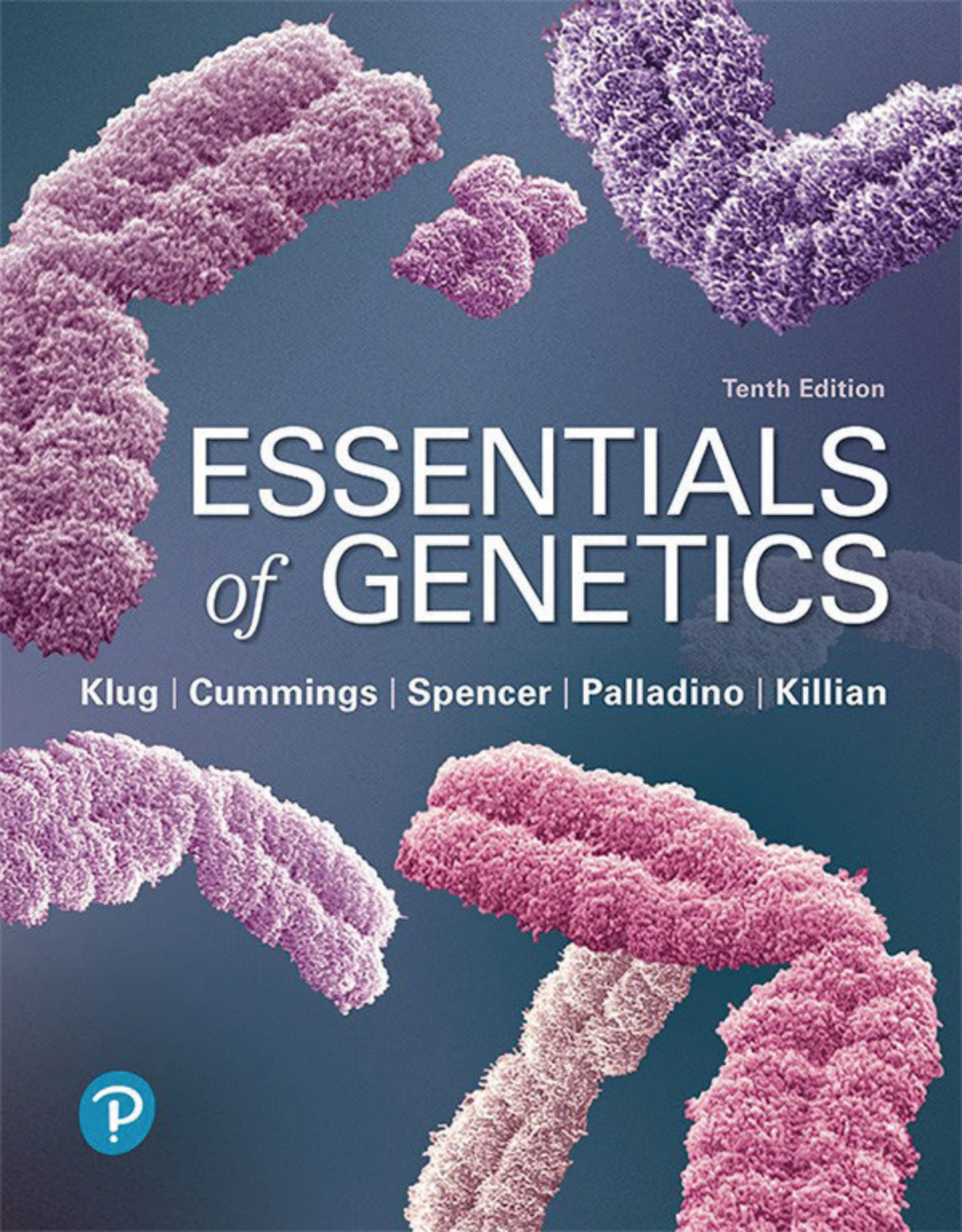
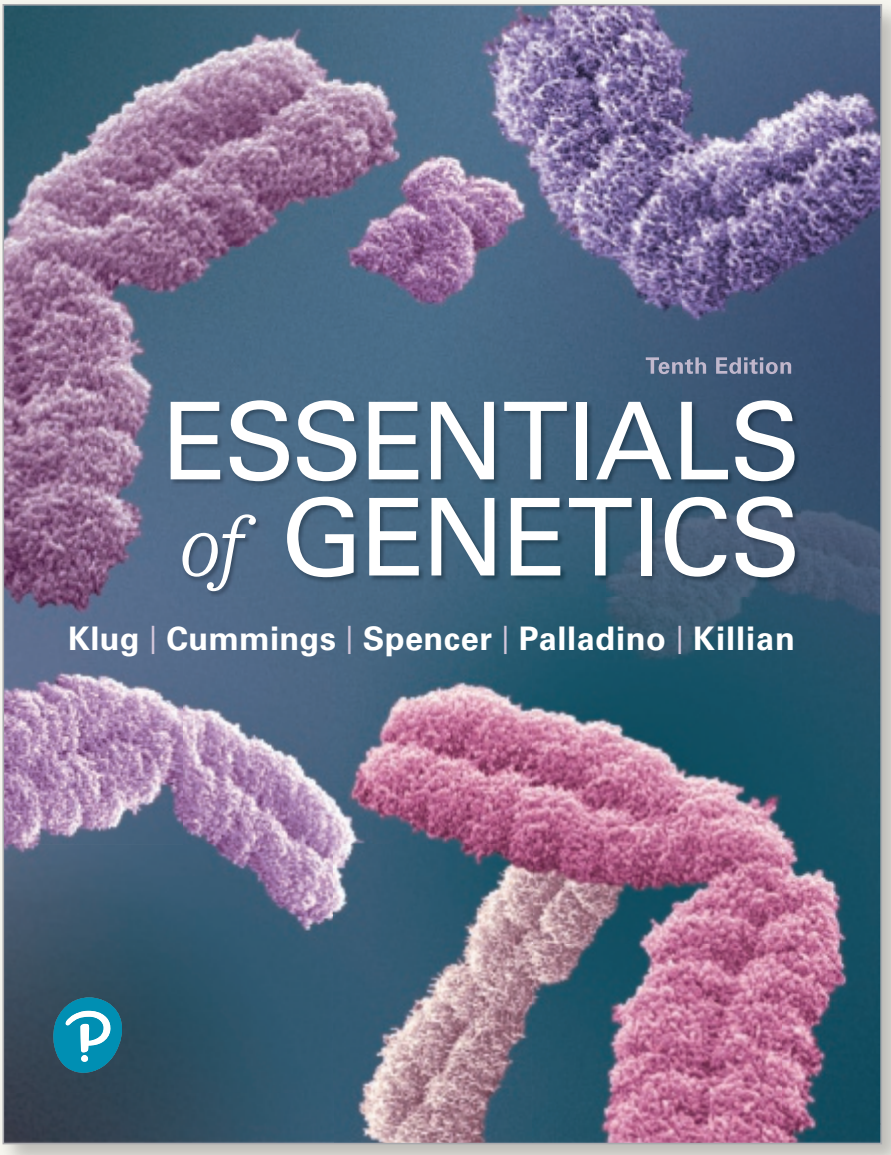

Tenth Edition
ESSENTIALS
of GENETICS
Klug | Cummings | Spencer | Palladino | Killian

Focus on essential genetic topics and explore the latest breakthroughs

Known for its focus on conceptual understanding, problem solving, and practical applications, the bestselling *Essentials of Genetics* strengthens problem-solving skills and explores the essential genetics topics that today's students need to understand. The 10th Edition has been extensively updated to provide comprehensive coverage of important, emerging topics such as CRISPR-Cas, epigenetics, and genetic testing. Mastering Genetics includes new tutorials on topics such as CRISPR-Cas and epigenetics, and new, mobile-ready Dynamic Study Modules, which prepare students for class and support the learning of key concepts.

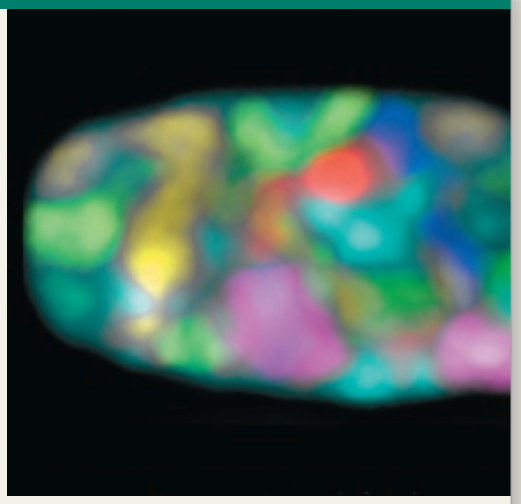


Pearson

Make genetics relevant . . .

16

Regulation of Gene Expression in Eukaryotes



Chromosome territories in a human fibroblast cell nucleus. Each chromosome is stained with a different-colored probe.

CHAPTER CONCEPTS

- While transcription and translation are tightly coupled in bacteria, in eukaryotes, these processes are spatially and temporally separated, and thus independently regulated.
- Chromatin remodeling, as well as modifications to DNA and histones, play important roles in regulating gene expression in eukaryotes.
- Eukaryotic transcription initiation requires the assembly of transcription regulatory proteins on DNA sites known as promoters, enhancers, and silencers.
- Following transcription, there are several mechanisms that regulate gene expression, referred to as posttranscriptional regulation.
- Alternative splicing allows for a single gene to encode different protein isoforms with different functions.
- RNA-binding proteins regulate mRNA stability, degradation, localization, and translation.
- Noncoding RNAs may regulate gene

Virtually all cells in a multicellular eukaryotic organism contain a complete genome; however, such organisms often possess different cell types with diverse morphologies and functions. This simple observation highlights the importance of the regulation of gene expression in eukaryotes. For example, skin cells and muscle cells differ in appearance and function because they express different genes. Skin cells express keratins, fibrous structural proteins that bestow the skin with protective properties. Muscle cells express high levels of myosin II, a protein that mediates muscle contraction. Skin cells do not express myosin II, and muscle cells do not express keratins.

In addition to gene expression that is cell-type specific, some genes are only expressed under certain conditions or at certain times. For example, when oxygen levels in the blood are low, such as at high altitude or after rigorous exercise, expression of the hormone erythropoietin is upregulated, which leads to an increase in red blood cell production and thus oxygen-carrying capacity.

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NEW! Regulation of gene expression has been expanded and is now divided into coverage of bacteria in Chapter 15 and coverage of eukaryotes in Chapter 16.

Coverage of CRISPR-Cas is expanded and integrated in multiple chapters – Chapters 1, 15, 17, and Special Topics Chapters ST3 and ST6.

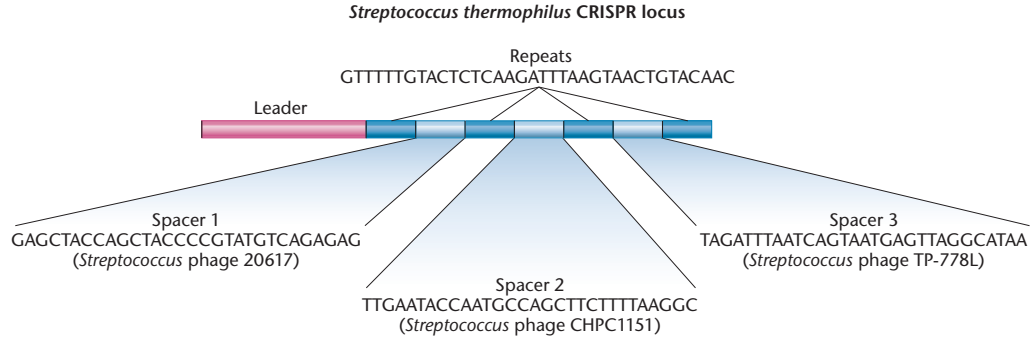

FIGURE 15.13 A CRISPR locus from the bacterium *Streptococcus thermophilus* (LMG18311). Spacer sequences are derived from portions of bacteriophage genomes and are flanked on either side by a repeat sequence. Only 3 of 33 total spacers in this CRISPR locus are shown.

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with current high interest topics

SPECIAL TOPICS IN MODERN GENETICS 2

Genetic Testing

SPECIAL TOPIC X

Earlier in the text (see Chapters 17 and 18), we reviewed essential concepts of recombinant DNA technology and genomic analysis. Because of the Human Genome Project and related advances in genomics, researchers have been making rapid progress in identifying genes involved in both single-gene diseases and complex genetic traits. As a result, **genetic testing**—the ability to analyze DNA, and increasingly RNA, for the purposes of identifying specific genes or sequences associated with different genetic conditions—has advanced very rapidly.

Genetic testing, including genomic analysis by DNA sequencing, is transforming medical diagnostics. Technologies for genetic testing have had major impacts on the diagnosis of disease and are revolutionizing medical treatments based on the development of specific and effective pharmaceuticals. In this Special Topics chapter we provide an overview of applications that are effective for the genetic testing of children and adults and examine historical and modern methods. We consider the impact of different genetic technologies on the diagnosis of human diseases and dis-

"Genetic testing, including genomic analysis by DNA sequencing, is transforming medical diagnostics. Technologies for genetic testing have had major

dystrophy. Other tests have been developed for disorders that may involve multiple genes such as certain types of cancers.

Gene tests are used for prenatal, childhood, and adult prognosis and diagnosis of genetic diseases; to identify carriers; and to identify genetic diseases in embryos created by *in vitro* fertilization, among other applications. For genetic testing of adults, DNA from white blood cells is commonly used. Alternatively, many genetic tests can be carried out on cheek cells, collected by swabbing the inside of the mouth, or on hair cells. Some genetic testing can be carried out on gametes.

What does it mean when a genetic test is performed for *prognostic* purposes, and how does this differ from a *diagnostic* test? A prognostic test predicts a person's likelihood of developing a particular genetic disorder. A diagnostic test for a genetic condition

NEW! Special Topics chapter on Genetic Testing guides students through the many contexts in which genetic testing is becoming prominent and explores many questions and ethical concerns related to its use.

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SPECIAL TOPICS IN MODERN GENETICS 4

Advances in Neurogenetics: The Study of Huntington Disease

As the result of groundbreaking advances in molecular genetics and genomics made since the 1970s, new fields in genetics and related disciplines have emerged. One new field is **neurogenetics**—the study of the genetic basis of normal and abnormal functioning of the nervous system, with emphasis on brain functions. Research in this field includes the genes associated with neurodegenerative disorders, with the ultimate goal of developing effective therapies to combat these devastating conditions. Of the many such diseases, including Alzheimer disease, Parkinson disease, and amyotrophic lateral sclerosis (ALS), **Huntington disease (HD)** stands out as a model for the genetic investigation of neurodegenerative disorders. Not only is it monogenic and 100 percent penetrant, but nearly all analytical approaches in molecular genetics have been successfully applied to the study of HD, validating its significance as a model for these diseases.

HD is an autosomal dominant disorder characterized by adult onset of defined and progressive behavioral changes, including uncontrolled movements (chorea), cognitive decline, and psychiatric disturbances, with death occurring within 10 to 15 years after symptoms appear. HD was one of the first examples of complete dominance in human inheritance, with no differences in phenotypes between homozygotes and heterozygotes. In the vast majority of cases, symptoms do not develop until about age 45. Overall, HD currently affects about 25,000 to 30,000 people in North America.

The disease is named after George Huntington, a nineteenth-century physician. He was not the first to describe the disorder,

"Driving with my father through a wooded road leading from Easthampton to Amagansett, we suddenly came upon two women, mother and daughter, both bowing, twisting, grimacing. I stared in wonderment, almost in fear. What could it mean?"

know about the molecular and cellular mechanisms associated with the disorder, particularly those discovered during the study of transgenic model systems. Finally, we will consider how this information is being used to develop a range of therapies.

ST 4.1 The Search for the Huntington Gene

Mapping the gene for Huntington disease was one of the first attempts to employ a method from a landmark 1980 paper by Botstein, White, and Davis in which the authors proposed that DNA sequence variations in humans could be detected as differences in the length of DNA fragments produced by cutting DNA with restriction enzymes. These differences, known as restriction fragment length polymorphisms (RFLPs), could be visualized using Southern blots (see Chapter 18 for a discussion of RFLPs, and Chapter 17 for a discussion of Southern blots). The authors estimated that a collection of about 150 RFLPs distributed across the genome could be used with pedigrees to detect linkage anywhere in the genome between an RFLP marker and a disease gene of interest. In practical terms, this meant that it would be possible to map a disease gene with no information about the gene, its gene product, or its function—an approach referred to as reverse genetics.

SPECIAL TOPIC 4

NEW! Special Topics chapter on Advances in Neurogenetics: The Study of Huntington Disease, explores how genetic analysis has informed scientists about the disease's causes, symptoms, and future treatment. All Special Topics chapters include a series of questions that help students review key ideas or facilitate personal contemplations and group discussions, and are assignable in Mastering Genetics.

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Explore the latest ethical considerations

GENETICS, ETHICS, AND SOCIETY

Down Syndrome and Prenatal Testing—The New Eugenics?

Down syndrome is the most common chromosomal abnormality seen in newborn babies. Prenatal diagnostic tests for Down syndrome have been available for decades, especially to older pregnant women who have an increased risk of bearing a child with Down syndrome. Scientists estimate that there is an abortion rate of about 30 percent for fetuses that test positive for Down syndrome in the United States, and rates of up to 85 percent in other parts of the world, such as Taiwan and France.

Some people agree that it is morally acceptable to prevent the birth of a genetically abnormal fetus. However, others argue that prenatal genetic testing, with the goal of eliminating congenital disorders, is unethical. In addition, some argue that prenatal genetic testing followed by selective abortion is eugenic. How does eugenics apply, if at all, to screening for Down syndrome and other human genetic disorders

The term *eugenics* was first defined by Francis Galton in 1883 as "the science which deals with all influences that improve the inborn qualities of a race; also with those that develop them to the utmost advantage." Galton believed that human traits such as intelligence and personality were hereditary and that humans could selectively mate with each other to create gifted groups of people—analogous to the creation of purebred dogs with specific traits. Galton did not propose coercion but thought that people would voluntarily select mates in order to enhance particular genetic outcomes for their offspring.

In the early to mid-twentieth century, countries throughout the world adopted eugenic policies with the aim of enhancing desirable human traits (positive eugenics) and eliminating undesirable ones (negative eugenics). Many countries, including Britain, Canada, and the United States, enacted compulsory sterilization programs for the "feebleminded," mentally ill, and criminals. The eugenic policies of Nazi Germany were particularly infamous, resulting in forced human genetic experimentation and the slaughter of tens of thousands of people with disabilities. The eugenics movement was discredited after World War II, and the evils perpetuated in its name have tainted the term *eugenics* ever since.

Given the history of the eugenics movement, is it fair to use the term *eugenics* when we speak about genetic testing for Down syndrome and other genetic disorders? Some people argue that it is not eugenic to select for healthy children because there is no coercion, the state is not involved, and the goal is the elimination of suffering. Others point out that such voluntary actions still constitute eugenics, since they involve a form of bioengineering for "better" human beings.

Now that we are entering an era of unprecedented knowledge about our genomes and our predisposition to genetic disorders, we must make decisions about whether our attempts to control or improve human genomes are ethical and what limits we should place on these efforts. The story of the eugenics movement provides us with a powerful cautionary tale about the potential misuses of genetic information.

Your Turn

Take time, individually or in groups, to consider the following questions. Investigate the references and links to help you discuss some of the ethical issues surrounding genetic testing and eugenics.

1. Do you think that modern prenatal and preimplantation genetic testing followed by selective abortion is eugenic? Why or why not?

For background on these questions, see McCabe, L., and McCabe, E. (2011). Down syndrome: Coercion and eugenics. *Genet. Med.* 13:708–710. *Another useful discussion can be found in* Wilkinson, S., (2015). Prenatal screening, reproductive choice, and public health. *Bioethics* 29:26–35.

2. If genetic technologies were more advanced than today, and you could choose the traits of your children, would you take advantage of that option? Which traits would you choose—height, weight, intellectual abilities, athleticism, artistic talents? If so, would this be eugenic? Would it be ethical?

To read about similar questions answered by groups of Swiss law and medical students, read Elger, B., and Harding, T., (2003). Huntington's disease: Do future physicians and lawyers think eugenically? *Clin. Genet.* 64:327–338.

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Genetics, Ethics, and Society essays provide synopses of ethical issues related to current findings in genetics that impact directly on society today. They include a section called *Your Turn*, which directs students to related resources of short readings and websites to support deeper investigation and discussion of the main topic of each essay.

Case Studies at the end of each chapter have been updated with new topics. Students can read and answer questions about a short scenario related to one of the chapter topics. Each Case Study links the coverage of formal genetic knowledge to everyday societal issues, and they include ethical considerations.

CASE STUDY To test or not to test

Thomas discovered a devastating piece of family history when he learned that his brother had been diagnosed with Huntington disease (HD) at age 49. This dominantly inherited autosomal condition usually begins around age 45 with progressive dementia, muscular rigidity, and seizures and ultimately leads to death when affected individuals are in their early 60s. There currently is no effective treatment or cure for this genetic disorder. Thomas, now 38, wonders what the chances are that he also has inherited the mutant allele for HD, leading him to discuss with his wife whether they should seek genetic counseling and whether he should undergo genetic testing. They have two teenage children, a boy and a girl.

1. If they seek genetic counseling, what issues would likely be discussed? Which of these pose grave ethical dilemmas?
2. If you were in Thomas's position, would you want to be tested and possibly learn that you were almost certain to develop the disorder sometime in the next 5–10 years?
3. If Thomas tests positive for the HD allele, should his children be told about the situation, and if so, at what age? Who should make the decision about having the son and daughter tested?

Fulda, K., and Lykens, K. (2006). Ethical issues in predictive genetic testing: A public health perspective. *J. Med. Ethics* 32:143–147.

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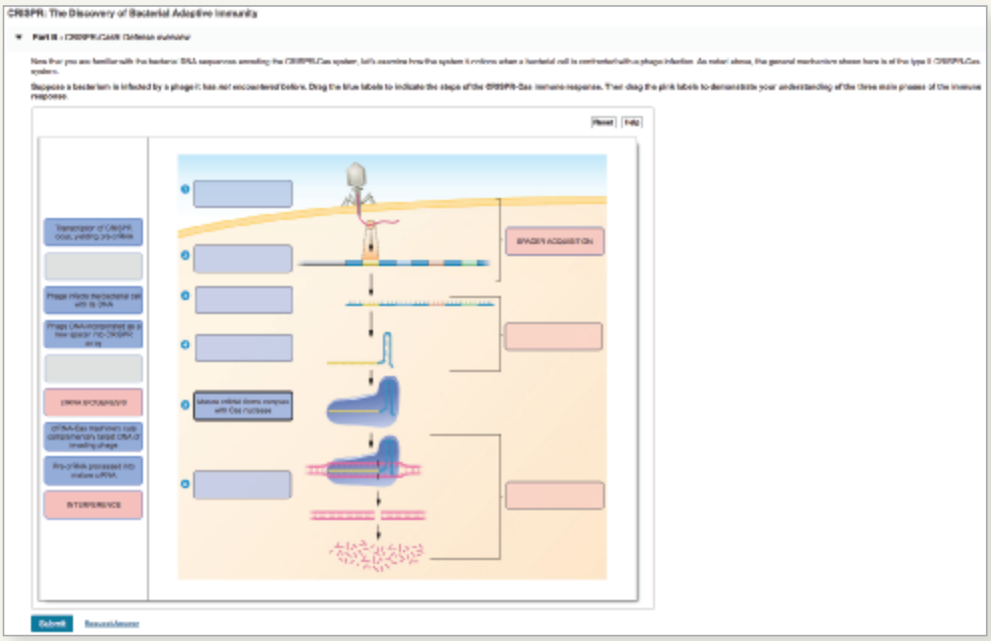
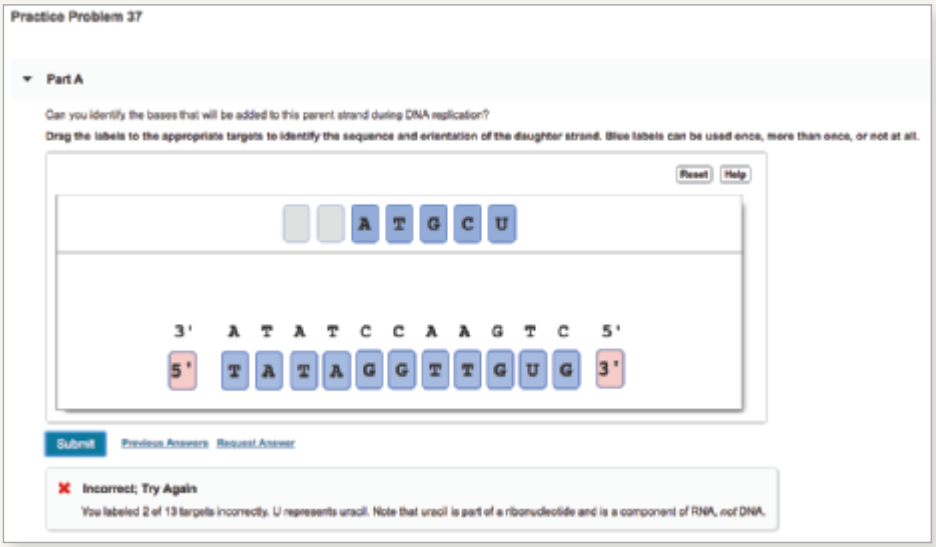
Learn genetics concepts and problem solving in Mastering Genetics



NEW! Tutorials have been added to the library on topics like CRISPR-Cas and epigenetics, to help students master important and challenging concepts.

A library of over 100 Practice Problems offers more opportunities to assign high quality problems for student homework or practice. These questions appear only in Mastering Genetics and include targeted wrong-answer feedback to help students learn from their mistakes. They are similar to end-of-chapter questions in terms of topic coverage and difficulty.



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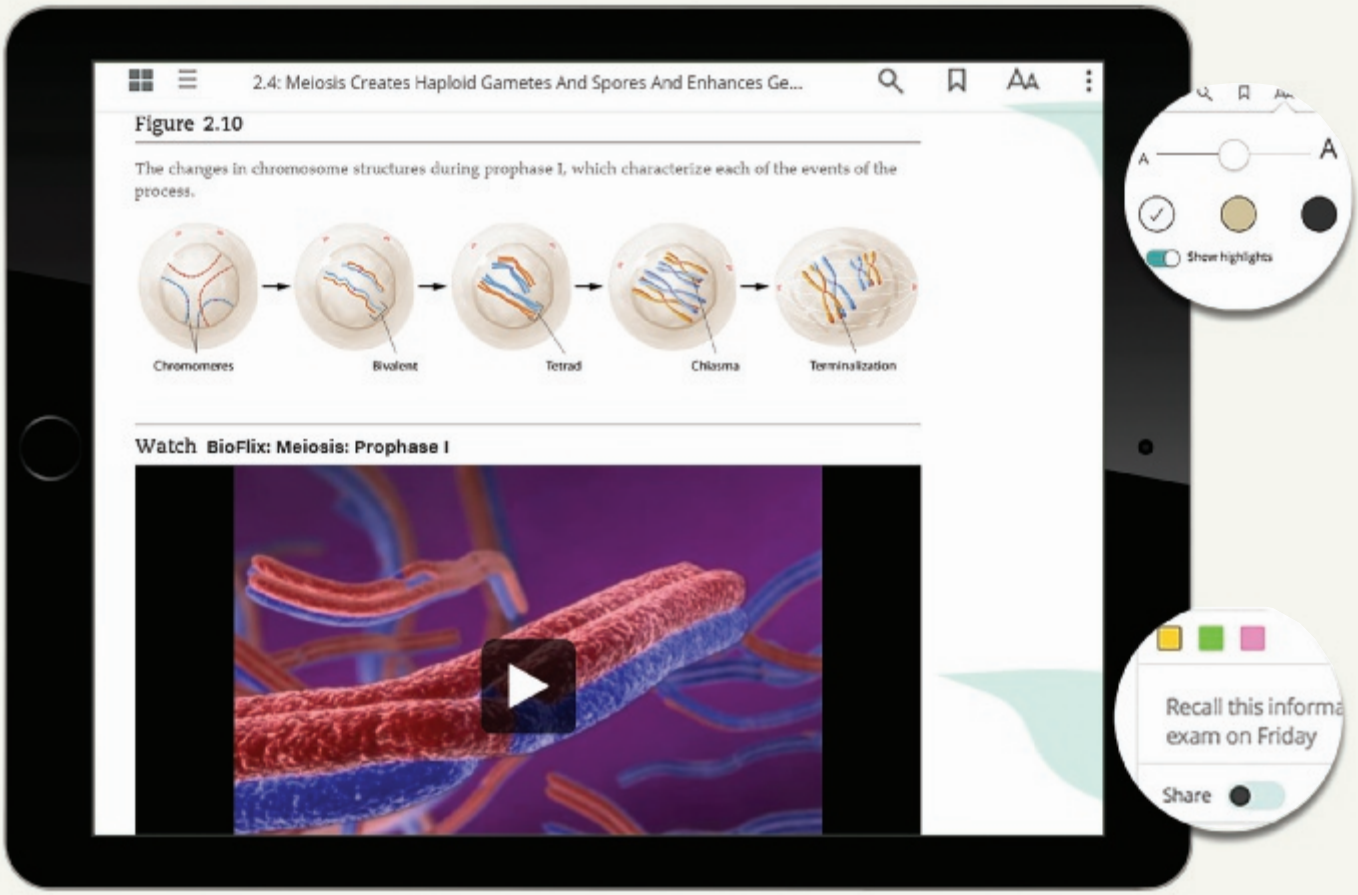
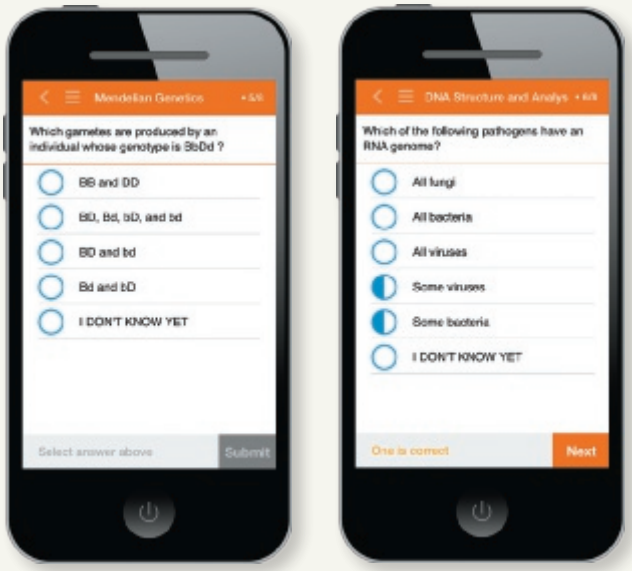
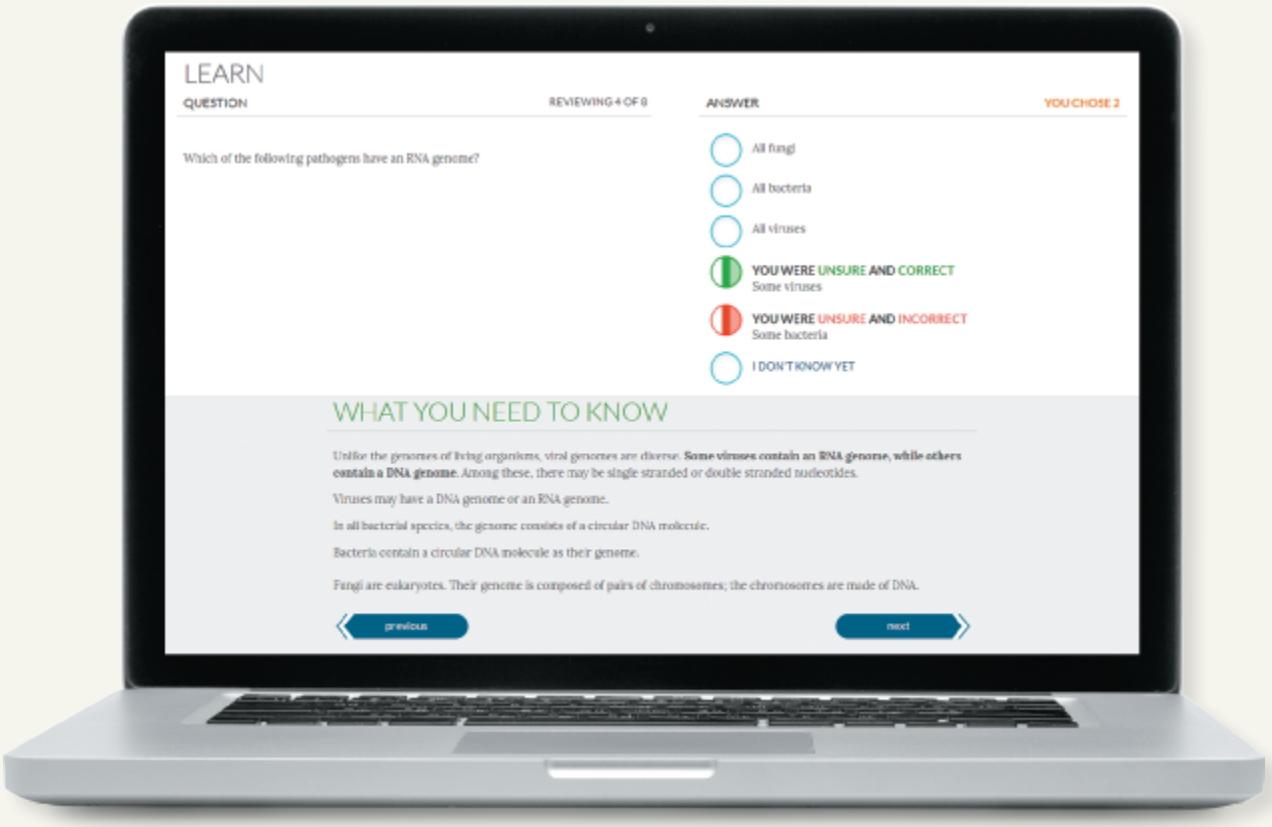
NEW! Pearson eText increases student engagement with embedded animations and videos. In addition, interactive Now Solve This problems help students build knowledge and develop problem-solving skills while learning chapter content.



Improve learning with Dynamic Study Modules

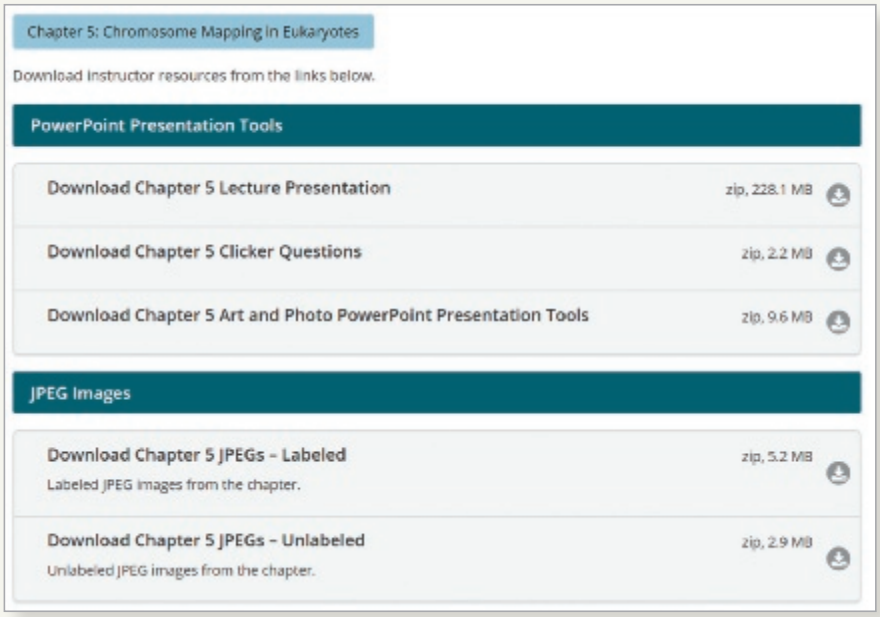


Dynamic Study Modules in Mastering Genetics help students study effectively—and at their own pace—by keeping them motivated and engaged. The assignable modules rely on the latest research in cognitive science, using methods—such as adaptivity, gamification, and intermittent rewards—to stimulate learning and improve retention of key concepts.

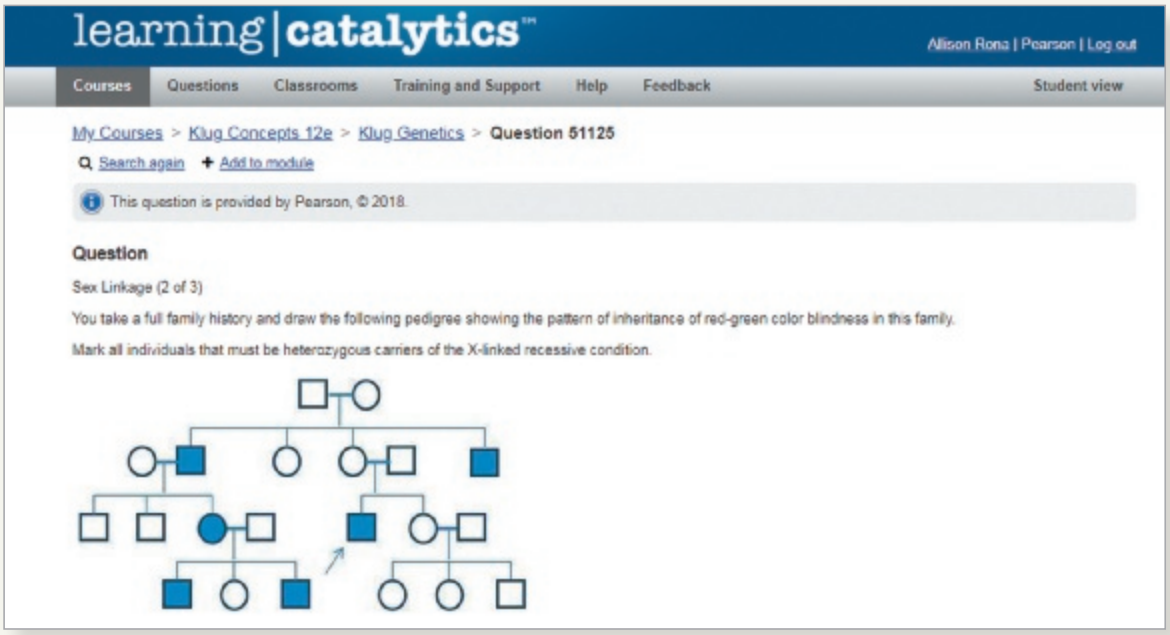


Each module poses a series of questions about a course topic. These question sets adapt to each student's performance and offer personalized, targeted feedback to help them master key concepts. With **Dynamic Study Modules,** students build the confidence they need to deepen their understanding, participate meaningfully, and perform better—in and out of class.

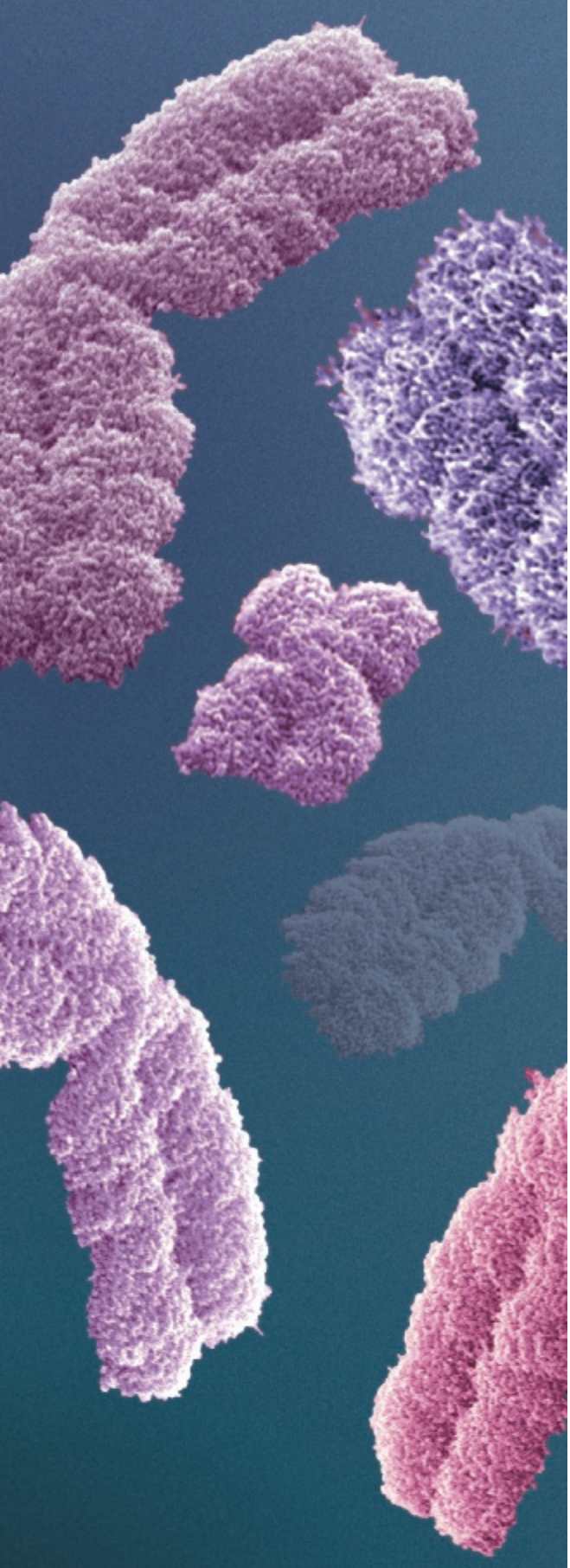
Instructor support you can rely on



Essentials of Genetics includes a full suite of instructor support materials in the Instructor Resources area in Mastering Genetics. Resources include lecture presentations, clicker questions, and art and photos in PowerPoint®; labeled and unlabeled JPEGs of images from the text; and a test bank.



Instructors also have access to Learning Catalytics. With Learning Catalytics, you'll hear from every student when it matters most. You can pose a variety of questions in class that help students recall ideas, apply concepts, and develop critical-thinking skills. Your students respond using their own smartphones, tablets, or laptops. You can monitor responses with real-time analytics and find out what your students do—and don't—understand. Then, you can adjust your teaching accordingly and even facilitate peer-to-peer learning, helping students stay motivated and engaged. Write your own questions, pull from a shared library of community-generated questions, or use Pearson's content clusters, which pose 2-5 questions about a single data set or scenario.



ESSENTIALS *of* GENETICS

Tenth Edition

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Michael R. Cummings is a Research Professor in the Department of Biological, Chemical, and Physical Sciences at Illinois Institute of Technology, Chicago, Illinois. For more than 25 years, he was a faculty member in the Department of Biological Sciences and in the Department of Molecular Genetics at the University of Illinois at Chicago. He has also served on the faculties of Northwestern University and Florida State University. He received his B.A. from St. Mary's College in Winona, Minnesota, and his M.S. and Ph.D. from Northwestern University in Evanston, Illinois. In addition to this text, he has written textbooks in human genetics and general biology. His research interests center on the molecular organization and physical mapping of the heterochromatic regions of human acrocentric chromosomes. At the undergraduate level, he teaches courses in molecular genetics, human genetics, and general biology, and has received numerous awards for teaching excellence given by university faculty, student organizations, and graduating seniors. When not teaching or writing, Dr. Cummings can often be found far offshore fishing for the one that got away.

Charlotte A. Spencer is a retired Associate Professor from the Department of Oncology at the University of Alberta in Edmonton, Alberta, Canada. She has also served as a faculty member in the Department of Biochemistry at the University of Alberta. She received her B.Sc. in Microbiology from the University of British Columbia and her Ph.D. in Genetics from the University of Alberta, followed by postdoctoral training at the Fred Hutchinson Cancer Research Center in Seattle, Washington. Her research interests involve the regulation of RNA polymerase II transcription in cancer cells, cells infected with DNA viruses, and cells traversing the mitotic phase of the cell cycle. She has taught undergraduate and graduate courses in biochemistry, genetics, molecular biology, and oncology. She has also written booklets in the Prentice Hall Exploring Biology series. When not writing and editing contributions to genetics textbooks, Dr. Spencer works on her hazelnut farm and enjoys the peace and quiet of a remote Island off the west coast of British Columbia.

Michael A. Palladino is Vice Provost for Graduate Studies, former Dean of the School of Science, and Professor of Biology at Monmouth University in West Long Branch, New Jersey. He received his B.S. degree in Biology from The College of New Jersey and his Ph.D. in Anatomy and Cell Biology from the University of Virginia. For more than 15 years he directed a laboratory of undergraduate student researchers supported by external funding from the National Institutes of Health, biopharma companies, and other agencies. He and his undergraduates studied molecular mechanisms involved in innate immunity of mammalian male reproductive organs and genes involved in oxygen homeostasis and ischemic injury of the testis. He has taught a wide range of courses including genetics, biotechnology, endocrinology, and cell and molecular biology. He has received several awards for research and teaching, including the 2009 Young Andrologist Award of the American Society of Andrology, the 2005 Distinguished Teacher Award from Monmouth University,

and the 2005 Caring Heart Award from the New Jersey Association for Biomedical Research. He is co-author of the undergraduate textbook *Introduction to Biotechnology*. He was Series Editor for the Benjamin Cummings *Special Topics in Biology* booklet series, and author of the first booklet in the series, *Understanding the Human Genome Project*. When away from the university or authoring textbooks, Dr. Palladino can often be found watching or playing soccer or attempting to catch most any species of fish in freshwater or saltwater.

Darrell J. Killian is an Associate Professor in the Department of Molecular Biology at Colorado College in Colorado Springs, Colorado. He received his B.A. degree in Molecular Biology and Biochemistry from Wesleyan University in Middletown, Connecticut, prior to working as a Research Technician in Molecular Genetics at Rockefeller University in New York, New York. He earned his Ph.D. in Developmental Genetics from New York University in New York, New York, and received his postdoctoral training at the University of Colorado–Boulder in the Department of Molecular, Cellular, and Developmental Biology. Prior to joining Colorado College, he was an Assistant Professor of Biology at the College of New Jersey in Ewing, New Jersey. His research focuses on the genetic regulation of animal development, and he has received funding from the National Institutes of Health and the National Science Foundation. Currently, he and his undergraduate research assistants are investigating the molecular genetic regulation of nervous system development using *C. elegans* and *Drosophila* as model systems. He teaches undergraduate courses in genetics, molecular and cellular biology, stem cell biology, and developmental neurobiology. When away from the classroom and research lab, Dr. Killian can often be found on two wheels exploring trails in the Pike and San Isabel National Forests.

Dedication

We dedicate this edition to our long-time colleague and friend Harry Nickla, who sadly passed away in 2017. With decades of experience teaching Genetics to students at Creighton University, Harry's contribution to our texts included authorship of the Student Handbook and Solutions Manual and the test bank, as well as devising many of the data-based problems found near the end of each chapter. He was also a source of advice during the planning session for each new edition. We always appreciated his professional insights, friendship, and conviviality. We were lucky to have him as part of our team, and we miss him greatly.

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Preface

Essentials of Genetics is written for courses requiring a text that is briefer and less detailed than its more comprehensive companion, *Concepts of Genetics*. While coverage is thorough and modern, *Essentials* is written to be more accessible to biology majors, as well as to students majoring in a number of other disciplines, including agriculture, animal husbandry, chemistry, nursing, engineering, forestry, psychology, and wildlife management. Because *Essentials of Genetics* is shorter than many other texts, it is also more manageable in one-quarter and trimester courses.

Goals

In this edition of *Essentials of Genetics*, the two most important goals have been to introduce pedagogic innovations that enhance learning and to provide carefully updated, highly accessible coverage of genetic topics of both historical and modern significance. As new tools and findings of genetics research continue to emerge rapidly and grow in importance in the study of all subdisciplines of biology, instructors face tough choices about what content is truly essential as they introduce the discipline to novice students. We have thoughtfully revised each chapter in light of this challenge, by selectively scaling back the detail or scope of coverage in the more traditional chapters in order to provide expanded coverage and broader context for the more modern, cutting-edge topics. Our aim is to continue to provide efficient coverage of the fundamental concepts in transmission and molecular genetics that lay the groundwork for more in-depth coverage of emerging topics of growing importance—in particular, the many aspects of the genomic revolution that is already relevant to our day-to-day lives.

While we have adjusted this edition to keep pace with changing content and teaching practices, we remain dedicated to the core principles that underlie this book. Specifically, we seek to

- Emphasize concepts rather than excessive detail.
- Write clearly and directly to students in order to provide understandable explanations of complex analytical topics.
- Emphasize problem solving, thereby guiding students to think analytically and to apply and extend their knowledge of genetics.
- Provide the most modern and up-to-date coverage of this exciting field.
- Propagate the rich history of genetics that so beautifully elucidates how information is acquired as the discipline develops and grows.
- Create inviting, engaging, and pedagogically useful figures enhanced by meaningful photographs to support student understanding.
- Provide outstanding interactive media support to guide students in understanding important concepts through animations, tutorial exercises, and assessment tools.

The above goals serve as the cornerstone of *Essentials of Genetics*. This pedagogic foundation allows the book to accommodate courses with many different approaches and lecture formats. While the book presents a coherent table of contents that represents one approach to offering a course in genetics, chapters are nevertheless written to be independent of one another, allowing instructors to utilize them in various sequences.

New to This Edition

In addition to updating information with new findings in all chapters throughout the text, four chapters are new to this edition.

- **Two new chapters expand the coverage of the regulation of gene expression** The topic of genetic regulation was previously covered in a single chapter, but has now been split into two new chapters. The first (Chapter 15) involves regulation in bacteria, while the second (Chapter 16) focuses on eukaryotes. The bacterial coverage represents the pioneering work in this field and then concludes with an introduction to CRISPR-Cas. The eukaryotic coverage focuses on the regulation of gene expression first at the level of transcription, and then post-transcriptionally, where the expanded coverage focuses on mechanisms that regulate RNA. Research into posttranscriptional regulation in the past 15 years has highlighted the importance of topics such as alternative splicing, mRNA stability and decay, and regulatory noncoding RNAs. Collectively, the addition of these two new chapters provides students and instructors with a thorough, up-to-date presentation of these important aspects of genetics.
- **Two new Special Topics in Modern Genetics chapters** Special Topics chapters are focused and flexible, providing abbreviated, cohesive coverage of important topics in genetics. There are seven Special Topics chapters in this edition, two of which are new. Special Topics Chapter 2—*Genetic Testing* explores how genetic testing is becoming prominent in many contexts and how its use raises many questions and ethical concerns. Special Topics Chapter 4—*Advances in Neurogenetics: The Study of Huntington Disease* illustrates the many advances that have been made in the study of Huntington disease, a

monogenic human disorder that has been subjected to analysis using multiple approaches involving molecular genetics. As such, the chapter exemplifies the growing body of information that has accrued regarding the causes, symptoms, and future treatment of this disorder.

- **Expanded coverage of CRISPR-Cas** Since the previous edition was published, techniques for genome editing have vastly improved due to CRISPR-Cas technology. Thus, we have integrated information about CRISPR-Cas in several different locations within the text. The impact of genome editing with CRISPR-Cas is briefly introduced in Chapter 1. Then, in Chapter 15, students learn how CRISPR-Cas was originally discovered as a bacterial system that regulates the gene expression of bacterial viruses (bacteriophages), providing an immunity against infection. The mechanism and applications to biotechnology are subsequently covered in Chapter 17. Finally, the use of CRISPR-Cas genome editing for gene therapy and the production of genetically modified foods is discussed in Special Topics Chapter 3—*Gene Therapy* and Special Topics Chapter 6—*Genetically Modified Foods*.
- **Increased emphasis on ethics** We recognize in this edition the importance of providing an increased emphasis on ethical considerations that genetics is bringing into everyday life. Regarding this point, we have converted the essay feature previously called *Genetics, Technology, and Society* to one with added emphasis on ethics and renamed it *Genetics, Ethics, and Society*. Approximately half the chapters have new or revised essays. In each case, a synopsis is presented of an ethical issue related to a current finding in genetics that impacts directly on society today. The feature then includes a section called *Your Turn*, which directs students to related resources of short readings and Web sites to support deeper investigation and discussion of the main topic of each essay. In addition, another feature called *Case Study*, which appears near the end of all chapters, has been recast with an increased focus on ethics. Both of these features increase the opportunities for active and cooperative learning as well.

New and Updated Coverage

Below is a chapter-by-chapter list of the most significant new and updated coverage present in this edition.

Ch. 1: Introduction to Genetics • New chapter introduction vignette emphasizing the significance of the discovery of CRISPR-Cas9, a powerful genome-editing system.

Ch. 2: Mitosis and Meiosis • New information on microtubules and microfilaments • Revised Figure 2.9 on Meiotic Prophase I • New Exploring Genomics (EG) entry: PubMed: Exploring and Retrieving Biomedical Literature • New Case Study (CS): Timing Is Everything

Ch. 3: Mendelian Genetics • New Table 3.2 on Dominant and Recessive Human Traits • New Now Solve This (NST) 3.5 on pedigree analysis

Ch. 4: Modification of Mendelian Ratios • New information in the "Mitochondria, Human Health, and Aging" section • New information on the *MERFF* mutation • New Genetics, Ethics, and Society (GES) entry: Mitochondrial Replacement and Three-Parent Babies

Ch. 5: Sex Determination and Sex Chromosomes • New information on Klinefelter syndrome • New GES: A Question of Gender: Sex Selection in Humans

Ch. 6: Chromosome Mutations: Variation in Number and Arrangement • Updated information on copy number variation • New GES: Down Syndrome and Prenatal Testing—The New Eugenics? • A new end of chapter problem involving mapping analysis in *Drosophila*.

Ch. 8: Genetic Analysis and Mapping in Bacteria and Bacteriophages • New GES: Multidrug-Resistant Bacteria: Fighting with Phage

Ch. 10: DNA Replication and Recombination • New details about DNA unwinding during replication • New section entitled "Telomeres in Disease, Aging, and Cancer" • Two new end of chapter problems involving telomeres and telomerase

Ch. 12: The Genetic Code and Transcription • Revised coverage of transcription and RNA processing in eukaryotes • New information on termination of transcription in bacteria • New section entitled "Why Do Introns Exist?" • New GES: Treating Duchene Muscular Dystrophy

Ch. 13: Translation and Proteins • Revised coverage of ribosome and tRNA structure • Revised coverage of translation in bacteria • Expanded coverage of translation in eukaryotes including new information on closed-loop translation, illustrated in a new figure (Fig. 13.10)

Ch. 14: Gene Mutation, DNA Repair, and Transposition • Reorganization of the section on mutation classification, including new table summaries • New and expanded coverage of human germ-line and somatic mutation rates • New, reorganized, and revised coverage of transposable elements, focusing on the major characteristics of retrotransposons and DNA transposons, as well as on how transposons create mutations • Three new figures and one new table

Ch. 15: Regulation of Gene Expression in Bacteria • New chapter that focuses specifically on gene regulation in bacteria • Expanded coverage on the roles of RNA in bacterial gene regulation • New coverage of CRISPR-Cas-mediated regulation of invading viral DNA sequences

Ch. 16: Regulation of Gene Expression in Eukaryotes • New chapter that focuses specifically on gene regulation in eukaryotes • Revised and expanded coverage of alternative splicing, including a new figure, and its relevance to human disease • Expanded coverage on RNA stability and RNA decay including a new figure (Fig. 16.11) • Updated information on noncoding RNAs that regulate gene expression • Enriched coverage of ubiquitin-mediated protein degradation, including a new figure (Fig. 16.14)

Ch. 17: Recombinant DNA Technology • Updated content on modern sequencing technologies including a new figure (Fig. 17.12) on third-generation sequencing (single-strand DNA sequencing) • New section, "Genome Editing with CRISPR-Cas," describes this system as a genome editing tool and includes a new figure (Fig. 17.16)

Ch. 18: Genomics, Bioinformatics, and Proteomics • A new section, "DNA Sequence Analysis Relies on Bioinformatics Applications and Genome Databases," integrating applications of bioinformatics, genome databases, and functional genomics for analyzing and understanding gene function by sequence analysis • Reorganized and revised content on the Human Genome Project, including a new end of chapter problem citing the PANTHER database as part of the Human Genome Project • Updated content on personal genome projects • New content on diploid genomes, mosaicism, and reference genomes and the pangenome to emphasize human genetic variations, including a new figure (Fig. 18.8) • Incorporated coverage of the Human Microbiome Project into a new section, "Metagenomics," and expanded content to include a new Figure (Fig. 18.9) displaying microbiome results of patients with different human disease conditions • A new section titled "RNA Sequencing" • A new section, "Synthetic Genomes and the Emergence of Synthetic Biology," including a new figure (Fig. 18.13) • New GES: Privacy and Anonymity in the Era of Genomic Big Data • Several new and revised end of chapter problems

Ch. 19: The Genetics of Cancer • Extended coverage of environmental agents that contribute to human cancers, including more information about both natural and human-made carcinogens • New subsection entitled "Tobacco Smoke and Cancer" explaining how a well-studied carcinogen induces a wide range of genetic effects that may lead to mutations and cancer

Ch. 20: Quantitative Genetics and Multifactorial Traits • Revised coverage of Expression QTLs (eQTLs) in the regulation of gene expression • New GES: Rice, Genes, and the Second Green Revolution • New CS: A Chance Discovery

Ch. 21: Population and Evolutionary Genetics • New figure (Fig. 21.7) on the relationship between genotype and allele frequency • Important modifications to Figures 21.8 and 21.9 illustrating allele selection • New figure (Fig. 21.13) on the impact of selection types on the phenotypic mean and variance • Revised text and figure (Fig. 21.24) on molecular clocks • Updated information about the origins of the human genome • New figure (Fig. 21.26) on hominin contributions to the genome of modern humans

Special Topic 1: Epigenetics • Revised, updated, and expanded coverage of epigenetic topics, including histone modifications, noncoding RNAs, assisted reproductive technologies, and the heritability of stress-induced behaviors • Updated coverage of epigenetics and cancer • New section on "Epigenetics and Monoallelic Gene Expression" • New figures on DNA methylation, chemical modification of histones, genomic imprinting, random autosomal monoallelic gene expression, imprinting in germ cells, and maternal behavior and stress responses in rat pups

Special Topic 2: Genetic Testing • New Special Topics chapter emphasizing modern approaches to genetic testing including prenatal genetic testing, noninvasive procedures for testing fetal DNA, testing using allele-specific oligonucleotides, microarrays, and genetic analysis by DNA and RNA sequencing • Includes coverage of the recommended uniform screening panel, undiagnosed diseases network, and genetic analysis for pathogen identification during infectious disease outbreaks • Section on genome-wide association studies incorporates approaches for genomic analysis of disease conditions at the population level • A range of ethical, social, and legal considerations are discussed

Special Topic 3: Gene Therapy • Updated information on gene therapy trials that are under way • An expanded section "Genome Editing" highlighting the application of the CRISPR-Cas system and describing some of the most promising trials under way in humans and animals • New ethical considerations of CRISPR-Cas and germ-line and embryo editing • New section, "RNA-Based Therapeutics," that includes coverage of antisense RNA; RNA interference; and updated trials for RNA-based therapeutics, including Spinraza as an antisense RNA modifying splicing for the treatment of spinal muscular atrophy • Updated content on roles of stem cells in gene therapy • New content on combining genome editing with immunotherapy

Special Topic 4: Advances in Neurogenetics: The Study of Huntington Disease • New Special Topics chapter that surveys the study of Huntington Disease (HD) from 1970 to the present • Coverage includes the genetic basis and progression of HD, the mapping and isolation of the gene responsible for the disorder, and information on the mutant gene product • Discussions

include information on the molecular and cellular alterations caused by the mutant protein, the use of transgenic animal models of HD, and the molecular and cellular approaches to therapy

Special Topic 5: DNA Forensics • New section entitled "DNA Phenotyping," describing a controversial forensic method, including descriptions of how law-enforcement agencies currently use this new technology

Special Topic 6: Genetically Modified Foods • New section, entitled "Gene Editing and GM Foods," describing how scientists are using the new techniques of gene editing (including ZFN, TALENS, and CRISPR-Cas) to create GM food plants and animals, and how these methods are changing the way in which GM foods are being regulated • A new box, "The New CRISPR Mushroom," describing the development and regulatory approval of the first CRISPR-created GM food to be cleared for human consumption

Special Topic 7: Genomics and Precision Medicine • New section, entitled "Precision Oncology," describing two targeted cancer immunotherapies: adoptive cell transfer and engineered T-cell therapy • Updated section, "Pharmacogenomics," including a discussion of new trends in preemptive gene screening for pharmacogenomic variants • New box, "Preemptive Pharmacogenomic Screening: The pGEN-4Kids Program," discussing preemptive gene screening that integrates DNA analysis into patient electronic health records

Emphasis on Concepts

Essentials of Genetics focuses on conceptual issues in genetics and uses problem solving to develop a deep understanding of them. We consider a concept to be a cognitive unit of meaning that encompasses a related set of scientifically derived findings and ideas. As such, a concept provides broad mental imagery, which we believe is a very effective way to teach science, in this case, genetics. Details that might be memorized, but soon forgotten, are instead subsumed within a conceptual framework that is more easily retained. Such a framework may be expanded in content as new information is acquired and may interface with other concepts, providing a useful mechanism to integrate and better understand related processes and ideas. An extensive set of concepts may be devised and conveyed to eventually encompass and represent an entire discipline—and this is our goal in this genetics textbook.

To aid students in identifying the conceptual aspects of a major topic, each chapter begins with a section called ***Chapter Concepts***, which identifies the most important ideas about to be presented. Then, throughout each chapter, ***Essential Points*** are provided that establish the key issues that have been discussed. And in the ***How Do We Know?*** question that starts each chapter's problem set, students are asked to identify the experimental basis of important genetic findings presented in the chapter. As an extension of the learning approach in biology called "Science as a Way of Knowing," this feature enhances students' understanding of many key concepts covered in each chapter. Finally, the second entry in each chapter's problem set is labeled as a **Concepts Question**, which asks the student to review and comment on specific aspects of the Chapter Concepts found at the beginning of each chapter.

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 also support this approach throughout the book.

Emphasis on Problem Solving

Helping students develop effective problem-solving skills is one of the greatest challenges of a genetics course. The feature called ***Now Solve This***, integrated throughout each chapter, asks students to link conceptual understanding in a more immediate way to problem solving. Each entry provides a problem for the student to solve that is closely related to the current text discussion. A pedagogic hint is then provided to aid in arriving at the correct solution. All chapters conclude with ***Insights and Solutions***, a popular and highly useful section that provides sample problems and solutions that demonstrate approaches useful in genetic analysis. These help students develop analytical thinking and experimental reasoning skills. Digesting the information in *Insights and Solutions* primes students as they move on to the lengthier ***Problems and Discussion Questions*** section that concludes each chapter. Here, we present questions that review topics in the chapter and problems that ask students to think in an analytical and applied way about genetic concepts. The addition of Mastering Genetics extends our focus on problem solving online, and it allows students to get help and guidance while practicing how to solve problems.

Continuing Features

The Tenth Edition has maintained several popular features that are pedagogically useful for students as they study genetics. Together, these create a platform that seeks to challenge students to think more deeply about, and thus understand more comprehensively, the information he or she has just finished studying.

- **Exploring Genomics** Appearing in numerous chapters, this feature illustrates the pervasiveness of genomics in the current study of genetics. Each entry asks students to access one or more genomics-related Web sites that collectively are among the best publicly available resources and databases. Students work through interactive exercises that ensure their familiarity with the type of

genomic or proteomic information available. Exercises instruct students on how to explore specific topics and how to access significant data. Questions guide student exploration and challenge them to further explore the sites on their own. Importantly, *Exploring Genomics* integrates genomics information throughout the text, as this emerging field is linked to chapter content. This feature provides the basis for individual or group assignments in or out of the classroom.

- **Case Studies** This feature, with an increased emphasis on ethical considerations, appears at the end of each chapter and provides the basis for enhanced classroom interactions. In each entry, a short scenario related to one of the chapter topics is presented, followed by several questions. These ask students to apply their newly acquired knowledge to real-life issues that may be explored in small-group discussions or serve as individual assignments.

For the Instructor

Mastering Genetics
http://www.masteringgenetics.com

Mastering Genetics engages and motivates students to learn and allows you to easily assign automatically graded activities. Tutorials provide students with personalized coaching and feedback. Using the gradebook, you can quickly monitor and display student results. Mastering Genetics easily captures data to demonstrate assessment outcomes. Resources include:

- In-depth tutorials that coach students with hints and feedback specific to their misconceptions.
- A new, robust library of **Practice Problems** offers more opportunities to assign challenging problems for student homework or practice. These questions include targeted wrong answer feedback to help students learn from their mistakes. They appear only in Mastering Genetics, and solutions are not included in the Student Solutions Manual.
- An item library of assignable questions including end of chapter problems, test bank questions, and reading quizzes. You can use publisher-created prebuilt assignments to get started quickly. Each question can be easily edited to match the precise language you use.
- A gradebook that provides you with quick results and easy-to-interpret insights into student performance.

Instructor Resources

The Instructor Resources, available for download in the Instructor area of Mastering Genetics, offer adopters of the text convenient access to a comprehensive and innovative set of lecture presentation and teaching tools. 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 test bank, the text's in-chapter text questions, and the student media practice questions.

TestGen EQ Computerized Testing Software

(ISBN: 0135272823 / 9780135272824)

Test questions are 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.

For the Student

Student Handbook and Solutions Manual

Authored by Michelle Gaudette, Tufts University, and Harry Nickla ***(ISBN: 0132300728 / 9780135300428)***

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.

Mastering Genetics
http://www.masteringgenetics.com

Used by over a million science students, the Mastering platform is the most effective and widely used online tutorial, homework, and assessment system for the sciences. Perform better on exams with Mastering Genetics. As an instructor-assigned homework system, Mastering Genetics is designed to provide students with a variety of assessments to help them understand key topics and concepts and to build problem-solving skills. Mastering Genetics tutorials guide

students through the toughest topics in genetics with self-paced tutorials that provide individualized coaching with hints and feedback specific to a student's individual misconceptions. Students can also explore Mastering Genetics' Study Area, which includes animations, the eText, *Exploring Genomics* exercises, and other study aids. The interactive eText 2.0 allows students to access their text on mobile devices, highlight text, add study notes, review instructor's notes, and search throughout the text, 24/7.

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