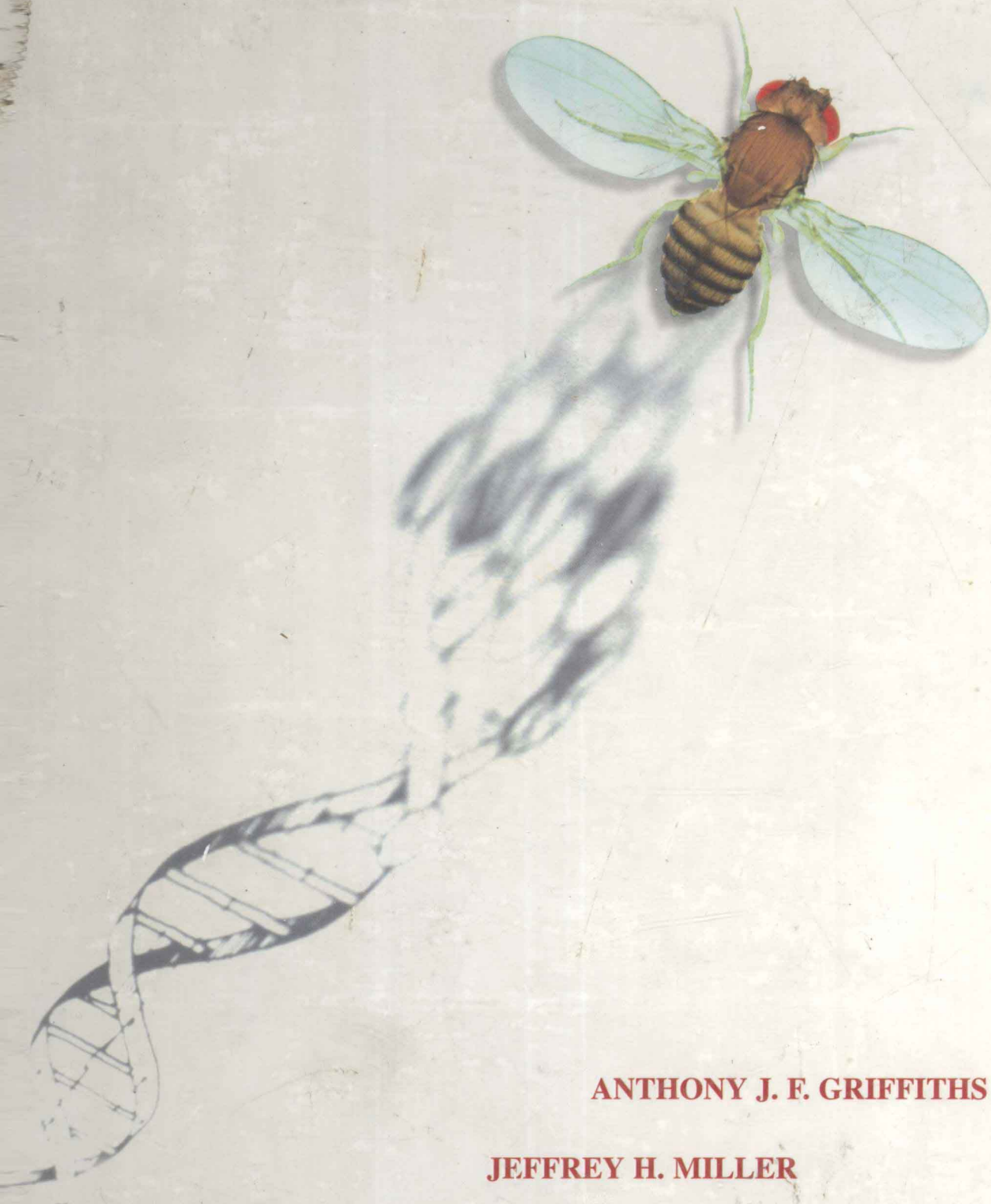

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Preface

Genetics and Education

Genetics has become an indispensable component of almost all research in modern biology and medicine. Research publications investigating any biological process, from the molecular level all the way to the population level, use the "genetic approach" to gain understanding of that process. Thus, no student of the life sciences can afford to be ignorant of the science of genetics.

Genetics has also risen to a position of prominence in human affairs. Special types of plants, animals, and microbes have been developed for human foods, drugs, and myriad other uses. Molecular genetics is the central foundation of the burgeoning biotechnology industry. At the philosophical level, genetics has presented humans with a large number of ethical dilemmas, which regularly surface in the media. Some examples are genetically modified foods, eugenics, privacy of genetic information about individuals, and loss of genetic diversity in nature. Students must be knowledgeable about genetics in order to understand these issues and make informed decisions about them. Lastly, genetic insight has radically affected the human worldview — the way we see ourselves in relation to other organisms.

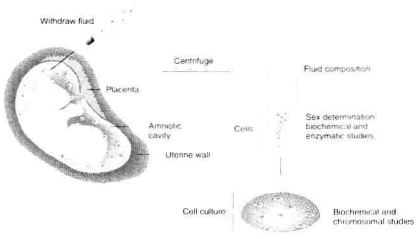


Figure 13-28

Figure 13-18

The Balanced Approach to Teaching Genetics

Genetics has risen to such prominence through the powerful merger of classical and molecular approaches. Each analytical approach has its unique strengths: classical (organismal) genetics is unparalleled in its ability to explore uncharted biological terrain; molecular genetics is equally unparalleled in its ability to unravel cellular mechanisms. It would be unthinkable to teach one without the other, and each is given due prominence in this book. Armed with both approaches, students are able to form an integrated view of genetic principles.

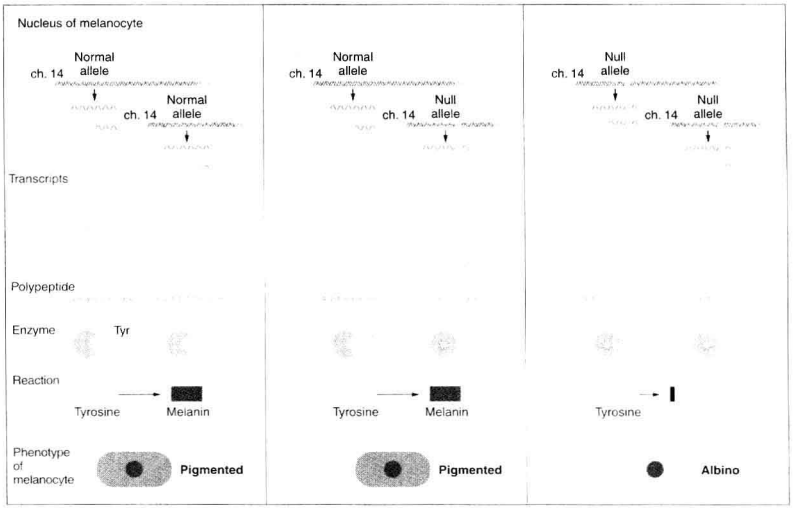


Figure 1-15

The partnership of classical and molecular genetics has always presented a teaching dilemma: which of the two partners should the student be introduced to first, the classical or the molecular? We believe that students begin much as biologists did at the turn of the century, asking general questions about the laws governing heredity. Therefore, the first half of the book introduces the intellectual framework of classical eukaryotic genetics in more or less historical sequence. However, molecular information is provided where appropriate. Our students' knowledge base and our years of teaching have together caused us to rethink the traditional organization. In this new edition we have integrated a good deal of molecular genetics into the early chapters. Thus, we reinforce the students' knowledge of DNA structure and function and avoid presenting the gene as an abstraction. The second half of the book pursues the details of molecular genetics.

Focus on Genetic Analysis

True to its title, the theme of this book is genetic analysis. This theme emphasizes our belief that the best way to understand genetics is by learning how genetic inference is made. On almost every page, we recreate the landmark experiments in genetics and have the students analyze the data and draw conclusions as if they had done the research themselves. This proactive process teaches students how to think like scientists. The modes of inference and the techniques of analysis are the keys to future exploration.

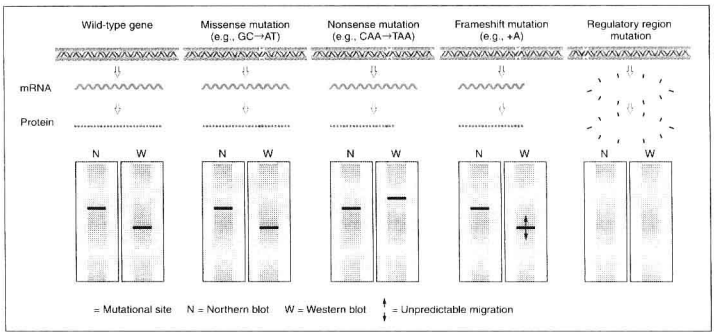


Figure 15-2

Similarly, quantitative analysis is central to the book because many of the new ideas in genetics, from the original conception of the gene to such modern techniques as SSLP mapping, are based on quantitative analysis. The problems at the end of each chapter provide students with the opportunity to test their understanding in quantitative analyses that effectively simulate the act of doing genetics.

Fostering Analytical Skills

A great deal of effort has been put into encouraging students to practice and hone their analytical and problem-solving skills. We provide a great variety of solved and unsolved problems and a wide range of study aids.

The **Problems** section continues to be one of the strengths of the book. Problems are generally arranged to start from simple and proceed to the more difficult. Particularly challenging problems are marked with an asterisk. All problems have been classroom tested. Answers to selected problems are found at the back of the book, and the full set of solutions is in the Student Companion, all prepared by Bill Fixsen (Harvard University).

54. To understand the genetic basis of locomotion in the diploid nematode *Caenorhabditis elegans*, recessive mutations were obtained, all making the worm "wiggle" ineffectually instead of moving with its usual smooth gliding motion. These mutations presumably affect the nervous or muscle systems. Twelve homozygous mutants were intercrossed, and the F_1 hybrids were examined to see if they wiggled. The results were as follows, where a plus sign means that the F_1 hybrid was wild type (gliding) and "w" means that the hybrid wiggled.

	1	2	3	4	5	6	7	8	9	10	11	12
1	w	+	+	+	w	+	+	+	+	+	+	+
2		w	+	+	+	w	+	w	+	w	+	+
3			w	w	+	+	+	+	+	+	+	+
4				w	+	+	+	+	+	+	+	+
5					w	+	+	+	+	+	+	+
6						w	+	w	+	w	+	+
7							w	+	+	+	w	w
8								w	+	w	+	+
9									w	+	+	+
10										w	+	+
11											w	w
12												w

a. Explain what this experiment was designed to test.

b. Use this reasoning to assign genotypes to all 12 mutants.

20. In *Drosophila*, a cross (cross 1) is made between two mutant flies, one homozygous for the recessive mutation bent wing (*b*) and the other homozygous for the recessive mutation eyeless (*e*). The mutations *e* and *b* are alleles of two different genes that are known to be very closely linked on the tiny autosomal chromosome 4. All the progeny were wild-type phenotype. One of the female progeny was crossed to a male of genotype *b e*/*b e*; call this cross 2. The progeny of cross 2 were mostly of the expected types, but there was also one rare female of wild-type phenotype.

a. Explain what the common progeny are expected to be from cross 2.

b. Could the rare wild-type female have arisen by

(1) crossing-over?

(2) nondisjunction?

Explain.

c. The rare wild-type female was testcrossed to a male of genotype *b e*/*b e* (cross 3). The progeny were

$\frac{1}{6}$ wild type
$\frac{1}{6}$ bent, eyeless
$\frac{1}{3}$ bent
$\frac{1}{3}$ eyeless

Which of the explanations in part b are compatible with this result? Explain the genotypes and phenotypes of progeny of cross 3 and their proportions.

Unpacking the Problem

1. Define *homozygous, mutation, allele, closely linked, recessive, wild type, crossing-over, nondisjunction, testcross, phenotype,* and *genotype.*

2. Does this problem concern sex linkage? Explain.

3. How many chromosomes does *Drosophila* have?

4. Draw a clear pedigree summarizing the results of crosses 1, 2, and 3.

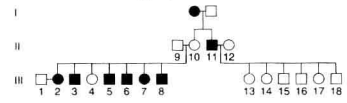
Most chapters have an exercise in problem solving called **Unpacking the Problem.** This exercise grew from the idea that a genetics problem represents only the tip of a vast iceberg of knowledge (we originally considered calling them "iceberg problems"). It is only when the underlying levels of knowledge are exposed that the problem can be solved in a constructive manner. The unpacking activities access this underlying knowledge without actually solving the problem. Some of the component questions in the unpacking exercise might sound trivial, but often these address the kind of fundamental levels of misunderstanding that prevent students from successfully solving problems.

The problems at the end of each chapter are prefaced by **Solved Problems** that illustrate the ways that geneticists apply principles to experimental data. Research in science education has shown that this application of principles is a process that professionals find second nature, whereas students find it a major stumbling block. The Solved Problems demonstrate this process and prepare the students for solving problems on their own.

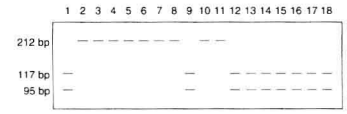
The **Chapter Integration Problems** are solved problems that emphasize concept integration both within and between chapters. These chapter integration problems help to show how one set of learned skills builds on and interacts with previous ones. They also enable students to develop a holistic perspective as they begin to organize diverse concepts into a coherent body of knowledge.

Chapter Integration Problem

1. The human pedigree below concerns a rare visual abnormality in which the person affected loses central vision while retaining peripheral vision.



a. What inheritance pattern is shown? Can it be explained by nuclear inheritance? Mitochondrial inheritance? Molecular geneticists studied the mitochondrial DNA of the 18 members of generations II and III. A restriction fragment 212 base pairs long of the mtDNA from each person was digested with another restriction enzyme, *Sfa*N1, with the following results:



b. What inheritance pattern is shown by these restriction fragments?

c. How does the restriction-pattern inheritance relate to the inheritance of the disease?

d. How can you explain individuals 4 and 10?

e. What is the likely nature of the mutation?

f. How would this analysis be useful in counseling this family?

◆ Solution ◆

a. On the basis of the pedigree alone, it is possible, but unlikely, that the disease is caused by a dominant nuclear allele. But we would have to invoke lack of penetrance in individual 10, who would have to carry the allele because it is passed on to her children. In addition, we have to explain the ratios in generation III. The matings 9 × 10 and 11 × 12 would have to be $A/a \times a/a$, and the phenotypic ratio of affected to normal then expected among the children in each family is 1:1. So overall this model is not an attractive one for explaining the results.

Solved Problems

2. A yeast plasmid carrying the yeast *leu2*$^+$ gene is used to transform nonrevertible haploid *leu2*$^-$ yeast cells. Several *leu*$^+$-transformed colonies appear on a medium lacking leucine. Thus, *leu2*$^+$ DNA presumably has entered the recipient cells, but now we have to decide what has happened to it inside these cells. Crosses of transformants to *leu2*$^-$ testers reveal that there are three types of transformants, A, B, and C, representing three different fates of the *leu2*$^+$ gene in the transformation. The results are:

Type A × *leu2*$^-$ ⟶ ½ *leu*$^+$
½ *leu*$^-$, × standard *leu2*$^-$
⟶ ¼ *leu*$^+$
¼ *leu*$^-$

Type B × *leu2*$^-$ ⟶ ½ *leu*$^+$
½ *leu*$^-$, × standard *leu2*$^-$
⟶ 100% *leu*$^+$
0% *leu*$^-$

Type C × *leu2*$^-$ ⟶ 100% *leu*$^+$

What three different fates of the *leu2*$^+$ DNA do these results suggest? Be sure to explain *all* the results according to your hypotheses. Use diagrams if possible.

◆ Solution ◆

If the yeast plasmid does not integrate, then it replicates independently of the chromosomes. In meiosis, the daughter plasmids would be distributed to the daughter cells, resulting in 100 percent transmission. This percentage was observed in transformant type C.

If one copy of the plasmid is inserted, in a cross with a *leu2*$^-$ line, the resulting offspring would have a ratio of 1 *leu*$^+$:1 *leu*$^-$. This ratio is seen in type A and type B.

When the resulting *leu*$^+$ cells are crossed with standard *leu2*$^-$ lines, the data from type A cells suggest that the inserted gene is segregating independently of the standard *leu2*$^+$ gene; so the *leu2*$^+$ transgene has inserted ectopically into another chromosome.

Key Concepts

When two haploid genomes containing different recessive mutations are combined in one cell and the phenotype is mutant, the mutations must be in the same gene (alleles).

When two haploid genomes containing different recessive mutations are combined in one cell and the phenotype is wild type, the mutations must be in different genes.

The phenotypes of some heterozygotes reveal types of dominance other than full dominance.

Some mutant alleles can kill the organism.

Most characters are determined by sets of genes that interact with one another and with the environment.

Modified monohybrid ratios reveal allelic interactions.

Modified dihybrid ratios reveal gene interactions.

The **Key Concepts** at the chapter openings give an overview of the main principles to be covered in the chapter, stated in simple prose without genetic terminology. These provide a strong pedagogic direction for the reader.

MESSAGE

The linear sequence of a protein folds up to yield a unique three-dimensional configuration. This configuration creates specific sites to which substrates bind and at which catalytic reactions take place. The three-dimensional structure of a protein, which is crucial for its function, is determined solely by the primary structure (linear sequence) of amino acids. Therefore, genes can control enzyme function by controlling the primary structure of proteins.

Throughout the chapters, boxed **Messages** provide convenient milestones at which the reader can pause and contemplate the material just presented.

Summary

After making dihybrid crosses of sweet pea plants, William Bateson and R. C. Punnett discovered deviations from the 9:3:3:1 ratio of phenotypes expected in the F_2 generation. The parental gametic types outnumbered the other two classes. Later, in his studies of two different autosomal genes in *Drosophila*, Thomas Hunt Morgan found a similar deviation from Mendel's law of independent assortment. Morgan postulated that the two genes were located on the same pair of homologous chromosomes. This relation is called linkage.

Linkage explains why the parental gene combinations stay together but not how the nonparental combinations arise. Morgan postulated that in meiosis there may be a physical exchange of chromosome parts by a process now called crossing-over. Thus, there are two types of meiotic recombination. Recombination by Mendelian independent assortment results in a recombinant frequency of 50 percent.

ing-over results in a recombinant frequency ev

Chapter **Summaries** provide a short distillation of the chapter material and an immediate reinforcement of the concepts. All these items are useful in text review, especially for exam study.

Now draw a concept map for this chapter, interrelating as many of the following terms as possible. Note that the terms are listed in no particular order.

genotype / phenotype / norm of reaction / environment / development / organism

A **Concept Map** exercise appears at the end of every chapter. Concept maps grew out of the constructivist movement in education, which asserts that student learning is most effective when new information is brought into direct conflict with previous understanding. Concept mapping can be a powerful method for visualizing and resolving such conflicts, while aiding concept integration.

New Features of the Seventh Edition

New emphasis on the integration of classical and molecular genetics

The linearity of the teaching process means that concepts have to be introduced one at a time to avoid bewilderment. Nevertheless, genetics is an integrated subject in which organismal and molecular manipulations go hand in hand. Therefore, integration is a key issue in teaching and a key goal in learning. This edition integrates organismal and molecular aspects of genetics wherever possible, starting in the early chapters. Chapter 1 presents the basics of DNA structure and function and uses albinism in humans as an example to establish the relationship between DNA, genes, proteins, and phenotype. In Chapter 2 we examine patterns of inheritance (both Mendelian and non-Mendelian) and explain them at the molecular level, using examples such as PKU. The complementarity of the two genetic approaches continues with the exploration of gene interactions at the molecular level, as exemplified by flower color in Blue-eyed Mary and sickle cell anemia in humans (Chapter 4).

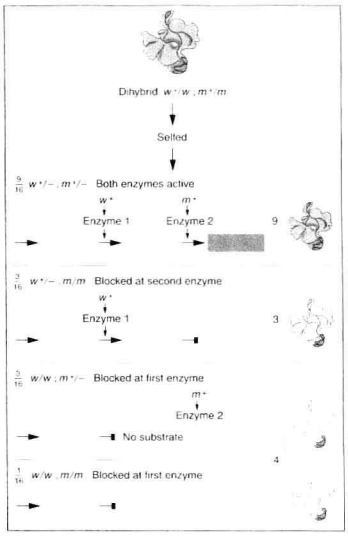


Figure 4-13

New chapter sequence for greater topic cohesiveness

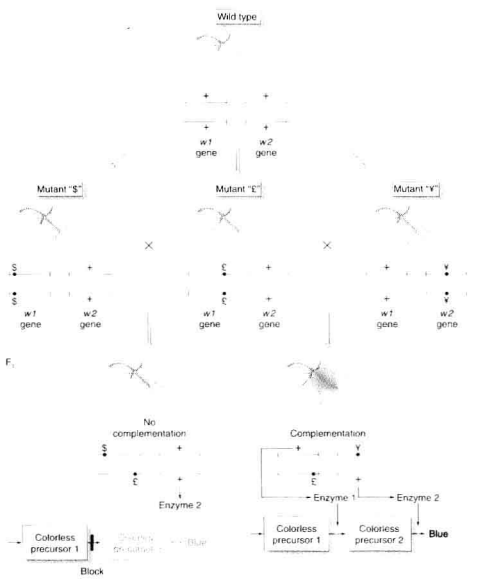


Figure 4-1

Our goal to better emphasize overarching principles has resulted in the relocation of several chapters. The book now consists of six major blocks that group topics related by common underlying principles: general aspects of inheritance are covered in Chapters 1–4; recombination and mapping of genes in Chapters 5–7; molecular genetics in Chapters 8–14; genetic change and variation in Chapters 15–20; developmental genetics in Chapters 21–23; and population genetic analysis in Chapters 24–26.

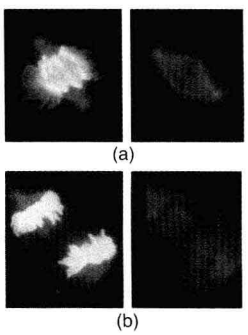


Figure 3-28

The benefits of this reorganization can be found throughout the text. Chapter 2, for instance, now covers both Mendelian genetics and sex-linked inheritance in order to emphasize general patterns of inheritance. Similarly, the incorporation of eukaryotic genome structure with the chromosomal basis of heredity in Chapter 3 provides mutual reinforcement of principles. Chapter 4's new title, Gene Interaction, reflects its inclusion of new material on complementation and its new emphasis on overarching principles of how genes interact. Chapter 11 now juxtaposes prokaryotic and eukaryotic gene regulation so that principles common to both processes emerge. Chapter 23, Developmental Genetics, now includes coverage of gene regulation during development, a topic previously covered in a separate chapter.

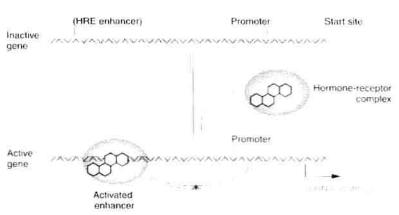


Figure 11-37

New streamlined and simplified explanations

In an effort to focus on overarching principles, we have examined the level of detail in many chapters and have chosen to simplify and streamline the coverage of several topics. This is particularly noticeable in Chapter 7 (Gene Transfer in Bacteria and Their Viruses), Chapter 9 (Genetics of DNA Function), Chapter 16 (Mechanisms of Gene Mutation), Chapter 19 (Mechanisms of Recombination), and Chapter 21 (Extranuclear Genes).

Two new chapters and updates throughout

Two new chapters have been introduced: Chapter 22, on Cancer as a Genetic Disease, and Chapter 26, on Evolutionary Genetics. Chapter 22 presents the integrated control mechanisms of cell proliferation and cell death and what happens when these mechanisms are disrupted. Chapter 26 discusses the evolutionary process in terms of both natural selection and random factors and includes sections on speciation and the origin of new genes. Notable updates in other chapters are: lod scores (Chapter 5); rolling circle replication and synteny (Chapter 8); functional genomics including yeast 2-hybrid analysis, micro arrays/DNA chips, and global regulation (Chapter 14); the molecular basis of chromosome rearrangements (Chapter 17); mitochondrial DNA, aging and human disease (Chapter 21); and programmed cell death (Chapter 24).

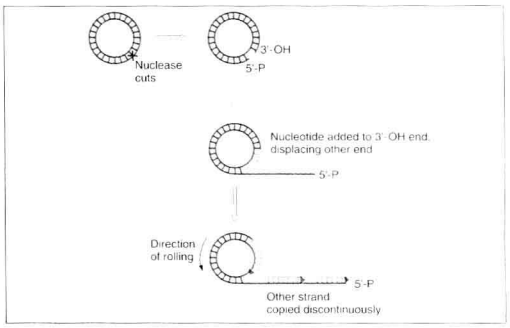


Figure 8-19

New problems

More than 50 new problems have been added, including many problems involving molecular analysis.

55. In corn, synthesis of purple pigment is controlled by two genes acting sequentially through colorless (white) intermediates:

$$\text{white 1} \xrightarrow{\text{gene } A} \text{white 2} \xrightarrow{\text{gene } B} \text{purple}$$

Recessive nonsense mutations (a^n and b^n) were obtained in genes *A* and *B*. Each of these mutations gave a white phenotype, and each could be suppressed by the nonsense-suppressor mutation T^s (wild-type allele T^+)

a. Would you expect T^s to be dominant to T^+? Explain.

b. A trihybrid A/a^n ; B/b^n ; T^s/T^+ is selfed. If all the genes are unlinked, what phenotypic ratio do you expect in the progeny? Explain, preferably with a diagram.

56. A plant believed to be heterozygous for a pair of alleles *B*/*b* (where *B* encodes yellow and *b* encodes bronze) was selfed and in the progeny there were 280 yellow and 120 bronze individuals. Do these results support the hypothesis that the plant is *B*/*b*?

57. A plant thought to be heterozygous for two independently assorting genes (*P*/*p* ; *Q*/*q*) was [illegible]

Supplements

The following supplementary materials are available to accompany *Introduction to Genetic Analysis.*

Solutions Manual
William Fixsen, Harvard University, 0-7167-3525-3
The *Solutions Manual* contains worked-out solutions to all the problems in the textbook.

Introduction to Genetic Analysis CD-ROM
(hybrid format for Windows and Macintosh)
Packaged with every copy of the textbook, this CD has over 30 original animations that are available in two formats. Topics such as transcription, complementation, and DNA replication bring the textbook figures to life. Students can view each animation as a series of steps that make up the process or watch the animation in its entirety.

Text and media come together by denoting figures of genetic processes that come to life as animations on the Freeman Genetics CD-ROM.

Introduction to Genetic Analysis Web Site
W. H. Freeman and Sumanas, Inc., with contributions from William Sofer, The State University of New Jersey at Rutgers

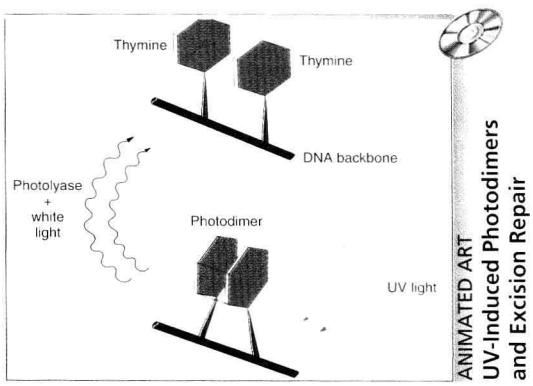


Figure 16-25

This multimedia learning tool complements and enriches the textbook. Practice tools such as interactive quizzes, tutorials, flashcards, key concepts, and Web links in every chapter help students review for exams. All text images will be available for downloading. The Introduction to Genetic Analysis Web site, at *www.whfreeman.com/iga/* will be updated regularly.

Essays on Genetics Education

Understanding Genetics: Ideas for Teachers

Anthony J. F. Griffiths and Jolie A. Mayer-Smith, both of the University of British Columbia

This biweekly electronic publication explores the problems faced by instructors in genetics education and offers creative and thoughtful strategies for promoting student understanding of genetics and learning in general. A complete collection of the essays will be available for purchase early in 2000.

Instructor Resource Manual and Test Bank

Sally Allen, University of Michigan, and Ewan Harrison

Printed: 0-7167-3530X; CD ROM: 0-7167-3528-8

The *Instructor Resource Manual* contains over 700 test questions in multiple-choice, true-false, and matching formats. It also contains complete sample exams and teaching hints. The electronic version (both Windows and Mac formats on one CD) allows instructors to download, edit, add, and re-sequence questions to suit their particular needs.

Online Testing

With Diploma, the computerized test bank package from Brownstone Research Group, instructors can create and administer exams on paper, over a network, and now, over the Internet as well. Instructors can include multimedia, graphics, movies, and sound in their questions. Security features allow instructors to restrict tests to specific computers or time blocks. The package also includes an impressive suite of grade book and question-analysis features.

Instructor's Resource CD-ROM

0-7167-3952-6

Our instructor's CD offers all the images from the textbook and the animations in two formats — as part of our Presentation Manager Pro software and in JPEG files. Presentation Manager lets instructors quickly prepare play lists of images for display during lectures. The JPEG files are for instructors who use commercially available presentation software.

Transparency Set

0-7167-3526-1

A full-color overhead transparency set of 150 key illustrations from the textbook is available free of charge to qualified adopters.

Course Syllabi

For a two-semester course, the entire text provides an appropriate course structure and syllabus that reflects the range of modern genetics. A syllabus for a one-semester course can be designed around selected chapters. One possible selection of chapters for a one-semester course is Chapters 1, 2, 3, 4, 5, 8, 11, 12, 15, 17, 18, 23, and 24. A one-semester course in molecular genetics could be based on Chapters 8 through 23.

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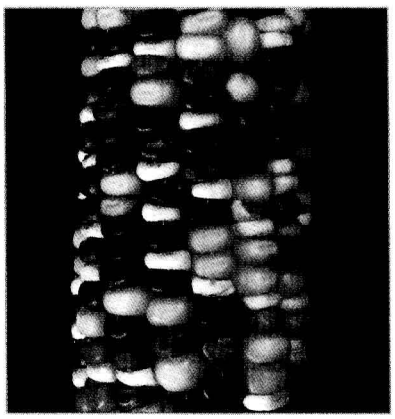
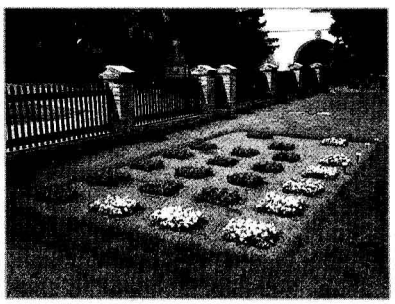
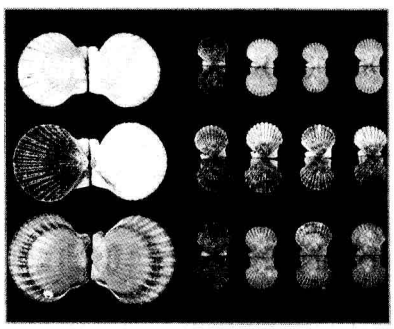
David E. Sheppard	*University of Delaware*
Laurence Von Kalm	*University of Central Florida*
Bruce Walsh	*University of Arizona*
Edmund J. Zimmerer	*Murray State University*

We believe this edition to be a true celebration of genetics. As authors, we hope that our love of the subject comes through and that the book will stimulate the reader to do some firsthand genetics, whether as professional scientist, student, amateur breeder, or naturalist. Failing this, we hope to impart some lasting impression of the incisiveness, elegance, and power of genetic analysis.

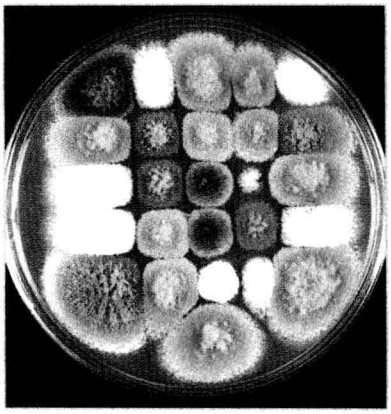
Contents

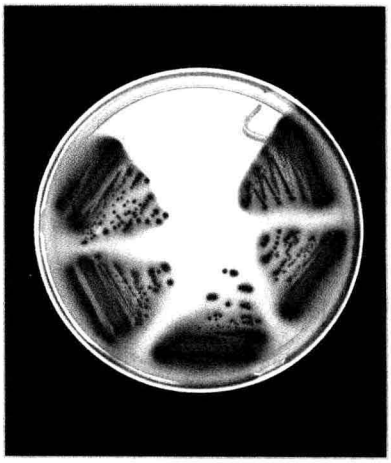


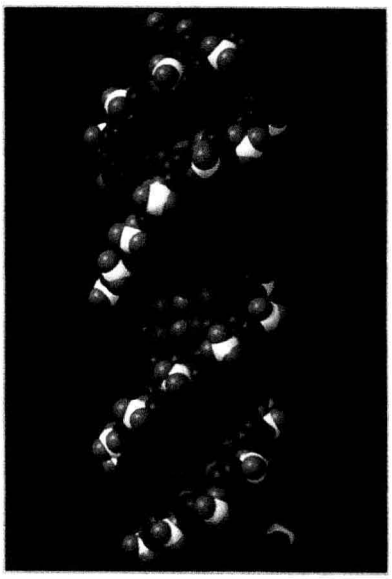


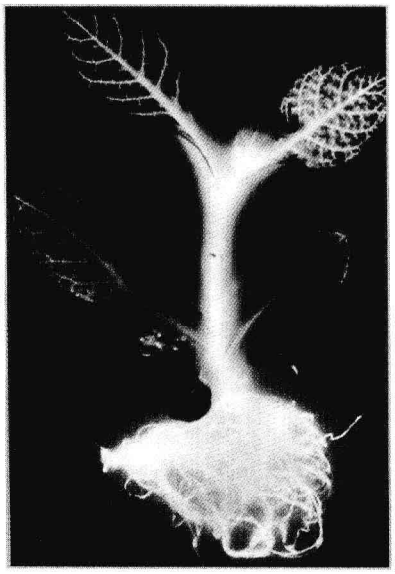


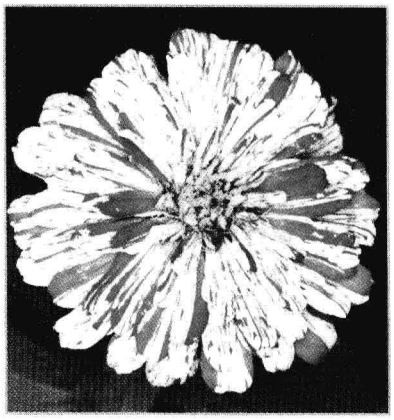
Contents in Brief

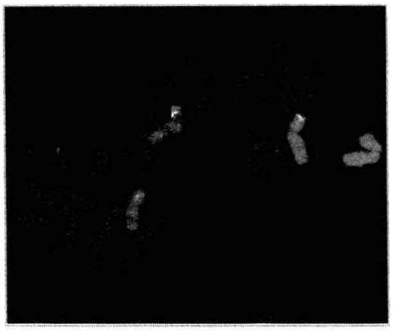


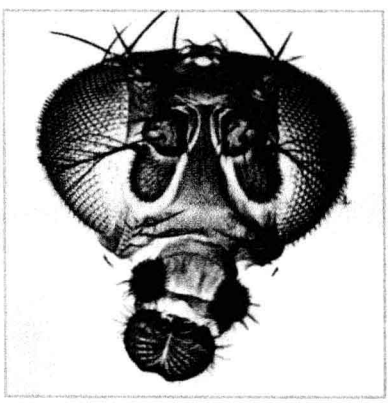


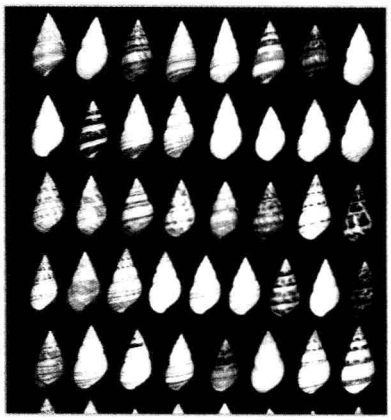




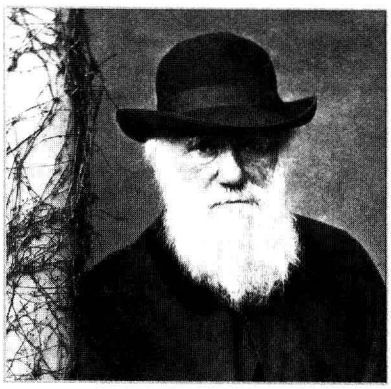






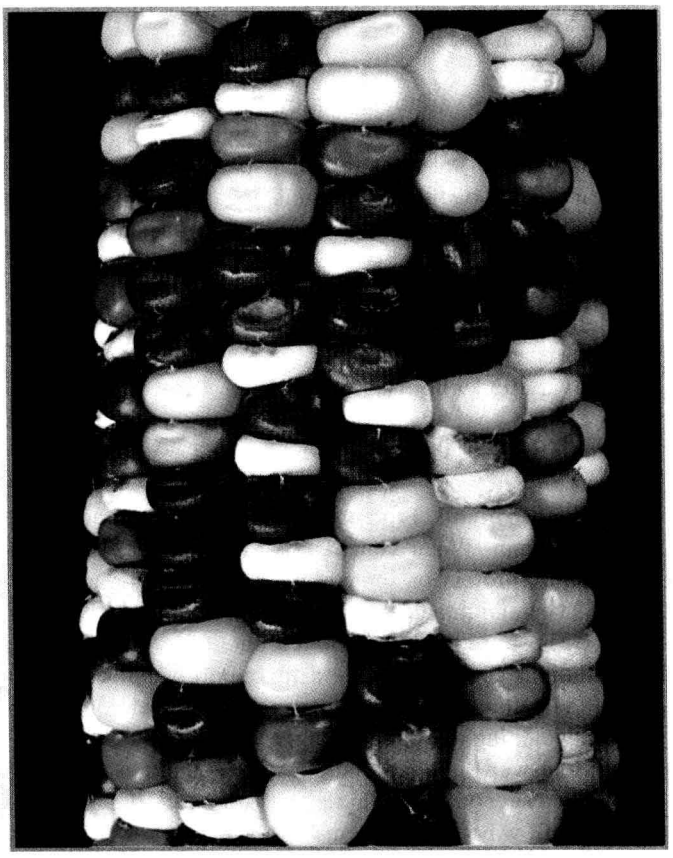


GENES AT THE POPULATION LEVEL



1

GENETICS AND THE ORGANISM



Genetic variation in the color of corn kernels.
Each kernel represents a separate individual with a distinct genetic makeup. The photograph symbolizes the history of humanity's interest in heredity. Humans were breeding corn thousands of years before the advent of the modern discipline of genetics. Extending this heritage, corn today is one of the main research organisms in classical and molecular genetics.
(William Sheridan, University of North Dakota; photograph by Travis Amos.)

Key Concepts

The hereditary material is DNA.

DNA is a double helix composed of two intertwined nucleotide chains oriented in opposite directions.

In the copying of DNA, the chains separate and serve as molds for making two identical daughter DNA molecules.

The functional units of DNA are genes.

A gene is a segment of DNA that can be copied to make RNA.

The nucleotide sequence in RNA is translated into the amino acid sequence of a protein.

Proteins are the main determinants of the basic structural and physiological properties of an organism.

The characteristics of a species are encoded by its genes.

Variation within a species may be from hereditary variation, environmental variation, or both.

Hereditary variation is caused by variant forms of genes (alleles).

Why study genetics? There are two basic reasons. First, genetics occupies a pivotal position in the entire subject of biology. Therefore, for any serious student of plant, animal, or microbial life, an understanding of genetics is essential. Second, genetics, like no other scientific discipline, is central to numerous aspects of human affairs. It touches our humanity in many different ways. Indeed, genetic issues seem to surface daily in our lives, and no thinking person can afford to be ignorant of its discoveries. In this chapter, we take an overview of the science of genetics, showing how it has come to occupy its crucial position. In addition, we provide a perspective from which to view the subsequent chapters.

First, we need to define what genetics is. Some define it as the study of heredity, but hereditary phenomena were of interest to humans long before biology or genetics existed as the scientific disciplines that we know today. Ancient peoples were improving plant crops and domesticated animals by selecting desirable individuals for breeding. They also must have puzzled about the inheritance of individuality in humans and asked such questions as, "Why do children resemble their parents?" and "How can various diseases run in families?" But these people could not be called geneticists. Genetics as a set of principles and analytical procedures did not begin until the 1860s, when an Augustinian monk named Gregor Mendel (Figure 1-1) performed a set of experiments that pointed to the existence of biological elements that we now call genes. The word *genetics* comes from the word "gene," and genes are the focus of the subject. Whether geneticists study at the molecular, cellular, organismal, family, population, or evolutionary level, genes are always central in their studies. Simply stated, genetics is the study of genes.

Figure 1-1 Gregor Mendel. (Moravian Museum, Brno.)

What is a gene? A gene is a section of a threadlike double helical molecule called **deoxyribonucleic acid,** abbreviated **DNA.** The discovery of genes and understanding their molecular structure and function have been sources of profound insight into two of the biggest mysteries of biology:

1. What makes a species what it is? We know that cats always have kittens and people always have babies. This common-sense observation naturally leads to questions about the determination of the properties of a species. The determination must be hereditary because, for example, the ability to have kittens is inherited by every generation of cats.

2. What causes variation within a species? We can distinguish each other as well as our own pet cat from other cats. Such differences within a species require explanation. Some of these distinguishing features are clearly familial; for example, animals of a certain unique color often have offspring with the same color, and, in human families, certain features such as the shape of the nose definitely "run in the family." Hence we might suspect that a hereditary component explains at least some of the variation within a species.

The answer to the first question is that genes dictate the inherent properties of a species. The products of most genes are specific **proteins.** Proteins are the main macromolecules of an organism. When you look at an organism, what you see is either protein or something that has been made by a protein. The amino acid sequence of a protein is encoded in a gene. The timing and rate of production of proteins and other cellular components are a function both of the genes within the cells and of the environment in which the organism is developing and functioning.

The answer to the second question is that any one gene can exist in several forms that differ from each other, generally in small ways. These forms of a gene are called **alleles.** Allelic variation causes hereditary variation within a species. At the protein level, allelic variation becomes protein variation.

The next two sections show how genes influence the inherent properties of a species and how allelic variation contributes to variation within a species. These sections are an overview; most of the details will be presented in later chapters.

Genes as determinants of the inherent properties of species

What is the nature of genes, and how do they perform their biological roles? Three fundamental properties are required of genes and the DNA of which they are composed.

1. *Replication.* Hereditary molecules must be capable of being copied at two key stages of the life cycle (Figure 1-2). The first stage is the production of the cell type that will ensure the continuation of a species from one generation to the next. In plants and animals, these cells are the gametes: egg and sperm. The other stage is when the first cell of a new organism undergoes multiple rounds of division to produce a multicellular organism. In plants and animals, this is the stage at which the fertilized egg, the **zygote,** divides repeatedly to produce the complex organismal appearance that we recognize.
2. *Generation of form.* The working structures that make up an organism can be thought of as form or substance. Looked at in this way, DNA has essential "information"; in other words, "that which is needed to give form."
3. *Mutation.* A gene that has changed from one allelic form into another has undergone mutation — an event that happens rarely but regularly. Mutation is not only a basis for variation within a species, but, over the long term, also the raw material for evolution.

We will examine replication and the generation of form in this section and mutation in the next.

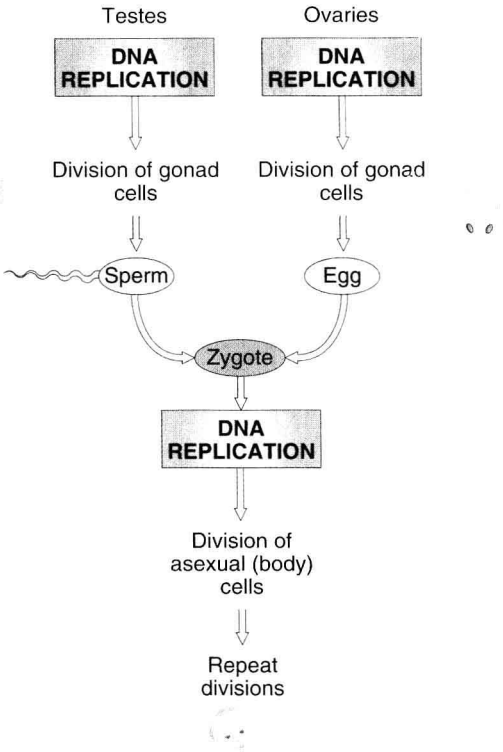


Figure 1-2 DNA replication is the basis of the perpetuation of life through time.

DNA and its replication

An organism's basic complement of DNA is called its **genome.** The body cells of most plants and animals contain two genomes (Figure 1-3). These organisms are **diploid.** The cells of most fungi, algae, and bacteria contain just one genome. These organisms are **haploid.** The genome itself is made up of one or more extremely long molecules of DNA that are organized into **chromosomes.** For instance, human body cells contain two sets of 23 chromosomes, for a total of 46. Genes are simply the functional regions of chromosomal DNA. Each chromosome in the genome carries a different array of genes. In diploid cells, each chromosome and its component genes are present twice. Two chromosomes with the same gene array are said to be **homologous.** When a cell divides, all the chromosomes (one or two genomes) are replicated; so each daughter cell contains the full complement. Therefore, the unit of replication is the chromosome; when a chromosome is replicated, all the genes of that chromosome are automatically replicated along with it.

To understand replication, we need to understand the basic nature of DNA. DNA is a linear, double-helical structure looking rather like a molecular spiral staircase. The double helix is composed of two intertwined chains of building blocks called **nucleotides.** Each nucleotide consists

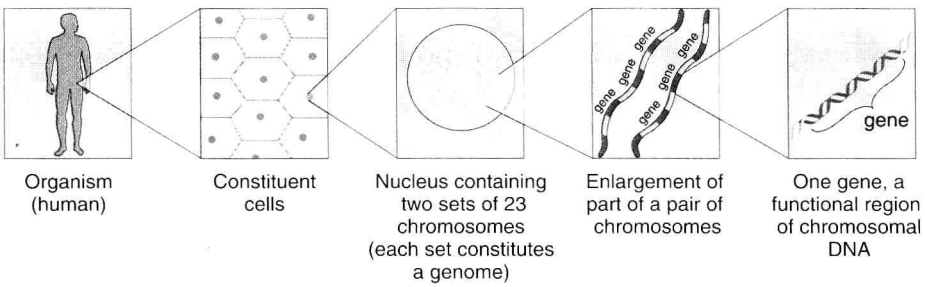


Figure 1-3 Successive enlargements bringing the genetic material of an organism into sharper focus.