

国外生命科学优秀教材

Concepts of Genetics

Ninth Edition

遗传学概要

影印版

William S. Klug
Michael R. Cummings
Charlotte A. Spencer
Michael A. Palladino



科学出版社



国外生命科学优秀教材

遗传学概要 (影印版)

Concepts of Genetics

NINTH EDITION

William S. Klug
THE COLLEGE OF NEW JERSEY

Michael R. Cummings
ILLINOIS INSTITUTE OF TECHNOLOGY

Charlotte A. Spencer

UNIVERSITY OF ALBERTA
常州大学图书馆
藏 A. Balladino
MONMOUTH UNIVERSITY

WITH CONTRIBUTIONS BY
SARAH M. WARD,
COLORADO STATE UNIVERSITY

Q3
=K66-2

科学出版社

北京

内 容 简 介

本书保持了前八版的编写特色：注重基础知识，概念清晰准确，探讨解决专业问题，师生易学易教。第九版内容拓展到基因组、生物信息、蛋白质组及相关前沿领域。该书在亚马逊专业教材销售排行榜长期名列前茅，被许多北美、欧洲高校教学选用。

第九版内容主要包括：遗传学概况，有丝分裂和减数分裂，孟德尔遗传学，孟德尔比率，真核生物染色体图谱，细菌和噬菌体的遗传分析，性别决定和性染色体，染色体突变：染色体数量和分布，核外遗传，DNA 结构与分析，DNA 复制与重组，染色体 DNA 的组织，重组 DNA 技术和基因克隆，遗传密码和转录，翻译，基因突变和 DNA 修复，原核生物基因表达，真核生物基因表达调控，模式生物的发育遗传学，癌症和细胞周期调控，基因组学，蛋白质组学，生物信息学，基因组动力学：转座子，免疫遗传学，真核病毒，基因组分析——基因功能，基因工程的应用及生物伦理剖析，数量遗传学和多因子性状，行为遗传学，群体遗传学，进化遗传学，保护遗传学。

本书适合生命科学相关专业教学选用，也可供从业人员参考使用。

Original edition, entitled CONCEPTS OF GENETICS, 9E, 9780321524041 by KLUG, WILLIAM S.; CUMMINGS, MICHAEL R.; SPENCER, CHARLOTTE A.; PALLADINO, MICHAEL A., published by Pearson Education, Inc, publishing as Benjamin, Copyright © 2009.

All rights reserved. No part of this book may be reproduced or transmitted in any form or by any means, electronic or mechanical, including photocopying, recording or by any information storage retrieval system, without permission from Pearson Education, Inc.

China edition published by PEARSON EDUCATION ASIA LTD., and SCIENCE PRESS LTD Copyright © 2011.

This edition is manufactured in the People's Republic of China, and is authorized for sale and distribution in the People's Republic of China exclusively (except Taiwan, Hong Kong SAR and Macau SAR).

本版本仅授权在中国地区（除台湾、香港与澳门）销售与发行。

图书在版编目(CIP)数据

遗传学概要=Concepts of Genetics: 第9版: 英文/(美)克卢格(Klug, W. S.)等著. —影印本.
—北京: 科学出版社, 2011.6

国外生命科学优秀教材

ISBN 978-7-03-031760-5

I. ①生… II. ①克… III. ①遗传学-教材-英文 IV. ①Q3

中国版本图书馆 CIP 数据核字(2011)第 119703 号

责任编辑：单冉东

责任印制：张克忠 / 封面设计：陈 敬

科学出版社出版

北京东黄城根北街16号

邮政编码：100717

http://www.sciencep.com

双青印刷厂印刷

科学出版社发行 各地新华书店经销

*

2011年7月第一版 开本：A4(890×1240)

2011年7月第一次印刷 印张：56

印数：1—3 000 字数：1 500 000

定价：138.00元

(如有印装质量问题，我社负责调换)

NOBEL PRIZES AWARDED FOR RESEARCH IN GENETICS OR GENETICS-RELATED AREAS

YEAR	RECIPIENTS	NOBEL PRIZE	RESEARCH TOPIC
2007	M. R. Capecchi M. J. Evans O. Smithies	Medicine or Physiology	Development of gene-targeting technology essential to the creation of knockout mice serving as animal models of human disease
2006	R. Kornberg	Chemistry	Molecular basis of eukaryotic transcription
2006	A. Z. Fire C. C. Mello	Medicine or Physiology	RNA interference (RNAi)
2002	S. Brenner H. R. Horvitz J. E. Sulston	Medicine or Physiology	Genetic regulation of organ development and programmed cell death
2001	L. Hartwell T. Hunt P. Nurse	Medicine or Physiology	Genes and regulatory molecules controlling the cell cycle
1999	G. Blobel	Medicine or Physiology	Genetically-encoded amino acid sequences in proteins that guide their cellular transport
1997	S. Prusiner	Medicine or Physiology	Prions—a new biological principle of infection
1995	E. B. Lewis C. Nusslein-Volhard E. Wieschaus	Medicine or Physiology	Genetic control of early development in <i>Drosophila</i>
1993	R. Roberts P. Sharp	Medicine or Physiology	RNA processing of split genes
	K. Mullis M. Smith	Chemistry	Development of polymerase chain reaction (PCR) and site-directed mutagenesis (SDM)
1989	J. M. Bishop H. E. Varmus	Medicine or Physiology	Role of retroviruses and oncogenes in cancer
	T. R. Cech S. Altman	Chemistry	Ribozyme function during RNA splicing
1987	S. Tonegawa	Medicine or Physiology	Genetic basis of antibody diversity
1985	M. S. Brown J. L. Goldstein	Medicine or Physiology	Genetic regulation of cholesterol metabolism
1983	B. McClintock	Medicine or Physiology	Mobile genetic elements in maize
1982	A. Klug	Chemistry	Crystalline structure analysis of significant complexes, including tRNA and nucleosomes
1980	P. Berg W. Gilbert F. Sanger	Chemistry	Development of recombinant DNA and DNA sequencing technology
1978	W. Arber D. Nathans H. O. Smith	Medicine or Physiology	Recombinant DNA technology using restriction endonuclease technology

YEAR	RECIPIENTS	NOBEL PRIZE	RESEARCH TOPIC
1976	B. S. Blumberg D. C. Gajdusek	Medicine or Physiology	Elucidation of the human prior-based diseases, kuru and Creutzfeldt-Jakob disease
1975	D. Baltimore R. Dulbecco H. Temin	Medicine or Physiology	Molecular genetics of tumor viruses
1972	G. M. Edelman R. R. Porter	Medicine or Physiology	Chemical structure of immunoglobulins
	C. Anfinsen	Chemistry	Relationship between primary and tertiary structure of proteins
1970	N. Borlaug	Peace Prize	Genetic improvement of Mexican wheat
1969	M. Delbrück A. D. Hershey S. E. Luria	Medicine or Physiology	Replication mechanisms and genetic structure of bacteriophages
1968	H. G. Khorana M. W. Nirenberg	Medicine or Physiology	Deciphering the genetic code
	R. W. Holley	Medicine or Physiology	Structure and nucleotide sequence of transfer RNA
1966	P. F. Rous	Medicine or Physiology	Viral induction of cancer in chickens
1965	F. Jacob A. M. Lwoff J. L. Monod	Medicine or Physiology	Genetic regulation of enzyme synthesis in bacteria
1962	F. H. C. Crick J. D. Watson M. H. F. Wilkins	Medicine or Physiology	Double helical model of DNA
	J. C. Kendrew M. F. Perutz	Chemistry	Three-dimensional structure of globular proteins
1959	A. Kornberg S. Ochoa	Medicine or Physiology	Biological synthesis of DNA and RNA
1958	G. W. Beadle E. L. Tatum	Medicine or Physiology	Genetic control of biochemical processes
	J. Lederberg	Medicine or Physiology	Genetic recombination in bacteria
	F. Sanger	Chemistry	Primary structure of proteins
1954	L. Pauling	Chemistry	Alpha helical structure of proteins
1946	H. J. Muller	Medicine or Physiology	X-ray induction of mutations in <i>Drosophila</i>
1933	T. H. Morgan	Medicine or Physiology	Chromosomal theory of genetics
1930	K. Landsteiner	Medicine or Physiology	Discovery of human blood groups

Preface

It is essential that textbook authors step back and look with fresh eyes as each edition of their work goes into planning and preparation. In doing so, they always need to pose two main questions: (1) How has the body of information in their field—in this case genetics—shifted since the last edition? (2) What pedagogic innovations might they add that will unquestionably enhance students' learning? The preparation of the 9th edition of *Concepts of Genetics*, now well into its third decade of providing support for students studying in this field, occasioned such a fresh look. And what we clearly saw is that in the past three years, the rapid expansion of the study of genomics, and the impact of that information at all levels in the field of genetics, represent the major advances in the field. In keeping with these observations, we have placed particular emphasis on genomics as we carefully revised and updated the entire text. This was accomplished not only by adding a new chapter related to genomics, but also by devising a new pedagogic feature that brings genomic information into each and every chapter in the text. Called *Exploring Genomics*, this innovation provides the information necessary for students to explore on the Web one or more databases closely related to the chapter topics being studied. We will discuss the details of our added coverage of genomic information as well as *Exploring Genomics* later in this preface.

The field of genetics has grown tremendously since our book was first published, both in what we know and what we want beginning students to comprehend. In creating this edition, we sought not only to continue to familiarize students with the most important discoveries of the past 150 years, but also to help them relate this information to the underlying genetic mechanisms that explain cellular processes, biological diversity, and evolution. We have also emphasized connections that link transmission genetics, molecular genetics, genomics, and proteomics.

In the first decade of this new millennium, discoveries in genetics continue to be numerous and profound. As students of genetics, the thrill of being part of this era must be balanced by a strong sense of responsibility and careful attention to the many scientific, social, and ethical issues that have already arisen, and others that will undoubtedly arise in the future. Policy makers, legislators, and an informed public will increasingly depend on knowledge of the details of genetics in order to address these issues. As a result, there has never been a greater need for a genetics textbook that clearly explains the principles of genetics.

Goals

In the 9th edition of *Concepts of Genetics*, as in all past editions, we had six major goals. Specifically, we sought to:

- Emphasize the basic concepts of genetics.

- Write clearly and directly to students in order to provide understandable explanations of complex, analytical topics.
- Establish a careful organization within and between chapters.
- Maintain constant emphasis on science as a way of illustrating how we know what we know.
- Propagate the rich history of genetics, which so beautifully illustrates how information is acquired during scientific investigation.
- Create inviting, engaging, and pedagogically useful full-color figures enhanced by equally helpful photographs to support concept development.

These goals collectively serve as the cornerstone of *Concepts of Genetics*. This pedagogic foundation allows the book to be used in courses with many different approaches and lecture formats. Although the chapters are presented in a coherent order that represents one approach to offering a course in genetics, they are nevertheless written to be independent of one another, allowing instructors to utilize them in various sequences. We believe that the varied approaches embodied in these goals together provide students with optimal support for their study of genetics.

Writing a textbook that achieves these goals and having the opportunity to continually improve on each new edition has been a labor of love for us. The creation of each of the nine editions is a reflection not only of our passion for teaching genetics, but also of the constructive feedback and encouragement provided by adopters, reviewers, and our students over the past three decades.

Major Innovations and Strengths of This Edition

- **Organization**—A revised organization, both within and between chapters, better illustrates how genetics is taught in the era of genomics. The introductory chapter provides an essential overview of molecular biology as a way to connect the early transmission genetics chapters to the molecular topics that follow. Enhanced coverage of model organisms is woven throughout many chapters but is especially prominent in the Introduction to Genetics (Chapter 1), as well as the chapters that consider Developmental Genetics of Model Organisms (Chapter 19) and the Genomic Analysis: Dissection of Gene Function (Chapter 23).

The table of contents marks a number of changes from the 8th edition. The chapter introducing Recombinant DNA and Gene Cloning, the foundation on which genomic information is initially obtained, is more suitably located in Part Two of the text (Chapter 13), which focuses specifically on DNA technology.

The chapters that address Developmental Genetics and Cancer and Cell-Cycle Regulation have been relocated so that they now follow one another (Chapters 19 and 20) and are placed just after the chapters on Gene Regulation (Chapters 17 and 18). This reorganization recognizes the common links between these topics and integrates them into more cohesive coverage.

- **Pedagogy**—For this edition we have created an exciting new feature that appears in every chapter: *Exploring Genomics*. The presence and execution of this feature confirm for students that genomics impacts every aspect of genetics. Introduced in each entry are one or more genomics-related Web sites that collectively are among the best publicly available resources and databases that scientists around the world rely on for current information in genomics. The student is led through a series of interactive exercises that ensure their familiarity with the type of genomic or proteomic information available through the site and with applications of this information. The exercises instruct students on how to explore specific topics and how to access significant data. Questions are provided to guide student exploration, and the student is challenged to further explore the sites on their own. Their participation in these entries ensures that students become knowledgeable about “cutting-edge” genetic topics in genomics, proteomics, bioinformatics, and related areas as well as introducing them to the impact and application of the field of genomics to every aspect of genetics. Most importantly, the *Exploring Genomics* feature integrates genomics throughout the text, and each exercise is connected to chapter content in order to expand or reinforce genomics-related topics from the chapter.

Another valued pedagogic feature, first introduced in the 8th edition, continues to appear in each chapter: *How Do We Know?* Previously appearing in the text of each chapter, entries have been consolidated and moved to the *Problems and Discussion Questions* section found at the end of each chapter. The *How Do We Know?* logo identifies this feature among the other problems. Each entry asks the student to identify and examine the experimental basis underlying important concepts and conclusions presented in the chapter. Addressing these questions will aid the student in more fully understanding, rather than only memorizing, the end-point of each body of research. This feature is an extension of the learning approach in biology often referred to as “Science as a Way of Knowing.”

Finally, a third feature, *Now Solve This*, has been maintained and is integrated within the text of each chapter. Each entry directs the student to a problem found at the end of the chapter that is closely related to the current text discussion. In each case, a pedagogic hint is provided to aid in solving the problem. This feature more closely links the text discussions to the problems.

All three of these features, which appear throughout each chapter, seek to challenge students to think more deeply about,

and thus understand more comprehensively, the information he or she has just finished studying.

- **New Chapters**—In keeping with our intent to offer information that represents the “cutting edge” of genetics and, as well, to increase the coverage of genomics and of model organisms utilized in genetic study, we have created two new chapters that ensure that we meet these goals. The first new chapter, *Genome Dynamics—Transposons, Immunogenetics, and Eukaryotic Viruses* (Chapter 22), provides modern coverage of three important topics essential to the study of modern genetics and also establishes that the genome is not a static entity. The second new chapter, *Genetics and Behavior* (Chapter 26), reflects our growing knowledge of the way genes impact many aspects of an organism’s existence within the environment in which it finds itself. This topic is as interesting as any in the text and is important because the findings surrounding it intersect our knowledge of our own species.

In addition, a third chapter, that relates to genomics, has received an important update and shift in emphasis: *Genomic Analysis—Dissection of Gene Function* (Chapter 23). This chapter establishes the important concept that genomic analysis allows us to explore more deeply the nature of the gene and how it functions. Relying on mutational studies of genomes, this topic represents one of the most important applications of genomic study.

We have also given particular attention to quantitative genetics, population genetics, and evolutionary genetics (Chapters 25, 27, and 28). The coverage in these chapters has been extensively reviewed, and their revision is the product of the best thinking of many colleagues specialized in these fields.

- **Modernization of Topics**—Although we have updated each chapter in the text so that we report the most current and significant findings in genetics, we have especially focused on modernizing the discussions found in the chapters entitled *Cancer and the Regulation of the Cell Cycle* (Chapter 20), *Genomics, Bioinformatics, and Proteomics* (Chapter 21), and *Applications and Ethics of Genetic Engineering and Biotechnology* (Chapter 24). An in-depth consideration of *Conservation Genetics* (Chapter 29) continues to be another hallmark of our modern genetic coverage. This field, which attempts to assess and maintain genetic diversity in endangered species, remains at the forefront of genetic studies.
- **New/Revised Genetics, Technology, and Society Essays**—We have added several new essays that relate genetics to popular culture topics, and we have revised many that embody recent findings in genetics and their impact on society. The four new essays consider *Gene Silencing* (Chapter 14), *Targeted Cancer Therapies* (Chapter 20), *The Quest for the \$1000 Genome* (Chapter 21), and *Genetics of Sexual Orientation* (Chapter 26). Those essays that have been updated

and refined include Tay-Sachs Disease (Chapter 3), Purebred Dogs (Chapter 4), Fragile Sites and Cancer (Chapter 8), Telomerase and Aging (Chapter 11), Prions and Mad Cow Disease (Chapter 15), Gene Regulation and Human Disorders (Chapter 18), and Stem Cell Wars (Chapter 19).

These new or revised essays supplement those that discuss edible vaccines, human sex selection, genetically modified foods, gene therapy, and endangered species such as the Florida panther, among other topics.

- **New Illustrations**—The 9th edition includes many new figures and refines many of the existing figures in order to enhance their pedagogic value and artistic quality. Many figures feature “flow diagrams” that visually guide a student through experimental protocols and techniques.
- **Section Numbers**—All major sections of each chapter are numbered, making it easier to assign and locate topics within chapters.
- **Instructor and Student Media Address Real Needs**—Support for lecture presentations and other teaching responsibilities has been increased, including electronic access to more text photos and tables and a greater variety of PowerPoint offerings on the book’s Instructor Resource Center on CD/DVD. Media found on the revamped Companion Web Site reflect the growing awareness that today’s students must use their limited study time as wisely as possible.

Emphasis on Concepts

Concepts of Genetics, as its title implies, emphasizes the conceptual framework of genetics. Our experience with this book, reinforced by the many adopters with whom we have been in contact over the years, demonstrates quite conclusively that students whose primary focus is on concepts more easily comprehend and take with them to succeeding courses the most important ideas in genetics as well as an analytic view of biological problem solving.

To aid students in identifying the conceptual aspects of a major topic, each chapter begins with a section called *Chapter Concepts*, which outlines the most important ideas about to be presented. In the *Problems and Discussion Questions* section, the *How Do We Know?* feature asks the student to connect concepts to experiments. In addition, the *Now Solve This* feature asks students to link conceptual understanding to problem solving in a more immediate way. Each chapter ends with a *Chapter Summary*, which enumerates the five to ten key points that have been discussed. Collectively, these features help to ensure that students easily become aware of and understand the major conceptual issues as they confront the extensive vocabulary and the many important details of genetics. Carefully designed figures support this approach throughout the book.

Problem Solving and Insights and Solutions

To optimize the opportunities for student growth in the important areas of problem solving and analytical thinking, each chapter ends with an extensive collection of *Problems and Discussion Questions*. These include several levels of difficulty, with the most challenging (*Extra-Spicy Problems*) located at the end of each section. Brief answers to approximately half the problems are presented in Appendix B. The *Student Handbook and Solutions Manual* answers every problem and is available to students when faculty decide that it is appropriate. As the reader familiar with previous editions will see, about 75 new problems appear throughout the text.

As an aid to the student in learning to solve problems, the *Problems and Discussion Questions* section of each chapter is preceded by what has become an extremely popular and successful section called *Insights and Solutions*. This expanded section poses problems or questions and provides detailed solutions or answers. The questions and their solutions are designed to stress problem solving, quantitative analysis, analytical thinking, and experimental rationale. Collectively, these constitute the cornerstone of scientific inquiry and discovery. These feature primes students for moving on to the *Problems and Discussion Questions*.

The Genetics MediaLab section is available on the Companion Web Site. Each MediaLab contains several Web-linked problems designed to enhance and extend the topics presented in the chapter. To complete these problems, students must actively participate in the exercises and virtual experiments. For reference, the estimated time required to solve the problem is noted at the beginning of the exercise.

Acknowledgments

Contributors

We begin with special acknowledgments to those who have made direct contributions to this text. We particularly thank Sarah Ward at Colorado State University for creating Chapter 29 on Conservation Genetics and also for providing revised drafts of the chapters involving Quantitative and Population Genetics. We also thank David Kass of Eastern Michigan University, Chaoyang Zeng of the University of Wisconsin at Milwaukee, and Virginia McDonough of Hope College for their most useful input into both text-related topics and the revision of the Companion Web Site. In addition, Amanda Norvell revised several sections emphasizing eukaryotic molecular genetics, and Janet Morrison revised numerous aspects of our coverage of evolutionary genetics. Amanda and Janet are colleagues from The College of New Jersey. Katherine Uyhazi, now at Yale University Medical School, wrote the *Genetics, Technology, and Society* essay on Quorum Sensing in Bacteria (Chapter 17) and helped revise several other essays. David Kass also contributed the essay on Gene Regulation and Human Disorders (Chapter 18). Tamara Mans, currently teaching at North Hennepin Community College, wrote the essay on the Genetics of Sexual Orientation (Chapter 26). She also helped revise numerous essays. Mark Shotwell at Slippery Rock University contributed several essays. As with previous editions, Elliott Goldstein from Arizona State University was always readily

available to consult with us concerning the most modern findings in molecular genetics. We also express special thanks to Harry Nickla, recently retired from Creighton University. In his role as author of the *Student Handbook and Solutions Manual* and the *Instructor's Resource Manual with Tests*, he has reviewed and edited the problems at the end of each chapter, and has written many of the new entries as well. He also provided the brief answers to selected problems that appear in Appendix B.

We are grateful to all of these contributors not only for sharing their genetic expertise, but for their dedication to this project as well as the pleasant interactions they provided.

Proofreaders and Accuracy Checking

Proofreading the manuscript of an 800+ page text deserves more thanks than words can offer. Our utmost appreciation is extended to the three individuals who confronted this task with patience, diligence, and good humor:

Tamara Horton Mans, *North Hennepin Community College*
Sudhir Nayak, *The College of New Jersey*
Michael Rossa, *Proofreader*

Reviewers

All comprehensive texts are dependent on the valuable input provided by many reviewers. While we take full responsibility for any errors in this book, we gratefully acknowledge the help provided by those individuals who reviewed the content and pedagogy of this and the previous edition:

Robert A. Angus, *University of Alabama, Birmingham*
Peta Bonham-Smith, *University of Saskatchewan*
Alan H. Christensen, *George Mason University*
Bert Ely, *University of South Carolina*
Elliott S. Goldstein, *Arizona State University*
Edward M. Golenberg, *Wayne State University*
Ashley Hagler, *University of North Carolina, Charlotte*
Jocelyn Krebs, *University of Alaska, Fairbanks*
Traci Lee, *University of Wisconsin, Parkside*
Paul F. Lurquin, *Washington State University*
Virginia McDonough, *Hope College*
Kim McKim, *Rutgers University*
Clint Magill, *Texas A&M University*
Harry Nickla, *Creighton University*
Mohamed Noor, *Duke University*
John C. Osterman, *University of Nebraska—Lincoln*
Gloria Regisford, *Prairie View A&M University*
Rodney Scott, *Wheaton College*
Barkur Shastri, *Oakland University*
Fang-sheng Wu, *Virginia Commonwealth University*
Chaoyang Zeng, *University of Wisconsin, Milwaukee*

Special thanks go to Mike Guidry of LightCone Interactive and Karen Hughes of the University of Tennessee for their original contributions to the media program.

As these acknowledgments make clear, a text such as this is a collective enterprise. All of the above individuals deserve to share in any success this text enjoys. We want them to know that our gratitude is equaled only by the extreme dedication evident in their efforts. Many, many thanks to them all.

Editorial and Production Input

At Benjamin Cummings, we express appreciation and high praise for the editorial guidance and seminal input of Gary Carlson, whose ideas and efforts have helped to shape and refine the features of this and the previous editions of the text. In addition, our editorial team—Deborah Gale, Executive Director of Development, Leata Holloway, Project Editor, and our Media Producer, Laura Tomassi—has provided valuable input into the current edition. They have worked tirelessly to ensure that the pedagogy and design of the book and media package are at the cutting edge of a rapidly changing discipline. We were most fortunate to benefit from superb developmental editing provided by Moira Nelson, who proved to us that you are never too old to learn how to write more clearly, and outstanding copyediting performed by Betty Pessagno, for which we are most grateful. We also appreciate the production efforts of Lori Newman and those at Preparé Inc., whose quest for perfection is reflected throughout the text. In particular, Rosaria Cassinese provided an essential measure of sanity to the otherwise chaotic process of production. Without their work ethic and dedication, the text would never have come to fruition. Lauren Harp has professionally and enthusiastically managed the marketing of the text. Kaci Smith, Editorial Assistant, has worked efficiently to provide assistance to the editorial staff. Finally, the beauty and consistent presentation of the art work is the product of Imagineering of Toronto. We particularly thank Victor Ayers for his efforts.

For the Student

Companion Web Site—www.geneticsplace.com

Respect for the students' increasingly valuable study time is evident in the features of the Companion Web Site, which have been designed to enable users of the 9th edition to focus on those chapter sections and topics where they need review or further explanation. The Online Study Guide provides students with a focused, section-by-section review of topic coverage that features concise summary points accompanied by key illustrations and probing review questions that offer hints and feedback. The Web Tutorials offer today's learners the opportunity to quickly and conveniently visualize complex topics and dynamic processes—or to simply re-familiarize themselves with concepts they may have learned earlier but are encountering for the first time in the context of a genetics course. The media's strict adherence to both the principles and specific lessons of the textbook means that students and instructors can be assured that study time is not being squandered on media that confuse students and emphasize extraneous topics. The media tab on the outside margin of this page appears throughout

the book to indicate when there is a Web Tutorial on a topic related to the coverage in the book.

All *Exploring Genomics* exercises also are available at the Companion Web Site, along with answers for each exercise. Although we have presented high-quality Web resources for *Exploring Genomics*, on occasion site addresses and navigation details may change that will affect instructions for an exercise. We encourage you to refer to *Exploring Genomics* at the Companion Web Site for the most up-to-date versions of these exercises. Another advantage of this approach is that for many exercises you can cut and paste nucleotide or amino acid sequence data to be analyzed rather than type long sequences from the exercise in the text into a Web site.

In addition, a Media Lab for each chapter is offered on the Companion Web Site for those who want to explore genetics beyond the boundaries of a book through the vast array of genetics-related resources available through the Web.

Student Handbook and Solutions Manual

Authored by Harry Nickla, Creighton University (Emeritus)
(0321544609)

This valuable handbook provides a detailed step-by-step solution or lengthy discussion for every problem in the text. The handbook also features additional study aids, including extra study problems, chapter outlines, vocabulary exercises, and an overview of how to study genetics.

For the Instructor

Instructor Resource Center on CD/DVD

(0321544633)

The Instructor Resource Center on CD/DVD for the 9th edition offers adopters of the text convenient access to the most comprehensive and innovative set of lecture presentation and teaching tools offered by any genetics textbook. Developed to meet the needs of veteran and newer instructors alike, these resources include:

- The JPEG files of all text line drawings with labels individually enhanced for optimal projection results (as well as unlabeled versions) and all text tables.
- Most of the text photos, including all photos with pedagogical significance, as JPEG files.
- The JPEG files of line drawings, photos, and tables preloaded into comprehensive PowerPoint® presentations for each chapter.

- A second set of PowerPoint® presentations consisting of a thorough lecture outline for each chapter augmented by key text illustrations.
- An impressive series of concise instructor animations adding depth and visual clarity to the most important topics and dynamic processes described in the text.
- The instructor animations preloaded into PowerPoint® presentation files for each chapter.
- PowerPoint® presentations containing a comprehensive set of in-class Classroom Response System (CRS) questions for each chapter.
- In Word files, a complete set of the assessment materials and study questions and answers from the testbank, the text's in-chapter text questions, and the student media practice questions, as well as files containing the entire *Instructor's Manual and Solutions Manual*.
- Finally, to help instructors keep track of all that is available in this media package, a printable Media Integration Guide in PDF format that lists each chapter's media offerings.

Instructor's Resource Manual with Tests

(0321548485)

This manual and testbank contains over 1000 questions and problems for use in preparing exams. The manual also provides optional course sequences, a guide to audiovisual supplements, and a section on searching the Web. The testbank portion of the manual is also available in electronic format.

TestGen EQ Computerized Testing Software

(0321550447)

In addition to the printed volume, the test questions are also available as part of the TestGen EQ Testing Software, a text-specific testing program that is networkable for administering tests. It also allows instructors to view and edit questions, export the questions as tests, and print them out in a variety of formats.

Transparencies

(0321544617)

The transparency package includes 275 figures from the text: 225 four-color transparencies from the text plus 50 transparency masters. The font size of the labels has been increased and boldfaced for easy viewing from the back of the classroom.

Brief Contents

PART ONE GENES, CHROMOSOMES, AND HEREDITY

- 1 Introduction to Genetics 1
- 2 Mitosis and Meiosis 18
- 3 Mendelian Genetics 42
- 4 Extensions of Mendelian Genetics 70
- 5 Chromosome Mapping in Eukaryotes 105
- 6 Genetic Analysis and Mapping in Bacteria and Bacteriophages 143
- 7 Sex Determination and Sex Chromosomes 173
- 8 Chromosome Mutations: Variation in Chromosome Number and Arrangement 198
- 9 Extranuclear Inheritance 227

PART TWO DNA: STRUCTURE, REPLICATION, AND VARIATION

- 10 DNA Structure and Analysis 245
- 11 DNA Replication and Recombination 278
- 12 DNA Organization in Chromosomes 302
- 13 Recombinant DNA Technology and Gene Cloning 322

PART THREE GENE EXPRESSION, REGULATION, AND DEVELOPMENT

- 14 The Genetic Code and Transcription 352
- 15 Translation and Proteins 381
- 16 Gene Mutation and DNA Repair 410
- 17 Regulation of Gene Expression in Prokaryotes 435
- 18 Regulation of Gene Expression in Eukaryotes 457
- 19 Developmental Genetics of Model Organisms 484
- 20 Cancer and Regulation of the Cell Cycle 511

PART FOUR GENOMICS

- 21 Genomics, Bioinformatics, and Proteomics 531
- 22 Genome Dynamics: Transposons, Immunogenetics, and Eukaryotic Viruses 574
- 23 Genomic Analysis—Dissection of Gene Function 605
- 24 Applications and Ethics of Genetic Engineering and Biotechnology 633

PART FIVE GENETICS OF ORGANISMS AND POPULATION

- 25 Quantitative Genetics and Multifactorial Traits 668
- 26 Genetics and Behavior 688
- 27 Population Genetics 710
- 28 Evolutionary Genetics 737
- 29 Conservation Genetics 762

Appendix A Glossary A-1

Appendix B Answers to Selected Problems A-18

Appendix C Selected Readings A-57

Credits C-1

Index I-1

Contents

Preface

PART ONE GENES, CHROMOSOMES, AND HEREDITY

1 Introduction to Genetics 1

1.1 Genetics Progressed from Mendel to DNA in Less Than a Century 2

Mendel's Work on Transmission of Traits 2

The Chromosome Theory of Inheritance:
Uniting Mendel and Meiosis 3

Genetic Variation 4

The Search for the Chemical Nature of Genes: DNA or Protein? 5

1.2 Discovery of the Double Helix Launched the Era of Molecular Genetics 5

The Structure of DNA and RNA 5

Gene Expression: From DNA to Phenotype 5

Proteins and Biological Function 6

Linking Genotype to Phenotype: Sickle-Cell Anemia 7

1.3 Development of Recombinant DNA Technology Began the Era of Cloning 8

1.4 The Impact of Biotechnology Is Continually Expanding 8

Plants, Animals, and the Food Supply 9

Who Owns Transgenic Organisms? 9

Biotechnology in Genetics and Medicine 10

1.5 Genomics, Proteomics, and Bioinformatics Are New and Expanding Fields 10

1.6 Genetic Studies Rely on the Use of Model Organisms 12

The Modern Set of Genetic Model Organisms 12

Model Organisms and Human Diseases 13

1.7 We Live in the Age of Genetics 14

The Nobel Prize and Genetics 14

Genetics and Society 15

GENETICS, TECHNOLOGY, AND SOCIETY

Genetics and Society: The Application and Impact of Science and Technology 15

EXPLORING GENOMICS

Internet Resources for Learning about the Genomes of Model Organisms 16

Chapter Summary 17

Problems and Discussion Questions 17

2 Mitosis and Meiosis 18

2.1 Cell Structure Is Closely Tied to Genetic Function 19

2.2 Chromosomes Exist in Homologous Pairs in Diploid Organisms 21

2.3 Mitosis Partitions Chromosomes into Dividing Cells 23

Interphase and the Cell Cycle 24

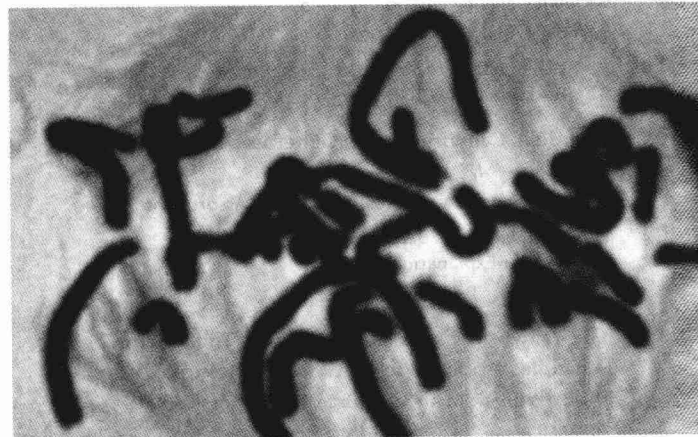
Prophase 24

Prometaphase and Metaphase 25

Anaphase 25

Telophase 26

Cell-Cycle Regulation and Checkpoints 27



- 2.4 Meiosis Reduces the Chromosome Number from Diploid to Haploid in Germ Cells and Spores** 28
 An Overview of Meiosis 28
 The First Meiotic Division: Prophase I 28
 Metaphase, Anaphase, and Telophase I 31
 The Second Meiotic Division 31
- 2.5 The Development of Gametes Varies in Spermatogenesis Compared to Oogenesis** 31
- 2.6 Meiosis Is Critical to the Successful Sexual Reproduction of All Diploid Organisms** 32
- 2.7 Electron Microscopy Has Revealed the Physical Structure of Mitotic and Meiotic Chromosomes** 34
 The Synaptonemal Complex 36
- GENETICS, TECHNOLOGY, AND SOCIETY**
 Breast Cancer: The Double-Edged Sword of Genetic Testing 37
- EXPLORING GENOMICS**
 PubMed: Exploring and Retrieving Biomedical Literature 38
- Chapter Summary 38
 Insights and Solutions 39
 Problems and Discussion Questions 40
 Extra-Spicy Problems 41

3

Mendelian Genetics 42

- 3.1 Mendel Used a Model Experimental Approach to Study Patterns of Inheritance** 43
- 3.2 The Monohybrid Cross Reveals How One Trait Is Transmitted from Generation to Generation** 43
 Mendel's First Three Postulates 45
 Modern Genetic Terminology 45
 Mendel's Analytical Approach 45
 Punnett Squares 46
 The Testcross: One Character 46
- 3.3 Mendel's Dihybrid Cross Generated a Unique F₂ Ratio** 47
 Mendel's Fourth Postulate: Independent Assortment 47
- How Mendel's Peas Become Wrinkled: A Molecular Explanation 48
 The Testcross: Two Characters 49
- 3.4 The Trihybrid Cross Demonstrates That Mendel's Principles Apply to Inheritance of Multiple Traits** 49
 The Forked-Line Method, or Branch Diagram 50
- 3.5 Mendel's Work Was Rediscovered in the Early Twentieth Century** 52

- 3.6 The Correlation of Mendel's Postulates with the Behavior of Chromosomes Provided the Foundation of Modern Transmission Genetics** 52
 The Chromosomal Theory of Inheritance 52
 Unit Factors, Genes, and Homologous Chromosomes 52
- 3.7 Independent Assortment Leads to Extensive Genetic Variation** 54
- 3.8 Laws of Probability Help to Explain Genetic Events** 54
 Conditional Probability 55
 The Binomial Theorem 55
- 3.9 Chi-Square Analysis Evaluates the Influence of Chance on Genetic Data** 56
 Chi-Square Calculations and the Null Hypothesis 57
 Interpreting Probability Values 58
- 3.10 Pedigrees Reveal Patterns of Inheritance of Human Traits** 59
 Pedigree Conventions 59
 Pedigree Analysis 60

GENETICS, TECHNOLOGY, AND SOCIETY

Tay-Sachs Disease: The Molecular Basis of a Recessive Disorder in Humans 61

EXPLORING GENOMICS

Online Mendelian Inheritance in Man 62

- Chapter Summary 63
 Insights and Solutions 63
 Problems and Discussion Questions 66
 Extra-Spicy Problems 68

4

Extensions of Mendelian Genetics 70

- 4.1 Alleles Alter Phenotypes in Different Ways** 71
- 4.2 Geneticists Use a Variety of Symbols for Alleles** 72
- 4.3 Neither Allele Is Dominant in Incomplete, or Partial, Dominance** 72
- 4.4 In Codominance, the Influence of Both Alleles in a Heterozygote Is Clearly Evident** 73
- 4.5 Multiple Alleles of a Gene May Exist in a Population** 74
 The ABO Blood Groups 74
 The A and B Antigens 75
 The Bombay Phenotype 76
 The *white* Locus in *Drosophila* 76



4.6	Lethal Alleles Represent Essential Genes	77
	Recessive Lethal Mutations	77
	Dominant Lethal Mutations	78
4.7	Combinations of Two Gene Pairs with Two Modes of Inheritance Modify the 9:3:3:1 Ratio	78
4.8	Phenotypes Are Often Affected by More Than One Gene	79
	Epistasis	79
	Novel Phenotypes	82
	Other Modified Dihybrid Ratios	84
4.9	Complementation Analysis Can Determine If Two Mutations Causing a Similar Phenotype Are Alleles	84
4.10	Expression of a Single Gene May Have Multiple Effects	84
4.11	X-Linkage Describes Genes on the X Chromosome	85
	X-Linkage in <i>Drosophila</i>	86
	X-Linkage in Humans	86
■	Lesch-Nyhan Syndrome: The Molecular Basis of a Rare X-Linked Recessive Disorder	88
4.12	In Sex-Limited and Sex-Influenced Inheritance, an Individual's Sex Influences the Phenotype	89
4.13	Genetic Background and the Environment May Alter Phenotypic Expression	90
	Penetrance and Expressivity	90
	Genetic Background: Suppression and Position Effects	91
	Temperature Effects—An Introduction to Conditional Mutations	91
	Nutritional Effects	92
	Onset of Genetic Expression	92
	Genetic Anticipation	93
	Genomic (Parental) Imprinting	93
■	GENETICS, TECHNOLOGY, AND SOCIETY	
	Improving the Genetic Fate of Purebred Dogs	94
■	EXPLORING GENOMICS	
	The Human Epigenome Project	95
	Chapter Summary	96

Insights and Solutions	97
Problems and Discussion Questions	98
Extra-Spicy Problems	102

5 Chromosome Mapping in Eukaryotes 105

5.1	Genes Linked on the Same Chromosome Segregate Together	106
	The Linkage Ratio	107
5.2	Crossing Over Serves as the Basis for Determining the Distance between Genes in Chromosome Mapping	109
	Morgan and Crossing Over	109
	Sturtevant and Mapping	109
	Single Crossovers	111
5.3	Determining the Gene Sequence during Mapping Requires the Analysis of Multiple Crossovers	112
	Multiple Exchanges	112
	Three-Point Mapping in <i>Drosophila</i>	113
	Determining the Gene Sequence	115
	A Mapping Problem in Maize	116
5.4	Interference Affects the Recovery of Multiple Exchanges	119
5.5	As the Distance between Two Genes Increases, the Results of Mapping Experiments Become Less Accurate	120
5.6	<i>Drosophila</i> Genes Have Been Extensively Mapped	121
5.7	Lod Score Analysis and Somatic Cell Hybridization Were Historically Important in Creating Human Chromosome Maps	121
5.8	Chromosome Mapping Is Now Possible Using DNA Markers and Annotated Computer Databases	124
5.9	Crossing Over Involves a Physical Exchange between Chromatids	125
5.10	Recombination Occurs between Mitotic Chromosomes	125
5.11	Exchanges Also Occur between Sister Chromatids	126
5.12	Linkage and Mapping Studies Can Be Performed in Haploid Organisms	127
	Gene-to-Centromere Mapping	129
	Ordered versus Unordered Tetrad Analysis	130
	Linkage and Mapping	130

- 5.13 Did Mendel Encounter Linkage?** 133
 Why Didn't Gregor Mendel Find Linkage? 133

EXPLORING GENOMICS

Human Chromosome Maps on the Internet 134

Chapter Summary 135

Insights and Solutions 135

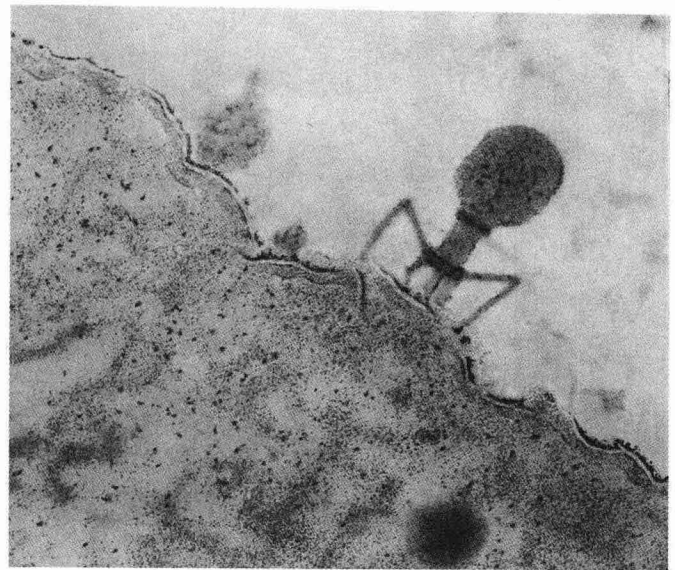
Problems and Discussion Questions 137

Extra-Spicy Problems 141

6

Genetic Analysis and Mapping in Bacteria and Bacteriophages 143

- 6.1 Bacteria Mutate Spontaneously and Grow at an Exponential Rate** 144
- 6.2 Conjugation Is One Means of Genetic Recombination in Bacteria** 145
F⁺ and *F*⁻ Bacteria 146
 Hfr Bacteria and Chromosome Mapping 147
 Recombination in *F*⁺ × *F*⁻ Matings: A Reexamination 151
 The *F*['] State and Merozygotes 151
- 6.3 Rec Proteins Are Essential to Bacterial Recombination** 151
- 6.4 The F Factor Is an Example of a Plasmid** 153
- 6.5 Transformation Is Another Process Leading to Genetic Recombination in Bacteria** 153
 The Transformation Process 154
 Transformation and Linked Genes 155
- 6.6 Bacteriophages Are Bacterial Viruses** 155
 Phage T4: Structure and Life Cycle 155
 The Plaque Assay 156
 Lysogeny 157
- 6.7 Transduction Is Virus-Mediated Bacterial DNA Transfer** 158
 The Lederberg-Zinder Experiment 158
 The Nature of Transduction 158
 Transduction and Mapping 160
- 6.8 Bacteriophages Undergo Intergenic Recombination** 160
 Bacteriophage Mutations 160
 Mapping in Bacteriophages 161
- 6.9 Intragenic Recombination Occurs in Phage T4** 161
 The *rII* Locus of Phage T4 162
 Complementation by *rII* Mutations 162



- Recombinational Analysis 163
 Deletion Testing of the *rII* Locus 163
 The *rII* Gene Map 164

GENETICS, TECHNOLOGY, AND SOCIETY

Bacterial Genes and Disease: From Gene Expression to Edible Vaccines 166

EXPLORING GENOMICS

Microbial Genome Program (MGP) 167

Chapter Summary 168

Insights and Solutions 168

Problems and Discussion Questions 169

Extra-Spicy Problems 171

7

Sex Determination and Sex Chromosomes 173

- 7.1 Life Cycles Depend on Sexual Differentiation** 174
Chlamydomonas 174
Zea mays 175
Caenorhabditis elegans 176
- 7.2 X and Y Chromosomes Were First Linked to Sex Determination Early in the Twentieth Century** 177
- 7.3 The Y Chromosome Determines Maleness in Humans** 178
 Klinefelter and Turner Syndromes 178
 47,XXX Syndrome 180
 47,XXY Condition 180
 Sexual Differentiation in Humans 181
 The Y Chromosome and Male Development 182

- 7.4 The Ratio of Males to Females in Humans Is Not 1.0** 183
- 7.5 Dosage Compensation Prevents Excessive Expression of X-Linked Genes in Humans and Other Mammals** 184
Barr Bodies 184
The Lyon Hypothesis 185
The Mechanism of Inactivation 186
- 7.6 The Ratio of X Chromosomes to Sets of Autosomes Determines Sex in *Drosophila*** 187
Dosage Compensation in *Drosophila* 189
Drosophila Mosaics 190
- 7.7 Temperature Variation Controls Sex Determination in Reptiles** 190

GENETICS, TECHNOLOGY, AND SOCIETY

A Question of Gender: Sex Selection in Humans 192

EXPLORING GENOMICS

The Ovarian Kaleidoscope Database (OKDB) 193

- Chapter Summary 194
Insights and Solutions 194
Problems and Discussion Questions 194
Extra-Spicy Problems 195

8

Chromosome Mutations: Variation in Chromosome Number and Arrangement 198

- 8.1 Specific Terminology Describes Variations in Chromosome Number** 199
Variation in the Number of Chromosomes Results from Nondisjunction 199
- 8.2 Monosomy, the Loss of a Single Chromosome, May Have Severe Phenotypic Effects** 200
- 8.3 Trisomy Involves the Addition of a Chromosome to a Diploid Genome** 200
Down Syndrome 201
Patau Syndrome 203
Edwards Syndrome 204
Viability in Human Aneuploidy 204
- 8.4 Polyploidy, in Which More Than Two Haploid Sets of Chromosomes Are Present, Is Prevalent in Plants** 205
Autopolyploidy 205
Allopolyploidy 206
Endopolyploidy 208

- 8.5 Variation Occurs in the Internal Composition and Arrangement of Chromosomes** 208
- 8.6 A Deletion Is a Missing Region of a Chromosome** 209
Cri du Chat Syndrome in Humans 210
Drosophila Heterozygous for Deficiencies May Exhibit Pseudodominance 210
- 8.7 A Duplication Is a Repeated Segment of the Genetic Material** 211
Gene Redundancy and Amplification: Ribosomal RNA Genes 211
The *Bar* Mutation in *Drosophila* 212
The Role of Gene Duplication in Evolution 213
- **Copy Number Variants (CNVs)—Duplications and Deletions of Specific DNA Sequences** 214
- 8.8 Inversions Rearrange the Linear Gene Sequence** 214
Consequences of Inversions during Gamete Formation 215
Position Effects of Inversions 216
Evolutionary Advantages of Inversions 217
- 8.9 Translocations Alter the Location of Chromosomal Segments in the Genome** 217
Translocations in Humans: Familial Down Syndrome 218
- 8.10 Fragile Sites in Humans Are Susceptible to Chromosome Breakage** 218
Fragile X Syndrome (Martin-Bell Syndrome) 219

GENETICS, TECHNOLOGY, AND SOCIETY

The Link between Fragile Sites and Cancer 220

EXPLORING GENOMICS

Atlas of Genetics and Cytogenetics in Oncology and Haematology 221

- Chapter Summary 222
Insights and Solutions 223
Problems and Discussion Questions 224
Extra-Spicy Problems 225

9

Extranuclear Inheritance 227

- 9.1 Organelle Heredity Involves DNA in Chloroplasts and Mitochondria** 228
Chloroplasts: Variegation in Four O'Clock Plants 228
Chloroplast Mutations in *Chlamydomonas* 228
Mitochondrial Mutations: The Case of *poky* in *Neurospora* 229
Petites in *Saccharomyces* 230
- 9.2 Knowledge of Mitochondrial and Chloroplast DNA Helps Explain Organelle Heredity** 231
Organelle DNA and the Endosymbiotic Theory 231
Molecular Organization and Gene Products of Chloroplast DNA 232

Molecular Organization and Gene Products
of Mitochondrial DNA 233

- 9.3 Mutations in Mitochondrial DNA Cause Human Disorders 234
- 9.4 Infectious Heredity Is Based on a Symbiotic Relationship between Host Organism and Invader 236
 - Kappa in *Paramecium* 236
 - Infective Particles in *Drosophila* 236
- 9.5 In Maternal Effect, the Maternal Genotype Has a Strong Influence during Early Development 237
 - Ephestia* Pigmentation 237
 - Limnaea* Coiling 238
 - Embryonic Development in *Drosophila* 239

GENETICS, TECHNOLOGY, AND SOCIETY
Mitochondrial DNA and the Mystery of the Romanovs 239

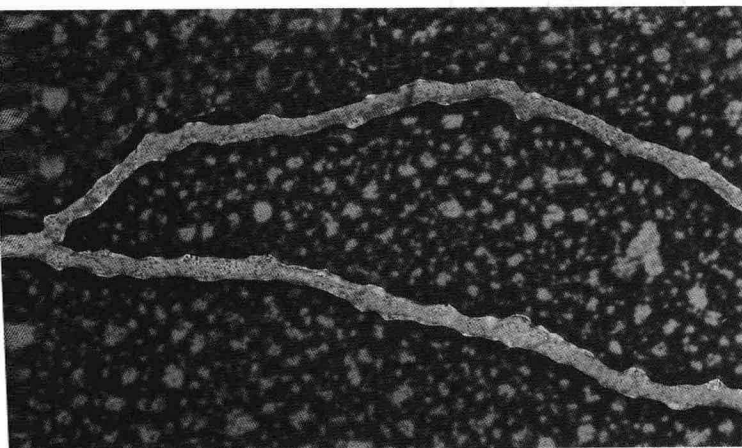
EXPLORING GENOMICS
Mitochondrial Genes and Mitomap 240

- Chapter Summary 241
- Insights and Solutions 242
- Problems and Discussion Questions 242
- Extra-Spicy Problems 243

PART TWO
DNA: STRUCTURE, REPLICATION, AND VARIATION

10
DNA Structure and Analysis 245

- 10.1 The Genetic Material Must Exhibit Four Characteristics 246



- 10.2 Until 1944, Observations Favored Protein as the Genetic Material 247
- 10.3 Evidence Favoring DNA as the Genetic Material Was First Obtained during the Study of Bacteria and Bacteriophages 247
 - Transformation: Early Studies 247
 - Transformation: The Avery, MacLeod, and McCarty Experiment 249
 - The Hershey-Chase Experiment 250
 - Transfection Experiments 251
- 10.4 Indirect and Direct Evidence Supports the Concept that DNA Is the Genetic Material in Eukaryotes 253
 - Indirect Evidence: Distribution of DNA 253
 - Indirect Evidence: Mutagenesis 253
 - Direct Evidence: Recombinant DNA Studies 254
- 10.5 RNA Serves as the Genetic Material in Some Viruses 254
- 10.6 Knowledge of Nucleic Acid Chemistry Is Essential to the Understanding of DNA Structure 255
 - Nucleotides: Building Blocks of Nucleic Acids 255
 - Nucleoside Diphosphates and Triphosphates 256
 - Polynucleotides 256
- 10.7 The Structure of DNA Holds the Key to Understanding Its Function 257
 - Base-Composition Studies 258
 - X-Ray Diffraction Analysis 259
 - The Watson-Crick Model 259
- 10.8 Alternative Forms of DNA Exist 262
- 10.9 The Structure of RNA Is Chemically Similar to DNA, but Single Stranded 263
- 10.10 Many Analytical Techniques Have Been Useful during the Investigation of DNA and RNA 264
 - Absorption of Ultraviolet Light 264
 - Sedimentation Behavior 264
 - Denaturation and Renaturation of Nucleic Acids 266
 - Molecular Hybridization 267
 - Fluorescent *in situ* Hybridization (FISH) 268
 - Reassociation Kinetics and Repetitive DNA 268
 - Electrophoresis of Nucleic Acids 270

GENETICS, TECHNOLOGY, AND SOCIETY
The Twists and Turns of the Helical Revolution 271

EXPLORING GENOMICS
Introduction to Bioinformatics: BLAST 272

- Chapter Summary 273
- Insights and Solutions 274
- Problems and Discussion Questions 275
- Extra-Spicy Problems 276