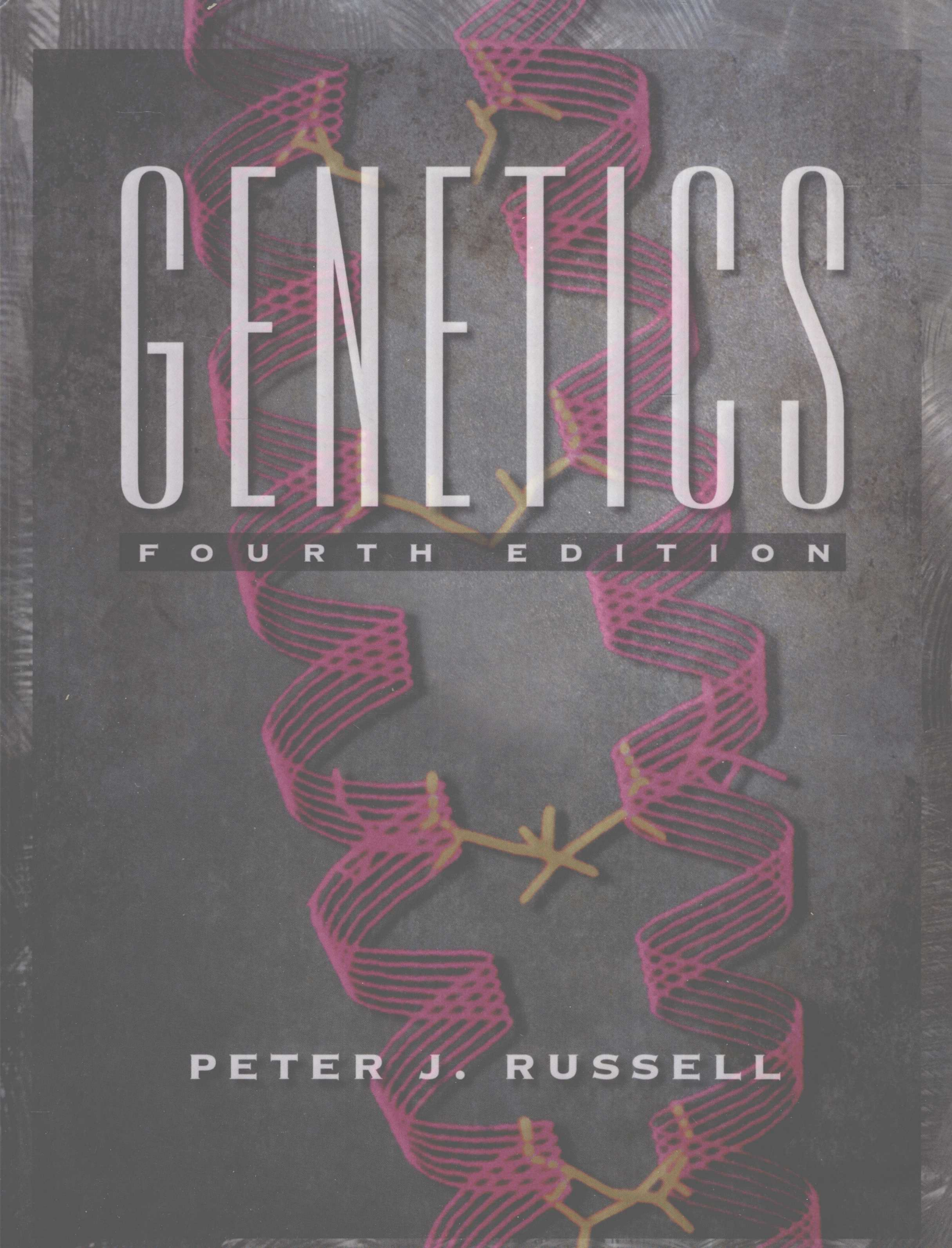
GENETICS

FOURTH EDITION



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GENETICS

Fourth Edition

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REED COLLEGE

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PREFACE

OVERVIEW OF THE TEXT

Genetics has long been one of the core areas in biology, and in the now thirty years since the DNA mysteries were first unravelled, our knowledge and understanding about genetics and its related disciplines has progressed at an incredible rate. The many applications of this exciting flood of discoveries continue to affect our daily lives and bring benefit to humanity.

Genetics, Fourth Edition, has been crafted carefully to present comprehensive, quality coverage of the field of genetics in an accessible manner. As with the previous editions, this new edition emphasizes an experimental, inquiry-based approach with solid treatment of many of the research experiments that have contributed to our knowledge of the subject. In this way students are exposed to the "process of science," that is, the way research scientists pose scientific hypotheses, design experiments to test those hypotheses, and analyze the data produced by the experiments. Learning about the formulation and study of scientific questions is of value not only in genetics, but in all other areas of biology, as well as in other fields of science. Great care has been taken to keep the text accessible to students by making it easy to read, with a consistent level of coverage and a logical development of ideas throughout. Many significant advances have been made in the field of genetics since the last edition, particularly those resulting from the use of molecular experimental approaches. The important advances have been included in this edition with an effort to ensure that the concepts being taught are not obscured by excessive facts and detail. *Genetics,* Fourth Edition, is an ideally suitable text for students who have had some background in biology and chemistry, and who are interested in learning the concepts of genetics from a research-oriented perspective.

This edition of *Genetics* retains the overall approach, logical progression of ideas, organization, and pedagogical features (such as "Principal Points," "Keynotes," "Summaries," and "Analytical Approaches for Solving Genetics Problems") that have made previous editions valued learning tools for students of genetics. It also retains a format that allows instructors to use the chapters out of sequence to accommodate various teaching strategies.

The fourth edition includes the following improvements:

- A new introductory chapter discusses the main branches of genetics, geneticists and their areas of research, and the main properties of genes in order to introduce this exciting, provocative field, and to acquaint the student with what is to come.
- All the molecular aspects of genetics are updated, so that the book continues to reflect our current understanding of genes at the molecular level.
- Human examples are used more extensively throughout the text, particularly with regard to new molecular understandings of various human genetic diseases.
- The coverage of recombinant DNA technology has been updated and expanded to reflect the rapid developments in that area. For example, a description of the cloning of the human cystic fibrosis gene is included as an illustration of how human genes may be isolated and studied.
- The chapters on "Transcription" and "RNA Molecules and RNA Processing" are now integrated into one chapter.
- The chapter on "Chromosomal Mutations" is now placed earlier in the text within the transmission genetics section to link the material more closely with the understanding of the relationship between genes and chromosomes developed in the early chapters. The involvement of triplet repeat mutations in certain human genetic diseases is included in this revised chapter.
- The chapters on genetics of populations and quantitative genetics have been revised by Robert Kaplan of Reed College and are now presented in the order "Population Genetics," then "Quantitative Genetics" to reflect the logical progression from studies of heredity in groups or individuals for traits determined by one or a few genes, to studies of heredity in groups or individuals for traits determined by many genes.
- New questions and problems have been added throughout the book, many of them supplied by reviewers.

- A new appendix has been added, detailing Barbara McClintock's extraordinary contributions to the study of genetics.
- Many new references have been added to the "suggested readings" that accompany each chapter.

ORGANIZATION AND COVERAGE

The three major areas of genetics—transmission genetics, molecular genetics, and population and quantitative genetics—are covered in 22 chapters. Chapter 1 is an introductory chapter designed to summarize the main branches of genetics, to explain what geneticists do and what their areas of research encompass, and to introduce the main properties of genes and the main experimental approaches used in genetics research today. The next seven chapters deal with transmission of the genetic material. Chapters 2 and 3 present the basic principles of genetics in relation to Mendel's laws. Chapter 2 is focused on Mendel's contributions to our understanding of the principles of heredity, while Chapter 3 covers mitosis and meiosis in the context of both animal and plant life cycles, the experimental evidence for the relationship between genes and chromosomes, and methods of sex determination. Mendelian genetics in humans is introduced in Chapter 2 with a focus on pedigree analysis and autosomal traits. The topic is continued in Chapter 3 with respect to sex-linked genes.

The exceptions to and extensions of Mendelian analysis (such as the existence of multiple alleles, the modification of dominance relationships, gene interactions and modified Mendelian ratios, essential genes and lethal alleles, and the relationship between genotype and phenotype) are described in Chapter 4. In Chapter 5, we describe how the order of and distance between the genes on eukaryotic chromosomes are determined in genetic experiments designed to quantify the crossovers that occur during meiosis. Chapter 6 considers advanced mapping analysis in eukaryotes, focusing on tetrad analysis, primarily in fungal systems; on mapping eukaryotic genes through mitotic analysis; and using somatic cell hybrids to map genes in human chromosomes. With the understanding of the relationship between genes and chromosomes obtained from Chapters 2 through 6, chromosomal mutations—changes in normal chromosome structure or chromosome number—are discussed in Chapter 7. Chromosomal mutations in eukaryotes and human disease syndromes that result from chromosomal mutations, including the recently discovered triplet repeat mutations, are emphasized. In Chapter 8, we discuss the ways of mapping genes in bacteriophages and in bacteria, which take advantage of the processes of transformation, conjugation, and transduction. Fine structure analysis of bacteriophage genes concludes this chapter.

Chapters 9 through 15 comprise the "molecular core" of *Genetics,* Fourth Edition, detailing the current level of our knowledge about the molecular aspects of genetics. In Chapter 9, we examine some aspects of gene function, such as the genetic control of the structure and function of proteins and enzymes, and the role of genes in directing and controlling biochemical pathways. A number of examples of human genetic diseases that result from enzyme deficiencies are described to reinforce the concepts. The discussion of gene function in Chapter 9 enables students to understand the important concept that genes specify proteins and enzymes, setting them up for the following chapters in which gene structure and expression is discussed.

In Chapter 10, we cover the structure of DNA, presenting the classical experiments that revealed DNA and RNA to be genetic material and that established the double helix model as the structure of DNA. The details of DNA structure and organization in prokaryotic and eukaryotic chromosomes are set out in Chapter 11. We cover DNA replication in prokaryotes and eukaryotes, and recombination between DNA molecules in Chapter 12.

After thoroughly explaining the nature of the gene and its relationship to chromosome structure, in Chapter 13 we discuss the first step in the expression of a gene—transcription. First, we describe the general process of transcription and then present the currently understood details of the transcription of messenger RNA, transfer RNA, and ribosomal RNA genes, and the processing of the initial transcripts to the mature RNAs for both prokaryotes and eukaryotes. In Chapter 14, we describe the structure of proteins, the evidence for the nature of the genetic code, and a detailed expression of our current knowledge of translation in both prokaryotes and eukaryotes. A brief discussion of how eukaryotic proteins are sorted to the compartments in which they function is also included in Chapter 14. In Chapter 15, we discuss recombinant DNA technology and other molecular techniques that are now essential tools of most areas of modern genetics. There are descriptions of the use of recombinant DNA technology to clone and characterize genes and to manipulate DNA, followed by a discussion of the applications of recombinant DNA technology in the analysis of biological processes, the diagnosis of human diseases, the isolation of human genes, the Human Genome Project (with the goal of mapping and sequencing the complete

genomes of humans and other selected organisms), forensics (DNA fingerprinting), gene therapy, the development of commercial products, and the genetic engineering of plants.

The next two chapters focus on regulation of gene expression in prokaryotes (Chapter 16) and eukaryotes (Chapter 17). In Chapter 16, we discuss the operon as a unit of gene regulation, the current molecular details in the regulation of gene expression in bacterial operons, and regulation of genes in bacteriophages. In Chapter 17, we explain how eukaryotic gene expression is regulated, stressing molecular changes that accompany gene regulation, short-term gene regulation in simple and complex eukaryotes, gene regulation in development and differentiation, and immunogenetics.

In Chapters 18 and 19, we describe some of the ways in which genetic material can change or be changed. Chapter 18 covers the processes of gene mutation, the procedures that screen for potential mutagens and carcinogens (the Ames test), some of the mechanisms that repair damage to DNA, and some of the procedures that are used to isolate particular classes of mutants from a heterogeneous population. Chapter 19 presents the structures and movements of transposable genetic elements in prokaryotes and eukaryotes, as well as discussions of tumor viruses, focusing on retroviruses and the oncogenes they may carry.

In Chapter 20, we address the organization and genetics of extranuclear genomes of mitochondria and chloroplasts. We cover the current molecular information about the organization of genes within the extranuclear genomes, and the classical genetic experiments that are used to study the inheritance of extranuclear genes. New to this chapter are discussions of RNA editing and genomic imprinting.

In Chapters 21 and 22, we describe the genetics of populations and quantitative genetics, respectively. In Chapter 21, "Population Genetics," we present the basic principles in population genetics, extending our studies of heredity from the individual organism to a population of organisms. This chapter includes an integrated discussion of the developing area of conservation genetics. In Chapter 22, "Quantitative Genetics," we consider the heredity of traits in groups of individuals that are determined by many genes simultaneously. In this chapter we also discuss heritability: the relative extent to which a characteristic is determined by genes or by the environment. Both Chapters 21 and 22 include discussions of the application of molecular tools to these areas of genetics. Chapter 21, for example, includes a section on measuring genetic variation with RFLPs and DNA sequencing, and a discussion of molecular evolution.

PEDAGOGICAL FEATURES

Because the field of genetics is complex, making the study of it potentially difficult, we have incorporated a number of special pedagogical features to assist students and to enhance their understanding and appreciation of genetic principles. These features have proved to be very effective in previous editions of this text:

- Each chapter opens with an outline of its contents and a section called "Principal Points." Principal Points are short summaries that alert students to the key concepts they will encounter in the material to come.
- Throughout each chapter, strategically placed "Keynote" summaries emphasize important ideas and critical points.
- Important terms and concepts—highlighted in bold—are clearly defined where they are introduced in the text. For easy reference, they are also compiled in a Glossary at the back of the book. The Glossary includes the page numbers on which the term and concepts are introduced so students can look up the definitions and easily find the text location to read more about them.
- Some chapters include boxes covering special topics related to chapter coverage. Some of these boxed topics are: *Genetic Terminology* (Chapter 2); *Denaturation-Renaturation Analysis of DNA* (Chapter 11); *Equilibrium Density Gradient Centrifugation* (Chapter 12); *Labeling of DNA* (Chapter 15); *Hardy, Weinberg, and the History of Their Contribution to Population Genetics* (Chapter 21); and *Analysis of Genetic Variation with Protein Electrophoresis* (Chapter 21).
- Chapter summaries close each chapter, further reinforcing the major points that have been discussed.
- With the exception of the introductory Chapter 1, all chapters conclude with a section entitled "Analytical Approaches for Solving Genetics Problems." Genetics principles have always been best taught with a problem-solving approach. However, beginning students often do not acquire the necessary experience with basic concepts that would enable them to attack assigned problems methodically. In the "analytical approaches" sections (pioneered in earlier editions of this text), typical genetic problems are talked through in step-by-step detail to help students understand how to tackle a genetics problem by applying fundamental principles.
- The problem sets that close the chapters include

approximately 530 questions and problems designed to give students further practice in solving genetics problems. The problems for each chapter represent a range of topics and difficulty. The answers to questions indicated by an asterisk (*) can be found at the back of the book, and answers to all questions are available in a separate supplement, the *Study Guide and Solutions Manual.*

- Comprehensive and up-to-date suggested readings for each chapter are listed at the back of the book.
- Special care has been taken to provide the most useful Index—extensive, accurate, and well cross-referenced.

SUPPLEMENTS

A *Study Guide and Solutions Manual* to accompany this text has been prepared by Bruce Chase of the University of Nebraska. In addition to detailed solutions for all the problems in the text, the *Guide* contains the following features for each chapter: a review of important terms and concepts; an "Analytical Approaches for Solving Genetics Problems" section, which provides guidance and tips on solving problems and avoiding common pitfalls; and additional questions for practice and review.

The study guide includes a variety of essays, multiple choice and matching questions, Key Terms and Concepts, Chapter Outline, Approach to Analytical Thinking and Comprehensive Solutions.

An Instructor's Manual/Test Bank, prepared by Holly Ahern of Adirondack Community College, is also available to adopters. The Instructor's Manual includes such features as Learning Objectives and Chapter Outlines. The Test Bank includes approximately 900 questions consisting of multiple choice, fill in the blanks and true/false .

A set of 134 full-color transparencies is available to adopters of the text.

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Peter J. Russell

Detailed Contents

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