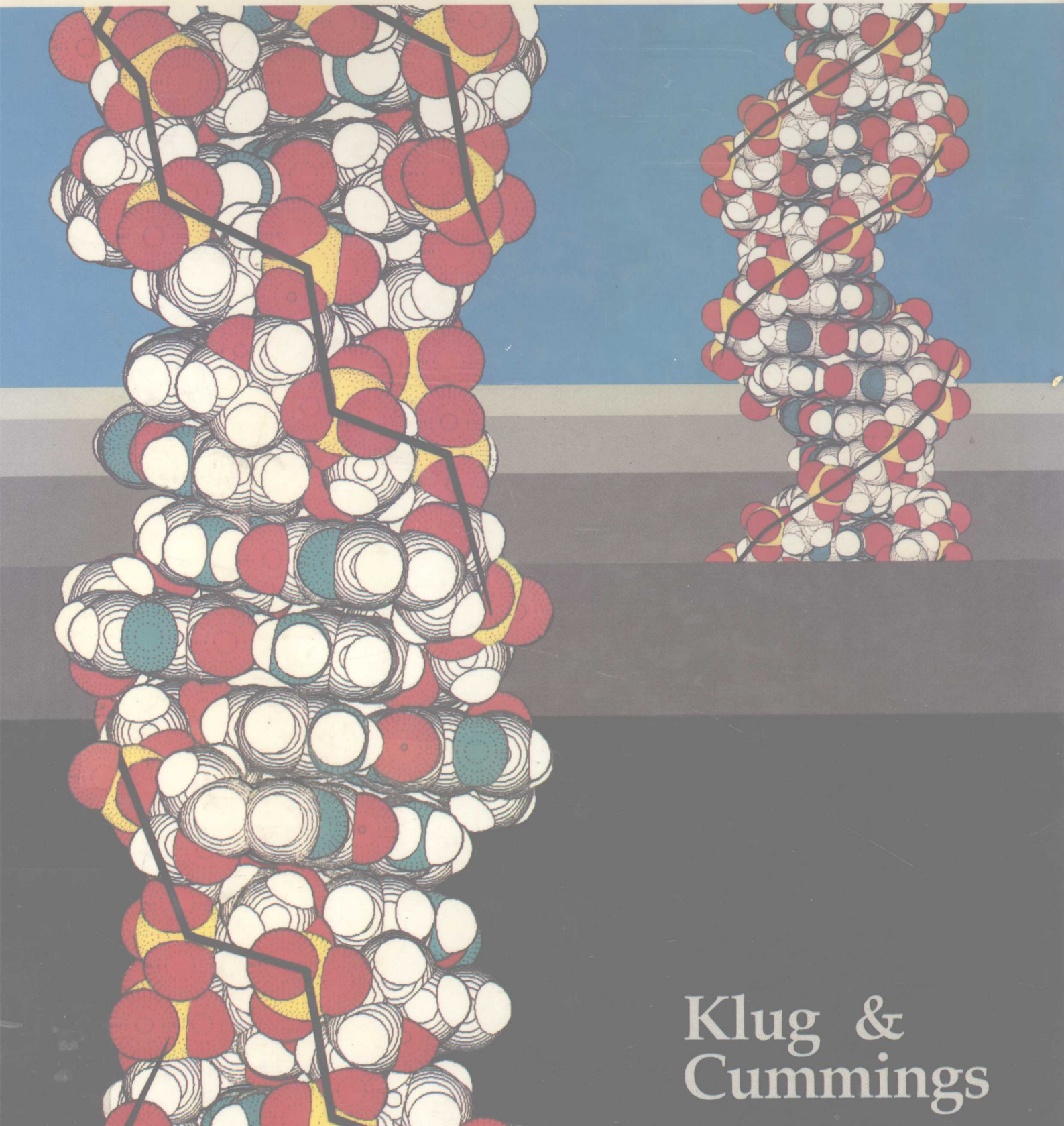
Concepts of GENETICS



Klug & Cummings

CONCEPTS OF GENETICS

WILLIAM S. KLUG
Trenton State College at Hillwood Lakes

and

MICHAEL R. CUMMINGS
University of Illinois, Chicago

CHARLES E. MERRILL PUBLISHING COMPANY
A Bell & Howell Company
Columbus • Toronto • London • Sydney

Cover Art: Z-DNA and B-DNA models computer generated by Gary J. Quigley, Massachusetts Institute of Technology. Courtesy of Alexander Rich, Department of Biology, Massachusetts Institute of Technology.

Published by
Charles E. Merrill Publishing Company
A Bell & Howell Company
Columbus, Ohio 43216

This book was set in Palatino.
Production Coordination: Rex Davidson
Copy Editor: Jo Ellen Gohr
Text Designer: Ann Mirels
Cover Designer: Tony Faiola

Library of Congress Catalog Card Number 82–060576
International Standard Book Number 0675–20010–5
Printed in the United States of America

4 5 6 7 8 9 10—87 86 85

CONCEPTS OF GENETICS

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
which bring beauty to living and justify their good deeds

The Authors

WILLIAM S. KLUG is presently serving as Professor of Biology and as Chairman of the Department of Biology at Trenton State College at Hillwood Lakes. He was previously a member of the faculty of Wabash College, Crawfordsville, Indiana. The focus of his research interest is on developmental genetics in *Drosophila,* with an emphasis on genes that control oogenesis and on ribosomal proteins. Dr. Klug is also interested in the use of the oogenesis system in carcinogenesis screening. He has numerous research publications in scientific journals.

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, Tallahassee. His research is concerned with differential gene expression during oogenesis, with emphasis on ribosomal RNA genes and mRNA populations. Dr. Cummings' other research interest is in the molecular consequences of aneuploidy, particularly in the molecular organization and regulation of gene expression in Down syndrome. He has published research papers in various scientific journals.

Preface

The title *Concepts of Genetics* conveys the basic pedagogic philosophy that has shaped this textbook. Throughout the text we have emphasized the concepts or general ideas that have been developed to explain genetic phenomena at all levels—the molecule, cell, organism, and population. The word *concept* implies that sense can be made of the abstract. Indeed, no discipline in biology better illustrates this transition in the acquisition of knowledge. We have attempted to minimize the presentation of highly technical and detailed information which for most students tends to blur the many important concepts and their logical development.

The information presented herein is a meld of the most significant findings in the science of heredity, including those in transmission genetics, cytogenetics, microbial genetics, molecular genetics, and population-evolutionary genetics. These include the original work of Mendel in the nineteenth century, the advances in genetic technology of the current and most recent decades, and those made between these periods.

Writing style has been carefully developed and edited solely with the student in mind. Our primary goals have been clear writing and meaningful figures, yet we have remained rigorous in our coverage of topics, believing that excess detail cannot be equated with rigor. This text is designed for use in all introductory genetics courses, whether offered in a semester, in a quarter, or in consecutive quarters.

For the instructor, we have created a flexible format and organization. The book is divided into five parts. The basic concepts of transmission genetics are discussed in Part 1. Within this section, meiosis is discussed before Mendelian genetics, which uses as its foundation the information about the behavior of chromosomes during gamete formation. Part 2 focuses on DNA—its structure, analysis, and replication. This section concludes with a chapter on the organization of DNA in chromosomes. Part 3 concentrates on genetic variability at both the chromosome and gene levels. The discussion of mutation and mutagenesis logically precedes the consideration of bacterial and viral genetics. These chapters provide a strong foundation for a more detailed consideration of molecular genetics in Part 4. Chapter 19 provides a modern synthesis of old and new findings concerning the organization of DNA into the functional unit of heredity, the gene. Part 5 includes discussions of developmental, somatic cell, and behavioral genetics as well as chapters on the genetics of populations and evolution. We believe that separate chapters on somatic cell genetics and behavior genetics are essential, since they represent two areas that are likely to expand our knowledge considerably over the next decade.

In general, the various parts and the chapters within them may be used interchangeably. This is particularly true of the first two parts, which stand completely independently of each other. Placing in-depth coverage of experimental methods and probability and statistics in Appendices A and B is in keeping with the idea of flexible reading assignments. Answers to selected problems (Appendix C) as well as an extensive glossary complete the text.

This work has been in progress for over five years, allowing several revisions and reviews for content and clarity. In the final draft, we were able to avoid the organizational idiosyncrasies of our own courses. Instead, we have written a text that will be easily adaptable to a wide variety of course formats. We have never, however, lost sight of our primary goal—to write a text that will provide students with a rewarding learning experience and that conveys clearly the concepts of genetics.

William S. Klug

Michael R. Cummings

Acknowledgements

Few, if any, textbooks are written without the support and professional contributions of many people. This book is no exception. First, we wish to thank Bob Lakemacher for his belief in this project. Also at Charles E. Merrill, we are extremely grateful to Jo Ellen Gohr, who edited and followed the manuscript through the production process, and to Meg Malde and Rex Davidson for their coordination efforts. It was a pleasure to deal with the many people at ANCO/Boston who rendered the more complex artwork.

A textbook depends in large part on the input provided by reviewers. We have been fortunate to have had the manuscript assessed at several stages during its preparation. While this advice has been instrumental in shaping the organization of the text, and in deciding to add or delete certain material, we take full responsibility for its content. Among others, the reviewers include Glen C. Bewley, North Carolina State University; Charles C. Biggers, Memphis State University; H. E. Brockman, Illinois State University; Raymond P. Canham, Southern Methodist University; Lee Ehrman, State University of New York, Purchase; B. W. Greer, Knox College; Elliot S. Goldstein, Arizona State University; Victor J. Hoff, Austin State University; R. D. Jackson, Texas Tech University; Diana Johnson, George Washington University; Gary Kikudome, University of Missouri, Columbia; Robert M. Kitchin, University of Wyoming; Walter Rothenbuhler, Ohio State University; Eliot Spiess, University of Illinois, Chicago; and Dana Wrensch, Ohio Wesleyan University.

The acquisition of photographs and permissions has placed us in contact with well over 100 geneticists. We are grateful to all those who dealt with us in a thoughtful and cooperative manner. Several have contributed above and beyond the call. In this regard, we thank Alexander Rich and those in his laboratory at the Massachusetts Institute of Technology who provided material for the cover and many fine micrographs. Likewise, we are indebted to Oscar Miller, Jr., University of Virginia; Jorge Yunis, University of Minnesota; and Arthur E. Greene, Institute of Medical Research, for their many contributions.

On a day-to-day basis, we were dependent on a secretarial staff whose dedicated efforts were essential to meeting the many deadlines during the past several years. This staff included Ruth Laskin and Saveria Symons at Trenton State College, and Helen Brown at the Institute for the Study of Developmental Disabilities. Their efficiency and pleasant manner aided immensely in the completion of this project.

The support of and discussions with colleagues have been no less important to us. We personally thank Tom Cole for his early participation in the conception of the project, Eliot Spiess for sharing his experiences as an

author and his expertise in population genetics, Karl Gottesman for discussions on writing style, and Al Eble and Joe Vena for their encouragement.

Finally, we would like to thank our spouses and families for their patience during periods of neglect.

To all of these people we offer our sincere gratitude for their part in making this textbook a reality.

William S. Klug

Michael R. Cummings

Contents in Brief

Contents

PART 2

DNA—The Chemical Basis of Heredity

PART 3

Genetic Variation

PART 4

Gene Structure, Function, and Regulation

PART 5

Genetics of Organisms and Populations

Appendices

1 An Introduction to Genetics

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, behavior, and ecology. 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, countless 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 which 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 which attract students to biology.

But there is still another reason why the study of genetics is so appealing. Since it began, the field has never stopped expanding. 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 only two years. Certainly, over the past four decades, no two-year period has passed without some of the newly acquired information causing significant excitement within the field and for biologists in general. And each advance becomes part of an ever-expanding cornerstone upon which further progress is made. It is particularly stimulating to be in the midst of these developments, whether you are studying or teaching genetics.

BASIC CONCEPTS OF GENETICS

In this introductory chapter we would like to review some of the simple but basic concepts in genetics which you have undoubtedly already studied. By reviewing