

CONCEPTS OF GENETICS

THIRD EDITION

CONCEPTS OF GENETICS

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Cover Illustration: Supplied by Lionel I. Rebhun at the University of Virginia, this micrograph was derived from fertilized eggs of the sea urchin *Strongylocentrotus purpuratus*. The eggs were processed after spindle formation had occurred with a buffer that preserves the cytoskeleton; fixed; and mounted on coverslips. The preparation was then exposed to an antibody which recognizes beta-tubulin and, thus, reveals microtubules. This antibody was visualized with a rhodamine-labelled secondary antibody. The chromosomes were stained with a DNA intercalating dye (Hoechst 33342) and the whole preparation photographed with a fluorescence microscope.

Text Illustrations: Hans & Cassady, Inc.

To those who mean the very most,
Cindy, Brad, and Dori
Lee Ann, Brendan, and Kerry

Captains and kings may rule the world,
but it is only the presence and memory of those we love
and with whom we share our intellectual and genetic heritage
that bring beauty to living and justify their good deeds

And to the memory of Bill Gray,
who understood this more so than most of us,
Rest easy, good friend.

WILLIAM S. KLUG is currently Professor and Chairman of the Department of Biology at Trenton State College at Hillwood Lakes, New Jersey. He was previously a member of the faculty of Wabash College.

MICHAEL R. CUMMINGS, currently Associate Professor of Biological Sciences and Research Associate Professor at the Institute for the Study of Developmental Disabilities, University of Illinois, Chicago, has also served on the faculties of Northwestern University and Florida State University.

Preface

While planning the third edition, it became clear that our goals for the third edition have not changed from the previous editions, namely to:

- Emphasize concepts rather than excess detail
- Write clearly and directly, to students rather than to researchers
- Provide a clear organizational framework within and between chapters
- Design meaningful illustrations
- Create extensive sets of challenging problems
- Propagate the rich history of genetics that so beautifully illustrates how a discipline develops and how science works

Feedback from a variety of sources has confirmed that we have been reasonably successful in achieving these goals, and that they should once again serve as the text's cornerstone.

The field of genetics has moved much too rapidly to allow a revision with only minor polishing and updating. The third edition reflects substantial reorganization; a considerable amount of new information; and many new features to enhance the pedagogical value of the book. As was true for the first two editions, this text will accommodate many different organizational approaches and lecture formats.

Chapters are written so that they are as independent of others as possible, allowing instructors to use them in various sequences. Within each chapter, central concepts are emphasized that will support a variety of lecture formats. We believe that this focus has contributed most to the success of *Concepts of Genetics*.

FEATURES

The major features that distinguish this edition from the second edition include

- A revised chapter organization
- A new, full-color art program
- Several new sections in each chapter which further emphasize problem solving as well as conceptual and analytical thinking
- Updating to include the most recent topics in the field of genetics, as well as expansion of several existing sections
- A new supplement package, which includes a *Student's Handbook: A Guide to Concepts and Problem Solving*, an *Instructor's Sourcebook*, and a package of full-color overhead transparencies

Organization

In response to comments from users and reviewers of the text over the past five years, we have rearranged the sequence of chapters.

Part One, which emphasizes transmission genetics and phenotypic variation, now includes the chapter entitled "Variations in Chromosome Number and Arrangement" (Chapter 7). In addition, while the topic of polygenic inheritance has been retained in Chapter 4 as it relates to "Modification of Mendelian Ratios," the remaining quantitative genetics topics have been moved and expanded upon in the new Chapter 23.

Part Two again begins with three chapters on DNA structure, analysis, and replication, but these are now followed immediately by the three chapters introducing the genetic code and its transcription and translation into proteins. This part concludes with the chapter discussing the process of mutation.

Part Three has been reorganized around the central theme of the organization and regulation of genetic information. Following the introduction of bacterial and viral genetic systems and the technology of recombinant DNA, the ensuing chapters pursue the topics of the structure and organization of the genome and the genes within it, as well as their regulation. The latter topic considers bacterial-viral modes separately from those of eukaryotes.

Part Four emphasizes the genetic basis of the development of organisms, as well as their behavior and organization into populations. The text concludes with the considerations of evolutionary genetics.

We have retained an appendix on experimental methodology, a glossary, and answers to selected problems.

Art Program

Redoing all artwork in a full-color format and adding a large number of color photographs and micrographs have undoubtedly enhanced the pedagogical value of the illustrations. The student will find a consistent assignment of color to each of the essential genetic molecules and components throughout the text (e.g., DNA, replicated DNA, the various RNAs, proteins, ribosomes, plasmids, etc.). Whenever possible, we have continued to use the "flow-chart diagrams" that were praised in the first two editions. We also have included a color photograph of nearly every plant that is discussed in the text.

Problem Solving and Conceptual and Analytic Thinking

Several new features added to each chapter capitalize on the way in which genetics lends itself to learning how to solve problems and to think conceptually and analytically. Initiating each chapter is a short section called "Chapter Concepts." In a few sentences, this section summarizes the most important ideas presented. At the conclusion of each chapter, a "Chapter Summary" enumerates the major points that have been discussed. Following the summary is a section entitled "Insights and Solutions," which presents students with solved problems and responses to analytical inquiries. This section emphasizes problem solving in those chapters lending themselves to quantitative analysis, and stresses analytical thinking and experimental rationale in other chapters. "Insights and Solutions" will teach students how to approach and solve problems, will stimulate analytical thinking, and will facilitate their success in the ensuing "Problems and Discussion Questions" section, which has been expanded with more challenging entries.

Updating

Every year new techniques are developed and new findings elevate certain topics to greater prominence. Some topics and techniques that have been introduced for the first time, or have been given greater attention, in this edition include

- DNA supercoiling
- gene conversion
- oncogenes
- transposable elements
- directed mutation
- retroviruses
- site-directed mutagenesis
- DNA fingerprinting
- polymerase chain reaction (PCR)
- restriction fragment length polymorphisms
- eukaryotic transcription factors
- centromere and telomere sequences
- human mitochondrial disorders

In addition, many other areas have been expanded, reflecting our growing knowledge of the genetics of eukaryotes. There are also several new sections—one in Chapter 1 that details the early history of genetics. A new section on probability has been added in Chapter 3.

Supplement Package

Student's Handbook: A Guide to Concepts and Problem Solving by Harry Nickla of Creighton University reviews vocabulary, concepts, and problem solving chapter by chapter. In addition, it provides a very detailed solution or lengthy discussion for every problem and question in the text, and supplies additional problems to be used for practice by students.

The *Instructor's Sourcebook*, also by Harry Nickla, contains questions and problems an instructor can use to prepare exams.

Overhead transparencies of approximately 100 illustrations from the text will be available to adopters of the text.

ACKNOWLEDGMENTS

No text of this breadth and depth could be the sole work of its authors. The field is just too extensive to be mastered by only two of us. While we take complete responsibility for any errors herein, we gratefully acknowledge the advice and contributions made by the reviewers of all three editions, particularly those who were involved in this edition:

Sidney L. Beck	De Paul University
Elliot S. Goldstein	Arizona State University
George Haughn	University of Saskatchewan
Dennis Hynes	California Polytechnic University, SLO
Keith K. Klein	Mankato State University
Harry Nickla	Creighton University
W. Stuart Riggsby	University of Tennessee
Robert M. Zarcaro	Providence College

One reviewer, Elliot Goldstein, has advised us during the preparation of all three editions. Another,

Harry Nickla, has written the *Student's Handbook* and the *Instructor's Sourcebook*. We thank them both for efforts far beyond reasonable expectations. Gratitude is also extended to Allan Gotthelf and James G. Lennox for reviewing and editing the new historical section in Chapter 1.

We also thank our colleagues and our secretarial staff, who together have bolstered our efforts with encouragement, specific discussions, and endless technical support. In particular, Dr. Jim Bricker, Ms. Bette Baier, and Mrs. Monica Zrada have provided invaluable assistance at Trenton State College. And to Linda Burroughs and Lisa Andrasz, who helped structure the Chapter Summaries.

At Macmillan Publishing, we express our appreciation to Cindy Brunk, whose design efforts have greatly enhanced the book; to JoEllen Gohr, who so skillfully guided the text through production; to Dick Morel and to the entire staff at Hans & Cassady, whose creative efforts are reflected in the many figures; to Mark Garrett, who coordinated the art program; to Yvonne Gerin and Chris Migdol, who researched the color photographs; and to Vickie Brewster, whose experience in genetics has enhanced the index, which she produced. Particular thanks are due to Bob Rogers, who has become a good friend as he coordinated all of these efforts in his role as Senior Editor. More than any other factor, it has been his belief in and support of this project that has brought the third edition to fruition. It has been a real pleasure working with all of the above individuals, who deserve to share in any success that this text enjoys. Our gratitude to them goes well beyond these written words.

William S. Klug
Michael R. Cummings

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**PART FOUR
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AND POPULATIONS**

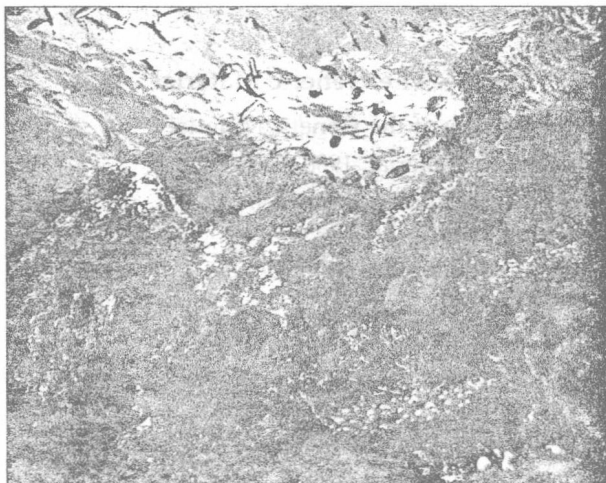
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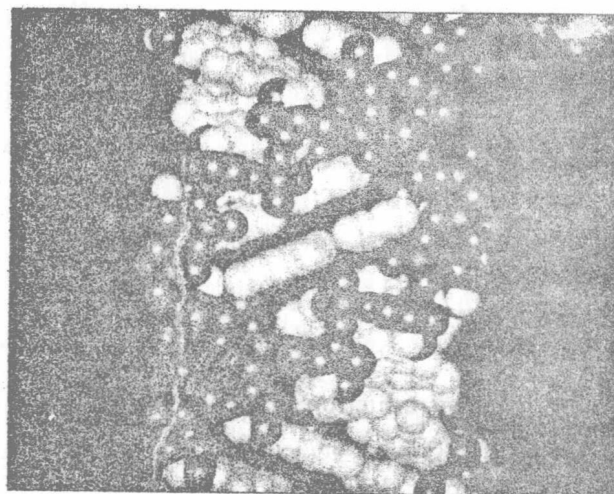
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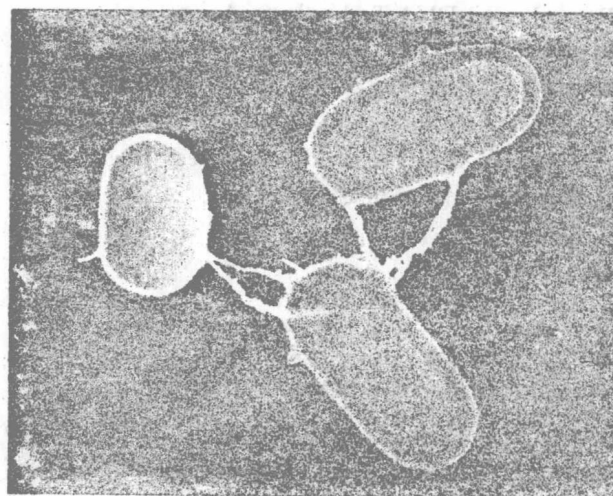
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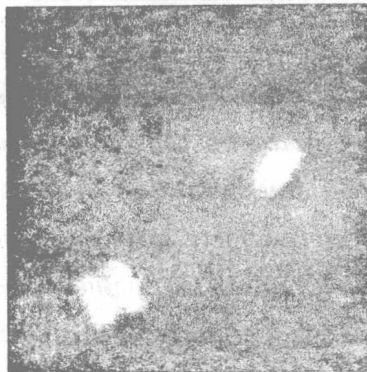
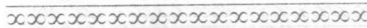
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1

An Introduction to Genetics

THE HISTORICAL CONTEXT OF GENETICS

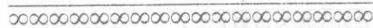
Prehistoric Times: Domesticated Animals and Cultivated Plants | The Greek Influence:
The Origin of Pangenesis and Inheritance of Acquired Characteristics | 1600–1850: The
Dawn of Modern Biology | Charles Darwin and Evolution

BASIC CONCEPTS OF GENETICS

INVESTIGATIVE APPROACHES IN GENETICS

THE SIGNIFICANCE OF GENETICS IN SOCIETY

Soviet Genetics: Science and Politics | Genetic Advances in Agriculture and Medicine



CHAPTER CONCEPTS

Genetics is the science of heredity. The discipline has a rich history and involves investigations of molecules, cells, organisms, and populations, utilizing many different experimental approaches. Because of the significance of genetic information and its expression at all levels of the function of individuals and during evolution, genetics has unified the study of biology and had a profound impact on human affairs.

Welcome to the study of the discipline of genetics. You are about to embark on the exploration of a subject that many students before you have found to be the most interesting and fascinating in the field of biology. This is not surprising because an understanding of genetic processes is fundamental to the comprehension of life itself. Genetic information directs cellular function, determines an organism's external appearance, and serves as the link between generations in every species. Knowing how these processes occur is important to understanding the living world. Knowledge of genetic concepts also helps us to understand the other disciplines of biology. The topics studied in genetics overlap directly with molecular biology, cell biology, physiology, evolution, ecology, systematics, and behavior. Usually, the study of each of these disciplines is incomplete without the knowledge of the genetic component underlying each of them. Genetics is therefore said to unify biology and serve as its "core."

Interest in and fascination with this discipline further stem from the fact that, in genetics, many initially vague and abstract concepts have been investigated in a logical fashion until they have become clearly and definitively understood. As a result, genetics has a rich history that exemplifies the nature of scientific discovery and the analytical approach used to acquire information. Scientific analysis, moving from the unknown to the known, is one of the major forces that attract students to biology.

But there is still another reason why the study of genetics is so appealing. Since its establishment, this field of study has expanded continuously. Every year large numbers of new findings are made. While it has been said that scientific knowledge doubles every ten years, one estimate holds that the doubling time in genetics is less than five years. Certainly, over the

past four decades, no five-year period has passed without some of the newly acquired information causing significant excitement for geneticists and for biologists in general. And each advance becomes part of an ever-expanding cornerstone upon which further progress is based. It is particularly stimulating to be in the midst of these developments, whether you are studying or teaching genetics.

THE HISTORICAL CONTEXT OF GENETICS

In the chapters that follow, we will discuss the behavior of chromosomes, the way in which genes present on them are transmitted from one generation to the next, and the way in which genetic information is stored, altered, expressed, and regulated. The most significant scientific findings that serve as the foundation for this information were obtained in the nineteenth century. As the twentieth century dawned, a period of integration of this knowledge occurred, clarifying the physical basis of living organisms and their relationship to one another. For example, the ideas that matter is composed of atoms, that cells are the fundamental units of living organisms, that cells contain nuclei that house threadlike structures called chromosomes, and that chromosomes are constant in number in a species and thus might be important in heredity together provided the basis for an important synthesis of ideas. When these ideas were combined with the newly rediscovered findings of Gregor Mendel and integrated with Darwin's theory of evolution and the origin of species, a more complete picture of life at the level of the individual and of the population emerged. On this foundation, the era of modern-day biology was initiated. And, beyond this introductory chapter, it is