909–1989
Principles of genetics / Eldon John Gardner, D. Peter Snustad, Michael J. Simmons.—8th ed.
p. cm.
Includes bibliographical references and index.
ISBN 0-471-50487-4
1. Genetics. I. Snustad, D. Peter. II. Simmons, Michael J. III. Title.
QH430.G37 1991
575.1—dc20 90-48758
CIP

10 9 8 7 6 5 4 3 2

The eighth edition
is dedicated to the memory of

Eldon John Gardner

who skillfully guided this textbook
through seven editions

PREFACE

The eighth edition is a continuing attempt to provide a true "principles of genetics" text that is up-to-date, readable, and challenging. Its major goal is to emphasize the central role that genetics plays in the lives of all organisms—plant, animal, and microbe—on planet Earth. In our complex modern society, with the very survival of our planet at stake, it is imperative that citizens be informed about scientific issues. Thus, our goal has been to write this text for students with only a minimal background in biology and chemistry. It is suitable for one-quarter, one-semester, or two-quarter courses in genetics that are designed for biology majors or for students from other disciplines. We have attempted to provide balanced coverage of many areas in genetics—classical, molecular, and population—but we recognize that each instructor will favor some topics over others. Consequently, this book has been written to allow instructors flexibility in the choice of topics and in the depth of coverage.

Genetics and related sciences have grown explosively in recent years, generating a large body of new and important information. As a result, the eighth edition has expanded considerably when compared with previous editions. A new introductory chapter attempts to convey the excitement of genetics today and to emphasize the central role that it plays in our lives. Two other new chapters cover the major developments in our knowledge of transposable genetic elements and the rapidly advancing methodologies of genetic engineering. Other chapters have been revised and expanded to incorporate new information about the structure and function of genes and their roles in development.

Compared with its predecessor, this edition has been substantially reorganized. The material on the structure and replication of DNA and chromosomes has been divided into two chapters, one focusing on prokaryotes and the other on eukaryotes. The analysis of genetic fine structure is included in two chapters, one dealing with classical studies and the other with molecular approaches, including cloning and sequencing. Also included in this molecular chapter are such important techniques as Southern, northern, and western blotting, PCR amplification of DNA, and oligonucleotide-directed site-specific mutagenesis. Regulation, which was treated in a single chapter in the previous edition, is now treated in four; two of these deal with the basic features of regulation, one focusing on prokaryotes and the other on eukaryotes, and two deal with special eukaryotic phenomena—the immune response and the genetic control of cell division. This last chapter includes an expanded discussion of the structure and function of oncogenes and protooncogenes.

Other changes have been made in the eighth edition to enhance its readability and focus. Multiple alleles are now treated in the same chapter as basic Mendelian genetics. The chapters on quantitative genetics and population and evolutionary genetics have been rewritten to provide more coherence to these somewhat difficult subjects. New material has been added to several other chapters; for example, the chapter on gene expression now contains sections on RNA splicing and the spliceosome, and the chapter on extrachromosomal inheritance contains new sections on the organization of mitochondrial and chloroplast genomes. As in previous editions, each chapter ends with an extensive set of problems and questions of varying difficulty, with the answers provided at the end of the book.

We believe that the eighth edition presents the basic principles of genetics in a clear and logical sequence, but we realize that many instructors will prefer other sequences. As mentioned earlier, this edition begins with a new introductory chapter that attempts to (1) convey some of the excitement that has resulted from recent developments in the field of genetics and (2) emphasize the important role that genetics plays in many aspects of our lives and, indeed, in the very survival of life on our planet. Chapter 2 describes the classic work of Gregor Mendel and the basic principles of Mendelian genetics that resulted from his work. Cells, chromosomes, and the process of meiosis that provide the basis for Mendel's laws are described in Chapter 3. The chromosomes and their behavior during gametogenesis are related to sex determination and sex-linked patterns of inheritance in Chapter 4. The molecular structure of genes and their modes of replication in prokaryotic chromosomes are introduced in Chapter 5. The complex structure and mechanism of replication of eukaryotic chromosomes are the subjects of Chapter 6. Chapter 7 covers the classical genetic topics of linkage, chromosome mapping, tetrad analysis, gene conversion, and the mechanism of crossing over. Recombination mechanisms unique to bacteria are covered in Chapter 8, and the structure and function of transposable genetic elements are the subjects of Chapter 9.

After considering the structure and recombination of genetic elements in Chapters 5 through 9, the focus shifts to the function of genetic elements in Chapter 10 (Gene Expression), which covers transcription, RNA processing ("splicing"), translation, and the genetic code. The mechanisms by which genes change or mutate are covered in Chapter 11. Chapters 12 and 13 dissect genes by both classical (Chapter 12) and molecular (Chapter 13) approaches. Chapter 13 also provides background information on the powerful tools of molecular genetics that are used in these dissetions: cloning vectors, Southern, northern, and western blots, PCR, and site-specific mutagenesis. Chapters 14 through 17 describe our present understanding of the various mechanisms by which gene expression and cell division are regulated. Control of gene expression in prokaryotes is the topic covered in Chapter 14, whereas Chapter 15 focuses on the regulation of

gene expression and development in eukaryotes. Although the genetic control of development in higher eukaryotes is much too complex to be covered in detail in an introductory genetics text, we hope the information presented in this chapter will give students an insight into the exciting developments in this field. Although much remains to be learned, molecular pictures of the genetic control of development in *Drosophila melanogaster* and the worm *Caenorhabditis elegans* have rapidly begun to emerge during the last few years.

The genetic control of the vast repertoire of antibodies that can be produced by higher animals was an enigma for decades. In the last few years, many of the mysteries of the immune system have been replaced by fascinating pictures of the genetic control of antibodies and T cell receptors; these new breakthroughs are described in Chapter 16. In Chapter 17, the genetic control of cell division is discussed, with emphases on the roles of protooncogenes and on the perturbations caused by oncogenes. The next two chapters discuss mutation in the broad sense of gross changes in chromosome structure (Chapter 18) and chromosome number (Chapter 19). In Chapter 20, the behavior of genes that are not located on the main nuclear chromosomes of the organism is considered, with an emphasis on the structure of the chromosomes of mitochondria and chloroplasts. Chapter 21 discusses those traits that are controlled by large numbers of genes and thus exhibit quantitative patterns of inheritance. In Chapter 22, the analysis of genes and their effects is extended to the level of the population; major emphases are the distributions of alleles in populations and the changes in these distributions (evolution) over time in response to factors such as mutation, genetic drift, and selection. Chapter 23 provides a brief introduction to the genetic factors that influence behavior.

Chapter 24 introduces the student to some of the present and future applications of genetic engineering. We realize that most introductory genetics classes will cover few, if any, of the topics discussed in Chapter 24. However, we believe that this chapter will be of considerable interest to many students, and we anticipate that those students who find genetics to be fascinating and challenging will read Chapter 24 on their own. Some instructors will probably choose to integrate certain topics from Chapter 24 into their courses. For example, a class with a plant genetics emphasis might appropriately include the material on *Agrobacterium tumefaciens* and plant transformation via the Ti plasmid. Other classes may choose to include the section on somatic-cell gene therapy, and so on. A glossary of important genetic terms, the answers to all the problems in the book, and an index complete the text.

Our thanks to students, teachers, and colleagues who have suggested improvements for the book. We especially thank Rayla Temin, R. H. Whalen, Linda Kosturko, Clint Magill, Peter Weijksnora, Charles Rodell, Hugh Stanley, Larry Puckett, Robert M. Fineman, Irene Uchida, Scott R. Woodward, John R. Simmons, and Franklin D. Enfield, who have all made valuable contributions. We also thank the following reviewers for their valuable suggestions and comments: Judith Van Houten, The University of Vermont; Robert Petters, Pennsylvania State University; Darrell S. English, Northern Arizona University; Romesh C. Mehra, Indiana University at South Bend, Glenn Wolfe, Kansas University; Paul J. Homsher, Old Dominion University; Paul A. Roberts, Oregon State University; Asim Esem, Virginia Polytechnic Institute and State University; Howard Laten, Loyola University; Charles H. Green, Glassboro State College; Alice S. Hunter, College of the Pacific; Martin Bard, Purdue University; John B. Jenkins, Swarthmore College; Kandy D. Baumgardner, Eastern Illinois University; Dwight E. Wilson, Rensselaer Polytechnic Institute; Carter Denniston and William R. Engels, University of Wisconsin, Madison; H. James Price, Texas A & M University; Peter Dawson, Oregon State University; Barry Bean, Lehigh University; Dean Whited, North Dakota State University.

We also gratefully acknowledge the invaluable assistance of Sally Cheney, editor; John Balbalis and John D. Woolsey, illustrators; Dawn L. Stanley, designer; Stella Kupferberg and John Schultz, photo researchers; Joe Ford, production manager; Marjorie Shustak, supervising copy editor; John Thomas, copy editor; Savoula Amanatidis, production supervisor; and Virginia McTavish, manuscript preparator.

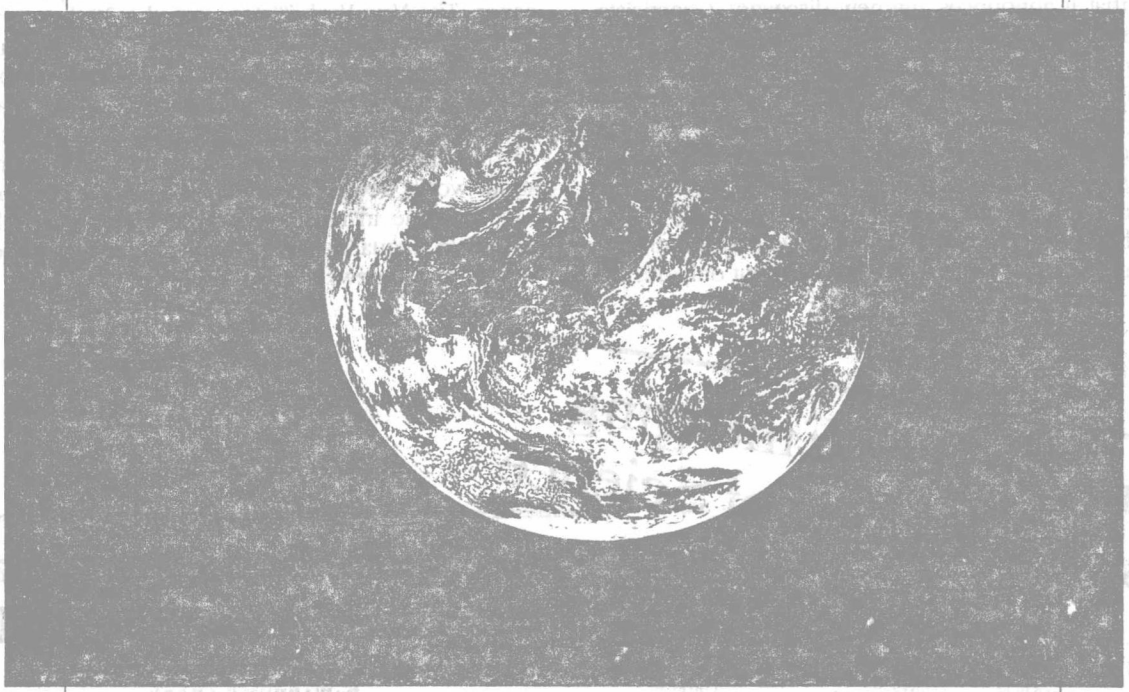
Credits for illustrations, tables, and quotations from other individuals or publications are given with the individual items.

MICHAEL J. SIMMONS
D. PETER SNUSTAD

BRIEF CONTENTS

CONTENTS

Chapter 1



Introduction

(Photo above): View of the Earth from the Apollo 8 spacecraft.

"*GENE Studies Emerging as Key Engine of Science*"—headline, *The New York Times*, September 6, 1988. The significance of this headline is not that it announces any new discovery. Geneticists have known for decades that genes play central roles in science and, in fact, in all aspects of the lives of all living organisms. The significance of this headline is that our society as a whole, or at least the more informed component of our society, now also recognizes the major roles that genes play in shaping our lives and the lives of all living creatures on planet Earth. Of course, such headlines have been commonplace in science journals for decades, but only during the last few years have these headlines appeared frequently in the popular press, particularly the news media. For example, a quick scan of one major newspaper, *The New York Times*, over the 30-day period from September 6, 1988, to October 6, 1988, revealed three major headlines focusing on genetics; one, *"Panel Backs Gene Transplant For Cancer Treatment Test,"* was on the front page (Fig. 1.1). In fact, as anyone who makes a significant effort to stay informed about world developments knows, similar headlines have become increasingly prevalent during the last decade in all quality newspapers as well as in the radio and television news media throughout the world.

Gene Studies Emerging As Key Engine of Science

By HAROLD M. SCHMECK Jr.

THE science of genetics is fast becoming what geneticists always knew it was: the central and most provocative science of life.

Toronto, president of the
the Toronto meeting wa
meetings, which are he
Four thousand scientist
tries attended, twice t
and four times the nur
resented three decade

Panel Backs Gene Transplant For Cancer Treatment Test

By WARREN E. LEARY

Special to The New York Times

WASHINGTON, Oct. 3 — An influential advisory committee recommended today that the Government for the first time approve the transplanting of foreign genes into human patients.

and the Food and Drug Administration.

But the scientist and his principal coworker on the cancer experiment, Dr. Steven Rosenberg, said they had indications from the remaining authorities

DNA Pioneer to Tackle Biggest Gene Project Ever

By HAROLD M. SCHMECK Jr.

DR. James D. Watson, who is about to assume a key role in the largest biological research project ever contemplated, sees that effort as the redemption of a longstanding bargain between scientists and other citizens.

"I think it is imperative to keep our bargain with the people who have supported biomedical research," he said in an interview Friday. The project will immensely expand scientists' knowledge of human genetics. Directly and indirectly, that knowledge will be used to seek new ways of curing or coping with important diseases by teasing out the secrets of the genes.

Dr. Watson was co-discoverer in 1953 of the structure of DNA, the substance of the genes in all living things. In 1962, he shared a Nobel Prize for that work with Dr. Francis H. C. Crick, now at the Salk Institute in La Jolla, Calif., and Dr. Maurice H. F. Wilkins of Cambridge University in England. The discovery showed how DNA functions as the repository of all hereditary information and led to a revolution in biology that continues to accelerate and expand.

The new job that Dr. Watson begins this week involves helping to organize a huge federally supported project to define all of the roughly 50,000 to 100,000 human genes. It is known as the genome project; a genome being the sum of all the genes of any organism. The project, if approved by Congress, is expected to take 15 years and to cost several billion dollars.

Dr. Watson has been appointed associate director of the National Institutes of Health for genome research, a newly created post to oversee the institutes' involvement with the genome project.

In his early years of research, Dr. Watson was known as a brash, witty and often abrasive enfant terrible. The new appointment seemingly completes his metamorphosis into a senior statesman of science.

"People know that I am interested in biology as a whole," he said. "Of course, I am also interested in human beings."

The project is a logical outcome of the discovery made in 1953, but until recently, the idea would have been derided as fantasy because it was so far beyond the technical capabilities of the science. Only a relatively few genes have been completely defined to date but the new project will try to define the chemical compusiton of them all. The effort is made possible now by a series of technical and

Continued on Page C16

The New York Times/Michael Shavel

Dr. James D. Watson, who pioneered research into the structure of DNA and now heads an effort to map the human genome.

Figure 1.1 Headlines in the *The New York Times* on September 6 and October 8, 1988, heralding some of the exciting recent developments in genetics. (Copyright 1988 by The New York Times Company. Photograph by Michael Shavel, New York Times Pictures. Reprinted with permission.)