Preface

Educators have increasingly focused on *scientific competency* as the goal of science education. Scientific competency refers to the learner's ability to use the concepts and methods of science in meaningful ways. Ideally, what a student should acquire in science education is knowledge, inquisitiveness, a desire to learn, and habits of critical thinking and quantitative reasoning that will last a lifetime.

Essential Genetics: A Genomics Perspective, Fifth Edition, is designed to help students achieve scientific competency. It is meant for the shorter, less comprehensive introductory course. The brevity of the text fits the pace of what can be covered in a typical one-semester or one-quarter course. The topics have been carefully chosen to help students master the following specific competencies:

- Understand the basic processes of gene transmission, mutation, expression, and regulation
- Analyze genetic processes using numerical relationships including ratios and proportions
- Learn to formulate genetic hypotheses in a statistical framework, work out their consequences, and test the results against observed data
- Develop basic skills in problem solving, including single-concept exercises, those requiring the application of several concepts in logical order, and numerical problems requiring some arithmetic for solution
- Interpret genetic data and make valid inferences to reveal the underlying causes
- Gain some sense of the social and historical context in which genetics has developed as well as an appreciation of current trends
- Become aware of some of the genetic resources and information that are available through the World Wide Web, and learn how to evaluate and integrate this information.

Genetics is an excellent subject for achieving scientific competency. It is integrative over a broad territory, ranging from molecular biology to evolutionary genetics. It is also quantitative, using concepts from probability, statistics, and computational biology. Fortunately, undergraduates come to a course in genetics highly motivated because of media reports about the human genome and genetic risk factors for disease, as well as many social and ethical controversies related to genetics such as genetic privacy, cloning, and stem-cell research. The challenges for the instructor are to sustain this motivation and to help students acquire the skills and habits of thought that constitute scientific competency.

What's New in the Fifth Edition?

The seamless integration of "molecular genetics" and "classical genetics" makes *Essential Genetics* unique. They are recognized as merely different aspects of the same things—the transmission, mutation, and function of the genetic material. These processes are manifested at the molecular level and can be studied by such techniques as DNA electrophoresis, as well as at the organismic level through their association with visible traits. These levels of genetic analysis are integrated even in the discussion of Mendel's experiments.

This fifth edition has been completely revised and updated. Each chapter has been thoroughly reworked. Nonessential or outdated material has been deleted, new methods and results have been added, and several of the chapters have been reorganized to allow smooth integration of the new material. The text has been analyzed, word by word and line by line, to condense, clarify, and update. To keep the book at a reasonable size, whenever new material has been added, an equivalent amount that is less relevant, of secondary importance, or no longer needed has been removed or abbreviated. Several new *human connection* boxes have been added, and there are more than 50 new or updated illustrations and tables. Hundreds of new, classroom-tested problems have been added, and the *Guide to Problem Solving* has been expanded.

A completely new section on **genetic analysis** has been included in Chapter 1, which illustrates

how geneticists obtain and analyze mutations to discover how biological processes are organized and how they function. The discussion focuses on the classical experiments of Beadle and Tatum, but emphasizes that the application of similar methods of genetic analysis has led to six Nobel Prizes. The discussion of transmission genetics in Chapter 2 is enlivened by a new section on the harmless **taster polymorphism** associated with a dislike of broccoli and by new information about the likely molecular basis of Mendel's green versus yellow difference in pea seeds. The discussion of cell division in Chapter 3 has been brought up to date with more information on **condensins, chromatin modification,** and **the architecture of the spindle.**

Chapter 5 includes a new section on genetic variation, focusing on **single-nucleotide polymorphisms (SNPs)** and the **oligonucleotide arrays (SNP chips)** used to detect them. The new section also includes a discussion of **copy-number polymorphisms (CNPs),** which prove to be far more widespread and prevalent than had been expected. Some CNPs are now known to be associated with conditions such as **autism.** The latest methods for **massively parallel DNA sequencing** are highlighted in Chapter 6. Some of the new sequencing machines can generate more than a billion base pairs of sequence in a single sequencing run. The discussion of microbial genomes in Chapter 7 is broadened to include differences in **genome size** among bacterial isolates of the same species and in the **pathogenicity islands** that allow some otherwise harmless bacteria to cause serious disease.

The discussion of transcription in Chapter 8 includes a completely new section on the structure and function of **RNA polymerase.** This chapter also includes a new discussion of mechanisms of **error correction in translation.** Chapter 9 contains expanded discussions of regulation of the lactose operon and its **multiple operators,** a new section on regulation of the **galactose genes in yeast,** and updated information on the mechanisms of **RNA interference (RNAi).**

Chapter 10, which deals with genomics and proteomics, has been completely revised to reflect these rapidly changing fields. The revision includes new sections on **comparative genomics** and **genome annotation.** Developmental genetics, the subject of Chapter 11, now includes a discussion of how the **principle of epistasis** is used to discover the order in which components act in a linear switch-regulation pathway, such as the developmental pathway controlling vulval development in the nematode. Chapter 12 includes a completely reorganized section on mechanisms of **DNA repair,** and Chapter 15 has a new section on the **special features of cancer cells** and the role of **p53 "poison subunits"** in cancer.

Chapters 14 and 15 summarize population and evolutionary genetics. Chapter 14 contains an expanded summary of the major computational methods for estimating **gene trees and species trees** by means of sequence comparisons among orthologous genes. Chapter 15 includes the principal findings that have emerged so far from studies of the **quantitative trait loci (QTLs)** that underlie the genetic basis of complex traits. New information on the possible relation between the serotonin reuptake transporter and depression comes from studies of genotype-by-environment interaction leading to **depression among habitual users of illicit drugs.**

Chapter Organization

To help the student keep track of the main concepts without being distracted by details, each chapter begins with a full-sentence **Outline** of the principal subjects to be discussed. The body of each chapter provides more detailed information and experimental evidence. An opening paragraph gives an overview of the chapter, illustrates the subject with some specific examples, and shows how the material is connected to genetics as a whole. The section and subsection **Headings** are in the form of complete sentences that encapsulate the main message. The text makes liberal use of **Numbered** and **Bulleted Lists** to aid students in organizing their learning, as well as **Summary Statements** set apart from the main text in order to emphasize important principles. A feature called ***A Moment to Think*** is designed to reinforce understanding. This is a problem integrated into the text in which the student is asked to interrupt studying by thinking about the concept just described and using it to solve an actual problem. Each chapter also includes ***the human connection.*** This special feature highlights a research paper in human genetics that reports a key experiment or raises important social, ethical, or legal issues. Each *human connection* has a brief introduction of its own, explaining the importance of the experiment and the context in which it was carried out. At the end of each chapter is a complete **Chapter Summary** in the form of bullet points highlighting the most important concepts and several different types and levels of **Problems.** At the back of the book are **Answers** to even-numbered problems, a complete **Glossary** of key terms in genetics, a compilation of frequently used **Word Roots** that will help students to understand key genetic terms and make them part of their vocabulary, and suggestions for **Further Reading.**

Contents

The organization of the chapters is that favored by the majority of instructors who teach genetics. An important feature is the presence of an introductory chapter providing a broad overview of the gene: what

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